

Section 3

Symptoms and diagnosis

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Experience of symptoms before diagnosis

Participants were asked in the questionnaire which symptoms they consistently experienced before diagnosis, they could choose from a set list of symptoms and could then specify other symptoms not listed. There were 25 participants (50.00%) that had no symptoms before diagnosis. Participants had a maximum of 12 symptoms, and a median of 0.50 (IQR=4.75).

Symptoms before diagnosis

The most common symptoms before diagnosis were dizziness (n=13, 26.00%), weakness of face, arm, or leg (n=10, 20.00%), confusion (n=9, 18.00%), and trouble walking (n=9, 18.00%).

Participants were asked a follow up question about their quality of life while experiencing these symptoms. Quality of life was rated on a Likert scale from one to seven, where one is “Life was very distressing” and seven is “Life was great”. Median quality of life is presented where five or more participants reported the symptom.

The median quality of life was between 1 and 4, for all of the symptoms listed in the questionnaire, this is in the “Life was very distressing” to “Life was average” range. The symptoms with the worst quality of life were , weakness of face, arm, or leg and, lack of coordination, trouble seeing in one or both eyes, trouble speaking, nausea and vomiting.

Symptoms leading to diagnosis

In the online questionnaire, participants were asked to select symptoms that they consistently experienced before diagnosis. In the structured interview, participants were asked to describe the symptoms that actually *led* to their diagnosis or triggered an event.

Most commonly participants strongly recalled their symptoms or how they came to be diagnosed (74.47%). Others had no symptoms (21.28%), or had an unclear recollection of their symptoms or how they came to be diagnosed (2.13%).

The most common symptoms leading to diagnosis were shortness of breath (17.02%), headache (12.77%), irregular heartbeat (12.77%), fatigue (10.64%), dizziness or fainting (10.64%), and chest pain (8.51%). There were 10 participants that described not noticing any symptoms.

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 (51.06%), having symptoms and not seeking medical attention initially (23.40%), and having no symptoms or not noticing any symptoms before diagnosis (21.28%).

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 being diagnosed in an emergency department (55.32%), a linear diagnosis after being referred to a specialist from their general practitioner (25.53%), and being diagnosed by their general practitioner during a routine check-up that was not related to symptoms (8.51 %).

Time from diagnostic test to receiving a diagnosis

Participants were asked in the online questionnaire how long they waited between diagnostic tests and getting a diagnosis.

Participants were most commonly diagnosed immediately at the consultation (n = 19, 38.00%). There were 15 participants (30.00%) that were diagnosed less than one week after diagnostic tests, 9 participants (18.00%) diagnosed between 1 and 2 weeks, 1 participant (2.00%) diagnosed between 2 and 3 weeks, 4 participants (8.00%) diagnosed between 3 and 4 weeks, and 2 participants (4.00%) diagnosed more than four weeks after diagnostic testing.

Diagnostic tests

Participants were asked in the questionnaire which diagnostic tests they had for their diagnosis with . They could choose from a set list of diagnostic tests, and could then specify other tests not listed. The number of tests per participant were counted using both tests from the set list and other tests specified.

Participants reported between 1 to 12 diagnostic tests (median=2.00 , IQR=4.00). The most common tests were blood tests (n=33, 66.00%), electrocardiogram (n=23, 46.00%), Echocardiogram (n=15, 30.00%), and Brain CT or MRI (n=14, 28.00%).

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.

Almost half of the participants were given their diagnosis by a Emergency doctor (n=17, 34.00%), and there were 15 participants (30.00%) given the diagnosis by a Cardiologist, 12 participants (24.00%) diagnosed by General practitioner (GP), and 4 participants (8.00%) by a Neurologist.

Participants were most commonly given their diagnosis in the Hospital (n=31, 63.27%), this was followed by General practice (GP) (n=10, 20.41%), and the Specialist clinic (n=8, 16.33%).

Year of diagnosis

In the online questionnaire, participants noted the approximate date of diagnosis, the year of diagnosis is presented in the table below.

Participants were diagnosed between 2001 to 2023. There were 27 participants (55.10%) that were diagnosed in the last five years.

Understanding of disease at diagnosis

Participants were asked in the structured interview how much they knew about their condition at diagnosis. The most common responses were knowing nothing or very little about the condition at diagnosis (61.70%) and knowing about the condition at diagnosis because they have a family history of the condition or that they know someone who has the condition (14.89%). Other themes included knowing a good amount about the condition at diagnosis with no reason provided (8.51%), and knowing about the condition due to professional background (6.38%).

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 19 participants (38.00%) who had enough support, 4 participants (8.00%) that had some support but it wasn't enough, and 27 participants (54.00%) had no support.

Information at diagnosis

Participants were asked in the online questionnaire how much information they or their family received at diagnosis.

There were 15 participants (35.71%) who had enough information, 19 participants (45.24%) that had Some information but it wasn't enough, and 8 participants (19.05%) had no information.

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 21 participants (42.00%) who had no out of pocket expenses, and 18 participants (36.00%) who did not know or could not recall. There were 4 participants (8.00%) that spent \$1 to \$250, 3 participants (6.00%) that spent between \$251 to \$500, and 4 participants (8.00%) that spent \$501 or more.

Burden of diagnostic costs

For 23 participants (67.65%) the cost was slightly or not at all significant. For 7 participants (20.59%) the out-of-pocket expenses were somewhat significant, and for 4 participants (11.76%), 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.

Despite 19 participant having confirmed their LPa status, participants most commonly reported that they had never had a conversation about biomarkers, genomic, or gene testing that might be relevant to treatment, (n=43, 86.00%). There were 4 participants (8.00%) who brought up the topic with their doctor, and 3 participants (6.00%) 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.

The majority of participants did not have any genetic or biomarker tests but would like to (n=38, 76.00%). There were 10 participants (20.00%) who did not have these tests and were not interested in them, and a total of 2 participants (4.00%) that had biomarker tests.

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 they had specific medical interventions they need to manage their condition (31.91%), that they were monitoring their condition until there is an exacerbation or progression (23.40%), and that their prognosis was positive, that their condition is manageable (21.28 %). Other themes included that there was uncertainty around prognosis (19.15%), that it was a lifelong condition (14.89%),

that they need to maintain a healthy lifestyle (12.77%), and that they would likely have a recurrence, or were in a cycle of recurrence (8.51%).

Biomarker tests

Participants were asked in the structured interview if they had any discussion about biomarkers that may be important to the management of their condition. The most common responses were that they did not have any tests and did not describe reasons (48.94%), that they did not have a test but would like to have this type of test (21.28%). This was followed by no test but family history was discussed (12.77%), and had a test and management of condition was not changed (6.38%).

Experience of symptoms before diagnosis

Participants were asked in the questionnaire which symptoms they consistently experienced before diagnosis, they could choose from a set list of symptoms and could then specify other symptoms not listed.

There were 25 participants (50.00%) that had no symptoms before diagnosis. Participants had a maximum of 12 symptoms, and a median of 0.50 (IQR=4.75).

Table 3.1: Number of symptoms per participant

Number of symptoms per participant	Number (n=50)	Percent
No symptoms	25	50.00
1 to 2	5	10.00
3 to 4	7	14.00
5 to 6	4	8.00
7 to 8	5	10.00
9 or more	4	8.00

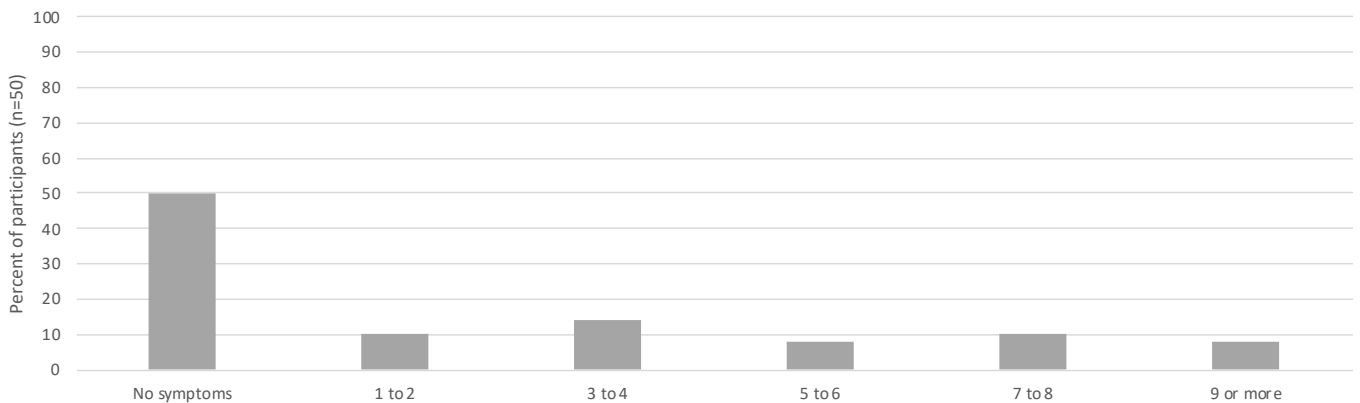


Figure 3.1: Number of symptoms per participant

Symptoms before diagnosis

The most common symptoms, consistently experienced before diagnosis were dizziness (n=13, 26.00%), weakness of face, arm, or leg (n=10, 20.00%), confusion (n=9, 18.00%), and trouble walking (n=9, 18.00%).

Participants were asked a follow up question about their quality of life while experiencing these symptoms. Quality of life was rated on a Likert scale from one to seven, where one is “Life was very distressing” and seven is “Life was great”. Median quality of life is

presented where five or more participants reported the symptom.

The median quality of life was between 1 and 4, for all of the symptoms listed in the questionnaire, this is in the “Life was very distressing” to “Life was average” range. The symptoms with the worst quality of life were, weakness of face, arm, or leg and, lack of coordination, trouble seeing in one or both eyes, trouble speaking, nausea and vomiting.

Table 3.2: Symptoms before diagnosis

Symptom	Number (n=50)	Percent	Quality of life	
			Mean	SD
No symptoms	25	50.00	NA	NA
Dizziness	13	26.00	2.00	1.00
Weakness of your face, arm, or leg	10	20.00	1.00	1.75
Confusion	9	18.00	1.50	1.25
Trouble walking	9	18.00	2.00	2.00
Lack of coordination	8	16.00	1.00	1.00
Headache	8	16.00	2.00	1.25
Memory loss	7	14.00	2.00	2.00
Blurred or double vision	7	14.00	1.00	0.50
Fatigue	7	14.00	2.00	2.50
Breathlessness	7	14.00	2.00	2.75
Trouble seeing in one or both eyes	6	12.00	1.00	0.75
Weakness	6	12.00	3.00	2.75
Trouble speaking	5	10.00	1.00	2.00
Nausea and vomiting	5	10.00	1.00	2.00
Swollen ankles, legs or stomach	4	8.00	NA	NA
Loss of appetite	4	8.00	NA	NA
Trouble understanding others talking	3	6.00	NA	NA
Weight gain	3	6.00	NA	NA
Persistent cough or wheeze	3	6.00	NA	NA

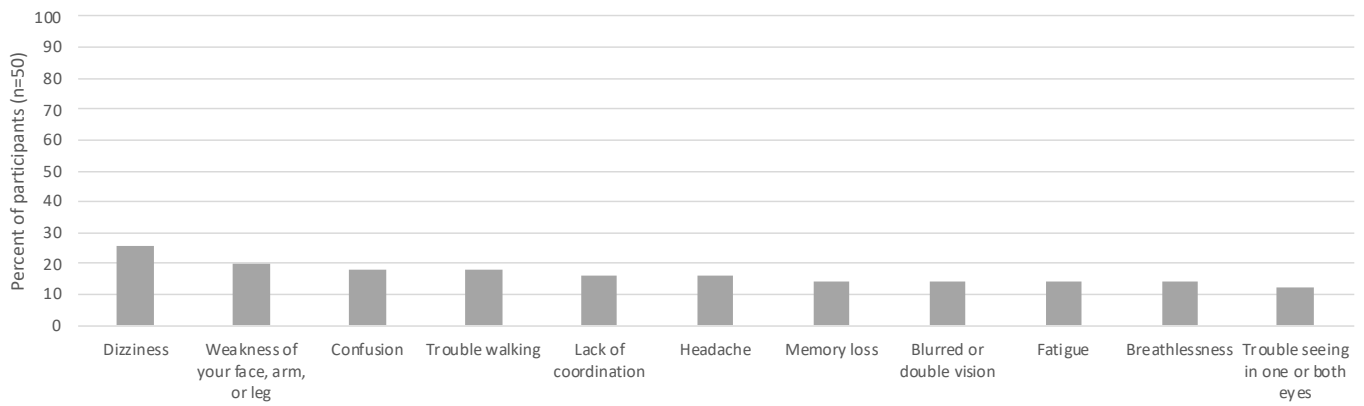


Figure 3.2: Symptoms before diagnosis

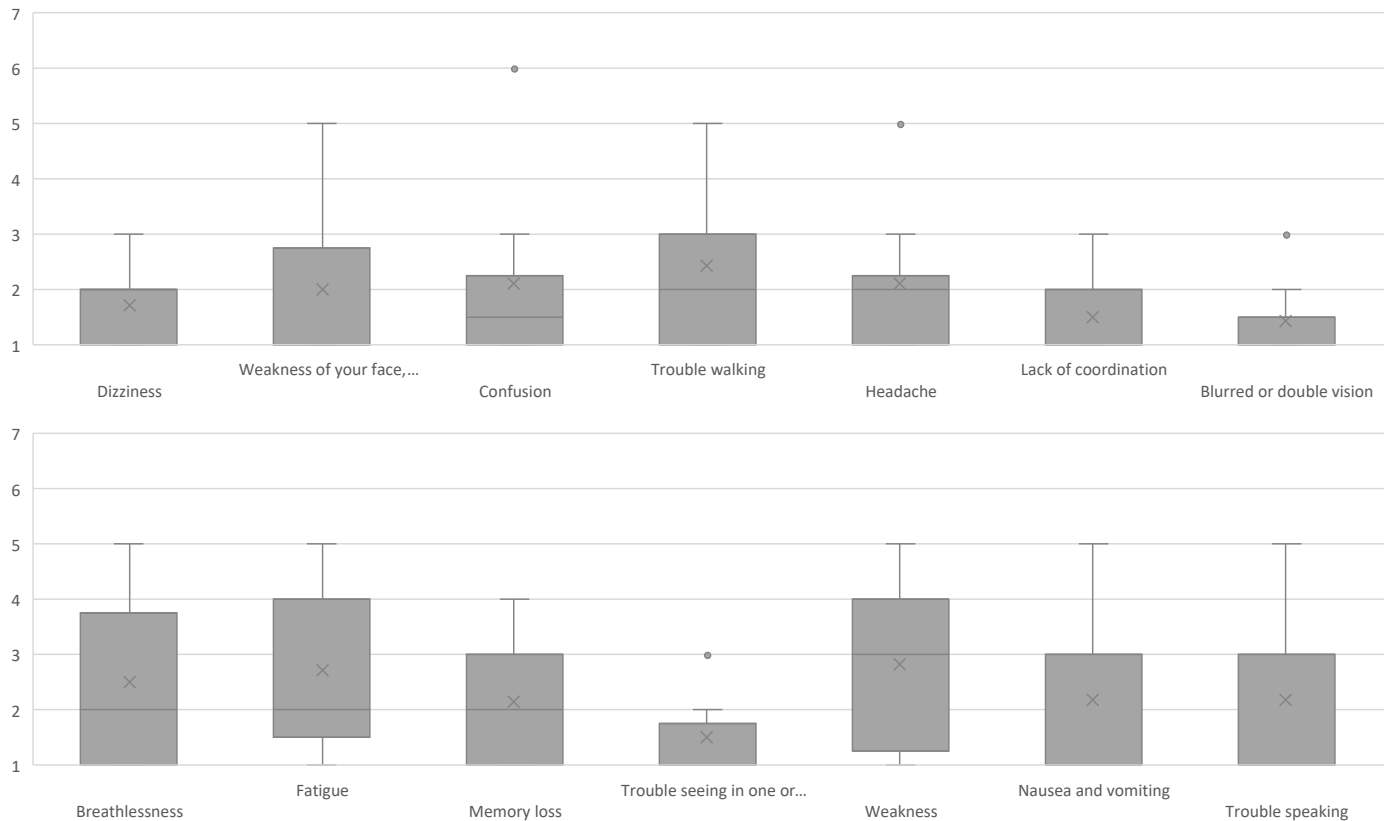


Figure 3.3: Quality of life from symptoms before diagnosis

Symptoms leading to diagnosis

In the online questionnaire, participants were asked to select symptoms that they consistently experienced before diagnosis. In the structured interview, participants were asked to describe the symptoms that actually *led* to their diagnosis or triggered an event.

Most commonly participants strongly recalled their symptoms or how they came to be diagnosed (74.47%). Others had no symptoms (21.28%), or had an unclear recollection of their symptoms or how they came to be diagnosed (2.13%).

The most common symptoms leading to diagnosis were shortness of breath (17.02%), headache (12.77%), irregular heartbeat (12.77%), fatigue (10.64%),

dizziness or fainting (10.64%), and chest pain (8.51%). There were 10 participants that described not noticing any symptoms.

Participant describes having no symptoms at the time

It's one of these like blood details or artifacts that where it just increases your risk factor and thankfully I have not had a cardiac event which can that's what it can lead to type of thing. So no, I haven't had any symptoms.

Participant 001_2023AUHBV

None at all. It was a circumstantial finding.

Participant 010_2023AUHBV

I didn't really have any symptoms until the actual stroke was happening.

Participant 047_2023AUHBV

I didn't notice any symptoms at all before I had a stroke. I went to sleep and I woke up and the whole right side of my body was paralysed. I laughed because I thought, "Pinch of the nerve," and then I realised that something more serious than that was going on. That's how I got a stroke.

Participant 048_2023AUHBV

Participant describes having shortness of breath which led to their diagnosis

Yeah, yes, it was breathlessness and just doing mild exercise caused me to become as you breath and my heart rate to go up more than you would expect, yeah.

Participant 017_2023AUHBV

I'll start at my 40s because that was my initial diagnosis, so in my 40s my daughter would have been about two, and there was one particular day when I was pushing the pram and I just felt like I couldn't breathe. I just literally had to hold onto the pram and just hold still for a few minutes until it passed, and then one particular day I think I had three episodes like that in the day, and I thought, "Okay, I need to go and see someone," because I do have a family history of AF from my brother had AF and my dad has AF. So I did, and initially I wore a Holter monitor for 48 hours and had the ECG and all that stuff, and then I went to a cardiologist and he diagnosed the VT.

Participant 023_2023AUHBV

Yeah. I was very short of breath. I've never been good at cardiac activity for most of my adult life. I've struggled with any kind of physical exercise. I've always put it down to the fact that I was just unfit. I always classed myself as an unfit person. I wasn't an overweight person. I'm not thin by any means, but I'm not chronically overweight or anything. There was one point where I was trying to get into the police force, so I was doing training. I was trying to run and improve my fitness.

Participant 036_2023AUHBV

The symptoms I had were just short of breath, bloating. Yeah, those were the main two. I didn't know it was heart failure obviously at the time until the doctor told me, but yeah I thought I was overweight.

Participant 038_2023AUHBV

Participant describes having headache which led to their diagnosis

Yes. I woke up that morning. I had a headache all day. I still went to work. It got worse by lunchtime, I was getting real bad, and then by I think one or two or three o'clock, I just couldn't see. I felt weird.

Participant 042_2023AUHBV

I recalled saying to her, "Got a bit of a headache, I might go and have to lie down, but call me tonight and let me know what we're doing." As I went to hang the phone up, I fell through a double glass window and that was the stroke.

Participant 050_2023AUHBV

Participant describes having irregular heartbeat which led to their diagnosis

I don't think there really wasn't mine. Like, no one's actually said what started it, but I had my third COVID vaccination and then within two or three days, the tachycardia started and it was just out of the way. All of a sudden my heart rate was 150 from just sitting on the couch. It just zoomed up and that's where it stayed.

Participant 008_2023AUHBV

Yeah. I had no idea. Because mine was pregnancy induced, every symptom that I had was a pregnancy symptom. Like a normal pregnancy symptom, so my symptoms were, I had difficulty breathing, and I had an elevated heart rate and swollen ankles. Which all three were very much just normal parts of pregnancy. And it was only the day after my little boy was born that I went, "Oh, I'm still struggling to breathe." And so then I mentioned it to the nurse saying, "It's probably nothing but I'm just having a little difficulty breathing." And then I was diagnosed the day following that.

Participant 035_2023AUHBV

Participant describes having fatigue which led to their diagnosis

I was exercising a lot, doing some extra labour work than normal and I was feeling. Extremely run down and exhausted, not recovering well from it that that was basically what started is being fatigue, extreme fatigue after slightly increasing activity.

Participant 003_2023AUHBV

I would say my early symptom without being aware of it was being tired.

Participant 005_2023AUHBV

I don't think I had any symptoms. I was a runner and the day before I had my condition I actually was actually really tired and didn't do much that day. Probably say I didn't feel well but didn't feel sick. Just it's, you know, it's just something that in hindsight I'd thought about. And then the next day I was still quite tired and made myself go for a run thinking that it would up my energy a little bit.

Participant 009_2023AUHBV

I had no symptoms at all. I had a massive heart attack... No warning whatsoever, except that I was very tired the night before.

Participant 033_2023AUHBV

Participant describes having dizziness or fainting which led to their diagnosis

I noticed that I had had dizziness for some time, and I had been monitored, and nothing came of it because

during the monitor, nothing was happening. Participant 034_2023AUHBV

I had dizziness and I kept falling over. I kept vomiting and my dizziness had increased a lot.

Participant 039_2023AUHBV

Participant describes having chest pain which led to their diagnosis

So I didn't really notice much in the initial stages. It basically came down to the fact where I had chest pain one afternoon. I basically just wandered into the hospital. Said to them that I had chest pain, which was traveling. It was actually traveling up into the jaw, through the chest, down my left leg, but down my right arm. Yeah. At the time, I think my blood pressure was 280 / 140, yeah.

Participant 018_2023AUHBV

I had pins and needles in in my arms and I had similar to to like a I was getting short pains into my chest area which. That was basically all the all of the symptoms that I know of now that would have been telling signs.

Participant 006_2023AUHBV

Table 3.3: Symptom recall

Symptom recall	All participants		Had LP(a) test		Did not had LP(a) test		High cholesterol under 50 years of age		Blood vessel conditions		Heart conditions		0 to 5 other conditions		6 to 11 other conditions	
	n=47	%	n=18	%	n=29	%	n=9	%	n=17	%	n=21	%	n=27	%	n=20	%
Symptom recall strong	35	74.47	14	77.78	21	72.41	4	44.44	12	70.59	19	90.48	21	77.78	14	70.00
No Symptoms	10	21.28	3	16.67	7	24.14	4	44.44	5	29.41	1	4.76	5	18.52	5	25.00
Symptom recall unclear	1	2.13	1	5.56	0	0.00	1	11.11	0	0.00	0	0.00	1	3.70	0	0.00
No particular comment	3	6.38	1	5.56	2	6.90	2	22.22	0	0.00	1	4.76	0	0.00	3	15.00

Symptom recall	All participants		Female		Male		Aged 25 to 44		Aged 45 and older		Regional or remote		Metropolitan		Mid to low socioeconomic status		Higher socioeconomic status	
	n=47	%	n=26	%	n=21	%	n=24	%	n=23	%	n=15	%	n=32	%	n=25	%	n=22	%
Symptom recall strong	35	74.47	20	76.92	15	71.43	16	66.67	19	82.61	8	53.33	27	84.38	18	72.00	17	77.27
No Symptoms	10	21.28	6	23.08	4	19.05	6	25.00	4	17.39	7	46.67	3	9.38	7	28.00	3	13.64
Symptom recall unclear	1	2.13	0	0.00	1	4.76	1	4.17	0	0.00	0	0.00	1	3.13	0	0.00	1	4.55
No particular comment	3	6.38	2	7.69	1	4.76	3	12.50	0	0.00	0	0.00	3	9.38	0	0.00	3	13.64

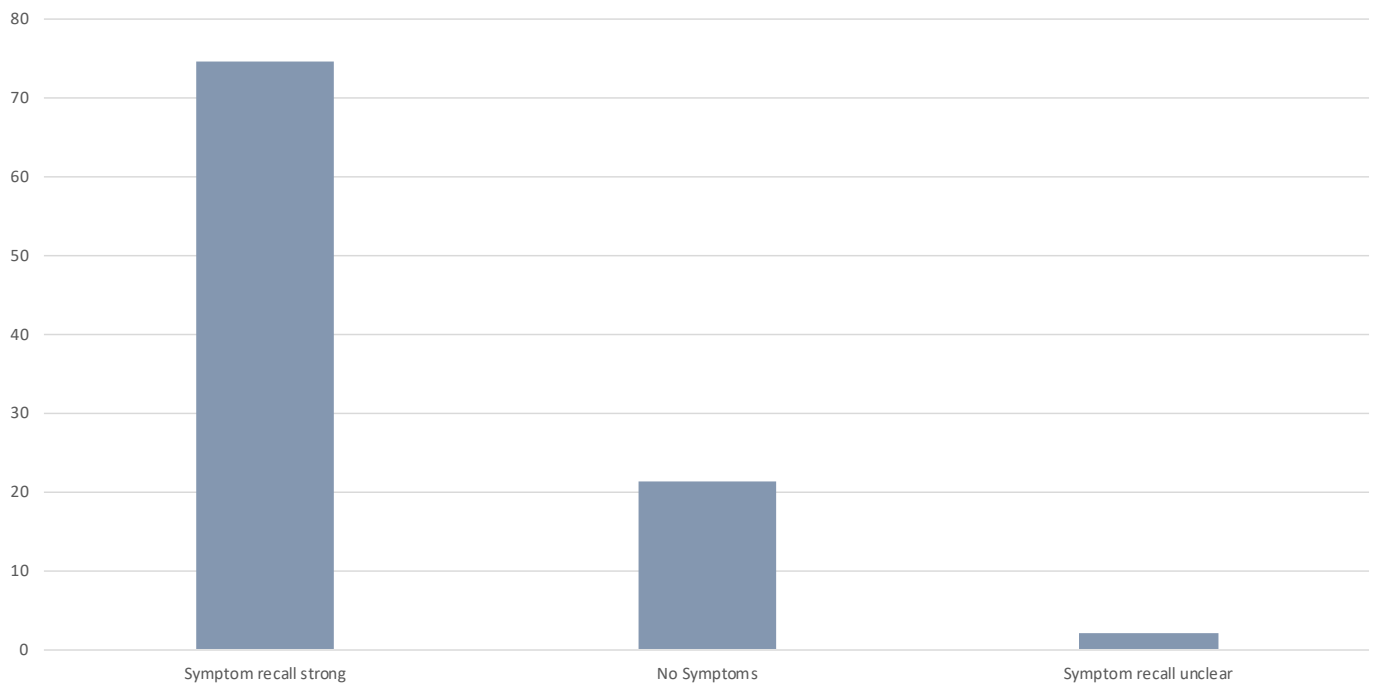


Figure 3.4: Symptom recall

Symptom recall	Reported less frequently	Reported more frequently
Symptom recall strong	High cholesterol under 50 years of age Regional or remote	Heart conditions
No Symptoms	Heart conditions Metropolitan	High cholesterol under 50 years of age Regional or remote

Table 3.4: Symptoms leading to diagnosis

Symptoms leading to diagnosis	All participants		Had LP(a) test		Did not had LP(a) test		High cholesterol under 50 years of age		Blood vessel conditions		Heart conditions		0 to 5 other conditions		6 to 11 other conditions	
	n=47	%	n=18	%	n=29	%	n=9	%	n=17	%	n=21	%	n=27	%	n=20	%
Participant describes having no symptoms	10	21.28	3	16.67	7	24.14	4	44.44	5	29.41	1	4.76	5	18.52	5	25.00
Participant describes having shortness of breath which led to their diagnosis	8	17.02	3	16.67	5	17.24	1	11.11	0	0.00	7	33.33	4	14.81	4	20.00
Participant describes having headache which led to their diagnosis	6	12.77	0	0.00	6	20.69	0	0.00	6	35.29	0	0.00	2	7.41	4	20.00
Participant describes having irregular heartbeat which led to their diagnosis	6	12.77	4	22.22	2	6.90	2	22.22	0	0.00	4	19.05	3	11.11	3	15.00
Participant describes having fatigue which led to their diagnosis	5	10.64	0	0.00	5	17.24	1	11.11	2	11.76	2	9.52	5	18.52	0	0.00
Participant describes having dizziness or fainting which led to their diagnosis	5	10.64	4	22.22	1	3.45	2	22.22	1	5.88	2	9.52	3	11.11	2	10.00
Participant describes having chest pain which led to their diagnosis	4	8.51	4	22.22	0	0.00	0	0.00	0	0.00	4	19.05	3	11.11	1	5.00

Symptoms leading to diagnosis	All participants		Female		Male		Aged 25 to 44		Aged 45 and older		Regional or remote		Metropolitan		Mid to low socioeconomic status		Higher socioeconomic status	
	n=47	%	n=26	%	n=21	%	n=24	%	n=23	%	n=15	%	n=32	%	n=25	%	n=22	%
Participant describes having no symptoms	10	21.28	6	23.08	4	19.05	6	25.00	4	17.39	7	46.67	3	9.38	7	28.00	3	13.64
Participant describes having shortness of breath which led to their diagnosis	8	17.02	5	19.23	3	14.29	5	20.83	3	13.04	2	13.33	6	18.75	2	8.00	6	27.27
Participant describes having headache which led to their diagnosis	6	12.77	4	15.38	2	9.52	3	12.50	3	13.04	0	0.00	6	18.75	4	16.00	2	9.09
Participant describes having irregular heartbeat which led to their diagnosis	6	12.77	4	15.38	2	9.52	5	20.83	1	4.35	2	13.33	4	12.50	3	12.00	3	13.64
Participant describes having fatigue which led to their diagnosis	5	10.64	5	19.23	0	0.00	1	4.17	4	17.39	2	13.33	3	9.38	2	8.00	3	13.64
Participant describes having dizziness or fainting which led to their diagnosis	5	10.64	3	11.54	2	9.52	2	8.33	3	13.04	3	20.00	2	6.25	4	16.00	1	4.55
Participant describes having chest pain which led to their diagnosis	4	8.51	1	3.85	3	14.29	3	12.50	1	4.35	0	0.00	4	12.50	2	8.00	2	9.09

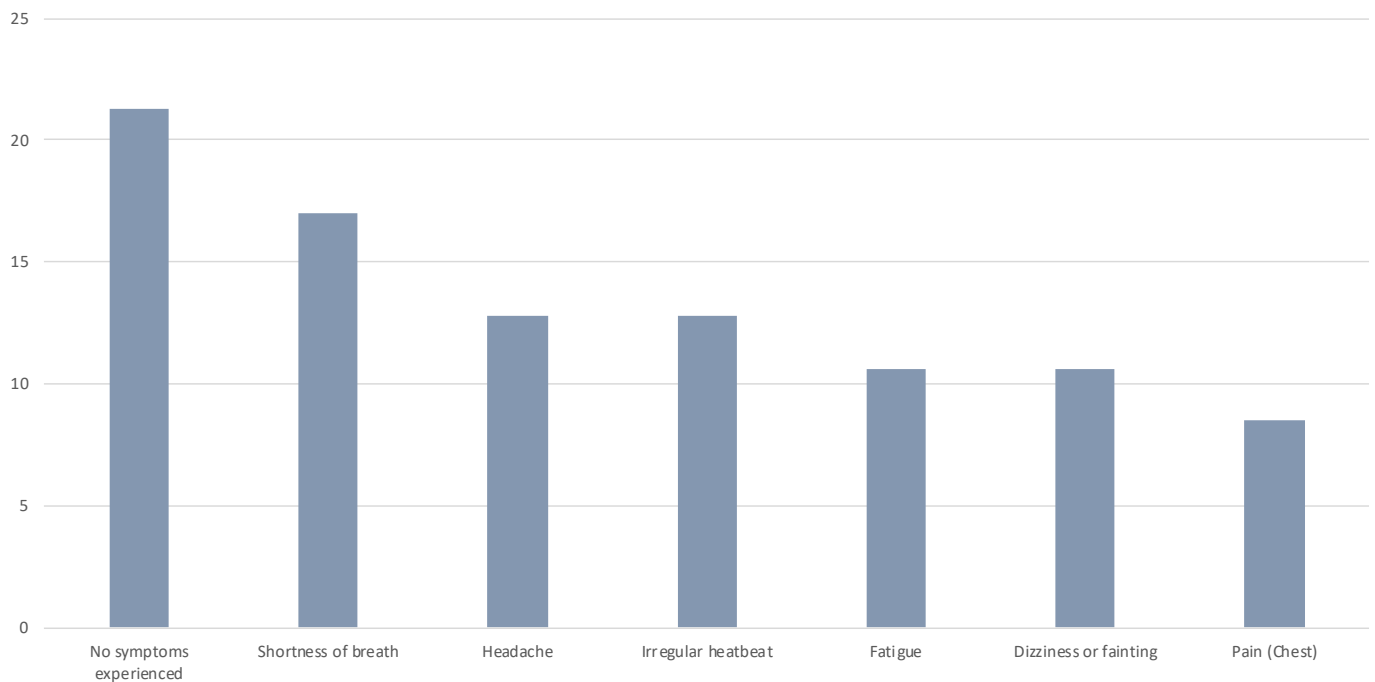


Figure 3.5: Symptoms leading to diagnosis

Table 3.5: Symptoms leading to diagnosis – subgroup variations

Symptoms leading to diagnosis	Reported less frequently	Reported more frequently
Participant describes having no symptoms	Heart conditions Metropolitan	High cholesterol under 50 years of age Regional or remote
Participant describes having shortness of breath which led to their diagnosis	Blood vessel conditions	Heart conditions Higher socioeconomic status
Participant describes having headache which led to their diagnosis	Had LP(a) test High cholesterol under 50 years of age Heart conditions Regional or remote	Blood vessel conditions
Participant describes having irregular heartbeat which led to their diagnosis	Blood vessel conditions	-
Participant describes having fatigue which led to their diagnosis	Had LP(a) test 6 to 11 other conditions Male	-
Participant describes having dizziness or fainting which led to their diagnosis	-	Had LP(a) test High cholesterol under 50 years of age
Participant describes having chest pain which led to their diagnosis	-	Had LP(a) test Heart conditions

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 (51.06%), having symptoms and not seeking medical attention initially (23.40%), and having no symptoms or not noticing any symptoms before diagnosis (21.28 %).

Participant describes having symptoms and seeking medical attention relatively soon

I don't think there really wasn't mine. Like, no one's actually said what started it, but I had my third COVID vaccination and then within two or three days, the tachycardia started and it was just out of the way. All of a sudden my heart rate was 150 from just sitting on the couch. It just zoomed up and that's where it stayed. Well, I went down to my local GP and they

took me into the nurses station and hooked me up to an ECG.

Participant 008_2023AUHBV

I basically just wandered into the hospital. Said to them that I had chest pain, which was traveling. It was actually traveling up into the jaw, through the chest, down my left leg, but down my right arm. Yeah. At the time, I think my blood pressure was 280 / 140, yeah.

Participant 018_2023AUHBV

Obviously, I was quite ill for a period. I should've gone to hospital straight away and I didn't unfortunately. I didn't go to the doctor until 24 hours later. I was very ill in those 24 hours. I finally went to the doctor and the doctor basically sent me to Emergency immediately. Then they did all the tests and I think they diagnosed it in Emergency within the first couple

of hours. Then, I woke up the following day on the stroke ward in LOCATION METROPOLITAN and the neurologist came and saw me that morning and advised me that I had a double occipital lobe stroke.
Participant 043_2023AUHBV

Participant describes having symptoms and not seeking medical attention initially

Right, so I just happened to be at the doctor getting scripts filled. I was just 53 at the time and had pretty good relationship with my doctor and we just got chatting about things and I happened to mention to her that my father had a heart attack at 53 and everyone always says, you know, you're so like your dad. So I said, you know, you know, should I be concerned or anything? And she said, have you any reason to be? And I said no, absolutely not. And so she said, well, there's no, there's no point. And she said we can do a stress test if you like. So I said, oh, well, I'm not go and do that.... So he said I'd refer you to a cardiologist. And I said, oh, I might just do that. I'm sure that was my father sitting on my shoulder and go and do that. Anyway, the cardiologist took me in and he said I don't think there's anything to be concerned about either. And so he did that. And then he came out a short time later and he actually apologized to me, said I am so sorry. And you have major blockages in major arteries.
Participant 005_2023AUHBV

Yeah. I was very short of breath. I've never been good at cardiac activity for most of my adult life. I've struggled with any kind of physical exercise. I've always put it down to the fact that I was just unfit. I always classed myself as an unfit person. I wasn't an

overweight person. I'm not thin by any means, but I'm not chronically overweight or anything. There was one point where I was trying to get into the police force, so I was doing training. I was trying to run and improve my fitness.
Participant 036_2023AUHBV

I was exercising a lot, doing some extra labour work than normal and I was feeling. Extremely run down and exhausted, not recovering well from it that that was basically what started is being fatigue, extreme fatigue after slightly increasing activity. After my children were born, I became more interested in health and what we were eating and I think I had a random blood test in it was like 2016 and it it it said high cholesterol...I was feeling healthy.
Participant 003_2023AUHBV

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

So I didn't have any signs or symptoms. It was just picked up in a general health check, my annual health check on my blood test.
Participant 020_2023AUHBV

Yeah. So for me it was quite different. So I didn't have any symptoms at all. I heart condition was picked up after I had a serious motorcycle accident and they noticed that my ECG's were quite abnormal. Obviously while they've they've caught me on the table doing whatever they got to do and that's that's when they found out that that I had what they call a right bundle branch blockage and and all that and then put me onto a cardiologist from there.
Participant 021_2023AUHBV

Table 3.6: Seeking medical attention

Seeking medical attention	All participants		Had LP(a) test		Did not had LP(a) test		High cholesterol under 50 years of age		Blood vessel conditions		Heart conditions		0 to 5 other conditions		6 to 11 other conditions	
	n=47	%	n=18	%	n=29	%	n=9	%	n=17	%	n=21	%	n=27	%	n=20	%
Participant describes having symptoms and seeking medical attention relatively soon	24	51.06	8	44.44	16	55.17	1	11.11	10	58.82	13	61.90	14	51.85	10	50.00
Participant describes having symptoms and not seeking medical attention initially	11	23.40	6	33.33	5	17.24	3	33.33	2	11.76	6	28.57	7	25.93	4	20.00
Participant describes having no symptoms or not noticing any symptoms before diagnosis	10	21.28	3	16.67	7	24.14	4	44.44	5	29.41	1	4.76	5	18.52	5	25.00
No particular comment	2	4.26	1	5.56	1	3.45	1	11.11	0	0.00	1	4.76	1	3.70	1	5.00

Seeking medical attention	All participants		Female		Male		Aged 25 to 44		Aged 45 and older		Regional or remote		Metropolitan		Mid to low socioeconomic status		Higher socioeconomic status	
	n=47	%	n=26	%	n=21	%	n=24	%	n=23	%	n=15	%	n=32	%	n=25	%	n=22	%
Participant describes having symptoms and seeking medical attention relatively soon	24	51.06	13	50.00	11	52.38	10	41.67	14	60.87	4	26.67	20	62.50	14	56.00	10	45.45
Participant describes having symptoms and not seeking medical attention initially	11	23.40	7	26.92	4	19.05	6	25.00	5	21.74	4	26.67	7	21.88	4	16.00	7	31.82
Participant describes having no symptoms or not noticing any symptoms before diagnosis	10	21.28	6	23.08	4	19.05	6	25.00	4	17.39	7	46.67	3	9.38	7	28.00	3	13.64
No particular comment	2	4.26	0	0.00	2	9.52	2	8.33	0	0.00	0	0.00	2	6.25	0	0.00	2	9.09

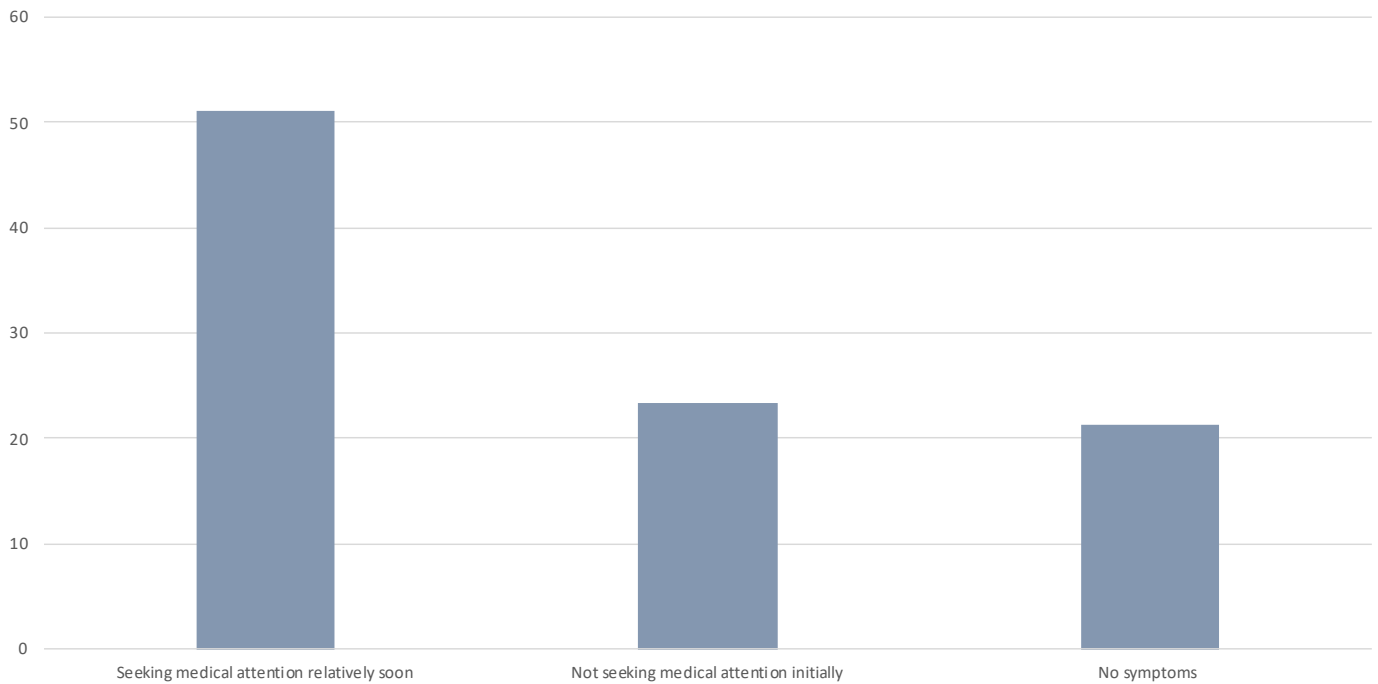


Figure 3.6: Seeking medical attention

Table 3.7: Seeking medical attention – subgroup variations

Seeking medical attention	Reported less frequently	Reported more frequently
Participant describes having symptoms and seeking medical attention relatively soon	High cholesterol under 50 years of age Regional or remote	Heart conditions Metropolitan
Participant describes having symptoms and not seeking medical attention initially	Blood vessel conditions	-
Participant describes having no symptoms or not noticing any symptoms before diagnosis	Heart conditions Metropolitan	High cholesterol under 50 years of age Regional or remote

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 being diagnosed in an emergency department (55.32%), a linear diagnosis after being referred to a specialist from their general practitioner (25.53%), and being diagnosed by their general practitioner during a routine check-up that was not related to symptoms (8.51 %).

Participant describes being diagnosed in an emergency department

I kept vomiting and my dizziness had increased a lot. I couldn't hold my baby because that's a newborn baby. I couldn't hold it. I went to the doctor's. I went to the hospital. Our doctors are in the hospital. My family has a history of stroke. I knew that I was possibly having a stroke. had a CT scan done after going to the hospital originally where they said, "You've got an aneurysm." That's when I was rushed there of course. From there I had an angiogram. Participant 039_2023AUHBV

Yes. I had high blood pressure, very high blood pressure. I collapsed when I had the stroke, and I was

diagnosed in the A&E department of the hospital. Participant 040_2023AUHBV

I was at work. My boss called an ambulance straight away. They took me to hospital straight away by ambulance. Then ambulance people took me there. They did a lot of tests. I think it was six hours of waiting and then they told me, "Yes, you've had a stroke."

Participant 042_2023AUHBV

I had a doctor's appointment in about three quarters of an hour, got out of the pool, got into the shower, went to wash my hair, started washing my hair, and an elephant sat on my chest. It was a massive heart attack. By the time the ambulance stabilised me, I had three ambulances here, it took them a while to stabilise me. I was taken to NAME HOSPITAL. The entire time in the ambulance they were trying to get permission to give me the drug. I got to the base, and I don't recall much about it except it was mad pain, of course, and I was there all night in massive pain. I had morphine pouring in. That was on a Wednesday. The Thursday morning the cardiologist, whom I'd never met before, came to me and said, "You've had a

massive heart attack and you need stents. Unfortunately today's list is full. You'll have to wait until the following Tuesday."
Participant 033_2023AUHBV

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

Well, the diagnosis came from visiting my cardiologist, and he indicated to me that I had atrial fibrillation, I had tachycardia. I had cardiovascular disease, and that's just to name a few.
Participant 031_2023AUHBV

There was a doctor there that asked me if I had a heart problem. It was during one of my hospital visits. He commented on my heart. I was almost due to have the baby, so I was more concerned with the baby at that point and didn't really think about my own health as such. He said, "Do you have a heart problem?" And, I said, "No." He said to me at the time, and I didn't remember this until all of this came up, but he said to me, "I'd like you to see a cardiologist after you've had the baby just to have a check-up." I sort of went, "Yeah. Yeah. Okay." Then it left my mind and I never thought anything of it again. It was just some doctor in the hospital he wasn't my doctor. Yeah, so anyway the doctor said, "You've got a heart murmur we're going to send you for some more tests." That just led from one thing to another, and obviously the diagnosis of HOCM. I was just very short of breath. Sorry I can't remember the original question.
Participant 036_2023AUHBV

I went back to my daughter's house and thought I suppose I should do something about this because I couldn't stop them, so I rang her and she came home and I went down to my GP, or the kids' GP because they live out on the other side of LOCATION METROPOLITAN. And he sent me for a blood test to see if I'd had a heart attack and the palpitations stopped so I drove home. The results were sent to my GP over on this side of LOCATION METROPOLITAN, and he said, "Nothing to worry about." The GP over where my daughter lives rang me up and said, "Get yourself in here." He sent me off to a cardiologist who said that I had ischemic heart disease and I needed to have an angiogram
Participant 037_2023AUHBV

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

So I didn't have any signs or symptoms. It was just picked up in a general health check, my annual health check on my blood test
Participant 020_2023AUHBV

Yeah, it was more of some of those routine year yearly GP visits and then they ordered those, you know, blood panel tests. Those were when my blood results were looking starting to look abnormal, especially you know like with the LDL for example.
Participant 001_2023AUHBV

Table 3.8: Diagnostic pathway

Diagnostic pathway	All participants		Had LP(a) test		Did not had LP(a) test		High cholesterol under 50 years of age		Blood vessel conditions		Heart conditions		0 to 5 other conditions		6 to 11 other conditions	
	n=47	%	n=18	%	n=29	%	n=9	%	n=17	%	n=21	%	n=27	%	n=20	%
Participant describes being diagnosed in an emergency department	26	55.32	13	72.22	13	44.83	4	44.44	11	64.71	11	52.38	14	51.85	12	60.00
Participant describes a linear diagnosis after being referred to a specialist from their general practitioner	12	25.53	3	16.67	9	31.03	0	0.00	4	23.53	8	38.10	9	33.33	3	15.00
Participant describes being diagnosed by their general practitioner during a routine check-up that was not related to symptoms	4	8.51	1	5.56	3	10.34	4	44.44	0	0.00	0	0.00	2	7.41	2	10.00
Participant describes a complex diagnosis, needing to see multiple specialists before diagnosis	1	2.13	0	0.00	1	3.45	0	0.00	1	5.88	0	0.00	0	0.00	1	5.00
No particular comment	4	8.51	1	5.56	3	10.34	1	11.11	1	5.88	2	9.52	2	7.41	2	10.00

Diagnostic pathway	All participants		Female		Male		Aged 25 to 44		Aged 45 and older		Regional or remote		Metropolitan		Mid to low socioeconomic status		Higher socioeconomic status	
	n=47	%	n=26	%	n=21	%	n=24	%	n=23	%	n=15	%	n=32	%	n=25	%	n=22	%
Participant describes being diagnosed in an emergency department	26	55.32	15	57.69	11	52.38	12	50.00	14	60.87	10	66.67	16	50.00	16	64.00	10	45.45
Participant describes a linear diagnosis after being referred to a specialist from their general practitioner	12	25.53	6	23.08	6	28.57	6	25.00	6	26.09	3	20.00	9	28.13	5	20.00	7	31.82
Participant describes being diagnosed by their general practitioner during a routine check-up that was not related to symptoms	4	8.51	3	11.54	1	4.76	4	16.67	0	0.00	1	6.67	3	9.38	3	12.00	1	4.55
Participant describes a complex diagnosis, needing to see multiple specialists before diagnosis	1	2.13	1	3.85	0	0.00	0	0.00	1	4.35	0	0.00	1	3.13	1	4.00	0	0.00
No particular comment	4	8.51	1	3.85	3	14.29	2	8.33	2	8.70	1	6.67	3	9.38	0	0.00	4	18.18

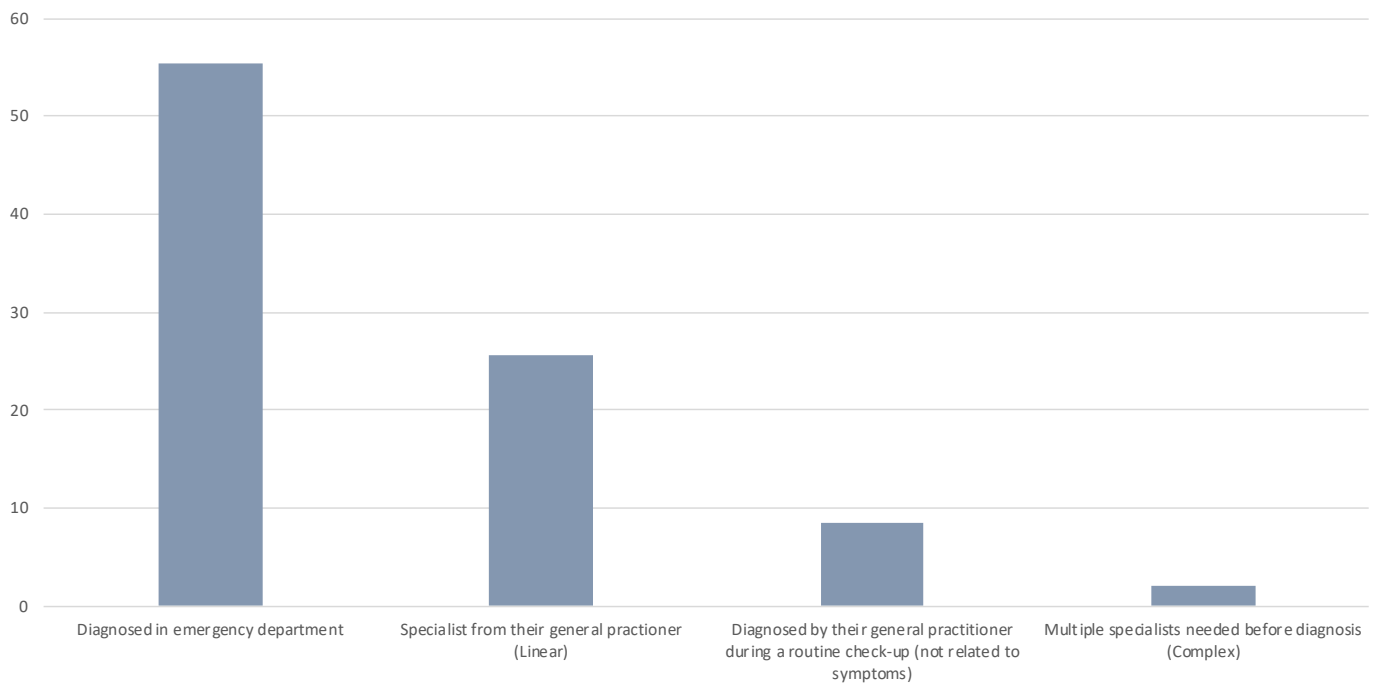


Figure 3.7: Diagnostic pathway

Table 3.9: Diagnostic pathway – subgroup variations

Diagnostic pathway	Reported less frequently	Reported more frequently
Participant describes being diagnosed in an emergency department	Did not had LP(a) test High cholesterol under 50 years of age	Had LP(a) test Regional or remote
Participant describes a linear diagnosis after being referred to a specialist from their general practitioner	High cholesterol under 50 years of age 6 to 11 other conditions	Heart conditions
Participant describes being diagnosed by their general practitioner during a routine check-up that was not related to symptoms	-	High cholesterol under 50 years of age

Timing of diagnosis

Time from diagnostic test to receiving a diagnosis

Participants were asked in the online questionnaire how long they waited between diagnostic tests and getting a diagnosis.

Participants were most commonly diagnosed immediately at the consultation (n = 19, 38.00%). There

were 15 participants (30.00%) that were diagnosed less than one week after diagnostic tests, 9 participants (18.00%) diagnosed between 1 and 2 weeks, 1 participant (2.00%) diagnosed between 2 and 3 weeks, 4 participants (8.00%) diagnosed between 3 and 4 weeks, and 2 participants (4.00%) diagnosed more than four weeks after diagnostic testing.

Table 3.10: Time from diagnostic test to diagnosis

Time from diagnosis test to diagnosis	Number (n=50)	Percent
Diagnosed immediately at the consultation	19	38.00
Less than 1 week	15	30.00
Between 1 and 2 weeks	9	18.00
Between 2 and 3 weeks	1	2.00
4 weeks or more	4	8.00
Not sure	2	4.00

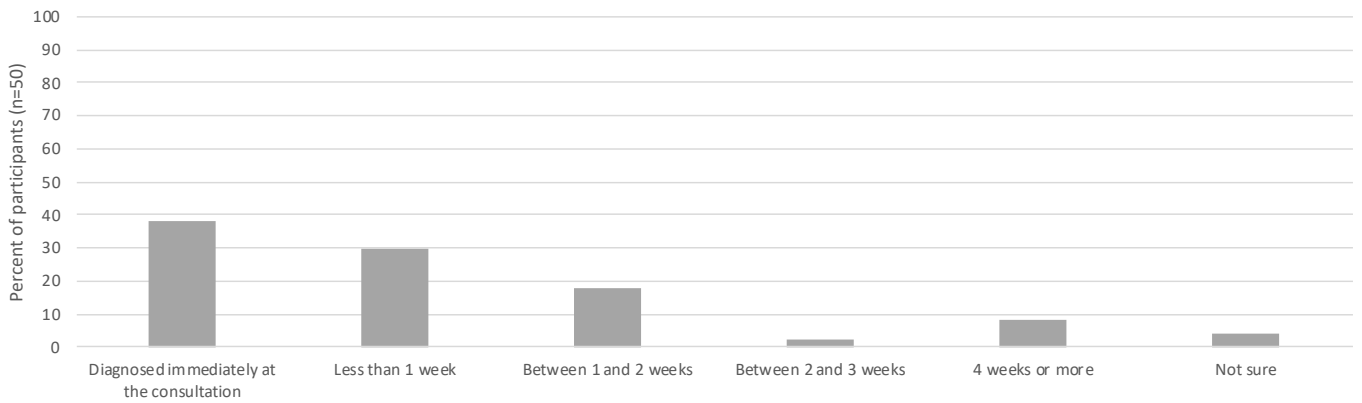


Figure 3.8: Time from diagnostic test to diagnosis

Diagnostic tests

Participants were asked in the questionnaire which diagnostic tests they had for their diagnosis. They could choose from a set list of diagnostic tests and could then specify other tests not listed. The number of tests per participant were counted using both tests from the set list and other tests specified.

Participants reported between 1 to 12 diagnostic tests (median=2.00 , IQR=4.00). The most common tests were blood tests (n=33, 66.00%), electrocardiogram (n=23, 46.00%), Echocardiogram (n=15, 30.00%), and Brain CT or MRI (n=14, 28.00%).

Table 3.11: Number of diagnostic tests

Number of diagnostic tests per participant	Number (n=50)	Percent
1 to 2	27	54.00
3 to 4	6	12.00
5 to 6	8	16.00
7 or more	9	18.00

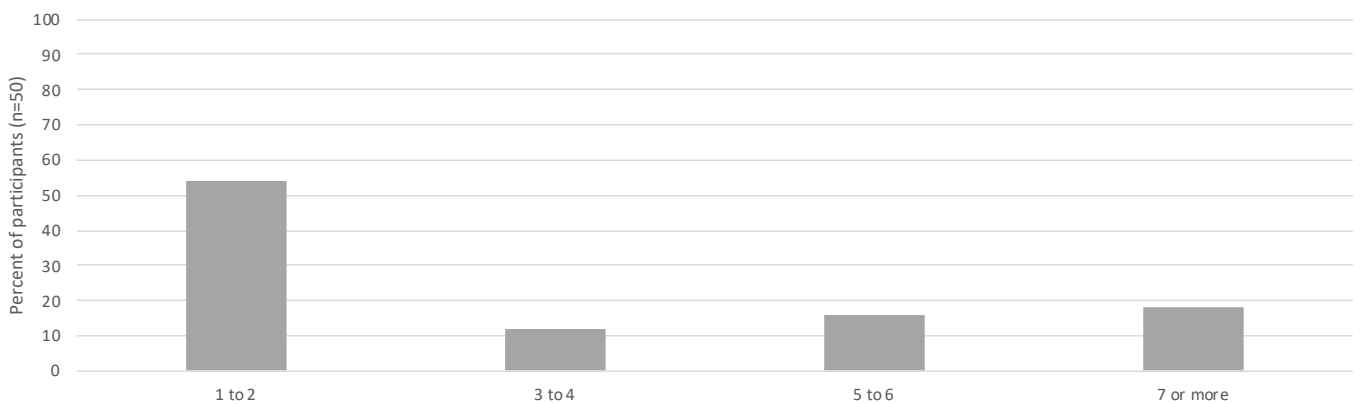


Figure 3.9: Number of diagnostic tests

Table 3.12: Diagnostic tests

Diagnostic tests	Number (n=50)	Percent
Blood test	33	66.00
Electrocardiogram (ECG)	23	46.00
Echocardiogram (Echo) (An ultrasound of your heart)	15	30.00
Brain CT or MRI	14	28.00
Coronary angiogram	10	20.00
Blood pressure monitoring	9	18.00
Physical exam	9	18.00
Assessment for rehabilitation	7	14.00
Communication assessment	7	14.00
Neuropsychological or cognitive assessment	7	14.00
Holter monitor (ECG) (A wearable device measuring your heart activity)	6	12.00
Swallowing tests	6	12.00
Screened for cognitive and perceptual deficits	5	10.00

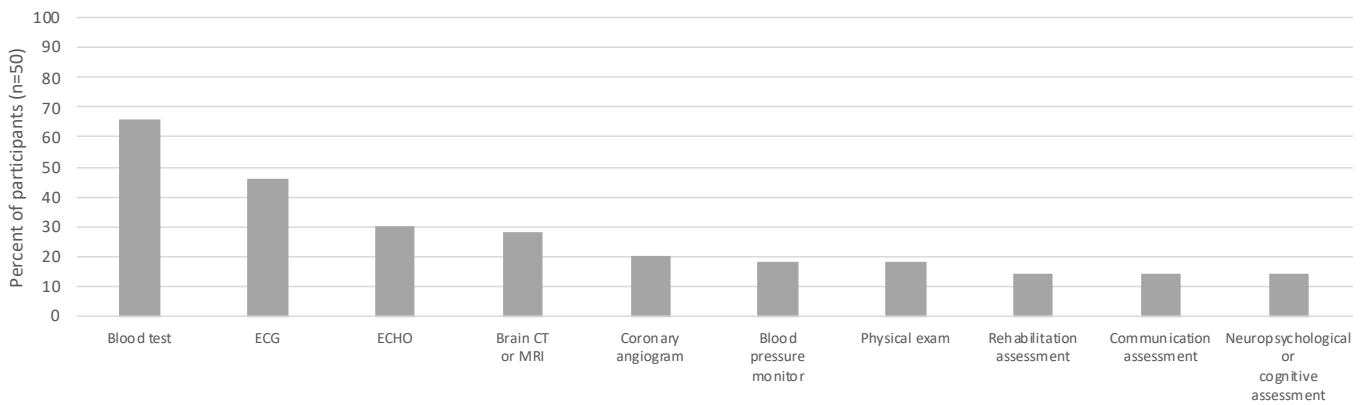


Figure 3.10: Diagnostic tests

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.

Almost half of the participants were given their diagnosis by a Emergency doctor (n=17, 34.00%), and there were 15 participants (30.00%) given the diagnosis by a Cardiologist, 12 participants (24.00%) diagnosed

by General practitioner (GP), and 4 participants (8.00%) by a Neurologist.

Participants were most commonly given their diagnosis in the Hospital (n=31, 63.27%), this was followed by General practice (GP) (n=10, 20.41%), and the Specialist clinic (n=8, 16.33%).

Table 3.13: Diagnosis provider

Health professional gave diagnosis	Number (n=50)	Percent
Emergency doctor	17	34.00
Cardiologist	15	30.00
General practitioner (GP)	12	24.00
Neurologist	4	8.00
Other specialist doctor	2	4.00

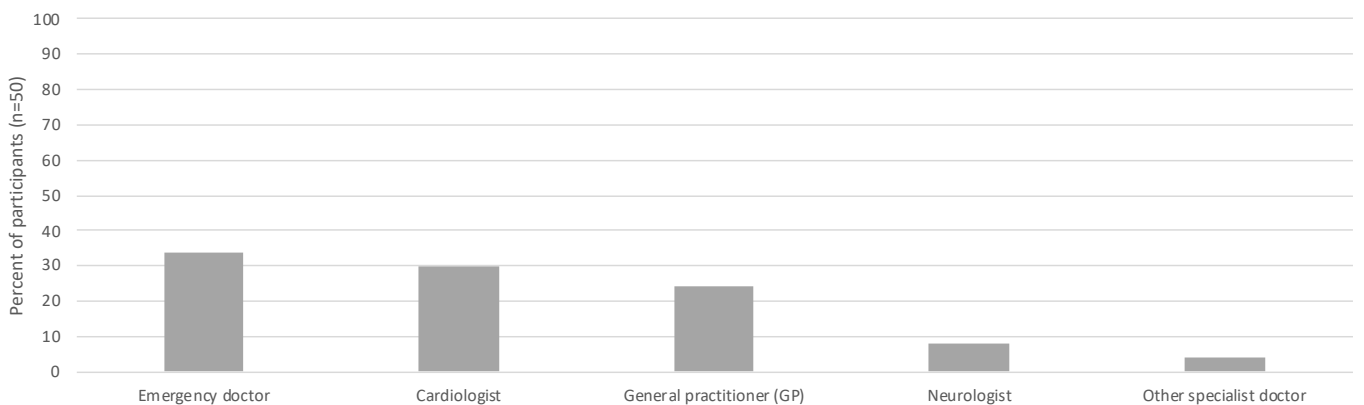


Figure 3.11: Diagnosis provider

Table 3.14: Diagnosis location

Location of diagnosis	Number (n=49)	Percent
General practice (GP)	10	20.41
Specialist clinic	8	16.33
Hospital	31	63.27

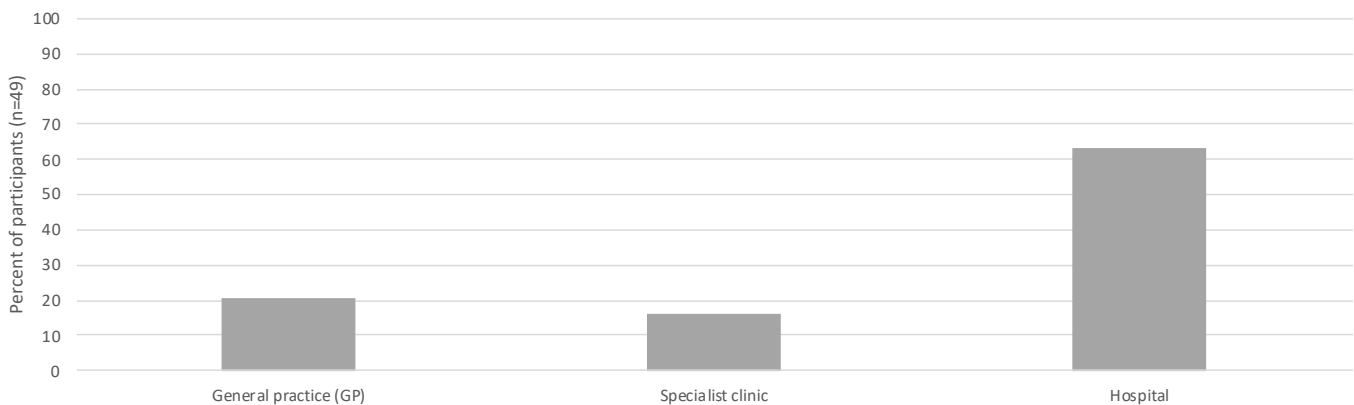


Figure 3.12: Diagnosis location

Year of diagnosis

In the online questionnaire, participants noted the approximate date of diagnosis, the year of diagnosis is presented in the table below.

Participants were diagnosed between 2001 to 2023. There were 27 participants (55.10%) that were diagnosed in the last five years.

Table 3.15 Year of diagnosis

Year of diagnosis	Number (n=49)	Percent
Before 2018	26	53.06
2018 to 2019	8	16.33
2020 to 2023	15	30.61

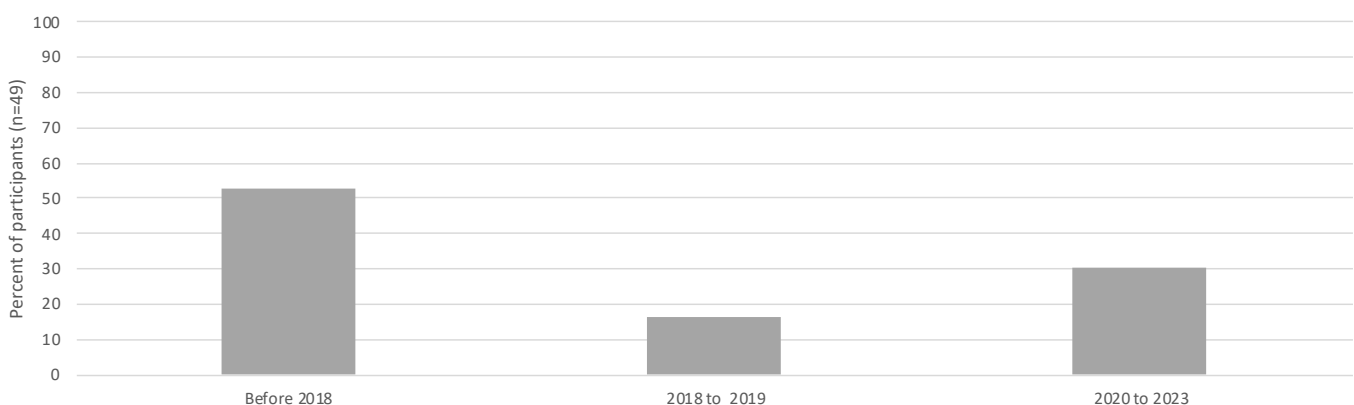


Figure 3.13: Year of diagnosis

Understanding of disease at diagnosis

Participants were asked in the structured interview how much they knew about their condition at diagnosis. The most common responses were knowing nothing or very little about the condition at diagnosis (61.70%) and knowing about the condition at diagnosis because they have a family history of the condition or that they know someone who has the condition (14.89%). Other themes included knowing a good amount about the condition at diagnosis with no reason provided (8.51%), and knowing about the condition due to professional background (6.38%).

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

Absolutely nothing. I was fit, I was going to the gym, I was eating properly and doing all the right things, and I thought I'd be okay, but I have a family history of heart problems, which sort of lingered in the background all the time.

Participant 031_2023AUHBV

Very little. In fact, not very much at all. It wasn't on my radar. I simply considered that stroke was a condition that elderly people experienced. I knew nothing about the technical aspects or the medical aspects.

Participant 045_2023AUHBV

Nothing really, not much. When they told me that I had it, they basically tried to get rid of me out of the ER straight away. I insisted I had no one to go home to because my partner and I weren't in a serious relationship, and I stayed overnight, and then my mum came and got me. She lived in LOCATION REGIONAL so I wouldn't let her drive up that night because she was distraught. I made her wait until the following morning, and then she took me home.

Participant 046_2023AUHBV

Participant describes knowing about the condition at diagnosis because they have a family history of the condition/know someone who has the condition

It it was assumed normal for my family, but not of no concern. So knowing my mom and my sister have high cholesterol, it was just, oh, well, that's because you related. So it was very, it was not an issue.

Participant 003_2023AUHBV

No, no, no. But I knew it was, it's in the family. So that was that was OK. You know, I knew it was in the town.

My, my mother died of a heart attack. Her mother died of a heart attack. Her brother died of a heart attack. My grandparents died of a heart attack. My father's brother died of a heart attack. So like everybody, everybody but my father died of a heart attack. He had a brain tumor. Yeah. Wow. You know it's it's very functional.

Participant 004_2023AUHBV

I knew a fair bit because my eldest brother had a triple bypass. I knew about cardiac artery disease to to a certain extent. And being the age that you are, you sort of know people that are mostly males, I might say. So I've learned a lot on the part, on the, on the, on the way. I didn't know anything about a FI can tell you, you know what I'm saying?

Participant 013_2023AUHBV

Participant describes knowing about the condition due to professional background

I knew a little bit about my I knew a little bit about coronary artery disease. Anyway, it's because I'm a PROFESSION, so it makes a bit of a difference. So I was very proactive in researching and and all that sort of thing. So I do my own literature reviews and things from back in nursing school. So I kind of was on top of it that. Participant 011_2023AUHBV

Table 3.16: Understanding of disease at diagnosis

Understanding of disease at diagnosis	All participants		Female		Male		Aged 25 to 44		Aged 45 and older		Regional or remote		Metropolitan		Mid to low socioeconomic status		Higher socioeconomic status	
	n=47	%	n=26	%	n=21	%	n=24	%	n=23	%	n=15	%	n=32	%	n=25	%	n=22	%
Participant describes knowing nothing or very little about the condition at diagnosis	29	61.70	15	57.69	14	66.67	15	62.50	14	60.87	11	73.33	18	56.25	13	52.00	16	72.73
Participant describes knowing about the condition at diagnosis because they have a family history of the condition/know someone who has the condition	7	14.89	5	19.23	2	9.52	3	12.50	4	17.39	0	0.00	7	21.88	6	24.00	1	4.55
Participant describes knowing a good amount about the condition at diagnosis (no reason provided)	4	8.51	3	11.54	1	4.76	3	12.50	1	4.35	1	6.67	3	9.38	1	4.00	3	13.64
Participant describes knowing about the condition due to professional background	3	6.38	2	7.69	1	4.76	1	4.17	2	8.70	2	13.33	1	3.13	3	12.00	0	0.00

Understanding of disease at diagnosis	All participants		Had LP(a) test		Did not had LP(a) test		High cholesterol under 50 years of age		Blood vessel conditions		Heart conditions		0 to 5 other conditions		6 to 11 other conditions	
	n=47	%	n=18	%	n=29	%	n=9	%	n=17	%	n=21	%	n=27	%	n=20	%
Participant describes knowing nothing or very little about the condition at diagnosis	29	61.70	11	61.11	18	62.07	4	44.44	11	64.71	14	66.67	20	74.07	9	45.00
Participant describes knowing about the condition at diagnosis because they have a family history of the condition/know someone who has the condition	7	14.89	3	16.67	4	13.79	2	22.22	3	17.65	2	9.52	3	11.11	4	20.00
Participant describes knowing a good amount about the condition at diagnosis (no reason provided)	4	8.51	3	16.67	1	3.45	2	22.22	0	0.00	2	9.52	1	3.70	3	15.00
Participant describes knowing about the condition due to professional background	3	6.38	0	0.00	3	10.34	1	11.11	2	11.76	0	0.00	2	7.41	1	5.00

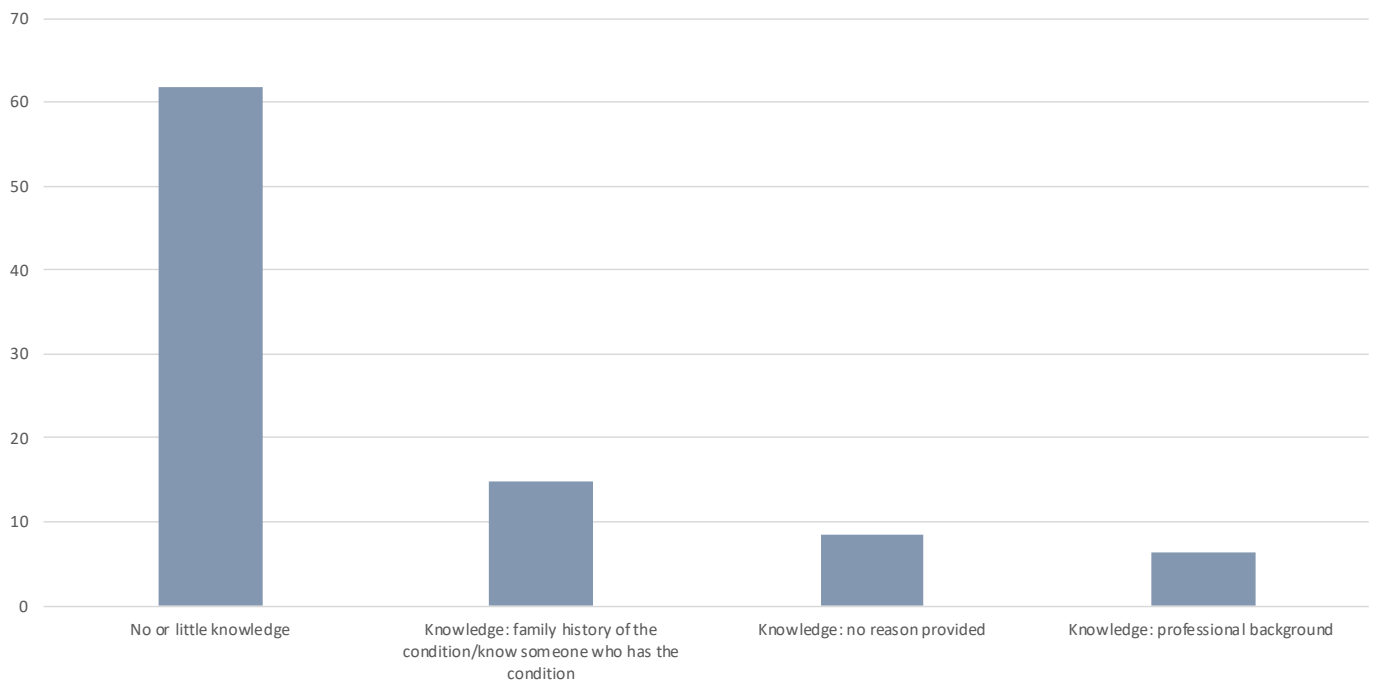


Figure 3.14 Understanding of disease at diagnosis

Table 3.17: Understanding of disease at diagnosis – subgroup variations

Understanding of disease at diagnosis	Reported less frequently	Reported more frequently
Participant describes knowing nothing or very little about the condition at diagnosis	High cholesterol under 50 years of age 6 to 11 other conditions	0 to 5 other conditions Regional or remote Higher socioeconomic status
Participant describes knowing about the condition at diagnosis because they have a family history of the condition/know someone who has the condition	Regional or remote Higher socioeconomic status	-
Participant describes knowing a good amount about the condition at diagnosis (no reason provided)	-	High cholesterol under 50 years of age

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 19 participants (38.00%) who had enough support, 4 participants (8.00%) that had some support but it wasn't enough, and 27 participants (54.00%) had no support.

Table 3.18: Emotional support at diagnosis

Emotional support at diagnosis	Number (n=50)	Percent
Enough support	19	38.00
Some support but it wasn't enough	4	8.00
No support	27	54.00



Figure 3.15: Emotional support at diagnosis

Information at diagnosis

Participants were asked in the online questionnaire how much information they or their family received at diagnosis.

There were 15 participants (35.71%) who had enough information, 19 participants (45.24%) that had Some information but it wasn't enough, and 8 participants (19.05%) had no information.

Table 3.19: Information at diagnosis

Information at diagnosis	Number (n=50)	Percent
Enough information	15	35.71
Some information but it wasn't enough	19	45.24
No information	8	19.05

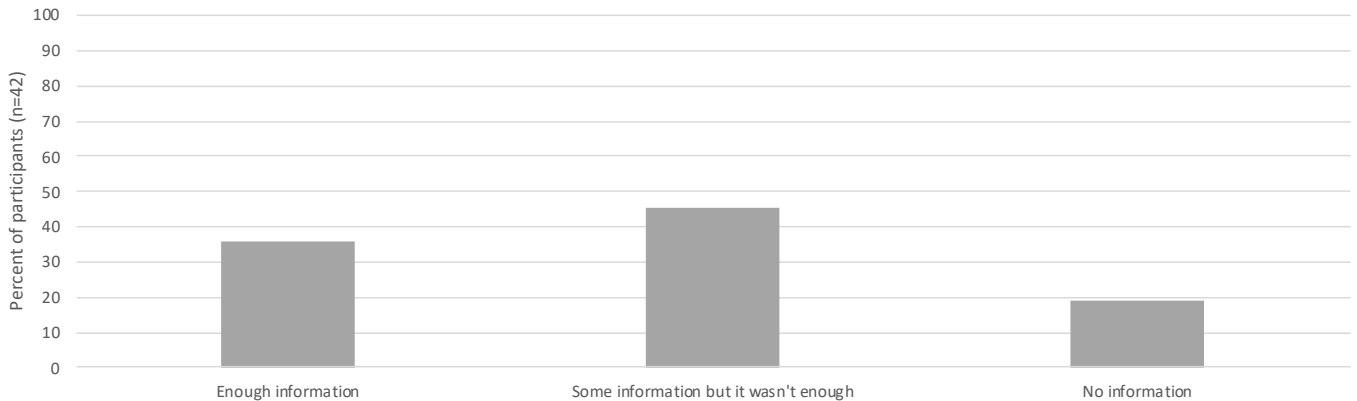


Figure 3.16: Information at diagnosis

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 21 participants (42.00%) who had no out of pocket expenses, and 18 participants (36.00%) who did not know or could not recall. There were 4 participants (8.00%) that spent \$1 to \$250, 3 participants (6.00%)

that spent between \$251 to \$500, and 4 participants (8.00%) that spent \$501 or more.

Burden of diagnostic costs

For 23 participants (67.65%) the cost was slightly or not at all significant. For 7 participants (20.59%) the out-of-pocket expenses were somewhat significant, and for 4 participants (11.76%), the burden of out-of-pocket expenses were moderately or extremely significant.

Table 3.20: Out of pocket expenses at diagnosis

Out of pocket expenses for diagnostic tests	Number (n=50)	Percent
\$0	21	42.00
\$1 to \$250	4	8.00
\$251 to \$500	3	6.00
\$501 or more	4	8.00
Not sure	18	36.00

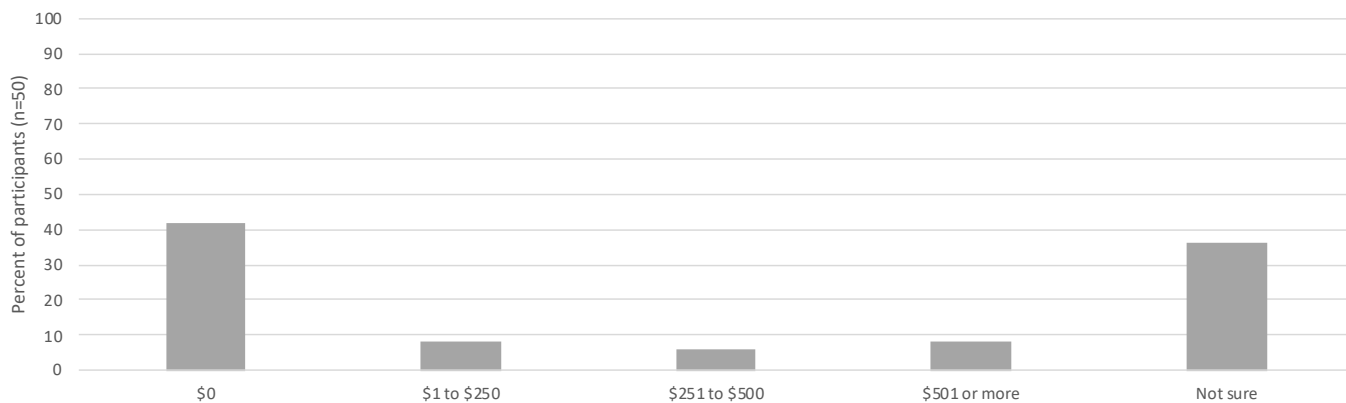


Figure 3.17: Out of pocket expenses at diagnosis

Table 3.21: Burden of diagnostic costs

Burden of diagnostic costs	Number (n=34)	Percent
Not at all significant	14	41.18
Slightly significant	9	26.47
Somewhat significant	7	20.59
Moderately significant	2	5.88
Extremely significant	2	5.88

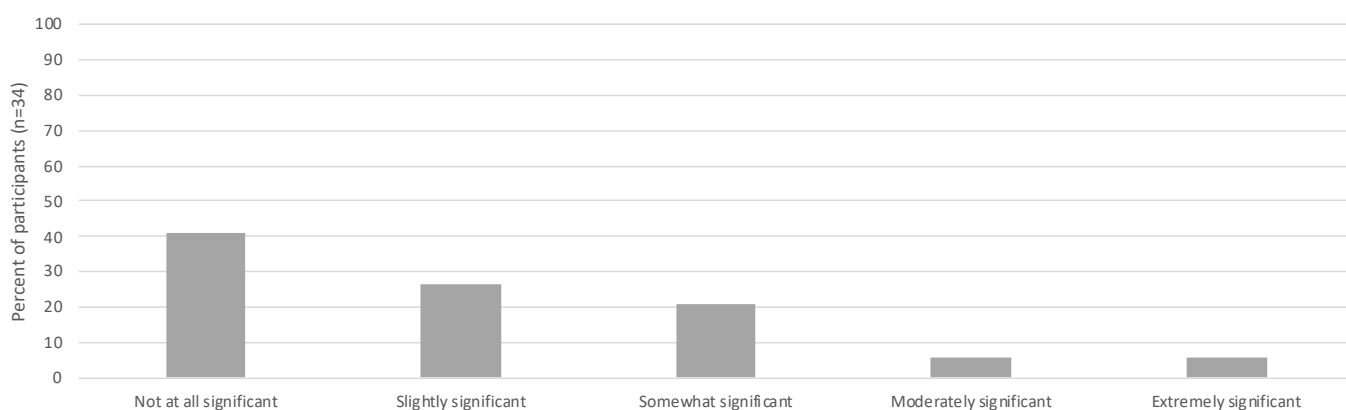


Figure 3.18: Burden of diagnostic costs

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.

Despite 19 participant having confirmed their LPA status, participants most commonly reported that they had never had a conversation about biomarkers, genomic, or gene testing that might be relevant to treatment, (n=43, 86.00%). There were 4 participants (8.00%) who brought up the topic with their doctor, and 3 participants (6.00%) 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.

The majority of participants did not have any genetic or biomarker tests but would like to (n=38, 76.00%). There were 10 participants (20.00%) who did not have these tests and were not interested in them, and a total of 2 participants (4.00%) that had biomarker tests.

Table 3.22: Discussions about biomarkers

Discussions about biomarkers	Number (n=50)	Percent
Participant brought up the topic with doctor for discussion	4	8.00
Doctor brought up the topic with participant for discussion	3	6.00
Participant had no discussion about this type of test	43	86.00

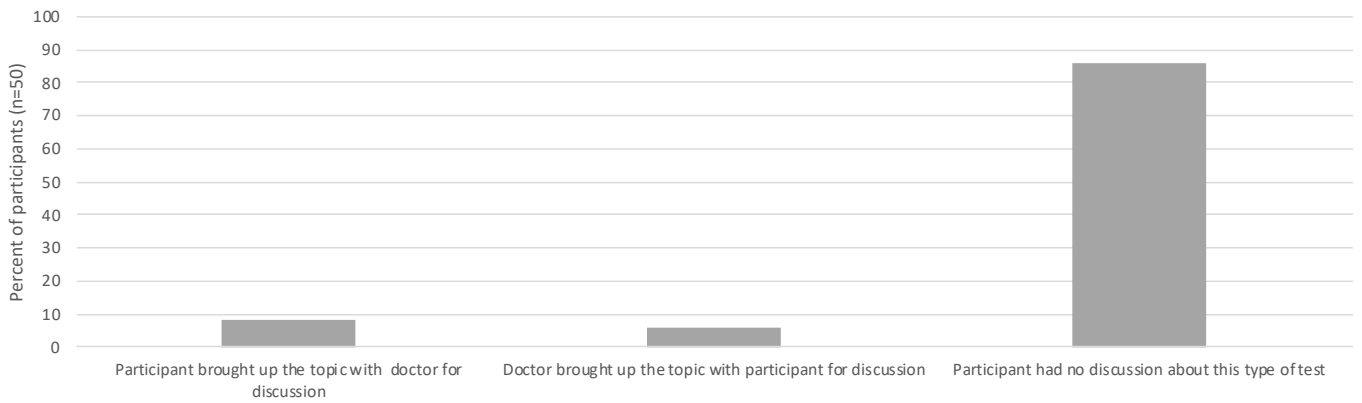


Figure 3.19: Discussions about biomarkers

Table 3.23: Experience of genetic tests and biomarkers

Experience of genetic tests and biomarkers	Number (n=50)	Percent
Participant had this test and did not have to pay out of pocket for it	2	4.00
Participant had this test through a clinical trial	0	0.00
Participant had this type of test and paid for it	0	0.00
Participant did not have this test and is not interested in it	10	20.00
Participant did not have this test but would like to	38	76.00

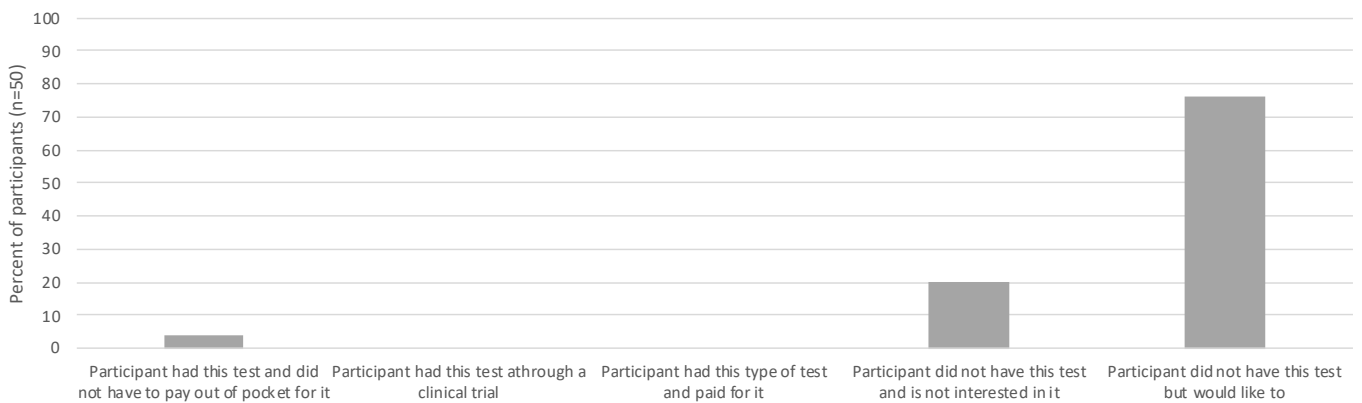


Figure 3.20: Experience of genetic tests and biomarkers

Biomarker status

Most commonly, participants had a family history of heart or blood vessel condition (n=25, 56.82%), followed by Lipoprotein A (LPa) status (n=19, 43.18%).

There were 7 participants that were and not sure (15.91%), and 2 participants that had no markers (4.55%).

Table 3.24: Biomarker status

Biomarkers	Number (n=44)	Percent
Family history	25	56.82
Lipoprotein A (LPa)	19	43.18
Not sure	7	15.91
I do not have any markers	2	4.55

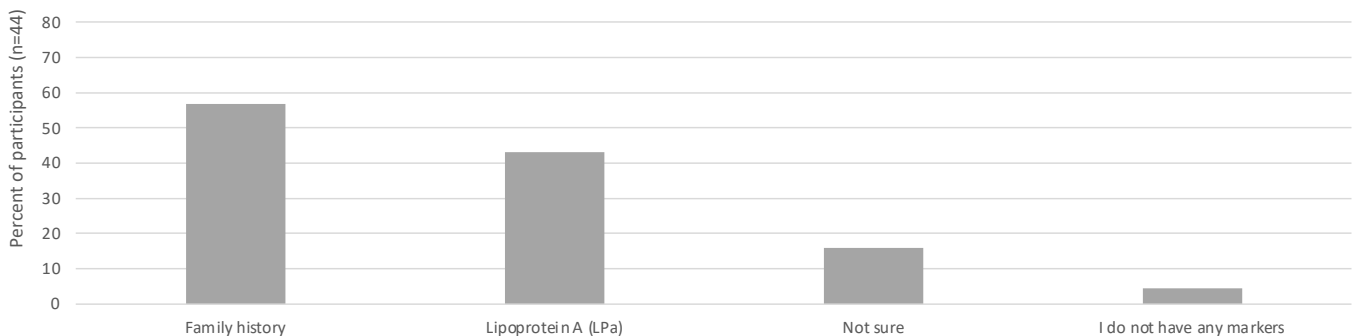


Figure 3.21: Biomarker status

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 they had specific medical interventions they need to manage their condition (31.91%), that they were monitoring their condition until there is an exacerbation or progression (23.40%), and that their prognosis was positive, that their condition is manageable (21.28 %). Other themes included that there was uncertainty around prognosis (19.15%), that it was a lifelong condition (14.89%), that they need to maintain a healthy lifestyle (12.77%), and that they would likely have a recurrence, or were in a cycle of recurrence (8.51%).

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

At the moment, it's looking okay. A year ago, I would not have said that, and neither would my cardiologist. It actually took about six months to get the heart rate and down and to get things under control. At the moment, I'm very happy with where I am. I'm back at work. I'm going to the gym a couple times a week, and so I feel okay mostly. I don't do very well in hot weather. As far as my prognosis is concerned, I know that I have to have a valve replacement in the near future, so when that is, it will depend on the ejection fraction. Participant 034_2023AUHBV

My outlook on the future is just living with my condition and continuing taking my medication to thin my blood. Just monitor my warfarin. Participant 049_2023AUHBV

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

Ohh yeah, yeah. Excuse me. Yeah. I mean as it's been monitored and you know every year since I, I, I usually I get about an annual reading at least at least once a year And so that always becomes the talking point in discussion. So the GP counseling is, you know they're taking on, on board a few other factors. I mean I'm now 44 years old. So they've kind of hinted, well, they've suggested and hinted that hey, you know, there's a point at which we would like you're pretty basically they're saying that you're pretty close to a point where we, we really recommend you know, a treatment, you know, some of the statins what have you to to lower your cholesterol, some kind of like in

that pretty close area. And so, yeah, I guess if it keeps repeating itself, they'll keep the volume up on that conversation as far as a pharmacological treatment. Participant 001_2023AUHBV

My current prognosis is that they really don't know what my prognosis is. I have yearly check-ups, and each yearly check-up there is more thickening in my heart. The prognosis is anywhere from it will stay the same as this for the rest of my life, to it could continue to thicken up and I may eventually need a heart transplant. And, anywhere in between those two. It's a very unpredictable disease what I have. They can't predict what's going to happen. They just have to watch and react to what does happen. Participant 036_2023AUHBV

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

I'm very stable. I'm a little bit boring. My prognosis, I have a great outlook in life. I don't see any issues going forward. Participant 039_2023AUHBV

I have been extremely lucky that I've managed to basically, I can do pretty much everything I used to do. The only thing I can't do is drive at night. I've behaviourally accommodated it to the point where I passed a special driving test to be allowed to drive during daylight hours. That's where I'm at physically. I don't believe that I'm particularly at risk of another stroke. Participant 046_2023AUHBV

Participant describes prognosis in relation to uncertainty around prognosis

They won't give me an answer. No one will give me an answer. When I had first went to LOCATION METROPOLITAN they wouldn't even give me six months. When I had my appointment on the 5th of October it was, it was like they both came in. It was like, "Well, I'm still here." Participant 033_2023AUHBV

Well NAME DOCTOR has never said and I've never asked him. He's never said what my prognosis is. Participant 037_2023AUHBV

Nothing. The doctors don't tell me a thing. My cardiologist, I believe, is a waste of \$500 every six months. I go in there, he tells me I'm too fat, which is

fair enough, and you get no information. No like, what's our next procedure, do an operation, or do we -- I know it's not a transplant, but how are we going to treat my heart? "We're going to give you this. We're going to give you that." It's nothing. It's usually a case of, "Oh, okay, well just keep watching what you eat. Lose some more weight. I'll see you in six months for another 500 bucks."

Participant 038_2023AUHBV

Participant describes prognosis in relation to it being a lifelong condition

Yeah. So, so I'm on, I'm a patient, I'm a heart patient. I'll, I'll always be a heart patient, I suppose somewhat. But as far as my condition goes, you know, I've had a CONDITION and there's a 20% chance I'll have another CONDITION. So I follow a pretty conservative heart program to try to avoid that from happening. And as far as my prognosis goes, really there's no, there's no expectation that that will cause me, you know, future issues, all the likes as long as I manage, you know, within within the guidelines I've been given, I suppose.

Participant 009_2023AUHBV

I don't know. I'm back to doing things now. I'm working again, but my outlook is pretty good. It's always been pretty good, to be honest, but I still suffer. I had a pretty ordinary day today, so yes. It's frustration is what my outlook is. Just continued frustration. I think now that I've accepted that this is how I have to be for the rest of my life.

Participant 043_2023AUHBV

I am two years passed stroke and I am able to work part-time. I have had a hole in my heart fixed that they found post-stroke. I am on preventive medication for life and I don't really see my situation improving at all from where it is now.

Participant 047_2023AUHBV

Participant describes prognosis in relation to maintaining a healthy lifestyle, including diet, exercise, reducing or quitting alcohol or smoking

It's very good. I keep my weight down, exercise, and stay on my medication.

Participant 005_2023AUHBV

My current outlook is from when I suffered. The condition is that I turned my I had to turn my whole lifestyle around. Because basically heart attacks were for older people and not for me. And that was my attitude. I was one of those disgusting smokers at the time because that all came about by the era that I grew up in. Drank alcohol, you know what I'm saying? Done nothing different to. Belly full, on, full felt lifestyle to I gave up smoking immediately. I didn't need any counseling. I didn't need to go and suck on a big bike in the end of the corridor. No, I just realized if I wanted to spend a lot of time with my children, well, I changed lifestyle straight away. And that lifestyle involved dietitian. Yeah, eating properly. Talking, buying a lot of stuff for myself, cooking properly.

Participant 006_2023AUHBV

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

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 008_2023AUHBV

Yes, I saw the neurosurgeon today and he said that I could have another one. They don't know why I had it. I've got no risk factors. They can't see on the MRI why I had a stroke. There's no haemorrhage, there's no aneurism and so they don't know what caused why?

Participant 041_2023AUHBV

Table 3.25: Understanding of prognosis

Understanding of prognosis	All participants		Female		Male		Aged 25 to 44		Aged 45 and older		Regional or remote		Metropolitan		Mid to low socioeconomic status		Higher socioeconomic status	
	n=47	%	n=26	%	n=21	%	n=24	%	n=23	%	n=15	%	n=32	%	n=25	%	n=22	%
Participant describes prognosis in relation to specific medical interventions they need to manage their condition	15	31.91	8	30.77	7	33.33	7	29.17	8	34.78	6	40.00	9	28.13	8	32.00	7	31.82
Participant describes prognosis in relation to monitoring their condition until there is an exacerbation or progression	11	23.40	6	23.08	5	23.81	8	33.33	3	13.04	6	40.00	5	15.63	8	32.00	3	13.64
Participant describes prognosis in a positive way, that their condition is manageable	10	21.28	6	23.08	4	19.05	4	16.67	6	26.09	3	20.00	7	21.88	7	28.00	3	13.64
Participant describes prognosis in relation to uncertainty around prognosis	9	19.15	6	23.08	3	14.29	6	25.00	3	13.04	2	13.33	7	21.88	3	12.00	6	27.27
Participant describes prognosis in relation to it being a lifelong condition	7	14.89	2	7.69	5	23.81	1	4.17	6	26.09	2	13.33	5	15.63	3	12.00	4	18.18
Participant describes prognosis in relation to maintaining a healthy lifestyle, including diet, exercise, reducing or quitting alcohol or smoking	6	12.77	3	11.54	3	14.29	4	16.67	2	8.70	3	20.00	3	9.38	4	16.00	2	9.09
Participant describes prognosis in relation to probable recurrence, or cycle of recurrence	4	8.51	2	7.69	2	9.52	2	8.33	2	8.70	0	0.00	4	12.50	1	4.00	3	13.64
No particular comment	4	8.51	2	7.69	2	9.52	2	8.33	2	8.70	1	6.67	3	9.38	3	12.00	1	4.55

Understanding of prognosis	All participants		Had LP(a) test		Did not had LP(a) test		High cholesterol under 50 years of age		Blood vessel conditions		Heart conditions		0 to 5 other conditions		6 to 11 other conditions	
	n=47	%	n=18	%	n=29	%	n=9	%	n=17	%	n=21	%	n=27	%	n=20	%
Participant describes prognosis in relation to specific medical interventions they need to manage their condition	15	31.91	6	33.33	9	31.03	3	33.33	3	17.65	9	42.86	8	29.63	7	35.00
Participant describes prognosis in relation to monitoring their condition until there is an exacerbation or progression	11	23.40	4	22.22	7	24.14	4	44.44	2	11.76	5	23.81	6	22.22	5	25.00
Participant describes prognosis in a positive way, that their condition is manageable	10	21.28	3	16.67	7	24.14	0	0.00	4	23.53	6	28.57	6	22.22	4	20.00
Participant describes prognosis in relation to uncertainty around prognosis	9	19.15	3	16.67	6	20.69	2	22.22	3	17.65	4	19.05	5	18.52	4	20.00
Participant describes prognosis in relation to it being a lifelong condition	7	14.89	3	16.67	4	13.79	0	0.00	6	35.29	1	4.76	5	18.52	2	10.00
Participant describes prognosis in relation to maintaining a healthy lifestyle, including diet, exercise, reducing or quitting alcohol or smoking	6	12.77	2	11.11	4	13.79	2	22.22	1	5.88	3	14.29	3	11.11	3	15.00
Participant describes prognosis in relation to probable recurrence, or cycle of recurrence	4	8.51	1	5.56	3	10.34	0	0.00	1	5.88	3	14.29	2	7.41	2	10.00
No particular comment	4	8.51	1	5.56	3	10.34	1	11.11	2	11.76	1	4.76	2	7.41	2	10.00

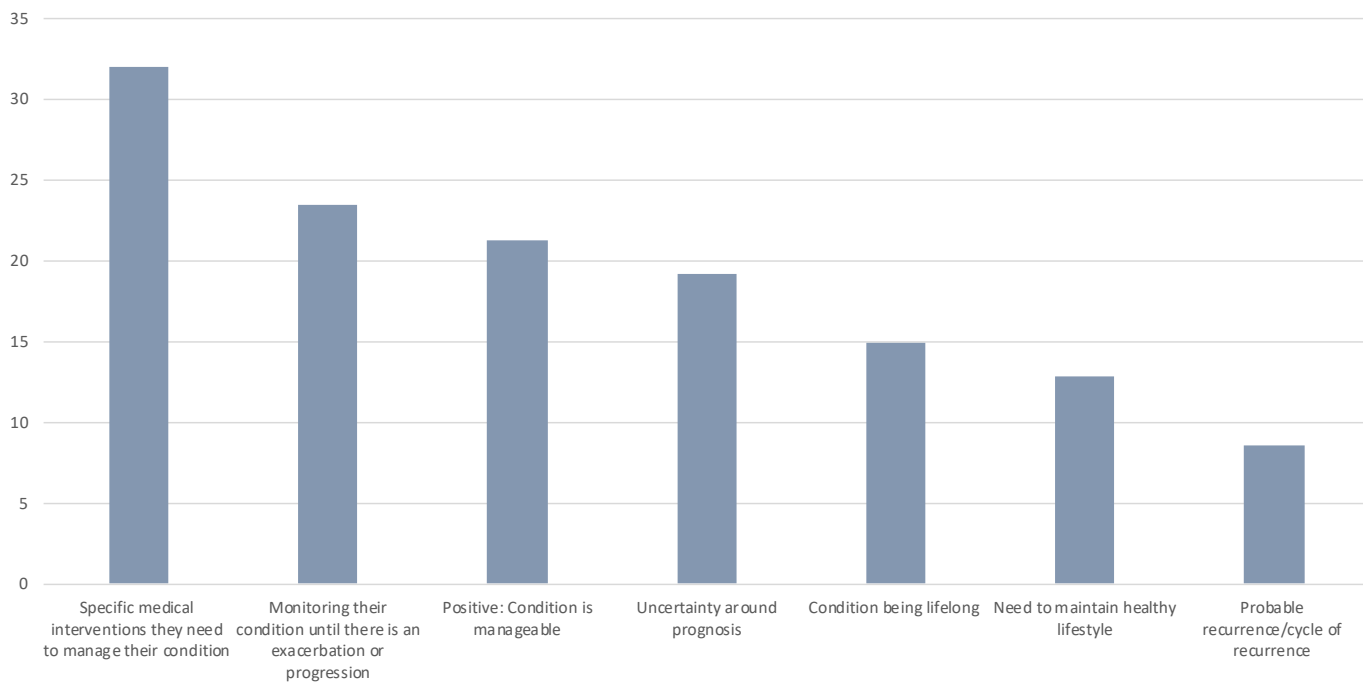


Figure 3.22: Understanding of prognosis

Table 3.26: Understanding of prognosis – subgroup variations

Understanding of prognosis	Reported less frequently	Reported more frequently
Participant describes prognosis in relation to specific medical interventions they need to manage their condition	Blood vessel conditions	Heart conditions
Participant describes prognosis in relation to monitoring their condition until there is an exacerbation or progression	Blood vessel conditions Aged 45 and older	High cholesterol under 50 years of age Regional or remote
Participant describes prognosis in a positive way, that their condition is manageable	High cholesterol under 50 years of age	-
Participant describes prognosis in relation to it being a lifelong condition	High cholesterol under 50 years of age Heart conditions Aged 25 to 44	Blood vessel conditions Aged 45 and older

Biomarker tests

Participants were asked in the structured interview if they had any discussion about biomarkers that may be important to the management of their condition. The most common responses were that they did not have any tests and did not describe reasons (48.94%), that they did not have a test but would like to have this type of test (21.28%). This was followed by no test but family history was discussed (12.77 %), and had a test and management of condition was not changed (6.38%).

Participant describes that they did not have a biomarker test, did not describe reasons

No, they they haven't done that...and no one else in my family has ever had. I haven't gone for genetic testing for that only because I suppose that this seems to be quite targeted to an event in my life as opposed to genetics.

Participant 014_2023AUHBV

Participant describes that they did not have a biomarker test, however would like to have this type of test

I don't know, I'm not speculating a bit, but yeah, they're probably just assuming that that's, I don't know, it's it's almost like, yeah, a lot of the population is affected with higher cholesterol. It's almost like one of these lifestyle things. But yeah, that's interesting you bring it up cuz I've definitely heard of hypercholesterol or the familial, yeah, there is like a familial link for some. There's like a portion of people percentage, yeah.

Participant 001_2023AUHBV

No, not at all. Never. And that's something I actually followed up myself to some extent, being a PROFESSION IN HEALTH and being in the role roles I've been in, I actually found out that there's only one genetic testing service in STATE, and so I rang them and I spoke to the registrar and said, is there any value in being tested? And basically he said no, you're diagnosed, you're treated, you don't have children, your family's got no symptoms. Because we went through all this. No, there's no, no point. And you know, it's very, very scarce resource. Don't waste it on someone who doesn't need it, which, you know, it made perfect sense to me. I don't want to be doing that. So yeah, no, nobody ever, ever said to me. Let's look at genetics, yeah.

Participant 010_2023AUHBV

No one's ever talked to me about any gene stuff, even though I've known that my father died from a heart attack and you know, at 50, something. And that, you know, all my siblings have heart conditions of 1 sort or another, but no one never talked about doing any sort of gene analysis or anything.

Participant 013_2023AUHBV

Participant describes that they did not have a biomarker test, however family history was discussed

Well they did ask me if my family had heart problems cuz it's to my dad's side. He also has the heart memory. He has heart publications, well all the time, but it was caused from drugs and alcohol on his behalf.

Participant 007_2023AUHBV

They did ask me a few questions and did the heart like heart conditions do, running my family through my pop. He has heart problems. Well, he did, and then he. Yeah. So I mentioned that to them and they did talk to me about it much, but I don't think we went any further with it just yet. They were very shocked by my age because I'm only 21. They were just very more shocked on what happened and where the like how did this come so early? Obviously like with no clogged arteries or anything like that, they just said it was very odd. So that they yeah, they just wanted me to come back for recent like for checkups every now and then to just. Keep on top of it, and obviously the more tests will be there.

Participant 029_2023AUHBV

No, that wouldn't be a bad idea though.

Participant 037_2023AUHBV

I should definitely have them because every woman in my family has had a stroke. I've got two girls so I think it would be really important for them to have that.

Participant 039_2023AUHBV

Participant describes that they did not have a biomarker test, however was motivated to research family history

Look, hindsight is a beautiful thing so in the heat of the moment, no, they didn't. It was all about just surviving really. Now that I am older and I do a lot of research and I'm a consumer consultant on other trials, et cetera, I know how important it is to have all that stuff done. I've done my own family tree, I've been able to see if anyone else died of a stroke or had a stroke or any of that. That doesn't seem to be relevant to my case.

Participant 050_2023AUHBV

Participant describes having had a biomarker test, and that management of condition was not changed

That's all I was being told. I've never been told anything else. The cause of my stroke, who knows? I don't know. I don't know why I had my stroke. I don't know anything. I've been just left in the never-nevers.

Participant 049_2023AUHBV

Participant describes having had a biomarker test, and as a result management of condition was changed

Yeah, that's right. He changed the way. So he started me on a statin...he actually went a lot more aggressive with my with his approach to me. So he increased my statin dose and I'm also on ezetimibe as well for that, yeah.

Participant 011_2023AUHBV

Participant describes having had a biomarker test, and as a result surveillance of condition was increased

I've had genetic testing for hypertrophic cardiomyopathy, and this is a progression of that...the heart failure was a progression of an existing diagnosis

Participant 032_2023AUHBV

Participant describes having had a biomarker test, to identify other family members at risk

What I understand is that it doesn't help me, but it helps identify my faulty gene, and it is more of a benefit to my direct family members to eliminate them from unnecessary screening for the disease.

Participant 036_2023AUHBV

Table 3.27: Biomarker tests

Biomarker tests	All participants		Female		Male		Aged 25 to 44		Aged 45 and older		Regional or remote		Metropolitan		Mid to low socioeconomic status		Higher socioeconomic status	
	n=47	%	n=26	%	n=21	%	n=24	%	n=23	%	n=15	%	n=32	%	n=25	%	n=22	%
Participant describes that they did not have a biomarker test, did not describe reasons	23	48.94	11	42.31	12	57.14	14	58.33	9	39.13	7	46.67	16	50.00	12	48.00	11	50.00
Participant describes that they did not have a biomarker test, however would like to have this type of test	10	21.28	5	19.23	5	23.81	4	16.67	6	26.09	1	6.67	9	28.13	7	28.00	3	13.64
Participant describes that they did not have a biomarker test, however family history was discussed	6	12.77	5	19.23	1	4.76	5	20.83	1	4.35	4	26.67	2	6.25	3	12.00	3	13.64
Participant describes that they did not have a biomarker test, however was motivated to research family history	1	2.13	1	3.85	0	0.00	0	0.00	1	4.35	0	0.00	1	3.13	1	4.00	0	0.00
Participant describes that they did not have a biomarker test, however BMI was discussed	1	2.13	1	3.85	0	0.00	1	4.17	0	0.00	1	6.67	0	0.00	1	4.00	0	0.00
Participant describes having had a biomarker test, and that management of condition was not changed	3	6.38	0	0.00	3	14.29	0	0.00	3	13.04	1	6.67	2	6.25	0	0.00	3	13.64
Participant describes having had a biomarker test, and as a result management of condition was changed	1	2.13	1	3.85	0	0.00	0	0.00	1	4.35	1	6.67	0	0.00	1	4.00	0	0.00
Participant describes having had a biomarker test, and as a result surveillance of condition was increased	1	2.13	1	3.85	0	0.00	0	0.00	1	4.35	0	0.00	1	3.13	0	0.00	1	4.55
Participant describes having had a biomarker test, to identify other family members at risk	1	2.13	1	3.85	0	0.00	0	0.00	1	4.35	0	0.00	1	3.13	0	0.00	1	4.55

Biomarker tests	All participants		Had LP(a) test		Did not had LP(a) test		High cholesterol under 50 years of age		Blood vessel conditions		Heart conditions		0 to 5 other conditions		6 to 11 other conditions	
	n=47	%	n=18	%	n=29	%	n=9	%	n=17	%	n=21	%	n=27	%	n=20	%
Participant describes that they did not have a biomarker test, did not describe reasons	23	48.94	8	44.44	15	51.72	2	22.22	11	64.71	10	47.62	14	51.85	9	45.00
Participant describes that they did not have a biomarker test, however would like to have this type of test	10	21.28	5	27.78	5	17.24	2	22.22	2	11.76	6	28.57	7	25.93	3	15.00
Participant describes that they did not have a biomarker test, however family history was discussed	6	12.77	4	22.22	2	6.90	3	33.33	0	0.00	3	14.29	3	11.11	3	15.00
Participant describes that they did not have a biomarker test, however was motivated to research family history	1	2.13	0	0.00	1	3.45	0	0.00	1	5.88	0	0.00	0	0.00	1	5.00
Participant describes that they did not have a biomarker test, however BMI was discussed	1	2.13	0	0.00	1	3.45	1	11.11	0	0.00	0	0.00	0	0.00	1	5.00
Participant describes having had a biomarker test, and that management of condition was not changed	3	6.38	1	5.56	2	6.90	1	11.11	2	11.76	0	0.00	2	7.41	1	5.00
Participant describes having had a biomarker test, and as a result management of condition was changed	1	2.13	0	0.00	1	3.45	0	0.00	1	5.88	0	0.00	0	0.00	1	5.00
Participant describes having had a biomarker test, and as a result surveillance of condition was increased	1	2.13	0	0.00	1	3.45	0	0.00	0	0.00	1	4.76	1	3.70	0	0.00
Participant describes having had a biomarker test, to identify other family members at risk	1	2.13	0	0.00	1	3.45	0	0.00	0	0.00	1	4.76	0	0.00	1	5.00

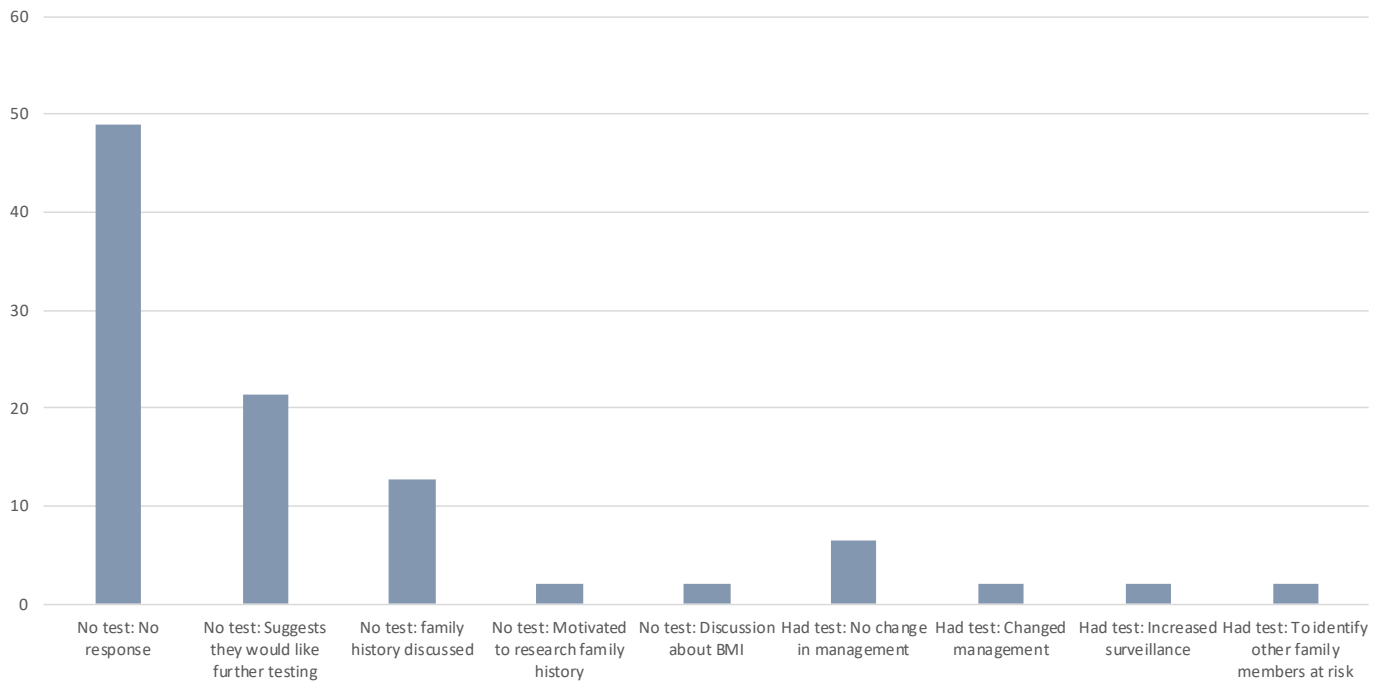


Figure 3.23: Biomarker tests

Table 3.28: Biomarker tests – subgroup variations

Biomarker tests	Reported less frequently	Reported more frequently
Participant describes that they did not have a biomarker test, did not describe reasons	High cholesterol under 50 years of age	Blood vessel conditions
Participant describes that they did not have a biomarker test, however would like to have this type of test	Regional or remote	-
Participant describes that they did not have a biomarker test, however family history was discussed	Blood vessel conditions	High cholesterol under 50 years of age Regional or remote