

## **Section 9**

### **Expectations and messages to decision-makers**

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

### **Expectations of future treatment**

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

### **Expectations of future information**

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

### **Expectations of future healthcare professional communication**

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

### **Expectations of future care and support**

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

### **What participants are grateful for in the health system**

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

## Values in making decisions

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

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

## Values for decision makers

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

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

## Time taking medication to improve quality of life

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

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

## Most effective form of medicine

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

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

## Messages to decision-makers

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

## Expectations of future treatment

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

### Future treatment will be more affordable

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

*Participant 013\_2023AUORC*

*Well, any new treatment that helps?...That isn't cost prohibitive, it would be welcome. But most people that have Scleroderma are limited in their income and so you know, these new treatments, like I said, there was that treatment they wanted to put me on that was \$46,000. Well, you could sell your house, but how long does that, how long does 40,000 last?*

*Participant 021\_2023AUDIS*

*With regards, I mean I feel very fortunate that I'm, I was approved for the Humira and the fact that it for me fortunately it's working better than I expected it to and I'm you know, I'm, I'm also fortunate that I'm in a financial position that I can afford to pay for it, whereas there are a lot of people out there that would have to. Would have to go without and we're lucky in that you know once it's approved through Medicare we only paid the dispensary cost. So you know in that in that regard you know there are aspects of I, I think I would really like to see.*

*Participant 005\_2023AUDSK*

### Future treatment will be more effective and/or targeted (personalised)

*It'd be good to see, in terms of medications, because I'm not sure if that's the only kind of treatment you're talking about, but in terms of medications, it would be reassuring if there were some that weren't just off-label use because nearly all the medications that we have that, that people with Ehlers-Danlos talk about is nearly all off-label use.*

*Participant 001\_2023AUDPA*

*So at the moment for instance speech, allied speech therapy, the wait list to get on to someone that actually understands this condition because there is specialist ways to treat but it's not just a general speech therapist that's required needs to understand the condition because' they have palate differences. So it's not just a regular speech pathologist that can do this they have to understand the communication disorders that go along with it as well. So that speech is probably the one of the biggest where we need more understanding.*

*Participant 025\_2023AUDPA*

*But I need some improvement. I mean, as I said, since I've been seeing my local doctor, well, the lumps kind of, he has put me take the job and put me on different things all the time. At the moment I'm not taking anything regularly except for, I mean I take generic, I take painkillers, zink and vitamin C and that kind of thing. But apart from that I'm not taking anything else.*

*Participant 023\_2023AUDSK*

Future treatments will include having choice (including availability/accessibility) and transparency/discussions in relation to treatment options (pathways)

*Yeah, yeah. Look, I suppose cost, cost is certainly a, a challenge or a barrier for some I think access to information about. What the options are and what the possible side effects of each pathway so that you can make informed decisions about what you're willing to, what you're willing to risk, but also kind of what your probabilities of success are. Yeah, I think, I think more information and more knowledgeable practitioners.*

*Participant 007\_2023AUDSK*

*So for new treatments, I would like to see obviously you know, if they could be subsidized. So the cost, the way that they are administered, you know, to be frank with you, like if it was an injection I wouldn't even care, at least it's something. So if it was oral or injection, I it wouldn't matter. You know, for something like really invasive, then yes, that would matter. But I couldn't imagine that being the case. I would like for health professionals, so for example, our hospital care team, to be aware of it and to talk about it and to offer it. Something that can be given in the home would be really important as well, just to not impact daily life.*

*Participant 021\_2023AUORC*

*So I'd like to see reduced side effects for most in the sense of a number of the treatments that are available to NAME have a significant side effect on reproduction health. She's nine years old and you know, not in the brain space right now to make be making decisions that may impact her in 10, 15 years, 20 years time when she may regret a decision that I've made or she actively made at nine years old that that might have an impact on the life that she wishes to have for herself. So, so a more available access to what possible side effects there are for an appropriate age group. So when you go and get your eyes done, they have a little kid booklet that tells you all about what they're going to do. Why isn't there any? Very simple, very direct communication in the drugs in the studies that target the age the under 12 age group if that's the demographic of the study or the treatment that's going to access it. Like there's lots of mumbo jumbo for parents to decide on but there's, there's no active information for NAME's brain capacity or.*

*Participant 080\_2023AUDIS*

**Future treatments will have fewer or less intense side effects/more discussion about side effects**

*So it would be nice if new treatments also considered more seriously. That the lived experience of a side effect is different perhaps, to the medical definition of a side effect. Participant 024\_2023AUDIS*

*Cost is definitely a big thing. The side effects to meds is also a thing like, how one medication could make another medication play up, or being on steroids. The weight gain, the reflux, the insomnia, how all that affects everything. Yes, side effects and cost are the main things I'd like to change, and the wait list. Like waiting for everything, because like in that time while I'm waiting, I'm declining more and more.*

*Participant 023\_2023AUDIS*

*So they need to be low cost, they need to have zero side effects and then they can most importantly make a difference like in terms of a substantial improvement like. Yeah, I think for me, I don't want something that might, you know, give me 10% less fatigue or even 20% fatigue because fatigue is fatigue. Like, if you still got 80% fatigue, which means you can't work and you can't do a lot of things, it's, you know, it just might mean I get one extra shower a week or something. Like, really, in this game of things, it's not enough.*

*Participant 010\_2023AUDIS*

**Future treatment will be easier to administer and/or able to administer at home and/or less invasive**

*If they had advanced that way that I could take a tablet and not have to have the amount of needles that I do. Yes, that would be good. Other than that, I don't really know. What else? If they had a cure, it would be great too. [laughs]*

*Participant 006\_2023AUDNS*

*Well, I would love an extended-release treatment or something that we only have to sort of give once or twice a day as opposed to every six hours they do have. They do actually have that available overseas. It's a 12 hour release, but it's, it's way too expensive for them to which I can understand, it's just it's not cost effective for them to get that here. But yeah, I guess yeah that would be the main one. Just I think. Yeah. NAME tends to sleep through. She sleeps through it when I give it to her now and the night and whatnot. But just I think. I don't know, as she gets older, it'd be nice for her not to get have to get up herself to do it, you know, just take it once and forget about it, yeah.*

*Participant 015\_2023AUORC*

**Future treatment will involve more clinical trials (including to access new technologies and treatments and funding)**

*Cost isn't an issue, I don't think. I think if you're looking at your life, you know it cost I don't think some a crazy issue as in we live in a pretty good place in Australia and we've got really good clean hospitals, clean places to live, all that kind of stuff. So yeah, that's not an issue. I think it's more. I think we need sort of more research on the...the rare forms of things that and this is this is unbiased obviously because you know...I want someone to bloody find what's wrong with you know you know you know how to kill it or how to how to deal with it for the rest of my life. So*

*yeah, my main concern would obviously be more research being done ... scenarios that obviously lead to, you know, clinical trials or positive outcomes for patients later down the track kind of things, yeah.*

*Participant 024\_2023AUORC*

*There's an incredible drug that treats cystic fibrosis that's been developed. But unfortunately it's not approved for youth, but under the age of 6 years old. So I'd like to see it approved down to the age of two. It's called Trikafta, so that I'm really putting a lot of effort into trying to advocate for Trikafta from 2 to 5 year olds at the.*

*Participant 029\_2023AUORC*

*Yeah, probably what I'd like to see particularly within Australia is, is probably more research so that we get a better understanding of. Why the condition occurs? Yeah, I guess that would be that would be probably my, my ultimate, you know, I think. You know not, not all medication works for everyone. So we can bring out a new medication and that may work for me, but it may not work for someone else. So I think ultimately for me, I would like to see more research into the condition and so that we can get a better understanding of why the condition is impacting so many people so.*

*Participant 001\_2023AUDSK*

**Future treatment will involve a more holistic approach**

*I would like to think that there's not a pill you have to take every day. Like I think there's something else that they can come up with and so maybe like less frequent medication like a tab, a needle or like a vaccination kind of thing, you know, a bit more like annually or biannually or something. I think that there needs to be more support, like more checking in, like whether it's a phone call or face to face.*

*Participant 078\_2023AUDIS*

*PARTICIPANT: Just holistic and family based care. Just understanding that a child is not just, you know, a pair of eyes or pair of ears or you know.*

*INTERVIEWER: But like how?*

*PARTICIPANT: All of those things interact with each other, yeah.*

*Participant 018\_2023AUDPA*

*What I would most like to see is a setup like the, the at the Children's Hospital, a specialized center where you get to see everybody in one day rather. So you still have you and that they do a like a holistic plan so they*

*work out the you know I need extra physio or extra OT and then you go off to your community person and yeah. So yeah, there's no consistency between the States and that's what I'd like is a national approach that it doesn't matter where you are, if you go somewhere you're going to receive the same standard and same type of assistance.*

*Participant 026\_2023AUORC*

**Future treatments will allow for a normal life/quality of life**

*Look, I think a reduction in side effects that he experiences, I guess as a result of his condition more than anything. So I mean if treatment's able to help give him some, you know, improved quality of life because I guess there's only so much we can do from a behavioural support you know, developmental sort of side of it. So if there's something that can help him to be able to get a bit of clarity and think a bit more clearly and, you know, be less impulsive and be calmer, happier, more content within himself, that would be fantastic.*

*Participant 031\_2023AUDPA*

*Reduced of potential side effects would be good, but the biggest thing would be accessibility and cost. Having it so you don't have to drive to a metropolitan area like Sydney or Melbourne, which are five and six hours away respectively from where I live, and be affordable and be covered so you're not out of pocket hundreds of dollars a week or a month. To have a quality of life and contribute to the community.*

*Participant 012\_2023AUDSK*

*Well, cost is obviously a big thing for me. I think anything that greatly improves someone quality of life for a condition like this shouldn't cost an arm and a leg and I think it should be readily available for anyone to be able to afford. But in terms of administering and all that kind of stuff, I don't have any issues with the way that I'm currently managing, so I can't think of anything extra that I would like to sort of change, no?*

*Participant 027\_2023AUDSK*

**Future treatments are important but we cannot ignore awareness and education**

*I think it would be more about trying to a greater access to health professionals who have knowledge about CHARGE syndrome. I think probably the main issue I've found is finding people who are experienced with this syndrome and know what to do? Yeah. I think the main thing would be more education and*

access to informed healthcare providers. Yeah. I can't think of anything else around that.

*Participant 089\_2023AUENM*

*I guess more probably that you like not necessarily that way, but supportive groups for people who are experiencing it. Because I do sometimes feel like you're not alone, but there's only a, you know, small amount of people that understand what you're talking about. Whereas you know, for example, Down syndrome, you know, everybody sort of knows about it and you know what to do with it, I guess.*

*Participant 034\_2023AUDPA*

*Because when you're sort of applying for therapies or like applying for NDIS because it's not a well known condition, they're just like, well, what does she need, doesn't know she needs or there's not a formula for them to go back to, to look at all this is what we do in this situation and this is the access you would need to dismiss this and this. It's like well what do you need without yeah what, what is the there's that lack of understanding some more recognition of the conditions and that so that there are less sort of less explanation of all the ins and outs and what, what it affects and how it works and, and things like that. So that aware as well.*

*Participant 017\_2023AUDPA*

**Future treatment will involve a multidisciplinary approach (communication)**

*Yeah. Yeah, nothing that I can think of. The only thing I'd like to see and I believe it's already started to happen and is that that they've got, they're following like the American path where they've now got clinics who specialize in 22Q11 and they have all the specialists on board in one place. And it's all about, it's*

*all about the actual diagnosis of 22Q,11, not about the individual issues that come up even though they're all traded together. It's, it's sort of looking at it I think holistically that's really the only thing I'm glad that I'm actually seeing now even though PATIENT being her age is a little bit past all that because she doesn't have to have you know more diagnostics and all that. It's good that it's coming through like that now for the younger kids through their per Children's Hospital, so, right. Yeah.*

*Participant 024\_2023AUDPA*

**Future treatment will be curative**

*I would like to see a cure rather than treatment because I don't understand the extent of how much the treatments are effective? I know that there's two choices for me, two or three choices for me at the moment. But other than that, I guess there's obviously different outliers with people's impact to it. So I wouldn't, I would have actually said probably cure because I think we've got enough treatments out there. I don't know what part you're trying to solve for in it .*

*Participant 004\_2023AUORC*

*PARTICIPANT: [laughs] A cure.*

*INTERVIEWER: A cure? Yes, that's what we want.*

*PARTICIPANT: I read about studies like they had been able to reverse, so not only stop but reverse the progression of fibrosis in mouse models. Again, I want that for me. I want to be able to bend and move again, because that's the problem...all the arteries into my lungs have gone fibrotic, hardened and thickened and narrowed. A cure, I'm going to the top.*

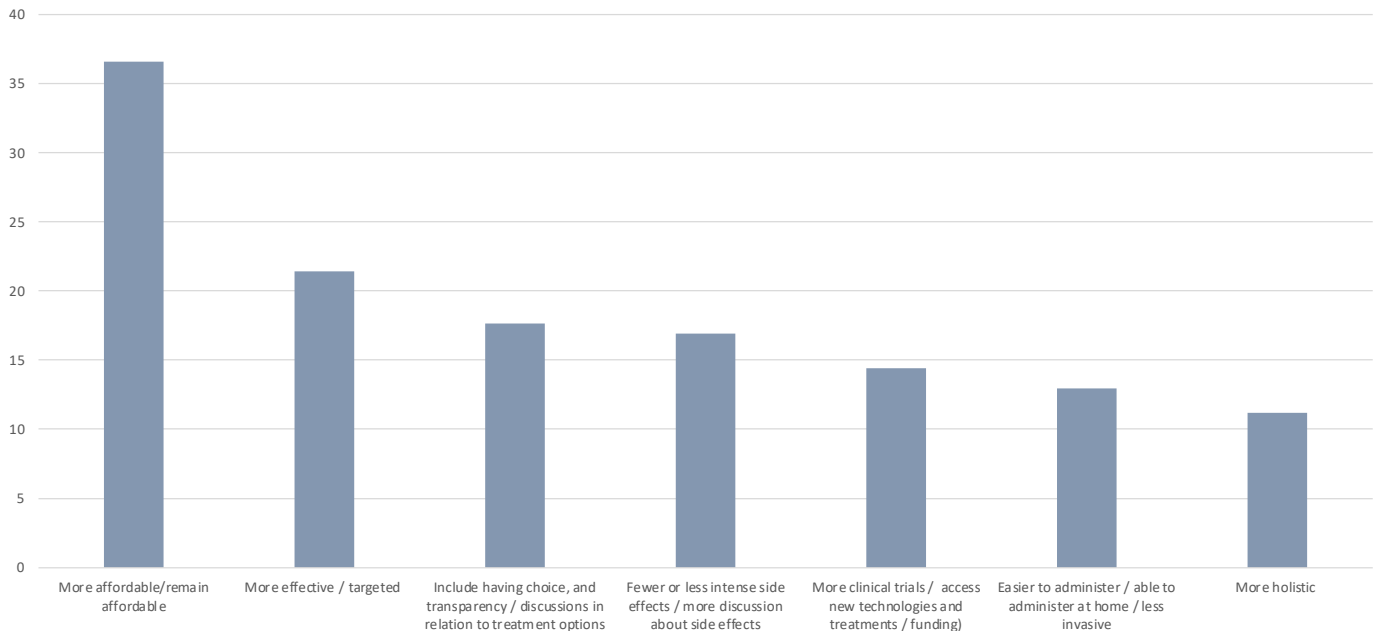
*Participant 008\_2023AUDIS*

**Table 9.1: Expectations of future treatment**

Expectations of future treatments	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
More affordable/remain affordable	147	36.57	24	35.82	33	40.74	24	25.26	17	53.13	38	40.00	11	34.38	103	38.43	44	32.84	106	36.05	39	36.79
More effective / targeted	86	21.39	4	5.97	18	22.22	26	27.37	5	15.63	32	33.68	1	3.13	69	25.75	17	12.69	64	21.77	22	20.75
Include having choice, and transparency / discussions in relation to treatment options	71	17.66	16	23.88	19	23.46	10	10.53	11	34.38	3	3.16	12	37.50	49	18.28	22	16.42	52	17.69	19	17.92
Fewer or less intense side effects / more discussion about side effects	68	16.92	8	11.94	23	28.40	18	18.95	7	21.88	6	6.32	6	18.75	52	19.40	16	11.94	53	18.03	14	13.21
More clinical trials / access new technologies and treatments / funding)	58	14.43	7	10.45	8	9.88	17	17.89	3	9.38	18	18.95	5	15.63	33	12.31	25	18.66	46	15.65	12	11.32
Easier to administer / able to administer at home / less invasive	52	12.94	10	14.93	11	13.58	8	8.42	7	21.88	6	6.32	10	31.25	38	14.18	14	10.45	39	13.27	12	11.32
More holistic	45	11.19	15	22.39	10	12.35	5	5.26	0	0.00	12	12.63	3	9.38	24	8.96	21	15.67	33	11.22	12	11.32

Expectations of future treatments	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
More affordable/remain affordable	147	36.57	31	31.96	50	38.17	39	34.21	27	45.00	76	38.38	66	33.67	37	33.33	110	37.80	68	34.00	79	39.11
More effective / targeted	86	21.39	13	13.40	29	22.14	32	28.07	12	20.00	47	23.74	39	19.90	26	23.42	60	20.62	44	22.00	42	20.79
Include having choice, and transparency / discussions in relation to treatment options	71	17.66	17	17.53	25	19.08	25	21.93	4	6.67	33	16.67	38	19.39	19	17.12	52	17.87	38	19.00	33	16.34
Fewer or less intense side effects / more discussion about side effects	68	16.92	10	10.31	27	20.61	24	21.05	7	11.67	31	15.66	35	17.86	16	14.41	52	17.87	36	18.00	32	15.84
More clinical trials / access new technologies and treatments / funding)	58	14.43	22	22.68	14	10.69	17	14.91	5	8.33	31	15.66	27	13.78	14	12.61	44	15.12	32	16.00	26	12.87
Easier to administer / able to administer at home / less invasive	52	12.94	12	12.37	18	13.74	12	10.53	10	16.67	29	14.65	22	11.22	8	7.21	44	15.12	24	12.00	28	13.86
More holistic	45	11.19	12	12.37	11	8.40	15	13.16	7	11.67	24	12.12	19	9.69	16	14.41	29	9.97	23	11.50	22	10.89



**Figure 9.1: Expectations of future treatment**

**Table 9.2: Expectations of future treatment – subgroup variations**

Expectations of future treatments	Reported less frequently	Reported more frequently
More affordable/remain affordable	Diseases of the nervous system Developmental anomalies Other rare condition Endocrine, nutritional or metabolic diseases Aged 65 plus	Diseases of the skin Endocrine, nutritional or metabolic diseases Diseases of the skin Other rare condition
More effective / targeted	Endocrine, nutritional or metabolic diseases	Diseases of the immune system
Include having choice, and transparency / discussions in relation to treatment options		
Fewer or less intense side effects / more discussion about side effects		Other rare condition
More clinical trials / access new technologies and treatments / funding)	Diseases of the skin	Developmental anomalies
Easier to administer / able to administer at home / less invasive		
More holistic		Diseases of the skin



## Expectations of future information

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

### **Future information will be more accessible/easy to find**

*I want information about it or the information from A-Z. Everything that I need to know, everything that the patient need to know. I would love to know about this. At least I can read about it. At least I can know and understand it more better.*

*Participant 001\_2023AUORC*

*It's not like we haven't got any information, it was just what the doctor told us. I think if there was something that could be provided a point of diagnosis in writing that would be really helpful or somewhere to go and look for information, so maybe be directed to a website or something. Because again, you know, you kind of have to do all of that yourself. I think information being available and relevant to the Australian community would be really important.*

*Participant 021\_2023AUORC*

*More of it and more accurate information and when you read stuff online, every site that you go to, none of them really marry up. So like some really formal type of information. That you that's readily accessible to people with the disease and certainly information on where you can get support from other people that also have the disease and others that understand the disease.*

*Participant 005\_2023AUDSK*

### **No particular comment - satisfied with information**

*All in terms of information, I think I got good information, so I don't think I want much to change consonant information.*

*Participant 006\_2023AUORC*

*No. I was pretty happy. I'm pretty happy with everything that I've I've been able to get hold of.*

*Participant 010\_2023AUDPA*

*No, I think I've always been able to find sort of the information that I needed, but I think that that's partly because most of it's on this website that was created by another parent and her child was the disease. So it's really focused on what a carer would want to see, which is probably different to if you're getting all your information from like a, you know, like a government website or something. Yeah, yeah.*

*Participant 079\_2023AUDIS*

### **Future information will provide more details about disease trajectory and what to expect**

*I think just in general there's a lack of information with HS and that would be really handy. Like even going into the surgery, they explained what the procedure was, but there was no information that I could look up and I explained this imagery is I find images really important. Not just not images. How do we how do we say this? All right. So I had the surgery and the doctors told me, Yep, we're just gonna go through we're gonna cut them open. And we're just going to lay them flat. You'll come back in in a week and we'll look at you. And one of the things that I noticed is because I've got dark skin, I was like, I couldn't tell if it was pus, but there was white all inside my in these craters. And I didn't know what they were. And so when I went to the nurse, she was just like, oh, that's just the layer of skin and it's just a little bit because of your pigment, it's just really obvious that it's white. And I was just like, 'oh, interesting', but this information wasn't widely available. There was nothing around I didn't know. I tried to look up what I was supposed to be looking at, and I couldn't find that information. And so when I ended up seeing the HS Connect roomed care, everything made sense. Just having information, obviously it's out there, it just needs to be distributed better or easily found.*

*Participant 026\_2023AUDSK*

*Some information was hard to source and even when we sourced it, it was a little bit difficult to understand how it actually affect us in a day-to-day life and some of the side effects would be written up in obviously medical language, which, you know, you think makes sense at the time. But then like one of the surgeries we did, they said, Oh, we'll do this particular surgery and it means she'll never vomit again, which was ideal*

*for us because she had such terrific reflux. But one of the side effects was afterwards is that they had to tighten her stomach so much that she actually couldn't drink water. So that was one of those things that we were like, oh, apparently we had it known. So I think the more the everyday life terms that a lot of parents sort of miss out when they're reading a lot of the information because they can't figure out how it actually affects their life.*

*Participant 032\_2023AUDPA*

**Future information will provide more details on subgroups and specific classifications of their condition**

*No. I think being pretty good presented. I think more so it's I think probably the thing I've seen the most is that because I've had this disease longer, I'm dismissed because I'm a longer patient and possibly at risk. There's more trials today for those that are in the newest stages, which I can completely understand because-- Then in equal side of things, I'm probably up to the stage of somebody that's in their six-- They don't look at the overall effects on my body as opposed to a new patient. I guess and age-wise, I suppose it's the age as well.*

*Participant 001\_2023AUDIS*

*More emphasis straight up on how different everybody can be, even though we're similar. What I find fascinating is how different everybody is, but at the same time, a lot of the same modalities and approaches and management techniques seem to work for all of us. That would be a really, really big one.*

*Participant 041\_2023AUDPA*

**Future information will help to inform the community and decision-makers about their condition (raise awareness)**

*I suppose I'd like to see more people understand what the condition cuz they just think you've got a bit of a pain in your neck. And get very knowing when they say, oh, you look really well, yeah, it's on time. That's not the problem. Yeah, that's very that can be very annoying.*

*Participant 005\_2023AUDNS*

*Oh, just more public awareness and what I said, more in the lower levels. By lower levels, I mean nurses, GP's, clinics, more awareness of the condition and more public awareness. Like when I try to explain it, some people say, Oh yeah, that's Downs syndrome.*

*And I said no, you know, everybody knows about Downs syndrome, everybody knows about this and that and the other, but nobody's ever heard it.*

*DiGeorge. I'm forever correcting and explaining.*

**Future information will be easier to understand**

*I just think the KISS principle is the most important. Just keep it simple, stupid. Because I think when people are first diagnosed they're very overwhelmed with all the information. So I think the simpler the the brochures are, the better. And the more contact information for people who can offer the people who are diagnosed support you know the you know. Making sure people feel connected if they feel overwhelmed technically, in whatever way, is simplest.*

*Participant 010\_2023AUORC*

*Very simple, very direct communication in the drugs in the studies that target the age the under 12 age group if that's the demographic of the study or the treatment that's going to access it. Like there's lots of mumbo jumbo for parents to decide on but there's there's there's no active information for NAME's brain capacity or appropriate. Participant 080\_2023AUDIS*

*It's in jargon I can understand it but you know I don't understand. You know like you look at articles and you know the specific you know you know a particular specialty and of course you know I'll know different words but as, as a therapist but I don't understand all the medical stuff behind it because it's written at that level that specialist you know of that specialty. So these are very much just like research things are looked at because you know just to see whether there is a question I should be asking if you know what I mean like yeah yeah. Could this be because?...*

*Participant 038\_2023AUDPA*

**Future information will provide more details about new treatments and/or trials**

*No. I'd like to know what research was happening. I have no idea, you know if there is anything new or if there is research happening that may impact me. It's just like there's void information on.*

*Participant 002\_2023AUDNS*

*I'd love to see a up to date listing of you know, the types and the genes involved. And symptoms you know because some, there's some that are very, very rare and they are actually life threatening. So it would*

*be nice and just to so because at the moment they just say 'Oh well CMT as a blanket', but there's you know four different sub subtypes with lots of. There's four different types with lots of subtypes below each one. Sit down please. So that and updated research. And you know, I'd like to see information of trials available, but including worldwide, you know, the opportunity to partake in. To be aware that a trial is about to happen and have the opportunity to partake if we choose to.*

*Participant 026\_2023AUORC*

**Future information will be more holistic (including emotional health)**

*Oh, I would love the international guidelines and to be accepted by the Australian medical community and adopted across the whole country so that we can get a uniformed approach. Instead of having this hotchpotch approach where oh we'll look at, we'll look at this. We'll look at speech today and now we'll look at immunology or we'll look at is we'll look at heart cardiac issues. We'll look at psychiatric. We need a, we need a uniformed allied health and, and clinicians coming together and working together, not just in their own discipline, but trying to understand that it, all the conditions are separate, but they all have to be managed together.*

*Participant 025\_2023AUDPA*

*We've had lots of really good Zoom conferences through Scleroderma Victoria that have dealt with a lot of really good issues like continence and intimacy and movement. I'd like to see more on mental health and mental health support. I know that's an area for millions of people with COVID affected and all that sort of thing. I could cry just thinking about what's ahead of me. I think you can't always explain it to your relatives or your husband or whatever, independent person, but again, they need to have knowledge of scleroderma. Probably more a holistic approach, you see one doctor for one area, and as I said, they treat each symptom virtually individually*

*Participant 008\_2023AUDIS*

*I think the stuff around intimacy and family support for others. When I started actually looking at that and seeking more information around that, it was like, oh, I thought I should have been thinking about this earlier. It's probably the stuff around quality of life rather than medical diagnosis and treatment. What other things can be useful for that for joy and happiness and all that thing, rather than just stopping degree disease progression and stuff.*

*Participant 026\_2023AUDIS*

**Future information will provide more details about symptom and side effect control**

*Anyhow, emails or brochures, anything like that, or like meetings would be amazing and I'd love to see someone do something on diet because nothing-- when I was at the support group, nothing came up about diet and I'd love to know if there are things that can help, you know, improve joint pain or fatigue and really focus on that would be amazing.*

*Participant 007\_2023AUDIS*

*Like I said, I think the information doesn't necessarily acknowledge the spectrum of impact. Yeah, It kind of talks about you know the bigger issues rather than you know when you've got a bunch of things at a milder level. So I would say that's an issue because anything that you see can be a little bit drastic and so it doesn't acknowledge the fact that these things also you know a lot of these symptoms pop up that but they're not necessarily at a clinical clinically significant level. But that doesn't mean that a parent wouldn't want to deal with them or that that that person who has that diagnosis wouldn't want to you know overcome that particular hurt or whatever it is. So I would say that's an issue and there is no one stop shop.*

*Participant 035\_2023AUDPA*

**Future information will be in a variety of formats**

*I'm very old fashioned so I'm not into all the online and support chat groups online and all that sort of stuff. I'm the old....printed brochure, talking to people face to face, and I still think that's pretty important face to face, even if you are doing telehealth and all that, nothing really beats that one-on-one to emulate the conversation, to actually get you to think about what you want to ask the doctor. I find with telehealth you always forget something and you tend to go off on tangents, I've found.*

*Participant 007\_2023AUORC*

*I think like I said, information needs to be presented in a more layman type access to. Information like handouts, little information sheets or would be good when you go to appointments and whatnot because you're given all this verbal information. How much do you actually retain once you walk out the door. So leaflets or where to access more information.*

*Participant 013\_2023AUDSK*

*I think like little videos, they are extremely powerful and I don't know that I actually have ever seen like kind of medical stuff put into layman's terms in a little video. I could be wrong. They could exist, but I haven't seen them because you know, when you like something is explained to you through little pictures and a video as if they're talking to children almost. But they're not like it's for adults. But I don't know like I've I'm a very visual person and I feel like it's easier to take on information and maintain information when you've got like it explained to in very simple terms with like little pictures and stuff you can pause and like write down almost like a little lecture I guess would be a great way for information.*

*Participant 023\_2023AUDPA*

**Future information will provide more details about the causes of their condition**

*What I'd really like to see is more research into why people get it, because I feel like there's no like, it just seems to be, you know, where as, as doctors, everyone...we're so, you know, we can make babies out of a test tube, but we can't work out why somebody has got this disease. You know, I think just more, yeah, more medical research into why and you know, why certain people get it and why other people don't. I don't know. That's what I would like to see.*

*Participant 017\_2023AUDSK*

*Yeah, the psychological impacts and what to and signs and what to look out for. I the...the fact that it's more known that it's it, you know more, more females are affected by it, it makes it hard to find information out. Between how this affects people of the different sexes even the flare zones can be different and I'm not saying that one's got it well I think actually think women have it worse than, than men cause they've got more places to get it. But having said that it's like yeah it and, and more education more information on as you were saying initially the genome testing because I'd love to know. I'd love to. I would actually. That's where I'd like the treatment to go is to maybe you know, if there could be some genetic, some genetic modification to delete it from the genetic code.*

*Participant 009\_2023AUDSK*

**Future information will be up-to-date/credible**

*Well, I must admit when I'm trying to digest something, I prefer it to be in hard copy than reading it online. I do read things online, but I tend to skim and I don't take it in as well as if I've got a hard copy that*

*I can go back to and mull over. I guess being able to ask questions of people who are informed, not who are ill-informed is another thing, people who you actually trust to know the answers.*

*Participant 004\_2023AUDIS*

*PARTICIPANT: Well, pretty much what like the underlying issue of the condition is our hair follicles under our skin, like that's what causes. Our condition. I mean, I want that to pass everywhere because all those doctors have misinformation saying we're overweight, like we don't shower enough and all this, blah blah, blah. Like I'm very hygienic. I had two showers a day only because recently I found only having one wasn't enough and I just felt gross still. But yeah.*

*INTERVIEWER: It sounds like you've got some solutions you're trying, which is good.*

*PARTICIPANT: Yeah, and like, they work for a little bit, but then, you know, go stop and change, see what works.*

*Participant 003\_2023AUDSK*

**Future information will include the ability to talk to/access to a health professional**

*Well I think the information brochure that they put in, they put out is really is good. It's very fun [sarcastic tone] I guess it's just I guess I'll go back to... it's just the communication with your specialist and GP. Yeah just and regular communication not wait for your appointment.*

*Participant 019\_2023AUDIS*

*PARTICIPANT: Yeah, no, look, not really, but like I said, NAME, that nurse that was at the clinic, the square during the clinic, once I did have my office. She was a great form of education. She was just absolutely wonderful and I think we need someone like her with her knowledge, yeah, to be available to help.*

*INTERVIEWER: That would be wonderful.*

*PARTICIPANT: Yeah, she, I really she did teach me a lot. And she, she, you know, she, she gave me a lot of like of where I could go and get all my dressings and my wound care, you know, she, she was like, I can't, you know, praise her enough.*

*Participant 015\_2023AUDIS*

*Know if there was a dedicated clinic to complex care, I mean even multisensory care would cover a whole lot of rare diseases, which would have a whole lot of similar issues that families are facing. If there was a that's where the information you would have one hub, you know one hub, they would know the experts. They*

could get that information for you. They could certainly help you on that pathway. There would be dedicated telehealth nurses coming out of that clinic who would be invaluable with the information that they would have access to. But I don't understand why that we don't have more clinics. We've got diabetic clinics. We've got bloody. What's the other one? Diabetes clinics. Sorry. You know, there's CP clinics.

Why isn't there a multisensory impaired clinic? You know, when there's so much challenges for them in their early years to get to get forward, I just, you know, I don't need to take.

Participant 028\_2023AUORC

Table 9.3: Expectations of future information

Expectations of future information	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
More accessible/easy to find/linkage to service	96	23.88	5	7.46	20	24.69	42	44.21	8	25.00	12	12.63	9	28.13	60	22.39	36	26.87	80	27.21	16	15.09
No particular comment - satisfied with experience	58	14.43	6	8.96	14	17.28	9	9.47	4	12.50	17	17.89	8	25.00	43	16.04	15	11.19	41	13.95	16	15.09
More details about disease trajectory and what to expect (incl. costs)	49	12.19	8	11.94	11	13.58	8	8.42	3	9.38	12	12.63	7	21.88	33	12.31	16	11.94	40	13.61	9	8.49
Used to raise awareness	48	11.94	4	5.97	10	12.35	9	9.47	7	21.88	14	14.74	4	12.50	35	13.06	13	9.70	41	13.95	7	6.60
More details on subgroups and specific classifications of their condition (including age-subgroups)	41	10.20	5	7.46	3	3.70	15	15.79	0	0.00	16	16.84	2	6.25	30	11.19	11	8.21	30	10.20	11	10.38
Easier to understand	32	7.96	5	7.46	5	6.17	2	2.11	5	15.63	8	8.42	7	21.88	21	7.84	11	8.21	21	7.14	10	9.43

Expectations of future information	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
More accessible/easy to find/linkage to service	96	23.88	28	28.87	25	19.08	32	28.07	11	18.33	51	25.76	45	22.96	24	21.62	72	24.74	42	21.00	54	26.73
No particular comment - satisfied with experience	58	14.43	13	13.40	20	15.27	13	11.40	12	20.00	26	13.13	31	15.82	19	17.12	39	13.40	28	14.00	30	14.85
More details about disease trajectory and what to expect (incl. costs)	49	12.19	12	12.37	17	12.98	14	12.28	6	10.00	16	8.08	32	16.33	12	10.81	37	12.71	24	12.00	25	12.38
Used to raise awareness	48	11.94	11	11.34	16	12.21	8	7.02	13	21.67	26	13.13	21	10.71	15	13.51	33	11.34	29	14.50	19	9.41
More details on subgroups and specific classifications of their condition (including age-subgroups)	41	10.20	8	8.25	12	9.16	12	10.53	9	15.00	19	9.60	21	10.71	5	4.50	36	12.37	16	8.00	25	12.38
Easier to understand	32	7.96	8	8.25	9	6.87	9	7.89	6	10.00	14	7.07	16	8.16	13	11.71	19	6.53	18	9.00	14	6.93

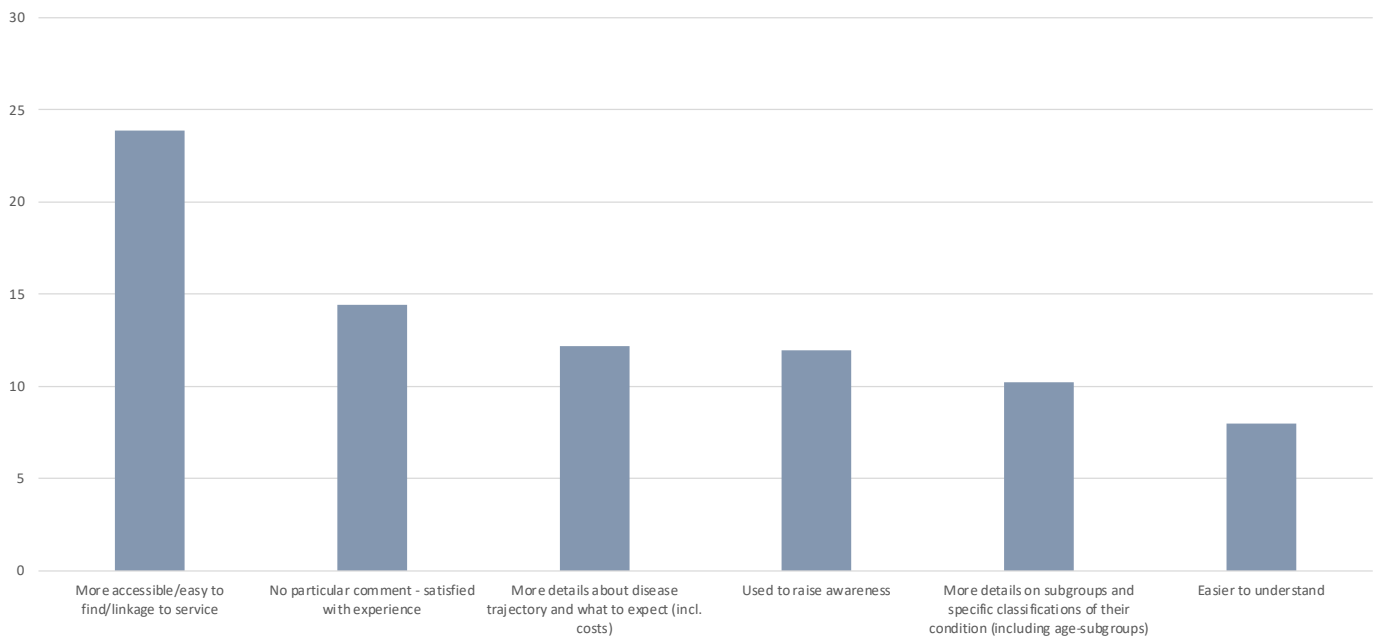


Figure 9.2: Expectations of future information

**Table 9.4: Expectations of future information – subgroup variations**

Expectations of future information	Reported less frequently	Reported more frequently
More accessible/easy to find/linkage to service	Developmental anomalies Endocrine, nutritional or metabolic diseases	Diseases of the nervous system Other rare condition
No particular comment - satisfied with experience		
More details about disease trajectory and what to expect (incl. costs)		
Used to raise awareness	Diseases of the skin	
More details on subgroups and specific classifications of their condition (including age-subgroups)		Other rare condition

### Expectations of future healthcare professional communication

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

#### Future communication will include health professionals with a better knowledge of the condition

*Oh well, I think that depends an enormous amount on the personalities involved. Some medicos are harder to deal with than others, in general, it's partly personality and language and things like that. I think it would be good if more doctors knew more about scleroderma, so when you said you had it, they knew what you were talking about, perhaps that would be something.*

*Participant 012\_2023AUDIS*

*Yes, I would like them not to Google the condition when you sit in front of them. Maybe if they if, just say, 'Look, I don't know this condition. I've not heard of it. But let me do some investigation and then I'll inform myself' But at the moment, most of the time, the parents or myself, even we go into a doctor and I've never heard that and they Google it in front of you. So if they're Googling you, what chance have we got?*

*Participant 025\_2023AUDPA*

#### Future communication will be more empathetic

*I can think of in terms of that, I mean some sort of think...some doctors have been insensitive in some of*

*their comments or the way they've spoken about some of the issues you might be facing, but I don't think that's, I don't think that's specific to charged into it. I think some doctors are just not very sensitive, yeah. That's not really specific to CHARGE syndrome, but I have experienced that in terms of my son, but I can't think of anything else apart from that.*

*Participant 089\_2023AUENM*

*In a human way, be aware that the child's in the room and can hear you, so be aware of that in the language that you use and. Maybe not focus so much on the deficit language, be accessible, and I know that's a really tricky one, but you know, try and they'll have some access availability so that people in between appointments if need a little check up can do that. But there's more in the communication. For me, it's just treating them like they're a human, not a pile of disabilities and things that need to be fixed that they have, you know, these kids and adults have, you know, feelings and they're a person.*

*Participant.95\_2023AUENM*

*Yes, don't blame me on people, people's weight and stop making them feel that something that your body creates or its own accord is their fault. The shame that is associated is insane. I know that people's diet and exercise and weight contribute. It is not something that we made ourselves have. It's not like smoking gives you lung cancer. Like, yes, it's your fault you did that. But it's not just unfit people that have this, it's not just people who can't afford to have a good diet.*

*Participant 018\_2023AUDSK*

**No particular comment - satisfied with communication**

*Not a thing. They're wonderful.*

*Participant 018\_2023AUORC*

*So far, no. I think that's just for my personal experience. I have a really good specialist, so. In my experience, I'm quite happy with the way that I communicate with my doctor, so nothing I would really change.*

*Participant 010\_2023AUDSK*

*Well, no, I suppose it depends on the doctors you get at the end of the day, so. If you get a good doctor, any, any, any, he gets on well with you and he can discuss options and, and be compassionate to you about it. I don't think there's any, any concerns but I've been pretty lucky with my referrals from my initial doctor and my current oncologist. So I think I've been pretty lucky in regards to the doctors I've got. I can't speak for everyone but yeah my, my scenario so far I've been pretty happy with my doctors so yeah.*

*Participant 024\_2023AUORC*

**Future communication will be more transparent and forthcoming**

*I would like them to explain why they make a decision and the reasoning behind it, rather than to just say, well look, this is what my recommendation is and you know, you either do it or you don't. You know to, to actually explain to you why, why a particular thing is essential or necessary. Because I think any, anyone, it doesn't matter who you are or what you are, if someone explains to you, you say to a child, I want you to go and tidy your bedroom and the kid says, well, why I like it like that. You know, if you say to them, well, because then you'll find it easier to find your toys that you're looking for, you know, it gives them a bit more motivation. But when you just do that, well, because I'm your mother and I said so, doesn't do much for communication. That's good. I, I get that kind of feeling from some of the doctors is that they do they have this patriarchal kind of thing of, well, I'm the doctor, I'm the one who's wearing the white coat, and this is what my you know what my demands are?*

*Participant 002\_2023AUDIS*

*Yeah, yeah. Was it? You see what they just say. You are all good. Your blood results are all good. See you next year. But they are not actually telling me what to avoid. What triggers it to be active. What should I do? What should I avoid? You know, you know, all those that information at least I know for sometimes, probably I'm doing something that makes it active. But I don't know, they never told me. They just say how you you're all good, not active, you're all fine.*

*Participant 001\_2023AUORC*

*Yes, be honest about the timeframe process, because that's why I said I was so ignorant. I've just gone in and gone, "Yep, yep, yep." Then again, I did jokingly say to them on the person that passed out in front of pathology lecture, "Don't tell me anything, just, just get it done." As I said, once I was on the queue, and things were moving, because you're in getting tests*

*done, and everything done week on week on week, that was one thing they did say, the good thing was on the very first visit with a rheumatologist, he spent two hours basically telling me I was potentially going to die, I'm having the transplant and I needed to understand the risks, but he did say, "Are you ready? You're going to be locked in a room and you're going to be on your own and you're going to be in there for weeks on end."*

*Participant 025\_2023AUDIS*

**Future communication will include listening to the patient**

*Oh, I'd like them not to treat us as if we're all stupid and inconvenient. I have been spoken down to more times than I care to remember by health professionals, and if I had no understanding of the words they were saying. I think a lot of the healthcare professionals and not her routine teams, although part of them, but certainly in the emergency rooms where they didn't engage me as an equal partner. They were telling me what was going to be done or telling me about her condition, but without actually providing information that would be useful and even you know. The case I had with her first year old, just when we told him that she wasn't sleeping and we couldn't get her to sleep, his response was, well, some children don't sleep in terms of some normal children don't sleep overnight. And I felt like saying, yeah, well, you come manage it then as opposed to actually looking for a solution to it. And that was really quite offensive and it is just that real, assuming that we know nothing about yeah. And I know people have different levels of understanding of the medical system and of medical terminology, but I just found they were very dismissive and often very dismissive and condescending. And I didn't didn't really need that.*

*Participant 090\_2023AUENM*

*Just the compassion side of things, I think like to know that someone's struggling so bad and they were in the beginning a little bit blasé. It wasn't until I had that second by the 1st ablation when he went in and said this is the worst case I've seen in a very long time. He was that that was the first time anyone that had, I felt like I'd been listened to properly. Do you know what I mean? Before that, it was like, I'll take this medicine, you'll be fine. It's like, no, you're not understanding. Yeah.*

*Participant 032\_2023AUORC*

*Yes. There's that saying about people with autism about like if you've met one person with autism, you've met one person with autism. I feel like the same applies to EDS, like don't make assumptions, just listen to your patients. I think also maybe like the pain scale, a pain scale thing. I've never seen anything formal about people with chronic pain, but I've seen loads of informal ones online where people have shared their versions of pain scales because I feel like it's just different. I feel like that might help be taken more seriously when you present to ED, for example and be like something's like, "My hip's out," and they're like, "Oh, your hip can't go out," and you're like, "No, no, no. My hip's out and this is where I'm sitting on the pain scale." To your GP or doctor. I feel like that would be really cool.*

*Participant 004\_2023AUDPA*

#### **Future communication will include a multidisciplinary and coordinated approach**

*Now they all seem to be talking to each other sometimes when I've been to heart specialist and then three months later I go to rheumatologist, they're supposed to like send a report on and sometimes they haven't received it for my appointment. So it feels like, well, that what that was a waste of time. Yeah, I mean, I know they eventually get it and they do read it, but yeah, I'd like to know that they're that's happening. I mean, I asked my GP and he reads them out to me. So yes. Yeah, of what? Different doctors have said so.*

*Participant 088\_2023AUENM*

*I would love to see everything put into the health portal and the patient government thing. I actually cancelled mine because I found it more detrimental than useful because so few people used it that most of my information wasn't in there and then when they did look at it, they had a very limited idea of my, my health issues. My GP wouldn't even use it. So I think if it would be an amazing tool if it was utilized mandatorily by all health professionals. But it sounds the way it is, yeah.*

*Participant 016\_2023AUDIS*

*Oh, look, in a perfect world, I'd have more consistency of the rheumatologist, but I get it's a public teaching hospital. It's just a turnover of people. In the past there has been, there was a lady there that's just retired, been there a long time, and then she was training someone else, but she's on maternity leave. I think, if there was sort of that one person that would be better than someone different each time you go.*

*Yes, they've got your file notes, but it's not quite the same.*

*Participant 017\_2023AUDIS*

#### **Future communication will allow people more time to meet with their clinician**

*PARTICIPANT: I think sometimes that, you know, because of their patient notes, you just, they're just too rushed and too, you know, you feel quite, you can feel quite just dismissed and because the next patient's coming along and yeah, like they wanna get you in and out. Yeah, that sort of thing and yeah, maybe a clearer pathway than you take this pill and they'll see you in six weeks and then you just let floundering with this lack of knowledge and information you know.*

*Participant 027\_2023AUORC*

*Yes, just communicating would be really good [laughs] just being able to have a little bit more access would be amazing. They're really good and do the best they can. I get that but yeah, it's hard. When I was diagnosed with that interstitial lung disease, they told me I had it and then you look it up and then you start to panic, and then I couldn't get hold of anyone to ask any questions so when you're diagnosed, it would be nice to be able to get information, you know, fairly straight away.*

*Participant 007\_2023AUDIS*

#### **Future communication will be more accessible to everyone**

*I think along the same lines, making sure that they're aware of this condition, and help people get the right treatment for it earlier on. I know people that were dismissed because of their condition and not given any further information and given medication that was never ever going to help them. I would like to see that information coming out to our local and our rural areas, your local GPs.*

*Participant 006\_2023AUDNS*

*Look, I've only, I've only had really good experiences with my doctors. If everyone could have that experience that would that would be fantastic.*

*Participant 032\_2023AUDSK*



**Future communication will include developing a care plan with follow-up**

*I'm really over the hospital system's rigidity in how everything's coordinated. And I know it comes back down to the state and the hospital that you're being monitored for. After being monitored in two states, I just find it really, really frustrating that you have no control over your scans, your blood tests and when your appointment is coming. I've lost all that ability to. And I'm constantly rescheduling when they give me shit time for it, especially because I have to fast for my, fast for my ultrasound. That's one thing that absolutely should see.*

*Participant 004\_2023AUORC*

*I don't think so. I think maybe just like I said before, with following up on following up on people so after their appointments. Just like a quick phone call, like saying at the second appointment after a diagnosis saying are you OK if we contact you in four weeks just to see how you're getting on with what we've discussed. Like instead of relying on the person to use, but instead of relying on the person to think about it, or for them to use the initiative, perhaps just following up. And then that way the person, like the patient, is at liberty to either answer and accept that help or they can just let it slide. Do you know what I mean?*

*Participant 029\_2023AUDPA*

**Future communication will be more holistic (including emotional health)**

*I would love them to take it seriously and not just wipe their hands, you know. Be proactive in assisting the patient to access the services that they need and have a regular follow up as to if those services are working, how they're going and if changes are needed. Whether that that start with your GP or your neurologist, but you know have a have a head person like a holistic health plan, but you know somebody that's invested in the best interest of the patient, not just the dollars that they can pay them.*

*Participant 026\_2023AUORC*

*Yes, I would say being like...just a hell of a lot more gentle with, with the delivery that there needs to be like an instant sort of healthcare support for the processing of that information and what it means. And it needs to be ongoing rather than just like here's your diagnosis, see you later that there really needs to be...the idea that, like, this isn't just something that happens to the individual, but it is also a, a whole family experience and therefore there needs to be support and mental health care for the whole family, including siblings and sibling services. Yeah, that kind of thing. Participant 018\_2023AUDPA*

*Yes. You know, I, I think they sort of just see it as their job to tell you what they need to tell you. They don't really go through how it can affect you and possible outcomes and anything like that. I think when someone's diagnosed they should be counselling or therapy offered maybe even to their carers as well, depending on if that's something they want or need. And I guess more awareness. There's been a couple times I've gone into hospital and said I've got NMO and I'm having a flare up. They sort of look at me and go, what's that? It's amazing how many health professionals don't even know what it is.*

*Participant 096\_2023AUDNS*

**Future communication will be more understandable**

*Keeping it simple. Participant 014\_2023AUDPA*

*Yeah, and that's very variable depending on the specialist. Some specialists just look and talk to me. Some specialists are good and you know and, and we'll talk to PATIENT in and, and explain things to her in a language that she understands and, and sometimes with those then I have to ask. I have to interrupt them and ask them to make sure that I've got it in the real language, if you know what I mean. So yes, so that that's, that's variable. So you know and others will talk and I'll be understanding them that I, I actually always through a consult will after they've said something. I will then put it into simpler, straightforward sentences for my daughter that is short enough to give her a picture. Like, you know, the doctor said this, he thinks whatever and make sure that she has understood it. And I look at the doctor to confirm that what I have conveyed to her is fair enough. Yeah. So I'm constantly have to do that. Generally allied health are better I think at communicating to PATIENT or the ones that we've chosen up. So, and I think also providing written information and visuals is something that is missing or that is often, Yeah. And even I find very useful when you know, when you when you're juggling so many multiple specialists and I'm a bit worn out right now, whereas once upon a time I was totally on top of everything. Nowadays, I really appreciate when they give me something written down as well, because like I've got so many other things I'm dealing with that to remember everything they've said, like I've understood it all at the time. But then you go home, you do all these other things and you think can't quite remember. So to have things that are written both at a level that I understand, but also having things in simpler format to show my daughter.*

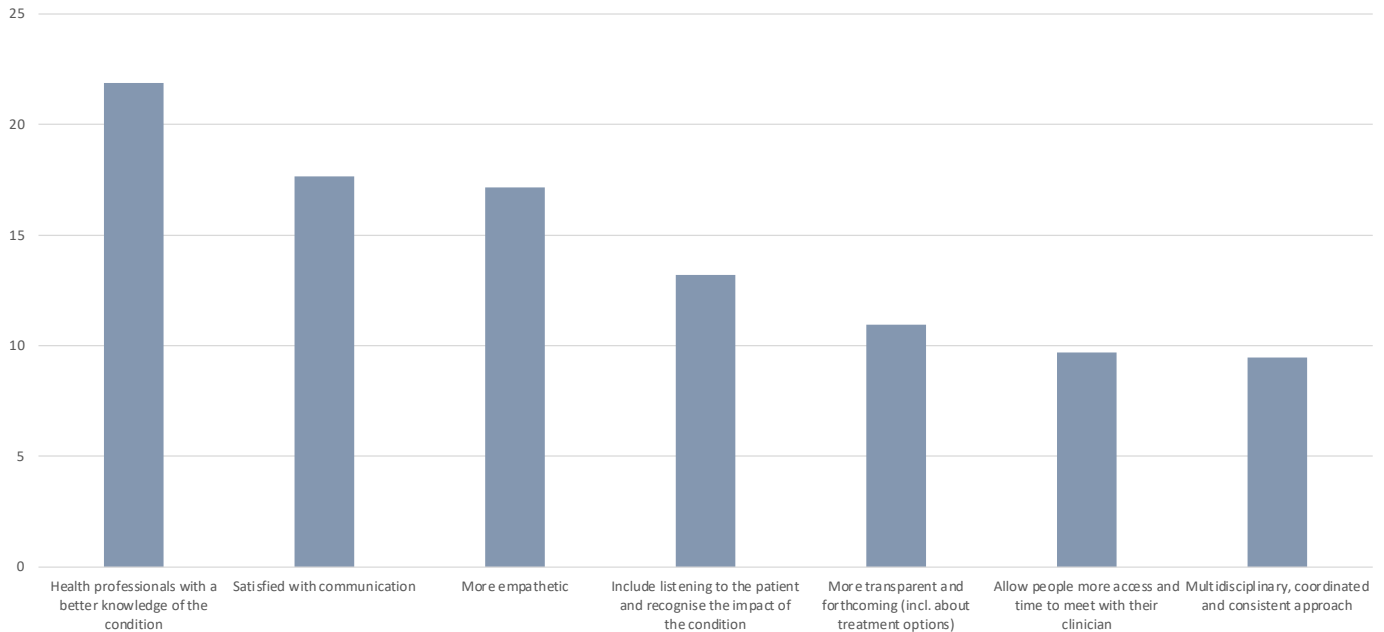
*Participant 038\_2023AUDPA*

**Table 9.5: Expectations of future healthcare professional communication**

Expectations of future communication	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Health professionals with a better knowledge of the condition	88	21.89	14	20.90	15	18.52	8	8.42	15	46.88	35	36.84	1	3.13	68	25.37	20	14.93	70	23.81	18	16.98
Satisfied with communication	71	17.66	4	5.97	12	14.81	23	24.21	5	15.63	20	21.05	7	21.88	45	16.79	26	19.40	48	16.33	23	21.70
More empathetic	69	17.16	3	4.48	17	20.99	19	20.00	10	31.25	10	10.53	10	31.25	47	17.54	22	16.42	54	18.37	14	13.21
Include listening to the patient and recognise the impact of the condition	53	13.18	5	7.46	19	23.46	7	7.37	5	15.63	10	10.53	7	21.88	45	16.79	8	5.97	43	14.63	9	8.49
More transparent and forthcoming (incl. about treatment options)	44	10.95	3	4.48	16	19.75	13	13.68	0	0.00	4	4.21	8	25.00	34	12.69	10	7.46	37	12.59	6	5.66
Allow people more access and time to meet with their clinician	39	9.70	3	4.48	9	11.11	21	22.11	0	0.00	2	2.11	4	12.50	34	12.69	5	3.73	30	10.20	9	8.49
Multidisciplinary, coordinated and consistent approach	38	9.45	3	4.48	15	18.52	3	3.16	2	6.25	9	9.47	6	18.75	30	11.19	8	5.97	29	9.86	8	7.55

Expectations of future communication	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Health professionals with a better knowledge of the condition	88	21.89	9	9.28	30	22.90	31	27.19	18	30.00	39	19.70	47	23.98	31	27.93	57	19.59	46	23.00	42	20.79
Satisfied with communication	71	17.66	24	24.74	18	13.74	15	13.16	14	23.33	33	16.67	37	18.88	20	18.02	51	17.53	30	15.00	41	20.30
More empathetic	69	17.16	18	18.56	22	16.79	22	19.30	7	11.67	31	15.66	37	18.88	15	13.51	54	18.56	39	19.50	30	14.85
Include listening to the patient and recognise the impact of the condition	53	13.18	7	7.22	18	13.74	22	19.30	6	10.00	23	11.62	30	15.31	17	15.32	36	12.37	28	14.00	25	12.38
More transparent and forthcoming (incl. about treatment options)	44	10.95	7	7.22	14	10.69	20	17.54	3	5.00	16	8.08	27	13.78	6	5.41	38	13.06	17	8.50	27	13.37
Allow people more access and time to meet with their clinician	39	9.70	2	2.06	20	15.27	12	10.53	5	8.33	26	13.13	13	6.63	7	6.31	32	11.00	19	9.50	20	9.90
Multidisciplinary, coordinated and consistent approach	38	9.45	5	5.15	12	9.16	14	12.28	7	11.67	21	10.61	17	8.67	11	9.91	27	9.28	18	9.00	20	9.90



**Figure 9.3: Expectations of future healthcare professional communication**

**Table 9.6: Expectations of future healthcare professional communication – subgroup variations**

Expectations of future communication	Reported less frequently	Reported more frequently
Health professionals with a better knowledge of the condition	Diseases of the nervous system Other rare condition Aged under 18	Diseases of the skin Endocrine, nutritional or metabolic diseases
Satisfied with communication	Developmental anomalies	Diseases of the skin Other rare condition
More empathetic	Developmental anomalies	Diseases of the skin Other rare condition
Include listening to the patient and recognise the impact of the condition	Developmental anomalies	Diseases of the immune system
More transparent and forthcoming (incl. about treatment options)	Diseases of the skin	Other rare condition
Allow people more access and time to meet with their clinician		Diseases of the nervous system

## Expectations of future care and support

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

### Future care and support will include more access to support services

*I think maybe access to the NDIS seems to be a real mixed bag on who gets approved and who doesn't. I know on the Scleroderma Australia website there are some guidelines, but if there was perhaps a person who was skilled in that who was available to assist or review applications before they went in or knew more about specific terminology or trigger things or what to say or what was not to say, I think that would be really helpful.*

*Participant 017\_2023AUDIS*

*I think charities have got better and some of the hospital systems have got better in that sense. It's funny because I've been treated when I've been in the hospital all of a sudden since, oh, you've got scleroderma. They're so like, wow, look at this. It's like some of them are really well-informed now. They know. They think it's great that they can see a patient like that, which it's good too. I think basically I've noticed that Queensland hasn't got a lot of support as opposed to Victoria and New South Wales and some other states.*

*Participant 001\_2023AUDIS*

*Just support in general, I guess you know. When in hospital because you do visit quite regularly, you know, the nurses go, oh, I haven't seen you for a little while or oh, you're back again, you know, so, so that is quite, quite regular. Yeah, I guess.*

*Participant 034\_2023AUDPA*

### Future care and support will include specialist clinics or services where they can talk to professionals (in person, phone, online)

*I mean, I know in America they have in one of the hospitals, they have an actual CHARGE syndrome clinic where there are professionals working there who are all experienced for the charge syndrome and they're all communicating with one another and working together, and I've always thought how great that sounded, that all these people came together in clinic and could work collaboratively instead of everybody working separately and searching out people with experience. Sorry, that would be pretty amazing. Someone else, yeah. I mean, communication is a big issue in CHARGE syndrome, so I've always wished that there was a service that had a instead of attending the speech general speech pathology like we do for people that are vision and hearing impaired, always thought it would be great if there was some sort of service that again where people had experience with CHARGE syndrome was focusing on communication in CHARGE syndrome, which is a bit different to somebody who's just vision and hearing impaired because it would take into account the other Physical impairments that you might have, yeah. I don't know. Nothing else really comes to me though apart from those two.*

*Participant 089\_2023AUENM*

*I've been thinking about this interesting development in. One Danish city from one Danish hospital. They offer a 24 hour clinic. Something like that needs to happen in Australia for the future because there's people like me who just live in misery. 24 hour care the 24 hour clinic, a specialized clinic from doctors and nurses who know all about it.*

*Participant 008\_2023AUDSK*

*Yeah, like proper support services, not just a group of people on Facebook that all have the same disease. So a network where you can actually. Like I've had, I've had times when I have had flares that are out of control. I can't, I don't know what to do. Like I've reached the point where I've literally, I end up at, at emergency because I don't know what to do. So, you know, having a support service, you know, like I don't know if you have it up in LOCATION, but like we have like a one 800 number in LOCATION that you can actually speak to registered nurses. Yes. Yeah. Having something like that specific to the disease that you*

*can make contact with someone and say, okay, what do I do? Yeah, yeah. Have I reached the point where I don't have a choice, I have to go to hospital or is there something else that I can try?*

*Participant 005\_2023AUDSK*

**Future care and support will include a multidisciplinary and coordinated approach**

*Again, that's a really hard question to answer specifically because it can be so broad. But even just in terms of treatment, access to more specialists, I mean, we struggle even accessing paediatrician and that shouldn't be that hard. But it is what it is. Yeah, just access to specialists really in a timely fashion is very major, yeah.*

*Participant 021\_2023AUDPA*

*Yeah, I definitely think like the dietitian support. Yeah, maybe that's just because I need to organize it, but that support for like ongoing management of someone who's having a restricted diet. Yeah, I think that that would because it's not a focus of it. It's not a focus of the gastroenterologist necessarily. But yeah, yeah, yeah, that's someone that maybe like it's maybe it's like more regular follow up to someone like that because you can always access it as a parent. Like if I book an appointment I can always access the service, but I don't. Yeah. But there's sort of no one doing that. Sort of following up to make sure that, yeah, like if I wasn't proactively doing that, nothing would happen.*

*Participant 079\_2023AUDIS*

**Future care and support will include health professionals with a better knowledge of the condition**

*Practitioner education, which is a bit more like, inside the Australian health system EDS Echo kind of thing. Practitioner education and client support. I don't know how else to put that. Support options for -- it's really hard because you can't create community out of a vacuum either. You kind of can for example, I tried to join the Australian Ehlers-Danlos thing. I signed up so many times and just keep getting these errors about your password and your thing. Then I tried to email someone and never got a response. It's like, does it actually exist?*

*Participant 001\_2023AUDPA*

*Maybe just awareness to more doctors and hospitals. I think when I was younger I saw a GP, they didn't even know what they were looking at and I think if they had*

*of known, I probably would have been able to get treatment like proper care a lot sooner. Maybe having a better skin clinic services within the public hospital so that people who have this disease don't have to fork out thousands and thousands to receive treatment for something that, you know they're born with, that they can't help that they have.*

*Participant 010\_2023AUDSK*

*I think the big thing, particularly being from a region is that the hospitals. Have the ability to tap into the specialists and are willing to because I've found sometimes I think oh no, well you're our patient and we're going to tell you what to do and you'll do it our way. And then you talk to the specialist in Brisbane they say well actually you should be doing this. So you you're sort of getting mixed messages. So that's probably something that needs to be worked on is educating regional hospitals and service providers about, you know, if there's any doubt, you don't decide, you get in touch with people that know rather than make a decision and then you've got two different opinions being given to the client.*

*Participant 007\_2023AUORC*

**Future care and support will include being able to connect with other patients through peer support (support groups, online forums)**

*I think I feel like something that was really lacking was the capacity or, or the information or like the invitation. I don't know whether there's anything out there around a support groups with people that have rarer set of conditions that can be that can be very isolating. I don't...it's possible that that's accessible in more metropolitan areas?...I'm not sure, but certainly out here in this rural space is very isolating.*

*Participant 007\_2023AUDSK*

*It would be lovely. It would be great to see more. I would like to actually see some, some more media stuff so that HS is normalized a little bit more. Yeah. So I would love to see some stuff on media so that it was it was normalized. That the community understood that the condition exists and that there were a lot of people in the community with the condition. I would like to see peer support groups. Maybe develop like face to face groups in communities would be great, particularly if there's communities that have, you know, high concentrations of the condition. I think, yeah, to be able to meet with others that understand and are walking that journey is absolutely valuable.*

*Participant 001\_2023AUDSK*

**Future care and support will include practical support (home care, transport, financial)**

*Our biggest cost comes from accessing poor quality dressings at astronomical prices, and we don't have access to specific dressings that have been created. But they're not approved or they're not allowed to be in Australia. So we're left without and it's not really fair. And Medicare won't cover any dressings at all. My previous surgeon, he tried. He tried to get me to go use the community nurse so that dressings will be covered, and every time he'd refer me, I was disbarred almost immediately from the service. Because you can dress yourself. You don't need the community nurse to dress you. Medicare doesn't cover dressings. It covers the nurses nurse doing the dressing, which isn't fair. It isn't right and it's now put us in this in more. In my instance, it's put us in a financial position where I have to. Cut back on groceries to cover dressings.*

*Participant 012\_2023AUDSK*

*Specific services. Maybe to service scleroderma, I'm talking for myself also, I need help to go out. I need help when I have to take an Uber to go somewhere to an appointment, I always have to ask for help, because I can't open the door, I can't put the seatbelt on. Even at the hospital, I think when you go to the hospital, there should be some people there, some attendant or, I don't know. When they see people with scleroderma coming at the reception over desk, they should be able to ask that person, "Do you need any help with the door? Do you want me to take you somewhere?" Of course, every time I go to hospital, I have to tell them I can't write, but they can see my hands so [unintelligible 01:00:26] [chuckles]. "Okay, don't worry, I will tell the doctor to fill up the application form for you." Maybe they should be observing more what's around them, pay more attention to disabled people because...I don't know what else to ask for.*

*Participant 020\_2023AUDIS*

*I think the cost of dressings and support that way. So nurses at GP clinics where you tend to go educate there, they have very limited dressing supplies, chemists don't have much and their prices are exorbitant. So like hospitals or the GP clinic, they need to help you access a wider range of dressings at a reasonable price.*

*Participant 013\_2023AUDSK*

**Future care and support will be more holistic (including emotional health)**

*The mental health services, I believe we all need them. Like, even if we say we don't like, when I first got diagnosed, I probably would have said no, I don't need that. Then like thinking about it, living with it, like with the diagnosis. And I'm like, yeah, OK, now I feel pretty crappy about myself.*

*Participant 003\_2023AUDSK*

*We've got I'd, I'd love CMT to have CMT Australia the support to be able to offer. You know, credible and good referrals, you know, to know this is if you're in this state, this is who you go to see, which is where we are looking at employing a Telehealth nurse to, to help. Begin the process of looking in. Yeah, starting these holistic health care so that, you know, it doesn't matter where we are, they can link us with services that we need.*

*Participant 026\_2023AUORC*

*I think the psychology services are really lacking in rural areas. So I think PATIENT could have done with more and, and that wasn't even really mentioned in this in Rome or in in by any of the health professionals that. I've, you know, I've come across writings that PATIENT'S written when I've been cleaning up and she's been very depressed. And so I think she did need a lot more support herself psychologically in her teens and her early adult that she wasn't getting and I wasn't really aware of. So to me that was a really big. A bit missing out of the whole thing because I think she went through a really hard time herself and was very, very lonely.*

*Participant 09\_2023AUDPA*

**Future care and support will include charities specific to condition**

*Well, I don't even know if there...I'm not aware if there even is a charity for Q22. I don't even know. That's bad. I don't even know if there is a charity, but. Yeah, maybe just I think the main thing I would like is just more awareness of the condition out in the community as a general. So whether that's, yeah, I didn't know the condition existed until I found out it was in my family. So just more general public awareness of it and the effect it can have because there are a lot of people out there who have it, who don't know they have it and might just think, like I did, that they're just not very clever.*

*Participant 029\_2023AUDPA*

*There is a CHARGE Syndrome association. I would like to see them get some funding because it it's, yeah, pretty much run solely by passionate volunteers who are all relatives of someone with CHARGE. But because no one knows about CHARGE, it you know, it doesn't attract any funding. It would be great if there could be some, some sort of cohesive umbrella for groups like that that you know they're so rare that it's very hard for them to attract notice and funding. But in a lot of ways they're more critical because those diseases are so rare. So, you know, if they could all kind of come together and I don't know, maybe get assistance so that they're, they're not all trying to replicate things that each group does. You know, like some admin stuff or you know, website maintenance that maybe that could all be funded better so that each little group isn't trying to do everything themselves.*

*Participant 091\_2023AUENM*

*It would be great if they had did have more charities in relation to 22 Q or you know, even just start talking about it in the media, you know, be more like bring it out more, you know, make people aware, you know, I mean they, they do it with, well, they started doing it with autism, haven't they? So just, yeah, making people aware of 22 Q.*

*Participant 028\_2023AUDPA*

### Future care and support will include more family and carer support

*Gosh, I've never really thought about it because it doesn't exist. It would be nice like a support system because they do have the clinic at our hospital and we're lucky that we do have that, but they isn't actually like support services for the family in that scenario. There's like it's a referral system pretty*

*much. So, you go there and they have a look at the kid and then they refer you to all the specialists that you need. So, I think the like the social support at home of how do you actually live with this condition is sort of missing there.*

*Participant 032\_2023AUDPA*

*Look, I think the, the thing that I sort of see in...like you know, the younger generation of, of babies that are being born now is just over and over severe parent overwhelmed, particularly when there are babies who are in hospital for long periods of time and I think there needs to be options for parents to be with critically ill babies living with them, you know, like that, yes, there's a room that's dedicated nursing or whatever it is. But like, in order for that attachment to form and really like the, the kids who have an attachment with CHARGE can be pulled out of themselves and do so well. They learn to speak, they learn to communicate, they learn to be like little amazing pocket rockets. But the kids that don't remain nonverbal and you know, don't develop social skills and, and that kind of thing and, and it's just like not fair. It's just not fair on them, yeah, so...You're just having someone also to support the parents with how you bond with a child who you can't give cuddles and kisses and tickle in the tummy and, you know, give them their fair foods and all of that. Yeah, beautiful.*

*Participant 018\_2023AUDPA*

*Yeah, I think that the carers of people with cystic fibrosis should receive some sort of government support funding, some funds for whether it's lost wages or that sort of thing. I know it's not considered a disability, but the level of care is still so high that I feel it should be considered for NDIS or something similar, yes. Yep.*

*Participant 029\_2023AUORC*

**Table 9.7: Expectations of future care and support**

Expectations of future care and support	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
More access to support services	92	22.89	13	19.40	30	37.04	24	25.26	5	15.63	15	15.79	5	15.63	63	23.51	29	21.64	70	23.81	21	19.81
Multidisciplinary and coordinated approach (incl. upskilling of staff)	59	14.68	7	10.45	15	18.52	14	14.74	0	0.00	20	21.05	3	9.38	38	14.18	21	15.67	48	16.33	11	10.38
Specialist clinics or services where they can talk to professionals (incl. nurse specialists)	56	13.93	6	8.96	10	12.35	17	17.89	9	28.13	7	7.37	7	21.88	40	14.93	16	11.94	38	12.93	18	16.98
Peer support, support groups and online forums	47	11.69	7	10.45	4	4.94	9	9.47	8	25.00	12	12.63	7	21.88	37	13.81	10	7.46	36	12.24	10	9.43
Health professionals with a better knowledge of the condition	39	9.70	8	11.94	20	24.69	5	5.26	3	9.38	1	1.05	2	6.25	32	11.94	7	5.22	35	11.90	4	3.77
Practical support	32	7.96	4	5.97	5	6.17	7	7.37	6	18.75	5	5.26	5	15.63	21	7.84	11	8.21	28	9.52	4	3.77
No particular comment - satisfied with experience	32	7.96	0	0.00	10	12.35	7	7.37	0	0.00	15	15.79	0	0.00	27	10.07	5	3.73	22	7.48	10	9.43

Expectations of future care and support	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
More access to support services	92	22.89	16	16.49	33	25.19	28	24.56	15	25.00	45	22.73	43	21.94	26	23.42	66	22.68	55	27.50	37	18.32
Multidisciplinary and coordinated approach (incl. upskilling of staff)	59	14.68	16	16.49	16	12.21	20	17.54	7	11.67	24	12.12	34	17.35	24	21.62	35	12.03	32	16.00	27	13.37
Specialist clinics or services where they can talk to professionals (incl. nurse specialists)	56	13.93	12	12.37	15	11.45	19	16.67	10	16.67	26	13.13	30	15.31	10	9.01	46	15.81	27	13.50	29	14.36
Peer support, support groups and online forums	47	11.69	5	5.15	16	12.21	15	13.16	11	18.33	27	13.64	20	10.20	14	12.61	33	11.34	17	8.50	30	14.85
Health professionals with a better knowledge of the condition	39	9.70	5	5.15	16	12.21	12	10.53	6	10.00	17	8.59	22	11.22	16	14.41	23	7.90	27	13.50	12	5.94
Practical support	32	7.96	9	9.28	12	9.16	10	8.77	1	1.67	16	8.08	16	8.16	5	4.50	27	9.28	14	7.00	18	8.91
No particular comment - satisfied with experience	32	7.96	4	4.12	8	6.11	11	9.65	9	15.00	19	9.60	13	6.63	8	7.21	24	8.25	14	7.00	18	8.91

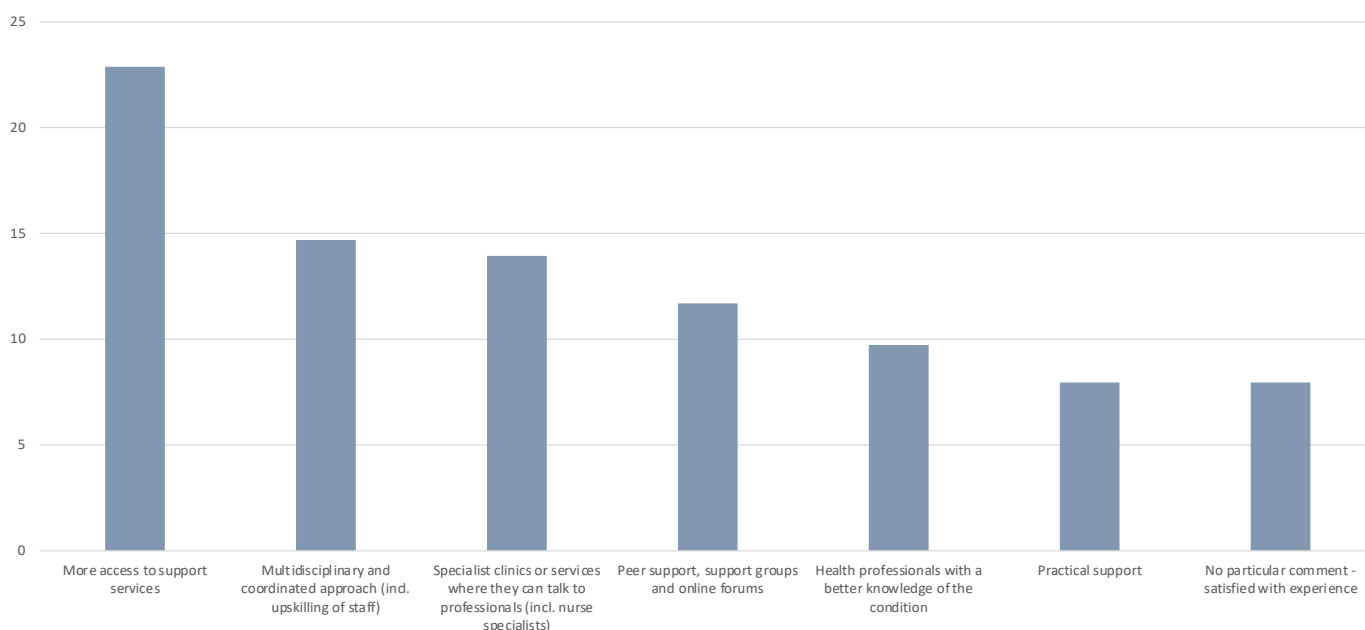


Figure 9.4: Expectations of future care and support

Table 9.8: Expectations of future care and support – subgroup variations

Expectations of future care and support	Reported less frequently	Reported more frequently
More access to support services		Diseases of the immune system
Multidisciplinary and coordinated approach (incl. upskilling of staff)	Diseases of the skin	
Specialist clinics or services where they can talk to professionals (incl. nurse specialists)		Diseases of the skin
Peer support, support groups and online forums		Diseases of the skin Other rare condition
Health professionals with a better knowledge of the condition		Diseases of the immune system
Practical support		Diseases of the skin
No particular comment - satisfied with experience		

### What participants are grateful for in the health system

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

### Participant describes being grateful for healthcare staff (including access to specialists)

*Yes, the nurses. The nurses are amazing. From the transfusion nurses, to the nurses that I do my six minute walk test with, to lung function nurses. All those little people, they do all the hard work. I take my hat off to them like, they are the best people because they've seen me progress to worse and like, "Oh, NAME, you've improved your six minute walk test." Those people that do those tests, and the nurses that*

*look after you in hospital, they are great people. I take my hats off to them any day.*

*Participant 023\_2023AUDIS*

*We used to have an adult cystic fibrosis ward at the HOSPITAL and it had specific cystic fibrosis nurses there. So they knew what our medications were. They knew they just were trained specifically in cystic fibrosis. So I found it really helpful that they had that there and that the nurses were not understood and they didn't have to ask all these stupid questions that other nurses do. They don't have that ward anymore. It's now a COVID ward, but when they did have it, I found it really helpful and that was great. And then also that they have a cystic fibrosis specialist team. Yeah, I think that's great as well.*

*Participant 013\_2023AUORC*

*My cardiologists have been amazing. Those last couple were really, really, really good.*

*Participant 032\_2023AUORC*

**Participant describes being grateful for low cost/free medical care through the government (Public health system in general)**

*Well, I have been grateful that a lot of the surgery that the surgeries I've had so far have not had to pay for, but they have been covered by Medicare when I've gone to the hospital, but that. That's where my gratitude ends, because from my experiences, from my accessibility, the healthcare system, the only thing I can be grateful for is that we are not yet an American healthcare.*

*Participant 012\_2023AUDSK*

*I guess, yeah, the fact that we do have a Medicare system that's yeah saved me a lot of money overall. Yeah. So, I'll say I'm and if you and the care physicians, OK. Yeah*

*Participant 005\_2023AUDNS*

*Oh, the virtually no cost to us. Yeah. Not, not direct medical costs, that's we're very grateful for that,*

*Participant 08\_2023AUDPA*

*Yeah, pretty much all of it. Because, you know, we wouldn't be able to afford to pay for surgery and hospital stays and you know, all those special like visiting all those specialists, that's just that would just be insane amount of money if we were paying and you know, we pay our taxes and we're very grateful that to have Medicare. You know what I mean? Yeah. So,*

*yeah, the cost, yeah, the cost and, and the care provided, very grateful.*

*Participant 023\_2023AUDPA*

**Participant describes being grateful for the entire health system**

*The fact that so much of it is completely free is just astounding just completely blows my mind and makes me very happy to pay taxes to say the weird thing to say. But. So much would have been free. So many of the people that I've interacted with have been just lovely and caring and supportive and helpful. Yeah, I'm very impressed with most, not all, but most of my interactions with the whole health system.*

*Participant 009\_2023AUDIS*

*Oh, boy, everything. It's cheap or free for a person like myself with a low income. The fact that I can see specialists once again—and also that I can involve myself in clinical trials because, for me, that's a way of being useful even within the limitations of my life. I feel at least of some use to somebody, even if the trials aren't any use to me down the track, they'll be useful for somebody.*

*Participant 004\_2023AUDIS*

*Very much so. I think I said it earlier as an Australian, I feel very fortunate that we do have a good healthcare system that we do have the option of having the drug approved so that we only pay dispensary costs, not the two and a half, \$1000. We don't have to have private insurance to access hospitals. We don't, you know at the dermatology clinic I don't pay for. I see some of the best. Doctors around in my state that that understand HS and it doesn't cost me anything. Sure there's a wait and you have to sit in line. You know the de roofing surgery that I'm waiting on at the moment will be through public health, through the plastic surgeons in public health. There's a wait. I have to wait for it. But I've waited 30 years to to get some help, so. You know, if I have to wait two more years for the surgery that sets me free, then so be it.*

*Participant 005\_2023AUDSK*

*I'm just, I'm thankful that we actually have a a health system where we can access pay for the treatments. I'm extremely thankful that we have PBS where I can pay \$30.00 a month for my Humira and not, you know, the two and a half thousand a month or whatever it is for it I'm thankful for chronic illness program that we have where I can access other services like counselling, physiotherapy, that sort of thing. Yeah.*

*Participant 001\_2023AUDSK*



### **Participant describes being grateful for timely access to treatment**

*I know this isn't the health system, but the NDIS has been amazing for us at least. I know not everyone has experienced that, but we certainly have and been well supported. Through that which has helped us a great deal. I am really grateful that when we have attended Emergency, it hasn't been a huge long wait. I mean, as she gets old it has been. But we were very grateful, particularly when she was younger. We were there quite a lot with aspiration or whatever, that she was triaged and expedited through the process because they could see. You know how complex she was. So very grateful for that and for our ambulance services. They were here really quick whenever we called them, so they're very, very grateful for that, yeah.*

*Participant 095\_2023AUENM*

*This is how quickly we're now able to like get this groups and everything and how we sometimes we can just go into a doctor and like have a walk in appointment where I know like other time other people may have had their life five days that's for one appointment. So I think it is well in one. In one particular instance was when they offered a, a CPAP machine through a charity at the Children's Hospital. We didn't take them up on that offer because we actually went out and bought her one ourselves. But I thought that that was actually really, really, really great. But we didn't want to take it because there was the next person that would have needed it from the charity. So I know now it's not a like Australian, you know, like government sort of thing, but I do think Australian general are very charitable when it comes to that sort of thing. So that to me is something that stands out in her medical history, that we've turned down something like that for somebody else.*

*Participant 037\_2023AUDPA*

*I guess probably quick and efficient care whe, when required. When she did have the heart issues and stuff, it was all dealt with very quickly and, and we didn't have to worry about anything at the time in regards to that. So that was there was no delays, nothing. It was all very, very prompt.*

*Participant 027\_2023AUDPA*

### **Participant describes being grateful for low cost/free medical treatments through the government**

*No, I'm pretty, I'm, I'm satisfied to the point that obviously my treatment you know, clean hospitals, great doctors, affordable. I'm pretty grateful for*

*where I live and yeah, lucky to be raised in Australia with all the support and, and you know, companies like you doing research and stuff like that. So yeah, it's very lucky and grateful, yeah.*

*Participant 024\_2023AUORC*

*Definitely being able to have the surgery not charged, that's been something that because that's the point of diagnosis. So that's kind of the most necessary part.*

*Participant 078\_2023AUDIS*

*The fact that there are public health outlets, you know that, that I've been able to access them freely. I was living in the States at a point of when I was pregnant with my daughter and it would have been I had C-section...My emergency caesarean would have cost me if I'd have stayed. And I, I think after then having recurring boils and those sorts of things any hospital or healthcare visit. It is not lost on me at all that that I do at least have the benefit of bulk billing and those sorts of things, PBS scripts, you know, being able to benefit from discounted pharmaceuticals. I think it is ...just be the education to go along with it for those healthcare providers.*

*Participant 015\_2023AUDSK*

*Definitely PBS medication. I'm like for other conditions I have. Quite a few non PBS medications. It makes big difference, yes, and I think easy access to the specialists and things like that as well.*

*Participant 019\_2023AUDSK*

### **Participant describes being grateful for timely access to diagnostics**

*I'm grateful I do the test every time. At least I'm being monitored. That's why I'm grateful that I get monitored every time, so if something goes wrong, at least I can catch it early.*

*Participant 001\_2023AUORC*

*I'm very grateful for the, very grateful for the fact that we got referred to a dermatologist very quickly from the GP and that we, the dermatologist was able to fit us in because he had a three month waiting, waiting list or 4 four month waiting list and because of his age and yeah, the...they spotted him in within like I think we got in within like 2 weeks. So I'm very grateful for that, very grateful for the, the way a child was prioritized.*

*Participant 009\_2023AUDSK*

Again, you know, newborn screening. Without that, I don't think that my child would have survived because they wouldn't have known what the condition was and how to manage it. I'm grateful for what resourcing is available in the tertiary care system to provide care for my child with their condition. I do think that comparatively, we do have a good health system, but I wouldn't say that it's great, to be honest. They're the things that I'm grateful for within the system that we have.

Participant 021\_2023AUORC

Participant describes being grateful for access to private healthcare/private insurance

Oh yes, because private therapy is like, I'm very grateful for being able to access good physiotherapy through private health. I had to use my chronic disease management plan, so even though it's only five, can't knock it, that's still helpful. I'm grateful to have had that towards the things that I've needed. Yes, they're probably the main things. I haven't needed to go to the public hospital much myself, but for my kids I have and that's been amazing and the genetics was covered through public health, so I am very grateful for that because I'm sure that would've been very expensive privately. I don't actually know if there is someone private who does it. I think only recently, maybe if someone went into genetics privately in [unintelligible 01:10:07] I think it was only at the time when I went through, it was only the public referral base.

Participant 004\_2023AUDPA

The ability to be able to go to a private hospital and have private, be privately health insured, you know grateful grateful. I've got that that service is available. I didn't get to choose my neurologist. It was, I was given one but, but I think, I think in the main I think the health service that I received was very was, was, you know, very good the fact that I had choice to go to a private hospital. I could have gone to a public hospital and, you know, possibly the public hospital would have been, would have been, okay, but I, I don't know that and I'll never know that but I'm grateful that I that I was able to go to a, to a private hospital. Participant 095\_2023AUDNS

Participant describes not being grateful for anything

Nothing, Nothing. As I said, public hospital. I've been at LOCATION, I've been at HOSPITAL. There's nothing, I mean nothing that they have helped me. And because I was able to cut and wash it and clean myself, even the nurses could get less about me.

Participant 023\_2023AUDSK

PARTICIPANT: Nope. Next question.

INTERVIEWER: Yeah, I thought so. No, it's fine.

PARTICIPANT: Gotta keep the sense of humor.

Participant 008\_2023AUDSK

I can't really say much at all.

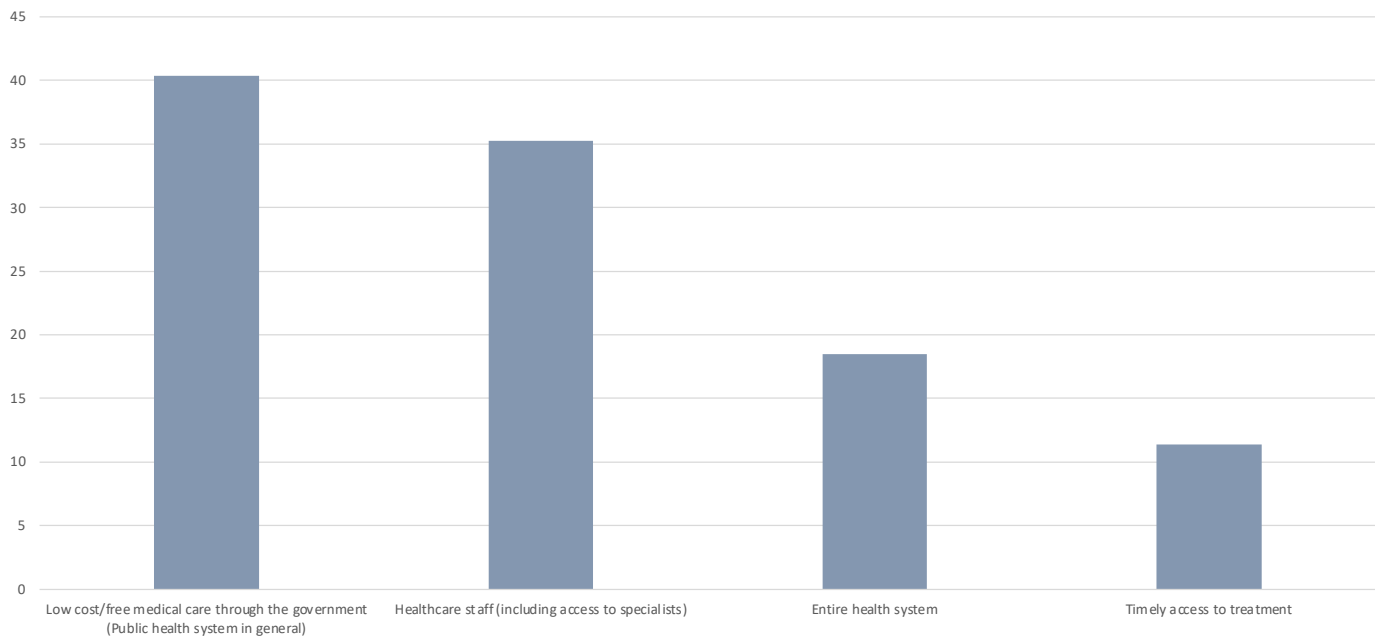
Participant 025\_2023AUDSK

Table 9.9: What participants are grateful for in the health system

What participants are grateful for in the health system	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=352	%	n=67	%	n=81	%	n=45	%	n=32	%	n=95	%	n=32	%	n=247	%	n=105	%	n=252	%	n=98	%
Low cost/free medical care through the government (Public health system in general)	142	40.34	19	28.36	40	49.38	17	37.78	17	54.84	36	37.89	13	41.94	106	42.91	36	34.29	117	46.43	25	25.51
Healthcare staff (including access to specialists)	124	35.23	23	34.33	28	34.57	14	31.11	10	32.26	32	33.68	17	54.84	83	33.60	41	39.05	91	36.11	32	32.65
Entire health system	65	18.47	7	10.45	9	11.11	6	13.33	5	16.13	33	34.74	5	16.13	50	20.24	15	14.29	46	18.25	19	19.39
Timely access to treatment	40	11.36	4	5.97	15	18.52	5	11.11	0	0.00	14	14.74	2	6.45	28	11.34	12	11.43	32	12.70	7	7.14

What participants are grateful for in the health system	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=352	%	n=69	%	n=116	%	n=108	%	n=59	%	n=172	%	n=172	%	n=100	%	n=252	%	n=176	%	n=176	%
Low cost/free medical care through the government (Public health system in general)	142	40.34	20	28.99	55	47.41	48	44.44	19	32.20	65	37.79	72	41.86	42	52.50	100	39.68	70	39.77	72	40.91
Healthcare staff (including access to specialists)	124	35.23	29	42.03	42	36.21	35	32.41	18	30.51	57	33.14	64	37.21	33	41.25	91	36.11	64	36.36	60	34.09
Entire health system	65	18.47	10	14.49	16	13.79	19	17.59	20	33.90	34	19.77	30	17.44	17	21.25	48	19.05	29	16.48	36	20.45
Timely access to treatment	40	11.36	6	8.70	16	13.79	13	12.04	5	8.47	23	13.37	15	8.72	12	15.00	28	11.11	18	10.23	22	12.50



**Figure 9.5: What participants are grateful for in the health system**

**Table 9.10: What participants are grateful for in the health system – subgroup variations**

What participants are grateful for in the health system	Reported less frequently	Reported more frequently
Low cost/free medical care through the government (Public health system in general)	Developmental anomalies Male Aged under 18	Diseases of the skin Regional or remote Other rare condition
Healthcare staff (including access to specialists)		Endocrine, nutritional or metabolic diseases Aged 65 plus
Entire health system		
Timely access to treatment	Diseases of the skin	

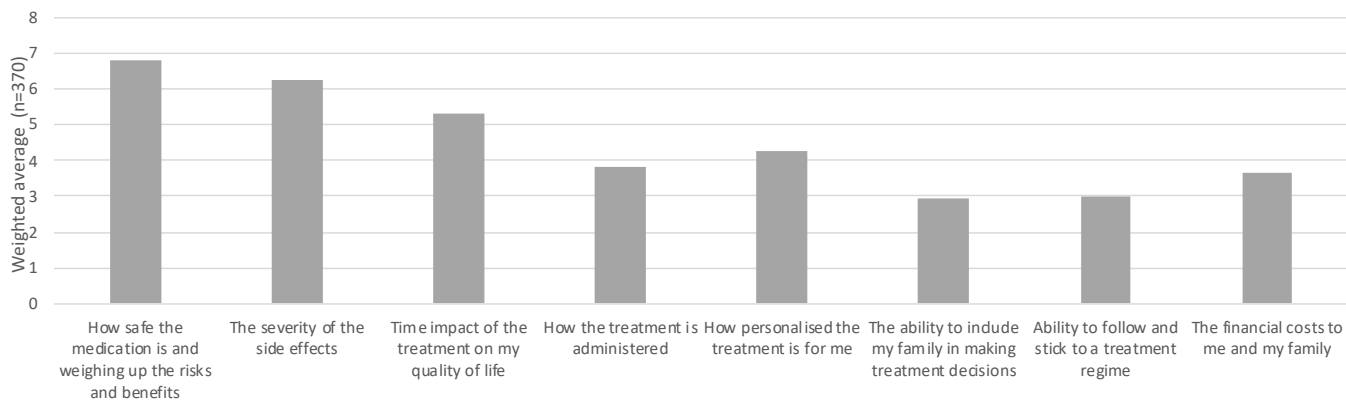
### Values in making decisions

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

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

**Table 9.11: Values in making decisions**

Values when making decisions	Weighted average (n=370)
How safe the medication is and weighing up the risks and benefits	6.79
The severity of the side effects	6.26
Time impact of the treatment on my quality of life	5.32
How the treatment is administered	3.83
How personalised the treatment is for me	4.24
The ability to include my family in making treatment decisions	2.93
Ability to follow and stick to a treatment regime	2.98
The financial costs to me and my family	3.68



**Figure 9.6: Values in making decisions**

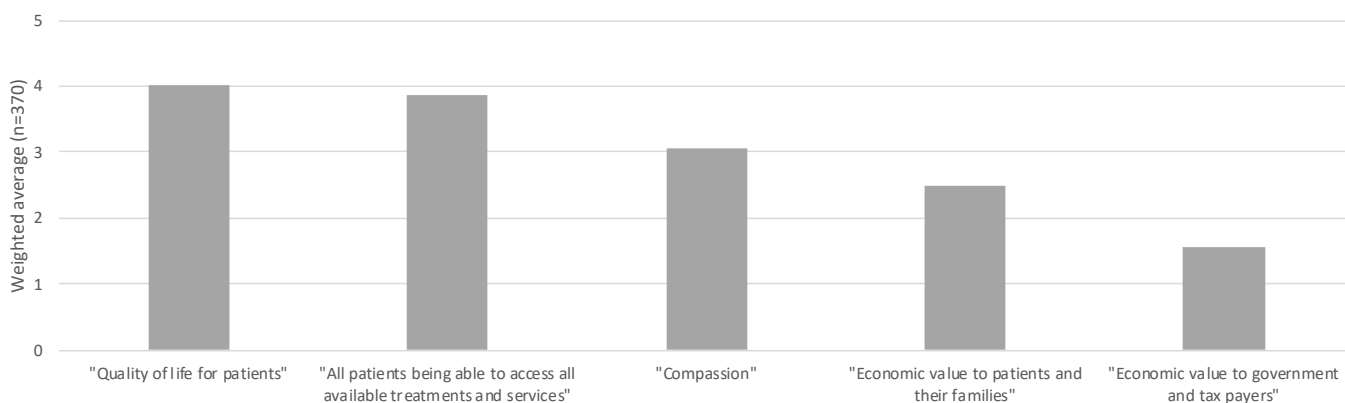
**Values for decision makers**

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

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

**Table 9.12: Values for decision makers**

Values for decision makers	Weighted average (n=370)
Economic value to government and tax payers	1.56
Economic value to patients and their families	2.49
Quality of life for patients	4.02
Compassion	3.07
All patients being able to access all available treatments and services	3.87



**Figure 9.7: Values for decision makers**

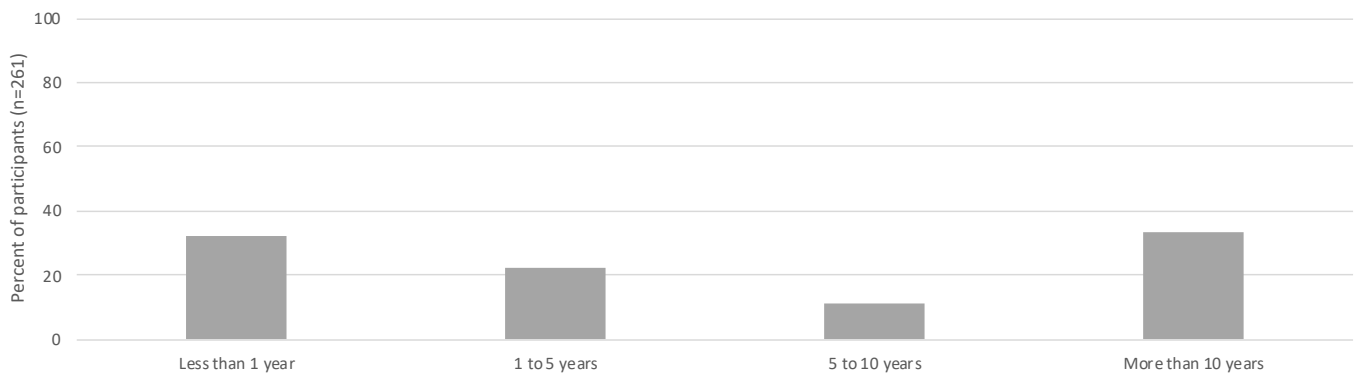
## Time taking medication to improve quality of life

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

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

**Table 9.13: Time taking treatment to improve quality of life**

Time taking medication to improve quality of life	Number (n=261)	Percent
Less than 1 year	84	32.18
1 to 5 years	59	22.61
5 to 10 years	30	11.49
More than 10 years	88	33.72



**Figure 9.8: Time taking treatment to improve quality of life**

## Most effective form of medicine

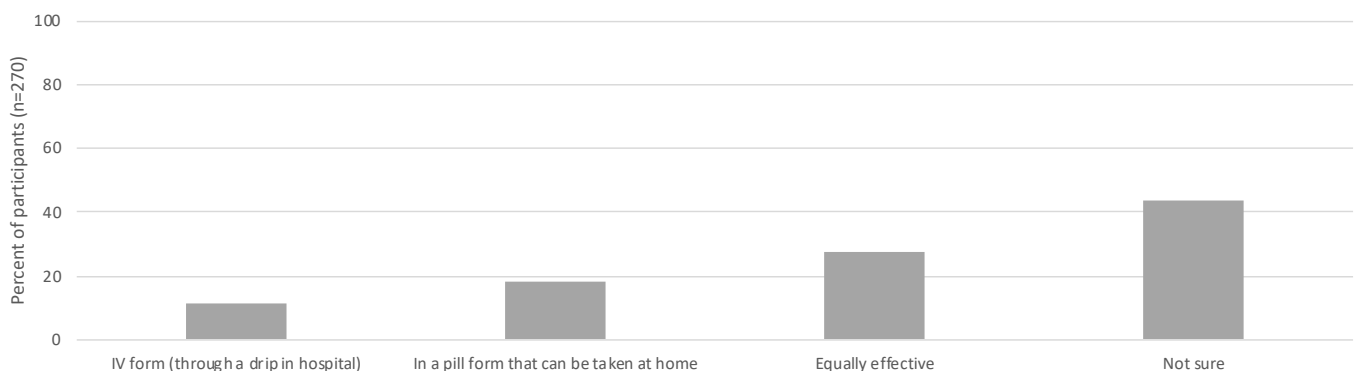
Participants were asked in the online questionnaire, in what form did they think medicine was most effective in.

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

There were 30 participants (11.11%) that thought that medicine delivered by IV was most effective,

**Table 9.14: Most effective form of medicine**

Treatment most effective in what form	Number (n=270)	Percent
IV form (through a drip in hospital)	30	11.11
In a pill form that can be taken at home	49	18.15
Equally effective	74	27.41
Not sure	117	43.33



**Figure 9.9: Most effective form of medicine**

## Messages to decision-makers

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

### Timely and equitable access to support, care and treatment

*I just think that everyone with the rare disease should have the exact same outcomes as other and like medical treatment etcetera and opportunities for medical allied health funding especially in endorse funding should have the same amount as people with well known conditions. Like the child with CP shouldn't be getting six times the amount of my child's funding just because they have cerebral palsy, which is more well known to the general population.*

*Participant 081\_2023AUDIS*

*I think they should have all the support and care that they need and be given all the resources that they need and support from the beginning of their journey right to the end all.*

*Participant 010\_2023AUORC*

*I wish there wasn't as many hoops, you know, like even, even when he was younger, to get help from the department that you know does respite care and, and things like that, like we were on waiting lists for that. And the NDIS is the same thing. Like if you don't, if you don't word it properly, you don't get the services that you need. Like it just needs to be more unified and fair so that people who need who have greater needs get them, and people are not routing the system when they're not.*

*Participant 040\_2023AUDPA*

### More clinical trials and/or new treatments

*Oh my God. Well, I've been trying to contact the health minister for the last several weeks about the access. Yeah, has potential access for this life changing drug and so that's my message, that there are 500 children in Australia waiting desperately for this drug, but it's been approved. And that, yeah, he,*

*he obviously, you know, as a teacher, there's a lot of money put into NDIS and it's that the whole system, in my opinion, is being completely abused. And there are children that are just want this drug that will just like change their daily living and their life expectancy and they're crazy. Like it must cost them so much money every time we're hospitalized.*

*Participant 023\_2023AUORC*

*We definitely need more options in Australia. And as I said that there's a stem cell, well it's gone from trial to the next phase now. So it's essentially it's work essentially they found a cure for it for sure. So...and it yeah... it's a stem cell treatment in the in the States and they're going into pods in, they're trying out to the UK now a couple of other places. So just you know I'd love to see more of that kind of.*

*Participant 015\_2023AUORC*

*Have access to just that. I said it'd be great to see Australia continue to get access to the new drugs, you know, as they're developed overseas because most people we kind of you hear about them and that the I guess that they're doing enough research and especially with children to be able to get that access for the for children as well and have to wait till you're an adult. Yeah.*

*Participant 079\_2023AUDIS*

### Help raise community awareness

*I think for it to be maybe more prominent and recognized that given no one really is, is that aware of it. I think that the deletion potentially more so, but I think, yeah, just some more awareness about it.*

*Participant 020\_2023AUDPA*

*I would like to see rare diseases, in general, better known. That's all. I'm very happy with the standard of care and the price of care. I would like more oversight by somebody or somebody who oversees the treatment. I would like more research and knowledge on rare diseases in general and my rare diseases specifically.*

*Participant 004\_2023AUDIS*

*Awareness, awareness, awareness and an awareness, that is the main thing. Because from that awareness, the treatments and, you know, diagnosis of different things could come through hell of a lot sooner. If they're aware that that could be there, they'd, they'd*

*come through sooner and the child will be treated quicker.*

*Participant 024\_2023AUDPA*

#### **Invest in research (including to find new treatments)**

*Money for research.*

*Participant 005\_2023AUDNS*

*Something. Try to find something, research, do something about it. Because I don't think I'm the only one. I think there's quite a few people that suffer from this condition.*

*Participant 023\_2023AUDSK*

*Oh, as much as possible, I suppose. I would say it would be a general thing. I don't know about scleroderma in particular, because nobody knows. I guess more money for research about the causes and potential cures or improvements in the condition for people. I guess that's what one could wish for, because there isn't any treatment that you can ask for, really.*

*Participant 012\_2023AUDIS*

*I would say, you need to put more money into HS research and there needs to be more help financially for things like dressings and at home care like for hexidine, those sorts of things.*

*Participant 001\_2023AUDSK*

#### **Invest in health professionals to service the patient population**

*I don't know about specifically for Scleroderma, but I think just in general, like here in CITY, we are lacking in staff and hospital resources and, and, and I know they're aware of that because there's a whole grand plan for the next 10 years. But like, yeah, it would be nice for things to be easier for everyone to access with less like shorter wait lists. And doctors having more time and more staff available.*

*Participant 024\_2023AUDIS*

*I would try and put it in the perspective of if they had a child or someone that they knew was cystic fibrosis, to think of it from that perspective, because otherwise they're just thinking, oh, just cystic fibrosis, don't really know what it is, don't care. And to think about the costs for people as someone that's already disadvantaged a lot by the sickness and everything that it has done to their life and everything. So trying to use the cost of that. And then also training the*

*doctors for and having specific cystic fibrosis doctors and nurses.*

*Participant 013\_2023AUORC*

*My message would be if we need more doctors, the wait lists are out of control. My idea would be that there was a coordinating person for complex cases where you're seeing multiple specialists, I guess again, my idea would be that when you're seeing these specialists, you see the same person for some kind of continuity. Yeah, continuity of care is a big factor for NAME. Yeah. She doesn't deal well with seeing new people continuously.*

*Participant 021\_2023AUDPA*

#### **Treatments need to be affordable**

*Maybe to tell him to give free treatment to people with who need the treatment.*

*Participant 001\_2023AUORC*

*That they should be able to get access, you know, like well, especially in the public system, free of charge. Yeah. There shouldn't be any barriers.*

*Participant 014\_2023AUDPA*

*So gosh, this put me on the spot. What would I say? I think overall my experience was not a bad one, and I know that there are other people that have experienced much worse with this condition. So for me it's kind of tricky I think. The, the main kind of sore points that came from my experience would have been probably the costs and the time it took and the frustration it took to to get the diagnosis. So if those points could be smoothed out, that would be fantastic. I understand there's lots of issues in the healthcare system with, you know, costs and that kind of thing. There's lots of talk about it, but if it didn't cost me over \$600 to get a diagnosis, that would have been lovely. And if I was just listened to by everyone that I spoke to the whole way through, that would have been fantastic as well, yeah.*

*Participant 027\_2023AUDSK*

#### **Increase investment (general)**

*More funding, more funding. Yeah, just, you know, more awareness, more funding. Yeah, it's just there seems to be more awareness in the states in the UK, yeah. And there's a lot of people being diagnosed with it in Australia. So why, you know, why aren't they, you know, looking to more, more funding. I get sicker than ...the fact that we have to, we have to virtually do well for anything the public does their own fundraising. It*

*shouldn't be. The government should be, you know, I'm sorry, there's a lot of people overseas that you know, need when there's a disaster, but start back at home, you know, put more into more into our self funding and our health care. That's what I'd say to them. Yeah.*

*Participant 019\_2023AUDIS*

*Please give us some funding and start recognizing that there's more and more people being diagnosed with it and we need help to-- Have professionals where these people can go so that mentally they're not crumbling. Just please help. Please help. Give them the information they need and don't let people suffer like they're suffering. I'm not as bad as a lot of them, so I think I'm doing all right, but some are really terrible.*

*Participant 007\_2023AUDIS*

*I think I could probably talk about a lot of things and it's probably not cardiomyopathy per se that only on its own. It's probably everyone with a heart condition or a cardiovascular disease or whatever it is that affects your cardio pulmonary activity, I guess it is more so than anything. Like I, I don't think people understand the cost of like not personal cost, but the cost to society and community that these conditions sort of bring to, you know, the state or the federal government or whatever. It's like it's astronomical and I don't understand why more isn't spent in the way of prevention and management so that it's not a matter of treating people at the most vulnerable, weakest and most detrimental point in their life where they need the most expensive and the most care.*

*Participant 030\_2023AUORC*

#### **Take the condition seriously**

*I guess maybe just greater recognition of what, what the condition is and what it entails for the people that do have it and like I said, it is such a broad area of issues that can crop up for different people and, and greater knowledge of what the condition entails.*

*Participant 027\_2023AUDPA*

*I think my predominant message would be people with complex disabilities are not the same as people with a list of all those disabilities just sort of added up because for a lot of people with charge, you know, it's like, oh, well, they're not quite deaf enough or they're not quite blind enough to receive this service or, you know, whatever it is, but it's like. Yes. But the impact of all of those senses being impaired and erratic means they need so much extra support.*

*Participant 018\_2023AUDPA*

*Everybody's different. Every disease is different. The issue we have is that some of those diseases are invisible and if you can't see it happening in the system, if you can't see the person-- I walked in and I had my leg from my toe up to my hip, people would look at me and go, "Okay, you've got that's you've got serious issues," or you've got braces on or whatever up your legs because you can't walk or you're having something else as an aid to help you to walk or whatever. They treat people differently if they've got those sorts of things to what is actually happening to other people like people who have invisible diseases.*

*Participant 005\_2023AUDPA*

#### **Invest in screening/early detection**

*Well, the diagnosis, the blood test that's really good is because of she was, I think it had only been developed then when NAME got it. I don't think it had been developed before that. So she couldn't have probably got it any earlier, I think. Just I guess. The doctors to be more aware of looking for these causes when, when a person presents with something a bit different if they get more look a bit harder and have a more coordinated plan and I just if the it's impossible for them to do this but just it'd be so good if doctors could just stay so that you can have the one doctor and not just be jumped from one doctor to another. And consistency in the health care is important, but that's probably out of the hands of government and just more psychology services in rural areas.*

*Participant 09\_2023AUDPA*

*Health minister, [silence] gosh. [silence] I guess it would be just raising awareness. I think I would be fortunate in my diagnosis. The only delay was access to specialists, whereas some people I think, struggle for years trying to get a diagnosis because it is rare and people aren't aware. I guess raising the awareness and the skill level because I think the earlier the diagnosis, the better the outcomes.*

*Participant 017\_2023AUDIS*

*Yeah, again, a better screening. Know if you call it screening policy or screening system for these kids, because they are mostly nonverbal, they cannot communicate, the behaviour is communication and sometimes they will have pain and they won't be able to articulate or show you that. A lot of GI issues, for example, get they're not even known about for many years until they get older and they're already struggled so. Better screening like I said, the early vision testing is very important, just a more specific where those these children go to learn. For example,*



*like would it be possible to have there's a very well established deaf schools, I'm not sure if there's as well as established deaf blind schools for example, if there is that haven't been told. Yeah, yeah, yeah.*

*Participant 094\_2023AUENM*

### **Compassionate and empathetic**

*Need more? I don't know. I don't know what I would say, actually. I just, it needs more spotlight. I think if you know, like if you really look at a lot of people, you would see it, you know, a lot more people. I think a lot more people just manage the symptoms, don't even know what's wrong with them. I didn't know 35 years what was wrong with me. So I think it's something that needs spotlight and needs to be people need to learn what it is, what to look out for. And then, you know, we need to be listened to. None of this. It's anxiety go away or true food go away, or they go on a liquid diet. Go away. Like just need to be more heard and more respected, you know, like and listen to compassionately. And not just in a textbook. But it's hard because I guess everyone is slightly different too.*

*Participant 078\_2023AUDIS*

*A bit of improvement. Well I guess the first thing would be have to change the, the mentality of the of the whole system which with doctors training for a start. So I don't think they're God's gift and, and then just professional drug dealers. You've got to change that and have some empathy for actual people who are dealing with shit rather than just, you know, write out scripts and then turn over, turn over pills for money. That is probably the, the most important part because nurses are better than doctors by country mile, you know? So start there and then, you know, might have a bit of a chance, yeah.*

*Participant 006\_2023AUDIS*

### **Invest in professional development so that clinicians understand the condition**

*I think that would be, what I would say is that they need to educate the doctors that it's not just some skin thing because I think the name Scleroderma. It, it puts them in that mindset of it's a skin thing when the skin part of it is negligible. Yes you know and, and so the name of the - and I'm not saying that they must go and change the name of it - but the name of it gives the illusion that it's a minor problem that's easily fixed and it's not. And they don't, they don't really know that. And next you're the rheumatologist, but if you any other doctor who interfaces with the person with Scleroderma, they don't give it any gravitas at all. It's*

*like, OK, so you've got Scleroderma. OK, well, now I'm talking about your kidneys today. I'm talking about your heart or I'm talking about, you know, they kind of OK, we don't need to talk about that. Where is a lot of my things could, quite well, the originating starting point of it was the scleroderma that they're just treating the symptom now. They're not treating the cause and they're not treating it holistically. They're just doing their tick and flick of their one particular thing and then they're going to go and have lunch, and that's as far as it goes.*

*Participant 002\_2023AUDIS*

*Oh God, yeah. I mean, I I've dealt with ministers before and they have no idea about most things. What would be my access to just so that there's so many different diseases out there which they know nothing about. And I think it's, it's you can't cure everything but you can. It's always about money, isn't it? The way it says, well, can you please give more money and educate people? But I don't know. And it's mainly educating the doctors, you know, get them more educated and the nurses. When I had my, when I had my hip thing, I think nobody knew in the hospital what an allograft was and they'd never had anybody have an allograft before and somebody was fine with a thing on it and it's just unknown, so. I suppose with rare things you can't. How do you speak to the just make more money available, I suppose for research.*

*Participant 024\_2023AUDSK*

### **Understand the financial implications (and provide financial support)**

*Make it affordable. People with chronic illness are penalized greatly because it's just not affordable.*

*Participant 016\_2023AUDIS*

*Well. They need access. Oh, that's one they need. They need subsidised access to, to dressings. They need extra sick leave. They need additional sick leave to for for their, their flares. Well, I don't care if it has to be that it has to be a doctor, certificate one or whatever, but they need additional and they need because it is a, it's a, it is a hidden disability and it needs to be much more made, much more aware of in the workplace that people have hidden disabilities and they get special consideration, parking, everything. Disabled parking is another one because when they're having an active flare they are as disabled as anybody else who is disabled. But when they're not having an active flare then yeah they they, they shouldn't be using it. But I think access to disabled parking when required should be available.*

*Participant 009\_2023AUDSK*

*I would like to say that don't forget our little rural communities and that our people that do suffer these rare conditions are really isolated more so because of our distance from everyone. They need to understand they need to be supportive. If someone does have to travel like us, have to do a 2,400-kilometer round trip for treatment, they need to be a little bit more supportive on the accommodation and that they can offer also the financial support with accommodation because that's something that they didn't do at the time when I was going down there. They wanted you to go down and back in a day and then travel over 100 kilometers to get home. It'd be different if we lived in those areas like Townsville all that, where you just drove to your house there. Then we also, besides the flight then had to drive as well. I think if they could take that into consideration for people in the future, that would be great.*

*Participant 006\_2023AUDNS*

#### **Grateful for the healthcare system and the treatment that they received**

*I'd say overall with very good. It's very good. I think a lot of what. We received in the first few years was about managing acute health issues and probably reactionary to what was going on and we received very good care, yeah, very good surgery, very good doctors, very good, mostly good hospital experiences. I think there probably could also be more focused on early intervention and I guess not just reacting to the health issues but also focusing on preparing for the future too, which only came later. The solving, once we work through the other health things that we're dealing with initially, yeah, I think that's pretty much it.*

*Participant 089\_2023AUENM*

*What would I say to him about what I've had? I'd say thank you.*

*Participant 018\_2023AUORC*

#### **Patient input and engagement in all aspects of decision-making**

*PARTICIPANT: How to express this? I'm not finding the right words. Targeted things that engage in proper community consultation to work out what's actually needed.*

*INTERVIEWER Yes, okay. Like reference groups type thing?*

*PARTICIPANT: Which pretty much is what you're doing.*

*Participant 001\_2023AUDPA*

*I'm bighting my tongue quite a bit there because I don't need to hear your ear off for the next hour about that, but we also don't. But you know what? I also believe we need to do a in education of doctors and clinicians. Lived experience needs to be part of the curriculum of when they're educated about different things. Like when you're becoming a GP, let's bring in some lived experience people to tell you...how they geneticists, genetic counselors, come and listen to the stories so that you don't make those same mistakes. Because these families are human beings with emotions, and the way you give a diagnosis could actually determine the path or trajectory they follow.*  
*Participant 025\_2023AUDPA*

#### **Holistic approach to the condition (including emotional support)**

*It needs to be family centered care. You need to understand that person is part of the family and the specific context of that family and how you can improve that individual's life but also support the family around that.*

*Participant 090\_2023AUENM*

*I would actually tell him that we have access to advices and, and the views relating to mental health issues and we also have access to the way to cure and go about the virus and we always have access to knowing that someday somehow you would actually be cured here.*

*Participant 009\_2023AUORC*

*Yeah, just that holistic care is, is really important and also just awareness by health practitioners on what 22 Q, what that involves, it's really helpful. I do find I have to educate everybody and you know that's a bit bearing in mind it's actually pretty common. I think, you know, I would advocate for money towards research like what you're doing, you know, potentially have capacity in the LOCATION health research system to be able to do linked to studies here in LOCATION, possibly not elsewhere because you can track kids a lot better here. And so, yeah, I would advocate for research money and and awareness and integrated services and also just acknowledgement of, yeah, when kids have a lot of minor symptoms that they add up to being a major symptom.*

*Participant 035\_2023AUDPA*

**Need for multidisciplinary and coordinated care}**

*I would say to him grateful for all that we have, but there is a huge room for improvement around coordinated care with all of the different specialists and access of care and treating the patient like a person in the room as well, particularly when they're a child.*

**Participant 095\_2023AUENM**

*How that would be done on not really for me to say and consistency of consistent care, not spasmodic and*

*obviously more funding, more funding for research and awareness.*

**Participant 08\_2023AUDPA**

*It's, it's a lifelong condition. It's not something that can be fixed. Our experience, I guess, is a lot different to other people's way they have to physically pay. I mean, they pay for lots of things for NAME, but if people have a lifelong condition. Yeah, make it the services available to them.*

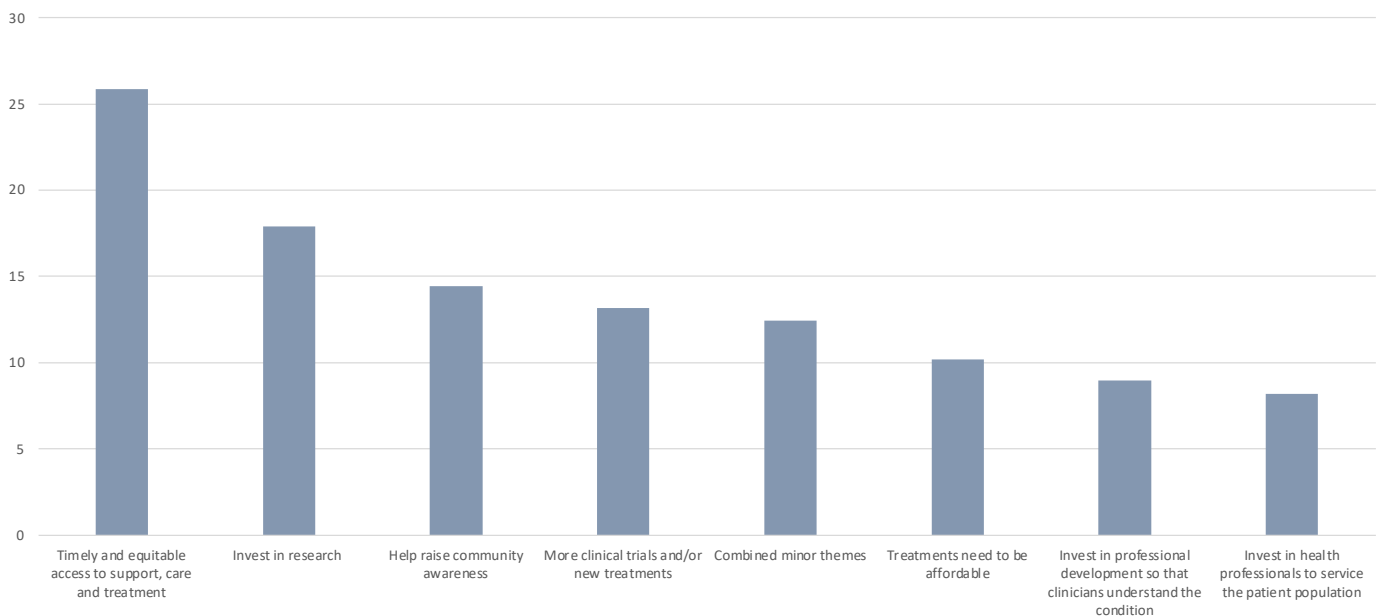
**Participant 015\_2023AUDPA**

**Table 9.15 Messages to decision-makers**

Message to decision-makers	All participants		Developmental anomalies		Diseases of the immune system		Diseases of the nervous system		Diseases of the skin		Endocrine, nutritional or metabolic diseases		Other rare condition		Person with condition		Family or carer		Female		Male	
	n=402	%	n=67	%	n=81	%	n=95	%	n=32	%	n=95	%	n=32	%	n=268	%	n=134	%	n=264	%	n=106	%
Timely and equitable access to support, care and treatment	104	25.87	11	16.42	26	32.10	19	20.00	7	21.88	27	28.42	14	43.75	80	29.85	24	17.91	78	26.53	25	23.58
Invest in research	72	17.91	5	7.46	17	20.99	9	9.47	14	43.75	23	24.21	4	12.50	62	23.13	10	7.46	55	18.71	17	16.04
Help raise community awareness	58	14.43	7	10.45	22	27.16	3	3.16	6	18.75	15	15.79	5	15.63	50	18.66	8	5.97	49	16.67	9	8.49
More clinical trials and/or new treatments	53	13.18	0	0.00	3	3.70	34	35.79	1	3.13	10	10.53	5	15.63	32	11.94	21	15.67	41	13.95	12	11.32
Combined minor themes	50	12.44	0	0.00	2	2.47	19	20.00	0	0.00	5	5.26	0	0.00	17	6.34	9	6.72	20	6.80	6	5.66
Treatments need to be affordable	41	10.20	6	8.96	8	9.88	4	4.21	5	15.63	12	12.63	6	18.75	31	11.57	10	7.46	29	9.86	12	11.32
Invest in professional development so that clinicians understand the condition	36	8.96	5	7.46	9	11.11	7	7.37	8	25.00	5	5.26	2	6.25	28	10.45	8	5.97	28	9.52	8	7.55
Invest in health professionals to service the patient population	33	8.21	5	7.46	7	8.64	0	0.00	1	3.13	16	16.84	4	12.50	22	8.21	11	8.21	27	9.18	6	5.66

Message to decision-makers	All participants		Aged under 18		Aged 18 to 44		Aged 45 to 64		Aged 65 plus		Trade or high school		University		Regional or remote		Metropolitan		Mid to low status		Higher status	
	n=402	%	n=97	%	n=131	%	n=114	%	n=60	%	n=198	%	n=196	%	n=111	%	n=291	%	n=200	%	n=202	%
Timely and equitable access to support, care and treatment	104	25.87	17	17.53	38	29.01	30	26.32	19	31.67	46	23.23	58	29.59	25	22.52	79	27.15	49	24.50	55	27.23
Invest in research	72	17.91	6	6.19	25	19.08	27	23.68	14	23.33	41	20.71	31	15.82	21	18.92	51	17.53	40	20.00	32	15.84
Help raise community awareness	58	14.43	6	6.19	23	17.56	20	17.54	9	15.00	35	17.68	23	11.73	23	20.72	35	12.03	38	19.00	20	9.90
More clinical trials and/or new treatments	53	13.18	18	18.56	16	12.21	10	8.77	9	15.00	29	14.65	22	11.22	10	9.01	43	14.78	25	12.50	28	13.86
Combined minor themes	50	12.44	7	7.22	5	3.82	10	8.77	4	6.67	11	5.56	14	7.14	7	6.31	19	6.53	12	6.00	14	6.93
Treatments need to be affordable	41	10.20	4	4.12	16	12.21	12	10.53	9	15.00	21	10.61	18	9.18	14	12.61	27	9.28	21	10.50	20	9.90
Invest in professional development so that clinicians understand the condition	36	8.96	4	4.12	15	11.45	12	10.53	5	8.33	17	8.59	19	9.69	12	10.81	24	8.25	22	11.00	14	6.93
Invest in health professionals to service the patient population	33	8.21	7	7.22	3	2.29	20	17.54	3	5.00	13	6.57	20	10.20	13	11.71	20	6.87	18	9.00	15	7.43



**Figure 9.10: Messages to decision-makers**

**Table 9.16: Messages to decision-makers – subgroup variations**

Message to decision-makers	Reported less frequently	Reported more frequently
Timely and equitable access to support, care and treatment		Other rare condition
Invest in research	Developmental anomalies Family or carer Aged under 18	Diseases of the skin
Help raise community awareness	Diseases of the nervous system	Diseases of the immune system
More clinical trials and/or new treatments	Developmental anomalies Diseases of the skin	Diseases of the nervous system
Combined minor themes	Developmental anomalies Diseases of the skin Other rare condition	
Treatments need to be affordable		
Invest in professional development so that clinicians understand the condition		Diseases of the skin
Invest in health professionals to service the patient population		