PERSONAL EXPERIENCE EXPECTATIONS KNOWLEDGE

PEEK

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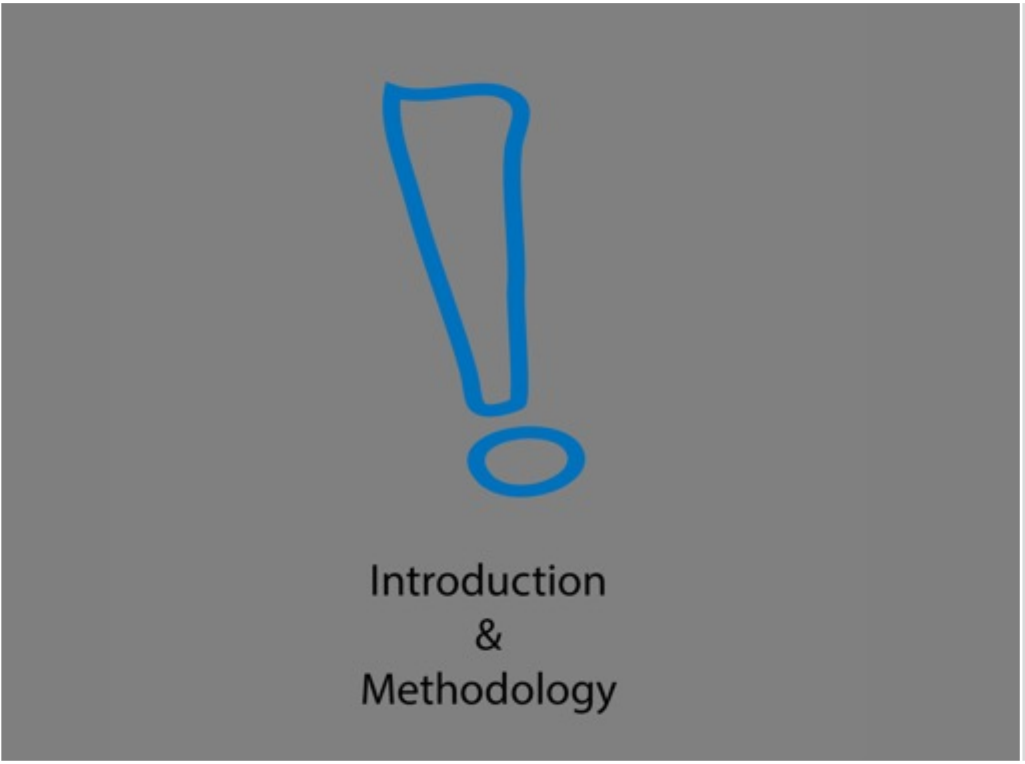
RARE & GENETIC CONDITIONS

AUSTRALIA 2024

Centre for Community-Driven Research

Section 1



This is where you can read about the PEEK methodology, information about the recruitment period and approach.

There is also a review of other mixed methodology studies in the field to identify the position of the PEEK study in the context of other literature. PEEK studies are often the largest of their kind in depth and participant numbers.

Full Report
Summary of Results
Infographic
Slide Set
Introduction & Methodology
Demographics
Symptoms & Diagnosis
Decision-making
Treatment
Communication & Information
Care & Support
Quality of Life
Expectations & Messages to Decision-Makers
Advice from current patients to patients in the future
Discussion
Where to from here?

The Centre for Community-Driven Research team for this study included:

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Thank you to each and every person that participated in this PEEK study and partners that supported the project and all of the health professionals across Australia that also helped to spread the word about this PEEK study.

This study was generously sponsored by a consortium including Illumina, Alexion, Novartis, Pfizer and Roche.

Funders provided an arm's length grant through Victorian Clinical Genetic Services in partnership with SWAN Australia and Genetic Alliance Australia, for the Centre for Community-Driven Research to report on the PEEK protocol data for people who have been diagnosed with rare or genetic conditions. The sponsors and partners had no input into the methodology, data collection, data analysis or reporting.

Position of this PEEK study

In this PEEK study, xxx people diagnosed with rare and genetic conditions throughout Australia participated in the study that included a qualitative structured interview and quantitative questionnaire. PEEK is largest study of rare diseases conducted in an Australian population. In addition, PEEK is a comprehensive study covering all aspects of disease experience from symptoms, diagnosis, treatment, healthcare communication, information provision, care and support, quality of life, and future treatment and care expectations.

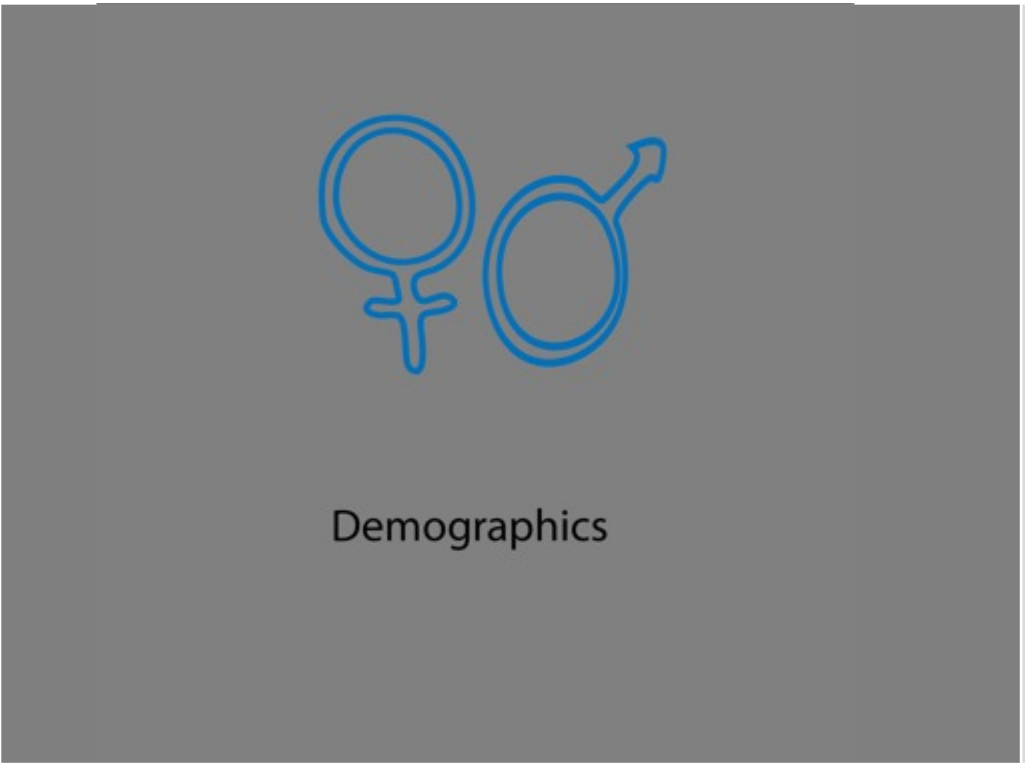
A comparison of studies is available in Section 1.

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

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).

Section 2



The demographic section is where you can find information about the study cohort. This is also where the sub-groups used throughout the study are described and where the baseline health of the cohort is described, using the SF36 questionnaire.

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

Demographics

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

Demographics

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

Subgroups

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

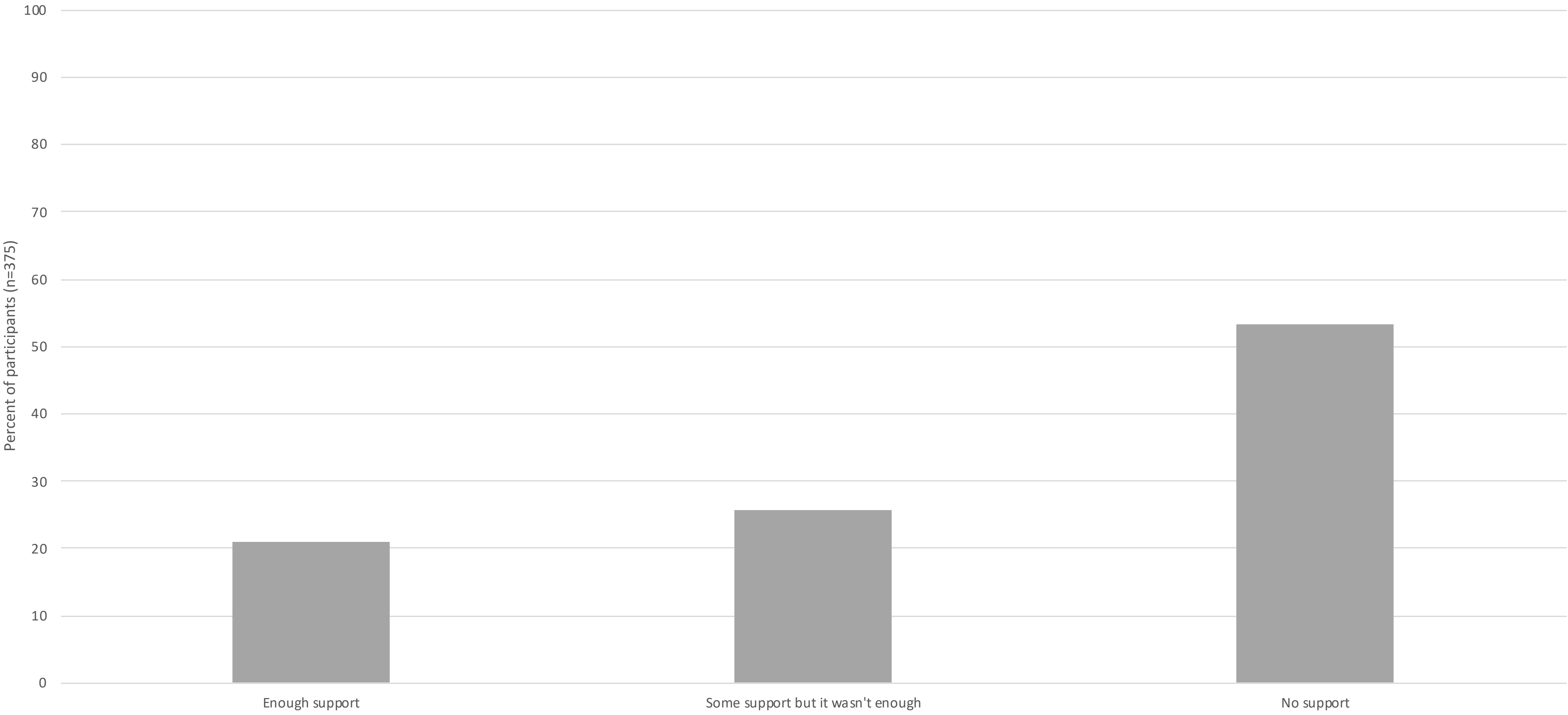
Skewed distribution, use median and IQR as central measure. Possible range 0-100

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,

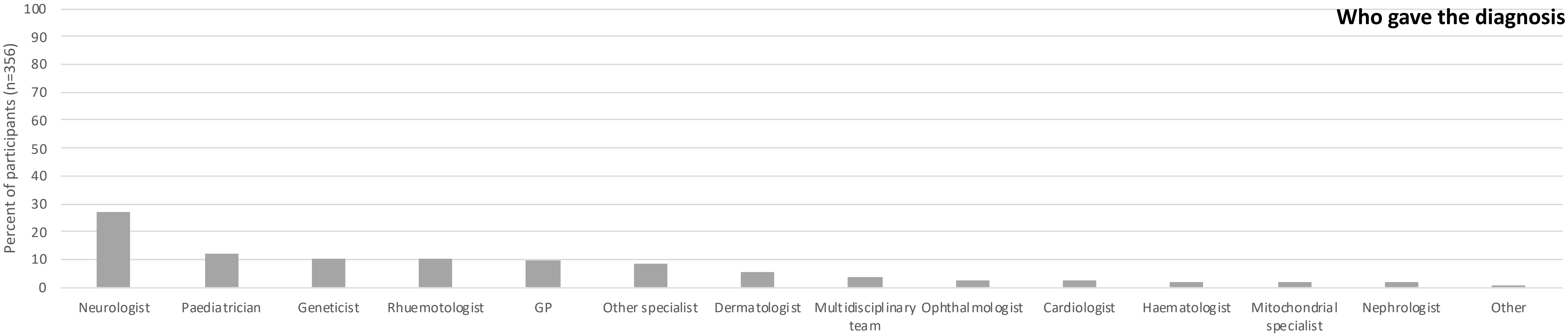
Baseline health

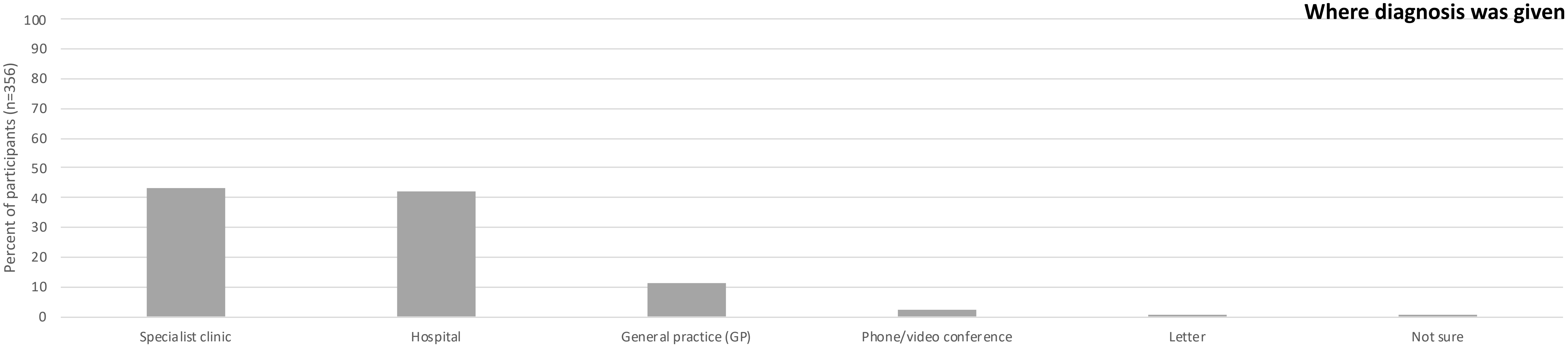
Section 3

The symptoms and diagnosis section is where you can find information about experience of symptoms before diagnosis, as well as the symptoms that led to the diagnosis. There is information about the diagnostic tests that were performed, where the tests were conducted and the time from tests to diagnosis. In this section, there is also information about the participant's understanding of their condition and their understanding of their prognosis.

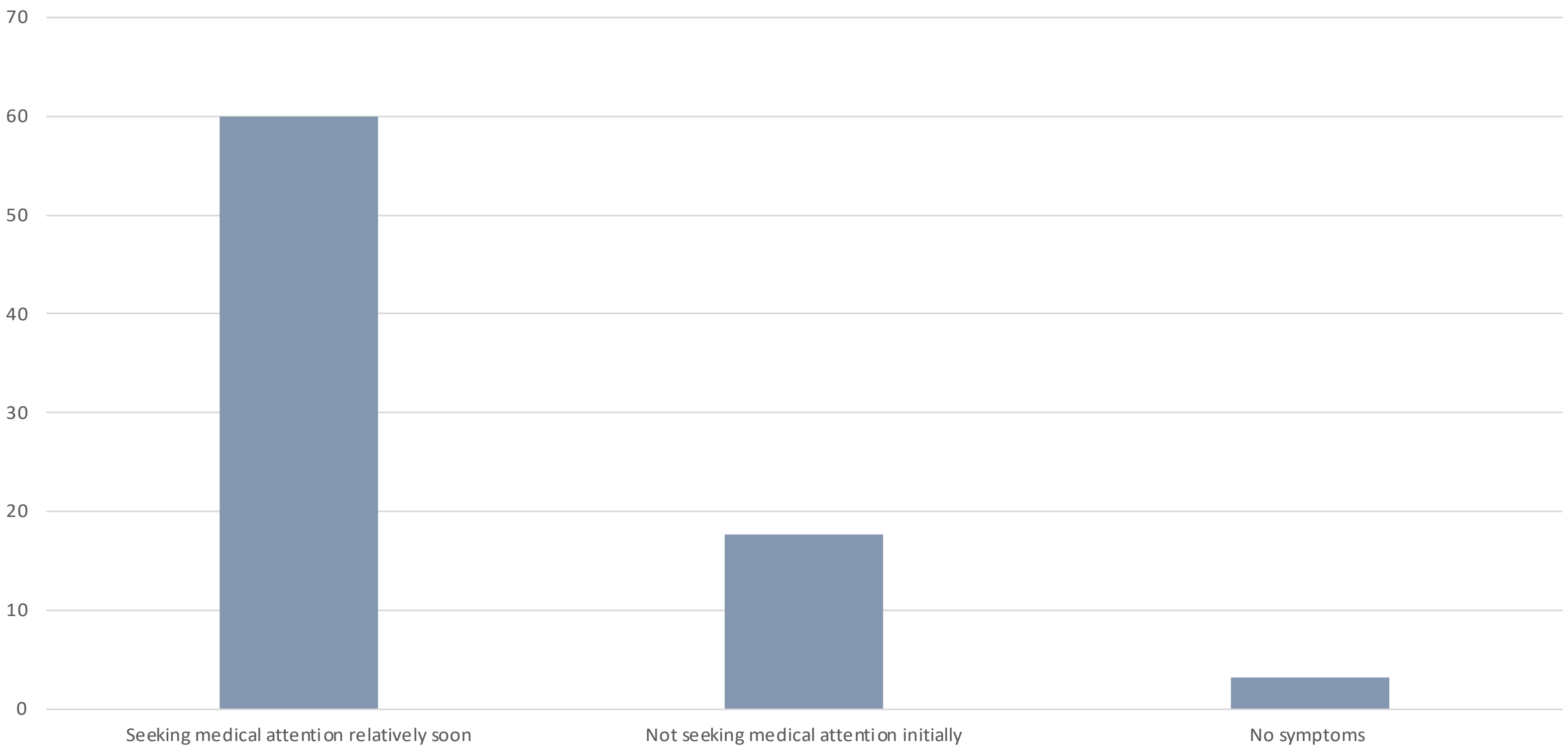


Support at diagnosis


Who gave the diagnosis
Percent of participants (n=356)
100
90
80
70
60
50
40
30
20
10
0
Neurologist
Paediatrician
Geneticist
Rhuemotologist
GP
Other specialist
Dermatologist
Multidisciplinary team
Ophthalmologist
Cardiologist
Haematologist
Mitochondrial specialist
Nephrologist
Other


Where diagnosis was given
Percent of participants (n=356)
100
90
80
70
60
50
40
30
20
10
0
Specialist clinic
Hospital
General practice (GP)
Phone/video conference
Letter
Not sure

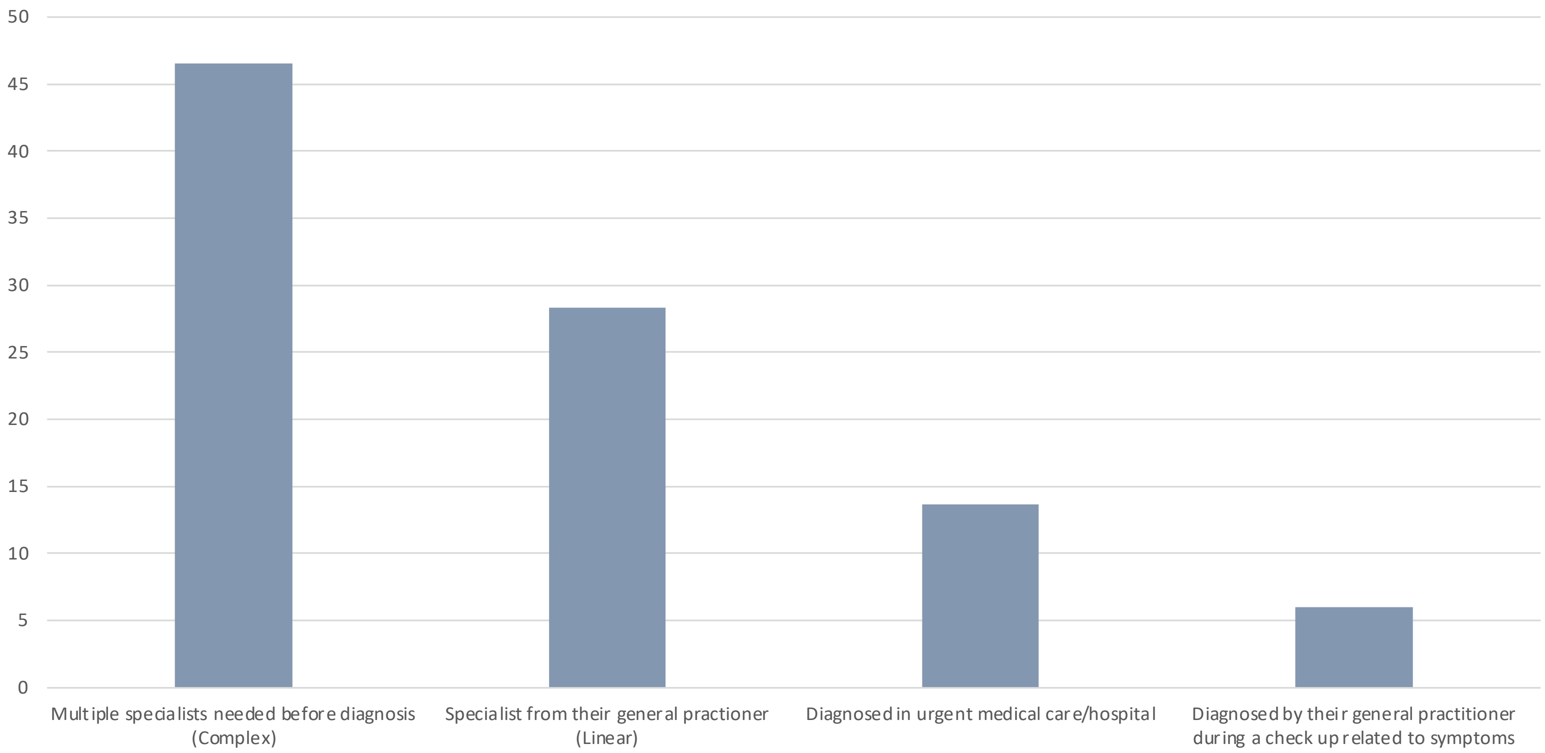
Diagnosis



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

Seeking medical attention (% of all participants)



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

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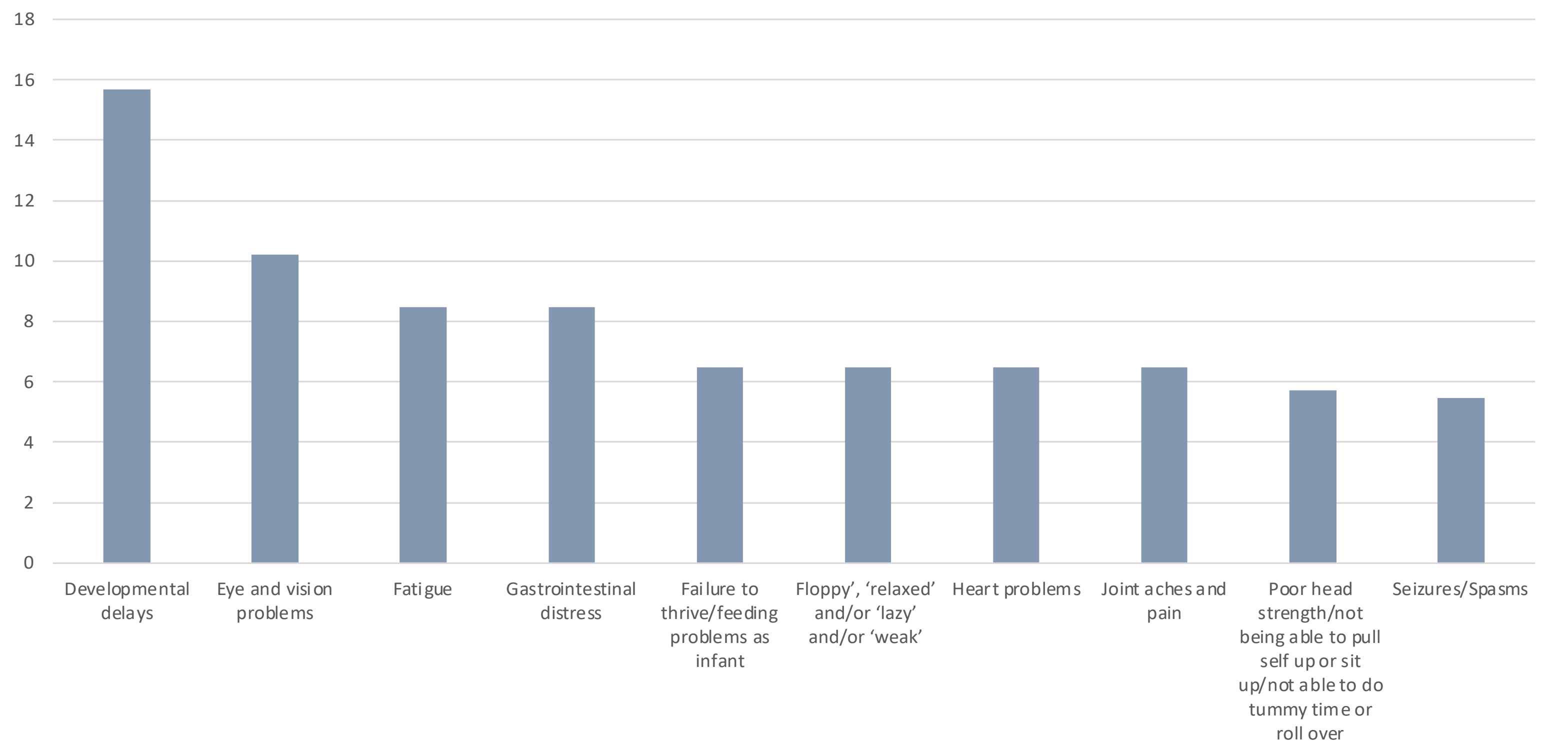
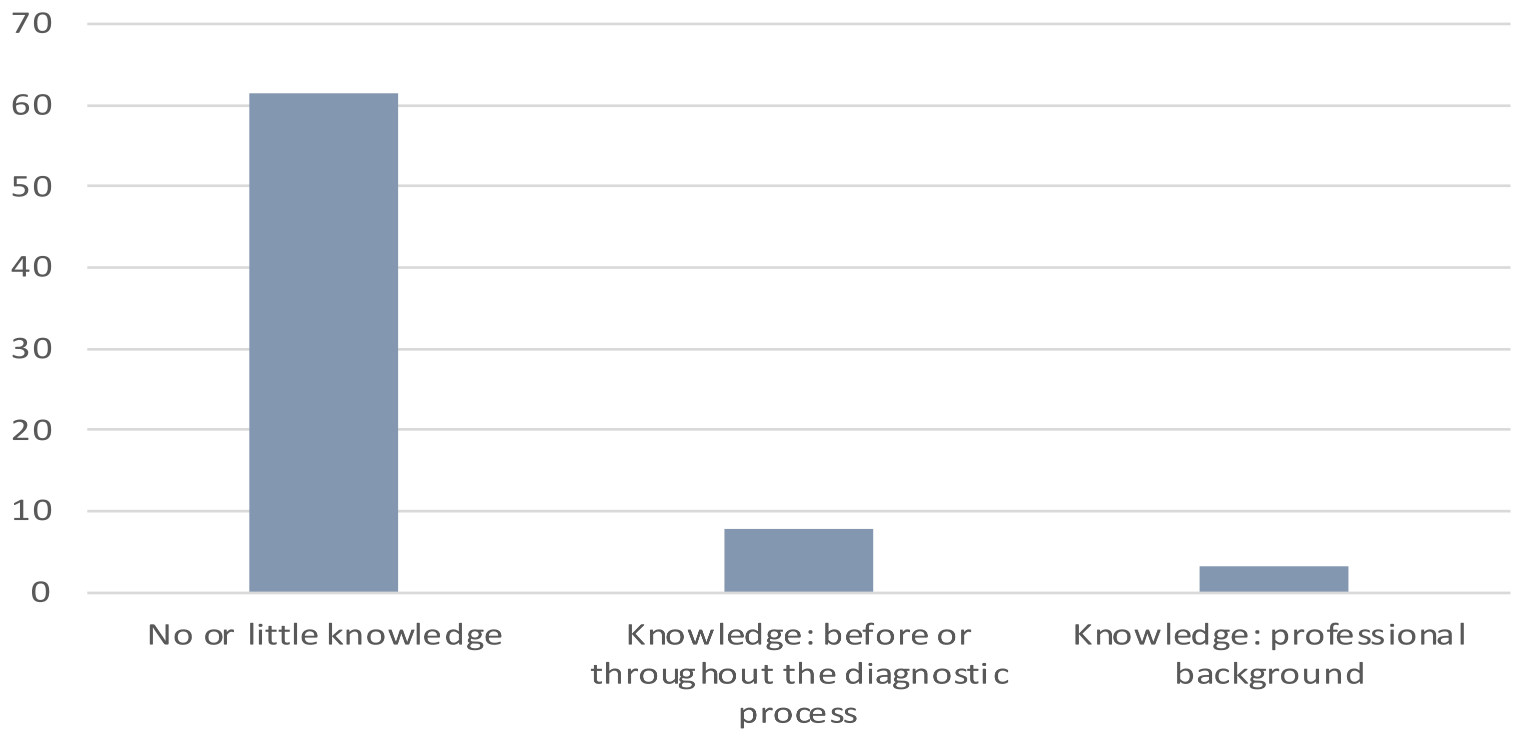
Diagnostic pathway



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. I couldn't sit until he was 2 1/2. So developmental milestones is definitely probably major indicator.
Participant 081_2023AUDIS

Symptoms leading to diagnosis (% of all participants)

Understanding of disease 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 didn't really know much. I had to do a lot of research after I was given the name of it, pretty much like they gave me a fact sheet. But it was like 2 sided that was it? Participant 003_2023AUDSK

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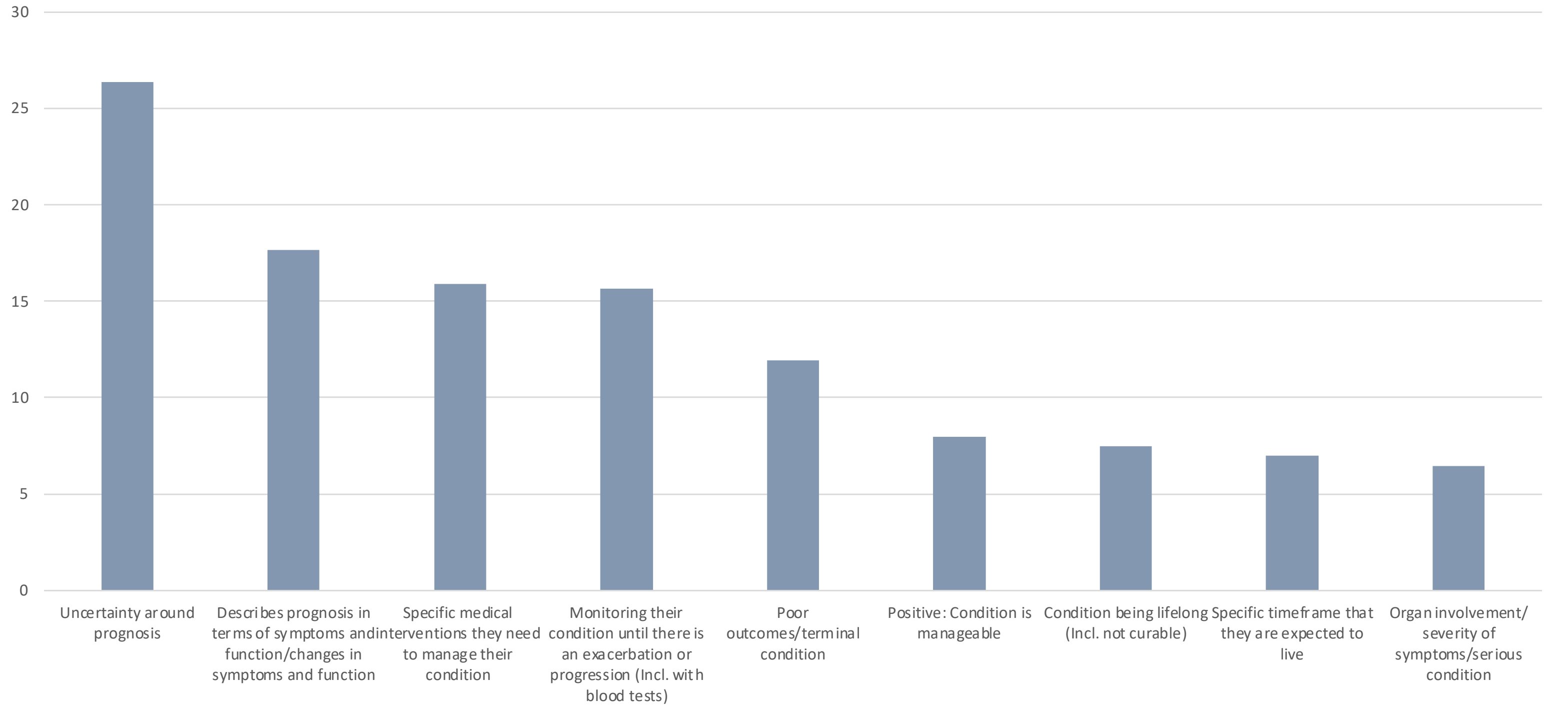
Understanding of prognosis

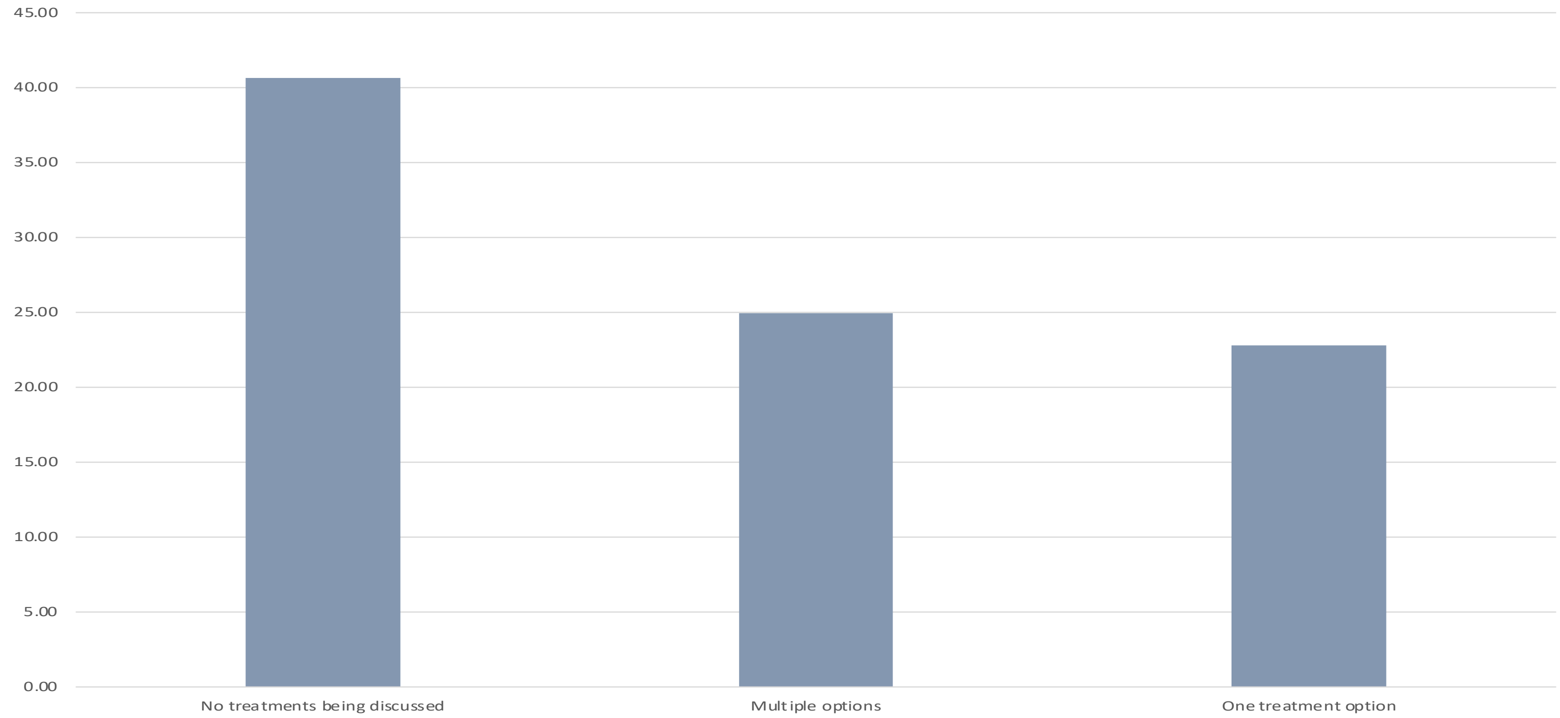


It's considered a rare disease in Australia. So the prognosis for NAME, who is typical in the terms that I am a carrier with a very mild version of the disease and she is a, is a, she has the disease full blown whereas where I don't really and then so her prognosis is unknown. Participant 080_2023AUDIS

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

Section 4

In the decision-making section, you can find information about the conversations that were had about treatment options at diagnosis, what is important to people when they make decisions about treatment, and whether decision-making changes over time.



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. For something fair, 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

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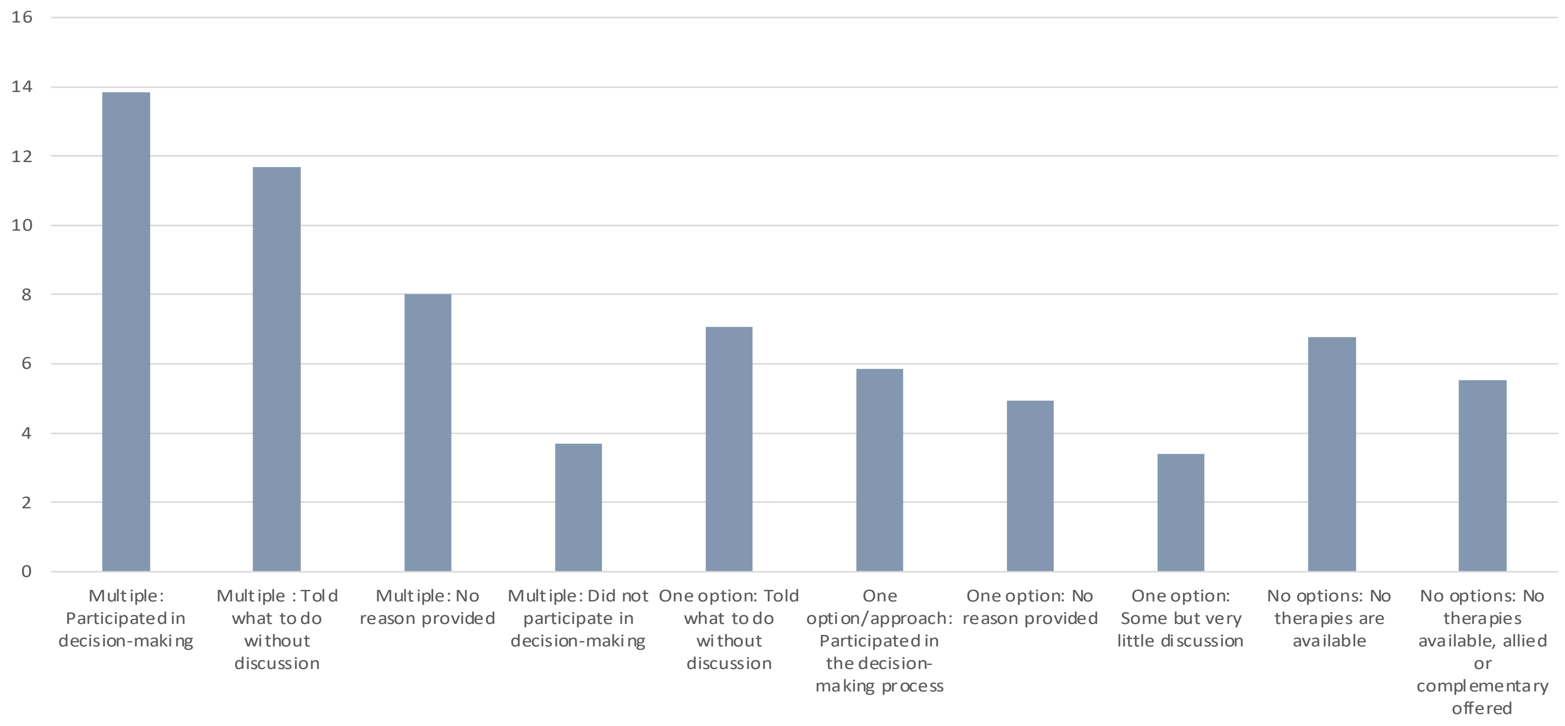
Discussions about treatment (% of all participants)



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

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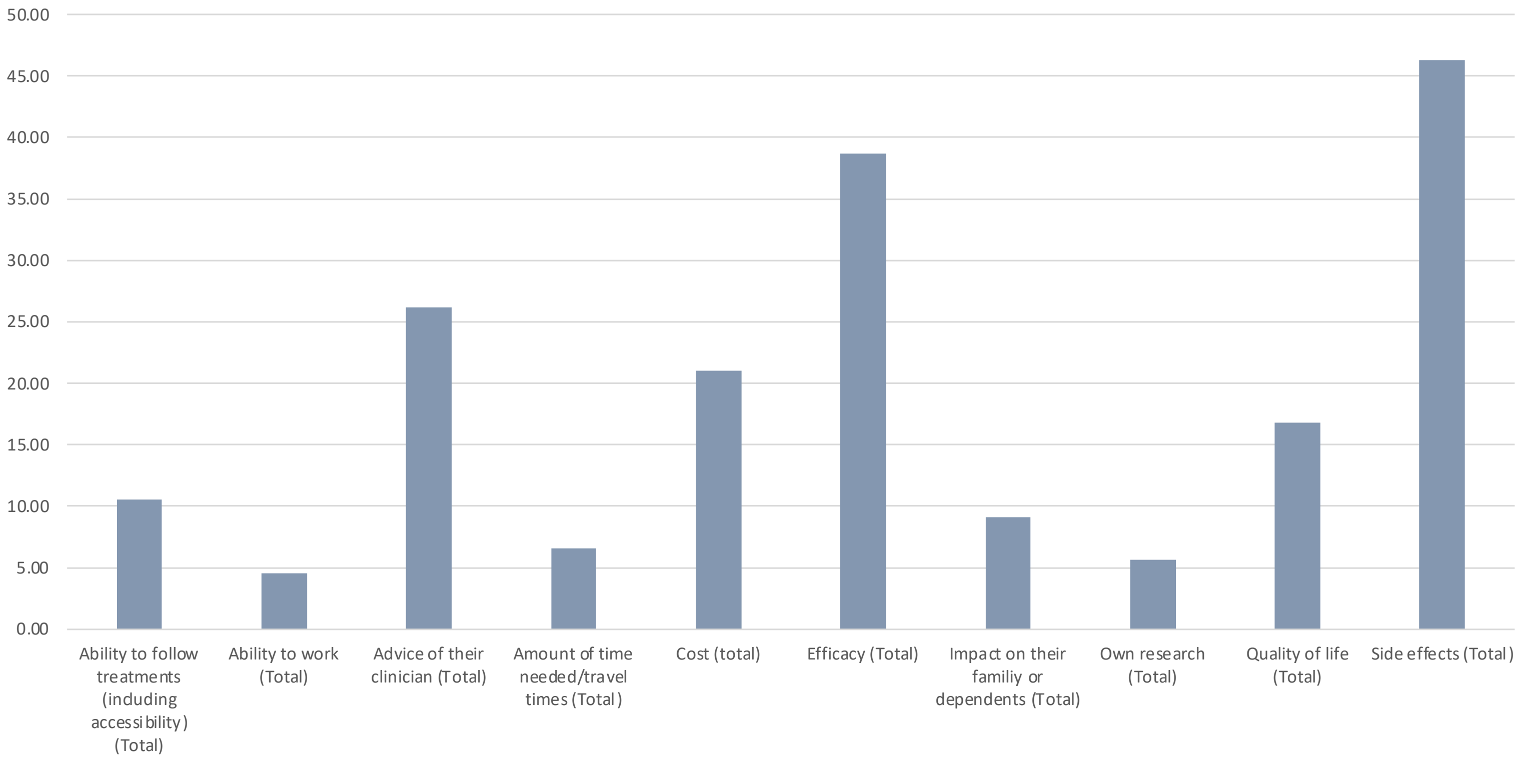
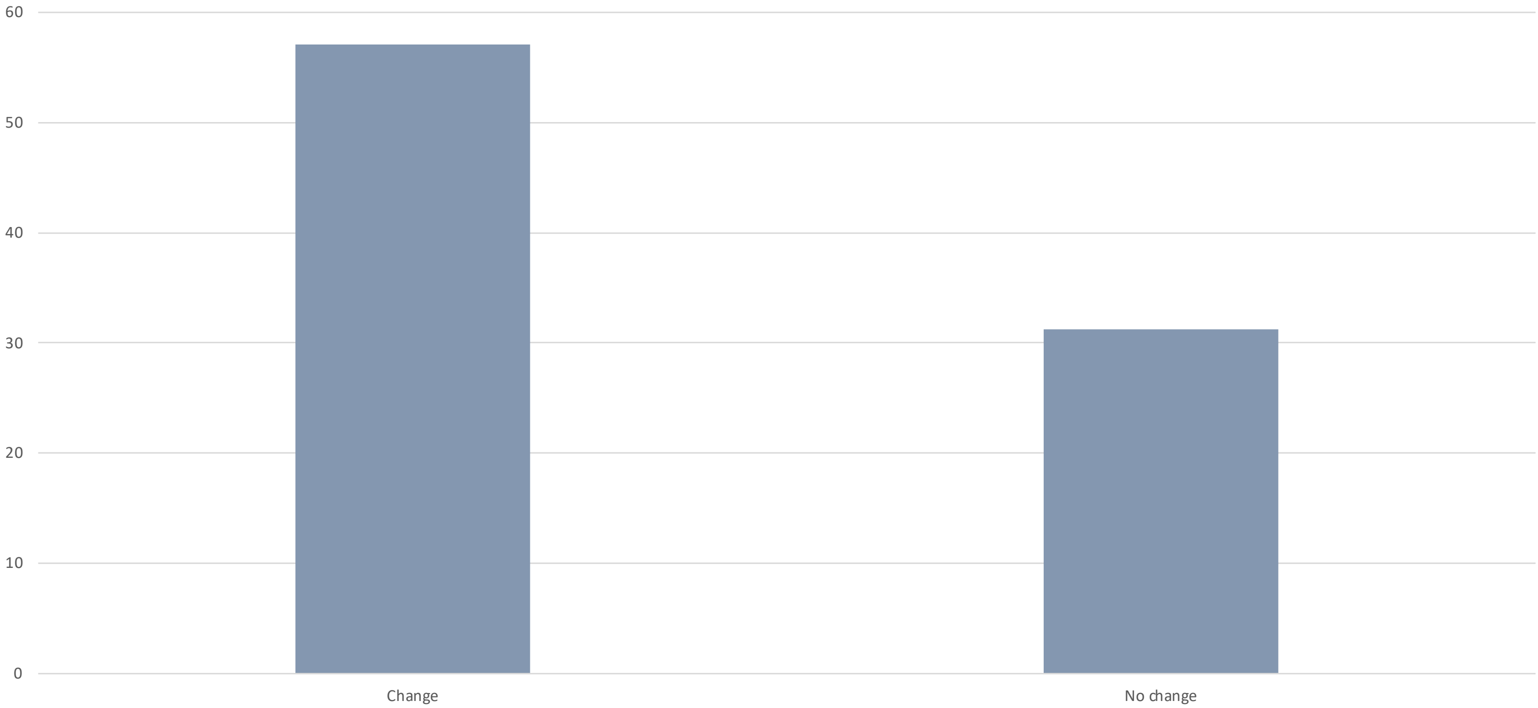
Participation in management discussed (% of all participants)



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

Considerations when making decisions about treatment

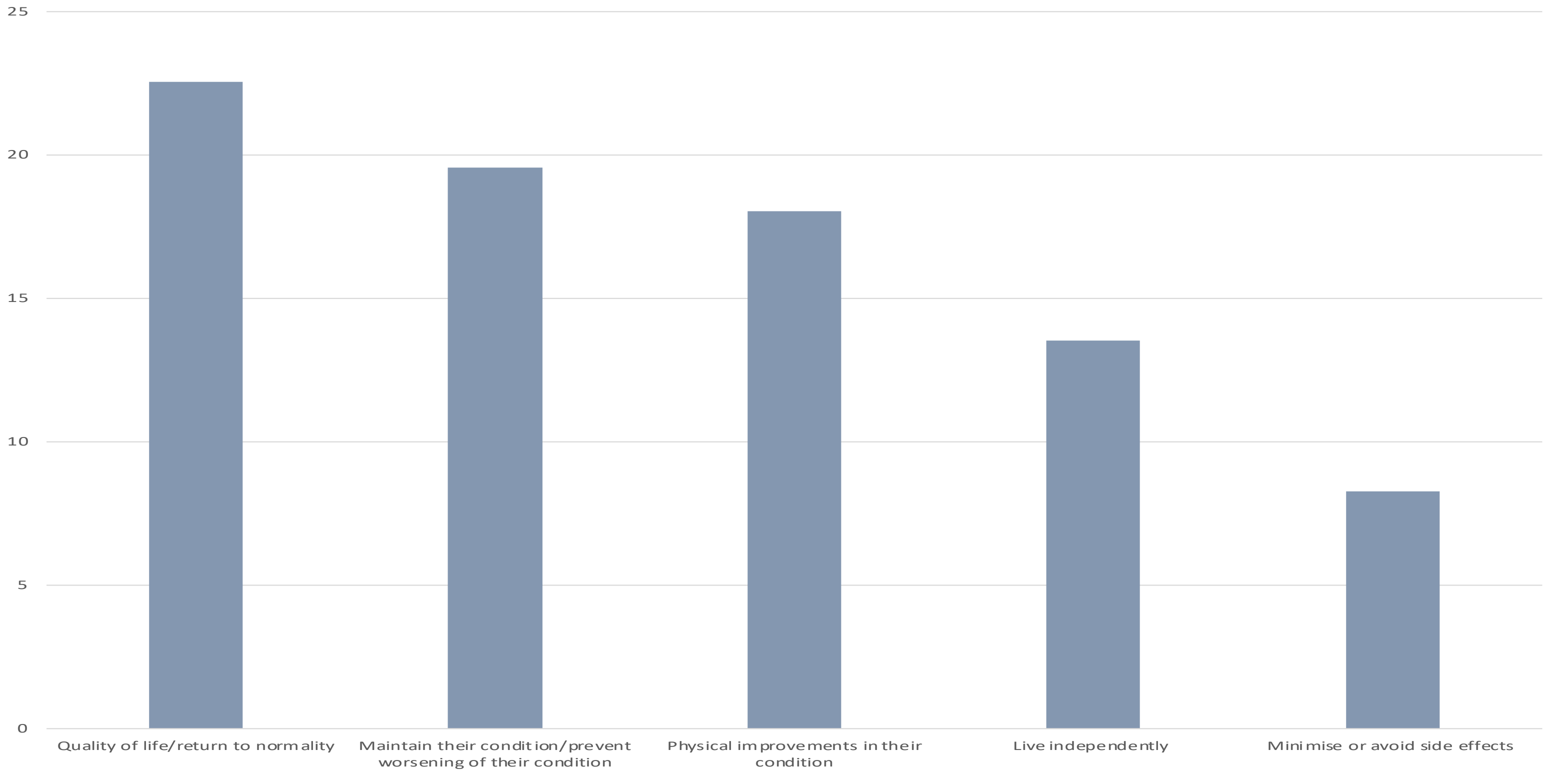


Look, I still think I make decisions in the same way, which is to get lots of different views and to get lots of different information and. Decide whether I trust the professional, whether the professional has expertise in that area, whether there's other evidence that supports that, you know, idea of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

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

Changes in decision-making over time (% of all participants)

Personal goals of treatment (% of all participants)



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 think 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

Section 5

Treatment

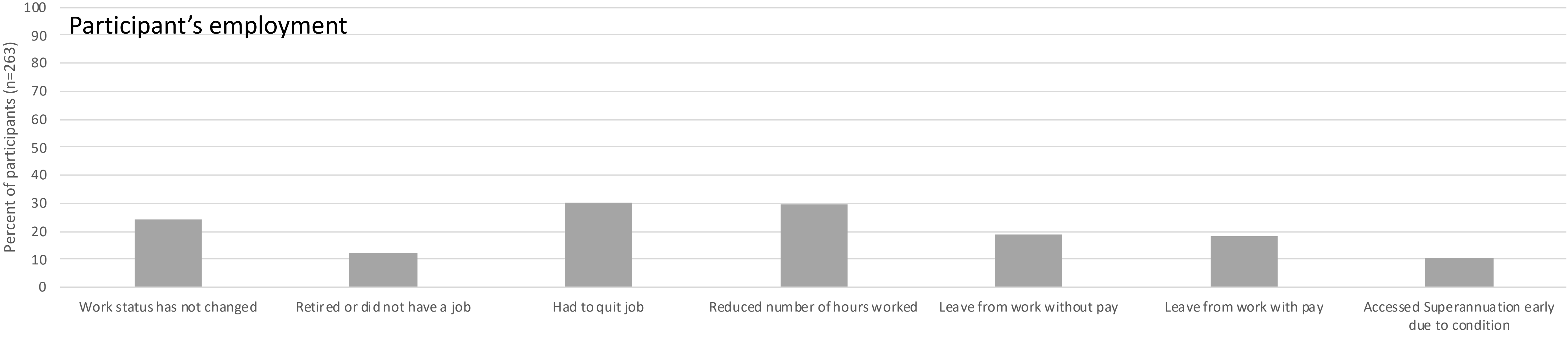
The treatment section is one of the largest sections in each study. Information is available in relation to the types of treatments people have experienced as well as rating quality of life and effectiveness for each treatment. There is information about the main providers of treatment, access to health professionals and information about which part of the health system participants were treated in. There is information about affordability of healthcare, any reduced income for the patient or their family, an additional costs or changes to employment. There is then also detailed information about the treatments that the participants have experienced, whether they had any side effects, their description of mild side effects and severe side effects, and any complementary therapies that they may have used. Information in relation to access to allied health is also available. Information about what needs to change to feel like treatment is effective, and adherence to treatment is also presented. In this section, there is also information on whether discussions about clinical trials were held with clinicians, and for those who have been on a trial, what their experience was like.

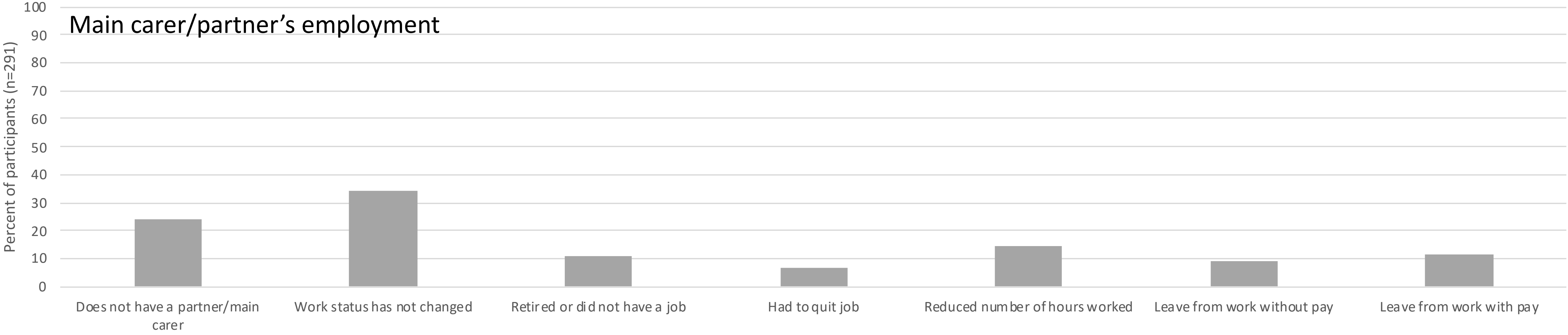
Access to healthcare

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

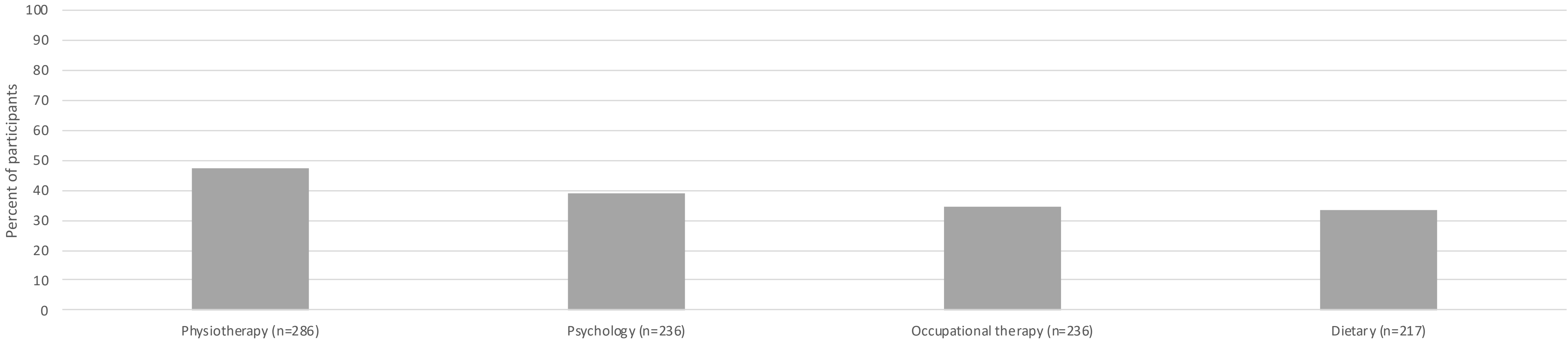
Affordability of healthcare	Response	Number	Percent
Delay or cancel healthcare appointments due to affordability	Never	215	59.56
	Rarely	44	12.19
	Sometimes	66	18.28
	Often	25	6.93
	Very often	11	3.05
Did not fill prescriptions due to cost	Never	260	72.02
	Rarely	45	12.47
	Sometimes	43	11.91
	Often	10	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

Employment status


Participant's employment

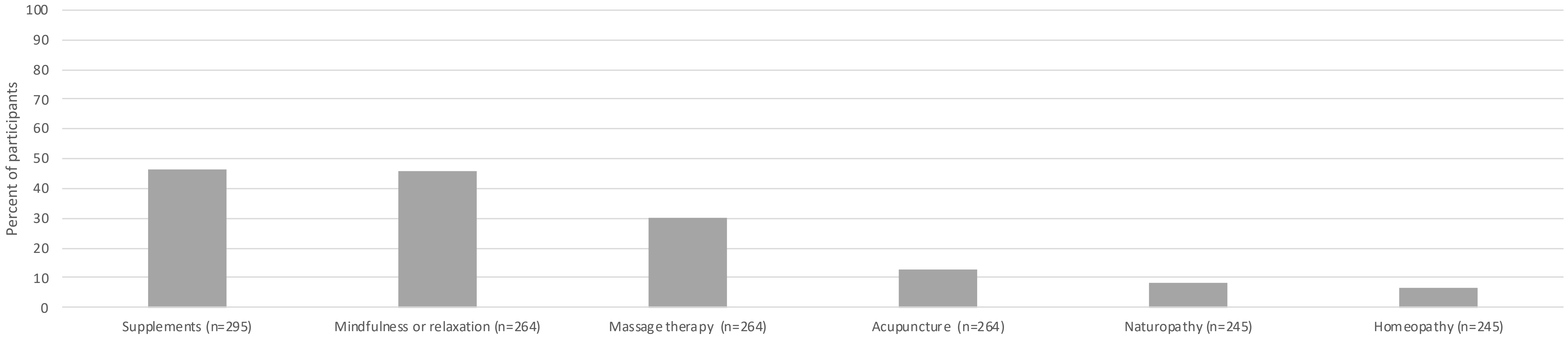

Main carer/partner's employment

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



Allied health

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



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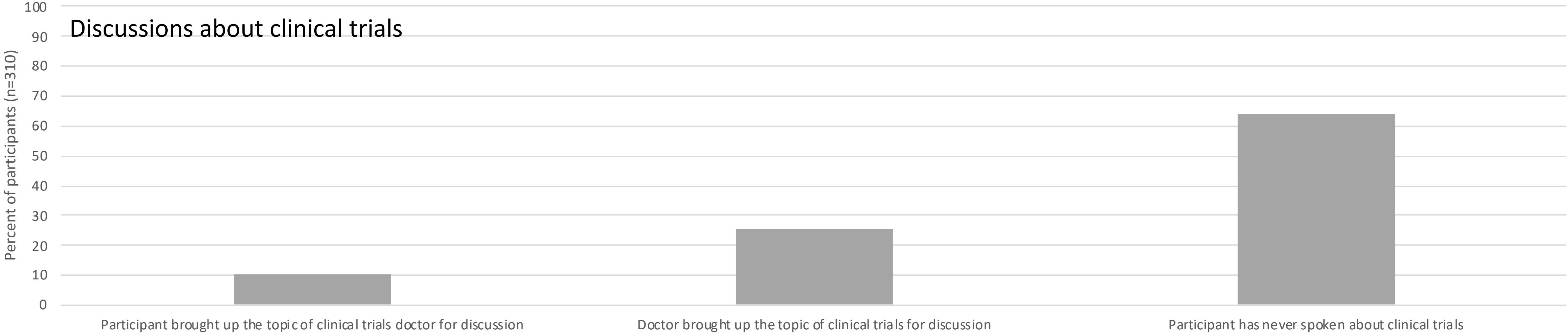
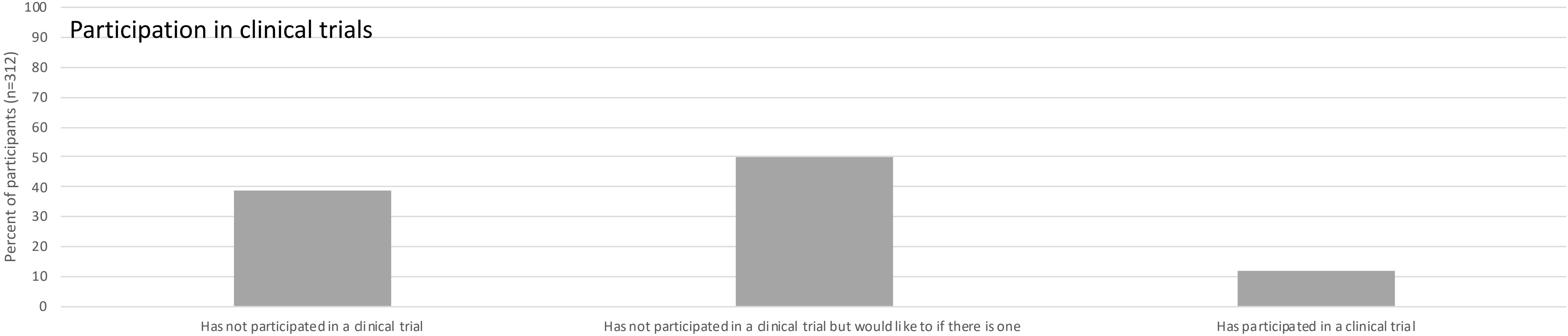
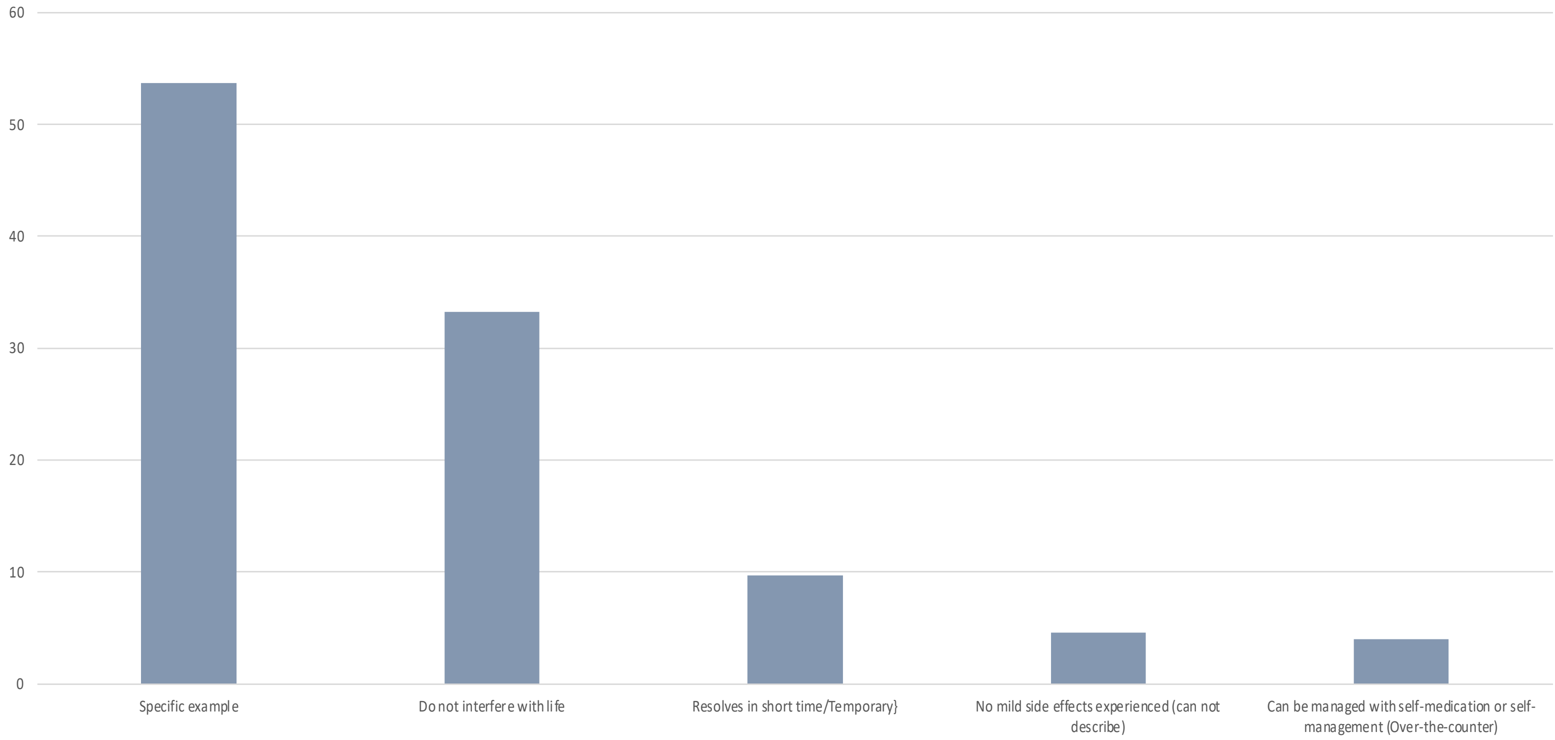
Complementary therapies

Clinical trials

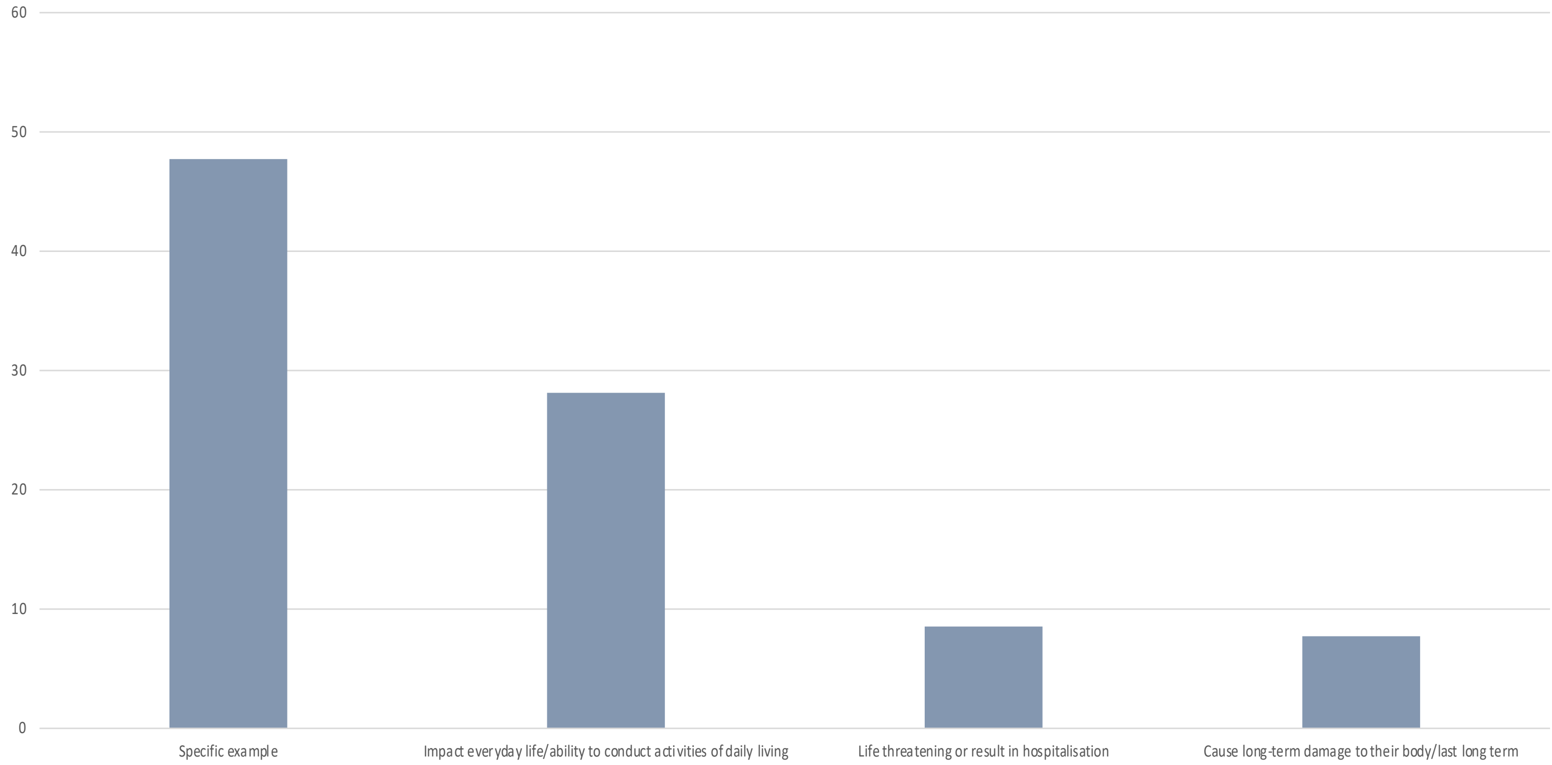

Discussions about clinical trials


Participation in clinical trials

Description of mild side effects (% of all participants)



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



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. Participant 024_2023AUDPA

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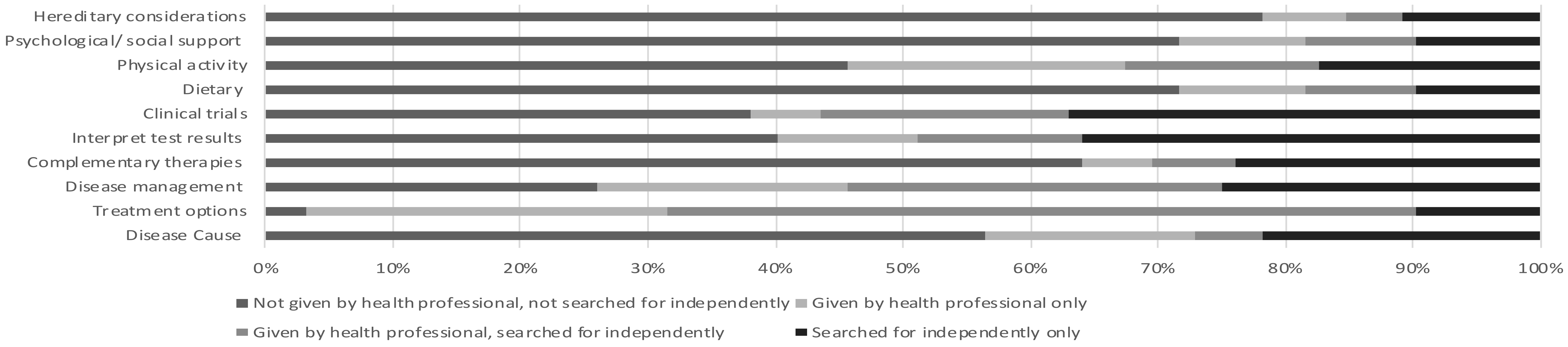
Description of severe side effects (% of all participants)

Section 6

Communication
&
Information

This section report access to information, information that was helpful and information that was not helpful, when participants feel they were most receptive to receiving information, information preferences, information topics for which information was received or searched for, as well as when it was received and additional information was required. This is also where there is information about healthcare professional communication and participant’s knowledge and confidence in managing their own health using the Partners in Health tool.



The largest gaps in information, where information was neither given to patients nor searched for independently were hereditary considerations (n = 72, 78.26%) and psychological/social support (n = 66, 71.74%), and dietary information (n = 66, 71.74%).

The topics that participants did not search for independently after not receiving information from healthcare professionals were treatment options (n = 26, 28.26%) and physical activity (n = 20, 21.74%).

The topics that participants were given most information from both healthcare professionals and searching independently for were treatment options (n = 54, 58.70%) and disease management (n = 27, 29.35%).

The topics that participants searched for independently after not receiving information from healthcare professionals were clinical trials (n = 34, 36.96%) and how to interpret test results (n = 33, 35.87%) (Table 6.28, Figure 6.47).

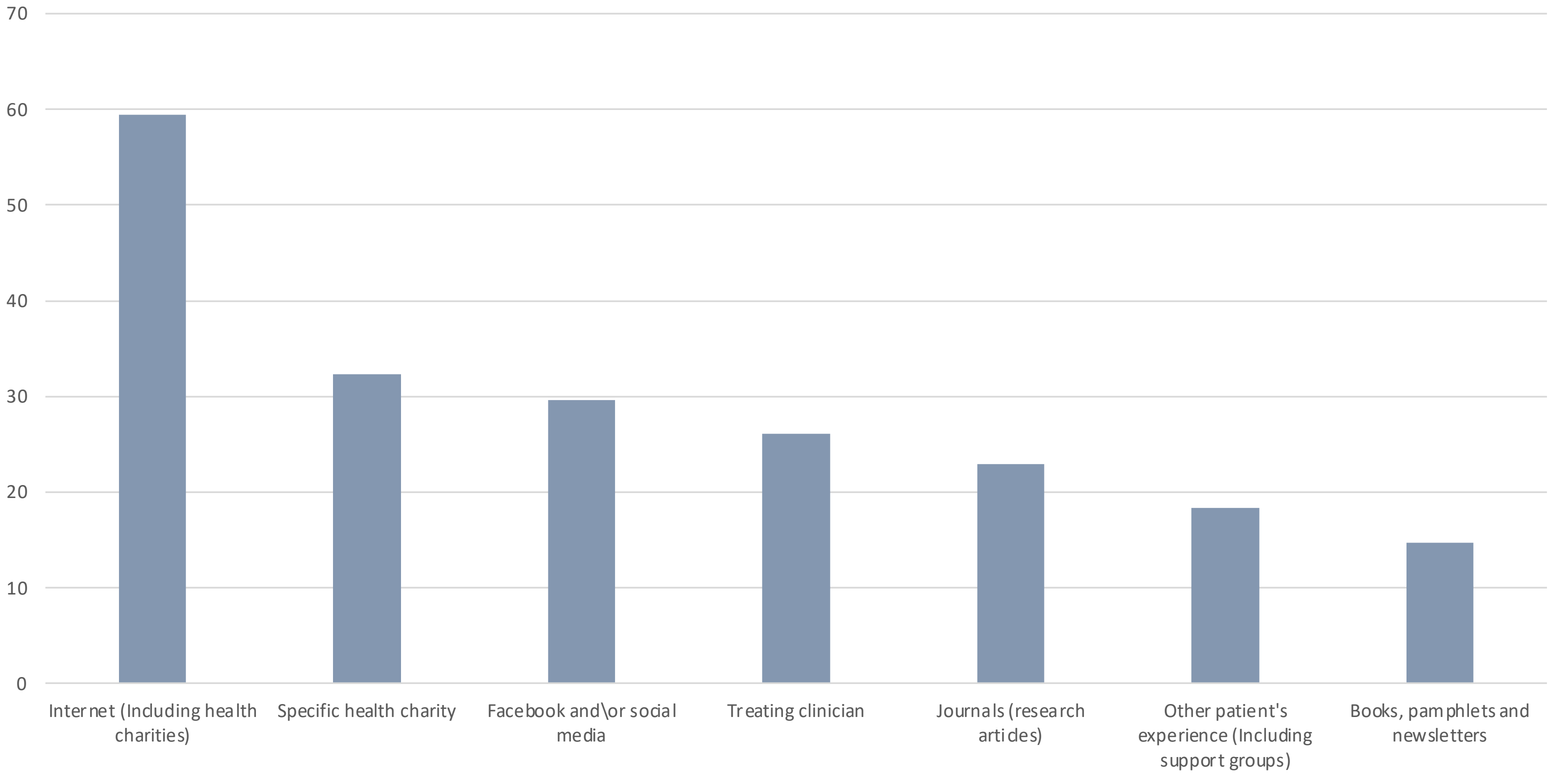
Information gaps

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

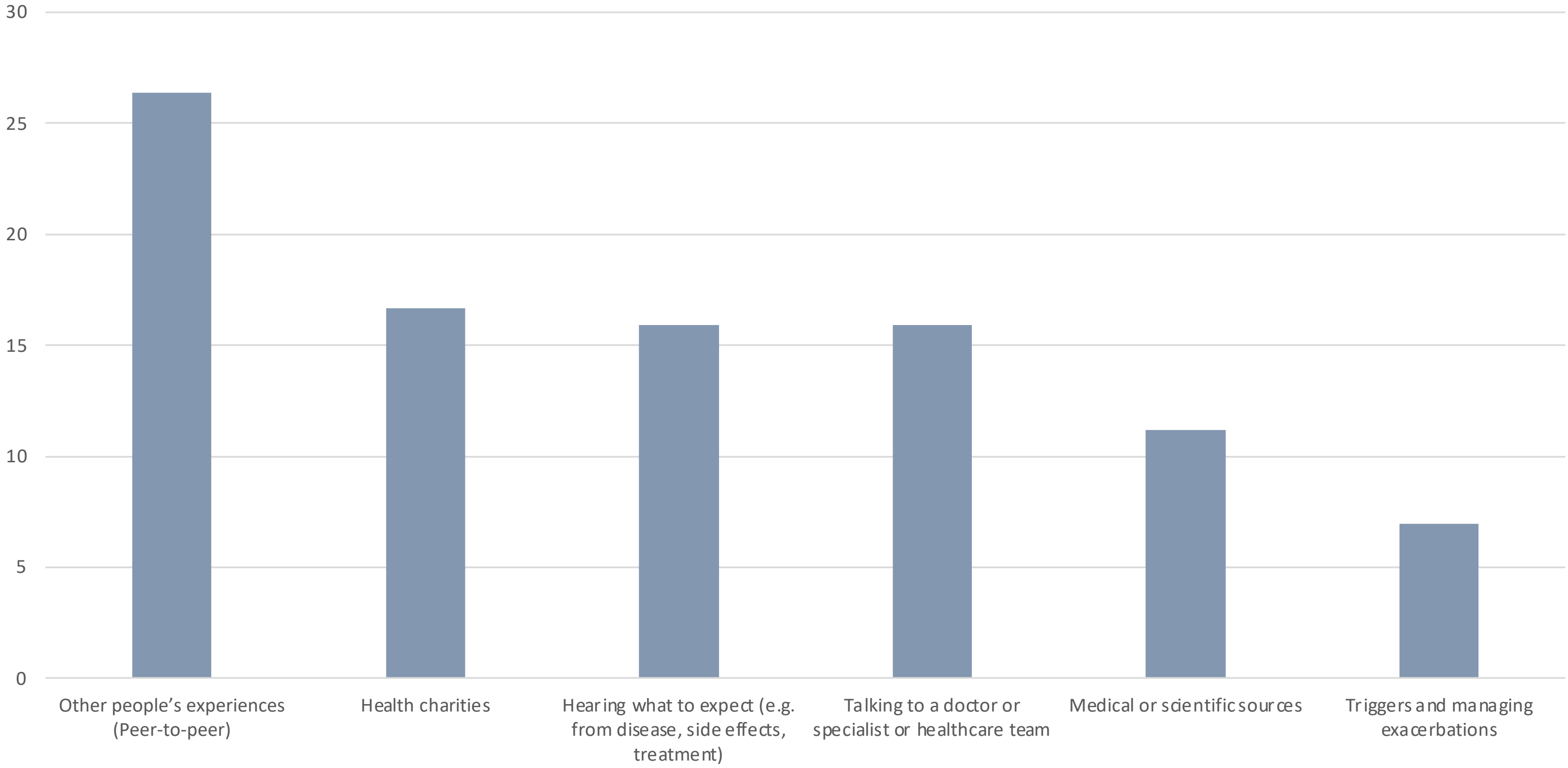
*Normal distribution use mean and SD as measure of central tendency

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.

Access to information– specific charity (% of all participants)



Just online it, it helped greatly. It it. It's clear it's out there. I knew I had it before I actually had a diagnosis, and I knew that for quite a few years and that. That to me was the frustrating part of everything was aligned, but the the the doctors didn't understand or know. The dermatologist I was originally seeing was the exact same, had didn't have knowledge of it. So it was frustrating. And then when you finally got the diagnosis, it was like, yes, someone that can actually understand and knows about what I've been. Yes, knowing I had. Participant 002_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

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

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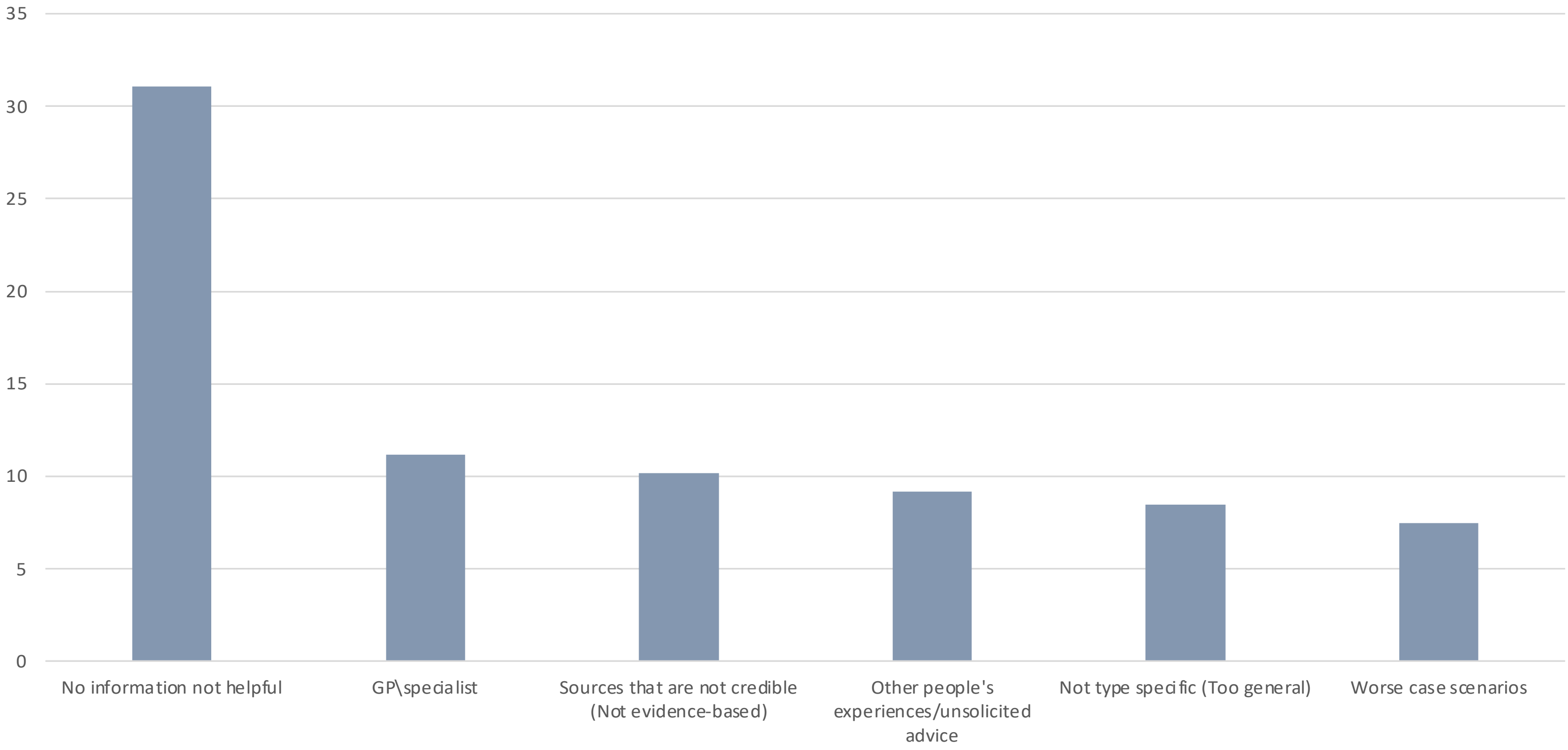
Information that has been helpful (% of all participants)



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

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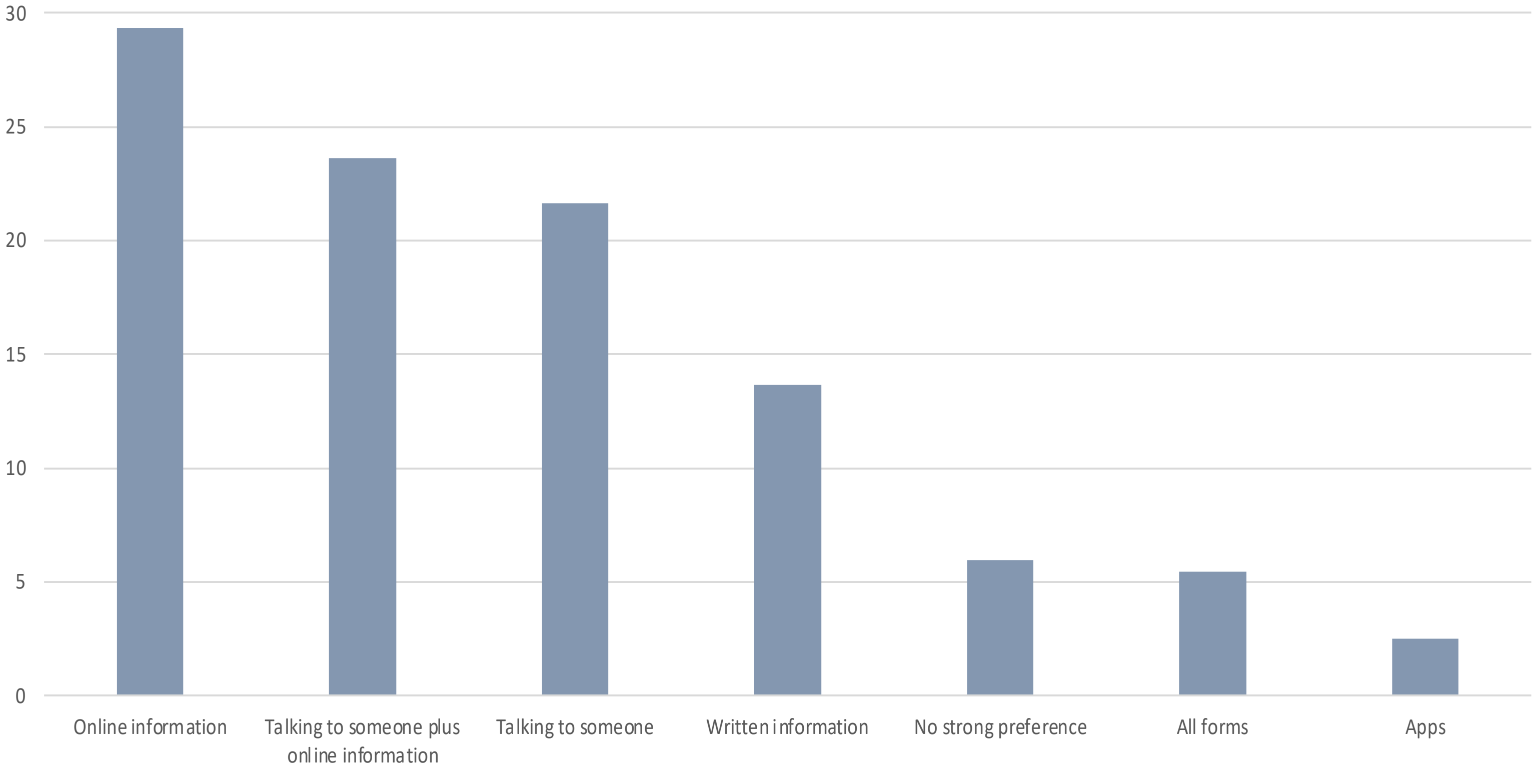
Information that has not been helpful (% of all participants)



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 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

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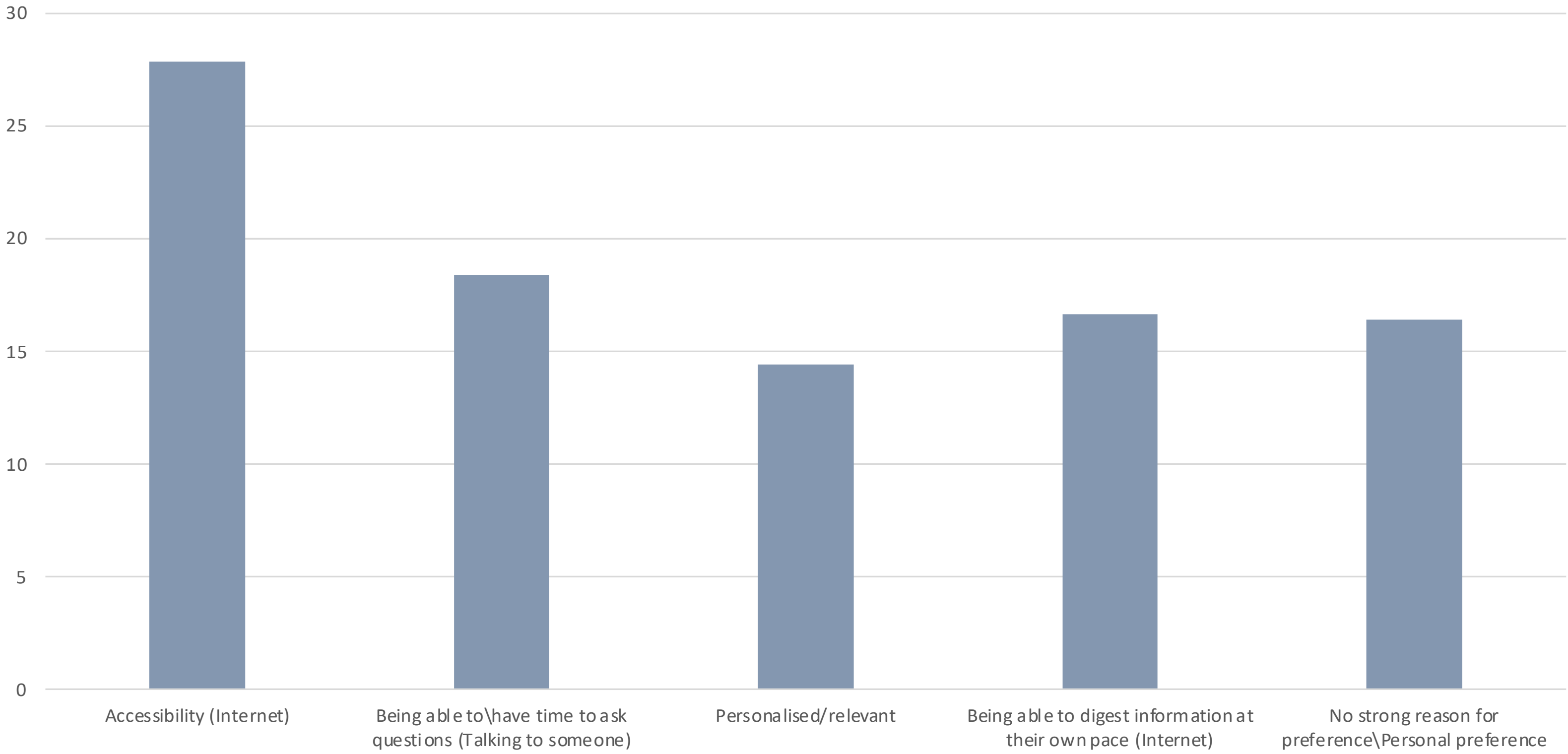
Information preferences (% of all participants)



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

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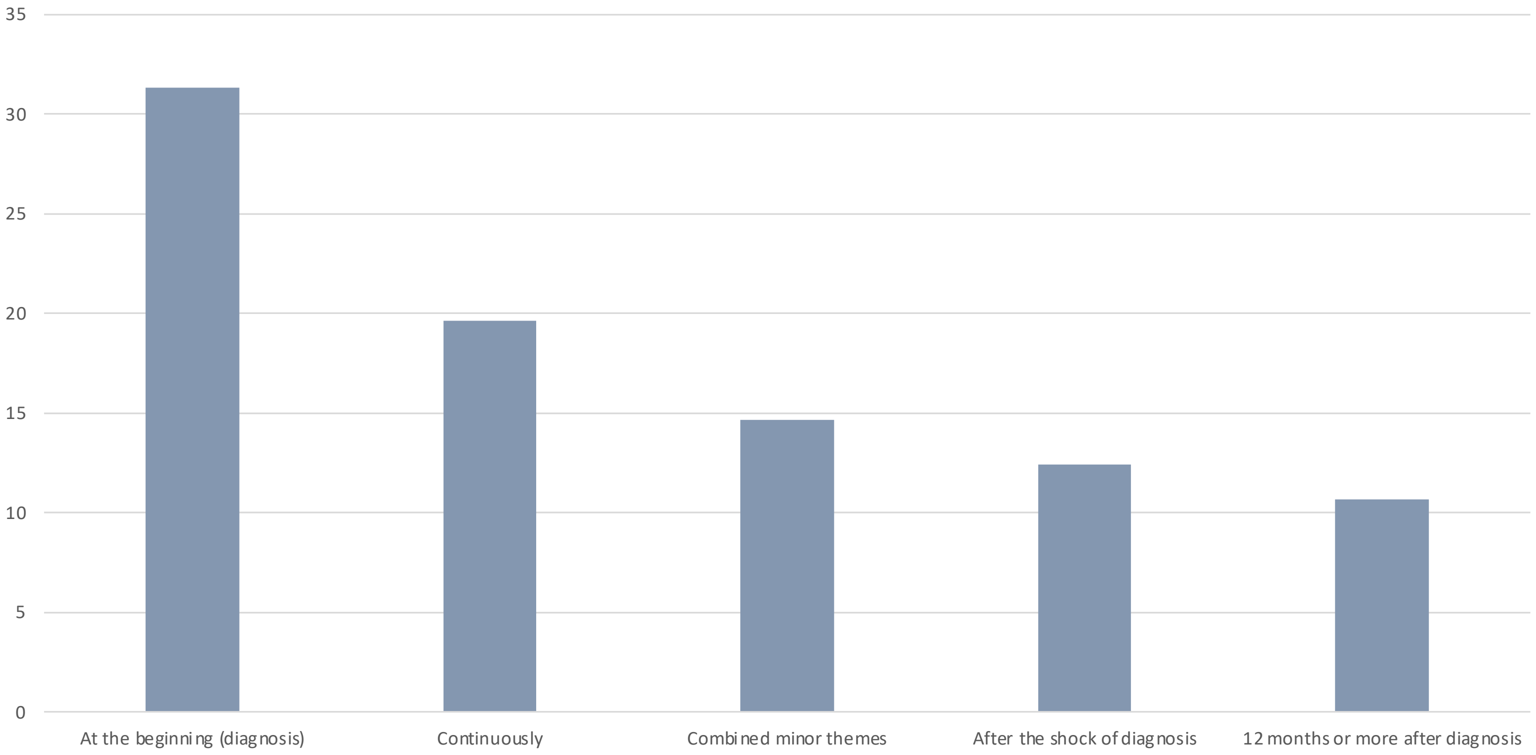
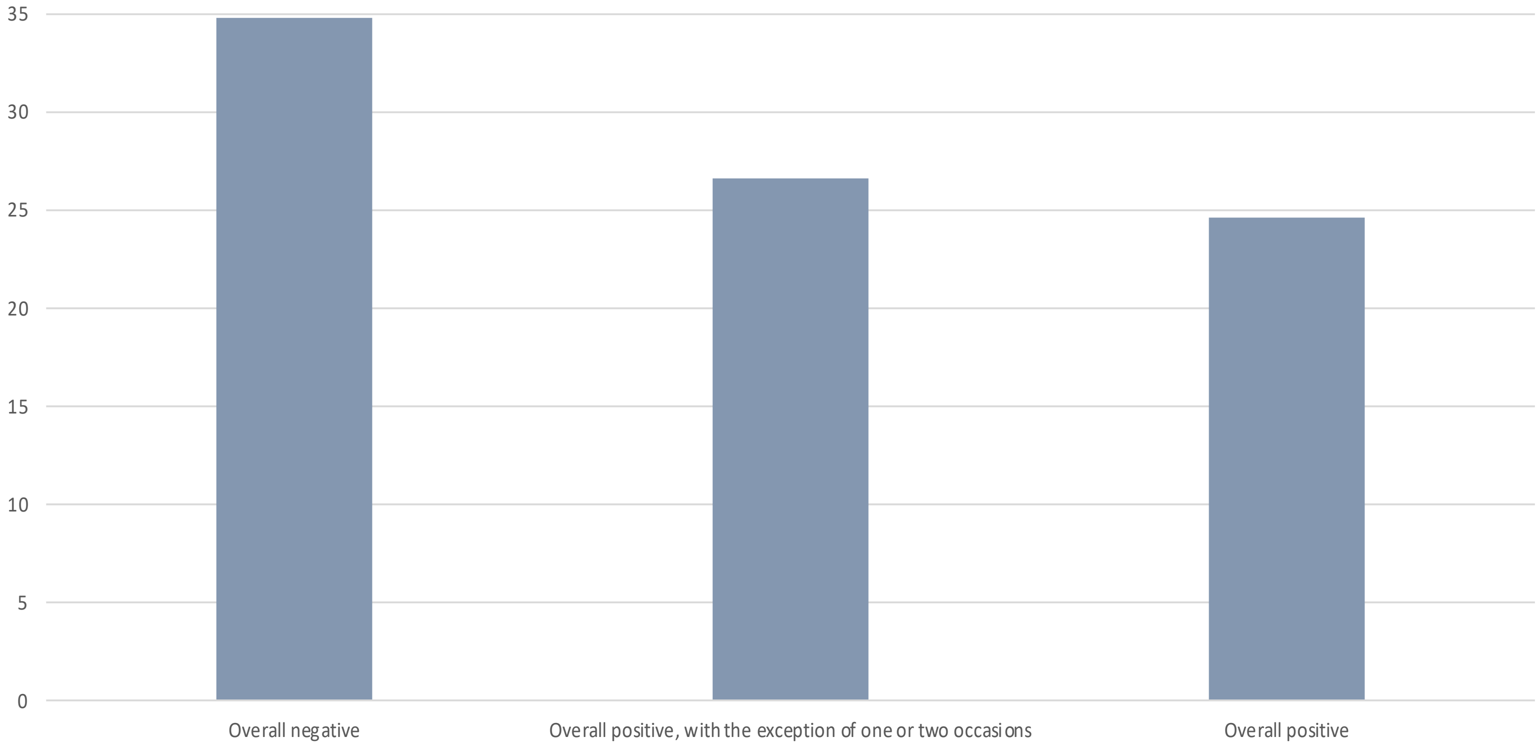
Information preference rationale (% of all participants)



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

Timing of information (% of all participants)



Crap not good. Like costed hundreds of dollars to see the dermatologist and I think he spent about 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.
Participant 006_2023AUDSK

Healthcare professional communication (% of all participants)

Section 7

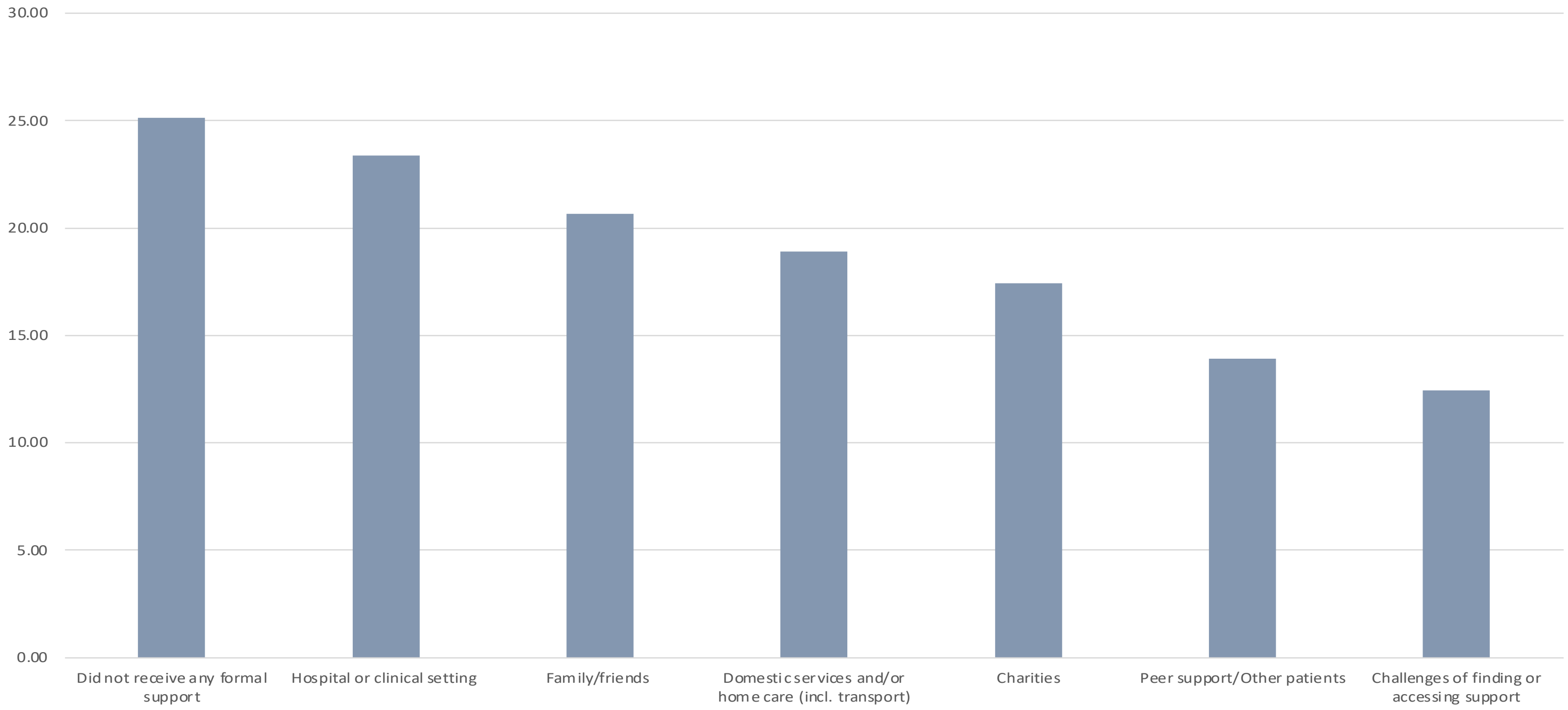
A Care Coordination questionnaire tool is used and reported in this section to demonstrate how people have been able to navigate the health system. There is also information about the care and support people have received, including from the charity sector.

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

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.

Care coordination



The most support, probably the only support I've really received has been from. My GP and 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

Care and support received (% of all participants)

Section 8

Quality of Life

In this section, information is available in relation to the impact that the condition or disease has on quality of life, regular activities to maintain mental health, regular activities to maintain physical health, impact on relationships, impact on family and relationships, cost considerations. The Fear of Progression tool is used and reported here to describe the level of anxiety that participants may have in relation to their condition.

Anxiety (measured by FOP)
Overall quality of life

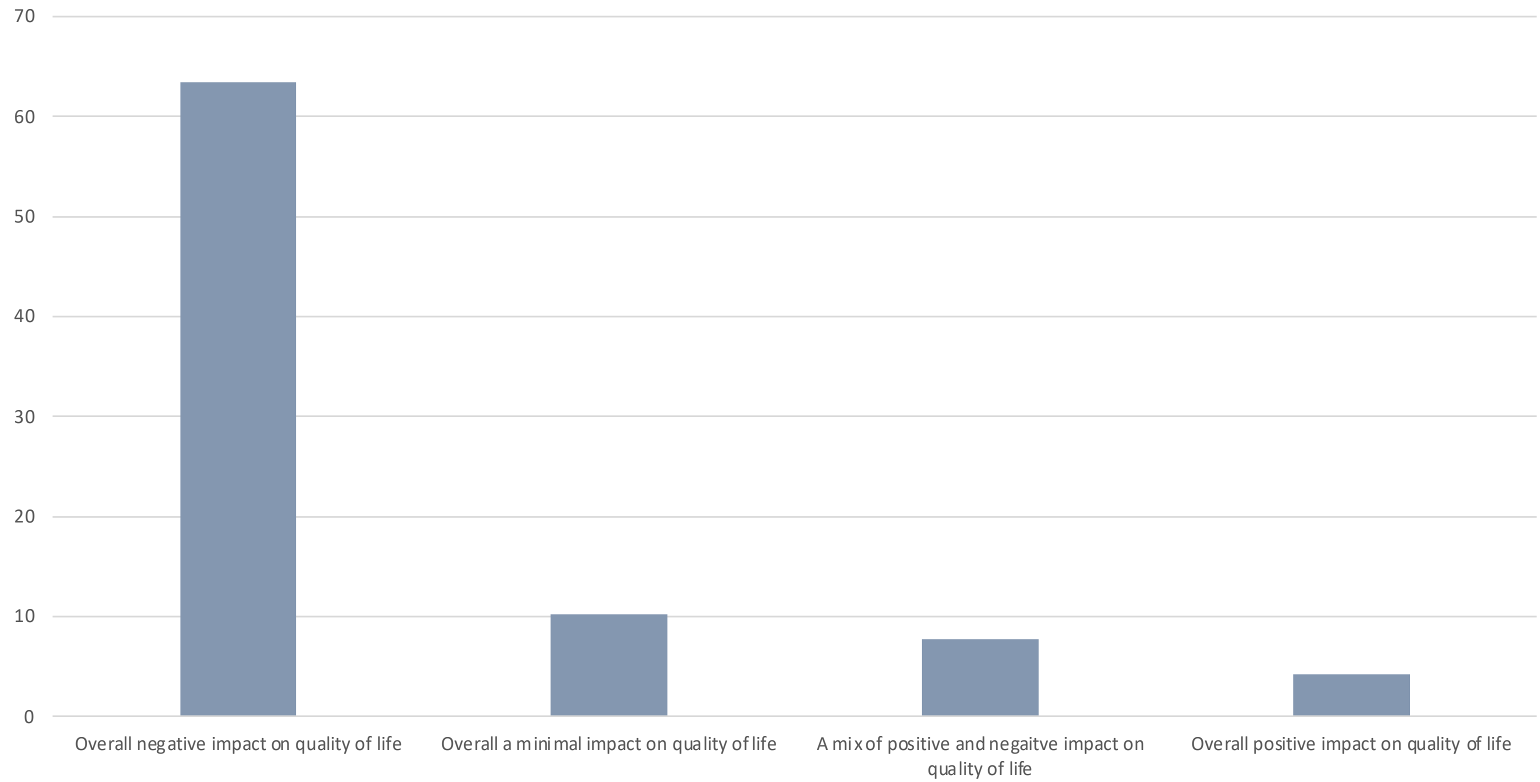
Fear of progression (n=370)	Mean	SD	Median	IQR	Possible range	Quintile
Total score*	37.09	10.40	37.00	14.75	12 to 60	3

*Normal distribution use mean and SD as measure of central tendency

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.

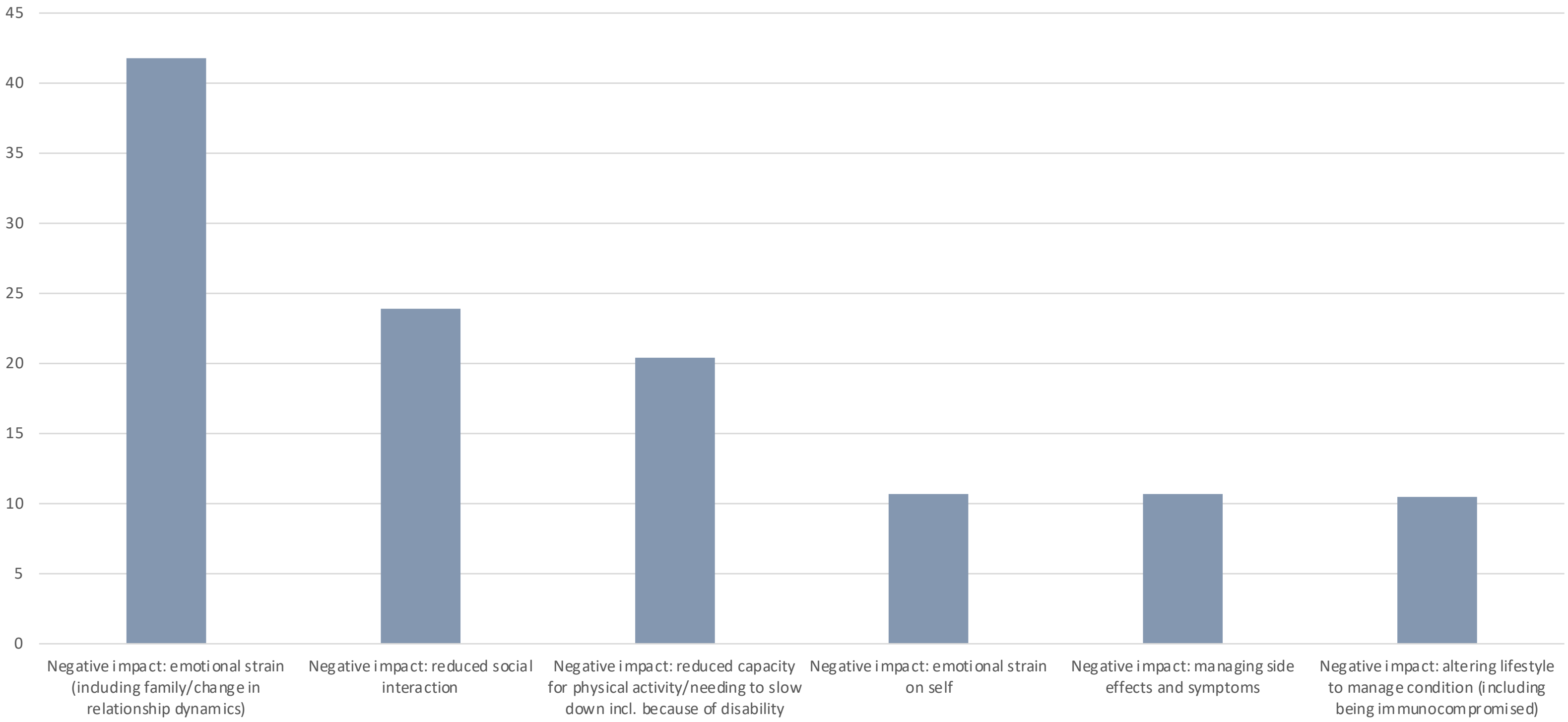
Impact of condition on quality of life	Number (n= 225)	Percent
1 Life is/was very distressing	46	20.44
2 Life is/was distressing	64	28.44
3 Life is/was a little distressing	56	24.89
4 Life is/was average	28	12.44
5 Life is/was good	21	9.33
6 Life is/was very good	8	3.56
7 Life is/was great	2	0.89

The average score was in the Life was a little distressing range (median=3.00, IQR=2.00).



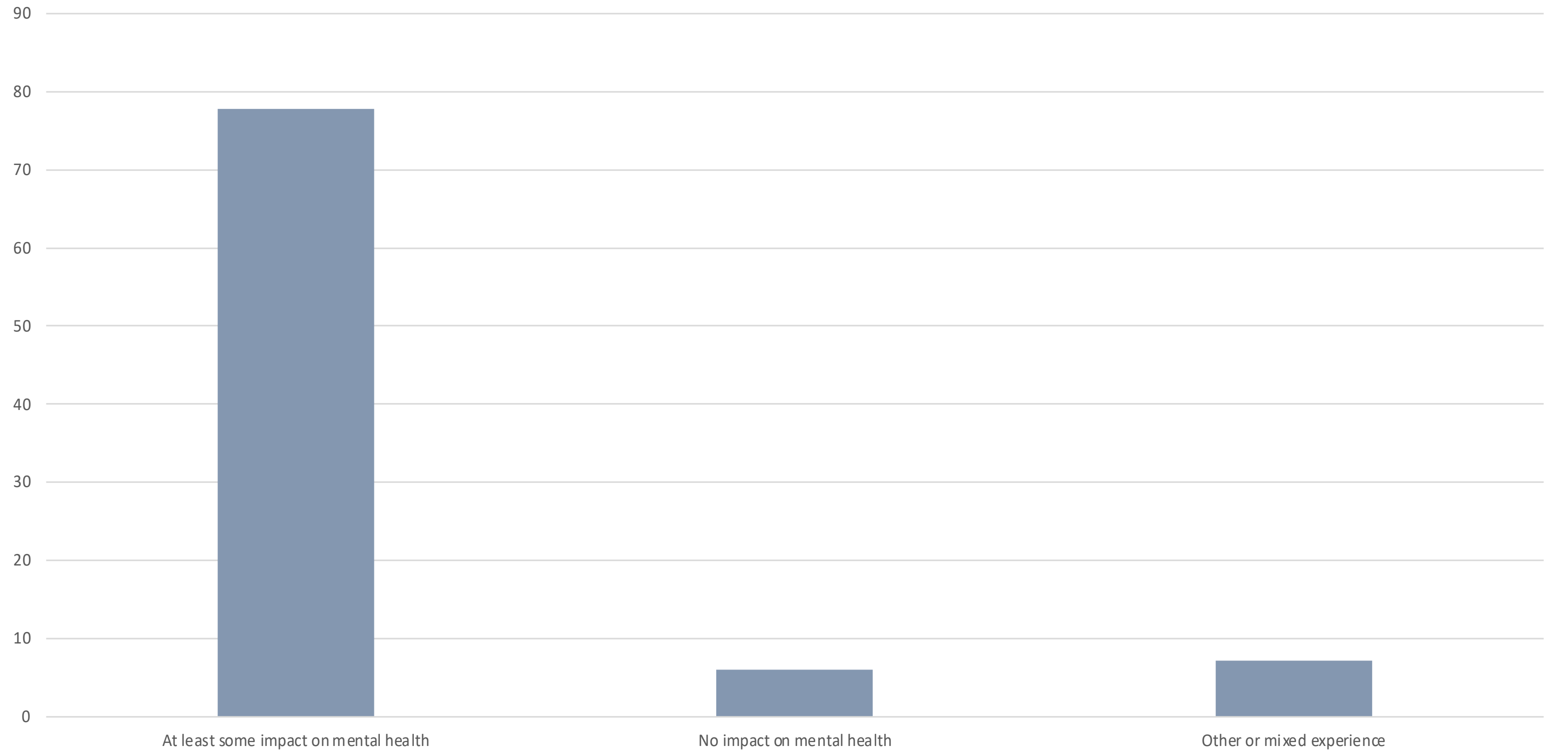
It has it, it's affected it. It's my partner has to be more aware. He has to be prepared to use his sick days to look after me rather than look after himself. It's affected relationships with family. It's left us financially struggling. It it on occasion it affects my relationship with my children because 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

Impact on quality of life (% of all participants)



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

Impact on quality of life (Main reasons) (% of all participants)



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

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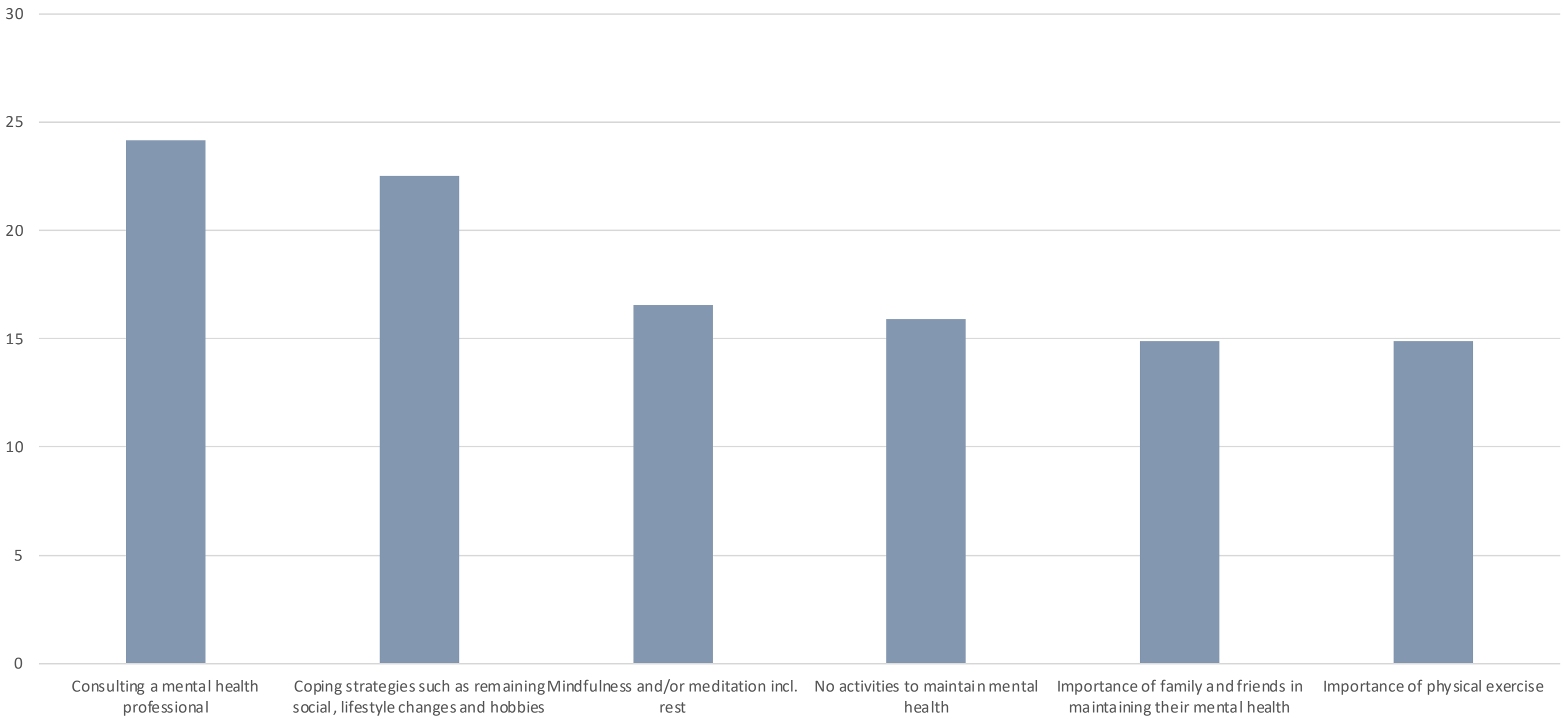
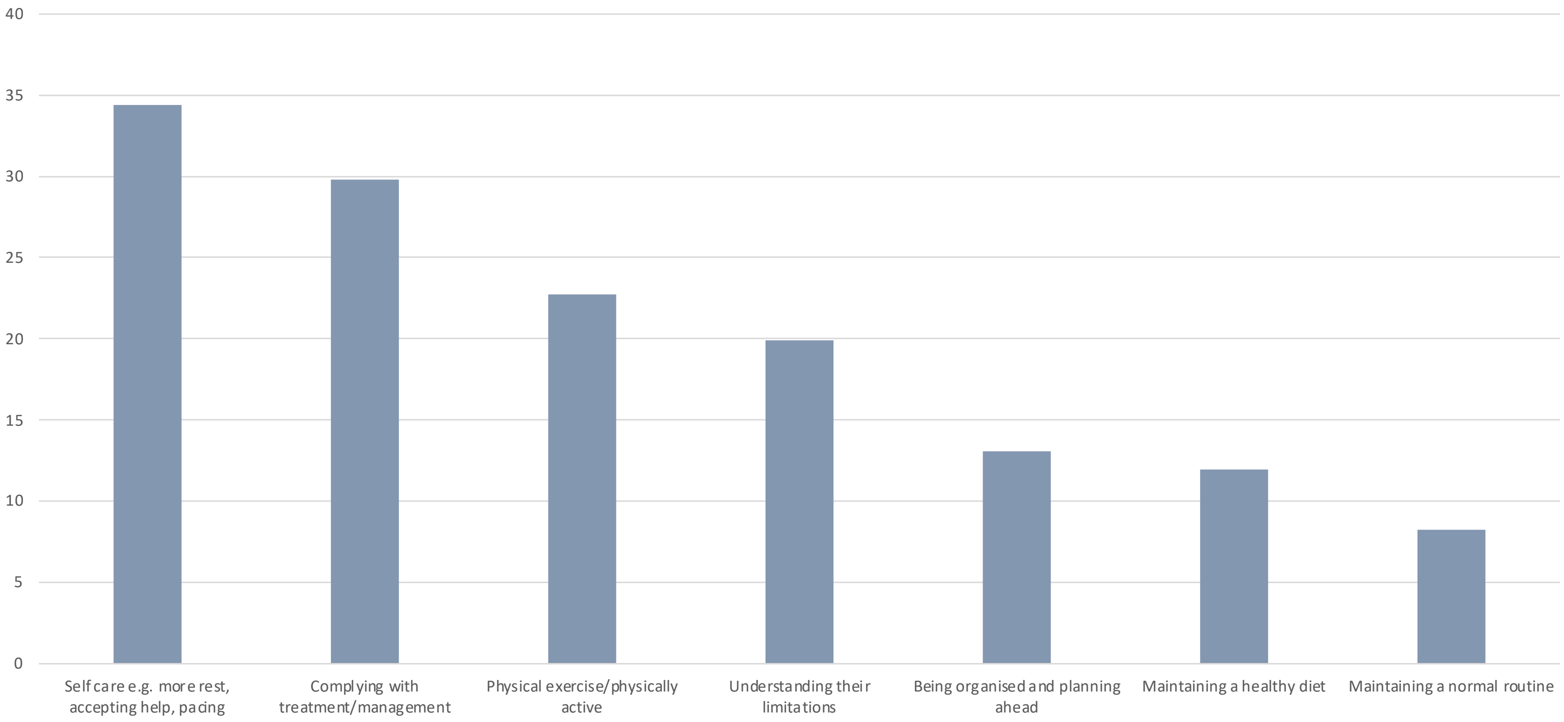
Impact on mental health (% of all participants)

Regular activities to maintain mental health (% of all participants)



Sometimes there's nothing I can do to control it. There's no 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



Yeah, so it did. It did affect me quite heavily at the start. I was a bit shocked by the diagnosis and it said it sent me downhill for a while. So I did go and talk to some grief counselors 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

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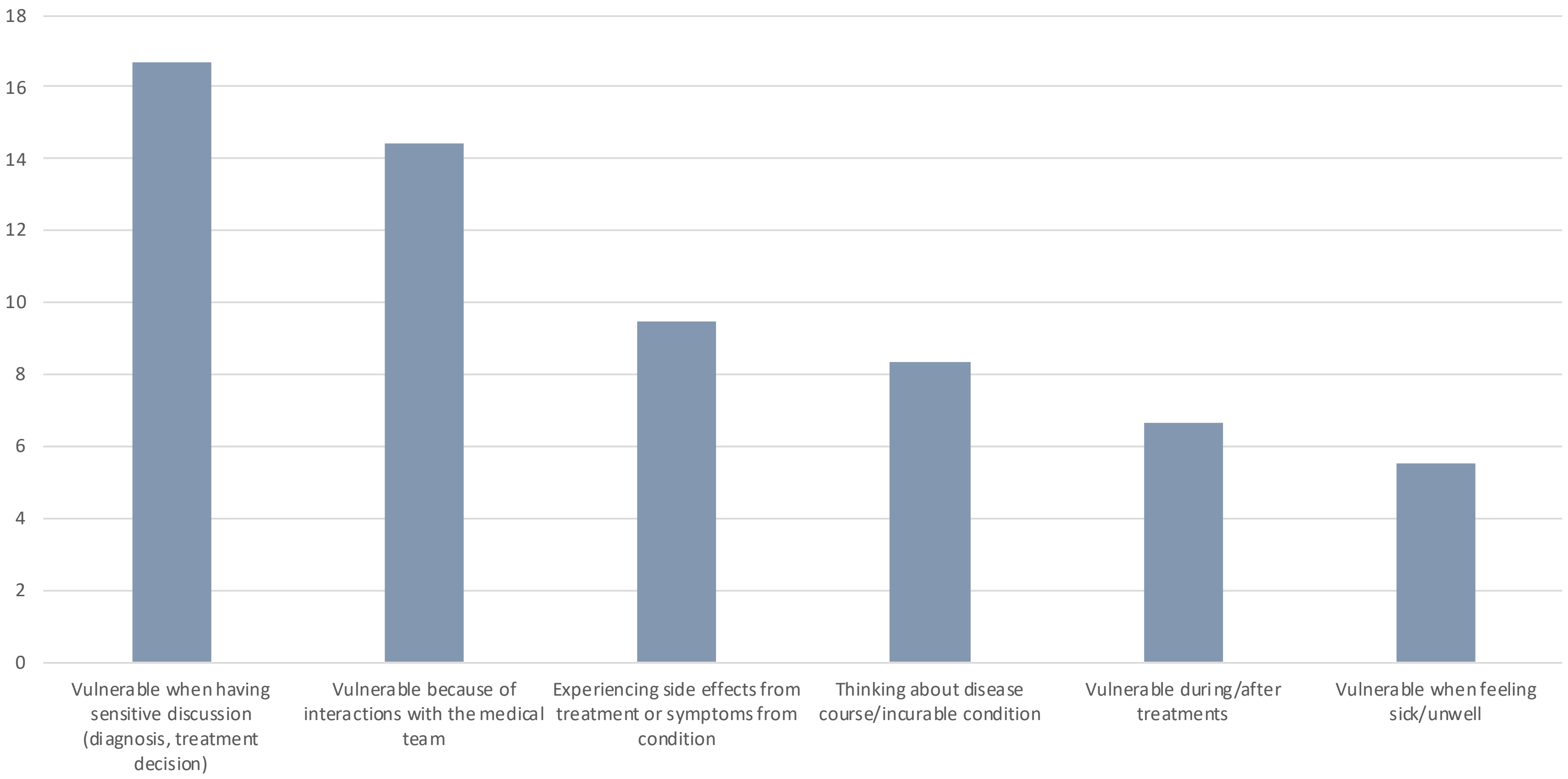
Regular activities to maintain general health (% of all participants)



Yeah, lots of times. I think for the 1st 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

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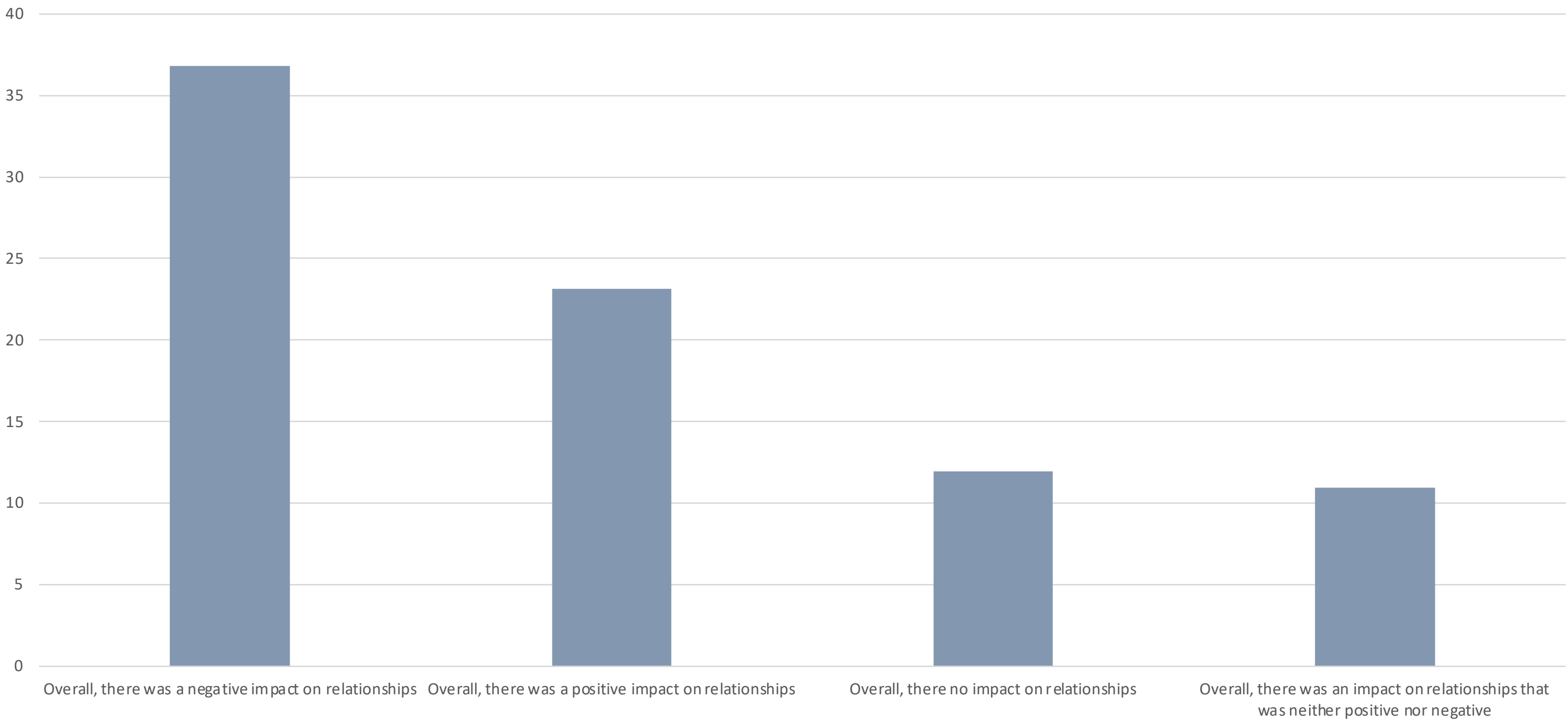
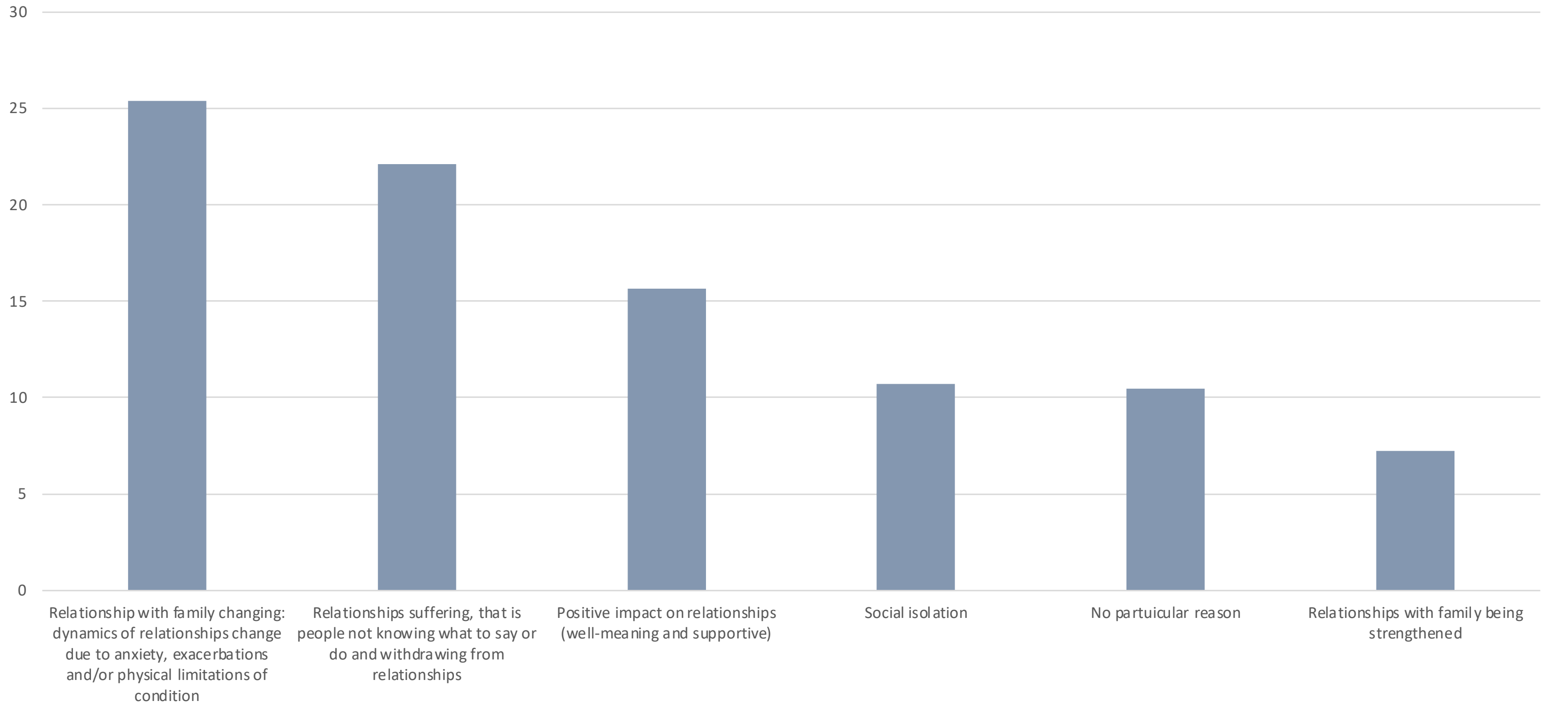
Vulnerability
% of all participants

Impact on relationships
% of all participants



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



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

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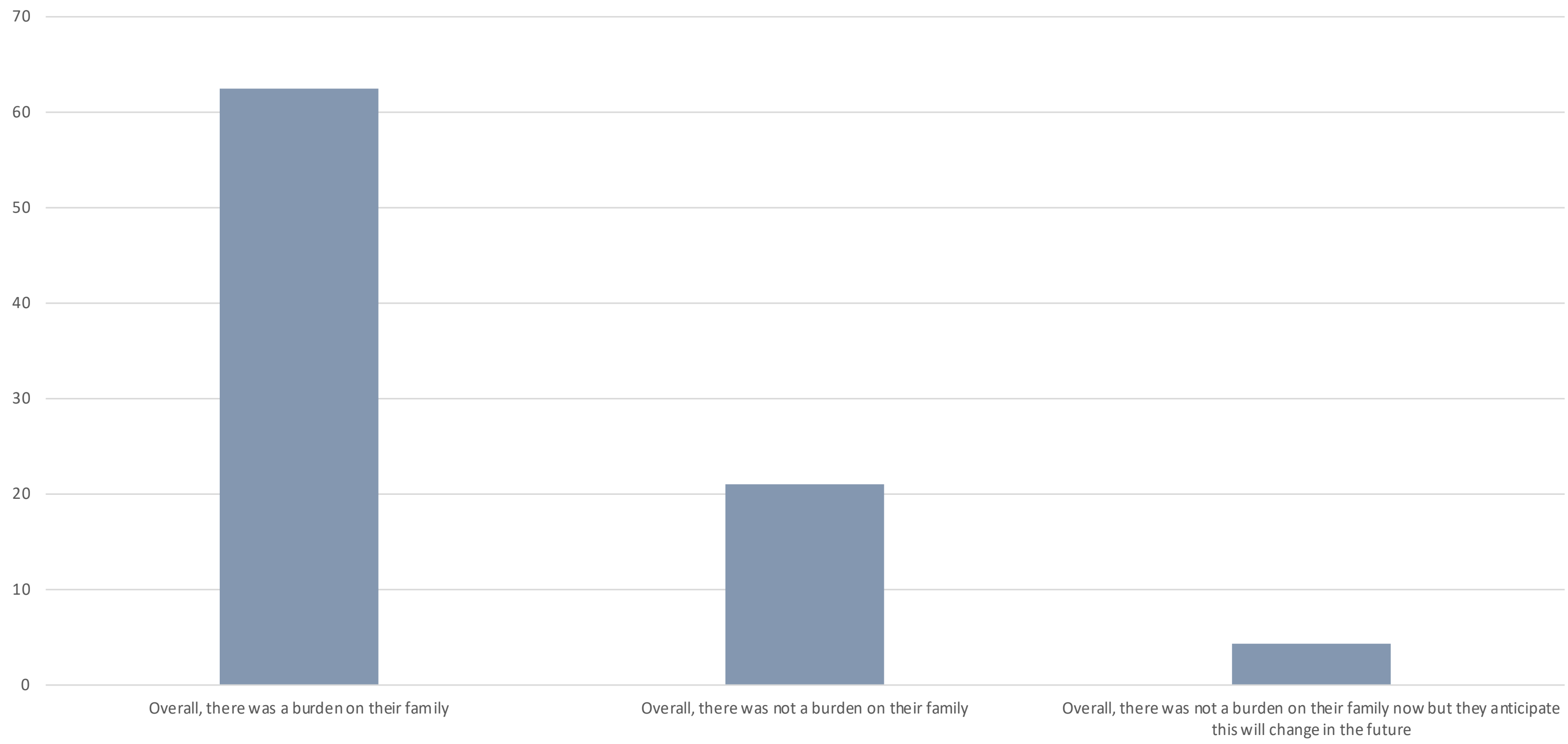
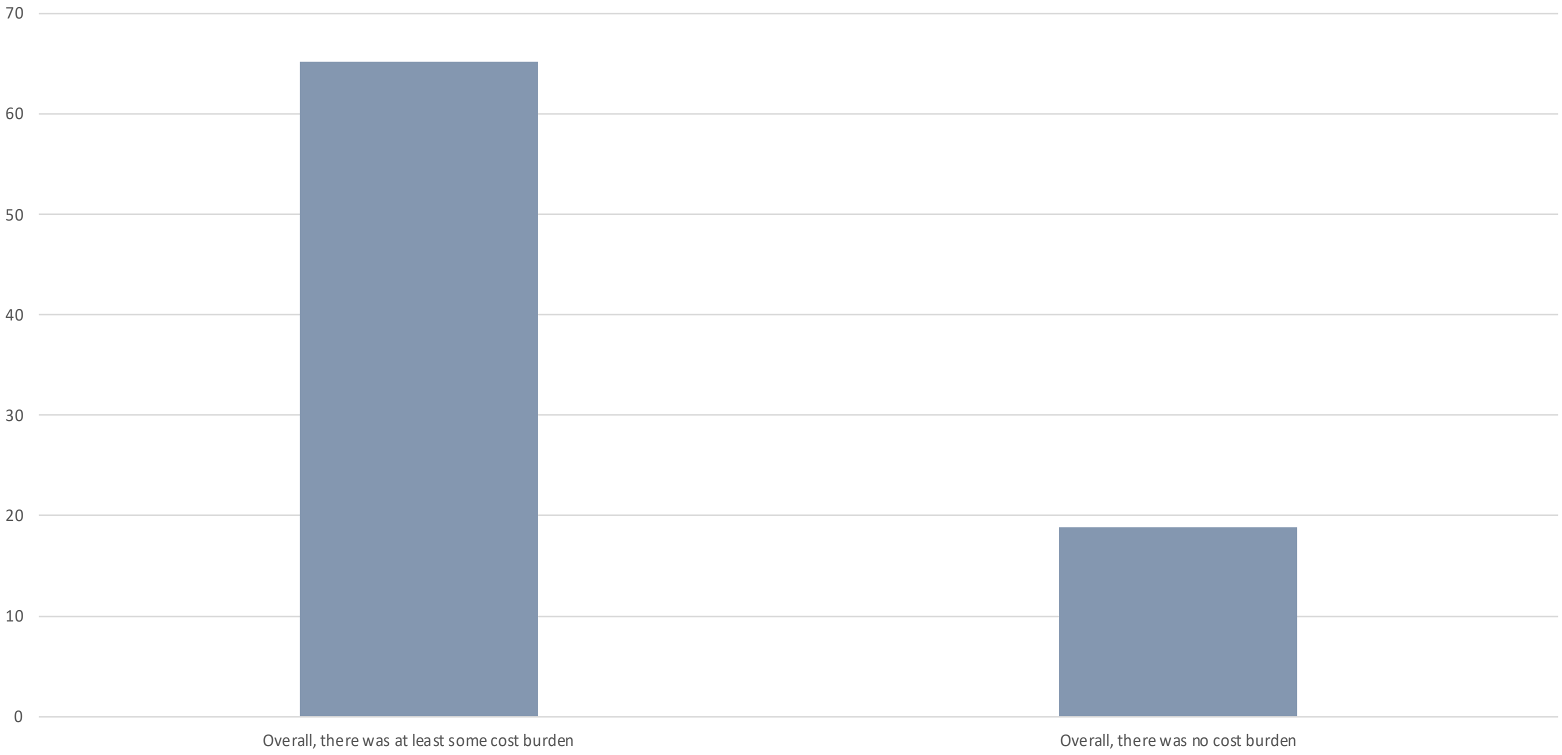
Impact on relationships - reasons (% of all participants)



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

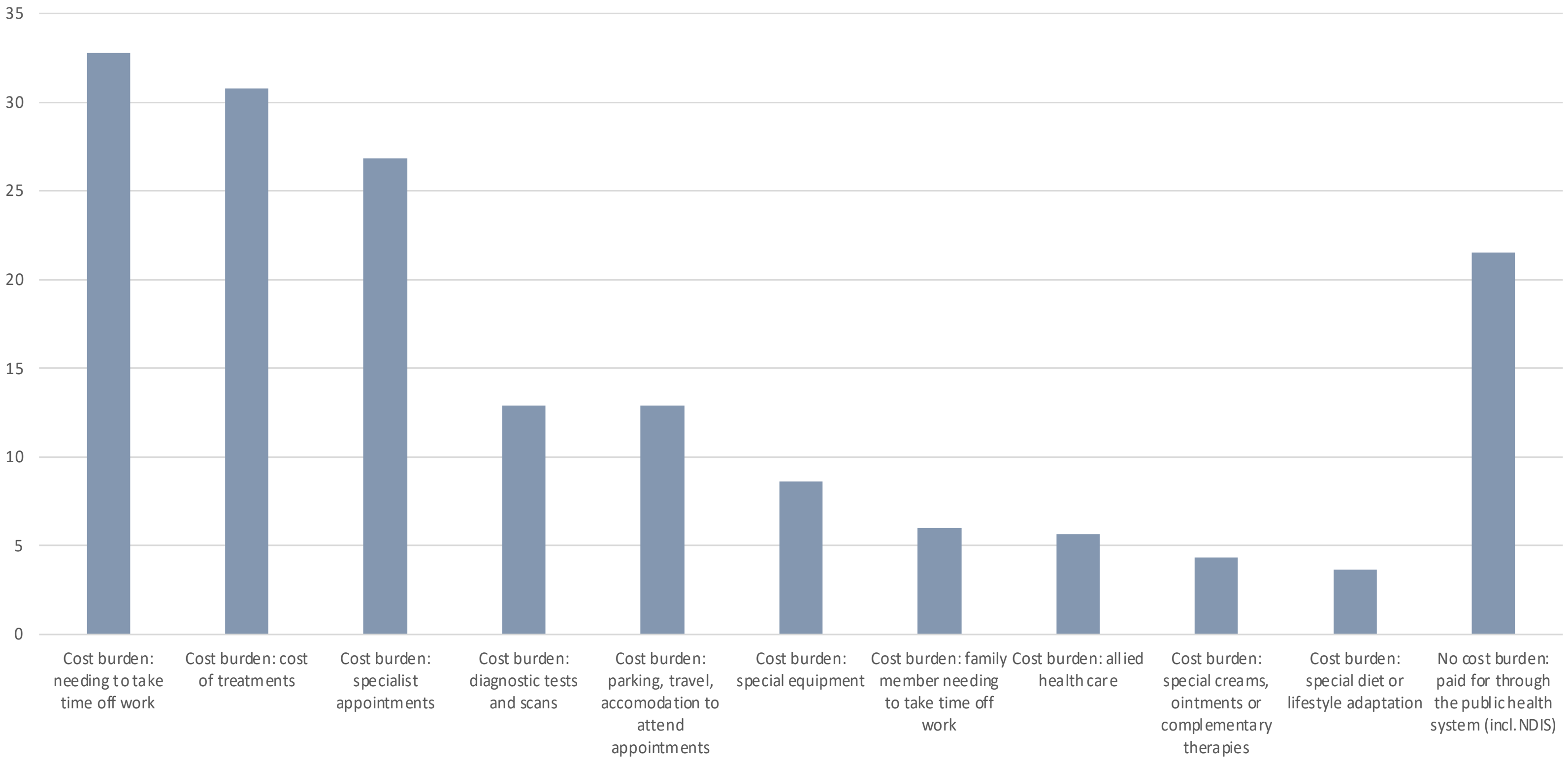
Burden on family (% of all participants)



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

Cost considerations (% of all participants)

Cost considerations - reasons (% of all participants)

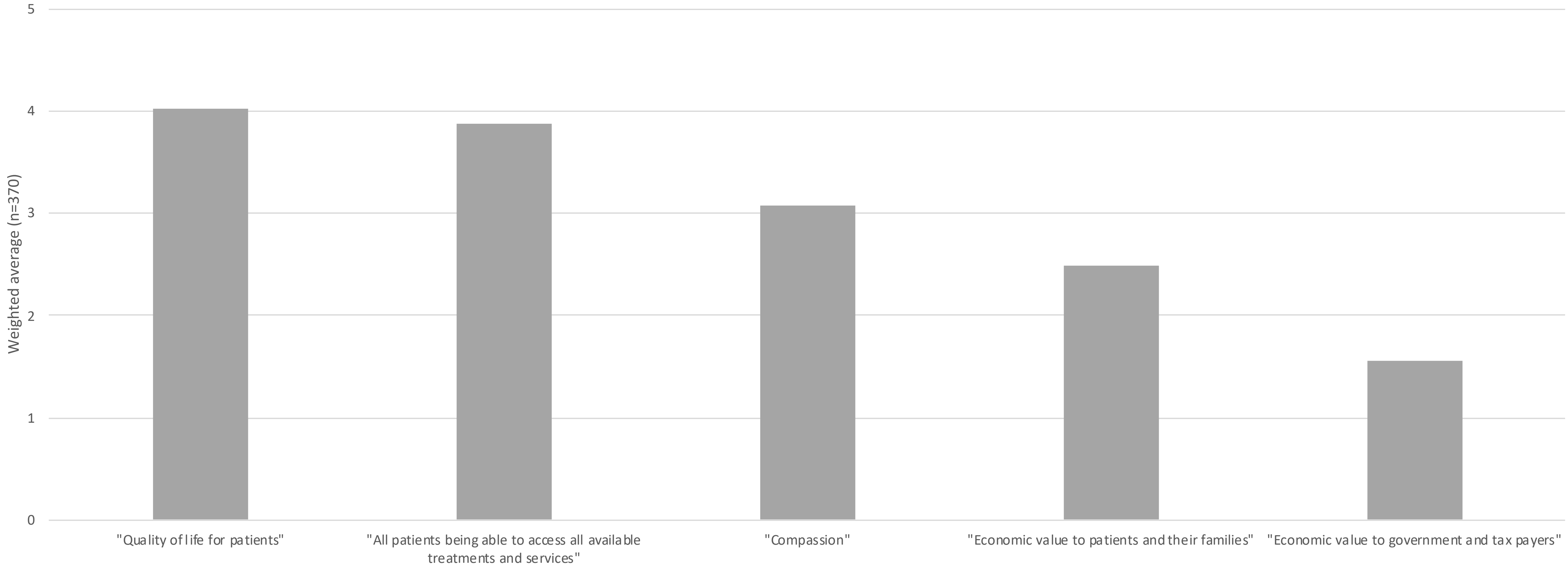


Probably the biggest one is the full-time off work, it's obviously very hard on the family. Also now, I find with fatigue and just chasing up medical appointments and things like that, that I only work part-time now. I work three days a week. Just financially that. I find that with scripts and seeking 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

Section 9

Expectations
&
Messages to Decision-Makers

By the time participants are asked questions about their expectations of the future, they have had the chance to talk about everything that they have been through. It means that they have been able to reflect on what worked for them and what was potentially not so great. In this section there is information about what participants would like to see from future treatments, information, care and communication. We present the cohorts message to decision-makers about their condition and the values that are important to them when they make decisions about treatment and care. In this section there is also information about what people have been grateful for.



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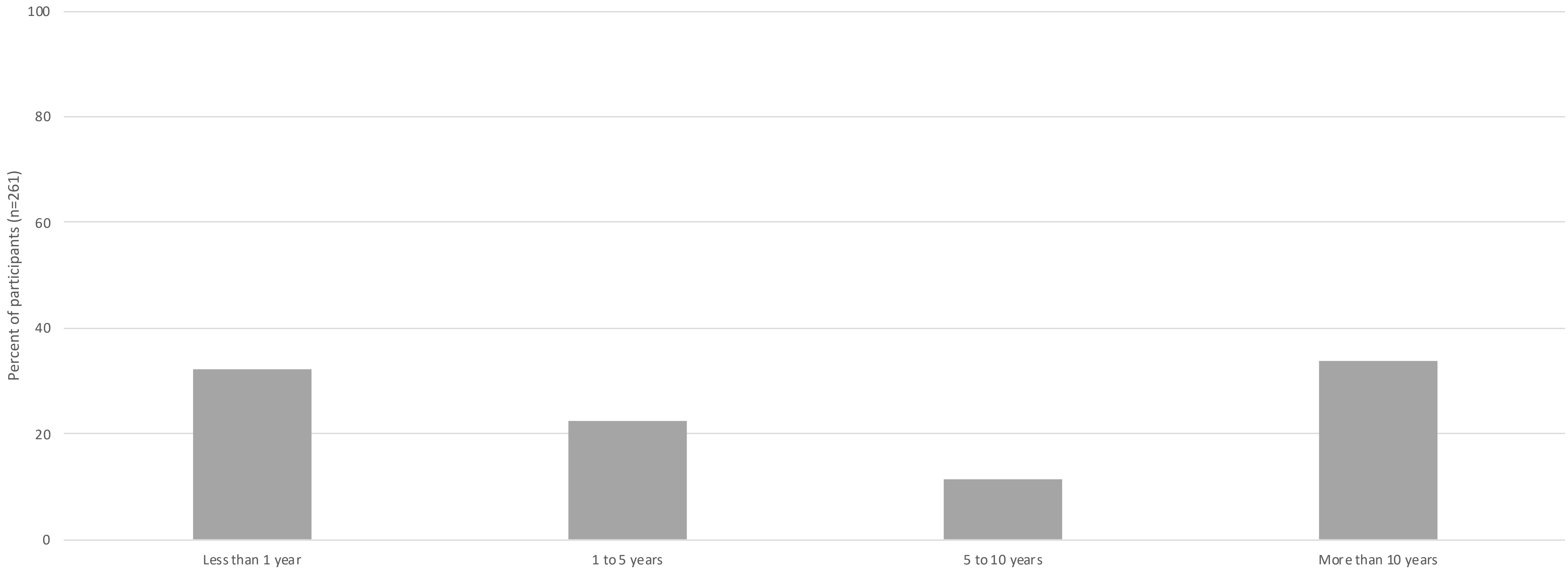
Values for decision makers



Values for decision to consider

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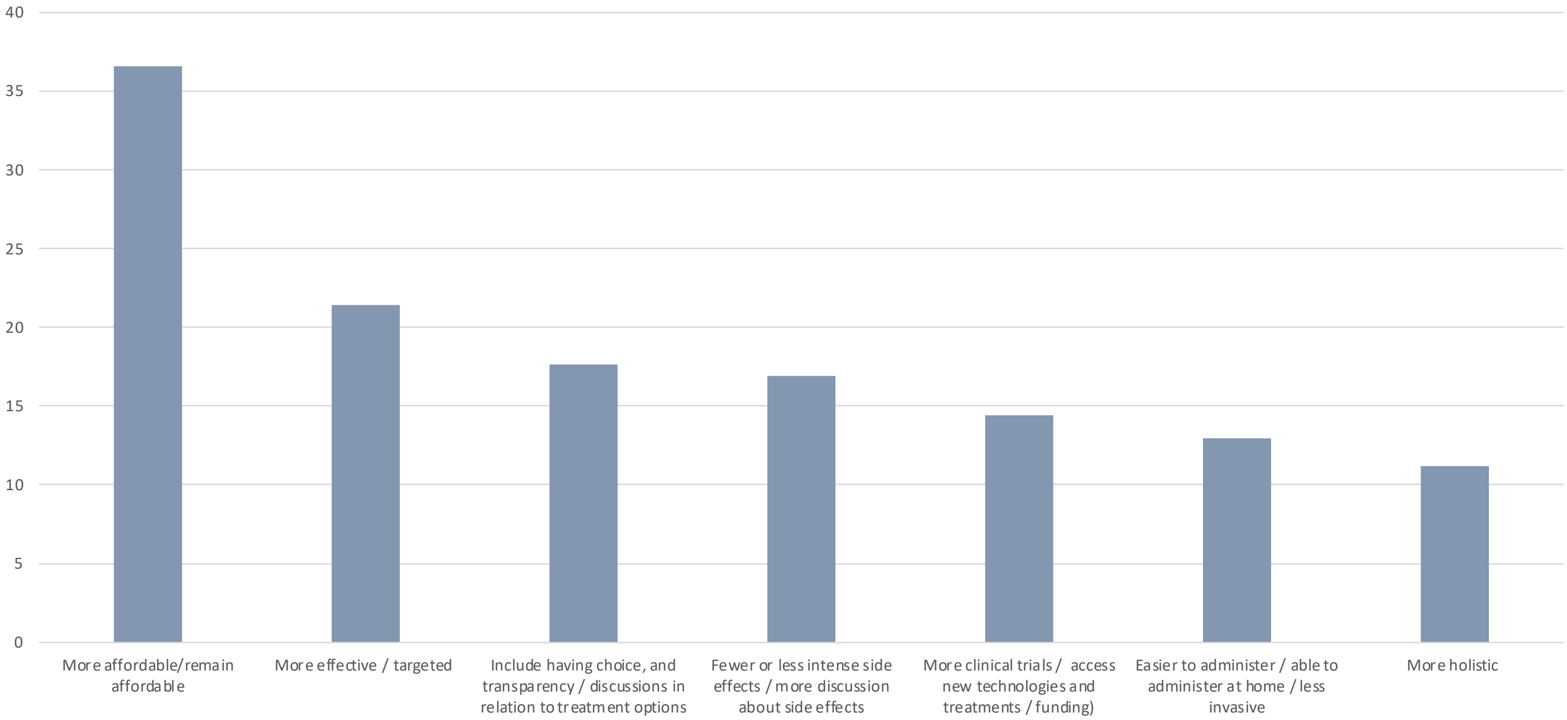

I'd like to see the cost reduce cuz yeah I don't have a healthcare card so the scripts are over 6 times the cost they would be if I did have a healthcare card. So I would like to see I guess if they could be cheaper for people that don't have access to a healthcare card, but doubt that and that's it. 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

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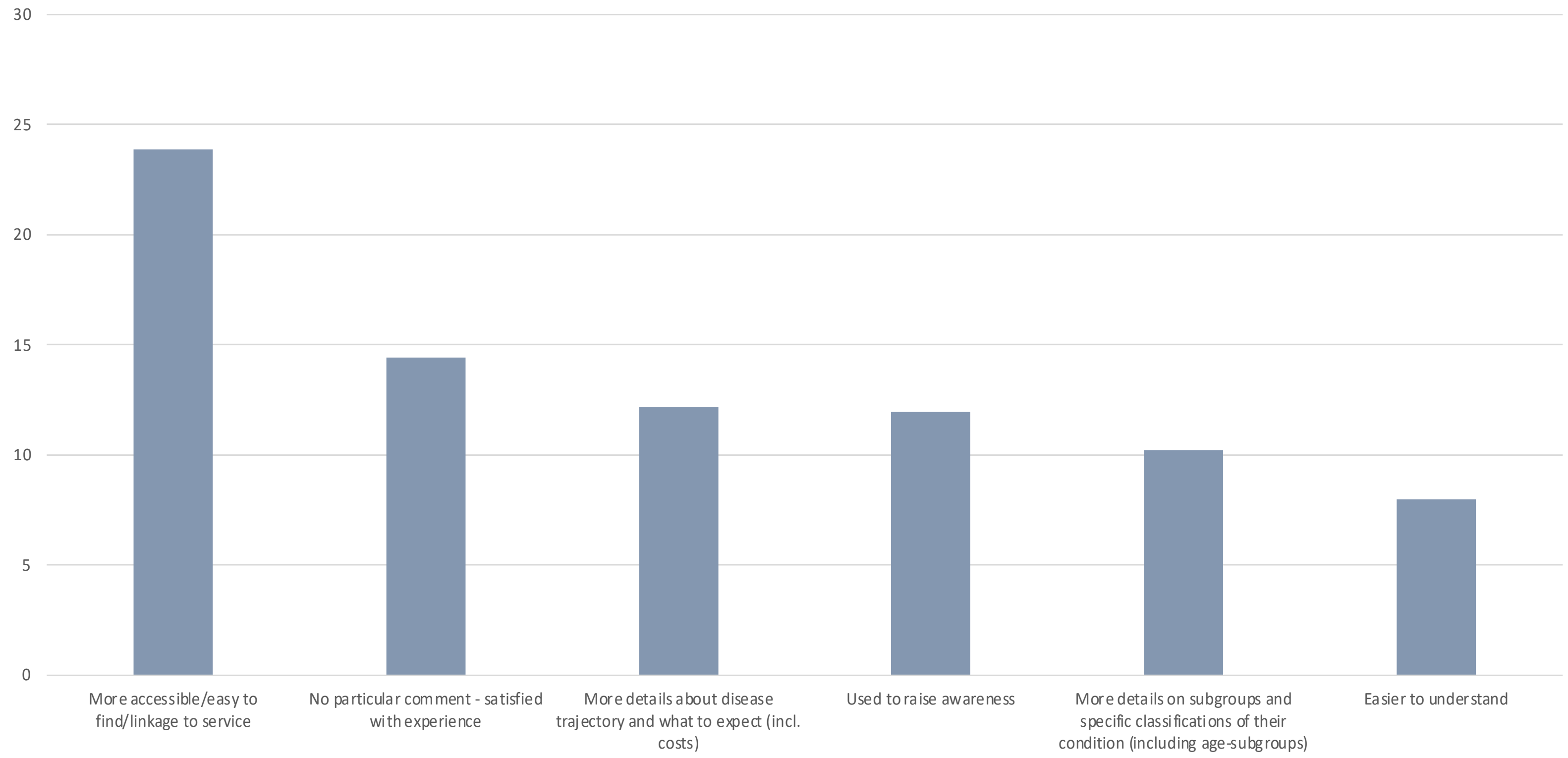
Expectations of future treatments (% of all participants)



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

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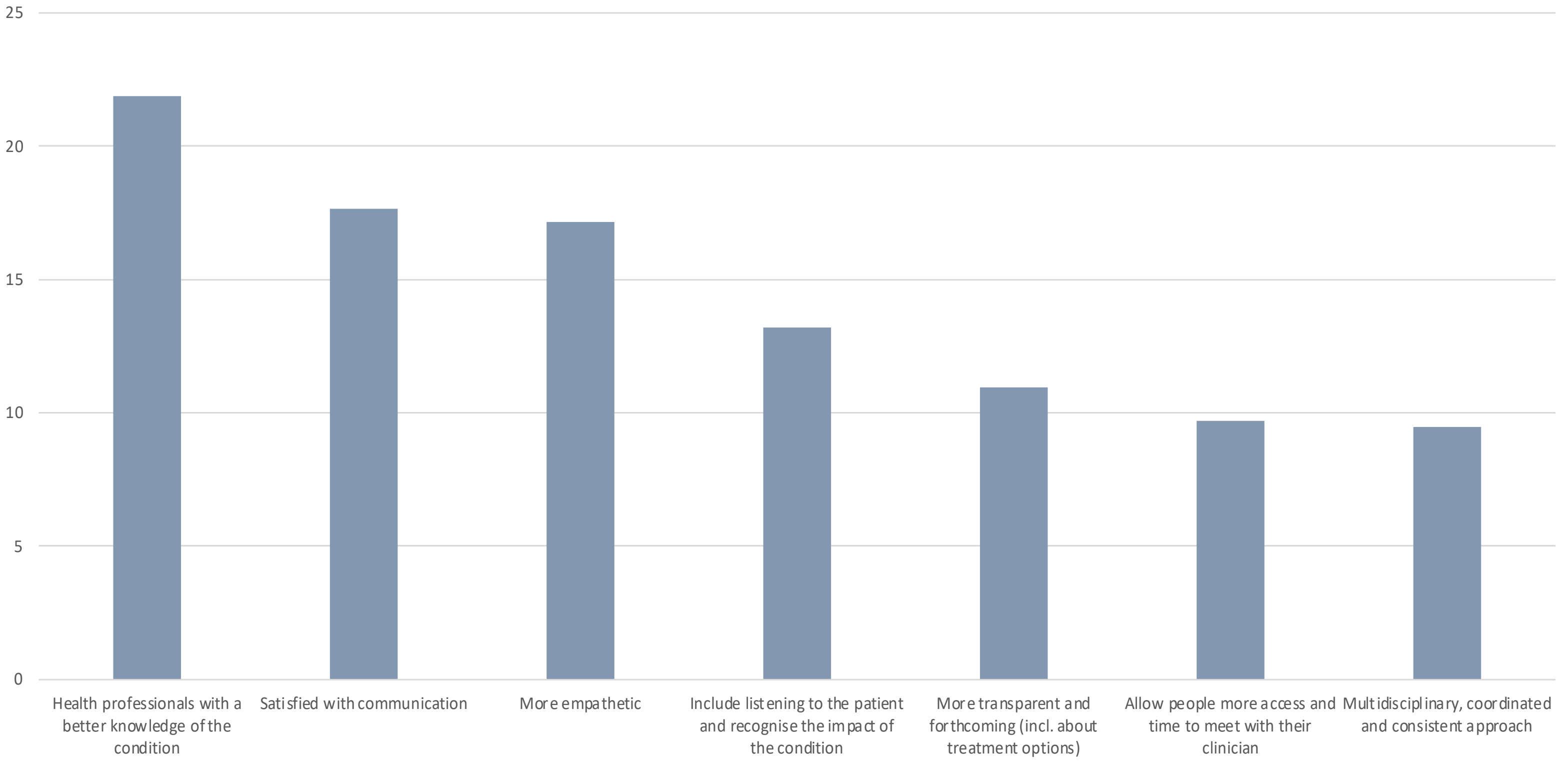
Expectations of future information



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

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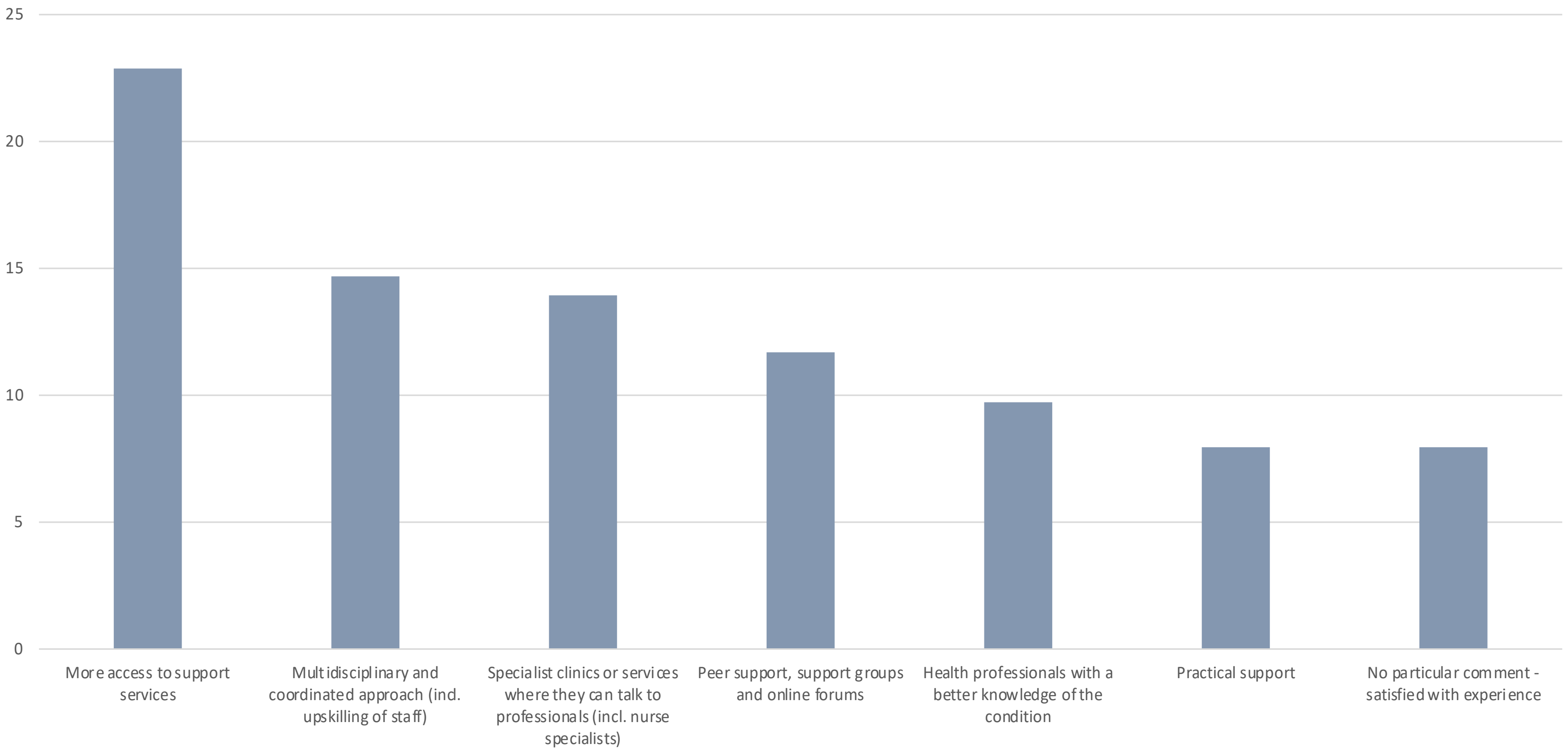
Expectations of future communication



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

Expectations of future care and support

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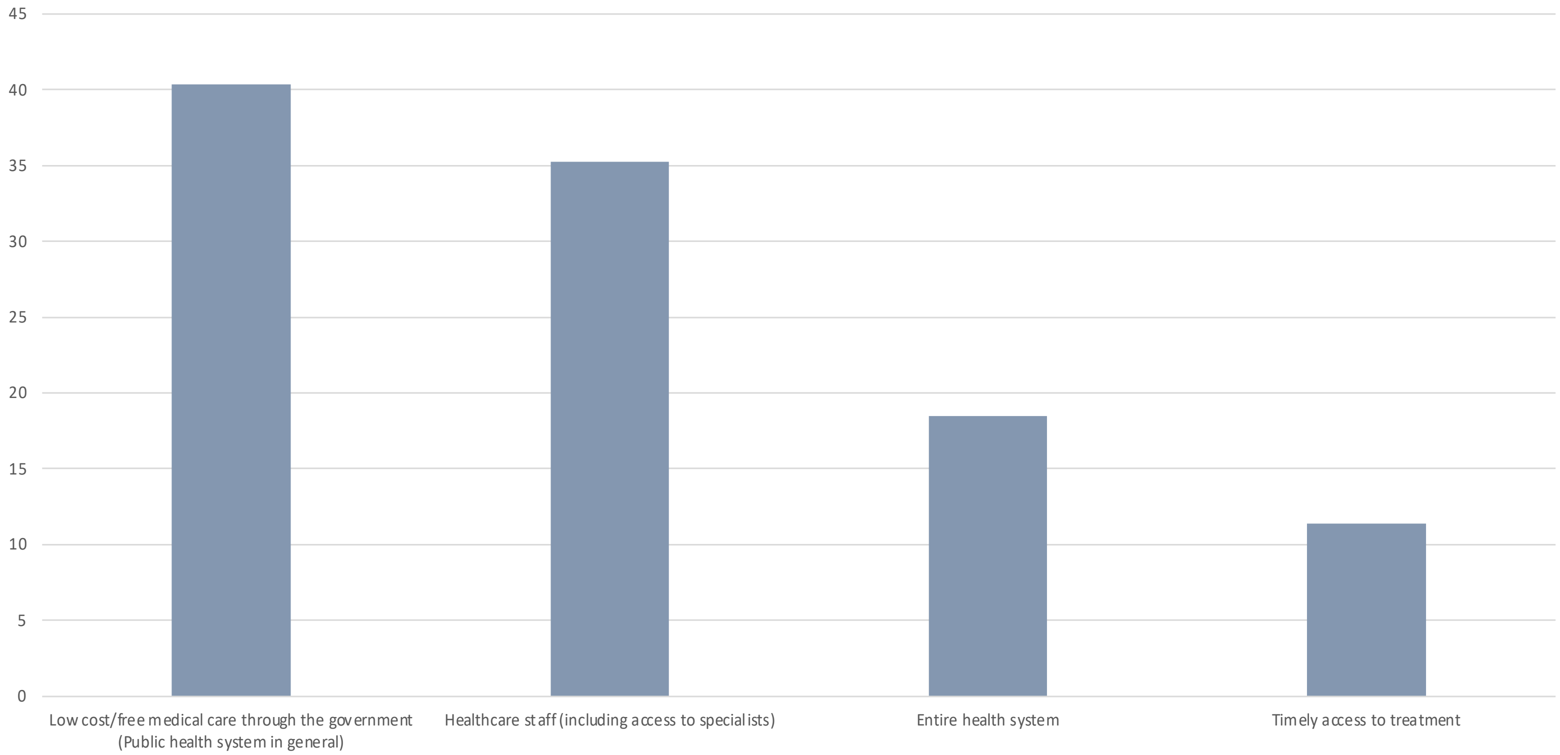

Yes, the nurses. The nurses are amazing. From the transfusion nurses, to the nurses that I do my six minute walk test with, to lung function nurses. All those little people, they do all the hard work. I take my hat off to them like, they are the best people because they've seen me progress to worse and 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 hat off to them any day. Participant 023_2023AUDIS

What people are grateful for

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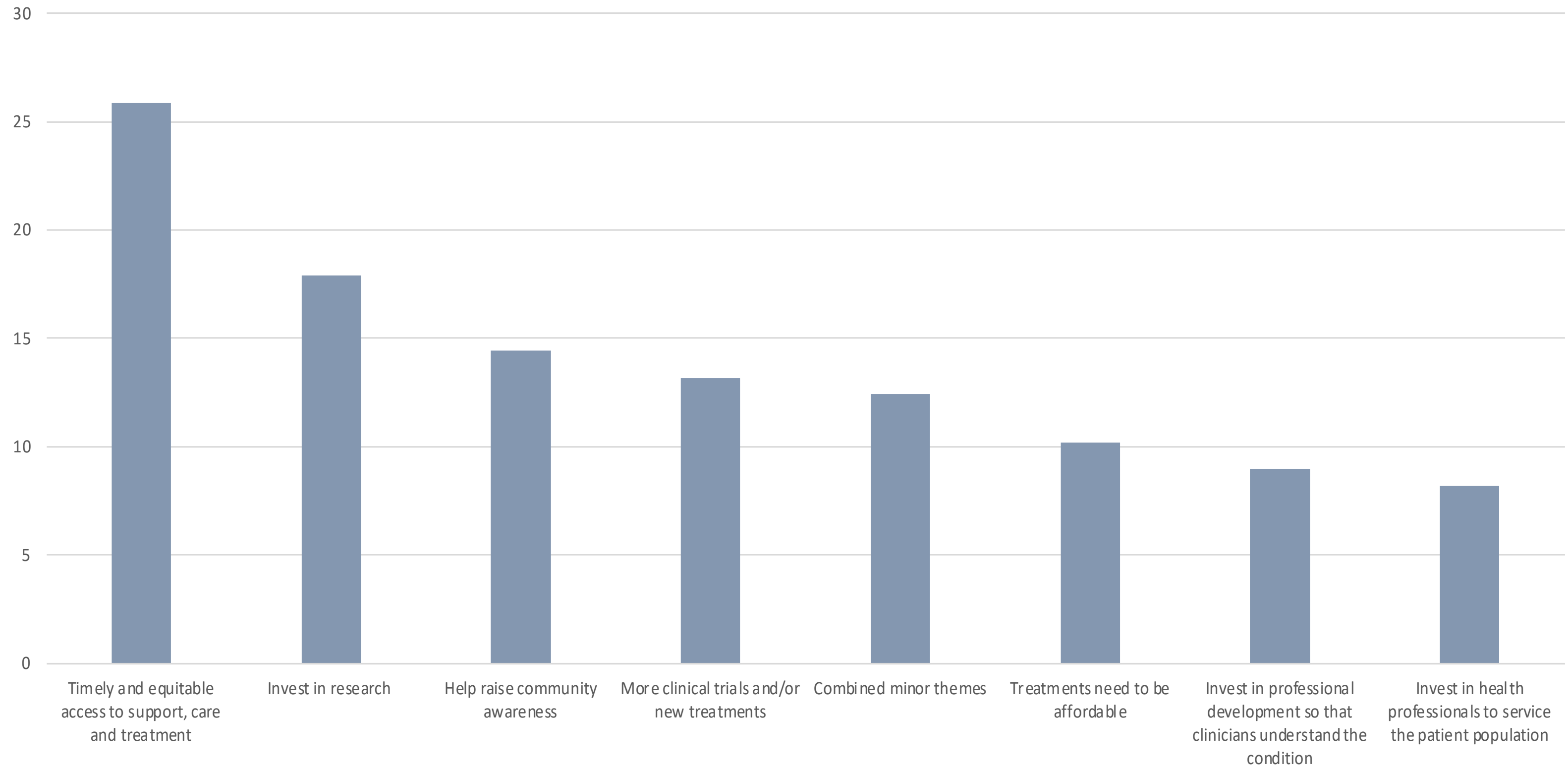
Message to decision-makers (% of all participants)

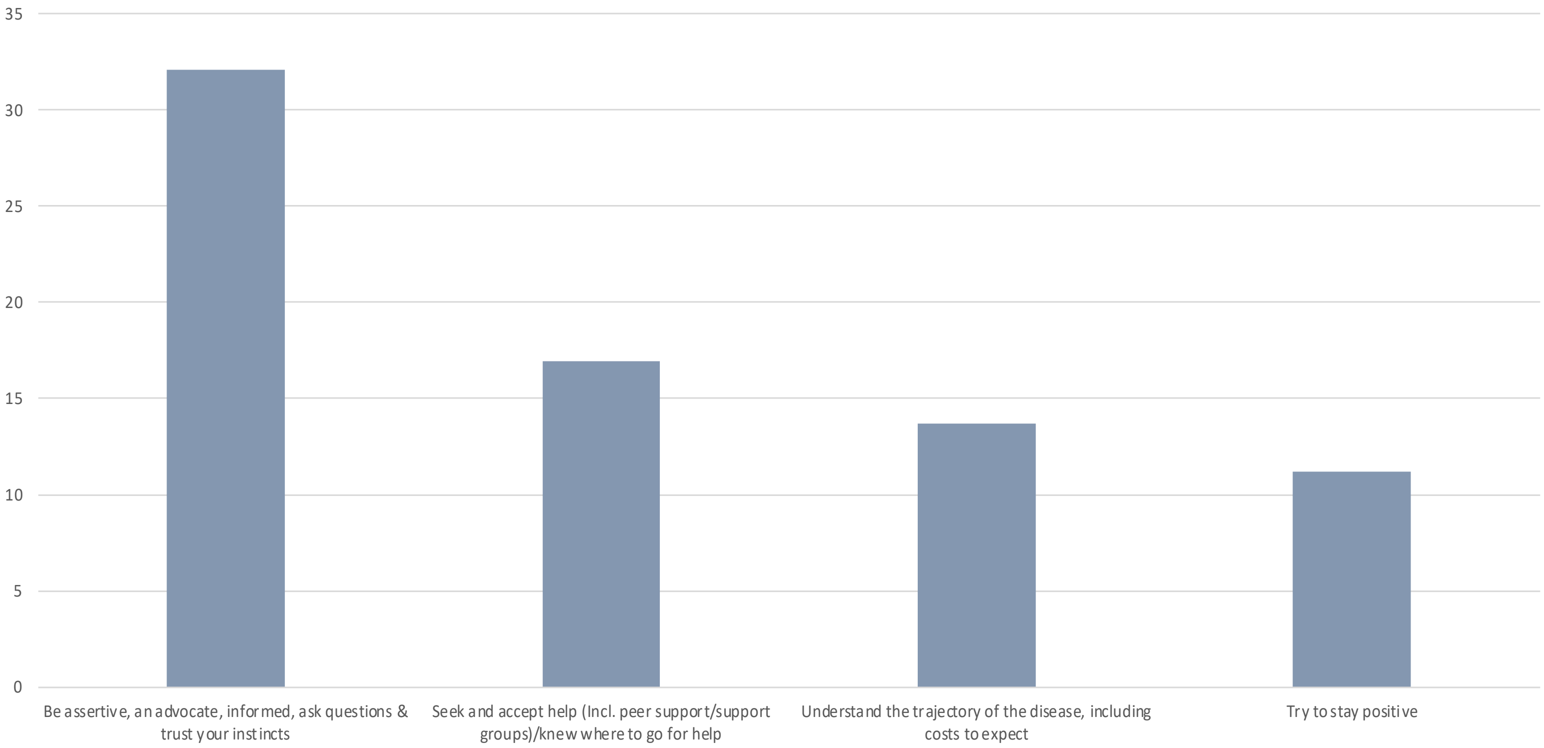


> ***I wish there wasn't as many hoops, you know, like even even when he was younger, to get help from. The department that you know does respite care and and things like that, like we were on waiting lists for that. And the NDIS is the same thing. Like if you don't, if you don't word it properly, you 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***

Section 10

Advice from current patients
to
patients in the future

In this section we present the response to the question 'what is your advice to other people who are diagnosed in the future?' This gives participants the chance to impart knowledge and for patients to feel connected with others in the community.



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

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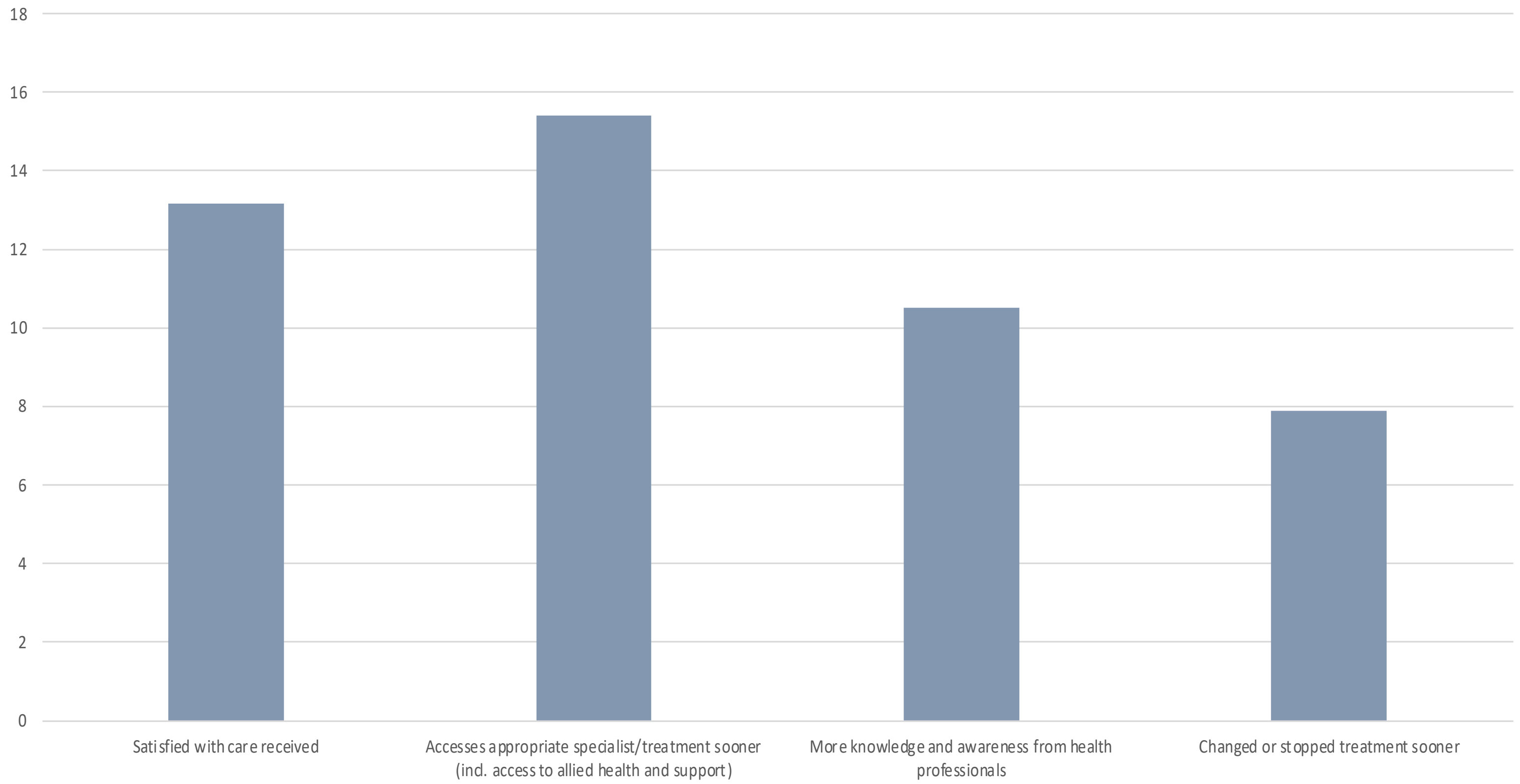
Wish they had known earlier (% of all participants)



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

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

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Anything to change about treatment or care (% of all participants)

- Discussion in the context of existing literature
- Key points
- How the PEEK study relates to existing literature
- Characterisation of the patient cohort

Discussion

Next steps

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.