CENTRE FOR COMMUNITY-DRIVEN RESEARCH

Personal Experience, Expectations and Knowledge (PEEK)

Rare and genetic conditions

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Thank you to each and every person that contributed information to this report.

CCDR dedicates this study to Heather Renton, founder of SWAN Australia in recognition of her service to the rare, genetic and undiagnosed conditions community.

PEEK study process information

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Summary of results

Executive summary

In this PEEK study, a total of 407 participants with rare diseases or carers to people with rare diseases were recruited into the study. The majority of participants lived in major cities, they lived in all levels of economic advantage. Most of the of participants identified as Caucasian/white, aged mostly between 35 and 64. Half of the participants had completed some university, and most were employed either full time or part time. Almost half of the participants were carers to family members or spouses.

Physical health interfered with work and other activities for participants in this study, they had poor energy levels and poor general health.

This is a group that had health conditions other than their condition to deal with, most often anxiety, sleep problems, and chronic pain.

Most participants sought medical attention after noticing symptoms and were diagnosed after their a complex pathway involving a number of specialists.

This is a cohort that was diagnosed by a specialist at a specialist clinic or in hospital. The majority did not have any out of pocket expenses at diagnosis, however, for those that did have out of pocket expenses it was a moderately significant burden.
This is a group that did not have enough emotional support at the time of diagnosis. This is a cohort that did not have conversations about biomarker/genomic/gene testing, though are interested in these types of tests.

This is a study cohort that had no or limited knowledge about their condition before they were diagnosed. This patient population that had uncertainty about their prognosis, or described their prognosis in terms of symptoms and function or changes in symptoms and function.

This is a patient population that had no discussions about treatment or were given multiple treatment options. Some participated in decision making but others were told what to do without discussion.

This is a study cohort that took into account side effects and efficacy as part of many considerations when making decisions about treatment.

Within this patient population, about half of the participants had changed decision making over time, this was linked to being more informed and assertive.

When asked about their personal goals of treatment or care participants most commonly described wanting quality of life or return to normality.

This is a group who felt they were mostly treated with respect throughout their experience.

Approximately two-thirds of this cohort had private health insurance, half were public patients treated in mostly in the public hospital system. This is a group that did not have trouble paying for healthcare appointments, prescriptions, and paying for basic essentials. Their monthly expenses due to their condition were somewhat of a burden.

Participants in this study had to quit, reduce hours, or take leave from work. Carers and family did not have to change employment status. The loss of family income was a burden.

Participants on average used one allied health service, one complementary therapy and made one lifestyle change.

More than a third had conversations about clinical trials, and the majority would take part in a clinical trial if there was a suitable one for them.

This is a patient population that described mild side effects using an example such as fatigue and as those which can be self-managed and do not interfere with daily life.

This is a study cohort that described severe side effects as symptoms such as pain, they also described severe side effects as those that impact everyday life and the ability to conduct activities of daily living.

This is a patient population which described adhering to treatments according to the advice or their doctor or that they would stick with it for 2 to 3 months. This is a study cohort that needed to see physical signs disappear to feel that treatment is working as well. If treatment did work, it would allow them to return to everyday activities

Participants in this study had very good knowledge about their condition, were average at coping with their condition, were good at recognizing and managing symptoms, and were very good at adhering to treatment.

Participants were given information about treatment options, disease management and , disease cause from health care professionals, and searched for the same topics most often. This is a group who accessed information from non-profit, charity or patient organisations most often.

This is a patient population that access information primarily through the internet, Facebook or social media, and from health charities.

This is a study cohort that found information from other people's experience to be helpful, and that no information was unhelpful.

This is a group that preferred online information, or talking to someone. This is a study cohort that generally felt most receptive to information from the beginning, at diagnosis.

Most participants described receiving an overall positive experience with health professional communication (some with a few exceptions) which was holistic, two way and comprehensive. For those that had a negative experience it was mostly because their healthcare professionals had a lack of knowledge about their condition.

The participants in this study experienced good quality of care, and average coordination of care. They had a moderate ability to navigate the healthcare system, and experienced moderate communication from healthcare professionals.

This is a patient population that did not have any formal support or found support in the clinical setting or from family and friends.

This is a patient population that experienced a negative impact on quality of life largely due to emotional strain on family, and changes to relationships.

Life was a little distressing for this group, due to having a rare disease

This is a study cohort that experienced at least some impact on their mental health and to maintain their mental health they used coping strategies such as consulting a mental health professional or remaining social, lifestyle changes and hobbies.

Within this patient population, participants described the importance of self-care, and complying with treatment in order to maintain their general health.

Participants in this study had felt vulnerable when having sensitive discussion about their condition. To manage vulnerability, they used self help methods such as resilience, acceptance and staying positive.

This cohort most commonly felt there was an overall negative impact on their relationships, due to people withdrawing from relationships or not knowing what to say.

Participants felt they were a burden on their family, due the extra household duties and responsibilities that their family must take on.

Most participants felt there was some cost burden which was from the costs of taking time off work and from the cost of treatments.
The participants in this PEEK study had moderate levels of anxiety in relation to their condition.

Participants would like future treatments to be more affordable, and more effective.

This is a study cohort that would like information to be more accessible and to provide more information about disease trajectory.

Participants in this study would like future communication to include health professionals with a better knowledge of their condition, and for more empathy.

Participants would like future treatments to include access to appropriate real-world support services.

This patient population was grateful for healthcare staff, including access to specialists.

Participants' message to decision-makers was the need for timely and equitable access to support, care and treatment

This is a patient population that wished had been more assertive, been an advocate, more informed and asked questions.

The aspect of care or treatment that participants in this study would most like to change is to accessed their specialist sooner, however, many wouldn't change any aspect of their treatment or care.

Section 1

Introduction and methods

Section 1 Introduction and methodology

Introduction

In Australia, a disease is considered rare if it affects less than 5 in 10,000 people. There are more than 7,000 rare diseases that are life threatening or chronically debilitating. Around 8% of Australians (2 million people) live with a rare disease.

A total of 407 participants with rare diseases or carers to people with rare diseases were recruited into this PEEK study. There were 392 that completed both parts of the study, 5 that completed or partially completed online questionnaire only and 10 participants that completed the interview only.

Personal Experience, Expectations and Knowledge (PEEK)

Patient Experience, Expectations and Knowledge (PEEK) is a research program developed by the Centre for Community-Driven Research (CCDR). The aim of PEEK is to conduct patient experience studies across several disease areas using a protocol that will allow for comparisons over time (both quantitative and qualitative components). PEEK studies give us a clear picture and historical record of what it is like to be a patient at a given point in time, and by asking patients about their expectations, PEEK studies give us a way forward to support patients and their families with treatments, information and care.

The research protocol used in PEEK studies is independently driven by CCDR. PEEK studies include a quantitative and qualitative component. The quantitative component is based on a series of validated tools. The qualitative component is the result of two years of protocol testing by CCDR to develop a structured interview that solicits patient experience data and provides patients with the opportunity to provide advice on what they would like to see in relation to future treatment, information and care. The structured interview has also been designed so that the outcomes of PEEK studies can inform policy, research, care, information, supportive care services and advocacy efforts.

Position of this study

A search was conducted in Pubmed (August 8, 2022) to identify studies of rare diseases with that described patient experience conducted in the past five years in Australia, and updated on January 4th 2023. The term “Rare disease” was searched in any field, and it is noted that not all rare diseases studies will be included using this search term, and the difficulty in searching using individual disease names. Interventional studies, meta-analysis studies, studies conducted in developing countries, and studies of less than five participants were excluded.

There were 201 studies identified, 52 studies used interviews, 30 studies used focus groups or other qualitative methods and 138 studies used questionnaires.

PEEK is largest study of rare diseases conducted in an Australian population with a total of 402 participants with rare diseases or carers to people with rare diseases were recruited into the study. There were 391 that completed or partially completed online questionnaires and 402 participants that were interviewed.

Section 2

Demographics

Section 2 Demographics

Participants

In this PEEK study, a total of 407 participants with rare diseases or carers to people with rare diseases were recruited into the study. There were 5 that completed or partially completed online questionnaires only and 10 participants that completed the interview only. There were 96 participants (23.59%) with diseases of the nervous system, 96 participants (23.59%) with endocrine, nutritional or metabolic diseases, 81 participants (16.71%) with diseases of the immune system, 68 participants (16.71%) with developmental anomalies, 34 participants (7.86%) with other rare condition, and 32 participants (7.86%) with diseases of the skin.

Demographics

There were 407 people with that took part in this study, 299 were females (73.83%). Participants were aged from infant to over 75 years of age, most were aged between 35 to 64 years (n=232, 64.09%).

Participants were most commonly from New South Wales (n=124, 30.47%), Queensland (n=92, 22.60%), and Victoria (n=91, 22.36%). Most participants were from major cities (n=295, 72.48%), and they lived in all levels of advantage, defined by Socio-economic Indexes for Areas (SEIFA) (www.abs.gov.au) with 204 participants (49.88%) from an area with a high SEIFA score of 7 to 10 (more advantage), and 203 participants (50.12%) from an area of mid to low SEIFA scores of 1 to 6 (less advantaged).

There were 201 participants (50.38%) that had completed university to at least an associate degree. There were 163 participants who were employed either full time (24.56%), or part time (23.10%). Almost half of the participants were carers to family members or spouses (n=192, 53.04%), and just under half of the participants carers to children (n=155, 42.82%).

Other health conditions

Participants were asked about health conditions, other than their rare disease that they had to manage. Participants could choose from a list of common health conditions and could specify other conditions.

The majority of participants had at least one other condition that they had to manage (n=287, 93.79%), the maximum number reported was 16 other conditions, with a median of 4.00 other conditions (IQR = 5.00). The most commonly reported health condition was anxiety (n=173, 56.54%), followed by sleep problems or insomnia (n=169, 55.23%), chronic pain (n=154, 50.33%), and depression (n=132, 43.14%).

Subgroup analysis

Comparisons were made by condition. There were 67 participants (16.46%) with developmental anomalies, 82 participants (20.15%) with diseases of the immune system, 99 participants (24.32%) with diseases of the nervous system, 32 participants (7.86%) with diseases of the skin, 95 participants (23.34%) with endocrine, nutritional or metabolic diseases , and 32 participants (7.86%) with other rare condition.

Baseline health

The Short Form Health Survey 36 (SF36) measures baseline health, or the general health of an individual. The SF36 comprises nine scales: physical functioning, role functioning/physical, role functioning/emotional, energy and fatigue, emotional well-being, social function, pain, general health, and health change from one year ago. The scale ranges from 0 to 100, a higher score denotes better health or function.

The overall scores for the cohort were in the second highest quintile for **SF36 Role functioning/emotional** (median=66.67, IQR=100.00), **SF36 Emotional well-being** (median=68.00, IQR=27.00), indicating good emotional role functioning, good emotional well-being.

The overall scores for the cohort were in the middle quintile for **SF36 Physical functioning** (median=55.00, IQR=60.00), **SF36 Social functioning** (median=50.00, IQR=50.00), **SF36 Pain** (median=55.00, IQR=45.00), **SF36 Health change** (median=50.00, IQR=25.00), indicating moderate physical functioning, moderate social functioning, moderate pain, about the same as a year ago.

The overall scores for the cohort were in the second lowest quintile for **SF36 Role functioning/physical** (median=25.00, IQR=100.00), **SF36 Energy/Fatigue** (median=30.00, IQR=35.00), **SF36 General health** (median=40.00, IQR=35.00), indicating poor physical role functioning, poor energy, poor general health.

Comparisons of SF36 have been made based on condition, participant type, gender, age, education, location and socioeconomic status.

SF36 Physical functioning scale measures health limitations in physical activities such as walking, bending, climbing stairs, exercise, and housework. On average, physical activities were moderately limited for participants in this study.

SF36 Role functioning/physical scale measures how physical health interferes with work or other activities. On average, physical health often interfered with work or other activities for participants in this study.

SF36 Role functioning/emotional scale measures how emotional problems interfere with work or other activities. On average, emotional problems sometimes interfered with work or other activities for participants in this study.

SF36 Energy/fatigue scale measures the proportion of energy or fatigue experienced. On average, participants were often fatigued.

The **SF36 Emotional well-being** scale measures how a person feels, for example happy, calm, depressed or anxious. On average, participants had good emotional well-being.

The **SF36 Social functioning** scale measures limitations on social activities due to physical or emotional problems. On average, social activities were moderately limited for participants in this study.

The **SF36 Pain** scale measures how much pain, and how pain interferes with work and other activities. On average, participants had moderate pain.

The **SF36 General health** scale measures perception of health. On average, participants reported poor health.

The **SF36 Health change** scale measures health compared to a year ago. On average, participants reported that their health is about the same as a year ago.

Section 3

Symptoms and diagnosis

Section 3: Symptoms and diagnosis

Symptoms leading to diagnosis

In the structured interview, participants were asked to describe the symptoms that actually *led* to their diagnosis. Most commonly participants strongly recalled their symptoms or how they came to be diagnosed (84.58%). Others had an unclear recollection of their symptoms or how they came to be diagnosed (7.46%), or had no symptoms that they felt specifically led to diagnosis (3.23%).

Symptoms leading to diagnosis: Seeking medical attention

Participants described when they sought medical attention after noticing symptoms. The most common responses were having symptoms and seeking medical attention relatively soon (59.95%), and having symptoms and not seeking medical attention initially (17.66%). Other themes included having no symptoms or not noticing any symptoms before diagnosis (3.23%).

Symptoms leading to diagnosis: Description of diagnostic pathway

In the structured interview, participants described their diagnostic pathway in the healthcare system. The most common descriptions were a complex diagnosis, needing to see multiple specialists before diagnosis (46.52%), and a linear diagnosis after being referred to a specialist from their general practitioner (28.36%). Other themes included being diagnosed in an emergency department/urgent care (13.68%), being diagnosed by their general practitioner during a routine check-up that was not related to symptoms (5.97%).

Diagnosis provider and location

Participants were asked in the online questionnaire, which healthcare professional gave them their diagnosis, and where they were given the diagnosis. Participants were most commonly given their diagnosis in the specialist clinic (n=154, 43.14%), this was followed by the hospital (n=151, 42.30%), and the general practice (GP) (n=40, 11.20%).

Understanding of disease at diagnosis

Participants were asked in the structured interview how much they knew about their condition at diagnosis. The most common response was knowing nothing or very little about the condition at diagnosis (61.44%) Others described knowing a good amount about the condition at diagnosis, for example they knew about the condition by learning about it before or during the diagnostic process (7.71%), and knowing about the condition due to professional background (3.23%).

Emotional support at diagnosis

Participants were asked in the online questionnaire how much emotional support they or their family received between diagnostic testing and diagnosis. There were 79 participants (21.07%) who had enough support, 96 participants (25.60%) that had some support but it wasn't enough, and 200 participants (53.33%) had no support.

Costs at diagnosis

Participants noted in the online questionnaire the amount of out-of-pocket expenses they had at diagnosis, for example doctors' fees, and diagnostic tests. There were 146 participants (53.09%) who had no out of pocket expenses, and 51 participants (18.55%) who did not know or could not recall. There were 34 participants (12.36%) that spent Less than $500, 13 participants (4.73%) that spent between $500 to $1000, and 31 participants (11.27%) that spent More than $1000.

Burden of diagnostic costs

In the follow-up question about the burden of costs at diagnosis, for 30 participants who had out of pocket expenses. For 65 participants (33.85%) the cost was slightly or not at all significant. For 40 participants (20.83%) the out-of-pocket expenses were somewhat significant, and for 87 participants (45.31%), the burden of out-of-pocket expenses were moderately or extremely significant.

Genetic tests and biomarkers

Participants answered questions in the online questionnaire about if they had any discussions with their doctor about biomarkers, genomic and gene testing that might be relevant to treatment. If they did have a discussion, they were asked if they brought up the topic or if their doctor did.

Most commonly, participants had never had a conversation about biomarkers, genomic, or gene testing that might be relevant to treatment, (n=211, 66.56%). There were 28 participants (8.83%) who brought up the topic with their doctor, and 78 participants (24.61%) whose doctor brought up the topic with them.

Participants were then asked if they had had any biomarker, genomic or gene testing. If they had testing, they were asked if they had it as part of a clinical trial, paid for it themselves or if they did not have to pay for it. Those that did not have the test were asked if they were interested in this type of test. A little over half of participants indicated that they did not have any genetic or biomarker tests but would like to (n=193, 60.88%.

Understanding of prognosis

Participants were asked in the structured interview to describe what their current understanding of their prognosis was. The most common responses were that there was uncertainty around prognosis (26.37%), in terms of symptoms and function/changes in symptoms and function (17.66%), and that they had specific medical interventions they need to manage their condition (15.92%). Other themes included that they were monitoring their condition until there is an exacerbation or progression (15.67%), and had poor outcomes, or a terminal condition (11.94%).

Section 4

Decision-making

Section 4 summary

Discussions about treatment

Participants were asked to recall what treatment options they were presented with and how they felt about the options. Participants most commonly were presented with multiple options (40.52%), and this was followed by no discussions about treatment (24.92%) and one treatment option (22.77 %).

Discussions about treatment (Participation in discussions)

For those presented with multiple treatment options, descriptions included participating in the decision-making process (13.85%) and being told what to do without discussion (11.69%). This was followed by not participating in the decision-making process (3.69%).

For those with a single treatment option, descriptions included being told what to do without discussion (7.08%) and participating in the discussion (5.85 %). Some participants were presented with no treatment options as no therapies are available but allied health or complementary support offered (5.54%), while others had no therapies or options presented.

Considerations when making decisions

Participants were asked in the structured interview what they considered when making decisions about treatment. The most common responses were side effects (46.31%), efficacy (38.64%), advice of their clinician (26.14%) and cost (21.02 %). Other themes quality of life (16.76%), impact on their family or dependents (9.09%), amount of time needed for treatment and travel times (6.53%), ability to follow treatments (10.51%), and ability to work (4.55%).

Decision-making over time

Participants were asked if the way they made decisions had changed over time. There were 201 participants (57.10%) that had changed the way they make decisions, and 110 participants (31.25%) had not changed the way they make decisions.

Where participants had changed the way they make decisions, the most common reasons were that they were more informed and/or more assertive (23.01%), more aware of their health, responsibilities and/or limitations (10.80%), and more cautious and considered (8.24 %). Other themes included more focused impact on quality of life (5.40%).

Where participants had not changed the way they make decisions, the most common reason was that they had always been informed/assertive (6.25%).

Personal goals of treatment or care

Participants were asked what their own personal goals of treatment or care were. The most common responses were to have quality of life/return to normality (22.56%), to maintain their condition or prevent worsening of their condition (19.55%) and have physical improvements in their condition (18.05 %). Other themes included the ability to live independently (13.53%) and wanting to minimise or avoid side effects (8.27%).

Section 5

Treatment

Section 5: Experience of treatment

Respect shown

Participants were asked to think about how respectfully they were treated throughout their experience, this question was asked in the online questionnaire. Just under half of the participants indicated that they had been treated with respect throughout their experience (n=133, 41.43%), and 134 participants (41.74%) were treated with respect with the exception of one or two occasions. There were 54 participants (16.82%) felt they had not been treated respectfully.

Health care system

In the online questionnaire, participants were asked questions about the healthcare system they used, about private insurance and about whether they were treated as a public or private patient. The majority of participants had private health insurance (n=201, 64.63%). The majority of participants were not asked if they wanted to be treated as a public or private patient (n=157, 60.15%), however, they were asked if they had private health insurance (n=153, 58.62%). Throughout their treatment, there were 71 participants (23.05%) that were treated as a private patient, 156 participants (50.65%) were mostly treated as a public patient, and there were 68 participants (22.08%) that were equally treated as a private and public patient. Throughout their treatment, there were 42 participants (11.73%) that were treated mostly in the private hospital system, 228 participants (63.69%) were mostly treated in the public system, and there were 88 participants (24.58%) that were equally treated in the private and public systems.

Affordability of healthcare

Participants were asked a series of questions about affordability of healthcare in the online questionnaire.

The first question was about having to delay or cancer healthcare appointments because they were unable to afford them. Almost all the participants never or rarely had to delay or cancel appointments due to affordability (n = 259, 71.75%).

The next question was about the ability to fill prescriptions. Almost all of the participants never or rarely were unable to fill prescriptions (n=66, 18.28%).

The third question was about the affordability of basic essentials such as such as food, housing and power. There were 36 participants (9.97%) that never or rarely had trouble paying for essentials, and 13 participants (3.60%) that sometimes found it difficult, and 48 participants (13.30%) often or very often found it difficult to pay for basic essentials.

The final question was about paying for additional carers for themselves or for their family, there were 74 participants (23.79%) that paid for additional carers due to their condition.

Cost of condition

In the online questionnaire, participants estimated the amount they spend per month due to their condition, including doctors' fees, transport, carers, health insurance gaps and complementary therapies.

The most common amount was between $1001 or more (n=32, 8.74%), followed by between $101 to $250 (n=61, 16.67%). There were 41 participants (11.20%), that spent $501 to $1000 a month.

Burden of cost

As a follow up question, for participants that had monthly expenses due to their condition, participants were asked if the amount spent was a burden.

The amount spent was an extremely significant or moderately significant burden for 102 participants (33.44%), somewhat significant for 77 participants (25.25%), and slightly or not at all significant for 126 participants (41.31%).

Changes to employment status

Participants were asked, in the online questionnaire, if they had any changes to their employment status due to their condition. Participants were able to choose multiple changes to employment.

Work status for 63 participants (23.95%) had not changed since diagnosis, and 33 participants (12.55%) were retired or did not have a job. There were 79 participants (30.04%) had to quit their job, 78 participants (29.66%) reduced the number of hours they worked, and 28 participants (10.65%) that accessed their superannuation early. There were 49 participants (18.63%) that took leave from work without pay, and 48 participants (18.25%) that took leave from work with pay.

Participants were asked, in the online questionnaire, if they had any changes to the employment status of their care or partner due to their condition. Participants were able to choose multiple changes to employment.

There were 71 participants (24.40%), without a main partner or carer. Most commonly, participants had partners or carers that did not change their work status due to their condition (n=100, 34.36%). There were 43 participants (14.78%) whose partners reduced the numbers of hours they worked, and 19 partners, (6.53%) that quit their job. The partners of 26 participants (8.93%) took leave without pay, and there were 34 partners (11.68%) that took leave with pay.

Reduced income due to condition

More than half of the participants (n=217, 57.05%) indicated in the online questionnaire that they had a reduced family income due to their condition.

Estimated reduction monthly income

As a follow up question, participants were asked if their family or household income had reduced due to their condition. Where a dollar amount was given, it is listed below.

Most commonly, participants were not sure about the amount their monthly income was reduced by $2501 to 5000 (n=32, 10.74%), or reduced by between $1501 to 2500 per month (n=38, 12.75%).

Burden of reduced income

Participants were then asked if this reduced family or household income was a burden.

For 22 of these participants (16.30%), the burden of this reduced income was extremely or moderately significant, for 28 participants (20.74%) the burden was somewhat significant, and for 85 participants (62.96%) the burden was slightly or not all significant.

Lifestyle changes

Participants were asked about any lifestyle changes they had made since diagnosis, the quality of life from these changes, and how effective they found them.

Most participants used at made at least one lifestyle change (n=204, 67.77%), and on average made 1 changes (median=1.00, IQR=1.00).

The most common lifestyle change used was diet changes (n=150, 51.02%), followed by exercise (n=146, 59.84%), and reduce alcohol (n=56, 22.95%)

Complementary therapies

Participants were asked about any complementary therapies they used to manage their condition, the quality of life from these changes, and how effective they found them.

Most participants used at made at least one complementary therapy (n=216, 68.35%), and on average used 1 therapies (median=1.00, IQR=2.00).

The most common complementary therapy used was supplements (n=136, 46.10%), followed by mindfulness or relaxation (n=121, 45.83%), and massage therapy (n=80, 30.30%).

Clinical trials

In the online questionnaire, participants were asked if they had discussions with their doctor about clinical trials, and if they did, who initiated the discussion.

There was a total of 111 participants (35.81%) that had discussions about clinical trials, 32 participants (10.32%) had brought up the topic with their doctor, and the doctor of 79 participants (25.48%) brought up the topic. The majority of participants had not spoken to anyone about clinical trials (n=199, 64.19%).

As a follow up question, participants were asked if they had taken part in a clinical trial, and if they had not taken part if they were interested in taking part.

There were 37 participants (11.86%) that had taken part in a clinical trial, 155 participants (49.68%) that would like to take part in a clinical trial if there was a suitable one, and 120 participants, that have not participated in a clinical trial and do not want to (38.46%).

Description of mild side effects

In the structured interview, participants were asked how they would describe the term 'mild side effects'. The most common descriptions of mild side effects were described using a specific example (53.69%), and those that do not interfere with life (33.24%). Other themes included those that are resolved in a short amount of time (9.66%) and those that can be managed with self-medication or self-management (3.98 %).

Description of severe side effects

In the structured interview, participants were asked how they would describe the term 'severe side effects'. The most common description of severe side effects were described using a specific example (47.73%), and those that impact everyday life or ability to conduct activities of daily living (28.13%). Other themes included those that are life threatening or result in hospitalisation (8.52%), those that cause long-term damage to their body (7.67%).

When a specific side effect was described, the most common examples were aches and pain (17.33%), emotional and mental impact (7.39%), and nausea with vomiting (6.53%). Other themes included fatigues (5.11%), gastrointestinal distress (4.83%), impact on sleep (4.26%), vision problems (3.98%), and impact on sleep (4.55%).

Adherence to treatment

Participants were asked in the structured interview what influences their decision to continue with a treatment regime. The most common responses were adhering to treatment for a specific amount of time (38.35%), adhering to treatment according to the advice of their specialist or as long as prescribed (36.08%), and adhering to treatment as long as side effects are tolerable (24.43 %). Other themes included never giving up on any treatment (11.36%), adhering to treatment as long as treatment is working (7.10%).

What needs to change to feel like treatment is working

Participants were asked to describe what needs to change to feel like treatment is effective. The most common responses were needing to see a specific symptom reduction (26.70%), needing to see needing to see physical signs and symptoms disappear or reduce side effects (25.85%), a needing to see improvements in general wellbeing (quality of life) (14.49%), needing to see evidence of stable disease (14.20%), needing to see a return to day-to-day functionality (14.20%), and needing to see improvement in pain levels (12.50%).

What it would mean if treatment worked

As a follow up question, participants were asked what it would mean to them if the treatment worked in the way they described. The most common responses were that it would allow them to do everyday activities/return to normal life (29.44%) and allow them to engage more with social activities and family life (11.67%). Other themes included allow them to return to work (9.44%), allow them to do more exercise (11.28%), will have a positive impact on their mental health (7.89%), allow the

Section 6

Information and communication

Section 6: Information and communication

Access to information

In the structured interview, participants were asked what information they had been able to access since they were diagnosed. The most common responses were the internet (Including health charities) (59.45%), from a specific health charity (32.34%) and from Facebook and\or social media (26.12%). Other themes included their treating clinician (25.62%), from journals (research articles) (22.89%), from other patient's experience (Including support groups) (18.41%), from books, pamphlets and newsletters (14.68%).

Information that was helpful

In the structured interview, participants were asked to describe what information they had found to be most helpful. The most common responses were other people's experiences (26.37%), health charity information (16.67%), hearing what to expect (e.g. from disease, side effects, treatment) (15.92%), and talking to a doctor or specialist or healthcare team (15.92%). Other themes included medical or scientific sources (11.19%), and information on triggers and managing exacerbations (6.97%).

Information that was not helpful

In the structured interview, participants were asked if there had been any information that they did not find to be helpful. The most common response was that there was no information that was not helpful (31.09%). The most common types of unhelpful information included information from their GP or specialist (11.94%), sources that are not credible (10.20%), other people's experiences (9.20 %), information that was not type specific or too general (8.46%). Other themes included a lack of new information (7.46%) and worse case scenarios (7.46%).

Information preferences

Participants were asked whether they had a preference for information online, talking to someone, in written (booklet) form or through a phone App. The most common responses were online information (29.35%), talking to someone plus online information (23.63%), and talking to someone (21.64 %). Other themes included written information (13.68%), all forms (5.47%), and apps (2.49%).

The main reasons for a preference for online information were accessibility (27.86%) and being able to digest information at their own pace (18.41%).

The main reasons for a preference for talking to someone was being able to have time to ask questions (18.41%), and that it was personalised (14.43%). The main reason for a preference for written information were written information is that they can refer back to/highlight important information (3.23%).

Timing of information

Participants in the structured interview were asked to reflect on their experience and to describe when they felt they were most receptive to receiving information. The most common times were at the beginning (diagnosis) (31.34%), continuously (19.65%), after the shock of diagnosis (12.44%) and 12 months or more after diagnosis (10.70 %).

Healthcare professional communication

Participants were asked to describe the communication that they had had with health professionals throughout their experience. The most common theme was that participants described having an overall negative (34.83%), overall positive (26.62%), and overall positive, with the exception of one or two occasions (24.63%).

Partners in health

The Partners in Health questionnaire (PIH) measures an individual's knowledge and confidence for managing their own health. The Partners in Health comprises a global score, 4 scales; knowledge, coping, recognition and treatment of symptoms, adherence to treatment and total score. A higher score denotes a better understanding and knowledge of disease.

The overall scores for the cohort were in the highest quintile for Partners in health: Knowledge (median=26.00, IQR=8.00), Partners in health: Adherence to treatment (median=14.00, IQR=4.00), indicating very good knowledge, very good adherence to treatment.

The overall scores for the cohort were in the second highest quintile for Partners in health:Recognition and management of symptoms (median=19.00, IQR=5.75), Partners in health:Total score (median=72.00, IQR=20.00) indicating good recognition and management of symptoms, good overall ability to manage their health.

The overall scores for the cohort were in the middle quintile for Partners in health:Coping (median=14.00, IQR=7.00), indicating moderate coping.

Ability to take medicine as prescribed

Participants were asked about their ability to take medicines as prescribed. The majority of the participants responded that they took medicine as prescribed all the time (n=173, 57.10%), and 120 participants (39.60%) responded that they took medicines as prescribed most of the time. There were 6 participants (1.98%) that sometimes took medicines as prescribed.

Information given by health professionals

Participants were asked about what type of information they were given by healthcare professionals, information about treatment options (n=188, 58.02%), disease management (n=147, 45.37%), disease cause (n=119, 36.73%) and, physical activity (n=85, 26.23%) were most frequently given to participants by healthcare professionals, and, information about interpret test results (n=54, 16.67%), clinical trials (n=43, 13.27%) and, complementary therapies (n=34, 10.49%) were given least often.

Information searched independently

Participants were then asked after receiving information from healthcare professionals, what information did they need to search for independently. The topics participants most often searched for were disease management (n=212, 65.43%), treatment options (n=210, 64.81%), disease cause (n=207, 63.89%) and, complementary therapies (n=167, 51.54%) were most frequently given to participants by healthcare professionals, and, information about clinical trials (n=123, 37.96%), interpret test results (n=120, 37.04%) and, hereditary considerations (n=103, 31.79%) were searched for least often.

Information gaps

The largest gaps in information, where information was neither given to patients nor searched for independently were clinical trials (n=177, 54.63%) and interpret test results (n=172, 53.09%).

The topics that participants did not search for independently after not receiving information from healthcare professionals were treatment options (n=66, 20.37%) and disease cause (n=58, 17.90%).

The topics that participants were given most information from both healthcare professionals and searching independently for were disease cause (n=146, 45.06%) and complementary therapies (n=145, 44.75%).

The topics that participants searched for independently after not receiving information from healthcare professionals were treatment options (n=122, 37.65%) and disease management (n=96, 29.63%).

Section 7

Care and support

Section 7: Experience of care and support

Care coordination

A Care Coordination questionnaire was completed by participants within the online questionnaire. The Care Coordination questionnaire comprises a total score, two scales (communication and navigation), and a single question for each relating to care-coordination and care received. A higher score denotes better care outcome.

The overall scores for the cohort were in the highest quintile for **Care coordination: Quality of care** global measure (median=7.00, IQR=3.00) indicating good quality of care. The overall scores for the cohort were in the highest quintile for **Care coordination: Communication** (median=36.00, IQR=13.00), **Care coordination: Navigation** (median=23.00, IQR=8.00), **Care coordination: Total score** (mean=58.51, SD=14.77), **Care coordination: Care coordination global measure** (median=6.00, IQR=4.00) indicating moderate communication, moderate communication, moderate care coordination, moderate care coordination.

The **Care coordination: communication** scale measures communication with healthcare professionals, measuring knowledge about all aspects of care including treatment, services available for their condition, emotional aspects, practical considerations, and financial entitlements. The average score indicates that participants had moderate communication with healthcare professionals.

The **Care coordination: navigation** scale navigation of the healthcare system including knowing important contacts for management of condition, role of healthcare professional in management of condition, healthcare professional knowledge of patient history, ability to get appointments and financial aspects of treatments. The average score indicates that participants had moderate navigation of the healthcare system.

The **Care coordination: total score** scale measures communication, navigation and overall experience of care coordination. The average score indicates that participants had moderate communication, navigation and overall experience of care coordination.

The **Care coordination: care coordination global measure** scale measures the participants overall rating of the coordination of their care. The average score indicates that participants scored rated their care coordination as moderate.

The **Care coordination: Quality of care global measure** scale measures the participants overall rating of the quality of their care. The average score indicates that participants rated their quality of care as good.

Experience of care and support

In the structured interview, participants were asked what care and support they had received since their diagnosis. This question aims to investigate what services patients consider to be support and care services. The most common responses were that they did not receive formal support (25.12%), found support and care from hospital or clinical setting (23.38%), family and friends (20.65%), and charities (17.41%). Other themes included peer support or other patients (13.93%), and challenges accessing support (12.44%).

Section 8

Quality of life

Section 8: Quality of life

Impact on quality of life

In the structured interview, participants were asked whether they felt that their condition had affected their quality of life. Most commonly, the descriptions suggested that there was an overall negative impact on quality of life (63.43%), followed by an overall minimal impact on quality of life (10.20%). Other themes included a mix of positive and negative impact on quality of life (7.71%), overall no impact on quality of life (2.74%), and overall positive impact on quality of life (4.23%).

The most common themes in relation to a negative impact on quality of life were emotional strain (including family/change in relationship dynamics) (41.79%), reduced social interaction (23.88 %) and reduced capacity for physical activity/needing to slow down (20.40%). Other themes included managing side effects and symptoms and emotional strain (respectively 10.70%), altering lifestyle to manage condition (including being immunocompromised) (10.45%), and managing fatigue (7.21%).

The most common theme in relation to a positive impact on quality of life was realising what is important (giving perspective/staying positive) (6.97%).

Impact on mental health

In the structured interview, participants were asked if there had been an impact on their mental health. Most commonly, the descriptions suggested that overall, there was at least some impact on mental health (77.84%), and overall, there was no impact on mental health (5.97%).

Regular activities to maintain mental health

In the structured interview, participants were asked what they needed to do to maintain their emotional and mental health. The most common response was consulting a mental health professional (24.17%), coping strategies such as remaining social, lifestyle changes and hobbies(22.52%), and mindfulness and/or meditation (16.56 %). Other themes included no activities to maintain mental health (15.89%), the importance of family and friends in maintaining their mental health (14.90%), and the importance of physical exercise (14.90%).

Regular activities to maintain health

In the structured interview, participants were asked what were some of the things they needed to do everyday to maintain their health? The most common activities for general health were self-care e.g. more rest, accepting help, pacing (34.38%), complying with treatment/management (29.83%), and doing physical exercise/physically active (22.73 %). Other themes included understanding their limitations (19.89%), maintaining a healthy diet (14.20%), being organised and planning ahead (11.93%), and maintaining a normal routine (8.24%).

Experience of vulnerability

In the structured interview, participants were asked if there had been times that they felt vulnerable. The most common responses were that they felt vulnerable when having sensitive discussion (diagnosis, treatment decision) (16.67%), because of interactions with the medical team(14.44%), and experiencing side effects from treatment or symptoms from condition (9.44 %). Other themes included thinking about disease course/incurable condition (8.33%), during or after treatments (6.67%), and when feeling sick/unwell (5.56%).

As a follow up question, participants described ways that they managed feelings of vulnerability. The most common ways to manage vulnerability were using self-help methods (resilience, acceptance, staying positive) (7.78%), and support from nurse or treatment team (3.89%). Other themes included getting support from family and friends (3.33%), and support from mental health professionals (2.22%).

Impact on relationships

Most commonly, the descriptions suggested that overall, there was a negative impact on relationships (36.82%), and overall, there was a positive impact on relationships (23.13%). Other themes included overall, no impact on relationships (11.91%), and overall, there was an impact on relationships that was neither positive nor negative (10.95%).

The most common themes in relation to having a negative impact on relationships was from the dynamics of relationships changing due to anxiety, exacerbations and/or physical limitations of condition (25.37%). from people not knowing what to say or do and withdrawing from relationships (22.14%). This was followed by social isolation (10.70 %). The most common reasons for a positive impact on relationships was that people were supportive and well-meaning (15.67%).

Burden on family

In the structured interview, participants were asked whether they felt that their condition placed additional burden on their family. Most commonly, the descriptions suggested that overall, there was a burden on their family (62.60.19%), overall, there was not a burden on their family now but they anticipate this will change in the future (4/26%), and overall, there was not a burden on their family (21.02.64 %).

The main reason that participant described their condition being a burden were the extra household duties and responsibilities that their family must take on(23.01%), and the mental/emotional strain placed on their family (9.94%). Others described the extra assistance needed getting to appointments (5.97 %) and that the burden on family was temporary or only during treatment (3.69 %).

Cost considerations

In the structured interview, participants were asked about any significant costs associated with having their condition. The most common descriptions were that overall, there was at least some cost burden (65.23%), and overall, there was no cost burden (18.87%).

Where participants described a cost burden associated with their condition, it was most commonly in relation to needing to take time off work (32.78%), the cost of treatments (including repeat scripts) (30.79%), and the cost specialist appointments (26.82 %). Other themes included diagnostic tests and scans (12.91%), the cost of parking and travel to attend appointments (including accommodation) (12.91%), needing to special equipment (8.61%), a family member needing to take time off work (5.96%) allied health care (5.63%), needing to special creams, ointments or complementary therapies (4.30%), and needing a special diet or lifestyle adaptation (3.64%).

Where participants described a cost burden associated with their condition, it was most commonly in relation to nearly everything was paid for through the public health system (21.52%).

Overall impact of condition on quality of life

In the online questionnaire, participants were asked to rate the overall impact their condition on quality of life. Quality of life was rated on a Likert scale from one to seven, where one is Life was very distressing and seven is life was great. The average score was in the Life was a little distressing range (median=3.00, IQR=2.00).

Fear of progression

The Fear of Progression questionnaire measures the level of anxiety people experience in relation to their conditions. The Fear of Progression questionnaire comprises a total score, between 12 and 60, with a higher score denoting increased anxiety. Summary statistics for the entire cohort are displayed in Table 8.10. Overall the entire cohort had a mean total score of 37.09 (SD = 10.40), which corresponds to moderate levels of anxiety.

On average, participants in the Diseases of the skin subgroup scored higher than participants in the Endocrine, nutritional or metabolic diseases subgroup. This indicates that participants in the Diseases of the skin subgroup had high levels of anxiety, and participants in the Endocrine, nutritional or metabolic diseases subgroup had moderate levels of anxiety.

On average, participants in the Female subgroup had a higher score compared to Male, however, both groups had moderate levels of anxiety.

On average, participants in the Aged 18 to 44 subgroup had a higher score compared to Aged 65 or older, however, both groups had moderate levels of anxiety.

Section 9

Expectations and messages to decision-makers

Section 9: Expectations of future treatment, care and support, information and communication

Expectations of future treatment

Participants were asked in the structured interview what their expectations of future treatments are. The most common responses were that future treatment will be more affordable (36.57%), be more effective and/or targeted (personalised) (21.39%) and will include having choice (including availability and accessibility) and transparency/discussions in relation to treatment options (pathways) (17.66 %). Other themes included have fewer or less intense side effects or more discussion about side effects (16.92%), involve more clinical trials (including to access new technologies and treatments and funding) (14.43%), be easier to administer or able to administer at home or be less invasive (12.94%) and involve a more holistic approach (11.19%).

Expectations of future information

Participants were asked in the structured interview if there was anything that they would like to see changed in the way information is presented or topics that they felt needed more information. The most common responses were that future information will be more accessible or easy to find (23.88%), and more details about disease trajectory and what to expect (12.19 %). Other themes included use information to help to inform the community and decision-makers about their condition (raise awareness) (11.94%), provide more details on subgroups and specific classifications of their condition (10.20%), and be easier to understand (7.96%). There were 58 participants (14.43%) who were satisfied with the information they received.

Expectations of future healthcare professional communication

Participants were asked in the structured interview what they would like to see in relation to the way that healthcare professionals communicate with patients. The most common expectations for future healthcare professional communication were that communication will include health professionals with a better knowledge of the condition (21.89%), be more empathetic (17.16%), and satisfied with experience (17.66 %). Other themes included be more transparent and forthcoming (10.95%), include listening to the patient (9.95%), allow people more time to meet with their clinician (9.70%), and include a multidisciplinary and coordinated approach (9.45%).

Expectations of future care and support

Participants were asked in the structured interview whether there was any additional care and support that they thought would be useful in the future, including support from local charities. The most common expectation for future care and support was that it will include more access to support services (22.89%), will include a multidisciplinary and coordinated approach (14.68 %) and will include specialist clinics or services where they can talk to professionals (in person, phone, online) (13.93%). Other themes included ill include being able to connect with other patients through peer support (support groups, online forums) (11.69%), will include health professionals with a better knowledge of the condition (9.70%), and will include practical support (home care, transport, financial) (7.96%). There were 32 participants (7.96%).) that were satisfied with their care and support and had no particular comment.

What participants are grateful for in the health system

Participants were asked in the structured interview what aspects of the health system that participants are grateful for. The most common responses were that participants were grateful for low cost or free medical care through the government (40.34%) – with the related theme os included timely access to treatment (11.36%). Other themes included being grateful for healthcare staff (including access to specialists) (35.23%), and the entire health system (18.47 %).

Values in making decisions

Participants were asked to rank what is important for them overall when they make decisions about treatment and care, where 1 is the most important and 8 is the least important. A weighted average is presented in the figure below. With a weighted ranking, the higher the score, the greater value it is to participants.

The most important aspects were “"How safe the medication is and weighing up the risks and benefits"”, and “"The severity of the side effects"”. The least important were “"Ability to follow and stick to a treatment regime"” and “"The ability to include my family in making treatment decisions"”.

Values for decision makers

Participants were asked to rank what is important for decision-makers to consider when they make decisions that impact treatment and care, where 1 is the most important and 5 is the least important. A weighted average is presented in the figure below. With a weighted ranking, the higher the score, the greater value it is to participants.

The most important values were “Quality of life for patients”, and “All patients being able to access all available treatments and services”. The least important was “Economic value to government and tax payers”.

Time taking medication to improve quality of life

Participants were asked in the online questionnaire, how many months or years would you consider taking a treatment, provided it gave you a good quality of life, even if it didn’t offer a cure.

The majority of participants (n = 88, 33.72%) would use a treatment for more than ten years for a good quality of life even if it didn’t offer a cure.

Most effective form of medicine

Participants were asked in the online questionnaire, in what form did they think medicine was most effective in.

There were 30 participants (11.11%) that thought that medicine delivered by IV was most effective, 49 participants (18.15%) thought that pill form was most effective, and 74 participants (27.41%) that thought they were equally effective. There were 117 participants (43.33%) that were not sure.

Messages to decision-makers

Participants were asked, “If you were standing in front of the health minister, what would your message be in relation to your condition?” The most common messages to the health minister were the need for timely and equitable access to support, care and treatment (25.87%), the need for more research investment (17.91%), and to help raise community awareness (14.43 %). Other themes included to invest in clinical trials (13.18%), that treatments need to be affordable (10.20%), and to invest in health professionals development (8.96%).

Section 10

Advice to others in the future: The benefit of hindsight

Section 10: Advice to others in the future

Anything participants wish they had known earlier

In the structured interview, participants were asked if there was anything they wish they had known earlier. The most common things that participants had wished they'd known earlier were to be assertive, an advocate, informed, and to ask questions (32.09%), to seek and accept help, including peer support and support groups (16.92%), to understand the trajectory of the disease (13.68%), and to try to stay positive (11.19 %).

Aspect of care or treatment they would change

In the structured interview, participants were asked if there was any aspect of their care or treatment they would change. The most common themes were that they would have liked to have had access to a specialist in their condition sooner (15.41%), that they would not change any aspect of their care or treatment and were satisfied with care and treatment received (13.16%), and they would have liked health care professionals to have had more knowledge and awareness of their condition (10.53 %). Other themes included they would have stopped or changed treatment sooner (7.89%), (5.64%), and they would have liked to have been diagnosed sooner (3.76%).

Section 1

Introduction and methods

Section 1 Introduction and methodology

Introduction

In Australia, a disease is considered rare if it affects less than 5 in 10,000 people. There are more than 7,000 rare diseases that are life threatening or chronically debilitating. Around 8% of Australians (2 million people) live with a rare disease.

A total of 407 participants with rare diseases or carers to people with rare diseases were recruited into this PEEK study. There were 392 that completed both parts of the study, 5 that completed or partially completed online questionnaire only and 10 participants that completed the interview only.

Personal Experience, Expectations and Knowledge (PEEK)

Patient Experience, Expectations and Knowledge (PEEK) is a research program developed by the Centre for Community-Driven Research (CCDR). The aim of PEEK is to conduct patient experience studies across several disease areas using a protocol that will allow for comparisons over time (both quantitative and qualitative components). PEEK studies give us a clear picture and historical record of what it is like to be a patient at a given point in time, and by asking patients about their expectations, PEEK studies give us a way forward to support patients and their families with treatments, information and care.

The research protocol used in PEEK studies is independently driven by CCDR. PEEK studies include a quantitative and qualitative component. The quantitative component is based on a series of validated tools. The qualitative component is the result of two years of protocol testing by CCDR to develop a structured interview that solicits patient experience data and provides patients with the opportunity to provide advice on what they would like to see in relation to future treatment, information and care. The structured interview has also been designed so that the outcomes of PEEK studies can inform policy, research, care, information, supportive care services and advocacy efforts.

Position of this study

A search was conducted in Pubmed (August 8, 2022) to identify studies of rare diseases with that described patient experience conducted in the past five years in Australia, and updated on January 4th 2023. The term “Rare disease” was searched in any field, and it is noted that not all rare diseases studies will be included using this search term, and the difficulty in searching using individual disease names. Interventional studies, meta-analysis studies, studies conducted in developing countries, and studies of less than five participants were excluded.

There were 201 studies identified, 52 studies used interviews, 30 studies used focus groups or other qualitative methods and 138 studies used questionnaires.

PEEK is largest study of rare diseases conducted in an Australian population with a total of 402 participants with rare diseases or carers to people with rare diseases were recruited into the study. There were 391 that completed or partially completed online questionnaires and 402 participants that were interviewed.

Introduction

In Australia, a disease is considered rare if it affects less than 5 in 10,000 people. There are more than 7,000 rare diseases that are life threatening or chronically debilitating. Around 8% of Australians (2 million people) live with a rare disease[1].

A total of 407 participants with rare diseases or carers to people with rare diseases were recruited into this PEEK study. There were 392 that completed both parts of the study, 5 that completed or partially completed online questionnaire only and 10 participants that completed the interview only.

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The research protocol used in PEEK studies is independently driven by CCDR. PEEK studies include a quantitative and qualitative component. The quantitative component is based on a series of validated tools. The qualitative component is the result of two years of protocol testing by CCDR to develop a structured interview that solicits patient experience data and provides patients with the opportunity to provide advice on what they would like to see in relation to future treatment, information and care. The structured interview has also been designed so that the outcomes of PEEK studies can inform policy, research, care, information, supportive care services and advocacy efforts.

Participants

To be eligible for the study, participants needed to have been diagnosed with a rare disease or be a carer to a person with a rare disease, have experienced the healthcare system in Australia, be 18 years of age or older, be able to speak English, and be able to give consent to participate in the study.

Ethics

Ethics approval for this study was granted (as a low or negligible risk research study) by the Centre for Community-Driven Research Ethics Committee (Reference CS_Q4_03).

Data collection

Data for the online questionnaire was collected using Zoho Survey (Zoho Corporation Pvt. Ltd. Pleasanton, California, USA, www.zoho.com/survey).

There were five researchers who conducted telephone interviews and used standardised prompts throughout the interview. The interviews were recorded and transcribed verbatim. Identifying names and locations were not included in the transcript. All transcripts were checked against the original recording for quality assurance.

Online questionnaire (quantitative)

The online questionnaire consisted of the 36-Item Short Form Health Survey (SF36) (RAND Health)[2], a modified Cancer Care Coordination Questionnaire for Patients (CCCQ)[3], the Short Fear of Progression Questionnaire (FOP12)[4], and the Partners in Health version 2 (PIH)[5]. In addition, investigator derived questions about demographics, diagnosis, treatment received and future treatment decisions making were included.

Structured Interview (qualitative)

Interviews were conducted via telephone by registered nurses who were trained in qualitative research. The first set of interview questions guided the patient through their whole experience from when symptoms were noticed up to the present day.

Questionnaire analysis

Statistical analysis was conducted using R included in the packages “car”, “dplyr” and “ggplot2” (R 3.3.3 GUI 1.69 Mavericks build (7328). The aim of the statistical analysis of the SF36, CCCQ, FOP12, and PIH responses was to identify variations by condition, type or participant (person with condition or carer), gender, age, education, location of residence, education status and socio-economic status. Scales and subscales were calculated according to reported instructions[2-5].

The Location of participants was evaluated by postcode using the Australian Statistical Geography Maps (ASGS) Remoteness areas accessed from the Australian Bureau of Statistics[6].

The level of socio-economic status of participants was evaluated by postcode using the Socio-economic Indexes for Areas (SEIFA) accessed from the Australian Bureau of Statistics[7].

For comparisons by condition and age, a one-way analysis of variance (ANOVA) analysis was conducted. A Tukey HSD test was used post-hoc to identify the source of any differences identified in the one-way ANOVA test. Where the assumptions for the one-way ANOVA were not met, a Kruskal-Wallis rank sum test on care was conducted with post-hoc pairwise comparisons using Wilcoxon rank sum test. When the assumption of equal variances were not met, a Welch one-way test was used with post-hoc pairwise t-tests with no assumption of equal variances.

For all other comparisons between groups, a two-sample t-test was used when assumptions for normality and variance were met, or when assumptions were not met, a Wilcoxon rank sum test with continuity correction was used. Questions where participants were asked to rank preferences were analysed using weighted averages. Weights were applied in reverse, the most preferred option was given the largest weight equal to the number of options, the least preferred option was given the lowest weight of 1.

Structured interviews analysis

A content analysis was conducted using conventional analysis to identify major themes from structured interviews. Text from the interviews were read line-by-line by the lead researcher and then imported into the custom PEEK analysis database. Each question within the interview was individually analysed. Initial categories and definitions were identified and registered in custom PEEK analysis database. The minimum coded unit was a sentence with paragraphs and phrases coded as a unit.

A second researcher verified the codes and definitions, and the text was coded until full agreement was reached using the process of consensual validation. Where a theme occurred less than 5 times it was not included in the study results, unless this result demonstrated a significant gap or unexpected result.

Data analysis and final reporting was completed in November 2023.

Position of this study

A search was conducted in Pubmed (August 8, 2022) to identify studies of rare diseases with that described patient experience conducted in the past five years in Australia, and updated on January 4th 2023 (Table 1.1). The term “Rare disease” was searched in any field, and it is noted that not all rare diseases studies will be included using this search term, and the difficulty in searching using individual disease names. Interventional studies, meta-analysis studies, studies conducted in developing countries, and studies of less than five participants were excluded. All studies identified are listed in Table 1.1 alongside the country where the study was conducted, the number of participants by data collection method, and the broad focus of the study.

There were 201 studies identified, 52 studies used interviews, 30 studies used focus groups or other qualitative methods and 138 studies used questionnaires.

The most common disease areas covered were endocrine, nutritional or metabolic diseases (n=43), developmental anomalies (n=35), diseases of the immune system (n=26), groups of diseases (n=23), diseases of the nervous system (n=17), neoplasms (n=13),and diseases of the respiratory system (n=10).

There were 4 studies conducted completely with an Australian population, one study of 25 participants that took part in focus groups, with 190 participants completing surveys about expectations of future care and treatment[8], one study of 20 participants that took part in focus groups also focused on future treatments and expectations[9], one study of 184 participants who completed surveys about decision making[10], and one study of 50 participants that completed surveys about health related quality of life[11].

PEEK is largest study of rare diseases conducted in an Australian population with a total of 407 participants with rare diseases or carers to people with rare diseases were recruited into the study. There were 392 that completed both parts of the study, 10 that completed or partially completed online questionnaire only and 5 participants that completed the interview only.

Table 1.1: PEEK position

Author (Year)	Disease category	Country	Interviews	Focus group	Surveys	Main focus
Babac (2018)[12,13]	Groups of diseases	Germany	68	0	0	Decision making
Guffon (2022)[14]	Endocrine, nutritional or metabolic diseases	France	55	0	0	unmet needs
Le Hénaff (2020)[15]	Diseases of the skin	France	50	0	0	Quality of life
Capella-Peris (2020)[16]	Diseases of the nervous system	USA	47	0	0	Symptoms and side effects
Hoffmann-Vold (2021)[17]	Diseases of the immune system	Multinational	47	0	0	Expectations
Moffatt (2019)[18-21]	Diseases of the circulatory system	Multinational	46	40	1203	Self-management
Kerr (2023)[22]	Multiple rare diseases	USA	45	0	0	Navigating healthcare
Pokrzywinski (2021)[23]	Endocrine, nutritional or metabolic diseases	Multinational	44	0	0	Quality of life
Peter (2022)[24]	Groups of diseases	UK	38	65	0	Symptoms and diagnosis
Harrington (2019)[25]	Endocrine, nutritional or metabolic diseases	USA	32	0	0	Symptoms and diagnosis
Eichler (2022)[26]	Endocrine, nutritional or metabolic diseases	Multi-national	31	0	0	Symptoms
Young (2022)[27]	Undiagnosed diseases	USA	30	0	0	Participation in research
Munro (2022)[28]	Groups of diseases	USA	29	0	29	Quality of life
Pompilus (2021)[29]	Neoplasms	USA	27	0	0	Quality of life
Bingaman (2022)[30]	Metabolic disorders	Multi-national	26	13	0	Symptoms
Kutsa (2022)[31]	Diseases of the immune system	USA	26	0	0	Parental coping
Vu Minh Arnell (2019)[32]	Developmental anomalies	Sweden	25	0	0	Quality of life
Pascoal (2022)[33]	Endocrine, nutritional or metabolic diseases	Portugal	23	0	23	Symptoms
Khanna (2020)[34]	Diseases of the immune system	Multinational	23	0	0	Symptoms and diagnosis
Lanar (2022)[35]	Endocrine, nutritional or metabolic diseases	Multinational	22	0	0	Quality of life
Gregersen (2022)[36]	Neoplasms	Denmark	22	0	0	Symptoms and diagnosis
Gumuchian (2018)[37]	Diseases of the skin	Canada	22	0	0	Quality of life
Nguyen (2022)[38]	Endocrine, nutritional or metabolic diseases	France	21	0	0	Quality of life
Iyer (2020)[39]	Diseases of the nervous system	USA	20	0	0	Self-management
Lewis (2020)[40]	Groups of diseases	UK	20	0	0	Symptoms and diagnosis
Hunter (2019)[41]	Diseases of the nervous system	USA	20	0	0	Quality of life
de Dios García-Díaz (2022)[42]	Endocrine, nutritional or metabolic diseases	Spain	20	0	0	Expectations
Güeita-Rodriguez (2020)[43]	Developmental anomalies	Spain	19	31	0	Care and support
Merker (2021)[44]	Developmental anomalies	USA	18	0	0	Symptoms and diagnosis
Powell (2022)[45]	Diseases of the nervous system	UK	18	0	0	Quality of life
Lyn (2020)[46]	Endocrine, nutritional or metabolic diseases	USA	17	19	0	Quality of life
Olivotto (2022)[47]	Diseases of the nervous system	Italy	16	0	16	Health-related quality of life
Plackowski (2022)[48]	Multiple rare diseases	USA	16	9	0	Self advocacy/lived experience
Granero-Molina (2020)[49]	Diseases of the immune system	Spain	16	6	0	Symptoms and diagnosis
Baumbusch (2018)[50]	Groups of diseases	Canada	16	0	0	Care and support
Jimenez-Moreno (2021)[51]	Diseases of the nervous system	UK	15	52	0	Decision making

Author (Year)	Disease category	Country	Interviews	Focus group	Surveys	Main focus
Simpson (2021)[52]	Groups of diseases	UK	15	0	0	Care and support
Stanarević Katavić (2019)[53]	Groups of diseases	Croatia	15	0	0	Self-management
Livermore (2019)[54]	Diseases of the immune system	UK	15	0	0	Quality of life
Grimstvedt (2021)[55]	Diseases of the nervous system	Norway	14	0	0	Treatment and management
Lee (2022)[56]	Groups of diseases	Canada	14	0	0	Symptoms and diagnosis
Deuitch (2021)[57]	Undiagnosed	USA	14	0	0	Care and support
Wheeden (2022)[58]	Endocrine, nutritional or metabolic diseases	Multinational	14	0	0	Symptoms and side effects
van Dongen (2022)[59]	Neoplasms	Netherlands	14	0	0	Condition management
Hausmann (2018)[60]	Diseases of the immune system	USA	12	0	0	Symptoms and diagnosis
Smits (2022)[61]	Groups of diseases	Netherlands	12	0	0	Care and support
Ford (2019)[62]	Diseases of the nervous system	UK	11	0	71	Symptoms and side effects
Honingh (2022)[63]	Endocrine, nutritional or metabolic diseases	Netherlands	11	0	0	Care and support
Adams (2018)[64]	Diseases of the skin	UK	11	0	0	Quality of life
McCausland (2018)[65]	Endocrine, nutritional or metabolic diseases	USA	10	0	341	Symptoms and diagnosis
Ragan (2021)[66]	Diseases of the digestive system	Canada	10	0	0	Self-management
Gaasterland (2019)[67]	Groups of diseases	Netherlands	10	0	0	Expectations
McLaughlin (2022)[68]	Diseases of the blood or blood-forming organs	UK	3	11	0	Symptoms
Eskes (2022)[69]	Endocrine, nutritional or metabolic diseases		2	37	0	Treatment
Erbis (2018)[70]	Diseases of the immune system	Germany	0	78	40	Expectations
Swezey (2019)[71]	Developmental anomalies	Multinational	0	75	0	Expectations
Dwyer (2022)[72]	Endocrine, nutritional or metabolic diseases	USA	0	58	0	Symptoms and diagnosis
Knoppers (2022)[73]	Diseases of the respiratory system	Canada	0	40	0	Expectations
Velvin (2021)[74]	Developmental anomalies	Norway	0	36	0	Treatment and management
Milette (2019)[75]	Diseases of the skin	Canada	0	34	0	Quality of life
Tikellis (2021)[8]	Diseases of the respiratory system	Australia	0	25	190	Expectations
Houdayer (2019)[76]	Undiagnosed	France	0	21	0	Symptoms and diagnosis
Long (2021)[9]	Endocrine, nutritional or metabolic diseases	Australia	0	20	0	Expectations
Milette (2020)[77]	Diseases of the skin	Canada	0	19	0	Care and support
Uhlenbusch (2019)[78]	Groups of diseases	Germany	0	18	0	Quality of life
Casassa (2021)[79]	Developmental anomalies	France	0	16	0	Quality of life
Bate (2019)[80]	Neoplasms	UK	0	10	18	Decision making
Younger (2022)[81]	Developmental anomalies	USA	0	10	0	multi-specilist clinic
Verger (2021)[82]	Groups of diseases	Spain	0	9	0	Care and support
Quinn (2020)[83]	Groups of diseases	Multinational	0	8	251	Self-management
Kocher (2021)[84]	Diseases of the immune system	Switzerland	0	5	101	Self-management
Mälstam (2018)[85]	Endocrine, nutritional or metabolic diseases	Sweden	0	5	0	Quality of life
Danvers (2022)[86]	Diseases of the immune system	France	0	*	0	Online support
Strobel (2022)[87]	Diseases of the immune system	Multi-national	0	*	0	Online support
Schwartz (2017)[88]	Groups of diseases	USA	0	0	3324	Quality of life

Author (Year)	Disease category	Country	Interviews	Focus group	Surveys	Main focus
Tse (2021)[89]	Diseases of the immune system	Multinational	0	0	3233	Expectations
Doser (2022)[90]	Developmental anomalies	Denmark	0	0	2467	Health-related quality of life
McMillan (2021)[91]	Diseases of the nervous system	Canada	0	0	1927	Treatment and management
Catto (2021)[92]	Neoplasms	UK	0	0	1796	Health-related quality of life
Spencer-Tansley (2022)[93]	Groups of diseases	UK	0	0	1795	Treatment and management
de Graaf (2021)[94]	Endocrine, nutritional or metabolic diseases	Multinational	0	0	1378	Expectations
Bogart (2017)[95]	Groups of diseases	USA	0	0	1218	Health-related quality of life
Wunsch (2022)[96]	Diseases of the immune system	Multi-national	0	0	1178	Health-related quality of life
Szczepura (2018)[97]	Groups of diseases	UK	0	0	1158	Symptoms and diagnosis
Bogart (2022)[98]	Groups of diseases	USA	0	0	1118	Care and support
Eichler (2022)[99,100]	Neoplasms	Germany	0	0	1113	Health-related quality of life
Springer (2020)[101]	Diseases of the immune system	USA	0	0	926	Symptoms and side effects
Thyen (2018)[102]	Developmental anomalies	Multinational	0	0	848	Health-related quality of life
Garrido-Estepa (2022)[103]	Injury, poisoning or certain other consequences of external causes	Spain	0	0	828	Health-related quality of life
Azar (2018)[104]	Diseases of the skin	Multinational	0	0	752	Treatment and management
Lancaster (2022)[105]	Diseases of the respiratory system	Multinational	0	0	739	Care and support
Cottin (2022)[106]	Diseases of the respiratory system	France	0	0	724	Symptoms and side effects
Hiremath (2018)[107]	Diseases of the digestive system	USA	0	0	632	Care and support
Duret (2020)[108]	Diseases of the immune system	France	0	0	632	Symptoms and side effects
Chu (2022)[109]	Diseases of the immune system	USA	0	0	629	Health-related quality of life
Damy (2022)[110]	Endocrine, nutritional or metabolic diseases	France	0	0	603	Symptoms and diagnosis
Webb (2021)[111]	Endocrine, nutritional or metabolic diseases	Multinational	0	0	598	Care and support
Montali (2021)[112]	Diseases of the digestive system	Multinational	0	0	569	Health-related quality of life
Kimura (2022)[113]	Certain infectious or parasitic diseases	Japan	0	0	538	Health-related quality of life
Hanisch (2018)[114,115]	Groups of diseases	Germany	0	0	484	Health-related quality of life
Tsai (2021)[116]	Developmental anomalies	USA	0	0	468	Expectations
Al Mukaddam (2022)[117]	Diseases of the musculoskeletal system or connective tissue	Multi-national	0	0	463	Health-related quality of life
Kreuter (2019)[118]	Diseases of the respiratory system	Germany	0	0	424	Health-related quality of life
Park (2019)[119]	Diseases of the immune system	Korea	0	0	360	Health-related quality of life
Mastboom (2018)[120]	Diseases of the musculoskeletal system or connective tissue	Netherlands	0	0	337	Health-related quality of life
Corneloup (2020)[121]	Diseases of the immune system	France	0	0	336	Health-related quality of life
Valassi (2022)[122]	Endocrine, nutritional or metabolic diseases	Multinational	0	0	320	Symptoms and diagnosis
Depping (2021)[123]	Groups of diseases	Germany	0	0	304	Expectations

Author (Year)	Disease category	Country	Interviews	Focus group	Surveys	Main focus
Shalhub (2020)[124]	Developmental anomalies	USA	0	0	300	Self-management
Pignolo (2020)[125]	Diseases of the musculoskeletal system or connective tissue	Multinational	0	0	299	Health-related quality of life
Wuyts (2018)[126]	Diseases of the respiratory system	Multinational	0	0	277	Health-related quality of life
Schuster (2022)[127]	Diseases of the nervous system	Multinational	0	0	275	Expectations
Hoyer (2022)[128]	Diseases of the respiratory system	Denmark	0	0	264	Health-related quality of life
Büttner (2022)[129]	Endocrine, nutritional or metabolic diseases	Germany	0	0	264	Health-related quality of life
Guilabert (2021)[130]	Groups of diseases	Spain	0	0	261	Care and support
Doser (2020)[131]	Developmental anomalies	Denmark	0	0	244	Health-related quality of life
Schut (2022)[132]	Neoplasms	Multinational	0	0	235	Health-related quality of life
Francisco (2020)[133]	Endocrine, nutritional or metabolic diseases	Portugal	0	0	209	Quality of life
Magliano (2021)[134]	Endocrine, nutritional or metabolic diseases	Italy	0	0	200	Quality of life
Maqhuzu (2020)[135]	Diseases of the respiratory system	Germany	0	0	194	Health-related quality of life
Ragusa (2020)[136]	Developmental anomalies	Italy	0	0	193	Quality of life
Graziano (2022)[137]	Diseases of the respiratory system	Italy	0	0	192	Health-related quality of life
Dinur (2020)[138]	Endocrine, nutritional or metabolic diseases	Israel	0	0	192	Quality of life
Warby (2019)[10]	Neoplasms	Australia	0	0	184	Decision making
Nicoloro-SantaBarbara (2017)[139]	Neoplasms	USA	0	0	180	Quality of life
Bernthal (2021)[140]	Diseases of the musculoskeletal system or connective tissue	Multinational	0	0	166	Health-related quality of life
Anghelina (2022)[141]	Neoplasms	USA	0	0	165	Health-related quality of life
Dimitrova (2021)[142]	Endocrine, nutritional or metabolic diseases	Switzerland	0	0	158	Health-related quality of life
Rihm (2022)[143]	Multiple rare diseases	Germany	0	0	149	Anxiety
Keller (2021)[144]	Endocrine, nutritional or metabolic diseases	Multinational	0	0	148	Health-related quality of life
Marques (2019)[145]	Developmental anomalies	Multinational	0	0	143	Care and support
Fjermestad (2018)[146]	Developmental anomalies	Norway	0	0	142	Health-related quality of life
Yuan (2020)[147]	Endocrine, nutritional or metabolic diseases	Netherlands	0	0	121	Health-related quality of life
Zöllner (2021)[148]	Developmental anomalies	Germany	0	0	121	Health-related quality of life
Witt (2019)[149]	Developmental anomalies	Germany	0	0	120	Health-related quality of life
Kuemmerle-Deschner (2020)[150]	Diseases of the immune system	Multinational	0	0	117	Health-related quality of life
Asperti (2022)[151]	Diseases of the immune system	Italy	0	0	116	Anxiety
Weidema (2020)[152,153]	Neoplasms	Multinational	0	0	115	Care and support
Hanisch (2019)[154]	Developmental anomalies	Germany	0	0	110	Health-related quality of life
Friedlander (2019)[155]	Groups of diseases	France	0	0	110	Health-related quality of life
Junker (2021)[156]	Diseases of the nervous system	Multinational	0	0	109	Health-related quality of life
Darmaun (2021)[157]	Developmental anomalies	France	0	0	108	Health-related quality of life
Polistena (2021)[158]	Endocrine, nutritional or metabolic diseases	Italy	0	0	106	Health-related quality of life

Author (Year)	Disease category	Country	Interviews	Focus group	Surveys	Main focus
Liu (2018)[159]	Endocrine, nutritional or metabolic diseases	USA	0	0	106	Treatment and management
Wiegand-Grefe (2022)[160]	Multiple rare diseases	Germany	0	0	100	Health-related quality of life
Schmidt (2022)[161]	Neoplasms	Germany	0	0	98	Health-related quality of life
van de Loo (2022)[162]	Groups of diseases	Netherlands	0	0	95	Health-related quality of life
Post (2021)[163]	Diseases of the nervous system	Multinational	0	0	95	Expectations
Chighizola (2021)[164]	Diseases of the immune system	Italy	0	0	92	Expectations
Bonnekoh (2022)[165]	Diseases of the skin	Germany	0	0	87	Health-related quality of life
Hyvönen (2020)[166]	Developmental anomalies	Finland	0	0	80	Health-related quality of life
Hanisch (2020)[167]	Developmental anomalies	Germany	0	0	79	Health-related quality of life
Lauby (2019)[168]	Diseases of the respiratory system	France	0	0	78	Health-related quality of life
Ramprasad (2021)[169]	Diseases of the nervous system	USA	0	0	77	Symptoms and side effects
Kodra (2020)[170]	Developmental anomalies	Italy	0	0	76	Health-related quality of life
Custers (2018)[171]	Diseases of the nervous system	Netherlands	0	0	76	Quality of life
Lee (2021)[172]	Diseases of the immune system	Canada	0	0	72	Health-related quality of life
Saad (2021)[173]	Developmental anomalies	France	0	0	72	Health-related quality of life
Thouvenin (2021)[174]	Diseases of the visual system	France	0	0	72	Health-related quality of life
Rabenstein (2021)[175]	Diseases of the nervous system	Germany	0	0	71	Treatment and management
Mengel (2020)[176]	Endocrine, nutritional or metabolic diseases	Germany	0	0	69	Symptoms and diagnosis
Theodore-Oklota (2022)[177]	Developmental anomalies	Multinational	0	0	68	Quality of life
Reisner (2022)[178]	Diseases of the skin	USA	0	0	66	Health-related quality of life
Camarata (2021)[179]	Endocrine, nutritional or metabolic diseases	USA	0	0	62	Health-related quality of life
Camarata (2022)[180]	Endocrine, nutritional or metabolic diseases	Multi-national	0	0	62	Health-related quality of life
Hahn (2018)[181]	Diseases of the immune system	Germany	0	0	61	Treatment and management
Underbjerg (2018)[182]	Endocrine, nutritional or metabolic diseases	Denmark	0	0	57	Health-related quality of life
De Sautu De Borbón (2021)[183]	Developmental anomalies	Spain	0	0	57	Health-related quality of life
Bahmer (2018)[184]	Diseases of the immune system	Germany	0	0	57	Treatment and management
Halimi (2018)[185]	Diseases of the circulatory system	France	0	0	55	Health-related quality of life
Biener (2021)[186]	Diseases of the immune system	Germany	0	0	54	Health-related quality of life
Hanisch (2018)[187]	Developmental anomalies	Germany	0	0	51	Symptoms and diagnosis
Xu (2021)[11]	Diseases of the immune system	Australia	0	0	50	Health-related quality of life
Yanes (2022)[188]	Endocrine, nutritional or metabolic diseases	Spain	0	0	50	Health-related quality of life
Crescimanno (2019)[189]	Diseases of the nervous system	Italy	0	0	48	Health-related quality of life
Defabianis (2022)[190]	Developmental anomalies	Italy	0	0	48	Health-related quality of life

Author (Year)	Disease category	Country	Interviews	Focus group	Surveys	Main focus
Murali (2022)[191]	Endocrine, nutritional or metabolic diseases	USA	0	0	47	Health-related quality of life
Izquierdo-García (2020)[192]	Endocrine, nutritional or metabolic diseases	Spain	0	0	46	Care and support
Oelerich (2020)[193]	Developmental anomalies	Germany	0	0	46	Health-related quality of life
Hanisch (2020)[194]	Diseases of the digestive system	Germany	0	0	44	Health-related quality of life
Hanisch (2019)[195]	Endocrine, nutritional or metabolic diseases	Germany	0	0	43	Health-related quality of life
Maffi (2022)[196]	Diseases of the nervous system	Italy	0	0	42	Symptoms and side effects
Konradi (2021)[197,198]	Diseases of the musculoskeletal system or connective tissue	USA	0	0	39	Quality of life
Peltola (2021)[199]	Neoplasms	France	0	0	38	Health-related quality of life
Chiu (2022)[200]	Multiple rare diseases	Hong Kong	0	0	36	Health-related quality of life
Gjørup (2021)[201]	Endocrine, nutritional or metabolic diseases/ Developmental anomalies	Denmark	0	0	35	Health-related quality of life
Herman (2022)[202]	Endocrine, nutritional or metabolic diseases	Slovenia	0	0	34	Covid
Nguyen (2019)[203]	Diseases of the circulatory system	USA	0	0	33	Health-related quality of life
Grimwood (2018)[204]	Diseases of the digestive system	France	0	0	33	Health-related quality of life
Quijada-Fraile (2021)[205]	Endocrine, nutritional or metabolic diseases	Spain	0	0	33	Symptoms and side effects
van de Loo (2022)[206]	Endocrine, nutritional or metabolic diseases	Netherlands	0	0	33	Health-related quality of life
Carey (2021)[207]	Developmental anomalies	USA	0	0	30	Symptoms and side effects
Kettenbach (2021)[208]	Diseases of the immune system	Germany	0	0	30	Symptoms and side effects
Ashtari (2022)[209]	Developmental anomalies	Multi-national	0	0	30	Online support
Holopainen (2019)[210]	Developmental anomalies	Finland	0	0	26	Symptoms and side effects
Stöberl (2019)[211]	Developmental anomalies	Switzerland	0	0	24	Symptoms and side effects
Harmon (2019)[212]	Developmental anomalies	USA	0	0	22	Health-related quality of life
Niekamp (2020)[213]	Developmental anomalies	Germany	0	0	21	Health-related quality of life
Morrison (2019)[214]	Endocrine, nutritional or metabolic diseases	Multinational	0	0	13	Quality of life
Lechevalier (2022)[215]	Diseases of the skin	France	0	0	11	Care and support
Planellas (2021)[216]	Endocrine, nutritional or metabolic diseases	Spain	0	0	6	Symptoms and side effects

* Analysis of social media comments number of participants not given

Abbreviations and terminology

ASGS	The Australian Statistical Geography Standard from the Australian Bureau of Statistics, defines remoteness and urban/rural definitions in Australia
CCDR	Centre for Community-Driven Research
dF	Degrees of Freedom. The number of values in the final calculation of a statistic that are free to vary.
f	The F ratio is the ratio of two mean square values, used in an ANOVA comparison. A large F ratio means that the variation among group means is more than you'd expect to see by chance.
FOP	Fear of Progression. Tool to measure anxiety related to progression
IQR	Interquartile range. A measure of statistical dispersion, being equal to the difference between 75th and 25th percentiles, or between upper and lower quartiles.
p	Probability value. A small *p*-value (typically ≤ 0.05) indicates strong. A large *p*-value (> 0.05) indicates weak evidence.
PEEK	Patient Experience, Expectations and Knowledge
PIH	Partners in Health
SD	Standard deviation. A quantity expressing by how much the members of a group digger from the mean value for the group/
SEIFA	Socio-Economic Indexes for Areas (SEIFA) ranks areas in Australia according to relative socio-economic advantage and disadvantage. This is developed by the Australian Bureau of Statistics.
SF36	Short Form Health Survey 36
t	t-Statistic. Size of the difference relative to the variation in your sample data.
Tukey HSD	Tukey's honestly significant difference test. It is used in this study to find 9significantly different means following an ANOVA test.
W	The W statistic is the test value from the Wilcoxon Rank sum test. The theoretical range of W is between 0 and (number in group one) x (number in group 2). When W=0, the two groups are exactly the same.
X^2	Chi-squared. Kruskal-Wallis test statistic approximates a chi-square distribution. The Chi-square test is intended to test how likely it is that an observed distribution is due to chance.

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Section 2

Demographics

Section 2 Demographics

Participants

In this PEEK study, a total of 407 participants with rare diseases or carers to people with rare diseases were recruited into the study. There were 5 that completed or partially completed online questionnaires only and 10 participants that completed the interview only. There were 96 participants (23.59%) with diseases of the nervous system, 96 participants (23.59%) with endocrine, nutritional or metabolic diseases, 81 participants (16.71%) with diseases of the immune system, 68 participants (16.71%) with developmental anomalies, 34 participants (7.86%) with other rare condition, and 32 participants (7.86%) with diseases of the skin.

Demographics

There were 407 people with that took part in this study, 299 were females (73.83%). Participants were aged from infant to over 75 years of age, most were aged between 35 to 64 years (n=232, 64.09%).

Participants were most commonly from New South Wales (n=124, 30.47%), Queensland (n=92, 22.60%), and Victoria (n=91, 22.36%). Most participants were from major cities (n=295, 72.48%), and they lived in all levels of advantage, defined by Socio-economic Indexes for Areas (SEIFA) (www.abs.gov.au) with 204 participants (49.88%) from an area with a high SEIFA score of 7 to 10 (more advantage), and 203 participants (50.12%) from an area of mid to low SEIFA scores of 1 to 6 (less advantaged).

There were 201 participants (50.38%) that had completed university to at least an associate degree. There were 163 participants who were employed either full time (24.56%), or part time (23.10%). Almost half of the participants were carers to family members or spouses (n=192, 53.04%), and just under half of the participants carers to children (n=155, 42.82%).

Other health conditions

Participants were asked about health conditions, other than their rare disease that they had to manage. Participants could choose from a list of common health conditions and could specify other conditions.

The majority of participants had at least one other condition that they had to manage (n=287, 93.79%), the maximum number reported was 16 other conditions, with a median of 4.00 other conditions (IQR = 5.00). The most commonly reported health condition was anxiety (n=173, 56.54%), followed by sleep problems or insomnia (n=169, 55.23%), chronic pain (n=154, 50.33%), and depression (n=132, 43.14%).

Subgroup analysis

Comparisons were made by condition. There were 67 participants (16.46%) with developmental anomalies, 82 participants (20.15%) with diseases of the immune system, 99 participants (24.32%) with diseases of the nervous system, 32 participants (7.86%) with diseases of the skin, 95 participants (23.34%) with endocrine, nutritional or metabolic diseases , and 32 participants (7.86%) with other rare condition.

Baseline health

The Short Form Health Survey 36 (SF36) measures baseline health, or the general health of an individual. The SF36 comprises nine scales: physical functioning, role functioning/physical, role functioning/emotional, energy and fatigue, emotional well-being, social function, pain, general health, and health change from one year ago. The scale ranges from 0 to 100, a higher score denotes better health or function.

The overall scores for the cohort were in the second highest quintile for **SF36 Role functioning/emotional** (median=66.67, IQR=100.00), **SF36 Emotional well-being** (median=68.00, IQR=27.00), indicating good emotional role functioning, good emotional well-being.

The overall scores for the cohort were in the middle quintile for **SF36 Physical functioning** (median=55.00, IQR=60.00), **SF36 Social functioning** (median=50.00, IQR=50.00), **SF36 Pain** (median=55.00, IQR=45.00), **SF36 Health change** (median=50.00, IQR=25.00), indicating moderate physical functioning, moderate social functioning, moderate pain, about the same as a year ago.

The overall scores for the cohort were in the second lowest quintile for **SF36 Role functioning/physical** (median=25.00, IQR=100.00), **SF36 Energy/Fatigue** (median=30.00, IQR=35.00), **SF36 General health** (median=40.00, IQR=35.00), indicating poor physical role functioning, poor energy, poor general health.

Comparisons of SF36 have been made based on condition, participant type, gender, age, education, location and socioeconomic status.

SF36 Physical functioning scale measures health limitations in physical activities such as walking, bending, climbing stairs, exercise, and housework. On average, physical activities were moderately limited for participants in this study.

SF36 Role functioning/physical scale measures how physical health interferes with work or other activities. On average, physical health often interfered with work or other activities for participants in this study.

SF36 Role functioning/emotional scale measures how emotional problems interfere with work or other activities. On average, emotional problems sometimes interfered with work or other activities for participants in this study.

SF36 Energy/fatigue scale measures the proportion of energy or fatigue experienced. On average, participants were often fatigued.

The **SF36 Emotional well-being** scale measures how a person feels, for example happy, calm, depressed or anxious. On average, participants had good emotional well-being.

The **SF36 Social functioning** scale measures limitations on social activities due to physical or emotional problems. On average, social activities were moderately limited for participants in this study.

The **SF36 Pain** scale measures how much pain, and how pain interferes with work and other activities. On average, participants had moderate pain.

The **SF36 General health** scale measures perception of health. On average, participants reported poor health.

The **SF36 Health change** scale measures health compared to a year ago. On average, participants reported that their health is about the same as a year ago.

Participants

In this PEEK study, a total of 402 participants with rare diseases or carers to people with rare diseases were recruited into the study. There were 5 that completed or partially completed online questionnaires only and 10 participants that completed the interview only.

There were 96 participants (23.59%) with diseases of the nervous system, 96 participants (23.59%) with endocrine, nutritional or metabolic diseases, 81 participants (16.71%) with diseases of the immune system, 68 participants (16.71%) with developmental anomalies, 34 participants (7.86%) with other rare condition, and 32 participants (7.86%) with diseases of the skin.

Table 2.1: Participants

Participants and diagnosis	Number (n=407)	Percent
Diseases of the nervous system	96	23.59
Endocrine, nutritional or metabolic diseases	96	23.59
Diseases of the immune system	81	19.90
Developmental anomalies	68	16.71
Other rare condition	34	8.35
Diseases of the skin	32	7.86

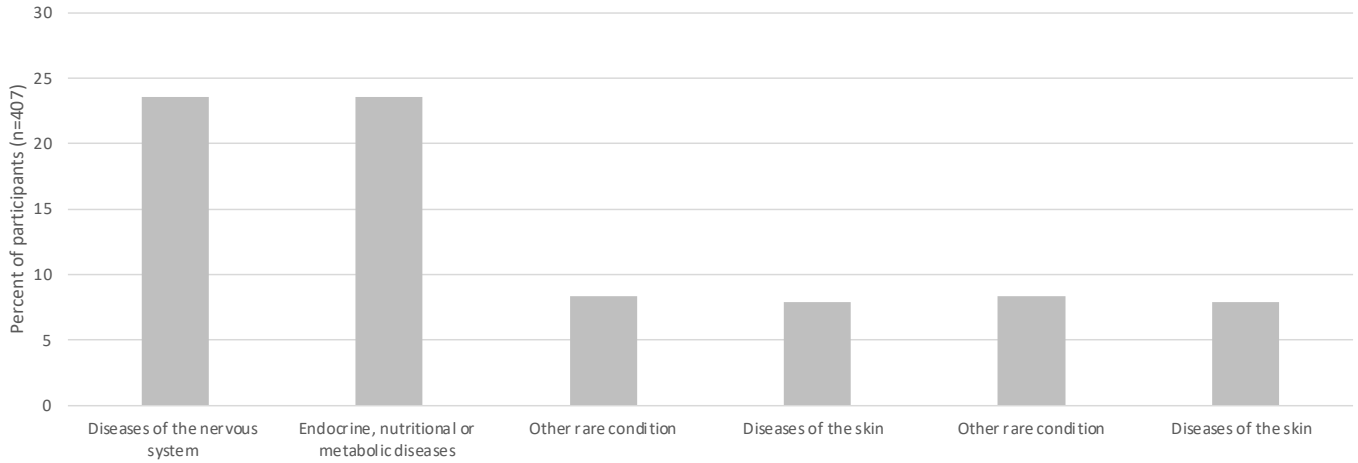


Figure 2.1: Participants

Demographics

There were 407 people with rare diseases that took part in this study, 299 were females (73.83%). Participants were aged from infant to over 75 years of age, most were aged between 35 to 64 years (n=232, 64.09%).

Participants were most commonly from New South Wales (n=124, 30.47%), Queensland (n=92, 22.60%), and Victoria (n=91, 22.36%). Most participants were from major cities (n=295, 72.48%), and they lived in all levels of advantage, defined by Socio-economic Indexes for Areas (SEIFA) (www.abs.gov.au) with 204 participants (49.88%) from an area with a high SEIFA score of 7 to 10 (more advantage), and 203 participants (50.12%) from an area of mid to low SEIFA scores of 1 to 6 (less advantaged).

There were 201 participants (50.38%) that had completed university to at least an associate degree. There were 163 participants who were employed either full time (24.56%), or part time (23.10%).

Almost half of the participants were carers to family members or spouses (n=192, 53.04%), most commonly carers to Children (n=155, 42.82%). The demographics of participants are listed in Table 2.2.

Table 2.2: Demographics

Demographic	Definition	Number	Percent
Gender (n=405)	Female	299	73.83
	Male	106	26.17
Age of person with condition (n=407)	Aged under 18	98	24.08
	18 to 24	14	3.44
	25 to 34	61	14.99
	35 to 44	58	14.25
	45 to 54	55	13.51
	55 to 64	61	14.99
	65 to 74	43	10.57
	75+	17	4.18
Location (n=407)	Major Cities of Australia	295	72.48
	Inner Regional Australia	77	18.92
	Outer Regional Australia	30	7.37
	Remote and very remote Australia	5	1.23
State (n=407)	Australian Capital Territory	14	3.44
	New South Wales	124	30.47
	Northern Territory	1	0.25
	Queensland	92	22.60
	South Australia	33	8.11
	Tasmania	10	2.46
	Victoria	91	22.36
	Western Australia	42	10.32
Socio-Economic Indexes for Areas (SEIFA) (n=407)	1	31	7.62
	2	27	6.63
	3	24	5.90
	4	29	7.13
	5	36	8.85
	6	56	13.76
	7	28	6.88
	8	49	12.04
	9	65	15.97
	10	62	15.23
Race/ethnicity (n=387)	Caucasian/White	350	90.44
	Asian	9	2.33
	Indigenous Australian/Torres Strait Islander	8	2.07
	Mixed race	6	1.55
	Pacific Islander	4	1.03
	Other	10	2.58
Education (n=399)	Less than high school degree	22	5.51
	High school degree or equivalent	70	17.54
	Some college but no degree	66	16.54
	Trade	34	8.52
	Trade or high school (Not specified)	6	1.50
	Associate degree	17	4.26
	Bachelor degree	97	24.31
	Graduate degree	78	19.55
	University (not specified)	9	2.26
Employment (n=342)	Currently receiving Centrelink support	44	12.87
	Disabled, unable to work	56	16.37
	Employed, working full time	84	24.56
	Employed, working part time	79	23.10
	Self employed	5	1.46
	Work in casual employment	10	2.92
	Engage in voluntary work	13	3.80
	Full/part time carer	34	9.94
	Full/part time study	16	4.68
	Not employed, looking for work	10	2.92
	Not employed, not looking for work	8	2.34
	Retired	63	18.42
Carer status (n=362)	I am not a carer	170	46.96
	Children	155	42.82
	Parents	20	5.52
	Spouse/Partner	19	5.25
	Grandchildren	14	3.87
	Other	6	1.66

Other health conditions

Participants were asked about health conditions, other than their primary rare disease that they had to manage. Participants could choose from a list of common health conditions and could specify other conditions.

The majority of participants had at least one other condition that they had to manage (n=287, 93.79%), the maximum number reported was 16 other conditions, with a median of 4.00 other conditions (IQR = 5.00). The most commonly reported health condition was anxiety (n=173, 56.54%), followed by sleep problems or insomnia (n=169, 55.23%), chronic pain (n=154, 50.33%), and depression (n=132, 43.14%).

Table 2.3: Number of other health conditions

Number of other conditions	Number (n=306)	Percent
No other conditions	19	6.21
1 to 2	56	18.30
3 to 4	62	20.26
5 to 6	65	21.24
7 to 8	48	15.69
9 to 10	28	9.15
11 or more	28	9.15

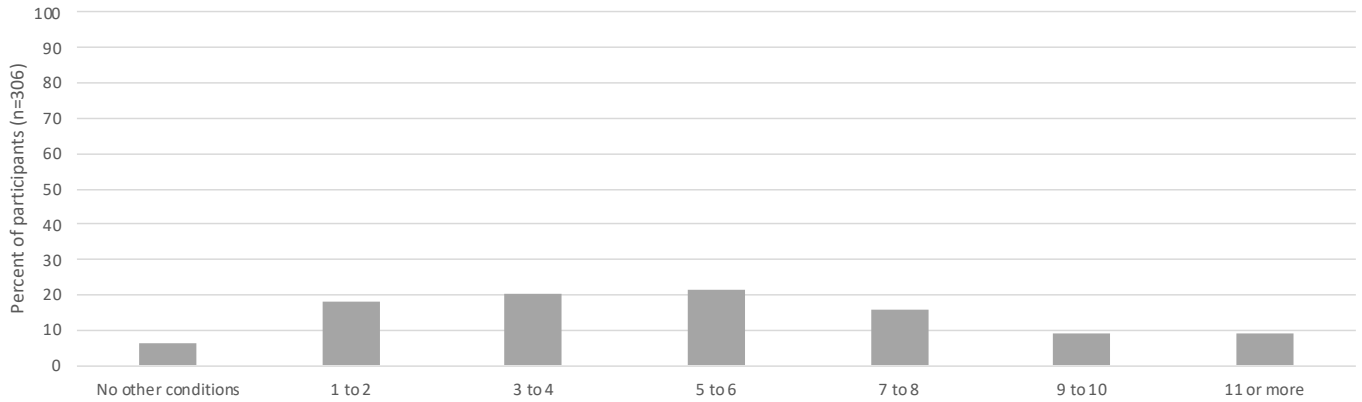


Figure 2.2: Number of other health conditions

Table 2.4: Other health conditions

Other conditions	Number (n=306)	Percent
Anxiety (Total)	173	56.54
Do you have anxiety (self diagnosed)	113	36.93
Do you have anxiety (diagnosed by a doctor)	106	34.64
Sleep problems or insomnia	169	55.23
Chronic pain	154	50.33
Depression (Total)	132	43.14
Depression (Self diagnosed)	74	24.18
Depression (Diagnosed by a doctor)	83	27.12
Arthritis	100	32.68
Hypertension	68	22.22
Asthma	49	16.01
High cholesterol	39	12.75
Atrial fibrillation	38	12.42
CODP (Chronic obstructive pulmonary disease)	22	7.19
Diabetes	22	7.19
Cancer	22	7.19
Stroke	16	5.23
Arrhythmias	15	4.90
Angina	14	4.58
Chronic heart failure	11	3.59
Chronic kidney disease	5	1.63

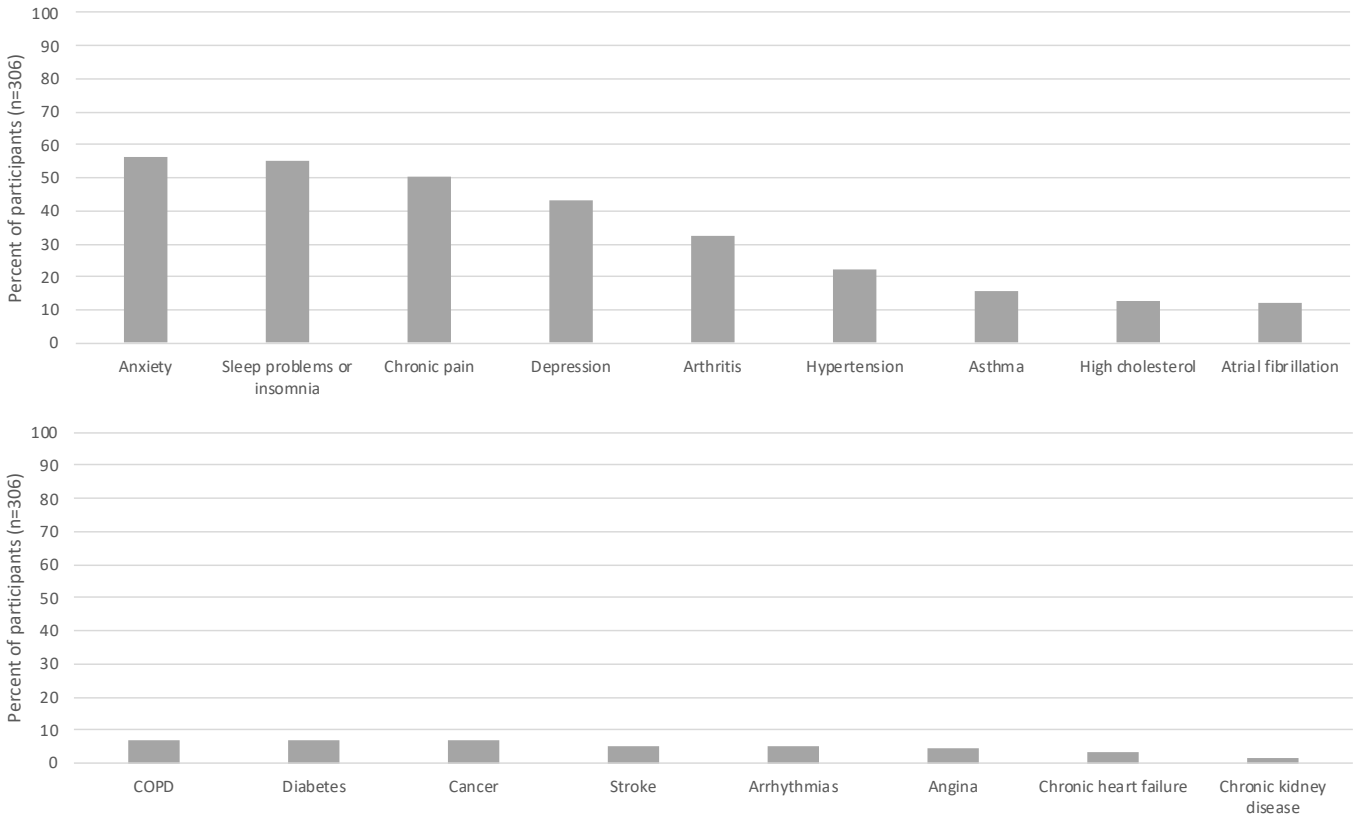


Figure 2.3: Other health conditions (% of all participants)

Subgroup analysis

Subgroup analysis are included throughout the study and the subgroups are listed in Table 2.5.

Comparisons were made by **condition**. There were 67 participants (16.46%) with developmental anomalies, 82 participants (20.15%) with diseases of the immune system, 99 participants (24.32%) with diseases of the nervous system, 32 participants (7.86%) with diseases of the skin, 95 participants (23.34%) with endocrine, nutritional or metabolic diseases, and 32 participants (7.86%) with other rare conditions.

Comparisons were made by **type of participant** there were 272 participants (66.83%) people with the condition themselves and 135 participants (33.17%) that were carers.

Comparisons were made by **gender**, there were 299 female participants (73.83%), and 106 male participants (26.17%).

Comparisons were made by **age** of person with condition. There were 99 participants (24.32%) aged under 18, 132 participants (32.43%) aged 18 to 44, 116 participants (28.50%) aged 45 to 64, and 60 participants (14.74%) aged 65 or older.

Comparisons were made by **education** status, between those with trade or high school qualifications (n=198, 49.62%), and those with a university qualification (n=201, 50.38%).

The **location** of participants was evaluated by postcode using the Australian Statistical Geography Maps (ASGS) Remoteness areas accessed from the Australian Bureau of Statistics. Those living in regional or remote areas (n=112, 27.52%) were compared to those living in a metropolitan area (n=295, 72.48%).

Comparisons were made by **socioeconomic status**, using the Socio-economic Indexes for Areas (SEIFA) (www.abs.gov.au), SEIFA scores range from 1 to 10, a higher score denotes a higher level of advantage. Participants with a mid to low SEIFA score of 1-6 (n=203, 49.88%) compared to those with a higher SEIFA score of 7-10 (n=204, 50.12%).

Table 2.5: Subgroups

Subgroup	Definition	Number	Percent
Condition (n=407)	Developmental anomalies	67	16.46
	Diseases of the immune system	82	20.15
	Diseases of the nervous system	99	24.32
	Diseases of the skin	32	7.86
	Endocrine, nutritional or metabolic diseases	95	23.34
	Other rare condition	32	7.86
Participant type (n=407)	Person with condition	272	66.83
	Carer	135	33.17
Gender (n=405)	Female	299	73.83
	Male	106	26.17
Age of person with condition (n=407)	Aged under 18	99	24.32
	Aged 18 to 44	132	32.43
	Aged 45 to 64	116	28.50
	Aged 65 or older	60	14.74
Education (n=399)	Trade or high school	198	49.62
	University	201	50.38
Location (n=404)	Regional or remote	112	27.52
	Metropolitan	295	72.48
Socioeconomic status (n=404)	Mid to low status	203	49.88
	Higher status	204	50.12

Baseline health

The Short Form Health Survey 36 (SF36) measures baseline health, or the general health of an individual. The SF36 comprises nine scales: physical functioning, role functioning/physical, role functioning/emotional, energy and fatigue, emotional well-being, social function, pain, general health, and health change from one year ago. The scale ranges from 0 to 100, a higher score denotes better health or function.

Summary statistics for the entire cohort are displayed alongside the possible range of each scale in Table 2.6, for scales with a normal distribution, the mean and SD should be used as a central measure, and median and IQR for scales that do not have a normal distribution.

The overall scores for the cohort were in the second highest quintile for **SF36 Role functioning/emotional** (median=66.67, IQR=100.00), **SF36 Emotional well-being** (median=68.00, IQR=27.00), indicating good emotional role functioning, good emotional well-being.

The overall scores for the cohort were in the middle quintile for **SF36 Physical functioning** (median=55.00, IQR=60.00), **SF36 Social functioning** (median=50.00, IQR=50.00), **SF36 Pain** (median=55.00, IQR=45.00), **SF36 Health change** (median=50.00, IQR=25.00), indicating moderate physical functioning, moderate social functioning, moderate pain, about the same as a year ago.

The overall scores for the cohort were in the second lowest quintile for **SF36 Role functioning/physical** (median=25.00, IQR=100.00), **SF36 Energy/Fatigue** (median=30.00, IQR=35.00), **SF36 General health** (median=40.00, IQR=35.00), indicating poor physical role functioning, poor energy, poor general health.

Comparisons of SF36 have been made based on condition, participant type, gender, age, education, location and socioeconomic status.

SF36 Physical functioning scale measures health limitations in physical activities such as walking, bending, climbing stairs, exercise, and housework. On average, physical activities were moderately limited for participants in this study.

SF36 Role functioning/physical scale measures how physical health interferes with work or other activities. On average, physical health often interfered with work or other activities for participants in this study.

SF36 Role functioning/emotional scale measures how emotional problems interfere with work or other activities. On average, emotional problems sometimes interfered with work or other activities for participants in this study.

SF36 Energy/fatigue scale measures the proportion of energy or fatigue experienced. On average, participants were often fatigued.

The **SF36 Emotional well-being** scale measures how a person feels, for example happy, calm, depressed or anxious. On average, participants had good emotional well-being.

The **SF36 Social functioning** scale measures limitations on social activities due to physical or emotional problems. On average, social activities were moderately limited for participants in this study.

The **SF36 Pain** scale measures how much pain, and how pain interferes with work and other activities. On average, participants had moderate pain.

The **SF36 General health** scale measures perception of health. On average, participants reported poor health.

The **SF36 Health change** scale measures health compared to a year ago. On average, participants reported that their health is about the same as a year ago.

Table 2.6: SF36 summary statistics

SF36 scale (n=383)	Mean	SD	Median	IQR	Possible range	Quintile
Physical functioning	54.32	32.69	55.00	60.00	0 to 100	3
Role functioning/physical	37.24	42.78	25.00	100.00	0 to 100	2
Role functioning/emotional	51.22	44.24	66.67	100.00	0 to 100	4
Energy/Fatigue	33.69	22.61	30.00	35.00	0 to 100	2
Emotional well-being	64.09	20.06	68.00	27.00	0 to 100	4
Social functioning	53.08	28.79	50.00	50.00	0 to 100	3
Pain	55.69	30.00	55.00	45.00	0 to 100	3
General health	41.64	24.02	40.00	35.00	0 to 100	2
Health change	44.76	24.74	50.00	25.00	0 to 100	3

Skewed distribution, use median and IQR as central measure. Possible range 0-100

SF36 by condition

Comparisons were made by **condition**. There were 59 participants (15.45%) with developmental anomalies, 77 participants (20.16%) with diseases of the immune system, 93 participants (24.35%) with diseases of the nervous system, 30 participants (7.85%) with diseases of the skin, 95 participants (24.87%) with endocrine, nutritional or metabolic diseases, and 28 participants (7.33%) with other rare condition.

Where the assumptions for normality of residuals were not met, a Kruskal-Wallis test was used. Post hoc pairwise comparisons using Wilcoxon rank sum test was used to identify the source of any differences identified in the Kruskal -Wallis test.

A Kruskal-Wallis test indicated a statistically significant difference in the SF36 Physical functioningscale between groups, $\chi^2(5) = 41.62$ $p<0.0001$. The largest significant difference was between Developmental anomalies (median = 80, IQR = 37.5), and Endocrine, nutritional or metabolic diseases (median = 45, IQR = 60, $p<0.0001$).

A Kruskal-Wallis test indicated a statistically significant difference in the SF36 Role functioning physicalscale between groups, $\chi^2(5) = 40.98$ $p<0.0001$. The largest significant difference was between Developmental anomalies (median = 75, IQR = 75), and Endocrine, nutritional or metabolic diseases (median = 0, IQR = 50, $p<0.0001$).

A Kruskal-Wallis test indicated a statistically significant difference in the SF36 Energy/fatiguescale between groups, $\chi^2(5) = 27.73$ $p<0.0001$. The largest significant difference was between Developmental anomalies (median = 40, IQR = 27.5), and Diseases of the immune system (median = 20, IQR = 30, $p = 0.0008$).

A Kruskal-Wallis test indicated a statistically significant difference in the SF36 Social functioningscale between groups, $\chi^2(5) = 16.89$ $P = 0.0047$. The largest significant difference was between Developmental anomalies (median = 62.5, IQR = 31.25), and Diseases of the immune system (median = 37.5, IQR = 37.5, $p = 0.0085$).

A Kruskal-Wallis test indicated a statistically significant difference in the SF36 Painscale between groups, $\chi^2(5) = 51.6$ $p<0.0001$. The largest significant difference was between Other rare condition(median = 85, IQR = 30.63), and Diseases of the immune system (median = 45, IQR = 32.5, $p<0.0001$).

A Kruskal-Wallis test indicated a statistically significant difference in the SF36 General healthscale between groups, $\chi^2(5) = 63.85$ $p<0.0001$. The largest significant difference was between Developmental anomalies (median = 55, IQR = 35), and Diseases of the immune system (median = 25, IQR = 30, $p<0.0001$).

SF36 Physical functioning scale measures health limitations in physical activities such as walking, bending, climbing stairs, exercise, and housework. On average, participants in the Developmental anomalies subgroup scored higher than participants in the Endocrine, nutritional or metabolic diseases subgroup. This indicates that physical activities were slightly limited for participants in the Developmental anomalies subgroup, and were moderately limited for participants in the Endocrine, nutritional or metabolic diseases subgroup.

SF36 Role functioning/physical scale measures how physical health interferes with work or other activities. On average, participants in the Developmental anomalies subgroup scored higher than participants in

the Endocrine, nutritional or metabolic diseases subgroup. This indicates that physical health seldom interfered with work or other activities for participants in the Developmental anomalies subgroup, and almost always interfered for participants in the Endocrine, nutritional or metabolic diseases subgroup.

SF36 Energy/fatigue scale measures the proportion of energy or fatigue experienced. On average, participants in the Developmental anomalies subgroup scored higher than participants in the Diseases of the immune system subgroup. This indicates that participants in the Developmental anomalies subgroup were often fatigued, and participants in the Diseases of the immune system subgroup were almost always fatigued.

SF36 Social functioning scale measures limitations on social activities due to physical or emotional problems. On average, participants in the Developmental anomalies subgroup scored higher than participants in the Diseases of the immune system subgroup. This indicates that social activities were moderately limited for participants in the Developmental anomalies subgroup, and limited for participants in the Diseases of the immune system subgroup.

SF36 Pain scale measures how much pain, and how pain interferes with work and other activities. On average, participants in the Other rare condition subgroup scored higher than participants in the Diseases of the immune system subgroup. This indicates that participants in the Other rare condition subgroup had no pain, and participants in the Diseases of the immune system subgroup had moderate pain.

SF36 General health scale measures perception of health. On average, participants in the Developmental anomalies subgroup scored higher than participants in the Diseases of the immune system subgroup. This indicates that participants in the Developmental anomalies subgroup had average health, and participants in the Diseases of the immune system subgroup had poor health.

SF36 Health change scale measures health compared to a year ago. On average, participants in the Developmental anomalies subgroup scored higher than participants in the Endocrine, nutritional or metabolic diseases subgroup. This indicates that participants in the Developmental anomalies subgroup reported that their health was about the same as it was a year ago, and participants in the Endocrine, nutritional or metabolic diseases subgroup reported somewhat worse health.

Table 2.7: SF36 by condition summary statistics and Kruskal Wallis test

SF36 scale	Group	Number (n=379)	Percent	Median	IQR	C^2	dF	p-value
Physical functioning	Developmental anomalies	59	15.45	80.00	37.50	41.62	5	<0.0001*
	Diseases of the immune system	77	20.16	50.00	35.00			
	Diseases of the nervous system	93	24.35	65.00	80.00			
	Diseases of the skin	30	7.85	62.50	50.00			
	Endocrine, nutritional or metabolic diseases	95	24.87	45.00	60.00			
	Other rare condition	28	7.33	80.00	53.75			
Role functioning physical	Developmental anomalies	59	15.45	75.00	75.00	40.98	5	<0.0001*
	Diseases of the immune system	77	20.16	0.00	25.00			
	Diseases of the nervous system	93	24.35	25.00	100.00			
	Diseases of the skin	30	7.85	25.00	50.00			
	Endocrine, nutritional or metabolic diseases	95	24.87	0.00	50.00			
	Other rare condition	28	7.33	62.50	100.00			
Role functioning emotional	Developmental anomalies	59	15.45	66.67	100.00	5.46	5	0.3619
	Diseases of the immune system	77	20.16	33.33	100.00			
	Diseases of the nervous system	93	24.35	66.67	100.00			
	Diseases of the skin	30	7.85	50.00	100.00			
	Endocrine, nutritional or metabolic diseases	95	24.87	33.33	100.00			
	Other rare condition	28	7.33	50.00	100.00			
Energy/fatigue	Developmental anomalies	59	15.45	40.00	27.50	27.73	5	<0.0001*
	Diseases of the immune system	77	20.16	20.00	30.00			
	Diseases of the nervous system	93	24.35	35.00	30.00			
	Diseases of the skin	30	7.85	25.00	33.75			
	Endocrine, nutritional or metabolic diseases	95	24.87	30.00	32.50			
	Other rare condition	28	7.33	47.50	36.25			
Emotional well-being	Developmental anomalies	59	15.45	64.00	20.00	10.79	5	0.0558
	Diseases of the immune system	77	20.16	68.00	24.00			
	Diseases of the nervous system	93	24.35	72.00	28.00			
	Diseases of the skin	30	7.85	56.00	27.00			
	Endocrine, nutritional or metabolic diseases	95	24.87	72.00	20.00			
	Other rare condition	28	7.33	64.00	29.00			
Social functioning	Developmental anomalies	59	15.45	62.50	31.25	16.89	5	0.0047*
	Diseases of the immune system	77	20.16	37.50	37.50			
	Diseases of the nervous system	93	24.35	62.50	37.50			
	Diseases of the skin	30	7.85	50.00	46.88			
	Endocrine, nutritional or metabolic diseases	95	24.87	50.00	50.00			
	Other rare condition	28	7.33	62.50	40.63			
Pain	Developmental anomalies	59	15.45	77.50	35.00	51.60	5	<0.0001*
	Diseases of the immune system	77	20.16	45.00	32.50			
	Diseases of the nervous system	93	24.35	57.50	45.00			
	Diseases of the skin	30	7.85	45.00	35.00			
	Endocrine, nutritional or metabolic diseases	95	24.87	55.00	45.00			
	Other rare condition	28	7.33	85.00	30.63			
General health	Developmental anomalies	59	15.45	55.00	35.00	63.85	5	<0.0001*
	Diseases of the immune system	77	20.16	25.00	30.00			
	Diseases of the nervous system	93	24.35	50.00	25.00			
	Diseases of the skin	30	7.85	30.00	23.75			
	Endocrine, nutritional or metabolic diseases	95	24.87	35.00	32.50			
	Other rare condition	28	7.33	55.00	30.00			
Health change	Developmental anomalies	59	15.45	50.00	0.00	18.62	5	0.0023*
	Diseases of the immune system	77	20.16	50.00	50.00			
	Diseases of the nervous system	93	24.35	50.00	25.00			
	Diseases of the skin	30	7.85	50.00	0.00			
	Endocrine, nutritional or metabolic diseases	95	24.87	25.00	25.00			
	Other rare condition	28	7.33	50.00	0.00			

Table 2.8: SF36 by condition one-way post hoc Wilcoxon rank sum test

SF36 scale		Developmental anomalies	Diseases of the immune system	Diseases of the nervous system	Diseases of the skin	Endocrine, nutritional or metabolic diseases
Physical functioning	Diseases of the immune system	<0.0001*	-	-	-	-
	Diseases of the nervous system	0.0020*	0.1020	-	-	-
	Diseases of the skin	0.0074*	0.1502	0.9606	-	-
	Endocrine, nutritional or metabolic diseases	<0.0001*	0.9606	0.1928	0.2457	-
	Other rare condition	0.6390	0.0020*	0.0520	0.0856	0.0042*
Role functioning/physical	Diseases of the immune system	<0.0001*	-	-	-	-
	Diseases of the nervous system	0.0041*	0.0426*	-	-	-
	Diseases of the skin	0.0041*	0.1454	0.6610	-	-
	Endocrine, nutritional or metabolic diseases	<0.0001*	0.6610	0.0757	0.2746	-
	Other rare condition	0.4843	0.0041*	0.1454	0.1178	0.0048*
Energy/Fatigue	Diseases of the immune system	0.0008*	-	-	-	-
	Diseases of the nervous system	0.2361	0.0037*	-	-	-
	Diseases of the skin	0.0286*	0.5163	0.0994	-	-
	Endocrine, nutritional or metabolic diseases	0.0305*	0.1134	0.1799	0.5407	-
	Other rare condition	0.4522	0.0019	0.0994	0.0286*	0.0286*
Social functioning	Diseases of the immune system	0.0085*	-	-	-	-
	Diseases of the nervous system	0.4338	0.0294	-	-	-
	Diseases of the skin	0.1573	0.5079	0.3404	-	-
	Endocrine, nutritional or metabolic diseases	0.0377*	0.4338	0.1775	0.9651	-
	Other rare condition	0.5079	0.2246	0.9651	0.5079	0.4542
Pain	Diseases of the immune system	<0.0001*	-	-	-	-
	Diseases of the nervous system	0.1258	<0.0001*	-	-	-
	Diseases of the skin	0.0002*	0.8722	0.0027*	-	-
	Endocrine, nutritional or metabolic diseases	0.0027*	0.0027	0.0829	0.0472*	-
	Other rare condition	0.4652	<0.0001*	0.0639	0.0002*	0.0027*
General health	Diseases of the immune system	<0.0001*	-	-	-	-
	Diseases of the nervous system	0.1687	0.0000*	-	-	-
	Diseases of the skin	0.0010*	0.1842	0.0025*	-	-
	Endocrine, nutritional or metabolic diseases	0.0004*	0.0066*	0.0018*	0.5193	-
	Other rare condition	0.8411	<0.0001*	0.1687	0.0011*	0.0011*
Health change	Diseases of the immune system	0.3359	-	-	-	-
	Diseases of the nervous system	0.2679	0.9669	-	-	-
	Diseases of the skin	0.9669	0.4694	0.3537	-	-
	Endocrine, nutritional or metabolic diseases	0.0045*	0.2998	0.1501	0.0261	-
	Other rare condition	0.4694	0.2679	0.1695	0.5049	0.0051*

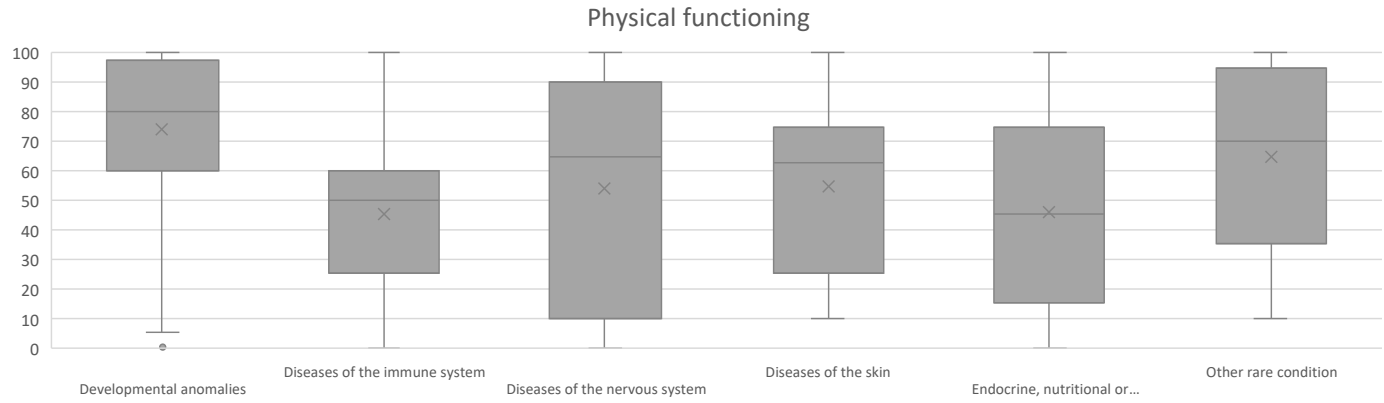


Figure 2.4: Boxplot of SF36 Physical functioning by condition

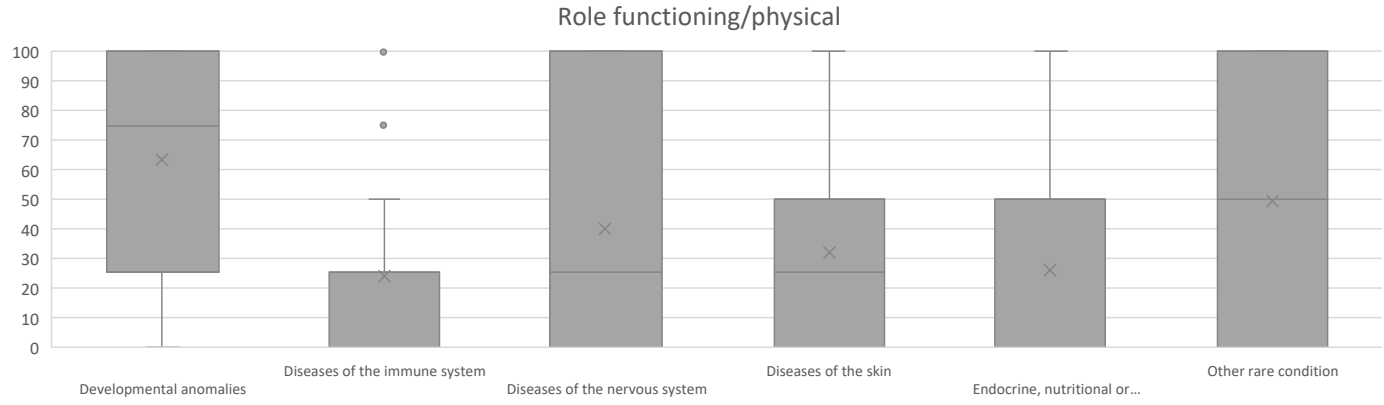


Figure 2.5: Boxplot of SF36 Role functioning/physical by condition

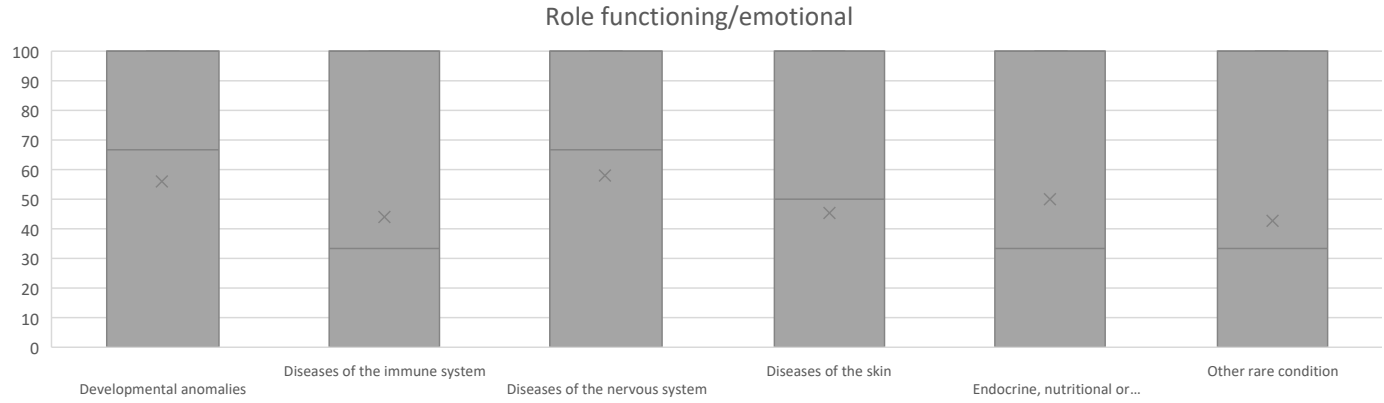


Figure 2.6: Boxplot of SF36 Role functioning/emotional by condition

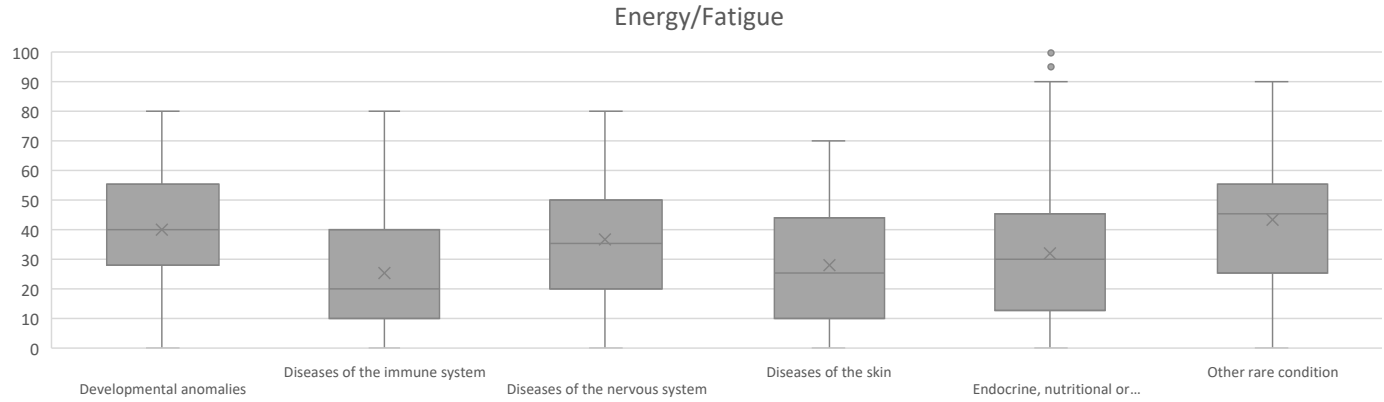


Figure 2.7: Boxplot of SF36 Energy/fatigue by condition

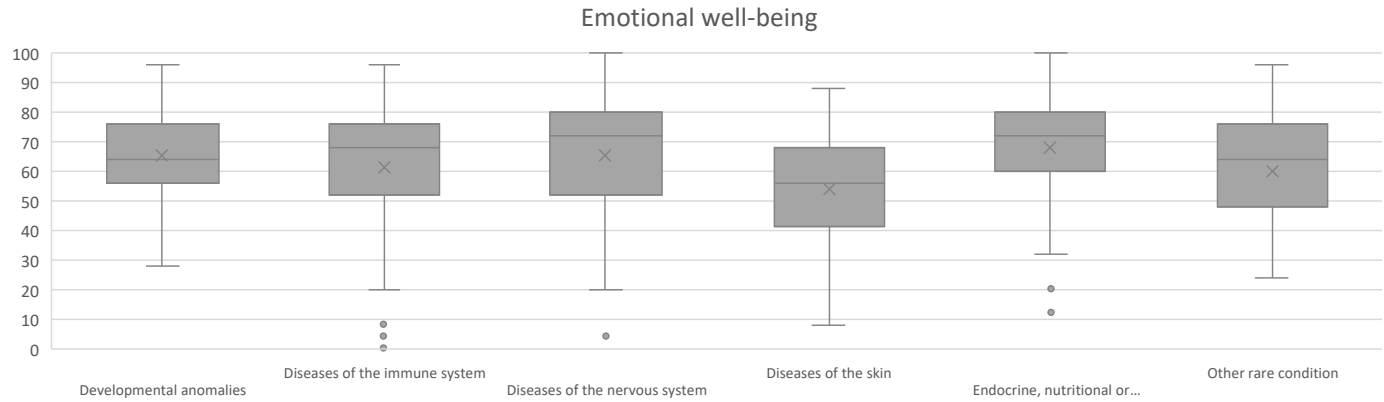


Figure 2.8: Boxplot of SF36 Emotional well-being by condition

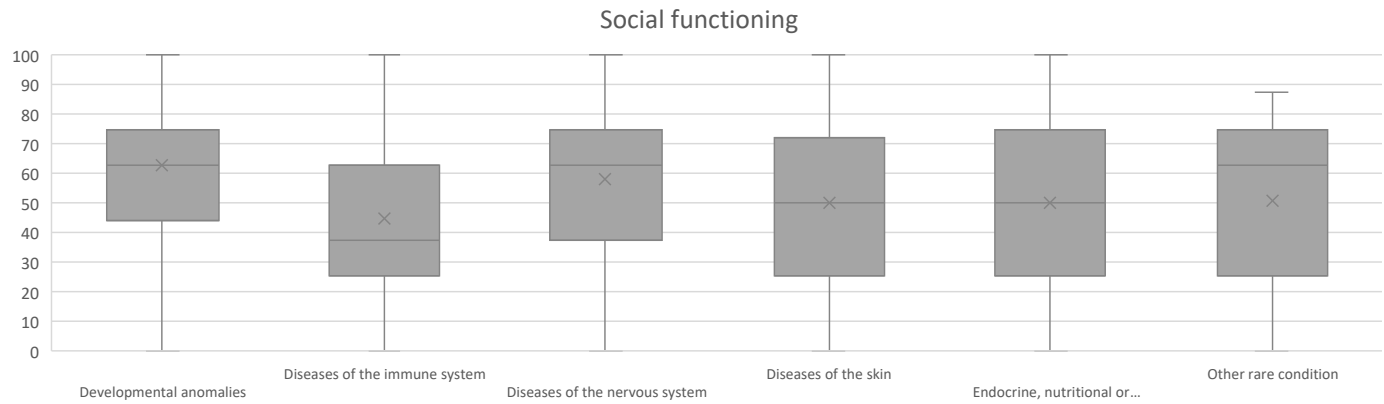


Figure 2.9: Boxplot of SF36 Social functioning by condition

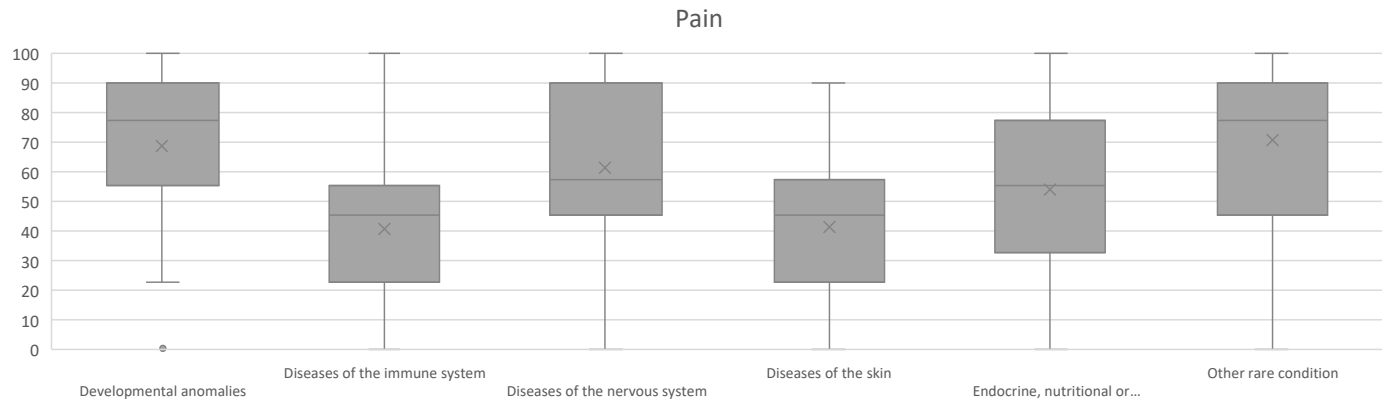


Figure 2.10: Boxplot of SF36 Pain by condition

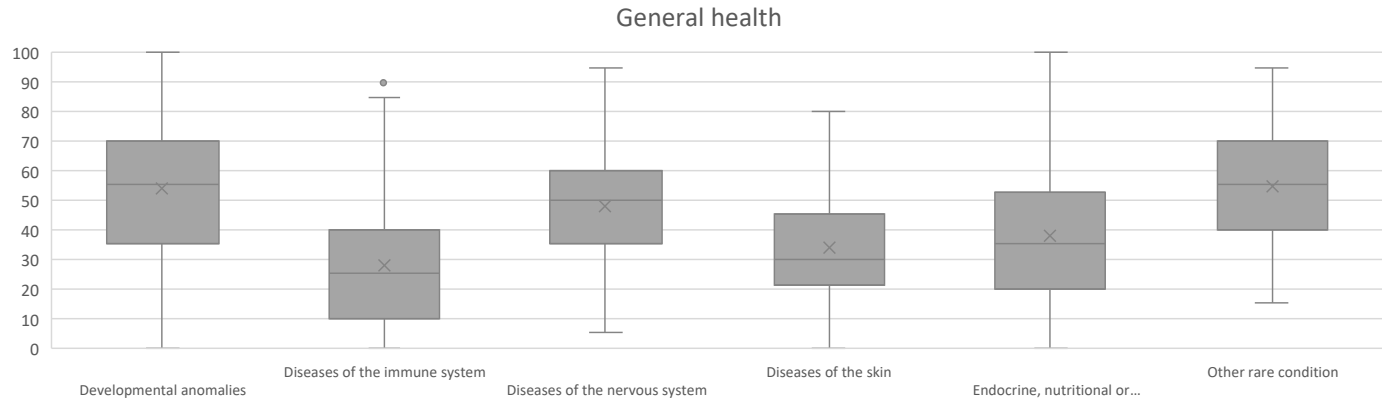


Figure 2.11: Boxplot of SF36 General health by condition

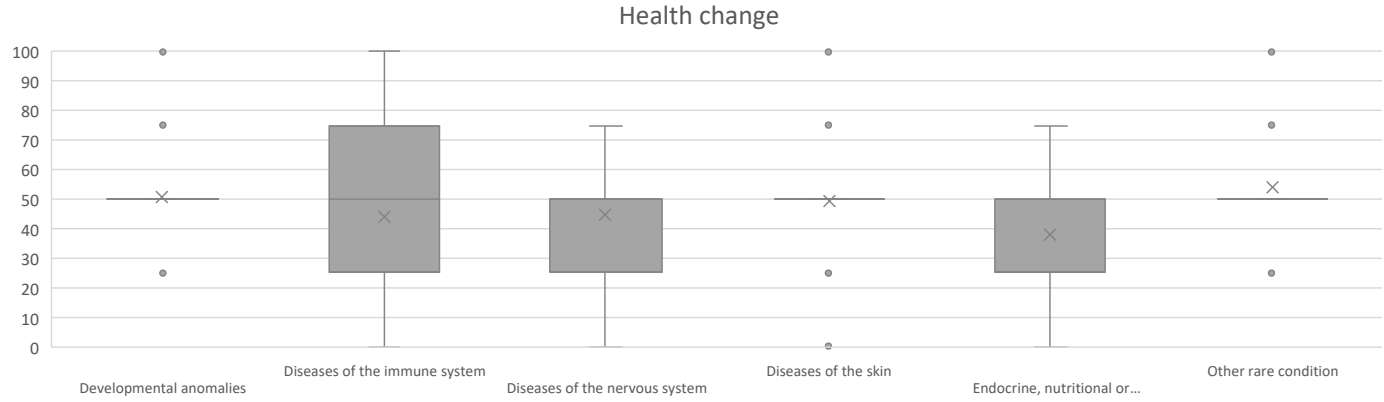


Figure 2.12: Boxplot of SF36 Health change by condition

SF36 by participant type

Comparisons were made by **type of participant** there were 256 participants (67.02%) with person with condition and, 126 participants (32.98%) with carer.

Assumptions for normality and variance for a two-sample t-test were not met, a Wilcoxon rank sum test with continuity correction was used.

Wilcoxon rank sum tests with continuity correction indicated that the median score for the **SF36 Physical functioning** scale [W = 9112.50, p<0.0001] was significantly lower for participants in the Person with condition subgroup (Median = 50.00, IQR = 50.00) compared to participants in the Carer subgroup (Median = 82.50, IQR = 45.00).

Wilcoxon rank sum tests with continuity correction indicated that the median score for the **SF36 Role functioning physical** scale [W = 10401.00, p<0.0001] was significantly lower for participants in the Person with condition subgroup (Median = 0.00, IQR = 50.00) compared to participants in the Carer subgroup (Median = 75.00, IQR = 100.00).

Wilcoxon rank sum tests with continuity correction indicated that the median score for the **SF36 Energy/fatigue** scale [W = 12864.00, p = 0.001] was

significantly lower for participants in the Person with condition subgroup (Median = 30.00, IQR = 30.00) compared to participants in the Carer subgroup (Median = 40.00, IQR = 28.75).

Wilcoxon rank sum tests with continuity correction indicated that the median score for the **SF36 Social functioning** scale [W = 12984.00, p = 0.002] was significantly lower for participants in the Person with condition subgroup (Median = 50.00, IQR = 50.00) compared to participants in the Carer subgroup (Median = 62.50, IQR = 37.50).

Wilcoxon rank sum tests with continuity correction indicated that the median score for the **SF36 Pain** scale [W = 10296.00, p<0.0001] was significantly lower for participants in the Person with condition subgroup (Median = 45.00, IQR = 45.00) compared to participants in the Carer subgroup (Median = 77.50, IQR = 45.00).

Wilcoxon rank sum tests with continuity correction indicated that the median score for the **SF36 General health** scale [W = 8901.00, p<0.0001] was significantly lower for participants in the Person with condition subgroup (Median = 32.50, IQR = 30.00) compared to participants in the Carer subgroup (Median = 55.00, IQR = 30.00).

Wilcoxon rank sum tests with continuity correction indicated that the median score for the **SF36 Health change** scale [W = 14486.00, p = 0.086] was significantly lower for participants in the Person with condition subgroup (Median = 50.00, IQR = 25.00) compared to participants in the Carer subgroup (Median = 50.00, IQR = 25.00).

Wilcoxon rank sum tests with continuity correction indicated that the median score for the **SF36 Physical functioning** scale [W = 9112.50, p<0.0001] was significantly lower for participants in the Person with condition subgroup (Median = 50.00, IQR = 50.00) compared to participants in the Carer subgroup (Median = 82.50, IQR = 45.00).

SF36 Physical functioning scale measures health limitations in physical activities such as walking, bending, climbing stairs, exercise, and housework. On average, participants in the Carer subgroup scored higher than participants in the Person with condition subgroup. This indicates that physical activities were not limited for participants in the Carer subgroup, and were slightly limited for participants in the Person with condition subgroup.

SF36 Role functioning/physical scale measures how physical health interferes with work or other activities. On average, participants in the Carer subgroup scored higher than participants in the Person with condition subgroup. This indicates that physical health seldom interfered with work or other activities for participants in the Carer subgroup, and almost always interfered for participants in the Person with condition subgroup.

SF36 Energy/fatigue scale measures the proportion of energy or fatigue experienced. On average, participants in the Carer subgroup had a higher score for energy/fatigue compared to Person with condition, however, both groups were often fatigued.

SF36 Social functioning scale measures **l**imitations on social activities due to physical or emotional problems. On average, participants in the Carer subgroup scored higher than participants in the Person with condition subgroup. This indicates that social activities were slightly limited for participants in the Carer subgroup, and moderately limited for participants in the Person with condition subgroup.

SF36 Pain scale measures how much pain, and how pain interferes with work and other activities. On average, participants in the Carer subgroup scored higher than participants in the Person with condition subgroup. This indicates that participants in the Carer subgroup had mild pain, and participants in the Person with condition subgroup had moderate pain.

SF36 General health scale measures perception of health. On average, participants in the Carer subgroup scored higher than participants in the Person with condition subgroup. This indicates that participants in the Carer subgroup had good health, and participants in the Person with condition subgroup had average health.

Table 2.9: SF36 by participant type summary statistics and Wilcoxon test

SF36 scale	Group	Number (n=382)	Percent	Median	IQR	W	p-value
Physical functioning	Person with condition	256	67.02	50.00	50.00	9112.50	<0.0001*
	Carer	126	32.98	82.50	45.00		
Role functioning/physical	Person with condition	256	67.02	0.00	50.00	10401.00	<0.0001*
	Carer	126	32.98	75.00	100.00		
Role functioning/emotional	Person with condition	256	67.02	50.00	100.00	15462.00	0.4863
	Carer	126	32.98	66.67	100.00		
Energy/Fatigue	Person with condition	256	67.02	30.00	30.00	12864.00	0.0013*
	Carer	126	32.98	40.00	28.75		
Emotional well-being	Person with condition	256	67.02	68.00	25.00	15302.00	0.4145
	Carer	126	32.98	68.00	23.00		
Social functioning	Person with condition	256	67.02	50.00	50.00	12984.00	0.0018*
	Carer	126	32.98	62.50	37.50		
Pain	Person with condition	256	67.02	45.00	45.00	10296.00	<0.0001*
	Carer	126	32.98	77.50	45.00		
General health	Person with condition	256	67.02	32.50	30.00	8901.00	<0.0001*
	Carer	126	32.98	55.00	30.00		
Health change	Person with condition	256	67.02	50.00	25.00	14486.00	0.0864
	Carer	126	32.98	50.00	25.00		

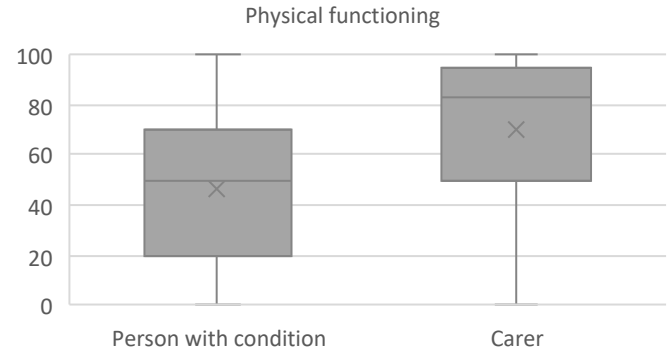


Figure 2.13: Boxplot of SF36 Physical functioning by participant type

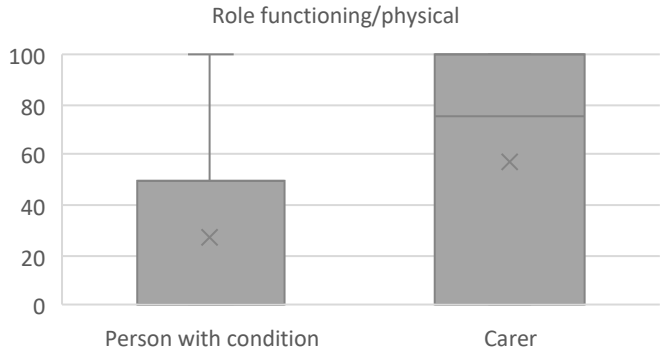


Figure 2.14: Boxplot of SF36 Role functioning/physical by participant type

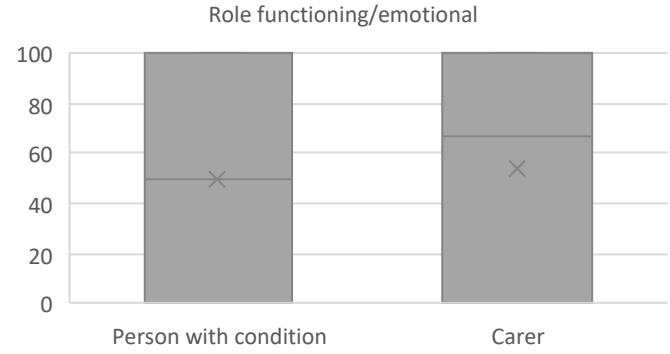


Figure 2.15: Boxplot of SF36 Role functioning/emotional by participant type

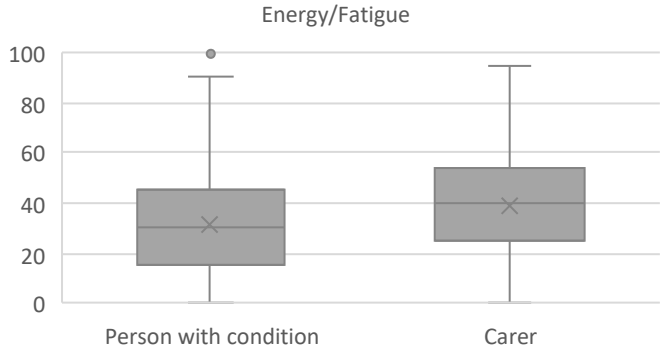


Figure 2.16: Boxplot of SF36 Energy/fatigue by participant type

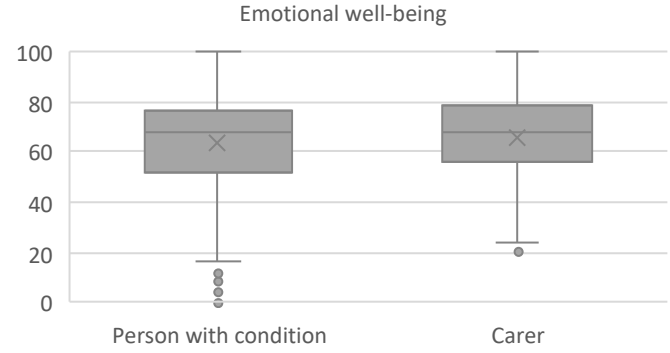


Figure 2.17: Boxplot of SF36 Emotional well-being by participant type

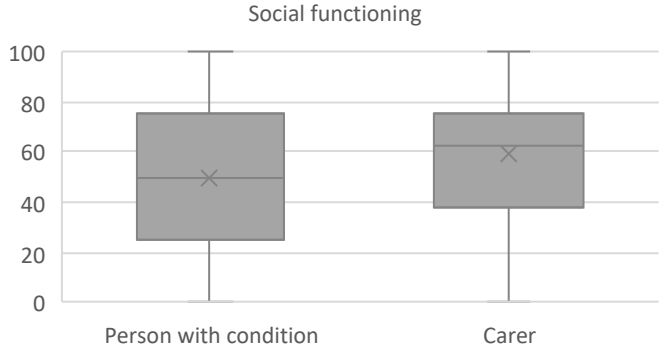


Figure 2.18: Boxplot of SF36 Social functioning by participant type

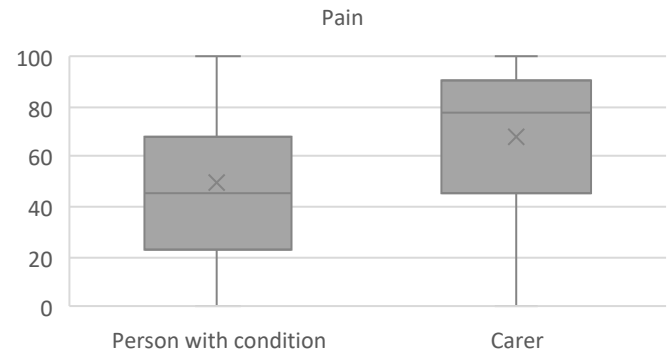


Figure 2.19: Boxplot of SF36 Pain by participant type

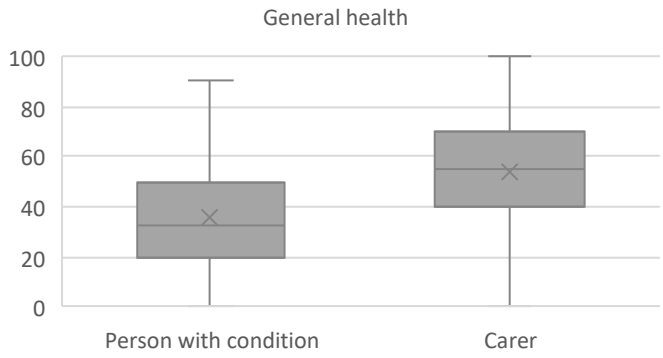


Figure 2.20: Boxplot of SF36 General health by participant type

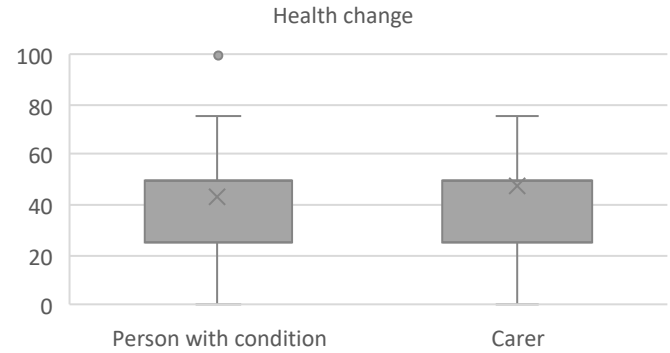


Figure 2.21: Boxplot of SF36 Health change by participant type

SF36 by gender

Comparisons were made by **gender**, there were 285 female participants (75.00%), and 95 male particpants (25.00%).

Assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used.

Wilcoxon rank sum tests with continuity correction indicated that the median score for the SF36 Physical functioning scale [W = 10896.00, p = 0.004] was significantly lower for participants in the Female subgroup (Median = 55.00, IQR = 60.00) compared to participants in the Male subgroup (Median = 65.00, IQR = 65.00).

Wilcoxon rank sum tests with continuity correction indicated that the median score for the SF36 Role functioning physical scale [W = 11180.00, p = 0.006] was significantly lower for participants in the Female subgroup (Median = 0.00, IQR = 75.00) compared to participants in the Male subgroup (Median = 50.00, IQR = 100.00).

Wilcoxon rank sum tests with continuity correction indicated that the median score for the SF36 Energy/fatigue scale [W = 10618.00, p = 0.002] was significantly lower for participants in the Female subgroup (Median = 30.00, IQR = 30.00) compared to participants in the Male subgroup (Median = 40.00, IQR = 32.50).

Wilcoxon rank sum tests with continuity correction indicated that the median score for the SF36 Social functioning scale [W = 10658.00, p = 0.002] was significantly lower for participants in the Female subgroup (Median = 50.00, IQR = 50.00) compared to participants in the Male subgroup (Median = 62.50, IQR = 50.00).

Wilcoxon rank sum tests with continuity correction indicated that the median score for the SF36 Pain scale [W = 9887.00, p<0.0001] was significantly lower for participants in the Female subgroup (Median = 45.00, IQR = 45.00) compared to participants in the Male subgroup (Median = 67.50, IQR = 45.00).

Wilcoxon rank sum tests with continuity correction indicated that the median score for the SF36 General health scale [W = 10317.00, p = 0.001] was significantly lower for participants in the Female subgroup (Median

= 35.00, IQR = 35.00) compared to participants in the Male subgroup (Median = 50.00, IQR = 40.00).

SF36 Physical functioning scale measures health limitations in physical activities such as walking, bending, climbing stairs, exercise, and housework. On average, participants in the Male subgroup scored higher than participants in the Female subgroup. This indicates that physical activities were slightly limited for participants in the Male subgroup, and were moderately limited for participants in the Female subgroup.

SF36 Role functioning/physical scale measures how physical health interferes with work or other activities. On average, participants in the Male subgroup scored higher than participants in the Female subgroup. This indicates that physical health sometimes interfered with work or other activities for participants in the Male subgroup, and almost always interfered for participants in the Female subgroup.

SF36 Energy/fatigue scale measures the proportion of energy or fatigue experienced. On average, participants in the Male subgroup had a higher score for energy/fatigue compared to Female, however, both groups were often fatigued.

SF36 Social functioning scale measures limitations on social activities due to physical or emotional problems. On average, participants in the Male subgroup scored higher than participants in the Female subgroup. This indicates that social activities were slightly limited for participants in the Male subgroup, and moderately limited for participants in the Female subgroup.

SF36 Pain scale measures how much pain, and how pain interferes with work and other activities. On average, participants in the Male subgroup scored higher than participants in the Female subgroup. This indicates that participants in the Male subgroup had mild pain, and participants in the Female subgroup had moderate pain.

SF36 General health scale measures perception of health. On average, participants in the Male subgroup scored higher than participants in the Female subgroup. This indicates that participants in the Male subgroup had average health, and participants in the Female subgroup had poor health.

Table 2.10: SF36 by gender summary statistics and T-test

SF36 scale	Group	Number (n=380)	Percent	Median	IQR	W	p-value
Physical functioning	Female	285	75.00	55.00	60.00	10896.00	0.0043*
	Male	95	25.00	65.00	65.00		
Role functioning/physical	Female	285	75.00	0.00	75.00	11180.00	0.0061*
	Male	95	25.00	50.00	100.00		
Role functioning/emotional	Female	285	75.00	66.67	100.00	12814.00	0.4083
	Male	95	25.00	66.67	100.00		
Energy/Fatigue	Female	285	75.00	30.00	30.00	10618.00	0.0016*
	Male	95	25.00	40.00	32.50		
Emotional well-being	Female	285	75.00	68.00	24.00	12124.00	0.1266
	Male	95	25.00	72.00	24.00		
Social functioning	Female	285	75.00	50.00	50.00	10658.00	0.0017*
	Male	95	25.00	62.50	50.00		
Pain	Female	285	75.00	45.00	45.00	9887.00	0.0001*
	Male	95	25.00	67.50	45.00		
General health	Female	285	75.00	35.00	35.00	10317.00	0.0005*
	Male	95	25.00	50.00	40.00		
Health change	Female	285	75.00	50.00	25.00	12593.00	0.2808
	Male	95	25.00	50.00	25.00		

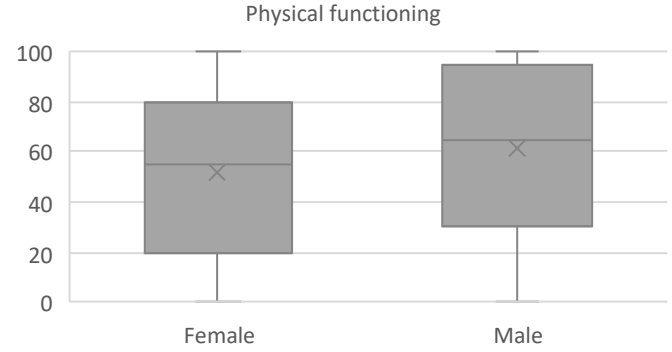


Figure 2.22: Boxplot of SF36 Physical functioning by gender

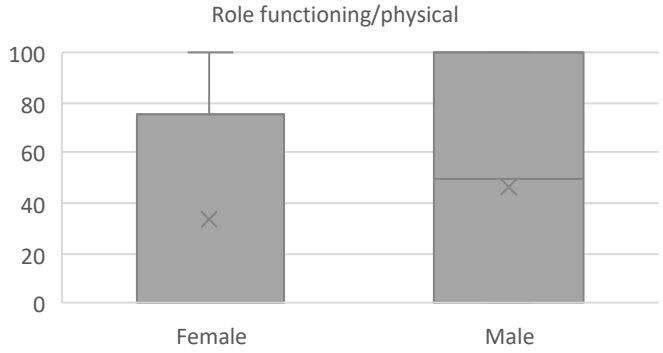


Figure 2.23: Boxplot of SF36 Role functioning/physical by gender

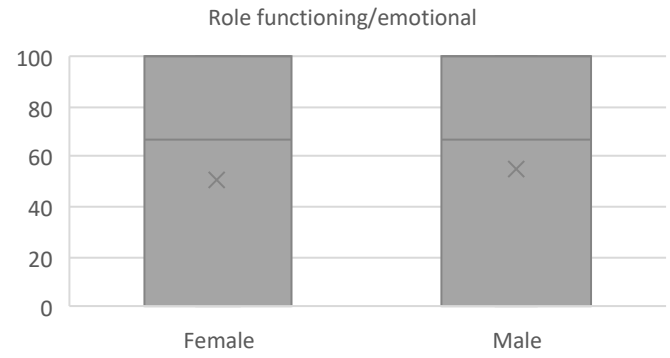


Figure 2.24: Boxplot of SF36 Role functioning/emotional by gender

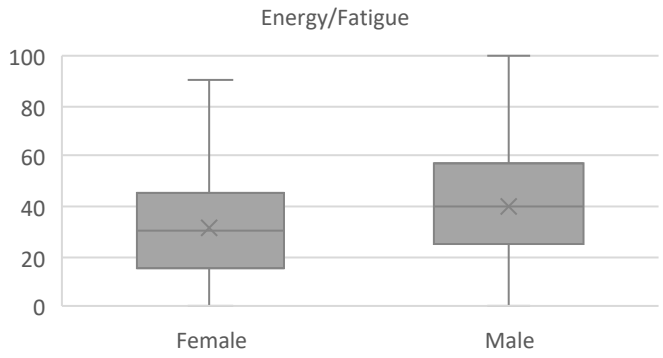


Figure 2.25: Boxplot of SF36 Energy/fatigue by gender

Figure 2.26: Boxplot of SF36 Emotional well-being by gende

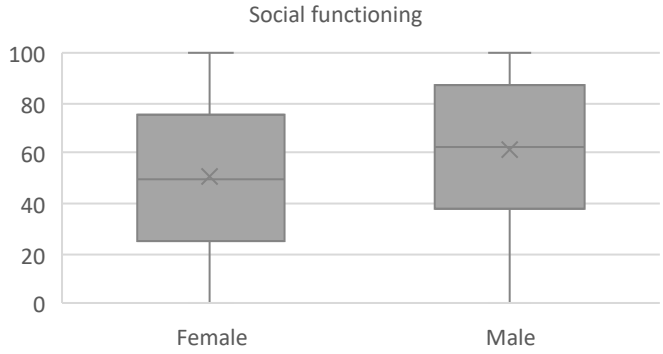


Figure 2.27: Boxplot of SF36 Social functioning by gender

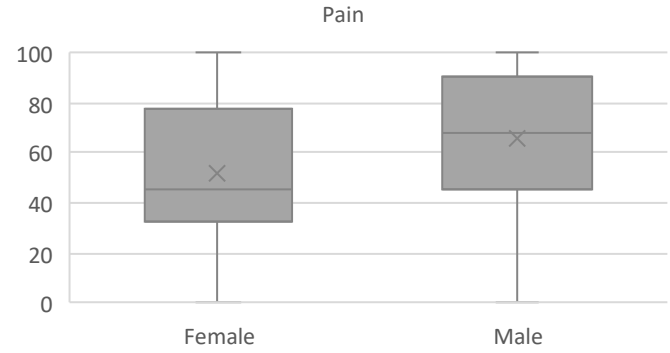


Figure 2.28: Boxplot of SF36 Pain by gender

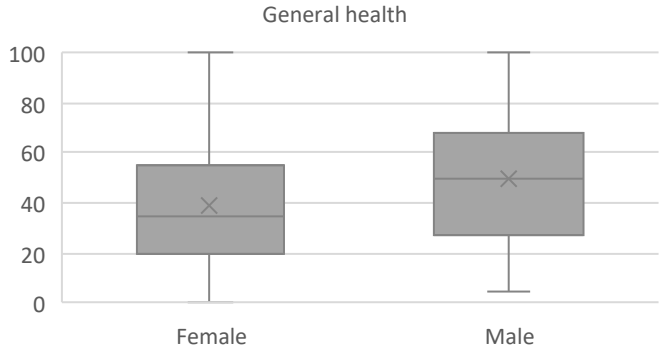


Figure 2.29: Boxplot of SF36 General health by gender

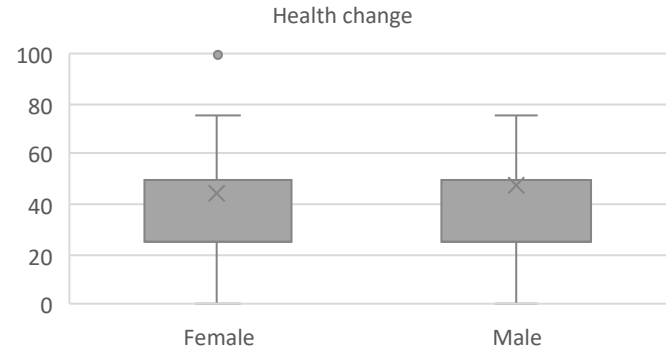


Figure 2.30: Boxplot of SF36 Health change by gender

SF36 by age

Comparisons were made by **age** of person with condition. There were 90 participants (23.56%) with aged under 18, 125 participants (32.72%) with aged 18 to 44, 109 participants (28.53%) with aged 45 to 64, and 58 participants (15.18%) with aged 65 or older.

The assumptions for normality of residuals was not met, a Kruskal-Wallis test was used. Post hoc pairwise comparisons using Wilcoxon rank sum test was used to identify the source of any differences identified in the Kruskal -Wallis test.

A Kruskal-Wallis test indicated a statistically significant difference in the SF36 Physical functioningscale between groups, $\chi 2(3) = 52.29$ $p<0.0001$

The largest significant difference was between Aged under 18(median = 85, IQR = 35), and Aged 45 to 64(median = 40, IQR = 55, $p<0.0001$). A Kruskal-Wallis test indicated a statistically significant difference in the SF36 Role functioning physicalscale between groups, $\chi 2(3) = 48.891$ $p<0.0001$

The largest significant difference was between Aged under 18(median = 75, IQR = 75), and Aged 65 or older(median = 0, IQR = 50, $p<0.0001$). A Kruskal-Wallis test indicated a statistically significant difference in the SF36 Energy/fatiguescale between groups, $\chi^2(3) = 9.2464$ $P = 0.0262$

The largest significant difference was between Aged under 18(median = 40, IQR = 33.75), and Aged 18 to 44(median = 30, IQR = 30, $p = 0.012$). A Kruskal-Wallis test indicated a statistically significant difference in the SF36 Emotional well-beingscale between groups, $\chi^2(3) = 12.741$ $P = 0.0052$

The largest significant difference was between Aged under 18(median = 72, IQR = 24), and Aged 18 to 44(median = 64, IQR = 28, $p = 0.014$). A Kruskal-Wallis test indicated a statistically significant difference in the SF36 Social functioningscale between groups, $\chi^2(3) = 10.418$ $P = 0.0153$

The largest significant difference was between Aged under 18(median = 62.5, IQR = 50), and Aged 18 to 44(median = 50, IQR = 50, $p = 0.0091$). A Kruskal-Wallis test indicated a statistically significant difference in the SF36 Painscale between groups, $\chi^2(3) = 33.501$ $p<0.0001$.

The largest significant difference was between Aged under 18(median = 77.5, IQR = 55), and Aged 18 to 44(median = 55, IQR = 45, $p<0.0001$). A Kruskal-Wallis test indicated a statistically significant difference in the SF36 General healthscale between groups, $\chi^2(3) = 58.747$ $p<0.0001$. The largest significant difference was between Aged under 18(median = 60, IQR = 30), and Aged 45 to 64(median = 30, IQR = 30, $p<0.0001$).

A Kruskal-Wallis test indicated a statistically significant difference in the SF36 Health changescale between groups, $\chi^2(3) = 11.104$ $P = 0.0112$. The largest significant difference was between Aged under 18(median = 50, IQR = 0), and Aged 65 or older(median = 25, IQR = 25, $p = 0.0046$).

SF36 Physical functioning scale measures health limitations in physical activities such as walking, bending, climbing stairs, exercise, and housework. On average, participants in the Aged under 18subgroup scored higher than participants in the Aged 45 to 64subgroup. This indicates that physical activities were not limited for participants in the Aged under 18subgroup, and were slightly limited for participants in the Aged 45 to 64subgroup.

SF36 Role functioning/physical scale measures how physical health interferes with work or other activities. On average, participants in the Aged under 18 subgroup scored higher than participants in the Aged 65 or older subgroup. This indicates that physical health sometimes interfered with work or other activities for participants in the Aged under 18subgroup, and almost always interfered for participants in the Aged 45 to 64 subgroup.

SF36 Energy/fatigue scale measures the proportion of energy or fatigue experienced. On average, participants in the Aged under 18 subgroup had a higher score for energy/fatigue compared to Aged 18 to 44, however, both groups were often fatigued.

SF36 Emotional well-being scale measures how a person feels, for example happy, calm, depressed or anxious. On average, participants in the Aged under 18 subgroup had a higher score for emotional well-being compared to Aged 18 to 44, however, both groups had good emotional well-being.

SF36 Social functioning scale measures limitations on social activities due to physical or emotional problems. On average, participants in the Aged under 18 subgroup scored higher than participants in the Aged 18 to 44 subgroup. This indicates that social activities were slightly limited for participants in the Aged under 18 subgroup, and moderately limited for participants in the Aged 18 to 44 subgroup.

SF36 Pain scale measures how much pain, and how pain interferes with work and other activities. On average, participants in the Aged under 18subgroup scored higher than participants in the Aged 18 to 44 subgroup. This indicates that participants in the Aged under 18subgroup had mild pain, and participants in the Aged 18 to 44 subgroup had moderate pain.

SF36 General health scale measures perception of health. On average, participants in the Aged under 18subgroup scored higher than participants in the Aged 45 to 64subgroup. This indicates that participants in the Aged under 18subgroup had average health, and participants in the Aged 45 to 64subgroup had poor health.

SF36 Health change scale measures health compared to a year ago. On average, participants in the Aged under 18subgroup scored higher than participants in the Aged 65 or older subgroup. This indicates that participants in the Aged under 18subgroup reported that their health was about the same as it was a year ago, and participants in the Aged 65 or oldersubgroup reported somewhat worse health.

Table 2.11: SF36 by age summary statistics and Kruskal Wallis test

SF36 scale	Group	Number (n=382)	Percent	Median	IQR	C^2	dF	p-value
Physical functioning	Aged under 18	90	23.56	85.00	35.00	52.29	3	<0.0001*
	Aged 18 to 44	125	32.72	50.00	60.00			
	Aged 45 to 64	109	28.53	40.00	55.00			
	Aged 65 or older	58	15.18	42.50	53.75			
Role functioning physical	Aged under 18	90	23.56	75.00	75.00	48.891	3	<0.0001*
	Aged 18 to 44	125	32.72	0.00	75.00			
	Aged 45 to 64	109	28.53	0.00	50.00			
	Aged 65 or older	58	15.18	0.00	50.00			
Role functioning emotional	Aged under 18	90	23.56	66.67	100.00	1.4131	3	0.7025
	Aged 18 to 44	125	32.72	33.33	100.00			
	Aged 45 to 64	109	28.53	66.67	100.00			
	Aged 65 or older	58	15.18	66.67	100.00			
Energy/fatigue	Aged under 18	90	23.56	40.00	33.75	9.2464	3	0.0262*
	Aged 18 to 44	125	32.72	30.00	30.00			
	Aged 45 to 64	109	28.53	30.00	35.00			
	Aged 65 or older	58	15.18	35.00	38.75			
Emotional well-being	Aged under 18	90	23.56	72.00	24.00	12.741	3	0.0052*
	Aged 18 to 44	125	32.72	64.00	28.00			
	Aged 45 to 64	109	28.53	68.00	28.00			
	Aged 65 or older	58	15.18	72.00	20.00			
Social functioning	Aged under 18	90	23.56	62.50	50.00	10.418	3	0.0153*
	Aged 18 to 44	125	32.72	50.00	50.00			
	Aged 45 to 64	109	28.53	50.00	50.00			
	Aged 65 or older	58	15.18	50.00	50.00			
Pain	Aged under 18	90	23.56	77.50	55.00	33.501	3	<0.0001*
	Aged 18 to 44	125	32.72	55.00	45.00			
	Aged 45 to 64	109	28.53	45.00	45.00			
	Aged 65 or older	58	15.18	55.00	55.00			
General health	Aged under 18	90	23.56	60.00	30.00	58.747	3	<0.0001*
	Aged 18 to 44	125	32.72	35.00	35.00			
	Aged 45 to 64	109	28.53	30.00	30.00			
	Aged 65 or older	58	15.18	35.00	38.75			
Health change	Aged under 18	90	23.56	50.00	0.00	11.104	3	0.0112*
	Aged 18 to 44	125	32.72	50.00	25.00			
	Aged 45 to 64	109	28.53	50.00	25.00			
	Aged 65 or older	58	15.18	25.00	25.00			

Table 2.12: SF36 by age one-way post hoc Wilcoxon rank sum test

SF36 scale		Aged under 18	Aged 18 to 44	Aged 45 to 64
Physical functioning	Aged 18 to 44	<0.0001*	-	-
	Aged 45 to 64	<0.0001*	0.1300	-
	Aged 65 or older	<0.0001*	0.5800	0.5900
Role functioning/physical	Aged 18 to 44	<0.0001*	-	-
	Aged 45 to 64	<0.0001*	0.1200	-
	Aged 65 or older	<0.0001*	0.5900	0.4900
Energy/Fatigue	Aged 18 to 44	0.0120*	-	-
	Aged 45 to 64	0.1880	0.3930	-
	Aged 65 or older	0.3980	0.3590	0.6860
Emotional well-being	Aged 18 to 44	0.1130	-	-
	Aged 45 to 64	0.3090	0.0140*	-
	Aged 65 or older	0.3090	0.0140*	0.8750
Social functioning	Aged 18 to 44	0.0091*	-	-
	Aged 45 to 64	0.0682	0.6163	-
	Aged 65 or older	0.0682	0.7714	0.7714
Pain	Aged 18 to 44	<0.0001*	-	-
	Aged 45 to 64	<0.0001*	0.1340	-
	Aged 65 or older	0.0006*	0.7703	0.4042
General health	Aged 18 to 44	<0.0001*	-	-
	Aged 45 to 64	<0.0001*	0.2600	-
	Aged 65 or older	<0.0001*	0.6900	0.2600
Health change	Aged 18 to 44	0.3059	-	-
	Aged 45 to 64	0.0574	0.3059	-
	Aged 65 or older	0.0046*	0.0754	0.3102

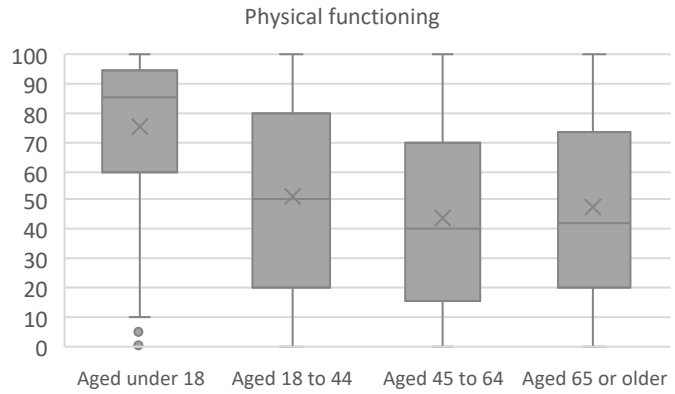


Figure 2.31: Boxplot of SF36 Physical functioning by age

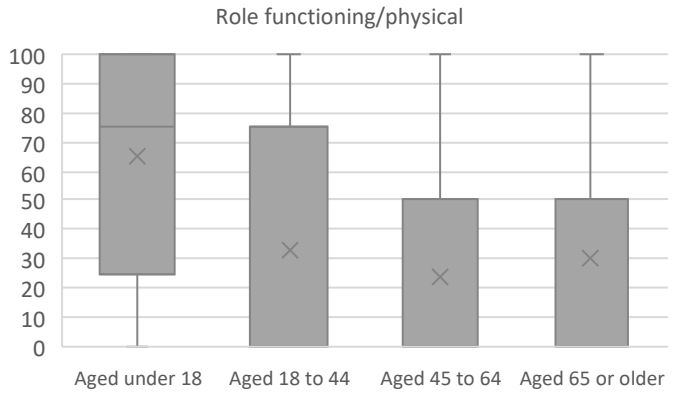


Figure 2.32: Boxplot of SF36 Role functioning/physical by age

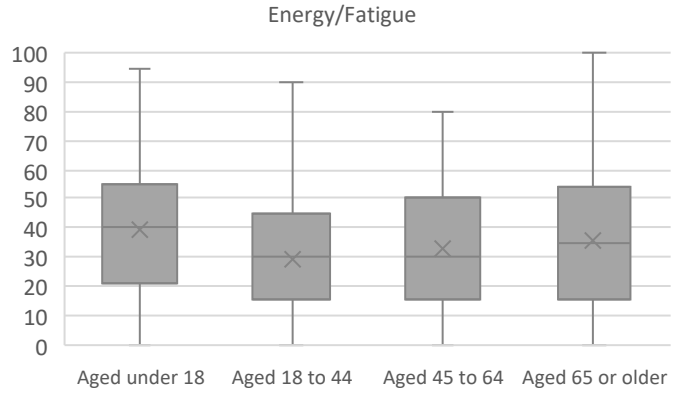


Figure 2.33: Boxplot of SF36 Role functioning/emotional by age

Figure 2.34: Boxplot of SF36 Energy/fatigue by age

Figure 2.35: Boxplot of SF36 Emotional well-being by age

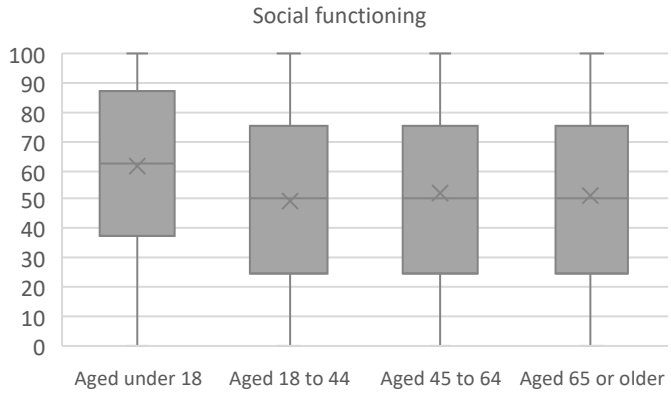


Figure 2.36: Boxplot of SF36 Social functioning by age

Figure 2.37: Boxplot of SF36 Pain by age

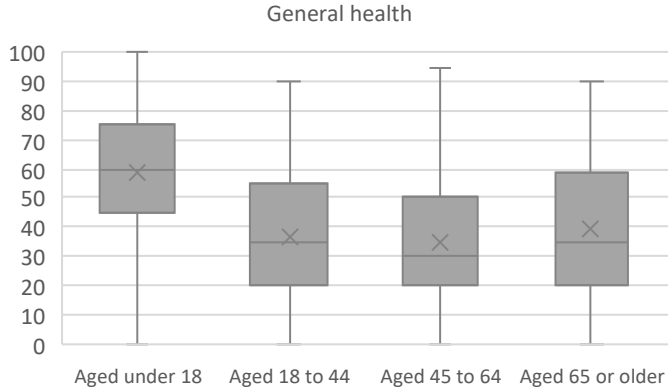


Figure 2.38: Boxplot of SF36 General health by age

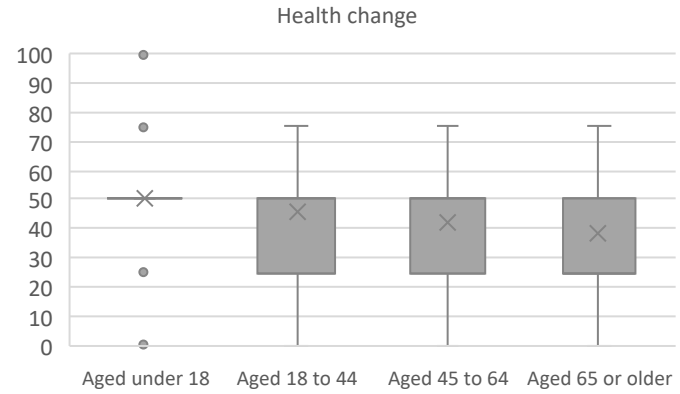


Figure 2.39: Boxplot of SF36 Health change by age

SF36 by education

Comparisons were made by **education** status, between those with trade or high school qualifications (n=185, 49.47%), and those with a university qualification (n=189, 50.53%).

Assumptions for normality and variance for a two-sample t-test were not met, a Wilcoxon rank sum test with continuity correction was used.

No significant differences were observed between participants by **education** for any of the SF36 scales.

Table 2.13: SF36 by education summary statistics and Wilcoxon test

SF36 scale	Group	Number (n=374)	Percent	Median	IQR	W	p-value
Physical functioning	Trade or high school	185	49.47	55.00	55.00	16016.00	0.1601
	University	189	50.53	60.00	60.00		
Role functioning/physical	Trade or high school	185	49.47	0.00	75.00	16742.00	0.4467
	University	189	50.53	25.00	100.00		
Role functioning/emotional	Trade or high school	185	49.47	33.33	100.00	16774.00	0.4728
	University	189	50.53	66.67	100.00		
Energy/Fatigue	Trade or high school	185	49.47	30.00	30.00	15916.00	0.1332
	University	189	50.53	35.00	35.00		
Emotional well-being	Trade or high school	185	49.47	64.00	24.00	16718.00	0.4638
	University	189	50.53	68.00	28.00		
Social functioning	Trade or high school	185	49.47	50.00	50.00	16817.00	0.5214
	University	189	50.53	50.00	37.50		
Pain	Trade or high school	185	49.47	55.00	55.00	16110.00	0.1870
	University	189	50.53	57.50	35.00		
General health	Trade or high school	185	49.47	40.00	35.00	15653.00	0.0796
	University	189	50.53	45.00	35.00		
Health change	Trade or high school	185	49.47	50.00	25.00	16794.00	0.4850
	University	189	50.53	50.00	25.00		

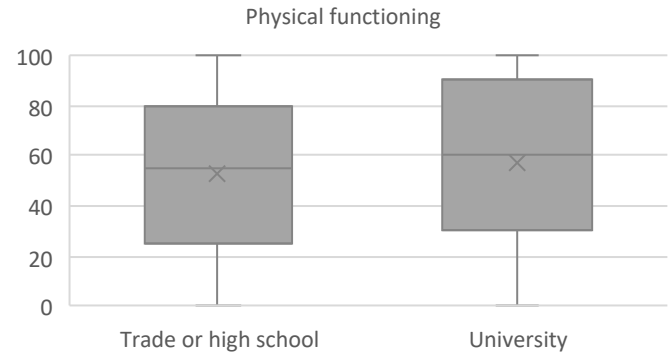


Figure 2.40: Boxplot of SF36 Physical functioning by education

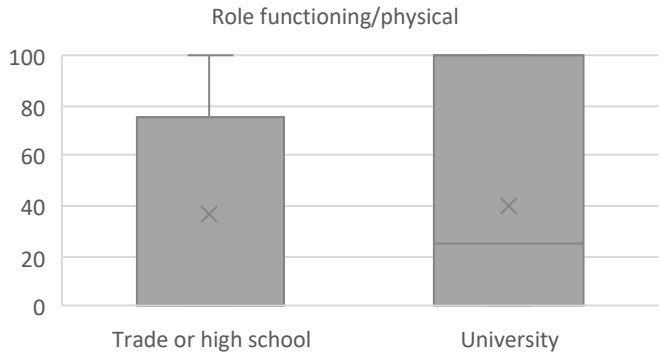


Figure 2.41: Boxplot of SF36 Role functioning/physical by education

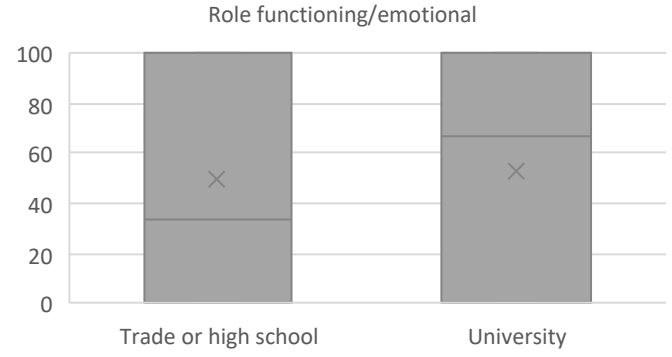


Figure 2.42: Boxplot of SF36 Role functioning/emotional by education

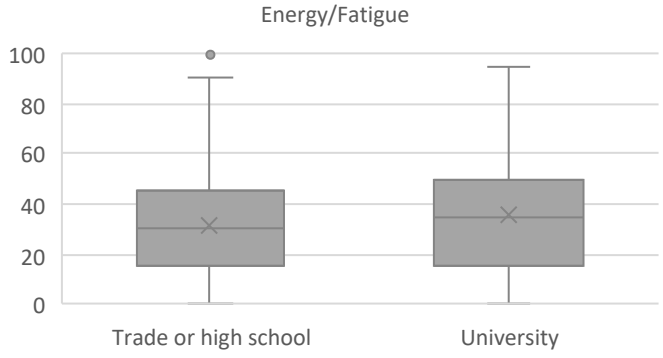


Figure 2.43: Boxplot of SF36 Energy/fatigue by education

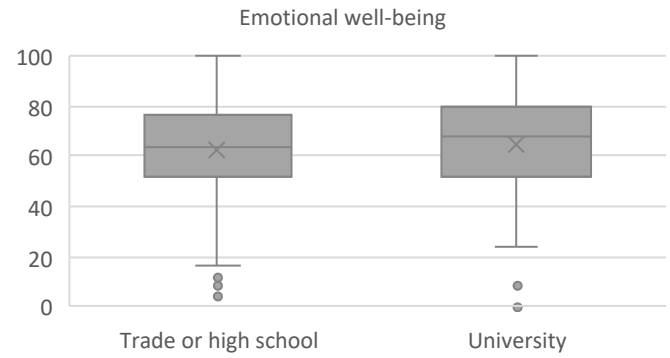


Figure 2.44: Boxplot of SF36 Emotional well-being by education

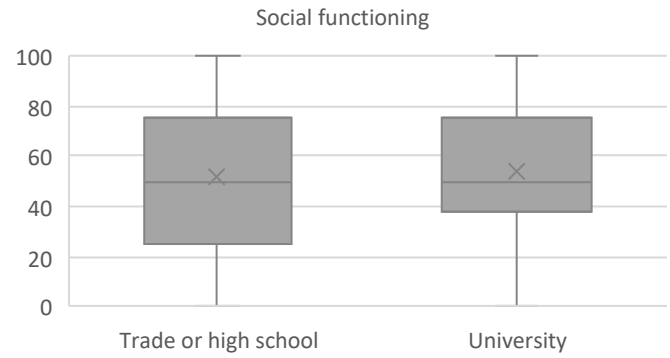


Figure 2.45: Boxplot of SF36 Social functioning by education

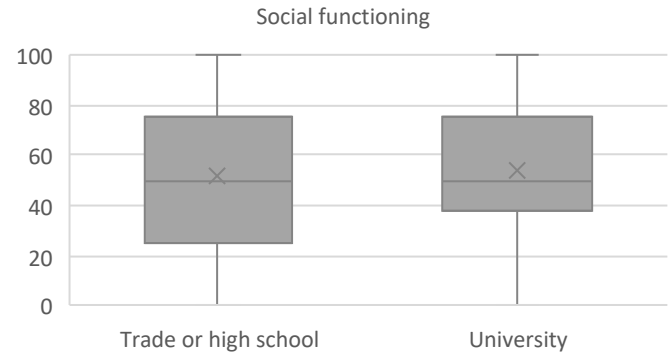


Figure 2.46: Boxplot of SF36 Pain by education

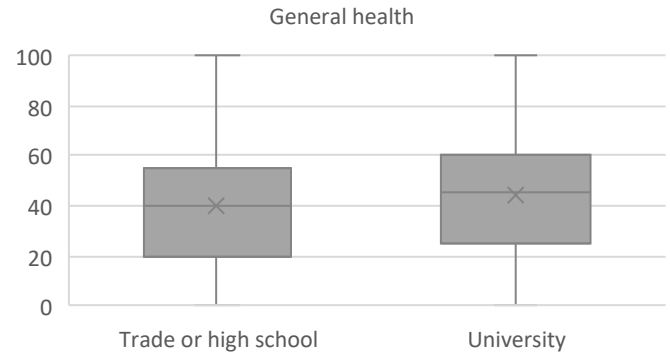


Figure 2.47: Boxplot of SF36 General health by education

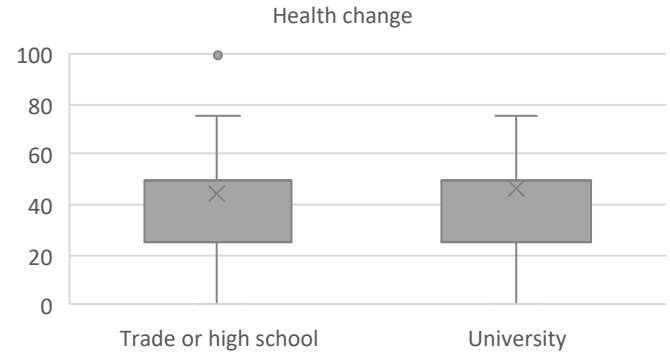


Figure 2.48: Boxplot of SF36 Health change by education

SF36 by location

The **location** of participants was evaluated by postcode using the Australian Statistical Geography Maps (ASGS) Remoteness areas accessed from the Australian Bureau of Statistics. Those living in regional or remote areas (n=107, 28.01%) were compared to those living in a metropolitan area (n=275, 71.99%).

Assumptions for normality and variance for a two-sample t-test were not met, a Wilcoxon rank sum test with continuity correction was used.

Wilcoxon rank sum tests with continuity correction indicated that the median score for the SF36 Pain scale [W = 12476.00, p = 0.020] was significantly lower for participants in the Regional or remote subgroup (Median = 55.00, IQR = 55.00) compared to participants in the Metropolitan subgroup (Median = 57.50, IQR = 56.25).

SF36 Pain scale measures how much pain, and how pain interferes with work and other activities. On average, participants in the Metropolitan subgroup had a higher score for Pain compared to Rural or remote , however, both groups had moderate pain.

Table 2.14: SF36 by location summary statistics and Wilcoxon test

SF36 scale	Group	Number (n=382)	Percent	Median	IQR	W	p-value
Physical functioning	Rural or remote	107	28.01	50.00	57.50	14163.00	0.5704
	Metropolitan	275	71.99	60.00	60.00		
Role functioning/physical	Rural or remote	107	28.01	0.00	75.00	13616.00	0.2229
	Metropolitan	275	71.99	25.00	100.00		
Role functioning/emotional	Rural or remote	107	28.01	33.33	100.00	14465.00	0.7869
	Metropolitan	275	71.99	66.67	100.00		
Energy/Fatigue	Rural or remote	107	28.01	35.00	32.50	14793.00	0.9340
	Metropolitan	275	71.99	30.00	35.00		
Emotional well-being	Rural or remote	107	28.01	68.00	24.00	14854.00	0.8841
	Metropolitan	275	71.99	68.00	28.00		
Social functioning	Rural or remote	107	28.01	50.00	50.00	14225.00	0.6125
	Metropolitan	275	71.99	50.00	37.50		
Pain	Rural or remote	107	28.01	55.00	55.00	12476.00	0.0205*
	Metropolitan	275	71.99	57.50	56.25		
General health	Rural or remote	107	28.01	35.00	35.00	13952.00	0.4320
	Metropolitan	275	71.99	40.00	35.00		
Health change	Rural or remote	107	28.01	50.00	25.00	13416.00	0.1563
	Metropolitan	275	71.99	50.00	25.00		

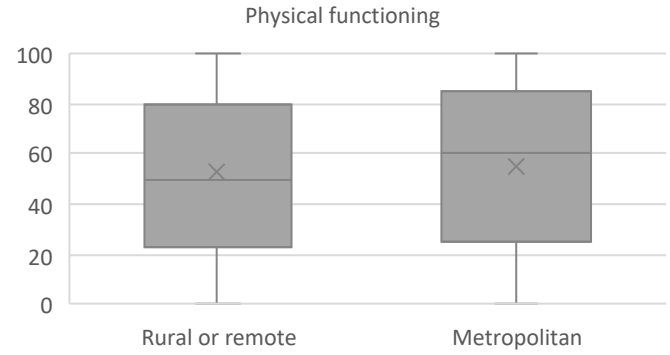


Figure 2.49: Boxplot of SF36 Physical functioning by location

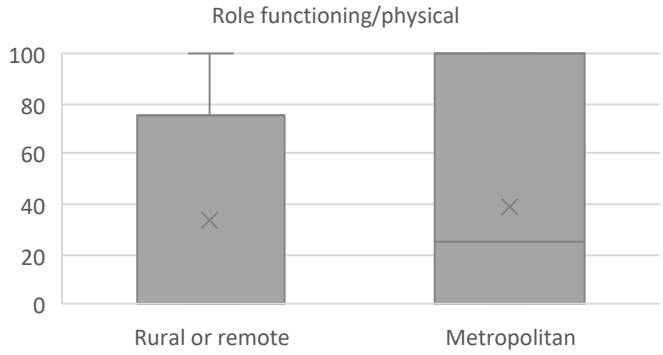


Figure 2.50: Boxplot of SF36 Role functioning/physical by location

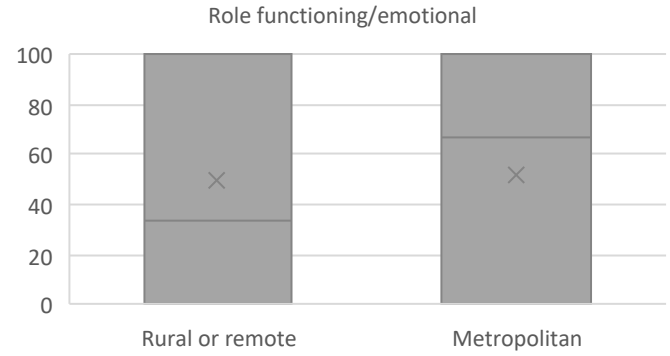


Figure 2.51: Boxplot of SF36 Role functioning/emotional by location

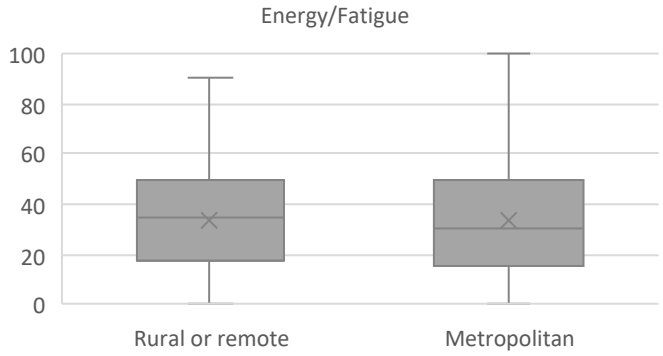


Figure 2.52: Boxplot of SF36 Energy/fatigue by location

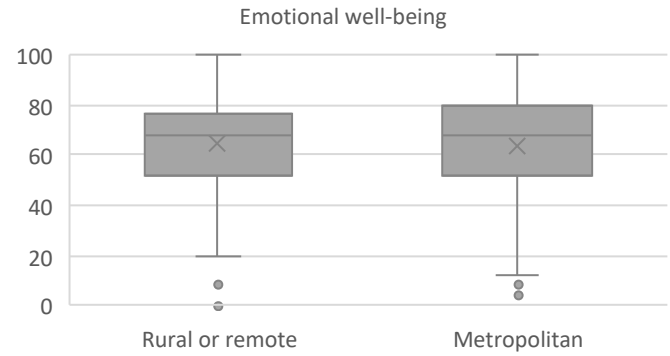


Figure 2.53: Boxplot of SF36 Emotional well-being by location

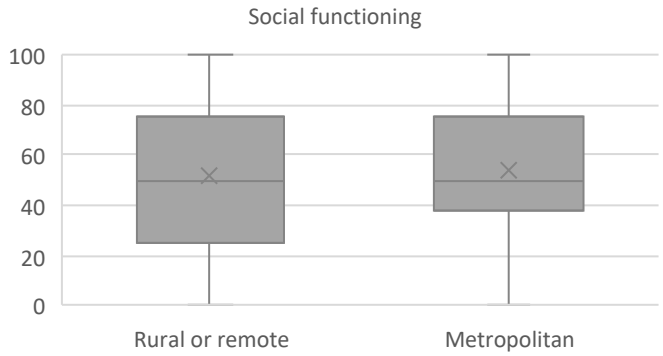


Figure 2.54: Boxplot of SF36 Social functioning by location

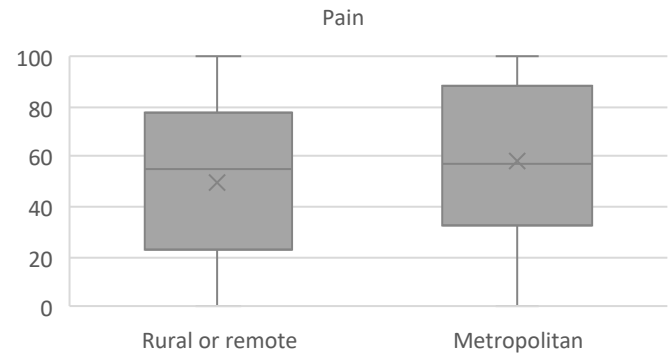


Figure 2.55: Boxplot of SF36 Pain by location

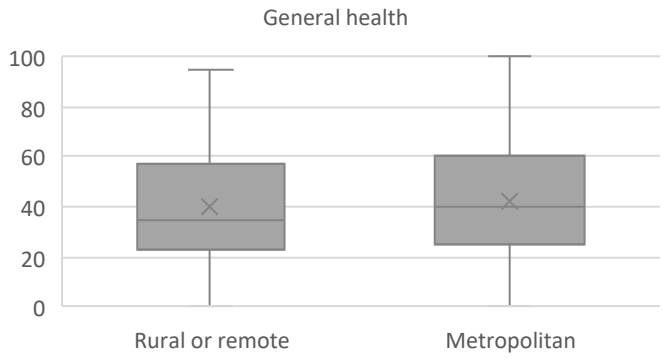


Figure 2.56: Boxplot of SF36 General health by location

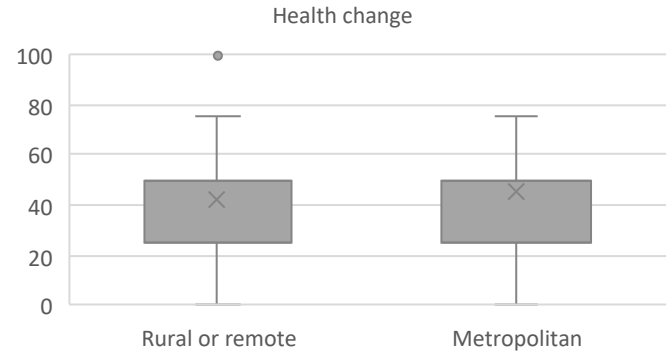


Figure 2.57: Boxplot of SF36 Health change by location

SF36 by socioeconomic status

Comparisons were made by **socioeconomic status**, using the Socio-economic Indexes for Areas (SEIFA) (www.abs.gov.au), SEIFA scores range from 1 to 10, a higher score denotes a higher level of advantage. Participants with a mid to low SEIFA score of 1-6 (n=191, 50.00%) compared to those with a higher SEIFA score of 7-10 (n=191, 50.00%).

Assumptions for normality and variance for a two-sample t-test were not met, a Wilcoxon rank sum test with continuity correction was used.

Wilcoxon rank sum tests with continuity correction indicated that the median score for the SF36 Physical functioning scale [W = 15642.00, p = 0.016] was significantly lower for participants in the Mid to low status subgroup (Median = 50.00, IQR = 57.50) compared to participants in the Higher status subgroup (Median = 65.00, IQR = 55.00).

Wilcoxon rank sum tests with continuity correction indicated that the median score for the SF36 Role functioning emotional scale [W = 16174.00, p = 0.042] was significantly lower for participants in the Mid to low status subgroup (Median = 33.33, IQR = 100.00) compared to participants in the Higher status subgroup (Median = 66.67, IQR = 100.00).

Wilcoxon rank sum tests with continuity correction indicated that the median score for the SF36 Emotional well-being scale [W = 15882.00, p = 0.029] was significantly lower for participants in the Mid to low status subgroup (Median = 64.00, IQR = 26.00) compared to participants in the Higher status subgroup (Median = 72.00, IQR = 24.00).

Wilcoxon rank sum tests with continuity correction indicated that the median score for the SF36 Social functioning scale [W = 15784.00, p = 0.022] was significantly lower for participants in the Mid to low status subgroup (Median = 50.00, IQR = 50.00) compared to participants in the Higher status subgroup (Median = 62.50, IQR = 37.50).

Wilcoxon rank sum tests with continuity correction indicated that the median score for the SF36 Pain scale [W = 15808.00, p = 0.024] was significantly lower for participants in the Mid to low status subgroup (Median = 45.00, IQR = 55.00) compared to participants in the Higher status subgroup (Median = 57.50, IQR = 55.00).

SF36 Physical functioning scale measures health limitations in physical activities such as walking, bending, climbing stairs, exercise, and housework. On average, participants in the Higher status subgroup scored higher than participants in the Mid to low status

subgroup. This indicates that physical activities were slightly limited for participants in the Higher status subgroup, and were moderately limited for participants in the Mid to low status subgroup.

SF36 Role functioning/emotional scale measures how emotional problems interfere with work or other activities. On average, participants in the Higher status subgroup scored higher than participants in the Mid to low status subgroup. This indicates that emotional health seldom interfered with work or other activities for participants in the Higher status subgroup, and often interfered for participants in the Mid to low status subgroup.

SF36 Emotional well-being scale measures how a person feels, for example happy, calm, depressed or anxious. On average, participants in the Higher status subgroup had a higher score for emotional well-being compared to Mid to low status, however, both groups had good emotional well-being.

SF36 Social functioning scale measures limitations on social activities due to physical or emotional problems. On average, participants in the Higher status subgroup scored higher than participants in the Mid to low status subgroup. This indicates that social activities were slightly limited for participants in the Higher status subgroup, and moderately limited for participants in the Mid to low status subgroup.

SF36 Pain scale measures how much pain, and how pain interferes with work and other activities. On average, participants in the Higher status subgroup had a higher score for Pain compared to Mid to low status, however, both groups had moderate pain.

SF36 General health scale measures perception of health. On average, participants in the Higher status subgroup had a higher score for general health compared to Mid to low status, however, both groups had poor health.

Table 2.15: SF36 by socioeconomic status summary statistics and Wilcoxon test

SF36 scale	Group	Number (n=382)	Percent	Median	IQR	W	p-value
Physical functioning	Mid to low status	191	50.00	50.00	57.50	15642.00	0.0159*
	Higher status	191	50.00	65.00	55.00		
Role functioning/physical	Mid to low status	191	50.00	0.00	75.00	16988.00	0.2111
	Higher status	191	50.00	25.00	100.00		
Role functioning/emotional	Mid to low status	191	50.00	33.33	100.00	16174.00	0.0423*
	Higher status	191	50.00	66.67	100.00		
Energy/Fatigue	Mid to low status	191	50.00	30.00	30.00	16513.00	0.1086
	Higher status	191	50.00	35.00	30.00		
Emotional well-being	Mid to low status	191	50.00	64.00	26.00	15882.00	0.0285*
	Higher status	191	50.00	72.00	24.00		
Social functioning	Mid to low status	191	50.00	50.00	50.00	15784.00	0.0218*
	Higher status	191	50.00	62.50	37.50		
Pain	Mid to low status	191	50.00	45.00	55.00	15808.00	0.0235*
	Higher status	191	50.00	57.50	55.00		
General health	Mid to low status	191	50.00	35.00	35.00	15757.00	0.0211*
	Higher status	191	50.00	40.00	35.00		
Health change	Mid to low status	191	50.00	50.00	25.00	17602.00	0.5305
	Higher status	191	50.00	50.00	25.00		

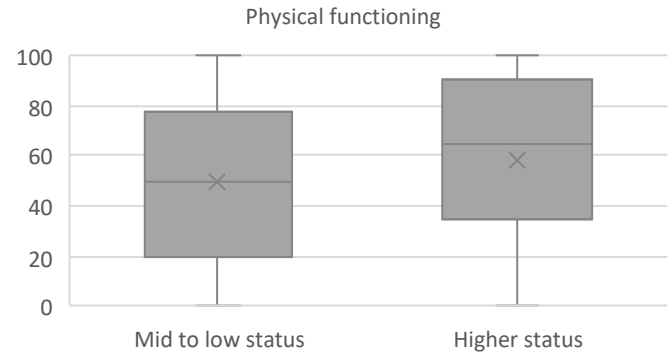


Figure 2.58: Boxplot of SF36 Physical functioning by socioeconomic status

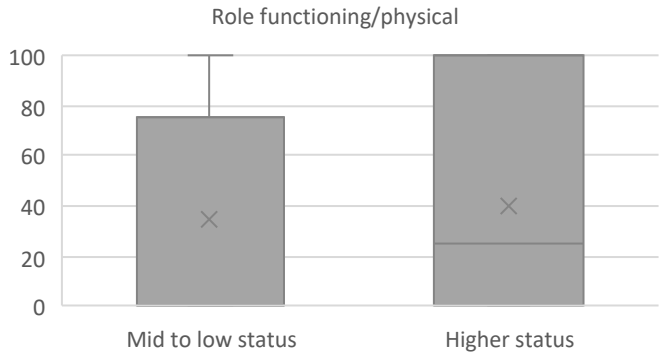


Figure 2.59: Boxplot of SF36 Role functioning/physical by socioeconomic status

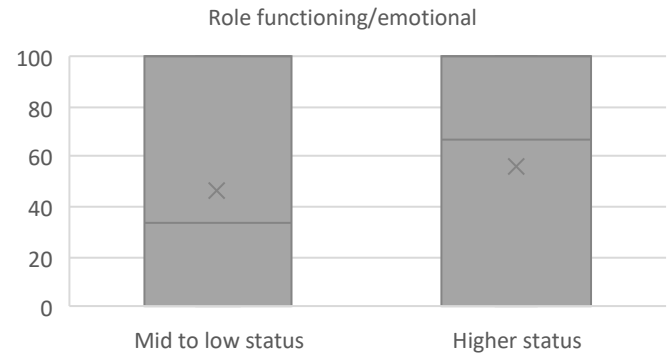


Figure 2.60: Boxplot of SF36 Role functioning/emotional by socioeconomic status

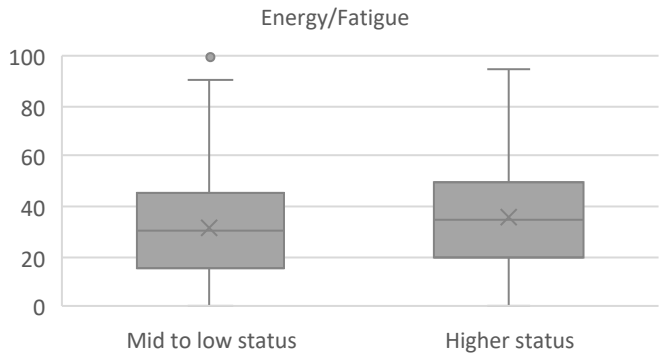


Figure 2.61: Boxplot of SF36 Energy/fatigue by socioeconomic status

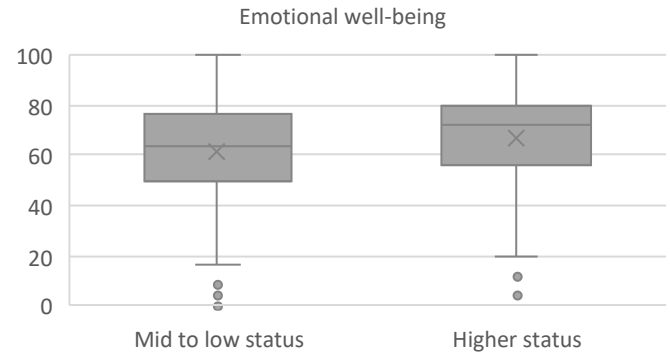


Figure 2.62: Boxplot of SF36 Emotional well-being by socioeconomic status

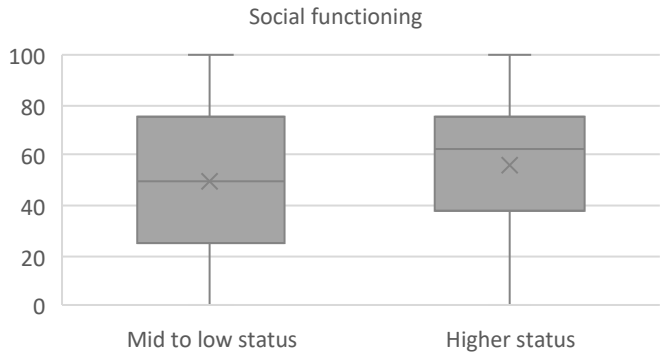


Figure 2.63: Boxplot of SF36 Social functioning by socioeconomic status

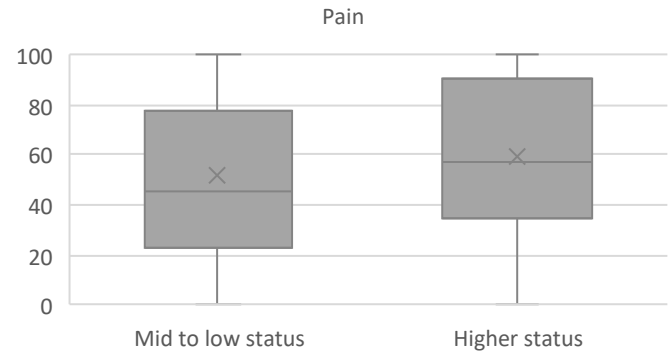


Figure 2.64: Boxplot of SF36 Pain by socioeconomic status

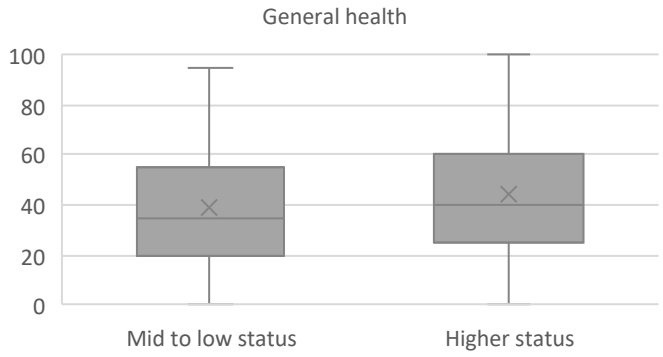


Figure 2.65: Boxplot of SF36 General health by socioeconomic status

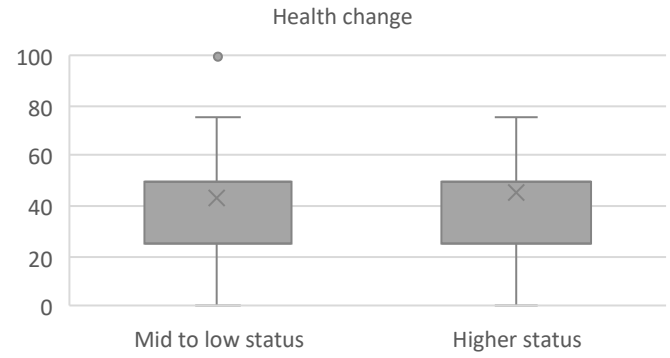


Figure 2.66: Boxplot of SF36 Health change by socioeconomic status

Section 3

Symptoms and diagnosis

Section 3: Symptoms and diagnosis

Symptoms leading to diagnosis

In the structured interview, participants were asked to describe the symptoms that actually *led* to their diagnosis. Most commonly participants strongly recalled their symptoms or how they came to be diagnosed (84.58%). Others had an unclear recollection of their symptoms or how they came to be diagnosed (7.46%), or had no symptoms that they felt specifically led to diagnosis (3.23%).

Symptoms leading to diagnosis: Seeking medical attention

Participants described when they sought medical attention after noticing symptoms. The most common responses were having symptoms and seeking medical attention relatively soon (59.95%), and having symptoms and not seeking medical attention initially (17.66%). Other themes included having no symptoms or not noticing any symptoms before diagnosis (3.23%).

Symptoms leading to diagnosis: Description of diagnostic pathway

In the structured interview, participants described their diagnostic pathway in the healthcare system. The most common descriptions were a complex diagnosis, needing to see multiple specialists before diagnosis (46.52%), and a linear diagnosis after being referred to a specialist from their general practitioner (28.36%). Other themes included being diagnosed in an emergency department/urgent care (13.68%), being diagnosed by their general practitioner during a routine check-up that was not related to symptoms (5.97%).

Diagnosis provider and location

Participants were asked in the online questionnaire, which healthcare professional gave them their diagnosis, and where they were given the diagnosis. Participants were most commonly given their diagnosis in the specialist clinic (n=154, 43.14%), this was followed by the hospital (n=151, 42.30%), and the general practice (GP) (n=40, 11.20%).

Understanding of disease at diagnosis

Participants were asked in the structured interview how much they knew about their condition at diagnosis. The most common response was knowing nothing or very little about the condition at diagnosis (61.44%) Others described knowing a good amount about the condition at diagnosis, for example they knew about the condition by learning about it before or during the diagnostic process (7.71%), and knowing about the condition due to professional background (3.23%).

Emotional support at diagnosis

Participants were asked in the online questionnaire how much emotional support they or their family received between diagnostic testing and diagnosis. There were 79 participants (21.07%) who had enough support, 96 participants (25.60%) that had some support but it wasn't enough, and 200 participants (53.33%) had no support.

Costs at diagnosis

Participants noted in the online questionnaire the amount of out-of-pocket expenses they had at diagnosis, for example doctors' fees, and diagnostic tests. There were 146 participants (53.09%) who had no out of pocket expenses, and 51 participants (18.55%) who did not know or could not recall. There were 34 participants (12.36%) that spent Less than $500, 13 participants (4.73%) that spent between $500 to $1000, and 31 participants (11.27%) that spent More than $1000.

Burden of diagnostic costs

In the follow-up question about the burden of costs at diagnosis, for 30 participants who had out of pocket expenses. For 65 participants (33.85%) the cost was slightly or not at all significant. For 40 participants (20.83%) the out-of-pocket expenses were somewhat significant, and for 87 participants (45.31%), the burden of out-of-pocket expenses were moderately or extremely significant.

Genetic tests and biomarkers

Participants answered questions in the online questionnaire about if they had any discussions with their doctor about biomarkers, genomic and gene testing that might be relevant to treatment. If they did have a discussion, they were asked if they brought up the topic or if their doctor did.

Most commonly, participants had never had a conversation about biomarkers, genomic, or gene testing that might be relevant to treatment, (n=211, 66.56%). There were 28 participants (8.83%) who brought up the topic with their doctor, and 78 participants (24.61%) whose doctor brought up the topic with them.

Participants were then asked if they had had any biomarker, genomic or gene testing. If they had testing, they were asked if they had it as part of a clinical trial, paid for it themselves or if they did not have to pay for it. Those that did not have the test were asked if they were interested in this type of test. A little over half of participants indicated that they did not have any genetic or biomarker tests but would like to (n=193, 60.88%.

Understanding of prognosis

Participants were asked in the structured interview to describe what their current understanding of their prognosis was. The most common responses were that there was uncertainty around prognosis (26.37%), in terms of symptoms and function/changes in symptoms and function (17.66%), and that they had specific medical interventions they need to manage their condition (15.92%). Other themes included that they were monitoring their condition until there is an exacerbation or progression (15.67%), and had poor outcomes, or a terminal condition (11.94%).

Symptoms leading to diagnosis

In the structured interview, participants were asked to describe the symptoms that actually *led* to their diagnosis. Most commonly participants strongly recalled their symptoms or how they came to be diagnosed (84.58%). Others had an unclear recollection of their symptoms or how they came to be diagnosed (7.46%), or had no symptoms that they felt specifically led to diagnosis (3.23%).

The most common symptoms leading to diagnosis were having developmental delays (15.67%), eye and visions problems (10.20%), and fatigue (8.46 %). Other themes included gastrointestinal distress (8.46%). Failure to thrive, 'floppy, relaxed, lazy or weak', heart problems and joint aches/pains were all noted at 6.47% respectively.

Participant describes having developmental delays which led to their diagnosis

And when PARTICIPANT was, as he was growing up, he wasn't reaching milestones as a typical child should. By the time he was almost two weeks. Oh no, it was just over 12 months. It would have been closer to 16 months. My husband's PROFESSION. We were posted to a different location, and we asked if we could move earlier because PARTICIPANT needed to see a paediatrician and it was quite urgent, so they moved us. Earlier we saw a paediatrician. PARTICIPANT was not walking, not talking, not babbling. He couldn't crawl. He was still eating mushy foods at about 16 months old. So we weren't aware that this was an issue as such because he was our first child. But the paediatrician said he's a boy and a baby and he'll develop in his own time. And I kind of said to my husband, I think there might be something more to it than that. So we got a second opinion and the results came back with the 22 Q deletion and from then we've been able to understand why he took so long to develop in certain areas and how we are able to help him develop
Participant 036_2023AUDPA

So PATIENT, he's 5 now, and he basically wasn't meeting any milestones like developmental milestones. So he could meet, he met a few, like holding his neck and yeah, it's pretty, pretty much…and he could like it had like suck and slow reflex, so he could see it and so on and forth. But he got to about just before six months and he couldn't sit. And of course he can't, like a lot of babies still can't sit at six months. But he was making no attempt. He couldn't really roll, he wouldn't like grab out the toys. So kind of like the whole general gross motor development. And at that point it wasn't…we didn't really notice any cognitive differences. And then pretty much just went from there and he wasn't meeting any milestones at any of the ages that he should have been. He couldn't sit until he was 2 1/2. So developmental milestones is definitely probably major indicator.
Participant 081_2023AUDIS

Participant describes having fatigue which led to their diagnosis

Well, the very first common symptoms I experienced was mainly fatigue and, you know, dark urine and also my stool was affected. And also had some other symptoms like a lot of appetite and abdominal pains. So the symptoms kept on coming and I was kind of not really knowing what was happening to me. So it just started little by little to it. It got severe.
Participant 006_2023AUORC

Oh, certainly fatigue. Yes. Fatigue was my biggest issue really. Still is, I think joint pain, my fingers mainly. I think that's probably all at the time. Yes.
Participant 013_2023AUDIS

Participant describes having eye and vision problems which led to their diagnosis

Well, he was diagnosed at one or two months old and it was pretty obvious that something was going on because before I gave birth, we found out through the ultrasounds that he had congenital heart defects and after he was born the the doctors kept picking up on more things that could be wrong so that that he couldn't, he couldn't hear and that he's he had issues with vision so. We didn't know that he had CHARGE syndrome before he was born, but we knew something was wrong. But it was pretty obvious the first month or two, so I think they were searching for what it was almost instantly.
Participant 089_2023AUENM

You said yeah. I think the first time I remember anything was in 2011. I went to my GP with eye pain and blurred vision. They sent me to an optometrist who looked at my eyes and was like, no, you're fine, all good. And then 2015 it happened again. So I went to my GP and was sent to HOSPITAL for admission and then they were like, Yep, it's optic neuritis, 3 days of steroids and a few MRI's.
Participant 096_2023AUDNS

I woke up and had lost my eyesight and before that I didn't even have a headache or anything, so even the night before, I didn't have…I was working, it was over Easter. I didn't have any symptoms at all and woke up and lost half my vision, the upper field of my at that time it was my right eye.
075_2023AUDNS

Participant describes having failure to thrive/feeding problems as infant which led to their diagnosis

So she started having infantile spasms and so and not quite hitting her developmental milestones. So we took her to emergency room in LOCATION Children's and she had massive feeding issues. She was underweight, had these infantile spasms and while we were in hospital they did their investigations on why she had the infantile spasms and they did some form of gene genetic test and discovered that she had a the mutation.
Participant 090_2023AUENM

Participant describes having heart problems which led to their diagnosis

Yeah, so yeah, NAME was, had a VSD when we left hospital we discovered that and then she had failure to thrive, obviously meaning that she wasn't getting enough food and things and then obviously over a course of time we went through the process of eliminating what was going on and then our GP. I said, look, I just want to do, I thought she had a submucus cleft and then the pediatrician was just like I just want to do this test to eliminate the chances of this condition called 22 Q. So she did that and then obviously the results came back as as positive.
Participant 034_2023AUDPA

The that cardiologist was thinking that PATIENT had DiGeorge syndrome so they did the open heart surgery six weeks later cause they needed him a little bit bigger. And when they went in they found that he had a smaller than normal thymus and that he had that band that went around the esophagus and the trachea which they ligated. So that was giving him a diagnosis of DiGeorge.
Participant 040_2023AUDPA

Participant describes having lumps, boils, and cysts which led to their diagnosis

It started when I was about 7 years old. I don't remember a lot from back then, but I had it started with like a boil on my I think it was my butt and my mum took me to the doctors and they basically they didn't do any testing. They just said it's staph and we'll treat her for staph but the treatment obviously didn't work and after maybe like six months or so, they kind of said it's not working and gave up and then that must be it…Yep. Sorry, I wasn't diagnosed until I was 21. So four years ago, yeah. So I went to my doctor for something completely different and she saw all my scarring and the current flares that I had.
Participant 014_2023AUDSK

Yeah, I probably had symptoms. I would say roughly I would say 14 years old. I used to have like I have really thick black hair and curly hair and I was under the impression that because I shaved my armpits that that was the reason I was getting HS under my armpits. I just thought, you know, I have curly hair, it's obviously curling inside and that's what's infecting. So that was probably my earliest recollection, and that was something that I just dismissed. I didn't think it was that big of a deal. I thought everyone got them and I kind of went on with life thinking that was really normal for at least 10 years. And I remember I was probably around 23 or 24 and I was in Europe with a friend and I was telling her about how I got a infected in my groin area, which was making it quite difficult for me to walk around and explore our trip.
Participant 026_2023AUDSK

Participant describes having seizures/Spasms which led to their diagnosis

I had a tremor in my head as in it shook from side to side. My neck was spasming constantly and twisting to one side. Most of the time, when I was at work, I had to write reports standing up and hold my head still. It was shaking that badly. Is that the information what we're after, that sort of thing?..That shaking and tremoring progressively, and the pain progressively got worse over probably about six months about the June, July month I had sought initially treatment from a massage treatment. We went to the doctor and they suggested we try some acupuncture. My doctor did acupuncture, so we tried some of that as well. Acupuncture actually relieved the symptoms for about 10 minutes. It did give me a bit of relief, and then it never…We just carried on. I continued to ignore the symptoms basically because at that point in time actually thought I had Parkinson's. That's how I sort of go with things. I didn't want to know about it, so I just carried on. I hadn't been sleeping very well. It was only…my husband and I worked for the same company and I was in a meeting and my husband after meeting just grabbed me and said, "No, enough is enough, and we are going down to the doctor's now to find out what's going on."
Participant 006_2023AUDNS

Participant describes their child being floppy', 'relaxed' and/or 'lazy' and/or 'weak' which led to their diagnosis

Yeah, he was a floppy baby. It was quite like his limbs, I noticed, especially his arms were quiet, hyper mobile. I guess you would. Say, yeah, yeah. And didn't have a lot of strength like and then obviously then was delayed in you know rolling and sitting. And crawling all these milestones. So that's what kind of made me or prompted me to get him checked out.
Participant 014_2023AUDPA

Well, around six to eight months I noticed when I'd pop NAME on his bottom he would just flop and then about 10 months I though maybe I'll introduce those little walker things. He wouldn't do anything. He would just pop his head forward. In bed too,
like when I would put NAME down for a nap ... He wouldn't move, he wouldn't get up on his cart. I thought, "No, he's just probably a really relaxed baby. 010_2023AUDNS

I had noticed that she had declined even more at that stage, so we put her in a seat that would some- what support her back, and she kind of just flopped to one side. Not completely fall down, but she was almost slouchy.
041_2023AUDNS

Participant describes having shortness of breath which led to their diagnosis

Yeah. So that was all happening in the Children's Hospital because we were there while she was recovering from the heart surgery and then she got and ended up back in the NICU. Yeah. So in hindsight. I think, you know, like she was having trouble breathing. And so in hindsight now knowing what I know about CHARGE and like she was probably having issues with her swallowing mechanism and she was asphyxiating. But that was never kind of discussed or picked up or and we didn't have an overseeing pediatrician like we were just admitted on the cardiac.
Participant 087_2023AUENM

Participant describes having weight loss which led to their diagnosis

During my diagnosis but I still wasn't suspecting anything like that. I'd never Googled or I just thought I was getting older and all of a sudden my hip started to go and I had a lot of hip pain and back pain. More in my hip sort of radiated to my back rather than I didn't realise it was my back causing it. I thought it was my my hip to be honest, and they said I had bone spurs and things like that and then that was about…My GP. Yeah, she wasn't suspecting Scleroderma. She just ordered an…like panel because I had shooting pain in my arms. My hands had swollen to the point I couldn't use them. I was in agony and I was beside myself. And that was just after a bacterial lung infection that had knocked me off my feet for weeks and I never really got over it. And then all these other things started going wrong. I was getting headaches, vertigo. My hands and legs were giving me huge trouble. I couldn't walk properly. Yeah, there was just and my digestive system started to shut down. So I was really struggling to swallow food, digest food. I'd lost a heap of weight, but I'd had trouble with that before, but they told me I just had a sensitive tummy. February my arm started to swell, my hands and fingers were swollen, I couldn't even hold the steering wheel. Driving a car was really impossible.
Participant 016_2023AUDIS

Table 3.1: Symptom recall

Symptom recall	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Symptom recall strong	340	84.58	53	79.10	75	92.59	90	94.74	25	78.13	80	84.21	17	53.13	227	84.70	113	84.33	260	88.44	78	73.58
Symptom recall unclear	30	7.46	9	13.43	2	2.47	4	4.21	2	6.25	6	6.32	7	21.88	16	5.97	14	10.45	14	4.76	16	15.09
No symptoms	13	3.23	2	2.99	0	0.00	0	0.00	0	0.00	4	4.21	7	21.88	11	4.10	2	1.49	6	2.04	7	6.60

Symptom recall	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Symptom recall strong	340	84.58	81	83.51	107	81.68	101	88.60	51	85.00	175	88.38	160	81.63	95	85.59	245	84.19	175	87.50	165	81.68
Symptom recall unclear	30	7.46	12	12.37	12	9.16	3	2.63	3	5.00	11	5.56	19	9.69	5	4.50	25	8.59	11	5.50	19	9.41
No symptoms	13	3.23	2	2.06	5	3.82	5	4.39	1	1.67	3	1.52	10	5.10	4	3.60	9	3.09	4	2.00	9	4.46

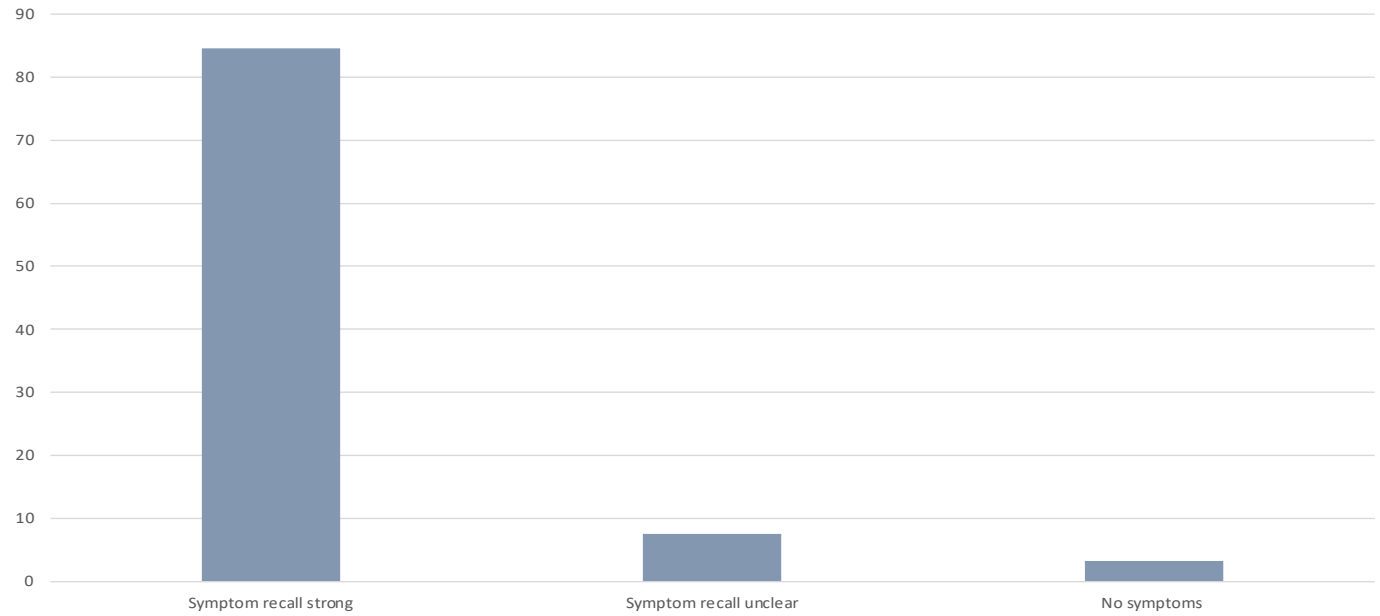


Figure 3.1: Symptom recall

Table 3.2: Symptom recall – sub group variations

Symptom recall	Reported less frequently	Reported more frequently
Symptom recall strong	Other rare condition Male	Diseases of the nervous system
Symptom recall unclear		Other rare condition
No Symptoms		Other rare condition

Table 3.3: Symptoms leading to diagnosis

Symptoms leading to diagnosis	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Developmental delays	63	15.67	21	31.34	1	1.23	36	37.89	0	0.00	2	2.11	3	9.38	22	8.21	41	30.60	45	15.31	17	16.04
Eye and vision problems	41	10.20	3	4.48	0	0.00	23	24.21	0	0.00	13	13.68	2	6.25	30	11.19	11	8.21	31	10.54	10	9.43
Fatigue	34	8.46	0	0.00	6	7.41	6	6.32	0	0.00	19	20.00	3	9.38	26	9.70	8	5.97	28	9.52	6	5.66
Gastrointestinal distress	34	8.46	8	11.94	8	9.88	0	0.00	0	0.00	14	14.74	4	12.50	22	8.21	12	8.96	26	8.84	7	6.60
Failure to thrive/feeding problems as infant	26	6.47	10	14.93	0	0.00	4	4.21	0	0.00	12	12.63	0	0.00	7	2.61	19	14.18	20	6.80	6	5.66
Floppy', 'relaxed' and/or 'lazy' and/or 'weak'	26	6.47	0	0.00	0	0.00	19	20.00	0	0.00	7	7.37	0	0.00	10	3.73	16	11.94	22	7.48	4	3.77
Heart problems	26	6.47	18	26.87	0	0.00	0	0.00	0	0.00	7	7.37	1	3.13	6	2.24	20	14.93	15	5.10	10	9.43
Joint aches and pain	26	6.47	1	1.49	17	20.99	0	0.00	0	0.00	7	7.37	1	3.13	25	9.33	1	0.75	25	8.50	1	0.94
Poor head strength/not being able to pull self up or sit up/not able to do tummy time or roll over	23	5.72	0	0.00	0	0.00	23	24.21	0	0.00	0	0.00	0	0.00	7	2.61	16	11.94	22	7.48	1	0.94
Seizures/Spasms	22	5.47	15	22.39	0	0.00	4	4.21	0	0.00	3	3.16	0	0.00	6	2.24	16	11.94	13	4.42	9	8.49

Symptoms leading to diagnosis	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Developmental delays	63	15.67	30	30.93	25	19.08	6	5.26	2	3.33	29	14.65	34	17.35	17	15.32	46	15.81	28	14.00	35	17.33
Eye and vision problems	41	10.20	7	7.22	21	16.03	13	11.40	0	0.00	17	8.59	24	12.24	9	8.11	32	11.00	19	9.50	22	10.89
Fatigue	34	8.46	1	1.03	12	9.16	12	10.53	9	15.00	18	9.09	14	7.14	8	7.21	26	8.93	16	8.00	18	8.91
Gastrointestinal distress	34	8.46	7	7.22	10	7.63	10	8.77	7	11.67	11	5.56	22	11.22	12	10.81	22	7.56	13	6.50	21	10.40
Failure to thrive/feeding problems as infant	26	6.47	14	14.43	8	6.11	2	1.75	2	3.33	17	8.59	9	4.59	7	6.31	19	6.53	14	7.00	12	5.94
Floppy', 'relaxed' and/or 'lazy' and/or 'weak'	26	6.47	15	15.46	6	4.58	4	3.51	1	1.67	13	6.57	13	6.63	8	7.21	18	6.19	10	5.00	16	7.92
Heart problems	26	6.47	15	15.46	6	4.58	2	1.75	3	5.00	13	6.57	13	6.63	3	2.70	23	7.90	9	4.50	17	8.42
Joint aches and pain	26	6.47	0	0.00	10	7.63	11	9.65	5	8.33	18	9.09	8	4.08	8	7.21	18	6.19	12	6.00	14	6.93
Poor head strength/not being able to pull self up or sit up/not able to do tummy time or roll over	23	5.72	15	15.46	8	6.11	0	0.00	0	0.00	13	6.57	10	5.10	5	4.50	18	6.19	13	6.50	10	4.95
Seizures/Spasms	22	5.47	13	13.40	5	3.82	2	1.75	2	3.33	11	5.56	11	5.61	9	8.11	13	4.47	11	5.50	11	5.45

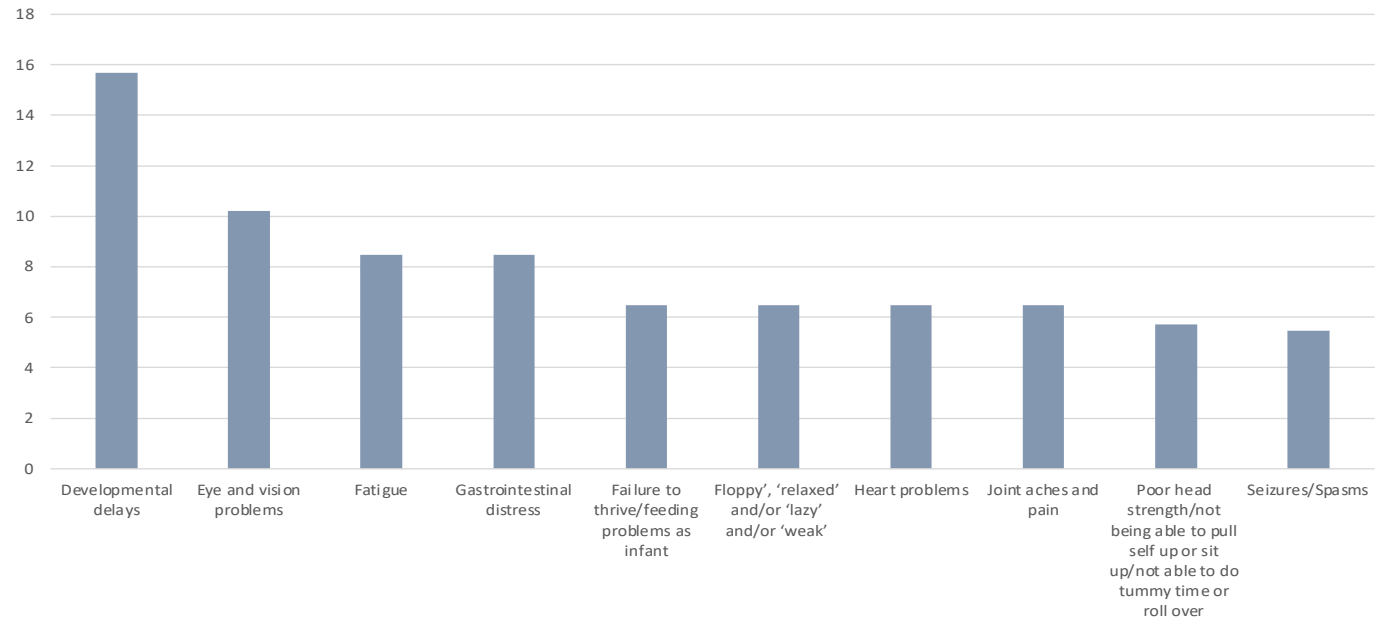


Figure 3.2: Symptoms leading to diagnosis

Table 3.4: Symptoms leading to diagnosis – subgroup variations

Symptoms leading to diagnosis	Reported less frequently	Reported more frequently
Developmental delays	Diseases of the immune system Diseases of the skin Endocrine, nutritional or metabolic diseases Aged 45 to 64 Aged 65 plus	Developmental anomalies Diseases of the nervous system Family or carer Aged under 18
Eye and vision problems	Diseases of the immune system Diseases of the skin Aged 65 plus	Diseases of the nervous system
Fatigue		Endocrine, nutritional or metabolic diseases
Gastrointestinal distress		
Failure to thrive/feeding problems as infant		
Floppy', 'relaxed' and/or 'lazy' and/or 'weak'		Diseases of the nervous system
Heart problems		Developmental anomalies
Joint aches and pain		Diseases of the immune system
Poor head strength/not being able to pull self up or sit up/not able to do tummy time or roll over		Diseases of the nervous system
Seizures/Spasms		Developmental anomalies

Symptoms leading to diagnosis: Seeking medical attention

Participants described when they sought medical attention after noticing symptoms. The most common responses were having symptoms and seeking medical attention relatively soon (59.95%) and having symptoms and not seeking medical attention initially (17.66%). Other themes included having no symptoms or not noticing any symptoms before diagnosis (3.23%).

Participant describes having symptoms and seeking medical attention relatively soon

Yes, when I was eighteen I was doing a high school certificate in NSW, which is, you know, the leaving high school, and I started getting the headaches, like migraines. I went to the doctor and the doctor said they see this neurologist. I went to the neurologist. The neurologist had absolutely no interest in the migraines, but he was interested in merely my feet and hands in particular. He insisted on doing a test which was a nerve conduction test and was extremely painful. He held me down because I was resistant to having this extremely painful test. I didn't know it was painful until he did it, and he held me down to do that and at that point, we said it was clear that my nerve conduction was slow. So I was 18. Yep.
Participant 017_2023AUORC

Yes, I had probably what you would call flare ups for many years which I didn't understand. I would go to the doctor and I would say things like all you've got an infection or would put you on a low dose antibiotic. Or periods of fatigue. I'm getting sick for long periods. Like I had respiratory infections for a long time and then finally when I turned 21, I was then able to sign myself into a hospital.
Participant 003_2023AUDIS

When I was around 10, I started having just some really bizarre issues. I would pass out for no reason, and then I would lose the use of my legs. I couldn't walk forward, but I could walk backwards. I could move my legs so it wasn't like I was paralyzed or anything. Obviously went and saw multiple doctors, multiple hospitals, numerous psychologists. I'm quite tall. I'm 6"2 now, and I was quite tall growing up, and they just put it down to that because they couldn't find anything to put it down to. That happened maybe once or twice, sometimes more every month, and up until I was 15.
Participant 004_2023AUDNS

Participant describes having symptoms and not seeking medical attention initially

Well, the very first common symptoms I experienced was mainly fatigue and, you know, dark urine and also my my stool was affected. And also had some other symptoms like a lot of appetite and abdominal pains. So the symptoms kept on coming and I was kind of not really knowing what was happening to me. So it just started little by little to it It got severe
Participant 006_2023AUORC

So for me this is really a bit confronting because I couldn't make sense of it as like I'm 51 and how, why would my needs now not be working because there were other people, you know, around my age…So that was a bit complex thing and I couldn't put the pieces together. I couldn't understand why this was happening and I was quite embarrassed and I felt quite humiliated that my body was failing me and no one could make sense of it because I was, you know, pretty useful in the sense of strong, healthy, flexible body. But no one said maybe you should get it checked out. I didn't even think about getting it checked out.
Participant 010_2023AUDIS

Participant describes having no symptoms or not noticing any symptoms before diagnosis

PARTICIPANT: No, I never had any symptom up to now.
INTERVIEWER: OK.
PARTICIPANT: Yeah, no symptom at all.
Participant 01_2023AUORC

Participant describes being diagnosed as a child

I don't really know when I noticed symptoms, I just always knew that I had it because I was diagnosed at birth. I think I probably noticed it most in like late high school or mid high school, just coughing and then being skinnier than everybody else, not being able to put on weight and then going to hospital when I got sick. I think that's it.
Participant 013_2023AUORC

Yeah, there isn't a lot.There because I I was diagnosed when I was 14 and it was so it was it was caught in a relatively relatively early but yeah I so I don't I don't recall having like a lot of a lot of you know kind of symptoms that that that couldn't be that couldn't be nailed down.
Participant 011_2023AUORC

Participant describes being diagnosed through surveillance

Yeah, in her circumstances, we actually found out six weeks prior to her being born that she was missing part of the brain. So we didn't know about the gene disorder, but we knew about the part of the brain, so we already were watching. I guess the symptoms to arise because there was a pre warning which had brain abnormalities.
Participant 016_2023AUORC

So we didn't really I guess because he was only three weeks old, a diagnosis. So since it was from the Heel Prick test, so we didn't really have a much of an opportunity to I guess see any symptoms at that point. He did end up afterwards after the diagnosis and dealt with a being diagnosed with things with the failure to thrive, but we didn't actually notice anything in that three weeks before we actually received the diagnosis.
Participant 020_2023AUORC

So my first child was actually diagnosed through all screens through newborn screening. So yeah, we received the results of the newborn screening and then the hospital contacted us regarding further follow up. But just prior to his newborn screening result, he was actually very unwell. So when he was a baby, when he was less than 24 hours old, he became critically unwell and they thought that he had sepsis, but he responded to the treatment that they provided. And then after I think about 8 days in care when we were discharged home, we got the phone call about newborn screening and then went back into the hospital to do follow up pathology. And then the kids hospital rang us and said that he been, you know, he. Had the condition. So we went in for further I guess genetic screening just to confirm that he had the condition alright.
Participant 021_2023AUORC

My daughter was diagnosed pre-birth, so about 12 week ultrasound the sonographer, person doing the ultrasound, is that what they're called, identified cardiac anomaly, suspected right aortic arch. The head sonographer person at the service did mention at the time, I think she'd just done a PD session or something on 22Q, that that anomaly was associated, she said, with DiGeorge syndrome. I said, of course, it's very unlikely to be, but we'll just bring me back for an early scan at 18 weeks just to be able to get a better visualization on the heart. Then, so we had an early, but within an 18-week scan where they confirmed the right aortic arch, we decided to do an amniote, and that confirmed that. Probably that 19 weeks by the time that happened.
Participant 067_2023AUDPA

Table 3.5: Seeking medical attention

Seeking medical attention	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Seeking medical attention relatively soon	241	59.95	36	53.73	58	71.60	72	75.79	11	34.38	57	60.00	7	21.88	158	58.96	83	61.94	184	62.59	56	52.83
Not seeking medical attention initially	71	17.66	7	10.45	17	20.99	10	10.53	16	50.00	13	13.68	8	25.00	63	23.51	8	5.97	55	18.71	15	14.15
No symptoms	13	3.23	0	0.00	0	0.00	2	2.11	0	0.00	5	5.26	6	18.75	10	3.73	3	2.24	7	2.38	6	5.66

Seeking medical attention	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Seeking medical attention relatively soon	241	59.95	60	61.86	72	54.96	76	66.67	33	55.00	125	63.13	114	58.16	65	58.56	176	60.48	120	60.00	121	59.90
Not seeking medical attention initially	71	17.66	3	3.09	29	22.14	20	17.54	19	31.67	39	19.70	30	15.31	21	18.92	50	17.18	37	18.50	34	16.83
No symptoms	13	3.23	2	2.06	5	3.82	5	4.39	1	1.67	4	2.02	8	4.08	4	3.60	9	3.09	5	2.50	8	3.96

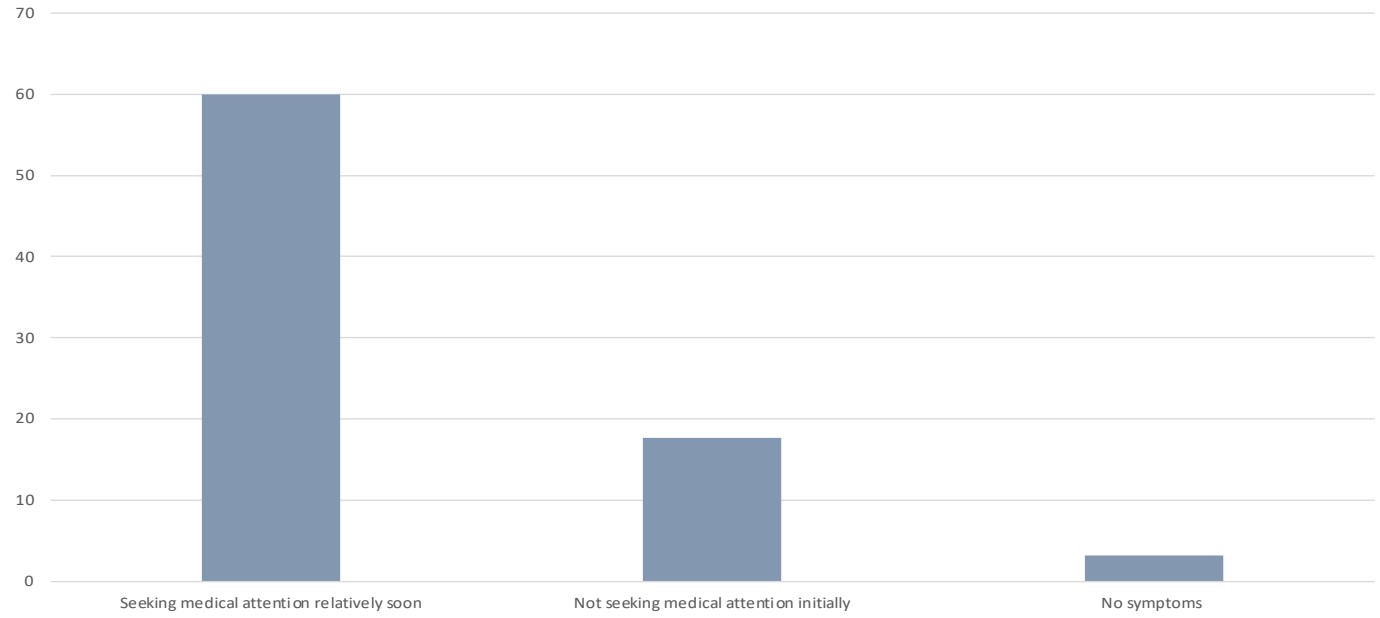


Figure 3.3: Seeking medical attention

Table 3.6: Seeking medical attention – subgroup variations

Seeking medical attention	Reported less frequently	Reported more frequently
Seeking medical attention relatively soon	Diseases of the skin Other rare condition	Diseases of the immune system Diseases of the nervous system
Not seeking medical attention initially	Family or carer Aged under 18	Diseases of the skin Aged 65 plus
No symptoms		Other rare condition

Symptoms leading to diagnosis: Description of diagnostic pathway

In the structured interview, participants described their diagnostic pathway in the healthcare system. The most common descriptions were a complex diagnosis, needing to see multiple specialists before diagnosis (46.52%), and a linear diagnosis after being referred to a specialist from their general practitioner (28.36%). Other themes included being diagnosed in an emergency department/urgent care (13.68%), being diagnosed by their general practitioner during a routine check-up that was not related to symptoms (5.97%).

Participant describes a complex diagnosis, needing to see multiple specialists before diagnosis

So gosh, so I've had a lot of over the years, I've had a lot of visits to infectious diseases departments and I've been put on different treatments and it was only in I think 2021 that I actually got a diagnosis, so. Yeah, I had a couple. They kind of over the years, they've kind of gone dormant for a while and then they come back up again. Or I always kind of had one or two hanging around and it was usually on my backside or on my inner sides. And they they kind of went through, you know, dormancy and then I'd have another one pop up. And I usually have that treated with any just antibiotics at the time through the GP. And anyhow, in 2021 I had three popped up on my low abdomen on my stomach. And then I also had a couple popped up under my bust, which I'd never had before and so. My GP referred me to infectious diseases at HOSPITAL and because with me there's no bacteria that that's in the pus and with me I had some really weird bacterias growing so I went to infectious diseases and I was treated by them. And she referred me to dermatology, the the infectious diseases, Dr. referred me to dermatology and then I was diagnosed.
Participant 017_2023AUDSK

I went to the GP and said my ring finger on my right hand is very white all the time and tingly and numb, so she referred me to a specialist hand surgeon. I saw him, he examined my hands, and he said, "No, I think all the blood's getting through. I think you are okay. You need to go to a rheumatologist." I got an appointment for the rheumatologist.In the meantime, I saw my GP. She said they'll need blood tests so I had the blood tests. By the time my appointment came along, I went to see a rheumatologist. He was holding my hands, and I'm thinking, "You're a nice friendly doctor." He was squeezing the top of my fingers to try and get a pinch fold. Then he said to me, "Do you get heartburn?"I said, "Actually, I get heartburn every day." Then he said to me, "Oh, we'll need to have blood tests done." I said, "I've already had them done." He looked on his computer screen and said, "Oh, that just confirms that you've got scleroderma."I'm like, "Oh, yes? I've never heard of that before." Then he said to me, "Oh, you're taking it very well." I thought, "Oh, oh, this must be serious." Then he also put me in touch with Scleroderma Victoria and said they've got a conference coming up, which I actually missed that year. Then I contacted someone from the information he gave me, and that was my beginning contact with support from Scleroderma Victoria and support groups.
Participant 008_2023AUDIS

We went to speech therapy and OT and through that initially and then with my GP getting referred to constantly was sick with ear infections, throat infections, things like that, like lots of upper respiratory style infections. My GP sent us onto an ENT which is Doctor NAME in CITY and Doctor NAME did his tonsils, adenoids, turbine, turbines, whatever it is operation. He also had an operation because he had very ears that stuck out quite a bit. So it's in between having the tonsil operation and the ear operation that he sent us to HOSPITAL for genetic testing because just wanted to look at like, I didn't even really know what he was looking for or anything like that. And it was going back to my GP and he said this has come through blah blah. He has 22 Q, 11. So then that started that whole journey and of course you just Google it. NAME looks like those children, he had the ears, like the back ears.
Participant 022_2023AUDPA

Heaps of hospital visits and ruling out with some neurologists, I don't know, ruling out other things, and finally got to this diagnosis but it's a long, long years. It takes years.
Participant 01_2023AUDNS

Participant describes a linear diagnosis after being referred to a specialist from their general practitioner

At the time, my doctor didn't know what was going on because they thought it was a post-encephalitis weird thing. They were just getting worse. The feet were getting worse, and the stiffness in my hands was getting worse. He said, "Look, I don't know what is going on, but I'll refer you to a specialist physician. He's very good diagnostic-wise. I saw him, and he looked at me and then said, "Look, I think I know what it is."He checked out the…I was getting a stiff neck and

my face was starting to get stiffer. He just did an assessment then and said, "Yes, I think I know what it is." He then sent me for some tests and diagnosed scleroderma then. It was about three, four months after I developed symptoms. That was very quick.
Participant 026_2023AUDIS

And so once you got to sort of kindergarten age, so as as she was growing or just like Okay, well we went to the hospital and they were like okay speeding issues, put her on solids early and then there was physical development. So I took it through the like to the physiotherapy to learning how to do those, my planning skills to be able to sit up and stand up and all that. And so then when she was into kindergarten, she was getting speech and so as part of the kindergarten sort of they call it like the healthy kids check kind of thing, Okay. So let's go to a pediatrician to see if there's something underlying for. The pediatrician did did urine test and did a blood test to see if there was anything underlying. And so then that came back, the urine was all clear. But then the blood test they did the DNA testing, microarray testing and that brought up the 22 Q.
Participant 017_2023AUDPA

I went to see my GP and after several blood tests, it was first thought that I had rheumatoid arthritis, which which I do have. But then when I went to see the rheumatoid arthritis specialist, I had an attack in his room and he said, Ohh no, I think you may have Scleroderma. And I'm like, oh gosh, what's that? So then he referred me then to a scleroderma specialist, DOCTOR. And yeah and then it was diagnosed from my visit with her that that I did have the scleroderma and Raynaud's.
Participant 015_2023AUDIS

So his school like his daycare basically flagged that he had like said hardly any words when he was supposed to have words and like he didn't walk until he was two and stuff like that. So the school suggested we go see a pediatrician. Actually first we saw a speech pathologist and then they suggested we see a pediatrician cuz he had hearing issues and we thought that was a speech issue. But then the, I think the speech, he just said go see a pediatrician in case. Yeah, in case there's anything else. And then the pediatrician saw some facial markers and you know, kind of put a few things together and said we need to do genetic testing because he suspected something and then he was right. And then he gave us our diagnosis.
Participant 023_2023AUDPA

Participant describes being diagnosed in an emergency department

Yeah, a bit of a funny story. So I was, I'm trying to think, I think I was 23 and I'd had an abscess and it was draining. You know, I had like been putting magnesium. I think that's what it's called on it with a patch. And then I went to have a shower, took the patch off and noticed that it was just like pure blood. Really bright red. And I was like, that's weird. And my mum was coming over to drop off some groceries because I'd just been made redundant with a loophole. So no payout. And yeah, I let her know and she's like, ohh, if it's still bleeding, you know, you will have to go up to the hospital kind of thing. So she left and then yeah, I went to kind of go change it and there was just so much blood coming out and it wasn't congealing or anything. It was just bright red running down my legs. So I rang my mum back and we went to the hospital and they put me through in the ER and we were just sitting there waiting and my mum was doing her pharmacology units, so we were just like, you know, laughing at all the funny stuff in her textbooks and things and a doctor. On her way out, popped her head in like literally running past. And she's like, I was supposed to, you know, have finished my shift like over an hour ago. I'm still trying to leave. Just thought I'd check in with you guys and we're like, well, no one's seen us yet. She asked me what, you know, have been going on and how I'd gotten there and all that stuff and it kind of just looked like a light bulb went off and she's like, I think you need to ask the doctor if it's HS. Well, we don't know what that is, but OK. Yeah. And at that point, I'd already been in surgery, have one, I think maybe one or two, It's all blur at the moment. I think it was one surgically removed by that point and they just didn't even question it, just took it out, got me out done. So here it was quite interesting that, yeah, if I hadn't gone to the hospital, I'd probably still be looking for answers. Participant 018_2023AUDSK

Yes, I was in hospital for my third admission in one year with episodes of pneumonia that didn't resolve. I'd never quite get over them before the next infection would kick in and I'd end up back in hospital again and. Participant 018_2023AUORC

Participant describes being diagnosed by their general practitioner during a routine check-up that was not related to symptoms

Yeah, sure. So I was probably around 18 when I was diagnosed. A family member had mentioned that they were positive and asked me to get tested. So I got tested at 18 and I think not knowing your body, you don't know any different of how you feel.
Participant 004_2023AUORC

OK, I just went to my GP to do some checkup to see if everything was fine. And there was an alteration on my blood testing like my iron was too high. OK. And then we did extra blood testing. Participant 05_2023AUORC

Participant describes that child was diagnosed by surveillance during pregnancy, new born screening, or at birth

I think it was through like a heel prick test or something like that when I was a baby and my sister who's older than me had cystic fibrosis as well. So I think they tested for that straight away because my sister had had it.
Participant 013_2023AUORC

They came back and she said, I've thoroughly checked him and there is nothing wrong with your son. You've been paranoid all through your pregnancy and you just need to enjoy your healthy son. So that was that. And then two days later there was a lot of whispering and you know, I didn't know. I didn't, I didn't connect, I didn't click and PATIENT hadn't passed meconium, and allegedly they're all doing these little internal tests without me knowing…and then suddenly I had a pediatrician. PATIENT had gone off the tests and I didn't know, you know, more blood tests and whatever…and next time I've got a pediatrician sitting on my bed saying now I just need you to just remain calm. Your son has been taken to NICU in Melbourne because all these sugar, sugar levels are dropping very quickly and it's quite concerning and we don't know why.
Participant 006_2023AUDPA

Yep. So when he was a newborn, he…everything kind of started out well, but we did notice he was really salty. Like his, like, you know, kiss a little newborn hands and and feed him stuff. He was really salty and his bowel motions were really like thick and greasy…So we did notice that, like, his bowel motions were just a lot bulkier than a normal newborn should have been. And then when he was about six weeks old, we got the phone call with the heel prick test results and then obviously they told us what was going on and we had to go for all the testing then.
Participant 025_2023AUORC

He was diagnosed through the heel prick test that's done at birth.
Participant 029_2023AUORC

Participant describes being diagnosed by their general practitioner during a check-up related to symptoms

I recall a long time physician GP she wrote it on a post it note, slipped it over to me and that was all that that was said about it. I I still remember looking at it going, I don't even know what that says and it took me ages…Super. You know, what does that mean? I mean it was probably 20, 20, 21 maybe. And so the Internet…I mean at least I didn't have a computer in my home. I was living by myself at that point …So there was no, there were no. Images, photos. What's life like? That was it? It was just a yellow post-it note. I still remember very clearly That was my diagnosis.
Participant 015_2023AUDSK

Well, all all the procedures was carried out by a doctor, you know, I had to seek medical attention when I noticed all all the symptoms and got into the clinic. I was kind of run. I was given some medication, you know, testing every other thing. Yeah, my it was a kind of blood test. The doctor took blood from veins and he sent it to the lab.
Participant 006_2023AUORC

I lived in LOCATION where it's cold. They just thought originally that was just because it was cold and I was just maybe reacting a bit more than other people. Then they took me on a trip to Queensland and it was a lot warmer. I walked into an air-conditioned shop and I just went black and purple and they thought that was a bit more than what we thought. When we got home they went to a GP and I was diagnosed straight away.
Participant 014_2023AUDIS

PARTICIPANT: Had for years, seven years, been complaining to my doctor that I was tired and didn't seem to matter how much sleep I got. I still was tired, and he said, "Oh, you're a young mum. You don't eat properly, you go to the gym and all this type of thing."Then one time when he was away, I saw a locum and I explained to her how I felt and she said, "I've got another patient that has similar symptoms for you and she's been diagnosed with scleroderma." She said, "It's a lot of tests, but she said, I think I'll run

them if you don't mind." She did and came back that my body doesn't absorb iron so basically, it's malabsorption, which is understandable now all these years down the road. I have no idea what my markers are or anything like that.
Participant 013_2023AUDIS

Table 3.7: Diagnostic pathway

Diagnostic pathway	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Multiple specialists needed before diagnosis (Complex)	187	46.52	20	29.85	48	59.26	34	35.79	12	37.50	67	70.53	6	18.75	134	50.00	53	39.55	140	47.62	46	43.40
Specialist from their general practitioner (Linear)	114	28.36	28	41.79	34	41.98	27	28.42	7	21.88	12	12.63	6	18.75	74	27.61	40	29.85	88	29.93	26	24.53
Diagnosed in urgent medical care/hospital	55	13.68	9	13.43	14	17.28	26	27.37	2	6.25	2	2.11	2	6.25	35	13.06	20	14.93	43	14.63	12	11.32
Diagnosed by their general practitioner during a check up related to symptoms	24	5.97	0	0.00	10	12.35	0	0.00	5	15.63	6	6.32	3	9.38	22	8.21	2	1.49	20	6.80	3	2.83

Diagnostic pathway	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Multiple specialists needed before diagnosis (Complex)	187	46.52	33	34.02	60	45.80	71	45.80	60	52.63	34	56.67	97	53.03	87	44.39	51	45.95	136	46.74	91	45.50
Specialist from their general practitioner (Linear)	114	28.36	34	35.05	33	25.19	31	25.19	32	28.07	15	25.00	48	23.74	64	32.65	36	32.43	78	26.80	61	30.50
Diagnosed in urgent medical care/hospital	55	13.68	14	14.43	21	16.03	22	16.03	17	14.91	3	5.00	32	15.66	23	11.73	15	13.51	40	13.75	30	15.00
Diagnosed by their general practitioner during a check up related to symptoms	24	5.97	2	2.06	9	6.87	6	6.87	9	7.89	4	6.67	3	4.55	11	5.61	3	7.21	16	5.50	12	6.00

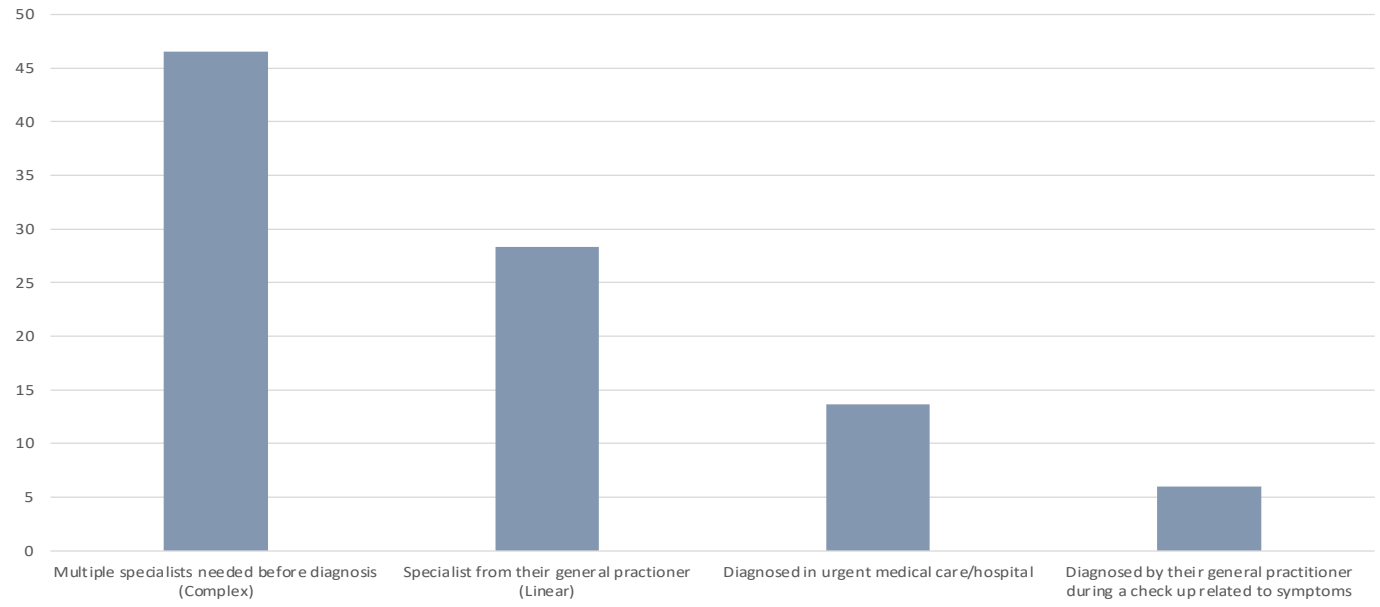


Figure 3.4: Diagnostic pathway

Table 3.8: Diagnostic pathway – subgroup variations

Diagnostic pathway	Reported less frequently	Reported more frequently
Multiple specialists needed before diagnosis (Complex)	Developmental anomalies Diseases of the nervous system Other rare condition Aged under 18	Diseases of the immune system Endocrine, nutritional or metabolic diseases
Specialist from their general practitioner (Linear)	Endocrine, nutritional or metabolic diseases	Developmental anomalies Diseases of the immune system
Diagnosed in urgent medical care/hospital	Endocrine, nutritional or metabolic diseases	Diseases of the nervous system
Diagnosed by their general practitioner during a check up related to symptoms		

Diagnosis provider and location

Participants were asked in the online questionnaire, which healthcare professional gave them their diagnosis, and where they were given the diagnosis.

Participants were most commonly given their diagnosis in the specialist clinic (n=154, 43.14%), this was followed by the hospital (n=151, 42.30%), and the general practice (GP) (n=40, 11.20%).

Figure 3.5: Diagnosis provider

Table 3.10: Diagnosis location

Location of diagnosis	Number (n=356)	Percent
Specialist clinic	154	43.14
Hospital	151	42.30
General practice (GP)	40	11.20
Phone/video conference	9	2.52
Letter	2	0.56
Not sure	1	0.28

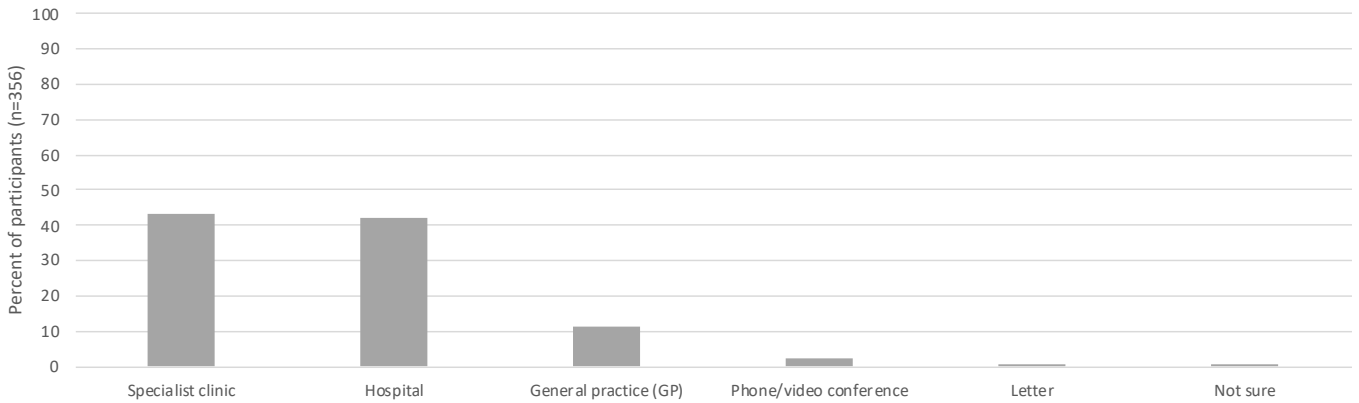


Figure 3.6: Diagnosis location

Understanding of disease at diagnosis

Participants were asked in the structured interview how much they knew about their condition at diagnosis. The most common response was knowing nothing or very little about the condition at diagnosis (61.44%) Others described knowing a good amount about the condition at diagnosis, for example they knew about the condition by learning about it before or during the diagnostic process (7.71%) and knowing about the condition due to professional background (3.23%).

Participant describes knowing nothing or very little about the condition at diagnosis

Not a lot until the specialist told me and actually, he didn't tell me in a very nice way. [laughs] I don't know. I can't remember what field he was in. I can't remember whether he was a rheumatologist or whether he was some sort of specialist in that sense. I really can't remember but now he basically just said I've got scleroderma and I went, what's that? [laughs] I didn't really know anything about anything because my doctor also didn't lead on much as well. I looked it up in the dictionary and got a hell of a fright.
Participant 01_2023AUDIS

I had never heard the word before, so I knew nothing about it.
Participant 004_2023AUDIS

Nothing. I'd never ever heard of it before. I'd never even come up on Google when I was researching like for myself, like what is wrong with me? Because it's just so similar to other cysts and things I guess in the beginning quite easily get confused with that, but no, it didn't even come up. I'd never heard of it.
Participant 006_2023AUDSK

It was only yeah. They handed us A2 page document saying there's fifty children around the world that had this condition. Yeah, and at that time. Just a couple of years ago now, there wasn't any, hardly any research.
Participant 016_2023AUORC

Pediatrician when he was born, when he was quite sick, the the doctor that said that he'd flagged the newborn screening didn't even know the name of the he didn't know what the condition was, so he couldn't give us any information either. So we didn't even have anything to research until the kids hospital called us a couple of weeks later.
Participant 021_2023AUORC

I didn't know anything about it before then. I've never heard of it. Completely new...shocked. I did hit the books so I did a lot of research. I helped inform my family a bit when we couldn't get to the doctors. I took that on myself a little bit. I ended up getting involved with AMDF. You've probably been in touch with them then.
056_2023AUENM

Not really a lot. When I was first diagnosed I was told very, very little. All I was told was that there was no definitive cure for the disease and no definitive cause, that was all I was told. It was more from groups on Facebook that's where I found help, which was absolutely perfect.
067_2023AUDNS

Participant describes knowing a good amount about the condition at diagnosis e.g. understood diagnosis and aspects of treatment

I think I knew a fair bit just because you know, I've had it for 10, 10 plus years. So I'm always reading and like how do I describe it. So I I had a pretty good knowledge. So he didn't have to explain much to me about the disease because I came in or at least suspecting what it was. I knew that there's no cure, there's no real way to kind of treat it. It's just you know eliminating foods from your diet, keeping it clean, maybe antibiotics will help. I know it's an auto inflammatory disease. Like I know almost everything there is to know about it, and I came in knowing that information. So yeah, I just, I I knew quite a fair bit about it.
Participant 010_2023AUDSK

Participant describes knowing about the condition by learning about it before or during the diagnostic process

I knew from just what I'd researched online after the integrative health GP had mentioned it. She basically said to me, "Do a bit of searching online. See if you feel like this is worth looking into," because she said some people don't want a formal diagnosis because it can affect travel insurance and things like that. When I looked into all of that, it really wasn't very much of a difference. I felt like it was worth getting diagnosed. I knew that it explained that my connective tissue and skin was made differently, and so felt like ah, that makes sense as to why I don't heal properly or I don't heal in the timeframe that they expect me to when I've had stitches.
Participant 004_2023AUDPA

I had actually Googled before I went to the doctors, just put hard skin Raynaud's and that was the first thing that came up. I had a breakthrough and realized, oh, that's not really that good, [chuckles] and then went to speak to the doctor. That's how I knew to mention that I did have Raynaud's so that he would hopefully, pick up the same, offer the test to see that. I had no idea other than that. Google was very scary back there. It's not so scary anymore when you do Google scleroderma now. [chuckles]
Participant 018_2023AUDIS

I knew a little bit about it because I'd googled some of the symptoms that they discussed with us and I realized this is probably what he had. So I was. I knew a bit about it and I sort of prepared that that was the diagnosis he would get.
Participant 089_2023AUENM

Participant describes knowing about the condition due to professional background

An awful lot because again, I had to do all the research. I did an entire, I did an entire degree in human physiology in order to save my own life. Like that is how far I had to go. So I understand it a great deal because I literally studied, you know, medical science for three years in order to be able to understand it. And, you know, I I can read and do read my proper academic journal articles and followed, you know, the the content on the the International Consortium's website and all of that. I know more than any practitioner of ever deal with by a massive, massive mark.
Participant 003_2023AUDPA

I have. I've got a health background. So I was hoping against hope that it wasn't, but I was suspecting that that's what my it might be. And his they suspect that his father may have. We suspect his father may have had it. Well, probably still has it.
Participant 009_2023AUDSK

Table 3.11: Understanding of disease at diagnosis

Understanding of disease at diagnosis	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
No or little knowledge	247	61.44	2	2.99	66	81.48	76	80.00	20	62.50	60	63.16	23	71.88	189	70.52	58	43.28	197	67.01	49	46.23
Knowledge: before or throughout the diagnostic process	31	7.71	3	4.48	5	6.17	9	9.47	4	12.50	8	8.42	2	6.25	20	7.46	11	8.21	24	8.16	7	6.60
Knowledge: professional background	13	3.23	1	1.49	1	1.23	1	1.05	1	3.13	7	7.37	2	6.25	7	2.61	6	4.48	10	3.40	3	2.83

Understanding of disease at diagnosis	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
No or little knowledge	247	61.44	42	43.30	90	68.70	87	76.32	28	46.67	126	63.64	118	60.20	69	62.16	178	61.17	130	65.00	117	57.92
Knowledge: before or throughout the diagnostic process	31	7.71	8	8.25	8	6.11	11	9.65	4	6.67	10	5.05	18	9.18	5	4.50	26	8.93	15	7.50	16	7.92
Knowledge: professional background	13	3.23	3	3.09	3	2.29	3	2.63	4	6.67	3	1.52	7	3.57	4	3.60	9	3.09	5	2.50	8	3.96

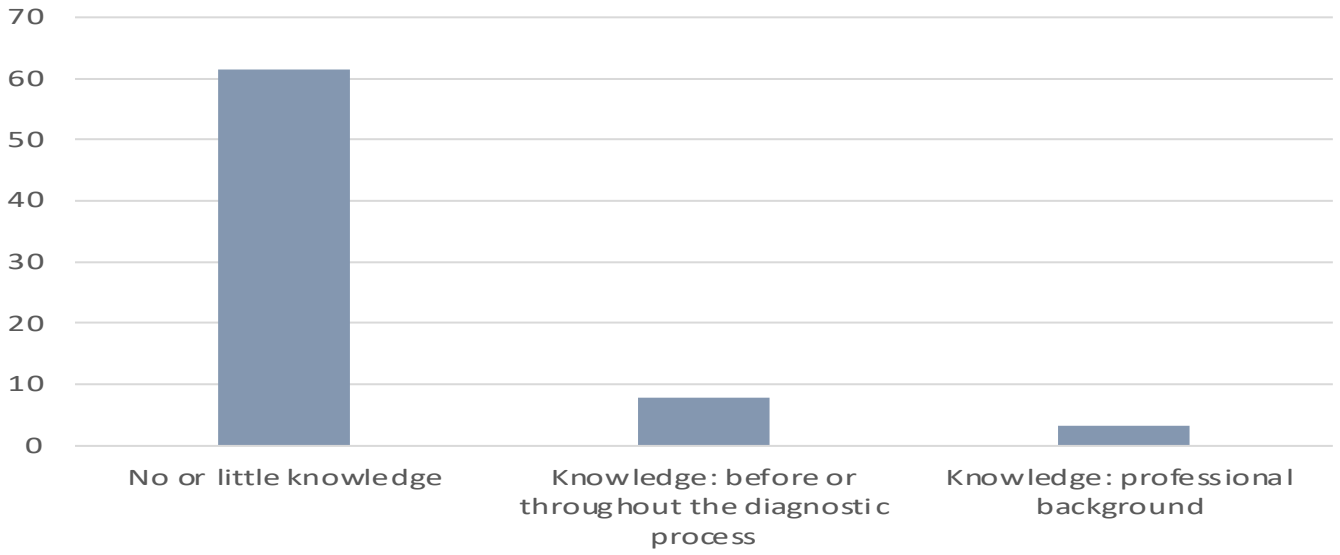


Figure 3.7: Understanding of disease at diagnosis

Table 3.12: Understanding of disease at diagnosis – subgroup variations

Understanding of disease at diagnosis	Reported less frequently	Reported more frequently
No or little knowledge	Developmental anomalies Family or carer Male Aged under 18 Aged 65 plus	Diseases of the immune system Diseases of the nervous system Other rare condition Aged 45 to 64
Knowledge: before or throughout the diagnostic process		

Emotional support at diagnosis

Participants were asked in the online questionnaire how much emotional support they or their family received between diagnostic testing and diagnosis.

There were 79 participants (21.07%) who had enough support, 96 participants (25.60%) that had some support, but it wasn't enough, and 200 participants (53.33%) had no support.

Table 3.13: Emotional support at diagnosis

Emotional support at diagnosis	Number (n=375)	Percent
Enough support	79	21.07
Some support but it wasn't enough	96	25.60
No support	200	53.33

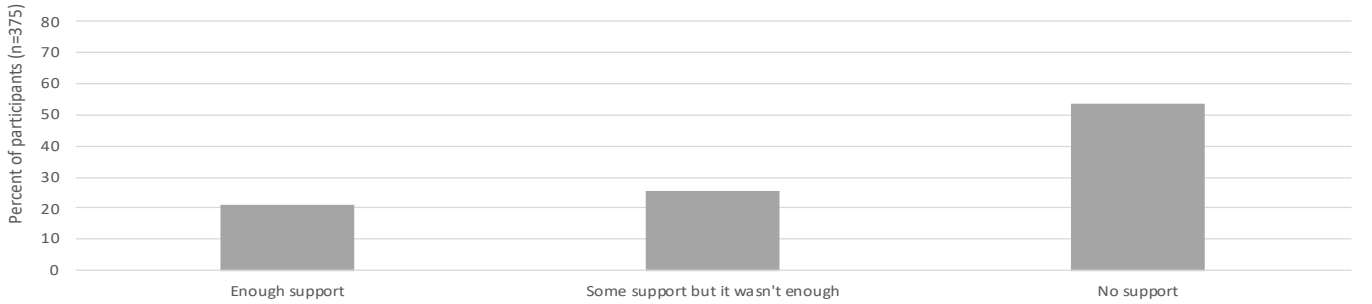


Figure 3.8: Emotional support at diagnosis

Costs at diagnosis

Out of pocket expenses at diagnosis

Participants noted in the online questionnaire the amount of out-of-pocket expenses they had at diagnosis, for example doctors' fees, and diagnostic tests.

There were 146 participants (53.09%) who had no out of pocket expenses, and 51 participants (18.55%) who did not know or could not recall. There were 34 participants (12.36%) that spent Less than $500, 13 participants (4.73%) that spent between $500 to $1000, and 31 participants (11.27%) that spent More than $1000.

Burden of diagnostic costs

In the follow-up question about the burden of costs at diagnosis, for 30 participants who had out of pocket expenses.

For 65 participants (33.85%) the cost was slightly or not at all significant. For 40 participants (20.83%) the out-of-pocket expenses were somewhat significant, and for 87 participants (45.31%), the burden of out-of-pocket expenses were moderately or extremely significant.

Table 3.14: Out of pocket expenses at diagnosis

Out of pocket expenses for diagnostic tests	Number (n=275)	Percent
$0	146	53.09
Less than $500	34	12.36
$500 to $1000	13	4.73
More than $1000	31	11.27
Not sure	51	18.55

Figure 3.9: Out of pocket expenses at diagnosis

Table 3.15: Burden of diagnostic costs

Burden of diagnostic costs	Number (n=192)	Percent
Not at all significant	25	13.02
Slightly significant	40	20.83
Somewhat significant	40	20.83
Moderately significant	47	24.48
Extremely significant	40	20.83

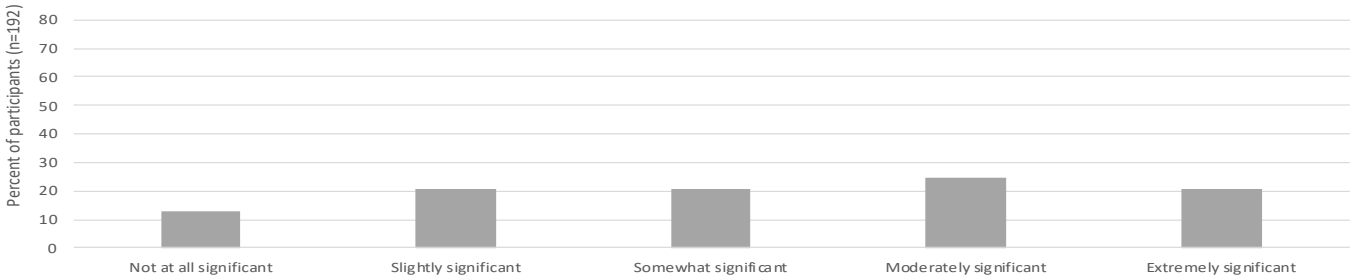


Figure 3.10: Burden of diagnostic costs

We understand that there is a lot of variation within rare and genetic conditions….

But should there be so much variation with the experience of genetic testing?

Have you had access to genetic testing?

No. Not at all.
028_2023AUDIS

No. I didn't remember. At that time, I was so ill. Not ill ill, but I wasn't concentrate what they're saying, a bit stressed
035_2023AUDIS

I've never had any gene tests done. Later after I was diagnosed, I did find out that my nan actually had lupus and they believe that's what brought her fate. They discovered she had lupus when she passed away, because she was bleeding somewhere and they couldn't find where the bleeding was coming from which was caused by her lupus. No one has actually done any digging into anything. It's just pretty much I was labelled with lupus and this is what it is, and now work at it from there.
036_2023AUDIS

Yes, they test us, me and my husband and my other kid because they said it's an inheritance. But there's none they found on my side. NAME DOCTOR said must be in your third generation. That's what NAME DOCTOR said.
042_2023AUDPA

Yes. We were already involved with NAME HOSPITAL Genetics because she's a carrier of cystic fibrosis. We were already in the system for that and then we saw them for genetic counselling for two years, and they…I think tested her to find out what the gene was…then we've also had me and my husband, had testing done to see if she got it from one of us, but we're both clear. She's just unlucky.
043_2023AUDPA

So, we didn't really have any formal genetic testing done until probably four years ago, which they lost the blood and then found the blood and we got the result a year ago. It largely was convoluted things, but he was confirmed to have TS2. I actually don't even understand why we actually did this formal testing because he fit all the criteria of TS2 anyway.
044_2023AUDPA

We were in the first trials with LOCATION when it first came to NAME HOSPITAL and CHILD'S NAME was one of the test participants in that and we had our DNA done, CHILD'S NAME's DNA done and we found a marker in chromosome 16, all the TSC2 marker….That we knew that she had TSC2 as opposed to TSC1, which was historically the milder form. We knew she was going to be presented with at least 80% of the diagnostic criteria within the TSC2. We knew it was going to be severe.
045_2023AUDPA

Not once. It wasn't until maybe a couple of years ago that I found out that my great-grandmother had lupus. No one has ever, ever mentioned that to me.
039_2023AUDIS

I don't know about the biomarker. I know he got genetically tested. They took his blood within the first three days that he was born. They sent that back to us and he tested positive for the gene deletion... I'm guessing it crosses over. Then my husband and I also got tested but we both tested negative for any of those abnormalities. We haven't done the kids, the other kids.
064_2023AUDPA

Yes, NAME DOCTOR had us do genetic testing for CHILD'S NAME. Because we have other children, CHILD'S NAME and I got it done. My husband and I got it done as well. NAME DOCTOR organised that for us pretty much straight away after our initial visit.
065_2023AUDPA

No, [chuckles] funny you ask. Not until I had split up with his father, remarried and I was 12 weeks pregnant with another child. [laughs] At that point, because we, after SON'S NAME's diagnosis, his father and I both went for renal scans and CAT scans. We don't have it, so it was a spontaneous mutation. I thought that was the end of the story, but as in, as I say, I was 12 weeks pregnant with NAME SECOND SON and the obstetricians, you give them your family history and she said, "You've got to have this baby tested." I said, "Why would I need to?" and she said, "No, it's a bigger issue than that."At that point, we got referred back to the genetics department of LOCATION…We had to get a sample from the baby and a sample from SON'S NAME and send them both to OVERSEAS LOCATION in the United States and work out what the mistake was for SON'S NAME before we then knew that NAME SECOND SON didn't have it.
066_2023AUDPA

Not aware of. Unless if they've done it, they haven't told me. I don't think I've ever been involved in that. They've asked me if anyone in my family's had it and I've said no. As far as I know, I'm the only one, but that's all.
012_2023AUDIS

No, but I'll be surprised if my son and daughter don't have it because my son has dreadful problems with his esophagus already and my daughter has problems with her joints and she's also lactose intolerant and gluten intolerance
013_2023AUDIS

Look, if that's a blood test, they may have done it, but no.
017_2023AUDIS

I did ask if I could provide my DNA for studies and stuff like that, which I have done, but nothing that other stuff. No.
018_22023AUDIS

Well, I should say that my auntie has lupus. My sister has MS and then my biological father, he was only diagnosed two years ago with the muscle condition one. Now me and him, my sister and auntie, so like they're not related. My sister isn't related to my real father and my auntie is from my mom's side. At first we all thought it was regarding mom's side of the family, and then only last year my real father was diagnosed with that one. Everyone's talked about doing those markers, but no one has followed through with it. They've even mentioned for my sister's girls to get it done so to see what their future may hold, but like they all mention it, but no one follows through with anything.
023_2023AUDIS

I can't remember but that's…Is that what that is? RNA, whatever that RNA polymerase…They sent me for a whole battery of pathology that I didn't even know… They've gone, oh, hang on a minute. We've got to look up the codes. They hadn't even seen the…they were all for autoimmune. She goes, all these codes are all for autoimmune.
025_2023AUDIS

Not at that stage, but since then, in the last six or three to six months, Doctor NAME, who's my rheumatologist, has asked if I would be willing to be part of the cohort study run through LOCATION, the exchange growth and interest rate. So they have a library of DNA samples and biodata Biobank sort of thing. And so I'm now in the process of getting the blood tests needed for that genetic testing as well.
009_2023AUDIS

No and this kind of makes me laugh because my husband battled the medical system for 20 years because I'm a researcher and we kept pounding at them and pounding at them that that the kind of drugs they wanted him to take made him violently ill anyway. Finally he through the cardiologist, got to a very, very specialist and the specialist did a DNA test and bingo, that was the reason we had battled the medical system for 20 years, so yeah. So when you say, you know, gene testing or anything. No, and I did see NAME the rheumatologist on Friday and I said, she said except for blood test, no, you've not had any DNA testing and so forth because she said that can be very expensive and often they only do that on trials.
003_2023AUDIS

Not since diagnosis. I've got a son who's 38, going to be turning 39 soon. He was born with a lot of fingers missing, and it was due to amniotic banding. Before we had our next child, we did have some genetic testing to see if it was anything genetic but it was not.
008_2023AUDIS

Interesting because, you know, of the fact was it was it caused by stress? Was it caused by, yeah, genetics, you know, not knowing, like because I'm in contact with my Mother's side like my sisters and brothers. I can easily get you know whatever from them. That's not that's not an issue. But they're all half. There's no, well they did say she did say that one sister and I are full but I think we've got..not that I know what my blood type is. Yeah. I'm not sure whether we're yeah whether we're full sisters or half sisters. There's a chance that there's one could be 1/2 sister, a bull sister. So yeah, I'm not really sure there so.
019_2023AUDIS

I asked about it and they told me it wasn't necessary.
016_2023AUDIS

Nothing for genetics. Two children since then, they didn't suggest doing, you know, any screening or anything like that. And I asked about genetic links and they said, you know, no, it's not the like if you have it, your kids are going to have it sort of thing.
024_2023AUDIS

I talked to them about it. It's always been the other way around. Like the level of ignorance in this country is just overwhelming. Like I am yet to meet a practitioner who'd even heard of it before I spoke to them. Even the guy who diagnosed me, I did the educating, not the other way around. And I said, given I've had bowel obstruction, bowel rupture situation, there's a serious chance it's vascular EDS. So I like that, that is a complication that's seen in that version and you know my my symptom pattern could be classical, could be classical, like could be vascular or could be hypermobile…So I asked, I asked for that testing and I was denied it because there's this ridiculous belief in Australia that like, you know, if you have an inborn genetic disorder of course you would have been diagnosed with a kid. So like they have no that the system is not built to understand that we have 20-30 forty year delays in diagnosing conditions in this country and so I wasn't eligible for the Royal Children's Service. And I begged and begged and begged and begged and my GP finally sent me to the one like adult clinic in Melbourne that you know, does this with the staff by registrars and the registrars hadn't heard of it either. And the registrar who was sent to do my consultation walked in with literally a print out from Wikipedia…you know of a patient with EDS in the Wikipedia print out looked at me, looked at the picture, looked at me and went you don't look like him, so you won't have it. And he refused me to test it after that. So for all I know, I've got VEDS and I'm on a ticking time bomb and I can't get anybody to agree to test me.
003_2023AUDPA

Well, that was all done by the geneticist. We've got the results. She explained some of that too, but at the time it's like everything else is there and without the other. I've been doing lots and lots of research trying to understand exactly what they were talking about. There's lots of stuff I still don't understand but I'm getting better.
005_2023AUDPA

So they don't really talk about anything. That's maybe one thing that probably called my interest about doing a survey or a theory on it. Because anyone talks about anything, they just go, Oh yeah, he's gonna see you later.
078_2023AUDIS

First we first saw the gastro. He did have some genetic testing done, but I don't think I ever got the results because I do remember asking the doctor and the doctor's NAME…and I remember, I think he just said, oh, nothing of interest came back. I think that's all he said.
002_2023AUENM

I know they can test for them. Through blood test to see if you carry any of the most common genes. I think it's 200 to 400 genes. You can get tested for that through a blood test. And if you do have cystic fibrosis, then the test is free. But if you don't have cystic fibrosis, it costs about $300 to get that test done. And there was no record of fibrosis in my family either. So that was like recorded. So they didn't think anything of it, they didn't even know what cystic fibrosis was until they had children with it.
013_22023AUORC

No, no. No one's ever talked about that, actually. So yeah, okay.
079_2023AUDIS

In the 12 months following, I was then offered genetic testing, and from that result I was labelled as a carrier for her genetic disease. In hindsight, I have been, told for the majority of my adult life that I have psoriasis. I've been treated as a psoriasis patient for the better part of 25 years and should have been treated differently.
080_2023AUDIS

Well, that's what I think the rheumatologist will be. He's the specialist, the GP basically…he didn't want to get into it too much with me, but he said with the way it is and your vitamin D the way they are, they seems to be the one that he's connected up. I'm guessing that I'll go to the rheumatologist and they'll then want to do further testing like that. I I would love to do it because I actually think I have something else…and I never crawled. I actually boot scooted like I sat up straight and pushed myself along with my arms and my legs. I had very heavy growth spurts and growing pains in my teenagers like I'm not big. But I'm a lot bigger than the rest of my family. My immediate family, anyway. I'm about 4 1/2 inches tall than my older brother, and I have all overgrowths on my bones, and I have backward joints and a lot of things like that, which from the being sick for two years and reading a lot suggests to me that I have some sort of connective tissue issue.
014_2023AUORC

Yes. So she did have have it confirmed that she had the the particular CARGE change on the particular charge gene…and then my husband and I were both test well once they found PATIENT's, and neither of us carry it, but that's very common in CHARGE, it's usually a spontaneous genetic mutation…Really just in helping us to understand whether it was…you know, in future family planning.
087_2023AUENM

So no, not at that time after receiving the confirmation of that I and it wasn't I wasn't sort of been freshly looking at you know what ALS was or what MND was. I think I'd already sort of piece two and two together by that stage. And while I was hoping that it wasn't, I think by the time I was given the diagnosis it was a shock. But it wasn't, you know, it it didn't come out of the blue because I'd looked at what those blood tests were for and, you know, that whole picture of things. So I'd already been doing a bit of reading anyway, and I was aware of those broad statistics that you know 90% sporadic and 10% me and hereditary. But so while I understood the hereditary part since then I I was obviously doing more reading…I was going to raise it with him anyway. So we were at the common place by the time that topic came up and he organized. So do you want me to talk about that process now? Because that's the other thing that's been left hanging. He organized a test and this, this came out more not because we have children. It came out because he was trying to understand the, you know, whether there's any somebody else like family history of of this condition…So I went and had that test. I think that was early mid-October I had that blood test. So then the results for that I followed up repeatedly with his officers to see what the results have become available and we've talked that they take a long, long time and I, they said, well, why don't we just send you a copy when it comes through? And I was thinking, well, I'm sure that's actually not what's supposed to happen and I assumed that if if they were just going to send them to me that you know, maybe that would just mean that it's all negative and that there was nothing concerning about it. His officers emailed me a copy of the, test results and one of them it there's no pathogenic variant detector. I've got it in front of me because it's in my follow up part and the second one said this particular results are just an expansion in this patient and that further and then it goes into talking about the the familial and it is inherited and all the rest of it now because we've got children of them very concerned about what that actually means and I asked my clinic coordinator, who's trying to get hold of Professor NAME. He's now my neurologist.
019_2023AUORC

Not prior to the diagnosis. Once we had the diagnosis we had the genetic testing, but there was no history of it in other our families. And to be honest, I had no idea. I guess it was even a thing even during I guess when I found out I was pregnant or even I guess I didn't discuss with the doctor prior to that, but I didn't even know it was really a thing people did because. We just didn't really have anything, I guess, in our family to warrant even thinking about genetic testing at that point.
020_2023AUORC

Yeah. So both my husband and I had to be screened to see what genetic mutations we had, you know, because it's a recessive condition. So obviously if they found it, then that would also mean. Diagnosis, but I think it was just for their information because my mutation is considered the common mutation and my husband's condition, mutation, sorry, was novel, so they'd never seen it before. But now they know that those two mutations combined cause the same outcome, I guess... So we did speak about that and then when we all were, you know, planning. To see if we would expand our family, we had genetic counselling as well.
021_2023AUORC

A little bit, but not much cuz it's sort of quite technical and detail....Pretty much it's limited in terms of if we know the tests find where there might be a variant or mutation. And then I guess the trio when pairs it to the, yeah, mother and father from what I understand, that's what it's called trio. So that's pretty much the extent of my understanding. So it's pretty limited pretty much because like most of the doctors or health professionals we are involved with don't know much about and you know that it's not generally part of their training
022_2023AUORC

No, I don't think it's hereditary or anything like that. So, but I don't know whether that's got anything to do with it.
024_2023AUORC

Never, never. They never, never said about it being genetic. They never told me getting genes…I see they didn't inquire about migraine history in my family, but I didn't have migraine history in my family and this is doesn't present with migraine, with the pain, the headache variety, but they do say that it's migraine affecting my balance system.
027_2023AUORC

Well, I'm like a massive advocate for, you know, a diagnosis regardless of whether it's a well known disease or disease causing mutation or whatever it may be just being in the fact that you know exactly what it's then and I do a lot of advocacy…. So I think it's better than being undiagnosed regardless of whether it's a totally rare random genetic mutation, but it is deemed to be the cause of the condition. Thing that at some point in the world there's going to be somewhere else with it as well. So PATIENT at the moment there's about 300 people will ride roughly that I know of. We're in a group. There's about 13, 14 in Australia and it's just it gives you something to bounce off. Like I know another kid that's almost exactly a year old and PATIENT. So I know and they're all very like very different kids and different mutations in terms of the RNA sequencing and whatnot, or if they're a translocation or if they're a complete deletion or a market deletion, you know. So that obviously differs on how their condition presents, but you can get a general gist. So we all know that our kids are, we all pretty much know that our kids are going to be nonverbal or boys are nonverbal. Girls are generally verbal. Some lose the skill, some don't. Some are minimally verbal, but mostly most girls were verbal, could communicate some and say some words or sign the boys. On the other hand, majority, let's say 90% cannot communicate verbally whatsoever. So that kind of like prepares you for the future so you know that you're going to work on instead of spoken communication. So I think it just increases quality of life to be honest.
081_2023AUDIS

They told us they were doing genetic testing. They didn't specify it was for CHARGE syndrome, but they didn't tell us that they were trying to search for a suspected genetic disorder.
089_2023AUENM

Yes and so he they ran his test first. And it takes about six months. There is a gene deletion sequence, but even then, only about 2/3 of people with charge display that sequence. So you can still have a diagnosis of charge even if that genetic sequence doesn't show up once they had him confirmed. They did a panel on myself and my husband, but they were fairly confident that they wouldn't find anything because neither of us have any of the diagnostic criteria and charge tends to be a first generation mutation.
091_2023AUENM

I can't remember. I know I had a few blood tests done, like when I first had optic neuritis, and they basically said all your tests don't really indicate anything in particular. Yeah. OK.
096_2023AUDNS
Well, when PARTICIPANT was diagnosed, it was an association so that there was no genetic component at that point or known component. I don't think the gene, the…gene was thought about until 2005 I think. And PARTICIPANT was born in 1992. So yeah, at that. It wasn't until that point that there was a genetic link. But that's my husband and I never got tested. But it was probably unlikely that it's come from us. It's probably a mutation as PARTICIPANT developed.
093_2023AUENM

Yeah, so we did very standard genetic testing, I feel, because we didn't know she was deaf. When she was born she started developing autism-like behaviours when she was very young. So I know they did just the microarray or the I forgot the names of them. We started with DNA testing, we never found anything. So first with saliva, then we started with blood for myself, my partner and her. Went down that road. I was aware of CHARGE syndrome from the very beginning because she has a missing semicircle canal in her ears on both sides, and the only syndrome related to that was CHARGE, and she has severe issues with her balance because of it. So I I knew about the…gene that's linked to that syndrome, so that I did, I did ask for them to run a full exome of that gene. I guess at that stage I was that was the third round I was feeling quite frustrated that we couldn't find anything. So we were linked in with the rare genetic disorder team at the LOCATION and they were very, very understanding and always took my concern seriously and it turned out that that gene test was also completely normal. And at that stage, I thought, okay, maybe I'm maybe I am just going crazy but with technology today they ordered a fourth round of testing and they found an upstream defect. So what that means is the gene that has all the coding information for the…gene is defect. So yeah, that's how we got diagnosed.
094_2023AUENM

My daughter also had a baby passed away at nine months, nine months of age and they somebody suggested a genetic test but then it was put down it was the death certificate was an upper respiratory tract infection. So nothing was ever followed up with a genetic test.
008_2023AUDPA

Well, I don't remember. I don't recall yeah talking about anything like that. Yeah, all I know is that they, they tested both me and my husband, like through would have been a blood test or saliva. No blood test. It was a blood test and that's when, yeah, we found out that my husband also has it, yeah, which he didn't know in the past.
027_2023AUDPA

I'm not sure. We've only ever done the one genetic testing and they said we'll go back when she's about eight or so. We must be due to go back now…I reckon that they said they wanted to see her, but I don't think I've been asked anything like that.
010_2023AUDPA

They just thought they'd do a bit more testing that we could say what if there was something underlying.
011_2023AUDPA

I did, yes...they didn't give me any clues. That's not long after we got a diagnosis.
043_2023AUDNS

Genetic tests and biomarkers

Participants answered questions in the online questionnaire about if they had any discussions with their doctor about biomarkers, genomic and gene testing that might be relevant to treatment. If they did have a discussion, they were asked if they brought up the topic or if their doctor did.

Most commonly, participants had never had a conversation about biomarkers, genomic, or gene testing that might be relevant to treatment, (n=211, 66.56%). There were 28 participants (8.83%) who brought up the topic with their doctor, and 78 participants (24.61%) whose doctor brought up the topic with them.

Participants were then asked if they had had any biomarker, genomic or gene testing. If they had testing, they were asked if they had it as part of a clinical trial, paid for it themselves or if they did not have to pay for it. Those that did not have the test were asked if they were interested in this type of test.

A little over half of participants indicated that they did not have any genetic or biomarker tests but would like to (n=193, 60.88%).

Table 3.16: Discussions about biomarkers

Discussions about biomarkers	Number (n=317)	Percent
Participant brought up the topic with doctor for discussion	28	8.83
Doctor brought up the topic with participant for discussion	78	24.61
Participant had no discussion about this type of test	211	66.56

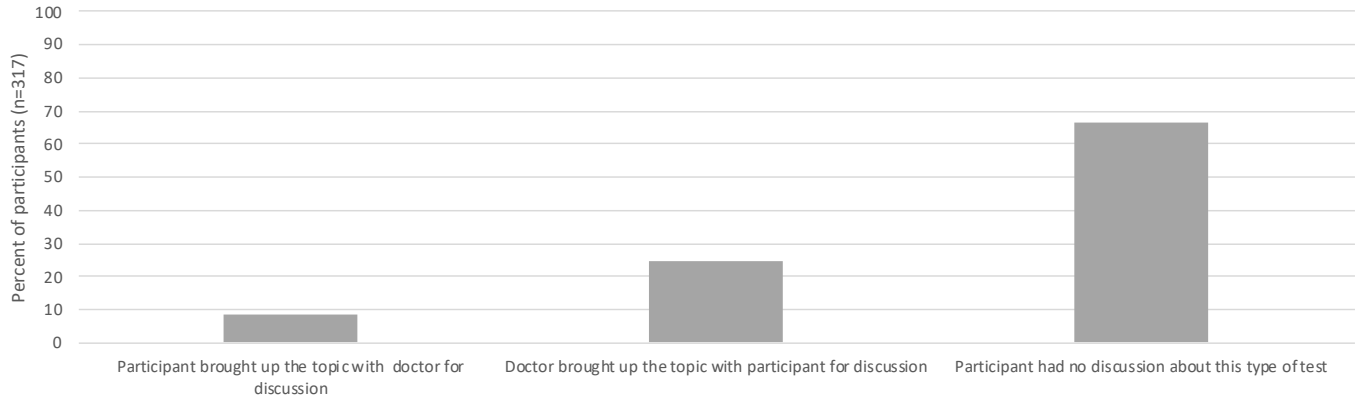


Figure 3.11: Discussions about biomarkers

Table 3.17: Experience of genetic tests and biomarkers

Experience of genetic tests and biomarkers	Number (n=317)	Percent
Participant had this test and did not have to pay out of pocket for it	60	18.93
Participant had this test athrough a clinical trial	21	6.62
Participant had this type of test and paid for it	18	5.68
Participant did not have this test and is not interested in it	25	7.89
Participant did not have this test but would like to	193	60.88

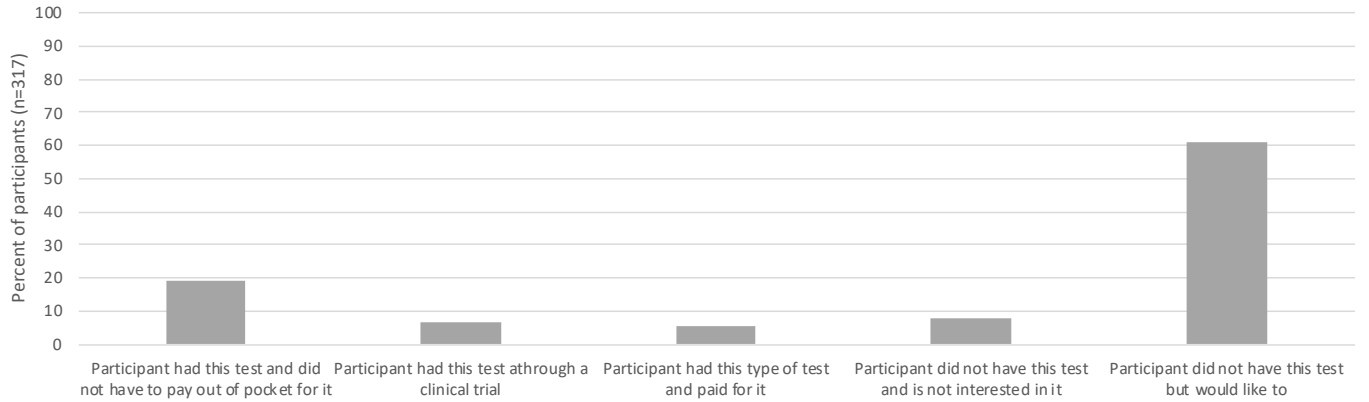


Figure 3.12: Experience of genetic tests and biomarkers

Understanding of prognosis

Participants were asked in the structured interview to describe what their current understanding of their prognosis was. The most common responses were that there was uncertainty around prognosis (26.37%), in terms of symptoms and function/changes in symptoms and function (17.66%), and that they had specific medical interventions they need to manage their condition (15.92%). Other themes included that they were monitoring their condition until there is an exacerbation or progression (15.67%), and had poor outcomes, or a terminal condition (11.94%).

Participant describes prognosis in relation to uncertainty around prognosis

It's considered a rare disease in Australia. So the prognosis for NAME, who is topical in the terms that I am a carrier with a very mild version of the disease and she is a, she has the disease full blown whereas where I don't really and then so her prognosis is unknown.
Participant 080_2023AUDIS

So I mean it's still positive, a positive in the sense that we're hoping we will get access to it and we'll get access to better medication in his lifetime. But it's just it's very hard to tell because it is such a varying disease across the board, but we just you know. We have no idea what it what it means for him in the future.
Participant 020_2023AUORC

Well, it's a bit tricky because I think this particular condition wasn't even discovered until 89. So there aren't a lot of older people with it. They have, well, my son has routine monitoring for the things that it might affect, like his heart and his eyes. And you know, he's ongoing blood testing. So we don't really know what the outlook is. We don't have any information really to go off.
Participant 021_2023AUORC

No I can't because it's such a fickle thing…I think my neurologist who I have a great deal of respect for, would say it's very difficult to make a prognosis as indefinite things, such and such will happen at such a time, or even what organs might be affected.
050_2023AUENM

Participant describes prognosis in relation to specific medical interventions they need to manage their condition

From what I understand, it's all about prevention, so it's really important to-- If you find the right immunosuppressant, you can live quite well, and you can pretty much-- As long as you can get on top of it early, from my understanding, and from what I've been through, I realised that it's very important that if something's going on, that you go and have treatments, like for example, steroids, IV steroids, and that helps you in the long term.
063_2023AUDNS

I'm supposed to be on managing the diabetes side because part of it I've got eye problems. That's being monitored annually by an Ophthalmologist. That's slightly deteriorating. I have diabetes. That's been monitored. I'm supposed to control that. I know I should better than I am. Exercising and the medication. I've had one medication to start with to control seizures. Then, after 12 months or so, they changed that to my current medication that I've been taking for about eight years now.
074_2023AUENM

Participant describes prognosis in terms of symptoms and function/changes in symptoms and function

So he's got a lot of different, different symptoms. So he's got problems with his heart, He's got small kidneys, he has problems with his teeth. He has an allergy which they can't get to the bottom of. That affects his esophagus, so he gets quite so. So he's on medication to keep the swelling and allergic reaction that he gets this in his esophagus down. A lot of people with 22 Q as adults suffer from obesity. He has trouble with knowing when he's had enough to eat, so he'll go for really long periods without eating and then eat too much. He has trouble like because he has an intellectual disability as well as his other problems and but he is quite high functioning and he lives independently but with support but he doesn't make good choices as far as what he eats goes. So he eats quite a childish diet. So he's not good at sitting down to a bowl of salad or something, but that's also difficult because of his problems with his teeth and he has with the problems he has with swallowing. He's getting as he gets older, he's getting particularly stiff. And his flexibility needs constant work.
Participant 026_2023AUDPA

Yeah, so I guess that's a lot of years ago since he was diagnosed. So from there he's developed well, what's come out I guess over time. So I guess it's been sort of as his age, it's kind of been, it's not that it's

progressive I guess, but you know with ageing and developmental sort of milestone sting start to become more apparent. So he's got a moderate intellectual delay, he's got a severe language disorder, expressive and receptive. When he was younger he was diagnosed also with dyspraxia. There is some impulsivity there, probably a ADHD.
Participant 031_2023AUDPA

Yeah, so I guess my vision in my right eye is still not up to what it was before my most recent flare up of optic neuritis, so. Like at the moment all I've had is optic right as flare ups and one flare up of a weak right arm. So yeah, basically just trying to get my vision back up to normal in my right eye and then we're back to normal somewhat.
Participant 096_2023AUDNS

Participant describes prognosis in relation to poor outcomes, or terminal condition

All right, so doctors don't have any prognosis at all. They pretty much tell me to go away because it's…I don't know of any answers, but my personal prognosis is I think the countdown's on and I think there's not long to go, to be honest.
Participant 006_2023AUDIS

Not so hot at the minute. I now have developed pulmonary arterial hypertension in the last three years. Three years ago, I started on one medication that helped. I got back to playing my tennis. Then after about a year, I deteriorated a bit, and I got another medication that helped. I went back for a little while. This is only social tennis. Now, I can't even walk up a flight of steps without stopping, so I'm hugely breathless. I have oxygen now. They're talking about a Hickman catheter, putting that in with a 24-hour infusion, and an assessment for a lung transplant. It's pretty crappy. [chuckles]
Participant 008_2023AUDIS

This is what I just tell myself as a result of the doctors and things, that it will never go away and that it will gradually just get worse. I say it isn't contagious and it isn't fatal, but it's constant and persistent and continually continually getting a bit worse.
Participant 012_2023AUDIS

When I was diagnosed, NAME said, "It'll probably shorten your life." et cetera. He said we would just go along because as I said earlier he explained that there was no medication he could give me, only painkillers and things like that. He didn't seem to know a real lot about it. When I went to him, he only had one other patient that had been diagnosed with it. He's an MS specialist actually, I think that's what he really is noted for. That's about it.
Participant 073_2023AUENM

Participant describes prognosis in a positive way, that their condition is manageable

Oh, I've got a really good prognosis. I think because I've been my best advocate over the years, the last 30 years. I've researched everything. At first, it was very hard to know anything because they didn't have much drug therapy, they didn't have any real knowledge. I did a lot of research and put myself forward for that thing in that sense. I've gone back to the rheumatologist that originally diagnosed me funny enough six or seven years ago and he's been very informed because I've traveled a lot with my husband being in the defense. It's been a multitude of steps, but I'm very well-informed today. I know everything basically.
Participant 01_2023AUDIS

Management, which is hopefully what I'm doing. I can live a fairly normal life again, just with good management. So, you know I, I see DOCTOR on a regular basis. I'm taking better care of myself now. So yes, I'm hoping to live a long life.
Participant 015_2023AUDIS

My current outlook is I'm, I'm really early stages, so it's a good outlook. My dermatologist and I are really just working on what she says is putting out small fires so that they don't grow into something larger. So just managing the condition in the in the kind of meantime and that's really it. I don't really. I haven't really looked much further than that.
Participant 027_2023AUDSK

Participant describes prognosis in relation to it being currently controlled

Well, I had the treatment and haven't needed more.
Participant 010_2023AUORC

Well, mine seems to be pretty dormant at the moment, so I went from what they call stage 3 or probably just stage one now. It's manageable. I'm not under a specialist or anything, I just manage it myself all.
Participant 013_2023AUDSK
Yeah, So it'll be it. It is. It is. It'll be a lifelong condition. There is as as things stand, there is no cure. But I. Apparently take medication that has greatly reduced the, you know, the symptoms and yeah, sort of,

fingers crossed, I guess, that that kind of continues to work.
Participant 032_2023AUDSK

Participant describes prognosis in relation to it being a lifelong condition

Yes, absolutely. Just my mindset is that this is lifelong and so learning how to manage it and be more preventative is helpful. My current goals are just building up strength slowly and then setting up more realistic expectations onto activity pipes and anything that's going to prevent injury really. That's been most effective so far, so I assume looking into the future that will be the same.
Participant 004_2023AUDPA

I don't know, like, it's a bit sad knowing that there's no cure and that I'm going to have it forever and it probably won't be fixed. So a bit depressing. But you know, I have a good partner who doesn't like judge me for the disease I have, so I think that's fine. The overall like pretty negative outlook. I don't think it'll ever be fixed, but it's just something I kind of have to manage.
Participant 010_2023AUDSK

Participant describes prognosis in relation to monitoring their condition until there is an exacerbation or progression

Yeah, just keep going. Basically I get reviewed every six months with my 3 specialists and it's a case of they usually just see me. Ohh, yeah. You're about to find nothing's changed. We'll see you in six months. I'm gonna say when I was diagnosed it was very much if you got anything you want to do, go and do it. And when you've done that we'll organise to put you on the pension because you know, the outlook then was maybe a couple of years.
Participant 007_2023AUORC

Prognosis. Nobody's really talked about a general prognosis. There's just bits of information in terms of talking to a cardiologist who's, you know, said as long as his heart is monitored, it shouldn't negatively impact his lifespan. It's more just talking to the individual doctors and therapists that he sees, and they give little bits of information about the area they're working with, but nobody's actually given a general overview of prognosis.
Participant 089_2023AUENM

Participant describes prognosis in relation to specific timeframe that they are expected to live

She's not expected to live a a full life. It's like the most sort of people with her condition, living into the sort of more early 30s, although treatments have gotten better since they were kids, I suppose. So I guess she's got a bit more of a, you know, she might have a longer life span and there's a lot in the words at the moment, but it is a progressive disease, so she's 6. And like usually by teenage sort of teen, mid teen years they need a kidney transplant. So yeah everything will sort of slowly decline and it affects all of her organs but the kidneys and the the kidneys are the and the eyes are the first affected and and throat too because you get muscle wastage. So dysphagia. So yeah so those things will sort of fail. Yep, but hopefully not for a while.
Participant 015_2023AUORC

Six months ago, my respiratory specialist said two years at best, I quite often, I've got more than that, but then there are days where I think I'm going to be pushing for two years. But yeah, it's sort of just take it as it is daily.
Participant 011_2023AUDIS

Oh well, yeah, like age expectant 38, 38 years old. But now if he gets this drug that's available it will significantly increase it into the 60s. But depends what happens between then and now.
Participant 023_2023AUORC

Participant describes prognosis in relation to probable recurrence, or cycle of recurrence

I guess from research that I've done myself. I I think that there's probably a a 10 to 20% chance that it may recur. I've had fairly drastic surgeries under both arms, which was which was at my own sort of request. Once I've done my own research, I wanted to treat it more aggressively then then then what the dermatologist was sort of looking at. Because going with the dermatologist for for a couple of years, it wasn't really having an impact. So, yeah, so in terms of I suppose outlook, right now I I have no symptoms whatsoever, but I'm conscious that at some stage in the future it may recur or it may turn up in the different in a different location.
Participant 007_2023AUDSK

At the moment, because I've had three ablation procedures, I'm pretty much good. I've had the occasional episode, but nothing like it was.
Participant 032_2023AUORC

Participant describes prognosis in relation to the stage of their condition

Yeah, they have. They've if I can get my current active flares under control and get. Back down to stage two. My prognosis is good. We've just got to eliminate the stage 3 flares that I've got so I can go on to medication to prevent them from getting that bad again. But it'll be trial and error as to what medications work… especially considering I can't stay on steroids forever. It's not good for my liver, it's not good for my head and it is contraindicated.
Participant 012_2023AUDSK

Yeah. So quite severe, so. She she cannot communicate in any way. She's best blind severe behavior issues. So I know they they classify severe CHARGE like in depending on what life face you're in. So with her age and with her behavior being so out of control she's she's pretty severe yeah.
Participant 094_2023AUENM

I mean, he's he's probably got a mild form, which is why they may not have picked it up until his developmental. Yeah, you know, delay was picked up because he, you know, he, he didn't have any heart conditions. He didn't have a clef palate, he didn't have a renal issue. So there was not, it was more for him, it was more of the the physical delay and now it's the developmental delay.
Participant 014_2023AUDPA

Participant describes prognosis in relation to support needed for school or independent living

Her learning's not the greatest. I think she's quite normal in being a teenage girl, but yeah, so the future, a lot of the time it's a lot of mental health issues can come with her syndrome as well, which I think is starting to take shake in her, which obviously will be a thing. I think that's like…recorded things that could possibly she could possibly have or possibly be wrong with her for her at the moment. That's mostly just her speech, her heart and the the the learning, her biggest issues. But we do have a support. We do go to like a group thing and it's very different. Like there's lots of kids there who aren't at NAME's level who are older than her, or there's kids there who are.
Participant 013_2023AUDPA

We had to pay privately for her to help us and the bill was like, you know, $2000 or something. But she helped us get all of our paperwork in place to go to NDIS and apply for NDIS. So he did get approved for NDIS. He got funding, so now we've got funding for lots of stuff that he can do now that he's left school. The thing for the major thing we went for was school labor support, which is so that he can have support in a workplace, but he has actually, we've managed to get him a job at COMPANY, so he doesn't actually have to be under disability employment. He's got it on his own standing and they've just made him from a casual to a permanent part time, which is wonderful in our eyes.
Participant 022_2023AUDPA

Yeah, Yeah, yeah, Yeah. OK. Well, I guess because it the condition is so variable with with people and it's dependent on their health conditions. I would expect just based on the multiple health conditions and comorbidities that she has, I would, unless things advance a lot in the future, I would be expecting that her lifespan might be somewhat reduced from average, but that I'm sort of unsure, you know, that's quite uncertain as a long term, you know, I'm talking long term, like I'm not immediately concerned in terms of quality of life. That is a constant struggle at the moment, trying to get adequate supports in place, dealing with all her comorbidities and her mental health issues and her multiple diagnosis and that affect her ability to function. Just getting her as independent, leading a satisfying quality of life that is a constant, just like everyday thing we're working on.
Participant 038_2023AUDPA

Participant describes prognosis in relation to allied health support

No, I'm not because. The doctor hasn't provided me with that sort of information. I've got a review coming up in October, but he because he didn't want to see me until October. He wanted to see what physiotherapy did for me. So I've had a number of of treatments in the meantime, but he said he'd want to leave it for six months before he sees me again.
Participant 095_2023AUDNS

She's about to turn five on Sunday. It's still on one of the fairly early days for her. In terms of the potential physical impacts of the condition, she's been very mildly affected. She has cardiac anomalies but hasn't required any intervention and no other physical issues. She has facial characteristics and fingers and things but not anything problematic. Her main things in the early years have been hypotonia and low muscle tone and fatiguing and so on, which is still a current issue, particular endurance and some speech delay, and now articulation difficulties that we've been working on with Speech Paths.
Participant 067_2023AUDPA

Yep, so we've been lucky to get…so he's had pretty significant therapy, I guess since he had the diagnosis, which has involved speech therapy, occupational therapy, a lot of work on his gut health. And for my bit of reading on the duplication, it seems to be something that's reasonably frequent. So I suppose his overall general health has really improved, which is great. More and more ear infections shall take a step back. He had to have a when he was two and a bit. He had a tonsillectomy and add noise out and grommets and address those issues.
Participant 020_2023AUDPA

Table 3.18: Understanding of prognosis

Understanding of prognosis	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Uncertainty around prognosis	106	26.37	8	11.94	16	19.75	22	23.16	2	6.25	50	52.63	8	25.00	71	26.49	35	26.12	74	25.17	32	30.19
Describes prognosis in terms of symptoms and function/changes in symptoms and function	71	17.66	17	25.37	17	20.99	18	18.95	3	9.38	13	13.68	3	9.38	39	14.55	32	23.88	55	18.71	16	15.09
Specific medical interventions they need to manage their condition	64	15.92	10	14.93	9	11.11	8	8.42	14	43.75	12	12.63	11	34.38	46	17.16	18	13.43	44	14.97	20	18.87
Monitoring their condition until there is an exacerbation or progression (Incl. with blood tests)	63	15.67	9	13.43	21	25.93	16	16.84	0	0.00	13	13.68	4	12.50	47	17.54	16	11.94	47	15.99	16	15.09
Poor outcomes/terminal condition	48	11.94	1	1.49	7	8.64	18	18.95	2	6.25	14	14.74	6	18.75	32	11.94	16	11.94	36	12.24	12	11.32
Positive: Condition is manageable	32	7.96	7	10.45	7	8.64	6	6.32	2	6.25	6	6.32	4	12.50	19	7.09	13	9.70	25	8.50	6	5.66
Condition being lifelong (Incl. not curable)	30	7.46	3	4.48	6	7.41	11	11.58	7	21.88	2	2.11	1	3.13	23	8.58	7	5.22	27	9.18	3	2.83
Specific timeframe that they are expected to live	28	6.97	1	1.49	2	2.47	10	10.53	0	0.00	10	10.53	5	15.63	17	6.34	11	8.21	15	5.10	13	12.26
Organ involvement/ severity of symptoms/serious condition	26	6.47	0	0.00	15	18.52	6	6.32	3	9.38	1	1.05	1	3.13	24	8.96	2	1.49	22	7.48	4	3.77

Understanding of prognosis	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Uncertainty around prognosis	106	26.37	25	25.77	26	19.85	34	29.82	21	35.00	51	25.76	49	25.00	28	25.23	78	26.80	52	26.00	54	26.73
Describes prognosis in terms of symptoms and function/changes in symptoms and function	71	17.66	22	22.68	22	16.79	17	14.91	10	16.67	35	17.68	36	18.37	20	18.02	51	17.53	37	18.50	34	16.83
Specific medical interventions they need to manage their condition	64	15.92	8	8.25	27	20.61	15	13.16	14	23.33	32	16.16	30	15.31	13	11.71	51	17.53	30	15.00	34	16.83
Monitoring their condition until there is an exacerbation or progression (Incl. with blood tests)	63	15.67	12	12.37	15	11.45	28	24.56	8	13.33	30	15.15	33	16.84	15	13.51	48	16.49	28	14.00	35	17.33
Poor outcomes/terminal condition	48	11.94	12	12.37	11	8.40	14	12.28	11	18.33	20	10.10	26	13.27	12	10.81	36	12.37	23	11.50	25	12.38
Positive: Condition is manageable	32	7.96	8	8.25	10	7.63	9	7.89	5	8.33	13	6.57	18	9.18	9	8.11	23	7.90	15	7.50	17	8.42
Condition being lifelong (Incl. not curable)	30	7.46	5	5.15	11	8.40	7	6.14	7	11.67	13	6.57	16	8.16	7	6.31	23	7.90	17	8.50	13	6.44
Specific timeframe that they are expected to live	28	6.97	9	9.28	5	3.82	6	5.26	8	13.33	11	5.56	16	8.16	3	2.70	25	8.59	10	5.00	18	8.91
Organ involvement/ severity of symptoms/serious condition	26	6.47	1	1.03	14	10.69	10	8.77	1	1.67	17	8.59	9	4.59	8	7.21	18	6.19	17	8.50	9	4.46

Figure 3.13: Understanding of prognosis

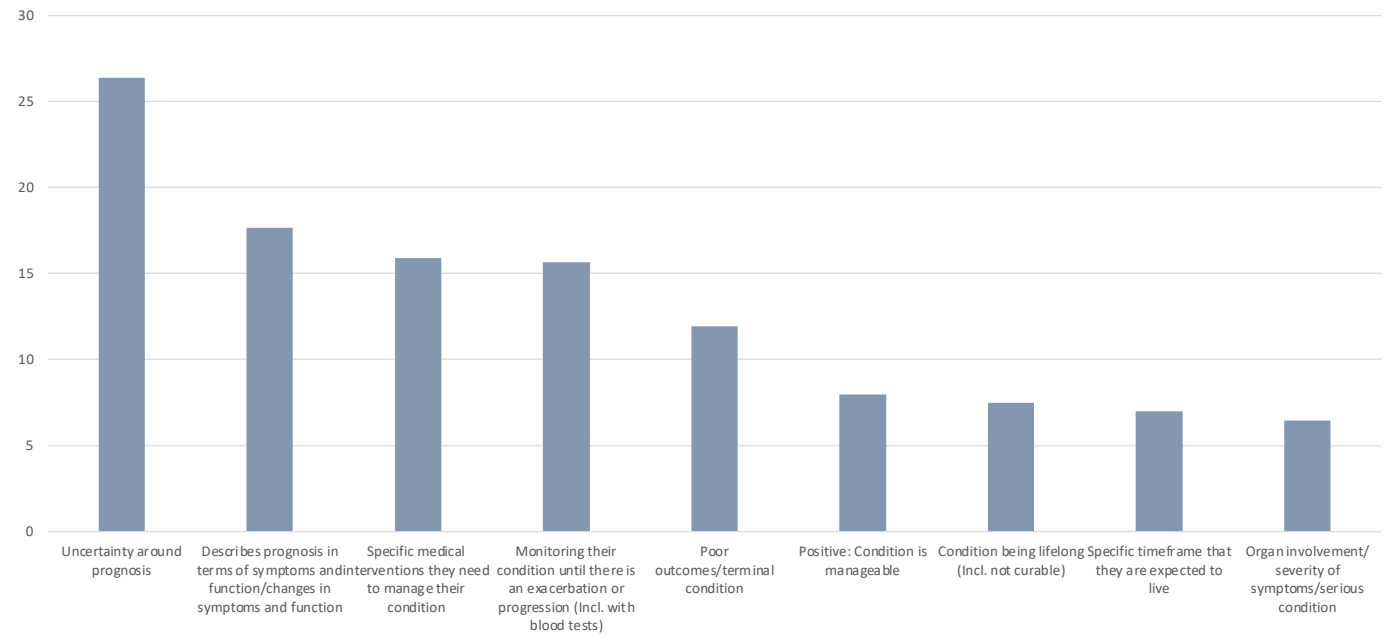


Table 3.19: Understanding of prognosis – subgroup variations

Understanding of prognosis	Reported less frequently	Reported more frequently
Uncertainty around prognosis	Developmental anomalies Diseases of the skin	Endocrine, nutritional or metabolic diseases
Describes prognosis in terms of symptoms and function/changes in symptoms and function		
Specific medical interventions they need to manage their condition		Diseases of the skin Other rare condition
Monitoring their condition until there is an exacerbation or progression (Incl. with blood tests)	Diseases of the skin	Diseases of the immune system
Poor outcomes/terminal condition	Developmental anomalies	
Positive: Condition is manageable		
Condition being lifelong (Incl. not curable)		Diseases of the skin
Specific timeframe that they are expected to live		
Organ involvement/ severity of symptoms/serious condition		Diseases of the immune system

Section 4

Decision-making

Section 4 summary

Discussions about treatment

Participants were asked to recall what treatment options they were presented with and how they felt about the options. Participants most commonly were presented with multiple options (40.52%), and this was followed by no discussions about treatment (24.92%) and one treatment option (22.77 %).

Discussions about treatment (Participation in discussions)

For those presented with multiple treatment options, descriptions included participating in the decision-making process (13.85%) and being told what to do without discussion (11.69%). This was followed by not participating in the decision-making process (3.69%).

For those with a single treatment option, descriptions included being told what to do without discussion (7.08%) and participating in the discussion (5.85 %). Some participants were presented with no treatment options as no therapies are available but allied health or complementary support offered (5.54%), while others had no therapies or options presented.

Considerations when making decisions

Participants were asked in the structured interview what they considered when making decisions about treatment. The most common responses were side effects (46.31%), efficacy (38.64%), advice of their clinician (26.14%) and cost (21.02 %). Other themes quality of life (16.76%), impact on their family or dependents (9.09%), amount of time needed for treatment and travel times (6.53%), ability to follow treatments (10.51%), and ability to work (4.55%).

Decision-making over time

Participants were asked if the way they made decisions had changed over time. There were 201 participants (57.10%) that had changed the way they make decisions, and 110 participants (31.25%) had not changed the way they make decisions.

Where participants had changed the way they make decisions, the most common reasons were that they were more informed and/or more assertive (23.01%), more aware of their health, responsibilities and/or limitations (10.80%), and more cautious and considered (8.24 %). Other themes included more focused impact on quality of life (5.40%).

Where participants had not changed the way they make decisions, the most common reason was that they had always been informed/assertive (6.25%).

Personal goals of treatment or care

Participants were asked what their own personal goals of treatment or care were. The most common responses were to have quality of life/return to normality (22.56%), to maintain their condition or prevent worsening of their condition (19.55%) and have physical improvements in their condition (18.05 %). Other themes included the ability to live independently (13.53%) and wanting to minimise or avoid side effects (8.27%).

Discussions about treatment

Participants were asked to recall what treatment options they were presented with and how they felt about the options. Participants most commonly were presented with multiple options (40.52%), and this was followed by no discussions about treatment (24.92%) and one treatment option (22.77 %).

Participant describes no treatments being discussed

Literally, he said, 'I do not deal' That's a quote. 'I do not deal with people like you, with people like you', meaning people with idiots, because the stigma around us is just overwhelmingly unbelievable. I do not deal with people like you. So now that I've delivered this diagnosis, I need you to not come back. And that was it.
Participant 003_2023AUDPA

I went to a rheumatologist, but I never was offered any treatment or like medication or anything in the beginning. I basically just was told there was no cure and I just have to learn to live with it. Which is fair enough probably because it's probably true, but I've been in hospital this year and I met a lady in there who said she's had lots of help. A lot of people get infusions and that, I've never been offered anything like that but that's okay. I'm managing.
Participant 013_2023AUDIS

At diagnosis, I was actually not given any options. I, the doctor that had diagnosed me, obviously heard of it and seen it, but she didn't give me any kind of like, this is what you can do for it. This is what can help. This is how you banded yourself. Like there was nothing. It was just this is what you have.
Participant 014_2023AUDSK

Participant describes multiple options being presented

Yep. So I think we, the gastroenterologist was just, she talked to us, I guess about the two ways you could treat it like either medication or diet management. And so we tried quite hard with the diet management at the start. So like you do a diet where you take out the top 4 triggers and then we kind of reintroduce food slowly to try and work out what he could have. So I think we, we always knew there were kind of two pathways and we've ended up kind of combining them.
Participant 079_2023AUDIS

Multiple options yes, but all of the treatment options were based around really different antibiotics or potentially hormonal treatments.
Participant 007_2023AUDSK

It was very murky in so far as he would come in to me and say I think it's this, I think I might try this treatment or that treatment. And it was very difficult to get information from him, and it was very difficult to have a discussion about the pros and cons of the different treatment options, he said to me at one stage I want to do X treatment on you, but I've got to make sure you have no cancers in your body. But more than that, he's very slippery in so far as he wouldn't stay long enough to sit down and have a chat right. So I found it very difficult…I found that very difficult, yeah.
Participant 095_2023AUDNS

Participant describes one option being presented

I honestly didn't pay that much attention because I thought it was not a big deal. Do you know what I mean? Like, I walked in there and she's like, 'Yep, you've got HS. If you take these tablets, we'll check you in six months to see if it worked or not'. And I kind of assumed that that was as easy as it was going to be …and I was just like, yep, no worries, thanks. I'll take the pills. And then after a certain period of time, it stopped working and I'd be like, crap.
Participant 026_2023AUDSK

Dr. NAME was a neurologist and he spoke with my GP about options. My GP was great in the sense that he didn't know anything about it but he certainly went and found out for us. He was quite concerned about it being treated with Botox. Then when I went down to CITY, the first neurologist that I saw there really I got absolutely nothing from that neurologist at all. His treatment was very different to what I'm receiving now. He provided no information or options of what else was available besides the Botox. He basically flew down to Brisbane. He injected three points and then that was it. There was no conversation, no anything.
Participant 006_2023AUDNS

Table 4.1: Discussions about treatment

Discussions about treatment	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=352	%	n=67	%	n=81	%	n=95	%	n=32	%	n=45	%	n=32	%	n=225	%	n=127	%	n=256	%	n=94	%
No treatments being discussed	81	24.92	23	34.33	6	7.41	34	35.79	2	6.25	5	11.11	11	34.38	41	18.22	40	31.50	57	22.27	24	25.53
Multiple options	132	40.62	5	7.46	47	58.02	35	36.84	17	53.13	19	42.22	9	28.13	107	47.56	25	19.69	102	39.84	30	31.91
One treatment option	74	22.77	8	11.94	20	24.69	21	22.11	9	28.13	7	15.56	9	28.13	53	23.56	21	16.54	58	22.66	16	17.02

Discussions about treatment	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=94	%	n=118	%	n=90	%	n=50	%	n=172	%	n=172	%	n=95	%	n=257	%	n=178	%	n=174	%
No treatments being discussed	81	24.92	34	36.17	24	20.34	15	16.67	8	16.00	38	22.09	43	25.00	18	18.95	63	24.51	33	18.54	48	27.59
Multiple options	132	40.62	17	18.09	51	43.22	46	51.11	18	36.00	66	38.37	66	38.37	40	42.11	92	35.80	76	42.70	56	32.18
One treatment option	74	22.77	16	17.02	27	22.88	17	18.89	14	28.00	41	23.84	33	19.19	16	16.84	58	22.57	38	21.35	36	20.69

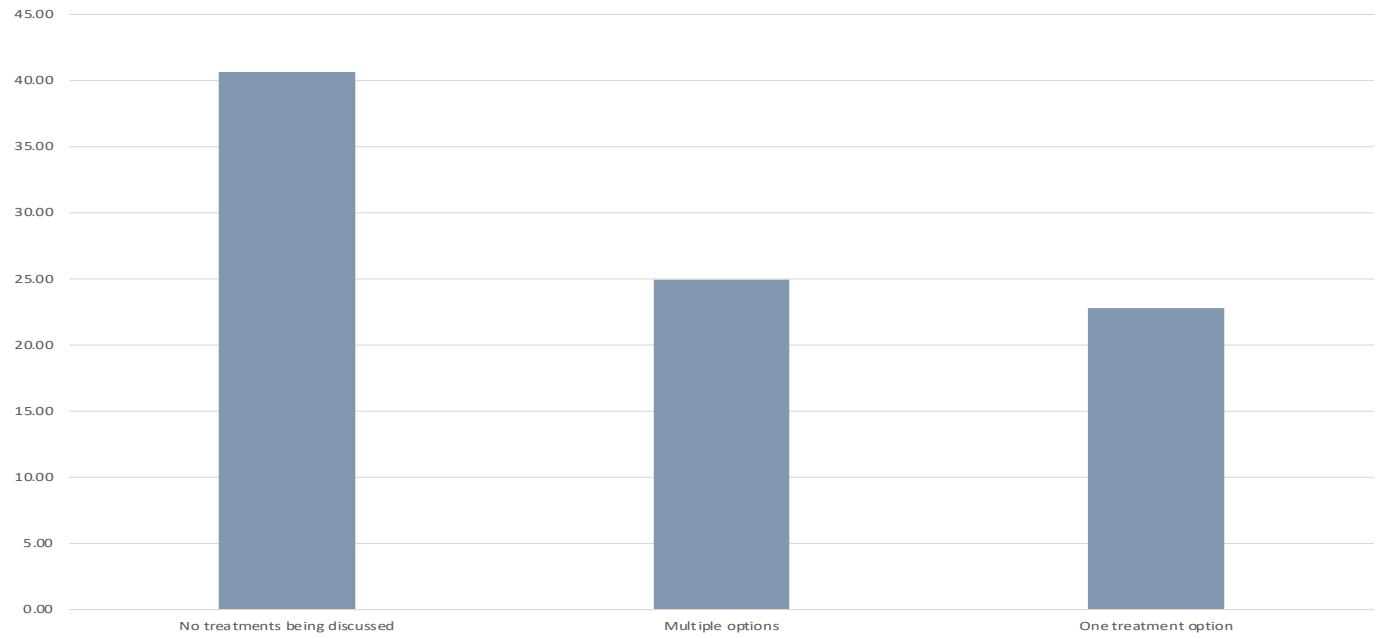


Figure 4.1: Discussions about treatment

Table 4.2: Discussions about treatment – subgroup variations

Discussions about treatment	Reported less frequently	Reported more frequently
No treatments being discussed	Diseases of the immune system Diseases of the skin Endocrine, nutritional or metabolic diseases	Diseases of the nervous system Aged under 18
Multiple options	Developmental anomalies Other rare condition Family or carer Aged under 18	Diseases of the immune system Diseases of the skin Aged 45 to 64
One treatment option	Developmental anomalies	

Discussions about treatment (Participation in discussions)

For those presented with multiple treatment options, descriptions included that they participated in the decision-making process (13.85%), and they were told what to do without discussion (11.69%). This was followed by not participating in the decision-making process (3.69%).

For those with a single treatment option, descriptions included being told what to do without discussion (7.08%), and participating in the discussion (5.85 %).

Some participants were presented with no treatment options as no therapies are available but allied health or complementary support offered (5.54%), while others had no therapies or options presented (6.77%).

Participant describes being presented with multiple options and participated in the decision-making process

The rheumatologist I had at that time, it's not the same, I have a different one now. The one I had at the time, she told me because there's no cure, there's lots of treatment available but is still on trial…it may work well, and others, there's no result. She told me at the beginning, I had to start the immunosuppressant which I'm still taking until now. Then she said maybe I have to do some infusion, which will help me. At the beginning I was scared and then I said, no, I don't want, because I had so much issues with my veins, and so much trouble getting blood tests done. I had a fear of going back and having incision, I don't know how many times, maybe once a month or I'm not sure. She said she will organize a plan for me, but I was so afraid because I had so much bad experience doing this. Even when I had to do some scan, CT scan, they had to find a vein and it was so hard. During that time I even had a surgery, I had to remove my right thyroid and I had such a bad experience. That's why I didn't want an infusion.
Participant 020_2023AUDIS

When I was, my doctor diagnosed me, she put me straight on doxycycline referred me to a dermatologist in Melbourne. This dermatologist was also doing studies on medications and treatments for HS saw a dermatologist initially who. I was not happy with because I hadn't…I went in with my own list of questions and I kind of was getting fobbed off and she was trying to push me towards one of these studies but wasn't prepared to answer the questions that I had there and then. So I had a discussion with my GP. We both felt it was was best that I asked to see a different dermatologist, which I did. So I had a consultation with this next dermatologist. I didn't fit the research program that they were doing at that time because I had only recently been diagnosed, so I was put on Humira. So I've probably been on Humira for what I don't know, for 2 1/2 years I think as far as. As far as most of the other treatments, the only other thing that we've discussed is the possibility of using sorry, my mind's going to go blank now. Steroid injections to help reduce some of the flaring or potential surgery, which is something personally I'm not keen to go down that track.
Participant 001_2023AUDSK

Participant describes being presented with multiple options, however, they were told what to do without discussion

The geneticist didn't do too much about the treatment planning. In his report, it just suggested linking into physiotherapy and some pain specialists perhaps, just from memory. It was quite a detailed report and it was just, "Consider these options," and then that was it. My GP at the time wasn't super helpful. [chuckles] I have changed GPs since then. Basically, I just run my own management now. Now that I know what I need, I know what to say, and what to ask for. That's made a big difference. Participant 004_2023AUDPA

He just wanted to cut. He just said antibiotics and then we'll cut it out. That was the end. And I thought, well, it keeps moving, so how do you just keep cutting it out? And then leaving it, they call it de-roofing. They re de-roof it. So they leave it open to to heal. And I go online have a look at some of the images. You'll be absolutely…and like some like I'm lucky I don't get it under my armpits yet or in my joints, you know, and guys can get it all over their head, on the back of their neck, wherever you've got hair. And women get in the under their boobs and and I've been lucky enough not to have that sort of thing. Mine's but lucky. I'm lucky but some it's retained to my butt, which is a bit more private. But some kids, even kids have it from an early age. Prepubescent, it's it's just awful for them and I don't know how they cope with it and nobody knew that it was acne, you know? But it's not just acne. Participant 024_2023AUDSK

Participant describes being presented with multiple options but did not participate in the decision-making process

This is about 7-8 years ago I went to HOSPITAL. You go there to outpatient. They always see a different person in about. They were kind of pimples in about a year and a half after I've been going there and they put me in all sort of… I can't even remember. Every time I went there, they put me on a different tablet and then they decided to do hydrocortisone injections on the buttocks area and they ruined my life. Not only damage the area, they created these lamps full of fluid that they were constantly oozing, oozing, oozing, oozing only a standard kind of decrease the oozing. I don't know if it is because what I'm taking at the moment, but it it's about 3-4 months ago that the ooze has decreased quite a lot.
Participant 031_2023AUDSK

The conversation was really the shoving medication down my throat. Like just it was initially taking steroids and taking like Prednisone and for reflux and taking a steroid puffer. But it so it's the whatever, the orange flixotide, but it was, yeah, but it was swallowing the flixotide instead of inhaling the flixotide.
Participant 078_2023AUDIS

Participant describes being presented with one option/approach, that they were told what to do without discussion

There was nothing. No, just they was just like, here's some drugs. There's no real, nothing we can do for you. Just try these drugs and see how we go. Then once, they didn't give me any other option. Participant 018_2023AUDIS

PARTICIPANT: I haven't really been given options. It's kind of this is what the plan is…You're kind of just being spoken to.
Participant 096_2023AUDNS

Participant describes being presented with one option/approach, and had some but very little discussion

Antibiotic and they don't work for me. They did and now they don't. But I have to have that conversation every time with the doctor and still given the same ones. So like I said, I've had no medical intervention or assistance really.
Participant 018_2023AUDSK

Well, I suppose the main one was the palate and really there was the operation available, but there was no, there was no God to say, you know, this is definitely going to fix her speech. You know, it depends some, some get, you know, better results than others. She's still got a tiny little gap. So therefore her, the discussions were about the way she's going to sound because she's quite nasally in her speech. You know, if they got a a bit of, you know, less of a clearance there and closed it a little bit more than she would be less, you know, nasal sounding. So that was one of the main things. The heart we didn't really have much of a conversation on apart from one doctor thought he heard a heart murmur and so we went through cardiology and they did a lot of scans and things, but they couldn't find anything there. Participant 024_2023AUDPA

Participant describes being presented with no options/approach as there were no therapies are available but allied health or complementary support offered

So there was no real treatment because there wasn't anything that needed to be treated. I guess once once we had her her diagnosis, it explained a lot of things like her, her delays and her size. So then from then on, we've just been able to go on to things like occupational therapy, speech therapy to try and help with those delays. Yeah, and medically…I said medically there hasn't been any problems as yet.
Participant 010_2023AUDPA

That was just with the geneticists, just sort of kept us up with the pediatrician. And then we had to start like therapy, OT physio, OT physio. Just trying to think what the others were, speech pathology. We went to programs for sensory perception at the hospital. Yeah, we've got individual education plans in progress in process. Yeah. So we work with that then. And cardiology testing as well.
Participant 11_2023AUDPA

Participant describes being presented with no options/approach as there were no therapies are available

Because it's vascular, there's not much I can do. You go, "Okay. I'm just one of those that sit in the corner and wait." Yes, you look normal, you look okay, fine but they've haven't discussed what options are available. To my daughter, they discussed with her the options of if and when she wants to have children, this is what you're going to do. For me and my son being male, there's not much that we need to discuss. Participant 005_2023AUDPA

It was a bit too it was almost a bit too much soft touch as far as you know how I left the hospital, what my understanding was was a little hazy at first but they did make it clear enough that there wasn't any medication available so they they there was no treatment available they they catch that in in. Participant 011_2023AUORC

Participant describes being presented with no options/approach as there were no therapies are available but monitoring of condition was offered

There was no intervention the first couple of years. It was just regular blood tests and monitoring every six, six or 12 months. So that took me up to maybe my late 20s or early 30s. No, it would have been late 20s. It took me up to my late 20s of just regular blood tests.
Participant 004_2023AUORC

So pretty much it was, we were given a 'well this is it and this is the main things that happens. So this is, this is the boxes that you need to go and tick off. You need to go and see a cardiologist and you need to go and have a ultrasound on your kidneys and you need to go and have a cervical spine, X-ray, and you need to, you know, go and see these people like these different specialists'. Then they said, you know, of course, she was only 12 at the time, but they said, you know, when it comes to planning a family, you know, you need to come back and see us, you know, and all of that sort of stuff. And that was pretty much it at the time. We were still under the well, we were still able to go to the HOSPITAL. So yeah we were we were pretty good. I'm sure there was a wait list but of course I can't remember too much about it but yeah so that. So that was pretty much the only the only feedback we were given. Here's your condition and this is the…to see we know what are the problems there are that we need to monitor on an ongoing basis. So that's all that pretty much was given to us at the time.
Participant 37_2023AUDPA

Table 4.3: Discussions about treatment (Participation in discussions)

Discussions about treatment (Participation in discussions)	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=352	%	n=352	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=247	%	n=105	%	n=252	%	n=98	%
Multiple: Participated in decision-making	45	13.85	1	1.49	15	18.52	15	15.79	6	18.75	6	13.33	2	6.25	35	15.56	10	7.87	36	14.06	9	9.57
Multiple : Told what to do without discussion	38	11.69	1	1.49	15	18.52	16	16.84	2	6.25	4	8.89	0	0.00	34	15.11	4	3.15	30	11.72	8	8.51
Multiple: No reason provided	26	8.00	2	2.99	11	13.58	2	2.11	5	15.63	3	6.67	3	9.38	22	9.78	4	3.15	20	7.81	6	6.38
Multiple: Did not participate in decision-making	12	3.69	0	0.00	0	0.00	4	4.21	2	6.25	5	11.11	1	3.13	10	4.44	2	1.57	8	3.13	4	4.26
One option: Told what to do without discussion	23	7.08	0	0.00	9	11.11	7	7.37	2	6.25	4	8.89	1	3.13	23	10.22	0	0.00	17	6.64	6	6.38
One option/approach: Participated in the decision-making process	19	5.85	0	0.00	2	2.47	12	12.63	1	3.13	2	4.44	2	6.25	8	3.56	11	8.66	15	5.86	4	4.26
One option: No reason provided	16	4.92	4	5.97	5	6.17	0	0.00	3	9.38	2	4.44	2	6.25	12	5.33	4	3.15	11	4.30	5	5.32
One option: Some but very little discussion	11	3.38	3	4.48	3	3.70	2	2.11	1	3.13	2	4.44	0	0.00	7	3.11	4	3.15	10	3.91	1	1.06
No options: No therapies are available	22	6.77	8	11.94	2	2.47	3	3.16	0	0.00	3	6.67	6	18.75	10	4.44	12	9.45	12	4.69	10	10.64
No options: No therapies available, allied or complementary offered	18	5.54	0	0.00	0	0.00	18	18.95	0	0.00	0	0.00	0	0.00	5	2.22	13	10.24	18	7.03	0	0.00

Discussions about treatment (Participation in discussions)	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=69	%	n=116	%	n=108	%	n=59	%	n=172	%	n=172	%	n=100	%	n=252	%	n=176	%	n=176	%
Multiple: Participated in decision-making	45	13.85	7	7.45	16	13.56	17	18.89	5	10.00	16	9.30	29	16.86	12	12.63	33	12.84	20	11.24	25	14.37
Multiple : Told what to do without discussion	38	11.69	2	2.13	13	11.02	16	17.78	7	14.00	19	11.05	19	11.05	12	12.63	26	10.12	20	11.24	18	10.34
Multiple: No reason provided	26	8.00	2	2.13	13	11.02	10	11.11	1	2.00	14	8.14	12	6.98	10	10.53	16	6.23	17	9.55	9	5.17
Multiple: Did not participate in decision-making	12	3.69	2	2.13	3	2.54	2	2.22	5	10.00	9	5.23	3	1.74	2	2.11	10	3.89	6	3.37	6	3.45
One option: Told what to do without discussion	23	7.08	0	0.00	8	6.78	8	8.89	7	14.00	17	9.88	6	3.49	6	6.32	17	6.61	15	8.43	8	4.60
One option/approach: Participated in the decision-making process	19	5.85	11	11.70	3	2.54	2	2.22	3	6.00	13	7.56	6	3.49	2	2.11	17	6.61	9	5.06	10	5.75
One option: No reason provided	16	4.92	1	1.06	7	5.93	5	5.56	3	6.00	8	4.65	8	4.65	3	3.16	13	5.06	7	3.93	9	5.17
One option: Some but very little discussion	11	3.38	1	1.06	7	5.93	1	1.11	2	4.00	5	2.91	6	3.49	4	4.21	7	2.72	8	4.49	3	1.72
No options: No therapies are available	22	6.77	11	11.70	5	4.24	1	1.11	5	10.00	10	5.81	12	6.98	6	6.32	16	6.23	9	5.06	13	7.47
No options: No therapies available, allied or complementary offered	18	5.54	12	12.77	4	3.39	2	2.22	0	0.00	7	4.07	11	6.40	2	2.11	16	6.23	3	1.69	15	8.62

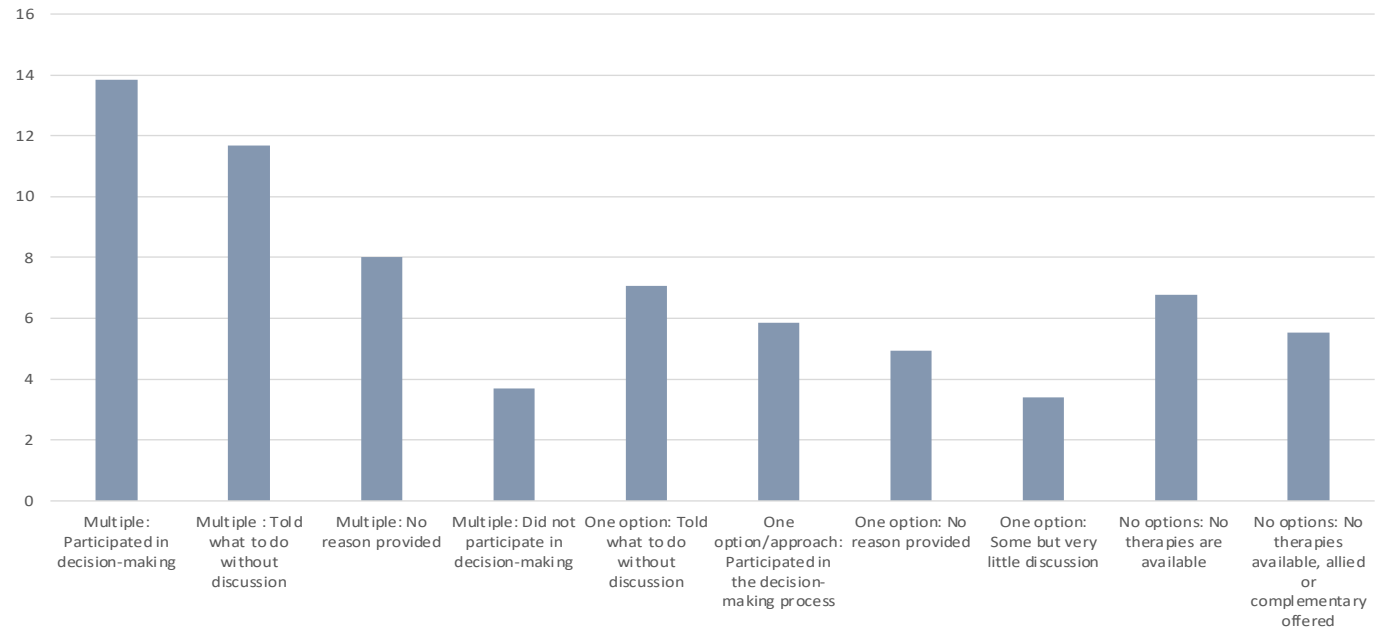


Figure 4.2: Discussions about treatment (Participation in discussions)

Table 4.4: Discussions about treatment (Participation in discussions) – subgroup variations

Discussions about treatment	Reported less frequently	Reported more frequently
No treatments being discussed	Diseases of the immune system Diseases of the skin Endocrine, nutritional or metabolic diseases	Diseases of the nervous system Aged under 18
Multiple options	Developmental anomalies Other rare condition Family or carer Aged under 18	Diseases of the immune system Diseases of the skin Aged 45 to 64
One treatment option	Developmental anomalies	

Considerations when making decisions

Participants were asked in the structured interview what they considered when making decisions about treatment. The most common responses were side effects (46.31%), efficacy (38.64%), advice of their clinician (26.14%) and cost (21.02 %). Other themes quality of life (16.76%), impact on their familiy or dependents (9.09%), amount of time needed for treatment and travel times (6.53%), ability to follow treatments (10.51%), and ability to work (4.55%).

Participant describes taking side effects into account when making decisions about treatments (Total)

About the side effects, because I live on my own and I don't want to feel more sick after that because there's no one to look after me if I get sick at home. That's the main thing I look about the side effects.
Participant 020_2023AUDIS

Side effects is a big one for me. Obviously I don't want to put on heaps of weight or feel nauseous, or if I can avoid some horrible side effects, I will and I guess not so much yet. But as I said in the future, like if I can be on them while pregnant or how long I have to be off them before being pregnant, yeah.
Participant 095_2023AUDNS

Well I just the side effects of different medications and what, what you know whether the side effects are worse than the and the actual thing but in the early …you have no choice sometimes whether yeah it's just mainly the side effects that the methotrexate when I took it 16 years ago it. It may…I got I got very sick sort of like chest and I also had mouth ulcers and stuff like that, all the side effects. And also it did something to my liver. So This is why I didn't want to take it again. Yeah. So I just, I just told him and he said I don't have to take it if I don't want to. It's up to me, which is you can advocate and say what, you know, whether I wanna take this medication or not, I, I do my research.
Participant 088_2023AUENM

Participant describes taking efficacy into account when making decisions about treatments (Total)

Medical and scientific evidence. Basically I read the publication. Yeah, that's how I decided it becomes difficult because still as of this date, there is only one drug approved in Australia for the treatment of HS. So…
Participant 008_2023AUDSK

Yeah. So in terms of making the decisions, I guess the efficacy and I guess I do some research in terms of…you know, not very academic research I must say, but I will look at any research papers that you know just through a search through a search engine and what their results were and also you know the period of time that it was done. Like, was it done last couple of years or was it done 10 years ago? It was ten years ago, I would ignore it and also look at different Scleroderma sites and talk to my GP. Trust my GP enormously and, you know, really talking to professionals such as the scleroderm clinic at Monash. So these are all the themes, yeah.
Participant 010_2023AUDIS

The efficacy of the treatment is is a is a big concern, you know particularly like coming from the, you know from the the experience of interferon where it was a very low like statistically very low success rate. Like I want to know that what I'm taking is going to have a you know, measurable, tangible, noticeable you know impact in my, in treatment yeah. And just the and also kind of you know absolutely tied to that is you know what are what are the potential side effects.
Participant 011_2023AUORC

Participant describes taking cost into account when making decisions about treatments (Total)

Probably cost is one currently that I've taken more of a consideration. Honestly with the side effects and everything it's it's low impact, I've got to go in open minded. There is no cure so I can't go when thinking it's going to fail, so I'll give everything a good go. And then ultimately wait to see if it pays off. With the cost component, it's more so of timing it so that I know I can afford it other than I wouldn't necessarily delay a treatment, I would just take note and make sure I got everything in order.
Participant 026_2023AUDSK

When it comes to, I think, cost when I could no longer work, a lot of decisions were around the cost of things as well because I was working full-time until three years ago where it was just too difficult. Thinking back now, I should have been more self-advocate about changing hours of work, and things like that, to assist with my ability to continue. At that stage, I was just so, "No I need to stop. I can't do it anymore."
Participant 026_2023AUDIS

Participant describes taking the advice of their clinician into account when making decisions about treatments (Total)

Well, I guess the main one is, will it help? [chuckles] I'm willing to try almost anything as long as it's prescribed by someone reputable or someone I know. If they said, "Try a particular medication," I'd try it or whatever it might be. No one has suggested anything very startling to me, I don't think so. That's my inclination in general in terms of, I've gone into various medical sampling tests and things over the years. When I'm asked by doctors or nurses or in a hospital if I'm willing to go into a test situation, I always say yes because I think it will do good for somebody if not for me. I tend to agree with trying anything that they suggest. Participant 012_2023AUDIS

I think I'm pretty trusting as a specialist and think they know what they're doing. I know you've got to advocate for yourself. If anything's not right, I have my list and I raise it at my appointments, trying to do the right thing. In regard to medication, I did at one point make an appointment and go and see the pharmacist, only because I was getting medications from different people and I wanted to make sure the combination of them and what time of day, can some be taken next to the others because there's so many. There's only so much during the day when you can take things. The pharmacist was good. I left a list and then went back and they said, yes, basically what I was doing was right, but I was a bit nervous about it. In regard to which medications, I trust them to be recommending [crosstalk].
Participant 017_2023AUDIS

I sort of put my hands in the doctor's hands because literally I don't know what else I can do because you know what I mean, what they sort of say, like I don't know what other, what other solutions there are, if you know what I mean…I've sort of asked questions, you know, why have I got it, all that kind of stuff…I think it's just bad luck of the draw kind of thing. So it's not hereditary, it's not something that you do. It's just literally, yeah. I've just caught it out of bad luck kind of thing. So yeah.
Participant 024_2023AUORC

Participant describes taking quality of life into account when making decisions about treatments (Total)

I think the impact that whatever I'm taking has on my health. Like obviously take Humira was I guess the big one, but I was….So the, the, the decision to take Humira was because I wanted to improve quality of life. Now I think there's an element of you know, quality over quantity and my, my quantity of quality of life at that time was not good at all. So my decision to take Humira was based on that.
Participant 001_2023AUDSK

Big side effects because there's a lot of treatments today that have a lot of side effects, and I have to weigh that up. I've already been on medication that has really affected me, and in the end, it had some negative results. I'm very well-informed now. I don't just sit there and take what's next. I just say, hang on a minute, how far is this going to take me, and is it really worth? I've always said to my doctors and physicians, whoever they are, I want quality of life, not quantity.I don't need to extend my life. I want to know that I'm going to enjoy my life. It's more important to me than ever. These last 30 years have taught me that. [laughs]
Participant 001_2023AUDIS

Quality of life, quality of life is because of effective treatment. So that I wouldn't say it's been a quality of life. It was just effectively treatment directly leads to quality of life improvement.
Participant 002_2023AUDSK

Participant describes taking the impact on their family or dependents into account when making decisions about treatments (Total)

Now we've got way too much going on, so we're having to make some prioritizing what feels either most important for her at the moment or what we're actually able to manage as a family. We're looking at, particularly at this point, it's about the school readiness and trying to prioritize the things that we think will help her fit in and thrive most in that environment, and having some of the other things take a back seat a bit more. We're fortunate that cost hasn't particularly been a factor because we've had good NDIS plans throughout and relatively speaking, financially okay. Even though we pay a fortune in dental treatment, we haven't had to use cost as a factor to decide not to have treatment.
Participant 067_2023AUDPA

The impact on the family as a whole in terms of like how we're going to manage. Like for example, I think it would be really good for her to do a sleep study, but I've never been able to take like because neither of my children will sleep without me and my husband can't really manage things when I'm not here because he struggles with all the neurodiversities in the house that like, we just haven't been able to do it because she can't sleep at a hospital for five months. So there's, you know, family management, there's affordability, there's the impact. Participant 018_2023AUDPA

A lot of, a lot of change over the years. It sort of depends on what point in time. Our main consideration of course is, is this the right thing for NAME and it's going to benefit him. Not OK well, everyone is doing this. This is what we should do, or this is what someone's recommended. It's like, is this going to benefit him or is this going to distress him or is it going to distress him for a small amount of time until he gets used to it, which is a lot of therapy. And then it's going to better fit him long term or is it going to be too much of a mental battle for the family and for myself for him to do this therapy that may or may not work?
Participant 081_2023AUDIS

Participant describes taking amount of time needed for treatment and travel times into account when making decisions about treatments (Total)

Side effects, any known side effects that they have at that point? Her age, the life, the effect on her fertility and her liver and any organs that may be life limiting access to the trial because often these things are done in Melbourne and Sydney and we live a long way away from that and what would be expected from us in person or financially comes into it as well and things like that. So access and any additional outcomes that would be expected as a result of testing the drug or the procedure.
Participant 080_2023AUDIS

Affordability. Local access, you know, like I don't want to be, you know, go driving an hour and a half to get to the treatment center because I have to manage my stress around this condition.
Participant 027_2023AUORC

Participant describes taking their ability to follow treatments into account when making decisions about treatments (Total)

The convenience because I mean say, well at the moment I'm basically taking the one pill…I mean that there was a time when I was taking over 40 pills a day. So it comes down to you know how convenient is it going to be and it is it easy to maintain and that's that's what I found because I'm just on the the one pill basically it's, it's it's not a problem at all.
Participant 007_2023AUORC

Probably the biggest thing is not overloading him with with too much that we've we've done a lot but just been conscious that yes still a 5 year old boy. Yeah, yeah. Became a little bit overwhelming there for a while with him.
Participant 00_2023AUDPA

Okay. I take into consideration like what, what is you know, that is evidence based I guess. And that I take into consideration all PATIENT's comorbidities and whether those have been considered when they're talking about treatment options or medication options. Whether that's going to impact on her quality of life and her function and how difficult things might be for her to tolerate or to follow through. And I'll, I'll, I'll just sort of, I'll bring up these issues, I'll question them. I will you know on her behalf, you know she's always there and also I'll explain things in language that she understands to make sure she's understanding what's going on. But we would generally, you know, always follow through with recommendations by the doctors unless we feel that they're really against her. They're not sort of considering her as a whole person, if you know what I mean.
Participant 038_2023AUDPA

Participant describes taking the ability to work into account when making decisions about treatments (Total)

Mostly work. So if I have to go in hospital trying to work that around to go into work and then, if I'm doing a treatment that's just like normal medications and whatnot, that would be working that around work as well, that's pretty much what I work it around.
Participant 013_2023AUORC

So when I'm employed, I feel psychologically, I feel a lot better about myself because I couldn't get a job before the pandemic. I couldn't get a job, numerous job, so I couldn't get a job. And then the pandemic happened and suddenly oh, you know, much more attractive of proposition for employment and just crazy. So now I'm employed, I feel much better about, you know, making contribution, paying tax, paying my way. Yeah. So if I'd only do a drug trial as long as it didn't compromise my health, my, you know, my mobility and my financial security…
Participant 002_2023AUDPA

I guess if treatment is going to affect my general life, like work like, you know, the doxazosin in that I was too sick to eat, or surgery, like how long will it take to recover? Time of work, that kind of thing.
Participant 006_2023AUDSK

Participant describes taking their own research into account when making decisions about treatments (Total)

I think about what would, what will happen if I do take it, what would happen if I didn't take it, so I can assess that comparison. I think about what the side effects would be. I think about where I ask about what the interactions would be with other medications that I'm on. I think about…How I take it. So there was one medication that was suggested to me at one point that I would have to self inject into my stomach and it was kind of off putting. So yeah. And I also use the squirt and the Facebook quote to get some idea of other people's reactions to it or how they've found that as well if I have the time to go away and do some research. Yeah, look into what other people's experiences have been.
Participant 009_2023AUDIS

Yeah. So in terms of making the decisions, I guess the efficacy and I guess I do some research in terms of…You know, not very academic research I must say, but I will look at any research papers that you know just through a search through a search engine and what their results were and also you know the period of time that it was done. Like, was it done last couple of years or was it done 10 years ago? It was ten years ago. I would ignore it and also look at different sites and talk to my GP. Trust my GP enormously and, you know, really talking to professionals.
Participant 010_2023AUDIS

Well, I take on board what he says because I have no option. I also have done a lot of reading on reputable websites on the Internet about seeing medication that he suggests. My observation is that there is not a lot of leeway with what I've got. There is also a lot….fairly large question mark over surgery anyway, so but I've had it done. I'm here. So great.
Participant 003_2023AUDNS

Table 4.5 Considerations when making decisions

Considerations about treatment	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=352	%	n=352	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=247	%	n=105	%	n=252	%	n=98	%
Ability to follow treatment (including accessibility) (Multiple)	31	8.81	5	7.46	5	6.17	7	15.56	1	3.23	9	9.47	4	12.90	21	8.50	10	9.52	22	8.73	9	9.18
Ability to follow treatment (including accessibility) (Single)	6	1.70	2	2.99	0	0.00	0	0.00	0	0.00	1	1.05	3	9.68	5	2.02	1	0.95	2	0.79	4	4.08
Ability to follow treatments (including accessibility) (Total)	37	10.51	7	10.45	5	6.17	7	15.56	1	3.23	10	10.53	7	22.58	26	10.53	11	10.48	24	9.52	13	13.27
Ability to work (Multiple)	13	3.69	3	4.48	0	0.00	3	6.67	4	12.90	2	2.11	1	3.23	8	3.24	5	4.76	9	3.57	4	4.08
Ability to work (Single)	3	0.85	0	0.00	1	1.23	0	0.00	0	0.00	0	0.00	2	6.45	2	0.81	1	0.95	2	0.79	1	1.02
Ability to work (Total)	16	4.55	3	4.48	1	1.23	3	6.67	4	12.90	2	2.11	3	9.68	10	4.05	6	5.71	11	4.37	5	5.10
Advice of their clinician (Multiple)	40	11.36	11	16.42	9	11.11	8	17.78	1	3.23	8	8.42	3	9.68	25	10.12	15	14.29	26	10.32	14	14.29
Advice of their clinician (Single)	52	14.77	20	29.85	11	13.58	7	15.56	1	3.23	12	12.63	1	3.23	28	11.34	24	22.86	36	14.29	16	16.33
Advice of their clinician (Total)	92	26.14	31	46.27	20	24.69	15	33.33	2	6.45	20	21.05	4	12.90	53	21.46	39	37.14	62	24.60	30	30.61
Amount of time needed/travel times (multiple)	19	5.40	8	11.94	1	1.23	1	2.22	3	9.68	3	3.16	3	9.68	8	3.24	11	10.48	11	4.37	8	8.16
Amount of time needed/travel times (single)	4	1.14	2	2.99	0	0.00	0	0.00	0	0.00	1	1.05	1	3.23	1	0.40	3	2.86	3	1.19	1	1.02
Amount of time needed/travel times (Total)	23	6.53	10	14.93	1	1.23	1	2.22	3	9.68	4	4.21	4	12.90	9	3.64	14	13.33	14	5.56	9	9.18
Cost (Multiple)	61	17.33	14	20.90	12	14.81	7	15.56	12	38.71	9	9.47	7	22.58	44	17.81	17	16.19	47	18.65	13	13.27
Cost (Single)	13	3.69	1	1.49	1	1.23	0	0.00	1	3.23	8	8.42	2	6.45	11	4.45	2	1.90	9	3.57	3	3.06
Cost (total)	74	21.02	15	22.39	13	16.05	7	15.56	13	41.94	17	17.89	9	29.03	55	22.27	19	18.10	56	22.22	16	16.33
Efficacy (Multiple)	110	31.25	26	38.81	23	28.40	16	35.56	8	25.81	26	27.37	11	35.48	73	29.55	37	35.24	72	28.57	37	37.76
Efficacy (Single)	26	7.39	3	4.48	9	11.11	0	0.00	2	6.45	12	12.63	0	0.00	22	8.91	4	3.81	21	8.33	5	5.10
Efficacy (Total)	136	38.64	29	43.28	32	39.51	16	35.56	10	32.26	38	40.00	11	35.48	95	38.46	41	39.05	93	36.90	42	42.86
Impact on their family or dependents (Multiple)	23	6.53	5	7.46	4	4.94	0	0.00	2	6.45	10	10.53	2	6.45	13	5.26	10	9.52	15	5.95	8	8.16
Impact on their family or dependents (Single)	9	2.56	0	0.00	3	3.70	0	0.00	0	0.00	6	6.32	0	0.00	7	2.83	2	1.90	8	3.17	1	1.02
Impact on their familiy or dependents (Total)	32	9.09	5	7.46	7	8.64	0	0.00	2	6.45	16	16.84	2	6.45	20	8.10	12	11.43	23	9.13	9	9.18
Own research (Multiple)	14	3.98	2	2.99	3	3.70	1	2.22	3	9.68	1	1.05	4	12.90	11	4.45	3	2.86	10	3.97	4	4.08
Own research (Single)	6	1.70	0	0.00	1	1.23	1	2.22	0	0.00	3	3.16	1	3.23	5	2.02	1	0.95	5	1.98	1	1.02
Own research (Total)	20	5.68	2	2.99	4	4.94	2	4.44	3	9.68	4	4.21	5	16.13	16	6.48	4	3.81	15	5.95	5	5.10
Quality of life (Multiple)	49	13.92	11	16.42	10	12.35	0	0.00	5	16.13	15	15.79	8	25.81	30	12.15	19	18.10	30	11.90	19	19.39
Quality of life (Single)	10	2.84	0	0.00	4	4.94	0	0.00	2	6.45	4	4.21	0	0.00	8	3.24	2	1.90	10	3.97	0	0.00
Quality of life (Total)	59	16.76	11	16.42	14	17.28	0	0.00	7	22.58	19	20.00	8	25.81	38	15.38	21	20.00	40	15.87	19	19.39
Side effects (Multiple)	133	37.78	25	37.31	33	40.74	17	37.78	12	38.71	32	33.68	14	45.16	88	35.63	45	42.86	94	37.30	38	38.78
Side effects (Single)	30	8.52	1	1.49	13	16.05	4	8.89	3	9.68	8	8.42	1	3.23	28	11.34	2	1.90	27	10.71	3	3.06
Side effects (Total)	163	46.31	26	38.81	46	56.79	21	46.67	15	48.39	40	42.11	15	48.39	116	46.96	47	44.76	121	48.02	41	41.84

Considerations about treatment	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=69	%	n=116	%	n=108	%	n=59	%	n=172	%	n=172	%	n=100	%	n=252	%	n=176	%	n=176	%
Ability to follow treatment (including accessibility) (Multiple)	31	8.81	1	1.45	3	2.59	1	0.93	1	1.69	1	0.58	5	2.91	1	1.25	5	1.98	2	1.14	4	2.27
Ability to follow treatment (including accessibility) (Single)	6	1.70	6	8.70	14	12.07	11	10.19	6	10.17	19	11.05	17	9.88	9	11.25	28	11.11	18	10.23	19	10.80
Ability to follow treatments (including accessibility) (Total)	37	10.51	3	4.35	5	4.31	4	3.70	1	1.69	4	2.33	9	5.23	5	6.25	8	3.17	7	3.98	6	3.41
Ability to work (Multiple)	13	3.69	1	1.45	1	0.86	0	0.00	1	1.69	2	1.16	1	0.58	0	0.00	3	1.19	0	0.00	3	1.70
Ability to work (Single)	3	0.85	4	5.80	6	5.17	4	3.70	2	3.39	6	3.49	10	5.81	5	6.25	11	4.37	7	3.98	9	5.11
Ability to work (Total)	16	4.55	8	11.59	13	11.21	9	8.33	10	16.95	18	10.47	22	12.79	9	11.25	31	12.30	21	11.93	19	10.80
Advice of their clinician (Multiple)	40	11.36	15	21.74	18	15.52	11	10.19	8	13.56	25	14.53	26	15.12	12	15.00	40	15.87	26	14.77	26	14.77
Advice of their clinician (Single)	52	14.77	23	33.33	31	26.72	20	18.52	18	30.51	43	25.00	48	27.91	21	26.25	71	28.17	47	26.70	45	25.57
Advice of their clinician (Total)	92	26.14	9	13.04	4	3.45	3	2.78	3	5.08	10	5.81	9	5.23	4	5.00	15	5.95	14	7.95	5	2.84
Amount of time needed/travel times (multiple)	19	5.40	3	4.35	1	0.86	0	0.00	0	0.00	1	0.58	3	1.74	2	2.50	2	0.79	2	1.14	2	1.14
Amount of time needed/travel times (single)	4	1.14	12	17.39	5	4.31	3	2.78	3	5.08	11	6.40	12	6.98	6	7.50	17	6.75	16	9.09	7	3.98
Amount of time needed/travel times (Total)	23	6.53	11	15.94	24	20.69	20	18.52	6	10.17	33	19.19	27	15.70	18	22.50	43	17.06	34	19.32	27	15.34
Cost (Multiple)	61	17.33	1	1.45	3	2.59	5	4.63	4	6.78	7	4.07	6	3.49	4	5.00	9	3.57	5	2.84	8	4.55
Cost (Single)	13	3.69	12	17.39	27	23.28	25	23.15	10	16.95	40	23.26	33	19.19	22	27.50	52	20.63	39	22.16	35	19.89
Cost (total)	74	21.02	25	36.23	38	32.76	30	27.78	17	28.81	48	27.91	62	36.05	32	40.00	78	30.95	45	25.57	65	36.93
Efficacy (Multiple)	110	31.25	3	4.35	6	5.17	10	9.26	7	11.86	11	6.40	15	8.72	8	10.00	18	7.14	11	6.25	15	8.52
Efficacy (Single)	26	7.39	28	40.58	44	37.93	40	37.04	24	40.68	59	34.30	77	44.77	40	50.00	96	38.10	56	31.82	80	45.45
Efficacy (Total)	136	38.64	6	8.70	8	6.90	5	4.63	4	6.78	10	5.81	12	6.98	6	7.50	17	6.75	13	7.39	10	5.68
Impact on their family or dependents (Multiple)	23	6.53	1	1.45	4	3.45	4	3.70	0	0.00	6	3.49	3	1.74	4	5.00	5	1.98	5	2.84	4	2.27
Impact on their family or dependents (Single)	9	2.56	7	10.14	12	10.34	9	8.33	4	6.78	16	9.30	15	8.72	10	12.50	22	8.73	18	10.23	14	7.95
Impact on their familiy or dependents (Total)	32	9.09	2	2.90	5	4.31	7	6.48	0	0.00	7	4.07	7	4.07	3	3.75	11	4.37	7	3.98	7	3.98
Own research (Multiple)	14	3.98	0	0.00	3	2.59	1	0.93	2	3.39	3	1.74	3	1.74	2	2.50	4	1.59	2	1.14	4	2.27
Own research (Single)	6	1.70	2	2.90	8	6.90	8	7.41	2	3.39	10	5.81	10	5.81	5	6.25	15	5.95	9	5.11	11	6.25
Own research (Total)	20	5.68	14	20.29	10	8.62	13	12.04	12	20.34	20	11.63	26	15.12	17	21.25	32	12.70	17	9.66	32	18.18
Quality of life (Multiple)	49	13.92	1	1.45	1	0.86	6	5.56	2	3.39	3	1.74	6	3.49	2	2.50	8	3.17	3	1.70	7	3.98
Quality of life (Single)	10	2.84	15	21.74	11	9.48	19	17.59	14	23.73	23	13.37	32	18.60	19	23.75	40	15.87	20	11.36	39	22.16
Quality of life (Total)	59	16.76	30	43.48	49	42.24	37	34.26	17	28.81	65	37.79	66	38.37	36	45.00	97	38.49	61	34.66	72	40.91
Side effects (Multiple)	133	37.78	2	2.90	14	12.07	11	10.19	3	5.08	18	10.47	12	6.98	12	15.00	18	7.14	18	10.23	12	6.82
Side effects (Single)	30	8.52	32	46.38	63	54.31	48	44.44	20	33.90	83	48.26	78	45.35	48	60.00	115	45.63	79	44.89	84	47.73
Side effects (Total)	163	46.31	2	2.90	4	3.45	1	0.93	7	11.86	12	6.98	2	1.16	4	5.00	10	3.97	8	4.55	6	3.41

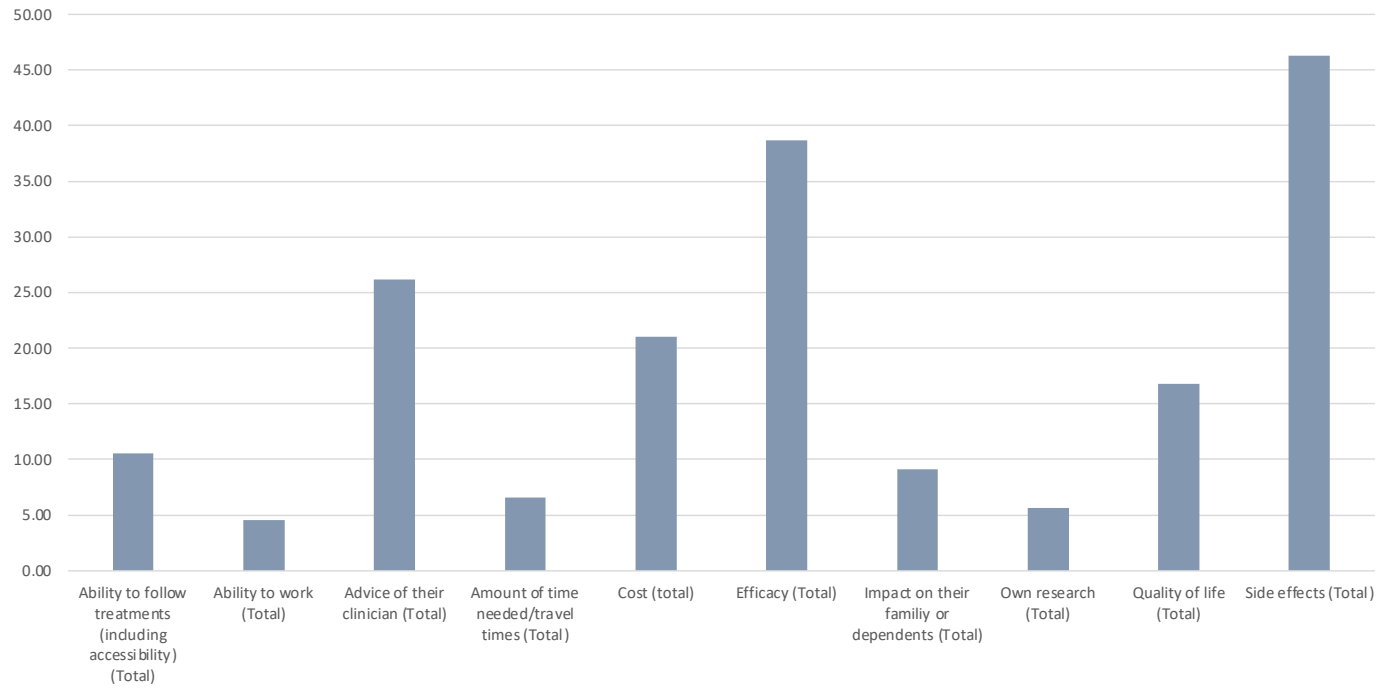


Figure 4.3 Considerations when making decisions

Table 4.6: Considerations when making decisions – subgroup variations

Discussions about treatment (Participation in discussions)	Reported less frequently	Reported more frequently
Ability to follow treatment (including accessibility) (Multiple)		
Ability to follow treatment (including accessibility) (Single)		
Ability to follow treatments (including accessibility) (Total)		Other rare condition
Ability to work (Multiple)		
Ability to work (Single)		
Ability to work (Total)		
Advice of their clinician (Multiple)		
Advice of their clinician (Single)	Diseases of the skin Other rare condition	Developmental anomalies
Advice of their clinician (Total)	Diseases of the skin Other rare condition	Developmental anomalies Family or carer
Amount of time needed/travel times (multiple)		
Amount of time needed/travel times (single)		
Amount of time needed/travel times (Total)		Aged under 18
Cost (Multiple)		Diseases of the skin
Cost (Single)		
Cost (total)		Diseases of the skin
Efficacy (Multiple)		
Efficacy (Single)		
Efficacy (Total)		Regional or remote
Impact on their family or dependents (Multiple)		
Impact on their family or dependents (Single)		
Impact on their familiy or dependents (Total)		
Own research (Multiple)		
Own research (Single)		
Own research (Total)		Other rare condition
Quality of life (Multiple)	Diseases of the nervous system	Other rare condition
Quality of life (Single)		
Quality of life (Total)	Diseases of the nervous system	
Side effects (Multiple)		
Side effects (Single)		
Side effects (Total)	Aged 65 plus	Diseases of the immune system Regional or remote

Decision-making over time

Participants were asked if the way they made decisions had changed over time. There were 201 participants (57.10%) that had changed the way they make decisions, and 110 participants (31.25%) had not changed the way they make decisions.

Where participants had changed the way they make decisions, the most common reasons were that they were more informed and/or more assertive (23.01%), more aware of their health, responsibilities and/or limitations (10.80%), and more cautious and considered (8.24 %). Other themes included more focused impact on quality of life (5.40%).

Where participants had not changed the way they make decisions, the most common reason was that they had always been informed/assertive (6.25%).

Changing over time as they are more informed and/or more assertive

No, very different. I'm a lot more informed now, and I understand my condition. I was struggling to cope with it at first. I didn't understand what was happening and I didn't know the outcome. I didn't know how it was going to end. Whereas now I'm a lot more informed. When the neurologist, we spoke about trying this new Botox, he made sure he had all the information there for me and then we decided that together. He put it forward. I think that I'm a lot more informed. Before I just went with whatever. If they said, "Go to this doctor and see this doctor," that's what I did. Whereas now, because I'm a lot more informed, I would never go to a neurologist that did not have the nerve conductivity equipment or just treated by just pure injection. I wouldn't do…I've got enough information for myself now to be able to make that decision.
Participant 006_2023AUDNS

I don't know. I think I'm a little more proactive in asking for what I want now rather than what I was in the beginning.
Participant 032_2023AUORC

Yes. Look, I just think I have got a lot more agency now. I just feel like now the ball is in my court a lot more than what it was. I suppose I'm more knowledgeable. I feel like when I'm discussing things with the doctors now it's more of an equal level after a team rather than just sitting there being passive. It's probably changed in that respect.
Participant 054_2023AUDPA

Changing over time as they are more aware of their health, responsibilities and/or limitations

Changed over time in the sense of from the what you… things like that, you know, as you grow older, you start to monitor your diet and your exercise, you know, and things like that, and actually try to do things to to keep yourself fit and healthy, I suppose when you're young, you think differently.
Participant 14_2023AUORC

No, no, no. I I'll look into things much more seriously now. I mean, I take responsibility for more on health. A lot more now. A lot more.
Participant 012_2023AUORC

Changing over time as they are more cautious and considered

Yeah, I think so. Yeah. I'm a little bit more cautious of some, what's in things, do I need to do it, that sort of thing. Yeah, yeah. And what…Yeah, how it's going to affect me? Yeah.
Participant 019_2023AUDIS

Yes, I'm sure it changed a lot because there's a lot more to take into consideration now. Yeah, I'm not, I'm not sure just say how my decision making's changed, but I'm sure it has changed just based on all the experiences. That we've had and the many more things that I have to take into consideration now when making most decisions. Yeah, I mean, even simple things like going on a holiday somewhere is much more complex than it used to be. So it involves, but I can't just make, make the decision that I would have made previously that, okay, I've got a holiday. There's a lot of other things that will be involved in trying to make it easier and make it work better... So yeah, I'd say a lot of things have changed with my decision making.
Participant 089_2023AUENM

Changing over time as they are more focused on how treatment impacts their family and dependents

I think my decision making process will have changed over time because it will have adapted to what is needed at that current point in time by the family and with the growing needs of the boys. So yeah, I feel it probably has changed, but it's still all vary based on PARTICIPANT.
Participant 036_2023AUDPA

Big time. It's changed. Yeah. I mean, initially it was my first child and it was a disease I've never heard of before in my life. So I could only do what the doctors suggested because I was terrified. But now I think about it when we're having a conversation and I asked them questions and I consider it for my family. If I have, you know, the opportunity, I'll try and do some research. If there's anything to research so that I can ask more informed questions, yeah. So I think it's changed definitely from just kind of doing what is suggested.
Participant 021_2023AUORC

Changing over time as their child gets older they take a greater part in decision making

I obviously didn't make any decisions until I was probably 15 or 16. Most of that was Mum and Dad. And then I was mum and dad and I had a pretty good relationship where it was they sort of thought that I understood what was going on at around 14 and 15 and 16. So then they sort of been started to include me in that. But then since about yeah 18, 19 it's up to me. But most of my decision making is pretty straightforward. It's do I basically keep staying with this doctor or do I go get an ECG or there..most of that's pretty self-explanatory and that doesn't require much decision making whatsoever. But I'm sure a time will come where I need to give it a little bit more critical thinking and then in that sense I'm a pretty pragmatic person and I like to think about things, probably overthink things sometimes and in that sense we'll we'll see what comes. But I imagine it'll be a pretty pragmatic and a an all inclusive sort of decision making process. It's not just me anymore it's my partner and mum and dad. All those people have a have a stake in my my health as well. So we'll, it'll be a sounding board sort of area.
Participant 030_2023AUORC

No, I would say it's changed. I just think, well, I mean now for example, she can actually weigh in and, you know, give us an indication of how she feels about a particular therapist or a particular doctor or, you know, whereas when she was a baby, we just were kind of head spinning, shooting in the dark, just trying to figure out what to do and, you know, kind of just feeling like, Oh my God, every decision is so critical. It's so life and death. Whereas now it often feels a little bit more like. There's time to just kind of figure it out right and constantly be in such a like frantic panic.
Participant 018_2023AUDPA

Changing over time as they are more accepting of their condition and choices available (however not by choice)

This might sound really bad. I'm probably not as optimistic about things as to having like.. I think people sell it to you that things are going to rapidly improve. And so perhaps I'm a little bit more not pessimistic, it's not the right word, but a bit more realistic I guess. Yeah, Okay, yeah.
Participant 020_2023AUDPA

No change in decision-making over time as they have always been informed/assertive

I approached it in the same way. And so I always make a decision, yeah, I make a decision by doing my research.
Participant 010_2023AUORC

I'll make decisions the same way, something that no… I think there was a couple of things that didn't agree with me, which I then spoke up and said no, don't you know, I don't think this...
Participant 005_2023AUDIS

Look, I still think I make decisions in the same way, which is to get lots of different views and to get lots of different information and decide whether I trust the professional, whether the professional has expertise in that area, whether there's other evidence that supports that, you know, idea of treatments.
Participant 010_2023AUDIS

Table 4.7: Decision-making over time

Decision-making over time	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=352	%	n=352	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=247	%	n=105	%	n=252	%	n=98	%
Change	201	57.10	35	52.24	50	61.73	29	64.44	19	59.38	42	44.21	26	83.87	143	57.89	58	55.24	148	58.73	51	52.04
No change	110	31.25	16	23.88	27	33.33	10	22.22	11	34.38	38	40.00	8	25.81	81	32.79	29	27.62	82	32.54	28	28.57

Decision-making over time	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=69	%	n=116	%	n=108	%	n=59	%	n=172	%	n=172	%	n=100	%	n=252	%	n=176	%	n=176	%
Change	201	57.10	44	63.77	64	55.17	66	61.11	27	45.76	92	53.49	107	62.21	59	73.75	142	56.35	99	56.25	102	57.95
No change	110	31.25	15	21.74	40	34.48	32	29.63	23	38.98	65	37.79	43	25.00	29	36.25	81	32.14	58	32.95	52	29.55

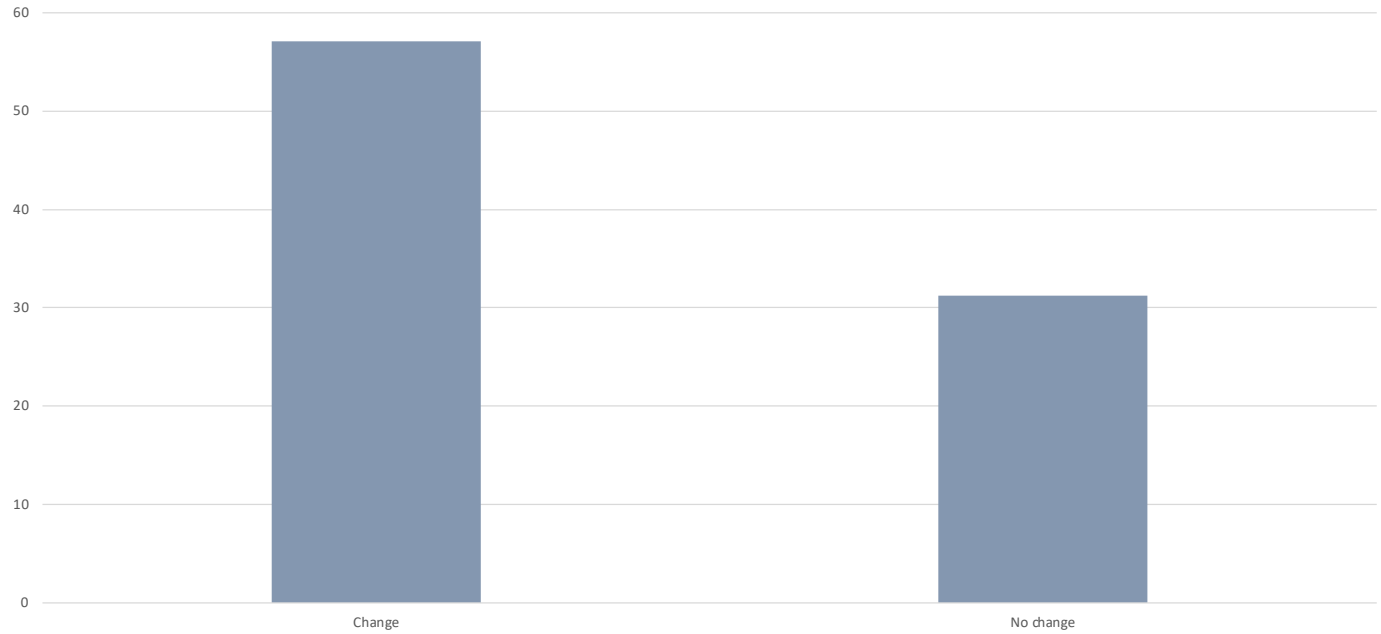


Figure 4.4: Decision-making over time

Table 4.8: Decision-making over time – subgroup variations

Theme	Reported less frequently	Reported more frequently
Change	Endocrine, nutritional or metabolic diseases Aged 65 plus	Other rare condition Regional or remote
No change		

Table 4.9: Decision-making over time (Reasons)

Decision-making over time (reasons)	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=352	%	n=352	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=247	%	n=105	%	n=252	%	n=98	%
Changing over time as they are more informed and/or more assertive	81	23.01	14	20.90	19	23.46	13	28.89	4	12.50	23	24.21	8	25.81	58	23.48	23	21.90	60	23.81	21	21.43
Changing over time as they are more aware of their health, responsibilities and/or limitations	38	10.80	5	7.46	5	6.17	7	15.56	7	21.88	5	5.26	9	29.03	26	10.53	12	11.43	22	8.73	14	14.29
Changing over time as they are more cautious and considered	29	8.24	4	5.97	16	19.75	2	4.44	2	6.25	3	3.16	2	6.45	22	8.91	7	6.67	28	11.11	1	1.02
Changing over time as they are more focused on quality of life or impact of side effects	19	5.40	3	4.48	3	3.70	2	4.44	2	6.25	9	9.47	0	0.00	13	5.26	6	5.71	13	5.16	6	6.12
No change in decision-making over time and there is no particular reason noted	54	15.34	5	7.46	8	9.88	8	17.78	8	25.00	19	20.00	6	19.35	42	17.00	12	11.43	38	15.08	16	16.33
No change in decision-making over time as they have always been informed/assertive	22	6.25	5	7.46	7	8.64	1	2.22	1	3.13	7	7.37	1	3.23	14	5.67	8	7.62	17	6.75	5	5.10

Decision-making over time (reasons)	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=69	%	n=116	%	n=108	%	n=59	%	n=172	%	n=172	%	n=100	%	n=252	%	n=176	%	n=176	%
Changing over time as they are more informed and/or more assertive	81	23.01	17	24.64	27	23.28	27	25.00	10	16.95	35	20.35	46	26.74	21	26.25	60	23.81	40	22.73	41	23.30
Changing over time as they are more aware of their health, responsibilities and/or limitations	38	10.80	9	13.04	11	9.48	11	10.19	7	11.86	19	11.05	18	10.47	15	18.75	23	9.13	23	13.07	15	8.52
Changing over time as they are more cautious and considered	29	8.24	6	8.70	12	10.34	8	7.41	3	5.08	17	9.88	12	6.98	6	7.50	23	9.13	15	8.52	14	7.95
Changing over time as they are more focused on quality of life or impact of side effects	19	5.40	2	2.90	6	5.17	6	5.56	5	8.47	10	5.81	8	4.65	6	7.50	13	5.16	6	3.41	13	7.39
No change in decision-making over time and there is no particular reason noted	54	15.34	8	11.59	23	19.83	15	13.89	8	13.56	34	19.77	20	11.63	13	16.25	41	16.27	28	15.91	26	14.77
No change in decision-making over time as they have always been informed/assertive	22	6.25	3	4.35	6	5.17	6	5.56	7	11.86	7	4.07	13	7.56	6	7.50	16	6.35	9	5.11	13	7.39

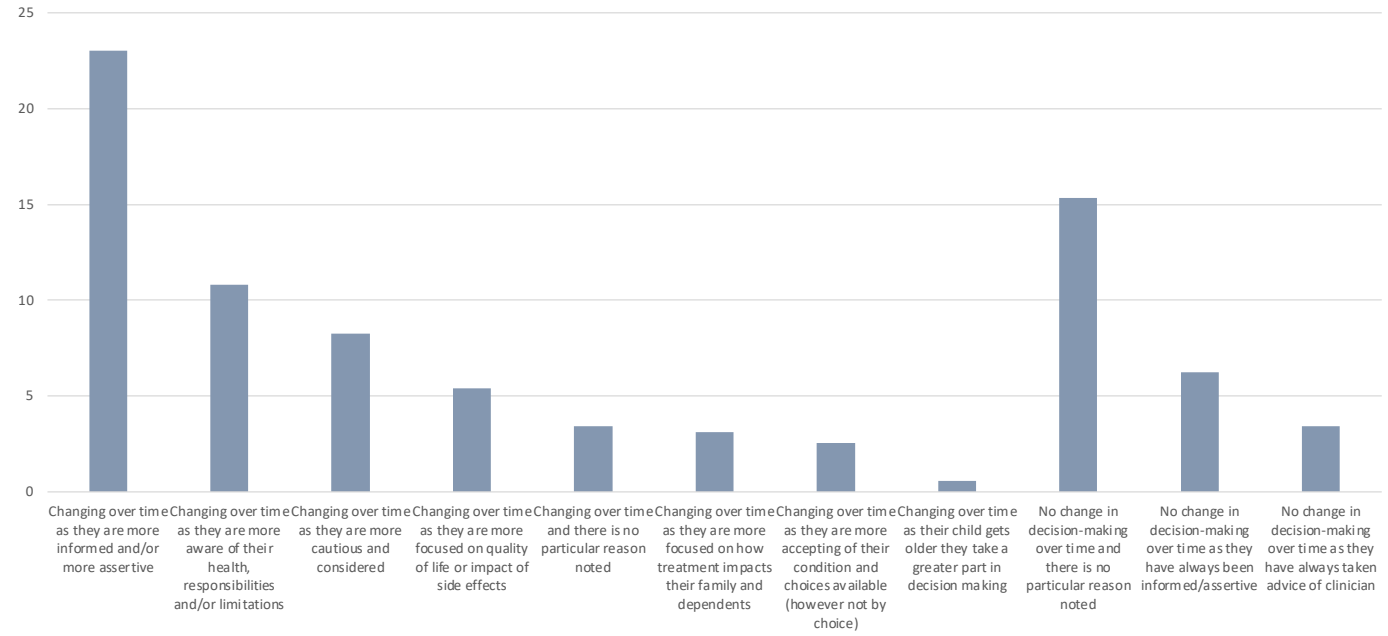


Figure 4.5: Decision-making over time (Reasons)

Table 4.10: Decision-making over time (Reasons) – subgroup variations

Decision-making over time (reasons)	Reported less frequently	Reported more frequently
Changing over time as they are more informed and/or more assertive	Diseases of the skin	
Changing over time as they are more aware of their health, responsibilities and/or limitations		Diseases of the skin Other rare condition
Changing over time as they are more cautious and considered		Diseases of the immune system
Changing over time as they are more focused on quality of life or impact of side effects	-	
No change in decision-making over time and there is no particular reason noted	-	
No change in decision-making over time as they have always been informed/assertive	-	

Personal goals of treatment or care

Participants were asked what their own personal goals of treatment or care were. The most common responses were to have quality of life/return to normality (22.56%), to maintain their condition or prevent worsening of their condition (19.55%), and have physical improvements in their condition (18.05 %). Other themes included the ability to live independently (13.53%) and wanting to minimise or avoid side effects (8.27%).

Participant describes wanting to improve their quality of life or return to normality

Yeah, it is. It's just to make his everyday life easier. Like we know even I haven't entered the physiotherapy side of things but, but you can tell when he hasn't done them. So our personal goal is to I guess get him to that, to be as healthy as he can be. So I can enjoy things you know, life as much as he can. That's pretty much our goal is just to make his life as easy as possible. And keep him health and his body as healthy as possible.
Participant 020_2023AUORC

Quality of life. Because I have things like central sleep apnea and digestive issues. I'm literally sleeping 16 hours a day and in extreme pain. And the more I do, the more I hurt. And yeah, it's it's like a catch 22 at the moment. So my quality of life is very poor and I'm very frustrated because I want to help myself, but no one's helping me. I, I would love to go back to work and I would love to. I'm happy to risk surgery. Whether it's going to have poor outcomes or not, it's my chance to have a better quality of life and I feel I should be able to sign a waiver to say I waive my right for something to go wrong. I, I accept that it can go wrong, but I want it done anyway and I'm not allowed to do that. But they could fix my back. I've got pinched nerves and bulging discs and bilaterally messed up back, and they could fix that. But they won't, because the risks too high. My neurologist won't do nerve biopsies because he's worried he'll disable me. But that's the only way I can get an answer.
Participant 016_2023AUDIS

Yeah, as as normal a life as possible.
Participant 015_2023AUDPA

Participant describes wanting to maintain their condition/prevent worsening of their condition

Oh look, I'd like to just get back to what I was before. For which started 2 1/2 years ago, at least close to it. I mean, the way I understand most of these things is they're not fixable, but they are manageable. And unfortunately, yeah, my, my management that I had unbeknownst to me already been doing got thrown into disarray and, and it's got on top of me a bit. But yeah, look my, my, I'm hoping I'm going to get back to almost normal again. Yeah, okay.
Participant 014_2023AUORC

I guess is really will become more independent. I guess it's the main goal and I guess stronger in terms of if there is going to be regression, we want to make her as strong as can be. So that's really the goal of therapy, yeah.
Participant 016_2023AUORC

Yeah, I guess I have a lot of side effects of treatment and I have to see other conditions as well and probably more than HS does alone. So I guess it's just management of everything more than anything. I think I'd like to be able to manage without it progressing too much worse and being able to manage whatever treatment I'm on.
Participant 019_2023AUDSK

Participants describe wanting to see physical improvements in their condition

I'd like to be more flexible and we did talk about with rheumatologist the fact that this condition causes you to have I call them elastic bands, okay, my rubber bands, they stretch out but they but they spring back and I I want something that will help me stretch them and keep them that way rather than this constant daily battle exercise that I have of going out and doing things and then the next day, I call it climbing Mount Everest, I wake up in the morning, I have to climb Mount Everest, and then tomorrow when I wake up, I have to climb it all over again. So that's that's what I want. I want that strength is not an issue. I can, you know, do weights and that sort of thing. It's that flexibility, tightness issue that bothers me.
Participant 003_2023AUDIS

Well, I my, my emphasis is to have less attacks. I know I think eventually I'm going to have to move to a warmer climate because I know that like I'm loving the, I love the hot weather. And I feel my body heals in the warmth. I have less you know I my fingertips don't ulcerate as much obviously less Raunaud's attacks and you… if with the less Raunaud's attacks I don't get the ulcers. So I just want to yeah I just want to have my goal is to yeah have less Raunaud's attacks because they're that's what I'm really struggling with.
Participant 015_2023AUDIS

Yes. So for me, I guess as a parent, my goal is just to allow my child to be a child as much as possible, so that's managing their condition the best way that I know how, with the information that I've got and the resources that I've got. But also, you know, focusing on the, I guess, the things that my son can do, I do have him, you know, attends different therapies to help improve his functioning, like his physical functioning or his gross motor. But I don't put a lot of pressure on him or on myself to achieve a certain goal because, you know, he's an individual. So I just want him to do the best that he can. But at the same time, you know, he's a kid, he needs to be a child. So I don't really know if that answers your question, but I try not to get too caught up in the shoulds or musts or if.
Participant 021_2023AUORC

Participant describes wanting to live independently

My goals are for her to be independent, like learn independent life skills that will help her when she is older and if no one is around, which won't be the case for a long time but I still would like her to think that she could have a job in a normal life and her own home if she wanted it. That's the most important thing to me.
Participant 013_2023AUDPA

PARTICIPANT: Everything really OK could be able to be, you know, obviously function you know society and, and making sure that he's, you know, I, like he's learning to the best of his ability and getting the support he needs with that. Yeah, you know and, you know, then grow into an independent, you know, adult.
INTERVIWER: Yep. So, yeah. Yep. Excellent. So have you had that sort of discussion about those goals with your permission?
PARTICIPANT: Well, yeah, I guess so. On and off. But I mean, at the moment, because he's still young and obviously needs support, there's not a lot, you know, that can be done at the moment, you know, apart from, I mean, some psychological support just with behavior and stuff. But yeah, there's not a lot more that at the moment we can do.
Participant 014_2023AUDPA

I guess our our goals for her she doesn't have like other than occupational therapy and speech therapy, there's no other treatment involved in her care at the moment, but she's not on any medication or there's no other ongoing things. So I guess her main goals that we have for her, you know, is to make her as prepared and capable to deal with, you know, the rest of her life and and set her up to be as functioning adult as she possibly can be.
Participant 027_2023AUDPA

Participant describes wanting to be less reliant on medication and to avoid hospitalisation

I just don't want to be on medication for the rest of my life. I keep trying to come off it myself, but it's not the best idea.
Participant 078_2023AUDIS

PARTICIPANT: My goal was just to stay out of hospital more, I guess.
INTERVIEWER: Okay, have you had a discussion about this with your clinician?
PARTICIPANT: Yes. Well, I certainly want to go to the hospital. Yeah. And I wanted to try to start a family this year. I was speaking to them about that recently.
Participant 013_2023AUORC

Participant describes wanting to minimise or avoid side effects of treatment for their condition

My personal is more…obviously, to be well-informed, but the treatment is certainly helping me now in making sure that I can walk and be physically fit. That's a big one because I've got to keep myself active. The other one is obviously to stop some of these issues in my body from affecting me and particularly in the cold, of course. It doesn't actually have to be cold. It's actually the first day of summer today, and it's actually cold.[laughter]
Participant 001_2023AUDIS

Nice to get them. Like, honestly, just get the pain to stop.
Participant 004_2023AUDSK

I think my main goal is to figure out how to work with bandaging, because I'm allergic to pretty much all adhesives, regardless of whether they contain silicone or allergen friendly or whatever it may be.
Participant 014_2023AUDSK

Participant describes wanting improvements in communication and engagement on community or school

Yeah, look better have the ability to self determine what she wants to do with her life and to be able to communicate that to the people around her and be able to access the appropriate support as, and when she needs it. But you know, basically for her to feel part of society and loved and have a good and happy life, same as for my other daughter.
Participant 018_2023AUDPA

There are multiple avenues of support that is required across the whole lifespan. So we were seeing up to 12 specialists for him…sometimes two or three a week. They come to my home or I'd be going to see specialists. And so life was very, very busy. I had a husband, I had a kid and I also was working from home and 18, you know. So I'm getting him onto eating properly or being [unintelligible] or doing speech therapy. We couldn't do it all and so I probably sacrificed some of his ability to eat safely and speech therapy by putting more energy into learning sign language and having him, helping him to to learn a total communication approach to life. I don't regret that in one little bit. But what I do know now is that he probably will never have a very safe swallow, to ever eat fully anyway. So if we would have to have pushed down that road or 18 normally, like everybody else, he would have been in hospital more times than he had been in the first five years. That would be gradually reducing over those years from, you know, five times a year to three times a year. So it was the right choice to make for us. Yeah, but this is what families are faced with the pressures of of my child must speak, my child must speak. What else? It's something, something will be sacrificed to meet that fully, that goal fully. So anything else that we did for goals, I think that's probably enough as an example from for for me.
Participant 028_2023AUORC

At the moment it's working on trying to increase her ability with speech and talking and language and being able to do things for herself. So you get a bit more independent and confident with that.
Participant 010_2023AUDPA

Participant describes wanting to see mental or emotional health improvements in their condition

Currently, I'm going to do everything I possibly can to keep myself healthy, fit, and eat well since my goal is to survive and get through this. I have to remain positive. Otherwise, I'm going to be like a little-- my other friends that have it and I don't want to get depressed. I'm just going to fight, keep fighting.
Participant 007_2023AUDIS

So our personal goals would be that he feels fulfilled in employment, that he feels like a valued member of society and that he is contributing in some way, but also has good mental health and some stress and pressures taken off him so that he feels that he is secure in his own living arrangements. So you know basically it's going to be at home with us for at least the next 10 years and then you know sort of working towards that independent living and and what that will look like we don't really know all.
Participant 022_2023AUDPA

Yeah sure. So with obviously with the the varying, the varying ailments or symptoms that that comes along with 22Q deletion you there's specific things. So we've wiped out a bunch of them already that she does, isn't affected by, but the ones moving forward, she has a thyroid disease. So I want her to be able to be able to get access to people readily and easily that can help her manage that because what happens when we're gone, she's got to have some some way of managing that. And also her, her psychiatric medication, for instance. We need to find something and people and professionals that know how to prescribe medicine properly to people with intellectual disabilities and not just throw drugs at them that are going to make them drowsy the whole time just you know subdue their personalities and things like that. It it's a bit…and there's a lot of autism and ADHD in in 22 Q as well. So there's a lot of all that's just sort of Ritalin outcome and this but that, that has massive side effects and that can also affect her personal life in social skills and ability to communicate as well. So they, they're the things I I want to say. I want to say her be able to access in my head a clinic of some sort…whereas coordinated approach, where they look at all the different sides of it, medically, socially, psychologically that that would be my ultimate goal and geez, wouldn't it be lovely if we could all have that.
Participant 025_2023AUDPA

Participant describes wanting to returning to work

So now, I mean, even back then, it was about finding answers. That was a goal. Finding answers, finding treatment, finding support, finding a therapist. Because back then we didn't have therapy money, you know, we just had nothing. And finding bowel support, finding enemas, you know, like finding help with all this. There's no continence nurses back then, but now his main goals are to be safe. Because he's not safe unless he's supported well to be healthy, and he's not healthy ever. So we aim to keep him to the best health as possible and to be have a healthy. So it's basically safe, healthy and happy and to have a meaningful day. So we've got four goals, but there's some other goals in NDIS land as well. You know about that, building his functional capacity to the best of his abilities. To ensure he has a meaningful day through social participation, civic and community access and economic participation. And he doesn't have a job, but I've certainly set up something for him with a ABN.
Participant 006_2023AUDPA

For me, going back to work is so that we could afford to do fun things again. So I love traveling. So yeah, my... I suppose my end goal is to get my life back, and in order to do that financially, I need to go back to work I suppose is is the driving thing about wanting to go to work, because hey, who, who wouldn't like to stay home every day and be well and enjoy it at the same time. So my big goal is to get my life back and be healthy.
Participant 031_2023AUORC

Table 4.11: Personal goals of treatment or care

Personal goals of treatment or care	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=266	%	n=67	%	n=81	%	n=45	%	n=32	%	n=9	%	n=32	%	n=176	%	n=90	%	n=200	%	n=64	%
Quality of life/return to normality	60	22.56	11	16.42	23	28.40	4	8.89	11	34.38	2	22.22	9	28.13	44	25.00	16	17.78	47	23.50	13	20.31
Maintain their condition/prevent worsening of their condition	52	19.55	2	2.99	17	20.99	11	24.44	10	31.25	1	11.11	11	34.38	44	25.00	8	8.89	40	20.00	11	17.19
Physical improvements in their condition	48	18.05	10	14.93	12	14.81	11	24.44	8	25.00	0	0.00	7	21.88	33	18.75	15	16.67	36	18.00	11	17.19
Live independently	36	13.53	16	23.88	10	12.35	6	13.33	0	0.00	2	22.22	2	6.25	17	9.66	19	21.11	31	15.50	5	7.81
Minimise or avoid side effects	22	8.27	1	1.49	7	8.64	6	13.33	5	15.63	0	0.00	3	9.38	21	11.93	1	1.11	20	10.00	2	3.13

Personal goals of treatment or care	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=266	%	n=66	%	n=102	%	n=76	%	n=22	%	n=132	%	n=134	%	n=75	%	n=191	%	n=143	%	n=123	%
Quality of life/return to normality	60	22.56	14	21.21	17	16.67	24	31.58	5	22.73	31	23.48	29	21.64	23	30.67	37	19.37	41	28.67	19	15.45
Maintain their condition/prevent worsening of their condition	52	19.55	5	7.58	23	22.55	20	26.32	4	18.18	24	18.18	28	20.90	16	21.33	36	18.85	31	21.68	21	17.07
Physical improvements in their condition	48	18.05	12	18.18	10	9.80	14	18.42	12	54.55	24	18.18	24	17.91	12	16.00	36	18.85	29	20.28	19	15.45
Live independently	36	13.53	10	15.15	15	14.71	9	11.84	2	9.09	14	10.61	22	16.42	9	12.00	27	14.14	21	14.69	15	12.20
Minimise or avoid side effects	22	8.27	1	1.52	10	9.80	9	11.84	2	9.09	10	7.58	12	8.96	3	4.00	19	9.95	11	7.69	11	8.94

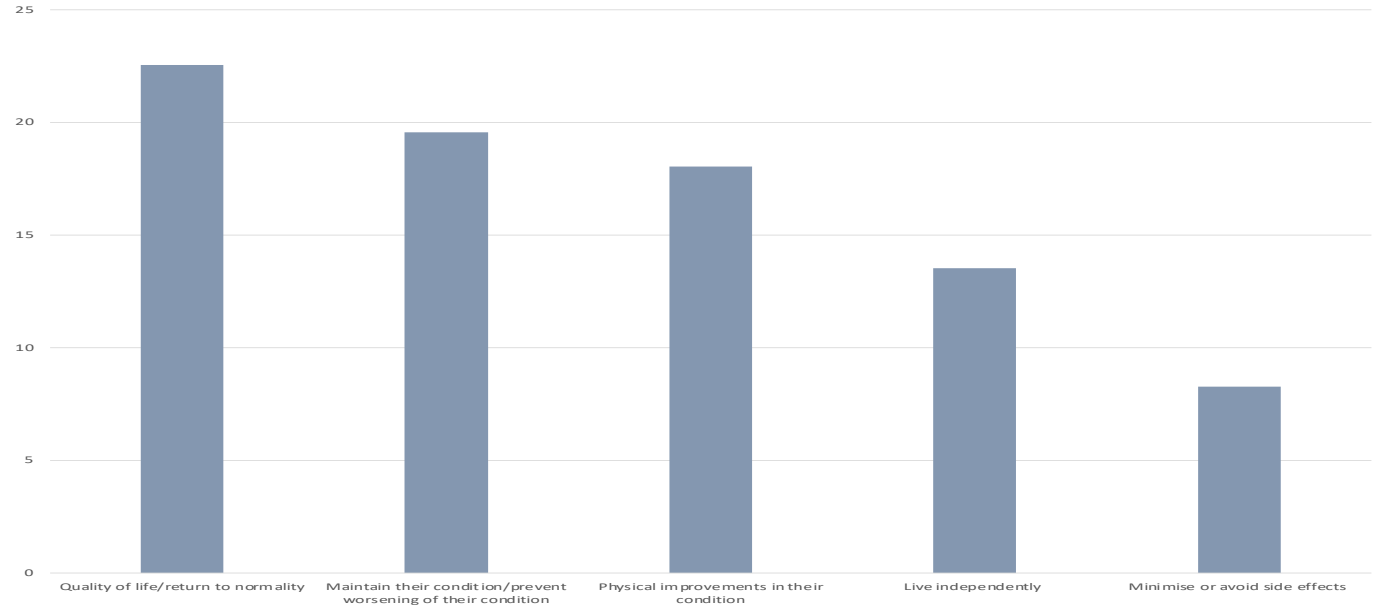


Figure 4.6: Personal goals of treatment or care

Table 4.12: Personal goals of treatment or care – subgroup variations

Personal goals of treatment or care	Reported less frequently	Reported more frequently
Quality of life/return to normality	Diseases of the nervous system	Diseases of the skin
Maintain their condition/prevent worsening of their condition	Developmental anomalies Family or carer Aged under 18	 Diseases of the skin Other rare condition
Physical improvements in their condition	Endocrine, nutritional or metabolic diseases	Aged 65 plus
Live independently	Diseases of the skin	Developmental anomalies
Minimise or avoid side effects		

Section 5

Treatment

Section 5: Experience of treatment

Respect shown

Participants were asked to think about how respectfully they were treated throughout their experience, this question was asked in the online questionnaire. Just under half of the participants indicated that they had been treated with respect throughout their experience (n=133, 41.43%), and 134 participants (41.74%) were treated with respect with the exception of one or two occasions. There were 54 participants (16.82%) felt they had not been treated respectfully.

Health care system

In the online questionnaire, participants were asked questions about the healthcare system they used, about private insurance and about whether they were treated as a public or private patient. The majority of participants had private health insurance (n=201, 64.63%). The majority of participants were not asked if they wanted to be treated as a public or private patient (n=157, 60.15%), however, they were asked if they had private health insurance (n=153, 58.62%). Throughout their treatment, there were 71 participants (23.05%) that were treated as a private patient, 156 participants (50.65%) were mostly treated as a public patient, and there were 68 participants (22.08%) that were equally treated as a private and public patient. Throughout their treatment, there were 42 participants (11.73%) that were treated mostly in the private hospital system, 228 participants (63.69%) were mostly treated in the public system, and there were 88 participants (24.58%) that were equally treated in the private and public systems.

Affordability of healthcare

Participants were asked a series of questions about affordability of healthcare in the online questionnaire.

The first question was about having to delay or cancer healthcare appointments because they were unable to afford them. Almost all the participants never or rarely had to delay or cancel appointments due to affordability (n = 259, 71.75%).

The next question was about the ability to fill prescriptions. Almost all of the participants never or rarely were unable to fill prescriptions (n=66, 18.28%).

The third question was about the affordability of basic essentials such as such as food, housing and power. There were 36 participants (9.97%) that never or rarely had trouble paying for essentials, and 13 participants (3.60%) that sometimes found it difficult, and 48 participants (13.30%) often or very often found it difficult to pay for basic essentials.

The final question was about paying for additional carers for themselves or for their family, there were 74 participants (23.79%) that paid for additional carers due to their condition.

Cost of condition

In the online questionnaire, participants estimated the amount they spend per month due to their condition, including doctors' fees, transport, carers, health insurance gaps and complementary therapies.

The most common amount was between $1001 or more (n=32, 8.74%), followed by between $101 to $250 (n=61, 16.67%). There were 41 participants (11.20%), that spent $501 to $1000 a month.

Burden of cost

As a follow up question, for participants that had monthly expenses due to their condition, participants were asked if the amount spent was a burden.

The amount spent was an extremely significant or moderately significant burden for 102 participants (33.44%), somewhat significant for 77 participants (25.25%), and slightly or not at all significant for 126 participants (41.31%).

Changes to employment status

Participants were asked, in the online questionnaire, if they had any changes to their employment status due to their condition. Participants were able to choose multiple changes to employment.

Work status for 63 participants (23.95%) had not changed since diagnosis, and 33 participants (12.55%) were retired or did not have a job. There were 79 participants (30.04%) had to quit their job, 78 participants (29.66%) reduced the number of hours they worked, and 28 participants (10.65%) that accessed their superannuation early. There were 49 participants (18.63%) that took leave from work without pay, and 48 participants (18.25%) that took leave from work with pay.

Participants were asked, in the online questionnaire, if they had any changes to the employment status of their care or partner due to their condition. Participants were able to choose multiple changes to employment.

There were 71 participants (24.40%), without a main partner or carer. Most commonly, participants had partners or carers that did not change their work status due to their condition (n=100, 34.36%). There were 43 participants (14.78%) whose partners reduced the numbers of hours they worked, and 19 partners, (6.53%) that quit their job. The partners of 26 participants (8.93%) took leave without pay, and there were 34 partners (11.68%) that took leave with pay.

Reduced income due to condition

More than half of the participants (n=217, 57.05%) indicated in the online questionnaire that they had a reduced family income due to their condition.

Estimated reduction monthly income

As a follow up question, participants were asked if their family or household income had reduced due to their condition. Where a dollar amount was given, it is listed below.

Most commonly, participants were not sure about the amount their monthly income was reduced by $2501 to 5000 (n=32, 10.74%), or reduced by between $1501 to 2500 per month (n=38, 12.75%).

Burden of reduced income

Participants were then asked if this reduced family or household income was a burden.

For 22 of these participants (16.30%), the burden of this reduced income was extremely or moderately significant, for 28 participants (20.74%) the burden was somewhat significant, and for 85 participants (62.96%) the burden was slightly or not all significant.

Lifestyle changes

Participants were asked about any lifestyle changes they had made since diagnosis, the quality of life from these changes, and how effective they found them.

Most participants used at made at least one lifestyle change (n=204, 67.77%), and on average made 1 changes (median=1.00, IQR=1.00).

The most common lifestyle change used was diet changes (n=150, 51.02%), followed by exercise (n=146, 59.84%), and reduce alcohol (n=56, 22.95%)

Complementary therapies

Participants were asked about any complementary therapies they used to manage their condition, the quality of life from these changes, and how effective they found them.

Most participants used at made at least one complementary therapy (n=216, 68.35%), and on average used 1 therapies (median=1.00, IQR=2.00).

The most common complementary therapy used was supplements (n=136, 46.10%), followed by mindfulness or relaxation (n=121, 45.83%), and massage therapy (n=80, 30.30%).

Clinical trials

In the online questionnaire, participants were asked if they had discussions with their doctor about clinical trials, and if they did, who initiated the discussion.

There was a total of 111 participants (35.81%) that had discussions about clinical trials, 32 participants (10.32%) had brought up the topic with their doctor, and the doctor of 79 participants (25.48%) brought up the topic. The majority of participants had not spoken to anyone about clinical trials (n=199, 64.19%).

As a follow up question, participants were asked if they had taken part in a clinical trial, and if they had not taken part if they were interested in taking part.

There were 37 participants (11.86%) that had taken part in a clinical trial, 155 participants (49.68%) that would like to take part in a clinical trial if there was a suitable one, and 120 participants, that have not participated in a clinical trial and do not want to (38.46%).

Description of mild side effects

In the structured interview, participants were asked how they would describe the term 'mild side effects'. The most common descriptions of mild side effects were described using a specific example (53.69%), and those that do not interfere with life (33.24%). Other themes included those that are resolved in a short amount of time (9.66%) and those that can be managed with self-medication or self-management (3.98 %).

Description of severe side effects

In the structured interview, participants were asked how they would describe the term 'severe side effects'. The most common description of severe side effects were described using a specific example (47.73%), and those that impact everyday life or ability to conduct activities of daily living (28.13%). Other themes included those that are life threatening or result in hospitalisation (8.52%), those that cause long-term damage to their body (7.67%).

When a specific side effect was described, the most common examples were aches and pain (17.33%), emotional and mental impact (7.39%), and nausea with vomiting (6.53%). Other themes included fatigues (5.11%), gastrointestinal distress (4.83%), impact on sleep (4.26%), vision problems (3.98%), and impact on sleep (4.55%).

Adherence to treatment

Participants were asked in the structured interview what influences their decision to continue with a treatment regime. The most common responses were adhering to treatment for a specific amount of time (38.35%), adhering to treatment according to the advice of their specialist or as long as prescribed (36.08%), and adhering to treatment as long as side effects are tolerable (24.43 %). Other themes included never giving up on any treatment (11.36%), adhering to treatment as long as treatment is working (7.10%).

What needs to change to feel like treatment is working

Participants were asked to describe what needs to change to feel like treatment is effective. The most common responses were needing to see a specific symptom reduction (26.70%), needing to see needing to see physical signs and symptoms disappear or reduce side effects (25.85%), a needing to see improvements in general wellbeing (quality of life) (14.49%), needing to see evidence of stable disease (14.20%), needing to see a return to day-to-day functionality (14.20%), and needing to see improvement in pain levels (12.50%).

What it would mean if treatment worked

As a follow up question, participants were asked what it would mean to them if the treatment worked in the way they described. The most common responses were that it would allow them to do everyday activities/return to normal life (29.44%) and allow them to engage more with social activities and family life (11.67%). Other themes included allow them to return to work (9.44%), allow them to do more exercise (11.28%), will have a positive impact on their mental health (7.89%), allow the

Respect shown

Participants were asked to think about how respectfully they were treated throughout their experience, this question was asked in the online questionnaire.

Just under half of the participants indicated that they had been treated with respect throughout their experience (n=133, 41.43%), and 134 participants (41.74%) were treated with respect with the exception of one or two occasions. There were 54 participants (16.82%) felt they had not been treated respectfully.

Table 5.1: Respect shown

Respect shown	Number (n=321)	Percent
Respect shown	133	41.43
Respect shown, with the exception of one or two occasions	134	41.74
Respect not shown	54	16.82

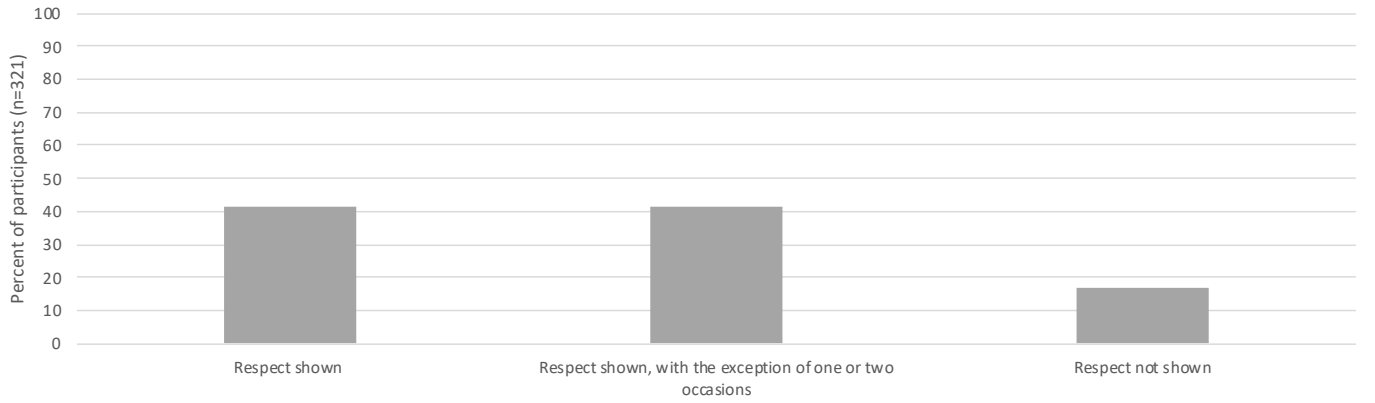


Figure 5.1: Respect shown

Health care system

In the online questionnaire, participants were asked questions about the healthcare system they used, about private insurance and about whether they were treated as a public or private patient.

The majority of participants had private health insurance (n=201, 64.63%). The majority of participants were not asked if they wanted to be treated as a public or private patient (n=157, 60.15%), however, they were asked if they had private health insurance (n=153, 58.62%).

Throughout their treatment, there were 71 participants (23.05%) that were treated as a private patient, 156 participants (50.65%) were mostly treated as a public patient, and there were 68 participants (22.08%) that were equally treated as a private and public patient.

Throughout their treatment, there were 42 participants (11.73%) that were treated mostly in the private hospital system, 228 participants (63.69%) were mostly treated in the public system, and there were 88 participants (24.58%) that were equally treated in the private and public systems.

Table 5.2: Health care system

Health care services	Response	Number	Percent
Private health insurance	No	110	35.37
	Yes	201	64.63
Asked whether you want to be treated as a public or private patient	No	157	60.15
	Yes	104	39.85
Asked whether you had private health insurance	No	108	41.38
	Yes	153	58.62
Throughout your treatment in hospital, have you most been treated as a public or a private patient	Equally as a public and private patient	68	22.08
	Private patient	71	23.05
	Public patient	156	50.65
	Not sure	13	4.22
Which hospital system have you primarily been treated in	Both public and private	88	24.58
	Private	42	11.73
	Public	228	63.69
	Not sure	0	0.00

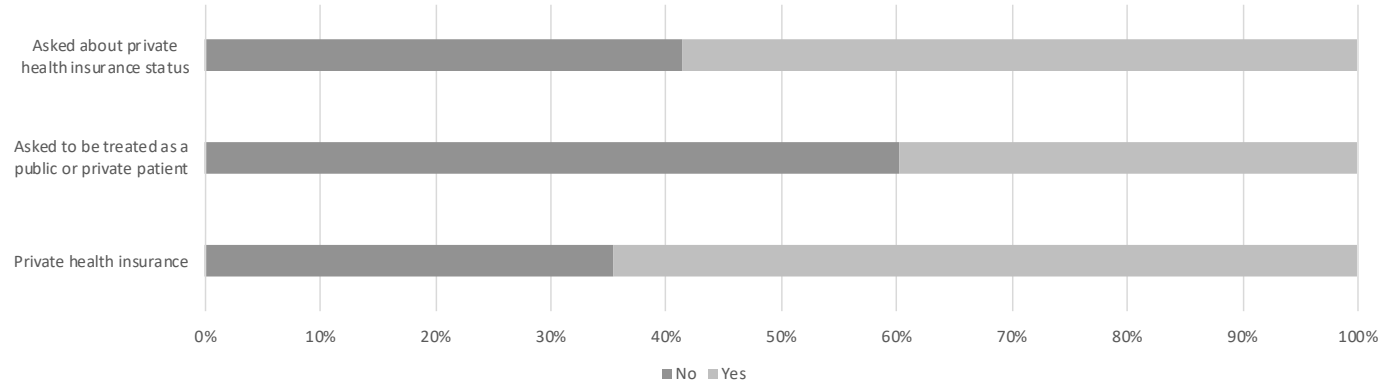


Figure 5.2: Health insurance

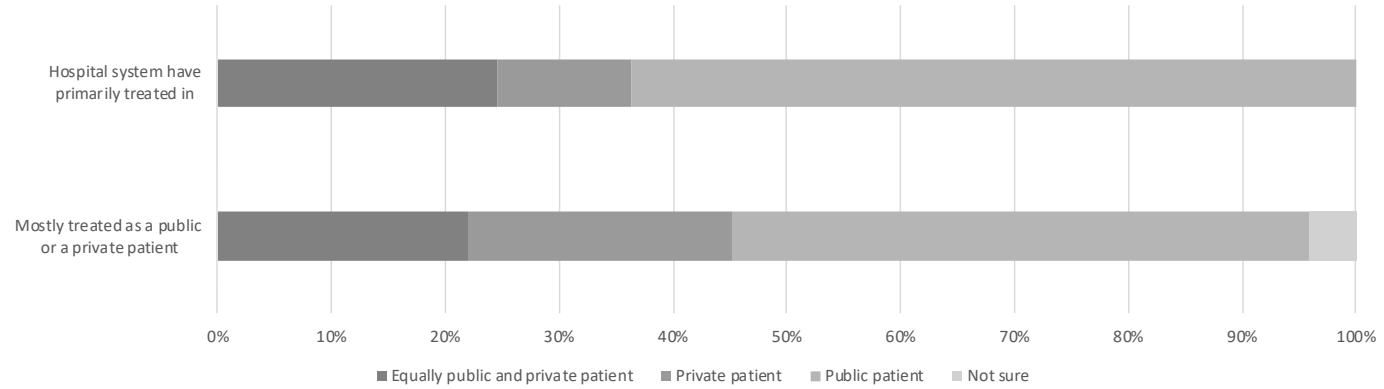


Figure 5.3: Hospital system

Affordability of healthcare

Participants were asked a series of questions about affordability of healthcare in the online questionnaire.

The first question was about having to delay or cancer healthcare appointments because they were unable to afford them. Almost all the participants never or rarely had to delay or cancel appointments due to affordability (n = 259, 71.75%).

The next question was about the ability to fill prescriptions. Almost all of the participants never or rarely were unable to fill prescriptions (n=66, 18.28%).

The third question was about the affordability of basic essentials such as such as food, housing and power. There were 36 participants (9.97%) that never or rarely had trouble paying for essentials, and 13 participants (3.60%) that sometimes found it difficult, and 48 participants (13.30%) often or very often found it difficult to pay for basic essentials.

The final question was about paying for additional carers for themselves or for their family, there were 74 participants (23.79%) that paid for additional carers due to their condition.

Table 5.3: Affordability of healthcare

Affordability of healthcare	Response	Number	Percent
Delay or cancel healthcare appointments due to affordability	Never	215	59.56
	Rarely	44	12.19
	Sometimes	66	18.28
	Often	25	6.93
	Very often	11	3.05
Did not fill prescriptions due to cost	Never	260	72.02
	Rarely	45	12.47
	Sometimes	43	11.91
	Often	10	2.77
	Very often	3	0.83
Difficult to pay for basic essentials	Never	172	47.65
	Rarely	52	14.40
	Sometimes	89	24.65
	Often	31	8.59
	Very often	17	4.71
Pay for additional carers for self or family	Yes	74	23.79
	No	237	76.21

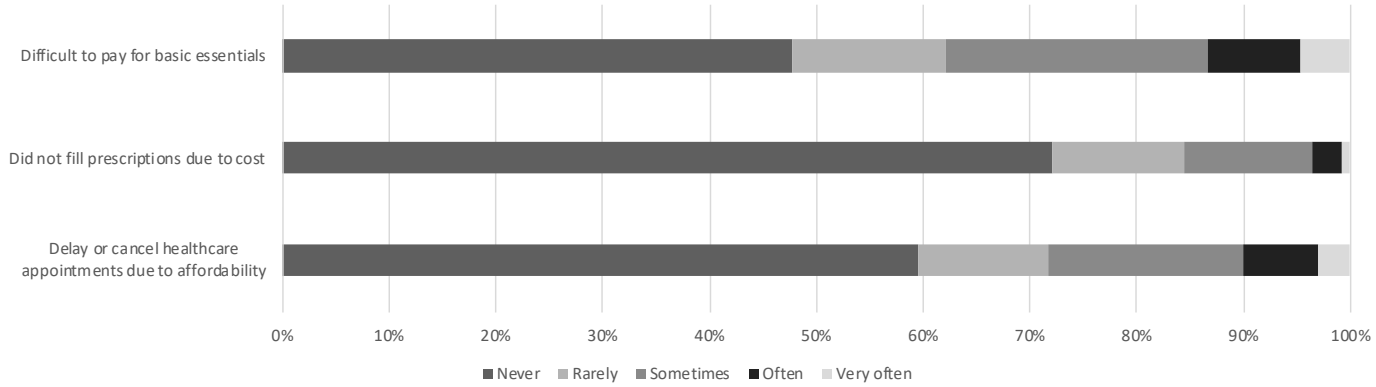


Figure 5.4: Affordability of healthcare

Cost of condition

In the online questionnaire, participants estimated the amount they spend per month due to their condition, including doctors' fees, transport, carers, health insurance gaps and complementary therapies. Where the response was given in a dollar amount, it is listed below.

The most common amount was between $1001 or more (n=32, 8.74%), followed by between $101 to $250 (n=61, 16.67%). There were 41 participants (11.20%), that spent $501 to $1000 a month.

Burden of cost

As a follow up question, for participants that had monthly expenses due to their condition, participants were asked if the amount spent was a burden.

The amount spent was an extremely significant or moderately significant burden for 102 participants (33.44%), somewhat significant for 77 participants (25.25%), and slightly or not at all significant for 126 participants (41.31%).

Table 5.4: Estimated monthly out of pocket expenses due to condition

Estimated monthly out of pocket expenses	Number (n=366)	Percent
$0	19	5.19
$100 or less	72	19.67
$1001 or more	32	8.74
$101 to $250	61	16.67
$251 to $500	79	21.58
$501 to $1000	41	11.20
Not sure of amount	62	16.94

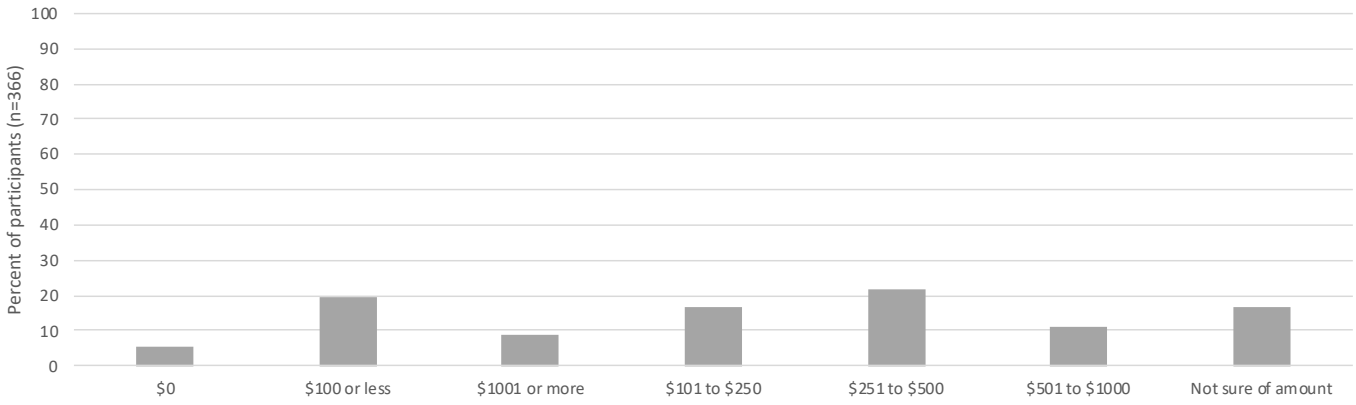


Figure 5.5: Estimated monthly out of pocket expenses due to condition

Table 5.5: Burden of out-of-pocket expenses due to condition

Burden of out of pocket expenses	Number (n=305)	Percent
Extremely significant	64	20.98
Moderately significant	38	12.46
Somewhat significant	77	25.25
Slightly significant	67	21.97
Not at all significant	59	19.34

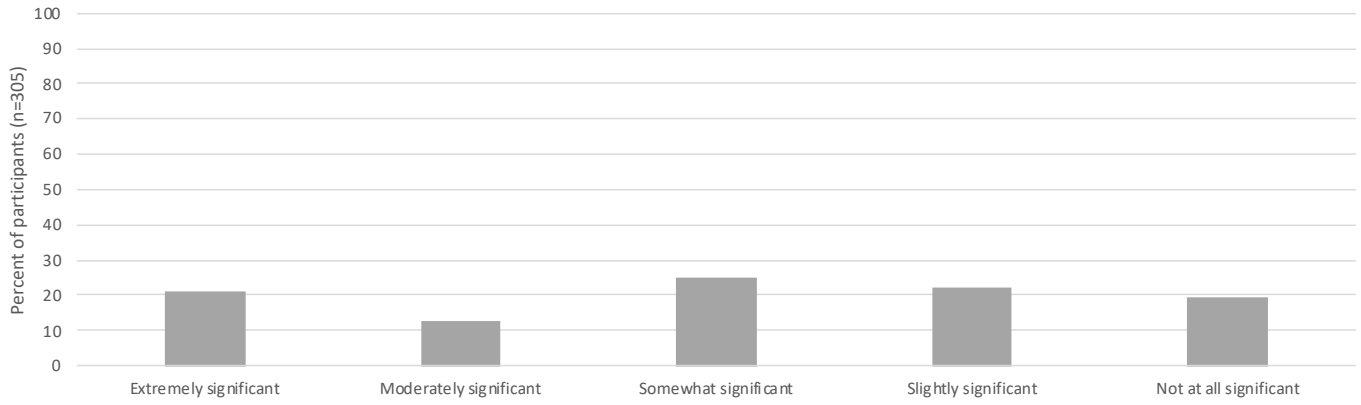


Figure 5.6: Burden of out-of-pocket expenses due to condition

Changes to employment status

Participants were asked, in the online questionnaire, if they had any changes to their employment status due to their condition. Participants were able to choose multiple changes to employment.

Work status for 63 participants (23.95%) had not changed since diagnosis, and 33 participants (12.55%) were retired or did not have a job. There were 79 participants (30.04%) had to quit their job, 78 participants (29.66%) reduced the number of hours they worked, and 28 participants (10.65%) that accessed their superannuation early. There were 49 participants (18.63%) that took leave from work without pay, and 48 participants (18.25%) that took leave from work with pay.

Changes to carer/partner employment status

Participants were asked, in the online questionnaire, if they had any changes to the employment status of their care or partner due to their condition. Participants were able to choose multiple changes to employment.

There were 71 participants (24.40%), without a main partner or carer. Most commonly, participants had partners or carers that did not change their work status due to their condition (n=100, 34.36%). There were 43 participants (14.78%) whose partners reduced the numbers of hours they worked, and 19 partners, (6.53%) that quit their job. The partners of 26 participants (8.93%) took leave without pay, and there were 34 partners (11.68%) that took leave with pay.

Table 5.6: Changes to employment status

Changes in work status due to condition	Number (n=263)	Percent
Work status has not changed	63	23.95
Retired or did not have a job	33	12.55
Had to quit job	79	30.04
Reduced number of hours worked	78	29.66
Leave from work without pay	49	18.63
Leave from work with pay	48	18.25
Accessed Superannuation early due to condition	28	10.65

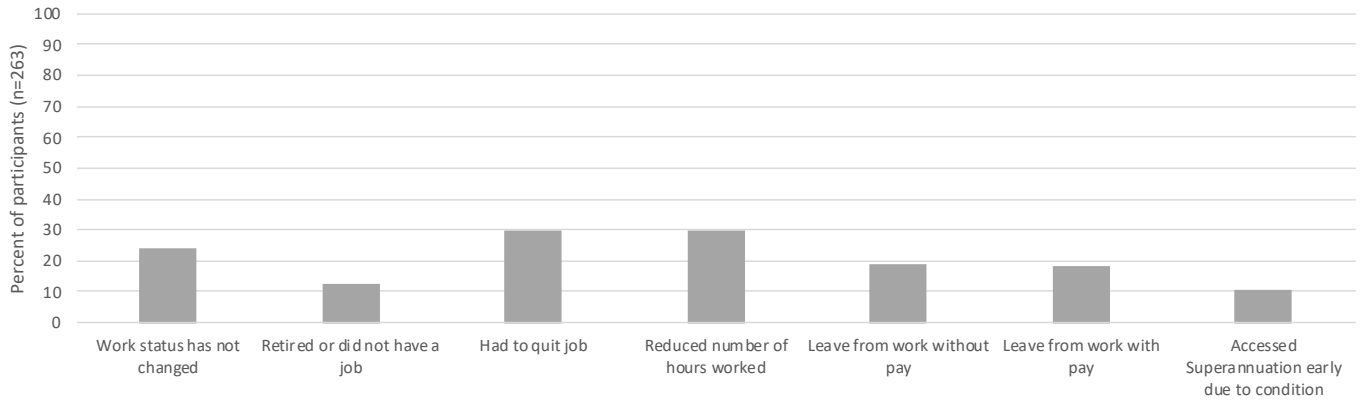


Figure 5.7: Changes to employment status

Table 5.7: Changes to care/partner employment status

Changes in partner or main carer work status due to condition	Number (n=291)	Percent
Does not have a partner/main carer	71	24.40
Work status has not changed	100	34.36
Retired or did not have a job	32	11.00
Had to quit job	19	6.53
Reduced number of hours worked	43	14.78
Leave from work without pay	26	8.93
Leave from work with pay	34	11.68

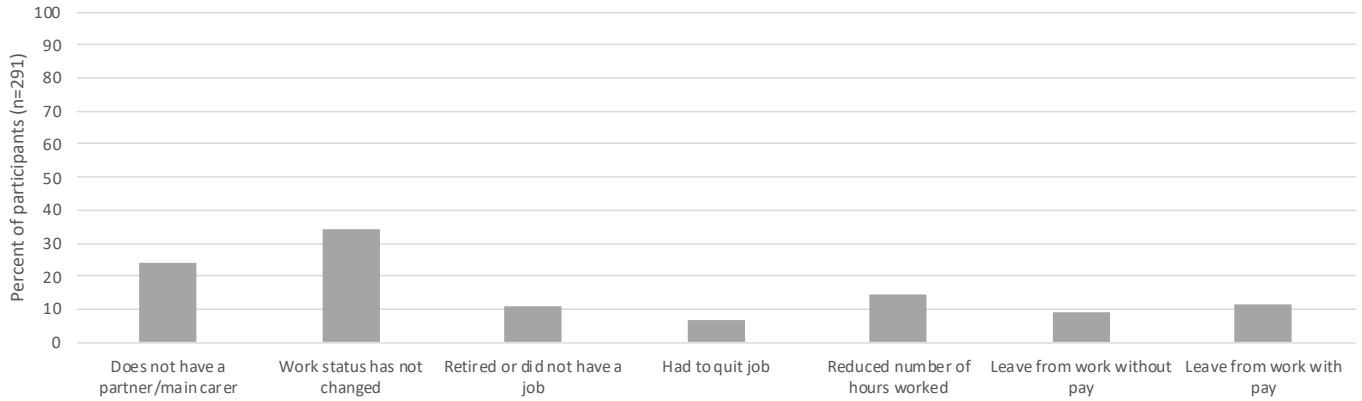


Figure 5.8: Changes to care/partner employment status

Reduced income due to condition

More than half of the participants (n=217, 57.05%) indicated in the online questionnaire that they had a reduced family income due to their condition.

Estimated reduction monthly income

As a follow up question, participants were asked if their family or household income had reduced due to their condition. Where a dollar amount was given, it is listed below.

Most commonly, participants were not sure about the amount their monthly income was reduced by $2501 to 5000 (n=32, 10.74%), or reduced by between $1501 to 2500 per month (n=38, 12.75%).

Burden of reduced income

Participants were then asked if this reduced family or household income was a burden.

For 22 of these participants (16.30%), the burden of this reduced income was extremely or moderately significant, for 28 participants (20.74%) the burden was somewhat significant, and for 85 participants (62.96%) the burden was slightly or not all significant.

Table 5.8: Estimated monthly loss of income

Estimated monthly loss of income	Number (n=298)	Percent
$0	128	42.95
$1501 to 2500	38	12.75
$2501 to 5000	32	10.74
$500 to 1500	29	9.73
More than $5000	15	5.03
Not sure/not specified	56	18.79

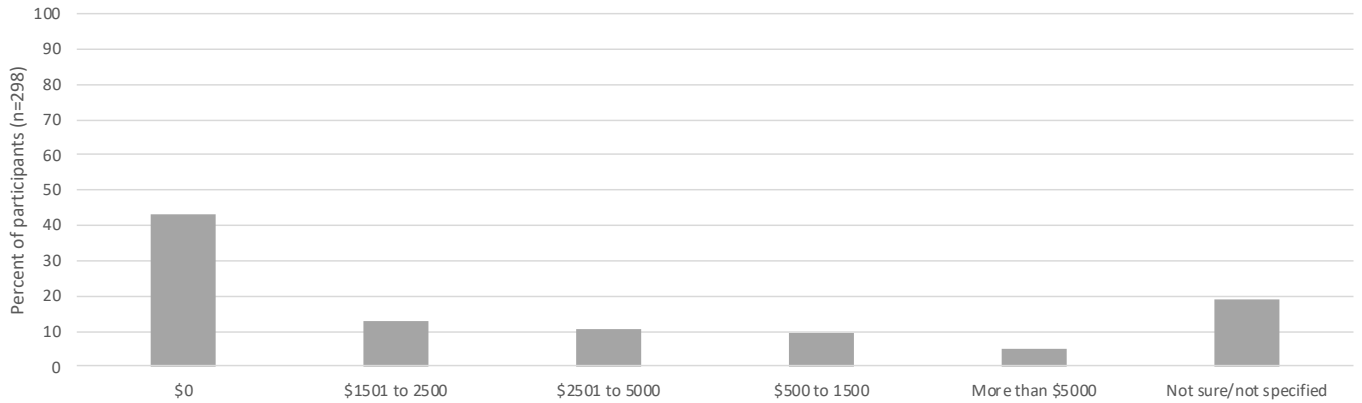


Figure 5.9: Estimated monthly loss of income

Table 5.9: Burden of reduced income

Burden of reduced income	Number (n=135)	Percent
Extremely significant	52	38.52
Moderately significant	33	24.44
Somewhat significant	28	20.74
Slightly significant	20	14.81
Not at all significant	2	1.48

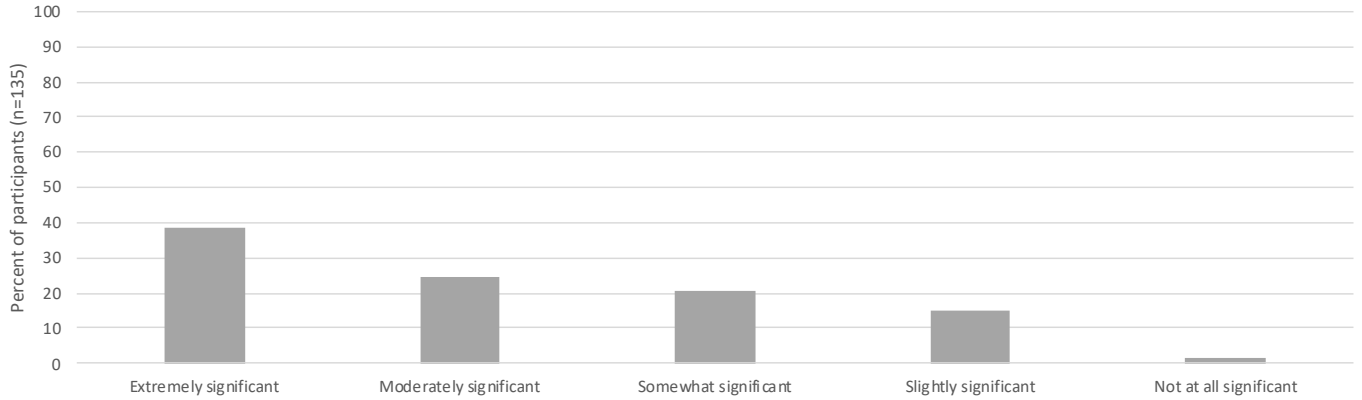


Figure 5.10: Burden of reduced income

Table 5.10: Allied health

Allied health	Number	Percent		Median quality of life	IQR	Median effectiveness	IQR
Physiotherapy (n=286)	135	135	47.20	4.00	4.00	2.00	1.50
Psychology (n=236)	92	92	38.98	2.50	3.00	2.00	2.00
Occupational therapy (n=236)	82	82	34.75	4.00	3.00	3.00	1.00
Dietary (n=217)	72	72	33.18	3.00	2.00	2.00	2.25
Speech therapy (n=286)	70	70	24.48	4.00	4.00	2.00	2.00
Podiatry (n=216)	67	67	31.02	4.00	4.00	1.50	1.50
Social work (n=236)	22	22	9.32	1.00	2.50	3.00	2.00

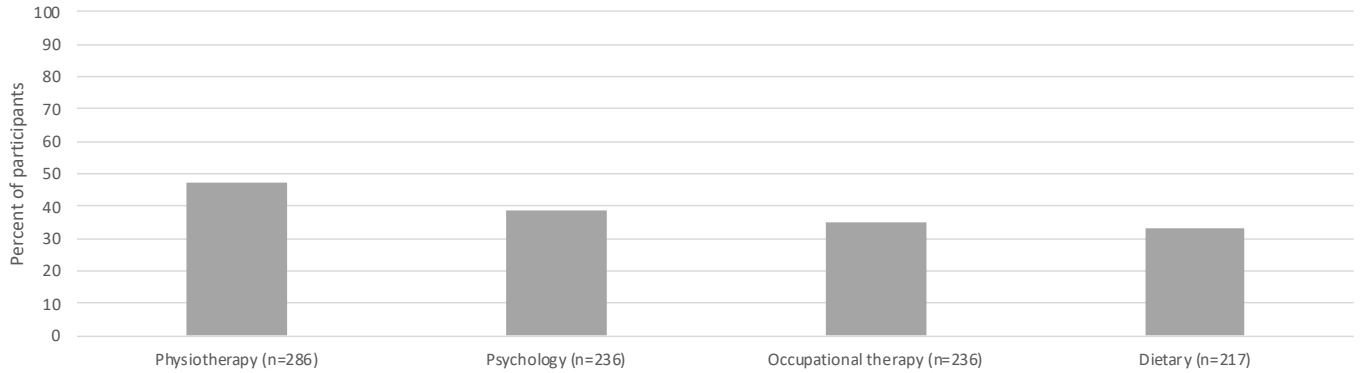


Figure 5.11: Allied health

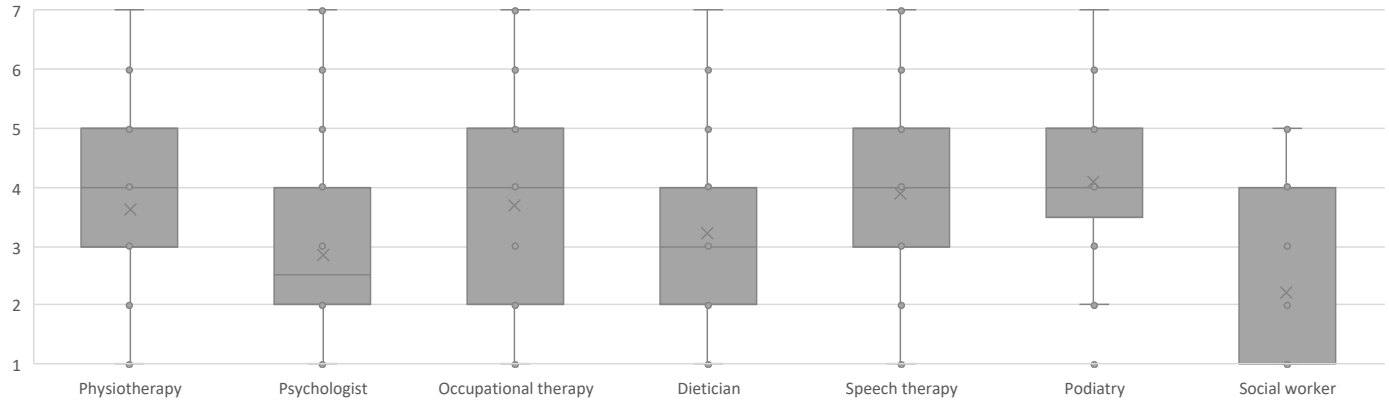


Figure 5.12: Quality of life from allied health

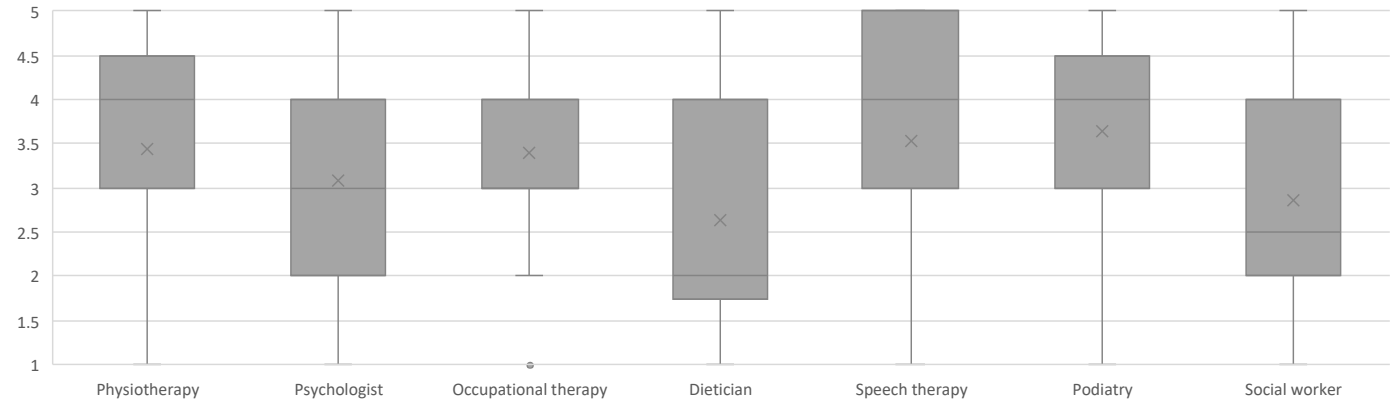


Figure 5.13: Effectiveness of allied health

Lifestyle changes

Participants were asked about any lifestyle changes they had made since diagnosis, the quality of life from these changes, and how effective they found them.

Most participants used at made at least one lifestyle change (n=204, 67.77%), and on average made 1 changes (median=1.00, IQR=1.00).

The most common lifestyle change used was diet changes (n=150, 51.02%), followed by exercise (n=146, 59.84%), and reduce alcohol (n=56, 22.95%).

On average, quality of life from diet changes was in the 'life was average' range (median=4.00, IQR=3.00), and was found to be ineffective (median=2.00, IQR=2.00).

On average, quality of life from exercise was in the 'life was average' range (median=4.00, IQR=3.00), and was found to be ineffective (median=2.00, IQR=2.00).

On average, quality of life from reducing alcohol was in the 'life was average' range (median=4.00, IQR=2.50), and was found to be ineffective (median=2.00, IQR=2.00).

On average, quality of life from quitting smoking was in the 'life was a little distressing' range (median=3.00, IQR=1.00), and was found to be ineffective (median=2.00, IQR=3.00).

Table 5.11: Lifestyle changes

Lifestyle changes	Number	Percent	Median quality of life	IQR	Median effectiveness	IQR
Diet changes (n=294)	150	51.02	4.00	3.00	2.00	2.00
Exercise (n=244)	146	59.84	4.00	3.00	2.00	2.00
Reduce alcohol (n=244)	56	22.95	4.00	2.50	2.00	2.00
Quit smoking (n=244)	19	7.79	3.00	1.00	2.00	3.00

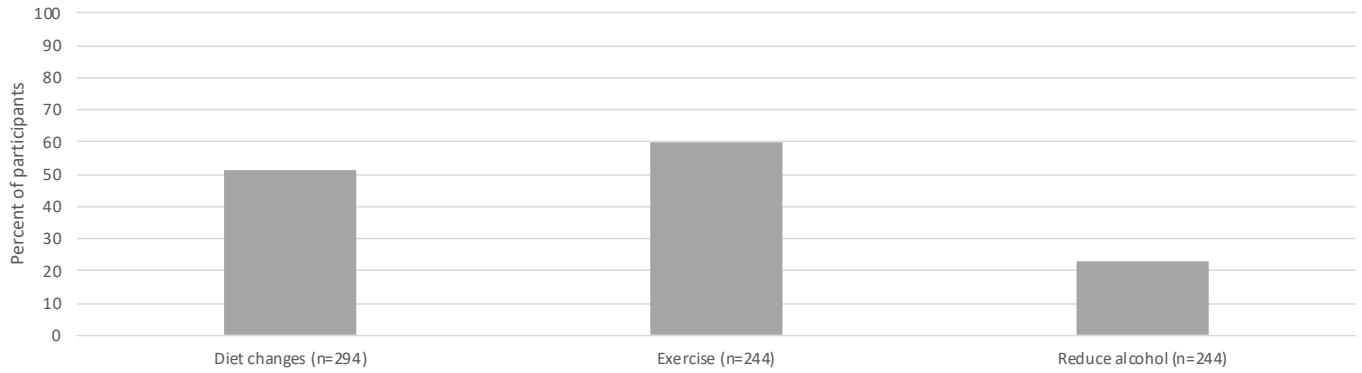


Figure 5.14: Lifestyle changes

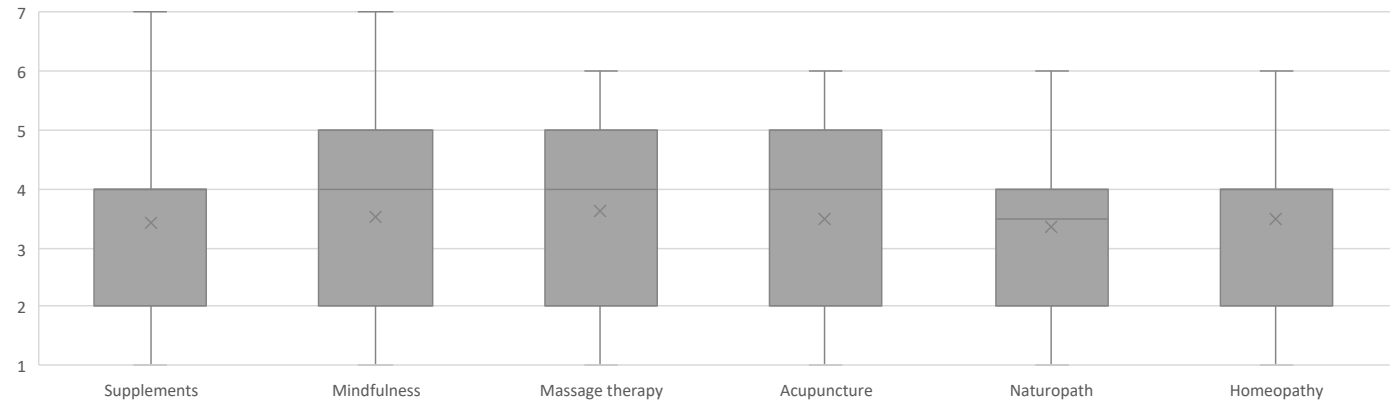


Figure 5.15: Quality of life from lifestyle changes

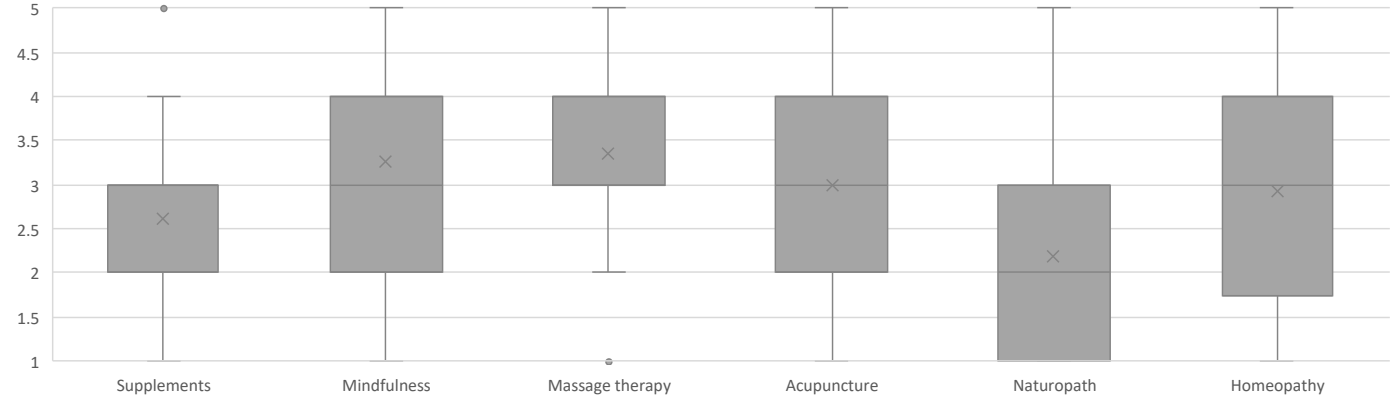


Figure 5.16: Effectiveness from lifestyle changes

Complementary therapies

Participants were asked about any complementary therapies they used to manage their condition, the quality of life from these changes, and how effective they found them.

Most participants used at made at least one complementary therapy (n=216, 68.35%), and on average used 1 therapies (median=1.00, IQR=2.00).

The most common complementary therapy used was supplements (n=136, 46.10%), followed by mindfulness or relaxation (n=121, 45.83%), and massage therapy (n=80, 30.30%).

On average, quality of life from supplements was in the 'life was average' range (median=4.00, IQR=3.00), and was found to be somewhat effective (median=2.00, IQR=1.00).

On average, quality of life from mindfulness or relaxation was in the 'life was average' range (median=4.00, IQR=3.00), and was found to be moderately effective (median=3.00, IQR=2.00).

On average, quality of life from massage therapy was in the 'life was average' range (median=4.00, IQR=3.00), and was found to be moderately effective (median=3.00, IQR=1.00).

On average, quality of life from acupuncture was in the 'life was average' range (median=4.00, IQR=3.00), and was found to be moderately effective (median=3.00, IQR=2.00).

On average, quality of life from naturopathy was in the 'life was a little distressing to average' range (median=3.50, IQR=2.00), and was found to be somewhat effective (median=2.00, IQR=2.00).

On average, quality of life from homeopathy was in the 'life was average' range (median=4.00, IQR=3.00), and was found to be somewhat effective (median=2.00, IQR=2.25).

Table 5.12: Complementary therapies

Complementary therapies	Number	Percent	Median quality of life	IQR	Median effectiveness	IQR
Supplements (n=295)	136	46.10	4.00	3.00	2.00	1.00
Mindfulness or relaxation (n=264)	121	45.83	4.00	3.00	3.00	2.00
Massage therapy (n=264)	80	30.30	4.00	3.00	3.00	1.00
Acupuncture (n=264)	34	12.88	4.00	3.00	3.00	2.00
Naturopathy (n=245)	20	8.16	3.50	2.00	2.00	2.00
Homeopathy (n=245)	16	6.53	4.00	3.00	2.00	2.25

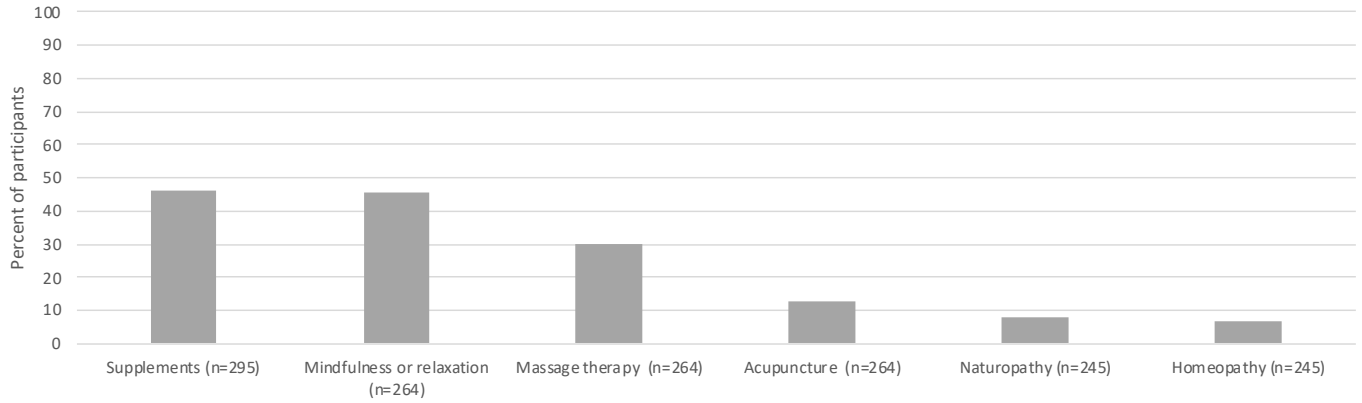


Figure 5.17: Complementary therapies

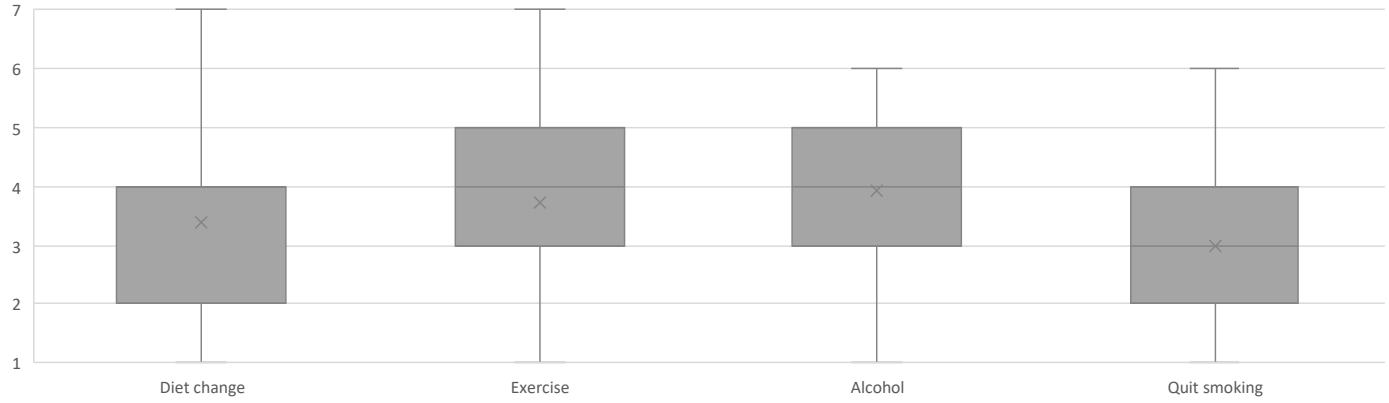


Figure 5.18: Quality of life from complementary therapies

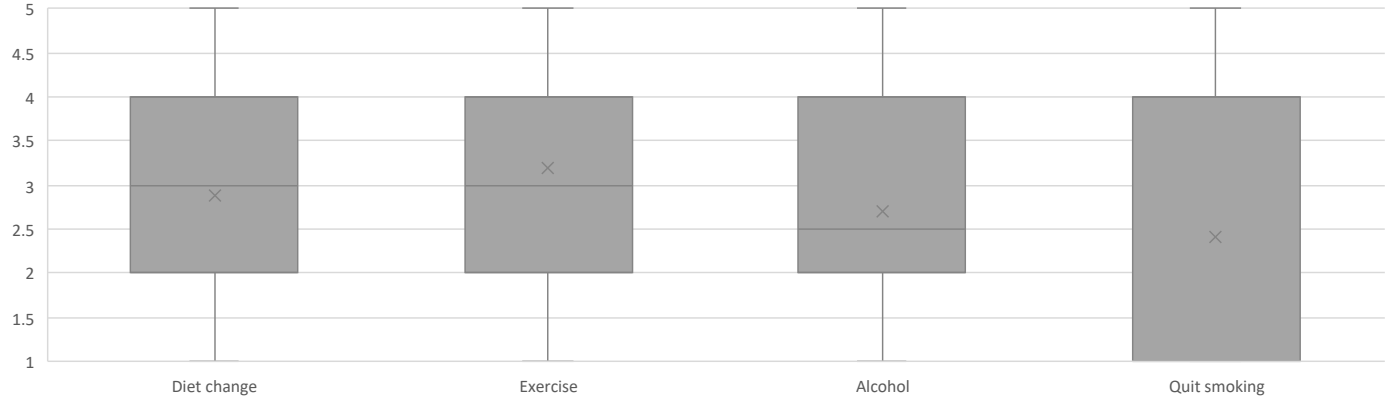


Figure 5.19: Effectiveness of complementary therapies

Clinical trials

Clinical trials discussions

In the online questionnaire, participants were asked if they had discussions with their doctor about clinical trials, and if they did, who initiated the discussion.

There was a total of 111 participants (35.81%) that had discussions about clinical trials, 32 participants (10.32%) had brought up the topic with their doctor, and the doctor of 79 participants (25.48%) brought up the topic. The majority of participants had not spoken to anyone about clinical trials (n=199, 64.19%).

Clinical trial participation

As a follow up question, participants were asked if they had taken part in a clinical trial, and if they had not taken part if they were interested in taking part.

There were 37 participants (11.86%) that had taken part in a clinical trial, 155 participants (49.68%) that would like to take part in a clinical trial if there was a suitable one, and 120 participants, that have not participated in a clinical trial and do not want to (38.46%).

Table 5.13: Clinical trial discussions

Clinical trial discussions	Number (n=310)	Percent
Participant brought up the topic of clinical trials doctor for discussion	32	10.32
Doctor brought up the topic of clinical trials for discussion	79	25.48
Participant has never spoken about clinical trials	199	64.19

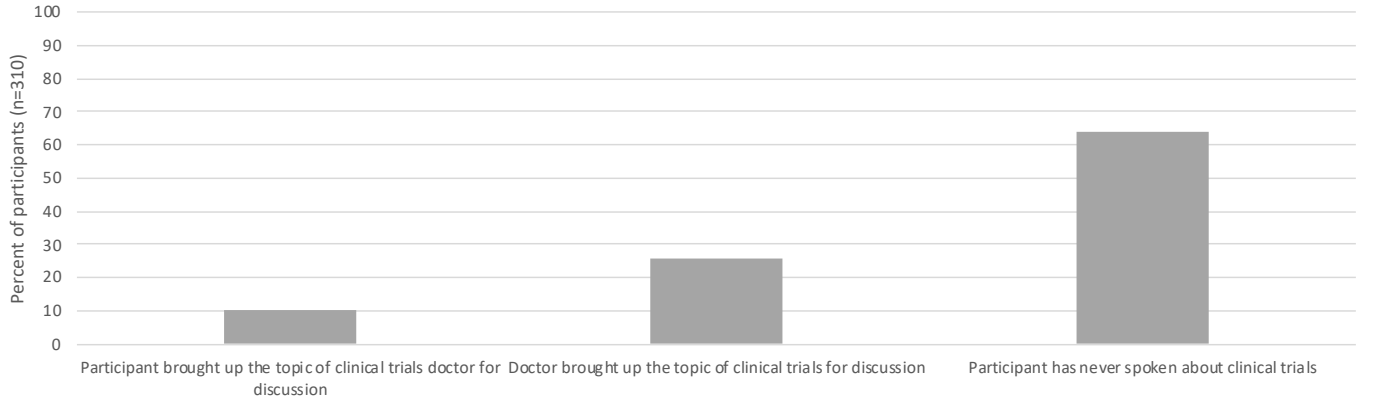


Figure 5.20: Clinical trial discussions

Table 5.14: Clinical trial participation

Clinical trial participation	Number (n=312)	Percent
Has not participated in a clinical trial	120	38.46
Has not participated in a clinical trial but would like to if there is one	155	49.68
Has participated in a clinical trial	37	11.86

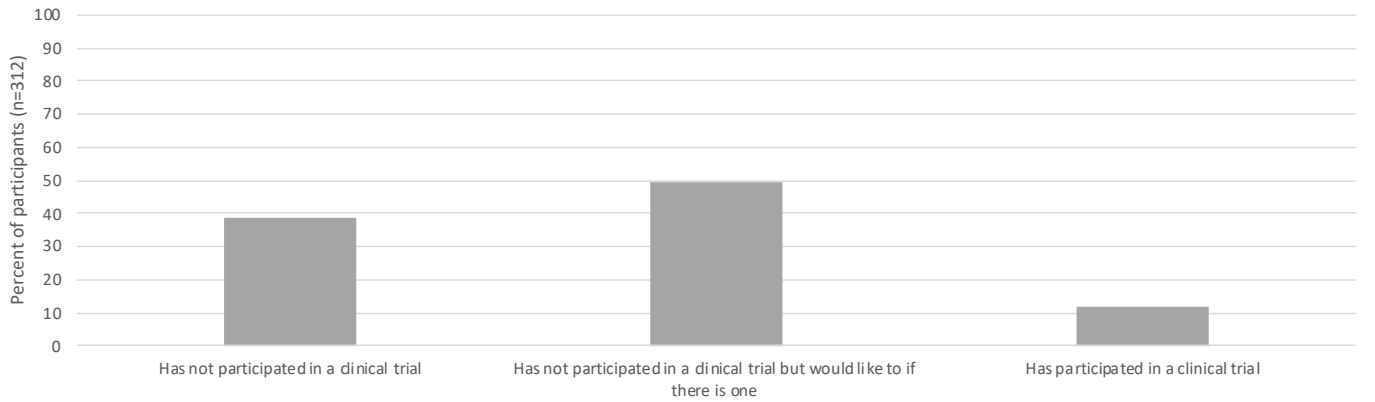


Figure 5.21: Clinical trial participation

Description of mild side effects

In the structured interview, participants were asked how they would describe the term 'mild side effects'. The most common descriptions of mild side effects were described using a specific example (53.69%), and those that do not interfere with life (33.24%).
Other themes included those that are resolved in a short amount of time (9.66%) and those that can be managed with self-medication or self-management (3.98 %).

When a specific side effect was described, the most common responses were fatigue and lethargy (12.50%), headaches (11.36%), aches/pain (10.51%) and gastrointestinal distress (9.38%). Other themes included lost of appetite (7.95%), skin rash, dry or itchy skin (7.67%) and nausea (3.98%).

Participant provides a specific side effect as an example

So mild would be like, I guess, you know, bruising from the the needle or yeah, the tummy discomfort from the antibiotics, that sort of thing. And just like if I was using creams, just the uncomfortableness of having it in the areas that they were in and having to sort of deal with that. Yeah.
Participant 022_2023AUDSK

Feeling a bit nauseous sometimes, yeah, but nothing that goes away or well, headaches. So just to get headaches from different medication, that was something that makes you sleep. I forget what it was called. It's melatonin.
Participant 088_2023AUENM

Fingers bending, the fibrosis, I suppose. I have lost muscle tone. I drop a lot of things, so that's a mild side effect.
Participant 008_2023AUDIS

A mild effect, I would say it's more like a headache. A severe thing is like constantly going back and forth to the toilet, because you can't take your medication and know you'd be out in public because you only think, oh, where's the toilet going to be?
Participant 023_2023AUDIS

Participant describes mild side effects as those that do not interfere with daily life

Don't really impact your ability to function normally, you know? So maybe feeling a bit squirmy in the stomach or yeah, having a bit of a headache or some itchy skin.
Participant 005_2023AUDSK

Basically he can like still go and do his go to school and do his things. It doesn't impact too much on his daily life.
Participant 009_2023AUDSK

Side effects, to me, I think that would be something that's, whilst you have side effects, doesn't necessarily impact your daily life too much. Yeah. Do you know, doesn't sort of decrease the quality of life I guess.
Participant 032_2023AUDSK

That you can continue to function. There's a little bit of with, a little bit of impact, but it's a manageable one and nobody external would be able to notice that you got.
Participant 092_2023AUENM

Participant describes mild side effects as those that can be self-managed

Well, you know, like I said, I had a, I had a sort of dull headache and I didn't feel the need to take anything. And then one night I had a more severe headache. But I just took two Panadol and I was fine and increased my water. So I drank a lot of water and I didn't have any severe.
Participant 010_2023AUORC

Well, I've only had the dry lips. Mine was just mild. Just Vaseline on the lips and just live with it.
Participant 013_2023AUDSK

So mild side effects I would kind of refer to as probably something like thrush and you can just use Canestan and you know sort it out you can, you can sort out the side effect.
Participant 017_2023AUDSK

That's an interesting question. Mild side effects I guess would be something that it would like mild side effect, something that you can kind of maybe manage daily on your own like with the correct mild side. Well, that's interesting. Something easily treatable at home, I guess.
Participant 029_2023AUDPA

Participant describes mild side effects as those that are resolved in a short amount of time

So I guess with mild side effects like things like you know, just I guess short term changes in like bowel habit, bowel habits, I guess it's hard to tell because of his age whether he gets it's just he, I don't even think he would know if he had a headache or anything like that. But short term I guess would be my description I guess of mild side effects or…Especially with like the liver enzyme changing and things like, I wouldn't call that a mild side effect, but if it was only short term then I would probably describe it more as mild. But I guess anything that's not going to cause long term damage or is only for a short period of time I would describe as mild side effects.
Participant 020_2023AUORC

A mild side effect might be a slight rash. That's not irritating. A mild side effect maybe something that dissipates really quickly and that doesn't affect the mental, mental and mental health wise. So not headaches or anxiety or depression. Yeah, a mild side effect of yeah not, not…doesn't make the lifestyle different.
Participant 023_2023AUORC

The mild side effect to me would be something that rarely impacts at all, maybe like slight nausea for like 10 minutes after taking it or whatever, but there's no actual impact on it.
Participant 025_2023AUORC

Maybe makes me drowsy, a bit drowsy. Something that's not added-- reacting in a minimal way but short-term as well.
Participant 058_2023AUDPA

Participants reports not experiencing any mild side effects

Mild side effects is level. He's very, very he's got a high tolerance, you know. I know it's really, really bad before he speaks up.
Participant 026_2023AUORC

She's very, like, resilient and guess she's been in the hospital so much. She's just used to everything, like nothing really bothers her.
Participant 013_2023AUDPA

In relation to her, well, she didn't really have any side effects from the medical procedures like that if her heart was repaired and she's been fine ever since.
Participant 027_2023AUDPA

Table 5.15: Description of mild side effects

Description of mild side effects	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=352	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=247	%	n=105	%	n=252	%	n=98	%
Specific example	189	53.69	34	50.75	51	62.96	32	71.11	15	48.39	36	37.89	21	67.74	129	52.23	60	57.14	134	53.17	53	54.08
Do not interfere with life	117	33.24	19	28.36	29	35.80	15	33.33	15	48.39	24	25.26	15	48.39	88	35.63	29	27.62	86	34.13	30	30.61
Resolves in short time/Temporary}	34	9.66	7	10.45	2	2.47	0	0.00	4	12.90	17	17.89	4	12.90	21	8.50	13	12.38	25	9.92	9	9.18
No mild side effects experienced (can not describe)	16	4.55	6	8.96	4	4.94	0	0.00	0	0.00	3	3.16	3	9.68	9	3.64	7	6.67	10	3.97	6	6.12
Can be managed with self-medication or self-management (Over-the-counter)	14	3.98	6	8.96	0	0.00	1	2.22	3	9.68	2	2.11	2	6.45	6	2.43	8	7.62	11	4.37	3	3.06

Description of mild side effects	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=352	%	n=69	%	n=116	%	n=108	%	n=59	%	n=172	%	n=172	%	n=100	%	n=252	%	n=176	%	n=176	%
Specific example	189	53.69	40	57.97	63	54.31	52	48.15	34	57.63	97	56.40	89	51.74	47	58.75	142	56.35	94	53.41	95	53.98
Do not interfere with life	117	33.24	22	31.88	40	34.48	34	31.48	21	35.59	50	29.07	65	37.79	30	37.50	87	34.52	51	28.98	66	37.50
Resolves in short time/Temporary}	34	9.66	7	10.14	14	12.07	8	7.41	5	8.47	18	10.47	16	9.30	13	16.25	21	8.33	15	8.52	19	10.80
No mild side effects experienced (can not describe)	16	4.55	5	7.25	5	4.31	3	2.78	3	5.08	10	5.81	6	3.49	7	8.75	9	3.57	8	4.55	8	4.55
Can be managed with self-medication or self-management (Over-the-counter)	14	3.98	6	8.70	3	2.59	3	2.78	2	3.39	5	2.91	9	5.23	5	6.25	9	3.57	9	5.11	5	2.84

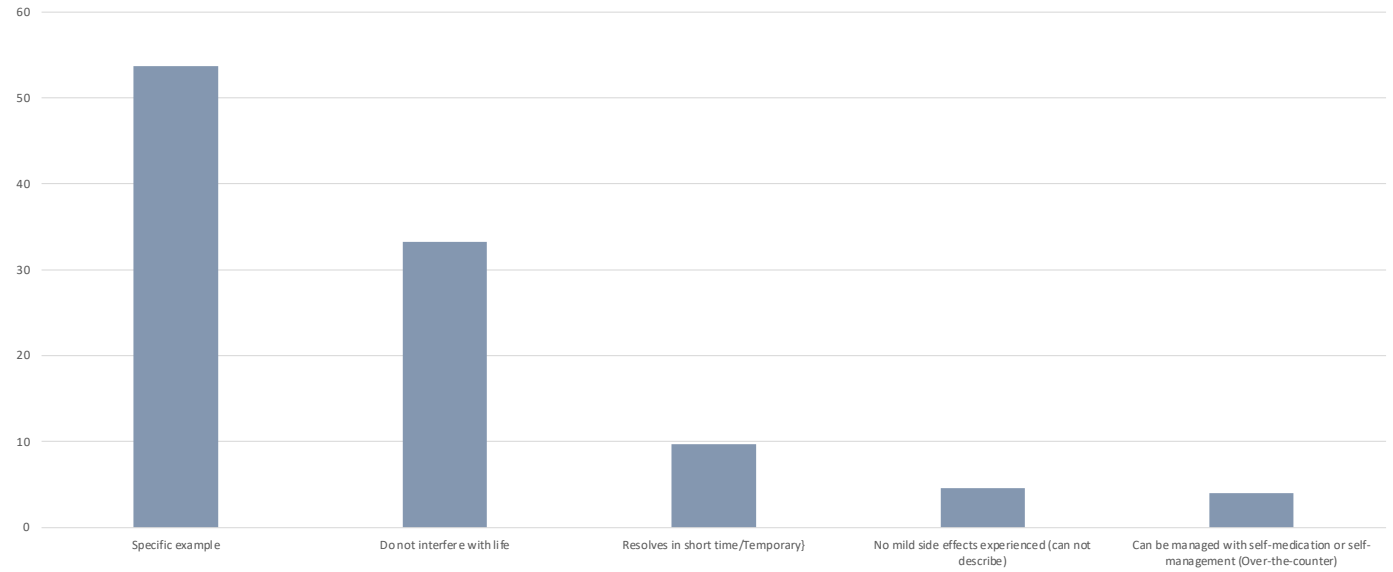


Figure 5.22: Description of mild side effects

Table 5.16: Description of mild side effects – subgroup variations

Description of mild side effects	Reported less frequently	Reported more frequently
Participant provides a specific side effect as an example	Endocrine, nutritional or metabolic diseases	Diseases of the nervous system Other rare condition
Participant describes mild side effects as those that do not interfere with daily life		Diseases of the skin Other rare condition
Participant describes mild side effects as those that can be self-managed		-

Table 5.17: Description of mild side effects (Specific side effects)

Description of mild side effects (Specific side effects)	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=352	%	n=352	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=247	%	n=105	%	n=252	%	n=98	%
Fatigue/lethargy	44	12.50	12	17.91	8	9.88	8	17.78	0	0.00	13	13.68	3	9.68	22	8.91	22	20.95	27	10.71	17	17.35
Headaches	40	11.36	4	5.97	15	18.52	5	11.11	2	6.45	8	8.42	6	19.35	29	11.74	11	10.48	32	12.70	7	7.14
Aches/pain	37	10.51	7	10.45	9	11.11	6	13.33	6	19.35	6	6.32	3	9.68	27	10.93	9	8.57	31	12.30	5	5.10
Gastrointestinal distress	33	9.38	3	4.48	9	11.11	7	15.56	1	3.23	10	10.53	3	9.68	29	11.74	4	3.81	28	11.11	5	5.10
Loss of appetite	28	7.95	1	1.49	13	16.05	4	8.89	2	6.45	5	5.26	3	9.68	25	10.12	3	2.86	21	8.33	7	7.14
Rash, dry or itchy skin/sensitive skin	27	7.67	2	2.99	8	9.88	2	4.44	10	32.26	1	1.05	4	12.90	22	8.91	5	4.76	21	8.33	6	6.12
Nausea	14	3.98	3	4.48	6	7.41	2	4.44	1	3.23	1	1.05	1	3.23	9	3.64	5	4.76	12	4.76	2	2.04

Description of mild side effects (Specific side effects)	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=69	%	n=116	%	n=108	%	n=59	%	n=172	%	n=172	%	n=100	%	n=252	%	n=176	%	n=176	%
Fatigue/lethargy	44	12.50	13	18.84	13	11.21	12	11.11	6	10.17	21	12.21	21	12.21	9	11.25	35	13.89	18	10.23	26	14.77
Headaches	40	11.36	6	8.70	16	13.79	12	11.11	6	10.17	19	11.05	21	12.21	14	17.50	26	10.32	25	14.20	15	8.52
Aches/pain	37	10.51	6	8.70	13	11.21	11	10.19	7	11.86	17	9.88	20	11.63	13	16.25	24	9.52	21	11.93	16	9.09
Gastrointestinal distress	33	9.38	3	4.35	7	6.03	10	9.26	13	22.03	16	9.30	16	9.30	4	5.00	29	11.51	13	7.39	20	11.36
Loss of appetite	28	7.95	0	0.00	11	9.48	13	12.04	4	6.78	16	9.30	12	6.98	5	6.25	23	9.13	13	7.39	15	8.52
Rash, dry or itchy skin/sensitive skin	27	7.67	5	7.25	15	12.93	5	4.63	2	3.39	13	7.56	14	8.14	10	12.50	17	6.75	18	10.23	9	5.11
Nausea	14	3.98	3	4.35	7	6.03	4	3.70	0	0.00	9	5.23	5	2.91	6	7.50	8	3.17	8	4.55	6	3.41

Figure 5.23: Description of mild side effects (Specific side effects)

Table 5.18: Description of mild side effects (Specific side effects) – subgroup variations

Description of mild side effects (Specific side effects)	Reported less frequently	Reported more frequently
Fatigue/lethargy	Diseases of the skin	
Gastrointestinal distress		Aged 65 plus
Rash, dry or itchy skin/sensitive skin		Diseases of the skin

Description of severe side effects

In the structured interview, participants were asked how they would describe the term 'severe side effects'. The most common description of severe side effects were described using a specific example (47.73%), and those that impact everyday life or ability to conduct activities of daily living (28.13%). Other themes included those that are life threatening or result in hospitalisation (8.52%), those that cause long-term damage to their body (7.67%).

When a specific side effect was described, the most common examples were aches and pain (17.33%), emotional and mental impact (7.39%), and nausea with vomiting (6.53%). Other themes included fatigues (5.11%), gastrointestinal distress (4.83%), impact on sleep (4.26%), vision problems (3.98%), and impact on sleep (4.55%).

Participant provides a specific side effect as an example

And I guess the severe one is the depression. But obviously I know what it is, I know what it feels like. I can pretty much handle it until it goes away.
Participant 014_2023AUDSK

Severe would just be mostly the pain, then the pain after I guess, the treatment…like the pain after the surgery or the excision and that sort of thing. That's probably been the worst.
Participant 022_2023AUDSK

Apart from like vomiting and nausea and gastrointestinal upset would be, we'll see…You would consider it mild, but in my child with their issues, it's actually quite a severe side effect. I can't really think of anything else.
Participant 021_2023AUORC

Massive, massive migraine, nauseous, vomiting. Can't cope with the vomiting and the really well, the migraines were like well I used to get migraines years ago. So every now and then I don't know seem to get it. But yeah some medications can give me headache, really bad headaches and I can't cope with that and I can't cope with the vomiting. But actually anything else is just yeah, you know.
Participant 088_2023AUENM

I would say it's that horrendous scream. He's been a really happy, really calm, really relaxed baby the whole way through. I think it's because of the steroids. He got more breakouts pretty badly. It seems that it never left that instant scream. He can scream for hours. I can be holding him or feeding him or patting him like it doesn't matter. He will now scream for hours at a time. If he's distracted in any way, he'll just scream so that's probably the most severe side effects for us.
Participant 064_2023AUDPA

Participant describes severe side effects as those that impact everyday life/ability to conduct activities of daily living

That would be medication when I tried the medication too fast and I just felt super spacey and couldn't walk properly and couldn't remember anything. I couldn't function and had to come off it. That was pretty severe. Also, with physical treatments, if I've say seen someone I'm not familiar with or it's just been a really difficult session because something was quite painful. Then there are times when the pain's too high, and I've had to stop treatment, or I've just had to sleep after. I've had to take stronger pain medication and sleep it off and not been able to function well for a couple of days, then I'd say that was probably a more severe side effect.
Participant 004_2023AUDPA

Well, like not being able to eat, or, yeah, not being able to get out of bed, or not being able to perform the normal functions like mild side effects. I'd say you should still be able to do things that you were already going to do, just maybe not at full 100% capacity. Yeah, with, you know, severe side effects. I feel like that's, you know, when you having something prevent you from being able to do something in order to try and fix another problem. You're then creating a myriad, you know, like a circle, a cycle where you're going to need to take something else for this. You know…it was like, well, here's some pills for your stomach, and then here's some pills for making those pills, for making you nauseous, for making your stomach feel better from the doxy. Participant 006_2023AUDSK

Daily life severe is when he can't go to school or he can't do his daily activities.
Participant 009_2023AUDSK

So severe side effects would mean that you couldn't go about your daily living, or your daily living would be severely impacted.
Participant 031_2023AUORC

Participant describes severe side effects as those that are life threatening or result in hospitalisation

Worst case scenario, allergy and an ambulance.
Participant 007_2023AUORC

Well, severe side effects like I said would be more along the life-threatening things which would require fairly urgent treatment or, or you know, to be looked at by someone. And PATIENT never really had anything like that. I suppose the worst side effects she would have had, it would be some pain when it comes to the operations on her legs. But apart from that and I wouldn't say that was severe, you know, more moderate. Yeah. And you know, Panadol and she's fixed like there was nothing really, you know, that would impact her greatly, really.
Participant 024_2023AUDPA

The severe side effects would be, in terms of the Afinitor which we keep an eye out for would be the effects of immuno-suppression. If he was to get sick, a simple cold could get a lot worse and he couldn't eat, he'd be hospitalised, so we just keep an eye. Not that we've ever had to do that, but I would think that that would be a severe symptom. The mild symptoms are skin reactions to the point that the dental debits that he got from the Vigabatrin. They're all things that we could deal with and fix. They're more severe stuff, anaphylactic reactions and severe immuno-suppression where we have, yes. I have to do a lot of things to help him out.
Participant 044_2023AUDPA

Participant describes severe side effects as those that cause long-term damage to their body

Yeah, kind of hinge it off what I said what we said about mild, it's like where it does interfere with your day-to-day functions. So yeah, where it's, where it's, it's having a negative and again fairly like noticeable, measurable and immediate or long-term impact on your quality of life. So it's, you know, it's having a negative impact.
Participant 011_2023AUORC

So when we were in hospital, one of the IV antibiotics he had a reaction to and it was like fever and labuored breathing and he came up in a rash and I would have…I describe that as a as a severe side effect… and it happened and they ended up taking him off it, but they weren't sure whether it was the reaction to the anaesthetic. But then the next time when he was on a different medication, it didn't happen at all. So I would describe that like the laboured breathing, the fevers, and the rash altogether. And just that really awful discomfort. I'm assuming he had a headache as well but like he was three at the time so he wouldn't have been able to communicate that but just overall was very miserable and also we deal can be with the risk of cataracts. I would describe the cataracts developing as a, as a quite a you know not a good side effect and I would also and I think like I guess very high level raised liver function over a long period of time that would lead to eventually to raise the liver. I would call that any…Yeah, in that category.
Participant 020_2023AUORC

Prednisone. Anything over 20 milligrams or even 15 milligrams. I don't sleep agitated. My hair grows and my face grows a nice downy peach fur. My bone density has dropped a couple of times, and I've had long stints on Prednisone, so yeah, I think Prednisone I would class as a severe side effect.
Participant 019_2023AUDPA

Yeah, well, liver failure, it would be severe and, and when the doctor tells you it will be fatal if you ever have this again, I think that's severe.
Participant 025_2023AUDPA

Participants reported not experiencing any severe side effects

Yeah, I haven't considered the…any treatment at the moment was I've been getting good results for the past 18 years, so.
Participant 001_2023AUORC

Well, I don't think I've actually experienced any from the medication anyway. Yeah, so that mean that's good so far.
Participant 003_2023AUDSK

Yeah. So I haven't had that experience, so.
Participant 010_2023AUDIS

Participant identifies severe side effects as impacting their everyday life by being bed ridden

Severe would probably be something where I'm in a bit more pain where I maybe need to lay down and get a heat pack out and put it on the area or I have an intense migraine. To the point where I need to turn off the lights and put myself to sleep and I can't really do much.
Participant 010_2023AUDSK

Yeah, so not something I've experienced with this particular medication, but I would class it as

something that makes me bedridden, home ridden. Something incredibly painful that doesn't allow me to get through my day and do my normal tasks. Yeah.
Participant 027_2023AUDSK

Mild is, with treatment I've had before, a bit of diarrhea or a bit of nausea, things like that. Severe side effects is where you just can't even move, [laughs] your whole body aches and you just can't move, you've got fatigue and you can't even get out of bed. That wasn't mild because I don't like staying in bed. [laughs]
Participant 001_2023AUDIS

Participant identifies severe side effects as requiring medical intervention

Something that you would have to seek regular treatment for, like someone else, like something that you..a side effect or something. Something that affects you that you need constant, constant and considerable assistance with managing all.
Participant 029_2023AUDPA

Well, something that requires, you know, a doctor's visit or an emergency presentation or more than just a day off school, like a day off school where I'd have to monitor her or something would be a severe side effect. I could…a day off school where she missed school and I couldn't.. I need to monitor her.
Participant 035_2023AUDPA

I think that's something that would interfere with his daily living, so something that required some intervention and he hasn't had anything like that.
Participant 040_2023AUDPA

Table 5.19: Description of severe side effects

Description of severe side effects	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=352	%	n=352	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=247	%	n=105	%	n=252	%	n=98	%
Specific example	168	47.73	34	50.75	45	55.56	32	71.11	11	35.48	32	33.68	14	45.16	112	45.34	56	53.33	117	46.43	49	50.00
Impact everyday life/ability to conduct activities of daily living	99	28.13	13	19.40	23	28.40	8	17.78	14	45.16	34	35.79	7	22.58	77	31.17	22	20.95	71	28.17	28	28.57
Life threatening or result in hospitalisation	30	8.52	6	8.96	10	12.35	3	6.67	3	9.68	5	5.26	3	9.68	21	8.50	9	8.57	23	9.13	7	7.14
Cause long-term damage to their body/last long term	27	7.67	9	13.43	10	12.35	1	2.22	1	3.23	3	3.16	3	9.68	17	6.88	10	9.52	20	7.94	7	7.14

Description of severe side effects	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=69	%	n=116	%	n=108	%	n=59	%	n=172	%	n=172	%	n=100	%	n=252	%	n=176	%	n=176	%
Specific example	168	47.73	36	52.17	54	46.55	51	47.22	27	45.76	84	48.84	81	47.09	47	58.75	121	48.02	88	50.00	80	45.45
Impact everyday life/ability to conduct activities of daily living	99	28.13	15	21.74	37	31.90	30	27.78	17	28.81	49	28.49	48	27.91	26	32.50	73	28.97	47	26.70	52	29.55
Life threatening or result in hospitalisation	30	8.52	7	10.14	7	6.03	11	10.19	5	8.47	16	9.30	14	8.14	7	8.75	23	9.13	16	9.09	14	7.95
Cause long-term damage to their body/last long term	27	7.67	8	11.59	9	7.76	7	6.48	3	5.08	13	7.56	14	8.14	12	15.00	15	5.95	15	8.52	12	6.82

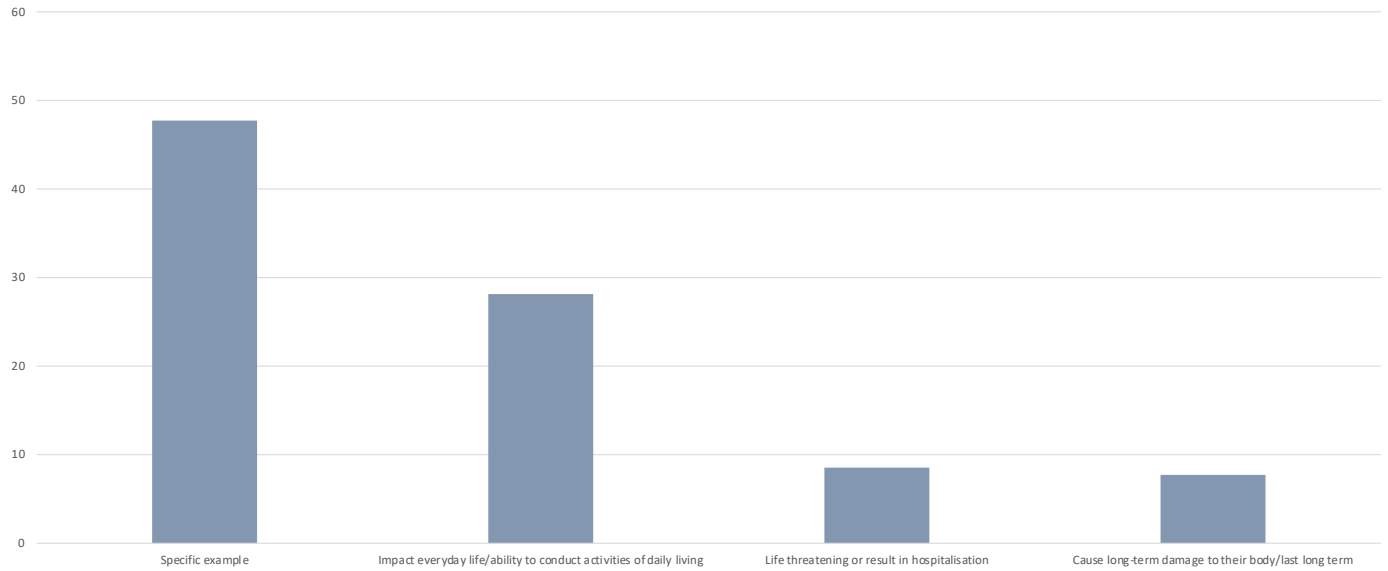


Figure 5.24: Description of severe side effects

Table 5.20: Description of severe side effects – subgroup variations

Description of severe side effects	Reported less frequently	Reported more frequently
Specific example	Diseases of the skin Endocrine, nutritional or metabolic diseases	Diseases of the nervous system Regional or remote
Impact everyday life/ability to conduct activities of daily living	Diseases of the nervous system	Diseases of the skin
Life threatening or result in hospitalisation		
Cause long-term damage to their body/last long term		

Table 5.21: Description of severe side effects (Specific example)

Description of severe side effects (Specific side effects)	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=352	%	n=352	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=247	%	n=105	%	n=252	%	n=98	%
Aches/pain	61	17.33	5	7.46	20	24.69	13	28.89	9	29.03	9	9.47	5	16.13	49	19.84	12	11.43	45	17.86	15	15.31
Emotion/mental impact	26	7.39	5	7.46	4	4.94	6	13.33	2	6.45	5	5.26	4	12.90	16	6.48	10	9.52	16	6.35	10	10.20
Nausea with vomiting	24	6.82	2	2.99	8	9.88	4	8.89	1	3.23	7	7.37	2	6.45	17	6.88	7	6.67	15	5.95	9	9.18
Fatigue/lethargy	18	5.11	0	0.00	8	9.88	6	13.33	1	3.23	1	1.05	2	6.45	16	6.48	2	1.90	15	5.95	3	3.06
Gastrointestinal distress	17	4.83	3	4.48	3	3.70	1	2.22	5	16.13	3	3.16	2	6.45	12	4.86	5	4.76	9	3.57	8	8.16
Impact on sleep	16	4.55	3	4.48	6	7.41	1	2.22	1	3.23	3	3.16	2	6.45	11	4.45	5	4.76	14	5.56	2	2.04

Description of severe side effects (Specific side effects)	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=69	%	n=116	%	n=108	%	n=59	%	n=172	%	n=172	%	n=100	%	n=252	%	n=176	%	n=176	%
Aches/pain	61	17.33	9	13.04	20	17.24	23	21.30	9	15.25	29	16.86	32	18.60	19	23.75	42	16.67	36	20.45	25	14.20
Emotion/mental impact	26	7.39	7	10.14	10	8.62	5	4.63	4	6.78	14	8.14	12	6.98	4	5.00	22	8.73	12	6.82	14	7.95
Nausea with vomiting	24	6.82	2	2.90	8	6.90	8	7.41	6	10.17	14	8.14	8	4.65	11	13.75	13	5.16	15	8.52	9	5.11
Fatigue/lethargy	18	5.11	1	1.45	8	6.90	8	7.41	1	1.69	9	5.23	9	5.23	5	6.25	13	5.16	10	5.68	8	4.55
Gastrointestinal distress	17	4.83	5	7.25	4	3.45	7	6.48	1	1.69	9	5.23	8	4.65	3	3.75	14	5.56	11	6.25	6	3.41
Impact on sleep	16	4.55	3	4.35	8	6.90	2	1.85	3	5.08	6	3.49	9	5.23	7	8.75	9	3.57	9	5.11	7	3.98

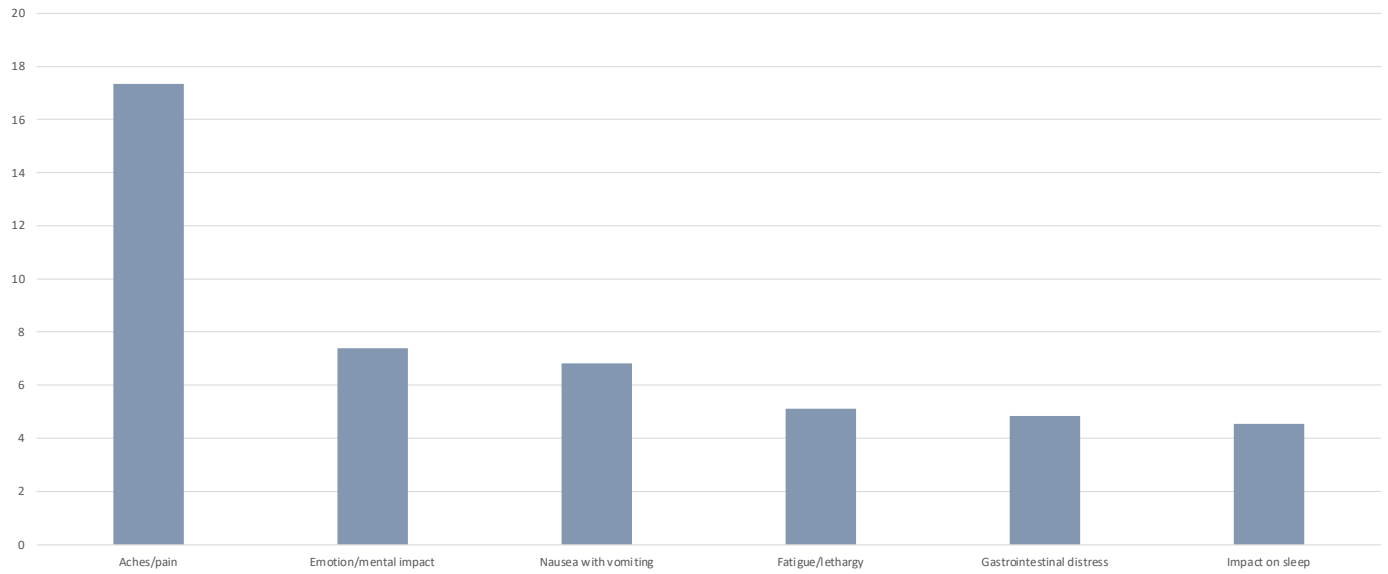


Figure 5.25: Description of severe side effects (Specific example)

Table 5.22: Description of severe side effects (Specific side effects)– subgroup variations

Description of severe side effects (Specific side effects)	Reported less frequently	Reported more frequently
Aches/pain		Diseases of the nervous system Diseases of the skin
Emotion/mental impact		
Nausea with vomiting		
Fatigue/lethargy		
Gastrointestinal distress		Diseases of the skin
Impact on sleep		

Adherence to treatment

Participants were asked in the structured interview what influences their decision to continue with a treatment regime. The most common responses were adhering to treatment for a specific amount of time (38.35%), adhering to treatment according to the advice of their specialist or as long as prescribed (36.08%), and adhering to treatment as long as side effects are tolerable (24.43 %). Other themes included never giving up on any treatment (11.36%), adhering to treatment as long as treatment is working (7.10%).

When participants stated a specific amount of time to adhere to a treatment, the most common amount of time was two to three months (14.20%), followed by one month (10.23%).

Participant describes adhering to treatment as per the advice of their specialist/as long as prescribed

Sorry, with the medications, we wouldn't stop anything without input from the doctors, from the medical team. So we persevered with those. We did raise our concerns quite frequently about various aspects of her treatment, but either the medical team thought it was worth continuing or we just hadn't got to the point yet where we had enough evidence. One of the issues we had as well with some of her treatments were that we were dependent on receiving…having ECGs to see whether they were having an impact and unfortunately the original neurologist was not supportive of doing ECGs. So we weren't really able to track her the efficacy of her treatments as closely as we would have liked.
Participant 090_2023AUENM

Okay. I usually stick to what the doctor-- as in if the doctor says, "Okay, you are on this medication." They usually give you two weeks to a month to -- if that's going to take a month to get into the system, I usually work on that particular length of time. If the doctor says this is X, you're going to take this for X and you're going to take it at this time, this time, and this time, that's what happened until the next time I see the doctor. The way I work where trying to take medication is concerned is, it might be a side effect or it might be that it's doing what it's supposed to in cleaning up my system so that it can start to work. It's based on what the doctor says as far as the medication is concerned. Basically, I do what the doctor tells me.
Participant 005_2023AUDPA

I'll keep using it until the next checkup because anytime I'm on a new medication, I have a checkup in, what, three to six months? So I won't stop until I'm actually given the clear, just in case there's some side effects.
Participant 026_2023AUDSK

Well, if there's any new treatment, so I'll stick to it till my doctor says I have to stop it. So it's just based on my doctor.
Participant 006_2023AUORC

Participant describes adhering to treatment for a specific amount of time

Usually at least a month for medications. When we try like new specialists or allied health, we obviously give them sort of like three to six months.
Participant 032_2023AUDPA

I don't know. I guess at least a few months to see. Well, it depends how long it takes the treatment to... So if she's taking her medicine, if it takes a good four weeks for it to start working or you've got to give it at least, you know, a couple of months.
Participant 010_2023AUDPA

I guess my rule for any medication is 3 months and if it's not working then it's probably not going to work. But yeah, this is my first lot of actual meds that I've tried.
Participant 014_2023AUDSK

Probably a month and then if it doesn't work, I kind of give up on it. And then when I go to the doctor's next day, I, you know, why do you stop that? And then if it's something that, if it's in regards to physio, they'll try and teach you a new technique of how to do that, or try and see if there's a different medication that does the same thing. Or just to clear the mucus out of the lungs. Yeah, they'll see if there is something else that they can do.
Participant 013_2023AUORC

Participant describes adhering to treatment as long as side effects are tolerable

If it wasn't showing any bad side effects, I'd probably persist, probably for about three months or so and then I'd be questioning. If we weren't seeing any positive improvement, I'd be questioning it with our paediatrician. I'd give it a little while, but if it had severe side effects, I'd be stopping pretty quick if I couldn't see a big change.
Participant 048_2023AUDPA

I will usually I follow the, the prescribed course unless I think I've had a big reaction to it, in which case I'll reach out to the prescriber and say this is what's going on. Can I stop this or should I be stopping this?
Participant 007_2023AUDSK

I would probably be like in constant consultation with the hospital, like the hospital care team, if I thought that it wasn't working. And I guess it depends on the severity of the side effects, whether I would cease it or continue it and try and push through. Yeah, I think I'd,

I'd throw the ball straight back to the kids hospital if I felt that it wasn't. I mean, he's a child. I don't want him to have any undue, you know side effects if he doesn't need them, so I don't really know how to answer that question other than say probably within a day or less depending on what the side effect was.
Participant 021_2023AUORC

Well, I'm not that kind of person. If I get a treatment and I'm told to take it from the beginning to the end, I take it until it's finished. I never, I never stop unless, unless it's giving me a severe side effect which really makes me ill, then I take it till it's finished.
Participant 010_2023AUORC

Participant describes not giving up on any treatment

I don't know. Because he's always wanted to persevere. He never gives up. Now he always sticks through with things. Yeah.
Participant 011_2023AUDPA

I've been on this injection for three years or something. I don't, I can't tell you. Like I said, I, I don't give up. I mean, you know, I, I was never a pill taker, but I started, you know, seeking assistance for mental health conditions when I was 30. And I understand I'm not medicated now, but I understand relapse and those sorts of things occur from people just saying and and I can tell you, there were times where I thought, 'what's this doing anyway?' But I never did. I mean, never missed a dose. Same with my, you know, contraceptive, those sorts of things. So how long before I gave up? Probably never until circumstances changed where it was, you know, time. So there's no real give up, but it's a more of a measured decision that I would, I would make.
Participant 015_2023AUDSK

I don't think I've ever given up on one. I'm compliant.
Participant 004_2023AUDIS

I probably haven't had any treatment as in, you know, any treatment that I've canned on. So, you know, my doctor has obviously put me through this first round of radiation. So I don't know what my next sort of outcome is going to be if it does start growing again or I find it somewhere else. So all we've really done so far is just had the radiation and that's about it. So I really don't know what my next sort of scenario will be whether the doctor's gonna try and cut it out or whether he's gonna, I don't know, put a probe in it and try and kill it from the inside or I'm not 100% sure at this stage, so yeah.
Participant 024_2023AUORC

Now, I wouldn't say we're actually ever really given up. We've always found persistence pays, and sometimes it might be frustrating for her, or she's a bit psych or lazy and doesn't want to do it, but so we find repetition working with us and therapists. It might take a few weeks to click on, but persistence pays, and as far as I can recall, we've never given up on any particular strategy or intervention.
Participant 022_2023AUORC

Participant describes adhering to treatment as long as treatment is working

That I guess would depend on what we are treating. We haven't had any issues with medication where we felt it was ineffective because he hasn't, he doesn't need to take a lot of medication. He, he was on medication for his reflux but that was very effective so you know that was a there was no discussion to be had it worked, kept taking it. I listen to the doctor's advice a lot. I would say if he's been taking a medication for something for three months and we're not seeing any improvement, I, I would, I'd take him off it because I think if you haven't seen any improvement in three months, then clearly either the medication's ineffective or you're treating the wrong thing. But we haven't had to do a lot medication wise it's, it's more been surgeries and because those surgeries have all been so successful you know we haven't really had to wonder if they were worth it.
Participant 091_2023AUENM

Well, the dose that they gave me in hospital kind of worked within that few hours, but then after that it come it came back and then after that the doses weren't really doing much of anything at all. So I don't know, I was on them for a few weeks before they changed to another one. But I guess I don't know because it wasn't working. My tolerance level of wanting to keep going with it wasn't good. OK, I think if it worked. I probably would have prolonged the medication, if you know what I mean.
Participant 032_2023AUORC

Participant describes needing to see test results/no evidence or reduction of disease in order to adhere to treatment

I just have, I haven't really challenged it. I've been told that I'm on this medication pretty much for life until they tell me otherwise. I think what helps is the regular blood tests of monitoring the levels of what is happening with my body.
Participant 004_2023AUORC

It's really hard to tell whether they're working or not because you need to have cultures after the medication is completed so it's not you…we don't usually question whether it's working or not, although we can tell. We are usually hopeful with symptoms decreasing, but it's more medications within sort of two or three days. We would know if he has like a really bad rash come up actually. Thinking about it, we've had a lot of drugs where the side effect might be really cranky, bad behaviour, and we probably put that one down as a mild because we have to just live with it. But, it's very obvious that it's for drugs and we, we usually stick it out with those, but yeah, it'd be the rash within to the 28, you know, 48 hours, yeah.
Participant 023_2023AUORC

Yeah. So normally at the beginning we did a few things for like a month at a time and then we gave up because it wasn't working, which was probably the wrong decision when you look back. Yeah, normally we try things for at least two months. Yeah, but it's tricky. It's a tricky one because you can't really, the outward symptoms don't always match what's happening on the inside. So we you typically have to do a scope to really know the answer. OK, yeah.
Participant 079_2023AUDIS

Participant is unable to answer because they have not had treatment and/or cannot answer hypothetical question

Actually there's nothing I can think of that I know has worked in the sense that it's really just being careful reducing these amounts, being slower or that sort of thing. That has always helped but I don't feel that anyone is ever given me, certainly no magic bullets. Not even something I could try and say, oh, well, it didn't work. The approach from everybody is the peripherals, the things that show and the things that I can say like the dryness and the cold of my mouth and the cold disease and the oesophageal problems and things. They're all peripheral in a way. They're the result of the combination of things…no one can treat that because there isn't any treatment as far as I know.
Participant 012_2023AUDIS

Yeah, I don't think I can really answer this question. It's been yeah, I've just never had to experience it. I can answer for my brother, but that's not me. And if we he tried a couple of different ones, but it was like quite extensive. You give it a bit of time and in his circumstance like he had the time to give it where it's not sort of effect. He wasn't working, he was still at school, but he was like taking time off. So in that sense it was like not as detrimental. Whereas like if I was working, I think you'd have to consider a much different approach. But I can't really give you much of an answer for that question.
Participant 030_2023AUORC

We haven't really had to do any of that, I don't think, to be honest. We haven't had to sort of go in that this isn't, you know, working. It's useless. Yeah, we haven't, we haven't experienced that. So yeah, I guess I can't, I can't really answer that one.
Participant 034_2023AUDPA

Table 5.23: Adherence to treatment

Adherence to treatment	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=352	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=247	%	n=105	%	n=252	%	n=98	%
Adhering to treatment for a specific amount of time	135	38.35	23	34.33	42	51.85	17	37.78	11	35.48	36	37.89	6	19.35	100	40.49	35	33.33	107	42.46	28	28.57
Advice of their specialist/as long as prescribed	127	36.08	26	38.81	27	33.33	13	28.89	18	58.06	30	31.58	13	41.94	93	37.65	34	32.38	89	35.32	37	37.76
As long as side effects are tolerable	86	24.43	20	29.85	22	27.16	17	37.78	9	29.03	10	10.53	8	25.81	57	23.08	29	27.62	62	24.60	24	24.49
Participant describes not giving up on any treatment	40	11.36	8	11.94	11	13.58	9	20.00	1	3.23	7	7.37	4	12.90	29	11.74	11	10.48	28	11.11	12	12.24
As long as treatment is working	25	7.10	6	8.96	4	4.94	5	11.11	4	12.90	4	4.21	2	6.45	15	6.07	10	9.52	16	6.35	9	9.18

Adherence to treatment	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=352	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=247	%	n=105	%	n=252	%	n=98	%
Adhering to treatment for a specific amount of time	135	38.35	23	34.33	42	51.85	17	37.78	11	35.48	36	37.89	6	19.35	100	40.49	35	33.33	107	42.46	28	28.57
Advice of their specialist/as long as prescribed	127	36.08	26	38.81	27	33.33	13	28.89	18	58.06	30	31.58	13	41.94	93	37.65	34	32.38	89	35.32	37	37.76
As long as side effects are tolerable	86	24.43	20	29.85	22	27.16	17	37.78	9	29.03	10	10.53	8	25.81	57	23.08	29	27.62	62	24.60	24	24.49
Participant describes not giving up on any treatment	40	11.36	8	11.94	11	13.58	9	20.00	1	3.23	7	7.37	4	12.90	29	11.74	11	10.48	28	11.11	12	12.24
As long as treatment is working	25	7.10	6	8.96	4	4.94	5	11.11	4	12.90	4	4.21	2	6.45	15	6.07	10	9.52	16	6.35	9	9.18

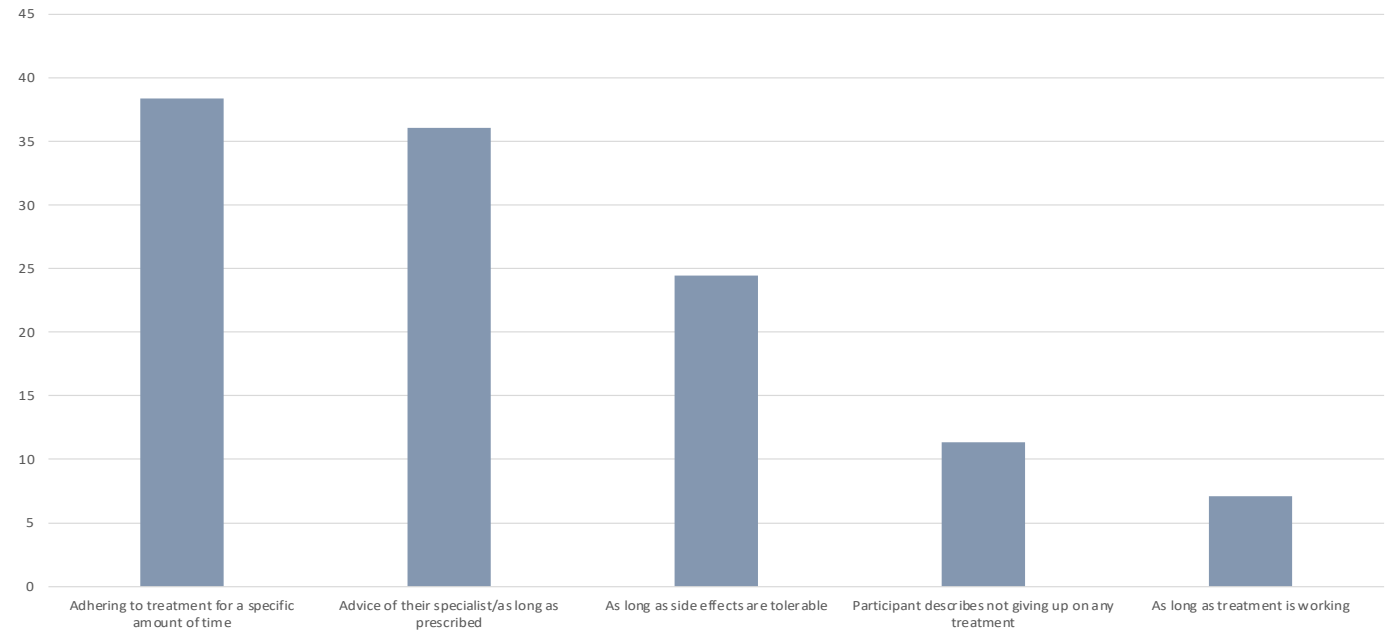


Figure 5.26: Adherence to treatment

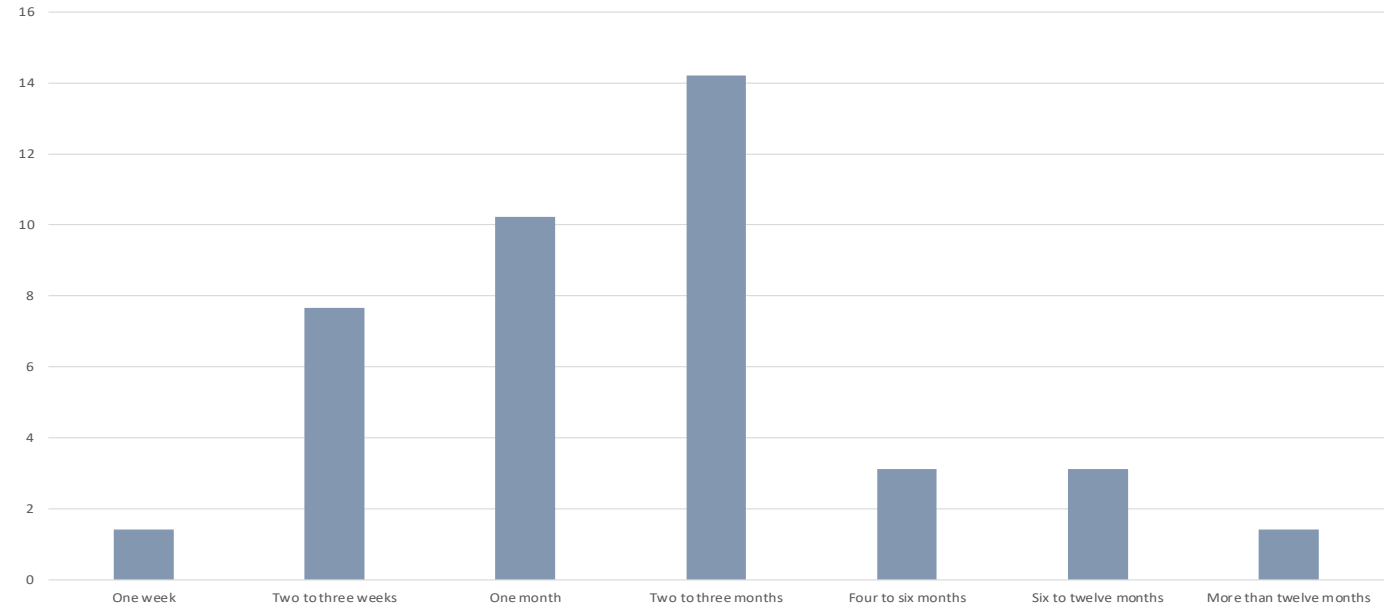


Figure 5.27: Adherence to treatment (Time to adhere to treatment)

Table 5.24: Adherence to treatment – subgroup variations

Adherence to treatment	Reported less frequently	Reported more frequently
Adhering to treatment for a specific amount of time	Other rare condition	Diseases of the immune system Regional or remote
Advice of their specialist/as long as prescribed		Diseases of the skin Regional or remote
As long as side effects are tolerable	Endocrine, nutritional or metabolic diseases Aged 65 plus	Diseases of the nervous system
Participant describes not giving up on any treatment		
As long as treatment is working		

What needs to change to feel like treatment is working

Participants were asked to describe what needs to change to feel like treatment is effective. The most common responses were needing to see a specific symptom reduction (26.70%), needing to see needing to see physical signs and symptoms disappear or reduce side effects (25.85%), a needing to see improvements in general wellbeing (quality of life) (14.49%), needing to see evidence of stable disease (14.20%), needing to see a return to day-to-day functionality (14.20%), and needing to see improvement in pain levels (12.50%).

When a specific side effect or symptom was described, the most common examples were emotional and mental impact (3.41%), and impact on sleep (2.84%).

Participants reported needing to see all physical signs and symptoms disappear

So I'm in the most recent medication. Obviously the Humira is probably the easiest one for me to talk about, and it has…there are a lot of side effects for Humira. I'm very fortunate that I am not one of the people that experiences any nasty side effects, but for me the improvement was that I wasn't getting 6 to 8 new lesions a day. Yeah, so now I might have three come up in a month. 4 come up in a month and generally speaking they're very mild. They come up and they go away as quickly as they come up. Occasionally they flare up, but generally that's, you know, it's it's once in a blue moon that it flares up the leg, the leg and the breast abscess that, as I said, there's been no change with those whatsoever. They still flare every week.
Participant 005_2023AUDSK

With it, yeah, one probably reduction in side effects or if they are quite severe, they've at least come down to a mild state. Like for example, we had hair loss when we first started that was pretty, pretty bad, but we just sort of waited it out and see if it got worse then we could always stop it, if not then but it went down to mild and also just obviously whatever you're treating. So like we're treating his seizures, obviously a reduction in seizures is always a good statistic and good see.
Participant 081_2023AUDIS

I guess it would just be that I can see some sort of visible positive difference, whether that's a reduction in negative symptoms, so whether something is actually, it was safe. It's his sleep apnoea, whether I can actually hear that it's getting better or hearing breathing better or whether it's something positive being added, like maybe he's starting to add a bit more communication in as a result of the speech pathology. So yeah, I think the main things would be that either a reduction in the negative symptoms or an addition of something that positive.
Participant 089_2023AUENM

I guess that you need to see that those symptoms are resolving, yeah, within the time frame mm.
Participant 038_2023AUDPA

Participant describes needing to see a reduction in a specific symptom

For me, for this condition, I would say pain relief is #1. And because it is kind of a visible condition, seeing like those boils on the skin, I'd say just a visible reduction in inflammation and redness, really. Yeah.
Participant 027_2023AUDSK

Yeah, definitely reduction in side effects. And for me it's things like it's easier to swallow, less pain, less rigidity, things sliding down a lot easier not getting that breathlessness feeling and that kind of heavy feeling that need to stretch. The specific symptoms. Yeah, just that those things ease and I can kind of eat or drink. Sometimes it's even drinking. Just like a normal person, I guess.
Participant 078_2023AUDIS

Going with this, yeah, the reduction will basically the reduction of the seizures. I think that's probably the in terms of the treatment that she has for that then, yeah.
Participant 016_2023AUORC

I understand about little people. You'd want to see progress against the goal that you had for the treatment. If you're taking an anti-constipation medication, you want to see that it's having the desired effect of relieving constipation. If you're doing speech therapy, you want to, over time, see some improvement in articulation or being able to apply the social and pragmatic communication skills in practice or whatever it might be.
Participant 067_2023AUDPA

Participants reported needing to experience evidence of stable disease/no disease progression

I think you just need to see some level of results and sometimes I think the blood test results are just a stronger indication of how I'm feeling day-to-day to attribute that. So I think the monitoring of the actual condition and knowing that, you know, when you do do my blood test, you are looking for viral load, you're looking for all the other impacts.
Participant 004_2023AUORC

Probably better monitoring instead of like here, take this medication and come back in 6 to 12 months. I should be seeing you to see how you're going and reassure you that yes, while you're not seeing signs, it is actually working. So better monitoring.
Participant 013_2023AUDSK

Yeah, you know. I can't really say that because a lot of these aren't standardized treatments, so you don't really have a particular measure. But if we were to say that, then we would say obviously blood test results. For example, if they're in hospital or they're having medication, we would be comparing blood work, which isn't my job, it's the doctor's job. If it's something like Allied health, then it would be about the improved level of functioning, which would be perceived by me. Or reported by the Allied health professional, because, again, these aren't standardized treatments. Really, I think that's it.
Participant 021_2023AUORC

Participants reported needing to experience an improvement in pain levels

Okay. As far as that's concerned, it's if I can feel there is a difference in whether the actual treatment is working. As in, if I have pain, if the pain is diminishing and I can feel that there's less pain because of the new treatment is working or something like that. As far as other medication is concerned, if I can feel that there's a marked improvement. Just to expand, because of some of the issues that my daughter's been having…When she was living at home, we changed from basically a normal diet to a gluten-free diet because she was put onto a strict diet and we worked too. We were cutting out gluten and et cetera. There was an improvement, the way she was feeling was improving in the way I was feeling. It's a good side effect in that I can feel something is changing to the better.
Participant 005_2023AUDPA

Probably in some situations less pain, and in other situations it's test results showing that things haven't progressed or things are getting better, so it's a big combination of things really, isn't it.
Participant 007_2023AUDIS

For me, it would be, I would need to have equal or better than relief from the symptoms. The fact that this particular Botox is less harmful is probably not useful for me if it's not providing me pain relief and relief from the spasming. It would have to do that. It would have to stop the pain and the spasming for me to consider it.
Participant 006_2023AUDNS

Participants reported needing to experience an improvement in general wellbeing (quality of life)

Positive outcomes and reaching milestones, I guess. Yeah. So positive improvement looks like, you know, our son's being happy, enjoying life, reengaging with community, doing hobbies and activities that he likes and finds meaningful and fulfilling. Yeah, and I guess maturing in a way that he's developing his self identity of who he is and developing as his own person.
Participant 031_2023AUDPA

Well, the aim is to have less spasms and better quality of life.
Participant 001_2023AUDNS

I guess just a sense of normality. Obviously, that's quite broad, but just a sense of normality, I guess, in the sense where it's helping whatever I'm trying to target to a point where I can get out in the community and actually exist rather than being stuck at home.
Participant 004_2023AUDNS

If it was a treatment for seizures, they would need to stop entirely. If it was a treatment for the spots on his face, they would have to go because he has these sensory problems and it would be more of a battle to get it on than it would be worth. Because his is only a mild case, it is hard to say. If it wasn't going to improve his life or if it wasn't going to make our lives easier to reduce stress or knowing that we weren't going to have to put up with many things and all that sort of stuff or because there's nothing that you could do about his sensory issues or autistic tendencies. It would have to make our lives easier for us to continue to take it. Not necessarily ours, but his life. It would have to mean that it was going to make his life better in the long run.
Participant 046_2023AUDPA

Participants reported needing to experience a return to day-to-day functionality

They don't feel like side effects, but anyway, they feel like just the effects. It's a hard thing to quantify because at the moment, everything that's involved in just having a day, like getting up, putting on clothes, washing myself is as much as I can handle in a day. I don't wash myself fully every day because if I do that, I can't do anything else. It's really hard. That's a really, really hard question because I don't know anything from being able to have a shower every day or have a shower whenever I felt like it without needing to ask someone for help. I would be able to clean my teeth every day without that having an impact on my fatigue and my arm. At the same time, if I had medication that was managing all of that, maybe I could do more than all of that. Maybe I could go out sometimes. It feels like an almost impossible question to answer.
Participant 001_2023AUDPA

Functions might be good. I just want to be able to be as normal as I can. For example, the pain medication, it's like, "Oh, I can get through a whole workday now," or I'd say muscle-wise like I'm able to garden once a week without pain or something like that. I have quite specific markers for my daily life. That's how I do it. I'm able to play with my children on the floor comfortably. My back must be good. My knees aren't popping out, great, that sort of thing. I tend to use the day-to-day functional things as my markers or able to walk to drop off rather than drive, those types of things. I'm doing well with it.
Participant 004_2023AUDPA

So one of the medications she's on is for anxiety, and that made a major difference. It allowed her to function. The other one she's on is for her thyroid. And again, you could see. I mean, you could see a quantitative difference there in her thyroid levels.
Participant 021_2023AUDPA

I'd like to be able to drink out of a cup in public and eat with a knife and fork. I can't. I cut all my food up with the pair of scissors, right? I eat it out of, I eat out of a bowl. That's alright. A lot of people lay out of a bowl, but I couldn't, I can't eat a steak, for instance. I mean, I probably couldn't cut it up with the..anyway. I don't.
Participant 003_2023AUDNS

Participants reported needing to experience a reduction in fatigue levels

If I'm taking a treatment that is making me feeling more fit, less tired. If I have more energy and if I feel less tense and stiff that's happening.
Participant 020_2023AUDIS

I guess I think like I know it's working if I feel well enough to do everyday life things. Like, I think I didn't realize quite how fatigued and sick I felt until I stopped feeling like that because, you know, it's been so such a long time.
Participant 024_2023AUDIS

For example, with the epilepsy meds, the drowsiness, I need to see that he can actually manage and get through the day without falling to sleep in class, and that's not going to impact on his learning.
Participant 063_2023AUDPA

Participants reported needing to experience improved mobility

To feel it has worked. Reduction of, reduction of symptoms, so reduction in in reports of pain, reduction in cramps. He's walking better.
Participant 026_2023AUORC

So a reduction in side effects go up with her medication, so the side effects went away. OK, well, that's working. Reports back from people she interacts with. So like schools saying yes, it's been a much improvement in her ability to engage in the classroom. Yeah. So positive, positive feedback or if she's being able to do things independently. So yeah, so seeing improvements in her in her life, like with the OT being able to all like physio with her now, being able to jump up and down or go up and down stairs properly or seeing that or people saying, oh, now she can do that or she's reading things, she's able to decode words or we see her working through her issues. So we're going, Oh yeah, that. So those things are working because we're seeing results.
Participant 017_2023AUDPA

My tolerance of exercise and my shortness of breath. So when I'm not well, it's a struggle to walk up one side of stairs. I'm acquiring anything, so at the moment, post COVID, I'm still struggling to bring a bag of groceries up my stairs. But when I'm well, I know I can walk up with two bags in each hand.
Participant 019_2023AUDPA

Table 5.25: What needs to change to feel like treatment is working

What needs to change to feel like treatment is working	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=352	%	n=352	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=247	%	n=105	%	n=252	%	n=98	%
Specific symptom reduction	94	26.70	31	46.27	18	22.22	15	33.33	4	12.90	19	20.00	7	22.58	51	20.65	43	40.95	66	26.19	27	27.55
Physical signs and symptoms disappear/reduce side effects	91	25.85	13	19.40	13	16.05	18	40.00	25	80.65	6	6.32	16	51.61	63	25.51	28	26.67	67	26.59	23	23.47
Improvement in general wellbeing (quality of life)	51	14.49	12	17.91	6	7.41	5	11.11	2	6.45	25	26.32	1	3.23	35	14.17	16	15.24	34	13.49	17	17.35
Evidence of stable disease/no disease progression	50	14.20	0	0.00	19	23.46	14	31.11	1	3.23	12	12.63	4	12.90	42	17.00	8	7.62	37	14.68	13	13.27
Return to day-to-day functionality	50	14.20	11	16.42	12	14.81	2	4.44	2	6.45	22	23.16	1	3.23	38	15.38	12	11.43	39	15.48	11	11.22
Improvement in pain levels	44	12.50	4	5.97	22	27.16	10	22.22	0	0.00	8	8.42	0	0.00	39	15.79	5	4.76	39	15.48	5	5.10

What needs to change to feel like treatment is working	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=69	%	n=116	%	n=108	%	n=59	%	n=172	%	n=172	%	n=100	%	n=252	%	n=176	%	n=176	%
Specific symptom reduction	94	26.70	30	43.48	23	19.83	26	24.07	15	25.42	45	26.16	48	27.91	28	35.00	66	26.19	48	27.27	46	26.14
Physical signs and symptoms disappear/reduce side effects	91	25.85	20	28.99	34	29.31	25	23.15	12	20.34	41	23.84	50	29.07	22	27.50	69	27.38	47	26.70	44	25.00
Improvement in general wellbeing (quality of life)	51	14.49	10	14.49	19	16.38	9	8.33	13	22.03	23	13.37	27	15.70	15	18.75	36	14.29	26	14.77	25	14.20
Evidence of stable disease/no disease progression	50	14.20	3	4.35	17	14.66	20	18.52	10	16.95	22	12.79	26	15.12	14	17.50	36	14.29	21	11.93	29	16.48
Return to day-to-day functionality	50	14.20	11	15.94	13	11.21	18	16.67	8	13.56	25	14.53	25	14.53	19	23.75	31	12.30	28	15.91	22	12.50
Improvement in pain levels	44	12.50	2	2.90	18	15.52	16	14.81	8	13.56	26	15.12	18	10.47	9	11.25	35	13.89	20	11.36	24	13.64

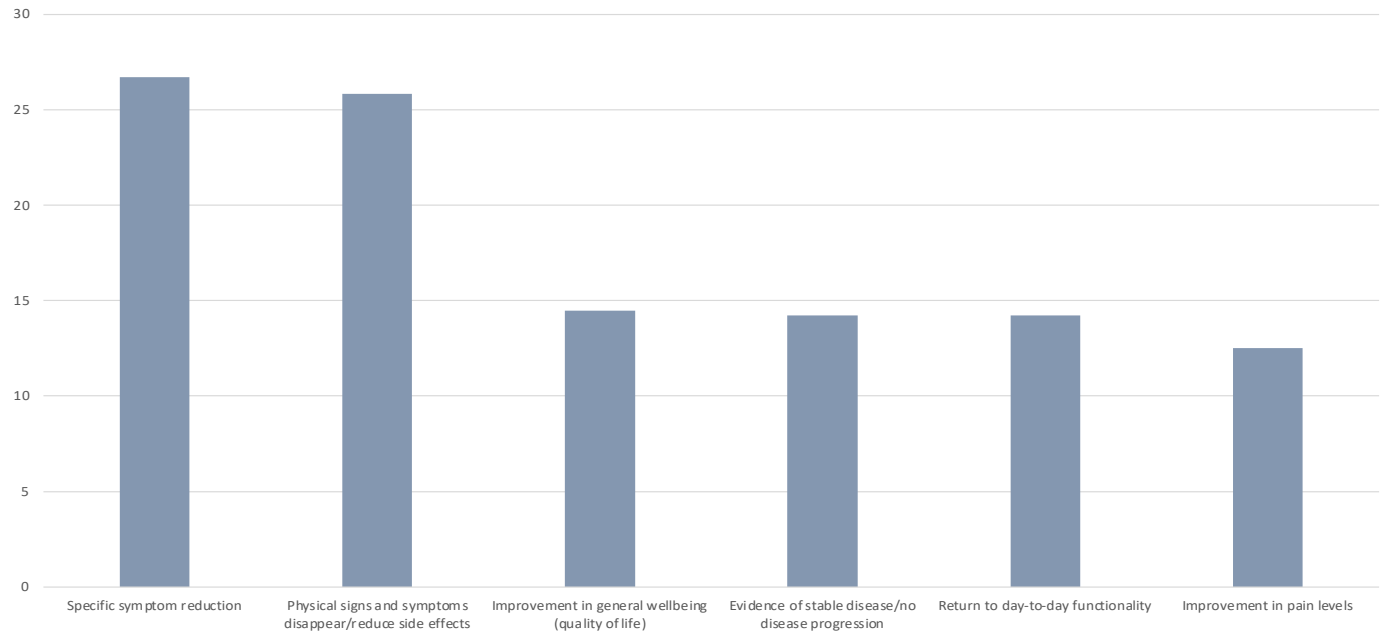


Figure 5.28: What needs to change to feel like treatment is working

Table 5.26: What needs to change to feel like treatment is working – subgroup variations

What needs to change to feel like treatment is working	Reported less frequently	Reported more frequently
Specific symptom reduction	Diseases of the skin	Developmental anomalies Family or carer Aged under 18
Physical signs and symptoms disappear/reduce side effects	Endocrine, nutritional or metabolic diseases	Diseases of the nervous system Diseases of the skin Other rare condition
Improvement in general wellbeing (quality of life)	Other rare condition	Endocrine, nutritional or metabolic diseases
Evidence of stable disease/no disease progression	Developmental anomalies Diseases of the skin	Diseases of the nervous system
Return to day-to-day functionality	Other rare condition	
Improvement in pain levels	Diseases of the skin Other rare condition	Diseases of the immune system

What it would mean if treatment worked

As a follow up question, participants were asked what it would mean to them if the treatment worked in the way they described. The most common responses were that it would allow them to do everyday activities/return to normal life (29.44%) and allow them to engage more with social activities and family life (11.67%). Other themes included allow them to return to work (9.44%), allow them to do more exercise (11.28%), will have a positive impact on their mental health (7.89%), allow them to do domestic tasks (6.77%), and lead to a reduction in symptoms/side effects (5.64%).

Allowing them to do everyday activities/return to normal life

Life. It would be life changing. I have to, I often say to my husband, I would just love to have one day of my life. Just one day. Like between now and death where I don't feel pain 24/7 so that would mean everything. To be able to live without the disease, it would probably one of the biggest breakthroughs I think for me. Like if I have to live with this disease for the rest of my life and I'm assuming I do one of the biggest things that could change my life is GP's understanding what the disease is.
Participant 005_2023AUDSK

If there was something I could take easily that wasn't going to have any other effects on my health that were detrimental in any way, and it just helped with my HS, it would help my life a lot…like I would have the security to be able to go to the beach or go swimming or wear a dress because you have to wear underwear. Just, just normal things like that that I can't do and, you know, feel more confident myself if you were being intimate with somebody. I know mine isn't very bad because nobody in my life has ever noticed. So I guess that's, you know, even in a relationship for three years, no one, no one noticed. So I guess that's something. Other people have it worse than me, but I am aware of it and it's just a conversation you don't want to have to have with somebody. Because it's not like it's an STD, but you just can't help but you lean that way, can you really, if you were uninformed. So yeah, it's always in the back of my head. So yeah, something like that would just make my life a lot easier.
Participant 006_2023AUDSK

Well, I guess because like the, the dizziness and the soreness and things like that, that kind of went away on its own after a few weeks. I think it only stuck around for maybe three weeks, four weeks maximum, and then it kind of settled down a bit. That was not too bad. I feel like I'm kind of OK now. And yeah, the depression, I guess that would be good. I guess it would just help with everyday life, trying to get through work and everything like that. Yeah.
Participant 014_2023AUDSK

Allowing them to engage more with social activities and family life

Look, I cope all right with what I've got at the moment, except when it starts to get quite inflamed and sore and if it becomes embarrassing or I can't go out or something like that, that's difficult from a bit of the social bit as well. And if I can't wear certain clothes it's just…and if I leak on the bed at night, well that's a pain because you've got to go change the sheets and all that stuff. If I go to friends places to stay, I've got to make sure I don't destroy the sheets. If something happens and you just don't know when it's gonna go pop, so. Yeah, you gotta make sure you take enough things away with you and underpants and pads and all that and actually wear your underpants to bed in case you do. But also you gotta make sure you are also get enough air in your bottom area so it doesn't keep doing that. It's one of those hard things so.
Participant 024_2023AUDSK

Well, firstly, it reduces the anxiety around food. You can actually have a life you can like…At the moment, I can't go out for dinner, I can't eat outside of the house. I can, but it's very difficult. Like it takes away the social side of eating and doing those kinds of things. So I guess you can just live a more full, normal life.
Participant 078_2023AUDIS

I'm able to actually have a few hours a day where I can be involved in things. For the last two 2 1/2 years I haven't been able to do pretty much anything. You know, I've been pretty much housebound. You know, where now I can actually get out and go shopping with my wife. And, you know, I'm not worried about having to run to the toilet every 10 minutes and, you know, stuff like that. I just want to be able…I don't want to exist. I want to live.
Participant 014_2023AUORC

I could do a lot. I could go out. I could feel. I could feel happy. So I could, yeah, as I said, there's just days where you just go, no, that's OK. I'll just, yeah, stay

home. I've become a bit of a, I'll just go out to do the grocery shop. I'll go out to do the market. That's it. We might go back to the kids, but yeah, whereas I used to be out all the time. You know, go out some things. But, but yeah, it it changes. You just go now. Not going to risk that because, you know, the first thing I look for is where, where's the closest loo? As soon as I get to a shopping center, use the loo. Yeah. And you know, I can I can empty my gut up to 10 times a day. Yeah. And to me that's not good. That's not healthy.
Participant 019_2023AUDIS

Allowing them to return to work

I could my enjoy my passion and what I've worked at for most of my working life. I'm a carpenter, enjoy my life, but I do like doing joining work. I have a workshop down the church. I can spend more time doing work that I enjoy and making things that are fun. At the moment because I had an accident and fell off of a ladder and broke my heel and aggravated some previous injuries. At the moment, I'm not enjoying any of it. Because as in the pain medications not doing what it's supposed to either. I'm trying to enjoy my enjoy life anyway
Participant 005_2023AUDPA

It would mean so much better. Like I I could go out and get an actual job which I can lift up my arms to reach something.
Participant 004_2023AUDSK

Probably a lot and I'd probably go back to work. [chuckles] I probably wouldn't go back to work because of lung disease now, it's a bit hard to cope. I suppose it's just, just everyday things like I've made adjustments in my own life to cope with it but I suppose just being able to put my hair up by myself, being able to wash my hair by myself. It's just those sort of things, I suppose. I still manage to do my makeup all by myself. I don't wear it often but when I do have to wear it, I can still manage to do it. [chuckles] My sister is always surprised, she's like, 'Who did your makeup? It looks fantastic." I'm like, "I did it myself."
Participant 018_2023AUDIS

It would be amazing. I'd be able to drive all the time, not take time off work. That's a big one. I only started a new job in Feb and I'm already negative like 80 hours sick leave because I've just been in hospital so much. I guess they're the main two for me not having to miss work and being able to drive all the time.
Participant 096_2023AUDNS

Allowing them to do more exercise

Walk further. Being able to breathe while I'm walking and up and down the stairs, in hills is another thing. When I started taking it, I was able to walk further.
Participant 004_2023AUDIS

Walk. I still love walking, you know, I can still walk, but I can't walk much, you know, sort of just getting up, gets out of breath and I have to stop, have a rest and then carry on. Yes, yes. Where I used to be so energetic, really, you know, and I can find my. I strengthen my hands. I've really decreased.
Participant 005_2023AUDIS

Oh, wow. That would be awesome. I'd be able to cook again, I'd be able to just clean and keep the house tidy. Sounds ridiculous, but you see things, and it's frustrating, you can't do anything about it. I would be able to do more exercise. I used to walk the dog every morning, probably for 45 minutes in a brisk walk. Now, I'd say it's more of a stroll than a walk, and I'm out for about 20 minutes. I'd like to be able to do more exercise and swim. I've lost…I can't move my shoulders or arms. I used to enjoy swimming, I go in the water, although [crosstalk] about the chlorine on my skin, so I can't go into those indoor pools. I can't actually swim breaststroke or freestyle or anything, but if [crosstalk] the mobility back, that would definitely be something I'd do.
Participant 017_2023AUDIS

That would mean a lot. If I feel less stiff I can do more stretching, more exercises and if I do more exercises, it'll help me to feel again less stiff than before and I can do more activity, more house chores and be more dependent.
Participant 020_2023AUDIS

A positive impact on their mental health

I would…I'd be working. I would be able to afford dental care. I could improve my mental health, my mental outlook. I could reconnect with my family. I could basically, in inverted commas, get a life. Yeah, I'm just hiding away from the world at the moment. That's what my life is.
Participant 008_2023AUDSK

I think it's, it's high impact because on a really bad day, I can't get off the couch, which means I can't feed my animals. I can't, you know, clean the house. I can't. Even focus on studying or working at that given time. So the less flare ups, the less energy my body has to fight the infections. I would feel much better, think

much clearer and, you know, be able to actually enjoy life because currently it's 5050, yeah.
Participant 026_2023AUDSK

Well, if this, if it was something like that, it would mean that he would be able to better express his needs and have his have his needs met better, probably reduce overall stress for him and everybody, and reduce his frustration. Yeah, it it would just mean that he would be able to function better and achieve more of his desires and more of what he wants, yeah.
Participant 089_2023AUENM

I guess it would just be, I mean it's psychologically for us it's just difficult seeing your child in pain and struggling because of medication that they're on to try and treat the condition that they there's no cure for. So if we're able to. If there was some other way around it or something that we could have to get rid of those unwanted side effects, then for us it would be just a major relief. You know, no parents would see their kid in pain or anything. So yeah, it's just be. It wouldn't change, I guess it wouldn't change what he can and can't do because he can't do much at the moment anyway. But it would just be a relief for us to see him more settled and happy.
Participant 029_2023AUORC

Allowing them to do domestic tasks

Just everything. If my pain's flared up, or my backs out or something like that, then I can't even unload the dishwasher, or I can't comfortably get my kids ready for bed, can't bath them. There's so many slow on effects from that type of thing. Often we push through pain so often that you do get used to it. I do notice that it will affect my mood, I'll be more irritable or more tired or I won't want to go out. I'm not going to book-- seeing my friends in my spare time. I'll just be resting. Again, that functional impact is the big one. It's day-to-day things.
Participant 004_2023AUDPA

I'd be able to go for walks again and be able to actually play with my kids properly clean the house. Stack the dishwasher. I can't even stack the bloody dishwasher at the moment. Be able to go horse riding again and and do the activities and and go back to competition. So like, I haven't been able to do that since I had the really bad flair in 2019. That's just gone from spot to spot to spot.
Participant 012_2023AUDSK

I can, you know, go back to doing, you know, cooking and baking. I can, you know, better help my parents with, you know, various gardening activities and things like that. I stopped. I stopped some of the voluntary work that I was doing before, and, you know, being able to pick that up again would be lovely. Having said that, I've found other ways to volunteer, obviously, so.
Participant 019_2023AUORC

About 100 things like work, have my 16 year old son live with me because I could care for him. I could do all his washing, you know, domestic chores and cooking. It would mean I could go back to doing all the sport I would normally do. It would mean I could see all my friends, so I could be social because I would have enough energy to do that. It would mean I could drive longer distances, which also would mean I could be more social because I could, you know, drive to see friends. That would also mean I have greater independence. Therefore I would have greater I would have income because at the moment I'm on a pension because I can't work. So it would change my life enormously. To increase the quality of life enormously.
Participant 010_2023AUDIS

Leading to a reduction in symptoms/side effects

More energy and you know, a knowledge that you know the medication has worked and that, what would you…it's going to give me more energy. I won't have brain fog, I won't have aching limbs. I won't have night sweats. I'm, you know, I'm going to be a much more productive member of society and to my family.
Participant 010_2023AUORC

It would mean a lot. It's, it's something that I deal with daily and kind of dealing with the pain. So it would allow me to just sort of not have to think about it. It would be great, yeah.
Participant 027_2023AUDSK

Look. If the treatment wasn't doing its job, you know all three in my life, I went to work pretty well, but in pain. So I did. I went to work, I went shopping…I, you know, I did a normal life, but I had a pain level that upset me. But it was also the social thing as well, so I sometimes, if I was looking bad, I wouldn't go out. So that that that wasn't because I couldn't go out. It was just because I made the choice that I didn't want people to see me. Yeah, and there was sometimes I didn't accept social events because I wasn't looking good, but I could have gone. It's just that I made the the choice but it was more more about pain for me like.
Participant 029_2023AUDSK

I'll be able to shower easy. The shortness of breath is the most annoying thing ever because I have to constantly stop. I got to remind myself to stop, to breathe. People look at you and think, "oh God, she's unfit." Well, I'm not. It's just I can't breathe, like my lungs are destroyed.
Participant 023_2023AUDIS

Table 5.27: What it would mean if treatment worked

What it would mean if treatment worked	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=180	%	n=67	%	n=31	%	n=9	%	n=32	%	n=9	%	n=32	%	n=100	%	n=80	%	n=121	%	n=57	%
Allowing them to do everyday activities/return to normal life	53	29.44	3	4.48	17	54.84	1	11.11	15	46.88	4	44.44	13	40.63	42	42.00	11	13.75	42	34.71	10	17.54
Allowing them to engage more with social activities and family life	21	11.67	2	2.99	7	22.58	0	0.00	8	25.00	0	0.00	4	12.50	19	19.00	2	2.50	18	14.88	2	3.51
Allowing them to return to work	17	9.44	1	1.49	4	12.90	0	0.00	6	18.75	1	11.11	5	15.63	15	15.00	2	2.50	10	8.26	7	12.28
Allowing them to do more exercise	16	8.89	0	0.00	8	25.81	0	0.00	2	6.25	1	11.11	5	15.63	10	10.00	6	7.50	13	10.74	3	5.26
A positive impact on their mental health	12	6.67	1	1.49	4	12.90	1	11.11	5	15.63	0	0.00	1	3.13	12	12.00	0	0.00	9	7.44	3	5.26

What it would mean if treatment worked	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=180	%	n=61	%	n=63	%	n=37	%	n=19	%	n=88	%	n=92	%	n=48	%	n=132	%	n=94	%	n=86	%
Allowing them to do everyday activities/return to normal life	53	29.44	10	16.39	18	28.57	15	40.54	10	52.63	24	27.27	29	31.52	14	29.17	39	29.55	28	29.79	25	29.07
Allowing them to engage more with social activities and family life	21	11.67	2	3.28	6	9.52	9	24.32	4	21.05	8	9.09	13	14.13	7	14.58	14	10.61	14	14.89	7	8.14
Allowing them to return to work	17	9.44	2	3.28	7	11.11	6	16.22	2	10.53	7	7.95	10	10.87	1	2.08	16	12.12	6	6.38	11	12.79
Allowing them to do more exercise	16	8.89	5	8.20	4	6.35	3	8.11	4	21.05	6	6.82	10	10.87	2	4.17	14	10.61	6	6.38	10	11.63
A positive impact on their mental health	12	6.67	0	0.00	3	4.76	8	21.62	1	5.26	6	6.82	6	6.52	0	0.00	12	9.09	7	7.45	5	5.81

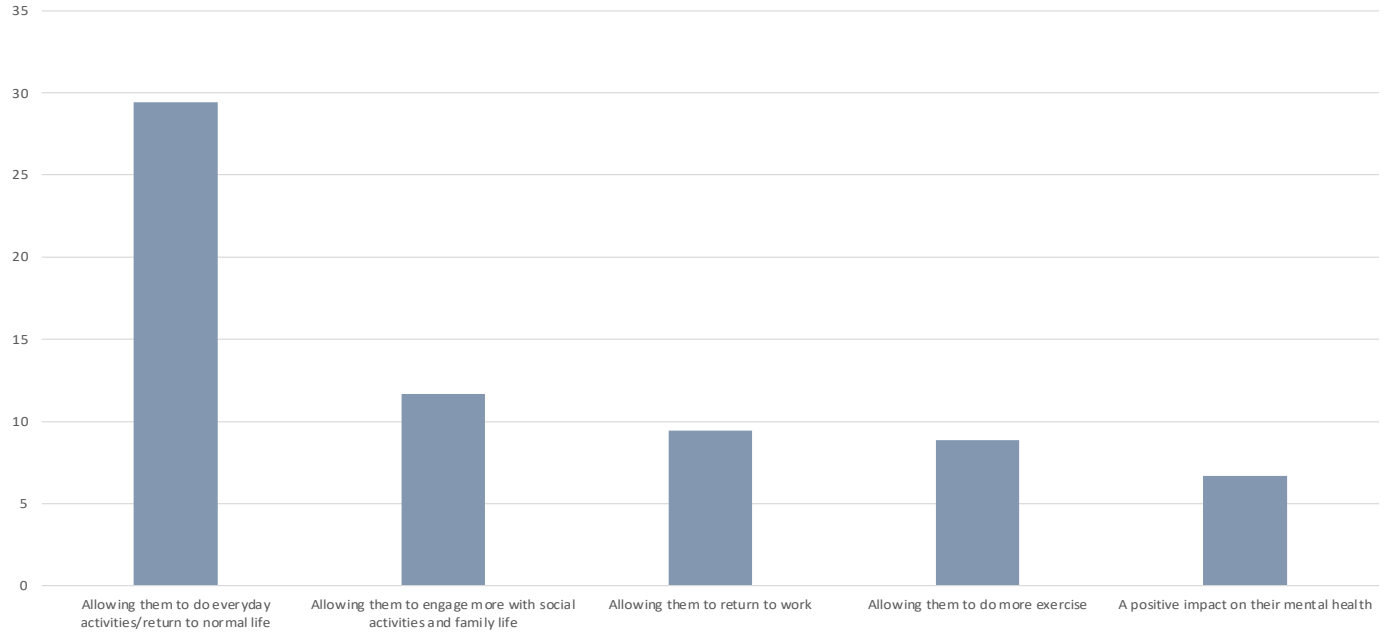


Figure 5.29: What it would mean if treatment worked

Table 5.28: What it would mean if treatment worked – subgroup variations

What it would mean if treatment worked	Reported less frequently	Reported more frequently
Allowing them to do everyday activities/return to normal life	Developmental anomalies Diseases of the nervous system Family or carer Male Aged under 18	Diseases of the immune system Diseases of the skin Endocrine, nutritional or metabolic diseases Other rare condition Person with condition Aged 45 to 64 Aged 65 plus
Allowing them to engage more with social activities and family life	Diseases of the nervous system Endocrine, nutritional or metabolic diseases	Diseases of the immune system Diseases of the skin Aged 45 to 64
Allowing them to return to work		Aged 45 to 64
Allowing them to do more exercise		Diseases of the immune system Aged 65 plus
A positive impact on their mental health		

Section 6

Information and communication

Section 6: Information and communication

Access to information

In the structured interview, participants were asked what information they had been able to access since they were diagnosed. The most common responses were the internet (Including health charities) (59.45%), from a specific health charity (32.34%) and from Facebook and\or social media (26.12%). Other themes included their treating clinician (25.62%), from journals (research articles) (22.89%), from other patient's experience (Including support groups) (18.41%), from books, pamphlets and newsletters (14.68%).

Information that was helpful

In the structured interview, participants were asked to describe what information they had found to be most helpful. The most common responses were other people's experiences (26.37%), health charity information (16.67%), hearing what to expect (e.g. from disease, side effects, treatment) (15.92%), and talking to a doctor or specialist or healthcare team (15.92%). Other themes included medical or scientific sources (11.19%), and information on triggers and managing exacerbations (6.97%).

Information that was not helpful

In the structured interview, participants were asked if there had been any information that they did not find to be helpful. The most common response was that there was no information that was not helpful (31.09%). The most common types of unhelpful information included information from their GP or specialist (11.94%), sources that are not credible (10.20%), other people's experiences (9.20 %), information that was not type specific or too general (8.46%). Other themes included a lack of new information (7.46%) and worse case scenarios (7.46%).

Information preferences

Participants were asked whether they had a preference for information online, talking to someone, in written (booklet) form or through a phone App. The most common responses were online information (29.35%), talking to someone plus online information (23.63%), and talking to someone (21.64 %). Other themes included written information (13.68%), all forms (5.47%), and apps (2.49%).

The main reasons for a preference for online information were accessibility (27.86%) and being able to digest information at their own pace (18.41%).

The main reasons for a preference for talking to someone was being able to have time to ask questions (18.41%), and that it was personalised (14.43%). The main reason for a preference for written information were written information is that they can refer back to/highlight important information (3.23%).

Timing of information

Participants in the structured interview were asked to reflect on their experience and to describe when they felt they were most receptive to receiving information. The most common times were at the beginning (diagnosis) (31.34%), continuously (19.65%), after the shock of diagnosis (12.44%) and 12 months or more after diagnosis (10.70 %).

Healthcare professional communication

Participants were asked to describe the communication that they had had with health professionals throughout their experience. The most common theme was that participants described having an overall negative (34.83%), overall positive (26.62%), and overall positive, with the exception of one or two occasions (24.63%).

Partners in health

The Partners in Health questionnaire (PIH) measures an individual's knowledge and confidence for managing their own health. The Partners in Health comprises a global score, 4 scales; knowledge, coping, recognition and treatment of symptoms, adherence to treatment and total score. A higher score denotes a better understanding and knowledge of disease.

The overall scores for the cohort were in the highest quintile for Partners in health: Knowledge (median=26.00, IQR=8.00), Partners in health: Adherence to treatment (median=14.00, IQR=4.00), indicating very good knowledge, very good adherence to treatment.

The overall scores for the cohort were in the second highest quintile for Partners in health:Recognition and management of symptoms (median=19.00, IQR=5.75), Partners in health:Total score (median=72.00, IQR=20.00) indicating good recognition and management of symptoms, good overall ability to manage their health.

The overall scores for the cohort were in the middle quintile for Partners in health:Coping (median=14.00, IQR=7.00), indicating moderate coping.

Ability to take medicine as prescribed

Participants were asked about their ability to take medicines as prescribed. The majority of the participants responded that they took medicine as prescribed all the time (n=173, 57.10%), and 120 participants (39.60%) responded that they took medicines as prescribed most of the time. There were 6 participants (1.98%) that sometimes took medicines as prescribed.

Information given by health professionals

Participants were asked about what type of information they were given by healthcare professionals, information about treatment options (n=188, 58.02%), disease management (n=147, 45.37%), disease cause (n=119, 36.73%) and, physical activity (n=85, 26.23%) were most frequently given to participants by healthcare professionals, and, information about interpret test results (n=54, 16.67%), clinical trials (n=43, 13.27%) and, complementary therapies (n=34, 10.49%) were given least often.

Information searched independently

Participants were then asked after receiving information from healthcare professionals, what information did they need to search for independently. The topics participants most often searched for were disease management (n=212, 65.43%), treatment options (n=210, 64.81%), disease cause (n=207, 63.89%) and, complementary therapies (n=167, 51.54%) were most frequently given to participants by healthcare professionals, and, information about clinical trials (n=123, 37.96%), interpret test results (n=120, 37.04%) and, hereditary considerations (n=103, 31.79%) were searched for least often.

Information gaps

The largest gaps in information, where information was neither given to patients nor searched for independently were clinical trials (n=177, 54.63%) and interpret test results (n=172, 53.09%).

The topics that participants did not search for independently after not receiving information from healthcare professionals were treatment options (n=66, 20.37%) and disease cause (n=58, 17.90%).

The topics that participants were given most information from both healthcare professionals and searching independently for were disease cause (n=146, 45.06%) and complementary therapies (n=145, 44.75%).

The topics that participants searched for independently after not receiving information from healthcare professionals were treatment options (n=122, 37.65%) and disease management (n=96, 29.63%).

Access to information

In the structured interview, participants were asked what information they had been able to access since they were diagnosed. The most common responses were the internet (Including health charities) (59.45%), from a specific health charity (32.34%) and from Facebook and\or social media (26.12%). Other themes included their treating clinician (25.62%), from journals (research articles) (22.89%), from other patient's experience (Including support groups) (18.41%), from books, pamphlets and newsletters (14.68%).

Participant describes accessing information through the internet in general

Mainly Google. I sought out possible causes. I sought out whether vaccines have anything to do with it. That's just recent. I sought out diets that might be good. That's just recent too.
Participant 060_2023AUDNS

The only information I've got initially is just about the CHARGE syndrome. Well, all of it really I've just got off the Internet.
Participant 09_2023AUDPA

The Internet. The American Natural Library of Medicine has a lot of information. Mayo Clinic has information not so many Australian websites.
Participant 003_2023AUDNS

Participant describes accessing information primarily through Facebook and/or social media

The biggest one was to get onto the support page…on Facebook with the registered nurses and just listening to everyone else, and seeing what everyone else was going through, that's where I got all the information from.
Participant 067_2023AUDNS

Yep. So CHARGE online community Facebook groups. And the CHARGE of Australasia is fairly active. They have like regular conferences and courses and that kind of thing. There's also a fairly big online like social media presence these days, not so much when PATIENT was a baby, but these days it's a bit more active and otherwise, just like mining through medical journals, really more in the early days, but yeah. These days I don't tend to freak myself out.
Participant 018_2023AUDPA

Participant describes accessing information from a specific health charity

The AMDF has been very good. They've produce booklets, and of course there's what's online as well.
053_2023AUENM

Well from the Scleroderma, NSW and Australia. I'm actually a support person for Scleroderma Australia in my area in, I mean, I don't have meetings because we tried to get together. There was five of us from a range, from a long area like you know, it was over 200 kilometers. There was only five of us in that area. And it was just too hard for everyone to get together because someone was always sick or couldn't do it. And it cancelled. And so I said to the I won't be doing the meetings like, but I'll be, I could be in the book as a support person and I will post out any brochures, which I do if anyone gives me a call.
Participant 088_2023AUENM

The 22 Q Australia Foundation is awesome. NAME is a wealth of information. We've also got a local WA group which has been amazing. But the end yeah, there's also there's international groups which have got a wealth of information as well. Education's a big one because she learns differently, and it's been really helpful to have that sort of information for her teachers.
Participant 021_2023AUDPA

Participant describes primarily accessing information through treating clinician

Well, a bulk of the information came from my personal research. And the other part of it came from my doctor, you know, most of the information I got from Google or random search and goggle and you know, I actually to read through people's experience, you know, to get us some clue and knowledge about what the thing is all about. So it's was mostly largely from the Internet. I got an idea of what this is.
Participant 006_2023AUORC

Yeah, sure. So I guess we sort of tried to speak to the various doctors that are involved in her care. So the genetic doctor plus our pediatrician plus the GP just to find out information, but also doing things like looking on the Internet. Looking up journal articles and also looking on the various social media support groups that have out there for that condition and also talking to a couple of sort of like patient support or advocacy groups that look into these sort of rare syndromes or

genetic conditions. And also got in contact with a researcher from somewhere in NSW who had an interest in the issue.
Participant 022_2023AUORC

Obviously the Internet is a major source of information, but when we first found out and went to the hospital, the hospital provided us with an in depth information booklet regarding the condition which was really helpful. And then the other main source of information for us is being his specialist team. Every time we talk to them, we have a list of questions and they answer them all for us. They're very thorough, yes. So that's been the main source, his care team and information.
Participant 029_2023AUORC

Lots of Google, pretty much everything we can find our hands on the Internet. We've joined sort of support groups online like on Facebooks and have gotten information from actual parents with experience that probably where we get a lot of our information from because there isn't a lot of information about the duplication online. So when we sort of done our own research, it's been there or it's with the specialists that we have a really close relationship that we can just sort of bounce ideas of one another.
Participant 032_2023AUDPA

Participant describes accessing information primarily through journals (research articles)

As much as I could from 2003. I began the database what was published on the disease up until up until the end of 2021. I was pretty update on everything that was published and I'm just not behind now. This is a lot that's being published now. I still have my own databases published stuff, so the stuff I look at is what's published in peer reviewed journals. Yeah, yeah.
Participant 008_2023AUDSK

Probably the biggest source has been online, so just searching and then the CHARGE Foundation in the US website journals again found them online though medical journal articles and mums and dads who have children. With charge or adults with charge syndrome that they care for is often the biggest access for information or strategies and all that kind of stuff, yes.
Participant 095_2023AUENM

Lots, lots and lots. So initial Google searches and sort of got logins and paid for subscriptions for medical journals where it's mentioned and talked about done the research on what the specific, I guess the variants are of it and your side effects for children and yeah there's lots of things.
Participant 020_2023AUDPA

So for me it's medical journals and stuff like that. I don't like doctor Google. It has to be like a peer reviewed thing or yeah, information that's credible. I've talked a lot with my GP and it's actually really interesting too, because my physio has never had anyone with POTS, but she's really into it now, so she's doing a lot of study into POTS as well. So we have quite good conversations about it, but sort of that's where it ends I suppose.
Participant 031_2023AUORC

Participant describes primarily accessing information through other patient's experience

So we, I go straight to the doctors first and foremost to get any information I need. Then I, I check on like the the medical databases for the university, so like bring up journal articles. I also every now and then I'll jump onto a forum just to get like the real world experience of people that have tried it. So I I kind of like to get a a full review of of treatments and ideas and and suggestions. To get all my information from.
Participant 025_2023AUORC

A lot from support groups, scleroderma support group. I actually was helping run the one here in CITY at one point, so that's been a major thing. Facebook, we got another support group on Facebook, a bit off the doctor's, GP. There was one at LOCATION where the group was, there was a rheumatologist there and she has a wealth of information. Bits and pieces, a lot of it's from other people.
Participant 007_2023AUDIS

I guess I just googled it and yeah, got the information off the Internet and the support group that we I don't always go every month. Just depends on what you know how I'm feeling and what I'm up to and they all know. They all know that as well and we just sort of it's just a best friendship and and all of that. So it's you know and you know someone says all they tried this will you know try that you know sort of creams and stuff like that or whatever.
Participant 019_2023AUDIS

Participant describes receiving information from books, pamphlets and newsletters

I try and look at the Scleroderma Association. Obviously, the Australian one, the US one, and the UK one predominantly, as the strong English-speaking, and look at things that are on their newsletters, I get information about different trials and things coming up. I am on a number of Facebook groups, which can be good, but I think you have to be careful how much of that you read. That can be really, really depressing. Really, some people are carrying on, and you think, "Oh my gosh, your symptoms are so mild, you have no idea." Other people, it's just so shocking and awful, some of the stuff they're going through.
Participant 017_2023AUDIS

Yeah, the pediatrician gave us pamphlets to start with like a, you know, medical information. Since then that was pretty much all I used. Since then I've also bought myself resources for 22 Q in education and learning to see how that's how they work and what's the best way to teach them. I've also now joined the 22 Q Australian New Zealand group so I get information from them as well and that's about as far as I've gone so far.
Participant 010_2023AUDPA

Okay, there are printed pamphlets. I know when I get my Botox. It's a public clinic that's St Vincent, and there are Dystonia pamphlets there, and I found them pretty light on, but they're good to give to family and friends to give them an idea. I have scraped the Internet, reading stuff, and I'm also trying to get into the NDIS, and that's been a huge scrape of the Internet. So I feel fairly well-informed, but there's nothing there that's very encouraging.
Participant 002_2023AUDNS

Participant describes accessing information from clinicians and researchers (including webinars/seminars/conferences)

After I actually had the surgery and for the first time I had a few weeks at home, I, I reached out and started sort of finding more, I suppose patient, not support groups, but information sites and and there you would there was a there was one, I think it's called my HS, where they actually then also ran webinars and information sessions hosted by different dermatologists and practitioners and and lived experienced people mainly from the state. And so I was able to link in with some of those to hear about other people's experiences and then I, yeah, joined a couple of Facebook groups. In which did sort of hear about people lived experience and and what they were trying. Sometimes they had different sort of suggestions for things like lotions and creams and stuff that the dermatologists hadn't come up with. So there, yeah, there were a couple of times I tried some of those things but on a minor scale, whereas some of the suggestions was pretty out there.
Participant 007_2023AUDSK

So there's a lot of really good resources for cystic fibrosis, like they've got the cystic fibrosis, fibrosis Australia and then all the states have their own little subsidiary branches of it that really help with connecting you with that information. We also get a lot from the clinic itself. And they have a lot of events quite regularly where I can't remember what they call them, but pretty much they're like open community forums, but they actually get the doctors that are out there, you know, doing all the research. They get families in to just have a talk and let everyone know what the the latest information available out there is, what's happening.
Participant 020_2023AUORC

Table 6.1: Access to information.

Access to information	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Internet (Including health charities)	239	59.45	24	35.82	60	74.07	62	65.26	21	65.63	54	56.84	18	56.25	180	67.16	59	44.03	179	60.88	59	55.66
Specific health charity	130	32.34	20	29.85	22	27.16	26	27.37	4	12.50	45	47.37	13	40.63	87	32.46	43	32.09	97	32.99	32	30.19
Facebook and\or social media	119	29.60	12	17.91	35	43.21	31	32.63	16	50.00	18	18.95	7	21.88	89	33.21	30	22.39	93	31.63	26	24.53
Treating clinician	105	26.12	14	20.90	22	27.16	20	21.05	6	18.75	24	25.26	19	59.38	69	25.75	36	26.87	82	27.89	23	21.70
Journals (research articles)	92	22.89	13	19.40	14	17.28	24	25.26	12	37.50	21	22.11	8	25.00	59	22.01	33	24.63	73	24.83	18	16.98
Other patient's experience (Including support groups)	74	18.41	15	22.39	18	22.22	10	10.53	12	37.50	8	8.42	11	34.38	53	19.78	21	15.67	58	19.73	16	15.09
Books, pamphlets and newsletters	59	14.68	14	20.90	20	24.69	3	3.16	0	0.00	16	16.84	6	18.75	42	15.67	17	12.69	41	13.95	16	15.09

Access to information	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Internet (Including health charities)	239	59.45	24	35.82	60	74.07	62	65.26	21	65.63	54	56.84	18	56.25	180	67.16	59	44.03	179	60.88	59	55.66
Specific health charity	130	32.34	20	29.85	22	27.16	26	27.37	4	12.50	45	47.37	13	40.63	87	32.46	43	32.09	97	32.99	32	30.19
Facebook and\or social media	119	29.60	12	17.91	35	43.21	31	32.63	16	50.00	18	18.95	7	21.88	89	33.21	30	22.39	93	31.63	26	24.53
Treating clinician	105	26.12	14	20.90	22	27.16	20	21.05	6	18.75	24	25.26	19	59.38	69	25.75	36	26.87	82	27.89	23	21.70
Journals (research articles)	92	22.89	13	19.40	14	17.28	24	25.26	12	37.50	21	22.11	8	25.00	59	22.01	33	24.63	73	24.83	18	16.98
Other patient's experience (Including support groups)	74	18.41	15	22.39	18	22.22	10	10.53	12	37.50	8	8.42	11	34.38	53	19.78	21	15.67	58	19.73	16	15.09
Books, pamphlets and newsletters	59	14.68	14	20.90	20	24.69	3	3.16	0	0.00	16	16.84	6	18.75	42	15.67	17	12.69	41	13.95	16	15.09

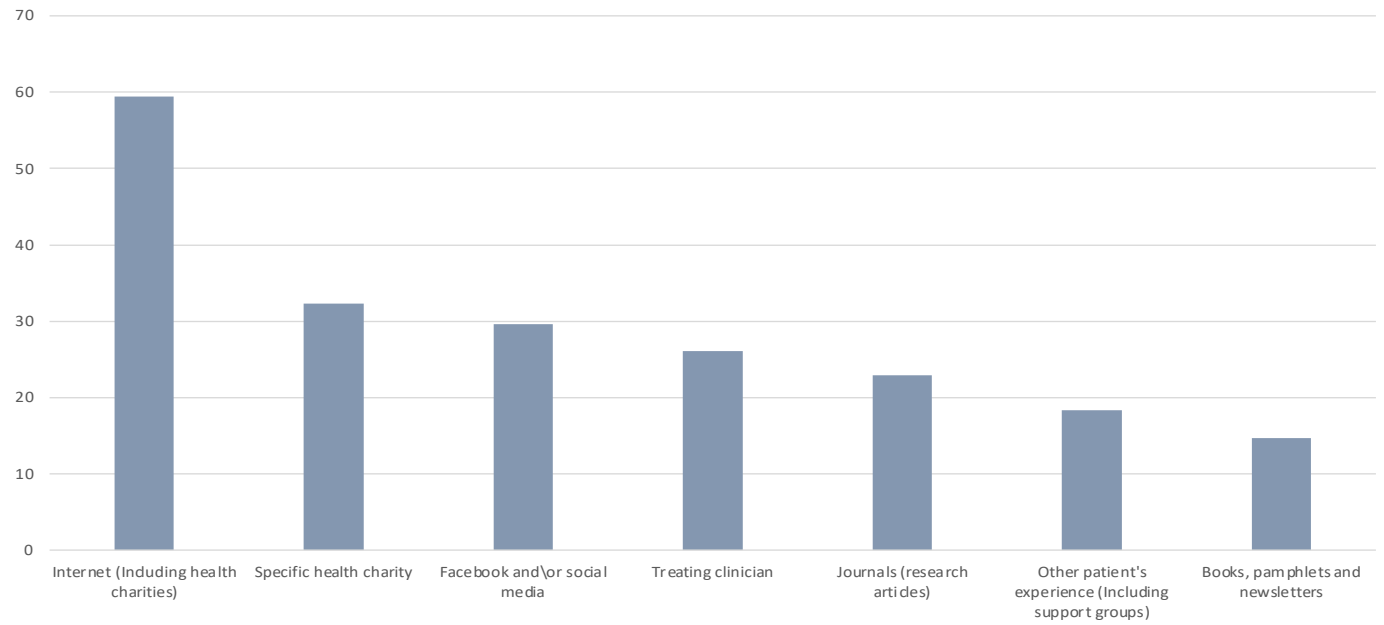


Figure 6.1: Access to information

Table 6.2: Access to information – subgroup variations

Access to information	Reported less frequently	Reported more frequently
Internet (Including health charities)	Developmental anomalies Family or carer Aged under 18	Diseases of the immune system
Specific health charity	Diseases of the skin	Endocrine, nutritional or metabolic diseases
Facebook and\or social media	Developmental anomalies Endocrine, nutritional or metabolic diseases Aged 65 plus	Diseases of the immune system Diseases of the skin
Treating clinician		Other rare condition
Journals (research articles)		Diseases of the skin
Other patient's experience (Including support groups)		Diseases of the skin Other rare condition
Books, pamphlets and newsletters	Diseases of the nervous system Diseases of the skin	Diseases of the immune system Aged 65 plus

Information that was helpful

In the structured interview, participants were asked to describe what information they had found to be most helpful. The most common responses were other people's experiences (26.37%), health charity information (16.67%), hearing what to expect (e.g. from disease, side effects, treatment) (15.92%), and talking to a doctor or specialist or healthcare team (15.92%). Other themes included medical or scientific sources (11.19%), and information on triggers and managing exacerbations (6.97%).

Participant describes other people's experiences as helpful (Peer-to-peer)

Yeah, made me feel I'm not alone. That was quite good because they said, they said most people don't talk about it, they just tend to hide it.
Participant 024_2023AUDSK

I suppose being in contact with the other parents and and finding out what's worked for them. And it was very interesting when we found out about. The, the gene that was probably responsible for most of the symptoms.
Participant 093_2023AUENM

Yeah, yeah. They're the people who know, you know it. I mean, you say 22 Q, the average person, they haven't got a clue and that's the most common genetic syndrome after Down syndrome. Yeah, when we're educating doctors, it's not a good thing. When you go and see a specialist and they Google 22 Q in front of you, it's not a good thing.
Participant 021_2023AUDPA

Going to the conferences is really good because you meet other families there as well as the kids, and they're all different ages. And yeah, so it's good to see how people are going.
Participant 026_2023AUDPA

Participant describes hearing what to expect (e.g. from disease, side effects, treatment) as being helpful

And I guess myself one has been just finding out about some of the potential symptoms and what the prognosis might be moving forward in terms of how people display, how they demonstrate the condition in labor life, what sort of symptoms have been.
Participant 022_2023AUORC

Just knowing what the hell scleroderma was and why the symptoms were what they were, very useful, worst case scenarios and best case scenarios and all that kind of thing and I said the wound stuff was very useful and more recently, information around disability. I've found that an interesting transition for me is now I am someone with a disability and the permission to be that person. Working with NDIS and disability support people and all that has been really useful and has made a big difference more recently. I'm someone with scleroderma but I am someone with a disability and my disability is caused by scleroderma as opposed to I'm someone with scleroderma if that makes sense.
Participant 026_2023AUDIS

What what sort of things that you see with a person with DiGeorge like the learning difficulties and the thought processes and yeah, understanding all that and seeing that that's very clearly what's happening with my daughter. So symptoms, I guess symptoms and examples of what you you will see and expect as normal and to not get frustrated because that person is doing their best. Yeah.
Participant 08_2023AUDPA

Participant describes talking to their doctor or specialist as helpful

I guess the information from the doctor from the specialist has been the most helpful because he's told me what to expect and everything like that, and I trust him. When he's told me this could happen, that could happen, that's been helpful in helping me understand what my body's going through and help me cope with the changes if that makes sense.
Participant 39_2023AUDIS

I think speaking directly to the the doctors or the specialists, so obviously there's there's a lot out there on types of nebulizers that are the best and what people have brought and have used for them, but I tend to always find what I speak directly to the doctors about. Is most helpful I suppose, cuz I can ask a question and have it answered, or they look into it specifically and get back to me instead of just looking at the general frequently asked sections.
Participant 025_2023AUORC

Receiving the information, what was the most helpful? I suppose some of the things that the neurologist just said to us where she has actually probably had some clients that she has seen that are in their teens and early 20s. Well, I guess helpful probably would be more that it's being more easing on our minds, that's positive information from her that she's had clients that are older than NAME, in their late teens and doing really well and they've continued to thrive regardless of having Leigh's disease.
Participant 066_2023AUENM

Participant describes health charities information as helpful

I would say the information located on the Hep Queensland website, I think it's a great overview and you can pass that to people that. Need to know, I need to teach themselves about it, because talking to potential partners about it, for some people it's a very big deal and it's it's a huge deal for them. For other people, it hasn't been a big deal. They've just gotten blood tests and checked their immunity, so. It's really interesting. I guess the stigma from that. I thought that maybe COVID would have helped that, but I think other people are more germaphobic than others, yeah.
Participant 004_2023AUORC

Yeah, so I contacted the CHARGE foundation and they sent a lot of helpful links to webinars. From the

leading experts on this field. A few journal articles, yeah, things like that.
Participant 094_2023AUENM

Oh look, I certainly don't want to discredit the information and the conversations that I'll have with my neurologist. I value those greatly because, I trust, that he's always looking for, the newest things available. I trust that he does keep me up-to-date on what's available out there, but from the support side of it, the Dystonia Network of Australia is just fantastic.
Participant 006_2023AUDNS

Participant describes information about triggers and managing or avoiding exacerbations

I guess the most helpful thing was I went to a workshop that the autoimmune resource centre ran on, it was health and looking after yourself and it was I guess it wasn't new information but it was just looking at things from a different perspective. It was about all the different symptoms that you can have and what different ways, I guess, you should go about tackling them. I guess I didn't really learn anything new because I'd already looked up a lot of my own symptoms.
Participant 31_2023AUDIS

More management plans. Knowing about the different types…what are the effects, whether is a one-off thing, whether it's relapsing form and management plan. What sensation will Residual symptoms, mainly residual symptoms because I need to work out whether is it residual symptoms or is it a relapse or whether I need to go to hospital. Actually, it's that kind of thing that sort of help me.
Participant 059_2023AUDNS

Participant describes no particular information being especially helpful

Not, no it's all very dismal prognosis. Very, very negative. What I can say as well, very, very frighteningly, is anything definitive about what your symptoms are or will be in the future? Do they get worse? What I mean is this deteriorating condition, it's gotten worse over the years. How much worse is it going to get and is it going to affect me so? That's it.
Participant 015_2023AUDSK

None.
Participant 006_2023AUDIS

I really do not have the answer to this question. That is probably my biggest problem with them, my condition is, I have to manage it myself and then try to seek out people with qualifications to help me. Then I find that I get nowhere. That probably the answer to one of your very first questions, that's probably my biggest thing. I need to have a team around me and I just don't have that.
Participant 014_2023AUDIS

Participant describes information specific to their condition (and sub-types) as helpful

After nine years of wondering where she fitted on the umbrella. It was it was comforting to at least know where she sat with her genetic diagnosis and that yes, we're doing what we can. Yes, we're following the guidelines. And it was, you know, great to know that there's someone else in the world with the same thing. It wasn't just us. So that was good for me. It was, I've wondered for five years whether I was more like NAME and why I wasn't responding to treatment and things like that. And I'm a bit annoyed that, you know, I wasn't offered this ten years ago and, you know, just a little things like that that that at least let her know, you know, what is the problem and why she is the way she. But we do have other genes that came up in the sequencing, mutations and they're not sure where they fit either. So, you know, there's still more science to come, yeah.
Participant 080_2023AUDIS

Yes. It's all helpful in different ways. Some of the information-- What's the information that's been-- I'm going to talk about types of information, I think. Because my manifestation is quite severe now, I mostly appreciate the information that incorporates that reality in its paradigm. I don't know how else to put it, so a lot of the time you'll come across something about movement in hypermobility and it's just really important to keep moving or do this, do this, do this. That information wasn't really very useful to me because too much of the wrong movement is actually just as bad if not more detrimental than no movement. So I like specific detailed stuff that, the difficulty there is that I don't have the cognitive capacity to read like whole papers and things, but in general, accessing all of that stuff was really important just in terms of getting a handle on it, not feeling alone, understanding that there are ways of managing all of that stuff, but really the most useful information that's come to me is what I get from my physiotherapist about how to manage my body.
Participant 041_2023AUDPA

Participant describes information from medical or scientific sources as helpful

When I looked up articles on what certain things did for my lungs, like some of the physio stuff, I…so I don't know why I didn't take it as seriously when they said, you know, it works and whatnot. And I just thought you just told me that because you want me to do it. I went and read articles about it and, you know, it was had facts behind it from people that had, you know, had scholarly stuff behind it. So I was like, oh, okay, well, yeah, that makes sense.
Participant 013_2023AUORC

Yeah, yeah. Medical, medical articles and having been researching medical things for so many years, I'm pretty well up on terms. And occasionally I'll stumble across a word I've never heard before. But not very often.
Participant 003_2023AUDIS

I think the National Library of Medicine. PubMed. Whether I would, especially when they'll give an abstract of an article, because I'm not particularly interested in waiting my way all the way through, but they will give an abstract of a scientific article. Like I was trying to work out whether coffee was a problem and some sites said don't drink coffee. So I found a site that said really just work it out for you because some people can't. Because I went on to decaf for a week, thought, you know, would for instance, would decaf coffee make a difference? No, it didn't.
Participant 003_2023AUDNS

Participant describes information about treatment options as helpful

I guess maybe the medication, the information about the medication on how it can like control the blood. Yeah, we don't really have much information.
Participant 003_2023AUORC

You know, there's definitely the the cleaning, you know, using the antibacterial stuff as much you know that's and then obviously new treatments, you know, like finding out people talking about what they're on and how good it's been and, you know, like I didn't know anything about this sort of stuff, so and I told them I was telling my GP. So yeah, not very good, yeah.
Participant 025_2023AUDSK

Participant describes information from international sources as being helpful

It's helped me in understanding part of what the issues are. The best bit of information I've actually found is I've been listening to EDS, I think it's the American version, and I actually have a monthly discussion/info program that's basically a TED Talk type where they're talking about various parts of various types of EDS and talking about what works and what doesn't work is what I've actually found most helpful. Dr. Google, not a good place to start.
Participant 005_2023AUDPA

Articles from the Mayo Clinic in America and Johns Hopkins, and a little bit from the Dystonia Support Network.
Participant 007_2023AUDNS

Participant describes information in lay language or that is easy to understand as helpful

PARTICIPANT The information can get through the UK Association website. They've got very easy-to-read one. They've been amazing. The forum has been helpful. Sorry, I forgot the question. It was about what?
INTERVIEWER Just when you've received information, what information has been most helpful?
PARTICIPANT Those sorts of things. The handouts break down what the issue is and what to do about it. They're quite clear and easy to use and easy to hand on to others if I've needed to.
Participant 004_2023AUDPA

I just wanted worded in layman terms, just just more about where the future studies are going how it's leaped forward since when I was first diagnosed to what's on offer now. There's heaps of clinical trials and better education. The doctors are actually at hospitals are being educated and I also work as a PROFESSION and I'm actually looking after patients. Who have this diagnosis? So it's getting out there.
Participant 013_2023AUDSK

Information that's written in like non-medical terms I guess if that's what you mean, easy to read, easy to understand, the medical journals get a little bit hard to read, a little bit too technical from me. Is that is that what you mean?
Participant 010_2023AUDPA

Table 6.3: Information that was helpful

Information that has been helpful	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Other people's experiences (Peer-to-peer)	106	26.37	13	19.40	24	29.63	33	34.74	9	28.13	18	18.95	9	28.13	71	26.49	35	26.12	81	27.55	24	22.64
Health charities	67	16.67	9	13.43	12	14.81	25	26.32	0	0.00	17	17.89	4	12.50	45	16.79	22	16.42	50	17.01	16	15.09
Hearing what to expect (e.g. from disease, side effects, treatment)	64	15.92	11	16.42	16	19.75	14	14.74	5	15.63	5	5.26	13	40.63	42	15.67	22	16.42	44	14.97	19	17.92
Talking to a doctor or specialist or healthcare team	64	15.92	6	8.96	11	13.58	20	21.05	2	6.25	17	17.89	8	25.00	45	16.79	19	14.18	45	15.31	19	17.92
Medical or scientific sources	45	11.19	2	2.99	10	12.35	1	1.05	3	9.38	28	29.47	1	3.13	37	13.81	8	5.97	34	11.56	11	10.38
Triggers and managing exacerbations	28	6.97	2	2.99	13	16.05	3	3.16	4	12.50	1	1.05	5	15.63	23	8.58	5	3.73	25	8.50	3	2.83

Information that has been helpful	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Other people's experiences (Peer-to-peer)	106	26.37	28	28.87	38	29.01	27	23.68	13	21.67	54	27.27	52	26.53	30	27.03	76	26.12	59	29.50	47	23.27
Health charities	67	16.67	17	17.53	18	13.74	15	13.16	17	28.33	33	16.67	33	16.84	16	14.41	51	17.53	34	17.00	33	16.34
Hearing what to expect (e.g. from disease, side effects, treatment)	64	15.92	16	16.49	25	19.08	12	10.53	11	18.33	29	14.65	34	17.35	20	18.02	44	15.12	33	16.50	31	15.35
Talking to a doctor or specialist or healthcare team	64	15.92	16	16.49	16	12.21	20	17.54	12	20.00	28	14.14	36	18.37	17	15.32	47	16.15	24	12.00	40	19.80
Medical or scientific sources	45	11.19	5	5.15	10	7.63	19	16.67	11	18.33	18	9.09	24	12.24	14	12.61	31	10.65	24	12.00	21	10.40
Triggers and managing exacerbations	28	6.97	4	4.12	10	7.63	12	10.53	2	3.33	15	7.58	13	6.63	4	3.60	24	8.25	12	6.00	16	7.92

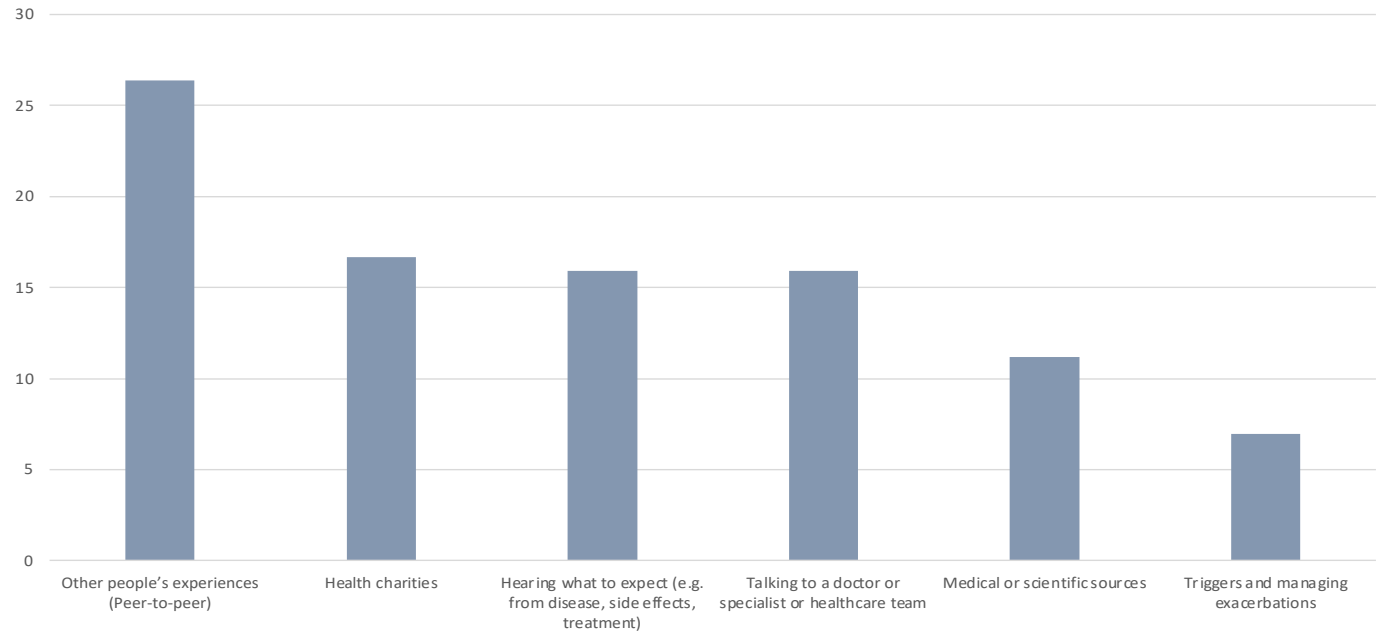


Figure 6.2: Information that was helpful

Table 6.4: Information that was helpful – subgroup variations

Information that has been helpful	Reported less frequently	Reported more frequently
Other people's experiences (Peer-to-peer)		
Health charities		
Hearing what to expect (e.g. from disease, side effects, treatment)		
Talking to a doctor or specialist or healthcare team		
Medical or scientific sources		
Triggers and managing exacerbations		

Information that was not helpful

In the structured interview, participants were asked if there had been any information that they did not find to be helpful. The most common response was that there was no information that was not helpful (31.09%). The most common types of unhelpful information included information from their GP or specialist (11.94%), sources that are not credible (10.20%), other people's experiences (9.20 %), information that was not type specific or too general (8.46%). Other themes included a lack of new information (7.46%) and worse case scenarios (7.46%).

Participant describes no information being not helpful

Not really, no, because I just all information is useful in some way and I and it didn't really. I didn't find it scary or anything. I think the only thing that was frustrating to me is what causes it. No one could tell me what caused it. So in the back of your mind you're always thinking, did I do something to cause it? But you know, they they just keep saying that it's just new in NAME'S sort of the first one in the line genetic line to get it out of nowhere, which is weird.
Participant 09_2023AUDPA

No. You always seem to gleam a little bit from it. Always got something in there that you didn't realise or remind you of, 'Oh, yes, that's right. I forgot about that' Sometimes you're gone, sometimes, I said... Sometimes it's a little bit overwhelming because there's so much. There's such a difference in symptoms between people with 22 Q that it's a very, very, very large field that can go wrong or can can affect the body in so many ways.
Participant 010_2023AUDPA

No, no, it's all been helpful. This was anything when you're researching or looking into anything. I always just sort of think everyone's experience is different for taking bits and pieces from other people's experiences. Or it's like a doctor, they may not have experienced it on a day-to-day. They're just going from the theory and nothing. That's been unhelpful because when the first time it was all helpful, you sort of want to cover the basis of everything just to see what is, what your focus is gonna be to find out how far reaching the condition was for her.
Participant 017_2023AUDPA

Participant describes the GP/specialist as being not helpful

Every service provider I ever visited, ever. Except for my current physician. OK?
Participant 015_2023AUDSK

The actual not paying attention. When you go in and you say, listen, there's something wrong, all right? And I think it's this or it's in this area and the GP goes, Nah, you don't know what you're talking about. We'll do this instead. And it's like, no, man, hang on. It's like the chest infections that I've been treated for, for the last six years. I kept saying to the doctor, it's not in the chest, it's in the throat, in the throat, somewhere in the throat there. I finally got a lung function test and my lungs, even though I've been an asthmatic since I was three months old, have operating at 98% and I have no scarring on them.
Participant 014_2023AUORC

Yeah, don't suggest moogoo for rough skin. This is a bit more than a bit of chafing, but this is what this twit who supposed that he was… I think he's what's running training wheels, I don't know, but he obviously know very much that was, was very much almost like you know give me a real medical diagnosis. I mean you know a bit of rough skin is not exactly being sick do you know what I mean? And I was actually having to educate this twit because he, he, when he was took…I was there about the kidney business and he was talking about my kidneys and saying I used to know your creatinine's 145 and you're geo fast 30 and blah blah blah blah and you know. Do I know what's caused the kidney injury? It must be a kidney injury that's happened at some point. And I said, well, I presume it's from the Scleroderma. He said no, that's just for the skin. If you tried moogoo, that's what he knew about Scleroderma. This was the doctors, the registrar.
Participant 002_2023AUDIS

Participant describes other people's experiences as being not helpful

Yeah, just a lot of, you know, the keyboard warriors with their personal advice when they're not clinicians, it's you see things that I can see, things that could be very harmful, but I choose not to engage in that kind of stuff.
Participant 018_2023AUDSK

Some things I suppose more, so you try to take some things on board from other people in forums, but some people can be quite one-minded. I think too, again, like I said, a lot of physicians or people that I've come across over the years they haven't treated you holistically.
Participant 001_2023AUDIS

PARTICIPANT: Yes, sometimes people go on and on about which medications they're on and stuff that's worked for them and rave on about it a bit too much and it, but one thing doesn't fit everyone, you know what I mean?
INTERVIEWER: That's right.
PARTICIPANT: Oh, you should see this doctor and this advice about, oh you should eat this and do that. That's not particularly helpful always because you get too much advice, do you know what I mean?
Participant 007_2023AUDIS

Not helpful as I told you, everybody's different. Some people can eat things and others they can't, but that's not helpful because it's not a true information. It's based on only a few people, a few opinion. It's not globally. Some people can eat anything. Information that will not be helpful.
Participant 020_2023AUDIS

Participant describes information that is not specific to their condition or sub-type as being not helpful (Too general)

If you search the symptoms of HS on Doctor Google, you have just about every disease ever known to men. And you're probably going to die from cancer.
Participant 005_2023AUDSK

Oh, probably a lot of stuff about life outcomes because they life expectancy is because they range so massively.
Participant 08_2023AUDPA

PARTICIPANT: Yes, when I, the very first time, this is back in 2014, I just Googled dystonia, and I don't know what site I got up, but it was just full of pictures of people in wheelchairs that were very contorted. And I went, "Oh my God, is that going to happen to me now?" I sort of spoke to the neurologist, and he said no, that's some other sort of thing, nothing else you've found. But it was just labeled as dystonia, and that was a shock to think, mm.
Participant 002_2023AUDNS

Participant describes a lack of new information as not helpful

Yeah, definitely. So like dreadful research studies or outdated information or lack of information is a real one. Like there's just not really anything out there. Yeah, I think that's probably it.
Participant 021_2023AUORC

A lot that you you read is outdated. You know series. Yeah, old papers of series haven't been updated. So you so you sort of get misinformation and what you know, a big one is you know lots of sites say that CMT is a form of muscular dystrophy, but it's not. So there's a lots of misinformation. Which can, you know, lead you down lots of wrong paths.
Participant 026_2023AUORC

Yeah, for sure. The Internet has been a terrible source of information. It's all outdated because it's…Because the medications and treatments with this progressing so quickly, when you research it on the Internet, everything is outdated and it's quite scary, particularly for someone who doesn't know much about the condition. It can be really depressing reading some of the material that's no longer actually applicable. But you don't know that until you you know you called by the. By the specialist team that it's not right anymore.
Participant 029_2023AUORC

Well, you open up websites sometimes and sometimes, you know, I suppose they just say the same old thing, so if that, you know, you're not learning anything new, so I just go on to the next one, so to speak. So, but nothing that's not negative.
Participant 016_2023AUDPA

Participant describes information about worse case scenarios and negative information as being not helpful

No, no, no, not at all. No, not that at all. I found the meetings are very much the opposite. In that, we all love so much. That is true. It's uplifting, but people, particularly there are online groups for scleroderma, and often, I think it's a good thing that people have somewhere they can vent or ask questions, but some people seem to be relentlessly negative. I find that difficult to cope with because I'm more a, I don't know, glass-half-full.
Participant 004_2023AUDIS

Probably some of the stuff on the Internet, like the horror stories and so much on the Internet where people are telling their story with the disease and everyone is different with Scleroderma, like you know you can't just read something on the Internet and think that's what's going to happen to you. Yeah, just the terrible pictures there and stuff.
Participant 022_2023AUDIS

PARTICIPANT: Yes, when I, the very first time, this is back in 2014, I just Googled dystonia, and I don't know what site I got up, but it was just full of pictures of people in wheelchairs that were very contorted. And I went, "Oh my God, is that going to happen to me now?" I sort of spoke to the neurologist, and he said no, that's some other sort of thing, nothing else you've found but it was just labeled as dystonia, and that was a shock to think, mm.
Participant 002_2023AUDNS

Participant describes information from sources that are not credible as not helpful (Not evidence-based)

There's a lot I would say out there online that hasn't been helpful when. When those potential boyfriends have gone searching the facts that they came up with and put in front of me really made me question like how is that true and how is that accurate? So I would give them a bit more, something a bit more accredited, but yeah, I think whether the information was out of date. I'm not sure, but I think that was definitely a hard part to counterbalance.
Participant 004_2023AUORC

I don't, I don't think these anti-inflammatory diets and give up milk, give up gluten and wheat are helpful. I think the research that I've read more in relation to how the genes are working and processing your DNA has been more educational.
Participant 013_2023AUDSK

I don't think so. As I said, she's selective in what she researches. She's not into populist treatments, if you like, from our alternative people.
Participant 062_2023AUDNS

Participant describes information that is not comprehensive as being not helpful

When you read like there's one set of papers that's available through them, the long version is actually fantastic and it's six pages long and it's written about how GPs in primary care can really cater to people like me. Oh, I was going to send you some of these things. Anyway, there's a short version of that paper and most GPs are going to jump to the short version of the paper, but there are some salient bits of information in that longer version that aren't in the short version that could cause someone with severe manifestation some issues if those nuances aren't. That's because of the complexity of the condition. I think that's just maybe the nature of the situation is that inevitably no information can cover the nuances of someone's individual expression, so generalizations can be just as, not harmful, probably too strong a word, troublesome as not having generalizations.
Participant 001_2023AUDPA

To be honest with the information from the medical, if we're looking at from that point of view, I find it's very limited because they don't know and some may may treat the condition like any other, like another condition. Yes. And that's the part that we find very difficult because we know these condition can be varying.
Participant 016_2023AUORC

There is, and again, it's with support groups that the newly diagnosed persons can sometimes overreact, and I, I sometimes don't find that helpful. I want to. I want to be caring and empathetic. But when you first diagnosed with Scleroderma, the natural thing is to Google and that's pretty much what everyone does and it's off three years if it hits your heart and lungs and you panic. And I did the exact same myself. So I feel like doctors could reassure patients better when they give that diagnosis and minimise the shock and the horror and the… but they don't. They don't give enough information about the disease when they diagnose you. I think that's the most unhelpful, that the reassurance could be a lot better in telling you that this, this disease affects everybody differently. And some people will live 30 years or 40 years with it, but they don't tell you that, they just tell you you have, and that's really frightening for a newly diagnosed person, it's mind bending. So I think yeah, that not giving enough information is not helpful. Yeah.
Participant 016_2023AUDIS

Participant describes feeling confident in deciding if something is not helpful (or not credible)

Well, any information that wasn't helpful to me during this. I just simply choose to ignore.
Participant 006_2023AUORC

No, not, not necessarily. I think you have to, you have to sort of siphon out what's what's right for you and what's not.
Participant 001_2023AUDSK

PARTICIPANT: Websites. So you know there again when you're reading when I'm reading anything from the web, I just take what I need, you know.
INTERVIEWER: Yes.

PARTICIPANT: And I don't worry about the others, the other things, you know, it's just what I need.
Participant 005_2023AUDIS

Table 6.5: Information that was not helpful

Information that has not been helpful	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
No information not helpful	125	31.09	19	28.36	17	20.99	27	28.42	8	25.00	43	45.26	11	34.38	88	32.84	37	27.61	88	29.93	36	33.96
GP\specialist	45	11.19	4	5.97	19	23.46	6	6.32	6	18.75	6	6.32	4	12.50	36	13.43	9	6.72	33	11.22	11	10.38
Sources that are not credible (Not evidence-based)	41	10.20	4	5.97	18	22.22	1	1.05	3	9.38	10	10.53	5	15.63	33	12.31	8	5.97	35	11.90	6	5.66
Other people's experiences/unsolicited advice	37	9.20	2	2.99	14	17.28	7	7.37	5	15.63	9	9.47	0	0.00	28	10.45	9	6.72	30	10.20	7	6.60
Not type specific (Too general)	34	8.46	3	4.48	8	9.88	15	15.79	1	3.13	6	6.32	1	3.13	25	9.33	9	6.72	31	10.54	3	2.83
Worse case scenarios	30	7.46	7	10.45	11	13.58	3	3.16	1	3.13	4	4.21	4	12.50	19	7.09	11	8.21	23	7.82	7	6.60

Information that has not been helpful	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher statu	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
No information not helpful	125	31.09	23	23.71	44	33.59	34	29.82	24	40.00	63	31.82	59	30.10	38	34.23	87	29.90	56	28.00	69	34.16
GP\specialist	45	11.19	7	7.22	9	6.87	20	17.54	9	15.00	19	9.60	26	13.27	20	18.02	25	8.59	26	13.00	19	9.41
Sources that are not credible (Not evidence-based)	41	10.20	6	6.19	11	8.40	20	17.54	4	6.67	15	7.58	25	12.76	13	11.71	28	9.62	24	12.00	17	8.42
Other people's experiences/unsolicited advice	37	9.20	6	6.19	13	9.92	12	10.53	6	10.00	15	7.58	21	10.71	6	5.41	31	10.65	15	7.50	22	10.89
Not type specific (Too general)	34	8.46	9	9.28	12	9.16	7	6.14	6	10.00	15	7.58	19	9.69	9	8.11	25	8.59	20	10.00	14	6.93
Worse case scenarios	30	7.46	8	8.25	7	5.34	12	10.53	3	5.00	16	8.08	13	6.63	8	7.21	22	7.56	14	7.00	16	7.92

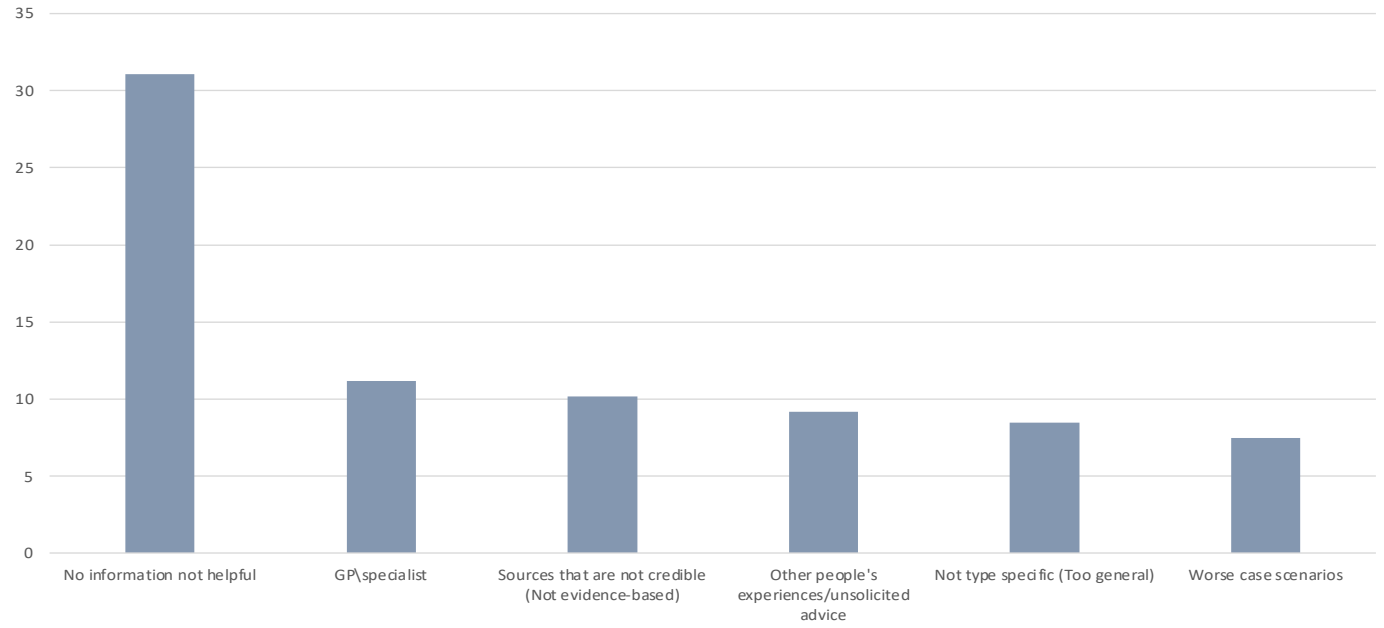


Figure 6.3: Information that was not helpful

Table 6.6: Information that was not helpful – subgroup variations

Information that has not been helpful	Reported less frequently	Reported more frequently
No information not helpful	Diseases of the immune system	Endocrine, nutritional or metabolic diseases Diseases of the immune system
GP\specialist		Diseases of the immune system
Sources that are not credible (Not evidence-based)		
Other people's experiences/unsolicited advice		
Not type specific (Too general)		
Worse case scenarios	Diseases of the nervous system Diseases of the skin	Diseases of the immune system Aged 65 plus

Information preferences

Participants were asked whether they had a preference for information online, talking to someone, in written (booklet) form or through a phone app. The most common responses were online information (29.35%),

talking to someone plus online information (23.63%), and talking to someone (21.64 %). Other themes included written information (13.68%), all forms (5.47%), and apps (2.49%).

The main reasons for a preference for online information were accessibility (27.86%), and being able to digest information at their own pace (18.41%).

The main reasons for a preference for talking to someone was being able to\have time to ask questions (18.41%), and that it was personalised (14.43%).

The main reason for a preference for written information were written information is that they can refer back to/highlight important information (3.23%).

Participant describes online information as main information preference

Well, I, I largely prefer online information because it's easily accessible, you can access it from anywhere at any time and you know, having to compare and hear from people who are first and who are first and experience about this is also helpful because the ideas and what they went through all brought together would provide a huge knowledge that can, you know, guiding the one through the process. And you know, it's easily accessible. That's it for me.
Participant 006_2023AUORC

Well, the option to talk to someone is probably limited. Yeah, quite happy to look stuff up online and read it at my leisure.
Participant 002_2023AUDPA

No, it's primarily online because I wanna know, well, there's not a lot great deal that even the like, the doctors and medical, medical people, they really don't know a great deal about it. Like my GP had to our GP had to to really look it up. The psychologist that we're seeing that my son's seeing doesn't know anything, didn't know anything about it. He and…he's had to to look it up. So it's a, you know, it's a, it's a condition that whilst it's common and it's very under diagnosed. And very few people know about it. Everyone knows about MS but nobody knows about HS.
Participant 009_2023AUDSK

Why online? Because I can access it as when I need it and nothing sort of presented. As a video or audio is much preferred than having to read because I'm with a special needs a child and I never have time to read anything, but I can get it in my headphones and put one thing. Participant 087_2023AUENM

Participant describes talking to someone plus online information as main information preference

I would say online is really good because you can access it 24/7 as long as you've got a good place to go. There are times when you just don't quite fit that mold or that doesn't quite add up, that it's great to be able to have someone to contact and clarify as well. For me, in-person they need to do the skin scoring and moderation and some of the clinical tests, but for the other stuff as a check-in in between, absolutely. Either phone or video; a Teams or Zoom type meeting, yes, it's really helpful.
Participant 017_2023AUDIS

And why I like to I guess online, because then I can do it in my own time. However, I really do like to talk to people about it because sometimes, especially face to face, you can sort of, you know, engage them I guess, and they can come up with. Different people have got different strategies of dealing with situations.
Participant 015_2023AUDPA

I like to be able to talk to someone because then that way I feel like I can carry on like in a flowchart kind of manner during the conversation, whereas if talking to someone online, you only ask a question and get a couple of different answers. With talking, it's a lot easier to branch to something else that's relatable or whatever. I do prefer it that way. If I'm wanting to do my own research, I do like the fact that I can look up stuff online as things pop into my head and I need to research. Probably a bit of both of those two.
Participant 040_2023AUDIS

Talking to someone if they're a specialist because the interaction back and forth to ask questions is helpful. Being able to access stuff online is convenient, provided you know the source and can interrogate that. Booklets in general are only useful for very high-level information, and I feel like I'm well past needing high-level information on things because I acquired that ages ago. If I have questions now, it's far more specific things that you won't find in the books.
Participant 067_2023AUDPA

Participant describes talking to someone as main information preference

Prefer. I actually prefer to be able to sit and talk with the doctors and nurses and that and then secondary to that would be information booklets that you can take away. But generally I'll just have the conversation and that's. That's enough for me to get what I feel I need to know. Participant 007_2023AUORC

I can't…I don't really have a preference. I think they all have their place. I mean, you can do research at any point in time, online or paper, you know, books or anything you know. But again, I don't think anything beats face to face. I think that or just talking to someone on the phone.
Participant 078_2023AUDIS

I would prefer face to face with with somebody that understood the condition and displayed a level of of competency around it that that, you know, that gave me reassurances and, and yeah, some, yeah, some empathy. I can't get that online. Yeah.
Participant 027_2023AUORC

Participant describes written information as main preference

I like something that I can read and that way I can refer back to it later. So if the doctors can e-mail me or send me a pamphlet or something like that. And then I would prefer that then face to face or in person just so I can refer back to it.
Participant 013_2023AUORC

Why I like I'm an old person, older person, so I like the physical booklets so I can read and highlight and go back and read again. I'm not a very clever learner online or reader online. I think it's all to do with our upbringing, I think. Even though I do do it, don't get me wrong, because I'm a researcher, I actually advocate and I do lots of research so I can do it. But personally, when like PATIENT's plan, I want to see every single therapy report in paper so I can read it, highlight it and you know, and that's the way I work. Whereas I don't find highlighting it online easy. So gaining it online. I find it. I do look online, but I always print it out. So and talking to families, of course it's great to talk to families, but for us it's not beneficial because PATIENT is so much worse than them. So I can help them, but they can't really help me. Does that make sense?
Participant 06_2023AUDPA

The booklets that they put out from Scleroderma Australia, they just have them online. I find them somewhat helpful. They give a little a broad scope of questions and answers. Not in-depth enough if it's actually happening to you, but sometimes that led me to being able to educate the doctors, "Look, this is what's they've said in the booklet, can we go further?" Sometimes it's led the conversation a bit better.
Participant 014_2023AUDIS

Participant describes prefering all forms of information

PARTICIPANT: All of the above. Everyone takes things in differently and sometimes you need to read it and see it and hear it five different ways for it to sink in because we are working in an area where there's not just one issue.
INTERVIEWER: Yep. Very good point.
PARTICIPANT: The thing is different personalities. Some people take it by diagram. Some people hear it, some people need it in paper.
Participant 092_2023AUENM

I don't have a preference because part of my pacing strategy is breaking things up into little bits and different things. I can't do too much of any one thing. I try not to preference in fact.
Participant 041_2023AUDPA

No, I tend to, I gather information from all sources, just sort of make up my mind on what I'm following, if you know what I mean. Yeah.
Participant 032_2023AUORC

Participant describes apps as main information preference

I guess. Maybe just like, yeah, phone apps are easy to use because I'm so used to doing it anyway. But I guess talking to someone is easier as well because then they can explain it to you. So yeah.
Participant 014_2023AUDSK

And why I prefer online and apps because I like to read things in my own time. I want them though, I want them to be from accredited sources. So for instance with this condition I, I like the fact that there's an international body and and also a body of professionals for this particular condition that are putting the information down the line. The problem is in that area, the professionals in, in our country and not adopting them, adopting these, So yeah, so that would be it. Online, I love it. Online and apps, I love apps.
Participant 025_2023AUDPA

Table 6.7: Information preferences

Information preferences	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Online information	118	29.35	16	23.88	29	35.80	26	27.37	11	34.38	29	30.53	7	21.88	83	30.97	35	26.12	90	30.61	28	26.42
Talking to someone plus online information	95	23.63	14	20.90	21	25.93	20	21.05	9	28.13	18	18.95	13	40.63	64	23.88	31	23.13	70	23.81	23	21.70
Talking to someone	87	21.64	5	7.46	16	19.75	25	26.32	4	12.50	31	32.63	6	18.75	68	25.37	19	14.18	66	22.45	21	19.81
Written information	55	13.68	7	10.45	17	20.99	11	11.58	5	15.63	10	10.53	5	15.63	43	16.04	12	8.96	46	15.65	9	8.49
No strong preference	24	5.97	1	1.49	2	2.47	7	7.37	1	3.13	11	11.58	2	6.25	18	6.72	6	4.48	16	5.44	8	7.55
All forms	22	5.47	4	5.97	3	3.70	5	5.26	2	6.25	5	5.26	3	9.38	15	5.60	7	5.22	18	6.12	4	3.77
Apps	10	2.49	2	2.99	4	4.94	0	0.00	1	3.13	3	3.16	0	0.00	8	2.99	2	1.49	9	3.06	1	0.94

Information preferences	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Online information	118	29.35	23	23.71	38	29.01	36	31.58	21	35.00	58	29.29	58	29.59	35	31.53	83	28.52	66	33.00	52	25.74
Talking to someone plus online information	95	23.63	24	24.74	31	23.66	30	26.32	10	16.67	47	23.74	47	23.98	26	23.42	69	23.71	42	21.00	53	26.24
Talking to someone	87	21.64	14	14.43	31	23.66	28	24.56	14	23.33	46	23.23	39	19.90	18	16.22	69	23.71	39	19.50	48	23.76
Written information	55	13.68	8	8.25	18	13.74	16	14.04	13	21.67	25	12.63	30	15.31	15	13.51	40	13.75	28	14.00	27	13.37
No strong preference	24	5.97	3	3.09	6	4.58	10	8.77	5	8.33	10	5.05	13	6.63	6	5.41	18	6.19	11	5.50	13	6.44
All forms	22	5.47	5	5.15	9	6.87	6	5.26	2	3.33	11	5.56	11	5.61	6	5.41	16	5.50	12	6.00	10	4.95
Apps	10	2.49	0	0.00	7	5.34	3	2.63	0	0.00	7	3.54	3	1.53	5	4.50	5	1.72	6	3.00	4	1.98

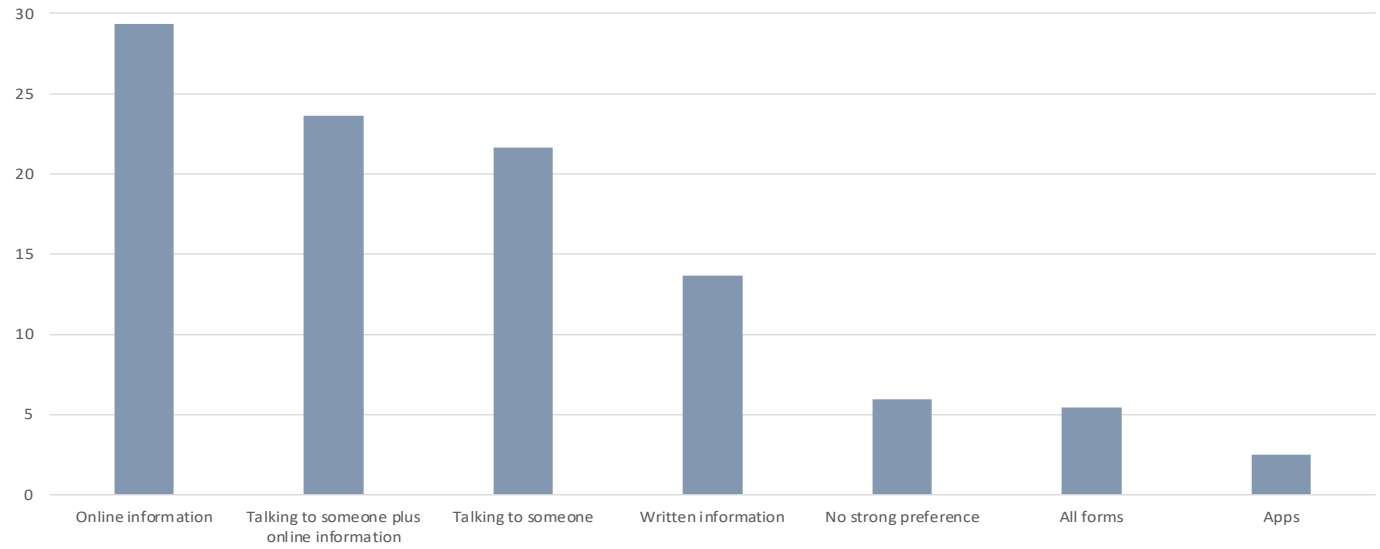


Figure 6.4: Information preferences

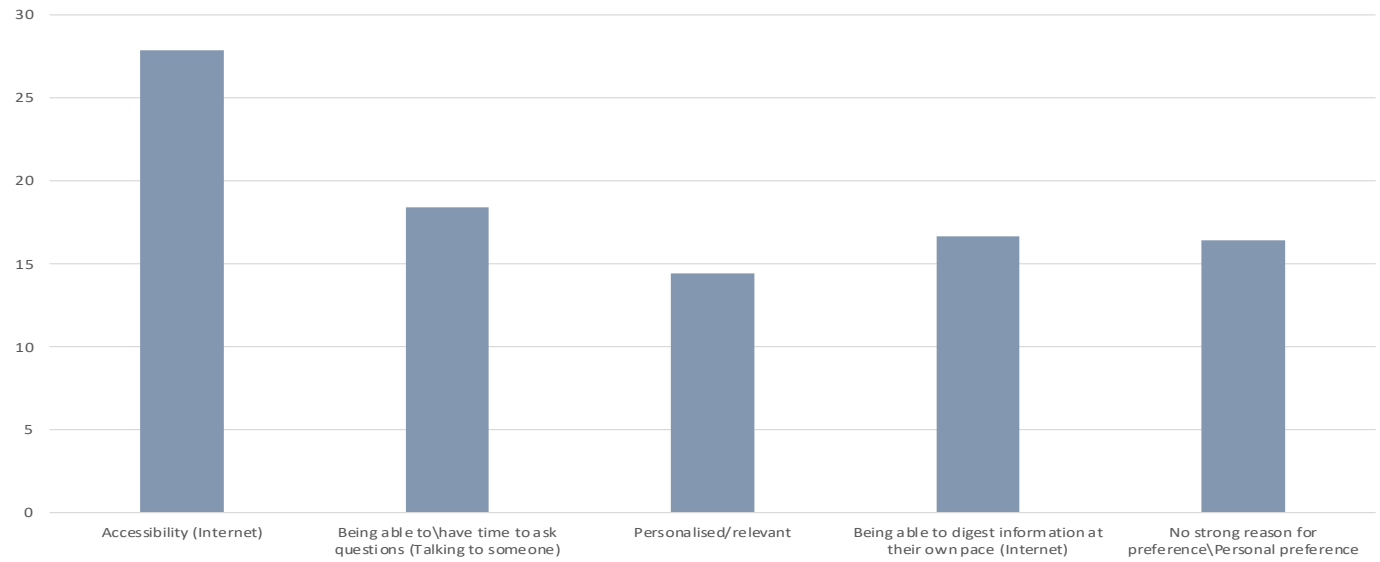


Figure 6.5: Reasons for information preferences by format

Table 6.8: Information preferences – subgroup variations

Information preferences	Reported less frequently	Reported more frequently
Online information		
Talking to someone plus online information		
Talking to someone		
Written information		
No strong preference		Other rare condition
All forms	Developmental anomalies	Endocrine, nutritional or metabolic diseases
Apps		

Timing of information

Participants in the structured interview were asked to reflect on their experience and to describe when they felt they were most receptive to receiving information. The most common times were at the beginning (diagnosis) (31.34%), continuously (19.65%), after the shock of diagnosis (12.44%) and 12 months or more after diagnosis (10.70 %).

Participant describes being receptive from the beginning (diagnosis)

Yeah, that's a really good question and I'm actually glad you asked, just cuz I was speaking to someone about this yesterday and I think, and it's related to my own, obviously my diagnosis, I don't think you necessarily give someone too…there can be too much information at the point of diagnosis. Like I, for the record, I mean this course is a record, but you know, I still maintain that I wasn't given enough information when I was when I was diagnosed.
Participant 011_2023AUORC

I'd say probably in the like month leading up to the diagnosis. It's when I thought I was pretty sure of what I had and I was just hungry for information. I was, you know, taking on any little snippet that I could find really heavily reading into things. So kind of a month leading up to my diagnosis and probably around that time, month after I'd say as well it's kind of settled down now.
Participant 027_2023AUDSK

Yeah, that's a great question. Obviously, initially, you know, you're trying to take as much information in and you need that information to try and understand when it's a rare disease because there's nothing, you can't really find anything. But at the same time your ability to absorb that information is really impaired. I would say at the very beginning it's really important because then you can kind of pace that information out if you're having a moment where you can understand it. But also, you know, when you're able to do a lot of self-directed research, you're open to the idea of receiving that information which will be at different times for different people depending on their processes, you know, how they cope with the information and grieving and all of that sort of stuff. So I would just say throughout, throughout the whole journey, like when my brain's working probably.
Participant 021_2023AUORC

All of information straight away-- I didn't share what was going on with many people but I wanted the information straight away. I wanted to know everything. Actually, that was when I found it best to talk to someone because you could ask specific questions.
Participant 065_2023AUDPA

Participant describes being receptive to information continuously throughout their experience or bit-by-bit so that it is digestible

Well, when someone had something decent to say. So anytime, anytime's a good time if it's helpful, if if they want to experiment, I'm fine with that too. You go for your life. It's going to work great if trial and error, so I'm always just have to do it. If someone's got a solution and you give it your best shot, go for it.
Participant 006_2023AUDIS

I think I've always been able to take the information in it's just been a lack of information more than anything, my parents were warned apparently when I was first diagnosed not to research it on their own because it was so broader disease that it would freak them out. Just stick to the…your fingers freeze, and don't smoke.
Participant 014_2023AUDIS

I have been fairly receptive to all along. I've just, I've wanted to know as much as I can. Yeah, yeah. I guess. I think at diagnosis I could have done with a whole lot more information than we were given, rather than having to go and find it myself.
Participant 021_2023AUDPA

Participant describes being receptive to information 12 months or more after diagnosis

Probably within the last year or two. I mean, I would have been diagnosed around four years ago. And I mean, it took a year. I was literally in tears about it for because this wasn't a sebaceous cyst anymore. This wasn't, do you know what I mean? Like, this was, this was serious, this was bad. This is not just gonna go away. And the doctors don't even know how to help you. So, you know, it was pretty upsetting. And yeah, I was pretty depressed about it because I was just like, what do I do? Like, there's nothing to do. Like, yeah. So yeah, I was pretty depressed about it.
Participant 006_2023AUDSK

Probably after like after about 18 months when it was out of major grief and is she going to die?
Participant 087_2023AUENM

Oh, probably in one to two years after diagnosis, because it all happened when her baby was born and it was life and death. Whether this baby would get through and she had massive heart surgeries, tube feeding, everything was just about the baby. It was only after that. That I could really sit back and say, yes, my daughter has has some. To George, now's my time to find out more about it. All right. If we hadn't had the baby born, I probably would have been on to it, you know, much sooner.
Participant 08_2023AUDPA

Participant describes being receptive to information after the shock of diagnosis

Yeah, I think, I think you want all the information at the beginning, but it's extremely overwhelming. So I think it was good that you sort of need a bit more time. So we definitely, I definitely like went back and saw like every time. You'd go back, you'd ask more questions or different questions. Yeah. So I'd say the first like probably the first three or four months after diagnosis look like, like I probably received a lot of information. I don't know if I took it all in properly. There's probably a little bit after that that you really need like continued access to it because eventually you, you know, you can move past the kind of initial stress of diagnosis and then you can. Sort of deal with the next step, yeah, okay.
Participant 079_2023AUDIS

Yeah, it's a good question. Definitely not within the first few days when we did receive most of the information that was really hard. We were to we were so overwhelmed with his diagnosis, it was impossible for me to focus on the info that we were being given by the team as like they're doing their best. But we just went in a position to be able to absorb all at that point. So I think it took us maybe nearly a week before we got over the shock of it all and were able to actually start to read the information with clear heads.
Participant 029_2023AUORC

Probably a couple of weeks after he was diagnosed. Because it was although we were reading it, it was finally then I really absorbed it because I could sit down calmly. Even now I still go back and re-read stuff I've read a hundred times and pick up different things and see it differently. A while after diagnosis I was able to absorb everything, I think.
Participant 048_2023AUDPA

Participant describes being receptive to information when emotionally and physically able to take in the information (eg when not having symptoms)

Probably when I started feeling a bit less fatigued and was a little less worried. You know, about the more serious outcomes. So once I wrap my head around what that actually looked like and that, yeah, and that I was starting to feel better than I was more able to process more information. Whereas when you like to people sometimes because you just got that time right there and duty of care and so on and so forth and they throw a lot of information at you, it's really hard to process that because you're processing the oh, there's actually something wrong in the moment.
Participant 024_2023AUDIS

Look at the beginning. It was very emotional I guess, you know, thinking that yeah, I've just had a child who now has this condition. Personally, I have a lot going on. I have a 15 month old daughter as well and my mum passed away like two days after she was born. So I you know, and my pregnancy was not planned, I guess. Not that we regretted in any way at all, but so I guess, you know, I was still grieving, but I was excited, you know, to have another baby. But then you know, this information bombarded us as well. Yeah, so definitely the beginning was the hardest and not knowing anything and not knowing anyone. You could sort of just talk to and not knowing, not having a clear answer I guess on you know she's got this condition you know and surgery would fix that sort of thing like whereas now, I'm more you know let's just go along and see what happens and when we come across it we'll we'll fix it. So I guess now I'm feeling, you know, not more supported, but more willing to take on the information, I guess.
Participant 034_2023AUDPA

Definitely not initially. The spectrum of possible things that could happen I found completely overwhelming and very scary. I don't even know how I logged on to the website, initially. I was very different to my husband. My husband wanted to know everything about it right away, whereas I didn't. I had to have a number of counselling sessions to get to a place where I had accepted the diagnosis. It was really only at that point where I could invite more information about it in. At what point was that? Maybe six months after the diagnosis. By the time she turned two just over a year after the diagnosis I was comfortable with it but maybe between six months up to eighteen months. Six months to twelve months, yes.
Participant 061_2023AUDPA

Participant describes being receptive to information during adolescence or adulthood (once they appreciated their personal responsibility for health)

You should answer this one. This is a good one for you. As remember we were talking about how you weren't interested and then all of a sudden you started becoming interested and you booked wanting to know the information that was being talked about. So, so how long ago was that do you think? So you're 20, I think 24, it was May, it was maybe 2014/2015. So I think I would have been 16 or 17 or 20, but anyway, but it was like this is I was leaving school, so whatever at age.
Participant 037_2023AUDPA

Participant describes being receptive to information when condition changes or there are new symptoms

Took a lot of information in when we got the, yeah, in the early, you know, days, there was a lot of lot of stuff to look at. There were a lot of medical interventions that needed to be addressed and checked. And also, being a child, you know, I was very, you know, wanted to make sure that we were getting whatever I could to help her along the way. I'm always open to getting information now, but I must admit I'm at a stage of quite a lot of fatigue and so tend to respond to things as they happen and I need to. And there's always there's a whole pile of things there that I want to read one day that you need to have some time out to yourself. So I haven't sat and done that cuz I'm too exhausted. But there was nothing. I was just gonna say I've lost it now, but anyway.
Participant 038_2023AUDPA

Well, I think when something changes in me or the condition, I sort of, I don't know what which it is exactly. It's sometimes it's hard to pinpoint, but yeah, when there's a change. To see change anyway, whether it's good or bad.
Participant 005_2023AUDNS

Participant describes being receptive to information five to six months after diagnosis

I think it would mean after a good few months, like four or five months before they before I'd kind of accepted it enough to hear what was being said. I suppose in the beginning, once something becomes too overwhelming, you shut off. You stop listening, you just can't hear it anymore. So after a few months, then like a few like Doctor visits and hearing it over and over again, it become easier to accept and then digest what was being said. Yeah?
Participant 025_2023AUORC

Probably three to six months after her diagnosis. I think once you accept it and you, you know, sort of get on with life, yeah, you stop that crisis management, then you become more receptive to it. And I think it also depends on the medical team you're working with. If you still, if you feel heard and listened to, I think it makes it easier to absorb the information. Because I do know a couple of times going into the neurologist that I was so focused on what I wanted to find out, that I wasn't really listening to what he was saying. Yes. Yeah. So you're right. Having those channels to follow up informally, the thing that I found really frustrating was trying to, you know, you confine to appointment times and sometimes you have a question that you don't need a whole appointment for, but you just need to clarify something.
Participant 090_2023AUENM

That's a good question because when you're in the full blown pots, you can't actually remember much or take much in. I think it wasn't until maybe six months down the track that I was able to fully get my head around it and then work out a way to move forward.
Participant 031_2023AUORC

Participant describes being receptive to information more than 6 months, less than 12 months after diagnosis

I that's tricky. I think definitely initially when she was diagnosed and stuff, I definitely wasn't in the right headspace to take on the information and understand it fully. But I think as she got older, probably from, you know, that 6 to 8 month mark when things started to calm down a little bit more. I was probably more proactive myself, trying to find out what this all

actually meant and what it meant for her for the future and what it was going to mean for us going forward. Participant 027_2023AUDPA

When do I feel like? Probably just more recently. Yes, it's probably around six, well, more than six to ten months. I think I needed to come to accepting the diagnosis first before receiving any more information. Participant 059_2023AUDNS

Definitely not in the first couple of weeks because it was all just overwhelming, and I was I guess in shock about it. Probably anywhere from six months onwards it started to sink in a bit, and I was able to take it all in and like, this is where we need to go, this is what we need to do now.
Participant 060_2023AUDPA

Participant describes being receptive to information two months after diagnosis

I mean, I didn't really think in what I had until. At least two months after I was diagnosed. I heard the doctor. I heard what he was saying. I'm like, great. So I have to have this for the rest of my life. Like there's no cure for it. That's when it sunk in. Like, damn, you can't fix me.
Participant 003_2023AUENM

Look, I'd probably say in the in the couple of months, proceed like following. The diagnosis was when I was really looking for information. Yeah, I I know I would have. Yeah, I suppose I would have liked to have had the confidence to act on that a bit sooner too, because it might not have gotten in the stage it did.
Participant 007_2023AUDSK

Probably sort of now. I only found out two months ago maybe... And I think I was just so angry that I hadn't actually been told formally that I had it. And now that they've confirmed it and I've started doing my own research, I think I'm sort of interested to learn as much as I can.
Participant 096_2023AUDNS

Table 6.9: Timing of information

Timing of information	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
At the beginning (diagnosis)	126	31.34	21	31.34	24	29.63	31	32.63	10	31.25	32	33.68	8	25.00	81	30.22	45	33.58	95	32.31	31	29.25
Continuously	79	19.65	21	31.34	16	19.75	7	7.37	11	34.38	16	16.84	8	25.00	53	19.78	26	19.40	54	18.37	24	22.64
Combined minor themes	59	14.68	10	14.93	10	12.35	12	12.63	6	18.75	12	12.63	9	28.13	42	15.67	17	12.69	44	14.97	15	14.15
After the shock of diagnosis	50	12.44	11	16.42	7	8.64	15	15.79	0	0.00	10	10.53	7	21.88	24	8.96	26	19.40	34	11.56	16	15.09
12 months or more after diagnosis	43	10.70	14	20.90	15	18.52	5	5.26	3	9.38	2	2.11	4	12.50	29	10.82	14	10.45	30	10.20	12	11.32

Timing of information	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
At the beginning (diagnosis)	126	31.34	31	31.96	43	32.82	37	32.46	15	25.00	57	28.79	67	34.18	36	32.43	90	30.93	57	28.50	69	34.16
Continuously	79	19.65	18	18.56	28	21.37	21	18.42	12	20.00	39	19.70	40	20.41	25	22.52	54	18.56	45	22.50	34	16.83
Combined minor themes	59	14.68	14	14.43	13	9.92	25	21.93	7	11.67	30	15.15	29	14.80	21	18.92	38	13.06	33	16.50	26	12.87
After the shock of diagnosis	50	12.44	22	22.68	13	9.92	10	8.77	5	8.33	23	11.62	25	12.76	14	12.61	36	12.37	24	12.00	26	12.87
12 months or more after diagnosis	43	10.70	10	10.31	13	9.92	14	12.28	6	10.00	25	12.63	18	9.18	10	9.01	33	11.34	22	11.00	21	10.40

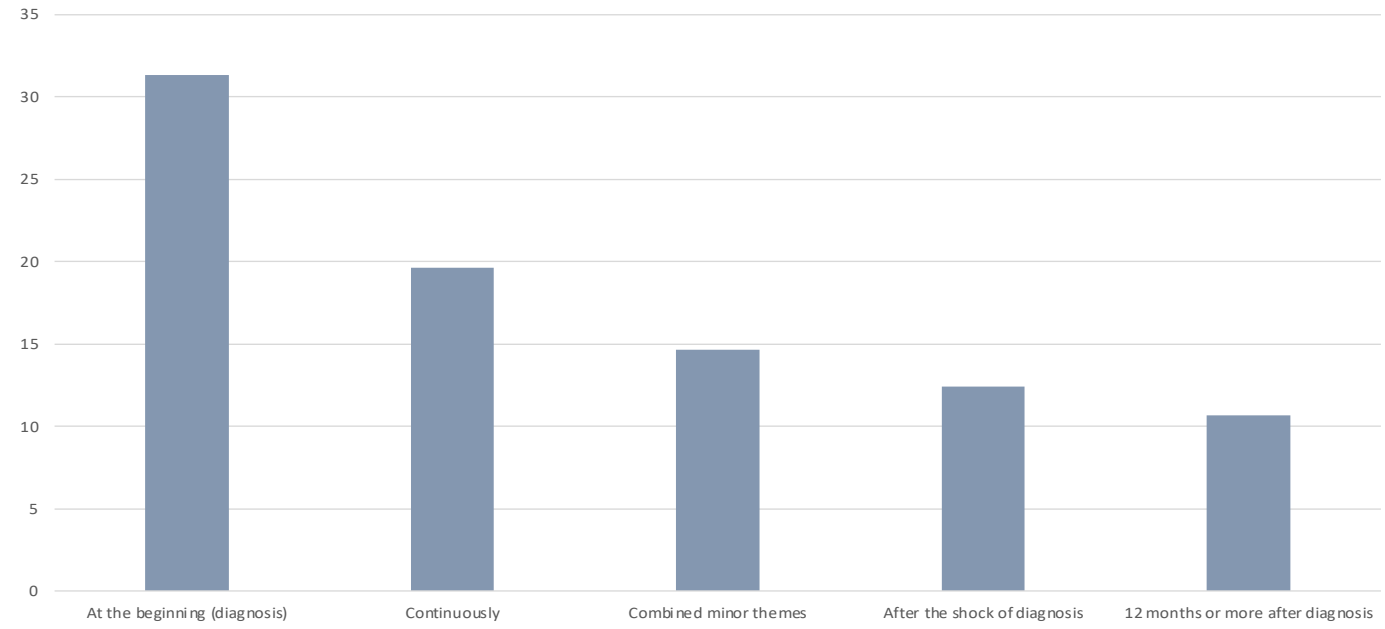


Figure 6.6: Timing of information

Table 6.10: Timing of information – subgroup variations

Timing of information	Reported less frequently	Reported more frequently
At the beginning (diagnosis)		
Continuously	Diseases of the nervous system	Developmental anomalies Diseases of the skin
Combined minor themes		Other rare condition
After the shock of diagnosis	Diseases of the skin	Aged under 18
12 months or more after diagnosis		Developmental anomalies

Healthcare professional communication

Participants were asked to describe the communication that they had had with health professionals throughout their experience. The most common theme was that participants described having an overall negative (34.83%), overall positive(26.62%), and overall positive, with the exception of one or two occasions (24.63%).

Participants described reasons for positive or negative communication with healthcare professionals.

Participants that had positive communication, described the reason for this was because of holistic with two way, supportive and comprehensive conversations (28.36%).

Participants that had negative communication, described the reason for this was because there were limits in understanding (33.33%), because of and dismissive (One way conversation) (16.42%). Other themes included limited in relation health professionals not having a lot of time (8.46%).

Participant describes communication with healthcare professionals as overall negative

Yeah, that there wasn't the communication wasn't great. Like I said there was a you know earlier in the interview there was there's basically a 10 year…where this is just ignored any even. Yes there was an actual medication available but there was still like treatment. There was treatment plans and and medical care that could have been provided. As far as counseling or just lifestyle stuff I kind of needed to know. So yeah, I'd say it is not not been very good.
Participant 011_2023AUORC

Crap, not good. Like costed hundreds of dollars to see the dermatologist and I think he spent about 9-9 to 10 minutes with me. Probably not even 10 minutes. Like, literally, like, just looked at me in and out because he's in demand and he's got a whole bunch of stuff going on. And other than that, the doctor, you know, if I'm like, oh, this is really bad, like, he doesn't want to have a look, he'll take my word for it.
Participant 006_2023AUDSK

Inadequate. I really felt that most of the time I was driving the understanding research, how to get help, who to get help from, what to do from professionals and that they would sort of. Not explain things like I was intelligent enough to like absorb the information. Yeah, and and therefore would miss things out and and not give me full picture.
Participant 087_2023AUENM

Yeah, very limited. And I have to say before we were diagnosed, there were some medical professionals who probably wasn't convinced of my concern. So yeah.
Participant 094_2023AUENM

Limited, I would say the pediatrician, you know, you get a small window every six months, so that's you know well, that's pretty much with all specialists. You just don't get their time. You just don't and you can't access them. In between, it's very difficult to communicate with them. In between the assigned appointments that you're given, yeah. So I, I feel like a lot more on researching information, finding information, going to them and talking about rather than the other way around
Participant 095_2023AUENM

Participant describes communication with healthcare professionals as overall positive

Brilliant. The Doctors are fantastic. Yeah. Any questions I've got that more than happy to ask, even if I ring up, but you know, short notice kind of thing that they're really good.
Participant 032_2023AUDSK

Yeah, really good. His team is great. We've got a phone number that we can call or text anytime 24/7 if we have any questions and we get responses straight away. And yeah, as I said earlier that every time we meet with his team and we've got questions, they've always been really, they've been really clear with this and able to answer everything that we've come to them with.
Participant 029_2023AUORC

It's been very good. The the pediatrician was very knowledgeable so was able to help. It was very good, but he said read it, that that she's not going to get all of that. That's just what could happen. You're best to just yeah, take it as it comes. And the doctor, our GP is very good. Whenever we need something, he's happy to to delve in and help with that or refer.
Participant 010_2023AUDPA

Oh, gosh, I couldn't speak highly enough of them. They're wonderful. Every person I've contacted with has has been, oh, they've been explicit but but kind. And I couldn't fault them. They were scientific, as I need them to be. And then at other times they're practical. Yeah. No, it's very good.
Participant 019_2023AUDPA

Participant describes communication with healthcare professionals as overall positive, with the exception of one or two occasions

In general it's been good. When it's somebody, I guess you know, being especially with Doctor NAME has been a really good experience. The surgeon prior wasn't was pretty good. It's more when it's somebody new, especially GP's wise or yeah, doctors that I've met through ED and that sort of thing. That's when I think. I've had the most negative experiences and it's been the most difficult, yeah.
Participant 022_2023AUDSK

I mean just very, very mixed, I think. When we've in general, when we've talked to people who are knowledgeable about it, it's quite positive. Yeah, it's quite positive and there's a lot that we've been able to learn. Yeah, I guess in general, quite positive. I think maybe there's more negative or confusing experiences have been with people who maybe don't know as much about CHARGE syndrome as a whole and they're more just focusing on the their smaller specialty. Yeah, I think, yeah, generally generally positive with most people who have had. With charge syndrome with health professionals.
Participant 089_2023AUENM

Well, that depends. Yeah. So once once I got a diagnosis, it was all really positive. The people that I have seen since then, I have been really good at communicating. Yeah, with the exception of one Doctor, he was quite a junior doctor. It was when the pandemic started and everyone was all over the place. And I did. I did do a phone appointment for that. And he didn't seem to know like the last year about the COVID vaccines and priority groups and all of those kinds of things. And he didn't seem to have any of the answers that I was looking for. But I like I said, I don't think he was. I think he was just kind of put in to the position to to do the cause. But

yeah, on the whole. Yeah, on the whole, really positive. And the same with the maternity doctors. People are pretty upfront about saying, you know, if they don't know anything and they've not heard of scleroderma, but also that they're going to go and consult with more senior people and come back for answers and things like that, which is which is good.
Participant 024_2023AUDIS

Some are good, some are not so good. Some medical professionals go above and beyond for us to explain things and make sure she's getting the right level care and things like that. Others, they can be quite dismissive because it's not, especially with the duplication, a lot of doctors, when you go into a new specialist, they're just like, 'Oh yeah, it's the it's the deletion' or they'll be like, 'oh, it's not as bad, it's the deletion. So you probably don't need help'. So it was a matter of finding our right team, which did take about five years to get it all the right people in there. So some of it's been really good and some of it has not.
Participant 032_2023AUDPA

Overall, great. Like my last cardiologist is amazing. She's been really, really good right back in the beginning. The cardiologist that I saw when I was first admitted to hospital weren't weren't really very nice. I didn't like them anywhere as much as my second and third cardiologist.
Participant 032_2023AUORC

Table 6.11: Healthcare professional communication.

Healthcare professional communication	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Overall negative	140	34.83	18	26.87	33	40.74	41	43.16	12	37.50	25	26.32	11	34.38	103	38.43	37	27.61	113	38.44	27	25.47
Overall positive, with the exception of one or two occasions	107	26.62	14	20.90	21	25.93	22	23.16	9	28.13	31	32.63	10	31.25	73	27.24	34	25.37	80	27.21	25	23.58
Overall positive	99	24.63	6	8.96	22	27.16	25	26.32	6	18.75	32	33.68	8	25.00	73	27.24	26	19.40	66	22.45	33	31.13

Healthcare professional communication	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher statu	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Overall negative	140	34.83	21	21.65	62	47.33	39	34.21	18	30.00	80	40.40	59	30.10	35	31.53	105	36.08	75	37.50	65	32.18
Overall positive, with the exception of one or two occasions	107	26.62	24	24.74	29	22.14	37	32.46	17	28.33	45	22.73	59	30.10	36	32.43	71	24.40	53	26.50	54	26.73
Overall positive	99	24.63	23	23.71	24	18.32	30	26.32	22	36.67	50	25.25	48	24.49	22	19.82	77	26.46	45	22.50	54	26.73

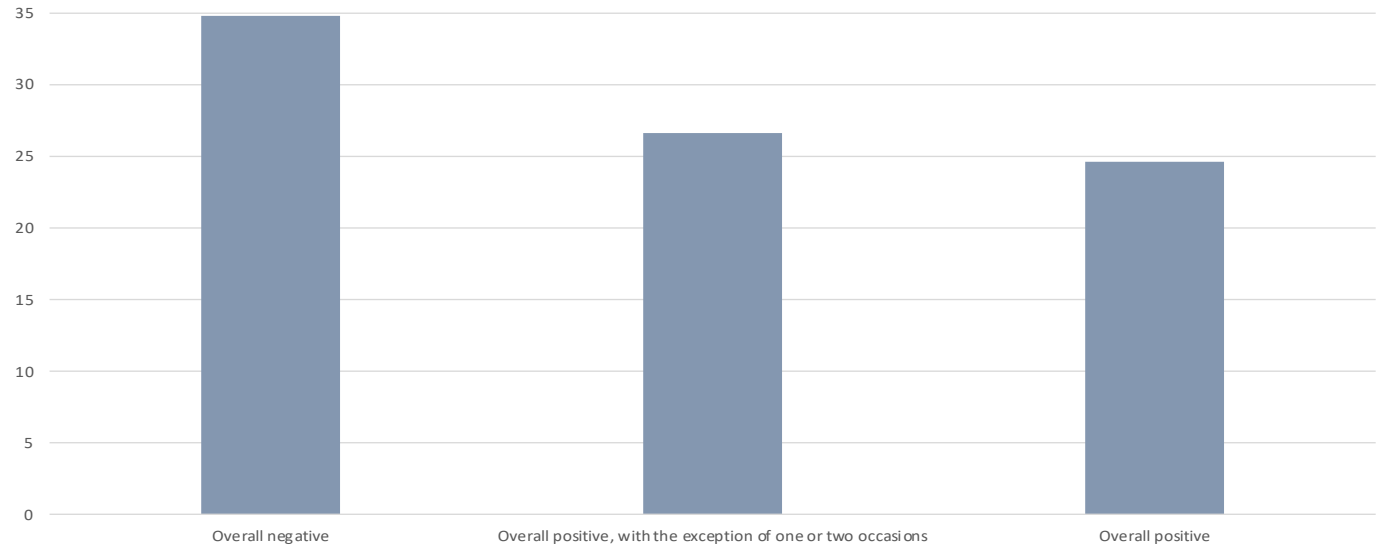


Figure 6.7: Healthcare professional communication

Table 6.12: Healthcare professional communication – subgroup variations

Healthcare professional communication	Reported less frequently	Reported more frequently
Overall negative	Aged under 18	Aged 18 to 44
Overall positive, with the exception of one or two occasions		
Overall positive	Developmental anomalies	Aged 65 plus

Table 6.13: Healthcare professional communication (Rationale for response)

Healthcare professional communication (reasons)	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Limited in understanding	134	33.33	20	29.85	38	46.91	30	31.58	13	40.63	27	28.42	6	18.75	96	35.82	38	28.36	109	37.07	24	22.64
Holistic (Two way, supportive and comprehensive conversations)	114	28.36	14	20.90	16	19.75	38	40.00	8	25.00	28	29.47	10	31.25	72	26.87	42	31.34	84	28.57	29	27.36
Dismissive (One way conversation/not empathetic)	66	16.42	10	14.93	19	23.46	16	16.84	3	9.38	11	11.58	7	21.88	49	18.28	17	12.69	52	17.69	14	13.21
Limited in not having time	34	8.46	5	7.46	9	11.11	8	8.42	1	3.13	6	6.32	5	15.63	25	9.33	9	6.72	26	8.84	8	7.55

Healthcare professional communication (reasons)	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Limited in understanding	134	33.33	23	23.71	41	31.30	47	41.23	23	38.33	72	36.36	59	30.10	37	33.33	97	33.33	67	33.50	67	33.17
Holistic (Two way, supportive and comprehensive conversations)	114	28.36	35	36.08	32	24.43	29	25.44	18	30.00	49	24.75	63	32.14	30	27.03	84	28.87	58	29.00	56	27.72
Dismissive (One way conversation/not empathetic)	66	16.42	10	10.31	33	25.19	17	14.91	6	10.00	33	16.67	33	16.84	16	14.41	50	17.18	35	17.50	31	15.35
Limited in not having time	34	8.46	7	7.22	11	8.40	13	11.40	3	5.00	19	9.60	15	7.65	7	6.31	27	9.28	18	9.00	16	7.92

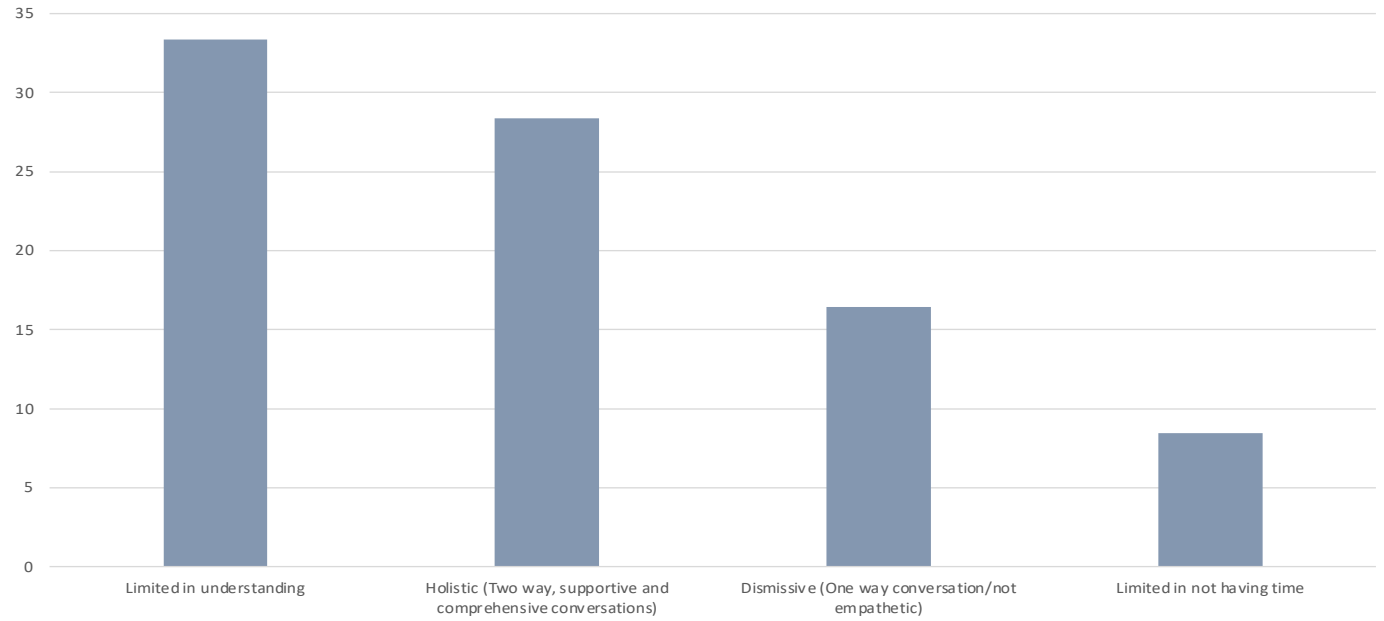


Figure 6.8: Healthcare professional communication (Rationale for response)

Table 6.14: Healthcare professional communication (Rationale for response) – subgroup variations

Healthcare professional communication (reasons)	Reported less frequently	Reported more frequently
Limited in understanding	Other rare condition Male	Diseases of the immune system
Holistic (Two way, supportive and comprehensive conversations)		Diseases of the nervous system
Dismissive (One way conversation/not empathetic)		
Limited in not having time		

Partners in health

The Partners in Health questionnaire (PIH) measures an individual's knowledge and confidence for managing their own health. The Partners in Health comprises a global score, 4 scales; knowledge, coping, recognition and treatment of symptoms, adherence to treatment and total score. A higher score denotes a better understanding and knowledge of disease. Summary statistics for the entire cohort are displayed alongside the possible range of each scale in the table below.

The overall scores for the cohort were in the highest quintile for Partners in health: Knowledge (median=26.00, IQR=8.00), Partners in health: Adherence to treatment (median=14.00, IQR=4.00), indicating very good knowledge, very good adherence to treatment.

The overall scores for the cohort were in the second highest quintile for Partners in health:Recognition and management of symptoms (median=19.00, IQR=5.75), Partners in health:Total score (median=72.00, IQR=20.00) indicating good recognition and management of symptoms, good overall ability to manage their health.

The overall scores for the cohort were in the middle quintile for Partners in health:Coping (median=14.00, IQR=7.00), indicating moderate coping.

Comparisons of Partners in health have been made based on condition, participant type, gender, age, education, location and socioeconomic status.

The **Partners in Health questionnaire (PIH)** measures an individual's knowledge and confidence for managing their own health.

The **Partners in health: knowledge** scale measures the participants knowledge of their health condition, treatments, their participation in decision making and taking action when they get symptoms. On average, participants in this study had very good knowledge about their condition and treatments.

The **Partners in health: coping** scale measures the participants ability to manage the effect of their health condition on their emotional well-being, social life and living a healthy life (diet, exercise, moderate alcohol and no smoking). On average, participants in this study had a moderate ability to manage the effects of their health condition.

The **Partners in health: treatment** scale measures the participants ability to take medications and complete treatments as prescribed and communicate with healthcare professionals to get the services that are needed and that are appropriate. On average participants in this study had a good ability to adhere to treatments and communicate with healthcare professionals.

The **Partners in health: recognition and management of symptoms** scale measures how well the participant attends all healthcare appointments, keeps track of signs and symptoms, and physical activities. On average participants in this study had very good recognition and management of symptoms.

The **Partners in health: total score** measures the overall knowledge, coping and confidence for managing their own health. On average participants in this study had good overall knowledge, coping and confidence for managing their own health.

Table 6.15: Partners in health summary statistics

Partners in health scale (n=362)	Mean	SD	Median	IQR	Possible range	Quintile
Knowledge	24.07	6.24	26.00	8.00	0 to 32	5
Coping	14.35	5.39	14.00	7.00	0 to 24	3
Recognition and management of symptoms	18.89	3.66	19.00	5.75	0 to 24	4
Adherence to treatment	13.12	3.18	14.00	4.00	0 to 16	5
Total score	70.44	14.39	72.00	20.00	0 to 96	4

Skewed distribution use median and IQR as measure of central tendency

Partners in health by condition

Comparisons were made by **condition**. There were 57 participants (15.75%) with developmental anomalies , 72 participants (19.89%) with diseases of the immune system , 93 participants (25.69%) with diseases of the nervous system , 29 participants (8.01%) with diseases of the skin , 86 participants (23.76%) with endocrine, nutritional or metabolic diseases , and 25 participants (6.91%) with other rare condition.

Assumptions for normality of residuals was not met, a Kruskal-Wallis test was used. Post hoc pairwise comparisons using Wilcoxon rank sum test was used to

identify the source of any differences identified in the Kruskal -Wallis test.

A Kruskal-Wallis test indicated a statistically significant difference in the **Partners in health: Knowledge scale** between groups, $\chi^2(5) = 11.38$ $p = 0.0443$. However, post hoc comparisons using the Tukey HSD test did not indicate any significant differences between groups.

A Kruskal-Wallis test indicated a statistically significant difference in the **Partners in health: Coping** scale between groups, $\chi^2(5) = 12.86$ $p = 0.0247$.

The largest significant difference was between participants in the Endocrine, nutritional or metabolic diseases subgroup (median = 15.00, IQR = 6.00), and participants in the Diseases of the skin subgroup (median = 11.00, IQR = 6.00, $p = 0.0160$).

A Kruskal-Wallis test indicated a statistically significant difference in the **Partners in health: Adherence to treatment** scale between groups, $\chi^2(5) = 12.99$ $p = 0.0235$. The largest significant difference was between participants in the Endocrine, nutritional or metabolic diseases subgroup (median = 15.00, IQR = 3.00), and participants in the Diseases of the skin subgroup (median = 12.00, IQR = 7.00, $p = 0.0097$).

A Kruskal-Wallis test indicated a statistically significant difference in the **Partners in health: Total score scale** between groups, $\chi^2(5) = 12.64$ $p = 0.0270$. The largest significant difference was between participants in the Endocrine, nutritional or metabolic diseases subgroup (median = 73.00, IQR = 13.00), and participants in the Diseases of the skin subgroup (median = 61.00, IQR = 16.00, $p = 0.0055$).

The **Partners in health: knowledge** scale measures the participants knowledge of their health condition, treatments, their participation in decision making and taking action when they get symptoms. On average, participants in the Diseases of the immune system subgroup scored higher than participants in the Diseases of the skin subgroup. This indicates that participants in the Diseases of the immune system subgroup had very good knowledge about their condition and treatments, and participants in the Diseases of the skin subgroup had good knowledge.

The **Partners in health: coping** scale measures the participants ability to manage the effect of their health condition on their emotional well-being, social life and living a healthy life (diet, exercise, moderate alcohol and no smoking). On average, participants in the Endocrine, nutritional or metabolic diseases subgroup scored higher than participants in the Diseases of the skin subgroup. This indicates that participants in the Endocrine, nutritional or metabolic diseases subgroup were good at coping with their condition, and participants in the Diseases of the skin subgroup were average at coping.

The **Partners in health: treatment** scale measures the participants ability to take medications and complete treatments as prescribed and communicate with healthcare professionals to get the services that are needed and that are appropriate. On average, participants in the Endocrine, nutritional or metabolic diseases subgroup scored higher than participants in the Diseases of the skin subgroup. This indicates that, treatment adherence was very good for participants in the Endocrine, nutritional or metabolic diseases subgroup, and good for participants in the Diseases of the skin subgroup.

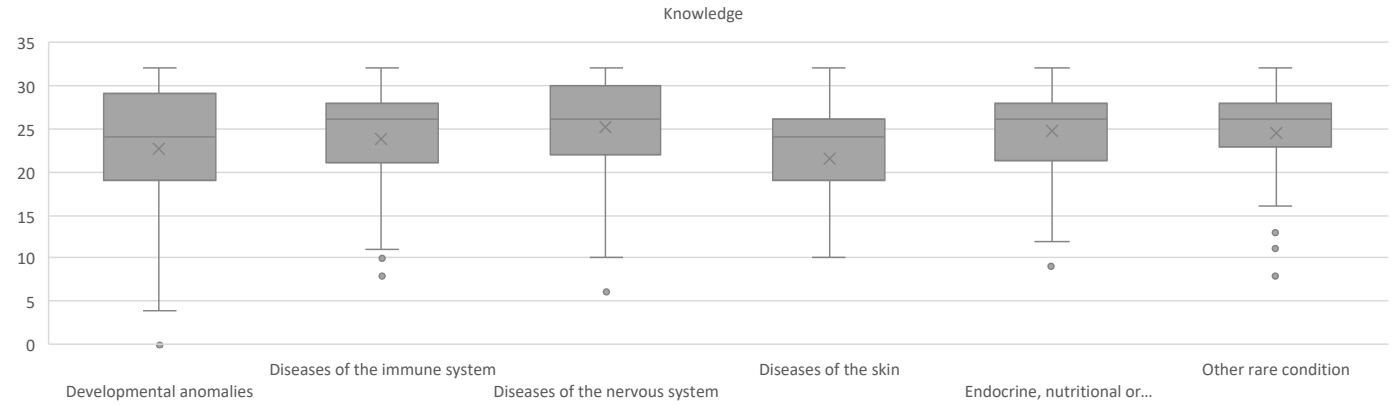
The **Partners in health: total score** measures the overall knowledge, coping and confidence for managing their own health. On average, participants in the Endocrine, nutritional or metabolic diseases subgroup had a higher score for quality of compared to the Diseases of the skin subgroup, however, both groups had good overall knowledge, coping and confidence for managing their own health.

Table 6.16: Partners in health by condition summary statistics and Kruskal-Wallis test

SF36 scale	Group	Number (n=362)	Percent	Median	IQR	C^2	dF	p-value
Knowledge	Developmental anomalies	57	15.75	24.00	10.00	11.38	5	0.0443*
	Diseases of the immune system	72	19.89	26.00	7.00			
	Diseases of the nervous system	93	25.69	26.00	8.00			
	Diseases of the skin	29	8.01	24.00	7.00			
	Endocrine, nutritional or metabolic diseases	86	23.76	26.00	6.75			
	Other rare condition	25	6.91	26.00	5.00			
Coping	Developmental anomalies	57	15.75	15.00	7.00	12.86	5	0.0247*
	Diseases of the immune system	72	19.89	14.00	8.00			
	Diseases of the nervous system	93	25.69	15.00	7.00			
	Diseases of the skin	29	8.01	11.00	6.00			
	Endocrine, nutritional or metabolic diseases	86	23.76	15.00	6.00			
	Other rare condition	25	6.91	16.00	8.00			
Recognition and management of symptoms	Developmental anomalies	57	15.75	20.00	6.00	4.29	5	0.5082
	Diseases of the immune system	72	19.89	19.00	5.00			
	Diseases of the nervous system	93	25.69	20.00	6.00			
	Diseases of the skin	29	8.01	18.00	3.00			
	Endocrine, nutritional or metabolic diseases	86	23.76	19.00	3.00			
	Other rare condition	25	6.91	20.00	5.00			
Adherence to treatment	Developmental anomalies	57	15.75	14.00	4.00	12.99	5	0.0235*
	Diseases of the immune system	72	19.89	14.00	3.00			
	Diseases of the nervous system	93	25.69	14.00	4.00			
	Diseases of the skin	29	8.01	12.00	7.00			
	Endocrine, nutritional or metabolic diseases	86	23.76	15.00	3.00			
	Other rare condition	25	6.91	13.00	5.00			
Total score	Developmental anomalies	57	15.75	72.00	26.00	12.64	5	0.0270*
	Diseases of the immune system	72	19.89	70.00	21.25			
	Diseases of the nervous system	93	25.69	73.00	17.00			
	Diseases of the skin	29	8.01	61.00	16.00			
	Endocrine, nutritional or metabolic diseases	86	23.76	73.00	13.00			
	Other rare condition	25	6.91	72.00	18.00			

Table6.17: Care coordination by condition one-way post hoc Wilcoxon rank sum test

Partners in health scale		Developmental anomalies	Diseases of the immune system	Diseases of the nervous system	Diseases of the skin	Endocrine, nutritional or metabolic diseases
Knowledge	Diseases of the immune system	0.6780	-	-	-	-
	Diseases of the nervous system	0.1670	0.2090	-	-	-
	Diseases of the skin	0.4710	0.2660	0.0820	-	-
	Endocrine, nutritional or metabolic diseases	0.3410	0.4710	0.4710	0.1670	-
	Other rare condition	0.4710	0.5580	0.7510	0.2090	0.8650
Coping	Diseases of the immune system	0.4870	-	-	-	-
	Diseases of the nervous system	0.4870	0.8630	-	-	-
	Diseases of the skin	0.0160*	0.0560	0.0480*	-	-
	Endocrine, nutritional or metabolic diseases	0.8070	0.4870	0.5680	0.0160*	-
	Other rare condition	0.8630	0.4870	0.4870	0.0450*	0.7750
Adherence to treatment	Diseases of the immune system	0.8147	-	-	-	-
	Diseases of the nervous system	0.8905	0.8147	-	-	-
	Diseases of the skin	0.1245	0.0888	0.1245	-	-
	Endocrine, nutritional or metabolic diseases	0.2674	0.2674	0.1737	0.0097*	-
	Other rare condition	0.8147	0.7740	0.8147	0.3752	0.3752
Total score	Diseases of the immune system	0.8255	-	-	-	-
	Diseases of the nervous system	0.8255	0.7054	-	-	-
	Diseases of the skin	0.1128	0.0716	0.0189*	-	-
	Endocrine, nutritional or metabolic diseases	0.7054	0.3654	0.8255	0.0055*	-
	Other rare condition	0.8255	0.8255	0.9396	0.0716	0.8255



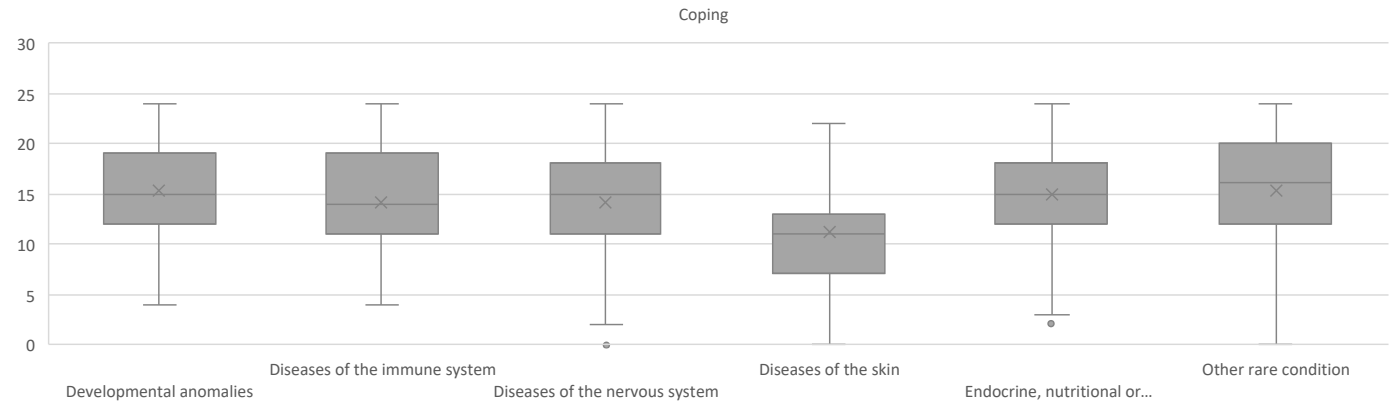


Figure 6.9: Boxplot of Partners in health: knowledge by condition

Figure 6.10: Boxplot of Partners in health: coping by condition

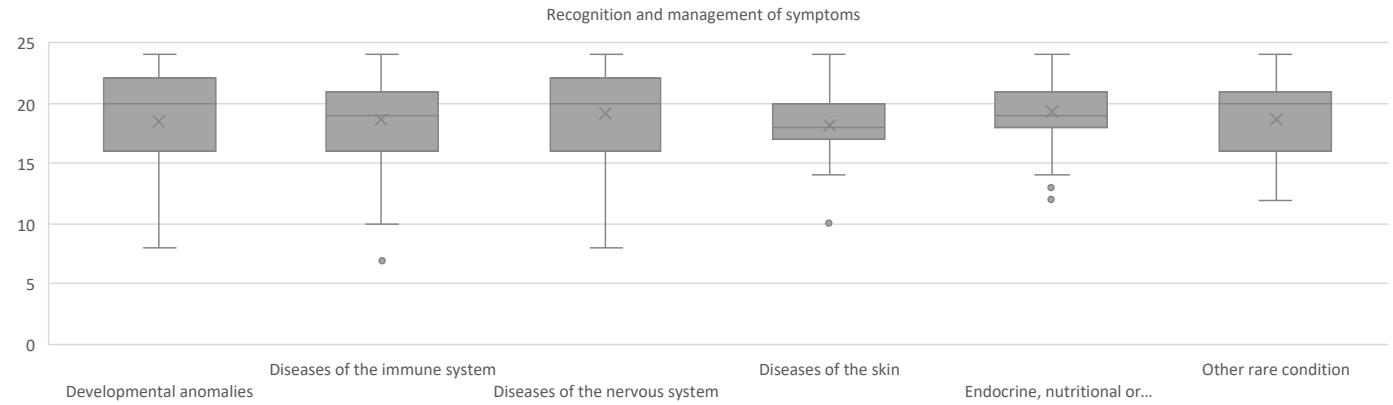


Figure 6.11: Boxplot of Partners in health: recognition and management of symptoms by condition

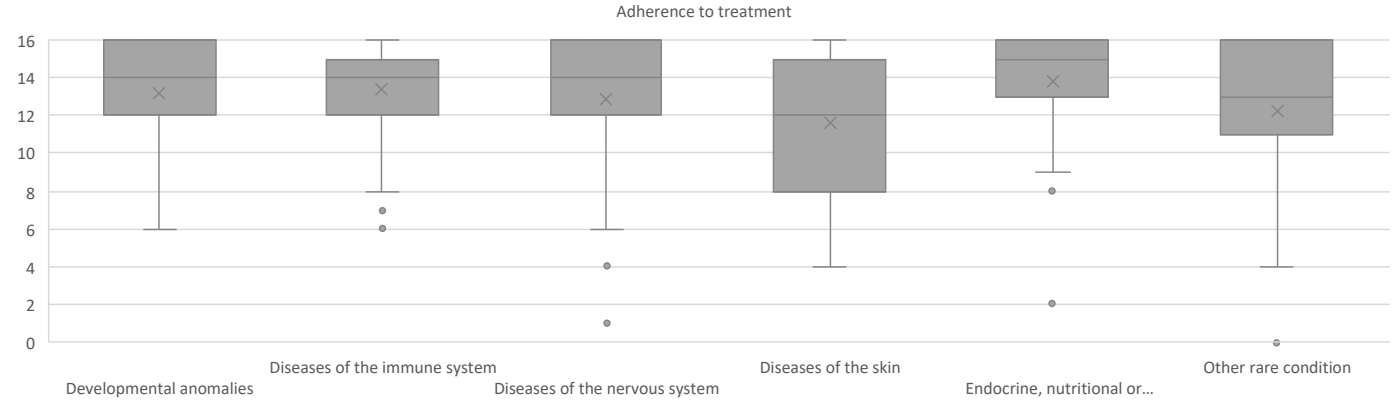


Figure 6.12: Boxplot of Partners in health: adherence to treatment by condition

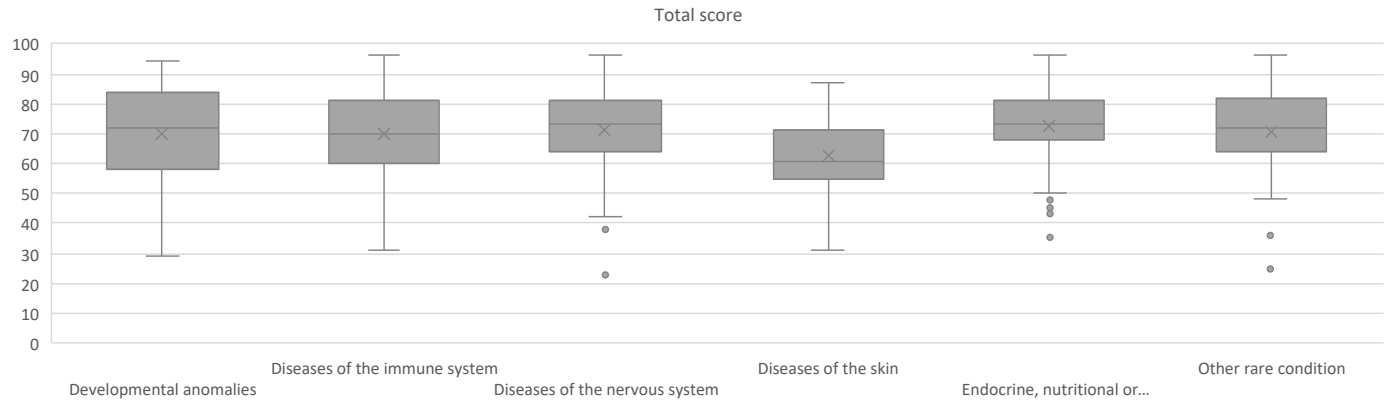


Figure 6.13: Boxplot of Partners in health Total score by condition

Partners in health by type of participant

Comparisons were made by **type of participant** there were 241 participants (66.57%) with person with condition and, 121 participants (33.43%) with carer.

Assumptions for normality and variance for a two-sample t-test were not met, a Wilcoxon rank sum test with continuity correction was used.
Wilcoxon rank sum tests with continuity correction indicated that the median score for the Partners in health Knowledge scale [W = 11891.00 , p = 0.0041] was significantly lower for participants in the Person with condition subgroup (Median = 25.00, IQR = 8.00) compared to participants in the Carer subgroup (Median = 27.00, IQR = 9.00.

Wilcoxon rank sum tests with continuity correction indicated that the median score for the Partners in health Recognition and management of symptoms scale [W = 11137.00 , p = 0.0002] was significantly lower for participants in the Person with condition subgroup (Median = 19.00, IQR = 5.00) compared to participants in the Carer subgroup (Median = 20.00, IQR = 5.00.

Wilcoxon rank sum tests with continuity correction indicated that the median score for the Partners in health Total score scale [W = 11925.00 , p = 0.0047] was significantly lower for participants in the Person with condition subgroup (Median = 71.00, IQR = 20.00) compared to participants in the Carer subgroup (Median = 75.00, IQR = 18.00.

The **Partners in health: knowledge** scale measures the participants knowledge of their health condition, treatments, their participation in decision making and taking action when they get symptoms. On average, participants in the Carer subgroup scored higher than participants in the Person with condition subgroup. This indicates that participants in the Carer subgroup had very good knowledge about their condition and treatments, and participants in the Person with condition subgroup had good knowledge.

The **Partners in health: recognition and management of symptoms** scale measures how well the participant attends all healthcare appointments, keeps track of signs and symptoms, and physical activities. On average, participants in the Carer subgroup scored higher than participants in the Person with condition subgroup. This indicates that recognition and management of symptoms was very good for participants in the Carer subgroup, and good for participants in the Person with condition subgroup.

The **Partners in health: total score** measures the overall knowledge, coping and confidence for managing their own health. On average, participants in the Carer subgroup had a higher score for quality of compared to the Person with condition subgroup, however, both groups had good overall knowledge, coping and confidence for managing their own health.

Table 6.18: Partners in health by type of participant summary statistics and Wilcoxon rank sum tests with continuity correction

Partners in health scale	Group	Number (n=362)	Percent	Median	IQR	W	p-value
Knowledge	Person with condition	241	66.57	25.00	8.00	11891	0.0041*
	Carer	121	33.43	27.00	9.00		
Coping	Person with condition	241	66.57	14.00	7.00	13584	0.2876
	Carer	121	33.43	15.00	8.00		
Recognition and management of symptoms	Person with condition	241	66.57	19.00	5.00	11137	0.0002*
	Carer	121	33.43	20.00	5.00		
Adherence to treatment	Person with condition	241	66.57	14.00	4.00	13414	0.2069
	Carer	121	33.43	14.00	4.00		
Total score	Person with condition	241	66.57	71.00	20.00	11925	0.0047*
	Carer	121	33.43	75.00	18.00		

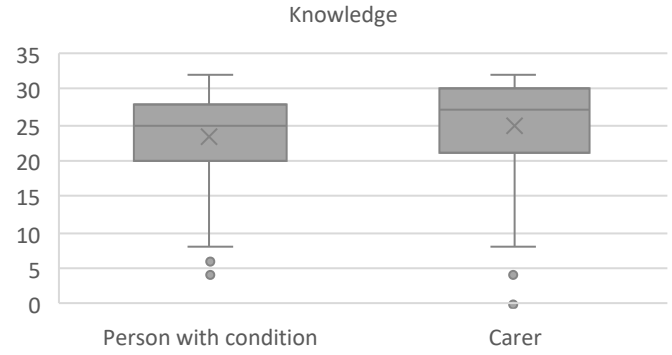


Figure 6.14: Boxplot of Partners in health: knowledge by type of participant

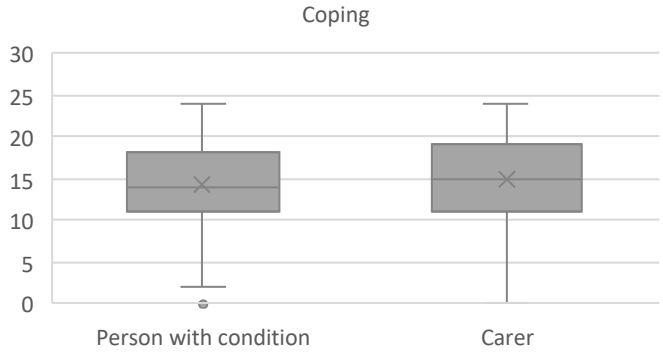


Figure 6.15: Boxplot of Partners in health: coping by type of participant

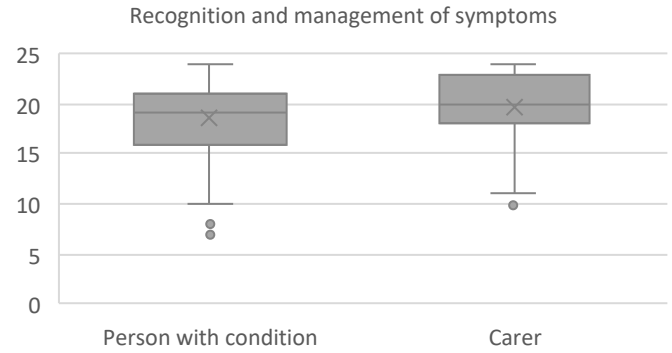


Figure 6.16: Boxplot of Partners in health: recognition and management of symptoms by type of participant

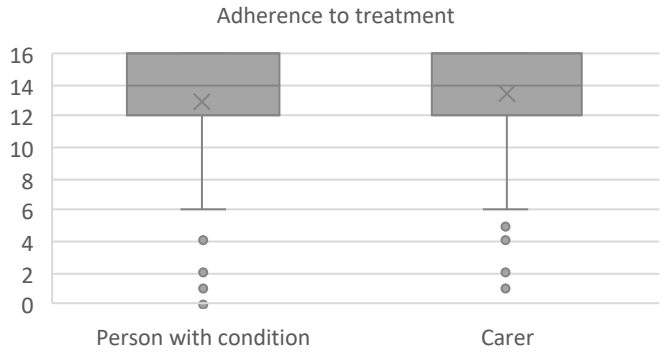


Figure 6.17: Boxplot of Partners in health: adherence to treatment by type of participant

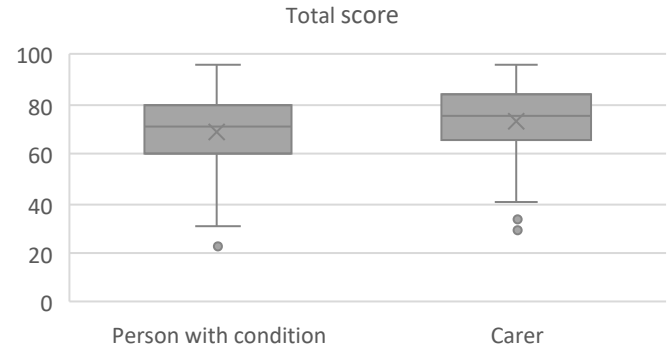


Figure 6.18: Boxplot of Partners in health Total score by type of participant

Partners in health by gender

Comparisons were made by **gender**, there were 272 female participants (75.56%), and 88 male particpants (24.44%).

Assumptions for normality and variance for a two-sample t-test were not met, a Wilcoxon rank sum test with continuity correction was used.

No significant differences were observed between participants by **gender** for any of the Partners in health scales.

Table 6.19: Partners in health by gender summary statistics and Wilcoxon test

Partners in health scale	Group	Number (n=360)	Percent	Median	IQR	W	p-value
Knowledge	Female	272	75.56	25.00	8.00	11443.00	0.5357
	Male	88	24.44	26.00	6.50		
Coping	Female	272	75.56	14.00	7.00	10584.00	0.1022
	Male	88	24.44	15.50	9.00		
Recognition and management of symptoms	Female	272	75.56	19.00	5.00	11131.00	0.3219
	Male	88	24.44	20.00	5.00		
Adherence to treatment	Female	272	75.56	14.00	4.00	11056.00	0.2750
	Male	88	24.44	14.00	4.00		
Total score	Female	272	75.56	71.00	20.00	10658.00	0.1228
	Male	88	24.44	73.50	18.25		

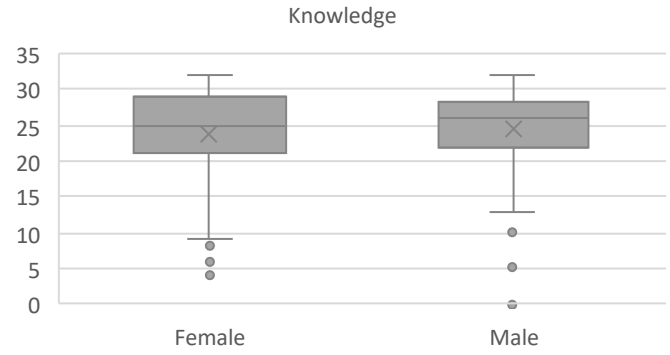


Figure 6.19: Boxplot of Partners in health: knowledge by gender

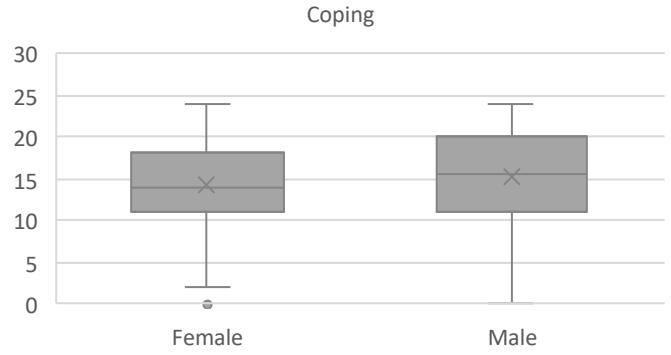


Figure 6.20: Boxplot of Partners in health: coping by gender

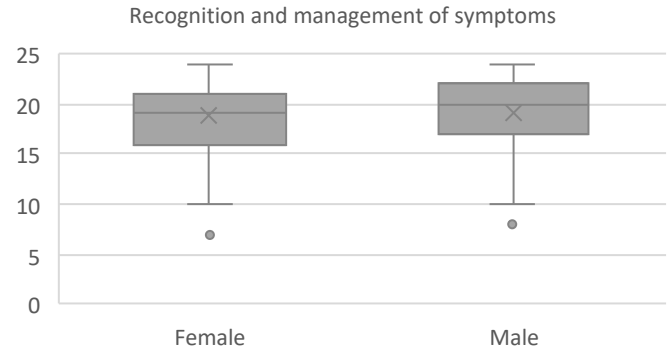


Figure 6.21: Boxplot of Partners in health: recognition and management of symptoms by gender

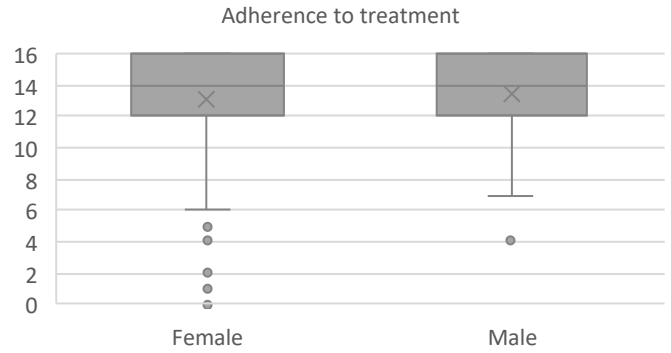


Figure 6.22: Boxplot of Partners in health: adherence to treatment by gender

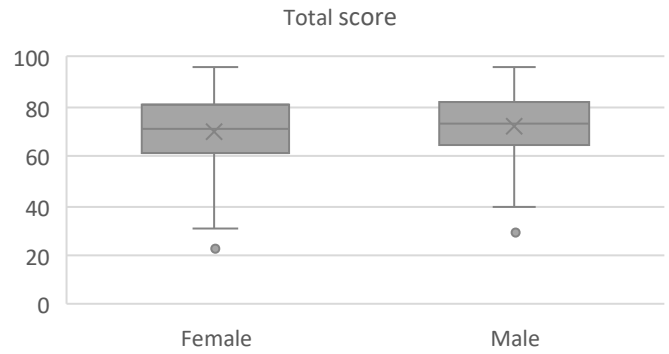


Figure 6.23: Boxplot of Partners in health Total score by gender

Partners in health by age

Comparisons were made by **age** of person with condition. There were 87 participants (24.03%) with aged under 18, 120 participants (33.15%) with aged 18 to 44, 102 participants (28.18%) with aged 45 to 64, and 53 participants (14.64%) with aged 65 or older.

Assumptions for normality of residuals was not met, a Kruskal-Wallis test was used. Post hoc pairwise comparisons using Wilcoxon rank sum test was used to identify the source of any differences identified in the Kruskal -Wallis test.

A Kruskal-Wallis test indicated a statistically significant difference in the Partners in health: Knowledge scale between groups, $\chi^2(3) = 29.64$ $p<0.0001$. The largest significant difference was between Aged under 18 (median = 26, IQR = 8.5), and Aged 18 to 44 (median = 24, IQR = 8, p = <0.0001).

A Kruskal-Wallis test indicated a statistically significant difference in the Partners in health: Coping scale between groups, $\chi2(3) = 8.34$ P = 0.0394. The largest significant difference was between Aged 65 or older (median = 17, IQR = 9.25), and Aged 18 to 44 (median = 12, IQR = 6, p = 0.037).

A Kruskal-Wallis test indicated a statistically significant difference in the Partners in health: Recognition and management of symptoms scale between groups, $\chi^2(3) = 29.24$ $p<0.0001$. The largest significant difference was between Aged under 18 (median = 20, IQR = 5.5), and Aged 18 to 44 (median = 19, IQR = 5, p = <0.0001).

A Kruskal-Wallis test indicated a statistically significant difference in the Partners in health: Adherence to treatment scale between groups, $\chi^2(3) = 25.21$ $p<0.0001$. The largest significant difference was between Aged 65 or older (median = 15, IQR = 2), and Aged 18 to 44 (median = 13, IQR = 4, p = 0.0001).

A Kruskal-Wallis test indicated a statistically significant difference in the Partners in health: Total score scale between groups, $\chi^2(3) = 32.1$ $p<0.0001$. The largest significant difference was between Aged under 18 (median = 74, IQR = 20.5), and Aged 18 to 44 (median = 69, IQR = 19, p = <0.0001).

The **Partners in health: knowledge** scale measures the participants knowledge of their health condition, treatments, their participation in decision making and taking action when they get symptoms. On average, participants in the Aged under 18subgroup scored higher than participants in the Aged 18 to 44subgroup. This indicates that participants in the Aged under 18subgroup had very good knowledge about their condition and treatments, and participants in the Aged 18 to 44subgroup had good knowledge.

The **Partners in health: coping** scale measures the participants ability to manage the effect of their health condition on their emotional well-being, social life and living a healthy life (diet, exercise, moderate alcohol and no smoking). On average, participants in the Aged 65 or older subgroup scored higher than participants in the Aged 18 to 44subgroup. This indicates that participants in the Aged 65 or older subgroup were good at coping with their condition, and participants in the Aged 18 to 44subgroup were average at coping.

The **Partners in health: recognition and management of symptoms** scale measures how well the participant attends all healthcare appointments, keeps track of signs and symptoms, and physical activities. On average, participants in the Aged under 18 subgroup scored higher than participants in the Aged 18 to 44 subgroup. This indicates that recognition and management of symptoms was very good for

participants in the Aged under 18 subgroup, and good for participants in the Aged 18 to 44 subgroup.

The **Partners in health:adherence to treatment** scale measures the participants ability to take medications and complete treatments as prescribed and communicate with healthcare professionals to get the services that are needed and that are appropriate. On average, participants in the Aged 65 or older subgroup had a higher total score for navigation compared to Aged 18 to 44, however both groups had very good treatment adherence.

The **Partners in health: total score** measures the overall knowledge, coping and confidence for managing their own health. On average, participants in the Aged under 18subgroup had a higher score for quality of compared to the Aged 18 to 44 subgroup, however, both groups had good overall knowledge, coping and confidence for managing their own health.

Table 6.20: Partners in health by age summary statistics and Kruskal-Wallis test

Partners in health scale	Group	Number (n=362)	Percent	Median	IQR	C^2	dF	p-value
Knowledge	Aged under 18	87	24.03	26.00	8.50	29.64	3	<0.0001*
	Aged 18 to 44	120	33.15	24.00	8.00			
	Aged 45 to 64	102	28.18	25.00	7.00			
	Aged 65 or older	53	14.64	28.00	5.25			
Coping	Aged under 18	87	24.03	16.00	7.00	8.34	3	0.0394*
	Aged 18 to 44	120	33.15	12.00	6.00			
	Aged 45 to 64	102	28.18	14.00	8.00			
	Aged 65 or older	53	14.64	17.00	9.25			
Recognition and management of symptoms	Aged under 18	87	24.03	20.00	5.50	29.24	3	<0.0001*
	Aged 18 to 44	120	33.15	19.00	5.00			
	Aged 45 to 64	102	28.18	19.00	5.00			
	Aged 65 or older	53	14.64	20.00	4.00			
Adherence to treatment	Aged under 18	87	24.03	14.00	4.00	25.21	3	<0.0001*
	Aged 18 to 44	120	33.15	13.00	4.00			
	Aged 45 to 64	102	28.18	14.00	4.00			
	Aged 65 or older	53	14.64	15.00	2.00			
Total score	Aged under 18	87	24.03	74.00	20.50	32.10	3	<0.0001*
	Aged 18 to 44	120	33.15	69.00	19.00			
	Aged 45 to 64	102	28.18	72.00	20.50			
	Aged 65 or older	53	14.64	80.00	15.75			

Table6.21: Care coordination by age one-way post hoc Wilcoxon rank sum test

Partners in health scale	Group	Aged under 18	Aged 18 to 44	Aged 45 to 64
Knowledge	Aged 18 to 44	<0.0001*	-	-
	Aged 45 to 64	0.0310*	0.0134	-
	Aged 65 or older	0.8293	0.0002*	0.0552
Coping	Aged 18 to 44	0.1950	-	-
	Aged 45 to 64	0.6000	0.2900	-
	Aged 65 or older	0.2900	0.0370*	0.2230
Recognition and management of symptoms	Aged 18 to 44	<0.0001*	-	-
	Aged 45 to 64	0.0067*	0.0245*	-
	Aged 65 or older	0.1183	0.0061*	0.3364
Adherence to treatment	Aged 18 to 44	0.0001*	-	-
	Aged 45 to 64	0.5543	0.0031*	-
	Aged 65 or older	0.4797	0.0001*	0.3661
Total score	Aged 18 to 44	<0.0001*	-	-
	Aged 45 to 64	0.0500*	0.0080*	-
	Aged 65 or older	0.8420	0.0001*	0.0590

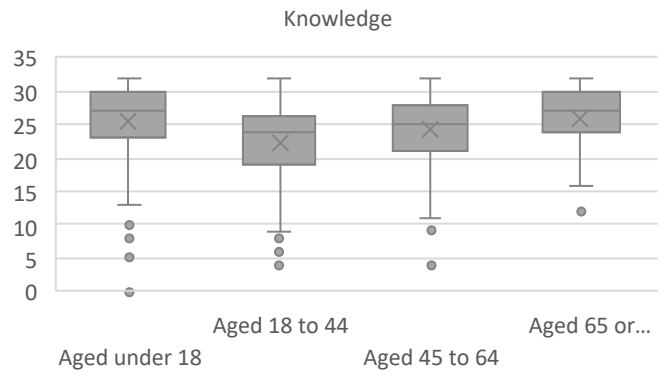


Figure 6.24: Boxplot of Partners in health: knowledge by age

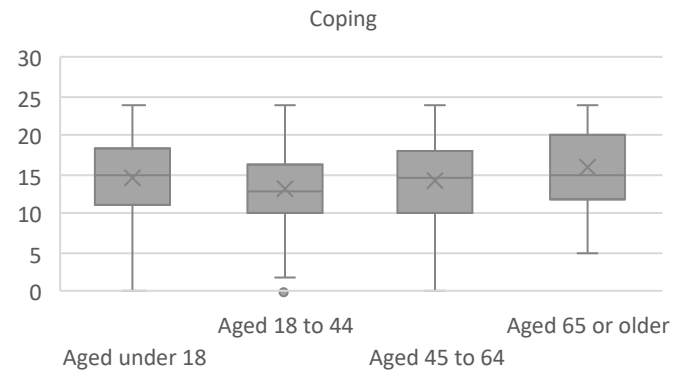


Figure 6.25: Boxplot of Partners in health: coping by age

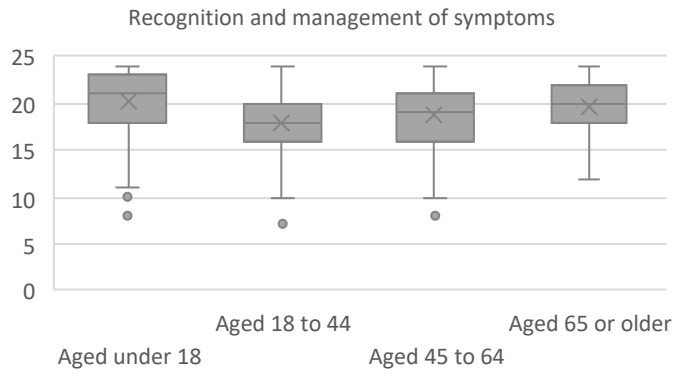


Figure 6.26: Boxplot of Partners in health: recognition and management of symptoms by age

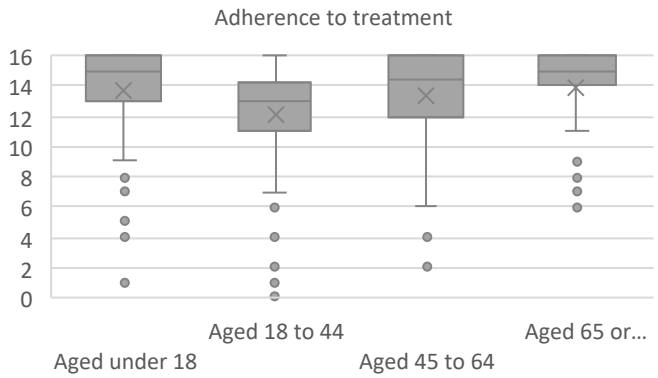


Figure 6.27: Boxplot of Partners in health: adherence to treatment by age

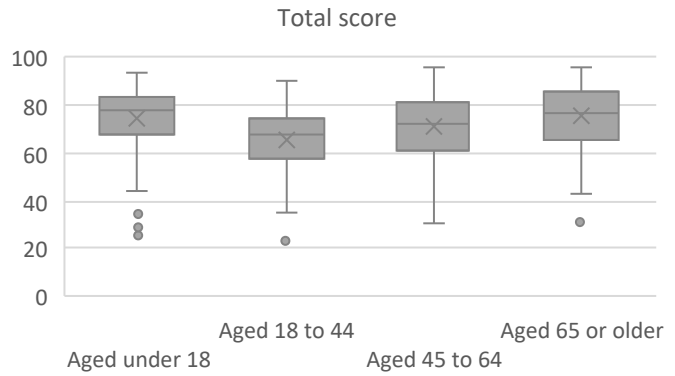


Figure 6.28: Boxplot of Partners in health Total score by age

Partners in health by education

Comparisons were made by **education** status, between those with trade or high school qualifications (n=175, 49.44%), and those with a university qualification (n=179, 50.56%).

Assumptions for normality and variance for a two-sample t-test were not met, a Wilcoxon rank sum test with continuity correction was used.

No significant differences were observed between participants by **education** for any of the Partners in health scales.

Table 6.22: Partners in health by education summary statistics and Wilcoxon test

Partners in health scale	Group	Number (n=354)	Percent	Median	IQR	W	p-value
Knowledge	Trade or high school	175	49.44	25.00	8.50	14980.00	0.4781
	University	179	50.56	26.00	8.00		
Coping	Trade or high school	175	49.44	14.00	7.00	14682.00	0.3073
	University	179	50.56	15.00	8.00		
Recognition and management of symptoms	Trade or high school	175	49.44	19.00	6.00	15652.00	0.9917
	University	179	50.56	20.00	4.00		
Adherence to treatment	Trade or high school	175	49.44	14.00	5.00	15165.00	0.5998
	University	179	50.56	14.00	4.00		
Total score	Trade or high school	175	49.44	71.00	20.50	14452.00	0.2084
	University	179	50.56	72.00	18.00		

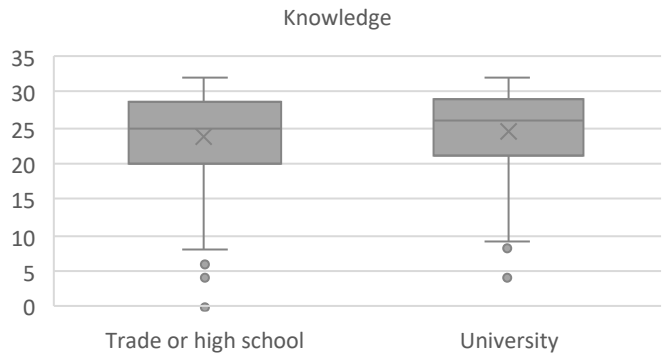


Figure 6.29: Boxplot of Partners in health: knowledge by education

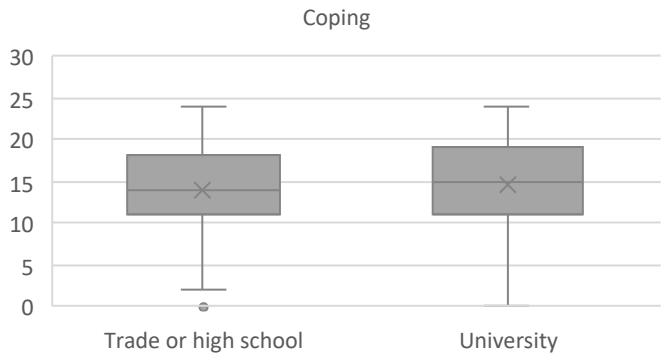


Figure 6.30: Boxplot of Partners in health: coping by education

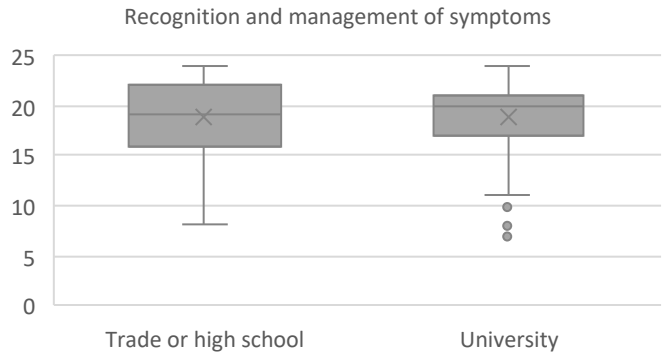


Figure 6.31: Boxplot of Partners in health: recognition and management of symptoms by education

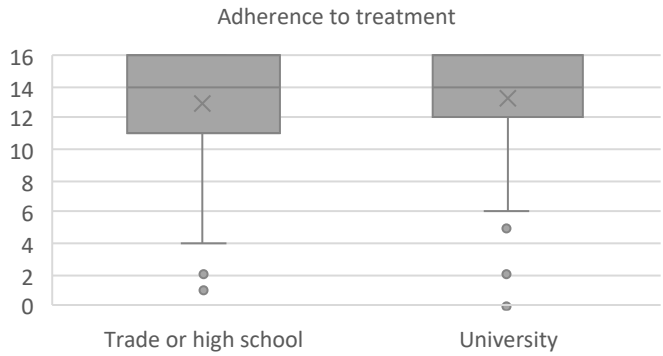


Figure 6.32: Boxplot of Partners in health: adherence to treatment by education

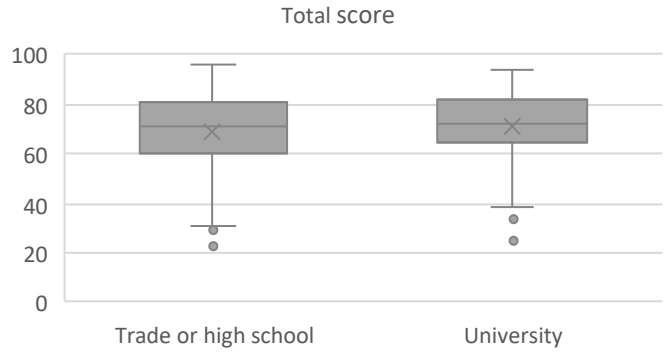


Figure 6.33: Boxplot of Partners in health Total score by education

Partners in health by location

The **location** of participants was evaluated by postcode using the Australian Statistical Geography Maps (ASGS) Remoteness areas accessed from the Australian Bureau of Statistics. Those living in regional or remote areas (n=103, 28.45%) were compared to those living in a metropolitan area (n=259, 71.55%).

Assumptions for normality and variance for a two-sample t-test were not met, a Wilcoxon rank sum test with continuity correction was used.

No significant differences were observed between participants by **location** for any of the Partners in health scales.

Table 6.23: Partners in health by location summary statistics and Wilcoxon test

Partners in health scale	Group	Number (n=362)	Percent	Median	IQR	W	p-value
Knowledge	Regional or remote	103	28.45	26.00	7.00	13282.00	0.9502
	Metropolitan	259	71.55	25.00	8.00		
Coping	Regional or remote	103	28.45	15.00	7.50	13535.00	0.8269
	Metropolitan	259	71.55	14.00	7.00		
Recognition and management of symptoms	Regional or remote	103	28.45	19.00	5.00	13667.00	0.7137
	Metropolitan	259	71.55	19.00	6.00		
Adherence to treatment	Regional or remote	103	28.45	14.00	4.00	14022.00	0.4399
	Metropolitan	259	71.55	14.00	4.00		
Total score	Regional or remote	103	28.45	73.00	17.50	13842.00	0.5758
	Metropolitan	259	71.55	72.00	20.50		

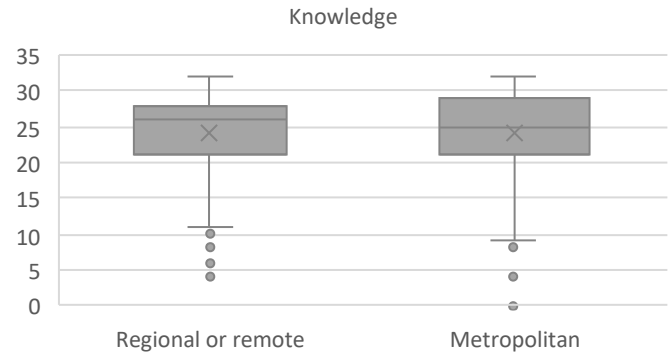


Figure 6.34: Boxplot of Partners in health: knowledge by location

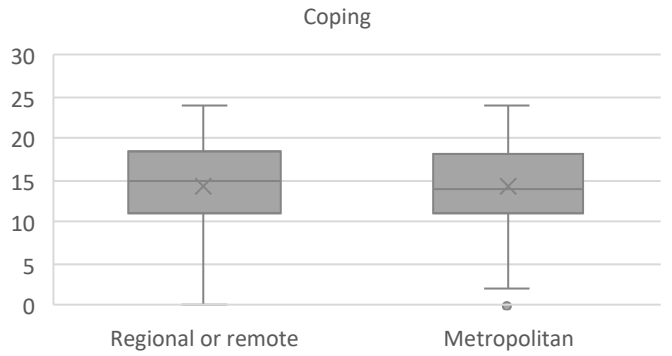


Figure 6.35: Boxplot of Partners in health: coping by location

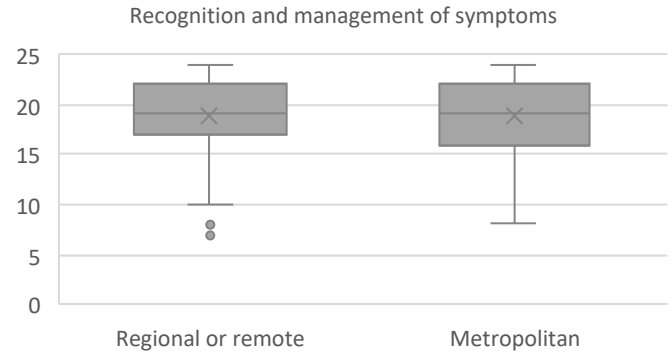


Figure 6.36: Boxplot of Partners in health: recognition and management of symptoms by location

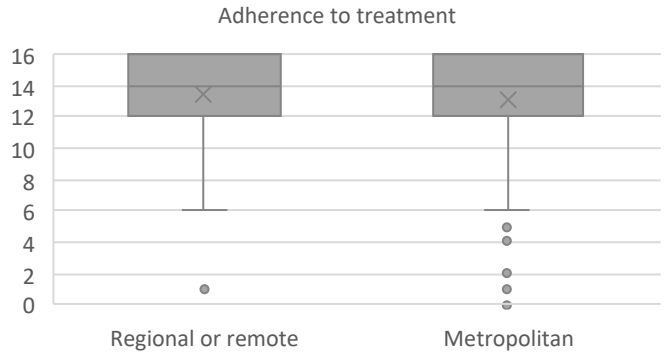


Figure 6.37: Boxplot of Partners in health: adherence to treatment by location

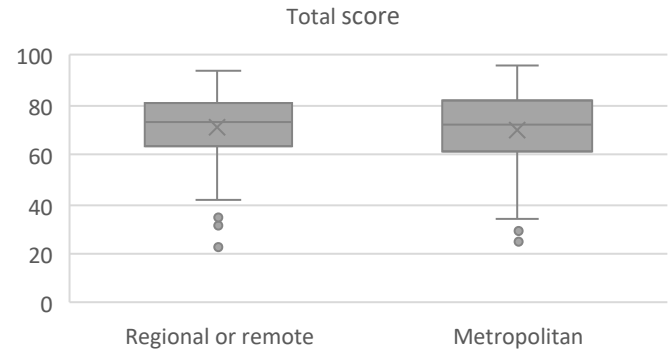


Figure 6.38: Boxplot of Partners in health Total score by location

Partners in health by socioeconomic status

Comparisons were made by **socioeconomic status**, using the Socio-economic Indexes for Areas (SEIFA) (www.abs.gov.au), SEIFA scores range from 1 to 10, a higher score denotes a higher level of advantage. Participants with a mid to low SEIFA score of 1-6 (n=184, 50.83%) compared to those with a higher SEIFA score of 7-10 (n=178, 49.17%).

Assumptions for normality and variance for a two-sample t-test were not met, a Wilcoxon rank sum test with continuity correction was used.

Wilcoxon rank sum tests with continuity correction indicated that the median score for the Partners in health Knowledge scale [W = 14326.00 , p = 0.0390] was significantly lower for participants in the Mid to low status subgroup (Median = 25.00, IQR = 8.00) compared to participants in the **Higher status** subgroup (Median = 26.00, IQR = 7.00.

Wilcoxon rank sum tests with continuity correction indicated that the median score for the Partners in health Recognition and management of symptoms scale [W = 14360.00 , p = 0.0420] was significantly lower for participants in the Mid to low status subgroup (Median = 19.00, IQR = 5.00) compared to participants in the Higher status subgroup (Median = 20.00, IQR = 5.00.

Wilcoxon rank sum tests with continuity correction indicated that the median score for the Partners in health Total score scale [W = 14090.00 , p = 0.0216] was significantly lower for participants in the Mid to low status subgroup (Median = 71.00, IQR = 21.25) compared to participants in the Higher status subgroup (Median = 73.00, IQR = 18.00.

The **Partners in health: knowledge** scale measures the participants knowledge of their health condition, treatments, their participation in decision making and taking action when they get symptoms. On average, participants in the Higher status subgroup scored higher than participants in the Mid to low status subgroup. This indicates that participants in the Higher status subgroup had very good knowledge about their condition and treatments, and participants in the Mid to low status subgroup had good knowledge.

The **Partners in health: recognition and management of symptoms** scale measures how well the participant attends all healthcare appointments, keeps track of signs and symptoms, and physical activities. On average, participants in the Higher status subgroup scored higher than participants in the Mid to low status subgroup. This indicates that recognition and management of symptoms was very good for participants in the Higher status subgroup, and good for participants in the Mid to low status subgroup.

The **Partners in health: total score** measures the overall knowledge, coping and confidence for managing their own health. On average, participants in the Higher status subgroup had a higher score for quality of compared to the Mid to low status subgroup, however, both groups had good overall knowledge, coping and confidence for managing their own health.

Table 6.24: Partners in health by socioeconomic status summary statistics and Wilcoxon test

Partners in health scale	Group	Number (n=362)	Percent	Median	IQR	W	p-value
Knowledge	Mid to low status	184	50.83	25.00	8.00	14326.00	0.03908
	Higher status	178	49.17	26.00	7.00		
Coping	Mid to low status	184	50.83	14.00	6.00	14620.00	0.07728
	Higher status	178	49.17	15.00	8.00		
Recognition and management of symptoms	Mid to low status	184	50.83	19.00	5.00	14360.00	0.04208
	Higher status	178	49.17	20.00	5.00		
Adherence to treatment	Mid to low status	184	50.83	14.00	4.00	15096.00	0.19158
	Higher status	178	49.17	14.00	4.00		
Total score	Mid to low status	184	50.83	71.00	21.25	14090.00	0.02168
	Higher status	178	49.17	73.00	18.00		

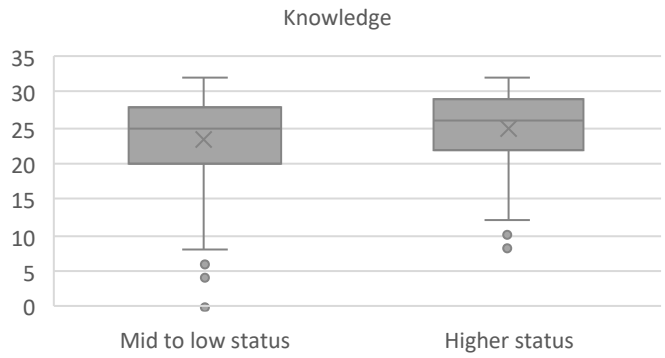


Figure 6.39: Boxplot of Partners in health: knowledge by socioeconomic status

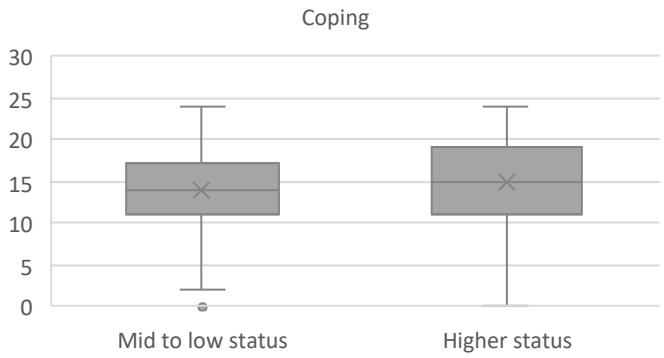


Figure 6.40: Boxplot of Partners in health: coping by socioeconomic status

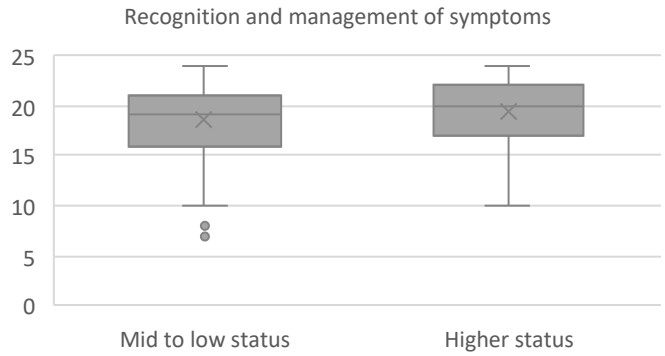


Figure 6.41: Boxplot of Partners in health: recognition and management of symptoms by socioeconomic status

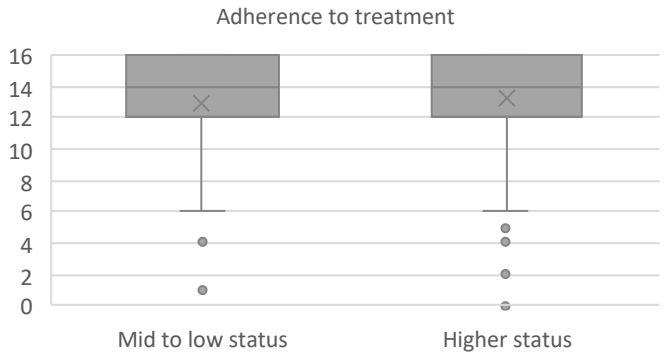


Figure 6.42: Boxplot of Partners in health: adherence to treatment by socioeconomic status

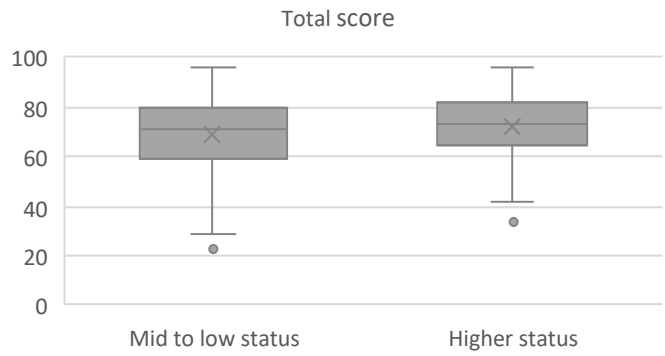


Figure 6.43: Boxplot of Partners in health Total score by socioeconomic status

Ability to take medicine as prescribed

Participants were asked about their ability to take medicines as prescribed. The majority of the participants responded that they took medicine as prescribed all the time (n=173, 57.10%), and 120 participants (39.60%) responded that they took medicines as prescribed most of the time. There were 6 participants (1.98%) that sometimes took medicines as prescribed.

Table 6.25: Ability to take medicine as prescribed

Ability to take medicine and stick to prescription	Number (n=303)	Percent
All of the time	173	57.10
Most of the time	120	39.60
Sometimes	6	1.98
Rarely	4	1.32

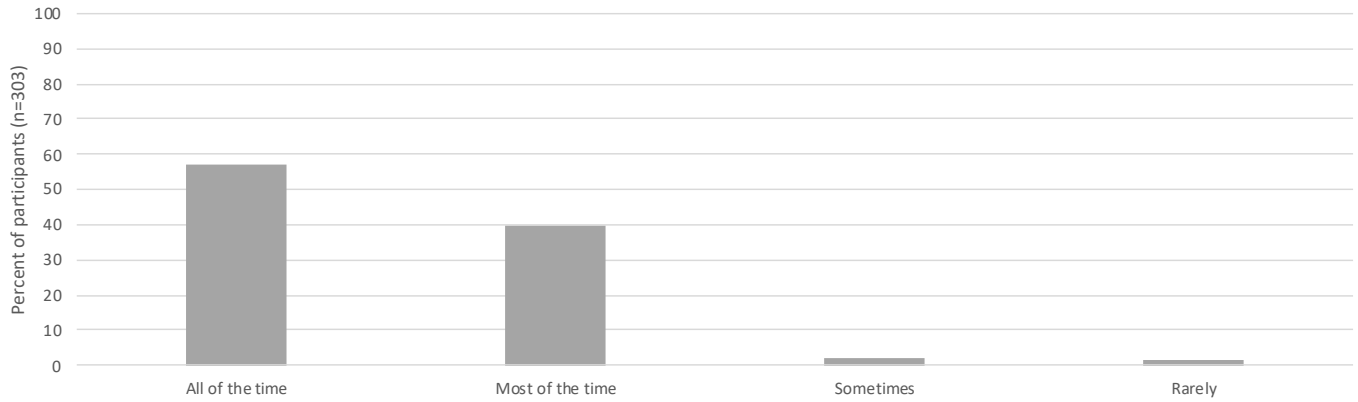


Figure 6.44: Ability to take medicine as prescribed

Information given by health professionals

Participants were asked about what type of information they were given by healthcare professionals, information about treatment options (n=188, 58.02%), disease management (n=147, 45.37%), disease cause (n=119, 36.73%) and, physical activity (n=85, 26.23%) were most frequently given to participants by healthcare professionals, and, information about interpret test results (n=54, 16.67%), clinical trials (n=43, 13.27%) and, complementary therapies (n=34, 10.49%) were given least often.

Table 6.26: Information given by health professionals

Information given by health professionals	Number (n=324)	Percent
Disease Cause	119	36.73
Treatment options	188	58.02
Disease management	147	45.37
Complementary therapies	34	10.49
Interpret test results	54	16.67
Clinical trials	43	13.27
Dietary	78	24.07
Physical activity	85	26.23
Psychological/ social support	69	21.30
Hereditary considerations	76	23.46

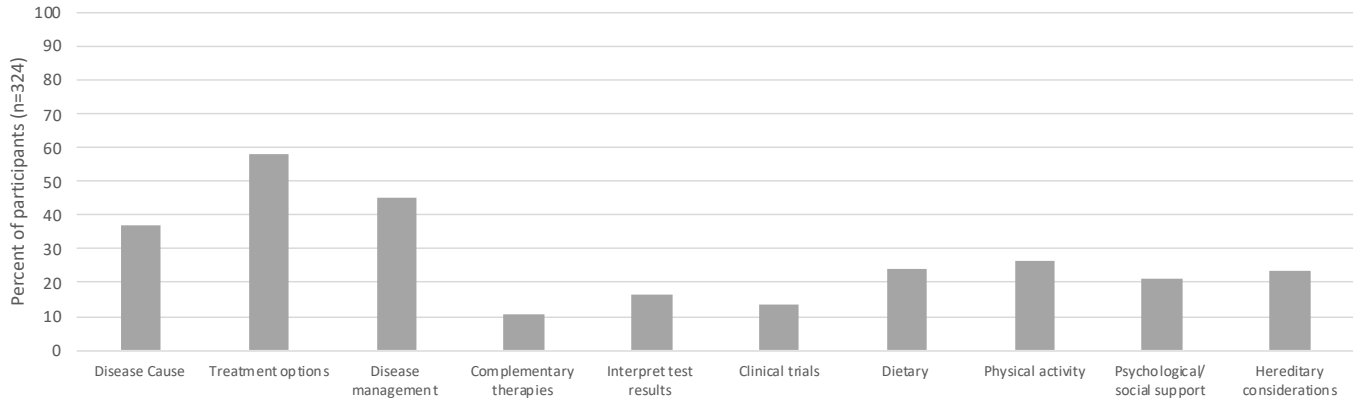


Figure 6.45: Information given by health professionals

Information searched independently

Participants were then asked after receiving information from healthcare professionals, what information did they need to search for independently. The topics participants most often searched for were disease management (n=212, 65.43%), treatment options (n=210, 64.81%), disease cause (n=207, 63.89%) and, complementary therapies (n=167, 51.54%) were most frequently given to participants by healthcare professionals, and, information about clinical trials (n=123, 37.96%), interpret test results (n=120, 37.04%) and, hereditary considerations (n=103, 31.79%) were searched for least often.

Table 6.27: Information searched for independently

Information searched independently	Number (n=324)	Percent
Disease Cause	207	63.89
Treatment options	210	64.81
Disease management	212	65.43
Complementary therapies	167	51.54
Interpret test results	120	37.04
Clinical trials	123	37.96
Dietary	155	47.84
Physical activity	138	42.59
Psychological/ social support	136	41.98
Hereditary considerations	103	31.79

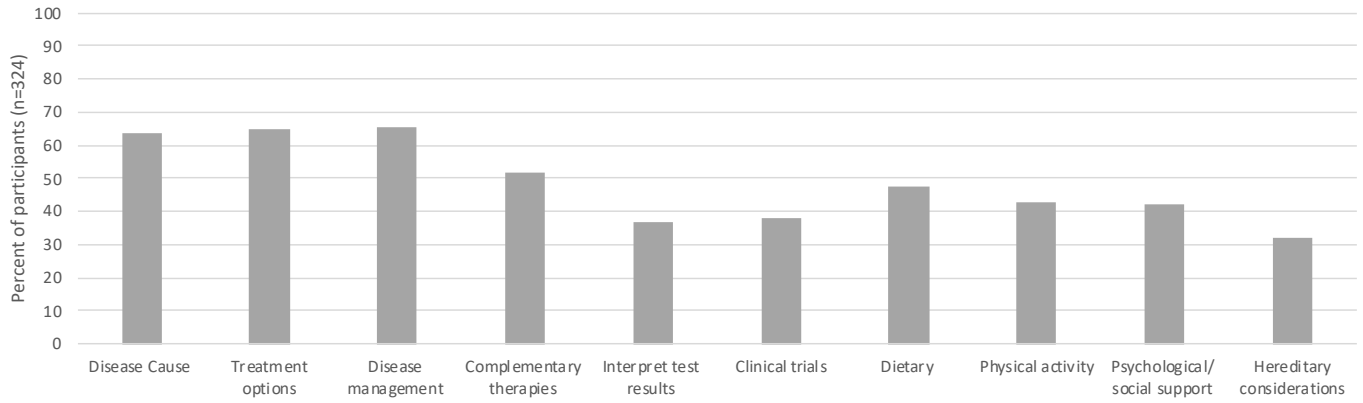


Figure 6.46: Information searched for independently

Information gaps

The largest gaps in information, where information was neither given to patients nor searched for independently were clinical trials (n=177, 54.63%) and interpret test results (n=172, 53.09%).

The topics that participants did not search for independently after not receiving information from healthcare professionals were treatment options (n=66, 20.37%) and disease cause (n=58, 17.90%).

The topics that participants were given most information from both healthcare professionals and searching independently for were disease cause (n=146, 45.06%) and complementary therapies (n=145, 44.75%).

The topics that participants searched for independently after not receiving information from healthcare professionals were treatment options (n=122, 37.65%) and disease management (n=96, 29.63%).

Table 6.28: Information gaps

Information topic	Not given by health professional, not searched for independently		Given by health professional only		Given by health professional, searched for independently		Searched for independently only	
	n=324	%	n=324	%	n=324	%	n=324	%
Disease cause	59	18.21	58	17.90	146	45.06	61	18.83
Treatment options	48	14.81	66	20.37	88	27.16	122	37.65
Disease management	61	18.83	51	15.74	116	35.80	96	29.63
Complementary therapies	145	44.75	12	3.70	145	44.75	22	6.79
How to interpret test results	172	53.09	32	9.88	98	30.25	22	6.79
Clinical trials	177	54.63	24	7.41	104	32.10	19	5.86
Dietary information	130	40.12	39	12.04	116	35.80	39	12.04
Physical activity	141	43.52	45	13.89	98	30.25	40	12.35
Psychological/social support	153	47.22	35	10.80	102	31.48	34	10.49
Hereditary considerations	169	52.16	52	16.05	79	24.38	24	7.41

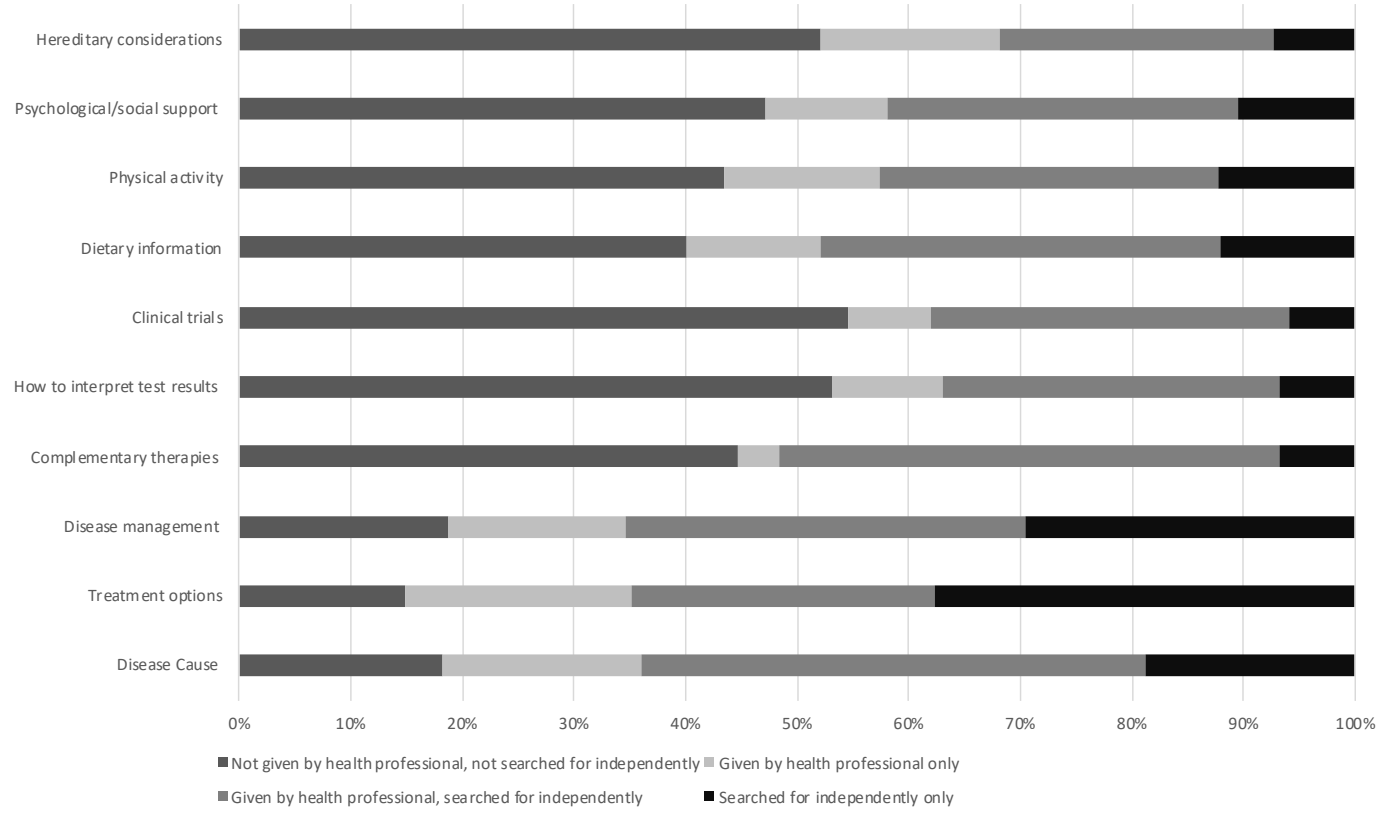


Figure 6.47: Information gaps

Most accessed information

Across all participants, information from Non-profit organisations, charity or patient organisations was most accessed followed by information from the Medical journals. Information from Government and from Pharmaceutical companies were least accessed.

Table 6.29: Most accessed information

Information source	Weighted average (n=321)
Non-profit organisations, charity or patient organisations	3.57
Government	2.45
Pharmaceutical companies	2.19
Hospital or clinic I am being treated in	3.15
Medical journals	3.34

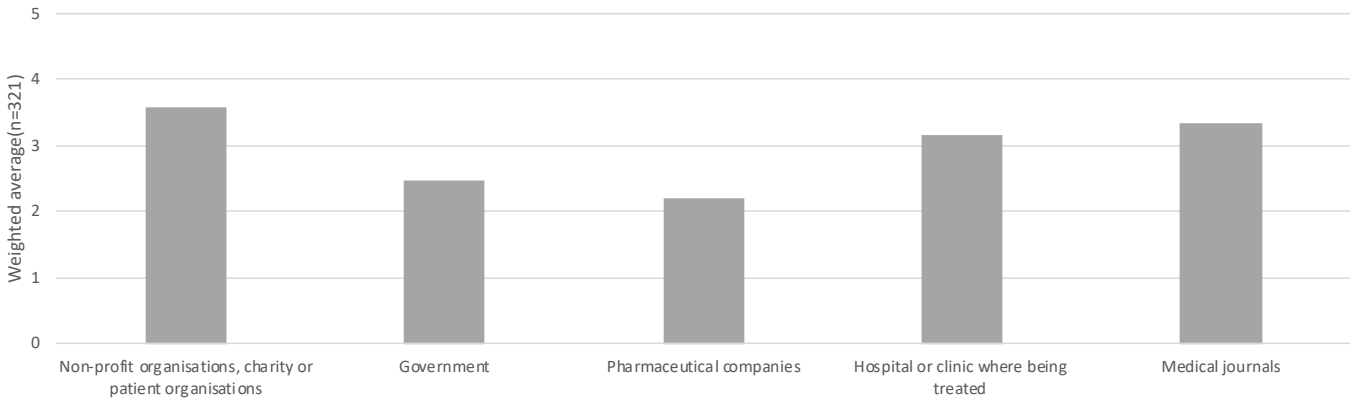


Figure 6.48: Most accessed information

My Health Record

My Health Record is an online summary of key health information, an initiative of the Australian Government. There were 114 participants (39.31%) had accessed My Health Record, 176 participants (60.69%) had not.

Of those that had accessed My Health Record, there were 71 participants (62.28%) who found it to be porr or very poor, 33participants (28.95%) who found it acceptable, and 10 participants (8.77%) who found it to be good or very good.

Table 6.30: Accessed My Health Record

How useful was "My health record"	Number (n=114)	Percent
Very poor	37	32.46
Poor	34	29.82
Acceptable	33	28.95
Good	9	7.89
Very good	1	0.88

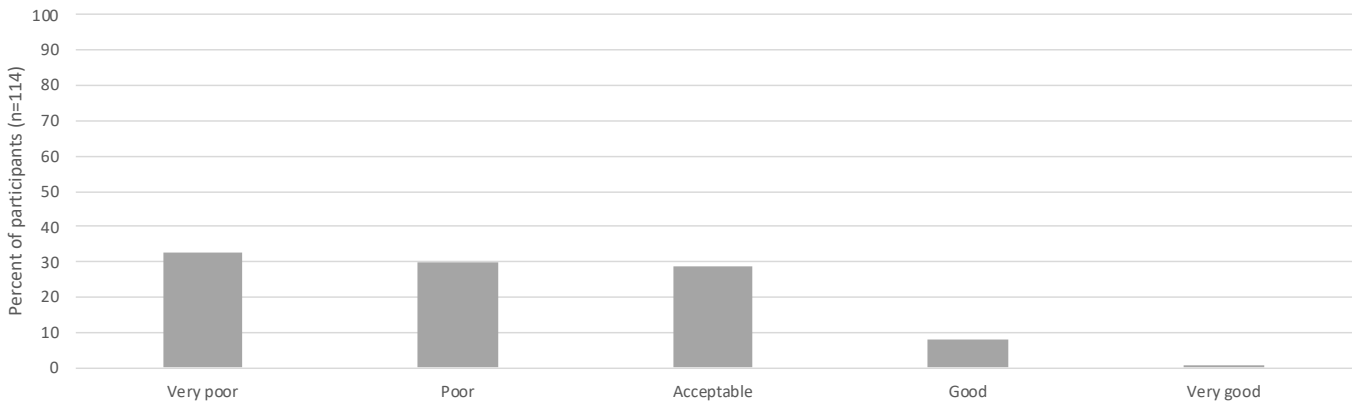


Figure 6.49: Accessed My Health Record

Table 6.31: How useful was My Health Record

Accessed "My health record"	Number (n=290)	Percent
Yes	114	39.31
No	145	50.00
Not sure	11	3.79
Doesn't know what 'My Health Record' is	20	6.90

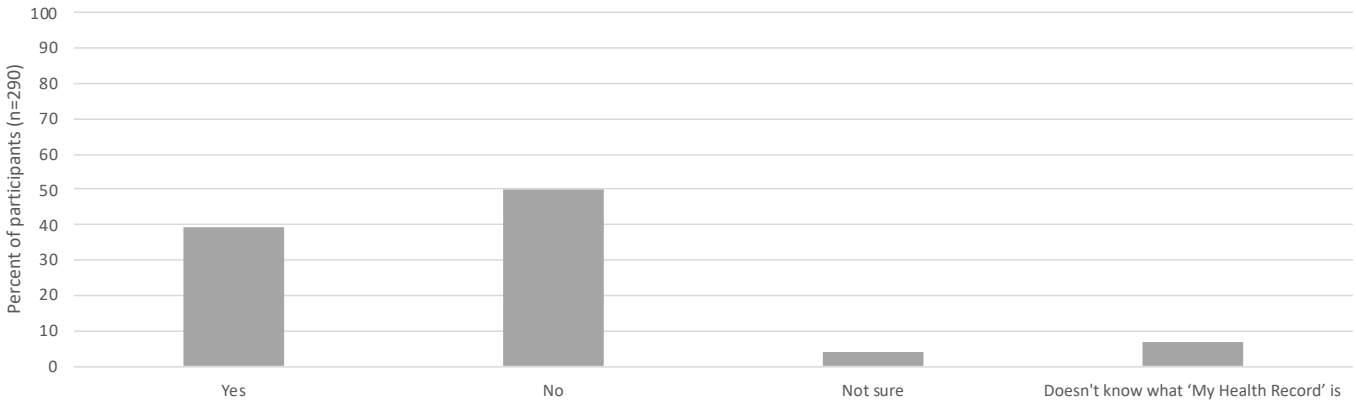


Figure 6.50: How useful was My Health Record

Section 7

Care and support

Section 7: Experience of care and support

Care coordination

A Care Coordination questionnaire was completed by participants within the online questionnaire. The Care Coordination questionnaire comprises a total score, two scales (communication and navigation), and a single question for each relating to care-coordination and care received. A higher score denotes better care outcome.

The overall scores for the cohort were in the highest quintile for **Care coordination: Quality of care** global measure (median=7.00, IQR=3.00) indicating good quality of care. The overall scores for the cohort were in the highest quintile for **Care coordination: Communication** (median=36.00, IQR=13.00), **Care coordination: Navigation** (median=23.00, IQR=8.00), **Care coordination: Total score** (mean=58.51, SD=14.77), **Care coordination: Care coordination global measure** (median=6.00, IQR=4.00) indicating moderate communication, moderate communication, moderate care coordination, moderate care coordination.

The **Care coordination: communication** scale measures communication with healthcare professionals, measuring knowledge about all aspects of care including treatment, services available for their condition, emotional aspects, practical considerations, and financial entitlements. The average score indicates that participants had moderate communication with healthcare professionals.

The **Care coordination: navigation** scale navigation of the healthcare system including knowing important contacts for management of condition, role of healthcare professional in management of condition, healthcare professional knowledge of patient history, ability to get appointments and financial aspects of treatments. The average score indicates that participants had moderate navigation of the healthcare system.

The **Care coordination: total score** scale measures communication, navigation and overall experience of care coordination. The average score indicates that participants had moderate communication, navigation and overall experience of care coordination.

The **Care coordination: care coordination global measure** scale measures the participants overall rating of the coordination of their care. The average score indicates that participants scored rated their care coordination as moderate.

The **Care coordination: Quality of care global measure** scale measures the participants overall rating of the quality of their care. The average score indicates that participants rated their quality of care as good.

Experience of care and support

In the structured interview, participants were asked what care and support they had received since their diagnosis. This question aims to investigate what services patients consider to be support and care services. The most common responses were that they did not receive formal support (25.12%), found support and care from hospital or clinical setting (23.38%), family and friends (20.65%), and charities (17.41%). Other themes included peer support or other patients (13.93%), and challenges accessing support (12.44%).

Care coordination

A Care Coordination questionnaire was completed by participants within the online questionnaire. The Care Coordination questionnaire comprises a total score, two scales (communication and navigation), and a single question for each relating to care-coordination and care received. A higher score denotes better care outcome. Summary statistics for the entire cohort are displayed alongside the possible range of each scale in the table below.

The overall scores for the cohort were in the highest quintile for **Care coordination: Quality of care** global measure (median=7.00, IQR=3.00) indicating good quality of care.

The overall scores for the cohort were in the highest quintile for **Care coordination: Communication** (median=36.00, IQR=13.00), **Care coordination: Navigation** (median=23.00, IQR=8.00), **Care coordination: Total score** (mean=58.51, SD=14.77), **Care coordination: Care coordination global measure** (median=6.00, IQR=4.00) indicating moderate communication, moderate communication, moderate care coordination, moderate care coordination.

Comparisons of Care co-ordination have been made based on condition, participant type, gender, age, education, location and socioeconomic status.

The **Care coordination: communication** scale measures communication with healthcare professionals, measuring knowledge about all aspects of care including treatment, services available for their condition, emotional aspects, practical considerations, and financial entitlements. The average score indicates that participants had moderate communication with healthcare professionals.

The **Care coordination: navigation** scale navigation of the healthcare system including knowing important contacts for management of condition, role of healthcare professional in management of condition, healthcare professional knowledge of patient history, ability to get appointments and financial aspects of treatments. The average score indicates that participants had moderate navigation of the healthcare system.

The **Care coordination: total score** scale measures communication, navigation and overall experience of care coordination. The average score indicates that participants had moderate communication, navigation and overall experience of care coordination.

The **Care coordination: care coordination global measure** scale measures the participants overall rating of the coordination of their care. The average score indicates that participants scored rated their care coordination as moderate.

The **Care coordination: Quality of care global measure** scale measures the participants overall rating of the quality of their care. The average score indicates that participants rated their quality of care as good.

Table 7.1: Care coordination summary statistics

Care coordination scale (n=368)	Mean	SD	Median	IQR	Possible range	Quintile
Communication	35.55	10.34	36.00	13.00	13 to 65	3
Navigation	22.96	6.07	23.00	8.00	7 to 35	3
Total score*	58.51	14.77	60.00	19.00	20 to 100	3
Care coordination global measure	5.79	2.60	6.00	4.00	1 to 10	3
Quality of care global measure	6.59	2.43	7.00	3.00	1 to 10	4

Normal distribution use mean and SD as measure of central tendency

Care coordination by condition

Comparisons were made by **condition**. There were 58 participants (15.89%) with developmental anomalies , 74 participants (20.27%) with diseases of the immune system , 92 participants (25.21%) with diseases of the nervous system , 27 participants (7.40%) with diseases of the skin , 92 participants (25.21%) with endocrine, nutritional or metabolic diseases , and 22 participants (6.03%) with other rare condition.

A one-way ANOVA test was used when the assumptions for response variable residuals were normally distributed and variances of populations were equal. A Tukey HSD test was used post hoc to identify the source of any differences identified in the one-way ANOVA test.

When the assumptions for normality of residuals was not met, a Kruskal-Wallis test was used. Post hoc

pairwise comparisons using Wilcoxon rank sum test was used to identify the source of any differences identified in the Kruskal -Wallis test.

A one way ANOVA test indicated a statistically significant difference in the Care coordination: Communication scale between groups, $F(5, 362) = 3.80$ $p = 0.0023$. The largest significant difference was between participants in the Developmental anomalies subgroup (median = 36.12, IQR = 10.27), and participants in the Endocrine, nutritional or metabolic diseases subgroup (median = 37.28, IQR = 9.81, $p<0.0000$).

A one way ANOVA test indicated a statistically significant difference in the Care coordination: Navigation scale between groups, $F(5, 362) = 7.06$ $p = <0.0001$. The largest significant difference was between participants in the Other rare condition subgroup (median = 23.44, IQR = 7.08), and participants in the Endocrine, nutritional or metabolic diseases subgroup (median = 24.38, IQR = 5.34, $p<0.0000$).

A one way ANOVA test indicated a statistically significant difference in the Care coordination: Total score scale between groups, $F(5, 362) = 5.95$ $p = <0.0001$. The largest significant difference was between participants in the Other rare condition subgroup (median = 58.56, IQR = 16.34), and participants in the Endocrine, nutritional or metabolic diseases subgroup (median = 61.66, IQR = 13.75, $p<0.0000$).

A Kruskal-Wallis test indicated a statistically significant difference in the Care coordination: Care coordination global measure scale between groups, $\chi^2(5) = 18.46$ $p = 0.0024$. The largest significant difference was between participants in the Diseases of the nervous system subgroup (median = 7.00, IQR = 3.00), and participants in the Diseases of the skin subgroup (median = 4.00, IQR = 2.50, $p = 0.0019$).

A Kruskal-Wallis test indicated a statistically significant difference in the Care coordination: Quality of care global measure scale between groups, $\chi^2(5) = 27.73$ $p = <0.0001$. The largest significant difference was between participants in the Developmental anomalies subgroup (median = 8.00, IQR = 3.00), and participants in the Diseases of the skin subgroup (median = 5.00, IQR = 3.00, $p = <0.0001$).

The **Care coordination: communication** scale measures communication with healthcare professionals, measuring knowledge about all aspects of care including treatment, services available for their condition, emotional aspects, practical considerations, and financial entitlements. On average, participants in the Diseases of the nervous system subgroup scored higher than participants in the Diseases of the immune system subgroup. This indicates that healthcare communication was average for participants in the Diseases of the nervous system subgroup, and poor for participants in the Diseases of the immune system subgroup.

The **Care coordination: navigation** scale measures the ability of a patient to navigate the healthcare system including knowing important contacts for management of condition, role of healthcare professional in management of condition, healthcare professional knowledge of patient history, ability to get appointments and financial aspects of treatments. On average, participants in the Developmental anomalies subgroup scored higher than participants in the Diseases of the immune system subgroup. This indicates that healthcare navigation was good for participants in the Developmental anomalies subgroup, and average for participants in the Diseases of the immune system subgroup.

The **Care coordination: total score** scale measures communication, navigation and overall experience of care coordination. On average, participants in the Developmental anomalies subgroup had a higher total score for navigation compared to Diseases of the immune system , however communication, navigation and overall experience of care coordination was average for both groups.

The **Care coordination: care coordination global measure** scale measures the participants overall rating of the coordination of their care. On average, participants in the Diseases of the nervous system subgroup scored higher than participants in the Diseases of the skin subgroup. This indicates that, overall care coordination was good for participants in the Diseases of the nervous system subgroup, and poor for participants in the Diseases of the skin subgroup.

The **Care coordination: Quality of care global measure** scale measures the participants overall rating of the quality of their care. On average, participants in the Developmental anomalies subgroup scored higher than participants in the Diseases of the skin subgroup. This indicates that, quality of care was good for participants in the Developmental anomalies subgroup, and average for participants in the Diseases of the skin subgroup.

Table 7.2: Care coordination by condition summary statistics and one-way ANOVA

Partners in health scale	Group	Number (n=368)	Percent	Mean	SD	Source of difference	Sum of squares	dF	Mean Square	f	p-value
Communication	Developmental anomalies	58	15.89	36.12	10.27	Between groups	1954.00	5	390.90	3.80	0.0023*
	Diseases of the immune system	74	20.27	32.21	9.07	Within groups	37246.00	362	102.90		
	Diseases of the nervous system	92	25.21	37.44	10.28	Total	39200.00	367			
	Diseases of the skin	27	7.40	31.19	11.64						
	Endocrine, nutritional or metabolic diseases	92	25.21	37.28	9.81						
	Other rare condition	22	6.03	35.12	11.74						
Navigation	Developmental anomalies	58	15.89	22.25	5.78	Between groups	1201.00	5	240.14	7.06	<0.0001*
	Diseases of the immune system	74	20.27	20.75	5.97	Within groups	12316.00	362	34.02		
	Diseases of the nervous system	92	25.21	24.61	5.70	Total	13517.00	367			
	Diseases of the skin	27	7.40	19.26	6.43						
	Endocrine, nutritional or metabolic diseases	92	25.21	24.38	5.34						
	Other rare condition	22	6.03	23.44	7.08						
Total score	Developmental anomalies	58	15.89	58.37	15.12	Between groups	6077.00	5	1215.40	5.95	<0.0001*
	Diseases of the immune system	74	20.27	52.96	13.13	Within groups	73947.00	362	204.30		
	Diseases of the nervous system	92	25.21	62.05	14.37	Total	80024.00	367			
	Diseases of the skin	27	7.40	50.44	15.08						
	Endocrine, nutritional or metabolic diseases	92	25.21	61.66	13.75						
	Other rare condition	22	6.03	58.56	16.34						

Table 7.3: Care coordination by condition one-way post hoc Tukey HSD test

Care coordination scale	Group	Difference	Upper	Lower	p adjusted
Communication	Diseases of the immune system - Developmental anomalies	-3.91	-9.07	1.24	0.2512
	Diseases of the nervous system - Developmental anomalies	1.32	-3.57	6.21	0.9720
	Diseases of the skin - Developmental anomalies	-4.94	-11.73	1.85	0.2982
	Endocrine, nutritional or metabolic diseases - Developmental anomalies	1.15	-3.72	6.03	0.9843
	Other rare condition - Developmental anomalies	-1.00	-7.97	5.97	0.9985
	Diseases of the nervous system - Diseases of the immune system	5.23	0.67	9.79	0.0141*
	Diseases of the skin - Diseases of the immune system	-1.02	-7.58	5.54	0.9977
	Endocrine, nutritional or metabolic diseases - Diseases of the immune system	5.07	0.52	9.62	0.0191*
	Other rare condition - Diseases of the immune system	2.91	-3.83	9.66	0.8186
	Diseases of the skin - Diseases of the nervous system	-6.26	-12.61	0.10	0.0564
	Endocrine, nutritional or metabolic diseases - Diseases of the nervous system	-0.16	-4.41	4.09	1.0000
	Other rare condition - Diseases of the nervous system	-2.32	-8.87	4.23	0.9126
	Endocrine, nutritional or metabolic diseases - Diseases of the skin	6.09	-0.25	12.44	0.0682
	Other rare condition - Diseases of the skin	3.93	-4.13	12.00	0.7285
	Other rare condition - Endocrine, nutritional or metabolic diseases	-2.16	-8.70	4.38	0.9345
Navigation	Diseases of the immune system - Developmental anomalies	-1.50	-4.46	1.47	0.6986
	Diseases of the nervous system - Developmental anomalies	2.37	-0.44	5.18	0.1545
	Diseases of the skin - Developmental anomalies	-2.99	-6.89	0.92	0.2442
	Endocrine, nutritional or metabolic diseases - Developmental anomalies	2.14	-0.67	4.94	0.2483
	Other rare condition - Developmental anomalies	1.19	-2.81	5.20	0.9570
	Diseases of the nervous system - Diseases of the immune system	3.86	1.24	6.49	0.0004*
	Diseases of the skin - Diseases of the immune system	-1.49	-5.26	2.28	0.8676
	Endocrine, nutritional or metabolic diseases - Diseases of the immune system	3.63	1.02	6.25	0.0012*
	Other rare condition - Diseases of the immune system	2.69	-1.19	6.57	0.3520
	Diseases of the skin - Diseases of the nervous system	-5.35	-9.01	-1.70	0.0005*
	Endocrine, nutritional or metabolic diseases - Diseases of the nervous system	-0.23	-2.67	2.21	0.9998
	Other rare condition - Diseases of the nervous system	-1.17	-4.94	2.59	0.9481
	Endocrine, nutritional or metabolic diseases - Diseases of the skin	5.12	1.47	8.77	0.0010*
	Other rare condition - Diseases of the skin	4.18	-0.46	8.82	0.1042
	Other rare condition - Endocrine, nutritional or metabolic diseases	-0.94	-4.70	2.82	0.9796
Total score	Diseases of the immune system - Developmental anomalies	-5.41	-12.67	1.85	0.2716
	Diseases of the nervous system - Developmental anomalies	3.69	-3.20	10.57	0.6432
	Diseases of the skin - Developmental anomalies	-7.92	-17.49	1.64	0.1685
	Endocrine, nutritional or metabolic diseases - Developmental anomalies	3.29	-3.58	10.17	0.7440
	Other rare condition - Developmental anomalies	0.19	-9.63	10.01	1.0000
	Diseases of the nervous system - Diseases of the immune system	9.10	2.67	15.52	0.0009*
	Diseases of the skin - Diseases of the immune system	-2.51	-11.75	6.73	0.9709
	Endocrine, nutritional or metabolic diseases - Diseases of the immune system	8.70	2.29	15.11	0.0017*
	Other rare condition - Diseases of the immune system	5.60	-3.90	15.11	0.5404
	Diseases of the skin - Diseases of the nervous system	-11.61	-20.56	-2.66	0.0032*
	Endocrine, nutritional or metabolic diseases - Diseases of the nervous system	-0.39	-6.38	5.59	1.0000
	Other rare condition - Diseases of the nervous system	-3.49	-12.72	5.73	0.8872
	Endocrine, nutritional or metabolic diseases - Diseases of the skin	11.22	2.27	20.16	0.0050*
	Other rare condition - Diseases of the skin	8.12	-3.25	19.48	0.3186
	Other rare condition - Endocrine, nutritional or metabolic diseases	-3.10	-12.31	6.12	0.9290

Table 7.4: Care coordination by condition summary statistics and Kruskal Wallis test

SF36 scale	Group	Number (n=368)	Percent	Median	IQR	C^2	dF	p-value
Care coordination global measure	Developmental anomalies	58	15.89	6.00	5.00	18.46	5	0.0024
	Diseases of the immune system	74	20.27	5.00	3.00			
	Diseases of the nervous system	92	25.21	7.00	3.00			
	Diseases of the skin	27	7.40	4.00	2.50			
	Endocrine, nutritional or metabolic diseases	92	25.21	7.00	3.75			
	Other rare condition	22	6.03	5.00	6.00			
Quality of care global measure	Developmental anomalies	58	15.89	8.00	3.00	27.73	5	<0.0001*
	Diseases of the immune system	74	20.27	7.00	3.25			
	Diseases of the nervous system	92	25.21	8.00	2.00			
	Diseases of the skin	27	7.40	5.00	3.00			
	Endocrine, nutritional or metabolic diseases	92	25.21	7.00	4.00			
	Other rare condition	22	6.03	7.00	4.00			

Table 7.5: Care coordination by condition one-way post hoc Wilcoxon rank sum test

SF36 scale		Developmental anomalies	Diseases of the immune system	Diseases of the nervous system	Diseases of the skin	Endocrine, nutritional or metabolic diseases
Coping	Diseases of the immune system	0.3338	-	-	-	-
	Diseases of the nervous system	0.7507	0.0400*	-	-	-
	Diseases of the skin	0.0400*	0.0720	0.0019*	-	-
	Endocrine, nutritional or metabolic diseases	0.8317	0.0743	0.8317	0.0049*	-
	Other rare condition	0.7789	0.9404	0.3338	0.3338	0.5369
Adherence to treatment	Diseases of the immune system	0.0442	-	-	-	-
	Diseases of the nervous system	0.4700	0.1470	-	-	-
	Diseases of the skin	<0.0001*	0.0038*	<0.0001*	-	-
	Endocrine, nutritional or metabolic diseases	0.4700	0.1470	0.9836	0.0001*	-
	Other rare condition	0.3781	0.9192	0.4700	0.0309*	0.4700

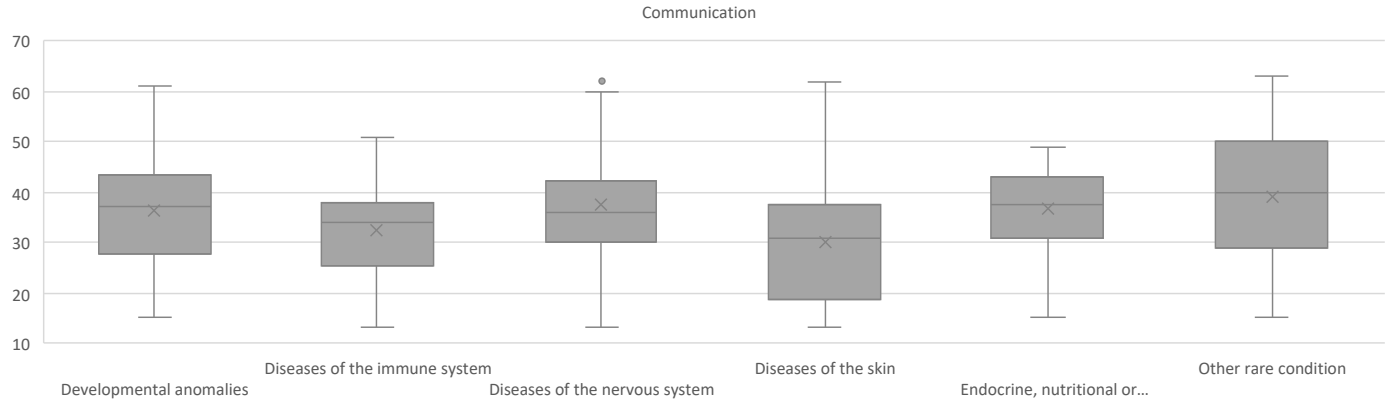


Figure 7.1: Boxplot of Care coordination: Communication by condition

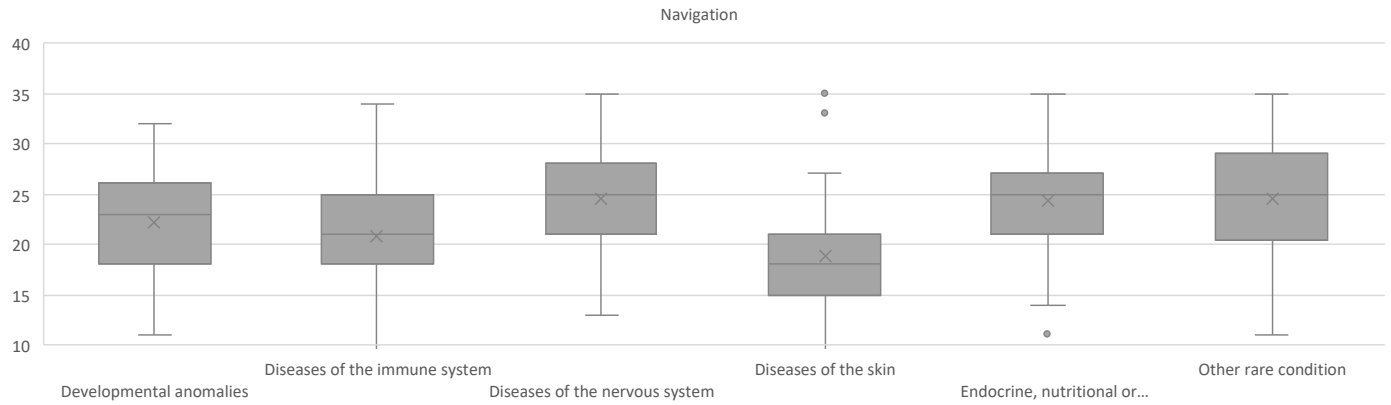


Figure 7.2: Boxplot of Care coordination: Navigation by condition

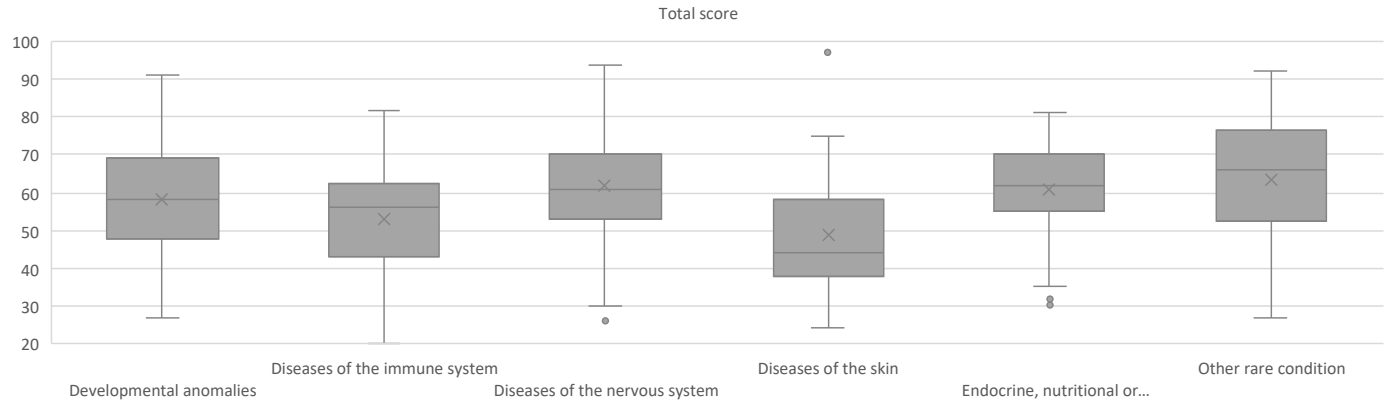


Figure 7.3: Boxplot of Care coordination: Total score by condition

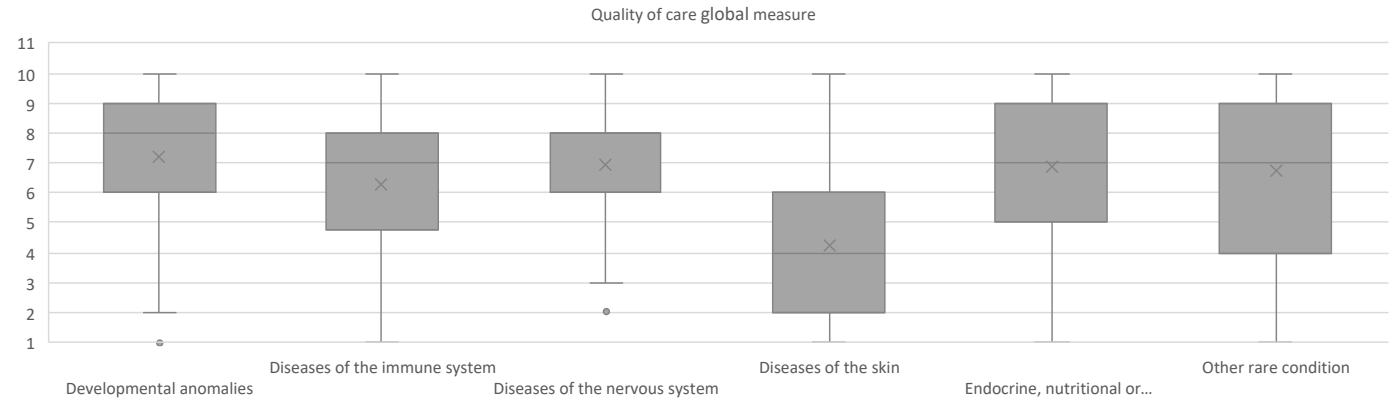


Figure 7.4: Boxplot of Care coordination: Care coordination global measure by condition

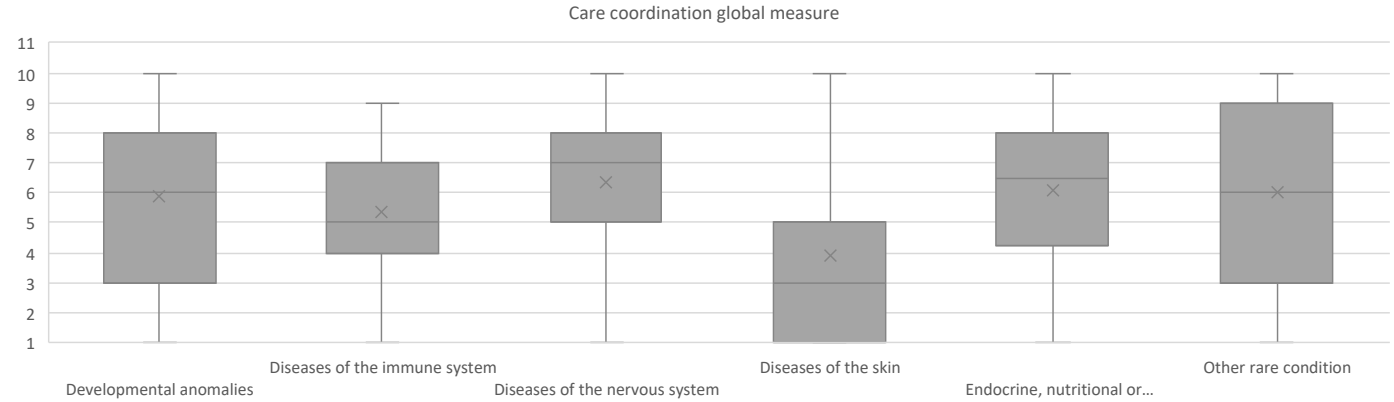


Figure 7.5: Boxplot of Care coordination: Quality of care global measure by condition

Care coordination by type of participant

Comparisons were made by **type of participant** there were 246 participants (66.85%) with person with condition and, 122 participants (33.15%) with carer.

A two-sample t-test was used when assumptions for normality and variance were met, or when assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used.

A two sample t-test indicated that the mean score for the Care coordination Communication scale [t(366) = -3.77 , p = 0.0002] was significantly lower for participants in the Person with condition subgroup (Mean = 34.15, SD = 9.89) compared to participants in the Carer subgroup (Mean = 38.39, SD = 10.67.)

Wilcoxon rank sum tests with continuity correction indicated that the median score for the Care coordination Navigation scale [W = 11900.00, p = 0.0012] was significantly lower for participants in the Person with condition subgroup (Median = 22.00, IQR = 7.75) compared to participants in the Carer subgroup (Median = 25.00, IQR = 7.00.

A two sample t-test indicated that the mean score for the Care coordination Total score scale [t(366) = -3.91 , p = 0.0001] was significantly lower for participants in the Person with condition subgroup (Mean = 56.43, SD = 14.22) compared to participants in the Carer subgroup (Mean = 62.70, SD = 15.02.)

Wilcoxon rank sum tests with continuity correction indicated that the median score for the Care coordination Care coordination global measure scale [W = 11419.00, p = 0.0002] was significantly lower for participants in the Person with condition subgroup (Median = 6.00, IQR = 4.00) compared to participants in the Carer subgroup (Median = 7.00, IQR = 3.00.

Wilcoxon rank sum tests with continuity correction indicated that the median score for the Care coordination Quality of care global measure scale [W = 10360.00, p<0.0000] was significantly lower for participants in the Person with condition subgroup (Median = 7.00, IQR = 4.00) compared to participants in the Carer subgroup (Median = 8.00, IQR = 2.00.

The **Care coordination: communication** scale measures communication with healthcare professionals, measuring knowledge about all aspects of care including treatment, services available for their condition, emotional aspects, practical considerations, and financial entitlements. On average, participants in the Carer subgroup had a higher score for communication compared to Person with condition, however, healthcare communication was average for both groups.

The **Care coordination: navigation** scale measures the ability of a patient to navigate the healthcare system including knowing important contacts for management of condition, role of healthcare professional in management of condition, healthcare professional knowledge of patient history, ability to get appointments and financial aspects of treatments. On average, participants in the Carer subgroup scored higher than participants in the Person with condition subgroup. This indicates that healthcare navigation was good for participants in the Carer subgroup, and average for participants in the Person with condition subgroup.

The **Care coordination: total score** scale measures communication, navigation and overall experience of care coordination. On average, participants in the Carer subgroup had a higher total score for navigation compared to Person with condition, however communication, navigation and overall experience of care coordination was average for both groups.

The **Care coordination: care coordination global measure** scale measures the participants overall rating of the coordination of their care. On average, participants in the Carer subgroup scored higher than participants in the Person with condition subgroup. This indicates that, overall care coordination was good for participants in the Carer subgroup, and average for participants in the Person with condition subgroup.

The **Care coordination: Quality of care global measure** scale measures the participants overall rating of the quality of their care. On average, participants in the Carer subgroup had a higher score for quality of compared to Person with condition, however, quality of care was good for both groups.

Table 7.6: Care coordination by type of participant summary statistics and T-test

Care coordination scale	Group	Number (n=368)	Percent	Mean	SD	T	dF	p-value
Communication	Person with condition	246	66.85	34.15	9.89	-3.77	366.00	0.0002*
	Carer	122	33.15	38.39	10.67			
Total score	Person with condition	246	66.85	56.43	14.22	-3.91	366.00	0.0001*
	Carer	122	33.15	62.70	15.02			

Table 7.7: Care coordination by type of participant summary statistics and Wilcoxon test

Care coordination scale	Group	Number (n=368)	Percent	Median	IQR	W	p-value
Navigation	Person with condition	246	66.85	22.00	7.75	11900.00	0.0012*
	Carer	122	33.15	25.00	7.00		
Care coordination global measure	Person with condition	246	66.85	6.00	4.00	11419.00	0.0002*
	Carer	122	33.15	7.00	3.00		
Quality of care global measure	Person with condition	246	66.85	7.00	4.00	10360.00	0.0000*
	Carer	122	33.15	8.00	2.00		

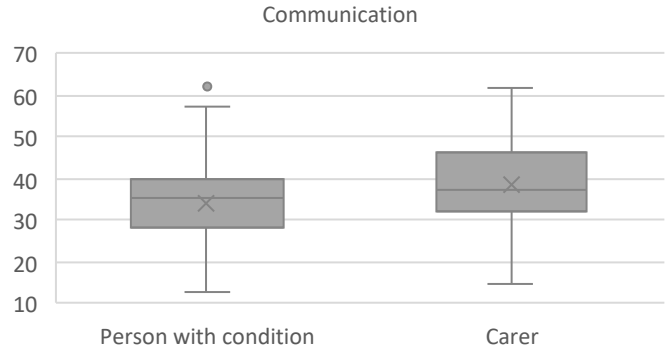


Figure 7.6: Boxplot of Care coordination: Communication by type of participant

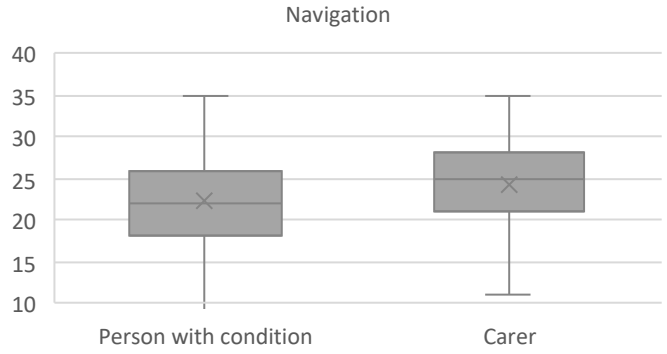


Figure 7.7: Boxplot of Care coordination: Navigation by type of participant

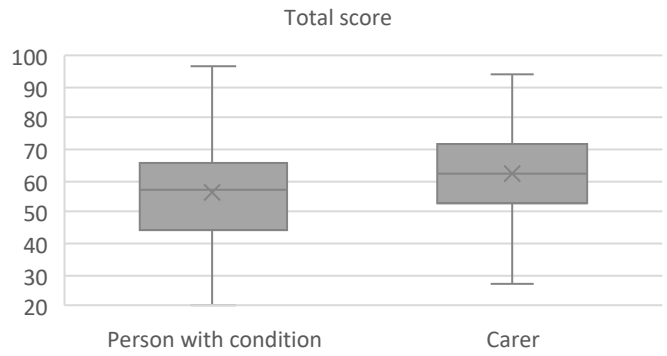


Figure 7.8: Boxplot of Care coordination: Total score by type of participant

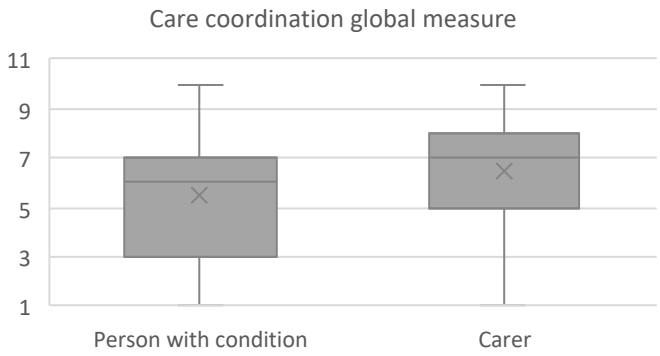


Figure 7.9: Boxplot of Care coordination: Care coordination global measure by type of participant

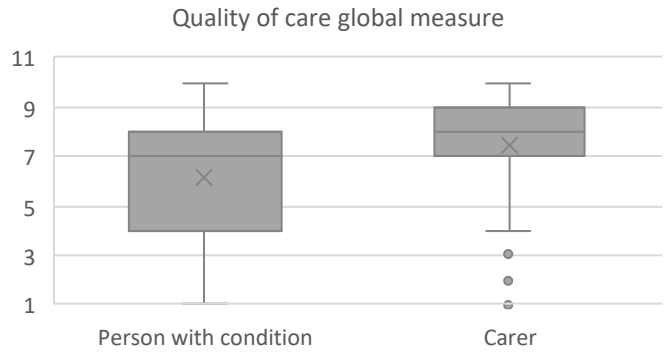


Figure 7.10: Boxplot of Care coordination: Quality of care global measure by type of participant

Care coordination by gender

Comparisons were made by **gender**, there were 274 female participants (74.86%) and 92 male participants (25.14%).

A two-sample t-test was used when assumptions for normality and variance were met, or when assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used.

A two sample t-test indicated that the mean score for the Care coordination Navigation scale [t(364) = -3.31 , p = 0.0010] was significantly lower for participants in the Female subgroup (Mean = 22.37, SD = 6.09) compared to participants in the Male subgroup (Mean = 24.76, SD = 5.70.)

Wilcoxon rank sum tests with continuity correction indicated that the median score for the Care coordination Communication scale [W = 10164.00 , p = 0.0054] was significantly lower for participants in the Female subgroup (Median = 35.00, IQR = 13.00) compared to participants in the Male subgroup (Median = 38.00, IQR = 12.00.

Wilcoxon rank sum tests with continuity correction indicated that the median score for the Care coordination Total score scale [W = 9613.00 , p = 0.0007] was significantly lower for participants in the Female subgroup (Median = 57.00, IQR = 20.00) compared to participants in the Male subgroup (Median = 64.00, IQR = 16.00.

Wilcoxon rank sum tests with continuity correction indicated that the median score for the Care coordination Care coordination global measure scale [W = 10117.00 , p = 0.0044] was significantly lower for participants in the Female subgroup (Median = 6.00, IQR = 4.00) compared to participants in the Male subgroup (Median = 7.00, IQR = 4.00.

Wilcoxon rank sum tests with continuity correction indicated that the median score for the Care coordination Quality of care global measure scale [W = 9653.00 , p = 0.0007] was significantly lower for participants in the Female subgroup (Median = 7.00, IQR = 3.00) compared to participants in the Male subgroup (Median = 8.00, IQR = 3.00.

The **Care coordination: communication** scale measures communication with healthcare professionals, measuring knowledge about all aspects of care including treatment, services available for their condition, emotional aspects, practical considerations, and financial entitlements. On average, participants in the Male subgroup had a higher score for communication compared to Female, however, healthcare communication was average for both groups.

The **Care coordination: navigation** scale measures the ability of a patient to navigate the healthcare system including knowing important contacts for management of condition, role of healthcare professional in management of condition, healthcare professional knowledge of patient history, ability to get appointments and financial aspects of treatments. On average, participants in the Male subgroup scored higher than participants in the Female subgroup. This indicates that healthcare navigation was good for participants in the Male subgroup, and average for participants in the Female subgroup.

The **Care coordination: total score** scale measures communication, navigation and overall experience of care coordination. On average, participants in the Male subgroup had a higher total score for navigation compared to Female, however communication, navigation and overall experience of care coordination was average for both groups.

The **Care coordination: care coordination global measure** scale measures the participants overall rating of the coordination of their care. On average, participants in the Male subgroup scored higher than participants in the Female subgroup. This indicates that, overall care coordination was good for participants in the Male subgroup, and average for participants in the Female subgroup.

The **Care coordination: Quality of care global measure** scale measures the participants overall rating of the quality of their care. On average, participants in the Male subgroup had a higher score for quality of compared to Female, however, quality of care was good for both groups.

Table 7.8: Care coordination by gender summary statistics and two-sample t-test

Care coordination scale	Group	Number (n=366)	Percent	Mean	SD	T	dF	p-value
Navigation	Female	274	74.86	22.37	6.09	-3.31	364.00	0.0010*
	Male	92	25.14	24.76	5.70			

Table 7.8: Care coordination by gender summary statistics and Wilcoxon test

Care coordination scale	Group	Number (n=366)	Percent	Median	IQR	W	p-value
Communication	Female	274	74.86	35.00	13.00	10164.00	0.0054*
	Male	92	25.14	38.00	12.00		
Total score	Female	274	74.86	57.00	20.00	9613.00	0.0007*
	Male	92	25.14	64.00	16.00		
Care coordination global measure	Female	274	74.86	6.00	4.00	10117.00	0.0044*
	Male	92	25.14	7.00	4.00		
Quality of care global measure	Female	274	74.86	7.00	3.00	9653.00	0.0007*
	Male	92	25.14	8.00	3.00		

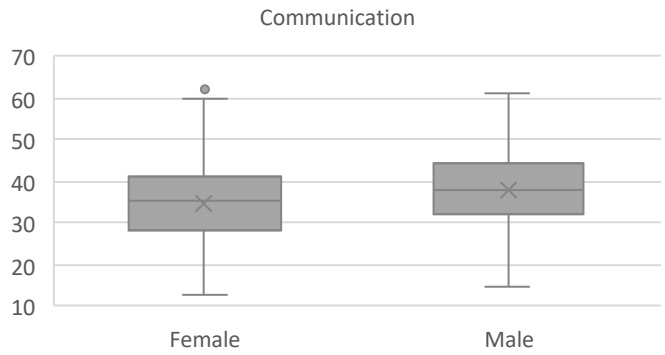


Figure 7.11: Boxplot of Care coordination: Communication by gender

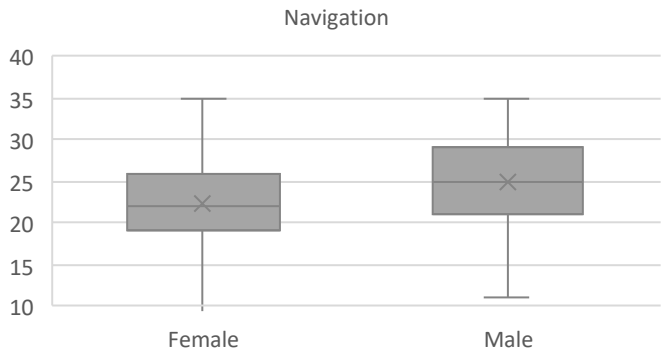


Figure 7.12: Boxplot of Care coordination: Navigation by gender

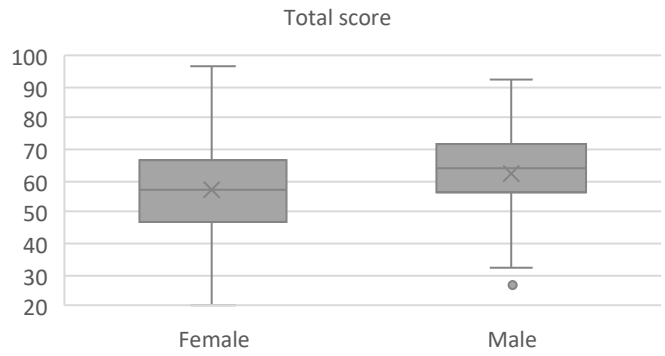


Figure 7.13: Boxplot of Care coordination: Total score by gender

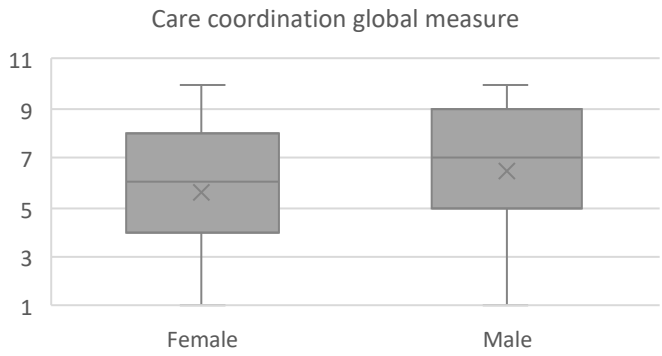


Figure 7.14: Boxplot of Care coordination: Care coordination global measure by gender

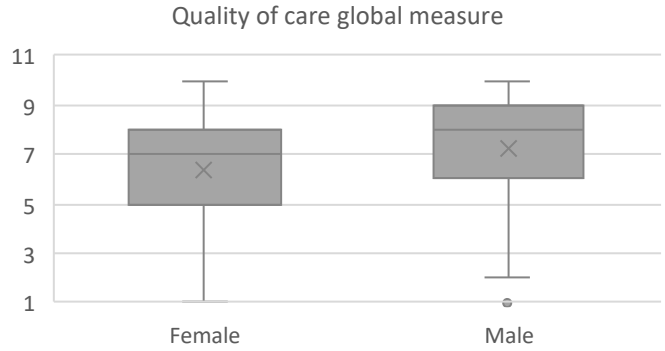


Figure 7.15: Boxplot of Care coordination: Quality of care global measure by gender

Care coordination by age

Comparisons were made by **age** of person with condition. There were 88 participants (23.91%) with aged under 18, 117 participants (31.79%) with aged 18 to 44, 105 participants (28.53%) with aged 45 to 64, and 58 participants (15.76%) with aged 65 or older.

A one-way ANOVA test was used when the assumptions for response variable residuals were normally distributed and variances of populations were equal. A Tukey HSD test was used post hoc to identify the source of any differences identified in the one-way ANOVA test.

When the assumptions for normality of residuals was not met, a Kruskal-Wallis test was used. Post hoc pairwise comparisons using Wilcoxon rank sum test was used to identify the source of any differences identified in the Kruskal -Wallis test.

A one way ANOVA test indicated a statistically significant difference in the Care coordination: Communication scale between groups, F(3, 364) = 13.90 p = <0.0001. The largest significant difference was between participants in the Aged 45 to 64 subgroup (median = 33.43, IQR = 9.02), and participants in the Aged 65 or oldersub group (median = 39.34, IQR = 9.79, p<0.0000).

A one way ANOVA test indicated a statistically significant difference in the Care coordination: Navigation scale between groups, F(3, 364) = 9.89 p = <0.0001.The largest significant difference was between participants in the Aged 45 to 64 subgroup (median = 22.37, IQR = 6.25), and participants in the Aged 65 or older subgroup (median = 24.83, IQR = 5.51, p<0.0001).

A one way ANOVA test indicated a statistically significant difference in the Care coordination: Total score scale between groups, F(3, 364) = 15.43 p = <0.0001. The largest significant difference was between participants in the Aged 45 to 64 subgroup (median = 55.80, IQR = 13.73), and participants in the Aged 65 or older subgroup (median = 64.17, IQR = 13.48, p<0.0000).

A Kruskal-Wallis test indicated a statistically significant difference in the Care coordination: Care coordination global measure scale between groups, $\chi^2(3) = 26.24$ P = <0.0001. The largest significant difference was between Aged under 18 (median = 7.5, IQR = 3), and Aged 18 to 44 (median = 5, IQR = 4, p = 0.0002).

A Kruskal-Wallis test indicated a statistically significant difference in the Care coordination: Quality of care global measure scale between groups, $\chi^2(3) = 41.88$ P = <0.0001. The largest significant difference was between Aged under 18 (median = 8, IQR = 2), and Aged 18 to 44 (median = 6, IQR = 4, p = <0.0001).

The **Care coordination: communication** scale measures communication with healthcare professionals, measuring knowledge about all aspects of care including treatment, services available for their condition, emotional aspects, practical considerations, and financial entitlements. On average, participants in the Aged under 18 subgroup scored higher than participants in the Aged 18 to 44 subgroup. This indicates that healthcare communication was average for participants in the Aged under 18 subgroup, and poor for participants in the Aged 18 to 44 subgroup.

The **Care coordination: navigation** scale measures the ability of a patient to navigate the healthcare system including knowing important contacts for management of condition, role of healthcare professional in management of condition, healthcare professional knowledge of patient history, ability to get appointments and financial aspects of treatments. On average, participants in the Aged under 18 subgroup scored higher than participants in the Aged 18 to 44 subgroup. This indicates that healthcare navigation was good for participants in the Aged under 18 subgroup, and average for participants in the Aged 18 to 44 subgroup.

The **Care coordination: total score** scale measures communication, navigation and overall experience of care coordination. On average, participants in the Aged under 18 subgroup had a higher total score for navigation compared to Aged 18 to 44, however communication, navigation and overall experience of care coordination was average for both groups.

The **Care coordination: care coordination global measure** scale measures the participants overall rating of the coordination of their care. On average, participants in the Aged under 18 subgroup scored higher than participants in the Aged 18 to 44 subgroup. This indicates that, overall care coordination was good for participants in the Aged under 18 subgroup, and average for participants in the Aged 18 to 44 subgroup.

The **Care coordination: Quality of care global measure** scale measures the participants overall rating of the quality of their care. On average, participants in the Aged under 18 subgroup scored higher than participants in the Aged 18 to 44 subgroup. This indicates that, quality of care was good for participants in the Aged under 18 subgroup, and average for participants in the Aged 18 to 44 subgroup.

Table 7.9: Care coordination by age summary statistics and one-way ANOVA

Care coordination scale	Group	Number (n=368)	Percent	Mean	SD	Source of difference	Sum of squares	dF	Mean Square	f	p-value
Communication	Aged under 18	88	23.91	39.77	11.15	Between groups	4030.00	3	1343.40	13.90	<0.0001*
	Aged 18 to 44	117	31.79	32.41	9.48	Within groups	35171.00	364	96.60		
	Aged 45 to 64	105	28.53	33.43	9.02	Total	39201.00	367			
	Aged 65 or older	58	15.76	39.34	9.79						
Navigation	Aged under 18	88	23.91	24.97	5.12	Between groups	1019.00	3	339.70	9.89	<0.0001*
	Aged 18 to 44	117	31.79	21.05	6.18	Within groups	12497.00	364	34.30		
	Aged 45 to 64	105	28.53	22.37	6.25	Total	13516.00	367			
	Aged 65 or older	58	15.76	24.83	5.51						
Total score	Aged under 18	88	23.91	64.74	15.30	Between groups	9027.00	3	3009.00	15.43	<0.0001*
	Aged 18 to 44	117	31.79	53.46	13.35	Within groups	70997.00	364	195.00		
	Aged 45 to 64	105	28.53	55.80	13.73	Total	80024.00	367			
	Aged 65 or older	58	15.76	64.17	13.48						

Table 7.10: Care coordination by age one-way post hoc Tukey HSD test

Care coordination scale	Group	Difference	Upper	Lower	p adjusted
Communication	Aged 18 to 44 - Aged under 18	-7.36	-10.94	-3.78	<0.0001*
	Aged 45 to 64 - Aged under 18	-6.34	-10.01	-2.68	0.0001*
	Aged 65 or older - Aged under 18	-0.43	-4.72	3.86	0.9940
	Aged 45 to 64 - Aged 18 to 44	1.02	-2.39	4.43	0.8677
	Aged 65 or older - Aged 18 to 44	6.93	2.86	11.01	0.0001*
	Aged 65 or older - Aged 45 to 64	5.92	1.77	10.07	0.0015*
Navigation	Aged 18 to 44 - Aged under 18	-3.91	-6.05	-1.78	<0.0001*
	Aged 45 to 64 - Aged under 18	-2.59	-4.78	-0.41	0.0125*
	Aged 65 or older - Aged under 18	-0.14	-2.70	2.42	0.9990
	Aged 45 to 64 - Aged 18 to 44	1.32	-0.71	3.35	0.3379
	Aged 65 or older - Aged 18 to 44	3.78	1.35	6.20	0.0004*
	Aged 65 or older - Aged 45 to 64	2.46	-0.02	4.93	0.0525
Total score	Aged 18 to 44 - Aged under 18	-11.28	-16.36	-6.19	<0.0001*
	Aged 45 to 64 - Aged under 18	-8.94	-14.15	-3.73	0.0001*
	Aged 65 or older - Aged under 18	-0.57	-6.66	5.53	0.9951
	Aged 45 to 64 - Aged 18 to 44	2.34	-2.51	7.18	0.5982
	Aged 65 or older - Aged 18 to 44	10.71	4.92	16.50	<0.0001*
	Aged 65 or older - Aged 45 to 64	8.37	2.48	14.27	0.0016*

Table 7.11: Care coordination by age summary statistics and Kruskal Wallis test

Care coordination scale	Group	Number (n=368)	Percent	Median	IQR	C^2	dF	p-value
Care coordination global measure	Aged under 18	88	23.91	7.50	3.00	26.24	3	<0.0001*
	Aged 18 to 44	117	31.79	5.00	4.00			
	Aged 45 to 64	105	28.53	5.00	4.00			
	Aged 65 or older	58	15.76	7.00	4.00			
Quality of care global measure	Aged under 18	88	23.91	8.00	2.00	41.88	3	<0.0001*
	Aged 18 to 44	117	31.79	6.00	4.00			
	Aged 45 to 64	105	28.53	7.00	4.00			
	Aged 65 or older	58	15.76	8.00	2.75			

Table 7.12: Care coordination by age one-way post hoc Wilcoxon rank sum test

SF36 scale		Aged under 18	Aged 18 to 44	Aged 45 to 64
Care coordination global measure	Aged 18 to 44	0.0002*	-	-
	Aged 45 to 64	0.0011*	0.5084	-
	Aged 65 or older	0.8844	0.0011*	0.0034*
Quality of care global measure	Aged 18 to 44	<0.0001*	-	-
	Aged 45 to 64	<0.0001*	0.6775	-
	Aged 65 or older	0.7505	0.0002*	0.0010*

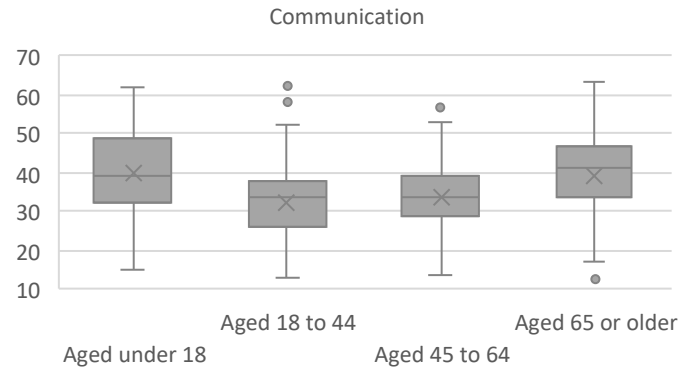


Figure 7.16: Boxplot of Care coordination: Communication by age

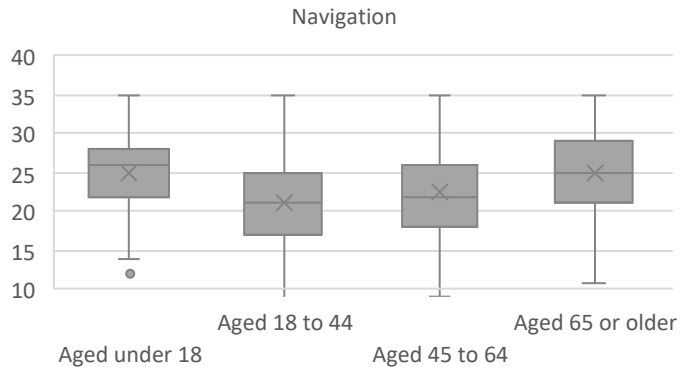


Figure 7.17: Boxplot of Care coordination: Navigation by age

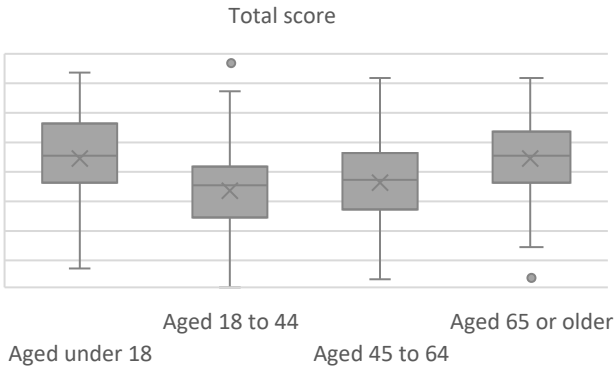


Figure 7.18: Boxplot of Care coordination: Total score by age

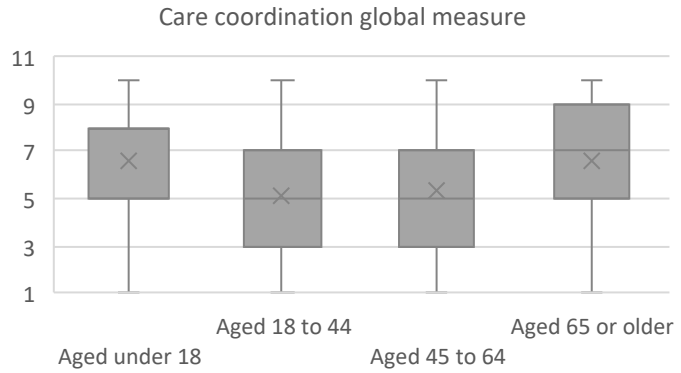


Figure 7.19: Boxplot of Care coordination: Care coordination global measure by age

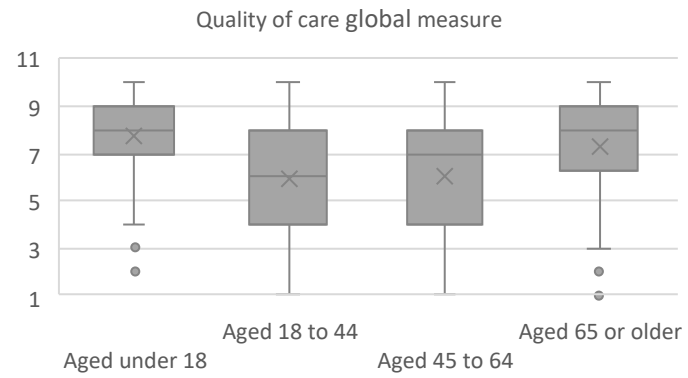


Figure 7.20: Boxplot of Care coordination: Quality of care global measure by age

Care coordination by education

Comparisons were made by **education** status, between those with trade or high school qualifications (n=176, 48.89%), and those with a university qualification (n=184, 51.11%).

A two-sample t-test was used when assumptions for normality and variance were met, or when assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used.

A two sample t-test indicated that the mean score for the Care coordination Navigation scale [t(358) = -2.37 , p = 0.0185] was significantly lower for participants in the Trade or high school subgroup (Mean = 22.11, SD = 6.42) compared to participants in the University subgroup (Mean = 23.62, SD = 5.65.)

The **Care coordination: navigation** scale measures the ability of a patient to navigate the healthcare system including knowing important contacts for management of condition, role of healthcare professional in management of condition, healthcare professional knowledge of patient history, ability to get appointments and financial aspects of treatments. On average, participants in the University subgroup scored higher than participants in the Trade or high school subgroup, healthcare system navigation was average for both groups.

Table 7.13: Care coordination by education summary statistics and T-test

Care coordination scale	Group	Number (n=360)	Percent	Mean	SD	T	dF	p-value
Communication	Trade or high school	176	48.89	35.22	11.00	-0.52	358	0.6030
	University	184	51.11	35.79	9.84			
Navigation	Trade or high school	176	48.89	22.11	6.42	-2.37	358	0.0185*
	University	184	51.11	23.62	5.65			

Table 7.14: Care coordination by education summary statistics and Wilcoxon test

Care coordination scale	Group	Number (n=360)	Percent	Median	IQR	W	p-value
Total score	Trade or high school	176	48.89	57.00	22.00	14736.00	0.1402
	University	184	51.11	60.00	17.00		
Care coordination global measure	Trade or high school	176	48.89	6.00	5.00	15139.00	0.2833
	University	184	51.11	6.00	4.00		
Quality of care global measure	Trade or high school	176	48.89	7.00	3.25	14812.00	0.1579
	University	184	51.11	7.00	2.25		

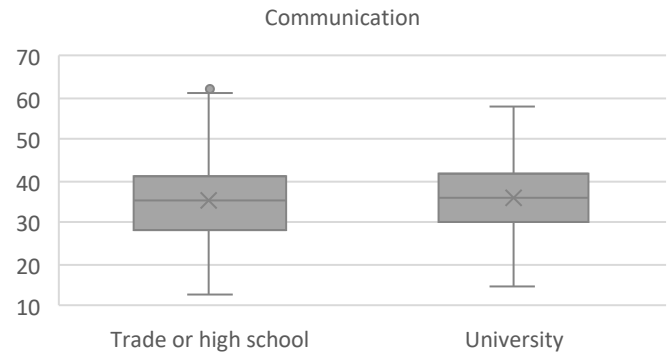


Figure 7.21: Boxplot of Care coordination: Communication by education

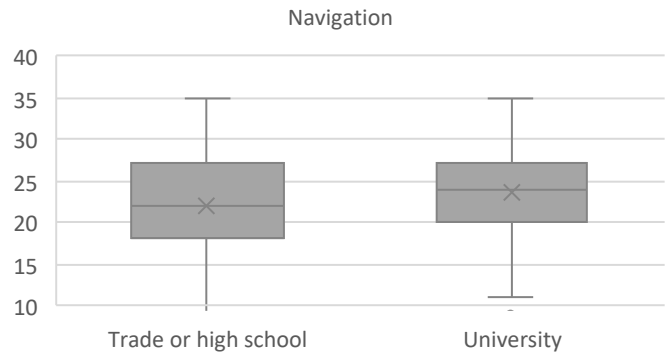


Figure 7.22: Boxplot of Care coordination: Navigation by education

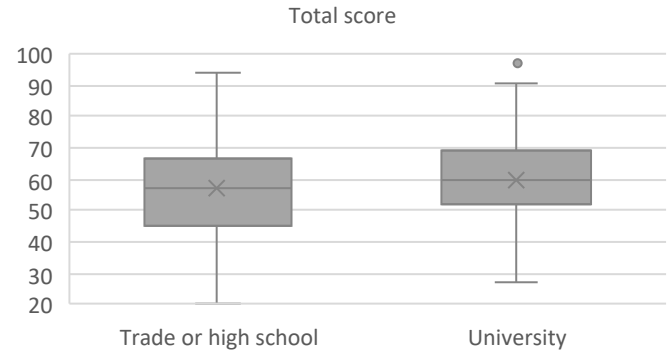


Figure 7.23: Boxplot of Care coordination: Total score by education

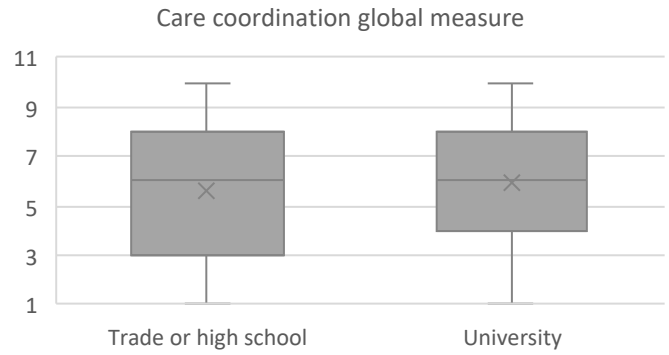


Figure 7.24: Boxplot of Care coordination: Care coordination global measure by education

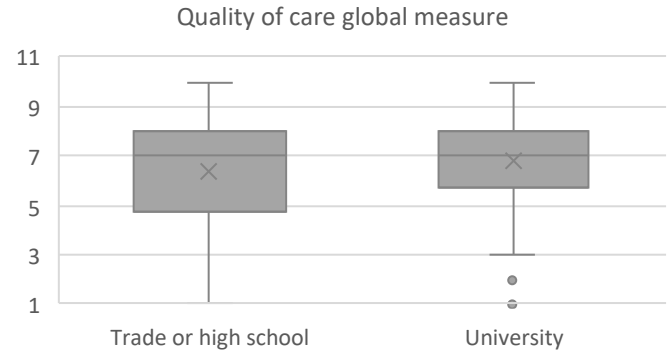


Figure 7.25: Boxplot of Care coordination: Quality of care global measure by education

Care coordination by location

The **location** of participants was evaluated by postcode using the Australian Statistical Geography Maps (ASGS) Remoteness areas accessed from the Australian Bureau of Statistics. Those living in regional or remote areas (n=102, 27.72%) were compared to those living in a metropolitan area (n=266, 72.28%).

A two-sample t-test was used when assumptions for normality and variance were met, or when assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used.

No significant differences were observed between participants by **location** for any of the Care coordination scales.

Table 7.15: Care coordination by location summary statistics and T-test

Care coordination scale	Group	Number (n=368)	Percent	Mean	SD	T	dF	p-value
Communication	Regional or remote	102	27.72	34.81	10.31	-0.85	366	0.3954
	Metropolitan	266	72.28	35.84	10.35			
Total score	Regional or remote	102	27.72	57.13	15.30	-1.12	366	0.2654
	Metropolitan	266	72.28	59.05	14.55			

Table 7.16: Care coordination by location summary statistics and Wilcoxon test

Care coordination scale	Group	Number (n=368)	Percent	Median	IQR	W	p-value
Navigation	Regional or remote	102	27.72	22.00	9.00	12548.00	0.2644
	Metropolitan	266	72.28	23.00	7.00		
Care coordination global measure	Regional or remote	102	27.72	6.00	4.75	13222.00	0.7047
	Metropolitan	266	72.28	6.00	4.00		
Quality of care global measure	Regional or remote	102	27.72	7.00	2.75	13300.00	0.7694
	Metropolitan	266	72.28	7.00	3.00		

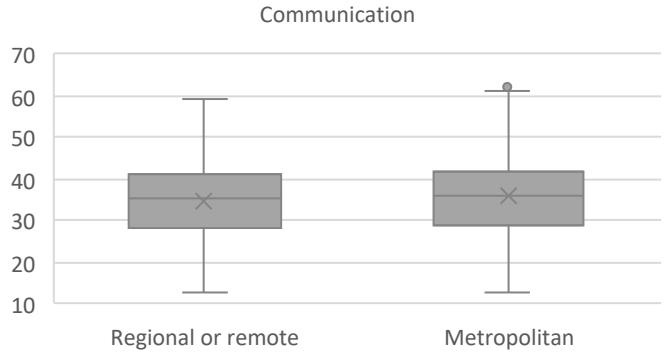


Figure 7.26: Boxplot of Care coordination: Communication by location

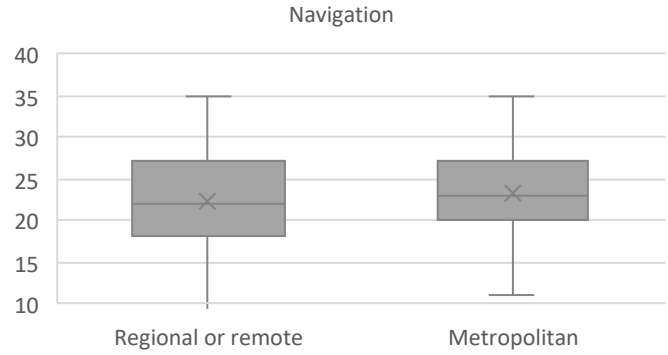


Figure 7.27: Boxplot of Care coordination: Navigation by location

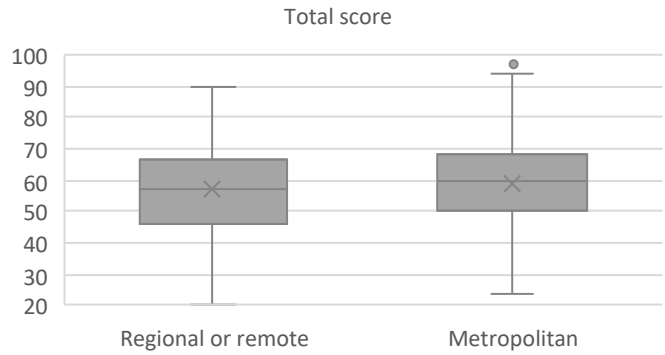


Figure 7.28: Boxplot of Care coordination: Total score by location

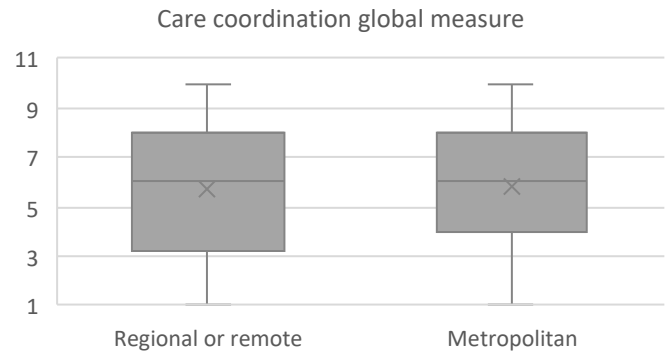


Figure 7.29: Boxplot of Care coordination: Care coordination global measure by location

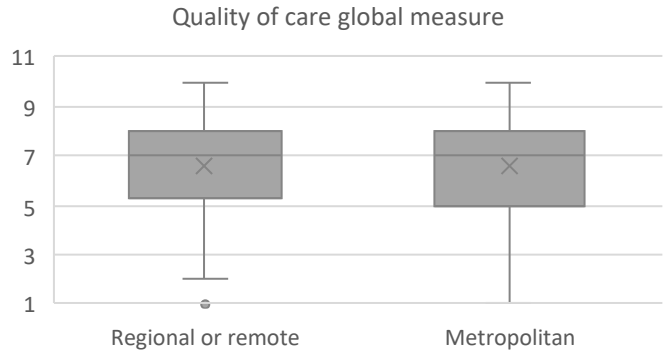


Figure 7.30: Boxplot of Care coordination: Quality of care global measure by location

Care coordination by socioeconomic status

Comparisons were made by **socioeconomic status**, using the Socio-economic Indexes for Areas (SEIFA) (www.abs.gov.au), SEIFA scores range from 1 to 10, a higher score denotes a higher level of advantage. Participants with a mid to low SEIFA score of 1-6 (n=182, 49.46%) compared to those with a higher SEIFA score of 7-10 (n=186, 50.54%).

A two-sample t-test was used when assumptions for normality and variance were met, or when assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used.

A two sample t-test indicated that the mean score for the Care coordination Communication scale [t(366) = -2.71 , p = 0.0071] was significantly lower for participants in the Mid to low status subgroup (Mean = 34.09, SD = 10.43) compared to participants in the Higher status subgroup (Mean = 36.98, SD = 10.07.)

A two sample t-test indicated that the mean score for the Care coordination Total score scale [t(366) = -3.78 , p = 0.0002] was significantly lower for participants in the Mid to low status subgroup (Mean = 55.63, SD = 15.02) compared to participants in the Higher status subgroup (Mean = 61.34, SD = 13.99.)

Wilcoxon rank sum tests with continuity correction indicated that the median score for the Care coordination Navigation scale [W = 12262.00 , p = <0.0001] was significantly lower for participants in the Mid to low status subgroup (Median = 21.00, IQR = 8.00) compared to participants in the Higher status subgroup (Median = 25.00, IQR = 7.00.

Wilcoxon rank sum tests with continuity correction indicated that the median score for the Care coordination Care coordination global measure scale [W = 13780.00 , p = 0.002] was significantly lower for participants in the Mid to low status subgroup (Median = 5.00, IQR = 5.00) compared to participants in the Higher status subgroup (Median = 7.00, IQR = 3.75.

Wilcoxon rank sum tests with continuity correction indicated that the median score for the Care coordination Quality of care global measure scale [W = 13838.00 , p = 0.0022] was significantly lower for participants in the Mid to low status subgroup (Median = 7.00, IQR = 3.00) compared to participants in the Higher status subgroup (Median = 8.00, IQR = 3.00.

The **Care coordination: communication** scale measures communication with healthcare professionals, measuring knowledge about all aspects of care including treatment, services available for their condition, emotional aspects, practical considerations, and financial entitlements. On average, participants in the Higher status subgroup had a higher score for communication compared to Mid to low status, however, healthcare communication was average for both groups.

The **Care coordination: navigation** scale measures the ability of a patient to navigate the healthcare system including knowing important contacts for management of condition, role of healthcare professional in management of condition, healthcare professional knowledge of patient history, ability to get appointments and financial aspects of treatments. On average, participants in the Higher status subgroup scored higher than participants in the Mid to low status subgroup. This indicates that healthcare navigation was good for participants in the Higher status subgroup, and average for participants in the Mid to low status subgroup.

The **Care coordination: total score** scale measures communication, navigation and overall experience of care coordination. On average, participants in the Higher status subgroup scored higher than participants in the Mid to low status subgroup. This indicates that communication, navigation and overall experience of care coordination was average for participants in the Higher status subgroup, and poor for participants in the Mid to low status subgroup.

The **Care coordination: care coordination global measure** scale measures the participants overall rating of the coordination of their care. On average, participants in the Higher status subgroup scored higher than participants in the Mid to low status subgroup. This indicates that, overall care coordination was good for participants in the Higher status subgroup, and average for participants in the Mid to low status subgroup.

The **Care coordination: Quality of care global measure** scale measures the participants overall rating of the quality of their care. On average, participants in the Higher status subgroup had a higher score for quality of compared to Mid to low status, however, quality of care was good for both groups.

Table 7.17: Care coordination by socioeconomic status summary statistics and T-test

Care coordination scale	Group	Number (n=368)	Percent	Mean	SD	T	dF	p-value
Communication	Mid to low status	182	49.46	34.09	10.43	-2.71	366	0.0071*
	Higher status	186	50.54	36.98	10.07			
Total score	Mid to low status	182	49.46	55.63	15.02	-3.78	366	0.0002*
	Higher status	186	50.54	61.34	13.99			

Table 7.18: Care coordination by socioeconomic status summary statistics and Wilcoxon test

Care coordination scale	Group	Number (n=368)	Percent	Median	IQR	W	p-value
Navigation	Mid to low status	182	49.46	21.00	8.00	12262.00	<0.0001*
	Higher status	186	50.54	25.00	7.00		
Care coordination global measure	Higher status	182	49.46	5.00	5.00	13780.00	0.002*
	Mid to low status	186	50.54	7.00	3.75		
Quality of care global measure	Higher status	182	49.46	7.00	3.00	13838.00	0.0022*
	Mid to low status	186	50.54	8.00	3.00		

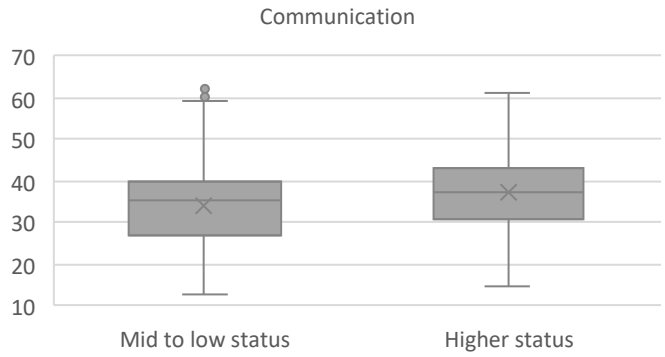


Figure 7.31: Boxplot of Care coordination: Communication by socioeconomic status

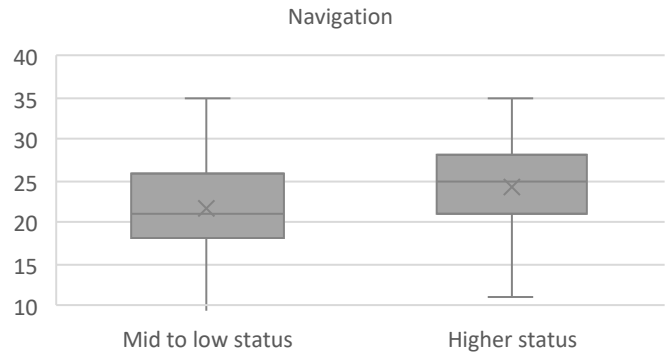


Figure 7.32: Boxplot of Care coordination: Navigation by socioeconomic status

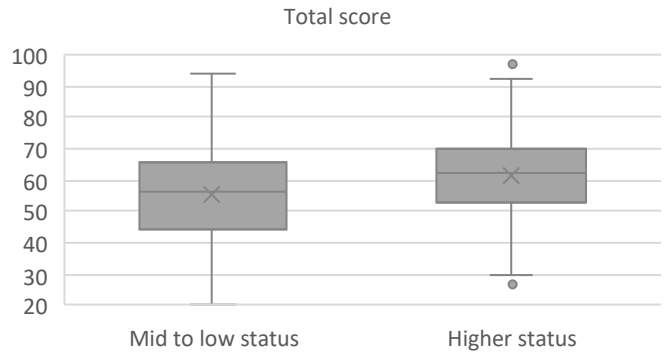


Figure 7.33: Boxplot of Care coordination: Total score by socioeconomic status

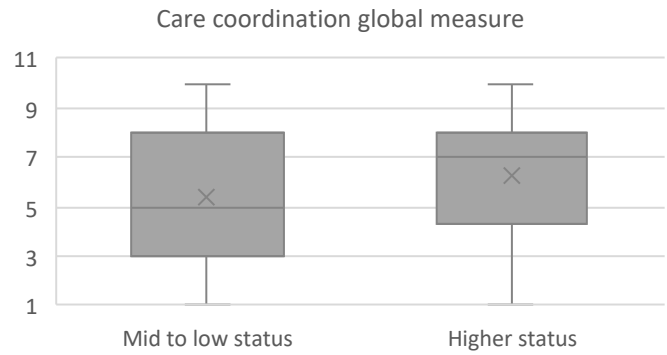


Figure 7.34: Boxplot of Care coordination: Care coordination global measure by socioeconomic status

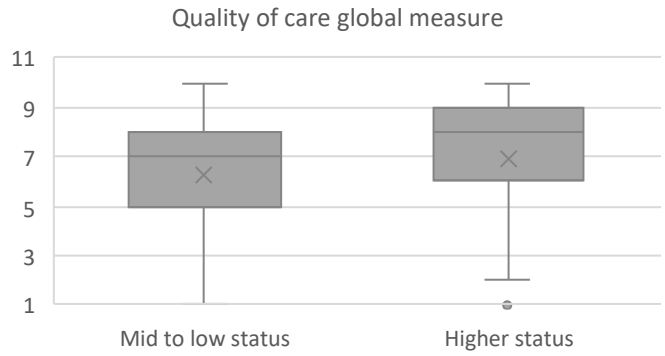


Figure 7.35: Boxplot of Care coordination: Quality of care global measure by socioeconomic status

Experience of care and support

In the structured interview, participants were asked what care and support they had received since their diagnosis. This question aims to investigate what services patients consider to be support and care services. The most common responses were that they did not receive formal support (25.12%), found support and care from hospital or clinical setting (23.38%), family and friends (20.65%), and charities (17.41%). Other themes included peer support or other patients (13.93%), and challenges accessing support (12.44%).

Participant describes getting care and support from hospital or clinical setting

The most support, probably the only support I've really received has been from my GP and my clinicians, my GP especially, he has been really good at trying to help me manage my pain. He's been really good at trying to…he tries to get me dressings and supplies and where he can he'll bulk on my appointments even though he's a private practice so that I can use the money I would have used on the appointment to go get medications or dressing.
Participant 012_2023AUDSK

PARTICIPANT: Yes, I've received support from the hospital when she was first born. They helped a lot. They helped us with hostel in the hospital so we could stay there. They helped. They gave us a grant. They helped us with the cost of the milk, they helped us with the cost of the machine, so it was cheaper.
INTERVIEWER: They, the hospital helped quite a lot when she was just born and I think any other support groups, patient groups.
PARTICIPANT: Oh well, yeah. Like the support groups like the Heart Kids and the 22 Q support groups, they have helped in like emotional support and just…
Participant 013_2023AUDPA

Yes. Well we've had a lot of support in terms of what the health system has provided us, like I've said with NAME NEUROLOGIST, and a lot of support from the local hospital who were very good when she used to come in with her seizures. We've had a lot support from the ECDP in the school there, I found them to be quite helpful. And the kindy have been really good because they have an extra person on staff to just have CHILD'S NAME at kindy, which is unbelievable experience for her.
Participant 057_2023AUDPA

Participant describes that they did not receive any formal support

No, not really. Yeah. I mean, when I had the interferon treatment, it was all through the hospital. So no, I wasn't aware of any other community support services that were available and none, none was made was, they didn't tell me about anything else was available. So, no, not not throughout.
Participant 011_2023AUORC

No, because it's not a recognized condition. Nobody knows about it. It's not on any of their lists at any of their foundations or their centers. Never heard of it. So it's not something they'll come and help out with. It's not on the list. All these joints are run by the government and the government gives them a list and if it's not on the list then you know and I feel like that's. You know that one of the things with this too, it's just like nobody knows about it hurt. No one's heard of it.
Participant 006_2023AUDSK

Not a bit, nothing.
067_2023AUENM

None, from nowhere.
Participant 061_2023AUDNS

Participant describes getting care and support from family and friends

Not really much. It's all just informal support from family and friends and just asking for help when we need it. Yes, that's really been it.
Participant 053_2023AUDPA

We definitely had that support there, which was very reassuring I guess to know you're in the hands of experts, and then also family looking after the other kids is an emotional support.
Participant 058_2023AUDPA

Again, my family just stepped up and were just a huge help. They were our number one support. The hospital has been fantastic. All his therapists are amazing. I know I can email or call anyone at any time, and they will help. It's never too much of an effort for them to shoot an email back or give me a call to say, "Maybe you should try this or give this a go." We've been just lucky with support, medical, and family wise.
Participant 060_2023AUDPA

Participant describes that they did not need or seek help or support

No, but like I was saying, thinking, I think I've been thinking. I think maybe in another year or so, I would. I think I will need somebody to clean. Yeah.
Participant 005_2023AUDIS

I can say that I haven't, because I have never asked for it, and I don't think anyone is particularly aware of it. I have had help and offers of what I can do in terms simply of age. Over 85 or whatever it is, there are various meetings at my local council. I've been given numbers to ring if I need help in the house and all sorts of things like that. That has nothing to do with the scleroderma, that's just to do with the age. As far as I am aware, as far as I can remember and think, I don't have any special treatment because of that. I don't think a lot of people are even aware of it.
Participant 012_2023AUDIS

No, but I guess I haven't reached out either
Participant 015_2023AUDIS

Participant describes getting care and support from charities

Community health, outreach programs which was the kids team in the early days. I did to get Cerebral Palsy Alliance when it was called the testing centre. Each program we were able to get into. All the way searching for programs and outside support. Really, that's about it. We had two close friends in the room that helped a little bit. They had their own issues. They didn't value the kids informally, more than likely have mental illness themselves. You got to be very careful who you bring into the home. Friends and family were just too far away.
Participant 045_2023AUDPA

The support from the helpline when I rang the Tuberous Sclerosis Association, TSA Australia helpline, a helpline or-- Then obviously friends and family support has been huge. One thing that practical support has been really at the time, the meals, people coming and just picking up the vacuum cleaner and doing that. That's what I found amazingly helpful and looking at the other two, like taking NAME and NAME for play dates and doing stuff for them. That really helped so much. The practical stuff is really helpful. Not asking me what can I do to help you but actually initiating and doing something blew me away. You don't really realise what you're needing until someone actually does it. [laughs].
Participant 048_2023AUDPA

Participant describes the challenges of finding or accessing support

No, I don't think so. I don't know I'm eligible for anything. I did briefly look into the NDIS while I was still recovering after my second pregnancy. I also needed to start the NDIS for my son. I did that and I know it's such a nightmare system and I just did not have the mental space at the time. I haven't looked into anything like that or assumed I wasn't eligible.
Participant 004_2023AUDPA

No, I couldn't. The hardest thing is I couldn't get any support because I had no diagnosis. You know, and that's like I spoke to the NDIS the other day. And they don't even have Paget's on their thing because it's for older people so, and because I'm only 51, they put me under osteoarthritis or something like that. So I've got some stuff I've got a doctor to fill out, and I've got some stuff I've got to fill out and everything like that to actually send it off to them. But until I got diagnosed, I couldn't get any help from anybody.
Participant 014_2023AUORC

No. I've tried to seek it out, but I haven't got any.
Participant 014_2023AUDIS

We haven't really had any. The doctor told us to apply for even the healthcare card for her. That's been taking a while. He said to apply for NDIS. Never heard back from them. There's otherwise no support for her. Otherwise, it's paediatrician support. We haven't received anything.
Participant 052_2023AUDPA

Participant describes getting care and support from psychologist or counselling service

No, except for. I don't know what they call themselves now, but veterans and veterans families counselling service through the Department of Veterans Affairs, they provided quite a bit of counselling for free. Actually, a lot of what we discussed was not HS related, but family related. You know how my family is and how I fit within it, but I think that was helpful. It's been many years since I did that part, okay.
Participant 008_2023AUDSK

We've received support and care from psychologists over the years, especially ones who've been specialists in autism spectrum disorder. That's been very important for all of us, to understand what we were doing where we were trying to go, what we were trying to achieve. That's probably been it for us. We've

not really had any support workers or other people involved.
Participant 044_2023AUDPA

Participant describes getting care and support from domestic services and/or home care

PARTICIPANT: Yes, I have. So with my local council I get home help. Which is supposed to be a fortnight. That's because of shortage of stuff is only once a month where they come in and they sweep my floors and then mop the floors and they clean the bathroom, so the handbags and the toilet and the shower.
INTERVIEWER: That's lovely. That's really helpful, isn't?
PARTICIPANT: It it's massively helpful. Yeah.
Participant 010_2023AUDIS

No, only when he needed dressings. Like I, I was a community nurse, a local community nurse. And when he needed the the dressings for the pilot of the sinus, you had to attend the dressing clinic once a week and they would give you supplies for six days. And then I could do the dressings at home sort of thing. But yes, we would go, I don't think community health ever came here. I think we were taking PATIENT to to the community Health Center. So I might have. I might have had one of the girlfriends. He had a couple of ingrown toenails that I think he had two operations on. And we were doing it at the clinic, but PATIENT was screaming the place down and the room we were doing it into was next to the the magistrate and the and they were saying you need to do it somewhere else. So I think NAME used to come and do the dressings here at home so he could scream and set off in it. I mean she you're doing the very best thing.
Participant 040_2023AUDPA

Apart from household help, no, I don't actually need anything else. There's a man from church comes to mow my lawn, which is tiny, but he does a much better job on the edges than I ever could. So I and I…a couple years ago I paid somebody to do some weeding, simply because it arranged so much like I normally no, I don't have any other care.
Participant 003_2023AUDNS

Participant describes getting care and support from peer support or other patients

PARTICIPANT: Not really. I am in a support group on Facebook, so I'm not sure if that counts.

INTERVIEWER: Well, you've mentioned it, so that's good that you've got support. So is there anything else you want to add? What kind of support is that?
PARTICIPANT: It's just people sharing their experience with HS. It's just something that makes me feel like, you know, I'm not alone with what I'm dealing with. I don't know anyone in my personal life who has this. So I joined a group where other people have it and we all kind of share our thoughts and experience and inspirational quotes and stuff like that to make sure that we're all okay, I guess.
Participant 010_2023AUDSK

Well, apart from all the specialists we've had, I can't remember if the OT and the speech, he I don't know if they came from the council or where they came from originally, but we haven't had a anybody assisting us as pointing us in the right direction or anything like that. We've just done it ourselves. And as I mentioned, we're part of the support group and we touch base with them from time to time.
Participant 093_2023AUENM

The most support came from when I first joined the support group, the patient support group, because I found people on a similar journey going through the same things where we could share information and and and and give and given advice on who to see, what to see, things like that. The rest of it's a bit hard because I've now been running the organization for over 15 years, so I'm supporting myself really. It's funny but yeah but no, the the support group is probably I would say my, my biggest place for to gather support. The next one would be from international professionals who I have now formed friendships and alliances with as well. So again I'm very fortunate in that area because of the connections I've made. Most families don't have that connection, but the support group and and and also passionate doctors. Now that we've got a couple of those here that we as a family have connected to and they've gone out there and learned about the condition and how it affects families and also looked at it more holistically than just a number.
Participant 025_2023AUDPA

Participant describes getting care and support from respite care

But we came home on hospital in the home and that transitioned to some sort of funding package that allowed us to have a respite. Carers come into the home on a regular basis in those first few years and they then had to be trained in suction and and oxygen

control and feeding, the tube feeding. And so they were really really useful to have other services and we didn't access any Council services. That was just wasn't worth it for us. You know, like cleaning the house and stuff. No was that respite care was the main services. He, he then had you know learning assistance at school he had. Now he's got this communication guide, which is like a support worker, the orientation, mobility, that's sort of like a service that we still use today, which helps him orientate in any situation he's in. So it's part of that planning to a transition. No, I think that's it.
Participant 028_2023AUORC

Yes, we were part of very special kids and the Starlight Foundation. So very special kids provide a family support worker for us and also they provide my oldest daughter with a, a friend to play with an adult young adult friend to take to the park and do those sorts of things. Also did respite care for maze and organized short notice respite care when we had our son recently. So that takes a lot of pressure off us knowing. That she could have. She was taken care of and we didn't have to worry about care arrangements for her.
Participant 090_2023AUENM

PARTICIPANT: So that's true, We have respite.
INTERVIEWER: Oh, yes, excellent.
PARTICIPANT: And that's been through the with the NDIS just and that's twice a week we have that.
INTERVIEWER: Excellent. Excellent.
PARTICIPANT: Just trying to think of anything else. No, I think that's about it.
Participant 015_2023AUDPA

Participant describes getting care and support from NDIS

Yes. Well, NDIS then would be the only one, but that was such a battle and it was rejected and my doctor said, "You have to do it again," so we had to do it again. That means thousands of doctors going to get the specialist report to get waiting for reports. Since it's been in place, it's only that I found fabulous carers that it's worked. Then nobody seems to be able to tell you do this with NDIS and you do that and they say things, "You've got to find a course person." Well, who's a course person? Right. It's not easy. Now that I have the support of NDIS and the care that I do, but if I ever have to move back to where I came from, it's going to be a nightmare because I have to get, well, it's an hour away, so maybe some of the carers might come, some possibly can't.
Participant 001_2023AUDNS

PARTICIPANT: No, I haven't applied for anything other than the NDIS.
INTERVIEWER: Yeah, Okay.
PARTICIPANT: Mainly because I know this is permanent and it's getting worse. So yeah,
Participant 002_2023AUDNS

Our only real support has been prior to the NDIS we had, I think, allocation of around $8,000 a year. It's only happened very recently before the NDIS. Prior to that, I think we had an allocation of about $2,000 a year which we couldn't really spend. We used to save it right until the last month or six weeks, and then spend it, get carers around, and blah, blah, this and that. Go to the beaches or do something for ourselves. Go for a meal or go to the beaches or something.
Participant 050_2023AUDPA

Participant describes getting care and support in the form of financial support including financial counselling

I pay, I think it's $15 a year to fibrosis QLD and they have like you pay that $15 and I think you get. Like $100 of your hospital parking covered for the year, you get $150.00 towards physical subsidy kind of thing. So like if you go to the gym or if you need choose for the gym or something like that, then they pay for $150.00 worth of that for the year. And also they cover $100 of medications or medical things that you need and they do have a thing that they can grant. A free physio implement. So something like if we needed something to like a nebulizer, I haven't used that myself, but but no, it's also an option. I think that's what they cover for and that's about it. I haven't really reached out to anybody else for any support. I tried to get a healthcare card but that was about it.
Participant 013_2023AUORC

Yeah, we receive cystic fibrosis QLD, sometimes do not for ages, but sometimes do like information for parents nights. We've been to some of those. They also provide us with financial assistance of, I think it's, what is it, $100 a year for sport, $50 a year for hospital parking, and every couple of years they help us with nebulizers of up to about $500.00. So we get that and we get family. He gets the carer allowance. Which is 120 fortnight from Centrelink. I get support online from families and people living with CF. That's the biggest support and the friendships are made from other mums.
Participant 023_2023AUORC

So while I was probably when he was about a year old, I thought I guess financial counselling, I don't know if that that's part of that because I couldn't work as much as I was intending to. So just help managing debt. I don't think, I don't think we've really thought any other sort of community support. Not that I can think of.
Participant 089_2023AUENM

Table 7.17: Experience of care and support

Experience of care and support	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Did not receive any formal support	101	25.12	9	13.43	33	40.74	16	16.84	14	43.75	21	22.11	8	25.00	85	31.72	16	11.94	84	28.57	17	16.04
Hospital or clinical setting	94	23.38	26	38.81	15	18.52	14	14.74	6	18.75	25	26.32	8	25.00	52	19.40	42	31.34	61	20.75	33	31.13
Family/friends	83	20.65	16	23.88	7	8.64	20	21.05	0	0.00	37	38.95	3	9.38	51	19.03	32	23.88	52	17.69	31	29.25
Domestic services and/or home care (incl. transport)	76	18.91	5	7.46	16	19.75	32	33.68	0	0.00	19	20.00	4	12.50	56	20.90	20	14.93	61	20.75	15	14.15
Charities	70	17.41	15	22.39	3	3.70	13	13.68	0	0.00	29	30.53	10	31.25	31	11.57	39	29.10	40	13.61	30	28.30
Peer support/Other patients	56	13.93	9	13.43	11	13.58	10	10.53	5	15.63	18	18.95	3	9.38	36	13.43	20	14.93	41	13.95	15	14.15
Challenges of finding or accessing support	50	12.44	11	16.42	7	8.64	17	17.89	0	0.00	12	12.63	3	9.38	25	9.33	25	18.66	34	11.56	15	14.15

Care and support received	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Did not receive any formal support	101	25.12	10	10.31	44	33.59	42	36.84	5	8.33	55	27.78	42	21.43	28	25.23	73	25.09	63	31.50	38	18.81
Hospital or clinical setting	94	23.38	33	34.02	22	16.79	19	16.67	20	33.33	44	22.22	47	23.98	28	25.23	66	22.68	42	21.00	52	25.74
Family/friends	83	20.65	23	23.71	20	15.27	22	19.30	18	30.00	37	18.69	39	19.90	19	17.12	64	21.99	39	19.50	44	21.78
Domestic services and/or home care (incl. transport)	76	18.91	13	13.40	26	19.85	26	22.81	11	18.33	43	21.72	39	19.90	17	15.32	59	20.27	38	19.00	38	18.81
Charities	70	17.41	29	29.90	14	10.69	14	12.28	13	21.67	33	16.67	31	15.82	23	20.72	47	16.15	24	12.00	46	22.77
Peer support/Other patients	56	13.93	11	11.34	18	13.74	13	11.40	14	23.33	26	13.13	30	15.31	13	11.71	43	14.78	21	10.50	35	17.33
Challenges of finding or accessing support	101	25.12	17	17.53	16	12.21	7	6.14	10	16.67	28	14.14	29	14.80	13	11.71	37	12.71	19	9.50	31	15.35

Figure 7.36: Experience of care and support

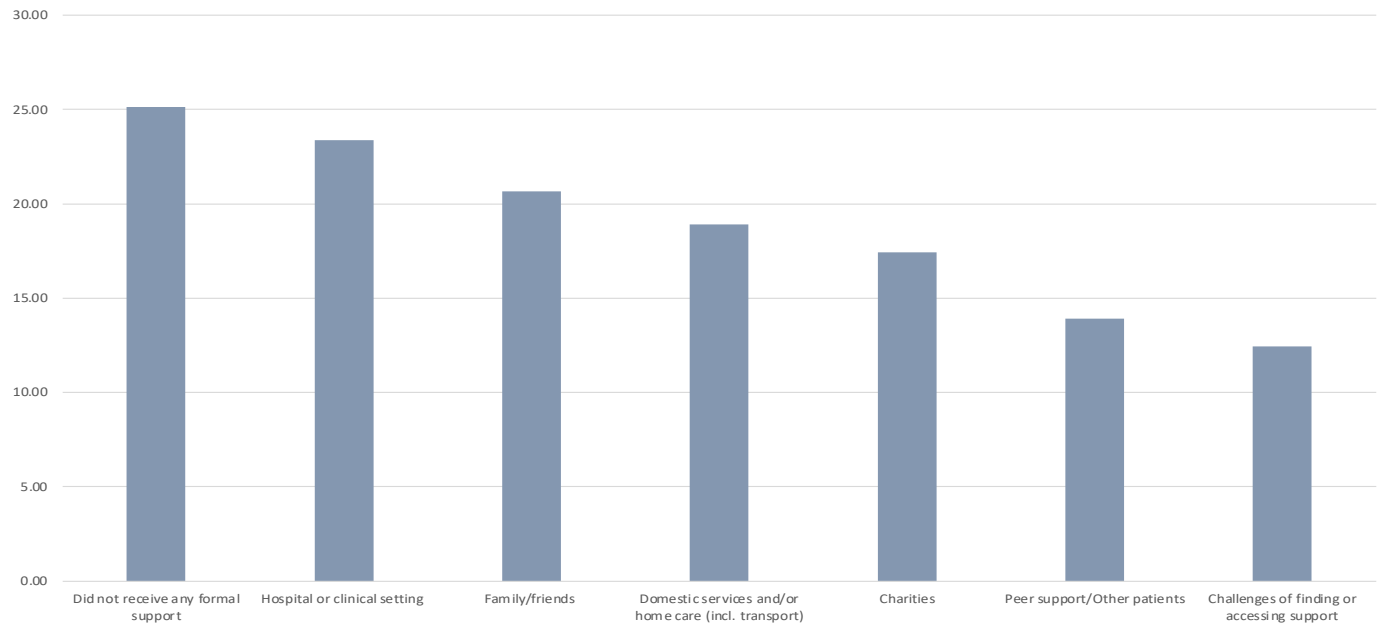


Table 7.18: Experience of care and support – subgroup variations

Care and support received	Reported less frequently	Reported more frequently
Did not receive any formal support	Developmental anomalies Aged under 18 Aged 65 plus	Diseases of the immune system Diseases of the skin Aged 45 to 64
Hospital or clinical setting		Developmental anomalies Aged under 18
Family/friends	Diseases of the immune system Diseases of the skin Other rare condition	Endocrine, nutritional or metabolic diseases
Domestic services and/or home care (incl. transport)	Developmental anomalies Diseases of the skin	Diseases of the nervous system
Charities	Diseases of the skin	Endocrine, nutritional or metabolic diseases Other rare condition Family or carer Male Aged under 18
Peer support/Other patients		
Challenges of finding or accessing support	Diseases of the skin Aged 18 to 44 Aged 45 to 64 Trade or high school University Regional or remote Metropolitan Mid to low status	

Section 8

Quality of life

Section 8: Quality of life

Impact on quality of life

In the structured interview, participants were asked whether they felt that their condition had affected their quality of life. Most commonly, the descriptions suggested that there was an overall negative impact on quality of life (63.43%), followed by an overall minimal impact on quality of life (10.20%). Other themes included a mix of positive and negative impact on quality of life (7.71%), overall no impact on quality of life (2.74%), and overall positive impact on quality of life (4.23%).

The most common themes in relation to a negative impact on quality of life were emotional strain (including family/change in relationship dynamics) (41.79%), reduced social interaction (23.88 %) and reduced capacity for physical activity/needing to slow down (20.40%). Other themes included managing side effects and symptoms and emotional strain (respectively 10.70%), altering lifestyle to manage condition (including being immunocompromised) (10.45%), and managing fatigue (7.21%).

The most common theme in relation to a positive impact on quality of life was realising what is important (giving perspective/staying positive) (6.97%).

Impact on mental health

In the structured interview, participants were asked if there had been an impact on their mental health. Most commonly, the descriptions suggested that overall, there was at least some impact on mental health (77.84%), and overall, there was no impact on mental health (5.97%).

Regular activities to maintain mental health

In the structured interview, participants were asked what they needed to do to maintain their emotional and mental health. The most common response was consulting a mental health professional (24.17%), coping strategies such as remaining social, lifestyle changes and hobbies(22.52%), and mindfulness and/or meditation (16.56 %). Other themes included no activities to maintain mental health (15.89%), the importance of family and friends in maintaining their mental health (14.90%), and the importance of physical exercise (14.90%).

Regular activities to maintain health

In the structured interview, participants were asked what were some of the things they needed to do everyday to maintain their health? The most common activities for general health were self-care e.g. more rest, accepting help, pacing (34.38%), complying with treatment/management (29.83%), and doing physical exercise/physically active (22.73 %). Other themes included understanding their limitations (19.89%), maintaining a healthy diet (14.20%), being organised and planning ahead (11.93%), and maintaining a normal routine (8.24%).

Experience of vulnerability

In the structured interview, participants were asked if there had been times that they felt vulnerable. The most common responses were that they felt vulnerable when having sensitive discussion (diagnosis, treatment decision) (16.67%), because of interactions with the medical team(14.44%), and experiencing side effects from treatment or symptoms from condition (9.44 %). Other themes included thinking about disease course/incurable condition (8.33%), during or after treatments (6.67%), and when feeling sick/unwell (5.56%).

As a follow up question, participants described ways that they managed feelings of vulnerability. The most common ways to manage vulnerability were using self-help methods (resilience, acceptance, staying positive) (7.78%), and support from nurse or treatment team (3.89%). Other themes included getting support from family and friends (3.33%), and support from mental health professionals (2.22%).

Impact on relationships

Most commonly, the descriptions suggested that overall, there was a negative impact on relationships (36.82%), and overall, there was a positive impact on relationships (23.13%). Other themes included overall, no impact on relationships (11.91%), and overall, there was an impact on relationships that was neither positive nor negative (10.95%).

The most common themes in relation to having a negative impact on relationships was from the dynamics of relationships changing due to anxiety, exacerbations and/or physical limitations of condition (25.37%). from people not knowing what to say or do and withdrawing from relationships (22.14%). This was followed by social isolation (10.70 %). The most common reasons for a positive impact on relationships was that people were supportive and well-meaning (15.67%).

Burden on family

In the structured interview, participants were asked whether they felt that their condition placed additional burden on their family. Most commonly, the descriptions suggested that overall, there was a burden on their family (62.60.19%), overall, there was not a burden on their family now but they anticipate this will change in the future (4/26%), and overall, there was not a burden on their family (21.02.64 %).

The main reason that participant described their condition being a burden were the extra household duties and responsibilities that their family must take on(23.01%), and the mental/emotional strain placed on their family (9.94%). Others described the extra assistance needed getting to appointments (5.97 %) and that the burden on family was temporary or only during treatment (3.69 %).

Cost considerations

In the structured interview, participants were asked about any significant costs associated with having their condition. The most common descriptions were that overall, there was at least some cost burden (65.23%), and overall, there was no cost burden (18.87%).

Where participants described a cost burden associated with their condition, it was most commonly in relation to needing to take time off work (32.78%), the cost of treatments (including repeat scripts) (30.79%), and the cost specialist appointments (26.82 %). Other themes included diagnostic tests and scans (12.91%), the cost of parking and travel to attend appointments (including accommodation) (12.91%), needing to special equipment (8.61%), a family member needing to take time off work (5.96%) allied health care (5.63%), needing to special creams, ointments or complementary therapies (4.30%), and needing a special diet or lifestyle adaptation (3.64%).

Where participants described a cost burden associated with their condition, it was most commonly in relation to nearly everything was paid for through the public health system (21.52%).

Overall impact of condition on quality of life

In the online questionnaire, participants were asked to rate the overall impact their condition on quality of life. Quality of life was rated on a Likert scale from one to seven, where one is Life was very distressing and seven is life was great. The average score was in the Life was a little distressing range (median=3.00, IQR=2.00).

Fear of progression

The Fear of Progression questionnaire measures the level of anxiety people experience in relation to their conditions. The Fear of Progression questionnaire comprises a total score, between 12 and 60, with a higher score denoting increased anxiety. Summary statistics for the entire cohort are displayed in Table 8.10. Overall the entire cohort had a mean total score of 37.09 (SD = 10.40), which corresponds to moderate levels of anxiety.

On average, participants in the Diseases of the skin subgroup scored higher than participants in the Endocrine, nutritional or metabolic diseases subgroup. This indicates that participants in the Diseases of the skin subgroup had high levels of anxiety, and participants in the Endocrine, nutritional or metabolic diseases subgroup had moderate levels of anxiety.

On average, participants in the Female subgroup had a higher score compared to Male, however, both groups had moderate levels of anxiety.

On average, participants in the Aged 18 to 44 subgroup had a higher score compared to Aged 65 or older, however, both groups had moderate levels of anxiety.

Impact on quality of life

In the structured interview, participants were asked whether they felt that their condition had affected their quality of life. Most commonly, the descriptions suggested that there was an overall negative impact on quality of life (63.43%), and this was followed by an overall minimal impact on quality of life (10.20%). Other themes included a mix of positive and negative impact on quality of life (7.71%), overall no impact on quality of life (2.74%), and overall positive impact on quality of life (4.23%).

The most common themes in relation to a negative impact on quality of life were emotional strain (including family/change in relationship dynamics) (41.79%), reduced social interaction (23.88 %) and reduced capacity for physical activity/needing to slow down (20.40%). Other themes included managing side effects and symptoms and emotional strain (respectively 10.70%), altering lifestyle to manage condition (including being immunocompromised) (10.45%), and managing fatigue (7.21%).

The most common theme in relation to a positive impact on quality of life was realising what is important (giving perspective/staying positive) (6.97%).

Participant describes a minimal impact on quality of life that has a general or temporary impact

Your quality to some extent, to some extent. I wouldn't go too far. A little bit, A little bit.
Participant 012_2023AUORC

Compared to my other conditions, I wouldn't say it's impacted with all that much.
Participant 019_2023AUDSK

No, it hasn't affected us too much. As I said, she, she lives life just like her brother and sister. She's very able. The only thing it is, is maybe being a little bit behind her peers, but as in family life, there's been no there's nothing that challenged us so far or made a massive difference.
Participant 10_2023AUDPA

Participant describes a negative impact on quality of life due to the emotional strain (including family/change in relationship dynamics)

It has it, it's affected it, it's my partner has to be more aware. He has to be prepared to use his sick days to look after me rather than look after himself. It's affected relationships with family. It's left us financially struggling. It, on occasion, it affects my relationship with my children because I'm there, supposed to be their full time carer and some days I can't care for myself. Mentally it affects most of our family because they can see the wounds, they can smell them, they want to help but they can't and that plays on them. Especially my parents. That plays on their mental state often and it just makes life harder.
Participant 012_2023AUDSK

As we just talked in the previous one, my 15 year old daughter, her life has been impacted as she's been expected to care in a way for her sister that wouldn't normally happen in a sibling relationship. She's been expected to provide medical support and you know she has had to get a job to in order to afford the things that she likes to do for myself as well and NAME's quality of life very much depends on how much lotion we can afford to purchase and vitamins and pills and doctor's appointments and her access to allied health and that sort of thing. So everybody else's quality of life is juggled whilst providing her the best opportunity to be as well as possible
Participant 80_2023AUDIS

I'm not an equal partner anymore. I can't do things. I can't just go and do the dishes. I can't cook dinner. I used to be able to help clean around the house. Can't do that. I feel like I'm just breathing through, just not participating and not giving everything I do as much as I can. It's hard. You can't really, you hurt your hands to do these things.
Participant 018_2023AUDIS

Participant describes a negative impact on quality of life due to reduced capacity for physical activity/needing to slow down

Absolutely because I guess the scleroderma in itself didn't…Apart from having to go to a specialist appointment, didn't really affect anything until the pulmonary arterial hypertension kicked in. Since then, I've been very limited. I've had not a bad life, but within limits. I haven't been able to do the things that I would have done with my family because I can't walk as far and I can't go up hills, and I need to sit down. I'm a person who likes to go out to concerts, plays, and art galleries. I still did those things, but I found that they took a lot more out of me and I couldn't do them as easily. I would be holding everybody up because I had to sit and rest. Now, I'm actually past being able

to do those things. This is the arthritis rather than-- They're telling me it's nothing to do with the progression of scleroderma.
Participant 004_2023AUDIS

Well, it's certainly affected my quality of life because I can't even cook as I enjoy doing or go out for meals and enjoy. I do that, but I don't know what I'm eating. Oh, fatigue, I don't think I've mentioned that, but fatigue has been a big thing in a general way with scleroderma. I get very tired and I, by and large, have a nap almost every afternoon. If I'm sitting at the computer, my head hits the computer because I'm just asleep, really. That's certainly something that's made a difference.
Participant 012_2023AUDIS

Participant describes a negative impact on quality of life due to reduced social interaction

There has been a huge amount of stress on everyone in the family, including our youngest child. I like…one of my siblings actually literally no longer talks to me anymore because she just feels that we're all just a bit high drama and, you know, there's just a huge narrowing of social circle because it's just very difficult in order to get out and relate, you know, we can't, we can't fit into what other people are doing. There's the fact that it's been impossible for my husband to see his family because they're in LOCATION and traveling with PATIENT in the middle of the global pandemic is impossible. There's and then there's like all the positives in that, like, we just….also sort of have…so we don't kind of embrace who we are and love people for who they are. Don't go around just people because they're in different or you know, it's opened up and you know, silent.
Participant 87_2023AUENM

Very, very much so. I feel it's destroyed my, it's destroyed all my friendships. I have no friends. I had a reasonably good friend group…A lot of people can't handle the fact that I got so sick so quick and I look different and I walk different and they get embarrassed. So I am extremely isolated. Even my adult and teenage children struggle to be in public with me because I walk slow and I have a limp and they find it hard to accept, so I'm extremely socially isolated too and I find it really hard to get out in public on my own. I need a support person with me and the availability of that is very limited. So I find I'm extremely socially isolated, like to the point where I have no social aspect in my life at all.
Participant 016_2023AUDIS

Participant describes a negative impact on quality of life due to managing side effects and symptoms

Look, while symptoms were present, yeah, absolutely not. I suppose because I've seen what it could be or how bad it could be. Not to that extent. But yeah, definitely very uncomfortable. It's very painful and it sort of, yeah, it certainly had a minimizing impact on, on what you did and then how well you felt to go and to go and do usual things. And I suppose in terms of family, yeah, as a single parent, that then also meant if I didn't feel up to it, that my daughter wouldn't go places because I wasn't in a position to be able to take her and support that.
Participant 007_2023AUDSK

It has affected in certain ways because of obviously not being able to go out and do things and, you know, sort of making sure he's OK sort of thing before we do things, especially with his OCD and his anxiety.
Participant 11_2023AUDPA

PARTICIPANT: Significantly.
INTERVIEWER: Yes.
PARTICIPANT: It impacts on her ability to have friends at school. She's not the same cognitively as her peers. Her learning is delayed. Yes, all areas of her life are impacted. She misses things because of seizures. She misses things because of appointments. She gives most things a go. For our family, the other two kids definitely misses out on stuff because of her, and you constantly think and analyse what you do and how to have minimal impact on them, or you think of the effects on them and try to make considerations for them as well.
Participant 53_2023AUDPA

Participant describes a negative impact on quality of life due to being unable to travel/adapt significantly in order to travel

It's affected my quality of life a great deal. Constant stress and worry. Not having the normal freedoms that you would have in retirement to just impulsively take off somewhere because you're always checking, can I do this? Is everything going to be alright at home while I'm away? Family relationship? Yeah, because sometimes family members don't always understand that how my daughter is. Behaving is part of to DiGeorge and I might think that I'm defending her, which I'm not. Yeah, that that would be an average, an average stress, not a high one. But the impact on my time and lack of freedom is very high.
Participant 8_2023AUDPA

Yeah it, it has expected yeah because we we really couldn't go on holidays so we've had to just be looking after both of our kids and so yeah it's it has affected probably our relationship and our what we had anticipated our life would be like. But we've just adapted to that and changed what we had, you know our sort of our expectations. We we changed them and altered them to just doing stuff around our place and and you know getting into boat because we live on this river and it's really nice area. So we've just do boating and taught our kids kayaking and just did sort of home like things around the home and around the local area so that we can still have a good life. We still have a great life and a good quality of life. But well, I guess we've just adapted our expectations and, yeah, altered, altered that to what we'd sort of first thought we might end up doing.
Participant 9_2023AUDPA

Participant describes a negative impact on quality of life due to the emotional strain on self

The culture has, I mean, this half of the people, they don't know what I've got. I don't tell them. I don't know. It's just the close friends that they know what is wrong with me. But I don't talk to anyone else about my condition because I'm I'm, I'm ashamed kind of, yeah.
Participant 023_2023AUDSK

I feel like it has, because I can't do the things that I used to be able to do. I'm probably not as much fun to live with nowadays as I used to be, because it gets you down. When I stay in bed, I probably have one day a fortnight at least, or one day a week sometimes where I'm in bed. Particularly in the winter, I would stay in bed all day, one day a week, because I feel terrible and that impacts my relationship with my husband because he's hanging around the house waiting for me to get up energy I suppose. My grandchildren, I've got five grandchildren. I can't play with them like I used to and they wear you out.
Participant 013_2023AUDIS

Participant describes a negative impact on quality of life due to altering lifestyle to manage condition (including being immunocompromised)

Well, if you could take it off them, you would. If you could wear the pain, you would. It's a significant cost to your emotional well-being. You feel guilty and you you just, you you shut the doors. Well, I shut the doors to doing. We've cut down on a lot of our not that we, you know, had a hugely social engagement calendar, but very very selective in in what things we go to now even or even more so. You would. Yeah. So you and if anyone's sick it's like don't come over don't you know and we don't go out. Yeah. So that's a, that's a but that's that's something that you just you have to accept. It's you don't it's not it's not worth taking a risk on that.
Participant 009_2023AUDSK

Yes, it has I, especially with, with Raynaud's like, I can't like, I will not go to cold climate. If we go out to dinner, we always have to make sure that we sit inside. That's, I mean, that's pretty. That's not really quality of life. Look, I've had to get a cleaner in the house. Because due to my ulcers, I can't clean my bathroom. I'd want to. I came to that realization about three years ago. I had to stop my my netball. Yep. Look it, yep, it stopped me from playing the sport that I love around the house. I can't, you know, because of the joint pain and the right rheumatoid. I can't open jars. There's lots of things that that my husband and the kids have to help me with, help have to help with. I can't tie shoe laces anymore or do buttons, so I don't buy shoes with laces anymore.
Participant 015_2023AUDIS

Participant describes a negative impact on quality of life due to fatigue

PARTICIPANT: Yes. I'm just so tired. Before this, I was an extremely active woman playing basketball three, four times a week, and my girls would play with me and we were in the same team. We ran a business and I worked full-time. Income was really good, things were going really great. That all stopped quite quickly post shoulder replacement and this fatigue, I started to get very fatigued and have joint pain, which I didn't know at that time what it was. I think tiredness is probably number one.
INTERVIEWER: Yes.
PARTICIPANT: It's hard. I still look after the kids, but limited time because I have crashes all the time, you know, where you are just drained. Definitely have, I can't walk as far now, get too tired. Definitely can't do as much as I…might be a bit of age in in there too though. I was thinking that I'm barely, I'm 62 now. Things happen when you get older as well, so I can't just blame that, but yes.
Participant 007_2023AUDIS

Yes, because I'm not as active as I used to be. I'm…I sit on the lounge. I'm tired after. If I sit down, I've got to get up to do something because I forgot to do something. It's like, oh geez, I got to end up getting

someone else to do it because I just don't have the energy left to do it. And yes, the grand, because the grand, the grandchildren don't live close. So when they come to visit, you know. It's like a it's like a hurricane hitting the house after two hours. I'm exhausted.
Participant 011_2023AUDIS

Participant describes a negative impact on quality of life due to inability to work/changes with their work

Yeah, yeah, I do. I guess, you know, her dad and my's relationship broke down and yeah, I said I haven't been able to work. Her sister has grown up not knowing anything different apart from going to a ton of appointments. So yeah, not quite the life I envisaged, but that's what we got.
Participant 21_2023AUDPA

She's a different child to my son who has no genetic conditions or health conditions. She's just a different child that we didn't anticipate but we have adapted, and we accept her the way that she can, I guess, interact with us. I guess other sort of flow on impact. Like I say that limits what the family can do we can just all go to the pool or something like that. It's quite a logistical exercise. We can't go out for dinner or breakfast because CHILD'S NAME might not tolerate sitting in a highchair and start crying and screaming and that's a way of communicating because we can't communicate with her. We can't help her work forever. It limits our interaction with the world that we would otherwise be having. What else? My work. I can't really advance with my work given that I can't work full time. I need to work part-time to have days off to fit in all of CHILD'S NAME's appointments. I also need to have a very flexible workplace so that when we have appointments at NAME HOSPITAL that are impossible to change. If you don't want to wait another four months, I need to have a workplace that understands that I can't go to work that day I've got an appointment at the NAME HOSPITAL. It also has to be flexible.
Participant 61_2023AUDPA

Participant describes a negative impact on quality of life due to having no respite, condition is always there

I guess, well, it affects it. I guess it affects everything because she requires I guess 24 hour care. So it's like anything you're doing, any decisions you make is revolved around her and how she's feeling or what, whatever. You know, making decisions to go to a party. Well, we know how she's feeling today. People is that.
Participant 16_2023AUORC

Well, this is a painful one. It affects us greatly. We're very isolated, I haven't in over five years. I'm with my son 24/7, I shouldn't say 24/7, I'm with him every night. I go to work. I don't have any family, my relationships have failed. My son is very isolated because of his autism and he very angry, he doesn't want tuberous sclerosis, he wants to be the same as every other neurotypical child. When to try to talk to him about getting the services that he needs, he rebels against it because again, expecting services means that there's something different about him and he doesn't want to be different. I'm trying to get help for him, psychological help. It has been a nightmare, an absolute nightmare and I'm fortunate because it's been probably eight years of hell trying to get someone to support him and help him. He no longer trusts or wants to have anything to do with a therapist of any description so that makes it really difficult to move forward.
Participant 59_2023AUDPA

Participant describes a negative impact on quality of life due to intimacy challenges

I don't live with my family, so, like, my mum's gone. Yeah, my dad wouldn't bother. Nothing to do with him, but I have a partner and yeah, since…like, this is going to be crazy, but like, we haven't slept together in over a year, probably 2 years, and we're still together and we're not seeing anyone else. And that is because he understands that like when you do stuff or get hot or sweaty down there, it flares up and then I'm screwed up for days, so… Like, we just need to wait till I can get this surgery and then and then like, maybe I want to have kids. But, you know, hormones and stuff just flare this shit right up. So I'd prefer to have it gone before I'm pregnant or something, you know?
Participant 006_2023AUDSK

Yeah, I guess. I guess it has in the sense that yeah, I just get tired. Yeah, physically with hubby. Yeah, yeah, that's non existent. As I keep saying, it's shut, it's closed.
Participant 019_2023AUDIS

Participant describes a positive impact on quality of life because it brings people together/highlights supportive relationships

It has probably affected the quality of my life somewhat. I probably struggle to be active because I am tired quite a lot. It can struggle, you know it depending on where where I'm at at the time. If I'm flaring I can struggle to do things around the home. So it has had an impact on, on day-to-day living, has impacted relationships, yes and no. You know, my husband has has been involved in my my diagnosis from the beginning and has a fair understanding of how it impacts me. So it hasn't been easy on him. So it has impacted the relationship to some degree, but at the same time he's been involved in it and he has an understanding of what I'm going through and what it's like for me, so.
Participant 001_2023AUDSK

Quality of life? In some ways, yes. Not with family. I feel like they've all been very supportive. But I do worry about my daughters having the same condition.
Participant 011_2023AUDSK

Of your family, I suppose, yeah, because we've had to readjust how I always imagined I would raise a kid. Like we can't do mud. All playgrounds are made of bark chips, and he can't go in bark chips like can't go in sand pits. So there was a lot of it was a big adjustment into he can't just go and be free in the wild. But once those, once you kind of just reshift your thought pattern, there's it doesn't really we still go camping, we still go swimming in creeks, we still do. We still do everything we would do. We're just aware of the extra precautions or extra physio. After an activity in terms of relationships, both our families are fine. They're not. I don't want to say they're not bothered by it, but it's it's as if it's not there. They pitch in, they do his meds, they do his physio. When he does sleepovers, it's just it's just become part of their world as well. We did have a few friends though, that we kind of never heard from again once we disclosed it. Which isn't uncommon obviously for diagnosis. A lot of people they just so they were in self-preservation. Perhaps they need to remove themselves from that kind of, I don't know, pain, I don't know where it comes from, but but I was like as a that that's not uncommon in any sort of diagnosis like this.
Participant 25_2023AUORC

Participant describes a positive impact on quality of life as they realise what is important (giving perspective/staying positive)

I've had an absolutely wonderful life. I've, you know, traveled and hiked and done every sport I wanted to. And so all of that's wonderful. There's been some parts, but they're really challenging for, like hiking... I had my pocket nebulizer with me and had to stop every half an hour to take a break. I guess other people might say that my quality of life has been compromised and I don't really think so because I've had more experiences than most people my age right now. I can't join my friends in things that I would normally do and that's the post COVID syndrome on my CF damaged lungs. So you know my lung function currently is down to 38% so at the moment. My quality of life has changed dramatically. I'm still doing a lot of things, but I'm not playing golf here, so that means I'm not that. That's a bit of social interaction that's gone. Mind you I've just last week I started meeting the girls for Devonshire tea after golf, skip the golf, but still go for morning tea. So finding a way around that and… Yeah, I'm not. I'm used to doing my big power walk around… but now it's a stroll down to the little cafe. So I'm still meeting that walking buddy. But it's not the the exercise has been changed.
Participant 18_2023AUORC

No, not at this stage. I think at…because it's, because it's early days. I haven't had, like we discussed earlier there, there haven't been major side effects or anything like that. Obviously, my parents would be upset when they first found out that it came back. But yeah, I think we've sort of moved on from that and we're just sort of taking one sort of day at a time kind of thing and just seeing where where the treatment takes me in, you know, in the next few months and just I suppose just seeing where. What my next step is kind of thing like you know if it does come back, if it doesn't come back or if it shrinks more or what my next sort of step is to to get rid of it. But quality of life I haven't really changed too much. Obviously there's just that sort of breathing issue and and sort of the cough but everyday life just goes on. You know I still wake up every morning grateful that I'm still here and shower, shower and eat and yeah, nothing's nothing's changed. Go to work and and try and keep my mind off it as much as possible. So yeah.
Participant 24_2023AUORC

Table 8.1: Impact on quality of life

Impact on quality of life	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Overall negative impact on quality of life	255	63.43	40	59.70	58	71.60	70	73.68	21	65.63	49	51.58	17	53.13	176	65.67	79	58.96	185	62.93	68	64.15
Overall a minimal impact on quality of life	41	10.20	10	14.93	8	9.88	4	4.21	2	6.25	13	13.68	4	12.50	27	10.07	14	10.45	25	8.50	16	15.09
A mix of positive and negaitve impact on quality of life	31	7.71	7	10.45	4	4.94	5	5.26	3	9.38	7	7.37	5	15.63	21	7.84	10	7.46	24	8.16	7	6.60
Overall positive impact on quality of life	17	4.23	3	4.48	0	0.00	9	9.47	1	3.13	4	4.21	0	0.00	11	4.10	6	4.48	15	5.10	2	1.89
Overall no impact on quality of life	11	2.74	2	2.99	1	1.23	2	2.11	1	3.13	3	3.16	2	6.25	8	2.99	3	2.24	9	3.06	2	1.89

Impact on quality of life	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Overall negative impact on quality of life	255	63.43	53	54.64	85	64.89	76	66.67	41	68.33	131	66.16	120	61.22	75	67.57	180	61.86	133	66.50	122	60.40
Overall a minimal impact on quality of life	41	10.20	12	12.37	14	10.69	7	6.14	8	13.33	16	8.08	25	12.76	14	12.61	27	9.28	20	10.00	21	10.40
A mix of positive and negaitve impact on quality of life	31	7.71	9	9.28	11	8.40	9	7.89	2	3.33	16	8.08	15	7.65	6	5.41	25	8.59	11	5.50	20	9.90
Overall positive impact on quality of life	17	4.23	6	6.19	5	3.82	4	3.51	2	3.33	8	4.04	9	4.59	3	2.70	14	4.81	9	4.50	8	3.96
Overall no impact on quality of life	11	2.74	2	2.06	5	3.82	2	1.75	2	3.33	7	3.54	4	2.04	3	2.70	8	2.75	4	2.00	7	3.47

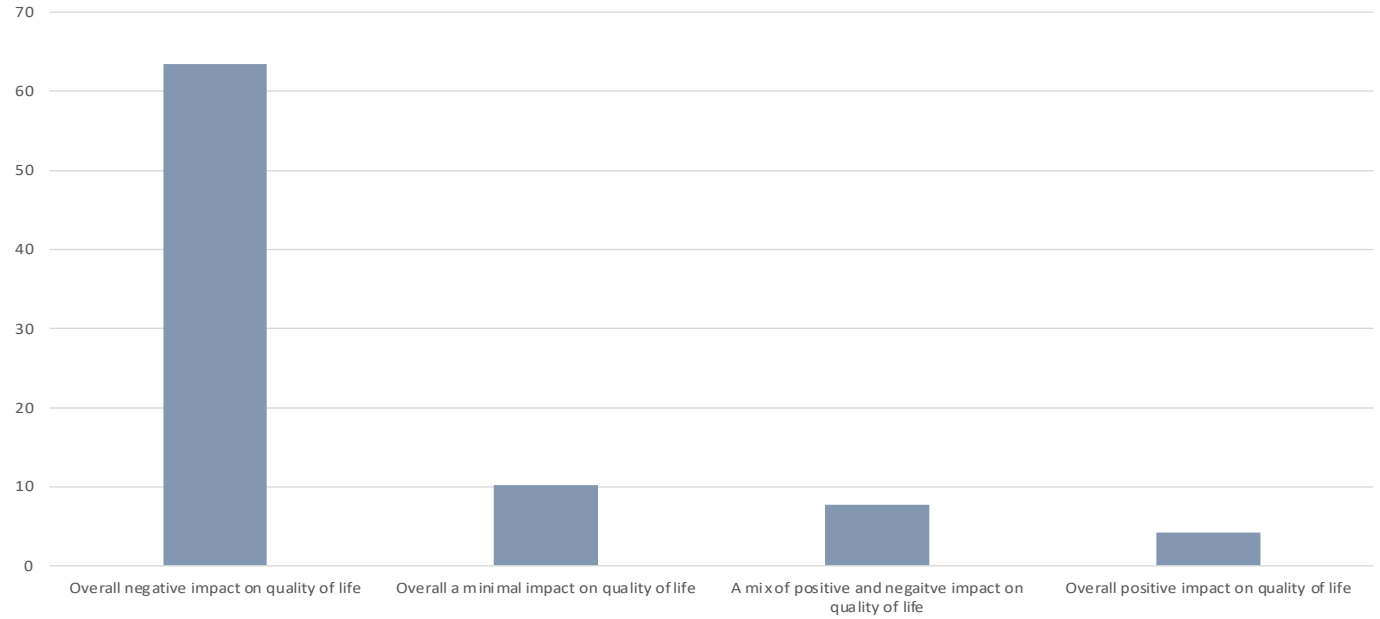


Figure 8.1: Impact on quality of life

Table 8.2: Impact quality of life – subgroup variations

Impact on quality of life	Reported less frequently	Reported more frequently
Overall negative impact on quality of life	Endocrine, nutritional or metabolic diseases Other rare condition	Diseases of the nervous system

Table 8.3: Impact on quality of life (Reasons)

Impact on quality of life (descriptions)	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Negative impact: emotional strain (including family/change in relationship dynamics)	168	41.79	34	50.75	44	54.32	37	38.95	7	21.88	34	35.79	12	37.50	109	40.67	59	44.03	129	43.88	38	35.85
Negative impact: reduced social interaction	96	23.88	16	23.88	19	23.46	21	22.11	5	15.63	26	27.37	9	28.13	61	22.76	35	26.12	73	24.83	22	20.75
Negative impact: reduced capacity for physical activity/needing to slow down incl. because of disability	82	20.40	8	11.94	31	38.27	15	15.79	7	21.88	18	18.95	3	9.38	69	25.75	13	9.70	61	20.75	21	19.81
Negative impact: emotional strain on self	43	10.70	5	7.46	4	4.94	4	4.21	3	9.38	18	18.95	9	28.13	31	11.57	12	8.96	29	9.86	14	13.21
Negative impact: managing side effects and symptoms	43	10.70	16	23.88	9	11.11	8	8.42	5	15.63	0	0.00	5	15.63	23	8.58	20	14.93	28	9.52	15	14.15
Negative impact: altering lifestyle to manage condition (including being immunocompromised)	42	10.45	9	13.43	8	9.88	19	20.00	1	3.13	0	0.00	5	15.63	23	8.58	19	14.18	32	10.88	10	9.43
Negative impact: unable to travel/adapt significantly in order to travel	39	9.70	10	14.93	3	3.70	3	3.16	1	3.13	18	18.95	4	12.50	21	7.84	18	13.43	27	9.18	12	11.32
Negative impact: fatigue	29	7.21	1	1.49	15	18.52	7	7.37	0	0.00	6	6.32	0	0.00	27	10.07	2	1.49	22	7.48	7	6.60
Positive impact: realise what is important (giving perspective/staying positive)	28	6.97	0	0.00	1	1.23	12	12.63	0	0.00	13	13.68	2	6.25	19	7.09	9	6.72	23	7.82	5	4.72

Impact on quality of life (descriptions)	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Negative impact: emotional strain (including family/change in relationship dynamics)	168	41.79	37	38.14	57	43.51	52	45.61	22	36.67	76	38.38	88	44.90	48	43.24	120	41.24	86	43.00	82	40.59
Negative impact: reduced social interaction	96	23.88	23	23.71	33	25.19	29	25.44	11	18.33	49	24.75	45	22.96	30	27.03	66	22.68	53	26.50	43	21.29
Negative impact: reduced capacity for physical activity/needing to slow down incl. because of disability	82	20.40	11	11.34	24	18.32	26	22.81	21	35.00	43	21.72	38	19.39	20	18.02	62	21.31	40	20.00	42	20.79
Negative impact: emotional strain on self	43	10.70	10	10.31	10	7.63	15	13.16	8	13.33	28	14.14	15	7.65	14	12.61	29	9.97	16	8.00	27	13.37
Negative impact: managing side effects and symptoms	43	10.70	14	14.43	14	10.69	8	7.02	7	11.67	21	10.61	22	11.22	13	11.71	30	10.31	18	9.00	25	12.38
Negative impact: altering lifestyle to manage condition (including being immunocompromised)	42	10.45	17	17.53	11	8.40	11	9.65	3	5.00	18	9.09	24	12.24	8	7.21	34	11.68	23	11.50	19	9.41
Negative impact: unable to travel/adapt significantly in order to travel	39	9.70	10	10.31	9	6.87	11	9.65	9	15.00	21	10.61	16	8.16	15	13.51	24	8.25	12	6.00	27	13.37
Negative impact: fatigue	29	7.21	1	1.03	9	6.87	11	9.65	8	13.33	14	7.07	15	7.65	10	9.01	19	6.53	17	8.50	12	5.94
Positive impact: realise what is important (giving perspective/staying positive)	28	6.97	8	8.25	12	9.16	6	5.26	2	3.33	13	6.57	15	7.65	6	5.41	22	7.56	11	5.50	17	8.42

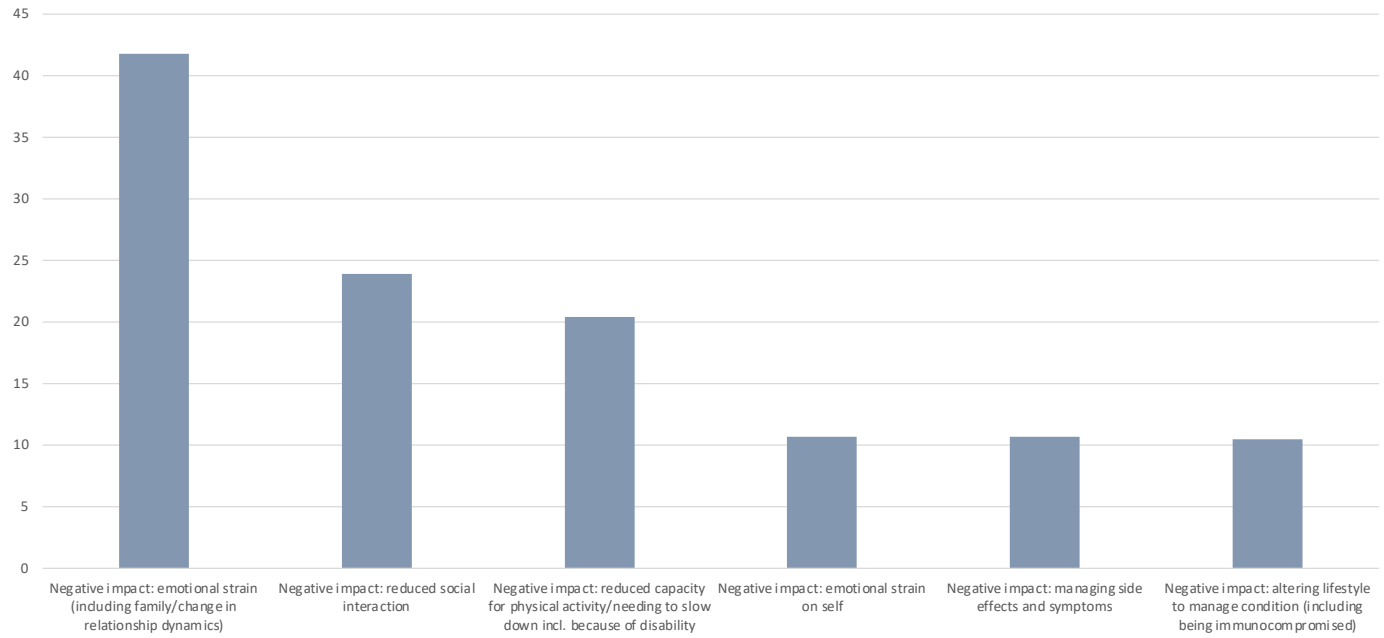


Figure 8.2: Impact on quality of life (Reasons)

Table 8.4: Impact on quality of life (Reasons)– subgroup variations

Impact on quality of life (descriptions)	Reported less frequently	Reported more frequently
Negative impact: emotional strain (including family/change in relationship dynamics)	Diseases of the skin	Diseases of the immune system
Negative impact: reduced social interaction		
Negative impact: reduced capacity for physical activity/needing to slow down incl. because of disability	Other rare condition Family or carer	Diseases of the immune system Aged 65 plus
Negative impact: emotional strain on self		Other rare condition
Negative impact: managing side effects and symptoms	Endocrine, nutritional or metabolic diseases	Developmental anomalies
Negative impact: altering lifestyle to manage condition (including being immunocompromised)	Endocrine, nutritional or metabolic diseases	
Negative impact: unable to travel/adapt significantly in order to travel		
Negative impact: fatigue		Diseases of the immune system
Positive impact: realise what is important (giving perspective/staying positive)		

Impact on mental health

In the structured interview, participants were asked if there had been an impact on their mental health. Most commonly, the descriptions suggested that overall, there was at least some impact on mental health (77.84%), and overall, there was no impact on mental health (5.97%).

Experience described suggests that overall, there was at least some impact on mental health

Yeah, 100% it does it affect because it affects yourself esteem. It makes you stress about the pain you're experiencing, or even just knowing that it's coming is sometimes worse. Stressing about money, taking time off of work, it's a lot of sometimes I think the mental

and emotional roller coaster puts you through sometimes worse than the physical, so that I know it's a lot worse for other people. Thankfully, I'm probably classed more as mild, especially these days. It's not as rampant as what it was, but yeah.
Participant 018_2023AUDSK

I would say it does have an impact on my mental health, especially before the diagnosis. Now that I have the diagnosis, it's it's settled a bit. The, the not knowing was the hardest part and separate to my diagnosis. I do actually see a therapist as well. It's not something I've actually spoken about with her, but this has sort of prompted me. To maybe do that in the future. So yeah.
Participant 027_2023AUDSK

Like, I think I'm a pretty stoic sort of person. But it does get to me every now and then especially if I come across an article and it you know when they're talking about mortality rates and things like that I get really depressed.
Participant 15_2023AUORC

Yes, it definitely has impacted my emotional, mental and physical health. Haven't had a decent night's sleep in seven years. And like, I try to do stuff for my own mental health, but it's very hard. There's a lot of things on my To Do List, and getting to any of my own is really very challenging.
Participant 87_2023AUENM

Experience described suggests that overall, there was no impact on mental health

Not particularly. I do what I do. I'm active. I've got a good brain, I manage. I have a job. And yeah, so yeah. So I'm quite active and we travel a lot.
Participant 003_2023AUDIS

No, no, no. I'm always, I'm very much for the kids and getting treatment and that. So I haven't really thought about myself. But no, I've never had to seek attention for that. Yeah, Okay.
Participant 11_2023AUDPA

For me personally, I just I'm, I'm one of those people. I just do what's needed to be done and you just move on. So yeah, I think, I think we've having the condition she's had. It's been a struggle to get across things to in the medical sense and in the in the in the health side of things. But other than that mental health wise with me really hasn't caused any issues. You know, people have often said to me and I don't know how you do it if it's all you've known. I mean, I gave birth to PATIENT when I was 18. So it is a lot known.
Participant 24_2023AUDPA

Table 8.5: Impact on mental health

Impact on mental health	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=352	%	n=67	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=247	%	n=105	%	n=252	%	n=98	%
At least some impact on mental health	274	77.84	36	53.73	72	88.89	34	75.56	26	83.87	77	81.05	29	93.55	209	84.62	65	61.90	205	81.35	67	68.37
No impact on mental health	21	5.97	3	4.48	1	1.23	6	13.33	1	3.23	9	9.47	1	3.23	14	5.67	7	6.67	13	5.16	8	8.16
Other or mixed experience	25	7.10	1	1.49	7	8.64	5	11.11	1	3.23	9	9.47	2	6.45	19	7.69	6	5.71	20	7.94	5	5.10

Impact on mental health	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=69	%	n=116	%	n=108	%	n=59	%	n=172	%	n=172	%	n=100	%	n=252	%	n=176	%	n=176	%
At least some impact on mental health	274	77.84	41	59.42	99	85.34	88	81.48	46	77.97	132	76.74	138	80.23	77	96.25	197	78.17	138	78.41	136	77.27
No impact on mental health	21	5.97	4	5.80	6	5.17	4	3.70	7	11.86	14	8.14	7	4.07	3	3.75	18	7.14	8	4.55	13	7.39
Other or mixed experience	25	7.10	1	1.45	3	2.59	15	13.89	6	10.17	8	4.65	13	7.56	9	11.25	16	6.35	15	8.52	10	5.68

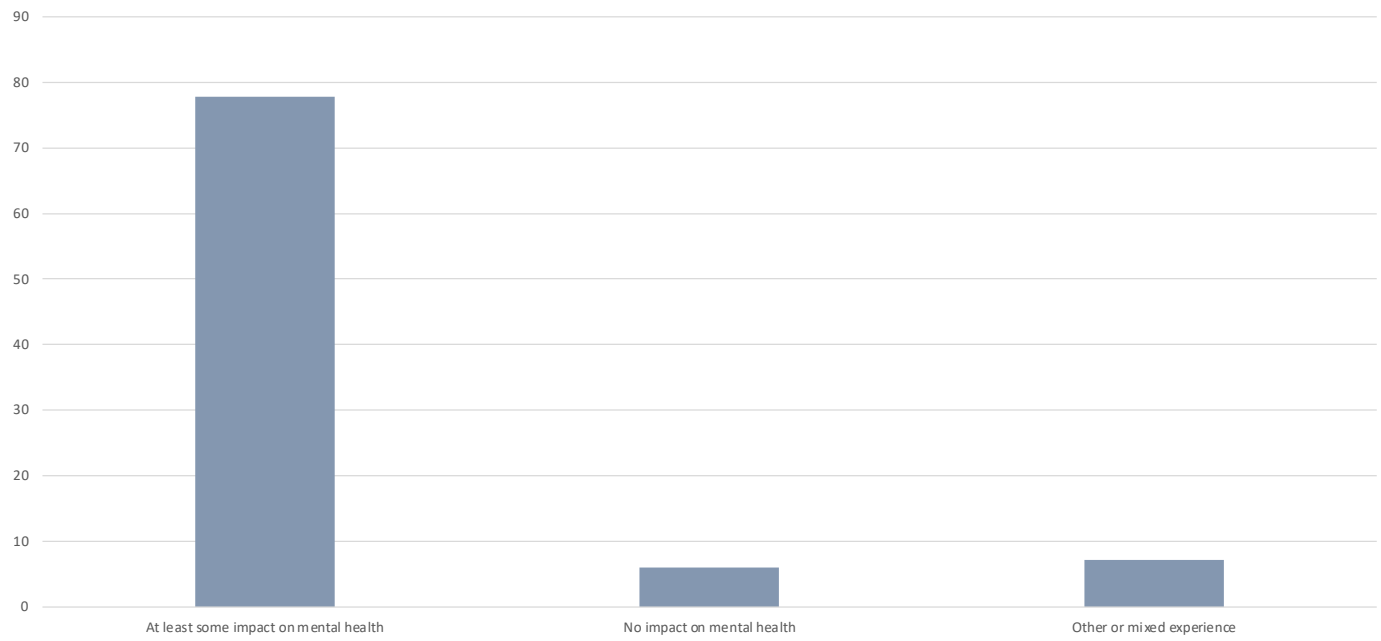


Figure 8.3: Impact on mental health

Table 8.6: Impact on mental health – subgroup variations

Impact on mental health	Reported less frequently	Reported more frequently
At least some impact on mental health	Developmental anomalies Family or carer Aged under 18	Diseases of the immune system Other rare condition Regional or remote

Regular activities to maintain mental health

In the structured interview, participants were asked what they needed to do to maintain their emotional and mental health. The most common response was consulting a mental health professional (24.17%), coping strategies such as remaining social, lifestyle changes and hobbies(22.52%), and mindfulness and/or meditation (16.56 %). Other themes included no activities to maintain mental health (15.89%), the importance of family and friends in maintaining their mental health (14.90%), and the importance of physical exercise (14.90%).

Participant describes consulting a mental health professional to maintain their mental health

Yeah, it definitely has an impact. It sort of makes you feel like. You're a bit less lovable. I don't know. There's just a lot going on, I guess. And yeah, I see a psychologist.
Participant 019_2023AUDSK

Negatively, at least half the time. And I've been seeing a psych for like, like a psychologist for two years. But then yeah, just working on self-care and all that fun stuff. But it's it's definitely a roller coaster of emotions and acceptance, like new acceptances. Like a lot of parents here, you know, this is the new normal, especially their children in palliative care and so on. But it's definitely hard, even if you have like you know, I was explaining my child as a fairly chill, easy going kid that doesn't have any overly complex medical issues. But when I talk about, you know, getting a feeding tube and doing this and doing that, it's so complex to other people but you just get so you what you're going through. But it's still definitely very hard on my mental health. Yeah.
Participant 81_2023AUDIS

Yeah, so it did. It did affect me quite heavily at the start. I was a bit shocked by the diagnosis and it sent me downhill for a while. So I did go and talk to some grief counsellors about it because I guess it was a form of grief that I was going through and that helped me a lot to manage, manage it and manage my thoughts around it. So it's a lot better now. But yeah, at the start it was quite intense. Participant 29_2023AUORC

Yeah, so I see a psychologist once a week, so it's just telehealth. But you know, I have a whole lot of other things going on. So we don't always necessarily talk about my illness, but certainly it has featured many times for the whole session, for the whole hour where I've just talked about the Scleroderma and and its really huge impact on my life.
Participant 010_2023AUDIS

Participant describes coping strategies such as remaining social, lifestyle changes and hobbies to maintain their mental health

Yeah, I do crocheting. I still pretty involved with my grandchildren although COVID put a spoke in the wheel for that for a very long time that we're kind of getting back to that again and just basically having to reset my brain to kind of well that's not going to happen again. So you know, stop looking for something that's not going to happen because you'll just become disappointed. So I I've, I've learned to just say to myself, 'OK, well that's not going to happen'. You can try about it. You can try…So just shut up, get on with it.
Participant 002_2023AUDIS

I'm lucky that we're now in the day of age where I can pick up the phone and I can FaceTime a friend and have a chat with her. I'm a big music lover so I just throw on the music, turn up loud and sing away, and I have my little dance party in my house. I go back to my essential oils and I use my relaxing oils to soothe and calm me. I've also learnt that I have to process the emotions through things.
Participant 023_2023AUDIS

Dystonia can cause anxiety and depression. I'm not depressed, but I am aware. I get anxious. However, I've got some nice books to read and if I find I'm getting a bit hit up right, you can go out and sit in the sun. You can go and weed, you can go and do this and then this afternoon you've got another book to read.
Participant 003_2023AUDNS

Participant describes mindfulness and/or meditation to maintain their mental health

Yeah, my, my mental health has been affected, that's for certain. And my emotions and everything has been affected. Yeah, I, I do certain activity once in a while, you know, to kind of, you know, soften the, the effects in my mental health, you know, let's say activity like yoga and meditation, just to calm the calm the tension down.
Participant 006_2023AUORC

It can affect my mental and emotional health and that can vary from day-to-day. I can have, you know, at the moment I have a flare and that can get me down because it's just kind of like you suddenly you're feeling well and then bang, you know, it's almost like there's no lit up. Like you, you just go for a day and you think you're doing really well and then suddenly you've got, you know, one flare pops up and. Your arms uncomfortable or whatever. So yeah, look, it can, but I think in that for me, I've had to put things in place to help me with my mental health. So that's for me that's talking about my condition. It's practicing mindfulness and mental health and wellbeing type of activities that keep me, keep me focused on the positive rather than allowing, you know, the focus to become the HS.
Participant 001_2023AUDSK

At the moment I'm not doing…I have to do a big work on myself to be able to look after myself, to be able to deal with all issues. It took me time, but I still have anxiety some days. Not every day like before, but I think it's something personal that we have to deal with. It's only me who can stop that anxiety from coming. I have to change my way of thinking to think more positively and to get in contact with people who are not helping me. That's how I was able to deal with this.
Participant 020_2023AUDIS

Participant describes no activities to maintain mental health to maintain their mental health

I mean, it is right now. It's certainly, yeah, we're having a flare up. It could be quite frustrating, painful and upsetting, but I don't. I don't seek out any.
Participant 011_2023AUDSK

No, because there isn't time. You know, we can always say yes, we will. I'll get to that. I'll get to that. But honestly and everything, yeah, something else. There's always something.
Participant 16_2023AUORC

I don't know. I don't…my daughter's studying SUBJECT at the moment and she's actually hammering me on that at the moment. She believes I should go. She said even though I'm strong and stubborn, that maybe I should talk to someone because she said the traumas I've been through, that maybe I should talk about it. I haven't actually done anything. I'm on a care plan

where I get 5 with my GP, where I get five allied health a year, which I really need about 50, but I get five. I spoke to them about that and they said, well, if I don't get my physiotherapist or podiatry or dietician and all the other allied health, that I could get some mental health support. I've got to weigh up what goes.
Participant 014_2023AUDIS

Participant describes the importance of family and friends in maintaining their mental health to maintain their mental health

Yes, it did affect my mental and emotional health prior to treatment. But you know as I said during treatment and prior to treatment and continuing after treatment I have a spiritual understanding that I live in the moment. I am very connected with my family and with my close friends.
Participant 010_2023AUORC

Sorry, I'm crying, but yeah, it affects my mental health a lot I would say. You know, I have ever…an episode at least twice a month where I break down about having the disease, because I just think, why me? Like why do I have this? And it's just it's ugly and I compare myself to other people all the time who, you know, don't have it. I like there's not much I can do for myself in terms of having disease because it's so hard to pull myself out of this like mental thought that it's ugly and it's something I should be ashamed of and it's just kind of, I need to let myself cry about it and then I move on because it's not going anywhere. So I just have to, you know, get over it. I'd say my partner is definitely someone who helps a lot with my mental health. He'll just remind me that, like, I'm still a great person with or without the disease. Yeah.
Participant 010_2023AUDSK

Yeah, I just said we've got really good support. So in the beginning when it was young and it would kind of just, I guess, build up and be a bit too much. We had ample, ample people like brothers, sisters, grandparents that we could just kind of say, can you just watch them for the night or can you come up? I'm just gonna go for a massage. Like, you know what I mean? Like, so we've been really lucky in the fact that at any stage we've had like full wrap around support for whatever we need.
Participant 25_2023AUORC

Participant describes the importance of physical exercise to maintain their mental health

Well, I can't walk very far because of my back and my legs and my arthritis, but try and do little walks. We try to get on the beach and just sit on the beach and have a walk around we…Yeah…visit friends or go out for dinner. Yeah. Doesn't really emotional how…that's more me. It's just more about, you know, finding a cure, which doesn't exist, you know, that sort of thing. Most things I can fix or have operations for. But this one is like, yeah, no, there's nothing.
Participant 024_2023AUDSK

I play a lot of sport. I do athletics, I do Pilates, I've always been active and I think that has helped a lot.
Participant 93_2023AUENM

For my, I mean my emotional…I was trying to say emotional and mental at the same time. Emotional health, I guess. I know that I need a good balance between alone time and making sure that I meet up with, you know, friends every now and then just for some social time. My mental health I really have focused on, so focusing on my physical health has also helped my mental health. Because I have, you know, I go for my walks and those sorts of things to help with my mental health. I, I don't handle being stuck at home very well. So yeah, like, it's the diagnosis I think has affected me more than I would care to admit. It's become like another thing. So yeah, I've just been focusing on if I eat right and exercise and things like that and help my mental health, that's gonna help me all over.
Participant 29_2023AUDPA

Table 8.7: Regular activities to maintain mental health

Regular activities to maintain mental health	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=302	%	n=67	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=204	%	n=98	%	n=214	%	n=86	%
Consulting a mental health professional	73	24.17	13	19.40	23	28.40	11	24.44	5	15.63	12	12.63	9	28.13	49	24.02	24	24.49	55	25.70	18	20.93
Coping strategies such as remaining social, lifestyle changes and hobbies	68	22.52	10	14.93	28	34.57	8	17.78	4	12.50	13	13.68	5	15.63	53	25.98	15	15.31	49	22.90	18	20.93
Mindfulness and/or meditation incl. rest	50	16.56	5	7.46	15	18.52	10	22.22	10	31.25	2	2.11	8	25.00	43	21.08	7	7.14	42	19.63	7	8.14
No activities to maintain mental health	48	15.89	5	7.46	9	11.11	11	24.44	7	21.88	9	9.47	7	21.88	35	17.16	13	13.27	29	13.55	19	22.09
Importance of family and friends in maintaining their mental health	45	14.90	3	4.48	18	22.22	6	13.33	4	12.50	9	9.47	5	15.63	36	17.65	9	9.18	37	17.29	8	9.30
Importance of physical exercise	45	14.90	4	5.97	7	8.64	10	22.22	5	15.63	12	12.63	7	21.88	32	15.69	13	13.27	31	14.49	14	16.28

Regular activities to maintain mental health	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=302	%	n=66	%	n=103	%	n=84	%	n=49	%	n=148	%	n=172	%	n=84	%	n=218	%	n=154	%	n=148	%
Consulting a mental health professional	73	24.17	17	25.76	28	27.18	20	23.81	8	16.33	28	18.92	44	25.58	18	21.43	55	25.23	32	20.78	41	27.70
Coping strategies such as remaining social, lifestyle changes and hobbies	68	22.52	8	12.12	16	15.53	24	28.57	20	40.82	31	20.95	34	19.77	17	20.24	51	23.39	36	23.38	32	21.62
Mindfulness and/or meditation incl. rest	50	16.56	4	6.06	25	24.27	14	16.67	7	14.29	24	16.22	25	14.53	16	19.05	34	15.60	28	18.18	22	14.86
No activities to maintain mental health	48	15.89	9	13.64	18	17.48	12	14.29	9	18.37	32	21.62	16	9.30	12	14.29	36	16.51	24	15.58	24	16.22
Importance of family and friends in maintaining their mental health	45	14.90	3	4.55	14	13.59	14	16.67	14	28.57	20	13.51	23	13.37	16	19.05	29	13.30	24	15.58	21	14.19
Importance of physical exercise	45	14.90	7	10.61	15	14.56	13	15.48	10	20.41	17	11.49	25	14.53	9	10.71	36	16.51	22	14.29	23	15.54

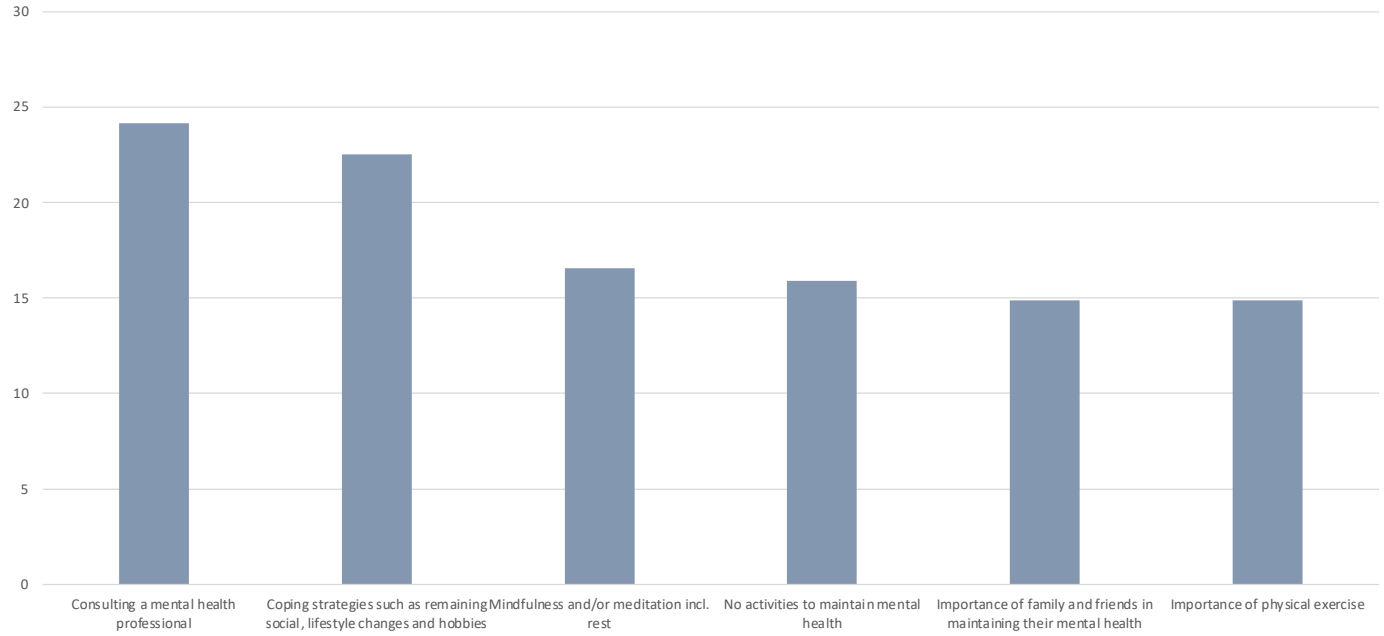


Figure 8.4: Regular activities to maintain mental health

Table 8.8: Regular activities to maintain mental health – subgroup variations

Regular activities to maintain mental health	Reported less frequently	Reported more frequently
Consulting a mental health professional	Endocrine, nutritional or metabolic diseases Diseases of the skin Aged under 18	Diseases of the immune system Aged 65 plus
Coping strategies such as remaining social, lifestyle changes and hobbies	Endocrine, nutritional or metabolic diseases Aged under 18	Diseases of the skin
Mindfulness and/or meditation incl. rest	Developmental anomalies Aged under 18	Aged 65 plus
No activities to maintain mental health		
Importance of family and friends in maintaining their mental health		
Importance of physical exercise		Diseases of the immune system

Regular activities to maintain health

In the structured interview, participants were asked what were some of the things they needed to do everyday to maintain their health? The most common activities for general health were self care e.g. more rest, accepting help, pacing (34.38%), complying with treatment/management (29.83%), and doing physical exercise/physically active (22.73 %). Other themes included understanding their limitations (19.89%), maintaining a healthy diet (14.20%), being organised and planning ahead (11.93%), and maintaining a normal routine (8.24%).

Participant describes the importance of self care e.g. more rest, accepting help, pacing in maintaining their general health

Sometimes there's nothing I can do to control it. There's no amount of pain medication is going to stop the nerve pain from happening when the infections are that bad. No amount of cleaning, irrigating, washing the wounds is going to get rid of the smell there's…It, it makes me feel very powerless, actually. If it's a mild flare I just have to push through. I just have to deal with the pain and then hope my partner comes home on time from work so I can then go and rest and physically sleep from the physical and mental exhaustion of being in constant pain.
Participant 012_2023AUDSK

Well, I if I, with the fibromyalgia, I have to sometimes I hit a wall of tiredness and I just have to have a sleep. So I do if I'm get, if I I'm not like that all the time, but when I do get like that, I do, don't get up and go and have a lay down for an hour or so and I'll get up and I feel better. So that's what I do for myself. If I'm not, you know, unwell, I'll have a, I will rest and I, my body tells me I need to go and lay down.
Participant 88_2023AUENM

I have to rest when I have to rest, simple as that. Put my feet up and make sure that I take care of that because rest is so important, and sitting aside and just taking time out. Just re-evaluating life and just go for a walk or do whatever you can to get your mind straight.
Participant 001_2023AUDIS

Participant describes the importance of complying with treatment/management in maintaining their general health

No, no. Well, that's, that's it. I mean I take, I'm probably taking about 10 pills a day now, so that's not too bad. But you know, apart from that, life just goes on. And I mean pills in the morning, pills at night, that's it. It's no great drama and it's, it's quick and easy to do.
Participant 007_2023AUORC

I mean, aside from giving him the medication, we have to keep up with the physio on his chest every day. That'll allow him to be able to become more active as he gets a bit older.
Participant 29_2023AUORC

For the moment, it's really just around compliance with existing treatments that that creates, you know, conflict, if I'm honest, you know, trying to get her to take her medication, to wear her retainer, to do her exercises, to do her homework, all those sorts of things. I think it's probably the main issue. Yeah, day-to-day it's just around compliance with things she's supposed to be doing to maintain her health, but also what she's supposed to be doing around her schooling.
Participant 35_2023AUDPA

Participant describes the importance of doing physical exercise/physically active in maintaining their general health

So I have to do exercise to keep my lungs healthy. I have to have a decent diet and you know, drink a lot of water and extra water and have medications every day. And then kind of manage that to how my wife is at that time health wise. So whether I need to increase any medications or add extra ones on or if I need to go into hospital. And yeah, medications in the other medications like doing physio, which takes about half an hour, 45 minutes in the morning and then in the afternoon as well, just to have a clear chest and be able to breathe normally.
Participant 13_2023AUORC

Well, I think it's just making sure you're getting up and moving every day. Some days you can do more than others, but I certainly can't just sit or lay. Even if you're feeling fatigued, I've got to move and stretch because then my joints just lock up, if you like. I think it's important that…yes, move every day, do some…As I say, I'll go for, I call it a stroll, it's not really a walk anymore. I get out and about and get some fresh air.
Participant 017_2023AUDIS

Well, like I said, she does look after herself. The onlyactivities I'm involved with is her financial

business. Everything else she seems to be, well, not really like she had to with this new job she's got. She does a lot of walking. And she needed new shoes and she didn't feel confident getting the right shoes for the job. So I had to take her shoe shopping. So, you know, things like that because, you know, she might go in and buy those because they look pretty, but they're not practical for what she needs, so to speak. So, you know, I do small things like that, but generally, no, she's pretty self-sufficient in in the choices she makes are quite reasonable.
Participant 16_2023AUDPA

Participant describes the importance of understanding their limitations in maintaining their general health

Pain relief. As I said, I had to change jobs to give up my career and go to a new career, and on any one day it could be dramatic changes to to anything. I'm very fortunate in my new job that I can choose to work from home if I need to because there are some days that putting a pair of pants on is not an option. So I'm very fortunate in that regard. But yeah, sometimes, sometimes every aspect of my life has to be altered. But on an everyday basis, the types of things that have changed my life is obviously changing my career and and pain, pain, pain and more pain.
Participant 005_2023AUDSK

I basically don't do anything in the evenings after work because I well, I need occasionally because otherwise I get too worn out. I try and plan activities so that I know that if I've got a couple of busy weeks then I'll make sure I have a weekend with nothing on to recover. I…when I wasn't able to walk much, I was having to manage that by arranging my life around not being able to walk. Very much. I don't. If I'm going to the I go to the theater regularly as friends, but they're kind enough now to go to matinee so I don't have to be out late at night. So it's more things like that to manage fatigue and tiredness, because I find if I'm if I'm not too tired, then I can cope with everything much better.
Participant 009_2023AUDIS

Well, since the ablation, there really isn't anything. I don't push it too hard. I sort of just listen to my body and if I feel like my heart's pounding too hard, I do stop. I don't push it as much as what I may have pushed previously. But if you have that fear that ohh God, if it goes too high, is it going to get stuck again? Yeah.
Participant 32_2023AUORC

Participant describes the importance of maintaining a healthy diet in maintaining their general health

Yeah, so I think just like making like reminding him because he's only….now I like to have his medication every day. So making sure that happens, organizing like the his day-to-day food so that he's got the right food to eat and that's pretty normal. But make like if he's got a birthday party or he has to go away on camp or you know, like making sure that or if he's staying at, you know, he wants to stay at a friend's house. It's like it's organizing you know him for you to take with him or organizing with the other adults that they'll have food that he can have.
Participant 79_2023AUDIS

I suppose, join the Pilates so that's keeping my body limber and active. Again, I got the NDIS funding so I managed to get a cleaner in to help in that respect. That's one thing that, I suppose, I do take care of. It's not me physically doing it, but I'm healthy in that respect. I think I'm just taking my drugs and trying to stay as healthy as possible, eat the best food I can, all that sort of stuff.
Participant 018_2023AUDIS

I need to plan things. Today, I'm talking with you and that's my day's events. Not overburden the whole thing. I was drinking a lot of alcohol earlier in my diagnosis and life and stuff and I've cut that right back. They're the things trying to eat well. I'm looking forward to the physio planning some more activities to get some fitness back because I have to work out how I can reinvent that from how I was physically active before. I'm looking forward to getting some ideas around that with him that'll be really positive. They're the sorts of thing and just being kind to yourself and being okay that if you need to just stop, that's all right. You're not being weak or pathetic or lazy or any of those sorts of things.
Participant 026_2023AUDIS

Participant describes the importance of being organised and planning ahead in maintaining their general health

So if I've got a break out. I need to make sure that I've got bandaging to be able to wear, I need to have… funnily enough, I haven't had any really strong pain relief for a very long for over a year now, and I'm quite happy about that. I need to be able to maybe cancel things in short notice if, if that's the case, you know. I need, I feel like I need certainly access to an endless supply of chlorhexidine and I have the biggest Band-

Aid and bandage collection in the world. So yeah, yeah, so I guess, I guess. Physically, to be able to, you know, wear undergarments, I need to be able to, you know, have enough padding to protect, you know, and I guess also obviously I need access to my medication and I need access, yeah.
Participant 017_2023AUDSK

Yeah. So yeah, on a day-to-day base, I guess is actually sit down with him every morning, get his nebulizers ready, get all his meds ready. We've got to actually make the salt tablets, sterilize all the equipment. Yeah, mainly just monitoring, making sure that the tablets are taken, you know, making sure everything's ordered in advance because some of the medications just take a little while to get in and yeah, just actually administering the the treatments essentially.
Participant 20_2023AUORC

Yes, that was just about, I guess, being organized to make sure she has everything she needs in terms of, you know, it might be exchange of clothes or support in terms of special shoes or things like that. And also it's been organized to attend the various appointments and pay the various bills and then also, you know, make calls and emails as needed to. Trace up results or get other questions answered.
Participant 22_2023AUORC

Participant describes the importance of maintaining a normal routine in maintaining their general health

Yeah, so that's very strict to keep routine, make sure her environment is safe. Very important that she doesn't get overwhelmed because then her behaviour gets worse. Yeah, she's just a lot of safety cleaning she engaged and things like fecal smearing, so…obviously having things, you know, clean up fast and make sure she doesn't ingest anything and get sick.
Participant 94_2023AUENM

I have a bit of a routine so I know my energy levels are best in the morning when I get up. It's keeping a routine and trying to keep mentally healthy and sticking to that routine so that if I'm exhausted, I know there'll be time in the afternoon to rest. Exercising to keep the joints mobile and also recognizing when your body's telling you I'm…I can't do this, you stop, you know, not keep pushing through.
Participant 007_2023AUDIS

So things like sticking to a routine and stuff like that, yeah, yeah, yeah. So trying to stick to a routine to make sure things run smoothly, very much having to warn him and show him photos of if we are doing something different to make sure that he's kind of semi familiar because thrusting something different upon him doesn't go very well. So just like you know, letting him know what's happening in advance day-to-day.
Participant 23_2023AUDPA

Participant describes the importance of socialising with friends and/or family in maintaining their general health

Some of the things I do, I think pacing myself and just both physically and mentally. So not looking too far ahead and just saying, all right, so today I need to deal with this by the end of the week. I need to do this, so not getting too far ahead of myself. And just, yeah, physically, just getting through the day. If it's been a hard day physically, I'm just like, okay, one thing after the other. And I just look forward to getting into bed at the end of the day and resting as much as possible. And yeah, so pacing myself and mentally, I think, I think my work actually helps me so as far as I'm physically able to work. And then it helps me mentally to just distract me and give me something to focus on. I'm pretty good sometimes. I'll take the dog out. Now that my mobility is a bit better. I'll take the dog out. I'll go for a walk. That's really good again because I'm a bit more mobile, will do things as a family, so….go out and do fun stuff on the weekend. So that's that's good.
Participant 022_2023AUDIS

Exercise. In fact, everything I just said exercise, social engagement, good diet, sleep. Exercise is huge, you know. Doing that something vigorous every day, yes.
Participant 19_2023AUDPA

I love my job. I've been in my job for 36 years with the same company. Very office-based job, so I'm very lucky that we have a cattle property, and on our weekends, we tend to get away up there. I walk every day, I swim most days. Sometimes when that pain did come back, you just have to be a bit kind to yourself and realize that you can't do everything when the pain is that bad. Like I said, I'm very lucky in the support that I have from friends and family. It's probably hard in my family to feel too sorry for yourself because they just bring you back to earth and basically we just…that's good too.
Participant 006_2023AUDNS

Participant describes the importance of using complementary therapies in maintaining their general health

I'll have to wear my orthotics and I have to do some fairly specific exercises because I have a joint go out. I have to get it to click back in or yeah, and then. I'll try and have a massage, a remedial massage as regularly as possible.
Participant 002_2023AUDPA

I just have to keep moving. If you don't keep moving then my lungs get tight and I can't breathe properly. My joints all get too sore and I can't move. So yeah, just sort of stay active or just move as much as possible. There's nothing much else apart from taking medication that I can do. Well, I give you pretty much regular massage on the shoulder, the neck, yeah, that's…
Participant 011_2023AUDIS

PARTICIPANT: When I feel like the anxiety is coming, I just stand up and open the door and look for some fresh air and do some stretching, or keep myself busy, although it is very hard for me to do cleaning. If I'm doing some cleaning or folding clothes or things like that, I just have to keep myself busy so I don't think about it, I don't overthink
INTERVIEWER: Do you still have a massage?
PARTICIPANT: Yes, I do.
INTERVIEWER: Yes. Do you find that's helping you to get through?
PARTICIPANT: It's helping me, yes, and also I have someone to talk to for one hour twice a week.
Participant 020_2023AUDIS

Participant describes the importance of a good hygiene routine to maintain their health

Yeah, look, all I do at the moment is, yeah, twice a day in the shower I will wash the sites or the potential sites with chlorhexidine, which is like a surgical scrub thing, but really. That's my only attempt at a preventative sort of measure, but I don't have any symptoms so I'm not needing to manage anything more than that at this point.
Participant 007_2023AUDSK

Definitely keeping the area clean. I have to take wet wipes with me wherever I go. So that if I get sweaty or is…you know, one of the scars or bumps kind of pairs open, I can clean it up straight away. There's not, you know, like I can't wear proper bras because it affects the area. So I'm always wearing like stretchy bras that don't have any wires or any like lace or scratchy things on them so that it doesn't aggravate. The area other than that, just keeping clean, like I have to shower twice a day, which is, you know, normal, but I just have to keep it very clean and I'm always very conscious of the area. Just yeah.
Participant 010_2023AUDSK

Good hygiene, as in making sure the affected areas are well cleaned and protected. I take vitamins to help counteract. I take medication when it's prescribed. Just looking after myself and getting on with life.
Participant 013_2023AUDSK

Participant describes the importance of mindfulness and/or meditation in maintaining their general health

So I think I touched on it. You gotta…we've learned that we have to take time out as parents and carers and whether that's together or separate, my husband and I, he goes away and has time with friends or mates. I do the same. And there's occasional time where we go on holidays by ourselves together, and that makes it easier when we do like have days or holidays or look after her every day and we can manage her and her and her mind and emotions and so, so that is using mindfulness, you know, also understanding and realizing when you're starting to get in a bad habit, let's say, you know, when things start to get tough, you drink too much or you eat too much, learning the signs and signals. So we now know what the triggers are because we're we have gone out and sought that help to to help us identify those those things and identify when it is becoming too much and asking for help from the professionals. Like we'll go to the GP and go we really need something here, can you help us? Because the GP's pretty much is your first point of call for most things.
Participant 25_2023AUDPA

I have a nice dog. I have a lovely little dog and I've trained her and now she can picks up sometimes when I'm having a spasm and then that's really that I try and stay put. If I do some meditation, I do some breathing. I do all my home programs from the physio and the speech therapist, I try and get out in the sunshine, and I go for walks when I can. I just work hard at being the healthiest and best I can. It doesn't have the right self.
Participant 001_2023AUDNS

Participant describes no activities to maintain their general health

No, no, I'm not doing anything because it's not active, so I don't even know what to do. I just continued my everyday life.
Participant 001_2023AUORC

I don't need to control anything yet.
Participant 005_2023AUORC

Nothing at the moment. It's just pretty normal actually, he just fits in. There's no medical or there is medical issues but then there's not a lot there at the moment.
Participant 51_2023AUDPA

Participant describes the importance of support from carer or support from parent to maintain health

Well, I'm here. Arms and legs and sensory interpretation and educational advocate and community rouser. And like all the things, it's like wiping your bum, to brushing your teeth, to rocking your sleep.
Participant 87_2023AUENM

I guess one of the like, you know, I've got a…I don't really know to be honest. Obviously I have to get her everywhere she needs to go and, and a sister getting ready to to be at school on time and you know, I'd be there to pick her up and all of those sorts of things. You know she doesn't have as much independence as her siblings do, just because she doesn't quite understand the risk factors and, and have the comprehension to be able to to do certain things that they do. So it's pretty much having to make sure that I'm always available if and when she needs me.
Participant 27_2023AUDPA

So she pretty much needs somebody with her every minute of the day so she can't sort of dress herself, toilet herself, change her feeding, pump bottles, extensions, provide medication, meeting she needs sort of assistance with. And then sort of obviously she's sort of globally delayed. So all the things at school, she needs to teach her aid to be with her as a lot of help with reading and writing and socially, she doesn't naturally understand that she needs help with making friends and things like that. And so she's pretty much supported all day, every day. Okay.
Participant 32_2023AUDPA

Table 8.9: Regular activities to maintain health

Regular activities to maintain general health	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=352	%	n=67	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=247	%	n=105	%	n=252	%	n=98	%
Self care e.g. more rest, accepting help, pacing	121	34.38	13	19.40	52	64.20	13	28.89	6	19.35	32	33.68	5	16.13	103	41.70	18	17.14	97	38.49	23	23.47
Complying with treatment/management	105	29.83	29	43.28	18	22.22	10	22.22	11	35.48	23	24.21	14	45.16	65	26.32	40	38.10	68	26.98	36	36.73
Physical exercise/physically active	80	22.73	7	10.45	22	27.16	13	28.89	3	9.68	28	29.47	7	22.58	63	25.51	17	16.19	56	22.22	24	24.49
Understanding their limitations	70	19.89	7	10.45	15	18.52	16	35.56	4	12.90	25	26.32	3	9.68	55	22.27	15	14.29	52	20.63	17	17.35
Being organised and planning ahead	46	13.07	22	32.84	7	8.64	2	4.44	3	9.68	4	4.21	8	25.81	18	7.29	28	26.67	25	9.92	20	20.41
Maintaining a healthy diet	42	11.93	5	7.46	12	14.81	5	11.11	4	12.90	10	10.53	6	19.35	32	12.96	10	9.52	28	11.11	14	14.29
Maintaining a normal routine	29	8.24	18	26.87	5	6.17	1	2.22	2	6.45	1	1.05	2	6.45	12	4.86	17	16.19	16	6.35	13	13.27

Regular activities to maintain general health	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=69	%	n=116	%	n=108	%	n=59	%	n=172	%	n=172	%	n=100	%	n=252	%	n=176	%	n=176	%
Self care e.g. more rest, accepting help, pacing	121	34.38	12	17.39	39	33.62	50	46.30	20	33.90	60	34.88	61	35.47	42	52.50	79	31.35	63	35.80	58	32.95
Complying with treatment/management	105	29.83	28	40.58	42	36.21	22	20.37	13	22.03	53	30.81	51	29.65	28	35.00	77	30.56	49	27.84	56	31.82
Physical exercise/physically active	80	22.73	11	15.94	24	20.69	30	27.78	15	25.42	37	21.51	41	23.84	22	27.50	58	23.02	41	23.30	39	22.16
Understanding their limitations	70	19.89	11	15.94	18	15.52	20	18.52	21	35.59	30	17.44	38	22.09	17	21.25	53	21.03	30	17.05	40	22.73
Being organised and planning ahead	46	13.07	21	30.43	13	11.21	8	7.41	4	6.78	23	13.37	23	13.37	8	10.00	38	15.08	19	10.80	27	15.34
Maintaining a healthy diet	42	11.93	8	11.59	11	9.48	12	11.11	11	18.64	20	11.63	21	12.21	11	13.75	31	12.30	18	10.23	24	13.64
Maintaining a normal routine	29	8.24	14	20.29	7	6.03	6	5.56	2	3.39	16	9.30	13	7.56	7	8.75	22	8.73	12	6.82	17	9.66

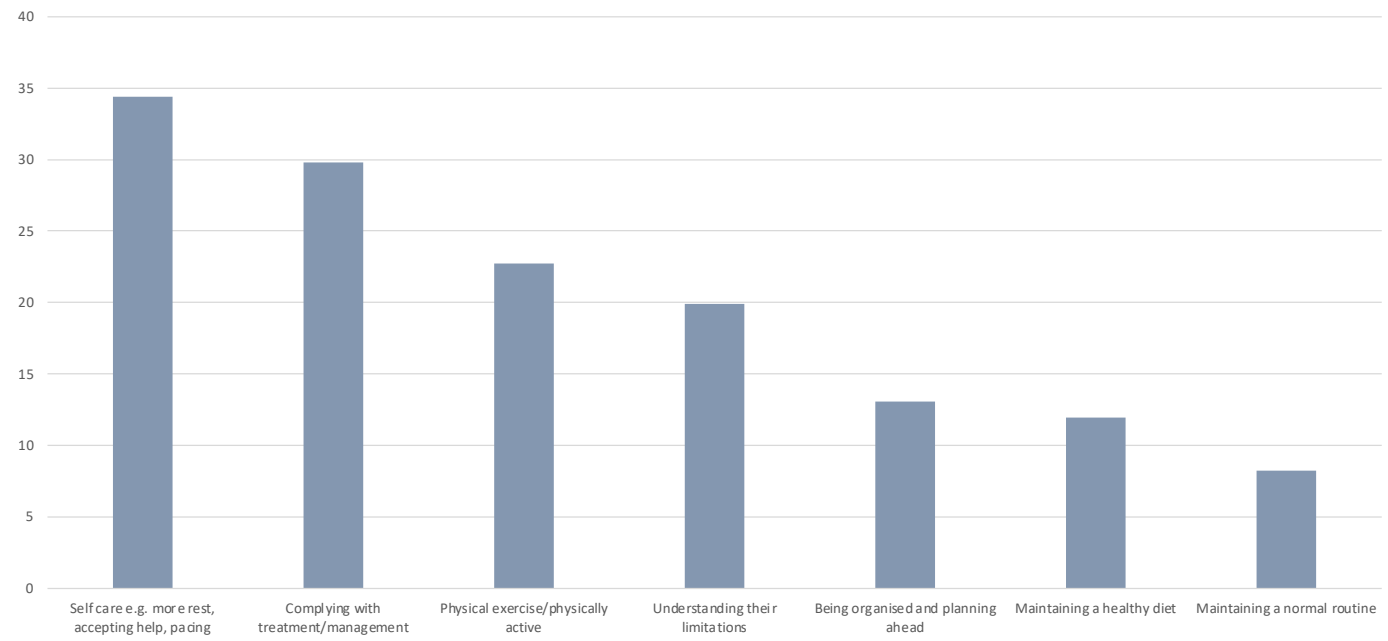


Figure 8.5: Regular activities to maintain health

Table 8.10: Regular activities to maintain health – subgroup variations

Regular activities to maintain general health	Reported less frequently	Reported more frequently
Self care e.g. more rest, accepting help, pacing	Developmental anomalies Diseases of the skin Other rare condition Family or carer Male Aged under 18	Diseases of the immune system Aged 45 to 64 Regional or remote
Complying with treatment/management		Developmental anomalies Other rare condition Aged under 18
Physical exercise/physically active	Developmental anomalies Diseases of the skin	
Understanding their limitations	Other rare condition	Diseases of the nervous system Aged 65 plus
Being organised and planning ahead		Developmental anomalies Other rare condition Family or carer Aged under 18
Maintaining a healthy diet		
Maintaining a normal routine		Developmental anomalies Aged under 18

Experience of vulnerability

In the structured interview, participants were asked if there had been times that they felt vulnerable. The most common responses were that they felt vulnerable when having sensitive discussion (diagnosis, treatment decision) (16.67%), because of interactions with the medical team(14.44%), and experiencing side effects from treatment or symptoms from condition (9.44 %). Other themes included thinking about disease course/incurable condition (8.33%), during or after treatments (6.67%), and when feeling sick/unwell (5.56%).

As a follow up question, participants described ways that they managed feelings of vulnerability. The most common ways to manage vulnerability were using self-help methods (resilience, acceptance, staying positive) (7.78%), and support from nurse or treatment team (3.89%). Other themes included getting support from family and friends (3.33%), and support from mental health professionals (2.22%).

Participant describes feeling vulnerable when having sensitive discussion (diagnosis, treatment decision)

When I was told that I had to, that I really only had Humira as my only option, I did feel very vulnerable then, yeah.
Participant 017_2023AUDSK

Anytime I kinda have to ask for help because I have to start from the beginning. And yeah, I've really gotta you wait for the 'no' and the antibiotics and the just the standard stuff that you know isn't gonna do anything, but you have to do it so they can tick off their list that they've tried that.
Participant 018_2023AUDSK

Yeah, lots of times. I think for the first 12 months when I was getting used to it, trying to figure out what it meant, trying to figure out how it was going to impact my future. And then when I went to see the neurologist and it seemed like every time I went to see him, he had another diagnosis for me. He was getting more, building up a extra long list of autoimmune conditions because at one stage he thought I had my as well. So I was getting all these extra, you know, diagnosis and. So those were some of the points and then when I started talking stem cell therapy and things that sounded really major and scary and whatever. So it was those kind of things when when I was a trigger point and when I was just getting used to the whole thing.
Participant 009_2023AUDIS

Probably only at first with the first rheumatologist that I had…He didn't fully listen, just gave me the bad news, and just sent me on my way. [chuckles]
Participant 018_2023AUDIS

Participant describes feeling vulnerable because of interactions with the medical team

The most vulnerable was that time I described being naked and being told that I need to quit smoking and lose weight and take Humira and then I'll be fine. I'm not sure they realise that. I left it on the opposite side of the city and it took me an hour and a half on public transport to get there. 30 minute walk. That's a lot of effort just for doctors and nurses to point their finger at you while you're naked and tell you you're the problem... That's why I've not seen a dermatologist since.
Participant 008_2023AUDSK

Really. They treated me like I was infectious in hospital, so no one knew the their treatment of my pain level. They made me feel like I was just a heroin addict who who's come in to get some end down. So I cried so hard and then you know I followed that up with that visit, and it was just, it was a joke. I'd never felt more, I'd never felt more unseen. Talked at like I was not a person of any circumstance. I wasn't even poorly dressed, I'd have to say if you thought I was anything other than a professional at that point. And the way he spoke to me in the presence of others, the nurse at the end of it, because he was just taking so long showing off in front of his registrar, this nurse came in and literally looked at me and said, you've taken enough of his time. And I remember laughing so hard because I was literally sitting there just being poked and prodded that no one was talking to me. You know, you regurgitated the lose weight, quit smoking. But wash yourself. And I remember just going 'You have no idea what I do' And I could be just, you know, do you want my skin to be red before you'd be satisfied that the amount of bleach I'm scrubbing into myself daily and all the while still feeling extremely dirty? It's really horrific. I've quit smoking for years and never saw a difference. I was not even overweight at that time. Like, there's no rhyme or reason at that point that I could say these are contributing factors.
Participant 015_2023AUDSK

Right back at the start. Definitely. Right back at the start. Even when I was first diagnosed, I just felt like I wasn't heard and I wasn't understood. And every time I mean my first, the gastroenterologist, he always said if you've got any questions, if you've got any concerns, just ring up, we'll help you out. But I would ring up and I would get met with like this block of, you know, the doctor has just told us to reassure you that, you know, when you're anxious, your symptoms are worse and you've just got to breathe through it, find some meditations and calming apps and, you know, work on your anxiety and your symptoms will go away. And it was that real dismissive like. That blocker, like there was no access because I knew what I was feeling. And I know the difference between anxiety and like an actual physical problem. And I know that anxiety can lead to things like having difficulty swallowing and whatever else. But the reality is you can still swallow when you're anxious. It will still go down. When you have an airway flare up. Nothing that you do, no amount of meditation is going to make your esophagus open up.
Participant 78_2023AUDIS

Participant describes feeling vulnerable experiencing side effects from treatment or symptoms from condition

Yeah, so I the vulnerability that I have, the disease come in my social and intimate life. So as I said, I have the disease in my breast area, which is very intimate area, so I find it hard to be intimate with my partner. Sometimes if I'm having a flare up, I don't want to get undressed. I don't want him to see me even though I know he doesn't care and he still loves me. But it's still hard for me to feel beautiful and wanted when I'm dealing with this. As well as just going out and going to the beach. Like I can't wear a bikini because then the disease is just kind'of on display. And I, you know, there was a time where I said, you know, like through it, like I'm going to wear a bikini. And then, you know,

I saw people looking at it and asking questions like what's on your chest? And yeah, it just sucks because I don't want people to point it out or look at it or anything like that and it just makes me feel embarrassed.
Participant 010_2023AUDSK

Oh yes. When I'm in pain, I think. When I'm in a lot of pain and I'm brought to tears by it, I feel quite vulnerable.
Participant 013_2023AUDIS

Yes, when I was in hospital the second time when it got really, really bad, it was very much so that no one understood what was going on, no one knew, and I was having spasm after spasm, seizure as well. It's almost like they didn't really believe me, which has been something my who'e life, but they were still willing to look after me, but the doctors hadn't passed on to the nurses, or whatever had happened, I don't know, but there was a communication miss and I ended up falling out of bed having a seizure and hitting my head on the floor, so they weren't looking after me properly, so that's definitely a vulnerable time I had.
Participant 004_2023AUDNS

Participant describes feeling vulnerable thinking about disease course/incurable condition

I probably think the most vulnerable I think felt is when I was first diagnosed, I guess there was an element of fear of the unknown and what was going on and what did this look like and what did this mean for me moving forward? So that would probably, yeah.
Participant 001_2023AUDSK

Yes, there has. Particularly when the condition was just getting out of control and you turn to a specialist for help and their answer is there's nothing I can do…and these are that, that they don't offer alternative suggestions. For instance, hang on, I'm going to call, I was having a chronic abscess that just would not heal. And the surgeon I saw just said there's nothing he can do anymore. And I but he didn't offer alternative solutions like go and see this person or what not. And that I still vividly remember being very vulnerable and alone in the world, thinking no one can help.
Participant 013_2023AUDSK

In the sense that I've been going and seeing a particular specialist and blah and blah and blah, and it feels like we're at the end of the line. There's no further that we can go because they're mandible jaw people, the fact that it's partly a muscle problem, they don't want to know about that. That's why I'm feeling a glimmer of hope with NAME's new guy, because he has actually got a plan, how much of it's muscular and how much of it is joint.
Participant 007_2023AUDNS

Participant describes feeling vulnerable during/after treatments

Yeah, I so, I had a fairly major surgery…and I went back for what I thought was my final checkup with the breast surgeon and at which time he decided to continue the surgery in the chair. So I actually witnessed the whole thing take place in front of my face and I was, I don't know if you've ever seen if you get cut open, but it's not a very nice thing to look at. It kind of all collapses open. And yeah, it kind of looks like something you would see in a slaughter movie. And I was literally holding my own breast while he cut it in front of my face. So I felt quite vulnerable then when I returned to the hospital, probably about 18 months later, and I was obviously very reluctant to go back when I returned to the hospital 18 months later because I needed another surgery…and I explained it to the nursing staff. They then informed me because of COVID restrictions, your husband has to leave after one hour and he has to sit in the waiting room and wait for you, and I explained how I was feeling. I asked to speak to the nursing unit manager, who then threatened to kick my husband out of the hospital, who is like my only support person. Even after explaining how stressed and vulnerable I felt, and I actually got out of bed and left the hospital about 35 minutes after the surgery, I pulled my own cannula out and left the hospital because that's how distraught I was when they kicked him out of the room rather than just having a little bit of. And by the way, I was in a single room. There was no reason for him to have to leave. He wasn't affecting anybody.
Participant 005_2023AUDSK

Oh yes, definitely in the first stages. I think again, when you don't understand what the medication's doing to you unless you've got somebody that you can call or go back to them immediately because sometimes you can't get in. That can be a bit difficult as well, so if you've got someone you can talk to, a good GP that's…My GP's very well-informed. I can book that and at least go to him and get some common knowledge and then they then try to then call the physician if they can or go from there.
Participant 001_2023AUDIS

It's really hard to say because OK, well, let's say when she's been in heart having heart surgery. OK, that's what you're most vulnerable.
Participant 33_2023AUDPA

Participant describes feeling vulnerable when feeling sick/unwell

In the beginning especially, and I was a lot younger back then, to be able to push for a diagnosis and actually tell them that I didn't feel what I thought was saying was correct. And definitely when I'm in the middle of a really awful flare up and you, you know, heading in to get help, I think that's when you're feeling the more when I'm feeling the most fragile because you're in so much pain, so.
Participant 022_2023AUDSK

Yeah, when I've been very sick. I live by myself. Yeah. Earlier this year I had a pneumovirus and I was absolutely sure it was COVID. I was so sick and but the tests had come back negative. And at the time I thought, you know, damn it, I'm pretty sick here. That was that was a bit scary. And you know, if that happens at night time when you go to lie down and you can't breathe lying down. I didn't go to hospital. I got better soon after that moment so I didn't have to go to hospital. That was actually just before my 70th birthday. And then three walks later I went on a hike and did 17 K's in soft sand in one day, which was hell. But anyway, that's beside the point. So I did get over it. But those moments when I'm, I'm very sick and, you know, I don't feel like treatment for doing anything. And that was at the time when COVID was on a bit of a rant. Yeah, and I really didn't want to go near a hospital because it was all everyone had. COVID.
Participant 19_2023AUDPA

When I was really sick and potentially couldn't advocate for myself?
Participant 31_2023AUORC

Table 8.11: Experience of vulnerability

Experience of vulnerability	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=180	%	n=67	%	n=31	%	n=9	%	n=32	%	n=9	%	n=32	%	n=100	%	n=80	%	n=121	%	n=57	%
Participant describes feeling vulnerable when having sensitive discussion (diagnosis, treatment decision)	30	16.67	10	14.93	11	35.48	2	22.22	4	12.50	0	0.00	3	9.38	22	22.00	8	10.00	23	19.01	7	12.28
Participant describes feeling vulnerable because of interactions with the medical team	26	14.44	5	7.46	10	32.26	3	33.33	6	18.75	0	0.00	2	6.25	22	22.00	4	5.00	23	19.01	3	5.26
Participant describes feeling vulnerable experiencing side effects from treatment or symptoms from condition	17	9.44	2	2.99	4	12.90	4	44.44	6	18.75	0	0.00	1	3.13	14	14.00	3	3.75	17	14.05	0	0.00
Participant describes feeling vulnerable thinking about disease course/incurable condition	15	8.33	2	2.99	6	19.35	3	33.33	2	6.25	0	0.00	2	6.25	12	12.00	3	3.75	11	9.09	3	5.26
Participant describes feeling vulnerable during/after treatments	12	6.67	2	2.99	2	6.45	0	0.00	3	9.38	0	0.00	5	15.63	9	9.00	3	3.75	8	6.61	4	7.02
Participant describes feeling vulnerable when feeling sick/unwell	10	5.56	3	4.48	3	9.68	0	0.00	1	3.13	1	11.11	2	6.25	7	7.00	3	3.75	9	7.44	1	1.75

Experience of vulnerability	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=180	%	n=61	%	n=63	%	n=37	%	n=19	%	n=88	%	n=92	%	n=48	%	n=132	%	n=94	%	n=86	%
Participant describes feeling vulnerable when having sensitive discussion (diagnosis, treatment decision)	30	16.67	7	11.48	8	12.70	12	32.43	3	15.79	12	13.64	18	19.57	8	16.67	22	16.67	18	19.15	12	13.95
Participant describes feeling vulnerable because of interactions with the medical team	26	14.44	3	4.92	7	11.11	9	24.32	7	36.84	15	17.05	11	11.96	4	8.33	22	16.67	12	12.77	14	16.28
Participant describes feeling vulnerable experiencing side effects from treatment or symptoms from condition	17	9.44	1	1.64	6	9.52	6	16.22	4	21.05	8	9.09	9	9.78	6	12.50	11	8.33	10	10.64	7	8.14
Participant describes feeling vulnerable thinking about disease course/incurable condition	15	8.33	3	4.92	1	1.59	7	18.92	4	21.05	4	4.55	11	11.96	6	12.50	9	6.82	9	9.57	6	6.98
Participant describes feeling vulnerable during/after treatments	12	6.67	2	3.28	5	7.94	4	10.81	1	5.26	7	7.95	5	5.43	5	10.42	7	5.30	8	8.51	4	4.65
Participant describes feeling vulnerable when feeling sick/unwell	10	5.56	2	3.28	4	6.35	2	5.41	2	10.53	5	5.68	5	5.43	4	8.33	6	4.55	7	7.45	3	3.49

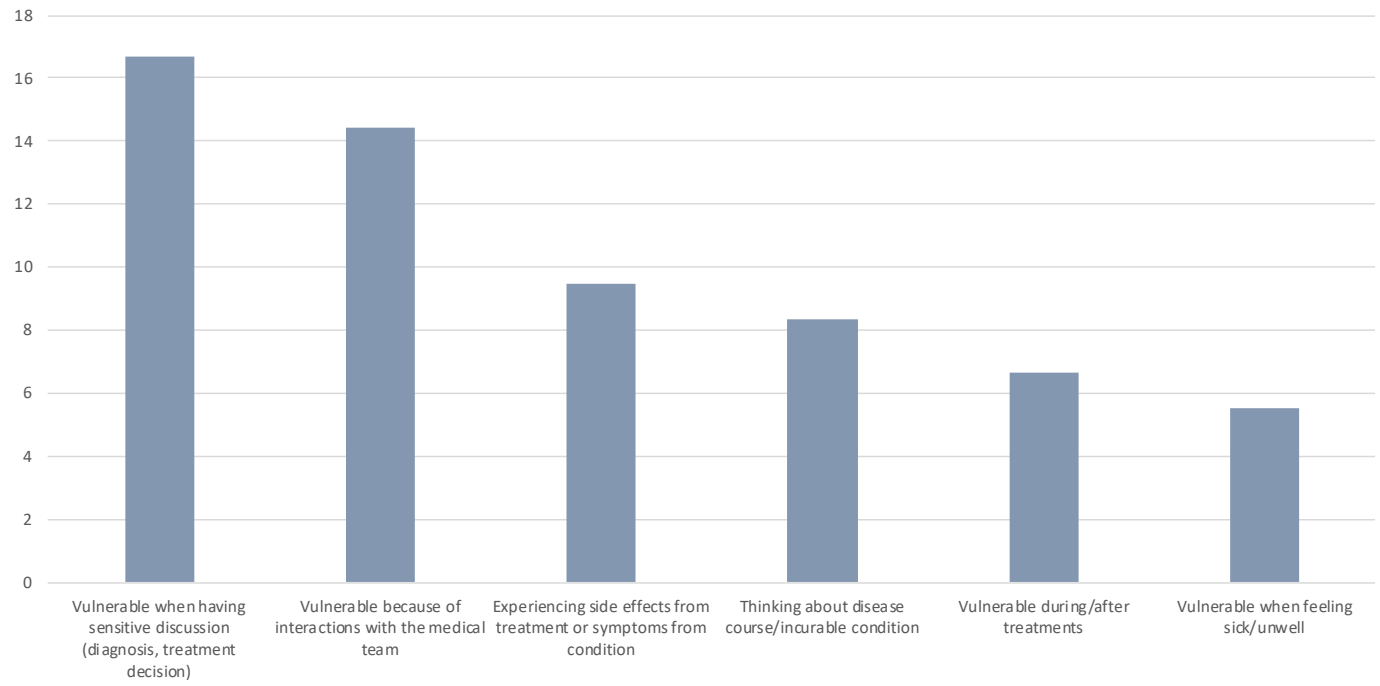


Figure 8.6: Experience of vulnerability

Table 8.12: Experience of vulnerability – subgroup variations

Experience of vulnerability	Reported less frequently	Reported more frequently
Participant describes feeling vulnerable when having sensitive discussion (diagnosis, treatment decision)	Endocrine, nutritional or metabolic diseases	Diseases of the immune system Aged 45 to 64
Participant describes feeling vulnerable because of interactions with the medical team	Endocrine, nutritional or metabolic diseases	Diseases of the immune system Diseases of the nervous system Aged 65 plus
Participant describes feeling vulnerable experiencing side effects from treatment or symptoms from condition	-	Diseases of the nervous system Aged 65 plus
Participant describes feeling vulnerable thinking about disease course/incurable condition	-	Diseases of the immune system Diseases of the nervous system Aged 45 to 64 Aged 65 plus

Table 8.13: Experience of vulnerability (details)

Methods to manage vulnerability	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=180	%	n=67	%	n=31	%	n=9	%	n=32	%	n=9	%	n=32	%	n=100	%	n=80	%	n=121	%	n=57	%
Participant describes using self-help methods (resilience, acceptance, staying positive) to manage the feeling of vulnerability	14	7.78	2	2.99	6	19.35	2	22.22	2	6.25	0	0.00	2	6.25	13	13.00	1	1.25	12	9.92	1	1.75
Participant describes support from nurse or treatment team to manage the feeling of vulnerability	7	3.89	0	0.00	4	12.90	1	11.11	0	0.00	0	0.00	2	6.25	5	5.00	2	2.50	5	4.13	1	1.75
Participant describes getting support from family and friends to manage the feeling of vulnerability	6	3.33	0	0.00	1	3.23	0	0.00	3	9.38	1	11.11	1	3.13	5	5.00	1	1.25	4	3.31	2	3.51
Participant describes support from mental health professionals to manage the feeling of vulnerability	4	2.22	2	2.99	1	3.23	0	0.00	1	3.13	0	0.00	0	0.00	2	2.00	2	2.50	3	2.48	1	1.75

Methods to manage vulnerability	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=180	%	n=61	%	n=63	%	n=37	%	n=19	%	n=88	%	n=92	%	n=48	%	n=132	%	n=94	%	n=86	%
Participant describes using self-help methods (resilience, acceptance, staying positive) to manage the feeling of vulnerability	14	7.78	1	1.64	2	3.17	6	16.22	5	26.32	9	10.23	5	5.43	4	8.33	10	7.58	8	8.51	6	6.98
Participant describes support from nurse or treatment team to manage the feeling of vulnerability	7	3.89	2	3.28	0	0.00	3	8.11	2	10.53	2	2.27	5	5.43	2	4.17	5	3.79	3	3.19	4	4.65
Participant describes getting support from family and friends to manage the feeling of vulnerability	6	3.33	0	0.00	4	6.35	2	5.41	0	0.00	4	4.55	2	2.17	0	0.00	6	4.55	5	5.32	1	1.16
Participant describes support from mental health professionals to manage the feeling of vulnerability	4	2.22	0	0.00	2	3.17	2	5.41	0	0.00	3	3.41	1	1.09	0	0.00	4	3.03	2	2.13	2	2.33

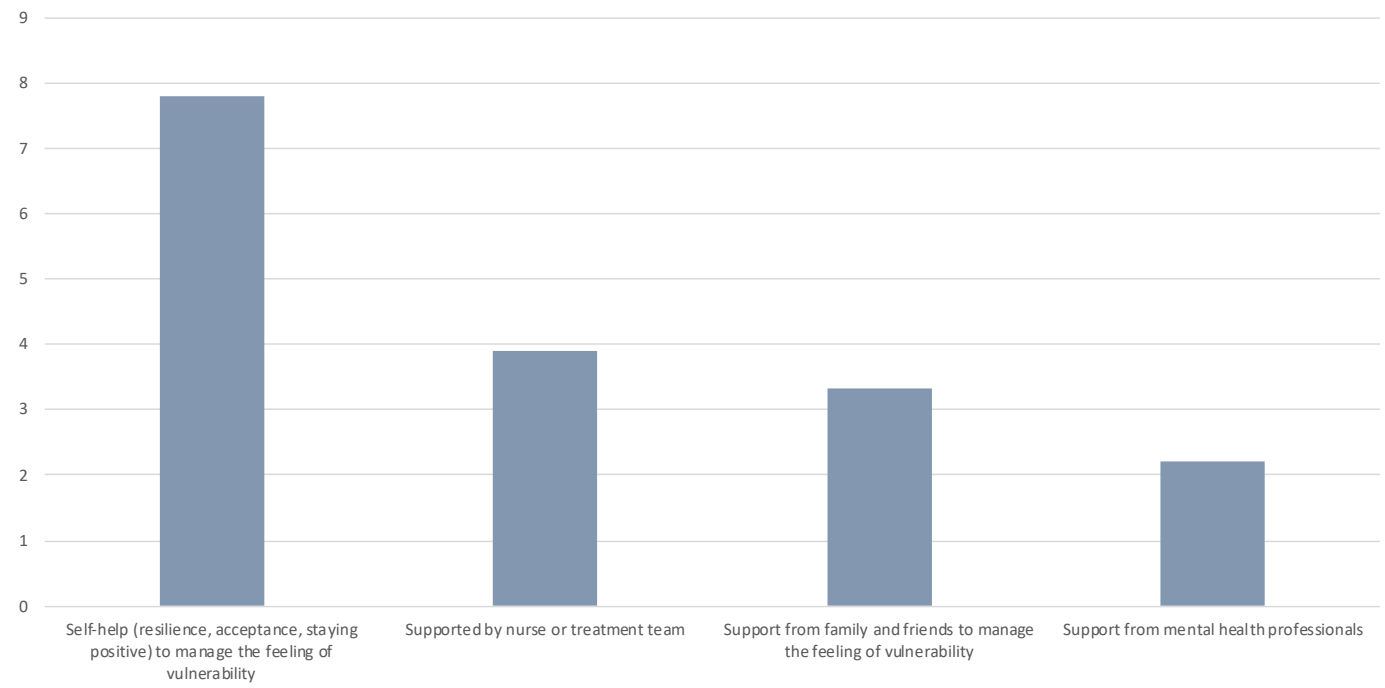


Figure 8.8: Experience of vulnerability (details)

Table 8.14: Experience of vulnerability (details) – subgroup variations

Methods to manage vulnerability	Reported less frequently	Reported more frequently
Participant describes using self-help methods (resilience, acceptance, staying positive) to manage the feeling of vulnerability	-	Diseases of the immune system Diseases of the nervous system Aged 65 plus

Impact on relationships

Most commonly, the descriptions suggested that overall, there was a negative impact on relationships (36.82%), and overall, there was a positive impact on relationships (23.13%). Other themes included overall, no impact on relationships (11.91%), and overall, there was an impact on relationships that was neither positive nor negative (10.95%).

The most common themes in relation to having a negative impact on relationships was from the dynamics of relationships changing due to anxiety, exacerbations and/or physical limitations of condition (25.37%). from people not knowing what to say or do and withdrawing from relationships (22.14%). This was followed by social isolation (10.70 %).

The most common reasons for a positive impact on relationships was that people were supportive and well-meaning (15.67%)

Participant describes that overall, there was a negative impact on relationships

Yes, yes. Especially with my friends. I wouldn't say the same about my family, but with my friends, a lot of things have been, you know, affected and a lot of things have changed. I don't associate much and I tend to shy away from certain activity, which I always engage with my friends, you know, like hanging out and taking drinks or drinking, and all these have been limited and, you know, caught short by the situation. Participant 006_2023AUORC

The only questions that got asked at the one of my face, oh my gosh, I hated going to Christmas. What's on your face? And it's like, Oh my gosh, I don't teach you about your conditions. Yeah. Like, half my family has psoriasis. So, I mean, they think they're worse off and they've got growing skin all the time. But I'm like…and you don't have any sympathy for me. Yeah. Participant 003_2023AUDSK

Yeah, I guess I would say it has. Because of making plans and having to cancel because you can't really do the things that you wanted to do. Yeah, I guess it does. And when people don't know why, you'll understand. It makes it difficult because then they just think that you're blowing them off for no reason and it's like, well, no. Participant 014_2023AUDSK

Yes. Yeah. I think it's just, I guess it's more because it's like invisible. I guess you can't tell unless you, you know what's going on inside the household. It's not a visible disability. And I think that that can be part of the problem in that family can be in denial or think that things will change and she'll get better and yes, and that's just how to fix us.
Participant 16_2023AUORC

Participant describes that overall, there no impact on relationships

They have to love me because they always will, no matter what. And if they didn't do this because they wouldn't be my friends.
Participant 002_2023AUDSK

Probably not. In my case, I have chronic fatigue syndrome, so I've already quite isolated and yeah, sort of focusing on just my main friendship.
Participant 019_2023AUDSK

No, not really, because they don't know about it. You know, it doesn't. I don't talk about it and I'm pretty good at hiding my emotions most of the time. A fairly happily happy person, outwardly.
Participant 024_2023AUDSK

No, I don't think so. No, because everybody who knows her just knows knows her the way she is. So I don't think it's affected anything. No.
Participant 27_2023AUDPA

Participant describes that overall, there was an impact on relationships that was both positive and negative

There are some things like I won't that I won't do so like I won't go to things like water parks and so there's there's some family things that I won't go to. Obviously there are lots of times I have to cancel because as I said pants are not an option and they kind of required when you leave the house. Very fortunate with regards to. So I've had like I've lost a lot of friendships over the years because you cancel all the time and then they just stop asking you to do things with them. I'm very fortunate in that, you know, both my first husband and my second husband are beautiful men. My first husband is still a beautiful man and, and so it didn't affect my physical relationships. But certainly had an impact on my ability to spend time with friends and things like that.
Participant 005_2023AUDSK

Yes, I think that people who don't have chronic illnesses don't genuinely understand how bad it is, and the people who do like you understand each other in a deeper way. But yeah, it impacts it. My closest friends, I don't feel bad about being like, I'm not up for it. I'm not coming out today, it's just not a 'me' day. And they will absolutely say yes, but with people who aren't the closest of friends, I think a lot of them think I've become very flaky over the years, but I don't really care because I don't need to explain myself. Slash if you know. If you don't know me well enough to know that I have HS, then I'm not really losing that but I do feel like there has been a bit of a roll your eyes at me sometimes and I'm OK with that.
Participant 026_2023AUDSK

To some extent it has. But I think now looking back, it's more my mental health that's been affected and and affects the social interactions. But I mean, you know, not, I wouldn't say or like greatly because our friends, our close friends and family understand and we've educated them about it and stuff. So they're pretty accepting.
Participant 14_2023AUDPA

Yes and no. It's made my partner and I get closer because he's had to be there for me a lot more. We were close anyway, but in that way, it's been really good for us. I have a few friends overseas. I have one specific friend who I've distanced myself from because she's all high and mighty studying psychology and she's telling me that it's in my head, all that stuff, and she has no idea what's actually going on because I haven't even seen her physically. Everyone has their opinions about it but she's probably the only one thus far that has really pushed it out too far.
Participant 004_2023AUDNS

Participant describes that overall, there was a positive impact on relationships

No, I don't think so. If anything, it's made it better. I'm quite open about it. I like to tell everyone I meet about it because no one talks about it so.
Participant 027_2023AUDSK

No, not at this stage. I think they're probably bought my family, probably closer. You know, thinking that maybe maybe I haven't spent enough time with my family over the years. And, you know, it's a good thing that now I'm bloody pulling my finger out and catching up with my family a bit more and yeah, probably probably bringing bringing us closer kind of thing. So, yeah.
Participant 24_2023AUORC

Not in a negative way. Probably in a positive way that people see-…We're from a small community, so a lot of people know…When they're walking down the street and word of mouth and stuff. People know how hard we have it and all the stuff that we go through. I think people appreciate and admire how strong and together we are. I think a lot of family love CHILD'S NAME that little bit more because she is the first grandchild of the family. I think because she's so special they want to protect her. Even my close girlfriends, she was treated differently like she was very well loved. Even at her school. People want to look after her and she's not just a normal child. I think she holds a place in people's hearts of everything that she's been through. She's the sweetest little thing. You would never think there's anything wrong with her. I think she feels that little bit more loved. That's all I think anyway. From other people. Yes.
Participant 43_2023AUDPA

I think especially for the family. That's what we thought before. Not just him, his character that had actually had nothing to do with his character, but that it was actually signed in his behaviour related to his condition. You educate the other family members to be more acceptive of what is different and learn them to understand this is not his character but it's part of the TSC. The same, I think with school. There was that one thing, once he was diagnosed, that I actually had a reason to go and sit down with the school and say, "Hey, he is because this, this and this and if we can do this, this and this, have little breaks in between, it'll make life so much easier." Has it changed the relationships? Yes, but I think in a positive way by educating all those around him to be more accepting of who he is.
Participant 49_2023AUDPA

Participant describes that overall, there was an impact on relationships that was neither positive nor negative

It's different but I'm very lucky in that I've got a very, very supportive family. I think it's different, but it's not better or worse.
Participant 017_2023AUDIS

In some ways, with family, with my one of my brothers it has in that when we first got diagnosed I actually let my brothers know that we've been diagnosed with it and my brother, who, actually had, I actually suggested he go and get checked out and he turned and just now the insurance would go through the roof. I told the insurance would bother him [unintelligible], it's the health.
Participant 005_2023AUDPA

It's different but I'm very lucky in that I've got a very, very supportive family. I think it's different, but it's not better or worse.
Participant 017_2023AUDIS

So yeah, I think, well, I think my family are amazing, but then sometimes like treat him a little bit differently to the other kids in the family. And I think that kid's lovely. But when it comes, when it comes to a choice of hanging out with someone you can understand versus someone you can't, it's a bit easier for them. And I don't blame them. It's a bit easier for them to hang out with someone or engage with someone that they can understand. So yes, I believe it affects him.
Participant 23_2023AUDPA

Table 8.15: Impact on relationships

Impact on relationships	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Overall, there was a negative impact on relationships	148	36.82	11	16.42	37	45.68	47	49.47	5	15.63	43	45.26	5	15.63	106	39.55	42	31.34	120	40.82	28	26.42
Overall, there was a positive impact on relationships	93	23.13	21	31.34	16	19.75	17	17.89	13	40.63	12	12.63	14	43.75	56	20.90	37	27.61	66	22.45	25	23.58
Overall, there no impact on relationships	48	11.94	4	5.97	9	11.11	12	12.63	5	15.63	12	12.63	6	18.75	39	14.55	9	6.72	36	12.24	12	11.32
Overall, there was an impact on relationships that was neither positive nor negative	44	10.95	16	23.88	10	12.35	5	5.26	2	6.25	8	8.42	3	9.38	23	8.58	21	15.67	23	7.82	21	19.81
Overall, there was an impact on relationships that was both positive and negative	25	6.22	5	7.46	0	0.00	8	8.42	3	9.38	5	5.26	4	12.50	15	5.60	10	7.46	19	6.46	6	5.66

Impact on relationships	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Overall, there was a negative impact on relationships	148	36.82	34	35.05	50	38.17	48	42.11	16	26.67	73	36.87	73	37.24	44	39.64	104	35.74	80	40.00	68	33.66
Overall, there was a positive impact on relationships	93	23.13	26	26.80	26	19.85	25	21.93	16	26.67	46	23.23	45	22.96	26	23.42	67	23.02	47	23.50	46	22.77
Overall, there no impact on relationships	48	11.94	5	5.15	20	15.27	15	13.16	8	13.33	26	13.13	21	10.71	9	8.11	39	13.40	23	11.50	25	12.38
Overall, there was an impact on relationships that was neither positive nor negative	44	10.95	16	16.49	6	4.58	14	12.28	8	13.33	26	13.13	18	9.18	11	9.91	33	11.34	19	9.50	25	12.38
Overall, there was an impact on relationships that was both positive and negative	25	6.22	9	9.28	12	9.16	2	1.75	2	3.33	10	5.05	15	7.65	9	8.11	16	5.50	10	5.00	15	7.43

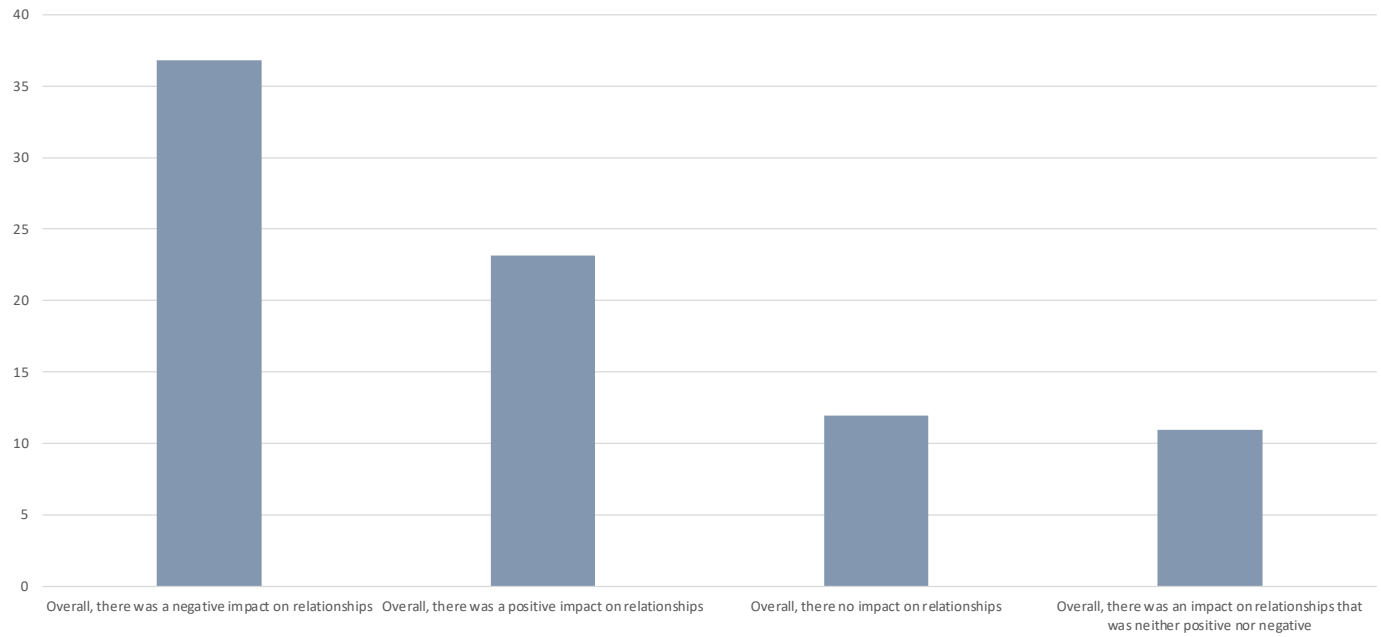


Figure 8.9: Impact on relationships

Table 8.16: Impact on relationships – subgroup variations

Impact on relationships	Reported less frequently	Reported more frequently
Overall, there was a negative impact on relationships	Developmental anomalies Diseases of the skin Other rare condition Male Aged 65 plus	Diseases of the nervous system
	Endocrine, nutritional or metabolic diseases	Diseases of the skin Other rare condition
Overall, there was a positive impact on relationships		
Overall, there no impact on relationships		Developmental anomalies
Overall, there was an impact on relationships that was neither positive nor negative		
Overall, there was an impact on relationships that was both positive and negative		Regional or remote

Table 8.17: Impact on relationships (Reason for impact)

Impact on relationships (reasons)	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Relationship with family changing: dynamics of relationships change due to anxiety, exacerbations and/or physical limitations of condition	102	25.37	22	32.84	26	32.10	22	23.16	6	18.75	19	20.00	7	21.88	63	23.51	39	29.10	78	26.53	23	21.70
Relationships suffering, that is people not knowing what to say or do and withdrawing from relationships	89	22.14	11	16.42	22	27.16	29	30.53	4	12.50	20	21.05	3	9.38	55	20.52	34	25.37	65	22.11	24	22.64
Positive impact on relationships (well-meaning and supportive)	63	15.67	22	32.84	11	13.58	17	17.89	3	9.38	8	8.42	2	6.25	28	10.45	35	26.12	42	14.29	21	19.81
Social isolation	43	10.70	0	0.00	17	20.99	8	8.42	0	0.00	18	18.95	0	0.00	40	14.93	3	2.24	38	12.93	5	4.72
No partuicular reason	42	10.45	11	16.42	4	4.94	16	16.84	5	15.63	2	2.11	4	12.50	23	8.58	19	14.18	32	10.88	10	9.43
Relationships with family being strengthened	29	7.21	6	8.96	2	2.47	6	6.32	1	3.13	9	9.47	5	15.63	16	5.97	13	9.70	19	6.46	10	9.43

Impact on relationships (reasons)	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Relationship with family changing: dynamics of relationships change due to anxiety, exacerbations and/or physical limitations of condition	102	25.37	29	29.90	24	18.32	34	29.82	15	25.00	51	25.76	51	26.02	32	28.83	70	24.05	53	26.50	41	20.30
Relationships suffering, that is people not knowing what to say or do and withdrawing from relationships	89	22.14	24	24.74	35	26.72	26	22.81	4	6.67	45	22.73	43	21.94	26	23.42	63	21.65	46	23.00	39	19.31
Positive impact on relationships (well-meaning and supportive)	63	15.67	28	28.87	20	15.27	8	7.02	7	11.67	29	14.65	34	17.35	18	16.22	45	15.46	29	14.50	44	21.78
Social isolation	43	10.70	2	2.06	18	13.74	17	14.91	6	10.00	27	13.64	16	8.16	11	9.91	32	11.00	19	9.50	24	11.88
No partuicular reason	42	10.45	14	14.43	14	10.69	10	8.77	4	6.67	14	7.07	27	13.78	11	9.91	31	10.65	23	11.50	28	13.86
Relationships with family being strengthened	29	7.21	9	9.28	6	4.58	8	7.02	6	10.00	10	5.05	17	8.67	10	9.01	19	6.53	11	5.50	7	3.47

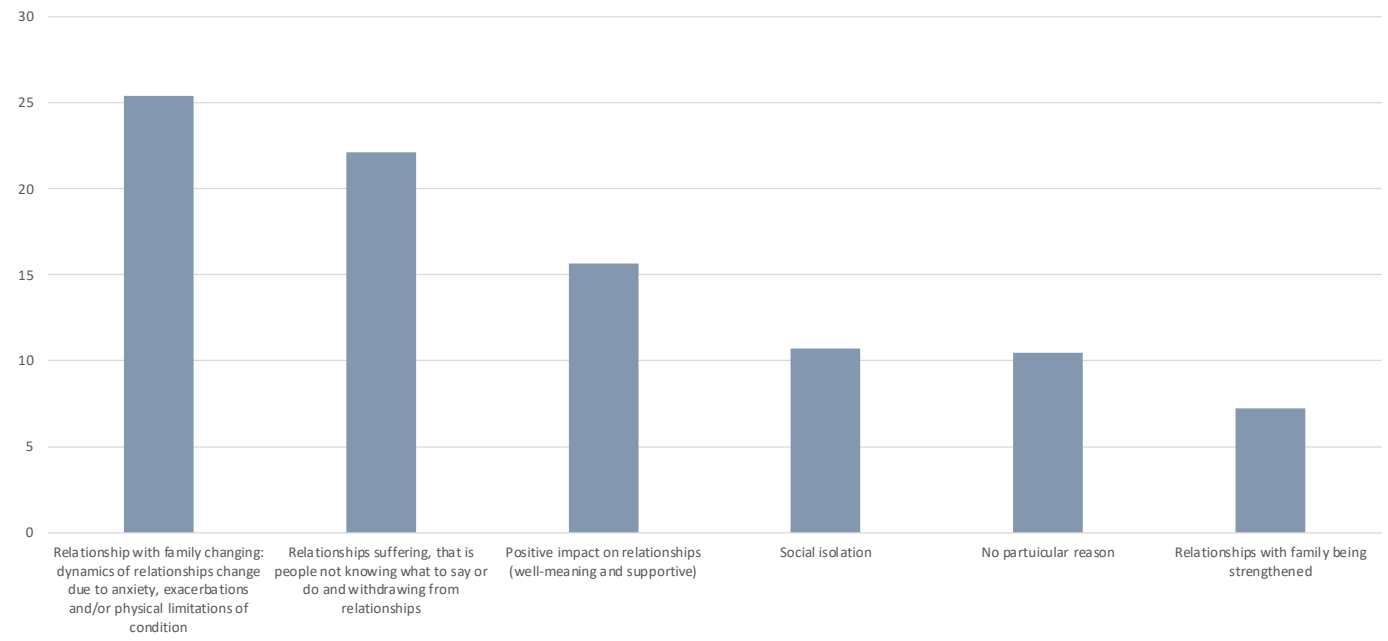


Figure 8.10: Impact on relationships

Table 8.18: Impact on relationships: Reason for impact – subgroup variations

Impact on relationships (reasons)	Reported less frequently	Reported more frequently
Relationship with family changing: dynamics of relationships change due to anxiety, exacerbations and/or physical limitations of condition		
Relationships suffering, that is people not knowing what to say or do and withdrawing from relationships	Other rare condition Aged 65 plus	
Positive impact on relationships (well-meaning and supportive)		Developmental anomalies Family or carer Aged under 18
Social isolation	Developmental anomalies Diseases of the skin Other rare condition	Diseases of the immune system
No partuicular reason		
Relationships with family being strengthened		

Burden on family

In the structured interview, participants were asked whether they felt that their condition placed additional burden on their family. Most commonly, the descriptions suggested that overall, there was a burden on their family (62.60.19%), overall, there was not a burden on their family now but they anticipate this will change in the future (4/26%), and overall, there was not a burden on their family (21.02.64 %).

The main reason that participant described their condition being a burden were the extra household duties and responsibilities that their family must take on(23.01%), and the mental/emotional strain placed on their family (9.94%). Others described the extra assistance needed getting to appointments (5.97 %) and that the burden on family was temporary or only during treatment (3.69 %).

Participant describes that overall, there was a burden on their family

My 15 year old daughter has missed out on having her mum because her mum has been trying to keep her little sister alive for nine years. It's affected, you know, I don't have many friends. I don't go out very often. You know, it's affected our entire lives. And NAME's particularly, you know, I would never label her a burden because she's, you know, a blessing. But it's the burden that she is unable to enjoy, the things that she would like to enjoy. She's unable to go to the places that she wants to go to because her disability prevents her from enjoying.
Participant 80_2023AUDIS

Yeah, I think so. I mean, it's better as the kids have gotten older, but you know, when they were younger they had to do lots of other things that they probably wouldn't have done otherwise, like learning how to tube feed and having emergency protocols stashed left, right and center in case the kids were sick, so. Yeah, I do feel like it's, I don't know if I don't know what the word burden is actually implying, whether it's like a negative thing or just additional load, but it's. Yeah, additional load, I would say all.
Participant 21_2023AUORC

No, the system's a burden, my son is not a burden. The system every, every corner of the system is a burden. No, absolutely not. What I say my son is a burden or or or charged in terms of burden. It's the system, it's the hurdles, it's the challenges, it's the inner, it's the gap, you know, it's the lack of services, it's everything is. It's like a research. It's lack of experts. All of those are, you know, what makes having CHARGE syndrome a burden on my family.
Participant 28_2023AUORC

I don't think so. I don't tend to rely on them a lot. Extra care assistance, I think they're just. Provide more amounts of care occasionally, which they would do for any child, many grandchild or nephew or whoever I'm seeking that from, so I don't think so, no.
Participant 89_2023AUENM

Participant describes that overall, there was not a burden on their family

No, I wouldn't say so. I guess I'm usually the carer, unfortunately. So, yeah.
Participant 014_2023AUDSK

No, I don't think so. No.
Participant 027_2023AUDSK

Them no. I'm fiercely independent with it. I have to be in a really bad way before I ask anyone for help. So you know, I'll get a taxi to hospital rather than ask my son to come and pick me up. And he would do it at a drop of a hat. But I'm very conscious of not wanting to be the single old lady, old lady living alone, needing help. You know, the needy other person. I don't want to be that. So I'll hang on doing whatever I can for myself for as long as I can. Yeah, okay. Nothing. Part of that keeps me well, get up, dress up, show up. Half the time now you might wake up feeling rotten, and if you stayed home, you'd feel rotten. Get up, dress up, show up and get out there. Half the time you feel better by the time you get out.
Participant 18_2023AUORC

Participant describes that overall, there was not a burden on their family now but they anticipate this will change in the future

At the moment we don't need a lot of carer assistance. I think as I get older I will, but the closest one's an hour away and then the other ones, he's he's about two hours away and the one in the middle is about an hour and a half. So you know they're, I don't need a lot. We don't need a lot of help from them yet.
Participant 024_2023AUDSK

It probably will be. If I don't get any better than where I'm at now, but everything's suggesting to me that I I can get better. So look if I'm stuck at this point, yes it will affect them fairly heavily. If I get past this which I think I'm gonna it. It's burning always up a fair bit, you know? Yeah.
Participant 14_2023AUORC

No not at this stage. Well not that I would say you know they haven't really sort of I don't think they've changed their life as much so but I'm not saying that later down the track they I may be a bit of a burden on them but early days I haven't had any sort of issues things sort of they've been pretty good that sort of set off you know if we can do anything for you let us know but. I don't think anything's changed at this stage, so yeah.
Participant 24_2023AUORC

Table 8.19: Burden on family

Burden on family	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=352	%	n=67	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=247	%	n=105	%	n=252	%	n=98	%
Overall, there was a burden on their family	220	62.50	48	71.64	56	69.14	24	53.33	15	48.39	55	57.89	22	70.97	155	62.75	65	61.90	152	60.32	67	68.37
Overall, there was not a burden on their family now but they anticipate this will change in the future	15	4.26	1	1.49	2	2.47	2	4.44	1	3.23	6	6.32	3	9.68	10	4.05	5	4.76	12	4.76	2	2.04
Overall, there was not a burden on their family	74	21.02	13	19.40	16	19.75	9	20.00	9	29.03	23	24.21	4	12.90	51	20.65	23	21.90	53	21.03	21	21.43

Burden on family	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=69	%	n=116	%	n=108	%	n=59	%	n=172	%	n=172	%	n=100	%	n=252	%	n=176	%	n=176	%
Overall, there was a burden on their family	220	62.50	43	62.32	76	65.52	66	61.11	35	59.32	111	64.53	105	61.05	63	78.75	157	62.30	108	61.36	112	63.64
Overall, there was not a burden on their family now but they anticipate this will change in the future	15	4.26	3	4.35	7	6.03	2	1.85	3	5.08	8	4.65	7	4.07	3	3.75	12	4.76	4	2.27	11	6.25
Overall, there was not a burden on their family	74	21.02	14	20.29	22	18.97	25	23.15	13	22.03	37	21.51	36	20.93	22	27.50	52	20.63	42	23.86	32	18.18

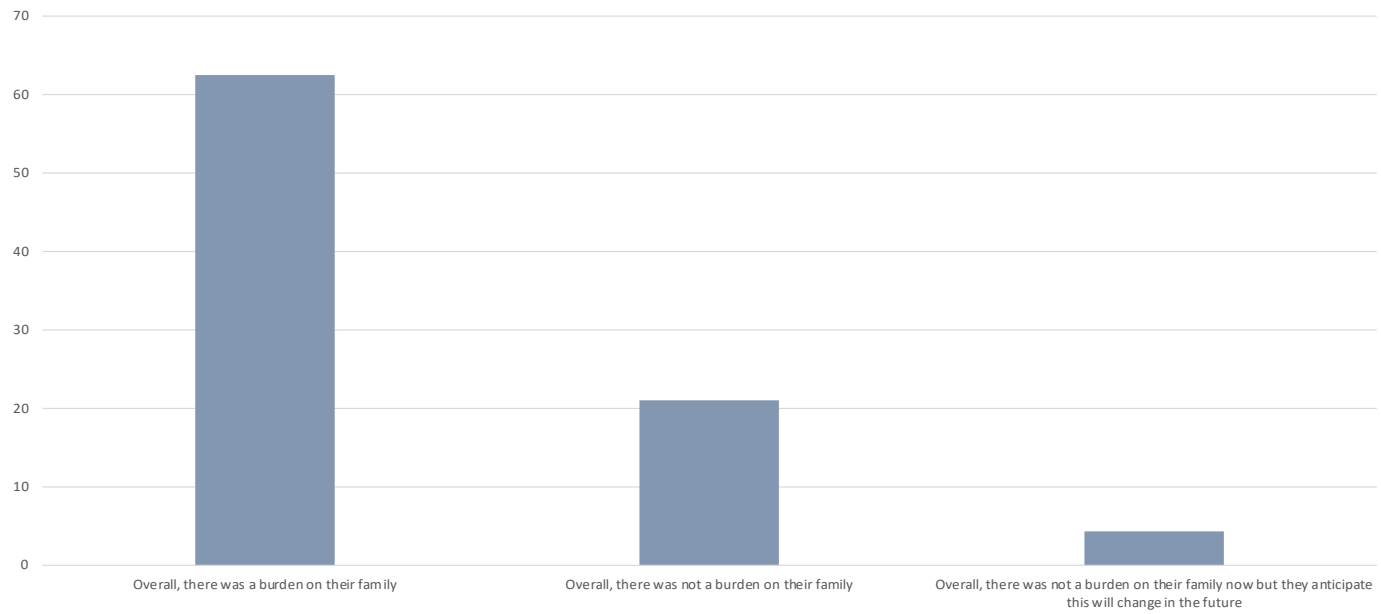


Figure 8.11: Burden on family

Table 8.20: Burden on family – subgroup variations

Burden on family	Reported less frequently	Reported more frequently
Overall, there was a burden on their family	Diseases of the skin	Regional or remote

Table 8.21: Burden on family (description)

Burden on family (description)	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=352	%	n=67	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=247	%	n=105	%	n=252	%	n=98	%
Extra household duties and responsibilities	81	23.01	21	31.34	21	25.93	7	15.56	5	16.13	23	24.21	4	12.90	53	21.46	28	26.67	57	22.62	24	24.49
Burden: no specific examples	73	20.74	11	16.42	21	25.93	7	15.56	5	16.13	18	18.95	11	35.48	54	21.86	19	18.10	50	19.84	22	22.45
Not a burden: no specific examples	48	13.64	8	11.94	12	14.81	8	17.78	4	12.90	16	16.84	0	0.00	34	13.77	14	13.33	35	13.89	13	13.27
Mental/emotional strain	35	9.94	11	16.42	2	2.47	3	6.67	3	9.68	10	10.53	6	19.35	20	8.10	15	14.29	22	8.73	13	13.27
Assistance getting to appointments	21	5.97	5	7.46	1	1.23	5	11.11	1	3.23	9	9.47	0	0.00	15	6.07	6	5.71	16	6.35	4	4.08
Temporary or only during treatment	13	3.69	2	2.99	1	1.23	1	2.22	3	9.68	0	0.00	6	19.35	10	4.05	3	2.86	8	3.17	5	5.10

Burden on family (description)	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=69	%	n=116	%	n=108	%	n=59	%	n=172	%	n=172	%	n=100	%	n=252	%	n=176	%	n=176	%
Extra household duties and responsibilities	81	23.01	21	30.43	23	19.83	24	22.22	13	22.03	38	22.09	42	24.42	23	28.75	58	23.02	31	17.61	50	28.41
Burden: no specific examples	73	20.74	13	18.84	26	22.41	19	17.59	15	25.42	37	21.51	35	20.35	21	26.25	52	20.63	43	24.43	30	17.05
Not a burden: no specific examples	48	13.64	9	13.04	13	11.21	14	12.96	12	20.34	18	10.47	29	16.86	17	21.25	31	12.30	29	16.48	19	10.80
Mental/emotional strain	35	9.94	9	13.04	13	11.21	5	4.63	8	13.56	15	8.72	18	10.47	10	12.50	25	9.92	12	6.82	23	13.07
Assistance getting to appointments	21	5.97	4	5.80	8	6.90	8	7.41	1	1.69	9	5.23	12	6.98	10	12.50	11	4.37	13	7.39	8	4.55
Temporary or only during treatment	13	3.69	1	1.45	7	6.03	4	3.70	1	1.69	6	3.49	7	4.07	4	5.00	9	3.57	4	2.27	9	5.11

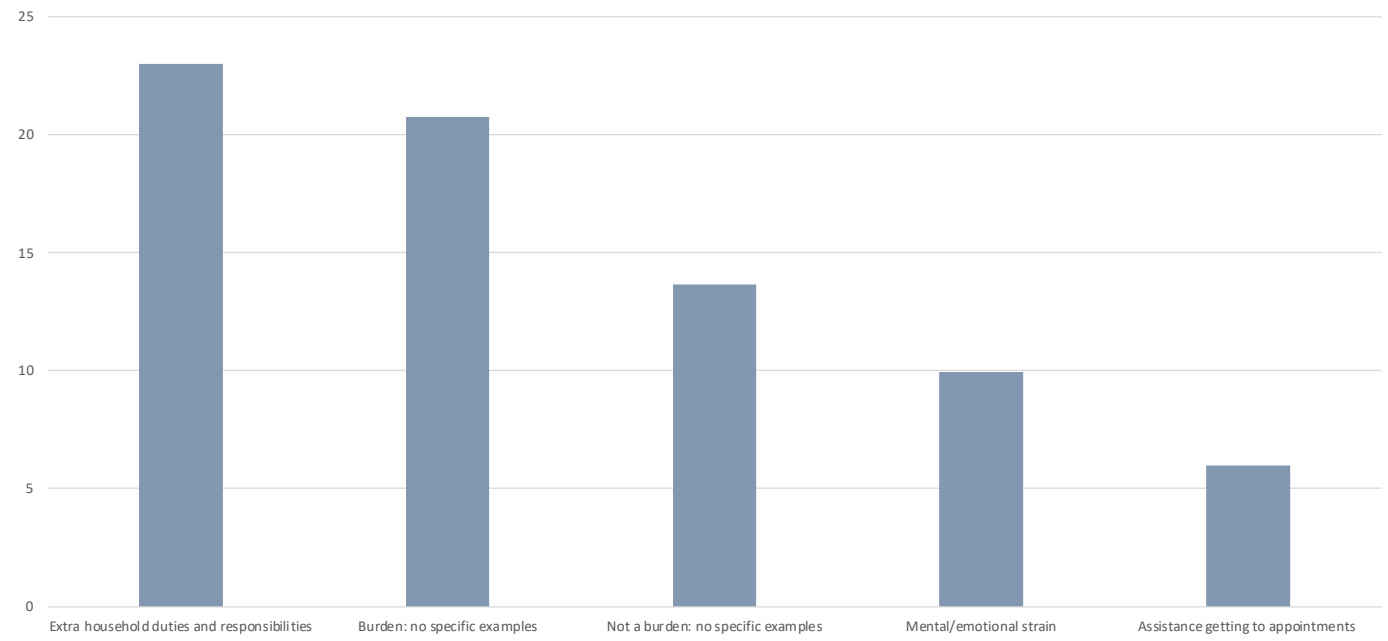


Figure 8.12: Burden on family (description)

Burden on family (description)	Reported less frequently	Reported more frequently
Extra household duties and responsibilities	Other rare condition	
Burden: no specific examples		Other rare condition
Not a burden: no specific examples	Other rare condition	
Mental/emotional strain		
Assistance getting to appointments		
Temporary or only during treatment		Other rare condition

Table 8.22: Burden on family (description)– subgroup variations

Cost considerations

In the structured interview, participants were asked about any significant costs associated with having their condition. The most common descriptions were that overall, there was at least some cost burden (65.23%), and overall, there was no cost burden (18.87%).

Where participants described a cost burden associated with their condition, it was most commonly in relation to needing to take time off work (32.78%), the cost of treatments (including repeat scripts) (30.79%), and the cost specialist appointments (26.82 %). Other themes included diagnostic tests and scans (12.91%), the cost of parking and travel to attend appointments (including accommodation) (12.91%), needing to special equipment (8.61%), a family member needing to take time off work (5.96%) allied health care (5.63%), needing to special creams, ointments or complementary therapies (4.30%), and needing a special diet or lifestyle adaptation (3.64%).

Where participants described a cost burden associated with their condition, it was most commonly in relation to nearly everything was paid for through the public health system (21.52%).

Cost burden in relation to needing to take time off work

So I think pretty much everything has been covered by Medicare or the NDIS. So in terms of accessing treatment any of the medical treatments or therapies that hasn't been usually costly but my income has been affected because instead of going back to work full time as I'd intended after his birth, I only ended up going back to work 2 days a week, just because it's quite difficult for one person to manage him. So say, if I left him with my partner five days a week while I worked, it's…that's quite a lot for him to manage and only recently has he been able to start childcare. We've been able to organise that just because of his additional need, so finances were affected in terms of

my income was greatly reduced, even though I didn't have extra costs with his therapies or medical treatments.
Participant 89_2023AUENM

There was definitely increased costs at birth because she was in hospital for such a large length of time. And she wasn't in private hospitals, so we had to pay out money for that, obviously and the private pediatrician that was, she was saying while she was in there. As I said though, she did gain NDIS funding. I'm gonna say early primary school. I can't really remember how old she was. So we've had that for a number of years to cover the cost of therapies and stuff going forward. So that's been very helpful. There would be, there would be probably a large chunk of time that I've personally had to take off from work to be home with her when she's unwell because she does, she doesn't get sick a lot, but when she does it tends to linger. So there are, you know, large links of time where, yeah, I've probably had to take a week off here and there to be at home with her. Yeah.
Participant 27_2023AUDPA

My experience is I'm very lucky that I have a dad who's still alive with some money. It's been very expensive. Yes. Yeah. I've had to stop working. I had to give up work.
Participant 002_2023AUDNS

Cost burden in relation to the cost of treatments (including repeat scripts)

It hasn't been too bad in the last two years since I got a higher paying job, but before that I pretty much just was paying bills and everything, week to week, and then my medical expenses came up, but I would rather just. I'd often go without medication because I couldn't afford it because of the amount of medications that we have to take. And then when it came to being in hospital with work, it was just pretty much borrowing money from my mom to cover bills and whatnot until I could go back to work, or until I could commit to the doctors to let me go to work while I was in there.
Participant 13_2023AUORC

Probably the biggest one is the full-time off work, it's obviously very hard on the family. Also now, I find with fatigue and just chasing up medical appointments and things like that, that I only work part-time now. I work three days a week. Just financially that. I find that with scripts and seeking treatment, very expensive. Just getting accommodation and things like that, going down to specialist appointments, I find very expensive as well. Time-wise, definitely it takes up way too much family time with conversations and just their support
Participant 014_2023AUDIS

Well, the surgery cost a fortune, but I mean, we were in that position of being able to afford it. My husband just said no, no, it's worth doing. The focused ultrasound would have been at least twice as much, which I didn't have. So, I'm fortunate that I could afford that. And as I said, my neurologist is no longer charging me, so I.
Participant 003_2023AUDNS

It's been pretty significant by the time you add up all the, the emergency room visits, the medications, the different diagnostic tests like halter monitors and echocardiograms and things like that. So even though we had private health, the out of pocket's been pretty, pretty significant, yeah. And then the ablations and things like that as well.
Participant 32_2023AUORC

Cost burden in relation to the cost specialist appointments

Yeah, so I did find, I did find the costs were substantial for me on a on a single income the you know this. And as a single parent, there's not a lot of extra. So the yeah…the constant outlay for medications, none of that stuff was like, I don't have a health care card or anything like that. So nothing like that was subsidized and then yeah. Seeing all your specialists, there was always a, a significant gap to pay and then I saw as I was lucky that I've been in my job for, for a while. So you know over the when I had the surgeries pretty much the six weeks that I needed to take off 3 after surgery I was able to absorb that through my sick leave. I was probably lucky in that sense.
Participant 007_2023AUDSK

Definitely a cost of even just to see a specialist. You might get a rebate back, but it's still quite expensive to go see the specialist. In the first place. Which is the reason why I haven't gone back and in there, cost of medication as well. Like it's, it's certainly not cheap.
Participant 011_2023AUDSK

It has put me into extreme financial hardship on every possible level and I find I'm often declining appointments, even specialist appointments. I've had to postpone multiple times because I just can't afford them, which is really disheartening when you wait six months to get in and then you can't afford to go.

Participant 016_2023AUDIS

Cost burden in relation to diagnostic tests and scans

The major cost is because you're treated as a private patient. So every time you go, there's the consultation fee and then the ongoing fee. That was the biggest killer tests. Sometimes they were both billed, sometimes that was out of pocket. Yes, I did have to take off days off work because I couldn't work. Yeah, yeah, every. I think the biggest thing is that all the appointments, it's, it's money, and a lot of it's not covered by Medicare or even your private health insurance. So there's a lot of money being spent upfront.
Participant 013_2023AUDSK

Yeah, I think, look, we live in a pretty lucky country with Medicare. A lot of it is covered. Little specialist fees can be a bit hefty when you're seeing them quite regularly, but I'm in a position now where it's only a couple of times a year that I have to see them. Initially, there were big costs, like it was all of the let's go see the heart guy. Let's go see the lung guy, let's go get echograms. Let's go get, you know, nuclear medicine. Let's try this medication, try that medication. I think, you know, after, once you get that diagnosis and you kind of managed after that, it's not too, but haven't been too bad. It all counts like I'm a single mom. It all counts now. I'm sort of managing now. Initially it was every time you turn around with another $300.00 with the hospital side of things and having the gastroscopes and all that kind of stuff, like, you know, it's covered by Medicare, so we're relatively lucky. But I have spoken to people in America, for example, and they don't have that. It's super expensive, but you know, they go without so much. So comparatively, you know, we're in a relatively lucky position. There are definitely costs involved, but I don't think once you've got your diagnosis and you're kind of on the right track, I don't think it's that bad. It's definitely plenty of worse things. Yeah, like at the moment I'm going through Melanoma stuff, so that's far more expensive, you know?
Participant 78_2023AUDIS

Yes, well the psychologist is very expensive. Yes, there have been quite a few costs but now it's been quite good and there were cardiology costs initially as well for all the scans, but now that I'm under that umbrella there, it's covered for that side of things. Still had the physio obviously isn't and the water therapy wasn't, and the psych isn't and the rheumatologist isn't covered because I see one privately as well. I am out of pocket quite a bit, and I couldn't possibly work because of my fatigue. I'm so tired all the time.
Participant 007_2023AUDIS

Cost burden in relation to the cost of parking and travel to attend appointments (including accommodation)

Initially the treatment that I was getting was in the public system so we weren't out of pocket in that way. The plane fares and that down to Brisbane they were covered by air travel with our local hospital but what they wouldn't cover at that point in time was someone to accompany me down to CITY. That was a added cost because my mum would come with me or one of my family members. We would pay for them to come down. My husband like I said at the time we had a young child so he would be at home and caring for her. We had cattle that we had to keep looking after as well. Then when we had NAME neurologist in town that was great. We would just go up there and then when he left I had to go back down to LOCATION even though I went privately because that was the closest treating neurologist. Our hospital would pay for my airfares down there and they also would pay at that time then for a support person. Every 12 weeks my treatment costs about $650 and I get about $350 of that back from Medicare. We're out of out of pocket about $300 depending on how it falls but four or five times a year. Then of course you've got you get travel upkeep. Then I see a physio once a month. That's covered by some of the private health care. We're lucky that financially we're able to afford that, and it doesn't put too much of a burden on our finances.
Participant 006_2023AUDNS

Yeah, luckily I haven't had many out of pocket like money costs. But yeah, time off work is very stressful and annoying. I guess that's sort of the main one and time traveling to and from the hospital. It's like nearly three hours return on public transport and then you've sort of been there for 5 minutes.
Participant 96_2023AUDNS

Cost burden in needing to special equipment

And then thankfully, if we had other kids, I don't know what we'd do because we don't have a support network to look after them, you know, it's like all of those things that are very quickly. And then you've got the nebulizer equipment, which isn't cheap but also because of, I guess, strict. Hiking protocols for CF, you've got to replace the parts every year, and some of them are covered under the they've got like an

equipment subsidy, so you get new mouthpieces and things every year through the hospital. But then you can't use the same nebulizer head for more than one medication. So for instance, NAME does at the moment. Without an exacerbation, he does hypertonic, saline and pulmicort, but he's got to have a different head for each one and it's cost the same to buy a new head as almost buying the whole new machine. But then you've got also the fact that they've now discontinued it, so you either have to go and source these the heads that they need to use because it has to. You have to push the medication through in a certain at a certain speed and time for it to be absorbed roughly. So you've got to source specific machines which the clinic have been trying to do, but then you've got to go pay for them as well, which is a couple $100 on top of that. And then you've got to have more than one and you know and then we've also got the added expenses of like all the, you know like the ongoing osmolax and…You know, just if like I guess that's kind of most of it because then you've got like we've obviously got PTSS and we get a subsidy for like heating and cooling as well, which is like $90.00 a year or half. I can't remember that you get to help with because of the excess you're sweating. You've got to try and keep them in the air con as much as possible during summer and but then you've got. I mean, the cost of living and electricity, you've got to actually, you know, it costs a lot more than you're actually getting subsidized. But I think it's a meds extra, like heating and cooling and then the nebulizers think. That's, yeah. And then, yeah, obviously time off illness for hospital stays and that's pretty much it that I can think of at this .0 and partly sorry, yeah.
Participant 20_2023AUORC

Costs. It's been very difficult. I'm I carer. And because I was a full time carer, I was had to quit my full time job. So not only did we lose a second income, the costs of the medications are all on PBS, which is fine, but when you've got ten a month, it sort of adds up. But the bigger costs are more the, the nebulizers. Like, they cost thousands of dollars, but then you need twice a year to change the heads, which are another like $500.00. And so it's those things, it's the extra, yeah. And then it's not listed on NDIS. So there's no. Yeah, it's very, it's tricky.
Participant 23_2023AUORC

Really hasn't been a lot of out of pocket. We've got private health cover, but trip was at the children. So apart from equipment, I don't know, like syringes and and different things, I, I don't know that there was and tubes and stuff. I don't think there was too much. Oh, that's right. I think because he was private, I think originally. We had a lot of pathology bills and if he'd been public we wouldn't have got any of those. Medibank Private has certainly helped a couple of times out with cochlear implant replacements, yeah, and even with schooling, apart from the regular costs, anybody else has. You know as far as surgeries go, I think we've, we've been very lucky and also when he was at sorry, I was trying to say when he was at LOCATION, I don't think we paid for occupational therapy sort of the oh sorry the occasional care which is sort of a bit like kinder. So I would say, you know, we've been quite fortunate in that regard.
Participant 93_2023AUENM

Cost burden in relation to a family member needing to take time off work

So iIn terms of cost, because I'm going through the public system, there's no cost with the consultations. Medications. I mean, my husband and I work full time, so we we're financially OK and the cost of medications isn't that bad, like, you know, it's not that bad. He had to take some time off work to help me when I had the surgery because I needed that dressing changed every couple of hours. Yeah, so he was the poor bunny who had to do it. And I've probably, I've never, you know, because you have paid sick leave. I probably can't. You know, I can't pinpoint time when I've lost money as such because I leave and I had to take time off work. But I, you know, I think maybe, maybe I could say I've probably lost up maybe 3 weeks. Work in the last three years into HS, so yeah.
Participant 017_2023AUDSK

Look, it's been it's, it's a very long journey for the most part. In my 20s, getting to the dermatologist, waiting three to six months, finding out it's three $400.00 and that most of that would be out of pocket. It's painful. So you delay it and you procrastinate going. As I've gotten older, I haven't done that, but the surgery was pretty much all private, so I think I coughed at around two and a half thousand plus not going to work, plus my partner staying home so he can take me back from the hospital. It's, it's quite it's currently quite expensive because. Of the surgery, but on a day-to-day basis with the appointments around me. I think I calculated it at roughly around $400.00 a month just for medical expenses.
Participant 026_2023AUDSK

Well, I had to give up my work because obviously, my wheelchair and I couldn't do it, so I had to give up work. Before that, you have to go to a specialist in LOCATION and we live in regional, LOCATION. I have to fly down and it's really expensive. I had to take my sister at the time because I didn't have any care. She had to take time off work. If I had to do a driving assessment, you have to go to the ophthalmologist. That's like, even though I'm in a bond, it's still hundreds of dollars. Neurologists are super expensive. Then you have to go to a psychologist or psychiatrist or that was at the beginning and that costs lots of money. It's just going for your driving license, $1,800 for an OT assessment. They've got to come to the house, get out of hospital jail card, you've got to have all this, then they've got to come and check and it costs so much money. Then medications, some on the PBS, but most of mine aren't. You go and get your scripts done and it's $250. That's not including the medical marijuana.
Participant 001_2023AUDNS

Cost burden in relation to allied health care

With the cost, yeah, we've had to pay everything along the way. We haven't had like the only time we got a bit of support when he was quite a bit younger. The kids at the local hospital that were near, they had like the allied health area there. But then when they get to the age of five, that's it. You can't be seen there anymore. So then we just had to go out and our own backs then and pay for OT and Physio and Health like the health fund paid a little bit and obviously Medicare paid for some things. But yeah, it's generally costs are on our back not they're not that many Medicare benefits for.
Participant 11_2023AUDPA

So everything we've done is privately, so cost would be speech therapy. Then that's not covered by Medicare, but it is partially covered by HBF. But once you reach the threshold, you don't get anymore cover for, you know, X amount of sessions in a year or within a six months or whatever. I can't even remember what it is now. So yeah, we've paid privately for speech OT. All of our doctors have been privately. We haven't gone through LOCATION Children's, His dental work, yeah, everything we've done is just private. So, yeah, it's been thousands and thousands of dollars.
Participant 22_2023AUDPA

Yeah, we've been very lucky that we've been able to afford her what we say. So I had to give up work after she was 18 months old. I was juggling the two, but it wasn't working so I gave up work. I've only just been able to start looking at going back to work probably the beginning of next year because you still need so much care. A lot of the specialists that we have are private because of the wait list for public entities. So, and then for the first few years we were paying all out of pocket costs for Private Allied Health because the duplication isn't necessarily recognized to the NDIS as a disability. So it wasn't until she was about how old is she now, 4 1/2 that we got an NDIS plan which still doesn't cover her support. So we use the NDIS package and then we use our own funds to pocket the rest of the year.
Participant 32_2023AUDPA

Cost burden in needing to special creams, ointments or complementary therapies

Across…I would definitely say there's, there's a few. So cost in terms of treatment is a big one, so seeing the specialists, I don't have health insurance, so I pay out of pocket to see a specialist, which is roughly $280 per session with this doctor. My medication costs $40 a month and you know I have to buy special bras which are very expensive like. Not very expensive, but you know, just constantly having to buy new ones as well that get a bit destroyed because I get puss and blood on them and they get a bit gross and I don't want to wear it so I have to buy new ones. Taking time off work is rare, like I won't normally take time off work for the disease. I just push through and go to work. Just trying to think, even like at home remedies. I'm always buying new creams, powders, things for the bath, antibiotics, stuff like that. I haven't even know how much that would amount to over the past 10 years. I've had probably a lot, definitely in the thousands that I've spent on all these different things to try for it.
Participant 010_2023AUDSK

Look, fortunate that the DOCTOR is part of the public system. The initial cost to the neurologist and the private rheumatologists were, gosh, I can't even remember now, but hundreds of dollars a visit. Whereas now, that's covered. Certainly, the medications add up and they can run to an expense. The massage is all out of pocket. Look, if I wasn't working, I don't think I wouldn't be able to have the massage or have those other things. I wouldn't have been able to have the wax bath and some of the other things I have. Whereas now, it's anything I need, absolutely. Even things like my complete wardrobe has changed. Now, I don't have anything with zips, buttons, all new shoes, because they've got to be slip

on; that all adds up. I can afford it because I'm still able to work, but if I understand people that can't work, that would be really tough.
Participant 017_2023AUDIS

Well, all of the above I we do go on to the PBS free list by late March, early April which gives you an indication as to how many drugs we take. But a lot of the medication is not on the PBS like TRENTAL is one that's about $58 every fortnight. The obviously the vitamins and that that we need as part of our treatment they obviously you have to pay for. So my chemist is usually about be between 120 and 150 week and between 20 and 80 the other week depending on when what medications have run out. So, yeah, and I mean I take things like laxativesl and you know, because of the drugs I get constipated, so you have to buy, you know, so there's all those sort of things and the expensive sunscreens because of the methotrexate and the specialist. You know, they don't bulk bill. So you you know, it might be a $300.00 bill and the shortfall might be 200 and and yeah it, yeah we've had some tough times but yeah look, I mean I I didn't say anything because my family would my daughter and son-in-law would just say, well you've only got to ask we'll help you. But I don't expect that. So you just manage, but they keep an eye on us and make sure we've got what we need without asking. So yeah.
Participant 021_2023AUDIS

Cost burden in needing to special diet or lifestyle adaptation

Yeah, it's, it can be quite expensive. Like we have like we're very lucky, we've got private health. I have a very flexible job. But yeah, it does a lot of, a lot of medical, like, you know, at times a lot of appointments and the medication. But because he's done so many diets, like eating a gluten and dairy free diet is just generally a lot more. Like my grocery bill is a lot more expensive now than it was before he was diagnosed. So that, like that is just generally that's more expensive than the medication. Yeah, yeah. OK.
Participant 79_2023AUDIS

So we were spending a lot. A lot of money on nappies until we got NDS funding when she was about, I don't know, 2 1/2. But before that we spent we go through three or four boxes of nappies a week. So that was a really big cost. Plus she's very has a very particular taste with things, so always sort of trying different things, buying things that she likes and of course as children that want to do, they're the most, you know, she likes the most expensive things. So having all these things that then she likes and then she doesn't like and, you know, spent all this money on, well she doesn't eat that much. So yeah, I find there's a lot of there's definitely a lot of expenses that we wouldn't have if she didn't have just, you know, just buying. Yeah. Particular things like we've had to air condition the whole house because she's, you know, intolerant to heat. We have the air conditioner running 24/7 in summer, so our power bills are astronomical. Yeah, yeah, OK.
Participant 15_2023AUORC

No cost burden and that nearly everything was paid for through the public health system

Most of my costs have been looked after by the PBS and that I'm very fortunate. You know, the pill, I believe in the United States is about $800 per pill. Yeah. So over a month period, that's about $24,000 exactly, which is a fair whack if you don't have a lot.
Participant 012_2023AUORC

Well, truly, I can't complain that I've had a lot of costs. I've been dealt with by the public hospital system, so I haven't had to pay the specialist. Now and again, I've been sent for something that I've had to pay for, but that's been not the rule, that's been the exception. If I had to pay for my medication, I would have stopped taking it 10 years ago because I can't afford them. The Macitentan itself is, I think it's something between $2,000 and $3,000 a month. I wouldn't be taking that if I had to pay for it. I feel very well supported there that I really…I mean there have been costs, but considering I'm an aged pensioner now that my medication cost very little and I reached the safety net very early.
Participant 004_2023AUDIS

Well, I haven't had any cost things that because I'm on the pension and I just get most of my medications on the PBS, which I'm very fortunate. And I thank God for that.
Participant 005_2023AUDIS

PARTICIPANT: We haven't had really that much cost because most of it's like being like bulk bill and everything. So that was sort of thing, especially like most doctors don't bulk build stuff anymore. So it seems like, yeah. My work's been pretty flexible when I had to take him to the doctors and stuff like that, or when we had like we had to go to Children's Hospital for scans and stuff like that. So they've been pretty flexible with that….Yeah, I so like when he was like

really young. Now it's like how he ever saw this thing and he how he ever goes to the doctors or anything like that.
Participant 7_2023AUDPA

Not too bad to be honest. Like a lot of the stuff you know has been, it goes through the public system or I haven't had to take a lot of time off or I've been given, you know, carers leave and things like that.
Participant 14_2023AUDPA

Well, we've been really fortunate in that most of it's been done through the that we've been bulk billed for most things. The main cost for us would have been our time and also the time that we have had to put into research this condition on our own and be the coordinators of the whole thing, both medically, physically, socially and mentally, so it's our time, our time has been so consumed with looking after our daughter on this condition that there has been some areas where we lack enjoyment and things like that.
Participant 25_2023AUDPA

No cost burden and that nearly everything was paid for through the private health system

I can consider the cost actually was I've got I've got a private health for everything is just covered, so I've never encountered any cost like out of pocket so far.
Participant 001_2023AUORC

Cost wise probably. I've got, I've got private medical, so it most of it, most of it's covered through the private health insurance. But medication? I don't have any. So now I've just got a Commonwealth Seniors card. I do get a bit of a deduction in some medications, which has been very helpful with the Humira, which would have cost me a lot of money.
Participant 024_2023AUDSK

No cost burden as participant was able to afford all costs

It hasn't been too bad because I haven't had that many treatments. The dermatologist appointments can be quite expensive, but it's it's manageable with me working at the moment. And the Humira injections that I'm about to start are very expensive, but they're on PBS them quite affordably.
Participant 019_2023AUDSK

I haven't really had an issue with the cost. I'm probably, I'm probably well off where. I've got private health cover, Medicare. I've covered most of the radiation, which I was, you know, very grateful for. So, yeah, I suppose at this stage money wasn't really an option. You know, scans, scans and all that were just part of part of the process. You know, pet scans. You know, it might have cost me $8000. The other scans are subsidized, didn't cost me a lot because I had private health cover. And the radiation, like you know, the lady did sort of say it was quite expensive but I I found it to be quite reasonable considering, you know, you're having radiation to save your life. The rebate after Medicare I thought was really good. So the costs weren't a real big factor of sort of more the outcome and making sure yeah I sort of knock this, knock this thing on the task as best I can kind of thing. So yeah.
Participant 24_2023AUORC

No cost burden: NDIS

The only the only cost would be me having the stop work to look after NAME because of the effect of the disorder, but other than that, I can't. I mean, she's covered under NDIS in terms of treatment. The other not really, I mean, the only cost is in terms of us raising funds now to get a treatment and a cure. But that's something different. Yeah, OK.
Participant 16_2023AUORC

So yeah, most of it's been covered. We haven't gone privately with anything and really we couldn't afford it back in the early days and now we have the NDIS. So it's a little bit different.
Participant 28_2023AUORC

Hasn't been a massive a lot of costs for us. She's covered by NDIS for therapies and things. So that's been, yeah, that's saved us a lot of cost. Maybe travel probably travel's the biggest cost and so far she's living a normal life as she as a brother and sister do, with maybe just a little bit of extra help and some extra therapies and things. Probably education. We get her tutored once a week with a tutor, so that's an extra cost for us and maybe education because she doesn't qualify for an aid for any of that, so for her schooling.
Participant 10_2023AUDPA

Yeah, very fortunate to have NDIS funding. Excellent. Excellent. Could not have done it without it. And also in terms of timing, our therapists have been very supportive to come and see him at his daycare. Excellent. So we haven't had to take the time off work.
Participant 20_2023AUDPA

Yeah, very costly. I think that my protect career potentials would be I couldn't keep a job of being continuously called out because she wasn't well that's been that was a big thing and yeah and footing the bill for all the specialists you know even though she's got and the health insurance as well. OK, that she's on NDIS now, which helps with certain things, but not with those things.
Participant 33_2023AUDPA

Table 8.23: Cost considerations

Cost considerations	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=302	%	n=67	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=204	%	n=98	%	n=214	%	n=86	%
Overall, there was at least some cost burden	197	65.23	29	43.28	61	75.31	36	80.00	24	75.00	25	26.32	22	68.75	149	73.04	48	48.98	149	69.63	46	53.49
Overall, there was no cost burden	57	18.87	10	14.93	12	14.81	7	15.56	4	12.50	15	15.79	9	28.13	38	18.63	19	19.39	38	17.76	19	22.09

Cost considerations	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=302	%	n=66	%	n=103	%	n=84	%	n=49	%	n=148	%	n=172	%	n=84	%	n=218	%	n=154	%	n=148	%
Overall, there was at least some cost burden	197	65.23	31	46.97	73	70.87	65	77.38	28	57.14	94	63.51	100	58.14	53	63.10	144	66.06	104	67.53	93	62.84
Overall, there was no cost burden	57	18.87	12	18.18	18	17.48	10	11.90	17	34.69	33	22.30	22	12.79	15	17.86	42	19.27	27	17.53	30	20.27

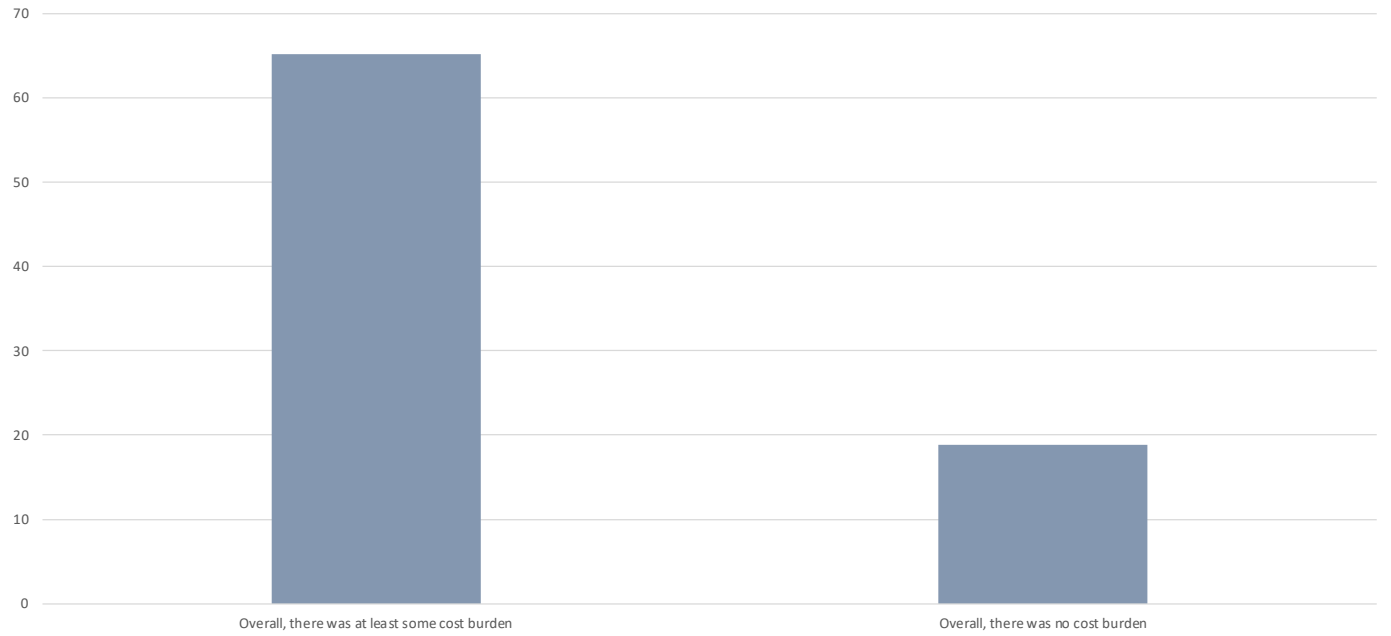


Figure 8.13: Cost considerations

Table 8.24: Cost considerations – subgroup variations

Cost considerations	Reported less frequently	Reported more frequently
Overall, there was at least some cost burden	Developmental anomalies Endocrine, nutritional or metabolic diseases Family or carer Male Aged under 18	Diseases of the immune system Diseases of the nervous system Aged 45 to 64
Overall, there was no cost burden		Aged 65 plus

Table 8.25: Cost considerations (Reasons for cost)

Cost considerations (reasons)	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=302	%	n=67	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=204	%	n=98	%	n=214	%	n=86	%
Cost burden: needing to take time off work	99	32.78	21	31.34	24	29.63	22	48.89	8	25.00	11	11.58	13	40.63	66	32.35	33	33.67	79	36.92	19	22.09
Cost burden: cost of treatments	93	30.79	6	8.96	34	41.98	17	37.78	15	46.88	8	8.42	13	40.63	79	38.73	14	14.29	75	35.05	18	20.93
Cost burden: specialist appointments	81	26.82	11	16.42	47	58.02	6	13.33	12	37.50	5	5.26	0	0.00	69	33.82	12	12.24	70	32.71	10	11.63
Cost burden: diagnostic tests and scans	39	12.91	7	10.45	18	22.22	3	6.67	1	3.13	7	7.37	3	9.38	31	15.20	8	8.16	30	14.02	9	10.47
Cost burden: parking, travel, accomodation to attend appointments	39	12.91	8	11.94	10	12.35	6	13.33	3	9.38	9	9.47	3	9.38	29	14.22	10	10.20	30	14.02	9	10.47
Cost burden: special equipment	26	8.61	8	11.94	2	2.47	3	6.67	6	18.75	4	4.21	3	9.38	14	6.86	12	12.24	23	10.75	3	3.49
Cost burden: family member needing to take time off work	18	5.96	0	0.00	2	2.47	8	17.78	2	6.25	6	6.32	0	0.00	10	4.90	8	8.16	13	6.07	5	5.81
Cost burden: allied health care	17	5.63	8	11.94	4	4.94	2	4.44	0	0.00	1	1.05	2	6.25	9	4.41	8	8.16	13	6.07	3	3.49
Cost burden: special creams, ointments or complementary therapies	13	4.30	1	1.49	4	4.94	1	2.22	6	18.75	0	0.00	1	3.13	11	5.39	2	2.04	10	4.67	3	3.49
Cost burden: special diet or lifestyle adaptation	11	3.64	3	4.48	5	6.17	0	0.00	0	0.00	2	2.11	1	3.13	7	3.43	4	4.08	8	3.74	3	3.49
No cost burden: paid for through the public health system (incl. NDIS)	65	21.52	21	31.34	9	11.11	7	15.56	4	12.50	16	16.84	8	25.00	33	16.18	32	32.65	38	17.76	26	30.23

Cost considerations (reasons)	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=302	%	n=66	%	n=103	%	n=84	%	n=49	%	n=148	%	n=172	%	n=84	%	n=218	%	n=154	%	n=148	%
Cost burden: needing to take time off work	99	32.78	22	33.33	39	37.86	28	33.33	10	20.41	41	27.70	55	31.98	27	32.14	72	33.03	52	33.77	47	31.76
Cost burden: cost of treatments	93	30.79	11	16.67	38	36.89	30	35.71	14	28.57	41	27.70	51	29.65	26	30.95	67	30.73	47	30.52	46	31.08
Cost burden: specialist appointments	81	26.82	8	12.12	33	32.04	28	33.33	12	24.49	46	31.08	35	20.35	25	29.76	56	25.69	47	30.52	34	22.97
Cost burden: diagnostic tests and scans	39	12.91	5	7.58	13	12.62	12	14.29	9	18.37	21	14.19	18	10.47	7	8.33	32	14.68	17	11.04	22	14.86
Cost burden: parking, travel, accomodation to attend appointments	39	12.91	8	12.12	10	9.71	12	14.29	9	18.37	15	10.14	23	13.37	9	10.71	30	13.76	24	15.58	15	10.14
Cost burden: special equipment	26	8.61	10	15.15	7	6.80	9	10.71	0	0.00	12	8.11	14	8.14	9	10.71	17	7.80	17	11.04	9	6.08
Cost burden: family member needing to take time off work	18	5.96	3	4.55	3	2.91	7	8.33	5	10.20	9	6.08	7	4.07	2	2.38	16	7.34	9	5.84	9	6.08
Cost burden: allied health care	17	5.63	5	7.58	5	4.85	4	4.76	3	6.12	9	6.08	8	4.65	3	3.57	14	6.42	8	5.19	9	6.08
Cost burden: special creams, ointments or complementary therapies	13	4.30	2	3.03	3	2.91	6	7.14	2	4.08	8	5.41	5	2.91	4	4.76	9	4.13	11	7.14	2	1.35
Cost burden: special diet or lifestyle adaptation	11	3.64	3	4.55	3	2.91	5	5.95	0	0.00	4	2.70	7	4.07	3	3.57	8	3.67	8	5.19	3	2.03
No cost burden: paid for through the public health system (incl. NDIS)	65	21.52	26	39.39	13	12.62	9	10.71	17	34.69	38	25.68	26	15.12	12	14.29	53	24.31	28	18.18	37	25.00

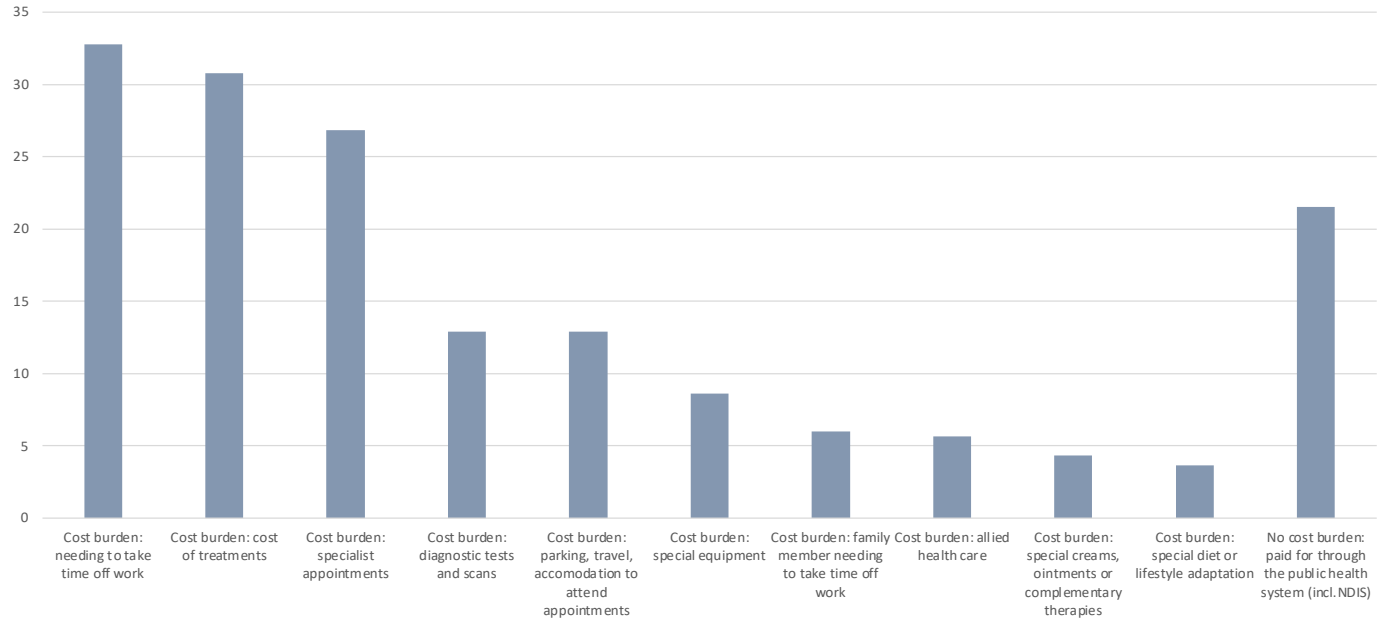


Figure 8.14: Cost considerations (Reasons for cost)

Table 8.26: Cost considerations (Reasons for cost)– subgroup variations

Cost considerations (reasons)	Reported less frequently	Reported more frequently
Cost burden: needing to take time off work	Endocrine, nutritional or metabolic diseases Male Aged 65 plus	Diseases of the nervous system
Cost burden: cost of treatments	Developmental anomalies Endocrine, nutritional or metabolic diseases Family or carer Aged under 18	Diseases of the immune system Diseases of the skin
Cost burden: specialist appointments	Developmental anomalies Diseases of the nervous system Endocrine, nutritional or metabolic diseases Other rare condition Family or carer Male Aged under 18	Diseases of the immune system Diseases of the skin
Cost burden: diagnostic tests and scans		
Cost burden: parking, travel, accomodation to attend appointments		
Cost burden: special equipment		Diseases of the skin
Cost burden: family member needing to take time off work		Diseases of the nervous system
Cost burden: allied health care		
Cost burden: special creams, ointments or complementary therapies		Diseases of the skin
Cost burden: special diet or lifestyle adaptation		
No cost burden: paid for through the public health system (incl.NDIS)	Diseases of the immune system Aged 45 to 64	Family or carer Aged under 18 Aged 65 plus

Overall impact of condition on quality of life

In the online questionnaire, participants were asked to rate the overall impact their condition on quality of life. Quality of life was rated on a Likert scale from one to seven, where one is Life was very distressing and seven is life was great. The average score was in the Life was a little distressing range (median=3.00, IQR=2.00).

Table 8.27: Overall impact of condition on quality of life

Impact of condition on quality of life	Number (n= 225)	Percent
1 Life is/was very distressing	46	20.44
2 Life is/was distressing	64	28.44
3 Life is/was a little distressing	56	24.89
4 Life is/was average	28	12.44
5 Life is/was good	21	9.33
6 Life is/was very good	8	3.56
7 Life is/was great	2	0.89

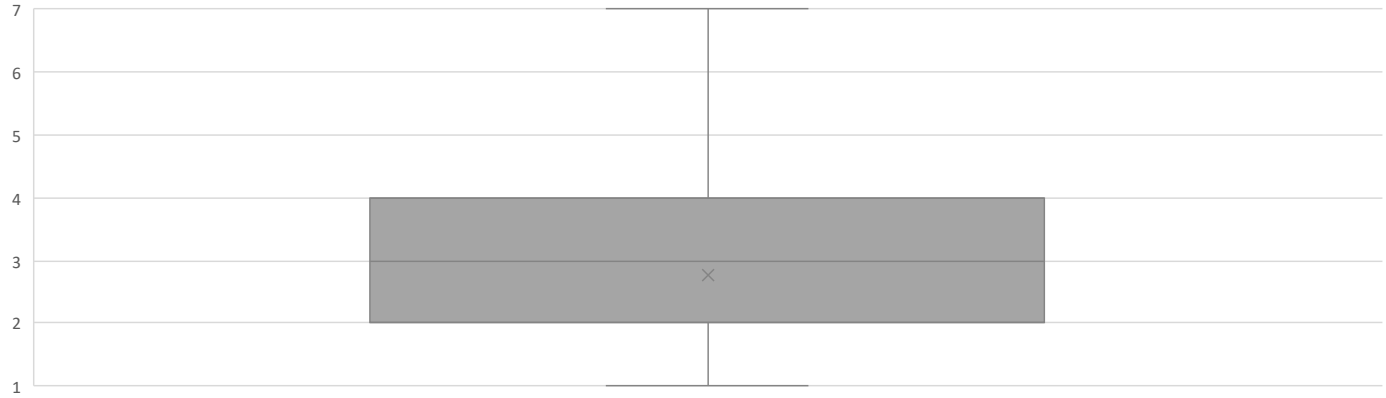


Figure 8.15: Overall impact of condition on quality of life

Experience of anxiety related to disease progression

Fear of progression

The Fear of Progression questionnaire measures the level of anxiety people experience in relation to their conditions. The Fear of Progression questionnaire comprises a total score, between 12 and 60, with a higher score denoting increased anxiety. Summary statistics for the entire cohort are displayed in Table 8.10. Overall the entire cohort had a mean total score of 37.09 (SD = 10.40), which corresponds to moderate levels of anxiety.

The **Fear of Progression** questionnaire measures the level of anxiety people experience in relation to their conditions. On average fear of progression score for participants in this study indicated moderate levels of anxiety.

Table 8.28: Fear of progression summary statistics

Fear of progression (n=370)	Mean	SD	Median	IQR	Possible range	Quintile
Total score*	37.09	10.40	37.00	14.75	12 to 60	3

*Normal distribution use mean and SD as measure of central tendency

Fear of progression by condition

Comparisons were made by **condition**. There were 57 participants (15.41%) with developmental anomalies , 72 participants (19.46%) with diseases of the immune system , 93 participants (25.14%) with diseases of the nervous system , 29 participants (7.84%) with diseases of the skin , 94 participants (25.41%) with endocrine, nutritional or metabolic diseases , and 25 participants (6.76%) with other rare condition.

A one way ANOVA test indicated a statistically significant difference in the Care coordination: Total score scale between groups, F(5, 364) = 4.29 p = 0.0008. The largest significant difference was between participants in the Diseases of the skin subgroup (median = 42.03, IQR = 9.76), and participants in the Endocrine, nutritional or metabolic diseases subgroup (median = 33.77, IQR = 9.10, p = 0.0024).

The **Fear of Progression** questionnaire measures the level of anxiety people experience in relation to their conditions. On average, participants in the Diseases of the skin subgroup scored higher than participants in the Endocrine, nutritional or metabolic diseases subgroup. This indicates that participants in the Diseases of the skin subgroup had high levels of anxiety, and participants in the Endocrine, nutritional or metabolic diseases subgroup had moderate levels of anxiety.

Table 8.29: Fear of progression total score by condition summary statistics and one-way ANOVA

Fear of progression	Group	Number (n=370)	Percent	Mean	SD	Source of difference	Sum of squares	dF	Mean Square	f	p-value
Total score	Developmental anomalies	57	15.41	36.26	10.46	Between groups	2217.00	5	443.40	4.29	0.0008*
	Diseases of the immune system	72	19.46	38.32	10.20	Within groups	37669.00	364	103.50		
	Diseases of the nervous system	93	25.14	38.82	9.95	Total	39886.00	369			
	Diseases of the skin	29	7.84	42.03	9.76						
	Endocrine, nutritional or metabolic diseases	94	25.41	33.77	9.10						
	Other rare condition	25	6.76	35.76	13.97						

Table 8.30: Fear of progression total score by condition one-way post hoc Tukey HSD test

Fear of progression	Group	Difference	Upper	Lower	p adjusted
Total score	Diseases of the immune system - Developmental anomalies	2.06	-3.11	7.22	0.8643
	Diseases of the nervous system - Developmental anomalies	2.55	-2.35	7.46	0.6693
	Diseases of the skin - Developmental anomalies	5.77	-0.88	12.42	0.1306
	Endocrine, nutritional or metabolic diseases - Developmental anomalies	-2.50	-7.39	2.40	0.6886
	Other rare condition - Developmental anomalies	-0.50	-7.49	6.49	0.9999
	Diseases of the nervous system - Diseases of the immune system	0.50	-4.08	5.07	0.9996
	Diseases of the skin - Diseases of the immune system	3.72	-2.69	10.13	0.5589
	Endocrine, nutritional or metabolic diseases - Diseases of the immune system	-4.55	-9.12	0.01	0.0510
	Other rare condition - Diseases of the immune system	-2.56	-9.33	4.21	0.8877
	Diseases of the skin - Diseases of the nervous system	3.22	-2.98	9.42	0.6728
	Endocrine, nutritional or metabolic diseases - Diseases of the nervous system	-5.05	-9.31	-0.79	0.0098*
	Other rare condition - Diseases of the nervous system	-3.06	-9.62	3.51	0.7659
	Endocrine, nutritional or metabolic diseases - Diseases of the skin	-8.27	-14.46	-2.08	0.0021*
	Other rare condition - Diseases of the skin	-6.27	-14.23	1.68	0.2134
	Other rare condition - Endocrine, nutritional or metabolic diseases	1.99	-4.56	8.55	0.9532

Figure 8.16: Boxplot of Fear of progression total score by condition

Fear of progression by type of participant

Comparisons were made by **type of participant** there were 249 participants (67.30%) with person with condition and, 121 participants (32.70%) with carer.

Assumptions for normality and variance were met, a two-sample t-test was used.

No significant differences were observed between participants by **type of participant** for any of the Fear of progression scales.

Table 8.31: Fear of progression total score by type of participant summary statistics and T-test

Fear of progression	Group	Number (n=370)	Percent	Mean	SD	T	dF	p-value
Total score	Person with condition	249	67.30	36.95	10.37	-0.36	368	0.7159
	Carer	121	32.70	37.37	10.48			

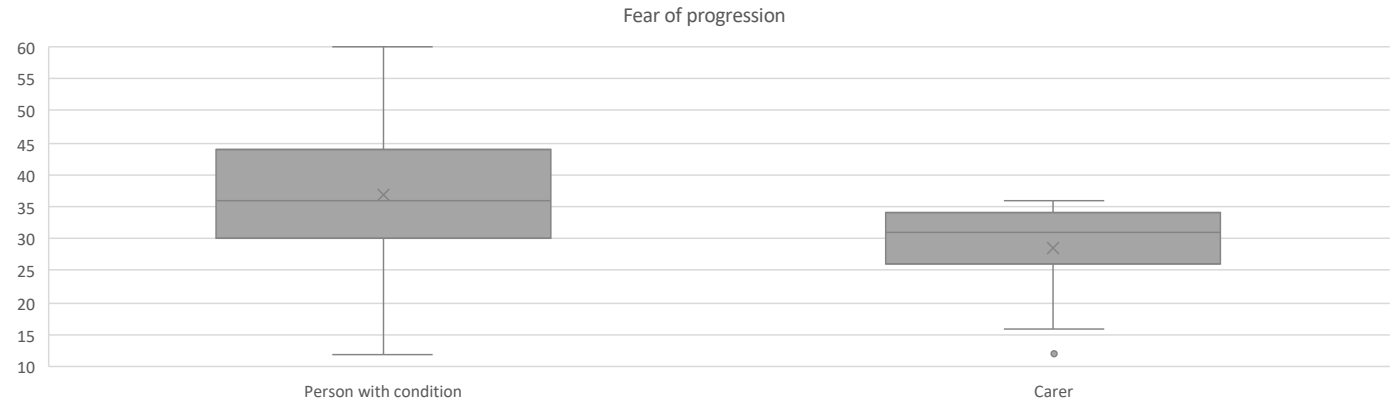


Figure 8.17: Boxplot of Fear of progression total score by type of participant

Fear of progression by gender

Comparisons were made by **gender**, there were 274 female participants (74.46%), and 94 male particpants (25.54%).

Assumptions for normality and variance were met, a two-sample t-test was used.

A two sample t-test indicated that the mean score for the **Fear of progression Total score** scale [t(366) = 2.87 , p = 0.0044] was significantly higher for participants in the Female subgroup (Mean = 37.99, SD = 9.97) compared to participants in the Male subgroup (Mean = 34.46, SD = 11.25.)

The **Fear of Progression** questionnaire measures the level of anxiety people experience in relation to their conditions. On average, participants in the Female subgroup had a higher score compared to Male, however, both groups had moderate levels of anxiety.

Table 8.32: Fear of progression total score by gender summary statistics and T-test

Fear of progression	Group	Number (n=368)	Percent	Mean	SD	T	dF	p-value
Total score	Female	274	74.46	37.99	9.97	2.87	366	0.0044*
	Male	94	25.54	34.46	11.25			

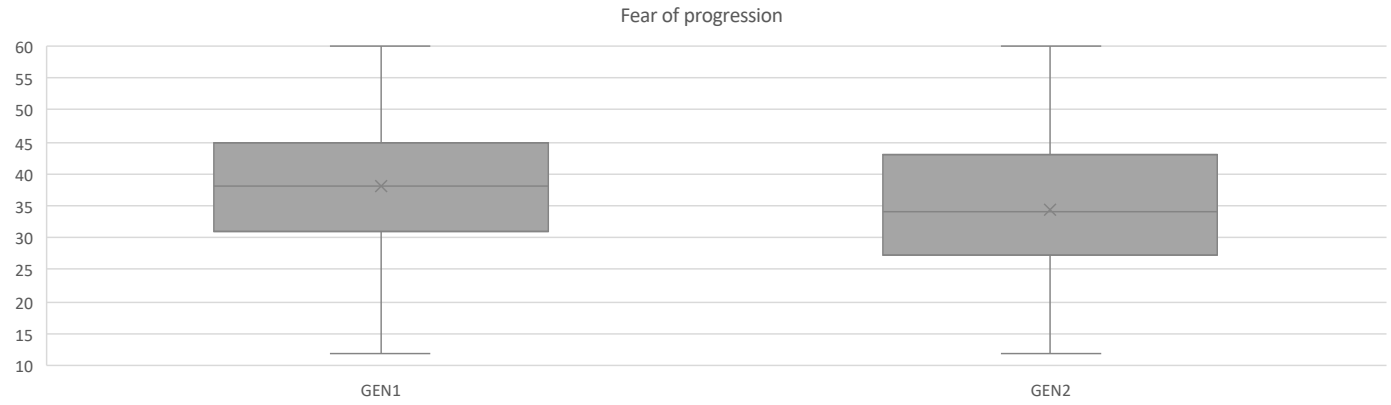


Figure 8.18: Boxplot of Fear of progression total score by gender

Fear of progression by age

Comparisons were made by **age** of person with condition. There were 87 participants (23.45%) with aged under 18, 120 participants (32.35%) with aged 18 to 44, 105 participants (28.30%) with aged 45 to 64, and 58 participants (15.63%) with aged 65 or older.

A one-way ANOVA test was used when the assumptions for response variable residuals were normally distributed and variances of populations were equal. A Tukey HSD test was used post hoc to identify the source of any differences identified in the one-way ANOVA test.

A one way ANOVA test indicated a statistically significant difference in the Care coordination: Total score scale between groups, F(3, 366) = 5.60, p = 0.0009. The largest significant difference was between participants in the Aged 18 to 44 subgroup (median = 39.36, IQR = 9.59), and participants in the Aged 65 or older subgroup (median = 32.74, IQR = 10.26, p = 0.0004).

The **Fear of Progression** questionnaire measures the level of anxiety people experience in relation to their conditions. On average, participants in the **Aged 18 to 44** subgroup had a higher score compared to **Aged 65 or older**, however, both groups had moderate levels of anxiety.

Table 8.33: Fear of progression total score by age summary statistics and one-way ANOVA

Fear of progression	Group	Number (n=370)	Percent	Mean	SD	Source of difference	Sum of squares	dF	Mean Square	f	p-value
Total score	Aged under 18	87	23.45	37.44	10.65	Between groups	1749.00	3	583.00	5.60	0.0009*
	Aged 18 to 44	120	32.35	39.36	9.59	Within groups	38137.00	366	104.20		
	Aged 45 to 64	105	28.30	36.61	10.48	Total	39886.00	369			
	Aged 65 or older	58	15.63	32.74	10.26						

Table 8.34: Fear of progression total score by age one-way post hoc Tukey HSD test

Fear of progression	Group	Difference	Upper	Lower	p adjusted
Total score	Aged 18 to 44 - Aged under 18	1.92	-1.79	5.63	0.5400
	Aged 45 to 64 - Aged under 18	-0.83	-4.65	2.99	0.9441
	Aged 65 or older - Aged under 18	-4.70	-9.16	-0.23	0.0350*
	Aged 45 to 64 - Aged 18 to 44	-2.75	-6.27	0.77	0.1842
	Aged 65 or older - Aged 18 to 44	-6.62	-10.83	-2.40	0.0004*
	Aged 65 or older - Aged 45 to 64	-3.87	-8.18	0.44	0.0962

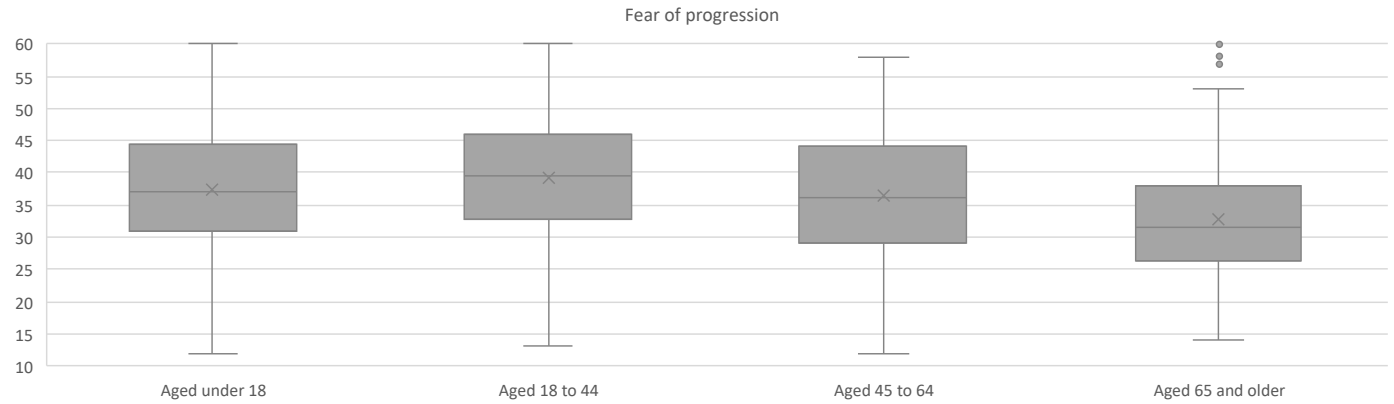


Figure 8.19: Boxplot of Fear of progression total score by age

Fear of progression by education

Comparisons were made by **education** status, between those with trade or high school qualifications (n=177, 48.90%), and those with a university qualification (n=185, 51.10%).
Assumptions for normality and variance were met, a two-sample t-test was used.

No significant differences were observed between participants by **education** for any of the Fear of progression scales.

Table 8.35: Fear of progression total score by education summary statistics and T-test

Fear of progression	Group	Number (n=362)	Percent	Mean	SD	T	dF	p-value
Total score	Trade or high school	177	48.90	37.54	10.90	0.76	360	0.4479
	University	185	51.10	36.70	9.98			

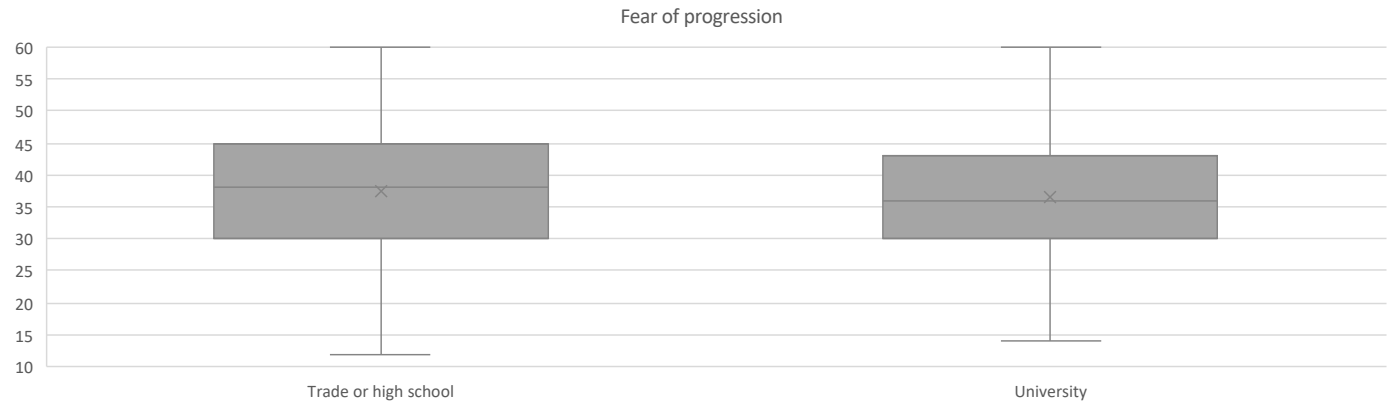


Figure 8.20: Boxplot of Fear of progression total score by education

Fear of progression by location

The **location** of participants was evaluated by postcode using the Australian Statistical Geography Maps (ASGS) Remoteness areas accessed from the Australian Bureau of Statistics. Those living in regional or remote areas (n=105, 28.38%) were compared to those living in a metropolitan area (n=265, 71.62%).

Assumptions for normality and variance were met, a two-sample t-test was used.

No significant differences were observed between participants by **location** for any of the Fear of progression scales.

Table 8.36: Fear of progression total score by location summary statistics and T-test

Fear of progression	Group	Number (n=370)	Percent	Mean	SD	T	dF	p-value
Total score	Regional or remote	105	28.38	37.19	10.23	0.12	368	0.9063
	Metropolitan	265	71.62	37.05	10.48			

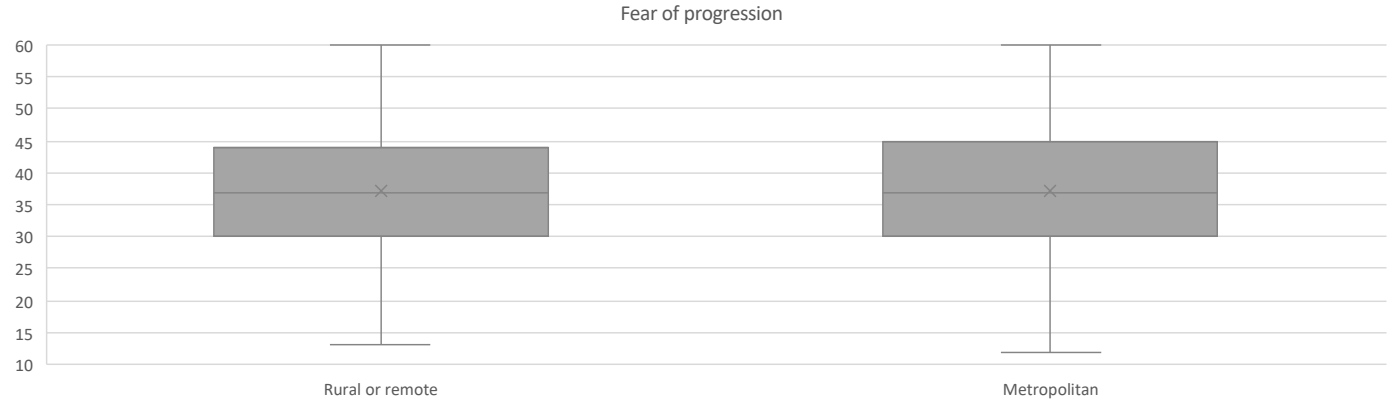


Figure 8.21: Boxplot of Fear of progression total score by location

Fear of progression by socioeconomic status

Comparisons were made by **socioeconomic status**, using the Socio-economic Indexes for Areas (SEIFA) (www.abs.gov.au), SEIFA scores range from 1 to 10, a higher score denotes a higher level of advantage. Participants with a mid to low SEIFA score of 1-6 (n=185, 50.00%) compared to those with a higher SEIFA score of 7-10 (n=185, 50.00%).

Assumptions for normality and variance were met, a two-sample t-test was used.

No significant differences were observed between participants by **socioeconomic status** for any of the Fear of progression scales.

Table 8.37: Fear of progression total score by socioeconomic status summary statistics and T-test

Fear of progression	Group	Number (n=370)	Percent	Mean	SD	T	dF	p-value
Total score	Mid to low status	185	50	37.71	10.46	1.15	368	0.2527
	Higher status	185	50	36.47	10.32			

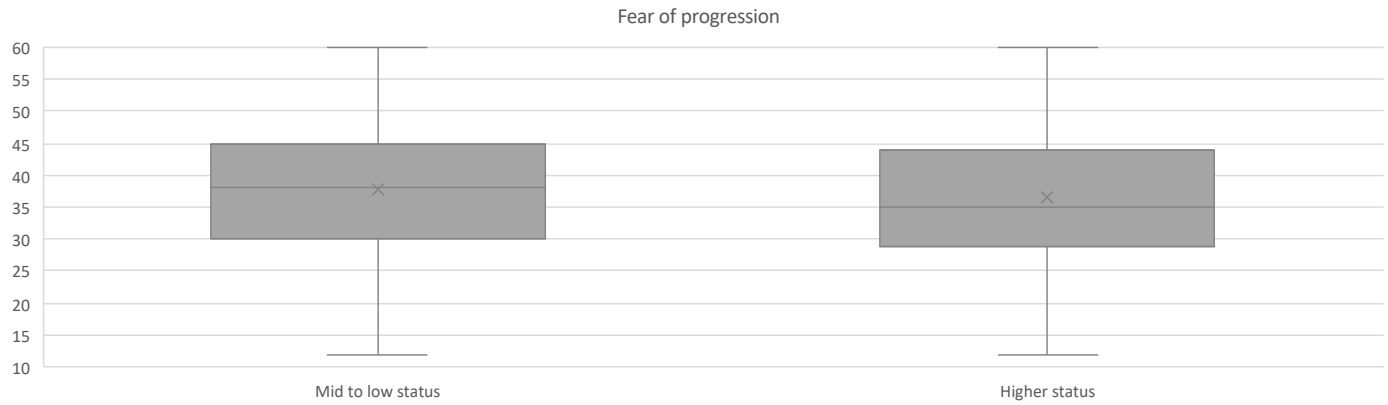


Figure 8.22: Boxplot of Fear of progression total score by socioeconomic status

Fear of progression individual questions

On average, participants scored in the "**Never**" range for the following questions: "Anxious if not experiencing any side effects think it doesn't work" (median=1.00, IQR=1.00).

On average, participants scored in the "**Sometimes**" range for the following questions:"Becomes anxious thinking that disease may progress" (median=3.00, IQR=1.00), "Is nervous prior to doctors appointments or periodic examinations" (median=3.00, IQR=2.00), "Afraid of pain" (median=3.00, IQR=2.00), "Has concerns about reaching professional and/or personal goals because of illness:" (median=3.00, IQR=2.00), "When anxious, has physical symptoms such as a rapid heartbeat, stomach ache or agitation" (median=3.00, IQR=2.00), "The possibility of relatives being diagnosed with this disease disturbs participant" (median=3.00, IQR=3.00), "Is disturbed that they may have to rely on strangers for activities of daily living" (median=3.00, IQR=2.00), "Afraid of severe medical treatments during the course of illness" (median=3.00, IQR=2.00), "Worried that treatment could damage their body" (median=3.00, IQR=2.00), "Worried about what will become of family if something should happen to participant" (median=3.00, IQR=3.00), "The thought that they might not be able to work due to illness disturbs participant" (median=3.00, IQR=3.00), "If a treatment and it is working well (limited side effects, no progression of disease), worry what will happen if treatment stopped" (median=3.00, IQR=3.00).

On average, participants scored in the "**Often**" range for the following questions:
"Worried that at some point in time will no longer be able to pursue hobbies because of illness" (median=4.00, IQR=2.00).

Table 38: Fear of progression by questions

Fear of progression (n=370)	Mean	SD	Median	IQR	Average response
Becomes anxious thinking that disease may progress	3.46	1.16	3	1.00	Sometimes
Is nervous prior to doctors appointments or periodic examinations	2.94	1.25	3	2.00	Sometimes
Afraid of pain	2.94	1.23	3	2.00	Sometimes
Has concerns about reaching professional and/or personal goals because of illness:	3.27	1.37	3	2.00	Sometimes
When anxious, has physical symptoms such as a rapid heartbeat, stomach ache or agitation	2.80	1.27	3	2.00	Sometimes
The possibility of relatives being diagnosed with this disease disturbs participant	2.77	1.44	3	3.00	Sometimes
Is disturbed that they may have to rely on strangers for activities of daily living	3.07	1.34	3	2.00	Sometimes
Worried that at some point in time will no longer be able to pursue hobbies because of illness	3.39	1.33	4	2.00	Often
Afraid of severe medical treatments during the course of illness	3.02	1.30	3	2.00	Sometimes
Worried that treatment could damage their body	2.99	1.31	3	2.00	Sometimes
Worried about what will become of family if something should happen to participant	3.29	1.42	3	3.00	Sometimes
The thought that they might not be able to work due to illness disturbs participant	3.16	1.55	3	3.00	Sometimes
If a treatment and it is working well (limited side effects, no progression of disease), worry what will happen if treatment stopped	2.63	1.39	3	3.00	Sometimes
Anxious if not experiencing any side effects think it doesn't work	1.76	1.11	1	1.00	Never

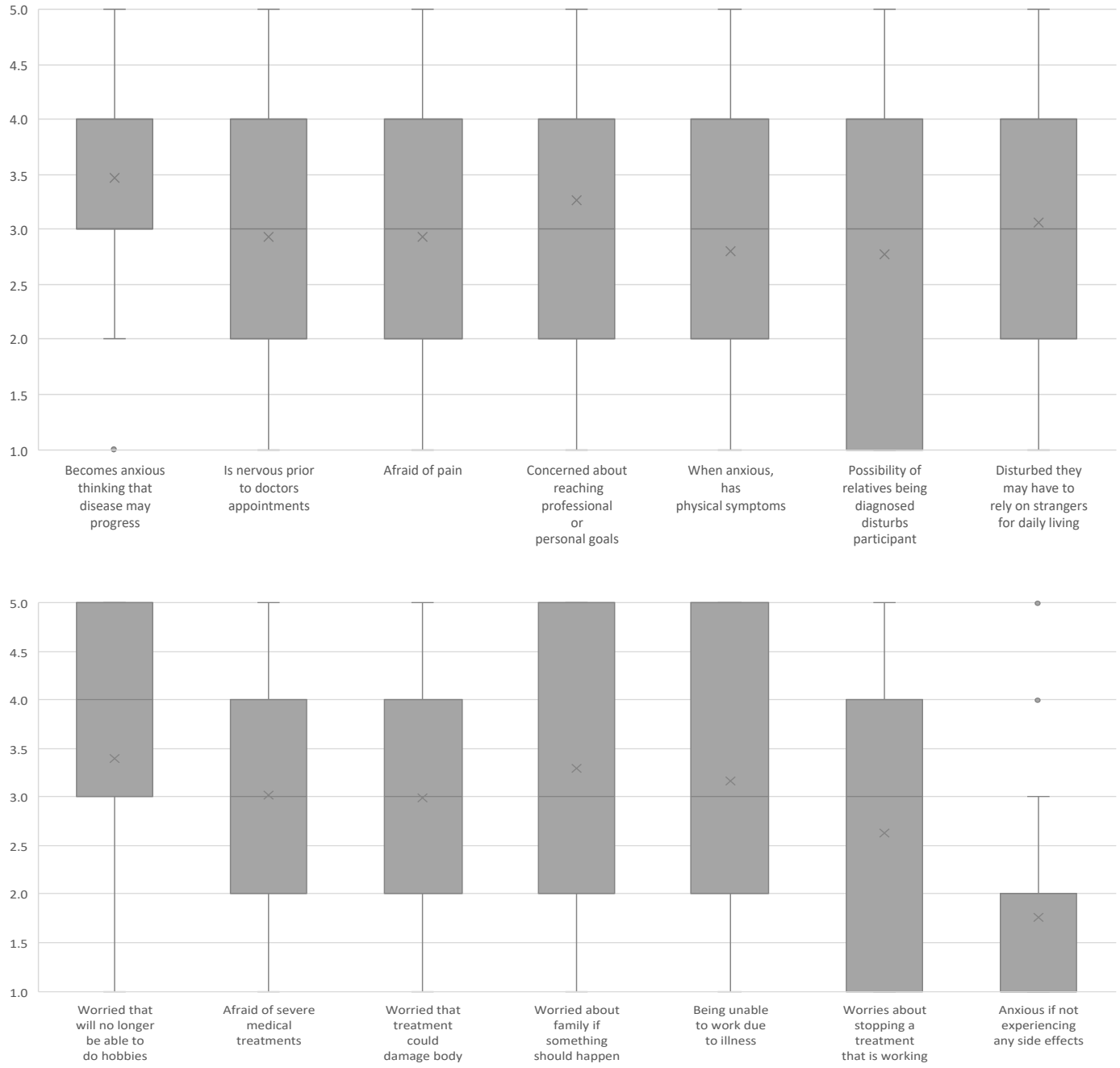


Figure 23: Box plot fear of progression by question

Section 9

Expectations and messages to decision-makers

Section 9: Expectations of future treatment, care and support, information and communication

Expectations of future treatment

Participants were asked in the structured interview what their expectations of future treatments are. The most common responses were that future treatment will be more affordable (36.57%), be more effective and/or targeted (personalised) (21.39%) and will include having choice (including availability and accessibility) and transparency/discussions in relation to treatment options (pathways) (17.66 %). Other themes included have fewer or less intense side effects or more discussion about side effects (16.92%), involve more clinical trials (including to access new technologies and treatments and funding) (14.43%), be easier to administer or able to administer at home or be less invasive (12.94%) and involve a more holistic approach (11.19%).

Expectations of future information

Participants were asked in the structured interview if there was anything that they would like to see changed in the way information is presented or topics that they felt needed more information. The most common responses were that future information will be more accessible or easy to find (23.88%), and more details about disease trajectory and what to expect (12.19 %). Other themes included use information to help to inform the community and decision-makers about their condition (raise awareness) (11.94%), provide more details on subgroups and specific classifications of their condition (10.20%), and be easier to understand (7.96%). There were 58 participants (14.43%) who were satisfied with the information they received.

Expectations of future healthcare professional communication

Participants were asked in the structured interview what they would like to see in relation to the way that healthcare professionals communicate with patients. The most common expectations for future healthcare professional communication were that communication will include health professionals with a better knowledge of the condition (21.89%), be more empathetic (17.16%), and satisfied with experience (17.66 %). Other themes included be more transparent and forthcoming (10.95%), include listening to the patient (9.95%), allow people more time to meet with their clinician (9.70%), and include a multidisciplinary and coordinated approach (9.45%).

Expectations of future care and support

Participants were asked in the structured interview whether there was any additional care and support that they thought would be useful in the future, including support from local charities. The most common expectation for future care and support was that it will include more access to support services (22.89%), will include a multidisciplinary and coordinated approach (14.68 %) and will include specialist clinics or services where they can talk to professionals (in person, phone, online) (13.93%). Other themes included ill include being able to connect with other patients through peer support (support groups, online forums) (11.69%), will include health professionals with a better knowledge of the condition (9.70%), and will include practical support (home care, transport, financial) (7.96%). There were 32 participants (7.96%).) that were satisfied with their care and support and had no particular comment.

What participants are grateful for in the health system

Participants were asked in the structured interview what aspects of the health system that participants are grateful for. The most common responses were that participants were grateful for low cost or free medical care through the government (40.34%) – with the related theme os included timely access to treatment (11.36%). Other themes included being grateful for healthcare staff (including access to specialists) (35.23%), and the entire health system (18.47 %).

Values in making decisions

Participants were asked to rank what is important for them overall when they make decisions about treatment and care, where 1 is the most important and 8 is the least important. A weighted average is presented in the figure below. With a weighted ranking, the higher the score, the greater value it is to participants.

The most important aspects were “"How safe the medication is and weighing up the risks and benefits"”, and “"The severity of the side effects"”. The least important were “"Ability to follow and stick to a treatment regime"” and “"The ability to include my family in making treatment decisions"”.

Values for decision makers

Participants were asked to rank what is important for decision-makers to consider when they make decisions that impact treatment and care, where 1 is the most important and 5 is the least important. A weighted average is presented in the figure below. With a weighted ranking, the higher the score, the greater value it is to participants.

The most important values were “Quality of life for patients”, and “All patients being able to access all available treatments and services”. The least important was “Economic value to government and tax payers”.

Time taking medication to improve quality of life

Participants were asked in the online questionnaire, how many months or years would you consider taking a treatment, provided it gave you a good quality of life, even if it didn’t offer a cure.

The majority of participants (n = 88, 33.72%) would use a treatment for more than ten years for a good quality of life even if it didn’t offer a cure.

Most effective form of medicine

Participants were asked in the online questionnaire, in what form did they think medicine was most effective in.

There were 30 participants (11.11%) that thought that medicine delivered by IV was most effective, 49 participants (18.15%) thought that pill form was most effective, and 74 participants (27.41%) that thought they were equally effective. There were 117 participants (43.33%) that were not sure.

Messages to decision-makers

Participants were asked, “If you were standing in front of the health minister, what would your message be in relation to your condition?” The most common messages to the health minister were the need for timely and equitable access to support, care and treatment (25.87%), the need for more research investment (17.91%), and to help raise community awareness (14.43 %). Other themes included to invest in clinical trials (13.18%), that treatments need to be affordable (10.20%), and to invest in health professionals development (8.96%).

Expectations of future treatment

Participants were asked in the structured interview what their expectations of future treatments are. The most common responses were that future treatment will be more affordable (36.57%), be more effective and/or targeted (personalised) (21.39%), and will include having choice (including availability and accessibility) and transparency/discussions in relation to treatment options (pathways) (17.66 %). Other themes included have fewer or less intense side effects or more discussion about side effects (16.92%), involve more clinical trials (including to access new technologies and treatments and funding) (14.43%), be easier to administer or able to administer at home or be less invasive (12.94%), and involve a more holistic approach (11.19%).

Future treatment will be more affordable

I'd like to see the cost reduce cuz yeah I don't have a healthcare card so the scripts are over 6 times the cost they would be if I did have a healthcare card. So I would like to see I guess if they could be cheaper for people that don't have access to a healthcare card, but doubt that and that's it.
Participant 013_2023AUORC

Well, any new treatment that helps?...That isn't cost prohibitive, it would be welcome. But most people that have Scleroderma are limited in their income and so you know, these new treatments, like I said, there was that treatment they wanted to put me on that was $46,000. Well, you could sell your house, but how long does that, how long does 40,000 last?
Participant 021_2023AUDIS

With regards, I mean I feel very fortunate that I'm, I was approved for the Humira and the fact that it for me fortunately it's working better than I expected it to and I'm you know, I'm, I'm also fortunate that I'm in a financial position that I can afford to pay for it, whereas there are a lot of people out there that would have to. Would have to go without and we're lucky in that you know once it's approved through Medicare we only paid the dispensary cost. So you know in that in that regard you know there are aspects of I, I think I would really like to see.
Participant 005_2023AUDSK

Future treatment will be more effective and/or targeted (personalised)

It'd be good to see, in terms of medications, because I'm not sure if that's the only kind of treatment you're talking about, but in terms of medications, it would be reassuring if there were some that weren't just off-label use because nearly all the medications that we have that, that people with Ehlers-Danlos talk about is nearly all off-label use.
Participant 001_2023AUDPA

So at the moment for instance speech, allied speech therapy, the wait list to get on to someone that actually understands this condition because there is specialist ways to treat but it's not just a general speech therapist that's required needs to understand the condition because' they have palate differences. So it's not just a regular speech pathologist that can do this they have to understand the communication disorders that go along with it as well. So that speech is probably the one of the biggest where we need more understanding.
Participant 025_2023AUDPA

But I need some improvement. I mean, as I said, since I've been seeing my local doctor, well, the lumps kind of, he has put me take the job and put me on different things all the time. At the moment I'm not taking anything regularly except for, I mean I take generic, I take painkillers, zink and vitamin C and that kind of thing. But apart from that I'm not taking anything else.
Participant 023_2023AUDSK

Future treatments will include having choice (including availability/accessibility) and transparency/discussions in relation to treatment options (pathways)

Yeah, yeah. Look, I suppose cost, cost is certainly a, a challenge or a barrier for some I think access to information about. What the options are and what the possible side effects of each pathway so that you can make informed decisions about what you're willing to, what you're willing to risk, but also kind of what your probabilities of success are. Yeah, I think, I think more information and more knowledgeable practitioners.
Participant 007_2023AUDSK

So for new treatments, I would like to see obviously you know, if they could be subsidized. So the cost, the way that they are administered, you know, to be frank with you, like if it was an injection I wouldn't even care, at least it's something. So if it was oral or injection, I it wouldn't matter. You know, for something like really invasive, then yes, that would matter. But I couldn't imagine that being the case. I would like for health professionals, so for example, our hospital care team, to be aware of it and to talk about it and to offer it. Something that can be given in the home would be really important as well, just to not impact daily life.
Participant 021_2023AUORC

So I'd like to see reduced side effects for most in the sense of a number of the treatments that are available to NAME have a significant side effect on reproduction health. She's nine years old and you know, not in the brain space right now to make be making decisions that may impact her in 10, 15 years, 20 years time when she may regret a decision that I've made or she actively made at nine years old that that might have an impact on the life that she wishes to have for herself. So, so a more available access to what possible side effects there are for an appropriate age group. So when you go and get your eyes done, they have a little kid booklet that tells you all about what they're going to do. Why isn't there any? Very simple, very direct communication in the drugs in the studies that target the age the under 12 age group if that's the demographic of the study or the treatment that's going to access it. Like there's lots of mumbo jumbo for parents to decide on but there's, there's no active information for NAME's brain capacity or.
Participant 080_2023AUDIS

Future treatments will have fewer or less intense side effects/more discussion about side effects

So it would be nice if new treatments also considered more seriously. That the lived experience of a side effect is different perhaps, to the medical definition of a side effect. Participant 024_2023AUDIS

Cost is definitely a big thing. The side effects to meds is also a thing like, how one medication could make another medication play up, or being on steroids. The weight gain, the reflux, the insomnia, how all that affects everything. Yes, side effects and cost are the main things I'd like to change, and the wait list. Like waiting for everything, because like in that time while I'm waiting, I'm declining more and more.
Participant 023_2023AUDIS

So they need to be low cost, they need to have zero side effects and then they can most importantly make a difference like in terms of a substantial improvement like. Yeah, I think for me, I don't want something that might, you know, give me 10% less fatigue or even 20% fatigue because fatigue is fatigue. Like, if you still got 80% fatigue, which means you can't work and you can't do a lot of things, it's, you know, it just might mean I get one extra shower a week or something. Like, really, in this game of things, it's not enough.
Participant 010_2023AUDIS

Future treatment will be easier to administer and/or able to administer at home and/or less invasive

If they had advanced that way that I could take a tablet and not have to have the amount of needles that I do. Yes, that would be good. Other than that, I don't really know. What else? If they had a cure, it would be great too. [laughs]
Participant 006_2023AUDNS

Well, I would love an extended-release treatment or something that we only have to sort of give once or twice a day as opposed to every six hours they do have. They do actually have that available overseas. It's a 12 hour release, but it', it's way too expensive for them to which I can understand, it's just it's not cost effective for them to get that here. But yeah, I guess yeah that would be the main one. Just I think. Yeah. NAME tends to sleep through. She sleeps through it when I give it to her now and the night and whatnot. But just I think. I don't know, as she gets older, it'd be nice for her not to get have to get up herself to do it, you know, just take it once and forget about it, yeah.
Participant 015_2023AUORC

Future treatment will involve more clinical trials (including to access new technologies and treatments and funding)

Cost isn't an issue, I don't think. I think if you're looking at your life, you know it cost I don't think some a crazy issue as in we live in a pretty good place in Australia and we've got really good clean hospitals, clean places to live, all that kind of stuff. So yeah, that's not an issue. I think it's more. I think we need sort of more research on the…the rare forms of things that and this is this is unbiased obviously because you know…I want someone to bloody find what's wrong with you know you know you know how to kill it or how to how to deal with it for the rest of my life. So

yeah, my main concern would obviously be more research being done … scenarios that obviously lead to, you know, clinical trials or positive outcomes for patients later down the track kind of things, yeah.
Participant 024_2023AUORC

There's an incredible drug that treats cystic fibrosis that's been developed. But unfortunately it's not approved for youth, but under the age of 6 years old. So I'd like to see it approved down to the age of two. It's called Trikafta, so that I'm really putting a lot of effort into trying to advocate for Trikafta from 2 to 5 year olds at the.
Participant 029_2023AUORC

Yeah, probably what I'd like to see particularly within Australia is, is probably more research so that we get a better understanding of. Why the condition occurs? Yeah, I guess that would be that would be probably my, my ultimate, you know, I think. You know not, not all medication works for everyone. So we can bring out a new medication and that may work for me, but it may not work for someone else. So I think ultimately for me, I would like to see more research into the condition and so that we can get a better understanding of why the condition is impacting so many people so.
Participant 001_2023AUDSK

Future treatment will involve a more holistic approach

I would like to think that there's not a pill you have to take every day. Like I think there's something else that they can come up with and so maybe like less frequent medication like a tab, a needle or like a vaccination kind of thing, you know, a bit more like annually or biannually or something. I think that there needs to be more support, like more checking in, like whether it's a phone call or face to face.
Participant 078_2023AUDIS

PARTICIPANT: Just holistic and family based care. Just understanding that a child is not just, you know, a pair of eyes or pair of ears or you know.
INTERVIEWER: But like how?
PARTICIPANT: All of those things interact with each other, yeah.
Participant 018_2023AUDPA

What I would most like to see is a setup like the, the at the Children's Hospital, a specialized center where you get to see everybody in one day rather. So you still have you and that they do a like a holistic plan so they work out the you know I need extra physio or extra OT and then you go off to your community person and yeah. So yeah, there's no consistency between the States and that's what I'd like is a national approach that it doesn't matter where you are, if you go somewhere you're going to receive the same standard and same type of assistance.
Participant 026_2023AUORC

Future treatments will allow for a normal life/quality of life

Look, I think a reduction in side effects that he experiences, I guess as a result of his condition more than anything. So I mean if treatment's able to help give him some, you know, improved quality of life because I guess there's only so much we can do from a behavioural support you know, developmental sort of side of it. So if there's something that can help him to be able to get a bit of clarity and think a bit more clearly and, you know, be less impulsive and be calmer, happier, more content within himself, that would be fantastic. Participant 031_2023AUDPA

Reduced of potential side effects would be good, but the biggest thing would be accessibility and cost. Having it so you don't have to drive to a metropolitan area like Sydney or Melbourne, which are five and six hours away respectively from where I live, and be affordable and be covered so you're not out of pocket hundreds of dollars a week or a month. To have a quality of life and contribute to the community.
Participant 012_2023AUDSK

Well, cost is obviously a big thing for me. I think anything that greatly improves someone quality of life for a condition like this shouldn't cost an arm and a leg and I think it should be readily available for anyone to be able to afford. But in terms of administering and all that kind of stuff, I don't have any issues with the way that I'm currently managing, so I can't think of anything extra that I would like to sort of change, no?
Participant 027_2023AUDSK

Future treatments are important but we cannot ignore awareness and education

I think it would be more about trying to a greater access to health professionals who have knowledge about CHARGE syndrome. I think probably the main issue I've found is finding people who are experienced with this syndrome and know what to do? Yeah. I think the main thing would be more education and

access to informed healthcare providers. Yeah. I can't think of anything else around that.
Participant 089_2023AUENM

I guess more probably that you like not necessarily that way, but supportive groups for people who are experiencing it. Because I do sometimes feel like you're not alone, but there's only a, you know, small amount of people that understand what you're talking about. Whereas you know, for example, Down syndrome, you know, everybody sort of knows about it and you know what to do with it, I guess.
Participant 034_2023AUDPA

Because when you're sort of applying for therapies or like applying for NDIS because it's not a well known condition, they're just like, well, what does she need, doesn't know she needs or there's not a formula for them to go back to, to look at all this is what we do in this situation and this is the access you would need to dismiss this and this. It's like well what do you need without yeah what, what is the there's that lack of understanding some more recognition of the conditions and that so that there are less sort of less explanation of all the ins and outs and what, what it affects and how it works and, and things like that. So that aware as well.
Participant 017_2023AUDPA

Future treatment will involve a multidisciplinary approach (communication)

Yeah. Yeah, nothing that I can think of. The only thing I'd like to see and I believe it's already started to happen and is that that they've got, they're following like the American path where they've now got clinics who specialize in 22Q11 and they have all the specialists on board in one place. And it's all about, it's all about the actual diagnosis of 22Q,11, not about the individual issues that come up even though they're all traded together. It's, it's sort of looking at it I think holistically that's really the only thing I'm glad that I'm actually seeing now even though PATIENT being her age is a little bit past all that because she doesn't have to have you know more diagnostics and all that. It's good that it's coming through like that now for the younger kids through their per Children's Hospital, so, right. Yeah.
Participant 024_2023AUDPA

Future treatment will be curative

I would like to see a cure rather than treatment because I don't understand the extent of how much the treatments are effective? I know that there's two choices for me, two or three choices for me at the moment. But other than that, I guess there's obviously different outliers with people's impact to it. So I wouldn't, I would have actually said probably cure because I think we've got enough treatments out there. I don't know what part you're trying to solve for in it .
Participant 004_2023AUORC

PARTICIPANT: [laughs] A cure.
INTERVIEWER: A cure? Yes, that's what we want.
PARTICIPANT: I read about studies like they had been able to reverse, so not only stop but reverse the progression of fibrosis in mouse models. Again, I want that for me. I want to be able to bend and move again, because that's the problem…all the arteries into my lungs have gone fibrotic, hardened and thickened and narrowed. A cure, I'm going to the top.
Participant 008_2023AUDIS

Table 9.1: Expectations of future treatment

Expectations of future treatments	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
More affordable/remain affordable	147	36.57	24	35.82	33	40.74	24	25.26	17	53.13	38	40.00	11	34.38	103	38.43	44	32.84	106	36.05	39	36.79
More effective / targeted	86	21.39	4	5.97	18	22.22	26	27.37	5	15.63	32	33.68	1	3.13	69	25.75	17	12.69	64	21.77	22	20.75
Include having choice, and transparency / discussions in relation to treatment options	71	17.66	16	23.88	19	23.46	10	10.53	11	34.38	3	3.16	12	37.50	49	18.28	22	16.42	52	17.69	19	17.92
Fewer or less intense side effects / more discussion about side effects	68	16.92	8	11.94	23	28.40	18	18.95	7	21.88	6	6.32	6	18.75	52	19.40	16	11.94	53	18.03	14	13.21
More clinical trials / access new technologies and treatments / funding)	58	14.43	7	10.45	8	9.88	17	17.89	3	9.38	18	18.95	5	15.63	33	12.31	25	18.66	46	15.65	12	11.32
Easier to administer / able to administer at home / less invasive	52	12.94	10	14.93	11	13.58	8	8.42	7	21.88	6	6.32	10	31.25	38	14.18	14	10.45	39	13.27	12	11.32
More holistic	45	11.19	15	22.39	10	12.35	5	5.26	0	0.00	12	12.63	3	9.38	24	8.96	21	15.67	33	11.22	12	11.32

Expectations of future treatments	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
More affordable/remain affordable	147	36.57	31	31.96	50	38.17	39	34.21	27	45.00	76	38.38	66	33.67	37	33.33	110	37.80	68	34.00	79	39.11
More effective / targeted	86	21.39	13	13.40	29	22.14	32	28.07	12	20.00	47	23.74	39	19.90	26	23.42	60	20.62	44	22.00	42	20.79
Include having choice, and transparency / discussions in relation to treatment options	71	17.66	17	17.53	25	19.08	25	21.93	4	6.67	33	16.67	38	19.39	19	17.12	52	17.87	38	19.00	33	16.34
Fewer or less intense side effects / more discussion about side effects	68	16.92	10	10.31	27	20.61	24	21.05	7	11.67	31	15.66	35	17.86	16	14.41	52	17.87	36	18.00	32	15.84
More clinical trials / access new technologies and treatments / funding)	58	14.43	22	22.68	14	10.69	17	14.91	5	8.33	31	15.66	27	13.78	14	12.61	44	15.12	32	16.00	26	12.87
Easier to administer / able to administer at home / less invasive	52	12.94	12	12.37	18	13.74	12	10.53	10	16.67	29	14.65	22	11.22	8	7.21	44	15.12	24	12.00	28	13.86
More holistic	45	11.19	12	12.37	11	8.40	15	13.16	7	11.67	24	12.12	19	9.69	16	14.41	29	9.97	23	11.50	22	10.89

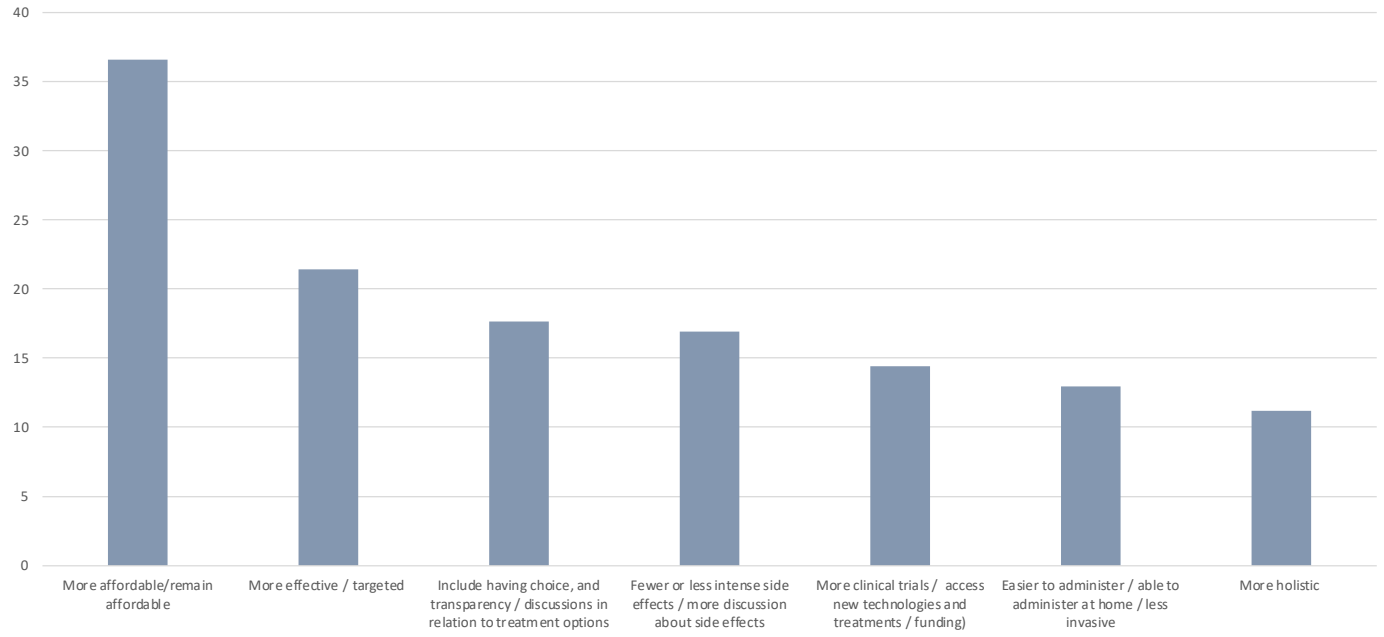


Figure 9.1: Expectations of future treatment

Table 9.2: Expectations of future treatment – subgroup variations

Expectations of future treatments	Reported less frequently	Reported more frequently
More affordable/remain affordable	Diseases of the nervous system Developmental anomalies Other rare condition Endocrine, nutritional or metabolic diseases Aged 65 plus	Diseases of the skin Endocrine, nutritional or metabolic diseases Diseases of the skin Other rare condition
More effective / targeted	Endocrine, nutritional or metabolic diseases	Diseases of the immune system
Include having choice, and transparency / discussions in relation to treatment options		
Fewer or less intense side effects / more discussion about side effects		Other rare condition
More clinical trials / access new technologies and treatments / funding)	Diseases of the skin	Developmental anomalies
Easier to administer / able to administer at home / less invasive		
More holistic		Diseases of the skin

Expectations of future information

Participants were asked in the structured interview if there was anything that they would like to see changed in the way information is presented or topics that they felt needed more information. The most common responses were that future information will be more accessible or easy to find (23.88%), and more details about disease trajectory and what to expect (12.19 %). Other themes included use information to help to inform the community and decision-makers about their condition (raise awareness) (11.94%), provide more details on subgroups and specific classifications of their condition (10.20%), and be easier to understand (7.96%). There were 58 participants (14.43%) who were satisfied with the information they received.

Future information will be more accessible/easy to find

I want information about it or the information from A-Z. Everything that I need to know, everything that the patient need to know. I would love to know about this. At least I can read about it. At least I can know and understand it more better.
Participant 001_2023AUORC

It's not like we haven't got any information, it was just what the doctor told us. I think if there was something that could be provided a point of diagnosis in writing that would be really helpful or somewhere to go and look for information, so maybe be directed to a website or something. Because again, you know, you kind of have to do all of that yourself. I think information being available and relevant to the Australian community would be really important.
Participant 021_2023AUORC

More of it and more accurate information and when you read stuff online, every site that you go to, none of them really marry up. So like some really formal type of information. That you that's readily accessible to people with the disease and certainly information on where you can get support from other people that also have the disease and others that understand the disease.
Participant 005_2023AUDSK

No particular comment - satisfied with information

All in terms of information, I think I got good information, so I don't think I want much to change consonant information.
Participant 006_2023AUORC

No. I was pretty happy. I'm pretty happy with everything that I've I've been able to get hold of.
Participant 010_2023AUDPA

No, I think I've always been able to find sort of the information that I needed, but I think that that's partly because most of it's on this website that was created by another parent and her child was the disease. So it's really focused on what a carer would want to see, which is probably different to if you're getting all your information from like a, you know, like a government website or something. Yeah, yeah.
Participant 079_2023AUDIS

Future information will provide more details about disease trajectory and what to expect

I think just in general there's a lack of information with HS and that would be really handy. Like even going into the surgery, they explained what the procedure was, but there was no information that I could look up and I explained this imagery is I find images really important. Not just not images. How do we how do we say this? All right. So I had the surgery and the doctors told me, Yep, we're just gonna go through we're gonna cut them open. And we're just going to lay them flat. You'll come back in in a week and we'll look at you. And one of the things that I noticed is because I've got dark skin, I was like, I couldn't tell if it was pus, but there was white all inside my in these craters. And I didn't know what they were. And so when I went to the nurse, she was just like, oh, that's just the layer of skin and it's just a little bit because of your pigment, it's just really obvious that it's white. And I was just like, 'oh, interesting', but this information wasn't widely available. There was nothing around I didn't know. I tried to look up what I was supposed to be looking at, and I couldn't find that information. And so when I ended up seeing the HS Connect roomed care, everything made sense. Just having information, obviously it's out there, it just needs to be distributed better or easily found.
Participant 026_2023AUDSK

Some information was hard to source and even when we sourced it, it was a little bit difficult to understand how it actually affect us in a day-to-day life and some of the side effects would be written up in obviously medical language, which, you know, you think makes sense at the time. But then like one of the surgeries we did, they said, Oh, we'll do this particular surgery and it means she'll never vomit again, which was ideal

for us because she had such terrific reflux. But one of the side effects was afterwards is that they had to tighten her stomach so much that she actually couldn't drink water. So that was one of those things that we were like, oh, apparently we had it known. So I think the more the everyday life terms that a lot of parents sort of miss out when they're reading a lot of the information because they can't figure out how it actually affects their life.
Participant 032_2023AUDPA

Future information will provide more details on subgroups and specific classifications of their condition

No. I think being pretty good presented. I think more so it's I think probably the thing I've seen the most is that because I've had this disease longer, I'm dismissed because I'm a longer patient and possibly at risk. There's more trials today for those that are in the newest stages, which I can completely understand because-- Then in equal side of things, I'm probably up to the stage of somebody that's in their six-- They don't look at the overall effects on my body as opposed to a new patient. I guess and age-wise, I suppose it's the age as well.
Participant 001_2023AUDIS

More emphasis straight up on how different everybody can be, even though we're similar. What I find fascinating is how different everybody is, but at the same time, a lot of the same modalities and approaches and management techniques seem to work for all of us. That would be a really, really big one.
Participant 041_2023AUDPA

Future information will help to inform the community and decision-makers about their condition (raise awareness)

I suppose I'd like to see more people understand what the condition cuz they just think you've got a bit of a pain in your neck. And get very knowing when they say, oh, you look really well, yeah, it's on time. That's not the problem. Yeah, that's very that can be very annoying.
Participant 005_2023AUDNS

Oh, just more public awareness and what I said, more in the lower levels. By lower levels, I mean nurses, GP's, clinics, more awareness of the condition and more public awareness. Like when I try to explain it, some people say, Oh yeah, that's Downs syndrome. And I said no, you know, everybody knows about Downs syndrome, everybody knows about this and that and the other, but nobody's ever heard it. DiGeorge. I'm forever correcting and explaining.
Participant 08_2023AUDPA

Future information will be easier to understand

I just think the KISS principle is the most important. Just keep it simple, stupid. Because I think when people are first diagnosed they're very overwhelmed with all the information. So I think the simpler the the brochures are, the better. And the more contact information for people who can offer the people who are diagnosed support you know the you know. Making sure people feel connected if they feel overwhelmed technically, in whatever way, is simplest.
Participant 010_2023AUORC

Very simple, very direct communication in the drugs in the studies that target the age the under 12 age group if that's the demographic of the study or the treatment that's going to access it. Like there's lots of mumbo jumbo for parents to decide on but there's there's there's no active information for NAME's brain capacity or appropriate. Participant 080_2023AUDIS

It's in jargon I can understand it but you know I don't understand. You know like you look at articles and you know the specific you know you know a particular specialty and of course you know I'll know different words but as, as a therapist but I don't understand all the medical stuff behind it because it's written at that level that specialist you know of that specialty. So these are very much just like research things are looked at because you know just to see whether there is a question I should be asking if you know what I mean like yeah yeah. Could this be because?...
Participant 038_2023AUDPA

Future information will provide more details about new treatments and/or trials

No. I'd like to know what research was happening. I have no idea, you know if there is anything new or if there is research happening that may impact me. It's just like there's void information on.
Participant 002_2023AUDNS

I'd love to see a up to date listing of you know, the types and the genes involved. And symptoms you know because some, there's some that are very, very rare and they are actually life threatening. So it would

be nice and just to so because at the moment they just say 'Oh well CMT as a blanket', but there's you know four different sub subtypes with lots of. There's four different types with lots of subtypes below each one. Sit down please. So that and updated research. And you know, I'd like to see information of trials available, but including worldwide, you know, the opportunity to partake in. To be aware that a trial is about to happen and have the opportunity to partake if we choose to.
Participant 026_2023AUORC

Future information will be more holistic (including emotional health)

Oh, I would love the international guidelines and to be accepted by the Australian medical community and adopted across the whole country so that we can get a uniformed approach. Instead of having this hotchpotch approach where oh we'll look at, we'll look at this. We'll look at speech today and now we'll look at immunology or we'll look at is we'll look at heart cardiac issues. We'll look at psychiatric. We need a, we need a uniformed allied health and, and clinicians coming together and working together, not just in their own discipline, but trying to understand that it, all the conditions are separate, but they all have to be managed together.
Participant 025_2023AUDPA

We've had lots of really good Zoom conferences through Scleroderma Victoria that have dealt with a lot of really good issues like continence and intimacy and movement. I'd like to see more on mental health and mental health support. I know that's an area for millions of people with COVID affected and all that sort of thing. I could cry just thinking about what's ahead of me. I think you can't always explain it to your relatives or your husband or whatever, independent person, but again, they need to have knowledge of scleroderma. Probably more a holistic approach, you see one doctor for one area, and as I said, they treat each symptom virtually individually
Participant 008_2023AUDIS

I think the stuff around intimacy and family support for others. When I started actually looking at that and seeking more information around that, it was like, oh, I thought I should have been thinking about this earlier. It's probably the stuff around quality of life rather than medical diagnosis and treatment. What other things can be useful for that for joy and happiness and all that thing, rather than just stopping degree disease progression and stuff.
Participant 026_2023AUDIS

Future information will provide more details about symptom and side effect control

Anyhow, emails or brochures, anything like that, or like meetings would be amazing and I'd love to see someone do something on diet because nothing-- when I was at the support group, nothing came up about diet and I'd love to know if there are things that can help, you know, improve joint pain or fatigue and really focus on that would be amazing.
Participant 007_2023AUDIS

Like I said, I think the information doesn't necessarily acknowledge the spectrum of impact. Yeah, It kind of talks about you know the bigger issues rather than you know when you've got a bunch of things at a milder level. So I would say that's an issue because anything that you see can be a little bit drastic and so it doesn't acknowledge the fact that these things also you know a lot of these symptoms pop up that but they're not necessarily at a clinical clinically significant level. But that doesn't mean that a parent wouldn't want to deal with them or that that that person who has that diagnosis wouldn't want to you know overcome that particular hurt or whatever it is. So I would say that's an issue and there is no one stop shop.
Participant 035_2023AUDPA

Future information will be in a variety of formats

I'm very old fashioned so I'm not into all the online and support chat groups online and all that sort of stuff. I'm the old….printed brochure, talking to people face to face, and I still think that's pretty important face to face, even if you are doing telehealth and all that, nothing really beats that one-on-one to emulate the conversation, to actually get you to think about what you want to ask the doctor. I find with telehealth you always forget something and you tend to go off on tangents, I've found.
Participant 007_2023AUORC

I think like I said, information needs to be presented in a more layman type access to. Information like handouts, little information sheets or would be good when you go to appointments and whatnot because you're given all this verbal information. How much do you actually retain once you walk out the door. So leaflets or where to access more information.
Participant 013_2023AUDSK

I think like little videos, they are extremely powerful and I don't know that I actually have ever seen like kind of medical stuff put into layman's terms in a little video. I could be wrong. They could exist, but I haven't seen them because you know, when you like something is explained to you through little pictures and a video as if they're talking to children almost. But they're not like it's for adults. But I don't know like I've I'm a very visual person and I feel like it's easier to take on information and maintain information when you've got like it explained to in very simple terms with like little pictures and stuff you can pause and like write down almost like a little lecture I guess would be a great way for information.
Participant 023_2023AUDPA

Future information will provide more details about the causes of their condition

What I'd really like to see is more research into why people get it, because I feel like there's no like, it just seems to be, you know, where as, as doctors, everyone…we're so, you know, we can make babies out of a test tube, but we can't work out why somebody has got this disease. You know, I think just more, yeah, more medical research into why and you know, why certain people get it and why other people don't. I don't know. That's what I would like to see.
Participant 017_2023AUDSK

Yeah, the psychological impacts and what to and signs and what to look out for. I the…the fact that it's more known that it's it, you know more, more females are affected by it, it makes it hard to find information out. Between how this affects people of the different sexes even the flare zones can be different and I'm not saying that one's got it well I think actually think women have it worse than, than men cause they've got more places to get it. But having said that it's like yeah it and, and more education more information on as you were saying initially the genome testing because I'd love to know. I'd love to. I would actually. That's where I'd like the treatment to go is to maybe you know, if there could be some genetic, some genetic modification to delete it from the genetic code.
Participant 009_2023AUDSK

Future information will be up-to-date/credible

Well, I must admit when I'm trying to digest something, I prefer it to be in hard copy than reading it online. I do read things online, but I tend to skim and I don't take it in as well as if I've got a hard copy that I can go back to and mull over. I guess being able to ask questions of people who are informed, not who are ill-informed is another thing, people who you actually trust to know the answers.
Participant 004_2023AUDIS

PARTICIPANT: Well, pretty much what like the underlying issue of the condition is our hair follicles under our skin, like that's what causes. Our condition. I mean, I want that to pass everywhere because all those doctors have misinformation saying we're overweight, like we don't shower enough and all this, blah blah, blah. Like I'm very hygienic. I had two showers a day only because recently I found only having one wasn't enough and I just felt gross still. But yeah.
INTERVIEWER: It sounds like you've got some solutions you're trying, which is good.
PARTICIPANT: Yeah, and like, they work for a little bit, but then, you know, go stop and change, see what works.
Participant 003_2023AUDSK

Future information will include the ability to talk to/access to a health professional

Well I think the information brochure that they put in, they put out is really is good. It's very fun [sarcastic tone] I guess it's just I guess I'll go back to… it's just the communication with your specialist and GP. Yeah just and regular communication not wait for your appointment.
Participant 019_2023AUDIS

PARTICIPANT: Yeah, no, look, not really, but like I said, NAME, that nurse that was at the clinic, the square during the clinic, once I did have my office. She was a great form of education. She was just absolutely wonderful and I think we need someone like her with her knowledge, yeah, to be available to help.
INTERVIEWER: That would be wonderful.
PARTICIPANT: Yeah, she, I really she did teach me a lot. And she, she, you know, she, she gave me a lot of like of where I could go and get all my dressings and my wound care, you know, she, she was like, I can't, you know, praise her enough.
Participant 015_2023AUDIS

Know if there was a dedicated clinic to complex care, I mean even multisensory care would cover a whole lot of rare diseases, which would have a whole lot of similar issues that families are facing. If there was a that's where the information you would have one hub, you know one hub, they would know the experts. They

could get that information for you. They could certainly help you on that pathway. There would be dedicated telehealth nurses coming out of that clinic who would be invaluable with the information that they would have access to. But I don't understand why that we don't have more clinics. We've got diabetic clinics. We've got bloody. What's the other one? Diabetes clinics. Sorry. You know, there's CP clinics. Why isn't there a multisensory impaired clinic? You know, when there's so much challenges for them in their early years to get to get forward, I just, you know, I don't need to take.
Participant 028_2023AUORC

Table 9.3: Expectations of future information

Expectations of future information	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
More accessible/easy to find/linkage to service	96	23.88	5	7.46	20	24.69	42	44.21	8	25.00	12	12.63	9	28.13	60	22.39	36	26.87	80	27.21	16	15.09
No particular comment - satisfied with experience	58	14.43	6	8.96	14	17.28	9	9.47	4	12.50	17	17.89	8	25.00	43	16.04	15	11.19	41	13.95	16	15.09
More details about disease trajectory and what to expect (incl. costs)	49	12.19	8	11.94	11	13.58	8	8.42	3	9.38	12	12.63	7	21.88	33	12.31	16	11.94	40	13.61	9	8.49
Used to raise awareness	48	11.94	4	5.97	10	12.35	9	9.47	7	21.88	14	14.74	4	12.50	35	13.06	13	9.70	41	13.95	7	6.60
More details on subgroups and specific classifications of their condition (including age-subgroups)	41	10.20	5	7.46	3	3.70	15	15.79	0	0.00	16	16.84	2	6.25	30	11.19	11	8.21	30	10.20	11	10.38
Easier to understand	32	7.96	5	7.46	5	6.17	2	2.11	5	15.63	8	8.42	7	21.88	21	7.84	11	8.21	21	7.14	10	9.43

Expectations of future information	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
More accessible/easy to find/linkage to service	96	23.88	28	28.87	25	19.08	32	28.07	11	18.33	51	25.76	45	22.96	24	21.62	72	24.74	42	21.00	54	26.73
No particular comment - satisfied with experience	58	14.43	13	13.40	20	15.27	13	11.40	12	20.00	26	13.13	31	15.82	19	17.12	39	13.40	28	14.00	30	14.85
More details about disease trajectory and what to expect (incl. costs)	49	12.19	12	12.37	17	12.98	14	12.28	6	10.00	16	8.08	32	16.33	12	10.81	37	12.71	24	12.00	25	12.38
Used to raise awareness	48	11.94	11	11.34	16	12.21	8	7.02	13	21.67	26	13.13	21	10.71	15	13.51	33	11.34	29	14.50	19	9.41
More details on subgroups and specific classifications of their condition (including age-subgroups)	41	10.20	8	8.25	12	9.16	12	10.53	9	15.00	19	9.60	21	10.71	5	4.50	36	12.37	16	8.00	25	12.38
Easier to understand	32	7.96	8	8.25	9	6.87	9	7.89	6	10.00	14	7.07	16	8.16	13	11.71	19	6.53	18	9.00	14	6.93

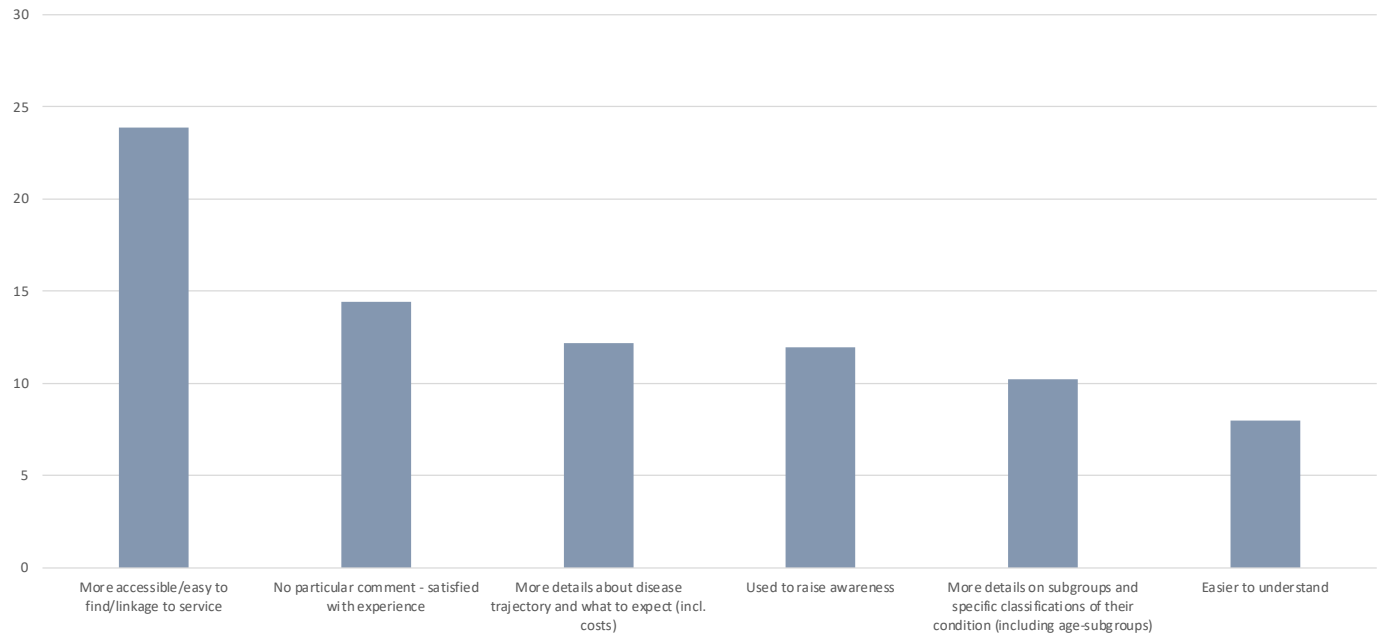


Figure 9.2: Expectations of future information

Table 9.4: Expectations of future information – subgroup variations

Expectations of future information	Reported less frequently	Reported more frequently
More accessible/easy to find/linkage to service	Developmental anomalies Endocrine, nutritional or metabolic diseases	Diseases of the nervous system Other rare condition
No particular comment - satisfied with experience		
More details about disease trajectory and what to expect (incl. costs)		
Used to raise awareness	Diseases of the skin	
More details on subgroups and specific classifications of their condition (including age-subgroups)		Other rare condition

Expectations of future healthcare professional communication

Participants were asked in the structured interview what they would like to see in relation to the way that healthcare professionals communicate with patients. The most common expectations for future healthcare professional communication were that communication will include health professionals with a better knowledge of the condition (21.89%), be more empathetic (17.16%), and satisfied with experience (17.66 %). Other themes included be more transparent and forthcoming (10.95%), include listening to the patient (9.95%), allow people more time to meet with their clinician (9.70%), and include a multidisciplinary and coordinated approach (9.45%).

Future communication will include health professionals with a better knowledge of the condition

Oh well, I think that depends an enormous amount on the personalities involved. Some medicos are harder to deal with than others, in general, it's partly personality and language and things like that. I think it would be good if more doctors knew more about scleroderma, so when you said you had it, they knew what you were talking about, perhaps that would be something.
Participant 012_2023AUDIS

Yes, I would like them not to Google the condition when you sit in front of them. Maybe if they if, just say, 'Look, I don't know this condition. I've not heard of it. But let me do some investigation and then I'll inform myself' But at the moment, most of the time, the parents or myself, even we go into a doctor and I've never heard that and they Google it in front of you. So if they're Googling you, what chance have we got?
Participant 025_2023AUDPA

Future communication will be more empathetic

I can think of in terms of that, I mean some sort of think…some doctors have been insensitive in some of their comments or the way they've spoken about some of the issues you might be facing, but I don't think that's, I don't think that's specific to charged into it. I think some doctors are just not very sensitive, yeah. That's not really specific to CHARGE syndrome, but I have experienced that in terms of my son, but I can't think of anything else apart from that.
Participant 089_2023AUENM

In a human way, be aware that the child's in the room and can hear you, so be aware of that in the language that you use and. Maybe not focus so much on the deficit language, be accessible, and I know that's a really tricky one, but you know, try and they'll have some access availability so that people in between appointments if need a little check up can do that. But there's more in the communication. For me, it's just treating them like they're a human, not a pile of disabilities and things that need to be fixed that they have, you know, these kids and adults have, you know, feelings and they're a person.
Participant.95_2023AUENM

Yes, don't blame me on people, people's weight and stop making them feel that something that your body creates or its own accord is their fault. The shame that is associated is insane. I know that people's diet and exercise and weight contribute. It is not something that we made ourselves have. It's not like smoking gives you lung cancer. Like, yes, it's your fault you did that. But it's not just unfit people that have this, it's not just people who can't afford to have a good diet.
Participant 018_2023AUDSK

No particular comment - satisfied with communication

Not a thing. They're wonderful.
Participant 018_2023AUORC

So far, no. I think that's just for my personal experience. I have a really good specialist, so. In my experience, I'm quite happy with the way that I communicate with my doctor, so nothing I would really change.
Participant 010_2023AUDSK

Well, no, I suppose it depends on the doctors you get at the end of the day, so. If you get a good doctor, any, any, any, he gets on well with you and he can discuss options and, and be compassionate to you about it. I don't think there's any, any concerns but I've been pretty lucky with my referrals from my initial doctor and my current oncologist. So I think I've been pretty lucky in regards to the doctors I've got. I can't speak for everyone but yeah my, my scenario so far I've been pretty happy with my doctors so yeah.
Participant 024_2023AUORC

Future communication will be more transparent and forthcoming

I would like them to explain why they make a decision and the reasoning behind it, rather than to just say, well look, this is what my recommendation is and you know, you either do it or you don't. You know to, to actually explain to you why, why a particular thing is essential or necessary. Because I think any, anyone, it doesn't matter who you are or what you are, if someone explains to you, you say to a child, I want you to go and tidy your bedroom and the kid says, well, why I like it like that. You know, if you say to them, well, because then you'll find it easier to find your toys that you're looking for, you know, it gives them a bit more motivation. But when you just do that, well, because I'm your mother and I said so, doesn't do much for communication. That's good. I, I get that kind of feeling from some of the doctors is that they do they have this patriarchal kind of thing of, well, I'm the doctor, I'm the one who's wearing the white coat, and this is what my you know what my demands are?
Participant 002_2023AUDIS

Yeah, yeah. Was it? You see what they just say. You are all good. Your blood results are all good. See you next year. But they are not actually telling me what to avoid. What triggers it to be active. What should I do? What should I avoid? You know, you know, all those that information at least I know for sometimes, probably I'm doing something that makes it active. But I don't know, they never told me. They just say how you you're all good, not active, you're all fine.
Participant 001_2023AUORC

Yes, be honest about the timeframe process, because that's why I said I was so ignorant. I've just gone in and gone, "Yep, yep, yep." Then again, I did jokingly say to them on the person that passed out in front of pathology lecture, "Don't tell me anything, just, just get it done." As I said, once I was on the queue, and things were moving, because you're in getting tests done, and everything done week on week on week, that was one thing they did say, the good thing was on the very first visit with a rheumatologist, he spent two hours basically telling me I was potentially going to die, I'm having the transplant and I needed to understand the risks, but he did say, "Are you ready? You're going to be locked in a room and you're going to be on your own and you're going to be in there for weeks on end."
Participant 025_2023AUDIS

Future communication will include listening to the patient

Oh, I'd like them not to treat us as if we're all stupid and inconvenient. I have been spoken down to more times than I care to remember by health professionals, and if I had no understanding of the words they were saying. I think a lot of the healthcare professionals and not her routine teams, although part of them, but certainly in the emergency rooms where they didn't engage me as an equal partner. They were telling me what was going to be done or telling me about her condition, but without actually providing information that would be useful and even you know. The case I had with her first year old, just when we told him that she wasn't sleeping and we couldn't get her to sleep, his response was, well, some children don't sleep in terms of some normal children don't sleep overnight. And I felt like saying, yeah, well, you come manage it then as opposed to actually looking for a solution to it. And that was really quite offensive and it is just that real, assuming that we know nothing about yeah. And I know people have different levels of understanding of the medical system and of medical terminology, but I just found they were very dismissive and often very dismissive and condescending. And I didn't didn't really need that.
Participant 090_2023AUENM

Just the compassion side of things, I think like to know that someone's struggling so bad and they were in the beginning a little bit blasé. It wasn't until I had that second by the 1st ablation when he went in and said this is the worst case I've seen in a very long time. He was that that was the first time anyone that had, I felt like I'd been listened to properly. Do you know what I mean? Before that, it was like, I'll take this medicine, you'll be fine. It's like, no, you're not understanding. Yeah.
Participant 032_2023AUORC

Yes. There's that saying about people with autism about like if you've met one person with autism, you've met one person with autism. I feel like the same applies to EDS, like don't make assumptions, just listen to your patients. I think also maybe like the pain scale, a pain scale thing. I've never seen anything formal about people with chronic pain, but I've seen loads of informal ones online where people have shared their versions of pain scales because I feel like it's just different. I feel like that might help be taken more seriously when you present to ED, for example and be like something's like, "My hip's out," and they're like, "Oh, your hip can't go out," and you're like, "No, no, no. My hip's out and this is where I'm sitting on the pain scale." To your GP or doctor. I feel like that would be really cool.
Participant 004_2023AUDPA

Future communication will include a multidisciplinary and coordinated approach

Now they all seem to be talking to each other sometimes when I've been to heart specialist and then three months later I go to rheumatologist, they're supposed to like send a report on and sometimes they haven't received it for my appointment. So it feels like, well, that what that was a waste of time. Yeah, I mean, I know they eventually get it and they do read it, but yeah, I'd like to know that they're that's happening. I mean, I asked my GP and he reads them out to me. So yes. Yeah, of what? Different doctors have said so.
Participant 088_2023AUENM

I would love to see everything put into the health portal and the patient government thing. I actually cancelled mine because I found it more detrimental than useful because so few people used it that most of my information wasn't in there and then when they did look at it, they had a very limited idea of my, my health issues. My GP wouldn't even use it. So I think if it would be an amazing tool if it was utilized mandatorily by all health professionals. But it sounds the way it is, yeah.
Participant 016_2023AUDIS

Oh, look, in a perfect world, I'd have more consistency of the rheumatologist, but I get it's a public teaching hospital. It's just a turnover of people. In the past there has been, there was a lady there that's just retired, been there a long time, and then she was training someone else, but she's on maternity leave. I think, if there was sort of that one person that would be better than someone different each time you go. Yes, they've got your file notes, but it's not quite the same.
Participant 017_2023AUDIS

Future communication will allow people more time to meet with their clinician

PARTICIPANT: I think sometimes that, you know, because of their patient notes, you just, they're just too rushed and too, you know, you feel quite, you can feel quite just dismissed and because the next patient's coming along and yeah, like they wanna get you in and out. Yeah, that sort of thing and yeah, maybe a clearer pathway than you take this pill and they'll see you in six weeks and then you just let floundering with this lack of knowledge and information you know.
Participant 027_2023AUORC

Yes, just communicating would be really good [laughs] just being able to have a little bit more access would be amazing. They're really good and do the best they can. I get that but yeah, it's hard. When I was diagnosed with that interstitial lung disease, they told me I had it and then you look it up and then you start to panic, and then I couldn't get hold of anyone to ask any questions so when you're diagnosed, it would be nice to be able to get information, you know, fairly straight away.
Participant 007_2023AUDIS

Future communication will be more accessible to everyone

I think along the same lines, making sure that they're aware of this condition, and help people get the right treatment for it earlier on. I know people that were dismissed because of their condition and not given any further information and given medication that was never ever going to help them. I would like to see that information coming out to our local and our rural areas, your local GPs.
Participant 006_2023AUDNS

Look, I've only, I've only had really good experiences with my doctors. If everyone could have that experience that would that would be fantastic.
Participant 032_2023AUDSK

Future communication will include developing a care plan with follow-up

I'm really over the hospital system's rigidity in how everything's coordinated. And I know it comes back down to the state and the hospital that you're being monitored for. After being monitored in two states, I just find it really, really frustrating that you have no control over your scans, your blood tests and when your appointment is coming. I've lost all that ability to. And I'm constantly rescheduling when they give me shit time for it, especially because I have to fast for my, fast for my ultrasound. That's one thing that absolutely should see.
Participant 004_2023AUORC

I don't think so. I think maybe just like I said before, with following up on following up on people so after their appointments. Just like a quick phone call, like saying at the second appointment after a diagnosis saying are you OK if we contact you in four weeks just to see how you're getting on with what we've discussed. Like instead of relying on the person to use, but instead of relying on the person to think about it, or for them to use the initiative, perhaps just following up. And then that way the person, like the patient, is at liberty to either answer and accept that help or they can just let it slide. Do you know what I mean?
Participant 029_2023AUDPA

Future communication will be more holistic (including emotional health)

I would love them to take it seriously and not just wipe their hands, you know. Be proactive in assisting the patient to access the services that they need and have a regular follow up as to if those services are working, how they're going and if changes are needed. Whether that that start with your GP or your neurologist, but you know have a have a head person like a holistic health plan, but you know somebody that's invested in the best interest of the patient, not just the dollars that they can pay them.
Participant 026_2023AUORC

Yes, I would say being like…just a hell of a lot more gentle with, with the delivery that there needs to be like an instant sort of healthcare support for the processing of that information and what it means. And it needs to be ongoing rather than just like here's your diagnosis, see you later that there really needs to be…the idea that, like, this isn't just something that happens to the individual, but it is also a, a whole family experience and therefore there needs to be support and mental health care for the whole family, including siblings and sibling services. Yeah, that kind of thing. Participant 018_2023AUDPA

Yes. You know, I, I think they sort of just see it as their job to tell you what they need to tell you. They don't really go through how it can affect you and possible outcomes and anything like that. I think when someone's diagnosed they should be counselling or therapy offered maybe even to their carers as well, depending on if that's something they want or need. And I guess more awareness. There's been a couple times I've gone into hospital and said I've got NMO and I'm having a flare up. They sort of look at me and go, what's that? It's amazing how many health professionals don't even know what it is.
Participant 096_2023AUDNS

Future communication will be more understandable

Keeping it simple. Participant 014_2023AUDPA

Yeah, and that's very variable depending on the specialist. Some specialists just look and talk to me. Some specialists are good and you know and, and we'll talk to PATIENT in and, and explain things to her in a language that she understands and, and sometimes with those then I have to ask. I have to interrupt them and ask them to make sure that I've got it in the real language, if you know what I mean. So yes, so that that's, that's variable. So you know and others will talk and I'll be understanding them that I, I actually always through a consult will after they've said something. I will then put it into simpler, straightforward sentences for my daughter that is short enough to give her a picture. Like, you know, the doctor said this, he thinks whatever and make sure that she has understood it. And I look at the doctor to confirm that what I have conveyed to her is fair enough. Yeah. So I'm constantly have to do that. Generally allied health are better I think at communicating to PATIENT or the ones that we've chosen up. So, and I think also providing written information and visuals is something that is missing or that is often, Yeah. And even I find very useful when you know, when you when you're juggling so many multiple specialists and I'm a bit worn out right now, whereas once upon a time I was totally on top of everything. Nowadays, I really appreciate when they give me something written down as well, because like I've got so many other things I'm dealing with that to remember everything they've said, like I've understood it all at the time. But then you go home, you do all these other things and you think can't quite remember. So to have things that are written both at a level that I understand, but also having things in simpler format to show my daughter.
Participant 038_2023AUDPA

Table 9.5: Expectations of future healthcare professional communication

Expectations of future communication	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Health professionals with a better knowledge of the condition	88	21.89	14	20.90	15	18.52	8	8.42	15	46.88	35	36.84	1	3.13	68	25.37	20	14.93	70	23.81	18	16.98
Satisfied with communication	71	17.66	4	5.97	12	14.81	23	24.21	5	15.63	20	21.05	7	21.88	45	16.79	26	19.40	48	16.33	23	21.70
More empathetic	69	17.16	3	4.48	17	20.99	19	20.00	10	31.25	10	10.53	10	31.25	47	17.54	22	16.42	54	18.37	14	13.21
Include listening to the patient and recognise the impact of the condition	53	13.18	5	7.46	19	23.46	7	7.37	5	15.63	10	10.53	7	21.88	45	16.79	8	5.97	43	14.63	9	8.49
More transparent and forthcoming (incl. about treatment options)	44	10.95	3	4.48	16	19.75	13	13.68	0	0.00	4	4.21	8	25.00	34	12.69	10	7.46	37	12.59	6	5.66
Allow people more access and time to meet with their clinician	39	9.70	3	4.48	9	11.11	21	22.11	0	0.00	2	2.11	4	12.50	34	12.69	5	3.73	30	10.20	9	8.49
Multidisciplinary, coordinated and consistent approach	38	9.45	3	4.48	15	18.52	3	3.16	2	6.25	9	9.47	6	18.75	30	11.19	8	5.97	29	9.86	8	7.55

Expectations of future communication	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Health professionals with a better knowledge of the condition	88	21.89	9	9.28	30	22.90	31	27.19	18	30.00	39	19.70	47	23.98	31	27.93	57	19.59	46	23.00	42	20.79
Satisfied with communication	71	17.66	24	24.74	18	13.74	15	13.16	14	23.33	33	16.67	37	18.88	20	18.02	51	17.53	30	15.00	41	20.30
More empathetic	69	17.16	18	18.56	22	16.79	22	19.30	7	11.67	31	15.66	37	18.88	15	13.51	54	18.56	39	19.50	30	14.85
Include listening to the patient and recognise the impact of the condition	53	13.18	7	7.22	18	13.74	22	19.30	6	10.00	23	11.62	30	15.31	17	15.32	36	12.37	28	14.00	25	12.38
More transparent and forthcoming (incl. about treatment options)	44	10.95	7	7.22	14	10.69	20	17.54	3	5.00	16	8.08	27	13.78	6	5.41	38	13.06	17	8.50	27	13.37
Allow people more access and time to meet with their clinician	39	9.70	2	2.06	20	15.27	12	10.53	5	8.33	26	13.13	13	6.63	7	6.31	32	11.00	19	9.50	20	9.90
Multidisciplinary, coordinated and consistent approach	38	9.45	5	5.15	12	9.16	14	12.28	7	11.67	21	10.61	17	8.67	11	9.91	27	9.28	18	9.00	20	9.90

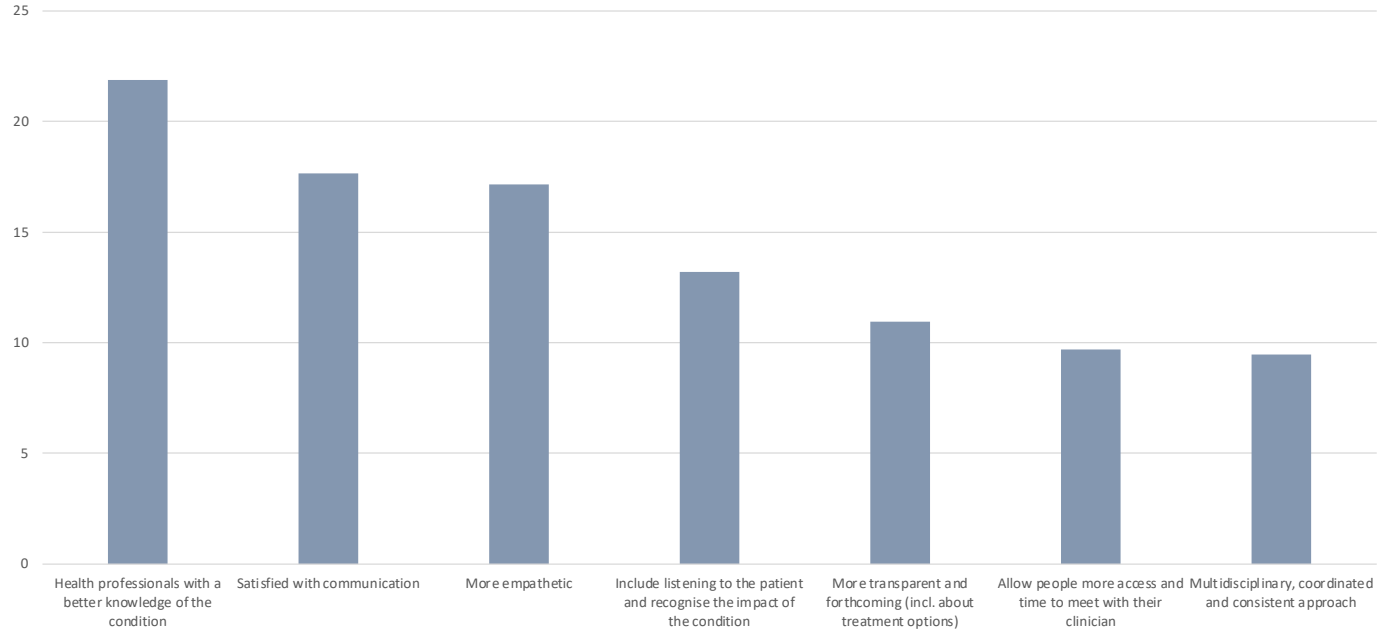


Figure 9.3: Expectations of future healthcare professional communication

Table 9.6: Expectations of future healthcare professional communication – subgroup variations

Expectations of future communication	Reported less frequently	Reported more frequently
Health professionals with a better knowledge of the condition	Diseases of the nervous system Other rare condition Aged under 18	Diseases of the skin Endocrine, nutritional or metabolic diseases
Satisfied with communication	Developmental anomalies	
More empathetic	Developmental anomalies	Diseases of the skin Other rare condition
Include listening to the patient and recognise the impact of the condition		Diseases of the immune system
More transparent and forthcoming (incl. about treatment options)	Diseases of the skin	Other rare condition
Allow people more access and time to meet with their clinician		Diseases of the nervous system

Expectations of future care and support

Participants were asked in the structured interview whether there was any additional care and support that they thought would be useful in the future, including support from local charities. The most common expectation for future care and support was that it will include more access to support services (22.89%), will include a multidisciplinary and coordinated approach (14.68 %) and will include specialist clinics or services where they can talk to professionals (in person, phone, online) (13.93%). Other themes included ill include being able to connect with other patients through peer support (support groups, online forums) (11.69%), will include health professionals with a better knowledge of the condition (9.70%), and will include practical support (home care, transport, financial) (7.96%). There were 32 participants (7.96%).) that were satisfied with their care and support and had no particular comment.

Future care and support will include more access to support services

I think maybe access to the NDIS seems to be a real mixed bag on who gets approved and who doesn't. I know on the Scleroderma Australia website there are some guidelines, but if there was perhaps a person who was skilled in that who was available to assist or review applications before they went in or knew more about specific terminology or trigger things or what to say or what was not to say, I think that would be really helpful.
Participant 017_2023AUDIS

I think charities have got better and some of the hospital systems have got better in that sense. It's funny because I've been treated when I've been in the hospital all of a sudden since, oh, you've got scleroderma. They're so like, wow, look at this. It's like some of them are really well-informed now. They know. They think it's great that they can see a patient like that, which it's good too. I think basically I've noticed that Queensland hasn't got a lot of support as opposed to Victoria and New South Wales and some other states.
Participant 001_2023AUDIS

Just support in general, I guess you know. When in hospital because you do visit quite regularly, you know, the nurses go, oh, I haven't seen you for a little while or oh, you're back again, you know, so, so that is quite, quite regular. Yeah, I guess.
Participant 034_2023AUDPA

Future care and support will include specialist clinics or services where they can talk to professionals (in person, phone, online)

I mean, I know in America they have in one of the hospitals, they have an actual CHARGE syndrome clinic where there are professionals working there who are all experienced for the charge syndrome and they're all communicating with one another and working together, and I've always thought how great that sounded, that all these people came together in clinic and could work collaboratively instead of everybody working separately and searching out people with experience. Sorry, that would be pretty amazing. Someone else, yeah. I mean, communication is a big issue in CHARGE syndrome, so I've always wished that there was a service that had a instead of attending the speech general speech pathology like we do for people that are vision and hearing impaired, always thought it would be great if there was some sort of service that again where people had experience with CHARGE syndrome was focusing on communication in CHARGE syndrome, which is a bit different to somebody who's just vision and hearing impaired because it would take into account the other Physical impairments that you might have, yeah. I don't know. Nothing else really comes to me though apart from those two.
Participant 089_2023AUENM

I've been thinking about this interesting development in. One Danish city from one Danish hospital. They offer a 24 hour clinic. Something like that needs to happen in Australia for the future because there's people like me who just live in misery. 24 hour care the 24 hour clinic, a specialized clinic from doctors and nurses who know all about it.
Participant 008_2023AUDSK

Yeah, like proper support services, not just a group of people on Facebook that all have the same disease. So a network where you can actually. Like I've had, I've had times when I have had flares that are out of control. I can't, I don't know what to do. Like I've reached the point where I've literally, I end up at, at emergency because I don't know what to do. So, you know, having a support service, you know, like I don't know if you have it up in LOCATION, but like we have like a one 800 number in LOCATION that you can actually speak to registered nurses. Yes. Yeah. Having something like that specific to the disease that you

can make contact with someone and say, okay, what do I do? Yeah, yeah. Have I reached the point where I don't have a choice, I have to go to hospital or is there something else that I can try?
Participant 005_2023AUDSK

Future care and support will include a multidisciplinary and coordinated approach

Again, that's a really hard question to answer specifically because it can be so broad. But even just in terms of treatment, access to more specialists, I mean, we struggle even accessing paediatrician and that shouldn't be that hard. But it is what it is. Yeah, just access to specialists really in a timely fashion is very major, yeah.
Participant 021_2023AUDPA

Yeah, I definitely think like the dietitian support. Yeah, maybe that's just because I need to organize it, but that support for like ongoing management of someone who's having a restricted diet. Yeah, I think that that would because it's not a focus of it. It's not a focus of the gastroenterologist necessarily. But yeah, yeah, yeah, that's someone that maybe like it's maybe it's like more regular follow up to someone like that because you can always access it as a parent. Like if I book an appointment I can always access the service, but I don't. Yeah. But there's sort of no one doing that. Sort of following up to make sure that, yeah, like if I wasn't proactively doing that, nothing would happen.
Participant 079_2023AUDIS

Future care and support will include health professionals with a better knowledge of the condition

Practitioner education, which is a bit more like, inside the Australian health system EDS Echo kind of thing. Practitioner education and client support. I don't know how else to put that. Support options for -- it's really hard because you can't create community out of a vacuum either. You kind of can for example, I tried to join the Australian Ehlers-Danlos thing. I signed up so many times and just keep getting these errors about your password and your thing. Then I tried to email someone and never got a response. It's like, does it actually exist?
Participant 001_2023AUDPA

Maybe just awareness to more doctors and hospitals. I think when I was younger I saw a GP, they didn't even know what they were looking at and I think if they had of known, I probably would have been able to get treatment like proper care a lot sooner. Maybe having a better skin clinic services within the public hospital so that people who have this disease don't have to fork out thousands and thousands to receive treatment for something that, you know they're born with, that they can't help that they have.
Participant 010_2023AUDSK

I think the big thing, particularly being from a region is that the hospitals. Have the ability to tap into the specialists and are willing to because I've found sometimes I think oh no, well you're our patient and we're going to tell you what to do and you'll do it our way. And then you talk to the specialist in Brisbane they say well actually you should be doing this. So you you're sort of getting mixed messages. So that's probably something that needs to be worked on is educating regional hospitals and service providers about, you know, if there's any doubt, you don't decide, you get in touch with people that know rather than make a decision and then you've got two different opinions being given to the client.
Participant 007_2023AUORC

Future care and support will include being able to connect with other patients through peer support (support groups, online forums)

I think I feel like something that was really lacking was the capacity or, or the information or like the invitation. I don't know whether there's anything out there around a support groups with people that have rarer set of conditions that can be that can be very isolating. I don't…it's possible that that's accessible in more metropolitan areas?...I'm not sure, but certainly out here in this rural space is very isolating.
Participant 007_2023AUDSK

It would be lovely. It would be great to see more. I would like to actually see some, some more media stuff so that HS is normalized a little bit more. Yeah. So I would love to see some stuff on media so that it was it was normalized. That the community understood that the condition exists and that there were a lot of people in the community with the condition. I would like to see peer support groups. Maybe develop like face to face groups in communities would be great, particularly if there's communities that have, you know, high concentrations of the condition. I think, yeah, to be able to meet with others that understand and are walking that journey is absolutely valuable.
Participant 001_2023AUDSK

Future care and support will include practical support (home care, transport, financial)

Our biggest cost comes from accessing poor quality dressings at astronomical prices, and we don't have access to specific dressings that have been created. But they're not approved or they're not allowed to be in Australia. So we're left without and it's not really fair. And Medicare won't cover any dressings at all. My previous surgeon, he tried. He tried to get me to go use the community nurse so that dressings will be covered, and every time he'd refer me, I was disbarred almost immediately from the service. Because you can dress yourself. You don't need the community nurse to dress you. Medicare doesn't cover dressings. It covers the nurses nurse doing the dressing, which isn't fair. It isn't right and it's now put us in this in more. In my instance, it's put us in a financial position where I have to. Cut back on groceries to cover dressings.
Participant 012_2023AUDSK

Specific services. Maybe to service scleroderma, I'm talking for myself also, I need help to go out. I need help when I have to take an Uber to go somewhere to an appointment, I always have to ask for help, because I can't open the door, I can't put the seatbelt on. Even at the hospital, I think when you go to the hospital, there should be some people there, some attendant or, I don't know. When they see people with scleroderma coming at the reception over desk, they should be able to ask that person, "Do you need any help with the door? Do you want me to take you somewhere?" Of course, every time I go to hospital, I have to tell them I can't write, but they can see my hands so [unintelligible 01:00:26] [chuckles]. "Okay, don't worry, I will tell the doctor to fill up the application form for you." Maybe they should be observing more what's around them, pay more attention to disabled people because…I don't know what else to ask for.
Participant 020_2023AUDIS

I think the cost of dressings and support that way. So nurses at GP clinics where you tend to go educate there, they have very limited dressing supplies, chemists don't have much and their prices are exorbitant. So like hospitals or the GP clinic, they need to help you access a wider range of dressings at a reasonable price.
Participant 013_2023AUDSK

Future care and support will be more holistic (including emotional health)

The mental health services, I believe we all need them. Like, even if we say we don't like, when I first got diagnosed, I probably would have said no, I don't need that. Then like thinking about it, living with it, like with the diagnosis. And I'm like, yeah, OK, now I feel pretty crappy about myself.
Participant 003_2023AUDSK

We've got I'd, I'd love CMT to have CMT Australia the support to be able to offer. You know, credible and good referrals, you know, to know this is if you're in this state, this is who you go to see, which is where we are looking at employing a Telehealth nurse to, to help. Begin the process of looking in. Yeah, starting these holistic health care so that, you know, it doesn't matter where we are, they can link us with services that we need.
Participant 026_2023AUORC

I think the psychology services are really lacking in rural areas. So I think PATIENT could have done with more and, and that wasn't even really mentioned in this in Rome or in in by any of the health professionals that. I've, you know, I've come across writings that PATIENT'S written when I've been cleaning up and she's been very depressed. And so I think she did need a lot more support herself psychologically in her teens and her early adult that she wasn't getting and I wasn't really aware of. So to me that was a really big. A bit missing out of the whole thing because I think she went through a really hard time herself and was very, very lonely.
Participant 09_2023AUDPA

Future care and support will include charities specific to condition

Well, I don't even know if there…I'm not aware if there even is a charity for Q22. I don't even know. That's bad. I don't even know if there is a charity, but. Yeah, maybe just I think the main thing I would like is just more awareness of the condition out in the community as a general. So whether that's, yeah, I didn't know the condition existed until I found out it was in my family. So just more general public awareness of it and the effect it can have because there are a lot of people out there who have it, who don't know they have it and might just think, like I did, that they're just not very clever.
Participant 029_2023AUDPA

There is a CHARGE Syndrome association. I would like to see them get some funding because it it's, yeah, pretty much run solely by passionate volunteers who are all relatives of someone with CHARGE. But because no one knows about CHARGE, it you know, it doesn't attract any funding. It would be great if there could be some, some sort of cohesive umbrella for groups like that that you know they're so rare that it's very hard for them to attract notice and funding. But in a lot of ways they're more critical because those diseases are so rare. So, you know, if they could all kind of come together and I don't know, maybe get assistance so that they're, they're not all trying to replicate things that each group does. You know, like some admin stuff or you know, website maintenance that maybe that could all be funded better so that each little group isn't trying to do everything themselves.
Participant 091_2023AUENM

It would be great if they had did have more charities in relation to 22 Q or you know, even just start talking about it in the media, you know, be more like bring it out more, you know, make people aware, you know, I mean they, they do it with, well, they started doing it with autism, haven't they? So just, yeah, making people aware of 22 Q.
Participant 028_2023AUDPA

Future care and support will include more family and carer support

Gosh, I've never really thought about it because it doesn't exist. It would be nice like a support system because they do have the clinic at our hospital and we're lucky that we do have that, but they isn't actually like support services for the family in that scenario. There's like it's a referral system pretty much. So, you go there and they have a look at the kid and then they refer you to all the specialists that you need. So, I think the like the social support at home of how do you actually live with this condition is sort of missing there.
Participant 032_2023AUDPA

Look, I think the, the thing that I sort of see in…like you know, the younger generation of, of babies that are being born now is just over and over severe parent overwhelmed, particularly when there are babies who are in hospital for long periods of time and I think there needs to be options for parents to be with critically ill babies living with them, you know, like that, yes, there's a room that's dedicated nursing or whatever it is. But like, in order for that attachment to form and really like the, the kids who have an attachment with CHARGE can be pulled out of themselves and do so well. They learn to speak, they learn to communicate, they learn to be like little amazing pocket rockets. But the kids that don't remain nonverbal and you know, don't develop social skills and, and that kind of thing and, and it's just like not fair. It's just not fair on them, yeah, so…You're just having someone also to support the parents with how you bond with a child who you can't give cuddles and kisses and tickle in the tummy and, you know, give them their fair foods and all of that. Yeah, beautiful.
Participant 018_2023AUDPA

Yeah, I think that the carers of people with cystic fibrosis should receive some sort of government support funding, some funds for whether it's lost wages or that sort of thing. I know it's not considered a disability, but the level of care is still so high that I feel it should be considered for NDIS or something similar, yes. Yep.
Participant 029_2023AUORC

Table 9.7: Expectations of future care and support

Expectations of future care and support	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
More access to support services	92	22.89	13	19.40	30	37.04	24	25.26	5	15.63	15	15.79	5	15.63	63	23.51	29	21.64	70	23.81	21	19.81
Multidisciplinary and coordinated approach (incl. upskilling of staff)	59	14.68	7	10.45	15	18.52	14	14.74	0	0.00	20	21.05	3	9.38	38	14.18	21	15.67	48	16.33	11	10.38
Specialist clinics or services where they can talk to professionals (incl. nurse specialists)	56	13.93	6	8.96	10	12.35	17	17.89	9	28.13	7	7.37	7	21.88	40	14.93	16	11.94	38	12.93	18	16.98
Peer support, support groups and online forums	47	11.69	7	10.45	4	4.94	9	9.47	8	25.00	12	12.63	7	21.88	37	13.81	10	7.46	36	12.24	10	9.43
Health professionals with a better knowledge of the condition	39	9.70	8	11.94	20	24.69	5	5.26	3	9.38	1	1.05	2	6.25	32	11.94	7	5.22	35	11.90	4	3.77
Practical support	32	7.96	4	5.97	5	6.17	7	7.37	6	18.75	5	5.26	5	15.63	21	7.84	11	8.21	28	9.52	4	3.77
No particular comment - satisfied with experience	32	7.96	0	0.00	10	12.35	7	7.37	0	0.00	15	15.79	0	0.00	27	10.07	5	3.73	22	7.48	10	9.43

Expectations of future care and support	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
More access to support services	92	22.89	16	16.49	33	25.19	28	24.56	15	25.00	45	22.73	43	21.94	26	23.42	66	22.68	55	27.50	37	18.32
Multidisciplinary and coordinated approach (incl. upskilling of staff)	59	14.68	16	16.49	16	12.21	20	17.54	7	11.67	24	12.12	34	17.35	24	21.62	35	12.03	32	16.00	27	13.37
Specialist clinics or services where they can talk to professionals (incl. nurse specialists)	56	13.93	12	12.37	15	11.45	19	16.67	10	16.67	26	13.13	30	15.31	10	9.01	46	15.81	27	13.50	29	14.36
Peer support, support groups and online forums	47	11.69	5	5.15	16	12.21	15	13.16	11	18.33	27	13.64	20	10.20	14	12.61	33	11.34	17	8.50	30	14.85
Health professionals with a better knowledge of the condition	39	9.70	5	5.15	16	12.21	12	10.53	6	10.00	17	8.59	22	11.22	16	14.41	23	7.90	27	13.50	12	5.94
Practical support	32	7.96	9	9.28	12	9.16	10	8.77	1	1.67	16	8.08	16	8.16	5	4.50	27	9.28	14	7.00	18	8.91
No particular comment - satisfied with experience	32	7.96	4	4.12	8	6.11	11	9.65	9	15.00	19	9.60	13	6.63	8	7.21	24	8.25	14	7.00	18	8.91

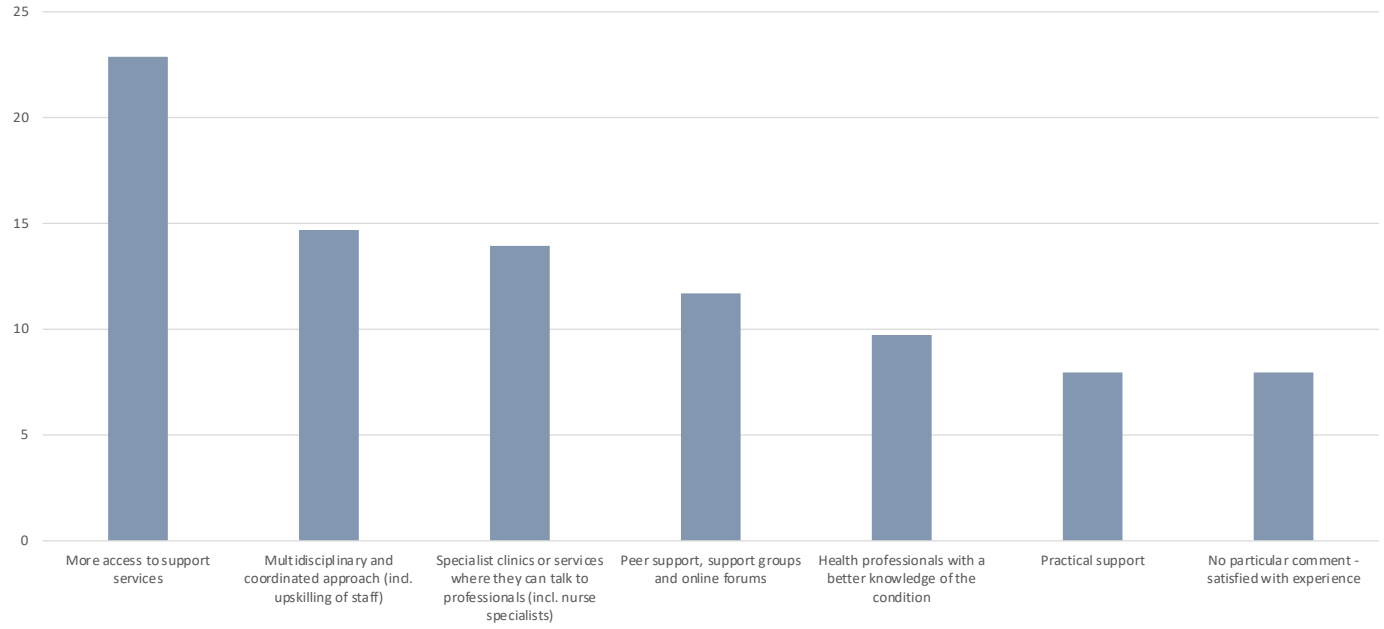


Figure 9.4: Expectations of future care and support

Table 9.8: Expectations of future care and support – subgroup variations

Expectations of future care and support	Reported less frequently	Reported more frequently
More access to support services		Diseases of the immune system
Multidisciplinary and coordinated approach (incl. upskilling of staff)	Diseases of the skin	
Specialist clinics or services where they can talk to professionals (incl. nurse specialists)		Diseases of the skin
Peer support, support groups and online forums		Diseases of the skin Other rare condition
Health professionals with a better knowledge of the condition		Diseases of the immune system
Practical support		Diseases of the skin
No particular comment - satisfied with experience		

What participants are grateful for in the health system

Participants were asked in the structured interview what aspects of the health system that participants are grateful for. The most common responses were that participants were grateful for low cost or free medical care through the government (40.34%) – with the related theme os included timely access to treatment (11.36%). Other themes included being grateful for healthcare staff (including access to specialists) (35.23%), and the entire health system (18.47 %).

Participant describes being grateful for healthcare staff (including access to specialists)

Yes, the nurses. The nurses are amazing. From the transfusion nurses, to the nurses that I do my six minute walk test with, to lung function nurses. All those little people, they do all the hard work. I take my hat off to them like, they are the best people because they've seen me progress to worse and like, "Oh, NAME, you've improved your six minute walk test." Those people that do those tests, and the nurses that

look after you in hospital, they are great people. I take my hats off to them any day.
Participant 023_2023AUDIS

We used to have an adult cystic fibrosis ward at the HOSPITAL and it had specific cystic fibrosis nurses there. So they knew what our medications were. They knew they just were trained specifically in cystic fibrosis. So I found it really helpful that they had that there and that the nurses were not understood and they didn't have to ask all these stupid questions that other nurses do. They don't have that ward anymore. It's now a COVID ward, but when they did have it, I found it really helpful and that was great. And then also that they have a cystic fibrosis specialist team. Yeah, I think that's great as well.
Participant 013_2023AUORC

My cardiologists have been amazing. Those last couple were really, really, really good.
Participant 032_2023AUORC

Participant describes being grateful for low cost/free medical care through the government (Public health system in general)

Well, I have been grateful that a lot of the surgery that the surgeries I've had so far have not had to pay for, but they have been covered by Medicare when I've gone to the hospital, but that. That's where my gratitude ends, because from my experiences, from my accessibility, the healthcare system, the only thing I can be grateful for is that we are not yet an American healthcare.
Participant 012_2023AUDSK

I guess, yeah, the fact that we do have a Medicare system that's yeah saved me a lot of money overall. Yeah. So, I'll say I'm and if you and the care physicians, OK. Yeah
Participant 005_2023AUDNS

Oh, the virtually no cost to us. Yeah. Not, not direct medical costs, that's we're very grateful for that,
Participant 08_2023AUDPA

Yeah, pretty much all of it. Because, you know, we wouldn't be able to afford to pay for surgery and hospital stays and you know, all those special like visiting all those specialists, that's just that would just be insane amount of money if we were paying and you know, we pay our taxes and we're very grateful that to have Medicare. You know what I mean? Yeah. So, yeah, the cost, yeah, the cost and, and the care provided, very grateful.
Participant 023_2023AUDPA

Participant describes being grateful for the entire health system

The fact that so much of it is completely free is just astounding just completely blows my mind and makes me very happy to pay taxes to say the weird thing to say. But. So much would have been free. So many of the people that I've interacted with have been just lovely and caring and supportive and helpful. Yeah, I'm very impressed with most, not all, but most of my interactions with the whole health system.
Participant 009_2023AUDIS

Oh, boy, everything. It's cheap or free for a person like myself with a low income. The fact that I can see specialists once again—and also that I can involve myself in clinical trials because, for me, that's a way of being useful even within the limitations of my life. I feel at least of some use to somebody, even if the trials aren't any use to me down the track, they'll be useful for somebody.
Participant 004_2023AUDIS

Very much so. I think I said it earlier as an Australian, I feel very fortunate that we do have a good healthcare system that we do have the option of having the drug approved so that we only pay dispensary costs, not the two and a half, $1000. We don't have to have private insurance to access hospitals. We don't, you know at the dermatology clinic I don't pay for. I see some of the best. Doctors around in my state that that understand HS and it doesn't cost me anything. Sure there's a wait and you have to sit in line. You know the de roofing surgery that I'm waiting on at the moment will be through public health, through the plastic surgeons in public health. There's a wait. I have to wait for it. But I've waited 30 years to to get some help, so. You know, if I have to wait two more years for the surgery that sets me free, then so be it.
Participant 005_2023AUDSK

I'm just, I'm thankful that we actually have a a health system where we can access pay for the treatments. I'm extremely thankful that we have PBS where I can pay $30.00 a month for my Humira and not, you know, the two and a half thousand a month or whatever it is for it I'm thankful for chronic illness program that we have where I can access other services like counselling, physiotherapy, that sort of thing. Yeah.
Participant 001_2023AUDSK

Participant describes being grateful for timely access to treatment

I know this isn't the health system, but the NDIS has been amazing for us at least. I know not everyone has experienced that, but we certainly have and been well supported. Through that which has helped us a great deal. I am really grateful that when we have attended Emergency, it hasn't been a huge long wait. I mean, as she gets old it has been. But we were very grateful, particularly when she was younger. We were there quite a lot with aspiration or whatever, that she was triaged and expedited through the process because they could see. You know how complex she was. So very grateful for that and for our ambulance services. They were here really quick whenever we called them, so they're very, very grateful for that, yeah.
Participant 095_2023AUENM

This is how quickly we're now able to like get this groups and everything and how we sometimes we can just go into a doctor and like have a walk in appointment where I know like other time other people may have had their life five days that's for one appointment. So I think it is well in one. In one particular instance was when they offered a, a CPAP machine through a charity at the Children's Hospital. We didn't take them up on that offer because we actually went out and bought her one ourselves. But I thought that that was actually really, really, really great. But we didn't want to take it because there was the next person that would have needed it from the charity. So I know now it's not a like Australian, you know, like government sort of thing, but I do think Australian general are very charitable when it comes to that sort of thing. So that to me is something that stands out in her medical history, that we've turned down something like that for somebody else.
Participant 037_2023AUDPA

I guess probably quick and efficient care whe, when required. When she did have the heart issues and stuff, it was all dealt with very quickly and, and we didn't have to worry about anything at the time in regards to that. So that was there was no delays, nothing. It was all very, very prompt.
Participant 027_2023AUDPA

Participant describes being grateful for low cost/free medical treatments through the government

No, I'm pretty, I'm, I'm satisfied to the point that obviously my treatment you know, clean hospitals, great doctors, affordable. I'm pretty grateful for where I live and yeah, lucky to be raised in Australia with all the support and, and you know, companies like you doing research and stuff like that. So yeah, it's very lucky and grateful, yeah.
Participant 024_2023AUORC

Definitely being able to have the surgery not charged, that's been something that because that's the point of diagnosis. So that's kind of the most necessary part.
Participant 078_2023AUDIS

The fact that there are public health outlets, you know that, that I've been able to access them freely. I was living in the States at a point of when I was pregnant with my daughter and it would have been I had C-section…My emergency caesarean would have cost me if I'd have stayed. And I, I think after then having recurring boils and those sorts of things any hospital or healthcare visit. It is not lost on me at all that that I do at least have the benefit of bulk billing and those sorts of things, PBS scripts, you know, being able to benefit from discounted pharmaceuticals. I think it is …just be the education to go along with it for those healthcare providers.
Participant 015_2023AUDSK

Definitely PBS medication. I'm like for other conditions I have. Quite a few non PBS medications. It makes big difference, yes, and I think easy access to the specialists and things like that as well.
Participant 019_2023AUDSK

Participant describes being grateful for timely access to diagnostics

I'm grateful I do the test every time. At least I'm being monitored. That's why I'm grateful that I get monitored every time, so if something goes wrong, at least I can catch it early.
Participant 001_2023AUORC

I'm very grateful for the, very grateful for the fact that we got referred to a dermatologist very quickly from the GP and that we, the dermatologist was able to fit us in because he had a three month waiting, waiting list or 4 four month waiting list and because of his age and yeah, the…the,y they spotted him in within like I think we got in within like 2 weeks. So I'm very grateful for that, very grateful for the, the way a child was prioritized.
Participant 009_2023AUDSK

Again, you know, newborn screening. Without that, I don't think that my child would have survived because they wouldn't have known what the condition was and how to manage it. I'm grateful for what resourcing is available in the tertiary care system to provide care for my child with their condition. I do think that comparatively, we do have a good health system, but I wouldn't say that it's great, to be honest. They're the things that I'm grateful for within the system that we have.
Participant 021_2023AUORC

Participant describes being grateful for access to private healthcare/private insurance

Oh yes, because private therapy is like, I'm very grateful for being able to access good physiotherapy through private health. I had to use my chronic disease management plan, so even though it's only five, can't knock it, that's still helpful. I'm grateful to have had that towards the things that I've needed. Yes, they're probably the main things. I haven't needed to go to the public hospital much myself, but for my kids I have and that's been amazing and the genetics was covered through public health, so I am very grateful for that because I'm sure that would've been very expensive privately. I don't actually know if there is someone private who does it. I think only recently, maybe if someone went into genetics privately in [unintelligible 01:10:07] I think it was only at the time when I went through, it was only the public referral base.
Participant 004_2023AUDPA

The ability to be able to go to a private hospital and have private, be privately health insured, you know grateful grateful. I've got that that service is available. I didn't get to choose my neurologist. It was, I was given one but, but I think, I think in the main I think the health service that I received was very was, was, you know, very good the fact that I had choice to go to a private hospital. I could have gone to a public hospital and, you know, possibly the public hospital would have been, would have been, okay, but I, I don't know that and I'll never know that but I'm grateful that I that I was able to go to a, to a private hospital.
Participant 095_2023AUDNS

Participant describes not being grateful for anything

Nothing, Nothing. As I said, public hospital. I've been at LOCATION, I've been at HOSPITAL. There's nothing, I mean nothing that they have helped me. And because I was able to cut and wash it and clean myself, even the nurses could get less about me.
Participant 023_2023AUDSK

PARTICIPANT: Nope. Next question.
INTERVIEWER: Yeah, I thought so. No, it's fine.
PARTICIPANT: Gotta keep the sense of humor.
Participant 008_2023AUDSK

I can't really say much at all.
Participant 025_2023AUDSK

Table 9.9: What participants are grateful for in the health system

What participants are grateful for in the health system	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=352	%	n=67	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=247	%	n=105	%	n=252	%	n=98	%
Low cost/free medical care through the government (Public health system in general)	142	40.34	19	28.36	40	49.38	17	37.78	17	54.84	36	37.89	13	41.94	106	42.91	36	34.29	117	46.43	25	25.51
Healthcare staff (including access to specialists)	124	35.23	23	34.33	28	34.57	14	31.11	10	32.26	32	33.68	17	54.84	83	33.60	41	39.05	91	36.11	32	32.65
Entire health system	65	18.47	7	10.45	9	11.11	6	13.33	5	16.13	33	34.74	5	16.13	50	20.24	15	14.29	46	18.25	19	19.39
Timely access to treatment	40	11.36	4	5.97	15	18.52	5	11.11	0	0.00	14	14.74	2	6.45	28	11.34	12	11.43	32	12.70	7	7.14

What participants are grateful for in the health system	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=69	%	n=116	%	n=108	%	n=59	%	n=172	%	n=172	%	n=100	%	n=252	%	n=176	%	n=176	%
Low cost/free medical care through the government (Public health system in general)	142	40.34	20	28.99	55	47.41	48	44.44	19	32.20	65	37.79	72	41.86	42	52.50	100	39.68	70	39.77	72	40.91
Healthcare staff (including access to specialists)	124	35.23	29	42.03	42	36.21	35	32.41	18	30.51	57	33.14	64	37.21	33	41.25	91	36.11	64	36.36	60	34.09
Entire health system	65	18.47	10	14.49	16	13.79	19	17.59	20	33.90	34	19.77	30	17.44	17	21.25	48	19.05	29	16.48	36	20.45
Timely access to treatment	40	11.36	6	8.70	16	13.79	13	12.04	5	8.47	23	13.37	15	8.72	12	15.00	28	11.11	18	10.23	22	12.50

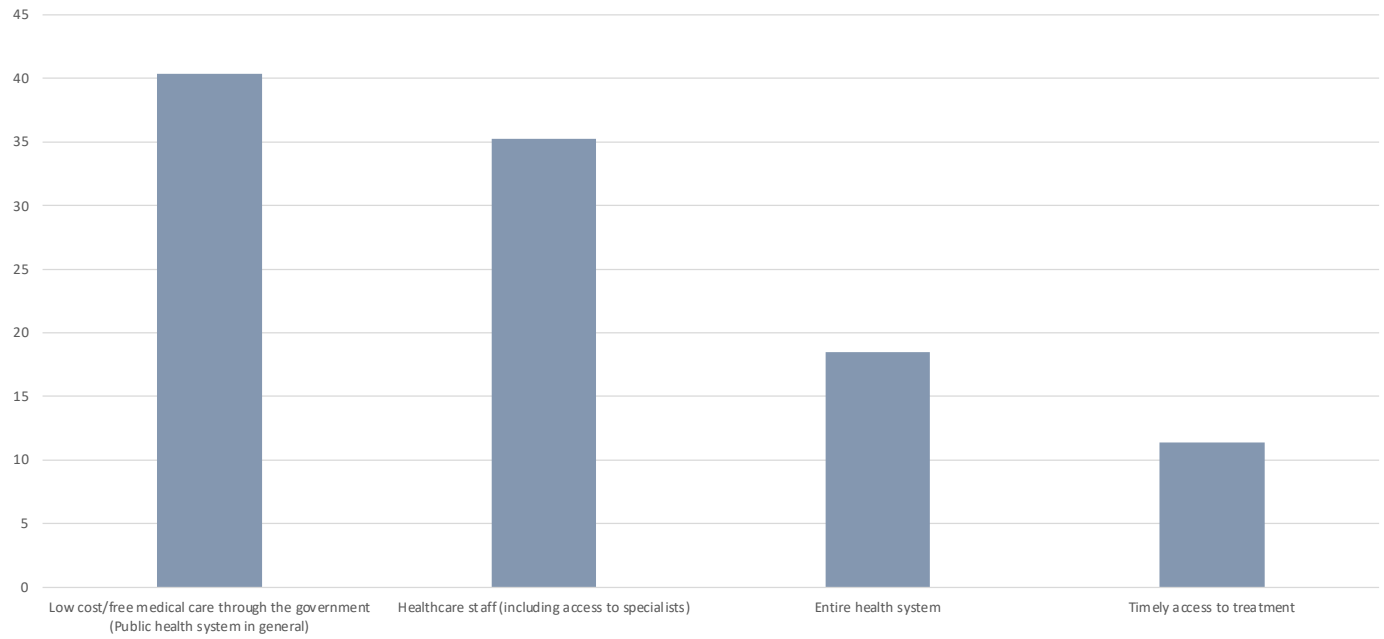


Figure 9.5: What participants are grateful for in the health system

Table 9.10: What participants are grateful for in the health system – subgroup variations

What participants are grateful for in the health system	Reported less frequently	Reported more frequently
Low cost/free medical care through the government (Public health system in general)	Developmental anomalies Male Aged under 18	 Diseases of the skin Regional or remote
Healthcare staff (including access to specialists)		Other rare condition
Entire health system		Endocrine, nutritional or metabolic diseases Aged 65 plus
Timely access to treatment	Diseases of the skin	

Values in making decisions

Participants were asked to rank what is important for them overall when they make decisions about treatment and care, where 1 is the most important and 8 is the least important. A weighted average is presented in the figure below. With a weighted ranking, the higher the score, the greater value it is to participants.

The most important aspects were “"How safe the medication is and weighing up the risks and benefits"”, and “"The severity of the side effects"”. The least important were “"Ability to follow and stick to a treatment regime"” and “"The ability to include my family in making treatment decisions"”.

Table 9.11: Values in making decisions

Values when making decisions	Weighted average (n=370)
How safe the medication is and weighing up the risks and benefits	6.79
The severity of the side effects	6.26
Time impact of the treatment on my quality of life	5.32
How the treatment is administered	3.83
How personalised the treatment is for me	4.24
The ability to include my family in making treatment decisions	2.93
Ability to follow and stick to a treatment regime	2.98
The financial costs to me and my family	3.68

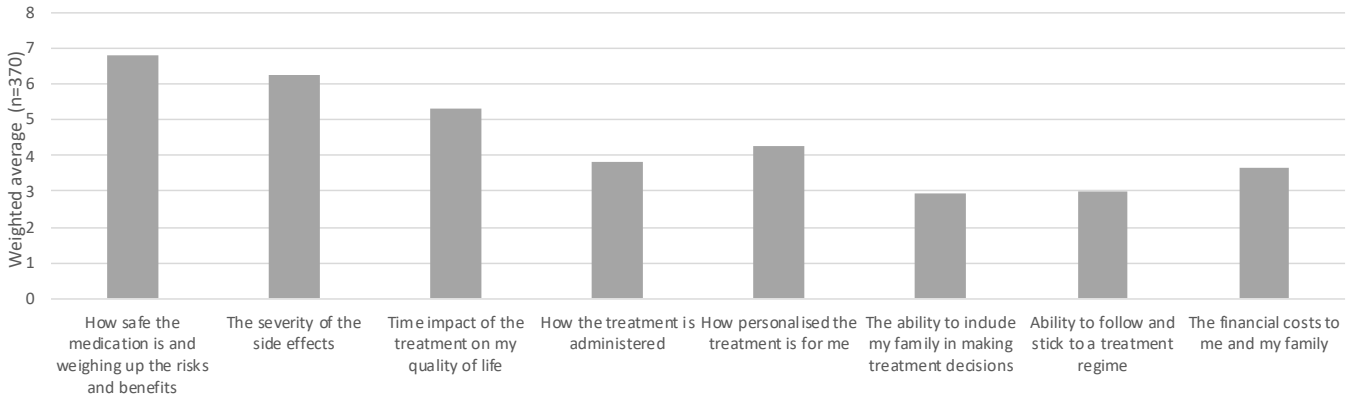


Figure 9.6: Values in making decisions

Values for decision makers

Participants were asked to rank what is important for decision-makers to consider when they make decisions that impact treatment and care, where 1 is the most important and 5 is the least important. A weighted average is presented in the figure below. With a weighted ranking, the higher the score, the greater value it is to participants.

The most important values were “Quality of life for patients”, and “All patients being able to access all available treatments and services”. The least important was “Economic value to government and tax payers”.

Table 9.12: Values for decision makers

Values for decision makers	Weighted average (n=370)
Economic value to government and tax payers	1.56
Economic value to patients and their families	2.49
Quality of life for patients	4.02
Compassion	3.07
All patients being able to access all available treatments and services	3.87

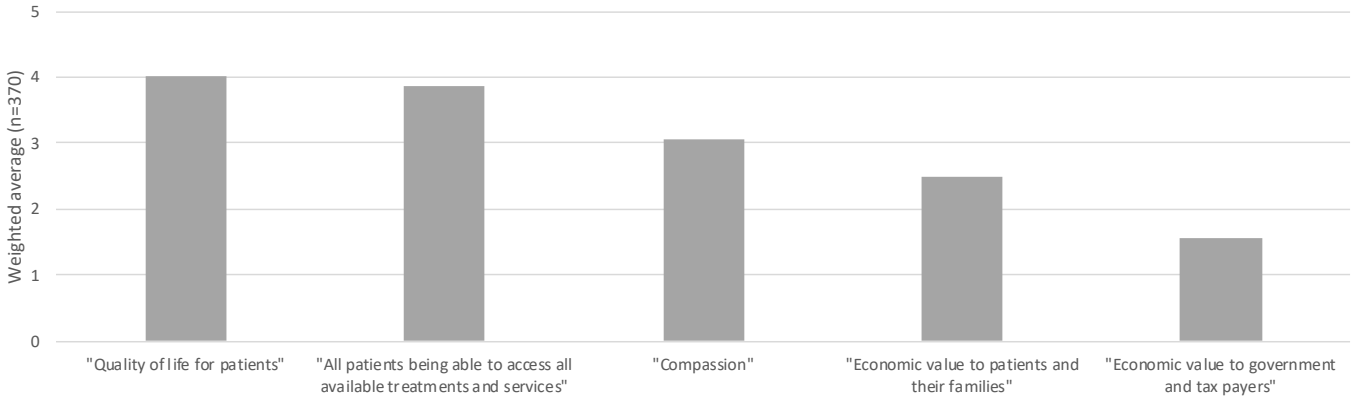


Figure 9.7: Values for decision makers

Time taking medication to improve quality of life

Participants were asked in the online questionnaire, how many months or years would you consider taking a treatment, provided it gave you a good quality of life, even if it didn't offer a cure.

The majority of participants (n = 88, 33.72%) would use a treatment for more than ten years for a good quality of life even if it didn't offer a cure.

Table 9.13: Time taking treatment to improve quality of life

Time taking medication to improve quality of life	Number (n=261)	Percent
Less than 1 year	84	32.18
1 to 5 years	59	22.61
5 to 10 years	30	11.49
More than 10 years	88	33.72

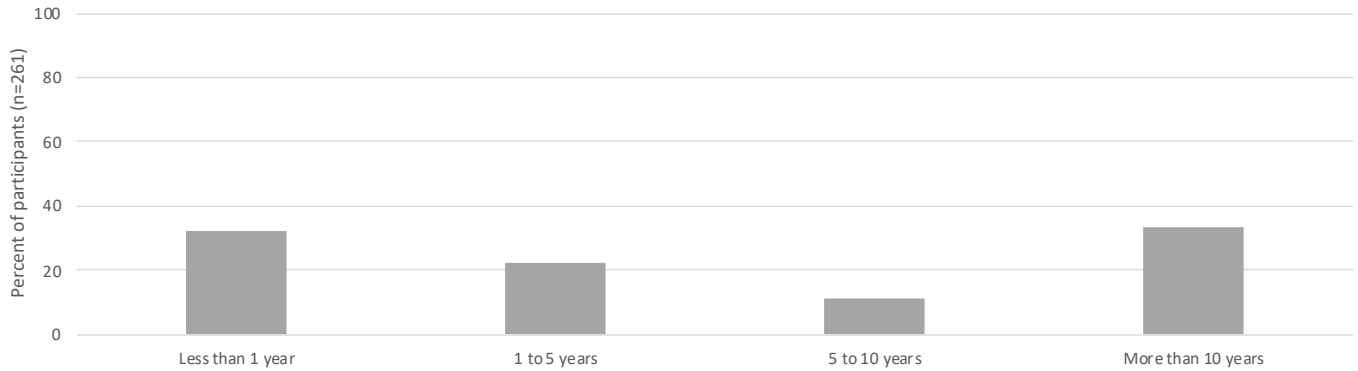


Figure 9.8: Time taking treatment to improve quality of life

Most effective form of medicine

Participants were asked in the online questionnaire, in what form did they think medicine was most effective in.

There were 30 participants (11.11%) that thought that medicine delivered by IV was most effective, 49 participants (18.15%) thought that pill form was most effective, and 74 participants (27.41%) that thought they were equally effective. There were 117 participants (43.33%) that were not sure.

Table 9.14: Most effective form of medicine

Treatment most effective in what form	Number (n=270)	Percent
IV form (through a drip in hospital)	30	11.11
In a pill form that can be taken at home	49	18.15
Equally effective	74	27.41
Not sure	117	43.33

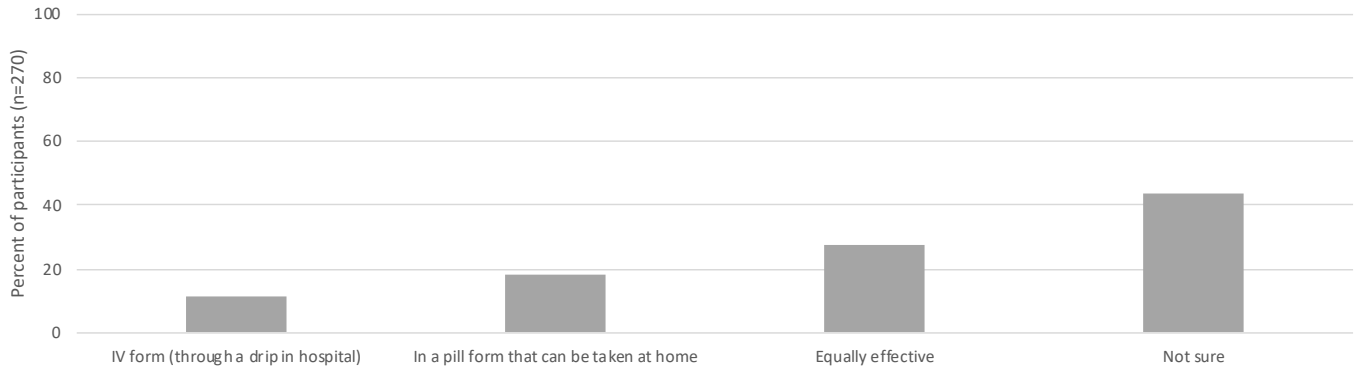


Figure 9.9: Most effective form of medicine

Messages to decision-makers

Participants were asked, "If you were standing in front of the health minister, what would your message be in relation to your condition?" The most common messages to the health minister were the need for timely and equitable access to support, care and treatment (25.87%), the need for more research investment (17.91%), and to help raise community awareness (14.43 %). Other themes included to invest in clinical trials (13.18%), that treatments need to be affordable (10.20%), and to invest in health professionals development (8.96%).

Timely and equitable access to support, care and treatment

I just think that everyone with the rare disease should have the exact same outcomes as other and like medical treatment etcetera and opportunities for medical allied health funding especially in endorse funding should have the same amount as people with well known conditions. Like the child with CP shouldn't be getting six times the amount of my child's funding just because they have cerebral palsy, which is more well known to the general population.
Participant 081_2023AUDIS

I think they should have all the support and care that they need and be given all the resources that they need and support from the beginning of their journey right to the end all.
Participant 010_2023AUORC

I wish there wasn't as many hoops, you know, like even, even when he was younger, to get help from the department that you know does respite care and, and things like that, like we were on waiting lists for that. And the NDIS is the same thing. Like if you don't, if you don't word it properly, you don't get the services that you need. Like it just needs to be more unified and fair so that people who need who have greater needs get them, and people are not routing the system when they're not.
Participant 040_2023AUDPA

More clinical trials and/or new treatments

Oh my God. Well, I've been trying to contact the health minister for the last several weeks about the access. Yeah, has potential access for this life changing drug and so that's my message, that there are 500 children in Australia waiting desperately for this drug, but it's been approved. And that, yeah, he, he obviously, you know, as a teacher, there's a lot of money put into NDIS and it's that the whole system, in my opinion, is being completely abused. And there are children that are just want this drug that will just like change their daily living and their life expectancy and they're crazy. Like it must cost them so much money every time we're hospitalized.
Participant 023_2023AUORC

We definitely need more options in Australia. And as I said that there's a stem cell, well it's gone from trial to the next phase now. So it's essentially it's work essentially they found a cure for it for sure. So…and it yeah… it's a stem cell treatment in the in the States and they're going into pods in, they're trying out to the UK now a couple of other places. So just you know I'd love to see more of that kind of.
Participant 015_2023AUORC

Have access to just that. I said it'd be great to see Australia continue to get access to the new drugs, you know, as they're developed overseas because most people we kind of you hear about them and that the I guess that they're doing enough research and especially with children to be able to get that access for the for children as well and have to wait till you're an adult. Yeah.
Participant 079_2023AUDIS

Help raise community awareness

I think for it to be maybe more prominent and recognized that given no one really is, is that aware of it. I think that the deletion potentially more so, but I think, yeah, just some more awareness about it.
Participant 020_2023AUDPA

I would like to see rare diseases, in general, better known. That's all. I'm very happy with the standard of care and the price of care. I would like more oversight by somebody or somebody who oversees the treatment. I would like more research and knowledge on rare diseases in general and my rare diseases specifically.
Participant 004_2023AUDIS

Awareness, awareness, awareness and an awareness, that is the main thing. Because from that awareness, the treatments and, you know, diagnosis of different things could come through hell of a lot sooner. If they're aware that that could be there, they'd, they'd

come through sooner and the child will be treated quicker.
Participant 024_2023AUDPA

Invest in research (including to find new treatments)

Money for research.
Participant 005_2023AUDNS

Something. Try to find something, research, do something about it. Because I don't think I'm the only one. I think there's quite a few people that suffer from this condition.
Participant 023_2023AUDSK

Oh, as much as possible, I suppose. I would say it would be a general thing. I don't know about scleroderma in particular, because nobody knows. I guess more money for research about the causes and potential cures or improvements in the condition for people. I guess that's what one could wish for, because there isn't any treatment that you can ask for, really.
Participant 012_2023AUDIS

I would say, you need to put more money into HS research and there needs to be more help financially for things like dressings and at home care like for hexidine, those sorts of things.
Participant 001_2023AUDSK

Invest in health professionals to service the patient population

I don't know about specifically for Scleroderma, but I think just in general, like here in CITY, we are lacking in staff and hospital resources and, and, and I know they're aware of that because there's a whole grand plan for the next 10 years. But like, yeah, it would be nice for things to be easier for everyone to access with less like shorter wait lists. And doctors having more time and more staff available.
Participant 024_2023AUDIS

I would try and put it in the perspective of if they had a child or someone that they knew was cystic fibrosis, to think of it from that perspective, because otherwise they're just thinking, oh, just cystic fibrosis, don't really know what it is, don't care. And to think about the costs for people as someone that's already disadvantaged a lot by the sickness and everything that it has done to their life and everything. So trying to use the cost of that. And then also training the doctors for and having specific cystic fibrosis doctors and nurses.
Participant 013_2023AUORC

My message would be if we need more doctors, the wait lists are out of control. My idea would be that there was a coordinating person for complex cases where you're seeing multiple specialists, I guess again, my idea would be that when you're seeing these specialists, you see the same person for some kind of continuity. Yeah, continuity of care is a big factor for NAME. Yeah. She doesn't deal well with seeing new people continuously.
Participant 021_2023AUDPA

Treatments need to be affordable

Maybe to tell him to give free treatment to people with who need the treatment.
Participant 001_2023AUORC

That they should be able to get access, you know, like well, especially in the public system, free of charge. Yeah. There shouldn't be any barriers.
Participant 014_2023AUDPA

So gosh, this put me on the spot. What would I say? I think overall my experience was not a bad one, and I know that there are other people that have experienced much worse with this condition. So for me it's kind of tricky I think. The, the main kind of sore points that came from my experience would have been probably the costs and the time it took and the frustration it took to to get the diagnosis. So if those points could be smoothed out, that would be fantastic. I understand there's lots of issues in the healthcare system with, you know, costs and that kind of thing. There's lots of talk about it, but if it didn't cost me over $600 to get a diagnosis, that would have been lovely. And if I was just listened to by everyone that I spoke to the whole way through, that would have been fantastic as well, yeah.
Participant 027_2023AUDSK

Increase investment (general)

More funding, more funding. Yeah, just, you know, more awareness, more funding. Yeah, it's just there seems to be more awareness in the states in the UK, yeah. And there's a lot of people being diagnosed with it in Australia. So why, you know, why aren't they, you know, looking to more, more funding. I get sicker than …the fact that we have to, we have to virtually do well for anything the public does their own fundraising. It

shouldn't be. The government should be, you know, I'm sorry, there's a lot of people overseas that you know, need when there's a disaster, but start back at home, you know, put more into more into our self funding and our health care. That's what I'd say to them. Yeah.
Participant 019_2023AUDIS

Please give us some funding and start recognizing that there's more and more people being diagnosed with it and we need help to-- Have professionals where these people can go so that mentally they're not crumbling. Just please help. Please help. Give them the information they need and don't let people suffer like they're suffering. I'm not as bad as a lot of them, so I think I'm doing all right, but some are really terrible.
Participant 007_2023AUDIS

I think I could probably talk about a lot of things and it's probably not cardiomyopathy per se that only on its own. It's probably everyone with a heart condition or a cardiovascular disease or whatever it is that affects your cardio pulmonary activity, I guess it is more so than anything. Like I, I don't think people understand the cost of like not personal cost, but the cost to society and community that these conditions sort of bring to, you know, the state or the federal government or whatever. It's like it's astronomical and I don't understand why more isn't spent in the way of prevention and management so that it's not a matter of treating people at the most vulnerable, weakest and most detrimental point in their life where they need the most expensive and the most care.
Participant 030_2023AUORC

Take the condition seriously

I guess maybe just greater recognition of what, what the condition is and what it entails for the people that do have it and like I said, it is such a broad area of issues that can crop up for different people and, and greater knowledge of what the condition entails.
Participant 027_2023AUDPA

I think my predominant message would be people with complex disabilities are not the same as people with a list of all those disabilities just sort of added up because for a lot of people with charge, you know, it's like, oh, well, they're not quite deaf enough or they're not quite blind enough to receive this service or, you know, whatever it is, but it's like. Yes. But the impact of all of those senses being impaired and erratic means they need so much extra support.
Participant 018_2023AUDPA

Everybody's different. Every disease is different. The issue we have is that some of those diseases are invisible and if you can't see it happening in the system, if you can't see the person-- I walked in and I had my leg from my toe up to my hip, people would look at me and go, "Okay, you've got that's you've got serious issues," or you've got braces on or whatever up your legs because you can't walk or you're having something else as an aid to help you to walk or whatever. They treat people differently if they've got those sorts of things to what is actually happening to other people like people who have invisible diseases.
Participant 005_2023AUDPA

Invest in screening/early detection

Well, the diagnosis, the blood test that's really good is because of she was, I think it had only been developed then when NAME got it. I don't think it had been developed before that. So she couldn't have probably got it any earlier, I think. Just I guess. The doctors to be more aware of looking for these causes when, when a person presents with something a bit different if they get more look a bit harder and have a more coordinated plan and I just if the it's impossible for them to do this but just it'd be so good if doctors could just stay so that you can have the one doctor and not just be jumped from one doctor to another. And consistency in the health care is important, but that's probably out of the hands of government and just more psychology services in rural areas.
Participant 09_2023AUDPA

Health minister, [silence] gosh. [silence] I guess it would be just raising awareness. I think I would be fortunate in my diagnosis. The only delay was access to specialists, whereas some people I think, struggle for years trying to get a diagnosis because it is rare and people aren't aware. I guess raising the awareness and the skill level because I think the earlier the diagnosis, the better the outcomes.
Participant 017_2023AUDIS

Yeah, again, a better screening. Know if you call it screening policy or screening system for these kids, because they are mostly nonverbal, they cannot communicate, the behaviour is communication and sometimes they will have pain and they won't be able to articulate or show you that. A lot of GI issues, for example, get they're not even known about for many years until they get older and they're already struggled so. Better screening like I said, the early vision testing is very important, just a more specific where those these children go to learn. For example,

like would it be possible to have there's a very well established deaf schools, I'm not sure if there's as well as established deaf blind schools for example, if there is that haven't been told. Yeah, yeah, yeah.
Participant 094_2023AUENM

Compassionate and empathetic

Need more? I don't know. I don't know what I would say, actually. I just, it needs more spotlight. I think if you know, like if you really look at a lot of people, you would see it, you know, a lot more people. I think a lot more people just manage the symptoms, don't even know what's wrong with them. I didn't know 35 years what was wrong with me. So I think it's something that needs spotlight and needs to be people need to learn what it is, what to look out for. And then, you know, we need to be listened to. None of this. It's anxiety go away or true food go away, or they go on a liquid diet. Go away. Like just need to be more heard and more respected, you know, like and listen to compassionately. And not just in a textbook. But it's hard because I guess everyone is slightly different too.
Participant 078_2023AUDIS

A bit of improvement. Well I guess the first thing would be have to change the, the mentality of the of the whole system which with doctors training for a start. So I don't think they're God's gift and, and then just professional drug dealers. You've got to change that and have some empathy for actual people who are dealing with shit rather than just, you know, write out scripts and then turn over, turn over pills for money. That is probably the, the most important part because nurses are better than doctors by country mile, you know? So start there and then, you know, might have a bit of a chance, yeah.
Participant 006_2023AUDIS

Invest in professional development so that clinicians understand the condition

I think that would be, what I would say is that they need to educate the doctors that it's not just some skin thing because I think the name Scleroderma. It, it puts them in that mindset of it's a skin thing when the skin part of it is negligible. Yes you know and, and so the name of the - and I'm not saying that they must go and change the name of it - but the name of it gives the illusion that it's a minor problem that's easily fixed and it's not. And they don't, they don't really know that. And next you're the rheumatologist, but if you any other doctor who interfaces with the person with Scleroderma, they don't give it any gravitas at all. It's like, OK, so you've got Scleroderma. OK, well, now I'm talking about your kidneys today. I'm talking about your heart or I'm talking about, you know, they kind of OK, we don't need to talk about that. Where is a lot of my things could, quite well, the originating starting point of it was the scleroderma that they're just treating the symptom now. They're not treating the cause and they're not treating it holistically. They're just doing their tick and flick of their one particular thing and then they're going to go and have lunch, and that's as far as it goes.
Participant 002_2023AUDIS

Oh God, yeah. I mean, I I've dealt with ministers before and they have no idea about most things. What would be my access to just so that there's so many different diseases out there which they know nothing about. And I think it's, it's you can't cure everything but you can. It's always about money, isn't it? The way it says, well, can you please give more money and educate people? But I don't know. And it's mainly educating the doctors, you know, get them more educated and the nurses. When I had my, when I had my hip thing, I think nobody knew in the hospital what an allograph was and they'd never had anybody have an allograph before and somebody was fine with a thing on it and it's just unknown, so. I suppose with rare things you can't. How do you speak to the just make more money available, I suppose for research.
Participant 024_2023AUDSK

Understand the financial implications (and provide financial support)

Make it affordable. People with chronic illness are penalized greatly because it's just not affordable.
Participant 016_2023AUDIS

Well. They need access. Oh, that's one they need. They need subsidised access to, to dressings. They need extra sick leave. They need additional sick leave to for for their, their flares. Well, I don't care if it has to be that it has to be a doctor, certificate one or whatever, but they need additional and they need because it is a, it's a, it is a hidden disability and it needs to be much more made, much more aware of in the workplace that people have hidden disabilities and they get special consideration, parking, everything. Disabled parking is another one because when they're having an active flare they are as disabled as anybody else who is disabled. But when they're not having an active flare then yeah they they, they shouldn't be using it. But I think access to disabled parking when required should be available.
Participant 009_2023AUDSK

I would like to say that don't forget our little rural communities and that our people that do suffer these rare conditions are really isolated more so because of our distance from everyone. They need to understand they need to be supportive. If someone does have to travel like us, have to do a 2,400-kilometer round trip for treatment, they need to be a little bit more supportive on the accommodation and that they can offer also the financial support with accommodation because that's something that they didn't do at the time when I was going down there. They wanted you to go down and back in a day and then travel over 100 kilometers to get home. It'd be different if we lived in those areas like Townsville all that, where you you just drove to your house there. Then we also, besides the flight then had to drive as well. I think if they could take that into consideration for people in the future, that would be great.
Participant 006_2023AUDNS

Grateful for the healthcare system and the treatment that they received

I'd say overall with very good. It's very good. I think a lot of what. We received in the first few years was about managing acute health issues and probably reactionary to what was going on and we received very good care, yeah, very good surgery, very good doctors, very good, mostly good hospital experiences. I think there probably could also be more focused on early intervention and I guess not just reacting to the health issues but also focusing on preparing for the future too, which only came later. The solving, once we work through the other health things that we're dealing with initially, yeah, I think that's pretty much it.
Participant 089_2023AUENM

What would I say to him about what I've had? I'd say thank you.
Participant 018_2023AUORC

Patient input and engagement in all aspects of decision-making

PARTICIPANT: How to express this? I'm not finding the right words. Targeted things that engage in proper community consultation to work out what's actually needed.
INTERVIEWER Yes, okay. Like reference groups type thing?
PARTICIPANT: Which pretty much is what you're doing.
Participant 001_2023AUDPA

I'm bighting my tongue quite a bit there because I don't need to hear your ear off for the next hour about that, but we also don't. But you know what? I also believe we need to do a in education of doctors and clinicians. Lived experience needs to be part of the curriculum of when they're educated about different things. Like when you're becoming a GP, let's bring in some lived experience people to tell you…how they geneticists, genetic counselors, come and listen to the stories so that you don't make those same mistakes. Because these families are human beings with emotions, and the way you give a diagnosis could actually determine the path or trajectory they follow.
Participant 025_2023AUDPA

Holistic approach to the condition (including emotional support)

It needs to be family centered care. You need to understand that person is part of the family and the specific context of that family and how you can improve that individual's life but also support the family around that.
Participant 090_2023AUENM

I would actually tell him that we have access to advices and, and the views relating to mental health issues and we also have access to the way to cure and go about the virus and we always have access to knowing that someday somehow you would actually be cured here.
Participant 009_2023AUORC

Yeah, just that holistic care is, is really important and also just awareness by health practitioners on what 22 Q, what that involves, it's really helpful. I do find I have to educate everybody and you know that's a bit bearing in mind it's actually pretty common. I think, you know, I would advocate for money towards research like what you're doing, you know, potentially have capacity in the LOCATION health research system to be able to do linked to studies here in LOCATION, possibly not elsewhere because you can track kids a lot better here. And so, yeah, I would advocate for research money and and awareness and integrated services and also just acknowledgement of, yeah, when kids have a lot of minor symptoms that they add up to being a major symptom.
Participant 035_2023AUDPA

Need for multidisciplinary and coordinated care}

I would say to him grateful for all that we have, but there is a huge room for improvement around coordinated care with all of the different specialists and access of care and treating the patient like a person in the room as well, particularly when they're a child.
Participant 095_2023AUENM

How that would be done on not really for me to say and consistency of consistent care, not spasmodic and obviously more funding, more funding for research and awareness.
Participant 08_2023AUDPA

It's, it's a lifelong condition. It's not something that can be fixed. Our experience, I guess, is a lot different to other people's way they have to physically pay. I mean, they pay for lots of things for NAME, but if people have a lifelong condition. Yeah, make it the services available to them.
Participant 015_2023AUDPA

Table 9.15 Messages to decision-makers

Message to decision-makers	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Timely and equitable access to support, care and treatment	104	25.87	11	16.42	26	32.10	19	20.00	7	21.88	27	28.42	14	43.75	80	29.85	24	17.91	78	26.53	25	23.58
Invest in research	72	17.91	5	7.46	17	20.99	9	9.47	14	43.75	23	24.21	4	12.50	62	23.13	10	7.46	55	18.71	17	16.04
Help raise community awareness	58	14.43	7	10.45	22	27.16	3	3.16	6	18.75	15	15.79	5	15.63	50	18.66	8	5.97	49	16.67	9	8.49
More clinical trials and/or new treatments	53	13.18	0	0.00	3	3.70	34	35.79	1	3.13	10	10.53	5	15.63	32	11.94	21	15.67	41	13.95	12	11.32
Combined minor themes	50	12.44	0	0.00	2	2.47	19	20.00	0	0.00	5	5.26	0	0.00	17	6.34	9	6.72	20	6.80	6	5.66
Treatments need to be affordable	41	10.20	6	8.96	8	9.88	4	4.21	5	15.63	12	12.63	6	18.75	31	11.57	10	7.46	29	9.86	12	11.32
Invest in professional development so that clinicians understand the condition	36	8.96	5	7.46	9	11.11	7	7.37	8	25.00	5	5.26	2	6.25	28	10.45	8	5.97	28	9.52	8	7.55
Invest in health professionals to service the patient population	33	8.21	5	7.46	7	8.64	0	0.00	1	3.13	16	16.84	4	12.50	22	8.21	11	8.21	27	9.18	6	5.66

Message to decision-makers	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Timely and equitable access to support, care and treatment	104	25.87	17	17.53	38	29.01	30	26.32	19	31.67	46	23.23	58	29.59	25	22.52	79	27.15	49	24.50	55	27.23
Invest in research	72	17.91	6	6.19	25	19.08	27	23.68	14	23.33	41	20.71	31	15.82	21	18.92	51	17.53	40	20.00	32	15.84
Help raise community awareness	58	14.43	6	6.19	23	17.56	20	17.54	9	15.00	35	17.68	23	11.73	23	20.72	35	12.03	38	19.00	20	9.90
More clinical trials and/or new treatments	53	13.18	18	18.56	16	12.21	10	8.77	9	15.00	29	14.65	22	11.22	10	9.01	43	14.78	25	12.50	28	13.86
Combined minor themes	50	12.44	7	7.22	5	3.82	10	8.77	4	6.67	11	5.56	14	7.14	7	6.31	19	6.53	12	6.00	14	6.93
Treatments need to be affordable	41	10.20	4	4.12	16	12.21	12	10.53	9	15.00	21	10.61	18	9.18	14	12.61	27	9.28	21	10.50	20	9.90
Invest in professional development so that clinicians understand the condition	36	8.96	4	4.12	15	11.45	12	10.53	5	8.33	17	8.59	19	9.69	12	10.81	24	8.25	22	11.00	14	6.93
Invest in health professionals to service the patient population	33	8.21	7	7.22	3	2.29	20	17.54	3	5.00	13	6.57	20	10.20	13	11.71	20	6.87	18	9.00	15	7.43

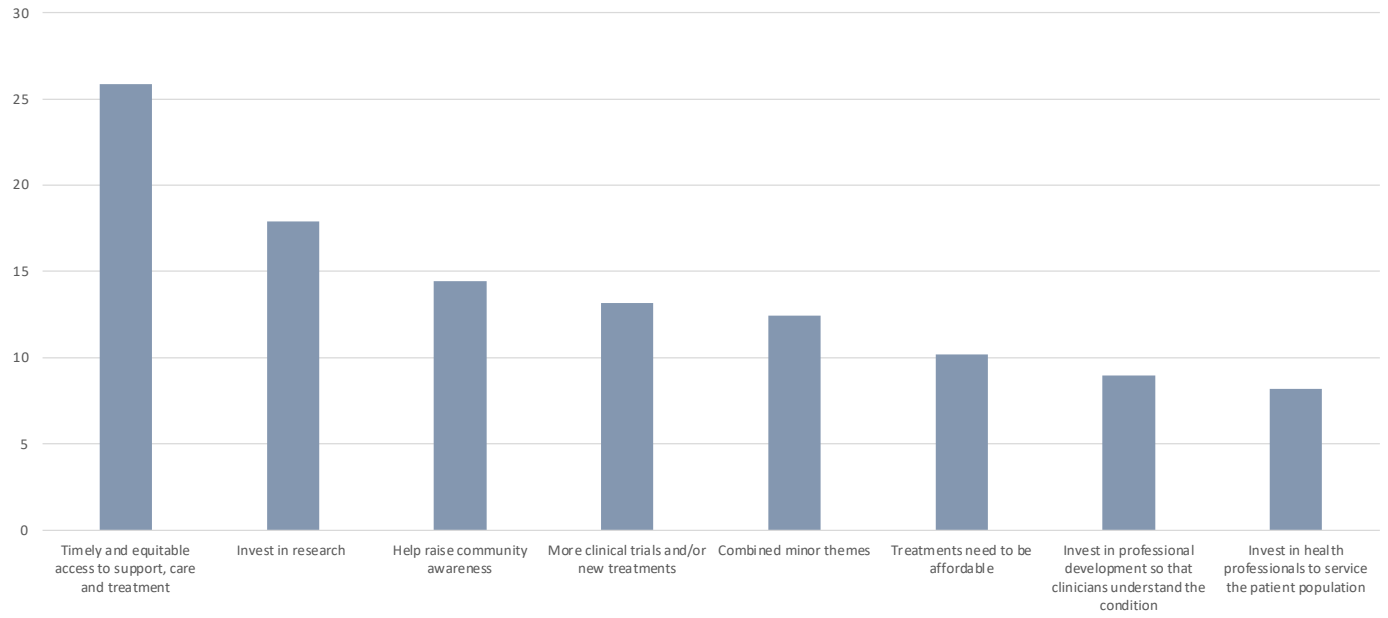


Figure 9.10: Messages to decision-makers

Table 9.16: Messages to decision-makers – subgroup variations

Message to decision-makers	Reported less frequently	Reported more frequently
Timely and equitable access to support, care and treatment		Other rare condition
Invest in research	Developmental anomalies Family or carer Aged under 18	Diseases of the skin
Help raise community awareness	Diseases of the nervous system	Diseases of the immune system
More clinical trials and/or new treatments	Developmental anomalies Diseases of the skin	Diseases of the nervous system
Combined minor themes	Developmental anomalies Diseases of the skin Other rare condition	
Treatments need to be affordable		
Invest in professional development so that clinicians understand the condition		Diseases of the skin
Invest in health professionals to service the patient population		

Section 10

Advice to others in the future: The benefit of hindsight

Section 10: Advice to others in the future

Anything participants wish they had known earlier

In the structured interview, participants were asked if there was anything they wish they had known earlier. The most common things that participants had wished they'd known earlier were to be assertive, an advocate, informed, and to ask questions (32.09%), to seek and accept help, including peer support and support groups (16.92%), to understand the trajectory of the disease (13.68%), and to try to stay positive (11.19 %).

Aspect of care or treatment they would change

In the structured interview, participants were asked if there was any aspect of their care or treatment they would change. The most common themes were that they would have liked to have had access to a specialist in their condition sooner (15.41%), that they would not change any aspect of their care or treatment and were satisfied with care and treatment received (13.16%), and they would have liked health care professionals to have had more knowledge and awareness of their condition (10.53 %). Other themes included they would have stopped or changed treatment sooner (7.89%), (5.64%), and they would have liked to have been diagnosed sooner (3.76%).

Anything participants wish they had known earlier

In the structured interview, participants were asked if there was anything they wish they had known earlier. The most common things that participants had wished they'd known earlier were to be assertive, an advocate, informed, and to ask questions (32.09%), to seek and accept help, including peer support and support groups (16.92%), to understand the trajectory of the disease (13.68%), and to try to stay positive (11.19 %).

Participant wishes they had known to be assertive, an advocate, informed, and ask questions

No, I think for me it's just about knowing how to navigate the hospital system, but you don't really know that until you're in and you experience it. And in saying that, it's because in order to get care for my child in the hospital system, I need to be very knowledgeable about what to ask for. Because there is only that one specialist team that knows what to do. And if they're not available and these guys are on call 24 hours a day, seven days a week, if they're not available or if there's a minor hiccup, then that can be life threatening for my child. So I think for me, if there's anything I wish that I knew it would just be about how to be really assertive and have the information to provide anybody at point of care around what my child condition is, and I've learned that over time now.
Participant 021_2023AUORC

I think I would have just liked to have known more about, yeah, self advocacy and communication than how to ask questions as doctors and be prepared for those kinds of things. Yeah, that would have been, been helpful.
Participant 024_2023AUDIS

Yeah, well, I reckon I've had optic neuritis between 10 and 15 times in the last or since about 2015. I wish when I first had it, I had sort of pushed for more tests to find out why. And I could have started on all these preventative drugs, you know, eight years ago rather than last year. Yeah, I wish I'd sort of known that. I always just thought I had optic neuritis flare ups and that was it. I wish I'd known they could have actually been causes and it could have been prevented or tried to be prevented.
Participant 096_2023AUDNS

Participant wishes they had understood the trajectory of the disease

I wish…yeah, I, I wish I would have known that he would be able to make progress in terms of moving and communication and hearing and vision. So yeah, all things that could have been helped at the time if I'd had more information. But yeah, I guess just that, that things could progress and get better. I wish I could have known that at the time.
Participant 089_2023AUENM

Yes. I wish I'd known that it was a significant problem and not just something that I could take one tablet twice a day and it would go away.
Participant 002_2023AUDIS

Would be nice to have known that. Well, I guess I knew from the beginning there's no cure. I would just like…be a cure so that…go through what we go through, yeah.
Participant 019_2023AUDIS

That 22 Q is so broad. Every single kid is unique. They do not know one kid shares the same set of symptoms. For 22 Q, you know it's not a death sentence. Yeah.
Participant 021_2023AUDPA

Participant wishes they had known to try to stay positive

No, it's probably just the old cliche of it's not as bad as it sounds. Once it becomes your routine, it's not. It's not the end, but no one's ready to hear that in the beginning. Like there's probably hundreds of people that did say that, but you're not ready to hear it at that time. So but you think this sort of, what's that?
Participant 025_2023AUORC

It gets easier. That's probably it. You do get the hang of it.
Participant 032_2023AUDPA

Yes, everything's got to be OK and everything will work out in the end. Yeah, that's quite I wish that we had known about the diagnosis earlier because we would have, it would have just opened a lot more doors to us in her early childhood and early schooling life.
Participant 037_2023AUDPA

Participant had no particular comment and were satisfied with experience

No because the issue with getting on with life, if you know "Okay," basically, sometimes you look at what- - As I said, there are people worse off than me. If they're having the gene dilution, having a gene mutation is basically a death sentence. That's actually what some people…sometimes, that is what I've thought about and this is why I'm actually the way I am. I think what I am at the moment is a miracle because as I said, if I had the mutation I wouldn't be alive. The point is it's the way you approach life is what matters and I get my ups and downs and all the rest of it. If you know the answers, then you're not going to ask the questions and if you're not going to ask questions, then you're not going to enjoy life.
Participant 005_2023AUDPA

Not, not necessarily. I think I'm I I've committed myself to learning more as and, and being involved more in things like clinical studies, I'm actually quite excited that there is a clinical study happening in Australia that's amazing. So yeah, so I think just yeah for me it's it's, it's been a continual, continual growth, continual learning and continue understanding about the condition and you know if there's more opportunities to do that then I'll be involved in that as well.
Participant 001_2023AUDSK

No, not really, because we were sort of on top of it ourselves and really researched what the options were. And I was actually the one that asked whether I could have an ablation to try and fix the problem. So that that was relatively early on in the diagnosis. I think that was like my third visit to the hospital. So I think, I don't know, I guess the natural progression of things they have to try medications to see if they work. So I think, yeah, no.
Participant 032_2023AUORC

Participant wishes they had been diagnosed sooner

Yes. I wish I knew what my condition was as a teenager. I wish I'd been taken seriously at 17 when I first saw a rheumatologist because I feel like that would've guided choices around even just like work choices or when I went to backpack around Europe. I would've made more sensible decisions on like luggage, or transport and just things. I feel like since diagnosis, now that I understand what it is, I know what to look for now and I'm just a bit more preventative and that's made a huge impact on quality of life. I feel like if I'd known sooner when I had all the symptoms then I feel like that would've been really helpful so I wouldn't have physio giving up on me because I wasn't responding to treatment how they thought I should. For example, I'd be able to say, "No, this is going to take longer" or "I just need to do this more slowly" or whatever
Participant 004_2023AUDPA

I just wish I'd been diagnosed earlier. I honestly think if I had been diagnosed with this when I had my first flare and then put on. The medication and the creams and had the area removed, I don't think that it would have progressed and I don't think I would have it anywhere else. I don't think I'll be sitting here talking to you today. I think because it took 15 years to diagnose and it was well rooted in my system and in my skin by that point that now it's a drama and nobody knows what they're doing and they're leaving bits and bobs when they cut it out and not getting it all.
Participant 006_2023AUDSK

Participant wishes they had known to ask for a second opinion or speak up during consultations

Second opinion. Getting a second opinion is really. What I could have been doing some stages. It's never too early to start intervention. It's never too early to start. It's never too early to start planning for the future. I think it's probably one of the most important takeaways from my journey. Yeah, definitely thinking more about the future I like. It's really hard in those early days, but seriously. What do you want for your kid in the future? Do you want him to be still, living at home and depending on you when they're 21, applying everything to that goal? Yeah.
Participant 028_2023AUORC

No, I just wish the rheumatologist that said you got skin thickening but you don't have Scleroderma wasn't such an idiot and actually said yes you got Scleroderma because you did ultrasounds on my arm and every and he said, 'oh, I see your skin. It's thick'. And he showed on the nurse who's doing the ultrasound. They put it on her arm and said I'll see how hers is only this thick. Yours is like twice as over, twice as thick, blah blah. And he said that you don't have Scleroderma, you just got skin thickening. So I was like on the way home in the car. I'm going. I'll look it up when I get home.
Participant 011_2023AUDIS

Participant wishes they had known the early signs and symptoms of their condition

Yes. I wish I knew the effects that it had on you. I probably maintained my health a lot better in that sense of what knowledge of what in the future. You

only look at the daily things, but you don't look at future. I think if that made me aware of more of the symptoms that I had, it probably could have been a lot better for me in the long run. Symptoms, particularly.
Participant 001_2023AUDIS

Of course, when I was 30 years old, somebody come along and say this is your trouble and this was why you've got these skin lesions and that's how we'll treat them.
Participant 003_2023AUDIS

Well, I wish I'd known back when I was having all those rashes that could have been…my rheumatologist later said, "Oh, yes, well, you can get that with scleroderma and that would've been in the active phase." Well, I spent years suffering with itchy skin, scratching, making myself bleed, and not being able to sleep because I was so itchy. That was nobody's fault that wasn't picked up, I think because who would've thought. I don't know. I hope somebody can change that.
Participant 004_2023AUDIS

Participant wishes they had known to look after emotional wellbeing

The emotional impact being diagnosed at a very formative age of 18, I think that there could have been a bit more intervention about the emotional impact. That I carried on my own for so long.
Participant 004_2023AUORC

Anything I know now well, for start that there was there would be some, some improvement, yeah. The importance of importance of of managing the whole person when they develop a sudden chronic illness like yeah the importance of that. It probably takes a team to to manage a chronic health person not just one doctor giving drugs that it probably. And that that team should be more interlinked rather than me having to go off and try to source this other help that was not linked with my that there wasn't a whole treatment plan. But I accessed mental health independently. It took me a long time to find effective mental health, but they still they still didn't. There was no link, real link between that and medical help, you know.
Participant 027_2023AUORC

Participant wishes they had known to pace themselves or know triggers and limitations

I think I've mentioned this before, but definitely like food eliminations because I didn't know when I was younger when I first had this disease, that certain things can make it worse. I also smoked when I was, you know, 16, 17, which is. Quite young, but I smoked cigarettes and I didn't realize that that was something that can aggravate the disease more, which I now do not smoke and I've seen improvement, slight improvement in the disease. So I guess just a bit more information about how to help or keep the disease calm.
Participant 010_2023AUDSK

I feel like, yeah, I feel like it would have helped me knowing a lot earlier as to, you know, what the causes are, how I can prevent them. And being at a younger age where I was probably a little bit more in control of my life and then control of my body. I mean, it's still something I can do. But, you know, I've got kids and family and money restrictions now, so yeah, yeah
Participant 011_2023AUDSK

Wow, a lot of things. I'm sure. I can't really think of them right now. Probably that…pace yourself. It's definitely a marathon. And just listen to your gut as well. Like people will be offering different things and just sort of know, you know, your child, don't, don't let people tell you that you don't. And trust yourself. I think it's all a bit woohoo, but I think that's what I wish I'd known. It's just, you know, it's not the sprint and just to pace yourself.
Participant 095_2023AUENM

Participant wishes they had known to seek and accept help, Including peer support and support groups

I wish that my neurologist, when he had told me that it may get worse, it may get better, he would have also told me that there's no treatment and there's no cure, but it's getting researched…Just like the fact that dystonia is very common, but very uncommon at the same time…Because I just felt alone, never heard of it, never knew anyone. I ended up making a social media account to connect with people. That was the first time I'd ever spoken to other people who had it.
Participant 004_2023AUDNS

It would've been helpful to see some more lived experience stories of what life looks like for people at different life stages and how they grew up. That would've been comforting even though obviously everyone's journey and experience is different. That would've been good information, having more information around the supports and typical funding that would be available, so that you know what pathways to pursue rather than having to do your own research about that.
Participant 067_2023AUDPA

Table 10.1: Anything participants wish they had known earlier

Anything participants wish they had known earlier	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Be assertive, an advocate, informed, ask questions & trust your instincts	129	32.09	4	5.97	32	39.51	26	27.37	13	40.63	42	44.21	12	37.50	96	35.82	33	24.63	100	34.01	29	27.36
Seek and accept help (Incl. peer support/support groups)/knew where to go for help	68	16.92	6	8.96	4	4.94	14	14.74	0	0.00	44	46.32	0	0.00	45	16.79	23	17.16	51	17.35	17	16.04
Understand the trajectory of the disease, including costs to expect	55	13.68	9	13.43	24	29.63	17	17.89	1	3.13	2	2.11	2	6.25	42	15.67	13	9.70	47	15.99	8	7.55
Try to stay positive	45	11.19	3	4.48	2	2.47	22	23.16	0	0.00	16	16.84	2	6.25	31	11.57	14	10.45	33	11.22	11	10.38

Anything participants wish they had known earlier	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Be assertive, an advocate, informed, ask questions & trust your instincts	129	32.09	24	24.74	46	35.11	40	35.09	19	31.67	60	30.30	67	34.18	37	33.33	92	31.62	59	29.50	70	34.65
Seek and accept help (Incl. peer support/support groups)/knew where to go for help	68	16.92	15	15.46	13	9.92	19	16.67	21	35.00	28	14.14	35	17.86	18	16.22	50	17.18	25	12.50	43	21.29
Understand the trajectory of the disease, including costs to expect	55	13.68	8	8.25	20	15.27	19	16.67	8	13.33	26	13.13	29	14.80	18	16.22	37	12.71	28	14.00	27	13.37
Try to stay positive	45	11.19	13	13.40	13	9.92	14	12.28	5	8.33	24	12.12	21	10.71	9	8.11	36	12.37	20	10.00	25	12.38

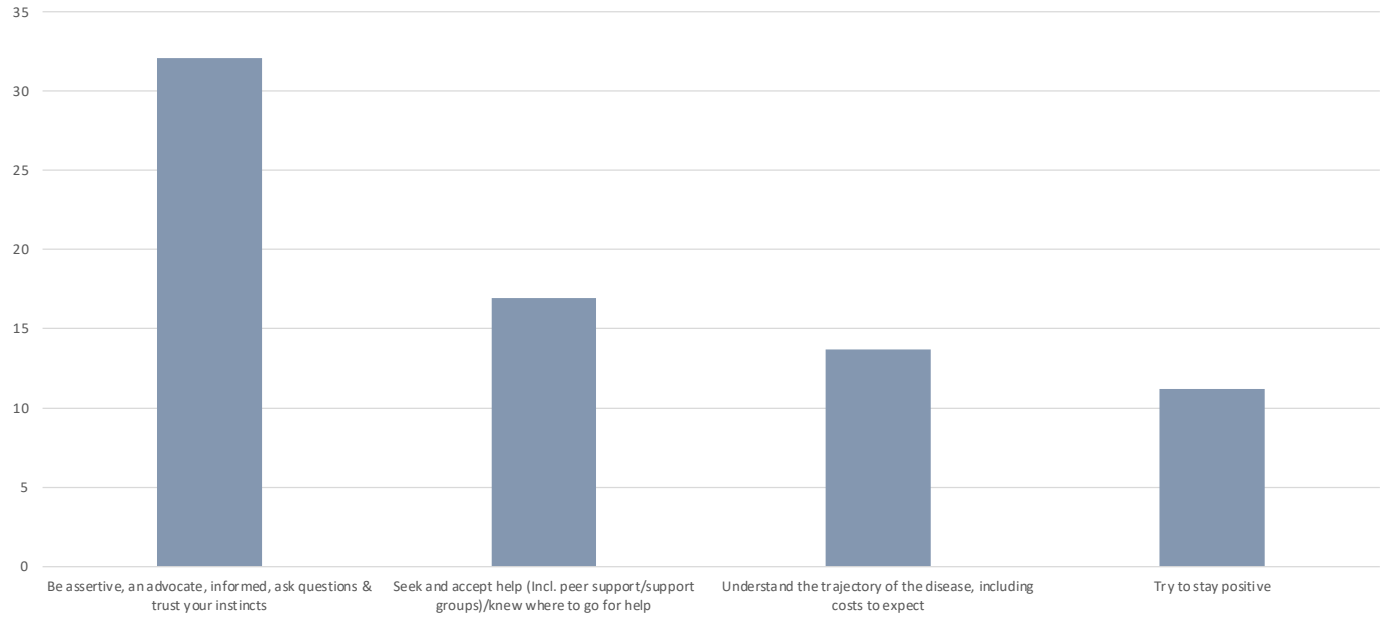


Figure 10.1: Anything participants wish they had known earlier

Table 10.2: Anything participants wish they had known earlier – subgroup variations

Anything participants wish they had known earlier	Reported less frequently	Reported more frequently
Be assertive, an advocate, informed, ask questions & trust your instincts	Developmental anomalies	Endocrine, nutritional or metabolic diseases
Seek and accept help (Incl. peer support/support groups)/knew where to go for help	Diseases of the immune system Diseases of the skin Other rare condition	Endocrine, nutritional or metabolic diseases Aged 65 plus
Understand the trajectory of the disease, including costs to expect	Diseases of the skin Endocrine, nutritional or metabolic diseases	Diseases of the immune system
Try to stay positive	Diseases of the skin	Diseases of the nervous system

Aspect of care or treatment they would change

In the structured interview, participants were asked if there was any aspect of their care or treatment they would change. The most common themes were that they would have liked to have had access to a specialist in their condition sooner (15.41%), that they would not change any aspect of their care or treatment and were satisfied with care and treatment received (13.16%), and they would have liked health care professionals to have had more knowledge and awareness of their condition (10.53 %). Other themes included they would have stopped or changed treatment sooner (7.89%), (5.64%), and they would have liked to have been diagnosed sooner (3.76%).

Participant would not change any aspect of their care or treatment and were satisfied with care and treatment received

No, no, wouldn't I think that I was provided with the best information at that given time. I have always been taken seriously by all the doctors that I saw. I can't fault them.
Participant 026_2023AUDSK

No, I don't think so. I think, I think they, they did everything really well for us. They met us where we needed to be met and I think, I think they've I think like the hospital have done a really good job. And on our side, we've done a really good job behind the scenes as well of keeping him on track and out of hospital admissions and well.
Participant 025_2023AUORC

No, no, I think, I think I made the choice, the right choice to go into a private hospital, not a government hospital. I'm, I'm very certain about that. I'm so bad. I insisted on that first night that the ambulance take me to a private hospital. Because I think I was in the best place I could possibly wish for, but I don't know. In my treatment, I don't know. I don't know. I was, I was quite confident with the clinical haematologist who I dealt with. She was fantastic. I had every, every respect for her. But as to…you know, there's certain aspects of my hospitalization that I'd change, but my treatment and management I don't think I've got reason to be concerned.
Participant 095_2023AUDNS

Participant would have liked to have access to a specialist in their condition, sooner

The only thing I would change is pushing to receive treatment a lot sooner, before it progressed to the stage that I'm at. But you know, when I was younger, I didn't have a choice. It was up to my parents to kind of look after me and and seek better help, but if I could go back I'd probably push harder to my parents to get me to seek.
Participant 010_2023AUDSK

Something that made me better. That's what I will change if I can find something that will improve the condition. Yes, by all mean, I close my eyes. I gave the study 10 minutes. These people don't know, they just don't treatment. As I said, I've seen so many doctors, so many specialists, no one seems to know or have they any idea about HS? So if you do know of someone that has got some idea, do send me a test message or an e-mail with the name and I wouldn't make an appointment to go and see them.
Participant 023_2023AUDSK

I would have gotten certain interventions earlier in terms of probably feeding therapy. Yeah, I think that's it. Just earlier intervention, possibly earlier OT intervention as well if I would have, if I could change things done more sooner, yeah.
Participant 089_2023AUENM

Participant would have liked health care professionals to have had more knowledge and awareness of their condition

Treatment. There needs to be a lot more research into it. It's not extremely rare and common condition. Now it needs more. More people need to look into it. The care I received, I can tell my two current providers the care I received. It could have been a lot better. It left a lot to be desired and that comes down to a lack of information available to medical practice to medical professionals on the condition. I've had to still go to the hospital tomorrow and I've been to the hospital multiple times for this condition and people still ask me, oh what's it? It just needs more awareness.
Participant 012_2023AUDSK

No, I don't think so. No. Other than that…if there was a GP that knew about scleroderma, again, because I don't see them that frequently, I would travel for that, but short of that, yes, someone who's going to advocate for you every time I'd take, and is willing to learn with you about what's happening.
Participant 017_2023AUDIS

Participant would not change any aspect of their care or treatment, with no reason given

No, I don't think so. No.
Participant 010_2023AUORC

No, I don't think so. I think we sort of went through the usual treatment option failure, yeah.
Participant 019_2023AUDSK

Participant would have stopped or changed treatment sooner

Maybe just coming away from your steroids really like finding some sort of treatment that's not steroid based.
Participant 078_2023AUDIS

No, I don't think so. I probably feel like I should have been offered more drugs, but I don't get off anything stronger than Panadol for pain and I don't even know if that's a good or bad thing. Anyhow, I'd certainly go back to my GP and tell him that I want to get some

steroids into me, but they're all scared to give you anything, I think.
Participant 013_2023AUDIS

I was on Prednisolone for so long that I have ongoing health issues from that. So weight, hair, like you think of all the side effects of Prednisolone. It's not pretty. So if I had my time again and I listened to my body, perhaps, I don't know. It's a hard one cuz a lot of my symptoms were similar to asthma symptoms so it's hard.
Participant 031_2023AUORC

Participant would have liked to be more assertive or supported

I probably would have advocated. I can see circumstances when I think back that I would have advocated differently. I would have declined certain things. I would have accepted other things that I didn't think were necessary at the time because I didn't think we had very long. I would have potentially moved closer to care earlier than I have because I didn't expect you to be here this long. Little things I would have changed earlier than I have.
Participant 080_2023AUDIS

I just, yeah. I think I would change it in asking more questions about my medication. I think that not just accepting the fact that that's the only thing I can have. Yeah. Yeah. You know, just. Yeah, I just, yeah. If I had the chance, I'd sort of, yeah.
Participant 019_2023AUDIS

I'll probably if I…I guess it's hard to say in hindsight, but I would have liked to have known earlier. So we could have pushed for those services earlier. You know, she was our first kid. We didn't know any different. You know, if I'd had my youngest first, then there would have been all sorts of red flags when we had Lily, but there wasn't. And the, you know, the healthcare professionals we saw up until that point didn't point anything out. And yeah, I think, as I said before, she's got really distinct facial features that scream 22 Q. If there had been one educated health professional there, I could have gone. Yep, that right there would have made a world difference.
Participant 021_2023AUDPA

Participant would have liked to have had a better understanding of their condition

PARTICIPANT: If I had more knowledge, yes, I would've looked after myself better if I was aware of the serious consequences.

INTERVIEWER: Is there anything else that you would change now that you know where you are and where you've got to where you were?
PARTICIPANT: I'd probably try and address my stress levels more. The cold and stress are the two worst things. I wouldn't let myself get cold and just put up with it. You go out somewhere, like a party may have it outside in winter and I'd end up with bronchitis. If I need those things, obviously I wouldn't have done them.
Participant 008_2023AUDIS

I'm not sure. I might have been made more aware, in particular, the problem I'm having the most now that I've done in a, almost, in a way the least about, because it's complicated and difficult is the whole problem with digestion. I feel that my oesophagus and my gut and all of that have narrowed. I know they have, or it got harder with the hardening of the flesh as it were. I'm finding it quite difficult. No one told me about that ahead of time really or explained what it meant, and no one has yet told me what I can do about it. I'm a bit fearful of that because I fear that the first thing I'm going to be told is I need an operation on the bowel or something to make functioning possible.
Participant 012_2023AUDIS

Information? Lack of the lack of information. Yeah, you wish you had more. Yeah, It's not to have through all the Internet to find out what how you handled surgery or that's why I made-up a brochure and gave it to him. But he thought that. I think you know, he thought that was funny and I are you also, to be fair, I think the neurologist is not sure what to do. He's fiddling too.
Participant 003_2023AUDNS

Participant switched health professionals

One, I would have gone harder with the physios earlier on, I just felt like we went. I guess I would have liked higher incentive, like being more fair with them and the dietitians as well. You know, and I guess that's where it is. It is coming from the allied health where they just come in and every week every time it was a new person. And, you know, I guess I would have changed, I would have, I would have just said straight off the bat, you need to either bring in the same person every time or I'm not talking to them because it's, you know, they're not reading the case notes, whatever. So I probably would have changed the way that we handled that, I guess, earlier on in the piece.
Participant 020_2023AUORC

I think I'd get a different respiratory specialist because he decided that I had sleep apnoea and that was the only thing he'd talked to me about. From then on, he just ignored [UNINTELLIGIBLE] altogether. And I'm like, my sleep apnoea is not the biggest issue here. So, but apart from. That, well, yeah, I the cardiologist also. So the rheumatologist wants me to get echocardiogram every year to keep an eye out for pulmonary arterial hypertension and cardiologist doesn't seem to believe that it's necessary for me to do that. So it's very hard to get appointments with him because he doesn't rank me as a high priority.
Participant 009_2023AUDIS

That was definitely go back to that first doctor. If I had someone that rather than just you would just walk in there, just touch your neck, shove a couple of needles in, and that would send you on your way. If we had further information from there, that would be the only thing I would change. If I would've had access to these wonderful neurologists that I had access to later on down the track, if I would've had access them to them first, that would've been so much better. Because I think those first 18 months were probably a couple of dark years that I didn't have to have if I had a neurologist that was going to treat me and explain my condition to me.
Participant 006_2023AUDNS

Participant would have liked to have been diagnosed sooner

Probably not waiting, like leaving it so long to get an answer in the beginning, because it obviously developed quite severely then, but otherwise. No, I'm glad I found the clinical trial and I'm glad we went down that road rather than it could have meant an awful lot more surgeries. So yeah, I think we did the right thing there.
Participant 022_2023AUDSK

Yes. So definitely a vision screening a lot earlier. Even though eyes appear to look normal sometimes it can be faults in the back that impacts what they see in their vision field. We never had any idea. So maybe in a lot earlier screening for vision, especially if you are deaf or have a hearing loss.
Participant 094_2023AUENM

Well, definitely. I would have liked people to diagnose this earlier and to have, yeah, been able to to have that communicated better, what people were looking for, why they were excluding things or yeah, I think that would have been. It helped me.
Participant 024_2023AUDIS

Table 10.3: Aspect of care or treatment they would change

Aspect of care or treatment they would change	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=266	%	n=66	%	n=102	%	n=76	%	n=22	%	n=132	%	n=134	%	n=75	%	n=191	%	n=143	%	n=123	%
Satisfied with care received	35	13.16	7	10.45	8	9.88	15	33.33	1	3.13	0	0.00	4	12.50	26	14.77	9	10.00	25	12.50	9	14.06
Accesses appropriate specialist/treatment sooner (incl. access to allied health and support)	41	15.41	17	25.37	6	7.41	2	4.44	7	21.88	3	33.33	6	18.75	21	11.93	20	22.22	31	15.50	9	14.06
More knowledge and awareness from health professionals	28	10.53	3	4.48	11	13.58	6	13.33	5	15.63	0	0.00	3	9.38	21	11.93	7	7.78	23	11.50	5	7.81
Changed or stopped treatment sooner	21	7.89	4	5.97	8	9.88	0	0.00	5	15.63	1	11.11	3	9.38	16	9.09	5	5.56	15	7.50	6	9.38

Aspect of care or treatment they would change	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=266	%	n=67	%	n=81	%	n=45	%	n=32	%	n=9	%	n=32	%	n=176	%	n=90	%	n=200	%	n=64	%
Satisfied with care received	35	13.16	6	9.09	14	13.73	10	13.16	5	22.73	18	13.64	17	12.69	8	10.67	27	14.14	17	11.89	18	14.63
Accesses appropriate specialist/treatment sooner (incl. access to allied health and support)	41	15.41	12	18.18	18	17.65	7	9.21	4	18.18	17	12.88	24	17.91	12	16.00	29	15.18	26	18.18	15	12.20
More knowledge and awareness from health professionals	28	10.53	4	6.06	12	11.76	8	10.53	4	18.18	15	11.36	13	9.70	10	13.33	18	9.42	18	12.59	10	8.13
Changed or stopped treatment sooner	21	7.89	5	7.58	6	5.88	8	10.53	2	9.09	10	7.58	11	8.21	6	8.00	15	7.85	12	8.39	9	7.32

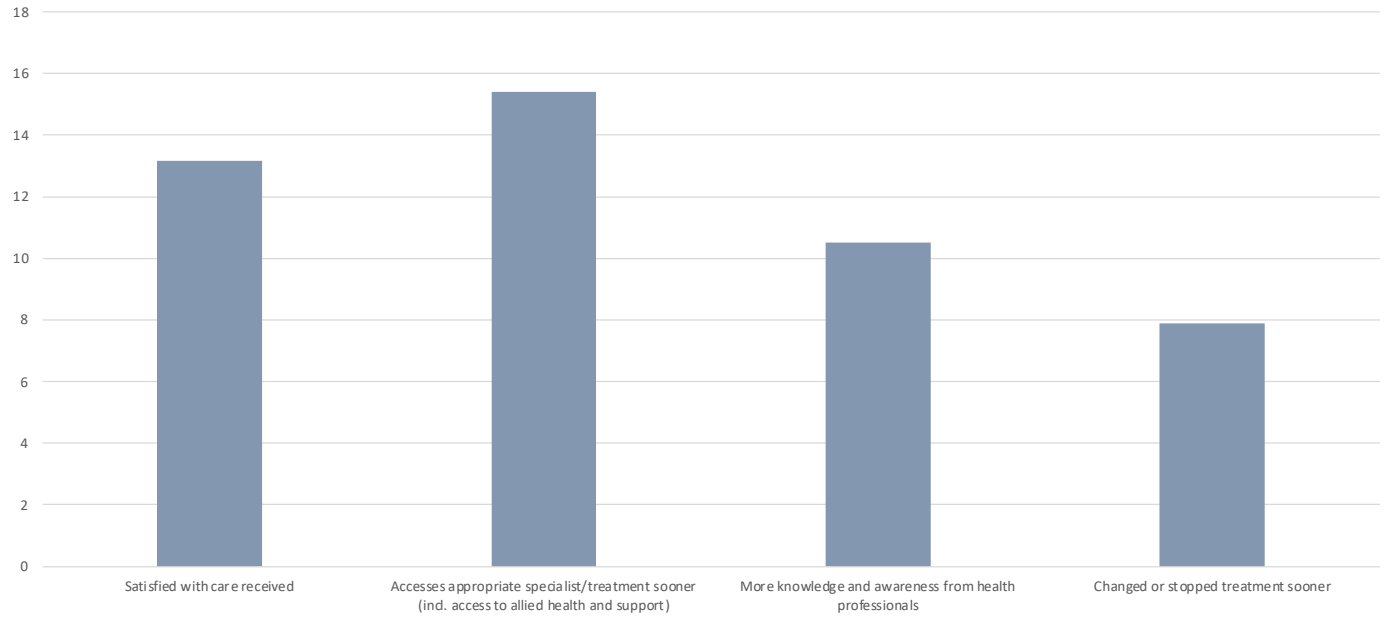


Figure 10.2: Aspect of care or treatment they would change

Table 10.4: Anything participants wish they had known earlier – subgroup variations

Aspect of care or treatment they would change	Reported less frequently	Reported more frequently
Satisfied with care received	Diseases of the skin Endocrine, nutritional or metabolic diseases	Diseases of the nervous system
Accesses appropriate specialist/treatment sooner (incl. access to allied health and support)	Diseases of the nervous system	Endocrine, nutritional or metabolic diseases
More knowledge and awareness from health professionals	Endocrine, nutritional or metabolic diseases	
Changed or stopped treatment sooner		

Section 11

Discussion

2024 PEEK Study in Rare and Genetic Conditions

Introduction

I think my predominant message would be people with complex disabilities are not the same as people with a list of all those disabilities just sort of added up because for a lot of people with CHARGE, you know, it's like, oh, well, they're not quite deaf enough or they're not quite blind enough to receive this service or, you know, whatever it is, but it's like, yes, but the impact of all of those senses being impaired and erratic means they need so much extra support. Participant 018_2023AUDPA

Patient Experience, Expectations and Knowledge (PEEK) is a research program developed by the Centre for Community-Driven Research (CCDR). The aim of PEEK is to conduct patient experience studies across several disease areas using a protocol that will allow for comparisons over time (both quantitative and qualitative components). PEEK studies give us a clear picture and historical record of what it is like to be a patient at a given point in time, and by asking patients about their expectations, PEEK studies give us a way forward to support patients and their families with treatments, information and care.

In this PEEK study, a total of 407 participants with rare diseases or carers to people with rare diseases were recruited into the study. There were 391 that completed or partially completed online questionnaires and 402 participants that were interviewed.

Background

In Australia, a disease is considered rare if it affects less than 5 in 10,000 people. There are more than 7,000 rare diseases that are life threatening or chronically debilitating. Around 8% of Australians (2 million people) live with a rare disease[1].

Demographics

The demographic data we collect in the PEEK study helps us to understand how our PEEK participants compares to people in Australia, and with people that have rare diseases.

In this PEEK study, the proportions of participants with rare diseases that lived in metropolitan areas, had non-school qualifications, and lived in all states of Australia, were similar to that of Australia. There were fewer that lived were in paid employment, and more that lived in areas with higher socioeconomic status, compared to the Australian population [2-4] .

Table 12.1: Demographics

Demographic	Australia %	Rare diseases PEEK %
Live in major cities	71	72
Non-school qualification	65	58
Higher socioeconomic status (7 to 10 deciles)	40	50
Employment (aged 15 to 64)	74	58
New South Wales	32	30
Victoria	26	22
Queensland	20	23
South Australia	7	8
Western Australia	10	10
Tasmania	2	2
Northern Territory	1	0
Australian Capital Territory	2	3

Health related quality of life

Health status

In PEEK studies we collect information about other health conditions that participants manage, as well as health-related quality of life (with the SF36 questionnaire). The purpose of this is to have an idea of the general health of the participants in the study. We can also compare this data with the Australian population, and with other studies with rare diseases participants.

Other health conditions

The National Health Survey was conducted in 2017 to 2018, it is an Australia wide survey conducted by the Australian Bureau of statistics. Almost half of the Australian population have one chronic condition[5]. Common chronic health conditions experienced in Australia in 2017-18 were: mental and behavioural conditions (20%), back problems (16%), arthritis (15%), asthma (11%), diabetes mellitus (5%), heart, stroke and vascular disease (5%), osteoporosis (4%), chronic obstructive pulmonary disease (COPD) (3%), cancer (2%), and kidney disease (1%)[5]. The Australian Bureau of statistics reports that 10% of Australians have depression or feelings of depression and 13.1% have an anxiety-related condition[5].

In this PEEK study, participants with rare diseases had higher levels of anxiety (57% compared to 13%), depression (43% compared to 10%), and arthritis (33% compared to 15%) compared to the Australian population. In addition, more PEEK participants with

rare diseases had chronic pain compared to PEEK participants with non-rare diseases.

A number of studies have described higher rates of anxiety and depression. Including people with Batten Disease[6], amyloidosis[7], rare diseases in general[8], fibrous dysplasia (FD) or McCune Albright syndrome (MAS) patients[9,10], pulmonary arterial hypertension[11], Leber's Hereditary Optic Neuropathy [12], mast cell disorders[13], mitochondrial disease[14], toxic oil syndrome[15], and neurofibromatosis[16]

Baseline health

The Short Form Health Survey 36 (SF36) measures baseline health, or the general health of an individual[17]. The SF36 comprises nine scales: physical functioning, role functioning/physical, role functioning/emotional, energy and fatigue, emotional well-being, social function, pain, general health, and health change from one year ago. The scale ranges from 0 to 100, a higher score denotes better health or function[17].

Population norms for the SF36 dimensions in Australia were assessed in the 1995 National health survey, while this was conducted 25 years ago, it can give an indication of how the breast cancer community in this PEEK study compares with the Australian population[18]. The PEEK participants with rare diseases on average had considerably lower scores for all SF36 domains with the exception of emotional well-being.

Health related quality of life data has been reported in a number of rare diseases in the last five years. Although the diseases are heterogeneous in nature, there are some commonly reported aspects about health-related quality of life. A review of the studies identified in the "Study Position" (see Section 1 of this report) identified that the majority of studies that collected health-related quality of life reported poor health-related quality of life compared to healthy populations, or poor scores for individual domains. People with urticarial vasculitis [19], mast cell disorders [20], idiopathic pulmonary fibrosis [21], and toxic oil syndrome [15] reported poor health-related quality of life across all domains, people with interstitial lung disease[22], idiopathic inflammatory myopathies[23], human T-lymphotropic virus type 1 (HTLV-1)-associated myelopathy (HAM)[24], tuberous sclerosis complex [25], systemic sclerosis , Sjogren's syndrome , lupus[26], Achondroplasia[27], nonsurgical hypoparathyroidism and pseudohypoparathyroidism[28], Beckwith-Wiedemann Syndrome[29], metabolic encephalopathy and arrhythmias[30], rare diseases [31-33], and carers to people with rare diseases[34] all reported poor quality of health compared to the general healthy population, and people with bladder cancer[35], and Fabry Disease [6] reported worse quality of life compared to other chronic conditions.

In this PEEK study, participants with diseases of the immune system, females, those aged 18 to 44, and those aged 45 to 64 tended to have poorer health related quality of life.

Several studies reported negative associations between symptoms and health-related quality of life. People with rare neurodegenerative diseases[37] who had more symptoms had worse quality of life. Fatigue was negatively associated with quality of life for people with sarcoma[38,39], neurofibromatosis 1 [40], PFAPA syndrome [41], and rare diseases[34]. Pain was negatively associated with quality of life for people with neurofibromatosis 1[40], skeletal dysplasia[42], sarcoma[8,39], amyloidosis[7], hereditary fructose intolerance[43], and rare diseases[34]. Poor joint function was associated with poor quality of life for people with fibro dysplasia ossificans[44].

Low scores in cognitive domains were reported for people with neurofibromatosis 1[40], hypoparathyroidism[45], and Sturge-Weber syndrome[46].

A number of studies reported that having a rare disease has a negative impact on emotional domains and mental health domains. Poor scores were reported in emotional domains for people with Skeletal dysplasia[42], Hypoparathyroidism[45], PFAPA syndrome[41], Fabry Disease[36], Acid sphingomyelinase deficiency[47], Duchenne muscular dystrophy[48], Neurofibromatosis 1[40], mitochondrial disease[49], Wilson's Disease[50], rare diseases[33,34], and care givers to people with fibro dysplasia ossificans[44]. Poor scores were reported in mental health domains for people with mitochondrial disease[49], Wilson's Disease[50], autoimmune liver diseases[51], caregivers to people with rare diseases of the respiratory system, rare diseases of the respiratory system[52], and neurofibromatosis 1[53].

People with skeletal dysplasia[42], hypoparathyroidism [45], acid sphingomyelinase deficiency[47], Duchenne muscular dystrophy[48], Pierre Robin sequence[54], amyloidosis[7], and sarcoma[38,39] had low scores for physical domains, and people with amyloidosis[7], hereditary fructose intolerance[43], toxic oil syndrome[15], fibro dysplasia ossificans[44], and skeletal dysplasia[42] had low scores for usual activities or self care.
In this PEEK study, participants on average had poor role functioning/physical, meaning physical health

often interfered with work or other activities. Participants had poor energy and were often fatigues, and they had poor general health.

There were some studies that reported health-related quality of life that was comparable to normal health populations, or that had good scores in certain domains. Children and adolescents with lymphedema reported good levels of health related quality of life[55-58], and children with achondroplasia had scores comparable to a health population for emotional domains[27]. The majority of people with Hereditary fructose intolerance reported no quality of life problems in any domain[43], as did siblings of children with rare diseases[33]. In the long term, people with insulinoma described health-related quality of life that is slightly better than the general population[59]. People with amyloidosis reported no problems with self-care domains[7], people with Wilson's disease had no problems in the physical domains[50], and siblings to children with rare diseases reported better scores in the social domain compared to healthy populations[33].

In this PEEK study, participants on average had good role functioning emotion, meaning that emotional problems rarely interfered with work or other activities for participants in this study. In addition they had good emotional well-being.

Summary of PEEK results

PEEK participants had poorer health related quality of life compared to a healthy population

PEEK most affected health related quality of life domains

- Role functioning physical
- Energy/fatigue
- General health

Subgroups most affected

- Diseases of the immune system
- Females
- Aged 18 to 44
- Aged 45 to 64

Summary of literature

People with rare diseases had poorer health related quality of life compared to a healthy population

Health-related quality of life domains that are often affected by rare diseases:

- Cognitive
- Social function
- Mental health
- Emotional well-being
- Physical activities

Symptoms that impact quality of life:

- Multiple symptoms
- Pain
- Fatigue
- Poor joint function

Screening and diagnosis

Screening and diagnosis

Heaps of hospital visits and ruling out with some neurologists, I don't know, ruling out other things, and finally got to this diagnosis but it's a long, long years. It takes years.
Participant 01_2023AUDNS

In other studies, people with rare diseases have described the pathway to diagnosis, these are often described in terms of delays, misdiagnosis and without adequate support. A number of studies described that people commonly were diagnosed years after first noticing symptoms and seeking medical attention[7,32,60-66]. Delays to diagnosis can be a result of doctors that are not familiar with the condition and associated symptoms, and the variability of the condition[66-70]. Delays were caused by the healthcare system, such as delays in specialist appointments and conditions of health insurance[32,67]. In addition, delays in diagnosis have been described as a result of patients not seeking medical attention[67-69,71]. For people seeking a diagnosis for a rare condition, many have described having numerous medical appointments with a number of specialists, or changing doctors until they find a diagnosis[64,66,67,70-73], often being misdiagnosed with other conditions[61,64,66-68,71,72,74-76].

Two studies reported promotors to diagnosis, including a study of metachromatic leukodystrophy[69] that reported educators and allied health workers noticed symptoms that led to diagnosis, and a study of rare cancers[71] reported that referral to a specialist cancer centre resulted in a quicker diagnosis.

The majority of participants in this PEEK study described noticing symptoms and seeking medical attention relatively soon. However, for almost half of the participants, the diagnostic pathway was complex and required multiple specialists before they got a diagnosis.

I recall a long-time physician GP she wrote it on a post it note, slipped it over to me and that was all that that was said about it. I I still remember looking at it going, I don't even know what that says and it took me ages…Super. You know, what does that mean? I mean it was probably 20, 20, 21 maybe. And so the Internet…I mean at least I didn't have a computer in my home. I was living by myself at that point …So there was no, there were no images, photos. What's life like? That was it? It was just a yellow post-it note. I still remember very clearly. That was my diagnosis. Participant 015_2023AUDSK

Summary of PEEK results

Diagnostic pathway

- 60% noticed symptoms and sought medical attention relatively soon
- 47% had a complex diagnostic pathway

Summary of literature

Barriers to diagnosis

- Doctors not familiar with the condition and symptoms
- Variability of the condition
- Healthcare system delays
- People not seeking medical attention
- Numerous medical appointments any doctors
- Misdiagnosis

Aids to Diagnosis

- Allied health
- Educators

Understanding, knowledge and support at diagnosis

Nothing. I'd never ever heard of it before. I'd never even come up on Google when I was researching like for myself, like what is wrong with me? Because it's just so similar to other cysts and things I guess in the beginning quite easily get confused with that, but no, it didn't even come up. I'd never heard of it. Participant 006_2023AUDSK

Understanding and knowledge

Knowledge about chronic disease before diagnosis varies between individuals. Some will gain information from family and friends with the condition, though it can result in misconceptions and misunderstandings[77,78]. Some people will seek out information about a possible diagnosis, or explore the reasons for symptoms, before receiving a final diagnosis[79,80] others, especially those who have symptoms for long periods before diagnosis, will gain information in terms of how to live with or adapt to symptoms they experience[81]. For some people, the first time they have heard of their chronic condition is when they are diagnosed[80]. At the time of diagnosis, it may be useful for the healthcare professional to talk about how much a patient knows about a condition so that appropriate information can be given, and correct misconceptions[80].

Knowledge about rare conditions is important for receiving a diagnosis, piece of mind, and having a basis for a management plan. For people with rare diseases a diagnosis has been described as important but not always able to give adequate answers for treatment and management of their condition[72]. Once diagnosed, people with rare conditions have struggled to find information about their condition, with unanswered questions about causes, treatment, management, symptom control, and how it differs from previous misdiagnosis[74,82].

In this PEEK study, 61% of participants with rare diseases had little to no understanding of their condition when they were diagnosed. In terms of genetic and biomarker testing, 67% of PEEK rare diseases participants had no discussions with their healthcare providers, and 69% did not have this type of test. In addition, more than a quarter were uncertain about the prognosis of their condition.

Well, it's a bit tricky because I think this particular condition wasn't even discovered until 89. So there aren't a lot of older people with it. They have, well, my son has routine monitoring for the things that it might affect, like his heart and his eyes. And you know, he's ongoing blood testing. So we don't really know what

the outlook is. We don't have any information really to go off.
Participant 021_2023AUORC

Support at diagnosis

When describing their diagnostic journey, some people with are diseases have described being reassured by healthcare professionals that were confident, that discussed steps of diagnosis and treatment as a team[82]. Others described anxiety due to misdiagnosis and delays, and a lack of information and psychological support to prepare for living with the condition [68,74,76,83-85]. Adding to stress and anxiety, some have described unsympathetic healthcare professionals, and being judged, not believed or blamed by healthcare professionals for their symptoms or symptoms of the person they care for[72,74-76]. While some describe relief of getting a diagnosis, there are others that describe shock, confusion a sense of loneliness, and a fear of unknown treatments ahead of them[67,75,76,84-86]. While a diagnosis may not always have a direct impact on clinical care, a diagnosis can also have a positive impact on behaviour changes for both the person with a rare condition and their family, and remove some difficulties in accessing support and services[86].

In this PEEK study, 79% of participants with rare diseases either had no support or not enough support at diagnosis.

Not a lot until the specialist told me and actually, he didn't tell me in a very nice way. [laughs] I don't know. I can't remember what field he was in. I can't remember whether he was a rheumatologist or whether he was some sort of specialist in that sense. I really can't remember but now he basically just said I've got scleroderma and I went, what's that? [laughs] I didn't really know anything about anything because my doctor also didn't lead on much as well. I looked it up in the dictionary and got a hell of a fright.
Participant 01_2023AUDIS

Summary of PEEK results

Understanding of condition

- Poor understanding of condition at diagnosis
- Poor understanding of prognosis
- Very few had discussions about biomarkers and genetic testing

Support at diagnosis

- The majority had either no emotional support or not enough emotional support at diagnosis.

Summary of literature

Supportive factors at diagnosis

- Reassured by healthcare professionals that discussed diagnosis and treatment as a team
- Receiving a diagnosis gives a sense of relief
- Removes difficulty in accessing support

Unsupportive factors at diagnosis

- Delays to diagnosis
- Misdiagnosis
- Lack of information
- Unsympathetic healthcare professionals
- Feeling judged, not believed or blamed for condition
- Fear of unknown after receiving a diagnosis
- Loneliness at diagnosis

Decision making

Decision making

I went to a rheumatologist, but I never was offered any treatment or like medication or anything in the beginning. I basically just was told there was no cure and I just have to learn to live with it. Which is fair enough probably because it's probably true, but I've been in hospital this year and I met a lady in there who said she's had lots of help. A lot of people get infusions and that, I've never been offered anything like that but that's okay. I'm managing.
Participant 013_2023AUDIS

The decision-making process in healthcare is an important component in care of chronic or serious illness[87]. Knowledge of prognosis, treatment options, symptom management, and how treatments are administered are important aspects of a person's ability to make decisions about their healthcare[88,89], highlighting the importance of healthcare professional communication. In addition, the role of family members in decision making is important, with many making decisions following consultation with family[90].

Confidence to take part in decision-making is increased by knowledge, being prepared with relevant questions

for their consultation, and summaries of previous consultations and results[91,92].

People with rare diseases have discussed their participation in treatment decision making, with a spectrum of involvement and descriptions of healthcare professional communication that are helpful or unhelpful. Some people with rare diseases have described not participating in treatment decisions, this was because they were told what to do without discussion, because of emergency situations, that they were not believed, or that they were happy and reassured to take their doctor's advise [70,93-95]. Others described the importance that their views should always be considered, the need for information provided in plain English and patient participation in multi-disciplinary team decisions via an nurse advocate [96]. One study described the importance of second opinions and being assertive and persistent when making treatment decisions[97], and another described the importance of the doctor-patient relationship in decision making participation[51].

Approximately a third of participants in this PEEK study were not given any treatment options at diagnosis, about a third had multiple options, and almost a fifth had one treatment option. Participation in decision making varied with some reporting that they had taken part in decision making, and others describing being told what to do without discussion, or having some but not enough discussion about the treatment or management of their condition. Some participants described that no treatments were available but they discussed allied health, monitoring, lifestyle changes or complementary therapies.

While some people with rare diseases described feeling informed by their doctor and that all options and relevant information was presented to them [73], others thought that information about treatment options had been withheld, or that their doctor had already made treatment decisions or were pushing for a particular treatment option [93-96]. One study noted that participants that had been diagnosed tens of years ago had no participation in decision making, while those diagnosed more recently were involved [96]. Another study described that participating in a support group improved knowledge about management and supported decision making[70].

Participants in this PEEK study described how decision making had changed over time. Approximately half of the participants described that decision making had changed over time. Participants changed the way that they made decisions over time because they became more informed and assertive, were more aware of their health and limitations, were more cautious or took the impact on family and dependents into account.

Yes. Look, I just think I have got a lot more agency now. I just feel like now the ball is in my court a lot more than what it was. I suppose I'm more knowledgeable. I feel like when I'm discussing things with the doctors now it's more of an equal level after a team rather than just sitting there being passive. It's probably changed in that respect.
Participant 054_2023AUDPA

Summary of PEEK results

Discussions about treatment and management of condition
- 33% no treatments discussed
- 32% multiple treatment options
- 18% on treatment option

Participation in decision making
- Took part in decision making
- Were told what to do without discussion
- Wanted more discussion about treatment and management
- Were offered allied health, monitoring, lifestyle advise or complementary therapies

Changes in decision making over time
- More informed and assertive
- More aware of health and responsibilities
- More cautious
- More focused on family and dependents

Summary of literature

Barriers to participating in decision making
- told what to do without discussion
- emergency situations
- happy and reassured to take their doctor's advise
- doctor withholding information or pushing for particular treatment
- Not being believed

Facilitators to Participate in decision making
- belief that own view should always be considered
- information in plain English
- patient participation in multi-disciplinary team decisions via a nurse advocate
- being well informed by doctor about all options
- Support group
- Second opinions
- Being assertive and persistent
- Good patient doctor relationships
- Taking part in support group

Considerations when making treatment decisions

Side effects is a big one for me. Obviously I don't want to put on heaps of weight or feel nauseous, or if I can avoid some horrible side effects, I will and I guess not so much yet. But as I said in the future, like if I can be on them while pregnant or how long I have to be off them before being pregnant, yeah.
Participant 095_2023AUDNS

Important considerations for PEEK participants with rare diseases in decision making were side effects, efficacy, cost, advice of their clinician, and quality of life. In other studies, people with rare diseases have discussed their considerations when making treatment decisions, this included taking the doctor's opinion into account, longevity, treatment effectiveness, location or travel to treatment centre, invasiveness or burden of treatment, impact on family and time off work, cost, type and severity of side effects, duration of side effects, improvement in symptoms, duration of improvements in condition, previous experience of treatments, and other people's experiences[85,95,98-100].

In terms of treatment goals, participants in this PEEK study had goals of quality of life, maintaining their condition, physical improvements in their condition, and to live independently.

Summary of PEEK results

Considerations when making treatment decisions

- Side effects
- Efficacy
- Cost
- Advice of their clinician
- Quality of life
- Treatment goals

Treatment goals

- Quality of life
- Maintaining their condition
- Physical improvements in their condition
- Live independently.

Summary of literature

Considerations when making treatment decisions

- doctor's opinion
- type and severity of side effects,
- duration of side effects,
- improvement in symptoms,
- duration of improvements in condition, and
- previous experience of treatments
- Longevity
- Treatment effectiveness
- Invasiveness or burden of treatment
- Impact on family and time off work
- Location
- Cost
- Other people's experiences

Treatment and healthcare provision

Treatment and healthcare provision

In this PEEK study, to get an insight healthcare access, information about access to health insurance, health system, and financial consequences from having a rare condition are collected.

Allied health is important to manage the physical, emotional, practical and financial consequences of rare diseases.

A review of the studies identified in the "Study Position" (see Section 1 of this report) gave little insight into allied health use in rare diseases. Some studies described that people with rare diseases would like more access to allied health, in particular psychological support, but also social work, dieticians, physiotherapy and rehabilitation specialists[8,60,105].

The majority of participants in this PEEK study had accessed allied health, the most common forms of allied health accessed were physiotherapy, psychology, occupational therapy, dietary, podiatry and speech therapy. Quality of life from allied health ranged from life was distressing (psychology) to life was average (physiotherapy, occupational therapy, speech therapy and podiatry). Effectiveness of these therapies were rated from ineffective (podiatry) to moderately effective (occupational therapy).

Other studies that reported patient satisfaction with allied health, people with Huntington's disease found speech therapy use as helpful, improving speech and language skills, and that groups sessions enabled them to meet other people in a similar situation[110], in another study, about half of those with a rare disease that accessed psychological support found it helpful[8]. People with Ehlers-Danlos Syndromes in a multi-

national study reported both positive and negative experiences of physical therapy for pain, noting better results from a physiotherapist that had knowledge and familiarity of Ehlers-Danlos Syndromes[70]. A French study of mucopolysaccharidoses described that they were most commonly referred to physiotherapy, followed by speech therapy, Orthoptics, and psychomotor therapy[84].

In other studies, people with rare diseases described the barriers they had to getting the healthcare services and treatments that they needed. Demographic factors such as not speaking the local language, being poor, having a low level of education and living in regional or remote areas were a barrier to access to healthcare services [71,84,101]. In addition, a lack of diagnosis was a barrier to accessing healthcare[2,66]. Having access to a specialist centre was a facilitator of access to healthcare and participants noted that they had improved coordination of appointments, improved access to allied health and support[51,71,102]. Patients or carers who were also healthcare professionals had better access to healthcare due to knowledge and professional contacts[101].

Patient treatment preferences

Clinical guidelines that are aligned to patient preferences are more likely to be used and lead to higher rates of patient compliance.[111-113] Patient preferences and priorities vary across different health issues, preferences are associated with health care service satisfaction, they refer to the perspectives, values or priorities related to health and health care, including opinions on risks and benefits, the impact on their health and lifestyle[111,114].

To help inform patient preferences in the rare diseases community, participants in this PEEK study discussed side effects and adherence to treatment. Mild side effects were described by providing examples, or as side effects that are self managed or do not interfere with life. In a similar way, participants describe severe side effects, broadly as those that impact every day life, or using the examples of specific side effects. Side effects were an important factor in treatment adherence.

Participants in this PEEK study described mild side effects using a specific example such as fatigue, gastrointestinal distress, and headaches, side effects that do not interfere with life, and side effects that can be self managed. They described severe side effects using a specific example such pain, fatigue and the emotional impact, and those that have an impact on everyday life or that are life threatening or require hospitalisation.

Participants in this PEEK study described their adherence to treatment. Most commonly they described following treatment according to the advice of their doctor. Others described sticking to a treatment for a specific amount of time, usually 2 to 3 months, or as long as the side effects are tolerable. Some described that they had not given up on any treatment

Side effects and problematic symptoms will vary across rare diseases, however [7,34,38-40,42,43,71,115], fatigue [34,38-41,71], and mental health problems were commonly reported across disease types[49-53].

Lifestyle changes

Many chronic diseases share the modifiable risk factors of poor diet, little exercise, smoking, and excessive alcohol consumption. Participants with rare diseases in this PEEK study most often made changes to their diet and exercise habits, and rated exercise as somewhat effective and diet as somewhat effective. PEEK participants with rare diseases made lifestyle changes at a similar rate to those with non-rare diseases[116-118].

Complementary therapies

Complementary therapies include taking supplements, mindfulness and relaxation techniques, massage therapy and acupuncture and many others. PEEK participants with rare diseases most commonly used supplements, mindfulness and relaxation techniques and massage therapy, they rated massage therapy and mindfulness as moderately effective and supplements as somewhat effective. Very few studies described access to complementary therapies, however, a multinational study of people with Ehlers-Danlos Syndromes described managing pain with dry needling, and acupuncture[70].Participants with rare diseases used complementary therapies at a similar rate to those with non-rare diseases.

Summary of PEEK results

Description of mild side effects

- Specific example such as fatigue, gastrointestinal distress, and headaches
- Do not interfere with life
- Can be self managed

Description of severe side effects

- Specific example such pain, fatigue and the emotional impact
- Has an impact on everyday life
- Life threatening or require hospitalisation

Adherence to treatment

- According to advice of their doctor
- Specific amount of time, usually 2 to 3 months
- Side effects are tolerable
- Does not give up on any treatment

Allied health

- 71% used at least one allied health service
- 47% physiotherapy
- 39% Psychology
- 35% occupational therapy
- 33% dietary
- 31% podiatry
- 24% speech therapy

Lifestyle changes

- 60% exercise
- 51% diet changes

Complementary therapies

- 46% supplements
- 46% mindfulness or relaxation
- 30% massage therapy

Summary of literature

Barriers Access to healthcare

- Do not speak local language
- Low income
- Low education attainment
- Regional or remote
- Lack of diagnosis

Facilitators Access to healthcare

- Access to a specialist clinic
- Patients that are also healthcare professionals

Common side effects and symptoms

- Fatigue
- Pain
- Mental health problems

Allied health Unmet needs

- psychological support
- social work
- dieticians
- physiotherapy
- rehabilitation specialists

Affordability of healthcare

Probably the biggest one is the full-time off work, it's obviously very hard on the family. Also now, I find with fatigue and just chasing up medical appointments and things like that, that I only work part-time now. I work three days a week. Just financially that. I find that with scripts and seeking treatment, very expensive. Just getting accommodation and things like that, going down to specialist appointments, I find very expensive as well. Time-wise, definitely it takes up way too much family time with conversations and just their support Participant 014_2023AUDIS

Almost half of the Australian population have private health insurance with hospital cover[103]. This can be used to partially or completely fund stays in public or private hospitals. Between 2006 and 2016, the proportion of private health care funded hospitalisations in public hospitals rose from about 8% to 14%[103]. In this PEEK study, a higher proportion had private health insurance compared to the Australian population.

People with rare diseases and carers to people with rare diseases have described lost educational opportunities due to the amount of time they missed at school [53,84,104], with implications on future earning potential. In addition, people with rare diseases and carers to people with rare diseases have described having to take time off work, reducing hours, changing roles or careers or quitting work as a result of a rare disease diagnosis[84,85,104]

In this PEEK study participants with rare diseases noted the cost of managing their condition. Approximately half of the participants in this PEEK study had no out of pocket expenses when they were diagnosed. However, for those that did have costs, for 45% this was a moderate or significant burden. The monthly expenses for managing their condition exceeded $250 for a third

of the participants, and was moderately to significantly a burden for 41% of participants. Costs were from time off work, treatment, specialist appointments, diagnostic tests and scans, and transport, parking and accommodation. In terms of employment changes, approximately a third had quit their job or reduced hours. In other studies, people with rare diseases have described the impact on employment from having a rare disease or caring for someone, describing reducing hours, taking less demanding jobs, or having to quit jobs, in addition to being overlooked for promotions or inclusion on specific projects[105-109]

Summary of PEEK results

Costs

- Time off work
- Treatment
- Specialist appointments
- Diagnostic tests and scans
- Transport, parking and accommodation

Changes in employment

- 30% Quit their job
- 30% had reduced hours that they worked

Summary of literature

Costs due to work and education

- Taking time off school
- Reduced education opportunities
- Taking time off work
- Reducing work hours
- Changing work role or career
- Quitting workforce

Clinical Trials

Clinical trials are essential for development of new treatments. The benefits to participants include access to new treatments, an active role in healthcare, and closer monitoring of health condition. The risks to participants include new treatment may not be as effective, and side effects.

In other studies, people with rare diseases described reasons for and against taking part in clinical research. One study described language, educational and socioeconomic barriers to taking part in research and noted the importance of having a good working relationship with their healthcare professionals in having access to taking part in research. [101]. The other study described that people with rare diseases were motivated to take part in research to help future generations, while those that had already taken part in clinical trials were reluctant to take part in more research as they had already done their part to help[100]. This study also described the opinion of their doctor and their family was important in their decision to take part in research[100].

The majority of participants in this PEEK study had not discussed clinical trials with their doctor, very few had taken part in a clinical trial though approximately half of the participants would take part in a clinical trial if one was available.

A search of the Australian New Zealand Clinical Trials Registry was conducted on 4 January 2023. The search term used was “rare disease”, and included any study that was conducted in Australia, and was open to recruitment in the last five years. A total of 74 studies were identified that had a target recruitment of between 4 and 20,000 participants (median=102), there were 35 studies that were international, and 39 studies that were conducted exclusively with in Australia. The most common types of studies were investigating drugs (n=47), followed by registries (n=9), and allied health (n=5). There were 4 studies investigating devices, 4 tissue banks, 3 surgical studies and 2 diagnostic studies.

There were 47 studies conducted in New South Wales, 44 studies in Victoria, 25 in Queensland, 24 in South Australia, 24 in Western Australia, 7 in the Australian Capital Territory, 7 in Tasmania and 5 in the Northern Territory.

There were 47 studies that included only adults, 8 that included only children and 19 that included both adults and children. The most common disease types that were investigated were rare cancers (n=29), diseases of the nervous system (n=8), endocrine, nutritional or metabolic diseases (n=7), and diseases of the blood or blood-forming organs (n=4).

Figure 12.1: Distribution of clinical trials for HER2 positive breast cancer in Australia 2016-2021

Table 11.2: Clinical trials

Summary of clinical trials		N=74	%
Disease area	Neoplasms/Cancer	29	39.19
	Diseases of the nervous system	8	10.81
	Endocrine, nutritional or metabolic diseases	7	9.46
	Diseases of the blood or blood-forming organs	4	5.41
	Developmental anomalies	3	4.05
	Diseases of the musculoskeletal system or connective tissue	3	4.05
	Rare diseases	3	4.05
	Diseases of the circulatory system	2	2.70
	Diseases of the ear or mastoid process	2	2.70
	Diseases of the skin	2	2.70
	Diseases of the visual system	2	2.70
	Injury, poisoning or certain other consequences of external causes	2	2.70
	Pregnancy, childbirth or the puerperium	2	2.70
	Diseases of the digestive system	1	1.35
	Diseases of the genitourinary system	1	1.35
	Diseases of the immune system	1	1.35
	Diseases of the respiratory system	1	1.35
	Mental, behavioural or neurodevelopmental disorders	1	1.35
Type of investigation	Drug	47	63.51
	Registry	9	12.16
	Allied health	5	6.76
	Device	4	5.41
	Tissue bank	4	5.41
	Surgery	3	4.05
	Diagnostic	2	2.70

Summary of PEEK results	Summary of literature
Clinical trial discussions and participation • 64% No discussions about clinical trials • 50% would take part in a clinical trial if one was available • 12% had taken part in a clinical trial	Barrier to take part in clinical research • Do not speak local language • Low socioeconomic status • Low education attainment • Previous participation in clinical research Promotor to take part in clinical research • Opinion of doctor • Opinion of family • Desire to help

Expectations for future treatments

So it would be nice if new treatments also considered more seriously. That the lived experience of a side effect is different perhaps, to the medical definition of a side effect. Participant 024_2023AUDIS

Participants in this PEEK study described their expectations for future treatments. The most common descriptions were that they expected future treatments to be more affordable, more effective or personalised, include choice, accessibility and discussions about treatments, have fewer or less intense side effects, will be easier to administer or less invasive and that they will have more access to clinical trials and access to new treatments and technologies.

In other studies, people with rare diseases described their expectations or priorities for future treatments. They described wanting effective treatments that were affordable and accessible, prevents clinical deterioration , improves life expectancy, treatments with fewer side effects treatments that are easier to administer, they wanted treatments that were holistic, in particular paying attention to mental and emotional health, treatments that gave symptom relief, that reduced short and long term side effects and improved quality of life, in addition, they noted the importance of prevention and early detection. [55-58,83,100,119-122]

Yeah, yeah. Look, I suppose cost, cost is certainly a a challenge or a barrier for some I think access to information about. What the options are and what the possible side effects of each pathway so that you can make informed decisions about what you're willing to, what you're willing to risk, but also kind of what your probabilities of success are. Yeah, I think, I think more information and more knowledgeable practitioners.
Participant 007_2023AUDSK

Summary of PEEK results	Summary of literature
Future treatments • More affordable • More effective or personalised • Choice, accessibility and discussions about treatments • Fewer or less intense side effects • Easier to administer or less invasive • More clinical trials and access to new treatments and technologies	**Future treatments** • Effective treatments • Affordable treatments • Accessible treatments • Holistic treatment • Symptom relief • Improves quality of life • Prevents clinical deterioration • Improves life expectancy • Fewer side effects • Easier administration

Information sources

People with rare diseases have described getting their information from a variety of sources, this includes social media, the internet, patient organisations, conferences, podcasts, videos, their healthcare professionals, medical journals, other patients, and from their own lived experiences[66 ,70,73,84,93,94,109,123-126]. One study noted that people who got information from a specialist treatment centre were more satisfied with their information compared to those that got information from a regional hospital.[71]

This is similar to the information sources for participants with rare diseases in this PEEK study. PEEK participants with rare diseases got information from a range of sources, including the internet, Facebook and social media, health charities, treating clinician, medical journals, other patients experience, books, pamphlets and newsletters. Most described a preference for talking to someone, online information or a combination of both, and also a preference for written information. Talking to someone was preferred because it gave them time to ask questions, online information was very accessible and written information made it easy to highlight information and refer back to.

Information that was not helpful

In other studies, people with rare diseases described information that was not helpful, this included not having enough information, a lack of information about what to expect, information that is withheld or underplayed, and information that is misleading, not relevant or inaccurate [96,125,127,128]. However, people with rare diseases described being able to critically evaluate information[124]. .

Similarly, PEEK participants with rare diseases found information from Most commonly, participants in this PEEK study described that no information not helpful. Others described unhelpful information as information from their GP or specialist, other people's experiences, and a lack of new information.

Information that was not helpful

In this PEEK study, participants described the following types of information as helpful; other people's experience and peer support, what to expect (for example disease course and treatments), talking to their doctor, and information from health charities.

In other studies, people with rare diseases described information from other people's experience as helpful, and having enough time to discuss information with their doctor as helpful[70,84]

Timing of information

In other studies, people with rare diseases described the need for information and support at all stages of their healthcare management, though it was described as most important at an earlier stage when seeking or having obtained a diagnosis [93,94,109].

Similarly, PEEK participants with rare diseases most commonly were receptive to information at diagnosis or after the shock of diagnosis had worn off, and continuously throughout their experience, some took more time to be receptive, needing up to wait for a year or more after diagnosis.

Information topics

In this PEEK study more than half of the participants searched for information about disease cause, treatment options, disease management, and complementary therapies. More than a third had searched for how to interpret test results, clinical trials, diet advice, physical activity and psychological or social support.

In other studies, people with rare diseases described the topics of information they needed or searched for independently. The topics included emotional and mental health support, disease management, treatment options, and side effects[34,70,71,129]. One study described that participants were satisfied with factual medical information about treatments and side effects[71].

Summary of PEEK results

Information sources
- Internet
- Facebook and social media
- Health charities
- Treating clinician
- Medical journals
- Other patients experience
- Books, pamphlets and newsletters

Information that is not helpful
- No information not helpful
- GP or specialist
- Other people's experiences
- Lack of new information

Information that was helpful
- Other people's experience, peer support
- What to expect
- Talking to their doctor
- Health charities

Timing of information
- From the beginning
- Continuously
- A year after diagnosis
- After the shock of diagnosis

Information topics most commonly searched for
- Disease cause
- Treatment options
- Disease management
- Complementary therapies
- How to interpret test results
- Clinical trials
- Diet advice
- physical activity
- psychological or social support

Summary of literature

Information sources
- Social media
- Internet
- Patient organisations
- Conferences, podcast and videos
- Healthcare professionals, treatment centre
- Medical journals
- Other patients
- Own lived experiences

Information that is not helpful
- Lack of information
- Information that is withheld or downplayed

Information that was helpful
- Able to critically evaluate information
- Other people's experience, peer support
- Enough time to discuss with doctor

Timing of information
- All the time
- Most important at diagnosis

Information searched for
- Emotional and mental health support
- Disease management
- Treatment options
- Side effects

Expectations for future information

It's not like we haven't got any information, it was just what the doctor told us. I think if there was something that could be provided a point of diagnosis in writing that would be really helpful or somewhere to go and look for information, so maybe be directed to a website or something. Because again, you know, you kind of have to do all of that yourself. I think information being available and relevant to the Australian community would be really important. Participant 021_2023AUORC

Participants in this PEEK study described their expectations for future information. They described wanting information to be more accessible and easy to find, they wanted information about their disease trajectory and what to expect, they wanted information specific to their condition, and they wanted to raise community awareness.

In other studies, people with rare diseases described their expectations or priorities for future information. This included details about what treatments are available and explanations when certain treatments are not suitable, what to expect, especially short and long term symptoms, information about transitioning from hospital to home, written information specific to their case, including treatment plans and test results, and information that is clear and jargon free [93,94,96,109,125,127,128,130-132]. In addition, people with rare diseases described wanting more community awareness and more education for healthcare professionals about rare diseases[97,133].

Summary of PEEK results

Future information

- Accessible and easy to find
- What to expect
- Specific to subgroup or condition
- Raise community awareness

Summary of literature

Future information

- All available treatments
- Explanations about suitability of treatments
- What to expect
- Short and long term symptoms
- Transition from hospital to home
- Written information specific to own case
- Plain language
- Community awareness
- Healthcare professional education

Communication and care coordination

Self-management

Self-management of chronic disease encompasses the tasks that an individual must do to live with their condition. Self-management is supported by education, support, and healthcare interventions. It includes regular review of problems and progress, setting goals, and providing support for problem solving[134]. Components of self-management include information, activation and collaboration[134].

Information is a key component of health self-management[135,136]. The types of information that help with self-management includes information about the condition, prognosis, what to expect, information about how to conduct activities of daily living with the condition, and information about lifestyle factors that can help with disease management[135,136].

Self-management of chronic disease encompasses the tasks that an individual must do to live with their condition. Self-management is supported by education, support, and healthcare interventions. It includes regular review of problems and progress, setting goals, and providing support for problem solving[134]. Components of self-management include information, activation and collaboration[134].

Activation (skills and knowledge)

Patient activation is the skills, knowledge, and confidence that a person has to manage their health and care; and is a key component to health self-management. Components of patient activation are support for treatment adherence and attendance at medical appointments, action plans to respond to signs and symptoms, monitoring and recording physiological measures to share with healthcare professionals, and psychological strategies such as problem solving and goal setting.

Patient activation is measured in the PEEK study using the Partners in Health questionnaire[137]. PEEK participants with rare diseases had very good knowledge about their condition, were average at coping with their condition, were good at recognizing and managing symptoms, and were very good at adhering to treatment.

Communication and collaboration

Yeah, really good. His team is great. We've got a phone number that we can call or text anytime 24/7 if we have any questions and we get responses straight away. And yeah, as I said earlier that every time we meet with his team and we've got questions, they've always been really, they've been really clear with this and able to answer everything that we've come to them with.
Participant 029_2023AUORC

Collaboration is an important part of health self-management, the components of collaboration include healthcare communication, details for available information, psychosocial and financial support [135,136] Communication between healthcare professionals and patients can impact the treatment adherence, self-management, health outcomes, and patient satisfaction[138-141].

An expert panel identified the fundamental elements of healthcare communication that encourages a caring, trusting relationship for patient and healthcare professional that enables communication, information sharing, and decision-making[142].

Building a relationship with patient, families and support networks is fundamental to establishing good communication[142]. Healthcare professionals should encourage discussion with patients to understand their concerns, actively listen to patients to gather

information using questions then summarising to ensure understanding[142]. It is important for healthcare professionals to understand the patient's perspective and to be sympathetic to their race, culture, beliefs, and concerns. It is important to share information using language that the patient can understand, encourage questions and make sure that the patient understands[142]. The healthcare professional should encourage patient participation in decision-making, agree on problems, check for willingness to comply with treatment and inform patient about any available support and resources[142]. Finally, the healthcare professional should provide closure, this is to summarise and confirm agreement with treatment plan and discuss follow up.

In this PEEK study, most participants described overall positive communication with healthcare professionals, though some of these described exceptions. Approximately a third described an overall negative experience of healthcare communication. When participants described positive communication, this was primarily because of two way, supportive and comprehensive conversations. Reasons for negative communication included healthcare professionals with a lack of knowledge about condition, having a lack of time in appointments, dismissive or one way conversations, and a lack of coordinated care and follow up.

In other studies, people with rare diseases described the positive impact that learning about the condition had on communication with healthcare professionals [133]. In addition, communication was improved when healthcare professionals were open, freely gave information, were confident and reassureing[73,82]. Other studies described barriers to communication including distrust of healthcare providers or healthcare system, healthcare professionals with inadequate knowledge of condition, healthcare professionals make assumptions about patient needs/preferences, and being embarrassed by condition or symptom, and embarassemnt[71,101]. In addition, people with rare diseases described that feeling that healthcare professional does not believe in patient symptoms, feeling judged by healthcare professionals, healthcare professionals that are biased to treatments and healthcare professionals that withheld information were all barriers to communication[72,74-76,93-96].

Inadequate. I really felt that most of the time I was driving the understanding research, how to get help, who to get help from, what to do from professionals and that they would sort of. Not explain things like I was intelligent enough to like absorb the information. Yeah, and and therefore would miss things out and and not give me full picture.
Participant 087_2023AUENM

Summary of PEEK results

Positive communication

- Two way, supportive and comprehensive conversations

Negative communication

- Lack of knowledge about condition
- Lack of time in appointments
- Dismissive, one way conversations
- Lack of coordinated care and follow up

Summary of literature

Positive communication

- Proactive patients learning about condition
- Healthcare professionals that are open and freely give information
- Healthcare professionals that are confident and reassuring

Negative communication

- Distrust of healthcare providers or healthcare system
- Healthcare professionals with inadequate knowledge of condition
- Embarrassment
- Healthcare professionals make assumptions about patient needs/preferences
- Feeling that healthcare professional does not believe in patient symptoms
- Feeling judged by healthcare professionals
- Healthcare professionals that are biased to treatments
- Healthcare professionals that withhold information

Expectations of future communication

Yes, I would like them not to Google the condition when you sit in front of them. Maybe if they if, just say, 'Look, I don't know this condition. I've not heard of it. But let me do some investigation and then I'll inform myself' But at the moment, most of the time, the parents or myself, even we go into a doctor and I've never heard that and they Google it in front of you. So if they're Googling you, what chance have we got?
Participant 025_2023AUDPA

In this PEEK study, participants wanted future communication to include healthcare professionals with better understanding of condition, more empathy, more transparent and forthcoming, for healthcare professionals will listen to patient, and to have a multidisciplinary and coordinated approach

Similarly, in other studies, people with rare diseases described their expectations or priorities for future healthcare professional communication. They described wanting a single point of communication to answer any questions they may have, more time in appointments, regular follow up, coordinated care, and that healthcare providers have empathy and kindness, help build trust between patient and provider, and work as a team with patient and family[72,96,97,143].

Summary of PEEK results	**Summary of literature**
Future communication • Healthcare professionals with better understanding of condition • More empathy • Communication will be more transparent and forthcoming • Healthcare professionals will listen to patient • Multidisciplinary and coordinated approach	Communication • Single point of contact • Coordinated care • Empathy, trust and kindness • Adequate time • Regular follow up • Work as team with patient and family

Care coordination

I guess as an overall picture, I would love some kind of coordination because 22Q is so broad and affects so many different aspects of the body. And you know, we say all these individual specialists separately, but none of them talk to each other. That lack of communication is pretty major…And I'm kinda hoping that the new 22Q clinic will help with some aspects of that, but like I said, we haven't quite got that far yet.
Participant 021_2023AUDPA

Parents of children with rare diseases described the barriers they faced in accessing specialist doctors, allied health, education and social support services. Sometimes the barriers were caused by not having a formal diagnosis, without a formal diagnosis they were not eligible for aid, though, some found that secondary diagnosis for example attention deficit hyperactivity disorder, or autism spectrum disorder allowed them to access services[72]. Others described he barrier of a lack of care-coordination, this led to families not knowing about what services were available[72]. Some described bureaucratic barriers, and having to argue with their doctors to get referrals[72].

People with rare diseases have described the difficulties in managing their condition in the healthcare system, and as a consequence have become their own health experts with unique knowledge about symptoms [93,94]. For some, self-advocating for their healthcare needs is a necessity, and adopt strategies such as preparing for medical appointments with questions and being assertive during medical appointments. [125,144]. Communication and collaboration with healthcare professionals was measured in this PEEK study by the Care Coordination questionnaire[145]. The participants in this study experienced good quality of care, and average coordination of care. They had a moderate ability to navigate the healthcare system, and experienced moderate communication from healthcare professionals.

Summary of PEEK results

Care coordination

- Moderate communication with healthcare professionals
- Moderate navigation of the healthcare system.
- Participants scored rated their care coordination as moderate.
- Participants rated their quality of care as good.

Summary of literature

Care coordination barriers

- Lack of formal diagnosis
- Lack of knowledge about available services
- Bureaucracy

Care coordination facilitator

- Having a secondary diagnosis when no formal diagnosis is given
- Patient becomes own health expert
- Being prepared and assertive in medical appointments

Care and support

Care and support

No, I couldn't. The hardest thing is I couldn't get any support because I had no diagnosis. You know, and that's like I spoke to the NDIS the other day. And they don't even have pageants on their thing because it's for older people so, and because I'm only 51, they put me under osteoarthritis or something like that. So I've got some stuff I've got a doctor to fill out, and I've got some stuff I've got to fill out and everything like that to actually send it off to them. But until I got diagnosed, I couldn't get any help from anybody. Participant 014_2023AUORC

In other studies, people with rare diseases described their support needs, including practical, psychological, social, information, financial, and healthcare coordination, access to healthcare support [32,125,130,133]. Others described the importance of family and friends for support [146]. People with rare diseases have described the importance of patient groups, often on social media for information and support. [70,109,125] Some described practical information about home care services, financial advice, managing symptoms[6,125,144]. Patient groups and social media were often described in terms of emotional support, allowing people with rare diseases to connect with other people in a similar situation[144]. However, finding the correct support group could be difficult for those with undiagnosed conditions, with having to determine the relevance of information for their condition, though the similarities of symptoms and managing symptoms was helpful[123]

In this PEEK study, a quarter of participants described not receiving any support, and more than 10% described the challenges they faced in accessing support. Others described getting support from the hospital or clinical setting, from family and friends, charities and other patients. Some described getting support from domestic services or home care.

Summary of PEEK results

Experience of care and support

- Did not receive support
- Had challenges accessing support
- Hospital or clinical setting
- Family and friends
- Charities
- Other patients
- Domestic services or home care

Summary of literature

Support given

- Patient groups
- Home care services
- Financial advice
- Managing symptoms
- Emotional support
- Family and friends

Support needs

- Practical
- Psychological
- Social
- Information
- Healthcare coordination,
- Access to healthcare support
- Financial

Expectations for future support

The mental health services, I believe we all need them. Like, even if we say we don't like, when I first got diagnosed, I probably would have said no, I don't need that. Then like thinking about it, living with it, like with the diagnosis. And I'm like, yeah, OK, now I feel pretty crappy about myself.
Participant 003_2023AUDSK

Participants in this PEEK study described their expectations for future support. They described wanting more access to support services, wanting access to specialist clinics to talk to healthcare professionals, healthcare professionals with better knowledge of their condition, they wanted care to be multidisciplinary and coordinated and access to peer support.

Other studies people with rare diseases described their expectations or priorities for future support. This included access to emotional and psychological support, access to peer support, and access to services that helped with activities of daily living[8,132,144,147,148].

Summary of PEEK results	**Summary of literature**
Expectations of future support • More access to support services • Specialist clinics or services where they can talk to professionals • Multidisciplinary and coordinated approach • Healthcare professionals with more knowledge • Peer support	Expectations of future support • Emotional and psychological support, • Peer support • Support for activities of daily living • Caregiver support

Quality of life

Quality of life

Well, it's certainly affected my quality of life because I can't even cook as I enjoy doing or go out for meals and enjoy. I do that, but I don't know what I'm eating. Oh, fatigue, I don't think I've mentioned that, but fatigue has been a big thing in a general way with scleroderma. I get very tired and I, by and large, have a nap almost every afternoon. If I'm sitting at the computer, my head hits the computer because I'm just asleep, really. That's certainly something that's made a difference. What was the question again then? How it has affected me?
Participant 012_2023AUDIS

The majority of participants in this PEEK study described a negative impact on their quality of life, the most common reasons were the emotional strain on family, a reduced capacity for physical activities, reduced social interactions, managing side effects and symptoms, being unable to travel or adapt to travel and the emotional stress on themselves.

Similarly, in other studies, people with rare diseases described negative impacts on their quality of life from the physical limitations it has on activities of daily life and on the ability to be social, the restrictions it places on the ability to travel, take holidays, or enjoy hobbies, and the emotional stress that it places on themselves and close family members[43,83,107,125,149,150]. In addition, people with rare diseases described the impact that symptoms such as pain have on quality of life, as well as frequent illness, communication problems, loss of autonomy, feelings of hopelessness and loss of control, and having to rely on others[66,70,71,84,99,115,133,146]. Others described that their quality of life was affected because of the stigma of having a rare disease, their body image, feelings of frustration about the unfairness of having condition, worry about the future, and the fear of relapses or flare-ups[66,70,71,84,99,115,133,146].

Summary of PEEK results

Quality of life

- 63% had an overall negative impact on quality of life
- The emotional strain on family
- Reduced capacity for physical activities
- Reduced social interactions
- Managing side effects and symptoms
- Unable to travel or adapt to travel
- Emotional stress on self

Summary of literature

Quality of Life

- Activities of daily living
- Travel restrictions
- Enjoyment of hobbies
- Emotional stress
- Impact on socialising
- Impact of symptoms – pain, motor skills
- Communication problems
- Hopelessness and loss of control
- Having to rely on others
- Stigma
- Body image
- Frustration at unfairness of illness
- Worry about future
- Fear of relapse or flare up

Activities to maintain general health

Well, I if I with the fibromyalgia, I have to sometimes I hit a wall of tiredness. And I just have to have a sleep. So I do if I'm get if I I'm not like that all the time, but when I do get like that. I do. Don't get up and go and have a lay down for an hour or so and I'll get up and I feel better. So that's what I do for myself. If I'm not, you know, unwell, I'll have a I will rest and I my body tells me I need to go and lay down.
Participant 88_2023AUENM

In this PEEK study, people with rare diseases described regular activities they do to maintain both mental and general health. They described consulting a mental health professional, remaining social, making lifestyle changes such as diet and exercise, hobbies, mindfulness and meditation, the importance of family and friends, accepting help and pacing themselves, complying with treatment and being organised and planning ahead.

In other studies, people described ways that they coped with having a rare disease or caring for someone with a rare disease. Some described being informed, organised and planning day to day activities, and having house rules to protect people with immunocompromised conditions[85,146]. Others described the importance of taking care of mental health by seeking help from healthcare professionals or remaining positive and using mindfulness techniques[71,85,97,146]. Some described the importance of enjoying life by getting out and about, or enjoy hobbies, sports and activities[85,146]. Having support from family, support groups and respite carers was an important coping mechanism, as was contributing back to society and helping others[70,84,85,146].

Summary of PEEK results

- Activities to cope with rare disease
- Consult a mental health professional
- Remaining social, lifestyle changes and hobbies
- Mindfulness and meditation
- Importance of family and friends
- Exercise
- Self care; rest, accept help and pacing
- Complying with treatment
- Healthy diet
- Being organised and planning ahead

Summary of literature

Activities to cope with rare disease

- Day to day planning
- House rules to protect immunocompromised child
- Keep informed/seek information
- Seek counselling/Take care of mental health
- Positive attitude and mindfulness
- Get out and about
- Enjoy hobbies, sports and activities
- Contribute to society and help others
- Join a support group
- Supportive family
- Caregivers get respite carers

Relationships

To some extent it has. But I think now looking back, it's more my mental health that's been affected and and affects the social interactions. But I mean, you know, not I wouldn't say or like greatly because our friends, our close friends and family understand and we've educated them about it and stuff. So they're pretty accepting.
Participant 14_2023AUDPA

In this PEEK study, participants most commonly described that having a rare disease had a negative impact on their relationships. This was from people not knowing what to say or do and withdrawing from relationships, the dynamics of relationships changing due to anxiety, exacerbations and/or physical limitations of condition, and social isolation. When participants described a positive impact, this was from family relationships being strengthened, and people that were well-meaning and supportive.

In other studies, people with rare diseases described negative impacts on relationships. Some described difficulties in forming romantic relationships, others the negative impact on intimate relationships[71,146]. The impact on the primary caregiver was difficult and in some cases led to divorce or separation, it also has a negative impact on other family members, in particular healthy siblings, and flare-ups or treatments can be disruptive to family life [84,115]. Friendships and the ability to socialise can be difficult, other people do not want to talk about health conditions, especially if embarrassing, some described difficulties in making friends and described experiencing teasing and bullying[71,99,146]

Summary of PEEK results

Impact on relationships

- 44% described an overall negative impact on relationships
- 9% described an overall positive impact on relationships

Negative impact

- People not knowing what to say or do and withdrawing from relationships
- Dynamics of relationships changing due to anxiety, exacerbations and/or physical limitations of condition
- Social isolation

Positive impact

- Family relationships being strengthened
- Well-meaning and supportive

Summary of literature

Relationships

- Not forming romantic relationships
- Divorce and separation
- Impact on intimate relationships.
- Impact on relationship with family member who is caregiver
- Negative impact on siblings
- Flare-ups and having treatments disruptive to family life
  People don't want to talk about diseases
- Want others to see past their condition but are often treated differently
- Affect on ability to make and maintain friendships
- Impact on social life
- Experienced teasing and bullying

Burden on family

No, the system's a burden, my son. Is not a burden. The system every. Every corner of the system is a burden. No, absolutely not. What I say my son is a burden or or or charged in terms of burden. It's the system, it's the hurdles, it's the challenges, it's the inner, it's the gap, you know, it's the lack of services, it's everything is. It's like a research. It's lack of experts. All of those are, you know, what makes having CHARGE syndrome a burden on my family.
Participant 28_2023AUORC

In this PEEK study, participants described that the rare disease was a burden on family because of the extra household duties and responsibilities that their family must take on, the extra assistance needed to get to appointments, and the emotional strain it placed on their family.

In other studies, people with rare diseases discussed the burden on their family, in particular feeling guilty and wanting their family members to have a break from being a carer and time to be just family[146]. Parents of children with rare disease described burden in terms of not meeting the needs of other family members, especially when child with rare diseases is hospitalised [85].

Summary of PEEK results	Summary of literature
Burden • The extra household duties and responsibilities that their family must take on • Extra assistance needed getting to appointments • Emotional strain placed on their family	Burden • Guilty about family needing to be carer • Caregiver not meeting needs of other family members

Anxiety associated with condition

The rates of depression and anxiety are higher in people with chronic conditions compared to the general population. In a meta-analysis of 20 qualitative studies, it was reported that people with chronic conditions experienced anxiety or depression as either as independent of their chronic condition or as a result of, or inter-related with the chronic disease, usually however, anxiety and depression develops as a consequence of being diagnosed with a chronic disease[151].

In this PEEK study, anxiety associated with breast cancer was measured by the fear of progression questionnaire[152]. On average participants in this PEEK study had a moderate fear of progression, they were most concerned that at some point in time will no longer be able to pursue hobbies because of illness. In other studies, people with rare diseases described the fear and anxiety that they have about their condition. The fear of the condition getting worse and the implications that will have on the ability to lead a normal or independent life, as well as contributing to feelings of depression

In other studies, people wih rare diseases described aspects of rare diseases that caused them anxiety, this included having dependency on others, limitations for daily activities, limitations for social interactions, side effects of treatment, and for caregivers, they described being stressed in general and about the shortened life of their child[34,99,146,153].

Summary of PEEK results	Summary of literature
Anxiety associated with condition • Moderate fear of progression • Often concerned that at some point in time will no longer be able to pursue hobbies because of illness	Anxiety associated with condition • High levels of caregiver stress • Dependency on others • Limitations for daily activities • Limitations for social interactions • Shortened lives • Side effects of treatment

Characterisation

In this PEEK study, a total of 407 participants with rare diseases or carers to people with rare diseases were recruited into the study. The majority of participants lived in major cities, they lived in all levels of economic advantage. Most of the of participants identified as Caucasian/white, aged mostly between 35 and 64. Half of the participants had completed some university, and most were employed either full time or part time. Almost half of the participants were carers to family members or spouses.

Physical health interfered with work and other activities for participants in this study, they had poor energy levels and poor general health.

This is a group that had health conditions other than their condition to deal with, most often anxiety, sleep problems, and chronic pain.

Most participants sought medical attention after noticing symptoms and were diagnosed after their a complex pathway involving a number of specialists.

This is a cohort that was diagnosed by a specialist at a specialist clinic or in hospital. The majority did not have any out of pocket expenses at diagnosis, however, for those that did have out of pocket expenses it was a moderately significant burden.
This is a group that did not have enough emotional support at the time of diagnosis. This is a cohort that did not have conversations about biomarker/genomic/gene testing, though are interested in these types of tests.

This is a study cohort that had no or limited knowledge about their condition before they were diagnosed. This patient population that had uncertainty about their prognosis, or described their prognosis in terms of symptoms and function or changes in symptoms and function.

This is a patient population that had no discussions about treatment or were given multiple treatment options. Some participated in decision making but others were told what to do without discussion.

This is a study cohort that took into account side effects and efficacy as part of many considerations when making decisions about treatment.

Within this patient population, about half of the participants had changed decision making over time, this was linked to being more informed and assertive.

When asked about their personal goals of treatment or care participants most commonly described wanting quality of life or return to normality.

This is a group who felt they were mostly treated with respect throughout their experience.

Approximately two-thirds of this cohort had private health insurance, half were public patients treated in mostly in the public hospital system. This is a group that did not have trouble paying for healthcare appointments, prescriptions, and paying for basic essentials. Their monthly expenses due to their condition were somewhat of a burden.

Participants in this study had to quit, reduce hours, or take leave from work. Carers and family did not have to change employment status. The loss of family income was a burden.

Participants on average used one allied health service, one complementary therapy and made one lifestyle change.

More than a third had conversations about clinical trials, and the majority would take part in a clinical trial if there was a suitable one for them.

This is a patient population that described mild side effects using an example such as fatigue and as those which can be self-managed and do not interfere with daily life.

This is a study cohort that described severe side effects as symptoms such as pain, they also described severe side effects as those that impact everyday life and the ability to conduct activities of daily living.

This is a patient population which described adhering to treatments according to the advice or their doctor or that they would stick with it for 2 to 3 months. This is a study cohort that needed to see physical signs disappear to feel that treatment is working as well. If treatment did work, it would allow them to return to everyday activities

Participants in this study had very good knowledge about their condition, were average at coping with their condition, were good at recognizing and managing symptoms, and were very good at adhering to treatment.

Participants were given information about treatment options, disease management and , disease cause from health care professionals, and searched for the same topics most often. This is a group who accessed information from non-profit, charity or patient organisations most often.

This is a patient population that access information primarily through the internet, Facebook or social media, and from health charities.

This is a study cohort that found information from other people's experience to be helpful, and that no information was unhelpful.

This is a group that preferred online information, or talking to someone. This is a study cohort that generally felt most receptive to information from the beginning, at diagnosis.

Most participants described receiving an overall positive experience with health professional communication (some with a few exceptions) which was holistic, two way and comprehensive. For those that had a negative experience it was mostly because their healthcare professionals had a lack of knowledge about their condition.

The participants in this study experienced good quality of care, and average coordination of care. They had a moderate ability to navigate the healthcare system, and experienced moderate communication from healthcare professionals.

This is a patient population that did not have any formal support or found support in the clinical setting or from family and friends.

This is a patient population that experienced a negative impact on quality of life largely due to emotional strain on family, and changes to relationships.

Life was a little distressing for this group, due to having a rare disease

This is a study cohort that experienced at least some impact on their mental health and to maintain their mental health they used coping strategies such as consulting a mental health professional or remaining social, lifestyle changes and hobbies.

Within this patient population, participants described the importance of self-care, and complying with treatment in order to maintain their general health.

Participants in this study had felt vulnerable when having sensitive discussion about their condition. To manage vulnerability, they used self help methods such as resilience, acceptance and staying positive.

This cohort most commonly felt there was an overall negative impact on their relationships, due to people withdrawing from relationships or not knowing what to say.

Participants felt they were a burden on their family, due the extra household duties and responsibilities that their family must take on.

Most participants felt there was some cost burden which was from the costs of taking time off work and from the cost of treatments.

The participants in this PEEK study had moderate levels of anxiety in relation to their condition.

Participants would like future treatments to be more affordable, and more effective.

This is a study cohort that would like information to be more accessible and to provide more information about disease trajectory.

Participants in this study would like future communication to include health professionals with a better knowledge of their condition, and for more empathy.

Participants would like future treatments to include access to appropriate real-world support services.

This patient population was grateful for healthcare staff, including access to specialists.

Participants' message to decision-makers was the need for timely and equitable access to support, care and treatment

This is a patient population that wished had been more assertive, been an advocate, more informed and asked questions.

The aspect of care or treatment that participants in this study would most like to change is to accessed their specialist sooner, however, many wouldn't change any aspect of their treatment or care.

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Section 12

Next steps

Next steps

At the end of each PEEK study, CCDR identifies three key areas that, if improved, would significantly increase the quality of life and/or the ability for individuals to better manage their own health.

In relation to this community, these three areas are:

1.Information
Information that provides families with more current and specific details about diagnosis, treatment, allied health management, prognosis, practical support and importantly, peer support. This may also include information that patients/families can take to clinicians to educate them on the condition, and to give to families and friends to help them understand the condition. Treatment information and decision tools should empower families to weigh up benefits, risks, side effects, quality of life, and to be informed about costs.

2. Care coordination
Pre and post diagnosis there is a complex health system that needs to be navigated to ensure patients are accessing allied health and supportive care. This patient population would benefit from health system navigation services to support timely diagnosis to and to provide holistic management of a broad range of rare diseases to patient and their families and ensure continuity of care across health and social services.

3. Support
Rare diseases have a negative impact on quality of life, mental health and relationships often due to the emotional and mental health strain. This group could benefit from emotional and mental health support both for patients, families and carers. Respite care will be an important aspect of this to allow carers time to access these services.

2024 PEEK study in Rare and Genetic Conditions

Data collected in this PEEK study also provides a basis on which future interventions and public health initiatives can be based. Some of the 2023 metrics that the sector can work together to improve upon are provided in Table 12.1

Table 12.1 Rare diseases 2024 Metrics

Measure	Detail	Mean	Median
Baseline health (SF36)	Physical functioning	54.32	55.00
	Role functioning/physical	37.24	25.00
	Role functioning/emotional	51.22	66.67
	Energy/fatigue	33.69	30.00
	Emotional well-being	64.09	68.00
	Social functioning	53.08	50.00
	Pain	55.69	55.00
	General health	41.64	40.00
	Health change	44.76	50.00
Knowledge of condition and treatments (Partners in Health)	Knowledge	24.07	26.00
	Coping	14.35	14.00
	Recognition and management of symptoms	18.89	19.00
	Adherence to treatment	13.12	14.00
	Total score	70.44	72.00
Care coordination scale	Communication	35.55	36.00
	Navigation	22.96	23.00
	Total score	58.51*	60.00
	Care coordination global measure	5.79	6.00
	Quality of care global measure	6.59	7.00
Fear of progression	Total Score	37.09*	37.00
		Percent	
Accessed My Health Record	-	39.31	-
Participants that had discussions about biomarkers/genetic tests	-	33.44	-