

## Section 8

### Quality of life

## **Section 8: Quality of life**

### **Impact on quality of life**

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

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

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

### **Impact on mental health**

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

### **Regular activities to maintain mental health**

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

### **Regular activities to maintain health**

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

### **Experience of vulnerability**

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

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

## **Impact on relationships**

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

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

## **Burden on family**

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

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

## **Cost considerations**

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

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

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

## **Overall impact of condition on quality of life**

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

## **Fear of progression**

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

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

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

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

## Impact on quality of life

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

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

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

### **Participant describes a minimal impact on quality of life that has a general or temporary impact**

*Your quality to some extent, to some extent. I wouldn't go too far. A little bit, A little bit.*  
**Participant 012\_2023AUORC**

*Compared to my other conditions, I wouldn't say it's impacted with all that much.*  
**Participant 019\_2023AUDSK**

*No, it hasn't affected us too much. As I said, she, she lives life just like her brother and sister. She's very able. The only thing it is, is maybe being a little bit behind her peers, but as in family life, there's been no there's nothing that challenged us so far or made a massive difference.*  
**Participant 10\_2023AUDPA**

### **Participant describes a negative impact on quality of life due to the emotional strain (including family/change in relationship dynamics)**

*It has it, it's affected it, it's my partner has to be more aware. He has to be prepared to use his sick days to look after me rather than look after himself. It's*

*affected relationships with family. It's left us financially struggling. It, on occasion, it affects my relationship with my children because I'm there, supposed to be their full time carer and some days I can't care for myself. Mentally it affects most of our family because they can see the wounds, they can smell them, they want to help but they can't and that plays on them. Especially my parents. That plays on their mental state often and it just makes life harder.*  
**Participant 012\_2023AUDSK**

*As we just talked in the previous one, my 15 year old daughter, her life has been impacted as she's been expected to care in a way for her sister that wouldn't normally happen in a sibling relationship. She's been expected to provide medical support and you know she has had to get a job to in order to afford the things that she likes to do for myself as well and NAME's quality of life very much depends on how much lotion we can afford to purchase and vitamins and pills and doctor's appointments and her access to allied health and that sort of thing. So everybody else's quality of life is juggled whilst providing her the best opportunity to be as well as possible*  
**Participant 80\_2023AUDIS**

*I'm not an equal partner anymore. I can't do things. I can't just go and do the dishes. I can't cook dinner. I used to be able to help clean around the house. Can't do that. I feel like I'm just breathing through, just not participating and not giving everything I do as much as I can. It's hard. You can't really, you hurt your hands to do these things.*  
**Participant 018\_2023AUDIS**

### **Participant describes a negative impact on quality of life due to reduced capacity for physical activity/needing to slow down**

*Absolutely because I guess the scleroderma in itself didn't...Apart from having to go to a specialist appointment, didn't really affect anything until the pulmonary arterial hypertension kicked in. Since then, I've been very limited. I've had not a bad life, but within limits. I haven't been able to do the things that I would have done with my family because I can't walk as far and I can't go up hills, and I need to sit down. I'm a person who likes to go out to concerts, plays, and art galleries. I still did those things, but I found that they took a lot more out of me and I couldn't do them as easily. I would be holding everybody up because I had to sit and rest. Now, I'm actually past being able*

*to do those things. This is the arthritis rather than-- They're telling me it's nothing to do with the progression of scleroderma.*

*Participant 004\_2023AUDIS*

*Well, it's certainly affected my quality of life because I can't even cook as I enjoy doing or go out for meals and enjoy. I do that, but I don't know what I'm eating. Oh, fatigue, I don't think I've mentioned that, but fatigue has been a big thing in a general way with scleroderma. I get very tired and I, by and large, have a nap almost every afternoon. If I'm sitting at the computer, my head hits the computer because I'm just asleep, really. That's certainly something that's made a difference.*

*Participant 012\_2023AUDIS*

**Participant describes a negative impact on quality of life due to reduced social interaction**

*There has been a huge amount of stress on everyone in the family, including our youngest child. I like...one of my siblings actually literally no longer talks to me anymore because she just feels that we're all just a bit high drama and, you know, there's just a huge narrowing of social circle because it's just very difficult in order to get out and relate, you know, we can't, we can't fit into what other people are doing. There's the fact that it's been impossible for my husband to see his family because they're in LOCATION and traveling with PATIENT in the middle of the global pandemic is impossible. There's and then there's like all the positives in that, like, we just....also sort of have...so we don't kind of embrace who we are and love people for who they are. Don't go around just people because they're in different or you know, it's opened up and you know, silent.*

*Participant 87\_2023AUENM*

*Very, very much so. I feel it's destroyed my, it's destroyed all my friendships. I have no friends. I had a reasonably good friend group...A lot of people can't handle the fact that I got so sick so quick and I look different and I walk different and they get embarrassed. So I am extremely isolated. Even my adult and teenage children struggle to be in public with me because I walk slow and I have a limp and they find it hard to accept, so I'm extremely socially isolated too and I find it really hard to get out in public on my own. I need a support person with me and the availability of that is very limited. So I find I'm extremely socially isolated, like to the point where I have no social aspect in my life at all.*

*Participant 016\_2023AUDIS*

**Participant describes a negative impact on quality of life due to managing side effects and symptoms**

*Look, while symptoms were present, yeah, absolutely not. I suppose because I've seen what it could be or how bad it could be. Not to that extent. But yeah, definitely very uncomfortable. It's very painful and it sort of, yeah, it certainly had a minimizing impact on, on what you did and then how well you felt to go and to go and do usual things. And I suppose in terms of family, yeah, as a single parent, that then also meant if I didn't feel up to it, that my daughter wouldn't go places because I wasn't in a position to be able to take her and support that.*

*Participant 007\_2023AUDSK*

*It has affected in certain ways because of obviously not being able to go out and do things and, you know, sort of making sure he's OK sort of thing before we do things, especially with his OCD and his anxiety.*

*Participant 11\_2023AUDPA*

**PARTICIPANT: Significantly.**

**INTERVIEWER: Yes.**

*PARTICIPANT: It impacts on her ability to have friends at school. She's not the same cognitively as her peers. Her learning is delayed. Yes, all areas of her life are impacted. She misses things because of seizures. She misses things because of appointments. She gives most things a go. For our family, the other two kids definitely misses out on stuff because of her, and you constantly think and analyse what you do and how to have minimal impact on them, or you think of the effects on them and try to make considerations for them as well.*

*Participant 53\_2023AUDPA*

**Participant describes a negative impact on quality of life due to being unable to travel/adapt significantly in order to travel**

*It's affected my quality of life a great deal. Constant stress and worry. Not having the normal freedoms that you would have in retirement to just impulsively take off somewhere because you're always checking, can I do this? Is everything going to be alright at home while I'm away? Family relationship? Yeah, because sometimes family members don't always understand that how my daughter is. Behaving is part of to DiGeorge and I might think that I'm defending her, which I'm not. Yeah, that that would be an average, an average stress, not a high one. But the impact on my time and lack of freedom is very high.*

*Participant 8\_2023AUDPA*

*Yeah it, it has expected yeah because we we really couldn't go on holidays so we've had to just be looking after both of our kids and so yeah it's it has affected probably our relationship and our what we had anticipated our life would be like. But we've just adapted to that and changed what we had, you know our sort of our expectations. We we changed them and altered them to just doing stuff around our place and and you know getting into boat because we live on this river and it's really nice area. So we've just do boating and taught our kids kayaking and just did sort of home like things around the home and around the local area so that we can still have a good life. We still have a great life and a good quality of life. But well, I guess we've just adapted our expectations and, yeah, altered, altered that to what we'd sort of first thought we might end up doing.*

*Participant 9\_2023AUDPA*

**Participant describes a negative impact on quality of life due to the emotional strain on self**

*The culture has, I mean, this half of the people, they don't know what I've got. I don't tell them. I don't know. It's just the close friends that they know what is wrong with me. But I don't talk to anyone else about my condition because I'm I'm, I'm ashamed kind of, yeah.*

*Participant 023\_2023AUDSK*

*I feel like it has, because I can't do the things that I used to be able to do. I'm probably not as much fun to live with nowadays as I used to be, because it gets you down. When I stay in bed, I probably have one day a fortnight at least, or one day a week sometimes where I'm in bed. Particularly in the winter, I would stay in bed all day, one day a week, because I feel terrible and that impacts my relationship with my husband because he's hanging around the house waiting for me to get up energy I suppose. My grandchildren, I've got five grandchildren. I can't play with them like I used to and they wear you out.*

*Participant 013\_2023AUDIS*

**Participant describes a negative impact on quality of life due to altering lifestyle to manage condition (including being immunocompromised)**

*Well, if you could take it off them, you would. If you could wear the pain, you would. It's a significant cost to your emotional well-being. You feel guilty and you you just, you you shut the doors. Well, I shut the doors to doing. We've cut down on a lot of our not that we, you know, had a hugely social engagement calendar,*

*but very very selective in in what things we go to now even or even more so. You would. Yeah. So you and if anyone's sick it's like don't come over don't you know and we don't go out. Yeah. So that's a, that's a but that's that's something that you just you have to accept. It's you don't it's not it's not worth taking a risk on that.*

*Participant 009\_2023AUDSK*

*Yes, it has I, especially with, with Raynaud's like, I can't like, I will not go to cold climate. If we go out to dinner, we always have to make sure that we sit inside. That's, I mean, that's pretty. That's not really quality of life. Look, I've had to get a cleaner in the house. Because due to my ulcers, I can't clean my bathroom. I'd want to. I came to that realization about three years ago. I had to stop my my netball. Yep. Look it, yep, it stopped me from playing the sport that I love around the house. I can't, you know, because of the joint pain and the right rheumatoid. I can't open jars. There's lots of things that that my husband and the kids have to help me with, help have to help with. I can't tie shoe laces anymore or do buttons, so I don't buy shoes with laces anymore.*

*Participant 015\_2023AUDIS*

**Participant describes a negative impact on quality of life due to fatigue**

*PARTICIPANT: Yes. I'm just so tired. Before this, I was an extremely active woman playing basketball three, four times a week, and my girls would play with me and we were in the same team. We ran a business and I worked full-time. Income was really good, things were going really great. That all stopped quite quickly post shoulder replacement and this fatigue, I started to get very fatigued and have joint pain, which I didn't know at that time what it was. I think tiredness is probably number one.*

*INTERVIEWER: Yes.*

*PARTICIPANT: It's hard. I still look after the kids, but limited time because I have crashes all the time, you know, where you are just drained. Definitely have, I can't walk as far now, get too tired. Definitely can't do as much as I...might be a bit of age in in there too though. I was thinking that I'm barely, I'm 62 now. Things happen when you get older as well, so I can't just blame that, but yes.*

*Participant 007\_2023AUDIS*

*Yes, because I'm not as active as I used to be. I'm...I sit on the lounge. I'm tired after. If I sit down, I've got to get up to do something because I forgot to do something. It's like, oh geez, I got to end up getting*

*someone else to do it because I just don't have the energy left to do it. And yes, the grand, because the grand, the grandchildren don't live close. So when they come to visit, you know. It's like a it's like a hurricane hitting the house after two hours. I'm exhausted.*

*Participant 011\_2023AUDIS*

**Participant describes a negative impact on quality of life due to inability to work/changes with their work**

*Yeah, yeah, I do. I guess, you know, her dad and my's relationship broke down and yeah, I said I haven't been able to work. Her sister has grown up not knowing anything different apart from going to a ton of appointments. So yeah, not quite the life I envisaged, but that's what we got.*

*Participant 21\_2023AUDPA*

*She's a different child to my son who has no genetic conditions or health conditions. She's just a different child that we didn't anticipate but we have adapted, and we accept her the way that she can, I guess, interact with us. I guess other sort of flow on impact. Like I say that limits what the family can do we can just all go to the pool or something like that. It's quite a logistical exercise. We can't go out for dinner or breakfast because CHILD'S NAME might not tolerate sitting in a highchair and start crying and screaming and that's a way of communicating because we can't communicate with her. We can't help her work forever. It limits our interaction with the world that we would otherwise be having. What else? My work. I can't really advance with my work given that I can't work full time. I need to work part-time to have days off to fit in all of CHILD'S NAME's appointments. I also need to have a very flexible workplace so that when we have appointments at NAME HOSPITAL that are impossible to change. If you don't want to wait another four months, I need to have a workplace that understands that I can't go to work that day I've got an appointment at the NAME HOSPITAL. It also has to be flexible.*

*Participant 61\_2023AUDPA*

**Participant describes a negative impact on quality of life due to having no respite, condition is always there**

*I guess, well, it affects it. I guess it affects everything because she requires I guess 24 hour care. So it's like anything you're doing, any decisions you make is revolved around her and how she's feeling or what, whatever. You know, making decisions to go to a*

*party. Well, we know how she's feeling today. People is that.*

*Participant 16\_2023AUORC*

*Well, this is a painful one. It affects us greatly. We're very isolated, I haven't in over five years. I'm with my son 24/7, I shouldn't say 24/7, I'm with him every night. I go to work. I don't have any family, my relationships have failed. My son is very isolated because of his autism and he very angry, he doesn't want tuberous sclerosis, he wants to be the same as every other neurotypical child. When to try to talk to him about getting the services that he needs, he rebels against it because again, expecting services means that there's something different about him and he doesn't want to be different. I'm trying to get help for him, psychological help. It has been a nightmare, an absolute nightmare and I'm fortunate because it's been probably eight years of hell trying to get someone to support him and help him. He no longer trusts or wants to have anything to do with a therapist of any description so that makes it really difficult to move forward.*

*Participant 59\_2023AUDPA*

**Participant describes a negative impact on quality of life due to intimacy challenges**

*I don't live with my family, so, like, my mum's gone. Yeah, my dad wouldn't bother. Nothing to do with him, but I have a partner and yeah, since...like, this is going to be crazy, but like, we haven't slept together in over a year, probably 2 years, and we're still together and we're not seeing anyone else. And that is because he understands that like when you do stuff or get hot or sweaty down there, it flares up and then I'm screwed up for days, so... Like, we just need to wait till I can get this surgery and then and then like, maybe I want to have kids. But, you know, hormones and stuff just flare this shit right up. So I'd prefer to have it gone before I'm pregnant or something, you know?*

*Participant 006\_2023AUDSK*

*Yeah, I guess. I guess it has in the sense that yeah, I just get tired. Yeah, physically with hubby. Yeah, yeah, that's non existent. As I keep saying, it's shut, it's closed.*

*Participant 019\_2023AUDIS*



Participant describes a positive impact on quality of life because it brings people together/highlights supportive relationships

*It has probably affected the quality of my life somewhat. I probably struggle to be active because I am tired quite a lot. It can struggle, you know it depending on where where I'm at at the time. If I'm flaring I can struggle to do things around the home. So it has had an impact on, on day-to-day living, has impacted relationships, yes and no. You know, my husband has been involved in my my diagnosis from the beginning and has a fair understanding of how it impacts me. So it hasn't been easy on him. So it has impacted the relationship to some degree, but at the same time he's been involved in it and he has an understanding of what I'm going through and what it's like for me, so.*

*Participant 001\_2023AUDSK*

*Quality of life? In some ways, yes. Not with family. I feel like they've all been very supportive. But I do worry about my daughters having the same condition.*

*Participant 011\_2023AUDSK*

*Of your family, I suppose, yeah, because we've had to readjust how I always imagined I would raise a kid. Like we can't do mud. All playgrounds are made of bark chips, and he can't go in bark chips like can't go in sand pits. So there was a lot of it was a big adjustment into he can't just go and be free in the wild. But once those, once you kind of just reshift your thought pattern, there's it doesn't really we still go camping, we still go swimming in creeks, we still do. We still do everything we would do. We're just aware of the extra precautions or extra physio. After an activity in terms of relationships, both our families are fine. They're not. I don't want to say they're not bothered by it, but it's it's as if it's not there. They pitch in, they do his meds, they do his physio. When he does sleepovers, it's just it's just become part of their world as well. We did have a few friends though, that we kind of never heard from again once we disclosed it. Which isn't uncommon obviously for diagnosis. A lot of people they just so they were in self-preservation. Perhaps they need to remove themselves from that kind of, I don't know, pain, I don't know where it comes from, but but I was like as a that that's not uncommon in any sort of diagnosis like this.*

*Participant 25\_2023AUORC*

Participant describes a positive impact on quality of life as they realise what is important (giving perspective/staying positive)

*I've had an absolutely wonderful life. I've, you know, traveled and hiked and done every sport I wanted to. And so all of that's wonderful. There's been some parts, but they're really challenging for, like hiking... I had my pocket nebulizer with me and had to stop every half an hour to take a break. I guess other people might say that my quality of life has been compromised and I don't really think so because I've had more experiences than most people my age right now. I can't join my friends in things that I would normally do and that's the post COVID syndrome on my CF damaged lungs. So you know my lung function currently is down to 38% so at the moment. My quality of life has changed dramatically. I'm still doing a lot of things, but I'm not playing golf here, so that means I'm not that. That's a bit of social interaction that's gone. Mind you I've just last week I started meeting the girls for Devonshire tea after golf, skip the golf, but still go for morning tea. So finding a way around that and... Yeah, I'm not. I'm used to doing my big power walk around... but now it's a stroll down to the little cafe. So I'm still meeting that walking buddy. But it's not the the exercise has been changed.*

*Participant 18\_2023AUORC*

*No, not at this stage. I think at...because it's, because it's early days. I haven't had, like we discussed earlier there, there haven't been major side effects or anything like that. Obviously, my parents would be upset when they first found out that it came back. But yeah, I think we've sort of moved on from that and we're just sort of taking one sort of day at a time kind of thing and just seeing where where the treatment takes me in, you know, in the next few months and just I suppose just seeing where. What my next step is kind of thing like you know if it does come back, if it doesn't come back or if it shrinks more or what my next sort of step is to to get rid of it. But quality of life I haven't really changed too much. Obviously there's just that sort of breathing issue and and sort of the cough but everyday life just goes on. You know I still wake up every morning grateful that I'm still here and shower, shower and eat and yeah, nothing's nothing's changed. Go to work and and try and keep my mind off it as much as possible. So yeah.*

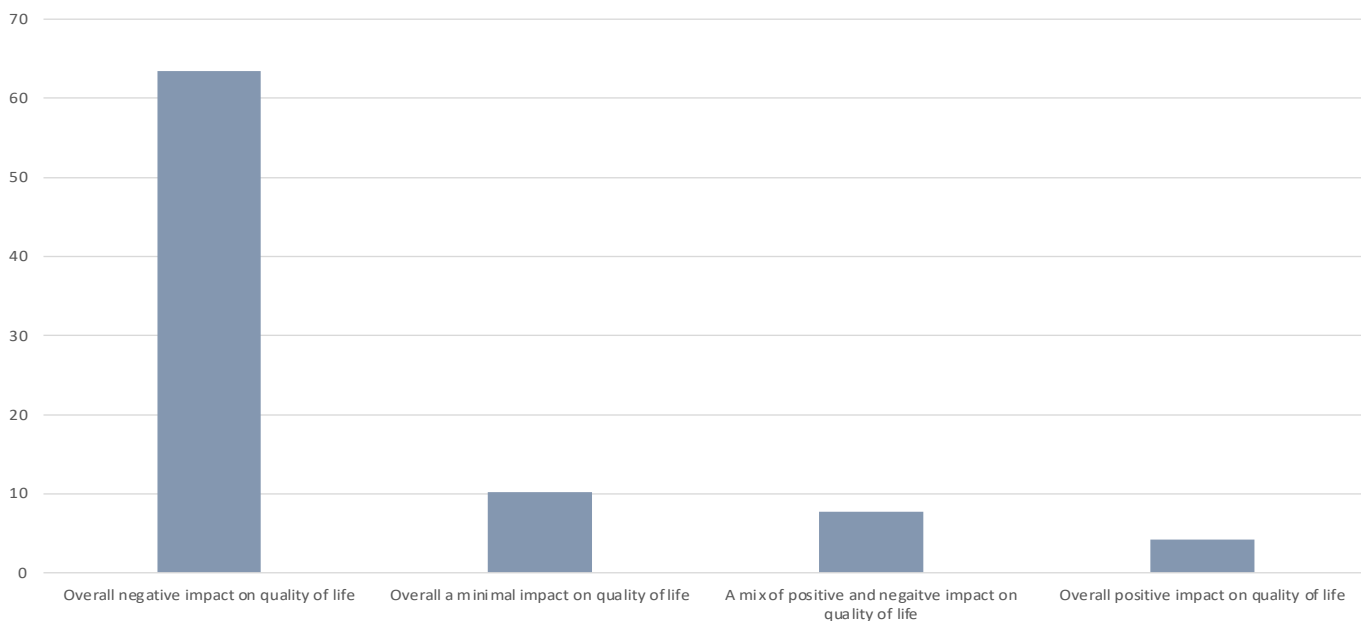
*Participant 24\_2023AUORC*

**Table 8.1: Impact on quality of life**

Impact on quality of life	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Overall negative impact on quality of life	255	63.43	40	59.70	58	71.60	70	73.68	21	65.63	49	51.58	17	53.13	176	65.67	79	58.96	185	62.93	68	64.15
Overall a minimal impact on quality of life	41	10.20	10	14.93	8	9.88	4	4.21	2	6.25	13	13.68	4	12.50	27	10.07	14	10.45	25	8.50	16	15.09
A mix of positive and negative impact on quality of life	31	7.71	7	10.45	4	4.94	5	5.26	3	9.38	7	7.37	5	15.63	21	7.84	10	7.46	24	8.16	7	6.60
Overall positive impact on quality of life	17	4.23	3	4.48	0	0.00	9	9.47	1	3.13	4	4.21	0	0.00	11	4.10	6	4.48	15	5.10	2	1.89
Overall no impact on quality of life	11	2.74	2	2.99	1	1.23	2	2.11	1	3.13	3	3.16	2	6.25	8	2.99	3	2.24	9	3.06	2	1.89

Impact on quality of life	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Overall negative impact on quality of life	255	63.43	53	54.64	85	64.89	76	66.67	41	68.33	131	66.16	120	61.22	75	67.57	180	61.86	133	66.50	122	60.40
Overall a minimal impact on quality of life	41	10.20	12	12.37	14	10.69	7	6.14	8	13.33	16	8.08	25	12.76	14	12.61	27	9.28	20	10.00	21	10.40
A mix of positive and negative impact on quality of life	31	7.71	9	9.28	11	8.40	9	7.89	2	3.33	16	8.08	15	7.65	6	5.41	25	8.59	11	5.50	20	9.90
Overall positive impact on quality of life	17	4.23	6	6.19	5	3.82	4	3.51	2	3.33	8	4.04	9	4.59	3	2.70	14	4.81	9	4.50	8	3.96
Overall no impact on quality of life	11	2.74	2	2.06	5	3.82	2	1.75	2	3.33	7	3.54	4	2.04	3	2.70	8	2.75	4	2.00	7	3.47



**Figure 8.1: Impact on quality of life**

**Table 8.2: Impact quality of life – subgroup variations**

Impact on quality of life	Reported less frequently		Reported more frequently	
	Endocrine, nutritional or metabolic diseases	Other rare condition	Diseases of the nervous system	
Overall negative impact on quality of life				

**Table 8.3: Impact on quality of life (Reasons)**

Impact on quality of life (descriptions)	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Negative impact: emotional strain (including family/change in relationship dynamics)	168	41.79	34	50.75	44	54.32	37	38.95	7	21.88	34	35.79	12	37.50	109	40.67	59	44.03	129	43.88	38	35.85
Negative impact: reduced social interaction	96	23.88	16	23.88	19	23.46	21	22.11	5	15.63	26	27.37	9	28.13	61	22.76	35	26.12	73	24.83	22	20.75
Negative impact: reduced capacity for physical activity/need to slow down incl. because of disability	82	20.40	8	11.94	31	38.27	15	15.79	7	21.88	18	18.95	3	9.38	69	25.75	13	9.70	61	20.75	21	19.81
Negative impact: emotional strain on self	43	10.70	5	7.46	4	4.94	4	4.21	3	9.38	18	18.95	9	28.13	31	11.57	12	8.96	29	9.86	14	13.21
Negative impact: managing side effects and symptoms	43	10.70	16	23.88	9	11.11	8	8.42	5	15.63	0	0.00	5	15.63	23	8.58	20	14.93	28	9.52	15	14.15
Negative impact: altering lifestyle to manage condition (including being immunocompromised)	42	10.45	9	13.43	8	9.88	19	20.00	1	3.13	0	0.00	5	15.63	23	8.58	19	14.18	32	10.88	10	9.43
Negative impact: unable to travel/adapt significantly in order to travel	39	9.70	10	14.93	3	3.70	3	3.16	1	3.13	18	18.95	4	12.50	21	7.84	18	13.43	27	9.18	12	11.32
Negative impact: fatigue	29	7.21	1	1.49	15	18.52	7	7.37	0	0.00	6	6.32	0	0.00	27	10.07	2	1.49	22	7.48	7	6.60
Positive impact: realise what is important (giving perspective/staying positive)	28	6.97	0	0.00	1	1.23	12	12.63	0	0.00	13	13.68	2	6.25	19	7.09	9	6.72	23	7.82	5	4.72

Impact on quality of life (descriptions)	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Negative impact: emotional strain (including family/change in relationship dynamics)	168	41.79	37	38.14	57	43.51	52	45.61	22	36.67	76	38.38	88	44.90	48	43.24	120	41.24	86	43.00	82	40.59
Negative impact: reduced social interaction	96	23.88	23	23.71	33	25.19	29	25.44	11	18.33	49	24.75	45	22.96	30	27.03	66	22.68	53	26.50	43	21.29
Negative impact: reduced capacity for physical activity/need to slow down incl. because of disability	82	20.40	11	11.34	24	18.32	26	22.81	21	35.00	43	21.72	38	19.39	20	18.02	62	21.31	40	20.00	42	20.79
Negative impact: emotional strain on self	43	10.70	10	10.31	10	7.63	15	13.16	8	13.33	28	14.14	15	7.65	14	12.61	29	9.97	16	8.00	27	13.37
Negative impact: managing side effects and symptoms	43	10.70	14	14.43	14	10.69	8	7.02	7	11.67	21	10.61	22	11.22	13	11.71	30	10.31	18	9.00	25	12.38
Negative impact: altering lifestyle to manage condition (including being immunocompromised)	42	10.45	17	17.53	11	8.40	11	9.65	3	5.00	18	9.09	24	12.24	8	7.21	34	11.68	23	11.50	19	9.41
Negative impact: unable to travel/adapt significantly in order to travel	39	9.70	10	10.31	9	6.87	11	9.65	9	15.00	21	10.61	16	8.16	15	13.51	24	8.25	12	6.00	27	13.37
Negative impact: fatigue	29	7.21	1	1.03	9	6.87	11	9.65	8	13.33	14	7.07	15	7.65	10	9.01	19	6.53	17	8.50	12	5.94
Positive impact: realise what is important (giving perspective/staying positive)	28	6.97	8	8.25	12	9.16	6	5.26	2	3.33	13	6.57	15	7.65	6	5.41	22	7.56	11	5.50	17	8.42

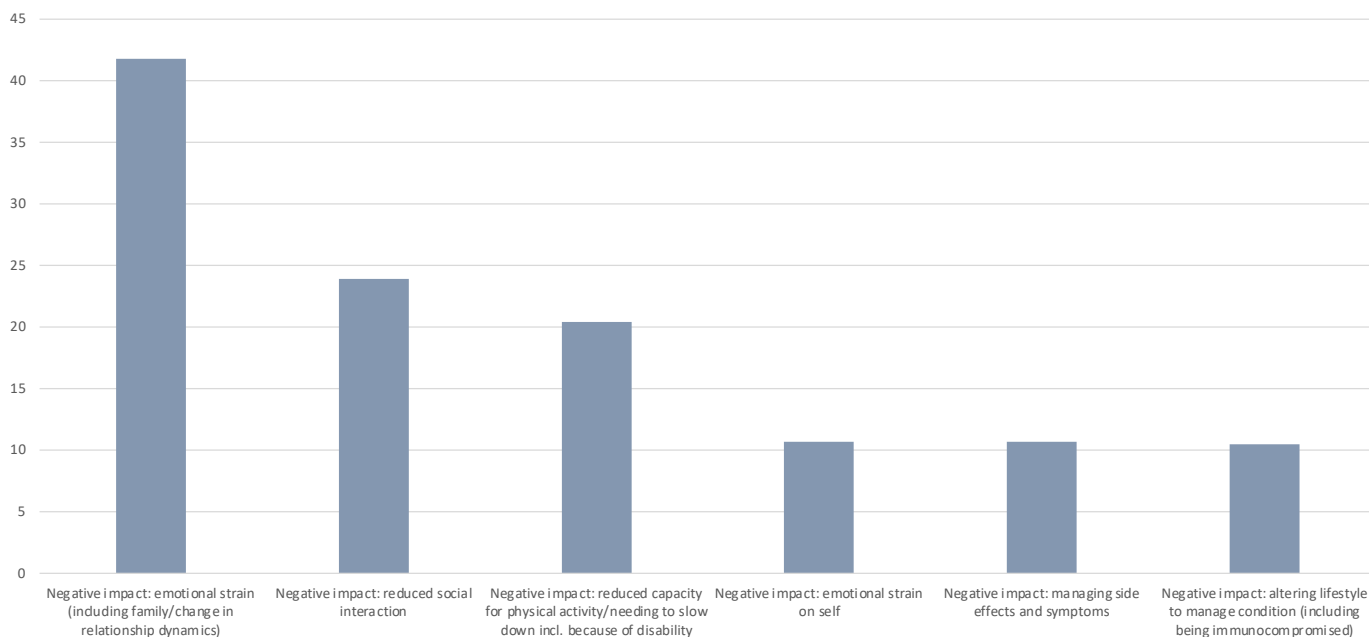


Figure 8.2: Impact on quality of life (Reasons)

Table 8.4: Impact on quality of life (Reasons)– subgroup variations

Impact on quality of life (descriptions)	Reported less frequently	Reported more frequently
Negative impact: emotional strain (including family/change in relationship dynamics)	Diseases of the skin	Diseases of the immune system
Negative impact: reduced social interaction		
Negative impact: reduced capacity for physical activity/need to slow down incl. because of disability	Other rare condition Family or carer	Diseases of the immune system Aged 65 plus
Negative impact: emotional strain on self		Other rare condition
Negative impact: managing side effects and symptoms	Endocrine, nutritional or metabolic diseases	Developmental anomalies
Negative impact: altering lifestyle to manage condition (including being immunocompromised)	Endocrine, nutritional or metabolic diseases	
Negative impact: unable to travel/adapt significantly in order to travel		
Negative impact: fatigue		Diseases of the immune system
Positive impact: realise what is important (giving perspective/staying positive)		

### Impact on mental health

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

Experience described suggests that overall, there was at least some impact on mental health

*Yeah, 100% it does it affect because it affects yourself esteem. It makes you stress about the pain you're experiencing, or even just knowing that it's coming is sometimes worse. Stressing about money, taking time off of work, it's a lot of sometimes I think the mental*

*and emotional roller coaster puts you through sometimes worse than the physical, so that I know it's a lot worse for other people. Thankfully, I'm probably classed more as mild, especially these days. It's not as rampant as what it was, but yeah.*  
**Participant 018\_2023AUDSK**

*I would say it does have an impact on my mental health, especially before the diagnosis. Now that I have the diagnosis, it's it's settled a bit. The, the not knowing was the hardest part and separate to my diagnosis. I do actually see a therapist as well. It's not something I've actually spoken about with her, but this has sort of prompted me. To maybe do that in the future. So yeah.*  
**Participant 027\_2023AUDSK**

*Like, I think I'm a pretty stoic sort of person. But it does get to me every now and then especially if I come across an article and it you know when they're talking about mortality rates and things like that I get really depressed.*  
**Participant 15\_2023AUORC**

*Yes, it definitely has impacted my emotional, mental and physical health. Haven't had a decent night's sleep in seven years. And like, I try to do stuff for my own mental health, but it's very hard. There's a lot of*

*things on my To Do List, and getting to any of my own is really very challenging.*  
**Participant 87\_2023AUENM**

**Experience described suggests that overall, there was no impact on mental health**

*Not particularly. I do what I do. I'm active. I've got a good brain, I manage. I have a job. And yeah, so yeah. So I'm quite active and we travel a lot.*  
**Participant 003\_2023AUDIS**

*No, no, no. I'm always, I'm very much for the kids and getting treatment and that. So I haven't really thought about myself. But no, I've never had to seek attention for that. Yeah, Okay.*  
**Participant 11\_2023AUDPA**

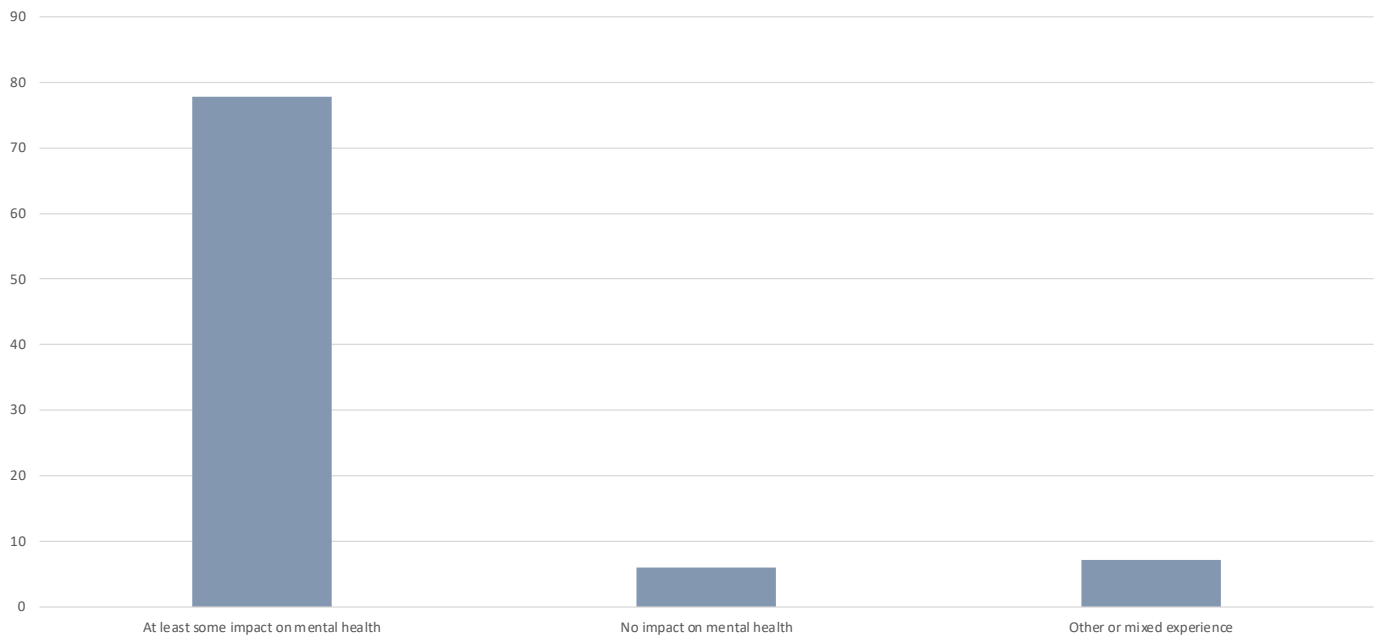
*For me personally, I just I'm, I'm one of those people. I just do what's needed to be done and you just move on. So yeah, I think, I think we've having the condition she's had. It's been a struggle to get across things to in the medical sense and in the in the in the health side of things. But other than that mental health wise with me really hasn't caused any issues. You know, people have often said to me and I don't know how you do it if it's all you've known. I mean, I gave birth to PATIENT when I was 18. So it is a lot known.*  
**Participant 24\_2023AUDPA**

**Table 8.5: Impact on mental health**

Impact on mental health	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=352	%	n=67	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=247	%	n=105	%	n=252	%	n=98	%
At least some impact on mental health	274	77.84	36	53.73	72	88.89	34	75.56	26	83.87	77	81.05	29	93.55	209	84.62	65	61.90	205	81.35	67	68.37
No impact on mental health	21	5.97	3	4.48	1	1.23	6	13.33	1	3.23	9	9.47	1	3.23	14	5.67	7	6.67	13	5.16	8	8.16
Other or mixed experience	25	7.10	1	1.49	7	8.64	5	11.11	1	3.23	9	9.47	2	6.45	19	7.69	6	5.71	20	7.94	5	5.10

Impact on mental health	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=69	%	n=116	%	n=108	%	n=59	%	n=172	%	n=172	%	n=100	%	n=252	%	n=176	%	n=176	%
At least some impact on mental health	274	77.84	41	59.42	99	85.34	88	81.48	46	77.97	132	76.74	138	80.23	77	96.25	197	78.17	138	78.41	136	77.27
No impact on mental health	21	5.97	4	5.80	6	5.17	4	3.70	7	11.86	14	8.14	7	4.07	3	3.75	18	7.14	8	4.55	13	7.39
Other or mixed experience	25	7.10	1	1.45	3	2.59	15	13.89	6	10.17	8	4.65	13	7.56	9	11.25	16	6.35	15	8.52	10	5.68



**Figure 8.3: Impact on mental health**

**Table 8.6: Impact on mental health – subgroup variations**

Impact on mental health	Reported less frequently	Reported more frequently
At least some impact on mental health	Developmental anomalies Family or carer Aged under 18	Diseases of the immune system Other rare condition Regional or remote

## Regular activities to maintain mental health

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

### Participant describes consulting a mental health professional to maintain their mental health

*Yeah, it definitely has an impact. It sort of makes you feel like. You're a bit less lovable. I don't know. There's just a lot going on, I guess. And yeah, I see a psychologist.*

*Participant 019\_2023AUDSK*

*Negatively, at least half the time. And I've been seeing a psych for like, like a psychologist for two years. But*

*then yeah, just working on self-care and all that fun stuff. But it's it's definitely a roller coaster of emotions and acceptance, like new acceptances. Like a lot of parents here, you know, this is the new normal, especially their children in palliative care and so on. But it's definitely hard, even if you have like you know, I was explaining my child as a fairly chill, easy going kid that doesn't have any overly complex medical issues. But when I talk about, you know, getting a feeding tube and doing this and doing that, it's so complex to other people but you just get so you what you're going through. But it's still definitely very hard on my mental health. Yeah.*

*Participant 81\_2023AUDIS*

*Yeah, so it did. It did affect me quite heavily at the start. I was a bit shocked by the diagnosis and it sent me downhill for a while. So I did go and talk to some grief counsellors about it because I guess it was a form of grief that I was going through and that helped me a lot to manage, manage it and manage my thoughts around it. So it's a lot better now. But yeah, at the start it was quite intense. Participant 29\_2023AUORC*

*Yeah, so I see a psychologist once a week, so it's just telehealth. But you know, I have a whole lot of other things going on. So we don't always necessarily talk about my illness, but certainly it has featured many times for the whole session, for the whole hour where I've just talked about the Scleroderma and and its really huge impact on my life.*

*Participant 010\_2023AUDIS*

**Participant describes coping strategies such as remaining social, lifestyle changes and hobbies to maintain their mental health**

*Yeah, I do crocheting. I still pretty involved with my grandchildren although COVID put a spoke in the wheel for that for a very long time that we're kind of getting back to that again and just basically having to reset my brain to kind of well that's not going to happen again. So you know, stop looking for something that's not going to happen because you'll just become disappointed. So I've, I've learned to just say to myself, 'OK, well that's not going to happen'. You can try about it. You can try...So just shut up, get on with it.*

*Participant 002\_2023AUDIS*

*I'm lucky that we're now in the day of age where I can pick up the phone and I can FaceTime a friend and have a chat with her. I'm a big music lover so I just throw on the music, turn up loud and sing away, and I have my little dance party in my house. I go back to my essential oils and I use my relaxing oils to soothe and calm me. I've also learnt that I have to process the emotions through things.*

*Participant 023\_2023AUDIS*

*Dystonia can cause anxiety and depression. I'm not depressed, but I am aware. I get anxious. However, I've got some nice books to read and if I find I'm getting a bit hit up right, you can go out and sit in the sun. You can go and weed, you can go and do this and then this afternoon you've got another book to read.*

*Participant 003\_2023AUDNS*

**Participant describes mindfulness and/or meditation to maintain their mental health**

*Yeah, my, my mental health has been affected, that's for certain. And my emotions and everything has been affected. Yeah, I, I do certain activity once in a while, you know, to kind of, you know, soften the, the effects in my mental health, you know, let's say activity like*

*yoga and meditation, just to calm the calm the tension down.*

*Participant 006\_2023AUORC*

*It can affect my mental and emotional health and that can vary from day-to-day. I can have, you know, at the moment I have a flare and that can get me down because it's just kind of like you suddenly you're feeling well and then bang, you know, it's almost like there's no lit up. Like you, you just go for a day and you think you're doing really well and then suddenly you've got, you know, one flare pops up and. Your arms uncomfortable or whatever. So yeah, look, it can, but I think in that for me, I've had to put things in place to help me with my mental health. So that's for me that's talking about my condition. It's practicing mindfulness and mental health and wellbeing type of activities that keep me, keep me focused on the positive rather than allowing, you know, the focus to become the HS.*

*Participant 001\_2023AUDSK*

*At the moment I'm not doing...I have to do a big work on myself to be able to look after myself, to be able to deal with all issues. It took me time, but I still have anxiety some days. Not every day like before, but I think it's something personal that we have to deal with. It's only me who can stop that anxiety from coming. I have to change my way of thinking to think more positively and to get in contact with people who are not helping me. That's how I was able to deal with this.*

*Participant 020\_2023AUDIS*

**Participant describes no activities to maintain mental health to maintain their mental health**

*I mean, it is right now. It's certainly, yeah, we're having a flare up. It could be quite frustrating, painful and upsetting, but I don't. I don't seek out any.*

*Participant 011\_2023AUDSK*

*No, because there isn't time. You know, we can always say yes, we will. I'll get to that. I'll get to that. But honestly and everything, yeah, something else. There's always something.*

*Participant 16\_2023AUORC*

*I don't know. I don't...my daughter's studying SUBJECT at the moment and she's actually hammering me on that at the moment. She believes I should go. She said even though I'm strong and stubborn, that maybe I should talk to someone because she said the traumas I've been through, that maybe I should talk about it. I haven't actually done anything. I'm on a care plan*

*where I get 5 with my GP, where I get five allied health a year, which I really need about 50, but I get five. I spoke to them about that and they said, well, if I don't get my physiotherapist or podiatry or dietician and all the other allied health, that I could get some mental health support. I've got to weigh up what goes.*

*Participant 014\_2023AUDIS*

**Participant describes the importance of family and friends in maintaining their mental health to maintain their mental health**

*Yes, it did affect my mental and emotional health prior to treatment. But you know as I said during treatment and prior to treatment and continuing after treatment I have a spiritual understanding that I live in the moment. I am very connected with my family and with my close friends.*

*Participant 010\_2023AUORC*

*Sorry, I'm crying, but yeah, it affects my mental health a lot I would say. You know, I have ever...an episode at least twice a month where I break down about having the disease, because I just think, why me? Like why do I have this? And it's just it's ugly and I compare myself to other people all the time who, you know, don't have it. I like there's not much I can do for myself in terms of having disease because it's so hard to pull myself out of this like mental thought that it's ugly and it's something I should be ashamed of and it's just kind of, I need to let myself cry about it and then I move on because it's not going anywhere. So I just have to, you know, get over it. I'd say my partner is definitely someone who helps a lot with my mental health. He'll just remind me that, like, I'm still a great person with or without the disease. Yeah.*

*Participant 010\_2023AUDSK*

*Yeah, I just said we've got really good support. So in the beginning when it was young and it would kind of just, I guess, build up and be a bit too much. We had ample, ample people like brothers, sisters, grandparents that we could just kind of say, can you*

*just watch them for the night or can you come up? I'm just gonna go for a massage. Like, you know what I mean? Like, so we've been really lucky in the fact that at any stage we've had like full wrap around support for whatever we need.*

*Participant 25\_2023AUORC*

**Participant describes the importance of physical exercise to maintain their mental health**

*Well, I can't walk very far because of my back and my legs and my arthritis, but try and do little walks. We try to get on the beach and just sit on the beach and have a walk around we...Yeah...visit friends or go out for dinner. Yeah. Doesn't really emotional how...that's more me. It's just more about, you know, finding a cure, which doesn't exist, you know, that sort of thing. Most things I can fix or have operations for. But this one is like, yeah, no, there's nothing.*

*Participant 024\_2023AUDSK*

*I play a lot of sport. I do athletics, I do Pilates, I've always been active and I think that has helped a lot.*

*Participant 93\_2023AUENM*

*For my, I mean my emotional...I was trying to say emotional and mental at the same time. Emotional health, I guess. I know that I need a good balance between alone time and making sure that I meet up with, you know, friends every now and then just for some social time. My mental health I really have focused on, so focusing on my physical health has also helped my mental health. Because I have, you know, I go for my walks and those sorts of things to help with my mental health. I, I don't handle being stuck at home very well. So yeah, like, it's the diagnosis I think has affected me more than I would care to admit. It's become like another thing. So yeah, I've just been focusing on if I eat right and exercise and things like that and help my mental health, that's gonna help me all over.*

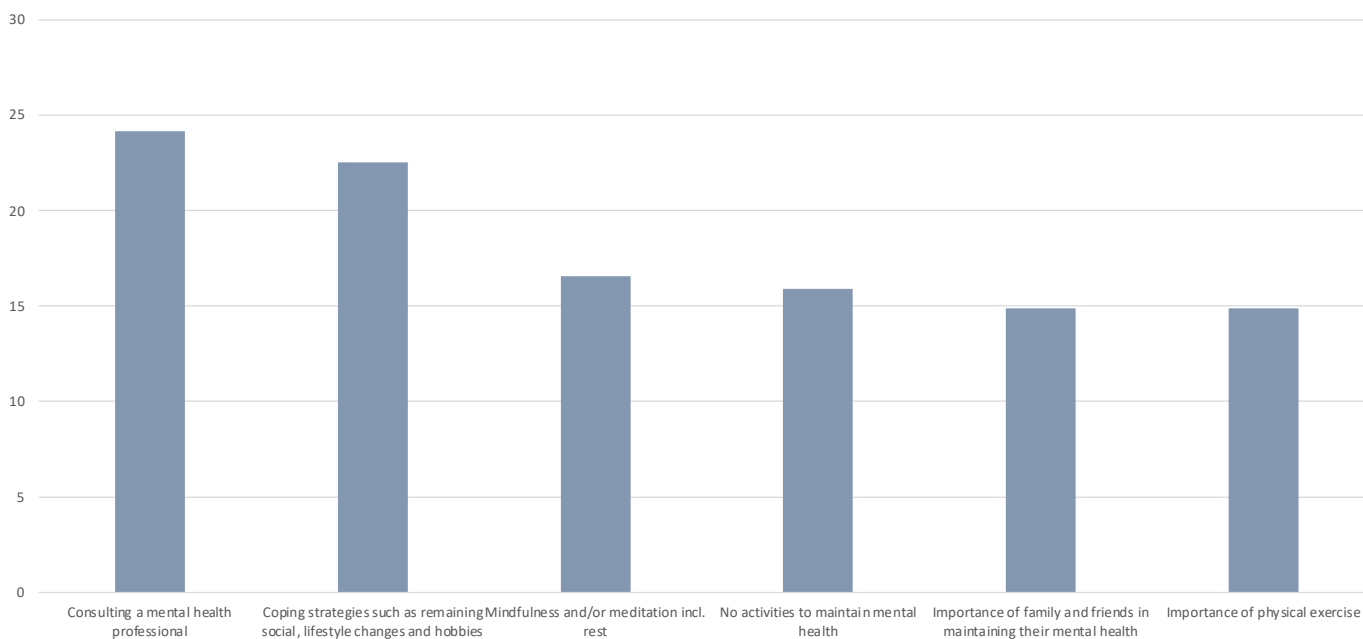
*Participant 29\_2023AUDPA*

**Table 8.7: Regular activities to maintain mental health**

Regular activities to maintain mental health	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=302	%	n=67	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=204	%	n=98	%	n=214	%	n=86	%
Consulting a mental health professional	73	24.17	13	19.40	23	28.40	11	24.44	5	15.63	12	12.63	9	28.13	49	24.02	24	24.49	55	25.70	18	20.93
Coping strategies such as remaining social, lifestyle changes and hobbies	68	22.52	10	14.93	28	34.57	8	17.78	4	12.50	13	13.68	5	15.63	53	25.98	15	15.31	49	22.90	18	20.93
Mindfulness and/or meditation incl. rest	50	16.56	5	7.46	15	18.52	10	22.22	10	31.25	2	2.11	8	25.00	43	21.08	7	7.14	42	19.63	7	8.14
No activities to maintain mental health	48	15.89	5	7.46	9	11.11	11	24.44	7	21.88	9	9.47	7	21.88	35	17.16	13	13.27	29	13.55	19	22.09
Importance of family and friends in maintaining their mental health	45	14.90	3	4.48	18	22.22	6	13.33	4	12.50	9	9.47	5	15.63	36	17.65	9	9.18	37	17.29	8	9.30
Importance of physical exercise	45	14.90	4	5.97	7	8.64	10	22.22	5	15.63	12	12.63	7	21.88	32	15.69	13	13.27	31	14.49	14	16.28

Regular activities to maintain mental health	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=302	%	n=66	%	n=103	%	n=84	%	n=49	%	n=148	%	n=172	%	n=84	%	n=218	%	n=154	%	n=148	%
Consulting a mental health professional	73	24.17	17	25.76	28	27.18	20	23.81	8	16.33	28	18.92	44	25.58	18	21.43	55	25.23	32	20.78	41	27.70
Coping strategies such as remaining social, lifestyle changes and hobbies	68	22.52	8	12.12	16	15.53	24	28.57	20	40.82	31	20.95	34	19.77	17	20.24	51	23.39	36	23.38	32	21.62
Mindfulness and/or meditation incl. rest	50	16.56	4	6.06	25	24.27	14	16.67	7	14.29	24	16.22	25	14.53	16	19.05	34	15.60	28	18.18	22	14.86
No activities to maintain mental health	48	15.89	9	13.64	18	17.48	12	14.29	9	18.37	32	21.62	16	9.30	12	14.29	36	16.51	24	15.58	24	16.22
Importance of family and friends in maintaining their mental health	45	14.90	3	4.55	14	13.59	14	16.67	14	28.57	20	13.51	23	13.37	16	19.05	29	13.30	24	15.58	21	14.19
Importance of physical exercise	45	14.90	7	10.61	15	14.56	13	15.48	10	20.41	17	11.49	25	14.53	9	10.71	36	16.51	22	14.29	23	15.54



**Figure 8.4: Regular activities to maintain mental health**

**Table 8.8: Regular activities to maintain mental health – subgroup variations**

Regular activities to maintain mental health	Reported less frequently	Reported more frequently
Consulting a mental health professional	Endocrine, nutritional or metabolic diseases Diseases of the skin Aged under 18	Diseases of the immune system Aged 65 plus
Coping strategies such as remaining social, lifestyle changes and hobbies	Endocrine, nutritional or metabolic diseases Aged under 18	Diseases of the skin
Mindfulness and/or meditation incl. rest	Developmental anomalies Aged under 18	Aged 65 plus
No activities to maintain mental health		
Importance of family and friends in maintaining their mental health		
Importance of physical exercise		Diseases of the immune system



## Regular activities to maintain health

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

**Participant describes the importance of self care e.g. more rest, accepting help, pacing in maintaining their general health**

*Sometimes there's nothing I can do to control it. There's no amount of pain medication is going to stop the nerve pain from happening when the infections are that bad. No amount of cleaning, irrigating, washing the wounds is going to get rid of the smell there's...It, it makes me feel very powerless, actually. If it's a mild flare I just have to push through. I just have to deal with the pain and then hope my partner comes home on time from work so I can then go and rest and physically sleep from the physical and mental exhaustion of being in constant pain.*

*Participant 012\_2023AUDSK*

*Well, I if I, with the fibromyalgia, I have to sometimes I hit a wall of tiredness and I just have to have a sleep. So I do if I'm get, if I I'm not like that all the time, but when I do get like that, I do, don't get up and go and have a lay down for an hour or so and I'll get up and I feel better. So that's what I do for myself. If I'm not, you know, unwell, I'll have a, I will rest and I, my body tells me I need to go and lay down.*

*Participant 88\_2023AUENM*

*I have to rest when I have to rest, simple as that. Put my feet up and make sure that I take care of that because rest is so important, and sitting aside and just taking time out. Just re-evaluating life and just go for a walk or do whatever you can to get your mind straight.*

*Participant 001\_2023AUDIS*

**Participant describes the importance of complying with treatment/management in maintaining their general health**

*No, no. Well, that's, that's it. I mean I take, I'm probably taking about 10 pills a day now, so that's not*

*too bad. But you know, apart from that, life just goes on. And I mean pills in the morning, pills at night, that's it. It's no great drama and it's, it's quick and easy to do.*

*Participant 007\_2023AUORC*

*I mean, aside from giving him the medication, we have to keep up with the physio on his chest every day. That'll allow him to be able to become more active as he gets a bit older.*

*Participant 29\_2023AUORC*

*For the moment, it's really just around compliance with existing treatments that that creates, you know, conflict, if I'm honest, you know, trying to get her to take her medication, to wear her retainer, to do her exercises, to do her homework, all those sorts of things. I think it's probably the main issue. Yeah, day-to-day it's just around compliance with things she's supposed to be doing to maintain her health, but also what she's supposed to be doing around her schooling.*

*Participant 35\_2023AUDPA*

**Participant describes the importance of doing physical exercise/physically active in maintaining their general health**

*So I have to do exercise to keep my lungs healthy. I have to have a decent diet and you know, drink a lot of water and extra water and have medications every day. And then kind of manage that to how my wife is at that time health wise. So whether I need to increase any medications or add extra ones on or if I need to go into hospital. And yeah, medications in the other medications like doing physio, which takes about half an hour, 45 minutes in the morning and then in the afternoon as well, just to have a clear chest and be able to breathe normally.*

*Participant 13\_2023AUORC*

*Well, I think it's just making sure you're getting up and moving every day. Some days you can do more than others, but I certainly can't just sit or lay. Even if you're feeling fatigued, I've got to move and stretch because then my joints just lock up, if you like. I think it's important that...yes, move every day, do some...As I say, I'll go for, I call it a stroll, it's not really a walk anymore. I get out and about and get some fresh air.*

*Participant 017\_2023AUDIS*

*Well, like I said, she does look after herself. The only activities I'm involved with is her financial*

*business. Everything else she seems to be, well, not really like she had to with this new job she's got. She does a lot of walking. And she needed new shoes and she didn't feel confident getting the right shoes for the job. So I had to take her shoe shopping. So, you know, things like that because, you know, she might go in and buy those because they look pretty, but they're not practical for what she needs, so to speak. So, you know, I do small things like that, but generally, no, she's pretty self-sufficient in in the choices she makes are quite reasonable.*

*Participant 16\_2023AUDPA*

**Participant describes the importance of understanding their limitations in maintaining their general health**

*Pain relief. As I said, I had to change jobs to give up my career and go to a new career, and on any one day it could be dramatic changes to to anything. I'm very fortunate in my new job that I can choose to work from home if I need to because there are some days that putting a pair of pants on is not an option. So I'm very fortunate in that regard. But yeah, sometimes, sometimes every aspect of my life has to be altered. But on an everyday basis, the types of things that have changed my life is obviously changing my career and and pain, pain, pain and more pain.*

*Participant 005\_2023AUDSK*

*I basically don't do anything in the evenings after work because I well, I need occasionally because otherwise I get too worn out. I try and plan activities so that I know that if I've got a couple of busy weeks then I'll make sure I have a weekend with nothing on to recover. I...when I wasn't able to walk much, I was having to manage that by arranging my life around not being able to walk. Very much. I don't. If I'm going to the I go to the theater regularly as friends, but they're kind enough now to go to matinee so I don't have to be out late at night. So it's more things like that to manage fatigue and tiredness, because I find if I'm if I'm not too tired, then I can cope with everything much better.*

*Participant 009\_2023AUDIS*

*Well, since the ablation, there really isn't anything. I don't push it too hard. I sort of just listen to my body and if I feel like my heart's pounding too hard, I do stop. I don't push it as much as what I may have pushed previously. But if you have that fear that ohh God, if it goes too high, is it going to get stuck again? Yeah.*

*Participant 32\_2023AUORC*

**Participant describes the importance of maintaining a healthy diet in maintaining their general health**

*Yeah, so I think just like making like reminding him because he's only....now I like to have his medication every day. So making sure that happens, organizing like the his day-to-day food so that he's got the right food to eat and that's pretty normal. But make like if he's got a birthday party or he has to go away on camp or you know, like making sure that or if he's staying at, you know, he wants to stay at a friend's house. It's like it's organizing you know him for you to take with him or organizing with the other adults that they'll have food that he can have.*

*Participant 79\_2023AUDIS*

*I suppose, join the Pilates so that's keeping my body limber and active. Again, I got the NDIS funding so I managed to get a cleaner in to help in that respect. That's one thing that, I suppose, I do take care of. It's not me physically doing it, but I'm healthy in that respect. I think I'm just taking my drugs and trying to stay as healthy as possible, eat the best food I can, all that sort of stuff.*

*Participant 018\_2023AUDIS*

*I need to plan things. Today, I'm talking with you and that's my day's events. Not overburden the whole thing. I was drinking a lot of alcohol earlier in my diagnosis and life and stuff and I've cut that right back. They're the things trying to eat well. I'm looking forward to the physio planning some more activities to get some fitness back because I have to work out how I can reinvent that from how I was physically active before. I'm looking forward to getting some ideas around that with him that'll be really positive. They're the sorts of thing and just being kind to yourself and being okay that if you need to just stop, that's all right. You're not being weak or pathetic or lazy or any of those sorts of things.*

*Participant 026\_2023AUDIS*

**Participant describes the importance of being organised and planning ahead in maintaining their general health**

*So if I've got a break out. I need to make sure that I've got bandaging to be able to wear, I need to have... funnily enough, I haven't had any really strong pain relief for a very long for over a year now, and I'm quite happy about that. I need to be able to maybe cancel things in short notice if, if that's the case, you know. I need, I feel like I need certainly access to an endless supply of chlorhexidine and I have the biggest Band-*

*Aid and bandage collection in the world. So yeah, yeah, so I guess, I guess. Physically, to be able to, you know, wear undergarments, I need to be able to, you know, have enough padding to protect, you know, and I guess also obviously I need access to my medication and I need access, yeah.*

*Participant 017\_2023AUDSK*

*Yeah. So yeah, on a day-to-day base, I guess is actually sit down with him every morning, get his nebulizers ready, get all his meds ready. We've got to actually make the salt tablets, sterilize all the equipment. Yeah, mainly just monitoring, making sure that the tablets are taken, you know, making sure everything's ordered in advance because some of the medications just take a little while to get in and yeah, just actually administering the the treatments essentially.*

*Participant 20\_2023AUORC*

*Yes, that was just about, I guess, being organized to make sure she has everything she needs in terms of, you know, it might be exchange of clothes or support in terms of special shoes or things like that. And also it's been organized to attend the various appointments and pay the various bills and then also, you know, make calls and emails as needed to. Trace up results or get other questions answered.*

*Participant 22\_2023AUORC*

**Participant describes the importance of maintaining a normal routine in maintaining their general health**

*Yeah, so that's very strict to keep routine, make sure her environment is safe. Very important that she doesn't get overwhelmed because then her behaviour gets worse. Yeah, she's just a lot of safety cleaning she engaged and things like fecal smearing, so...obviously having things, you know, clean up fast and make sure she doesn't ingest anything and get sick.*

*Participant 94\_2023AUENM*

*I have a bit of a routine so I know my energy levels are best in the morning when I get up. It's keeping a routine and trying to keep mentally healthy and sticking to that routine so that if I'm exhausted, I know there'll be time in the afternoon to rest. Exercising to keep the joints mobile and also recognizing when your body's telling you I'm...I can't do this, you stop, you know, not keep pushing through.*

*Participant 007\_2023AUDIS*

*So things like sticking to a routine and stuff like that, yeah, yeah, yeah. So trying to stick to a routine to make sure things run smoothly, very much having to*

*warn him and show him photos of if we are doing something different to make sure that he's kind of semi familiar because thrusting something different upon him doesn't go very well. So just like you know, letting him know what's happening in advance day-to-day.*

*Participant 23\_2023AUDPA*

**Participant describes the importance of socialising with friends and/or family in maintaining their general health**

*Some of the things I do, I think pacing myself and just both physically and mentally. So not looking too far ahead and just saying, all right, so today I need to deal with this by the end of the week. I need to do this, so not getting too far ahead of myself. And just, yeah, physically, just getting through the day. If it's been a hard day physically, I'm just like, okay, one thing after the other. And I just look forward to getting into bed at the end of the day and resting as much as possible. And yeah, so pacing myself and mentally, I think, I think my work actually helps me so as far as I'm physically able to work. And then it helps me mentally to just distract me and give me something to focus on. I'm pretty good sometimes. I'll take the dog out. Now that my mobility is a bit better. I'll take the dog out. I'll go for a walk. That's really good again because I'm a bit more mobile, will do things as a family, so...go out and do fun stuff on the weekend. So that's that's good.*

*Participant 022\_2023AUDIS*

*Exercise. In fact, everything I just said exercise, social engagement, good diet, sleep. Exercise is huge, you know. Doing that something vigorous every day, yes.*

*Participant 19\_2023AUDPA*

*I love my job. I've been in my job for 36 years with the same company. Very office-based job, so I'm very lucky that we have a cattle property, and on our weekends, we tend to get away up there. I walk every day, I swim most days. Sometimes when that pain did come back, you just have to be a bit kind to yourself and realize that you can't do everything when the pain is that bad. Like I said, I'm very lucky in the support that I have from friends and family. It's probably hard in my family to feel too sorry for yourself because they just bring you back to earth and basically we just...that's good too.*

*Participant 006\_2023AUDNS*

**Participant describes the importance of using complementary therapies in maintaining their general health**

*I'll have to wear my orthotics and I have to do some fairly specific exercises because I have a joint go out. I have to get it to click back in or yeah, and then. I'll try and have a massage, a remedial massage as regularly as possible.*

*Participant 002\_2023AUDPA*

*I just have to keep moving. If you don't keep moving then my lungs get tight and I can't breathe properly. My joints all get too sore and I can't move. So yeah, just sort of stay active or just move as much as possible. There's nothing much else apart from taking medication that I can do. Well, I give you pretty much regular massage on the shoulder, the neck, yeah, that's...*

*Participant 011\_2023AUDIS*

*PARTICIPANT: When I feel like the anxiety is coming, I just stand up and open the door and look for some fresh air and do some stretching, or keep myself busy, although it is very hard for me to do cleaning. If I'm doing some cleaning or folding clothes or things like that, I just have to keep myself busy so I don't think about it, I don't overthink*

*INTERVIEWER: Do you still have a massage?*

*PARTICIPANT: Yes, I do.*

*INTERVIEWER: Yes. Do you find that's helping you to get through?*

*PARTICIPANT: It's helping me, yes, and also I have someone to talk to for one hour twice a week.*

*Participant 020\_2023AUDIS*

**Participant describes the importance of a good hygiene routine to maintain their health**

*Yeah, look, all I do at the moment is, yeah, twice a day in the shower I will wash the sites or the potential sites with chlorhexidine, which is like a surgical scrub thing, but really. That's my only attempt at a preventative sort of measure, but I don't have any symptoms so I'm not needing to manage anything more than that at this point.*

*Participant 007\_2023AUDSK*

*Definitely keeping the area clean. I have to take wet wipes with me wherever I go. So that if I get sweaty or is...you know, one of the scars or bumps kind of pairs open, I can clean it up straight away. There's not, you know, like I can't wear proper bras because it affects the area. So I'm always wearing like stretchy bras that*

*don't have any wires or any like lace or scratchy things on them so that it doesn't aggravate. The area other than that, just keeping clean, like I have to shower twice a day, which is, you know, normal, but I just have to keep it very clean and I'm always very conscious of the area. Just yeah.*

*Participant 010\_2023AUDSK*

*Good hygiene, as in making sure the affected areas are well cleaned and protected. I take vitamins to help counteract. I take medication when it's prescribed. Just looking after myself and getting on with life.*

*Participant 013\_2023AUDSK*

**Participant describes the importance of mindfulness and/or meditation in maintaining their general health**

*So I think I touched on it. You gotta...we've learned that we have to take time out as parents and carers and whether that's together or separate, my husband and I, he goes away and has time with friends or mates. I do the same. And there's occasional time where we go on holidays by ourselves together, and that makes it easier when we do like have days or holidays or look after her every day and we can manage her and her and her mind and emotions and so, so that is using mindfulness, you know, also understanding and realizing when you're starting to get in a bad habit, let's say, you know, when things start to get tough, you drink too much or you eat too much, learning the signs and signals. So we now know what the triggers are because we're we have gone out and sought that help to to help us identify those those things and identify when it is becoming too much and asking for help from the professionals. Like we'll go to the GP and go we really need something here, can you help us? Because the GP's pretty much is your first point of call for most things.*

*Participant 25\_2023AUDPA*

*I have a nice dog. I have a lovely little dog and I've trained her and now she can pick up sometimes when I'm having a spasm and then that's really that I try and stay put. If I do some meditation, I do some breathing. I do all my home programs from the physio and the speech therapist, I try and get out in the sunshine, and I go for walks when I can. I just work hard at being the healthiest and best I can. It doesn't have the right self.*

*Participant 001\_2023AUDNS*

**Participant describes no activities to maintain their general health**

No, no, I'm not doing anything because it's not active, so I don't even know what to do. I just continued my everyday life.

Participant 001\_2023AUORC

I don't need to control anything yet.

Participant 005\_2023AUORC

Nothing at the moment. It's just pretty normal actually, he just fits in. There's no medical or there is medical issues but then there's not a lot there at the moment.

Participant 51\_2023AUDPA

Participant describes the importance of support from carer or support from parent to maintain health

Well, I'm here. Arms and legs and sensory interpretation and educational advocate and community rouser. And like all the things, it's like wiping your bum, to brushing your teeth, to rocking your sleep.

Participant 87\_2023AUENM

I guess one of the like, you know, I've got a...I don't really know to be honest. Obviously I have to get her

everywhere she needs to go and, and a sister getting ready to to be at school on time and you know, I'd be there to pick her up and all of those sorts of things. You know she doesn't have as much independence as her siblings do, just because she doesn't quite understand the risk factors and, and have the comprehension to be able to to do certain things that they do. So it's pretty much having to make sure that I'm always available if and when she needs me.

Participant 27\_2023AUDPA

So she pretty much needs somebody with her every minute of the day so she can't sort of dress herself, toilet herself, change her feeding, pump bottles, extensions, provide medication, meeting she needs sort of assistance with. And then sort of obviously she's sort of globally delayed. So all the things at school, she needs to teach her aid to be with her as a lot of help with reading and writing and socially, she doesn't naturally understand that she needs help with making friends and things like that. And so she's pretty much supported all day, every day. Okay.

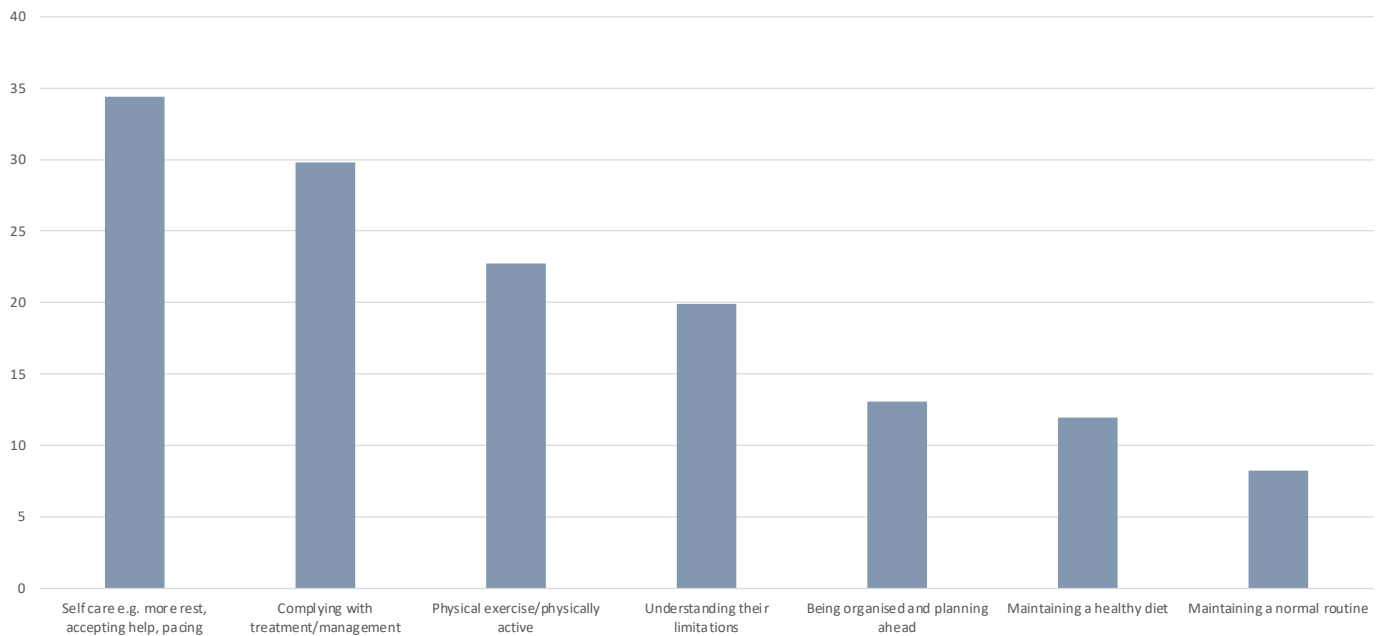
Participant 32\_2023AUDPA

Table 8.9: Regular activities to maintain health

Regular activities to maintain general health	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=352	%	n=67	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=247	%	n=105	%	n=252	%	n=98	%
Self care e.g. more rest, accepting help, pacing	121	34.38	13	19.40	52	64.20	13	28.89	6	19.35	32	33.68	5	16.13	103	41.70	18	17.14	97	38.49	23	23.47
Complying with treatment/management	105	29.83	29	43.28	18	22.22	10	22.22	11	35.48	23	24.21	14	45.16	65	26.32	40	38.10	68	26.98	36	36.73
Physical exercise/physically active	80	22.73	7	10.45	22	27.16	13	28.89	3	9.68	28	29.47	7	22.58	63	25.51	17	16.19	56	22.22	24	24.49
Understanding their limitations	70	19.89	7	10.45	15	18.52	16	35.56	4	12.90	25	26.32	3	9.68	55	22.27	15	14.29	52	20.63	17	17.35
Being organised and planning ahead	46	13.07	22	32.84	7	8.64	2	4.44	3	9.68	4	4.21	8	25.81	18	7.29	28	26.67	25	9.92	20	20.41
Maintaining a healthy diet	42	11.93	5	7.46	12	14.81	5	11.11	4	12.90	10	10.53	6	19.35	32	12.96	10	9.52	28	11.11	14	14.29
Maintaining a normal routine	29	8.24	18	26.87	5	6.17	1	2.22	2	6.45	1	1.05	2	6.45	12	4.86	17	16.19	16	6.35	13	13.27

Regular activities to maintain general health	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=69	%	n=116	%	n=108	%	n=59	%	n=172	%	n=172	%	n=100	%	n=252	%	n=176	%	n=176	%
Self care e.g. more rest, accepting help, pacing	121	34.38	12	17.39	39	33.62	50	46.30	20	33.90	60	34.88	61	35.47	42	52.50	79	31.35	63	35.80	58	32.95
Complying with treatment/management	105	29.83	28	40.58	42	36.21	22	20.37	13	22.03	53	30.81	51	29.65	28	35.00	77	30.56	49	27.84	56	31.82
Physical exercise/physically active	80	22.73	11	15.94	24	20.69	30	27.78	15	25.42	37	21.51	41	23.84	22	27.50	58	23.02	41	23.30	39	22.16
Understanding their limitations	70	19.89	11	15.94	18	15.52	20	18.52	21	35.59	30	17.44	38	22.09	17	21.25	53	21.03	30	17.05	40	22.73
Being organised and planning ahead	46	13.07	21	30.43	13	11.21	8	7.41	4	6.78	23	13.37	23	13.37	8	10.00	38	15.08	19	10.80	27	15.34
Maintaining a healthy diet	42	11.93	8	11.59	11	9.48	12	11.11	11	18.64	20	11.63	21	12.21	11	13.75	31	12.30	18	10.23	24	13.64
Maintaining a normal routine	29	8.24	14	20.29	7	6.03	6	5.56	2	3.39	16	9.30	13	7.56	7	8.75	22	8.73	12	6.82	17	9.66



**Figure 8.5: Regular activities to maintain health**

**Table 8.10: Regular activities to maintain health – subgroup variations**

Regular activities to maintain general health	Reported less frequently	Reported more frequently
Self care e.g. more rest, accepting help, pacing	Developmental anomalies Diseases of the skin Other rare condition Family or carer Male Aged under 18	Diseases of the immune system Aged 45 to 64 Regional or remote
Complying with treatment/management		Developmental anomalies Other rare condition Aged under 18
Physical exercise/physically active	Developmental anomalies Diseases of the skin	
Understanding their limitations	Other rare condition	Diseases of the nervous system Aged 65 plus
Being organised and planning ahead		Developmental anomalies Other rare condition Family or carer Aged under 18
Maintaining a healthy diet		
Maintaining a normal routine		Developmental anomalies Aged under 18

## Experience of vulnerability

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

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

family and friends (3.33%), and support from mental health professionals (2.22%).

### Participant describes feeling vulnerable when having sensitive discussion (diagnosis, treatment decision)

*When I was told that I had to, that I really only had Humira as my only option, I did feel very vulnerable then, yeah.*

*Participant 017\_2023AUDSK*

*Anytime I kinda have to ask for help because I have to start from the beginning. And yeah, I've really gotta you wait for the 'no' and the antibiotics and the just the standard stuff that you know isn't gonna do anything, but you have to do it so they can tick off their list that they've tried that.*

*Participant 018\_2023AUDSK*

*Yeah, lots of times. I think for the first 12 months when I was getting used to it, trying to figure out what it meant, trying to figure out how it was going to impact my future. And then when I went to see the neurologist and it seemed like every time I went to see him, he had another diagnosis for me. He was getting more, building up a extra long list of autoimmune conditions because at one stage he thought I had my as well. So I was getting all these extra, you know, diagnosis and. So those were some of the points and then when I started talking stem cell therapy and things that sounded really major and scary and whatever. So it was those kind of things when when I was a trigger point and when I was just getting used to the whole thing.*

*Participant 009\_2023AUDIS*

*Probably only at first with the first rheumatologist that I had...He didn't fully listen, just gave me the bad news, and just sent me on my way. [chuckles]*

*Participant 018\_2023AUDIS*

**Participant describes feeling vulnerable because of interactions with the medical team**

*The most vulnerable was that time I described being naked and being told that I need to quit smoking and lose weight and take Humira and then I'll be fine. I'm not sure they realise that. I left it on the opposite side of the city and it took me an hour and a half on public transport to get there. 30 minute walk. That's a lot of effort just for doctors and nurses to point their finger at you while you're naked and tell you you're the problem... That's why I've not seen a dermatologist since.*

*Participant 008\_2023AUDSK*

*Really. They treated me like I was infectious in hospital, so no one knew the their treatment of my pain level. They made me feel like I was just a heroin addict who who's come in to get some end down. So I cried so hard and then you know I followed that up with that visit, and it was just, it was a joke. I'd never felt more, I'd never felt more unseen. Talked at like I was not a person of any circumstance. I wasn't even poorly dressed, I'd have to say if you thought I was anything other than a professional at that point. And the way he spoke to me in the presence of others, the nurse at the end of it, because he was just taking so long showing off in front of his registrar, this nurse came in and literally looked at me and said, you've taken enough of his time. And I remember laughing so hard because I was literally sitting there just being poked and prodded that no one was talking to me.*

*You know, you regurgitated the lose weight, quit smoking. But wash yourself. And I remember just going 'You have no idea what I do' And I could be just, you know, do you want my skin to be red before you'd be satisfied that the amount of bleach I'm scrubbing into myself daily and all the while still feeling extremely dirty? It's really horrific. I've quit smoking for years and never saw a difference. I was not even overweight at that time. Like, there's no rhyme or reason at that point that I could say these are contributing factors.*

*Participant 015\_2023AUDSK*

*Right back at the start. Definitely. Right back at the start. Even when I was first diagnosed, I just felt like I wasn't heard and I wasn't understood. And every time I mean my first, the gastroenterologist, he always said if you've got any questions, if you've got any concerns, just ring up, we'll help you out. But I would ring up and I would get met with like this block of, you know, the doctor has just told us to reassure you that, you know, when you're anxious, your symptoms are worse and you've just got to breathe through it, find some meditations and calming apps and, you know, work on your anxiety and your symptoms will go away. And it was that real dismissive like. That blocker, like there was no access because I knew what I was feeling. And I know the difference between anxiety and like an actual physical problem. And I know that anxiety can lead to things like having difficulty swallowing and whatever else. But the reality is you can still swallow when you're anxious. It will still go down. When you have an airway flare up. Nothing that you do, no amount of meditation is going to make your esophagus open up.*

*Participant 78\_2023AUDIS*

**Participant describes feeling vulnerable experiencing side effects from treatment or symptoms from condition**

*Yeah, so I the vulnerability that I have, the disease come in my social and intimate life. So as I said, I have the disease in my breast area, which is very intimate area, so I find it hard to be intimate with my partner. Sometimes if I'm having a flare up, I don't want to get undressed. I don't want him to see me even though I know he doesn't care and he still loves me. But it's still hard for me to feel beautiful and wanted when I'm dealing with this. As well as just going out and going to the beach. Like I can't wear a bikini because then the disease is just kind'of on display. And I, you know, there was a time where I said, you know, like through it, like I'm going to wear a bikini. And then, you know,*

*I saw people looking at it and asking questions like what's on your chest? And yeah, it just sucks because I don't want people to point it out or look at it or anything like that and it just makes me feel embarrassed.*

*Participant 010\_2023AUDSK*

*Oh yes. When I'm in pain, I think. When I'm in a lot of pain and I'm brought to tears by it, I feel quite vulnerable.*

*Participant 013\_2023AUDIS*

*Yes, when I was in hospital the second time when it got really, really bad, it was very much so that no one understood what was going on, no one knew, and I was having spasm after spasm, seizure as well. It's almost like they didn't really believe me, which has been something my who'e life, but they were still willing to look after me, but the doctors hadn't passed on to the nurses, or whatever had happened, I don't know, but there was a communication miss and I ended up falling out of bed having a seizure and hitting my head on the floor, so they weren't looking after me properly, so that's definitely a vulnerable time I had.*

*Participant 004\_2023AUDNS*

**Participant describes feeling vulnerable thinking about disease course/incurable condition**

*I probably think the most vulnerable I think felt is when I was first diagnosed, I guess there was an element of fear of the unknown and what was going on and what did this look like and what did this mean for me moving forward? So that would probably, yeah.*

*Participant 001\_2023AUDSK*

*Yes, there has. Particularly when the condition was just getting out of control and you turn to a specialist for help and their answer is there's nothing I can do...and these are that, that they don't offer alternative suggestions. For instance, hang on, I'm going to call, I was having a chronic abscess that just would not heal. And the surgeon I saw just said there's nothing he can do anymore. And I but he didn't offer alternative solutions like go and see this person or what not. And that I still vividly remember being very vulnerable and alone in the world, thinking no one can help.*

*Participant 013\_2023AUDSK*

*In the sense that I've been going and seeing a particular specialist and blah and blah and blah, and*

*it feels like we're at the end of the line. There's no further that we can go because they're mandible jaw people, the fact that it's partly a muscle problem, they don't want to know about that. That's why I'm feeling a glimmer of hope with NAME's new guy, because he has actually got a plan, how much of it's muscular and how much of it is joint.*

*Participant 007\_2023AUDNS*

**Participant describes feeling vulnerable during/after treatments**

*Yeah, I so, I had a fairly major surgery...and I went back for what I thought was my final checkup with the breast surgeon and at which time he decided to continue the surgery in the chair. So I actually witnessed the whole thing take place in front of my face and I was, I don't know if you've ever seen if you get cut open, but it's not a very nice thing to look at. It kind of all collapses open. And yeah, it kind of looks like something you would see in a slaughter movie. And I was literally holding my own breast while he cut it in front of my face. So I felt quite vulnerable then when I returned to the hospital, probably about 18 months later, and I was obviously very reluctant to go back when I returned to the hospital 18 months later because I needed another surgery...and I explained it to the nursing staff. They then informed me because of COVID restrictions, your husband has to leave after one hour and he has to sit in the waiting room and wait for you, and I explained how I was feeling. I asked to speak to the nursing unit manager, who then threatened to kick my husband out of the hospital, who is like my only support person. Even after explaining how stressed and vulnerable I felt, and I actually got out of bed and left the hospital about 35 minutes after the surgery, I pulled my own cannula out and left the hospital because that's how distraught I was when they kicked him out of the room rather than just having a little bit of. And by the way, I was in a single room. There was no reason for him to have to leave. He wasn't affecting anybody.*

*Participant 005\_2023AUDSK*

*Oh yes, definitely in the first stages. I think again, when you don't understand what the medication's doing to you unless you've got somebody that you can call or go back to them immediately because sometimes you can't get in. That can be a bit difficult as well, so if you've got someone you can talk to, a good GP that's...My GP's very well-informed. I can book that and at least go to him and get some common knowledge and then they then try to then call the physician if they can or go from there.*

*Participant 001\_2023AUDIS*



*It's really hard to say because OK, well, let's say when she's been in heart having heart surgery. OK, that's what you're most vulnerable.*

*Participant 33\_2023AUDPA*

**Participant describes feeling vulnerable when feeling sick/unwell**

*In the beginning especially, and I was a lot younger back then, to be able to push for a diagnosis and actually tell them that I didn't feel what I thought was saying was correct. And definitely when I'm in the middle of a really awful flare up and you, you know, heading in to get help, I think that's when you're feeling the more when I'm feeling the most fragile because you're in so much pain, so.*

*Participant 022\_2023AUDSK*

*Yeah, when I've been very sick. I live by myself. Yeah. Earlier this year I had a pneumovirus and I was absolutely sure it was COVID. I was so sick and but the*

*tests had come back negative. And at the time I thought, you know, damn it, I'm pretty sick here. That was that was a bit scary. And you know, if that happens at night time when you go to lie down and you can't breathe lying down. I didn't go to hospital. I got better soon after that moment so I didn't have to go to hospital. That was actually just before my 70th birthday. And then three weeks later I went on a hike and did 17 K's in soft sand in one day, which was hell. But anyway, that's beside the point. So I did get over it. But those moments when I'm, I'm very sick and, you know, I don't feel like treatment for doing anything. And that was at the time when COVID was on a bit of a rant. Yeah, and I really didn't want to go near a hospital because it was all everyone had. COVID.*

*Participant 19\_2023AUDPA*

*When I was really sick and potentially couldn't advocate for myself?*

*Participant 31\_2023AUORC*

**Table 8.11: Experience of vulnerability**

Experience of vulnerability	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=180	%	n=67	%	n=31	%	n=9	%	n=32	%	n=9	%	n=32	%	n=100	%	n=80	%	n=121	%	n=57	%
Participant describes feeling vulnerable when having sensitive discussion (diagnosis, treatment decision)	30	16.67	10	14.93	11	35.48	2	22.22	4	12.50	0	0.00	3	9.38	22	22.00	8	10.00	23	19.01	7	12.28
Participant describes feeling vulnerable because of interactions with the medical team	26	14.44	5	7.46	10	32.26	3	33.33	6	18.75	0	0.00	2	6.25	22	22.00	4	5.00	23	19.01	3	5.26
Participant describes feeling vulnerable experiencing side effects from treatment or symptoms from condition	17	9.44	2	2.99	4	12.90	4	44.44	6	18.75	0	0.00	1	3.13	14	14.00	3	3.75	17	14.05	0	0.00
Participant describes feeling vulnerable thinking about disease course/incurable condition	15	8.33	2	2.99	6	19.35	3	33.33	2	6.25	0	0.00	2	6.25	12	12.00	3	3.75	11	9.09	3	5.26
Participant describes feeling vulnerable during/after treatments	12	6.67	2	2.99	2	6.45	0	0.00	3	9.38	0	0.00	5	15.63	9	9.00	3	3.75	8	6.61	4	7.02
Participant describes feeling vulnerable when feeling sick/unwell	10	5.56	3	4.48	3	9.68	0	0.00	1	3.13	1	11.11	2	6.25	7	7.00	3	3.75	9	7.44	1	1.75
Experience of vulnerability	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=180	%	n=61	%	n=63	%	n=37	%	n=19	%	n=88	%	n=92	%	n=48	%	n=132	%	n=94	%	n=86	%
Participant describes feeling vulnerable when having sensitive discussion (diagnosis, treatment decision)	30	16.67	7	11.48	8	12.70	12	32.43	3	15.79	12	13.64	18	19.57	8	16.67	22	16.67	18	19.15	12	13.95
Participant describes feeling vulnerable because of interactions with the medical team	26	14.44	3	4.92	7	11.11	9	24.32	7	36.84	15	17.05	11	11.96	4	8.33	22	16.67	12	12.77	14	16.28
Participant describes feeling vulnerable experiencing side effects from treatment or symptoms from condition	17	9.44	1	1.64	6	9.52	6	16.22	4	21.05	8	9.09	9	9.78	6	12.50	11	8.33	10	10.64	7	8.14
Participant describes feeling vulnerable thinking about disease course/incurable condition	15	8.33	3	4.92	1	1.59	7	18.92	4	21.05	4	4.55	11	11.96	6	12.50	9	6.82	9	9.57	6	6.98
Participant describes feeling vulnerable during/after treatments	12	6.67	2	3.28	5	7.94	4	10.81	1	5.26	7	7.95	5	5.43	5	10.42	7	5.30	8	8.51	4	4.65
Participant describes feeling vulnerable when feeling sick/unwell	10	5.56	2	3.28	4	6.35	2	5.41	2	10.53	5	5.68	5	5.43	4	8.33	6	4.55	7	7.45	3	3.49

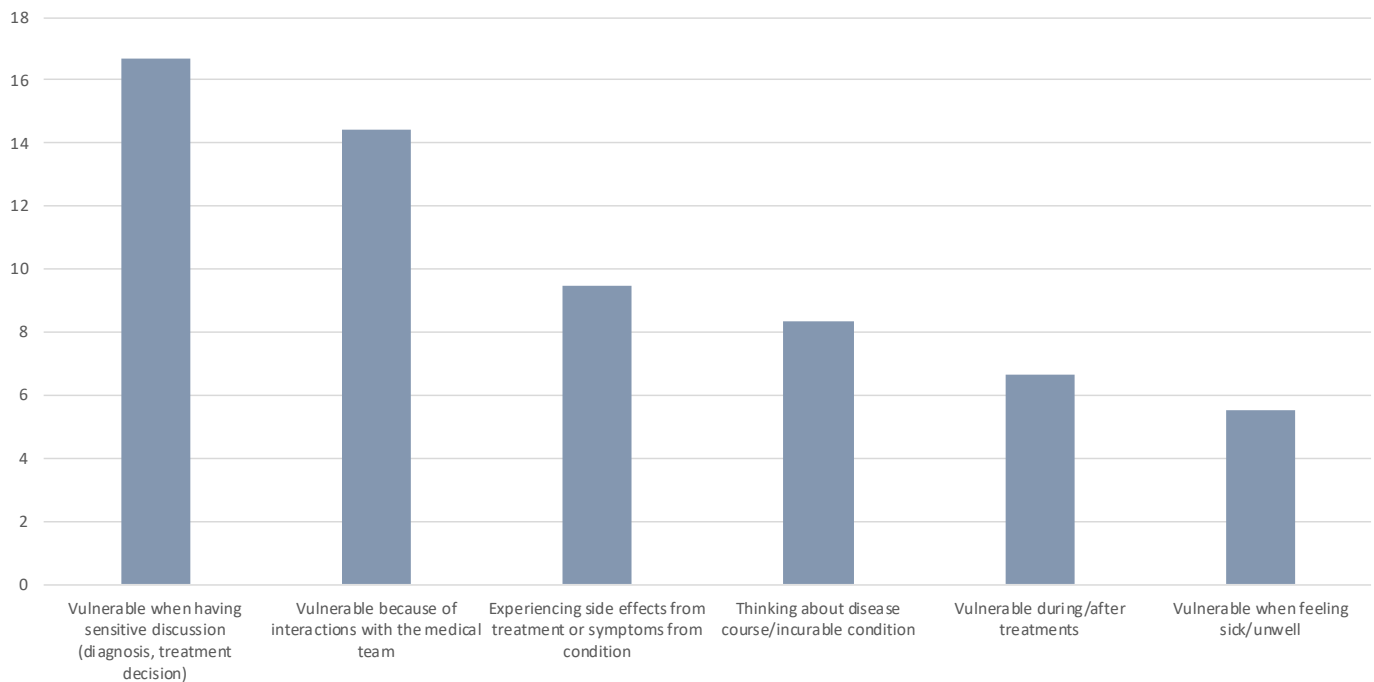


Figure 8.6: Experience of vulnerability

Table 8.12: Experience of vulnerability – subgroup variations

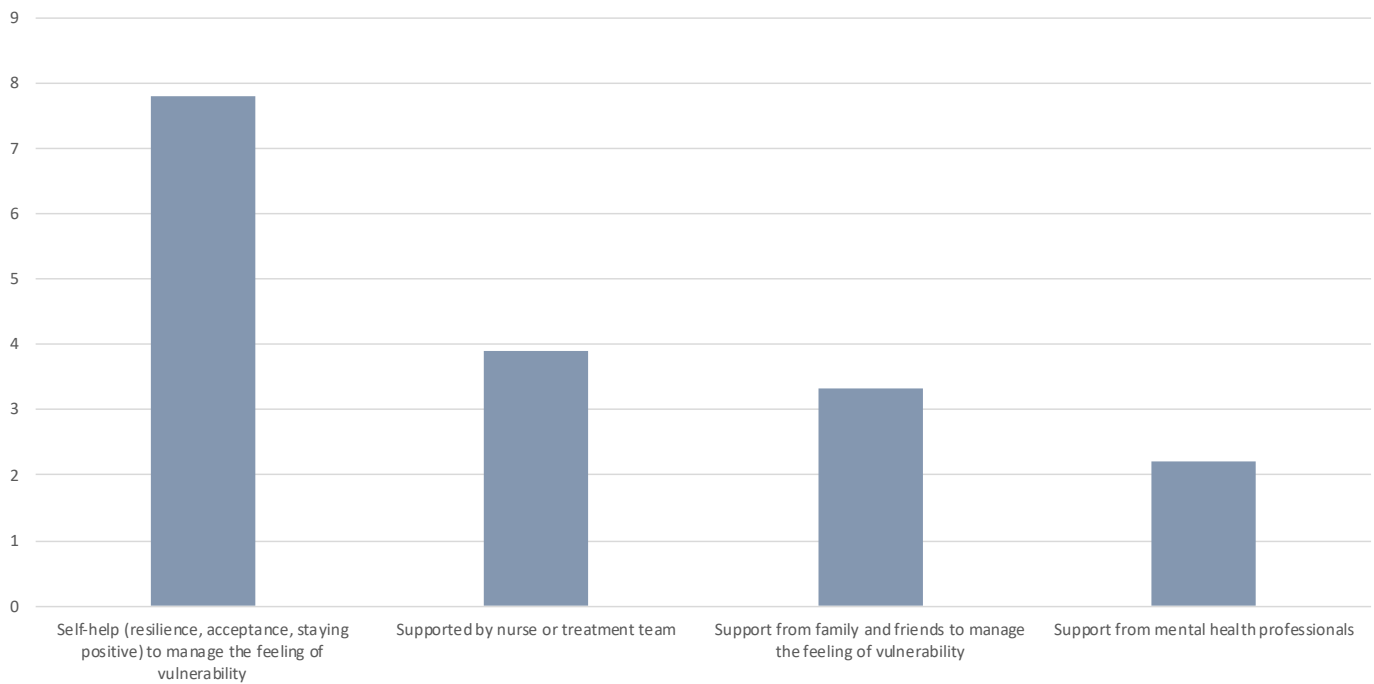
Experience of vulnerability	Reported less frequently	Reported more frequently
Participant describes feeling vulnerable when having sensitive discussion (diagnosis, treatment decision)	Endocrine, nutritional or metabolic diseases	Diseases of the immune system Aged 45 to 64
Participant describes feeling vulnerable because of interactions with the medical team	Endocrine, nutritional or metabolic diseases	Diseases of the immune system Diseases of the nervous system Aged 65 plus
Participant describes feeling vulnerable experiencing side effects from treatment or symptoms from condition	-	Diseases of the nervous system Aged 65 plus
Participant describes feeling vulnerable thinking about disease course/incurable condition	-	Diseases of the immune system Diseases of the nervous system Aged 45 to 64 Aged 65 plus

Table 8.13: Experience of vulnerability (details)

Methods to manage vulnerability	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=180	%	n=67	%	n=31	%	n=9	%	n=32	%	n=9	%	n=32	%	n=100	%	n=80	%	n=121	%	n=57	%
Participant describes using self-help methods (resilience, acceptance, staying positive) to manage the feeling of vulnerability	14	7.78	2	2.99	6	19.35	2	22.22	2	6.25	0	0.00	2	6.25	13	13.00	1	1.25	12	9.92	1	1.75
Participant describes support from nurse or treatment team to manage the feeling of vulnerability	7	3.89	0	0.00	4	12.90	1	11.11	0	0.00	0	0.00	2	6.25	5	5.00	2	2.50	5	4.13	1	1.75
Participant describes getting support from family and friends to manage the feeling of vulnerability	6	3.33	0	0.00	1	3.23	0	0.00	3	9.38	1	11.11	1	3.13	5	5.00	1	1.25	4	3.31	2	3.51
Participant describes support from mental health professionals to manage the feeling of vulnerability	4	2.22	2	2.99	1	3.23	0	0.00	1	3.13	0	0.00	0	0.00	2	2.00	2	2.50	3	2.48	1	1.75

Methods to manage vulnerability	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=180	%	n=61	%	n=63	%	n=37	%	n=19	%	n=88	%	n=92	%	n=48	%	n=132	%	n=94	%	n=86	%
Participant describes using self-help methods (resilience, acceptance, staying positive) to manage the feeling of vulnerability	14	7.78	1	1.64	2	3.17	6	16.22	5	26.32	9	10.23	5	5.43	4	8.33	10	7.58	8	8.51	6	6.98
Participant describes support from nurse or treatment team to manage the feeling of vulnerability	7	3.89	2	3.28	0	0.00	3	8.11	2	10.53	2	2.27	5	5.43	2	4.17	5	3.79	3	3.19	4	4.65
Participant describes getting support from family and friends to manage the feeling of vulnerability	6	3.33	0	0.00	4	6.35	2	5.41	0	0.00	4	4.55	2	2.17	0	0.00	6	4.55	5	5.32	1	1.16
Participant describes support from mental health professionals to manage the feeling of vulnerability	4	2.22	0	0.00	2	3.17	2	5.41	0	0.00	3	3.41	1	1.09	0	0.00	4	3.03	2	2.13	2	2.33



**Figure 8.8: Experience of vulnerability (details)**

**Table 8.14: Experience of vulnerability (details) – subgroup variations**

Methods to manage vulnerability	Reported less frequently	Reported more frequently
Participant describes using self-help methods (resilience, acceptance, staying positive) to manage the feeling of vulnerability	-	Diseases of the immune system Diseases of the nervous system Aged 65 plus

## Impact on relationships

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

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

The most common reasons for a positive impact on relationships was that people were supportive and well-meaning (15.67%)

**Participant describes that overall, there was a negative impact on relationships**

*Yes, yes. Especially with my friends. I wouldn't say the same about my family, but with my friends, a lot of things have been, you know, affected and a lot of*

*things have changed. I don't associate much and I tend to shy away from certain activity, which I always engage with my friends, you know, like hanging out and taking drinks or drinking, and all these have been limited and, you know, caught short by the situation.*  
**Participant 006\_2023AUORC**

*The only questions that got asked at the one of my face, oh my gosh, I hated going to Christmas. What's on your face? And it's like, Oh my gosh, I don't teach you about your conditions. Yeah. Like, half my family has psoriasis. So, I mean, they think they're worse off and they've got growing skin all the time. But I'm like...and you don't have any sympathy for me. Yeah.*  
**Participant 003\_2023AUDSK**

*Yeah, I guess I would say it has. Because of making plans and having to cancel because you can't really do the things that you wanted to do. Yeah, I guess it does. And when people don't know why, you'll understand. It makes it difficult because then they just think that you're blowing them off for no reason and it's like, well, no.*  
**Participant 014\_2023AUDSK**

*Yes. Yeah. I think it's just, I guess it's more because it's like invisible. I guess you can't tell unless you, you know what's going on inside the household. It's not a visible disability. And I think that that can be part of the problem in that family can be in denial or think that things will change and she'll get better and yes, and that's just how to fix us.*

*Participant 16\_2023AUORC*

**Participant describes that overall, there no impact on relationships**

*They have to love me because they always will, no matter what. And if they didn't do this because they wouldn't be my friends.*

*Participant 002\_2023AUDSK*

*Probably not. In my case, I have chronic fatigue syndrome, so I've already quite isolated and yeah, sort of focusing on just my main friendship.*

*Participant 019\_2023AUDSK*

*No, not really, because they don't know about it. You know, it doesn't. I don't talk about it and I'm pretty good at hiding my emotions most of the time. A fairly happily happy person, outwardly.*

*Participant 024\_2023AUDSK*

*No, I don't think so. No, because everybody who knows her just knows knows her the way she is. So I don't think it's affected anything. No.*

*Participant 27\_2023AUDPA*

**Participant describes that overall, there was an impact on relationships that was both positive and negative**

*There are some things like I won't that I won't do so like I won't go to things like water parks and so there's there's some family things that I won't go to. Obviously there are lots of times I have to cancel because as I said pants are not an option and they kind of required when you leave the house. Very fortunate with regards to. So I've had like I've lost a lot of friendships over the years because you cancel all the time and then they just stop asking you to do things with them. I'm very fortunate in that, you know, both my first husband and my second husband are beautiful men. My first husband is still a beautiful man and, and so it didn't affect my physical relationships. But certainly had an impact on my ability to spend time with friends and things like that.*

*Participant 005\_2023AUDSK*

*Yes, I think that people who don't have chronic illnesses don't genuinely understand how bad it is, and the people who do like you understand each other in a deeper way. But yeah, it impacts it. My closest friends, I don't feel bad about being like, I'm not up for it. I'm not coming out today, it's just not a 'me' day. And they will absolutely say yes, but with people who aren't the closest of friends, I think a lot of them think I've become very flaky over the years, but I don't really care because I don't need to explain myself. Slash if you know. If you don't know me well enough to know that I have HS, then I'm not really losing that but I do feel like there has been a bit of a roll your eyes at me sometimes and I'm OK with that.*

*Participant 026\_2023AUDSK*

*To some extent it has. But I think now looking back, it's more my mental health that's been affected and and affects the social interactions. But I mean, you know, not, I wouldn't say or like greatly because our friends, our close friends and family understand and we've educated them about it and stuff. So they're pretty accepting.*

*Participant 14\_2023AUDPA*

*Yes and no. It's made my partner and I get closer because he's had to be there for me a lot more. We were close anyway, but in that way, it's been really good for us. I have a few friends overseas. I have one specific friend who I've distanced myself from because she's all high and mighty studying psychology and she's telling me that it's in my head, all that stuff, and she has no idea what's actually going on because I haven't even seen her physically. Everyone has their opinions about it but she's probably the only one thus far that has really pushed it out too far.*

*Participant 004\_2023AUDNS*

**Participant describes that overall, there was a positive impact on relationships**

*No, I don't think so. If anything, it's made it better. I'm quite open about it. I like to tell everyone I meet about it because no one talks about it so.*

*Participant 027\_2023AUDSK*

*No, not at this stage. I think they're probably bought my family, probably closer. You know, thinking that maybe maybe I haven't spent enough time with my family over the years. And, you know, it's a good thing that now I'm bloody pulling my finger out and catching up with my family a bit more and yeah, probably probably bringing bringing us closer kind of thing. So, yeah.*

*Participant 24\_2023AUORC*

*Not in a negative way. Probably in a positive way that people see...We're from a small community, so a lot of people know...When they're walking down the street and word of mouth and stuff. People know how hard we have it and all the stuff that we go through. I think people appreciate and admire how strong and together we are. I think a lot of family love CHILD'S NAME that little bit more because she is the first grandchild of the family. I think because she's so special they want to protect her. Even my close girlfriends, she was treated differently like she was very well loved. Even at her school. People want to look after her and she's not just a normal child. I think she holds a place in people's hearts of everything that she's been through. She's the sweetest little thing. You would never think there's anything wrong with her. I think she feels that little bit more loved. That's all I think anyway. From other people. Yes.*

*Participant 43\_2023AUDPA*

*I think especially for the family. That's what we thought before. Not just him, his character that had actually had nothing to do with his character, but that it was actually signed in his behaviour related to his condition. You educate the other family members to be more acceptive of what is different and learn them to understand this is not his character but it's part of the TSC. The same, I think with school. There was that one thing, once he was diagnosed, that I actually had a reason to go and sit down with the school and say, "Hey, he is because this, this and this and if we can do this, this and this, have little breaks in between, it'll make life so much easier." Has it changed the relationships? Yes, but I think in a positive way by educating all those around him to be more accepting of who he is.*

*Participant 49\_2023AUDPA*

*Participant describes that overall, there was an impact on relationships that was neither positive nor negative*

*It's different but I'm very lucky in that I've got a very, very supportive family. I think it's different, but it's not better or worse.*

*Participant 017\_2023AUDIS*

*In some ways, with family, with my one of my brothers it has in that when we first got diagnosed I actually let my brothers know that we've been diagnosed with it and my brother, who, actually had, I actually suggested he go and get checked out and he turned and just now the insurance would go through the roof. I told the insurance would bother him [unintelligible], it's the health.*

*Participant 005\_2023AUDPA*

*It's different but I'm very lucky in that I've got a very, very supportive family. I think it's different, but it's not better or worse.*

*Participant 017\_2023AUDIS*

*So yeah, I think, well, I think my family are amazing, but then sometimes like treat him a little bit differently to the other kids in the family. And I think that kid's lovely. But when it comes, when it comes to a choice of hanging out with someone you can understand versus someone you can't, it's a bit easier for them. And I don't blame them. It's a bit easier for them to hang out with someone or engage with someone that they can understand. So yes, I believe it affects him.*

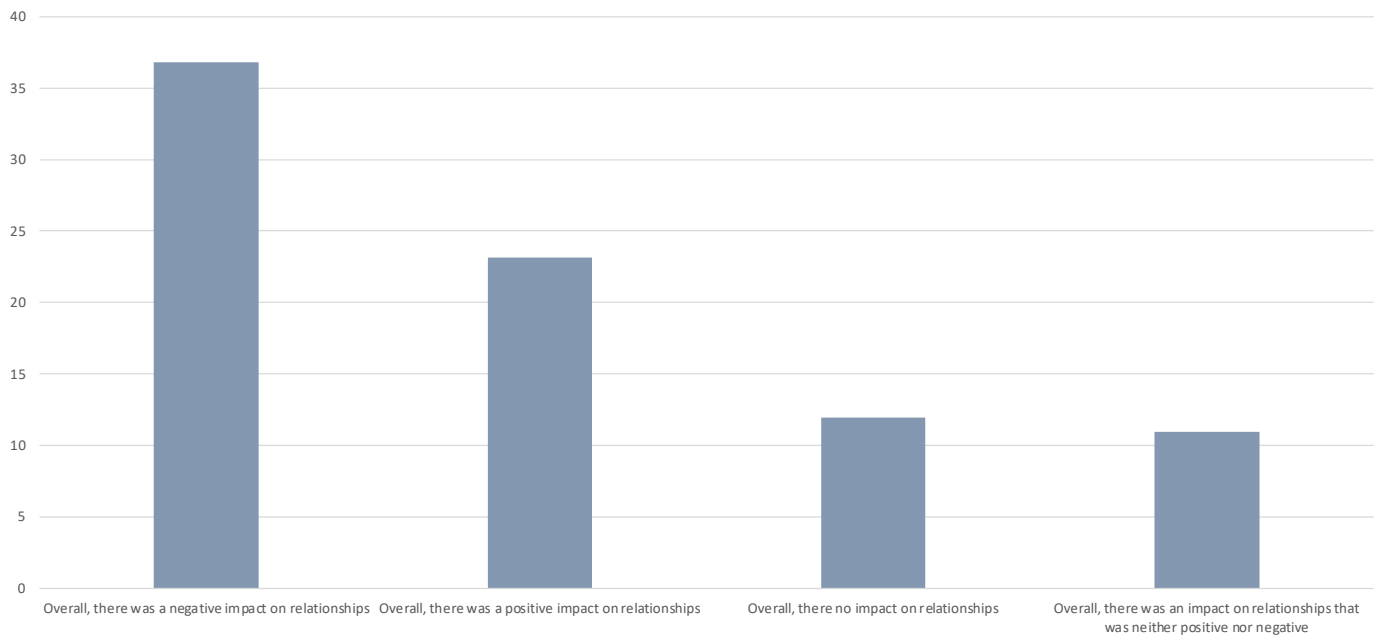
*Participant 23\_2023AUDPA*

**Table 8.15: Impact on relationships**

Impact on relationships	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Overall, there was a negative impact on relationships	148	36.82	11	16.42	37	45.68	47	49.47	5	15.63	43	45.26	5	15.63	106	39.55	42	31.34	120	40.82	28	26.42
Overall, there was a positive impact on relationships	93	23.13	21	31.34	16	19.75	17	17.89	13	40.63	12	12.63	14	43.75	56	20.90	37	27.61	66	22.45	25	23.58
Overall, there no impact on relationships	48	11.94	4	5.97	9	11.11	12	12.63	5	15.63	12	12.63	6	18.75	39	14.55	9	6.72	36	12.24	12	11.32
Overall, there was an impact on relationships that was neither positive nor negative	44	10.95	16	23.88	10	12.35	5	5.26	2	6.25	8	8.42	3	9.38	23	8.58	21	15.67	23	7.82	21	19.81
Overall, there was an impact on relationships that was both positive and negative	25	6.22	5	7.46	0	0.00	8	8.42	3	9.38	5	5.26	4	12.50	15	5.60	10	7.46	19	6.46	6	5.66

Impact on relationships	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Overall, there was a negative impact on relationships	148	36.82	34	35.05	50	38.17	48	42.11	16	26.67	73	36.87	73	37.24	44	39.64	104	35.74	80	40.00	68	33.66
Overall, there was a positive impact on relationships	93	23.13	26	26.80	26	19.85	25	21.93	16	26.67	46	23.23	45	22.96	26	23.42	67	23.02	47	23.50	46	22.77
Overall, there no impact on relationships	48	11.94	5	5.15	20	15.27	15	13.16	8	13.33	26	13.13	21	10.71	9	8.11	39	13.40	23	11.50	25	12.38
Overall, there was an impact on relationships that was neither positive nor negative	44	10.95	16	16.49	6	4.58	14	12.28	8	13.33	26	13.13	18	9.18	11	9.91	33	11.34	19	9.50	25	12.38
Overall, there was an impact on relationships that was both positive and negative	25	6.22	9	9.28	12	9.16	2	1.75	2	3.33	10	5.05	15	7.65	9	8.11	16	5.50	10	5.00	15	7.43



**Figure 8.9: Impact on relationships**

**Table 8.16: Impact on relationships – subgroup variations**

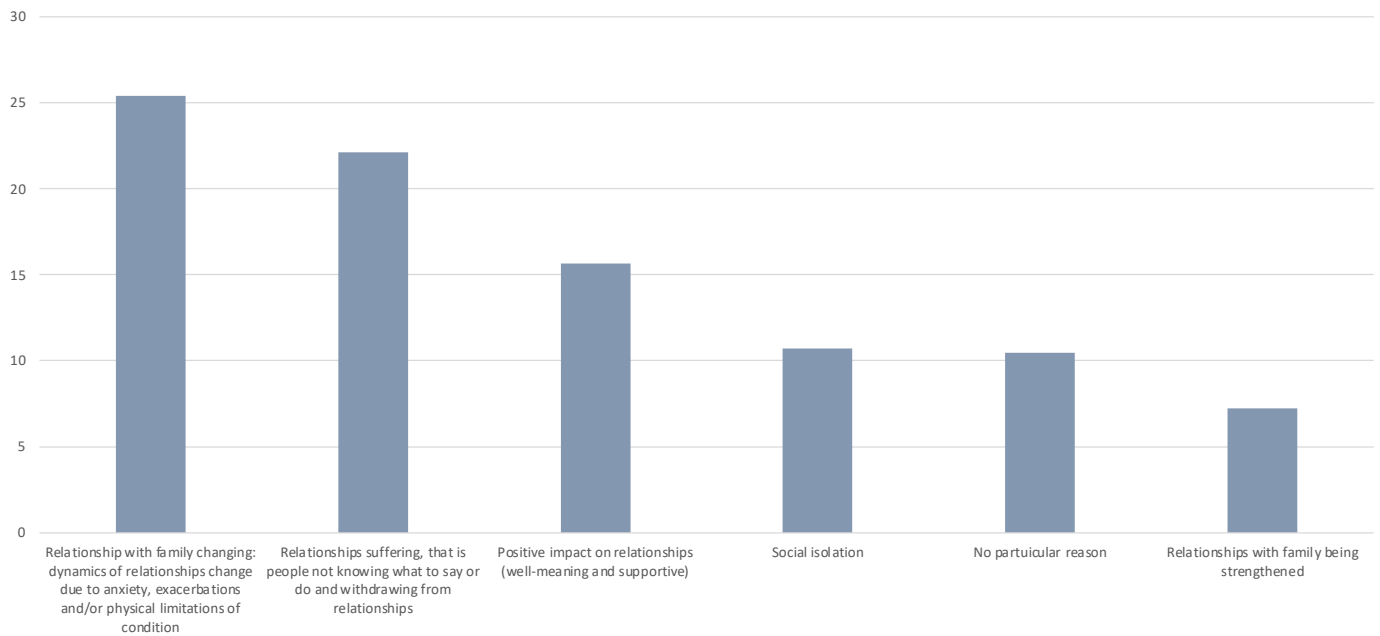
Impact on relationships	Reported less frequently	Reported more frequently
Overall, there was a negative impact on relationships	Developmental anomalies Diseases of the skin Other rare condition Male Aged 65 plus	
	Endocrine, nutritional or metabolic diseases	Diseases of the nervous system Diseases of the skin Other rare condition
Overall, there was a positive impact on relationships		
Overall, there no impact on relationships		Developmental anomalies
Overall, there was an impact on relationships that was neither positive nor negative		
Overall, there was an impact on relationships that was both positive and negative		Regional or remote

**Table 8.17: Impact on relationships (Reason for impact)**

Impact on relationships (reasons)	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Relationship with family changing: dynamics of relationships change due to anxiety, exacerbations and/or physical limitations of condition	102	25.37	22	32.84	26	32.10	22	23.16	6	18.75	19	20.00	7	21.88	63	23.51	39	29.10	78	26.53	23	21.70
Relationships suffering, that is people not knowing what to say or do and withdrawing from relationships	89	22.14	11	16.42	22	27.16	29	30.53	4	12.50	20	21.05	3	9.38	55	20.52	34	25.37	65	22.11	24	22.64
Positive impact on relationships (well-meaning and supportive)	63	15.67	22	32.84	11	13.58	17	17.89	3	9.38	8	8.42	2	6.25	28	10.45	35	26.12	42	14.29	21	19.81
Social isolation	43	10.70	0	0.00	17	20.99	8	8.42	0	0.00	18	18.95	0	0.00	40	14.93	3	2.24	38	12.93	5	4.72
No particular reason	42	10.45	11	16.42	4	4.94	16	16.84	5	15.63	2	2.11	4	12.50	23	8.58	19	14.18	32	10.88	10	9.43
Relationships with family being strengthened	29	7.21	6	8.96	2	2.47	6	6.32	1	3.13	9	9.47	5	15.63	16	5.97	13	9.70	19	6.46	10	9.43

Impact on relationships (reasons)	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Relationship with family changing: dynamics of relationships change due to anxiety, exacerbations and/or physical limitations of condition	102	25.37	29	29.90	24	18.32	34	29.82	15	25.00	51	25.76	51	26.02	32	28.83	70	24.05	53	26.50	41	20.30
Relationships suffering, that is people not knowing what to say or do and withdrawing from relationships	89	22.14	24	24.74	35	26.72	26	22.81	4	6.67	45	22.73	43	21.94	26	23.42	63	21.65	46	23.00	39	19.31
Positive impact on relationships (well-meaning and supportive)	63	15.67	28	28.87	20	15.27	8	7.02	7	11.67	29	14.65	34	17.35	18	16.22	45	15.46	29	14.50	44	21.78
Social isolation	43	10.70	2	2.06	18	13.74	17	14.91	6	10.00	27	13.64	16	8.16	11	9.91	32	11.00	19	9.50	24	11.88
No particular reason	42	10.45	14	14.43	14	10.69	10	8.77	4	6.67	14	7.07	27	13.78	11	9.91	31	10.65	23	11.50	28	13.86
Relationships with family being strengthened	29	7.21	9	9.28	6	4.58	8	7.02	6	10.00	10	5.05	17	8.67	10	9.01	19	6.53	11	5.50	7	3.47



**Figure 8.10: Impact on relationships**

**Table 8.18: Impact on relationships: Reason for impact – subgroup variations**

Impact on relationships (reasons)	Reported less frequently	Reported more frequently
Relationship with family changing: dynamics of relationships change due to anxiety, exacerbations and/or physical limitations of condition		
Relationships suffering, that is people not knowing what to say or do and withdrawing from relationships	Other rare condition Aged 65 plus	
Positive impact on relationships (well-meaning and supportive)		Developmental anomalies Family or carer Aged under 18
Social isolation	Developmental anomalies Diseases of the skin Other rare condition	
No particular reason		Diseases of the immune system
Relationships with family being strengthened		

## Burden on family

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

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

**Participant describes that overall, there was a burden on their family**

*My 15 year old daughter has missed out on having her mum because her mum has been trying to keep her*

*little sister alive for nine years. It's affected, you know, I don't have many friends. I don't go out very often. You know, it's affected our entire lives. And NAME's particularly, you know, I would never label her a burden because she's, you know, a blessing. But it's the burden that she is unable to enjoy, the things that she would like to enjoy. She's unable to go to the places that she wants to go to because her disability prevents her from enjoying.*

*Participant 80\_2023AUDIS*

*Yeah, I think so. I mean, it's better as the kids have gotten older, but you know, when they were younger they had to do lots of other things that they probably wouldn't have done otherwise, like learning how to tube feed and having emergency protocols stashed left, right and center in case the kids were sick, so. Yeah, I do feel like it's, I don't know if I don't know what the word burden is actually implying, whether it's like a negative thing or just additional load, but it's. Yeah, additional load, I would say all.*

*Participant 21\_2023AUORC*

No, the system's a burden, my son is not a burden. The system every, every corner of the system is a burden. No, absolutely not. What I say my son is a burden or or or charged in terms of burden. It's the system, it's the hurdles, it's the challenges, it's the inner, it's the gap, you know, it's the lack of services, it's everything is. It's like a research. It's lack of experts. All of those are, you know, what makes having CHARGE syndrome a burden on my family.

Participant 28\_2023AUORC

I don't think so. I don't tend to rely on them a lot. Extra care assistance, I think they're just. Provide more amounts of care occasionally, which they would do for any child, many grandchild or nephew or whoever I'm seeking that from, so I don't think so, no.

Participant 89\_2023AUENM

Participant describes that overall, there was not a burden on their family

No, I wouldn't say so. I guess I'm usually the carer, unfortunately. So, yeah.

Participant 014\_2023AUDSK

No, I don't think so. No.

Participant 027\_2023AUDSK

Them no. I'm fiercely independent with it. I have to be in a really bad way before I ask anyone for help. So you know, I'll get a taxi to hospital rather than ask my son to come and pick me up. And he would do it at a drop of a hat. But I'm very conscious of not wanting to be the single old lady, old lady living alone, needing help. You know, the needy other person. I don't want to be that. So I'll hang on doing whatever I can for myself for as long as I can. Yeah, okay. Nothing. Part of that keeps me well, get up, dress up, show up. Half

the time now you might wake up feeling rotten, and if you stayed home, you'd feel rotten. Get up, dress up, show up and get out there. Half the time you feel better by the time you get out.

Participant 18\_2023AUORC

Participant describes that overall, there was not a burden on their family now but they anticipate this will change in the future

At the moment we don't need a lot of carer assistance. I think as I get older I will, but the closest one's an hour away and then the other ones, he's he's about two hours away and the one in the middle is about an hour and a half. So you know they're, I don't need a lot. We don't need a lot of help from them yet.

Participant 024\_2023AUDSK

It probably will be. If I don't get any better than where I'm at now, but everything's suggesting to me that I I can get better. So look if I'm stuck at this point, yes it will affect them fairly heavily. If I get past this which I think I'm gonna it. It's burning always up a fair bit, you know? Yeah.

Participant 14\_2023AUORC

No not at this stage. Well not that I would say you know they haven't really sort of I don't think they've changed their life as much so but I'm not saying that later down the track they I may be a bit of a burden on them but early days I haven't had any sort of issues things sort of they've been pretty good that sort of set off you know if we can do anything for you let us know but. I don't think anything's changed at this stage, so yeah.

Participant 24\_2023AUORC

Table 8.19: Burden on family

Burden on family	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=352	%	n=67	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=247	%	n=105	%	n=252	%	n=98	%
Overall, there was a burden on their family	220	62.50	48	71.64	56	69.14	24	53.33	15	48.39	55	57.89	22	70.97	155	62.75	65	61.90	152	60.32	67	68.37
Overall, there was not a burden on their family now but they anticipate this will change in the future	15	4.26	1	1.49	2	2.47	2	4.44	1	3.23	6	6.32	3	9.68	10	4.05	5	4.76	12	4.76	2	2.04
Overall, there was not a burden on their family	74	21.02	13	19.40	16	19.75	9	20.00	9	29.03	23	24.21	4	12.90	51	20.65	23	21.90	53	21.03	21	21.43

Burden on family	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=69	%	n=116	%	n=108	%	n=59	%	n=172	%	n=172	%	n=100	%	n=252	%	n=176	%	n=176	%
Overall, there was a burden on their family	220	62.50	43	62.32	76	65.52	66	61.11	35	59.32	111	64.53	105	61.05	63	78.75	157	62.30	108	61.36	112	63.64
Overall, there was not a burden on their family now but they anticipate this will change in the future	15	4.26	3	4.35	7	6.03	2	1.85	3	5.08	8	4.65	7	4.07	3	3.75	12	4.76	4	2.27	11	6.25
Overall, there was not a burden on their family	74	21.02	14	20.29	22	18.97	25	23.15	13	22.03	37	21.51	36	20.93	22	27.50	52	20.63	42	23.86	32	18.18



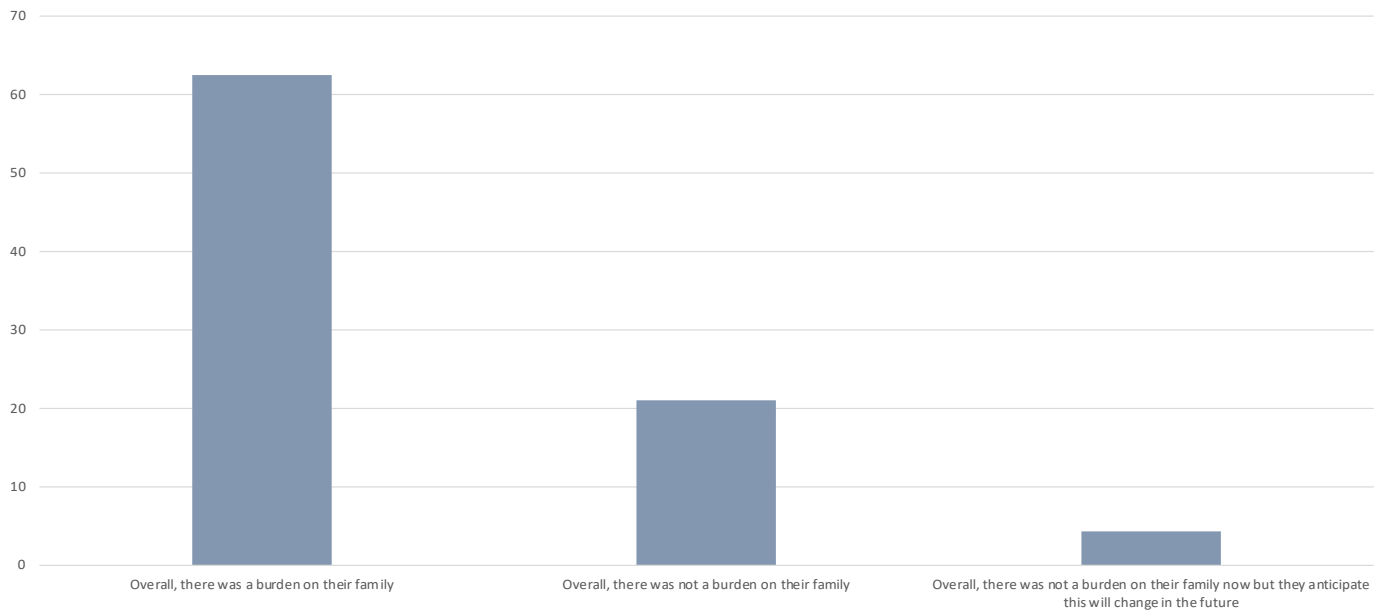


Figure 8.11: Burden on family

Table 8.20: Burden on family – subgroup variations

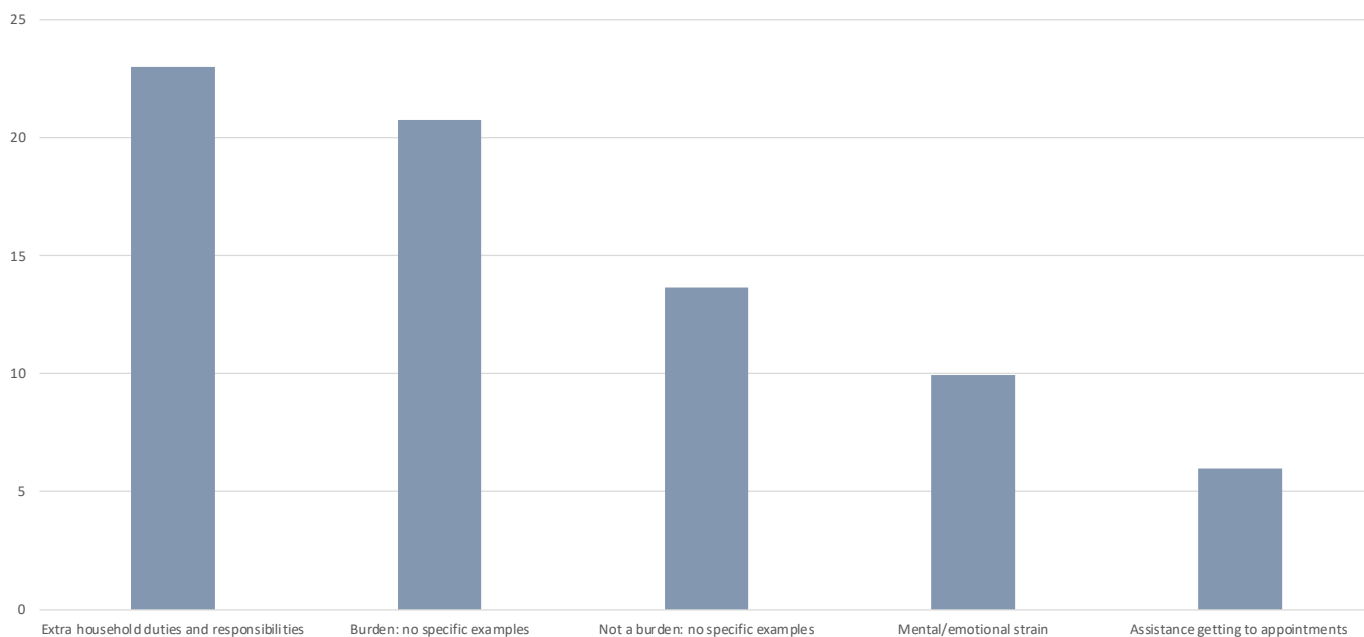
Burden on family	Reported less frequently	Reported more frequently
Overall, there was a burden on their family	Diseases of the skin	Regional or remote

Table 8.21: Burden on family (description)

Burden on family (description)	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=352	%	n=67	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=247	%	n=105	%	n=252	%	n=98	%
Extra household duties and responsibilities	81	23.01	21	31.34	21	25.93	7	15.56	5	16.13	23	24.21	4	12.90	53	21.46	28	26.67	57	22.62	24	24.49
Burden: no specific examples	73	20.74	11	16.42	21	25.93	7	15.56	5	16.13	18	18.95	11	35.48	54	21.86	19	18.10	50	19.84	22	22.45
Not a burden: no specific examples	48	13.64	8	11.94	12	14.81	8	17.78	4	12.90	16	16.84	0	0.00	34	13.77	14	13.33	35	13.89	13	13.27
Mental/emotional strain	35	9.94	11	16.42	2	2.47	3	6.67	3	9.68	10	10.53	6	19.35	20	8.10	15	14.29	22	8.73	13	13.27
Assistance getting to appointments	21	5.97	5	7.46	1	1.23	5	11.11	1	3.23	9	9.47	0	0.00	15	6.07	6	5.71	16	6.35	4	4.08
Temporary or only during treatment	13	3.69	2	2.99	1	1.23	1	2.22	3	9.68	0	0.00	6	19.35	10	4.05	3	2.86	8	3.17	5	5.10

Burden on family (description)	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=69	%	n=116	%	n=108	%	n=59	%	n=172	%	n=172	%	n=100	%	n=252	%	n=176	%	n=176	%
Extra household duties and responsibilities	81	23.01	21	30.43	23	19.83	24	22.22	13	22.03	38	22.09	42	24.42	23	28.75	58	23.02	31	17.61	50	28.41
Burden: no specific examples	73	20.74	13	18.84	26	22.41	19	17.59	15	25.42	37	21.51	35	20.35	21	26.25	52	20.63	43	24.43	30	17.05
Not a burden: no specific examples	48	13.64	9	13.04	13	11.21	14	12.96	12	20.34	18	10.47	29	16.86	17	21.25	31	12.30	29	16.48	19	10.80
Mental/emotional strain	35	9.94	9	13.04	13	11.21	5	4.63	8	13.56	15	8.72	18	10.47	10	12.50	25	9.92	12	6.82	23	13.07
Assistance getting to appointments	21	5.97	4	5.80	8	6.90	8	7.41	1	1.69	9	5.23	12	6.98	10	12.50	11	4.37	13	7.39	8	4.55
Temporary or only during treatment	13	3.69	1	1.45	7	6.03	4	3.70	1	1.69	6	3.49	7	4.07	4	5.00	9	3.57	4	2.27	9	5.11



**Figure 8.12: Burden on family (description)**

Burden on family (description)	Reported less frequently	Reported more frequently
Extra household duties and responsibilities	Other rare condition	
Burden: no specific examples		Other rare condition
Not a burden: no specific examples	Other rare condition	
Mental/emotional strain		
Assistance getting to appointments		
Temporary or only during treatment		Other rare condition

**Table 8.22: Burden on family (description)– subgroup variations**

### Cost considerations

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

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

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

#### Cost burden in relation to needing to take time off work

*So I think pretty much everything has been covered by Medicare or the NDIS. So in terms of accessing treatment any of the medical treatments or therapies that hasn't been usually costly but my income has been affected because instead of going back to work full time as I'd intended after his birth, I only ended up going back to work 2 days a week, just because it's quite difficult for one person to manage him. So say, if I left him with my partner five days a week while I worked, it's...that's quite a lot for him to manage and only recently has he been able to start childcare. We've been able to organise that just because of his additional need, so finances were affected in terms of*

*my income was greatly reduced, even though I didn't have extra costs with his therapies or medical treatments.*

*Participant 89\_2023AUENM*

*There was definitely increased costs at birth because she was in hospital for such a large length of time. And she wasn't in private hospitals, so we had to pay out money for that, obviously and the private pediatrician that was, she was saying while she was in there. As I said though, she did gain NDIS funding. I'm gonna say early primary school. I can't really remember how old she was. So we've had that for a number of years to cover the cost of therapies and stuff going forward. So that's been very helpful. There would be, there would be probably a large chunk of time that I've personally had to take off from work to be home with her when she's unwell because she does, she doesn't get sick a lot, but when she does it tends to linger. So there are, you know, large links of time where, yeah, I've probably had to take a week off here and there to be at home with her. Yeah.*

*Participant 27\_2023AUDPA*

*My experience is I'm very lucky that I have a dad who's still alive with some money. It's been very expensive. Yes. Yeah. I've had to stop working. I had to give up work.*

*Participant 002\_2023AUDNS*

#### **Cost burden in relation to the cost of treatments (including repeat scripts)**

*It hasn't been too bad in the last two years since I got a higher paying job, but before that I pretty much just was paying bills and everything, week to week, and then my medical expenses came up, but I would rather just. I'd often go without medication because I couldn't afford it because of the amount of medications that we have to take. And then when it came to being in hospital with work, it was just pretty much borrowing money from my mom to cover bills and whatnot until I could go back to work, or until I could commit to the doctors to let me go to work while I was in there.*

*Participant 13\_2023AUORC*

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

*accommodation and things like that, going down to specialist appointments, I find very expensive as well. Time-wise, definitely it takes up way too much family time with conversations and just their support*

*Participant 014\_2023AUDIS*

*Well, the surgery cost a fortune, but I mean, we were in that position of being able to afford it. My husband just said no, no, it's worth doing. The focused ultrasound would have been at least twice as much, which I didn't have. So, I'm fortunate that I could afford that. And as I said, my neurologist is no longer charging me, so I.*

*Participant 003\_2023AUDNS*

*It's been pretty significant by the time you add up all the, the emergency room visits, the medications, the different diagnostic tests like halter monitors and echocardiograms and things like that. So even though we had private health, the out of pocket's been pretty, pretty significant, yeah. And then the ablations and things like that as well.*

*Participant 32\_2023AUORC*

#### **Cost burden in relation to the cost specialist appointments**

*Yeah, so I did find, I did find the costs were substantial for me on a on a single income the you know this. And as a single parent, there's not a lot of extra. So the yeah...the constant outlay for medications, none of that stuff was like, I don't have a health care card or anything like that. So nothing like that was subsidized and then yeah. Seeing all your specialists, there was always a, a significant gap to pay and then I saw as I was lucky that I've been in my job for, for a while. So you know over the when I had the surgeries pretty much the six weeks that I needed to take off 3 after surgery I was able to absorb that through my sick leave. I was probably lucky in that sense.*

*Participant 007\_2023AUDSK*

*Definitely a cost of even just to see a specialist. You might get a rebate back, but it's still quite expensive to go see the specialist. In the first place. Which is the reason why I haven't gone back and in there, cost of medication as well. Like it's, it's certainly not cheap.*

*Participant 011\_2023AUDSK*

*It has put me into extreme financial hardship on every possible level and I find I'm often declining appointments, even specialist appointments. I've had to postpone multiple times because I just can't afford them, which is really disheartening when you wait six months to get in and then you can't afford to go.*

### *Participant 016\_2023AUDIS*

#### **Cost burden in relation to diagnostic tests and scans**

*The major cost is because you're treated as a private patient. So every time you go, there's the consultation fee and then the ongoing fee. That was the biggest killer tests. Sometimes they were both billed, sometimes that was out of pocket. Yes, I did have to take off days off work because I couldn't work. Yeah, yeah, every. I think the biggest thing is that all the appointments, it's, it's money, and a lot of it's not covered by Medicare or even your private health insurance. So there's a lot of money being spent upfront.*

### *Participant 013\_2023AUDSK*

*Yeah, I think, look, we live in a pretty lucky country with Medicare. A lot of it is covered. Little specialist fees can be a bit hefty when you're seeing them quite regularly, but I'm in a position now where it's only a couple of times a year that I have to see them. Initially, there were big costs, like it was all of the let's go see the heart guy. Let's go see the lung guy, let's go get echograms. Let's go get, you know, nuclear medicine. Let's try this medication, try that medication. I think, you know, after, once you get that diagnosis and you kind of managed after that, it's not too, but haven't been too bad. It all counts like I'm a single mom. It all counts now. I'm sort of managing now. Initially it was every time you turn around with another \$300.00 with the hospital side of things and having the gastroscopes and all that kind of stuff, like, you know, it's covered by Medicare, so we're relatively lucky. But I have spoken to people in America, for example, and they don't have that. It's super expensive, but you know, they go without so much. So comparatively, you know, we're in a relatively lucky position. There are definitely costs involved, but I don't think once you've got your diagnosis and you're kind of on the right track, I don't think it's that bad. It's definitely plenty of worse things. Yeah, like at the moment I'm going through Melanoma stuff, so that's far more expensive, you know?*

### *Participant 78\_2023AUDIS*

*Yes, well the psychologist is very expensive. Yes, there have been quite a few costs but now it's been quite good and there were cardiology costs initially as well for all the scans, but now that I'm under that umbrella there, it's covered for that side of things. Still had the physio obviously isn't and the water therapy wasn't, and the psych isn't and the rheumatologist isn't covered because I see one privately as well. I am out*

*of pocket quite a bit, and I couldn't possibly work because of my fatigue. I'm so tired all the time.*

### *Participant 007\_2023AUDIS*

#### **Cost burden in relation to the cost of parking and travel to attend appointments (including accommodation)**

*Initially the treatment that I was getting was in the public system so we weren't out of pocket in that way. The plane fares and that down to Brisbane they were covered by air travel with our local hospital but what they wouldn't cover at that point in time was someone to accompany me down to CITY. That was a added cost because my mum would come with me or one of my family members. We would pay for them to come down. My husband like I said at the time we had a young child so he would be at home and caring for her. We had cattle that we had to keep looking after as well. Then when we had NAME neurologist in town that was great. We would just go up there and then when he left I had to go back down to LOCATION even though I went privately because that was the closest treating neurologist. Our hospital would pay for my airfares down there and they also would pay at that time then for a support person. Every 12 weeks my treatment costs about \$650 and I get about \$350 of that back from Medicare. We're out of out of pocket about \$300 depending on how it falls but four or five times a year. Then of course you've got you get travel upkeep. Then I see a physio once a month. That's covered by some of the private health care. We're lucky that financially we're able to afford that, and it doesn't put too much of a burden on our finances.*

### *Participant 006\_2023AUDNS*

*Yeah, luckily I haven't had many out of pocket like money costs. But yeah, time off work is very stressful and annoying. I guess that's sort of the main one and time traveling to and from the hospital. It's like nearly three hours return on public transport and then you've sort of been there for 5 minutes.*

### *Participant 96\_2023AUDNS*

#### **Cost burden in needing to special equipment**

*And then thankfully, if we had other kids, I don't know what we'd do because we don't have a support network to look after them, you know, it's like all of those things that are very quickly. And then you've got the nebulizer equipment, which isn't cheap but also because of, I guess, strict. Hiking protocols for CF, you've got to replace the parts every year, and some of them are covered under the they've got like an*

equipment subsidy, so you get new mouthpieces and things every year through the hospital. But then you can't use the same nebulizer head for more than one medication. So for instance, NAME does at the moment. Without an exacerbation, he does hypertonic, saline and pulmicort, but he's got to have a different head for each one and it's cost the same to buy a new head as almost buying the whole new machine. But then you've got also the fact that they've now discontinued it, so you either have to go and source these the heads that they need to use because it has to. You have to push the medication through in a certain at a certain speed and time for it to be absorbed roughly. So you've got to source specific machines which the clinic have been trying to do, but then you've got to go pay for them as well, which is a couple \$100 on top of that. And then you've got to have more than one and you know and then we've also got the added expenses of like all the, you know like the ongoing osmolax and... You know, just if like I guess that's kind of most of it because then you've got like we've obviously got PTSS and we get a subsidy for like heating and cooling as well, which is like \$90.00 a year or half. I can't remember that you get to help with because of the excess you're sweating. You've got to try and keep them in the air con as much as possible during summer and but then you've got. I mean, the cost of living and electricity, you've got to actually, you know, it costs a lot more than you're actually getting subsidized. But I think it's a meds extra, like heating and cooling and then the nebulizers think. That's, yeah. And then, yeah, obviously time off illness for hospital stays and that's pretty much it that I can think of at this .0 and partly sorry, yeah.

*Participant 20\_2023AUORC*

Costs. It's been very difficult. I'm I carer. And because I was a full time carer, I was had to quit my full time job. So not only did we lose a second income, the costs of the medications are all on PBS, which is fine, but when you've got ten a month, it sort of adds up. But the bigger costs are more the, the nebulizers. Like, they cost thousands of dollars, but then you need twice a year to change the heads, which are another like \$500.00. And so it's those things, it's the extra, yeah. And then it's not listed on NDIS. So there's no. Yeah, it's very, it's tricky.

*Participant 23\_2023AUORC*

Really hasn't been a lot of out of pocket. We've got private health cover, but trip was at the children. So apart from equipment, I don't know, like syringes and and different things, I, I don't know that there was and

tubes and stuff. I don't think there was too much. Oh, that's right. I think because he was private, I think originally. We had a lot of pathology bills and if he'd been public we wouldn't have got any of those. Medibank Private has certainly helped a couple of times out with cochlear implant replacements, yeah, and even with schooling, apart from the regular costs, anybody else has. You know as far as surgeries go, I think we've, we've been very lucky and also when he was at sorry, I was trying to say when he was at LOCATION, I don't think we paid for occupational therapy sort of the oh sorry the occasional care which is sort of a bit like kinder. So I would say, you know, we've been quite fortunate in that regard.

*Participant 93\_2023AUENM*

**Cost burden in relation to a family member needing to take time off work**

So in terms of cost, because I'm going through the public system, there's no cost with the consultations. Medications. I mean, my husband and I work full time, so we we're financially OK and the cost of medications isn't that bad, like, you know, it's not that bad. He had to take some time off work to help me when I had the surgery because I needed that dressing changed every couple of hours. Yeah, so he was the poor bunny who had to do it. And I've probably, I've never, you know, because you have paid sick leave. I probably can't. You know, I can't pinpoint time when I've lost money as such because I leave and I had to take time off work. But I, you know, I think maybe, maybe I could say I've probably lost up maybe 3 weeks. Work in the last three years into HS, so yeah.

*Participant 017\_2023AUDSK*

Look, it's been it's, it's a very long journey for the most part. In my 20s, getting to the dermatologist, waiting three to six months, finding out it's three \$400.00 and that most of that would be out of pocket. It's painful. So you delay it and you procrastinate going. As I've gotten older, I haven't done that, but the surgery was pretty much all private, so I think I coughed at around two and a half thousand plus not going to work, plus my partner staying home so he can take me back from the hospital. It's, it's quite it's currently quite expensive because. Of the surgery, but on a day-to-day basis with the appointments around me. I think I calculated it at roughly around \$400.00 a month just for medical expenses.

*Participant 026\_2023AUDSK*

*Well, I had to give up my work because obviously, my wheelchair and I couldn't do it, so I had to give up work. Before that, you have to go to a specialist in LOCATION and we live in regional, LOCATION. I have to fly down and it's really expensive. I had to take my sister at the time because I didn't have any care. She had to take time off work. If I had to do a driving assessment, you have to go to the ophthalmologist. That's like, even though I'm in a bond, it's still hundreds of dollars. Neurologists are super expensive. Then you have to go to a psychologist or psychiatrist or that was at the beginning and that costs lots of money. It's just going for your driving license, \$1,800 for an OT assessment. They've got to come to the house, get out of hospital jail card, you've got to have all this, then they've got to come and check and it costs so much money. Then medications, some on the PBS, but most of mine aren't. You go and get your scripts done and it's \$250. That's not including the medical marijuana.*

*Participant 001\_2023AUDNS*

#### **Cost burden in relation to allied health care**

*With the cost, yeah, we've had to pay everything along the way. We haven't had like the only time we got a bit of support when he was quite a bit younger. The kids at the local hospital that were near, they had like the allied health area there. But then when they get to the age of five, that's it. You can't be seen there anymore. So then we just had to go out and our own backs then and pay for OT and Physio and Health like the health fund paid a little bit and obviously Medicare paid for some things. But yeah, it's generally costs are on our back not they're not that many Medicare benefits for.*

*Participant 11\_2023AUDPA*

*So everything we've done is privately, so cost would be speech therapy. Then that's not covered by Medicare, but it is partially covered by HBF. But once you reach the threshold, you don't get anymore cover for, you know, X amount of sessions in a year or within a six months or whatever. I can't even remember what it is now. So yeah, we've paid privately for speech OT. All of our doctors have been privately. We haven't gone through LOCATION Children's, His dental work, yeah, everything we've done is just private. So, yeah, it's been thousands and thousands of dollars.*

*Participant 22\_2023AUDPA*

*Yeah, we've been very lucky that we've been able to afford her what we say. So I had to give up work after she was 18 months old. I was juggling the two, but it*

*wasn't working so I gave up work. I've only just been able to start looking at going back to work probably the beginning of next year because you still need so much care. A lot of the specialists that we have are private because of the wait list for public entities. So, and then for the first few years we were paying all out of pocket costs for Private Allied Health because the duplication isn't necessarily recognized to the NDIS as a disability. So it wasn't until she was about how old is she now, 4 1/2 that we got an NDIS plan which still doesn't cover her support. So we use the NDIS package and then we use our own funds to pocket the rest of the year.*

*Participant 32\_2023AUDPA*

#### **Cost burden in needing to special creams, ointments or complementary therapies**

*Across...I would definitely say there's, there's a few. So cost in terms of treatment is a big one, so seeing the specialists, I don't have health insurance, so I pay out of pocket to see a specialist, which is roughly \$280 per session with this doctor. My medication costs \$40 a month and you know I have to buy special bras which are very expensive like. Not very expensive, but you know, just constantly having to buy new ones as well that get a bit destroyed because I get puss and blood on them and they get a bit gross and I don't want to wear it so I have to buy new ones. Taking time off work is rare, like I won't normally take time off work for the disease. I just push through and go to work. Just trying to think, even like at home remedies. I'm always buying new creams, powders, things for the bath, antibiotics, stuff like that. I haven't even know how much that would amount to over the past 10 years. I've had probably a lot, definitely in the thousands that I've spent on all these different things to try for it.*

*Participant 010\_2023AUDSK*

*Look, fortunate that the DOCTOR is part of the public system. The initial cost to the neurologist and the private rheumatologists were, gosh, I can't even remember now, but hundreds of dollars a visit. Whereas now, that's covered. Certainly, the medications add up and they can run to an expense. The massage is all out of pocket. Look, if I wasn't working, I don't think I wouldn't be able to have the massage or have those other things. I wouldn't have been able to have the wax bath and some of the other things I have. Whereas now, it's anything I need, absolutely. Even things like my complete wardrobe has changed. Now, I don't have anything with zips, buttons, all new shoes, because they've got to be slip*

on; that all adds up. I can afford it because I'm still able to work, but if I understand people that can't work, that would be really tough.

Participant 017\_2023AUDIS

Well, all of the above I we do go on to the PBS free list by late March, early April which gives you an indication as to how many drugs we take. But a lot of the medication is not on the PBS like TRENAL is one that's about \$58 every fortnight. The obviously the vitamins and that that we need as part of our treatment they obviously you have to pay for. So my chemist is usually about be between 120 and 150 week and between 20 and 80 the other week depending on when what medications have run out. So, yeah, and I mean I take things like laxatives and you know, because of the drugs I get constipated, so you have to buy, you know, so there's all those sort of things and the expensive sunscreens because of the methotrexate and the specialist. You know, they don't bulk bill. So you you know, it might be a \$300.00 bill and the shortfall might be 200 and and yeah it, yeah we've had some tough times but yeah look, I mean I I didn't say anything because my family would my daughter and son-in-law would just say, well you've only got to ask we'll help you. But I don't expect that. So you just manage, but they keep an eye on us and make sure we've got what we need without asking. So yeah.

Participant 021\_2023AUDIS

#### Cost burden in needing to special diet or lifestyle adaptation

Yeah, it's, it can be quite expensive. Like we have like we're very lucky, we've got private health. I have a very flexible job. But yeah, it does a lot of, a lot of medical, like, you know, at times a lot of appointments and the medication. But because he's done so many diets, like eating a gluten and dairy free diet is just generally a lot more. Like my grocery bill is a lot more expensive now than it was before he was diagnosed. So that, like that is just generally that's more expensive than the medication. Yeah, yeah. OK.

Participant 79\_2023AUDIS

So we were spending a lot. A lot of money on nappies until we got NDS funding when she was about, I don't know, 2 1/2. But before that we spent we go through three or four boxes of nappies a week. So that was a really big cost. Plus she's very has a very particular taste with things, so always sort of trying different things, buying things that she likes and of course as children that want to do, they're the most, you know,

she likes the most expensive things. So having all these things that then she likes and then she doesn't like and, you know, spent all this money on, well she doesn't eat that much. So yeah, I find there's a lot of there's definitely a lot of expenses that we wouldn't have if she didn't have just, you know, just buying. Yeah. Particular things like we've had to air condition the whole house because she's, you know, intolerant to heat. We have the air conditioner running 24/7 in summer, so our power bills are astronomical. Yeah, yeah, OK.

Participant 15\_2023AUORC

#### No cost burden and that nearly everything was paid for through the public health system

Most of my costs have been looked after by the PBS and that I'm very fortunate. You know, the pill, I believe in the United States is about \$800 per pill. Yeah. So over a month period, that's about \$24,000 exactly, which is a fair whack if you don't have a lot.

Participant 012\_2023AUORC

Well, truly, I can't complain that I've had a lot of costs. I've been dealt with by the public hospital system, so I haven't had to pay the specialist. Now and again, I've been sent for something that I've had to pay for, but that's been not the rule, that's been the exception. If I had to pay for my medication, I would have stopped taking it 10 years ago because I can't afford them. The Macitentan itself is, I think it's something between \$2,000 and \$3,000 a month. I wouldn't be taking that if I had to pay for it. I feel very well supported there that I really...I mean there have been costs, but considering I'm an aged pensioner now that my medication cost very little and I reached the safety net very early.

Participant 004\_2023AUDIS

Well, I haven't had any cost things that because I'm on the pension and I just get most of my medications on the PBS, which I'm very fortunate. And I thank God for that.

Participant 005\_2023AUDIS

PARTICIPANT: We haven't had really that much cost because most of it's like being like bulk bill and everything. So that was sort of thing, especially like most doctors don't bulk build stuff anymore. So it seems like, yeah. My work's been pretty flexible when I had to take him to the doctors and stuff like that, or when we had like we had to go to Children's Hospital for scans and stuff like that. So they've been pretty flexible with that....Yeah, I so like when he was like

*really young. Now it's like how he ever saw this thing and he how he ever goes to the doctors or anything like that.*

*Participant 7\_2023AUDPA*

*Not too bad to be honest. Like a lot of the stuff you know has been, it goes through the public system or I haven't had to take a lot of time off or I've been given, you know, carers leave and things like that.*

*Participant 14\_2023AUDPA*

*Well, we've been really fortunate in that most of it's been done through the that we've been bulk billed for most things. The main cost for us would have been our time and also the time that we have had to put into research this condition on our own and be the coordinators of the whole thing, both medically, physically, socially and mentally, so it's our time, our time has been so consumed with looking after our daughter on this condition that there has been some areas where we lack enjoyment and things like that.*

*Participant 25\_2023AUDPA*

**No cost burden and that nearly everything was paid for through the private health system**

*I can consider the cost actually was I've got I've got a private health for everything is just covered, so I've never encountered any cost like out of pocket so far.*

*Participant 001\_2023AUORC*

*Cost wise probably. I've got, I've got private medical, so it most of it, most of it's covered through the private health insurance. But medication? I don't have any. So now I've just got a Commonwealth Seniors card. I do get a bit of a deduction in some medications, which has been very helpful with the Humira, which would have cost me a lot of money.*

*Participant 024\_2023AUDSK*

**No cost burden as participant was able to afford all costs**

*It hasn't been too bad because I haven't had that many treatments. The dermatologist appointments can be quite expensive, but it's it's manageable with me working at the moment. And the Humira injections that I'm about to start are very expensive, but they're on PBS them quite affordably.*

*Participant 019\_2023AUDSK*

*I haven't really had an issue with the cost. I'm probably, I'm probably well off where. I've got private health cover, Medicare. I've covered most of the radiation, which I was, you know, very grateful for. So,*

*yeah, I suppose at this stage money wasn't really an option. You know, scans, scans and all that were just part of part of the process. You know, pet scans. You know, it might have cost me \$8000. The other scans are subsidized, didn't cost me a lot because I had private health cover. And the radiation, like you know, the lady did sort of say it was quite expensive but I I found it to be quite reasonable considering, you know, you're having radiation to save your life. The rebate after Medicare I thought was really good. So the costs weren't a real big factor of sort of more the outcome and making sure yeah I sort of knock this, knock this thing on the task as best I can kind of thing. So yeah.*

*Participant 24\_2023AUORC*

**No cost burden: NDIS**

*The only the only cost would be me having the stop work to look after NAME because of the effect of the disorder, but other than that, I can't. I mean, she's covered under NDIS in terms of treatment. The other not really, I mean, the only cost is in terms of us raising funds now to get a treatment and a cure. But that's something different. Yeah, OK.*

*Participant 16\_2023AUORC*

*So yeah, most of it's been covered. We haven't gone privately with anything and really we couldn't afford it back in the early days and now we have the NDIS. So it's a little bit different.*

*Participant 28\_2023AUORC*

*Hasn't been a massive a lot of costs for us. She's covered by NDIS for therapies and things. So that's been, yeah, that's saved us a lot of cost. Maybe travel probably travel's the biggest cost and so far she's living a normal life as she as a brother and sister do, with maybe just a little bit of extra help and some extra therapies and things. Probably education. We get her tutored once a week with a tutor, so that's an extra cost for us and maybe education because she doesn't qualify for an aid for any of that, so for her schooling.*

*Participant 10\_2023AUDPA*

*Yeah, very fortunate to have NDIS funding. Excellent. Excellent. Could not have done it without it. And also in terms of timing, our therapists have been very supportive to come and see him at his daycare. Excellent. So we haven't had to take the time off work.*

*Participant 20\_2023AUDPA*



Yeah, very costly. I think that my protect career potentials would be I couldn't keep a job of being continuously called out because she wasn't well that's been that was a big thing and yeah and footing the bill for all the specialists you know even though she's

got and the health insurance as well. OK, that she's on NDIS now, which helps with certain things, but not with those things.  
Participant 33\_2023AUDPA

Table 8.23: Cost considerations

Cost considerations	All participants	Developmental anomalies	Diseases of the immune system	Diseases of the nervous system	Diseases of the skin	Endocrine, nutritional or metabolic diseases	Other rare condition	Person with condition	Family or carer	Female	Male
	n=302 %	n=67 %	n=81 %	n=45 %	n=32 %	n=95 %	n=32 %	n=204 %	n=98 %	n=214 %	n=86 %
Overall, there was at least some cost burden	197 65.23	29 43.28	61 75.31	36 80.00	24 75.00	25 26.32	22 68.75	149 73.04	48 48.98	149 69.63	46 53.49
Overall, there was no cost burden	57 18.87	10 14.93	12 14.81	7 15.56	4 12.50	15 15.79	9 28.13	38 18.63	19 19.39	38 17.76	19 22.09

Cost considerations	All participants	Aged under 18	Aged 18 to 44	Aged 45 to 64	Aged 65 plus	Trade or high school	University	Regional or remote	Metropolitan	Mid to low status	Higher status
	n=302 %	n=66 %	n=103 %	n=84 %	n=49 %	n=148 %	n=172 %	n=84 %	n=218 %	n=154 %	n=148 %
Overall, there was at least some cost burden	197 65.23	31 46.97	73 70.87	65 77.38	28 57.14	94 63.51	100 58.14	53 63.10	144 66.06	104 67.53	93 62.84
Overall, there was no cost burden	57 18.87	12 18.18	18 17.48	10 11.90	17 34.69	33 22.30	22 12.79	15 17.86	42 19.27	27 17.53	30 20.27

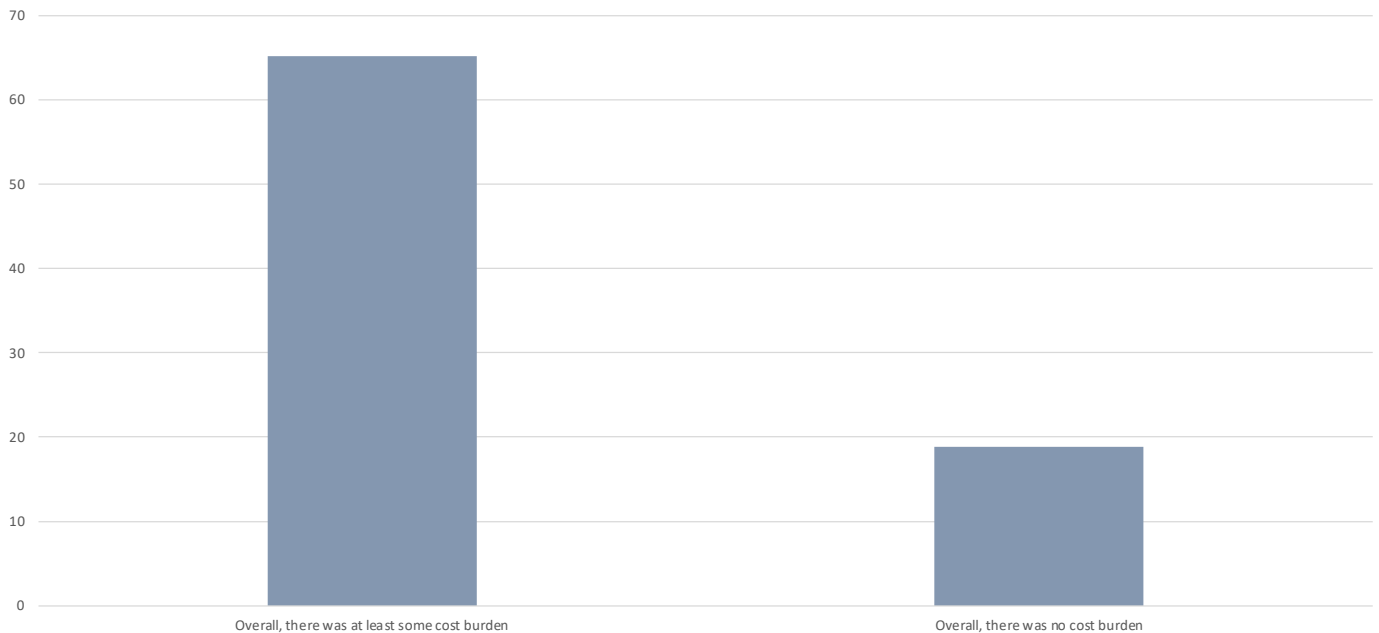


Figure 8.13: Cost considerations

Table 8.24: Cost considerations – subgroup variations

Cost considerations	Reported less frequently	Reported more frequently
Overall, there was at least some cost burden	Developmental anomalies Endocrine, nutritional or metabolic diseases Family or carer Male	Diseases of the immune system Diseases of the nervous system Aged 45 to 64
Overall, there was no cost burden	Aged under 18	Aged 65 plus

Table 8.25: Cost considerations (Reasons for cost)

Cost considerations (reasons)	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=302	%	n=67	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=204	%	n=98	%	n=214	%	n=86	%
Cost burden: needing to take time off work	99	32.78	21	31.34	24	29.63	22	48.89	8	25.00	11	11.58	13	40.63	66	32.35	33	33.67	79	36.92	19	22.09
Cost burden: cost of treatments	93	30.79	6	8.96	34	41.98	17	37.78	15	46.88	8	8.42	13	40.63	79	38.73	14	14.29	75	35.05	18	20.93
Cost burden: specialist appointments	81	26.82	11	16.42	47	58.02	6	13.33	12	37.50	5	5.26	0	0.00	69	33.82	12	12.24	70	32.71	10	11.63
Cost burden: diagnostic tests and scans	39	12.91	7	10.45	18	22.22	3	6.67	1	3.13	7	7.37	3	9.38	31	15.20	8	8.16	30	14.02	9	10.47
Cost burden: parking, travel, accomodation to attend appointments	39	12.91	8	11.94	10	12.35	6	13.33	3	9.38	9	9.47	3	9.38	29	14.22	10	10.20	30	14.02	9	10.47
Cost burden: special equipment	26	8.61	8	11.94	2	2.47	3	6.67	6	18.75	4	4.21	3	9.38	14	6.86	12	12.24	23	10.75	3	3.49
Cost burden: family member needing to take time off work	18	5.96	0	0.00	2	2.47	8	17.78	2	6.25	6	6.32	0	0.00	10	4.90	8	8.16	13	6.07	5	5.81
Cost burden: allied health care	17	5.63	8	11.94	4	4.94	2	4.44	0	0.00	1	1.05	2	6.25	9	4.41	8	8.16	13	6.07	3	3.49
Cost burden: special creams, ointments or complementary therapies	13	4.30	1	1.49	4	4.94	1	2.22	6	18.75	0	0.00	1	3.13	11	5.39	2	2.04	10	4.67	3	3.49
Cost burden: special diet or lifestyle adaptation	11	3.64	3	4.48	5	6.17	0	0.00	0	0.00	2	2.11	1	3.13	7	3.43	4	4.08	8	3.74	3	3.49
No cost burden: paid for through the public health system (incl. NDIS)	65	21.52	21	31.34	9	11.11	7	15.56	4	12.50	16	16.84	8	25.00	33	16.18	32	32.65	38	17.76	26	30.23

Cost considerations (reasons)	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=302	%	n=66	%	n=103	%	n=84	%	n=49	%	n=148	%	n=172	%	n=84	%	n=218	%	n=154	%	n=148	%
Cost burden: needing to take time off work	99	32.78	22	33.33	39	37.86	28	33.33	10	20.41	41	27.70	55	31.98	27	32.14	72	33.03	52	33.77	47	31.76
Cost burden: cost of treatments	93	30.79	11	16.67	38	36.89	30	35.71	14	28.57	41	27.70	51	29.65	26	30.95	67	30.73	47	30.52	46	31.08
Cost burden: specialist appointments	81	26.82	8	12.12	33	32.04	28	33.33	12	24.49	46	31.08	35	20.35	25	29.76	56	25.69	47	30.52	34	22.97
Cost burden: diagnostic tests and scans	39	12.91	5	7.58	13	12.62	12	14.29	9	18.37	21	14.19	18	10.47	7	8.33	32	14.68	17	11.04	22	14.86
Cost burden: parking, travel, accomodation to attend appointments	39	12.91	8	12.12	10	9.71	12	14.29	9	18.37	15	10.14	23	13.37	9	10.71	30	13.76	24	15.58	15	10.14
Cost burden: special equipment	26	8.61	10	15.15	7	6.80	9	10.71	0	0.00	12	8.11	14	8.14	9	10.71	17	7.80	17	11.04	9	6.08
Cost burden: family member needing to take time off work	18	5.96	3	4.55	3	2.91	7	8.33	5	10.20	9	6.08	7	4.07	2	2.38	16	7.34	9	5.84	9	6.08
Cost burden: allied health care	17	5.63	5	7.58	5	4.85	4	4.76	3	6.12	9	6.08	8	4.65	3	3.57	14	6.42	8	5.19	9	6.08
Cost burden: special creams, ointments or complementary therapies	13	4.30	2	3.03	3	2.91	6	7.14	2	4.08	8	5.41	5	2.91	4	4.76	9	4.13	11	7.14	2	1.35
Cost burden: special diet or lifestyle adaptation	11	3.64	3	4.55	3	2.91	5	5.95	0	0.00	4	2.70	7	4.07	3	3.57	8	3.67	8	5.19	3	2.03
No cost burden: paid for through the public health system (incl. NDIS)	65	21.52	26	39.39	13	12.62	9	10.71	17	34.69	38	25.68	26	15.12	12	14.29	53	24.31	28	18.18	37	25.00

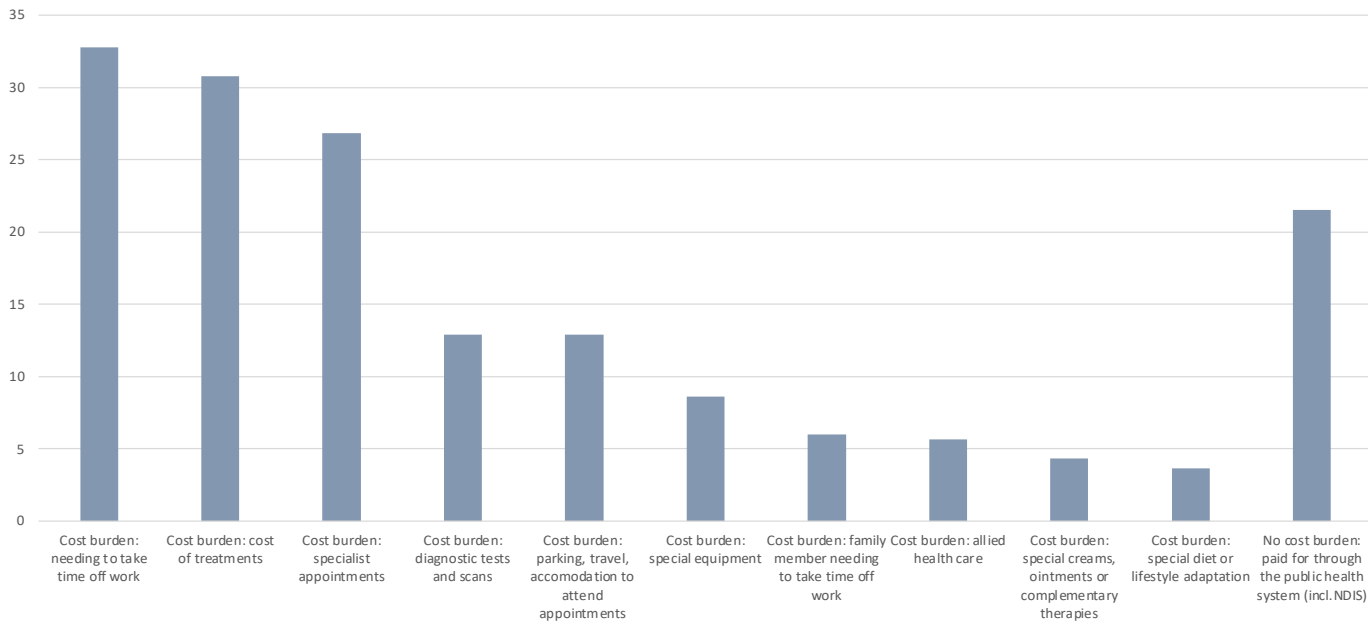


Figure 8.14: Cost considerations (Reasons for cost)

Table 8.26: Cost considerations (Reasons for cost)– subgroup variations

Cost considerations (reasons)	Reported less frequently	Reported more frequently
Cost burden: needing to take time off work	Endocrine, nutritional or metabolic diseases Male Aged 65 plus	Diseases of the nervous system
Cost burden: cost of treatments	Developmental anomalies Endocrine, nutritional or metabolic diseases Family or carer Aged under 18	Diseases of the immune system Diseases of the skin
Cost burden: specialist appointments	Developmental anomalies Diseases of the nervous system Endocrine, nutritional or metabolic diseases Other rare condition Family or carer Male Aged under 18	Diseases of the immune system Diseases of the skin
Cost burden: diagnostic tests and scans		
Cost burden: parking, travel, accomodation to attend appointments		
Cost burden: special equipment		Diseases of the skin
Cost burden: family member needing to take time off work		Diseases of the nervous system
Cost burden: allied health care		
Cost burden: special creams, ointments or complementary therapies		Diseases of the skin
Cost burden: special diet or lifestyle adaptation		
No cost burden: paid for through the public health system (incl.NDIS)	Diseases of the immune system Aged 45 to 64	Family or carer Aged under 18 Aged 65 plus

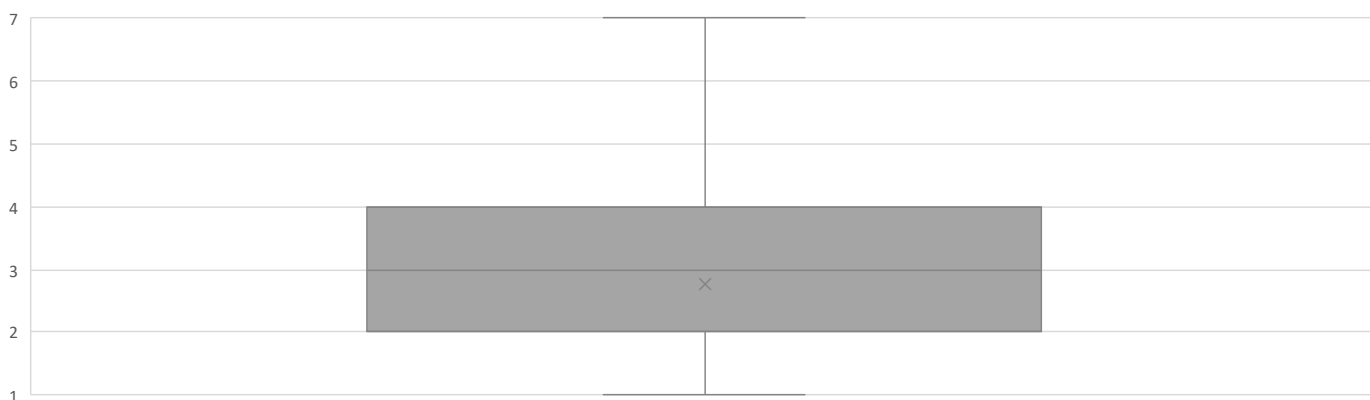
### Overall impact of condition on quality of life

In the online questionnaire, participants were asked to rate the overall impact their condition on quality of life. Quality of life was rated on a Likert scale from one to seven, where one is Life was very

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

**Table 8.27: Overall impact of condition on quality of life**

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



**Figure 8.15: Overall impact of condition on quality of life**

## Experience of anxiety related to disease progression

### Fear of progression

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

of 37.09 (SD = 10.40), which corresponds to moderate levels of anxiety.

The **Fear of Progression** questionnaire measures the level of anxiety people experience in relation to their conditions. On average fear of progression score for participants in this study indicated moderate levels of anxiety.

**Table 8.28: Fear of progression summary statistics**

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

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

### Fear of progression by condition

Comparisons were made by **condition**. There were 57 participants (15.41%) with developmental anomalies , 72 participants (19.46%) with diseases of the immune system , 93 participants (25.14%) with diseases of the nervous system , 29 participants (7.84%) with diseases of the skin , 94 participants (25.41%) with endocrine, nutritional or metabolic diseases , and 25 participants (6.76%) with other rare condition.

A one way ANOVA test indicated a statistically significant difference in the Care coordination: Total score scale between groups,  $F(5, 364) = 4.29$   $p = 0.0008$ . The largest significant difference was between participants in the Diseases of the skin subgroup

(median = 42.03, IQR = 9.76), and participants in the Endocrine, nutritional or metabolic diseases subgroup (median = 33.77, IQR = 9.10,  $p = 0.0024$ ).

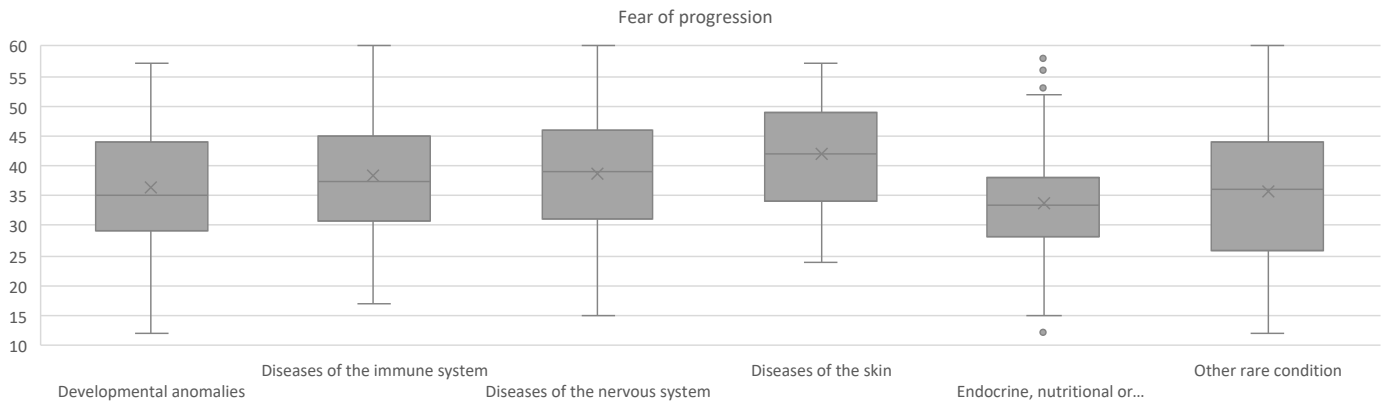
The **Fear of Progression** questionnaire measures the level of anxiety people experience in relation to their conditions. On average, participants in the Diseases of the skin subgroup scored higher than participants in the Endocrine, nutritional or metabolic diseases subgroup. This indicates that participants in the Diseases of the skin subgroup had high levels of anxiety, and participants in the Endocrine, nutritional or metabolic diseases subgroup had moderate levels of anxiety.

**Table 8.29: Fear of progression total score by condition summary statistics and one-way ANOVA**

Fear of progression	Group	Number (n=370)	Percent	Mean	SD	Source of difference	Sum of squares	dF	Mean Square	f	p-value
Total score	Developmental anomalies	57	15.41	36.26	10.46	Between groups	2217.00	5	443.40	4.29	0.0008*
	Diseases of the immune system	72	19.46	38.32	10.20	Within groups	37669.00	364	103.50		
	Diseases of the nervous system	93	25.14	38.82	9.95	Total	39886.00	369			
	Diseases of the skin	29	7.84	42.03	9.76						
	Endocrine, nutritional or metabolic diseases	94	25.41	33.77	9.10						
	Other rare condition	25	6.76	35.76	13.97						

**Table 8.30: Fear of progression total score by condition one-way post hoc Tukey HSD test**

Fear of progression	Group	Difference	Upper	Lower	p adjusted
Total score	Diseases of the immune system - Developmental anomalies	2.06	-3.11	7.22	0.8643
	Diseases of the nervous system - Developmental anomalies	2.55	-2.35	7.46	0.6693
	Diseases of the skin - Developmental anomalies	5.77	-0.88	12.42	0.1306
	Endocrine, nutritional or metabolic diseases - Developmental anomalies	-2.50	-7.39	2.40	0.6886
	Other rare condition - Developmental anomalies	-0.50	-7.49	6.49	0.9999
	Diseases of the nervous system - Diseases of the immune system	0.50	-4.08	5.07	0.9996
	Diseases of the skin - Diseases of the immune system	3.72	-2.69	10.13	0.5589
	Endocrine, nutritional or metabolic diseases - Diseases of the immune system	-4.55	-9.12	0.01	0.0510
	Other rare condition - Diseases of the immune system	-2.56	-9.33	4.21	0.8877
	Diseases of the skin - Diseases of the nervous system	3.22	-2.98	9.42	0.6728
	Endocrine, nutritional or metabolic diseases - Diseases of the nervous system	-5.05	-9.31	-0.79	0.0098*
	Other rare condition - Diseases of the nervous system	-3.06	-9.62	3.51	0.7659
	Endocrine, nutritional or metabolic diseases - Diseases of the skin	-8.27	-14.46	-2.08	0.0021*
	Other rare condition - Diseases of the skin	-6.27	-14.23	1.68	0.2134
	Other rare condition - Endocrine, nutritional or metabolic diseases	1.99	-4.56	8.55	0.9532



**Figure 8.16: Boxplot of Fear of progression total score by condition**

### Fear of progression by type of participant

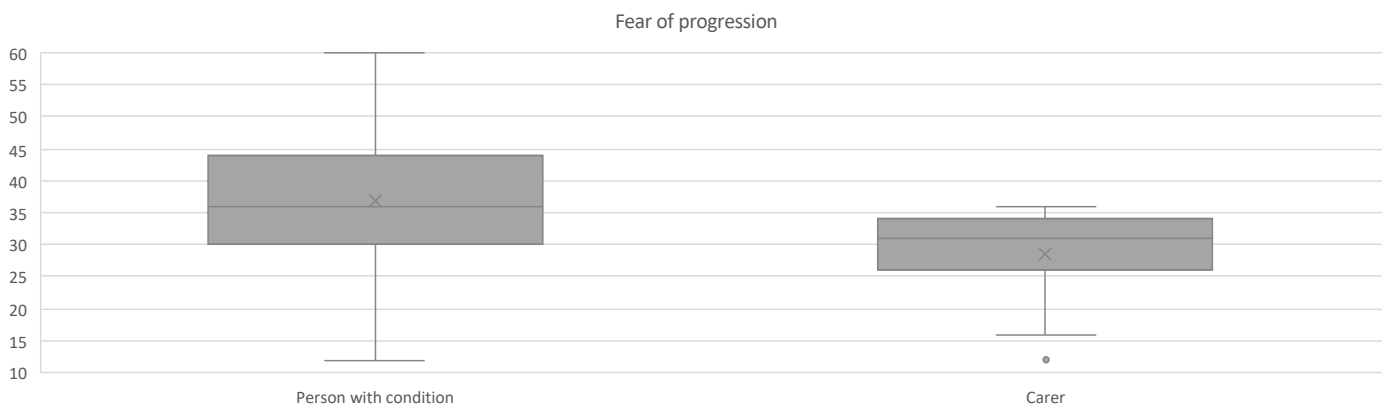
Comparisons were made by **type of participant** there were 249 participants (67.30%) with person with condition and, 121 participants (32.70%) with carer.

No significant differences were observed between participants by **type of participant** for any of the Fear of progression scales.

Assumptions for normality and variance were met, a two-sample t-test was used.

**Table 8.31: Fear of progression total score by type of participant summary statistics and T-test**

Fear of progression	Group	Number (n=370)	Percent	Mean	SD	T	dF	p-value
Total score	Person with condition	249	67.30	36.95	10.37	-0.36	368	0.7159
	Carer	121	32.70	37.37	10.48			



**Figure 8.17: Boxplot of Fear of progression total score by type of participant**

## Fear of progression by gender

Comparisons were made by **gender**, there were 274 female participants (74.46%), and 94 male participants (25.54%).

Assumptions for normality and variance were met, a two-sample t-test was used.

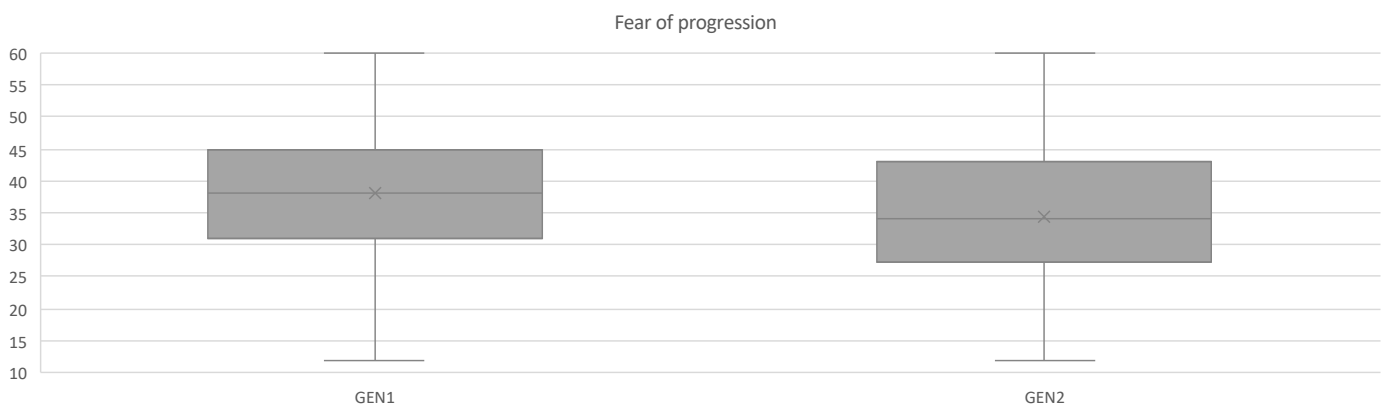
A two sample t-test indicated that the mean score for the **Fear of progression Total score** scale [ $t(366) = 2.87$ ,  $p = 0.0044$ ] was significantly higher for participants in

the Female subgroup (Mean = 37.99, SD = 9.97) compared to participants in the Male subgroup (Mean = 34.46, SD = 11.25.)

The **Fear of Progression** questionnaire measures the level of anxiety people experience in relation to their conditions. On average, participants in the Female subgroup had a higher score compared to Male, however, both groups had moderate levels of anxiety.

**Table 8.32: Fear of progression total score by gender summary statistics and T-test**

Fear of progression	Group	Number (n=368)	Percent	Mean	SD	T	dF	p-value
Total score	Female	274	74.46	37.99	9.97	2.87	366	0.0044*
	Male	94	25.54	34.46	11.25			



**Figure 8.18: Boxplot of Fear of progression total score by gender**

## Fear of progression by age

Comparisons were made by **age** of person with condition. There were 87 participants (23.45%) with aged under 18, 120 participants (32.35%) with aged 18 to 44, 105 participants (28.30%) with aged 45 to 64, and 58 participants (15.63%) with aged 65 or older.

A one-way ANOVA test was used when the assumptions for response variable residuals were normally distributed and variances of populations were equal. A Tukey HSD test was used post hoc to identify the source of any differences identified in the one-way ANOVA test.

A one way ANOVA test indicated a statistically significant difference in the Care coordination: Total

score scale between groups,  $F(3, 366) = 5.60$ ,  $p = 0.0009$ . The largest significant difference was between participants in the Aged 18 to 44 subgroup (median = 39.36, IQR = 9.59), and participants in the Aged 65 or older subgroup (median = 32.74, IQR = 10.26,  $p = 0.0004$ ).

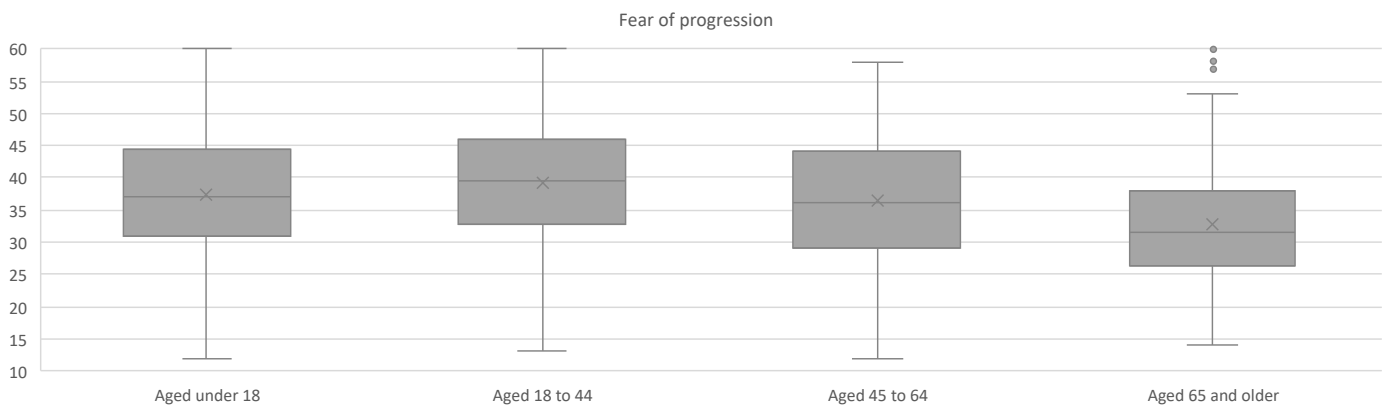
The **Fear of Progression** questionnaire measures the level of anxiety people experience in relation to their conditions. On average, participants in the **Aged 18 to 44** subgroup had a higher score compared to **Aged 65 or older**, however, both groups had moderate levels of anxiety.

**Table 8.33: Fear of progression total score by age summary statistics and one-way ANOVA**

Fear of progression	Group	Number (n=370)	Percent	Mean	SD	Source of difference	Sum of squares	dF	Mean Square	f	p-value
Total score	Aged under 18	87	23.45	37.44	10.65	Between groups	1749.00	3	583.00	5.60	0.0009*
	Aged 18 to 44	120	32.35	39.36	9.59	Within groups	38137.00	366	104.20		
	Aged 45 to 64	105	28.30	36.61	10.48	Total	39886.00	369			
	Aged 65 or older	58	15.63	32.74	10.26						

**Table 8.34: Fear of progression total score by age one-way post hoc Tukey HSD test**

Fear of progression	Group	Difference	Upper	Lower	p adjusted
Total score	Aged 18 to 44 - Aged under 18	1.92	-1.79	5.63	0.5400
	Aged 45 to 64 - Aged under 18	-0.83	-4.65	2.99	0.9441
	Aged 65 or older - Aged under 18	-4.70	-9.16	-0.23	0.0350*
	Aged 45 to 64 - Aged 18 to 44	-2.75	-6.27	0.77	0.1842
	Aged 65 or older - Aged 18 to 44	-6.62	-10.83	-2.40	0.0004*
	Aged 65 or older - Aged 45 to 64	-3.87	-8.18	0.44	0.0962



**Figure 8.19: Boxplot of Fear of progression total score by age**

### Fear of progression by education

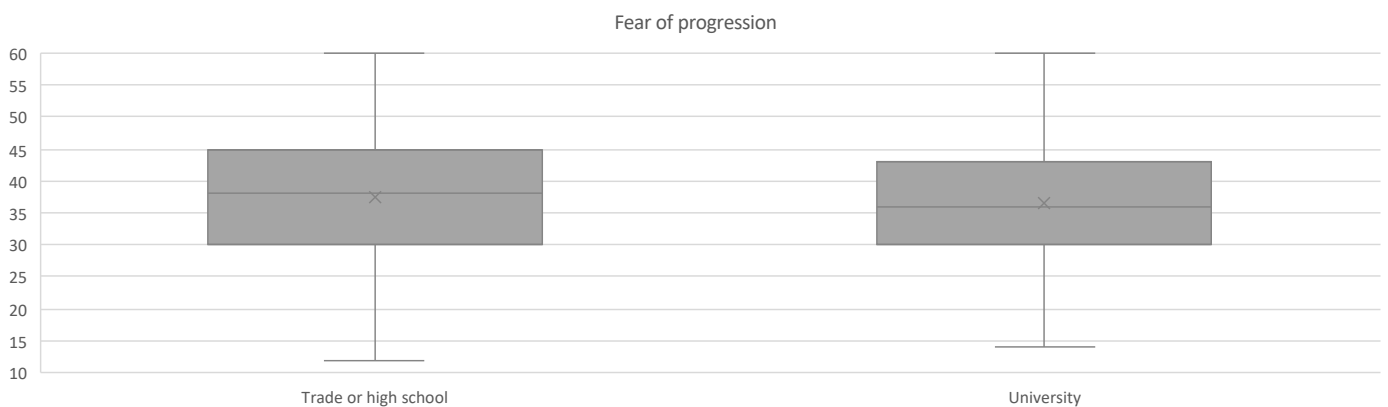
Comparisons were made by **education** status, between those with trade or high school qualifications (n=177, 48.90%), and those with a university qualification (n=185, 51.10%).

Assumptions for normality and variance were met, a two-sample t-test was used.

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

**Table 8.35: Fear of progression total score by education summary statistics and T-test**

Fear of progression	Group	Number (n=362)	Percent	Mean	SD	T	dF	p-value
Total score	Trade or high school	177	48.90	37.54	10.90	0.76	360	0.4479
	University	185	51.10	36.70	9.98			



**Figure 8.20: Boxplot of Fear of progression total score by education**

## Fear of progression by location

The **location** of participants was evaluated by postcode using the Australian Statistical Geography Maps (ASGS) Remoteness areas accessed from the Australian Bureau of Statistics. Those living in regional or remote areas (n=105, 28.38%) were compared to those living in a metropolitan area (n=265, 71.62%).

Assumptions for normality and variance were met, a two-sample t-test was used.

No significant differences were observed between participants by **location** for any of the Fear of progression scales.

**Table 8.36: Fear of progression total score by location summary statistics and T-test**

Fear of progression	Group	Number (n=370)	Percent	Mean	SD	T	dF	p-value
Total score	Regional or remote	105	28.38	37.19	10.23	0.12	368	0.9063
	Metropolitan	265	71.62	37.05	10.48			



**Figure 8.21: Boxplot of Fear of progression total score by location**

## Fear of progression by socioeconomic status

Comparisons were made by **socioeconomic status**, using the Socio-economic Indexes for Areas (SEIFA) ([www.abs.gov.au](http://www.abs.gov.au)), SEIFA scores range from 1 to 10, a higher score denotes a higher level of advantage. Participants with a mid to low SEIFA score of 1-6 (n=185, 50.00%) compared to those with a higher SEIFA score of 7-10 (n=185, 50.00%).

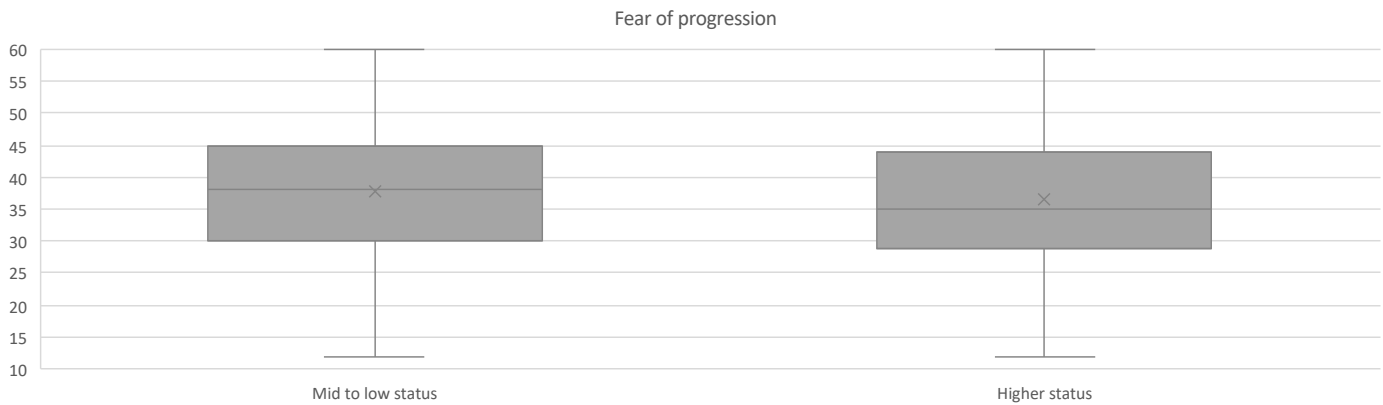
Assumptions for normality and variance were met, a two-sample t-test was used.

No significant differences were observed between participants by **socioeconomic status** for any of the Fear of progression scales.

**Table 8.37: Fear of progression total score by socioeconomic status summary statistics and T-test**

Fear of progression	Group	Number (n=370)	Percent	Mean	SD	T	dF	p-value
Total score	Mid to low status	185	50	37.71	10.46	1.15	368	0.2527
	Higher status	185	50	36.47	10.32			





**Figure 8.22: Boxplot of Fear of progression total score by socioeconomic status**

### Fear of progression individual questions

On average, participants scored in the **“Never”** range for the following questions: “Anxious if not experiencing any side effects think it doesn’t work” (median=1.00, IQR=1.00).

On average, participants scored in the **“Sometimes”** range for the following questions: “Becomes anxious thinking that disease may progress” (median=3.00, IQR=1.00), “Is nervous prior to doctors appointments or periodic examinations” (median=3.00, IQR=2.00), “Afraid of pain” (median=3.00, IQR=2.00), “Has concerns about reaching professional and/or personal goals because of illness:” (median=3.00, IQR=2.00), “When anxious, has physical symptoms such as a rapid heartbeat, stomach ache or agitation” (median=3.00, IQR=2.00), “The possibility of relatives being diagnosed with this disease disturbs participant” (median=3.00, IQR=3.00), “Is disturbed that they may have to rely on

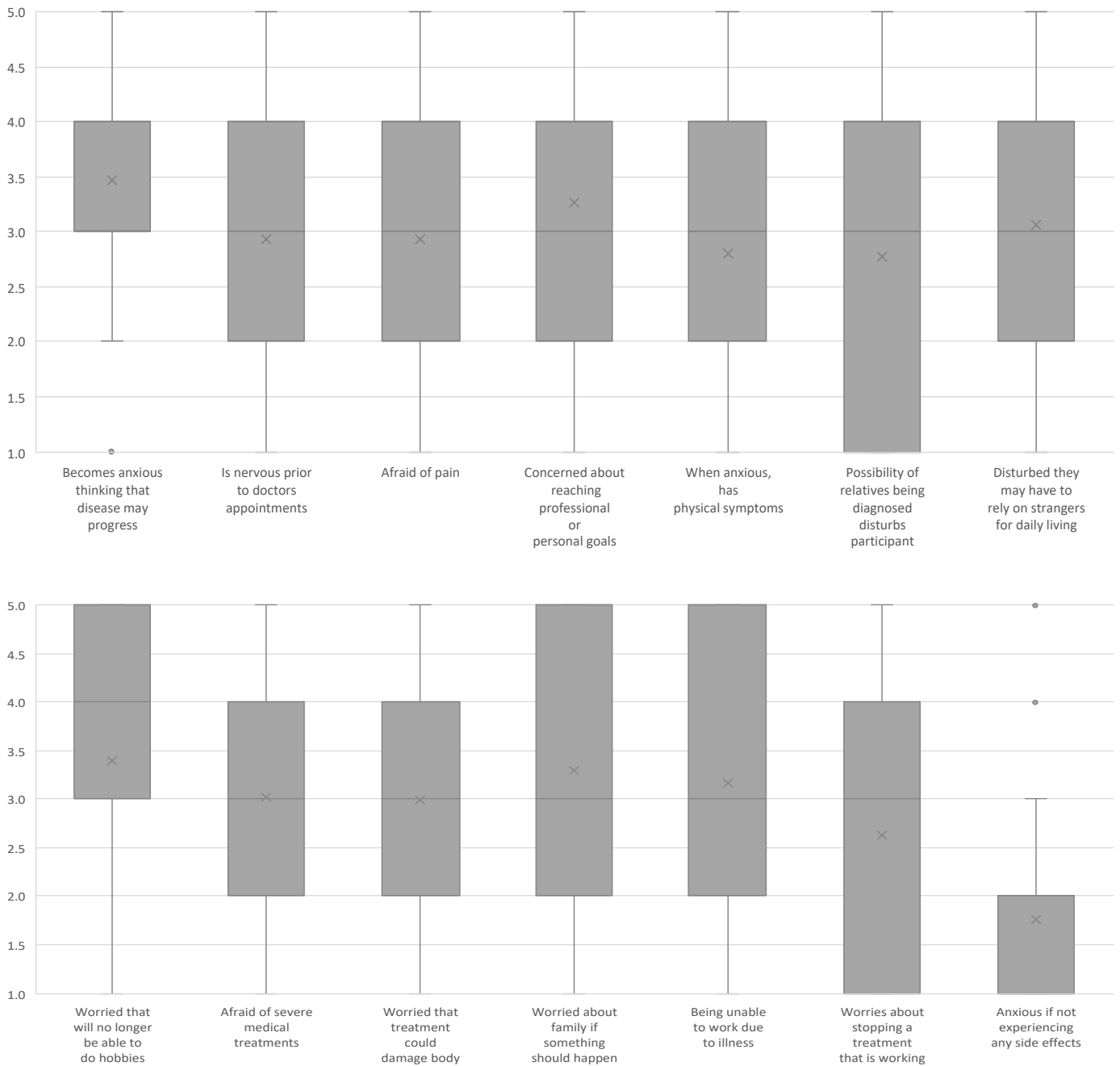
strangers for activities of daily living” (median=3.00, IQR=2.00), “Afraid of severe medical treatments during the course of illness” (median=3.00, IQR=2.00), “Worried that treatment could damage their body” (median=3.00, IQR=2.00), “Worried about what will become of family if something should happen to participant” (median=3.00, IQR=3.00), “The thought that they might not be able to work due to illness disturbs participant” (median=3.00, IQR=3.00), “If a treatment and it is working well (limited side effects, no progression of disease), worry what will happen if treatment stopped” (median=3.00, IQR=3.00).

On average, participants scored in the **“Often”** range for the following questions:

“Worried that at some point in time will no longer be able to pursue hobbies because of illness” (median=4.00, IQR=2.00).

**Table 38: Fear of progression by questions**

Fear of progression (n=370)	Mean	SD	Median	IQR	Average response
Becomes anxious thinking that disease may progress	3.46	1.16	3	1.00	Sometimes
Is nervous prior to doctors appointments or periodic examinations	2.94	1.25	3	2.00	Sometimes
Afraid of pain	2.94	1.23	3	2.00	Sometimes
Has concerns about reaching professional and/or personal goals because of illness:	3.27	1.37	3	2.00	Sometimes
When anxious, has physical symptoms such as a rapid heartbeat, stomach ache or agitation	2.80	1.27	3	2.00	Sometimes
The possibility of relatives being diagnosed with this disease disturbs participant	2.77	1.44	3	3.00	Sometimes
Is disturbed that they may have to rely on strangers for activities of daily living	3.07	1.34	3	2.00	Sometimes
Worried that at some point in time will no longer be able to pursue hobbies because of illness	3.39	1.33	4	2.00	Often
Afraid of severe medical treatments during the course of illness	3.02	1.30	3	2.00	Sometimes
Worried that treatment could damage their body	2.99	1.31	3	2.00	Sometimes
Worried about what will become of family if something should happen to participant	3.29	1.42	3	3.00	Sometimes
The thought that they might not be able to work due to illness disturbs participant	3.16	1.55	3	3.00	Sometimes
If a treatment and it is working well (limited side effects, no progression of disease), worry what will happen if treatment stopped	2.63	1.39	3	3.00	Sometimes
Anxious if not experiencing any side effects think it doesn't work	1.76	1.11	1	1.00	Never



**Figure 23: Box plot fear of progression by question**