Section 3

Symptoms and diagnosis

Section 3: Symptoms and diagnosis

Symptoms leading to diagnosis

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

Symptoms leading to diagnosis: Seeking medical attention

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

Symptoms leading to diagnosis: Description of diagnostic pathway

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

Diagnosis provider and location

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

Understanding of disease at diagnosis

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

Emotional support at diagnosis

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

Costs at diagnosis

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

Burden of diagnostic costs

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

Genetic tests and biomarkers

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

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

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

Understanding of prognosis

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

Symptoms leading to diagnosis

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

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

Participant describes having developmental delays which led to their diagnosis

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

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

Participant describes having fatigue which led to their diagnosis

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

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

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

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

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

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

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

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

Participant describes having heart problems which led to their diagnosis

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

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

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

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

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

Participant describes having seizures/Spasms which led to their diagnosis

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

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

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

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

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

Participant describes having shortness of breath which led to their diagnosis

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

Participant describes having weight loss which led to their diagnosis

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

Table 3.1: Symptom recall

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

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

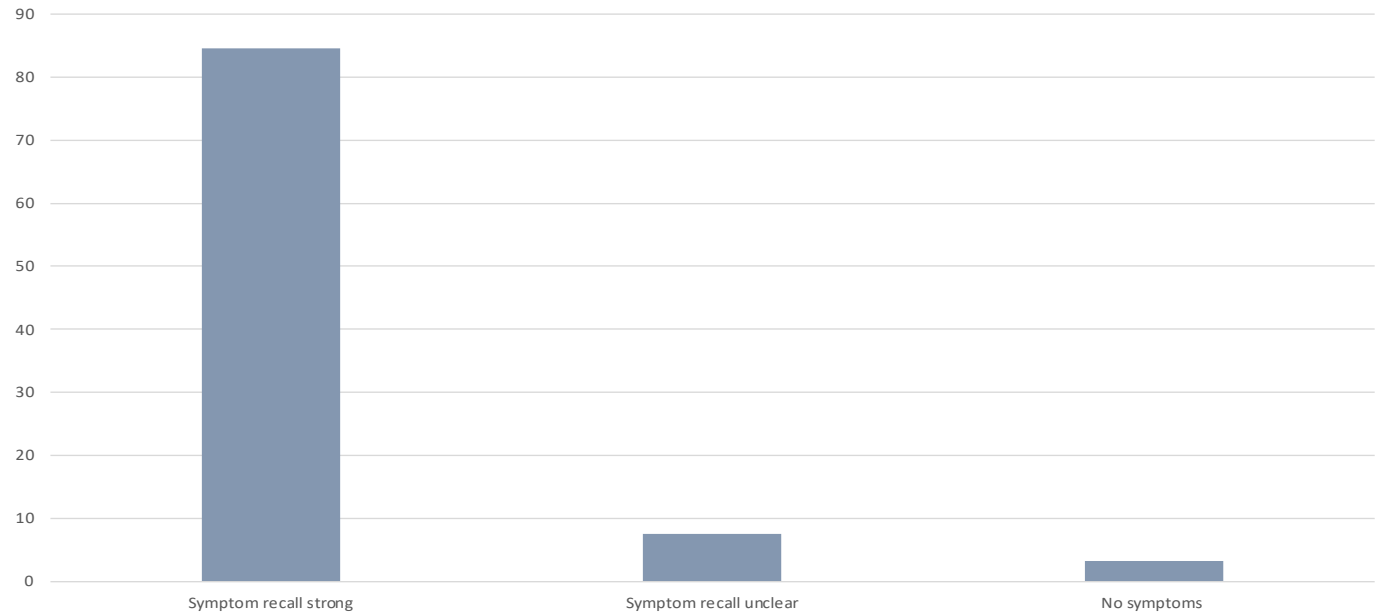

Figure 3.1: Symptom recall

Table 3.2: Symptom recall – sub group variations

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

Table 3.3: Symptoms leading to diagnosis

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

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

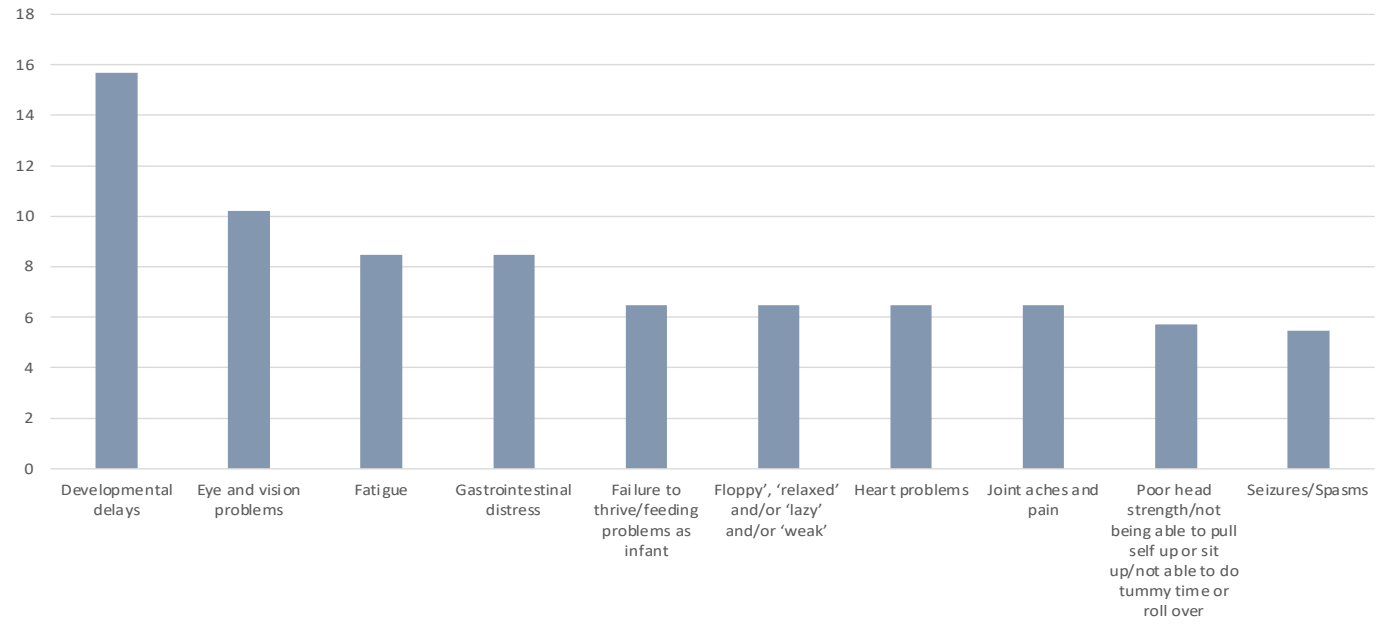

Figure 3.2: Symptoms leading to diagnosis

Table 3.4: Symptoms leading to diagnosis – subgroup variations

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

Symptoms leading to diagnosis: Seeking medical attention

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

Participant describes having symptoms and seeking medical attention relatively soon

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

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

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

Participant describes having symptoms and not seeking medical attention initially

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

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

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

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

Participant describes being diagnosed as a child

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

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

Participant describes being diagnosed through surveillance

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

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

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

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

Table 3.5: Seeking medical attention

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

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

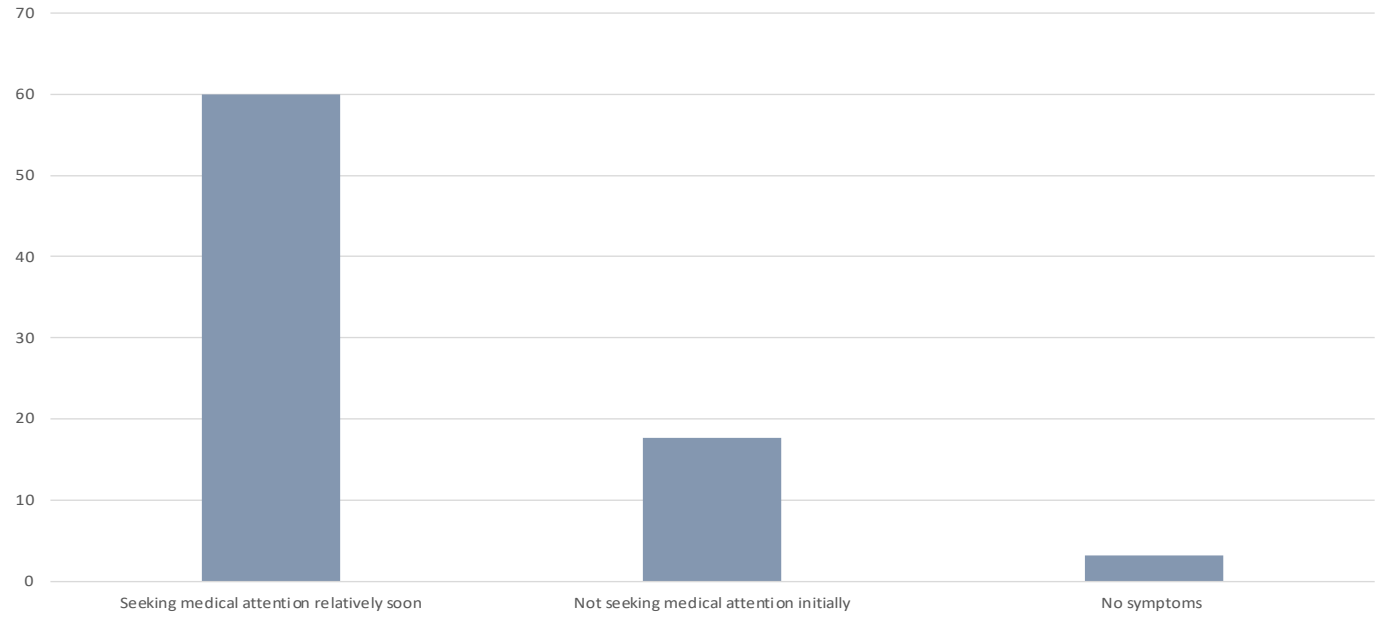

Figure 3.3: Seeking medical attention

Table 3.6: Seeking medical attention – subgroup variations

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

Symptoms leading to diagnosis: Description of diagnostic pathway

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

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

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

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

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

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

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

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

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

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

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

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

Participant describes being diagnosed in an emergency department

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Table 3.7: Diagnostic pathway

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

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

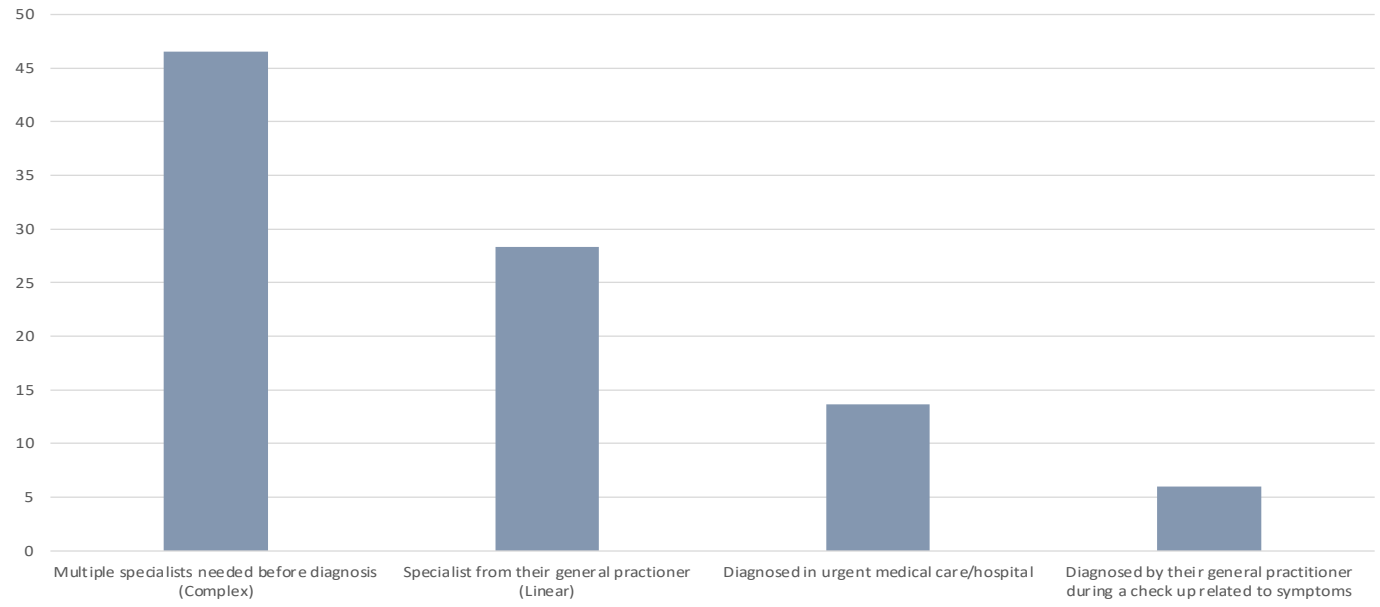

Figure 3.4: Diagnostic pathway

Table 3.8: Diagnostic pathway – subgroup variations

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

Diagnosis provider and location

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

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

Figure 3.5: Diagnosis provider

Table 3.10: Diagnosis location

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

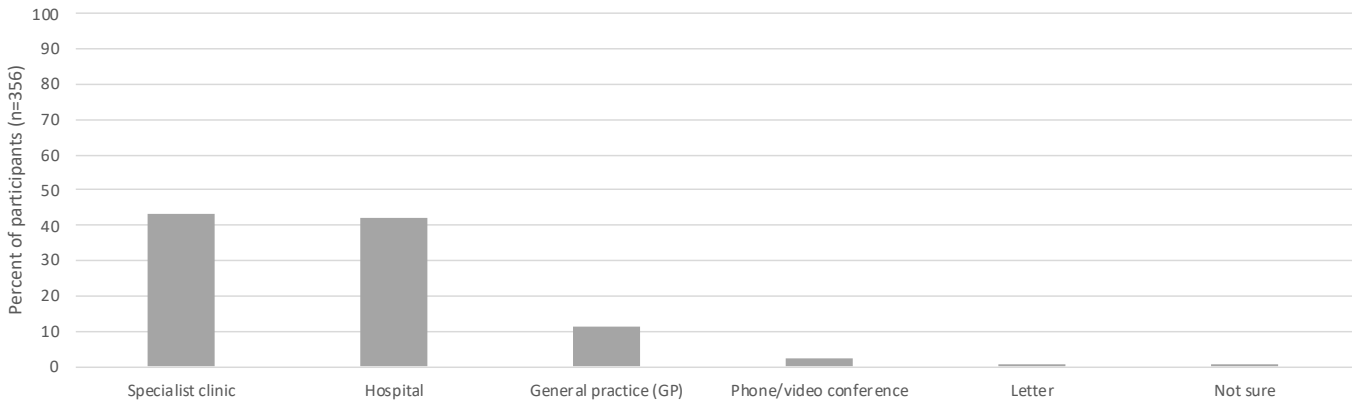

Figure 3.6: Diagnosis location

Understanding of disease at diagnosis

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Participant describes knowing about the condition due to professional background

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

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

Table 3.11: Understanding of disease at diagnosis

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

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

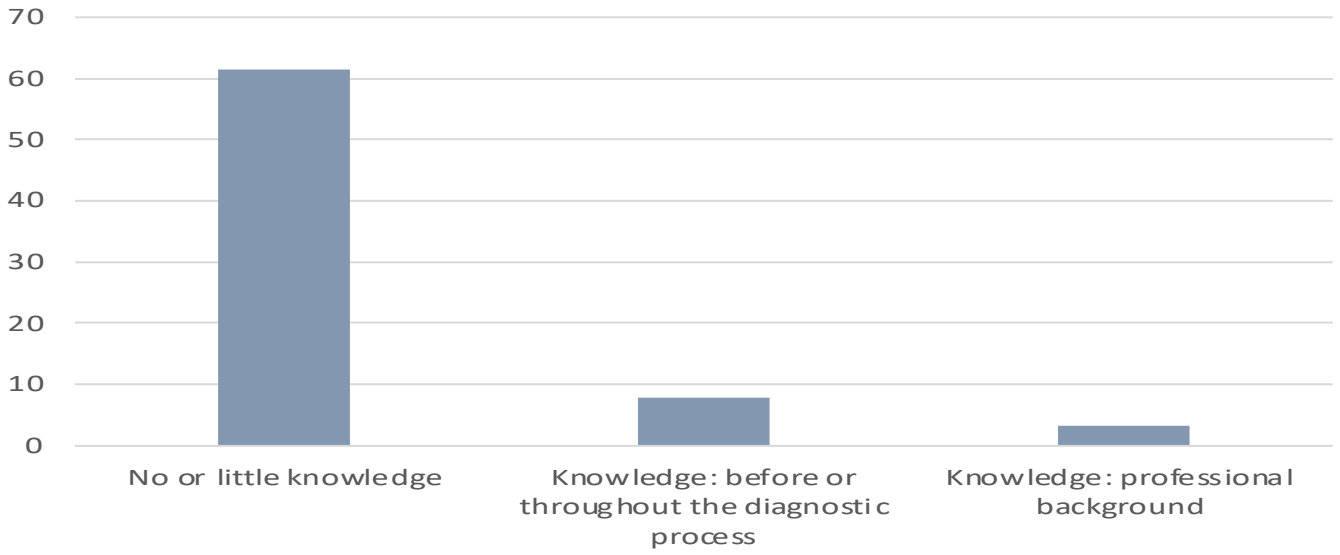

Figure 3.7: Understanding of disease at diagnosis

Table 3.12: Understanding of disease at diagnosis – subgroup variations

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

Emotional support at diagnosis

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

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

Table 3.13: Emotional support at diagnosis

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

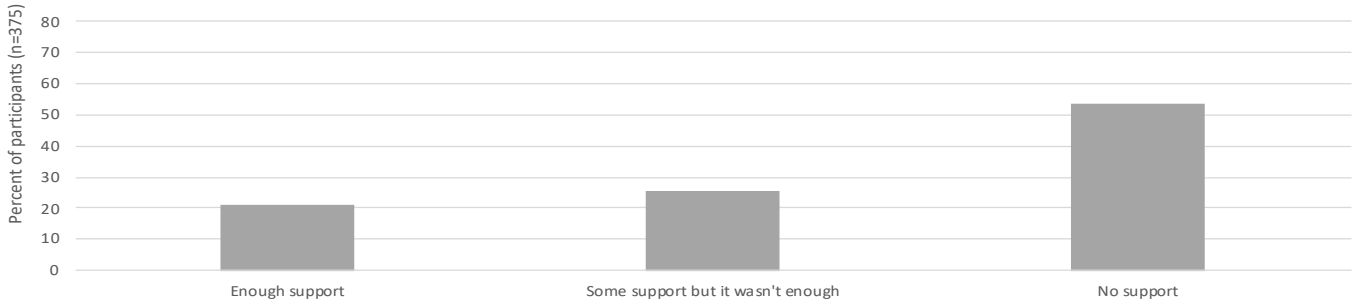

Figure 3.8: Emotional support at diagnosis

Costs at diagnosis

Out of pocket expenses at diagnosis

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

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

Burden of diagnostic costs

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

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

Table 3.14: Out of pocket expenses at diagnosis

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

Figure 3.9: Out of pocket expenses at diagnosis

Table 3.15: Burden of diagnostic costs

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

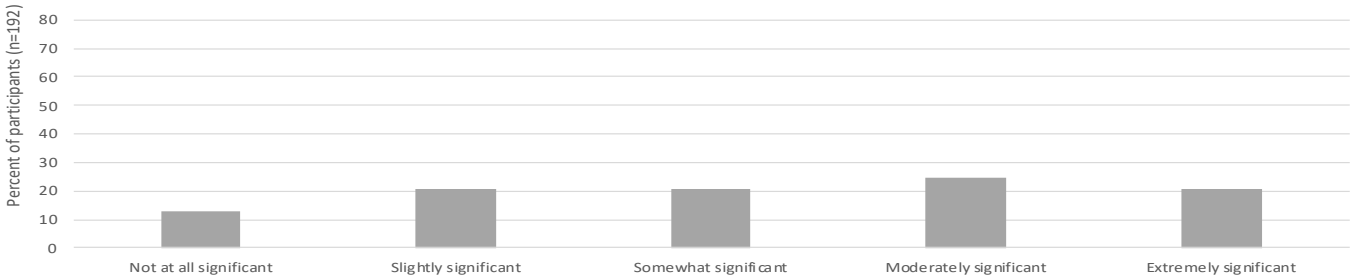

Figure 3.10: Burden of diagnostic costs

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

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

Have you had access to genetic testing?

No. Not at all.
028_2023AUDIS

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Genetic tests and biomarkers

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

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

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

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

Table 3.16: Discussions about biomarkers

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

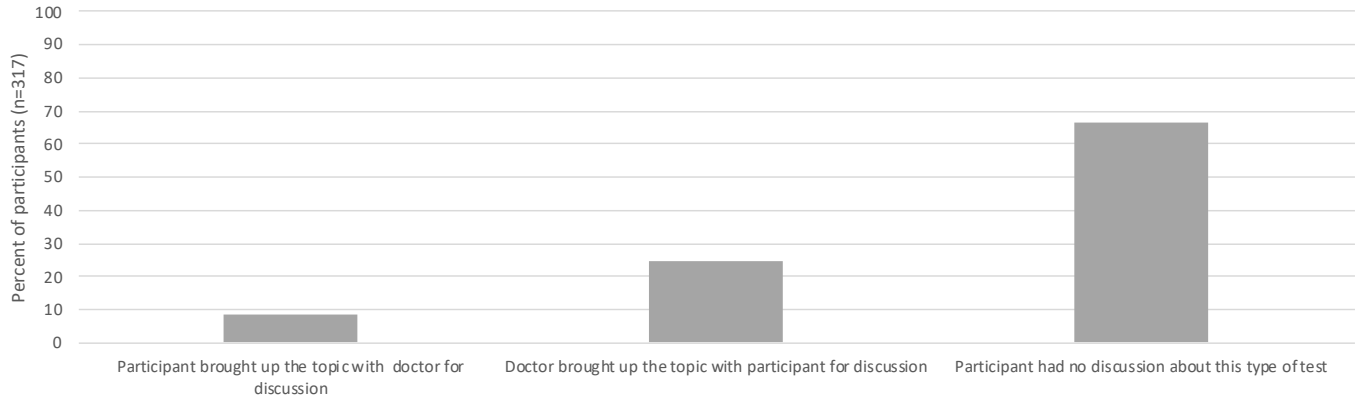

Figure 3.11: Discussions about biomarkers

Table 3.17: Experience of genetic tests and biomarkers

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

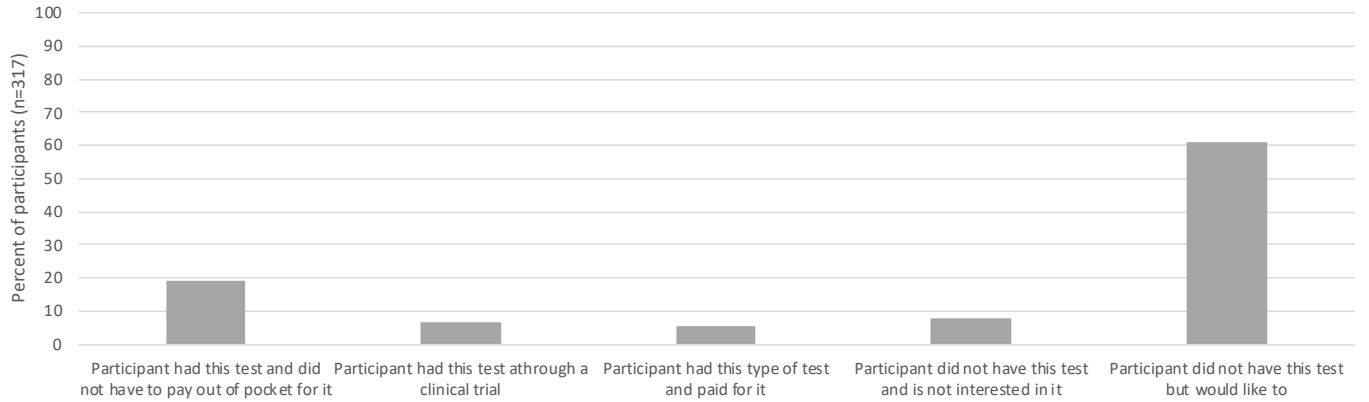

Figure 3.12: Experience of genetic tests and biomarkers

Understanding of prognosis

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

Participant describes prognosis in relation to uncertainty around prognosis

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Participant describes prognosis in relation to it being currently controlled

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

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

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

Participant describes prognosis in relation to it being a lifelong condition

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

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

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

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

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

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

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

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

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

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

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

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

Participant describes prognosis in relation to the stage of their condition

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

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

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

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

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

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

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

Participant describes prognosis in relation to allied health support

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

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

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

Table 3.18: Understanding of prognosis

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

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

Figure 3.13: Understanding of prognosis

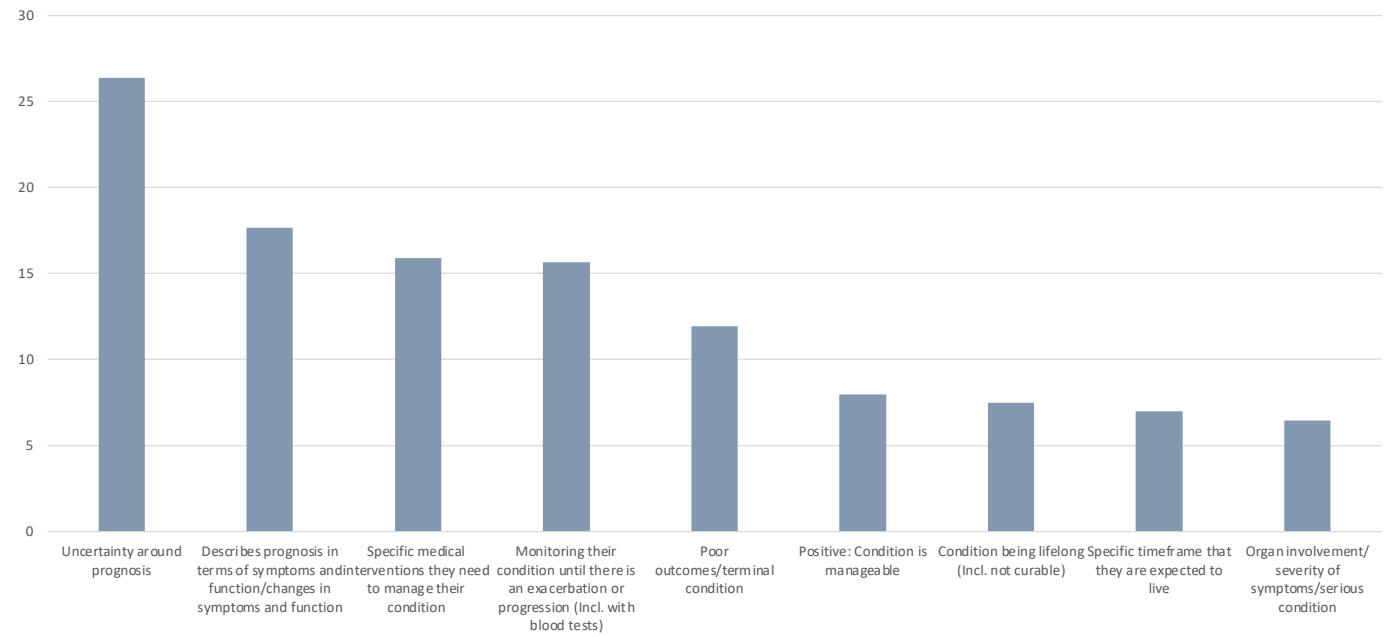

Table 3.19: Understanding of prognosis – subgroup variations

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