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%).