Summary of results

Executive summary

There were 36 participants in the study from across Australia, 28 diagnosed with amyloidosis, and eight carers to people with amyloidosis. The majority of participants were from Queensland and New South Wales, and most lived in major cities, they lived in all levels of advantage. Most of the of participants identified as Caucasian or white, aged mostly between 65 and 74. Half of the participants had completed some university, and most were retired.

Participants in this PEEK study were most commonly diagnosed with ATTR, either hereditary or wild type. Most of the participants also had other health conditions they had to manage, approximately 44% of the participants had anxiety and/or depression.

This is a patient population that experienced fatigue as the most common symptom leading to diagnosis. They most commonly had five or six diagnostic tests to get their diagnosis, and were diagnosed more than a year after first noticing symptoms. They had out of pocket expenses for their diagnosis, but usually the cost wasn't a significant burden. Most participants felt they had enough emotional support and information from healthcare professionals at the time of diagnosis.

This is a patient population that experienced excessive weight loss, breathlessness and tiredness as key symptoms leading to their diagnosis. Half of the participants described seeking medical attention relatively soon after they started experiencing symptoms.

This is a study cohort that described knowing nothing or very little about their condition prior to diagnosis.

This is a patient population that had conversations about treatment where multiple options were presented. They mostly took quality of life, efficacy of treatment, and side effects into consideration when making treatment decisions, their decision making had not changed over time. They commonly did not have many discussions about biomarkers and were not sure if they had any.

This is a group who felt they were treated with respect throughout their experience. They were most commonly treated for ATTR-CM with loop-acting diuretics, and doxycycline; and were most commonly treated for AL amyloidosis with melphalan and dexamethasone. Half of this study population made lifestyle changes following diagnosis, and most used complementary therapies to manage their amyloidosis

Most of the participants in this study population reported having discussions about clinical trials with their clinician and though only one had taken part in a clinical trial. Participants in this study would be willing to participate if there was a suitable trial for them.

This is a patient population that described mild side effects as fatigue and diarrhoea. They described severe side effects as pain, neuropathy, nausea and vomiting.

Within this patient population, most participants adhered to treatment at the advice of their clinician or as long as it was prescribed. They felt that evidence of stable disease and an improvement in general well-being were needed to feel like treatment was effective.

This is a patient population that primarily needed the advice of their clinician as well as information about side effects, scientific evidence and clinical advice or expertise in order to feel comfortable trying new treatments.

The cohort was split between people who did not need support to have treatment at home, and those who needed the support from family or friends, regular check-ups from a GP or nurse, and someone to call if they had a question or issue.

Participants in this study had excellent knowledge about their condition and treatments, an excellent ability to adhere to treatments and communicate with healthcare professionals, excellent recognition and management of

symptoms, and a very good ability to manage the effects of their health condition on emotional well-being, social life and healthy behaviours.

This is a patient population that primarily accessed information through the internet, books, pamphlets and newsletters as well as from specific health charities. They found information from reliable sources and from their doctors helpful, and preferred to get information by talking to someone. They were most receptive to information at the time of diagnosis.

The participants in this PEEK study had very good communication, navigation and overall experience of care coordination. They mostly experienced positive communication from health care professionals with holistic, two way, and supportive conversations.

This is a patient population that experienced support and care from family and friends, through hospital or clinical settings, peer support and charities though some reported the challenges of finding or accessing support.

This is a patient population where their condition had an impact on their mental and emotional health, and it had a negative impact on their quality of life. The participants in this PEEK study had moderate levels of anxiety in relation to their condition. They managed their general health by understanding their limitations.

This is a group who would most like to control heart and lung symptoms. The most important aspect for making decisions about their own treatment was medication safety, and they thought that decision-makers should consider quality of life when making decisions about treatment for people with amyloidosis.

This is a patient population that would like future treatments to be more affordable, and more effective.

This is a study cohort did not have any recommendations for information about their condition, but want more access to support services. They would like health professionals to have more knowledge of their condition.

This is a patient population that felt grateful for healthcare staff and the entire health system in general.

This is a patient population that wanted to tell patients and families in the future that they should seek peer support and join support groups, as well as seeking and accepting support in general.

Section 1 Summary: Introduction and methodology

About this condition

Amyloidosis is a heterogeneous disease, where amyloid deposits form and accumulate in tissues and organs of the body. It can be acquired or hereditary, localised or systemic. The amyloid deposits can accumulate in the heart, kidneys, spleen, nerves, and blood vessels [1].

Participants

To be eligible for the study, participants needed to have been diagnosed with ATTR-CM or AL amyloidosis, or be a carer to someone diagnosed with either condition, have experienced the healthcare system in Australia, be 18 years of age or older, be able to speak English, and be able to give consent to participate in the study. Initial recruitment commenced in July 2019 to October 2019 and recommenced April 2020 to June 2020.

Personal Experience, Expectations and Knowledge (PEEK): Study position

In this PEEK study, 28 people diagnosed with amyloidosis, and 8 carers to people with amyloidosis throughout Australia participated in the study that included a qualitative structured interview and quantitative questionnaire. This study in amyloidosis is therefore the largest mixed methods study reported in an Australian population, and it includes the most patient interviews worldwide. In addition, PEEK is a comprehensive study covering all aspects of disease experience from symptoms, diagnosis, treatment, healthcare communication, information provision, care and support, quality of life, and future treatment and care expectations.

Section 2 Summary: Demographics

Participants

- In this PEEK study, 28 participants with amyloidosis, and 8 carers to people with amyloidosis were recruited into the study, 14 females (38.89%) and 22 males (61.11%), aged mostly between 55 and 74 (n=27, 75.00%), and most participants identified as Caucasian/white (n=33, 91.67%).

- Participants were most frequently from Queensland (n=14, 38.89%), New South Wales (n=11, 30.56%), and Western Australia (n=6, 16.67%). Most participants were from major cities (n=27, 75.00%) and they lived in all levels of advantage, defined by Socio-economic Indexes for Areas (SEIFA) with 25 participants (69.44%) from an area with a high SEIFA score of 7 to 10 (more advantage), and 11 participants (30.56%) from an area of mid to low SEIFA scores of 1 to 6 (less advantaged).

Baseline health

- The Short Form Health Survey 36 (SF36) measures baseline health, or the general health of an individual. The SF36 comprises nine scales: physical functioning, role functioning/physical, role functioning/emotional, energy and fatigue, emotional well-being, social function, pain, general health, and health change from one year ago. The scale ranges from 0 to 100, where a higher score denotes better health or function.

- The **"SF36 Role functioning/emotional"** scale measures how emotional problems interfere with work or other activities. On average, any emotional problems of the participants in this study slightly interfered with work or other activities. The **"SF36 Emotional well-being"** scale measures how a person feels, for example happy, calm, depressed or anxious. On average, the participants in this study participants felt happy and calm most of the time, and anxious and depressed a little of the time.

- The **"SF36 Physical functioning"** measures health limitations in physical activities such as walking, bending, climbing stairs, exercise, and housework. On average, physical activities for participants in this study moderately limited. The "**SF36 Role functioning/physical"** scale measures how physical health interferes with work or other activities. On average, physical health of the participants in this study interfered quite a bit with work or other activities.

- The **"SF36 Social functioning"** scale measures the limitations on social activities due to physical or emotional problems. On average for the participants in this study, social activities were slightly limited

- The "SF36 Role Energy/Fatigue" scale measures the amount of energy or fatigue. On average the participants in this study had moderate energy/fatigue, that is, felt tired some of the time and had energy some of the time.

- The **"SF36 Pain"** scale measures the amount of pain, and how pain interferes with work and other activities. On average, the participants in this study had moderate pain.

Section 3 Summary: Symptoms and diagnosis

Symptoms

- Participants had between zero and 13 symptoms (median = 5.00, IQR = 3.00), most commonly three to four symptoms (n=6, 21.43%) (Table 3.1). The most common symptoms for all participants were fatigue (n=18, 64.29%), being short of breath (n=16, 57.14%), limb weakness (n=16, 57.14%), and light-headedness (n=16, 57.14%).

- The median quality of life was between 1.00 and 4.00, for all of the symptoms listed in the questionnaire, this is in the "Life was very distressing" to "Life was average" range. Median quality of life for the most common symptoms (fatigue, short of breath, light-headedness, and limb weakness) was between 3.00 and 4.00, in the life was a little distressing.

Symptoms leading to diagnosis

- In the online questionnaire, participants were asked to select every symptom that they had at diagnosis. In the structured interview, participants were asked to describe the symptoms that actually *led* to their diagnosis. The most common symptom leading to diagnosis was excessive weight loss (n=8, 22.22%). There were seven participants (19.44%) who described experiencing breathlessness and four participants (11.11%) who described having tiredness. A final four participants (11.11%) identified a specific physical sensation, such as numbness or tingling in their fingers or toes, which led to their diagnosis.

- When discussing symptoms leading to their diagnosis, participants described how soon after experiencing symptoms they sought medical attention. There were five participants (13.89%) that described having symptoms and not seeking medical attention initially but recognising the importance of those symptoms in hindsight. An additional three participants (8.33%) also mentioned having symptoms and not seeking medical attention initially, but they provided no reason for this.

- Overall, 18 participants (50.00%) described having symptoms and seeking medical attention relatively soon. There were eight participants (22.22%) that described having symptoms and not seeking medical attention initially, and a final five participants (13.89%) that described having no symptoms or not noticing them prior to diagnosis.

- There were nine participants (25.00%) that described a diagnostic pathway that required appointments with a general practitioner and two or more specialists. There were also nine participants (25.00%) who described receiving a diagnosis following referral from their general practitioner to a specialist. A final six participants (16.67%) described receiving diagnosis following a specialist ordering tests. They made no mention of a GP referral.

- When discussing symptoms, overall participants had either a strong recollection of symptoms (69.44%) or describes not experiencing any symptoms prior to diagnosis (11.11%).

Diagnostic tests

- Participants had between one and 11 diagnostic tests, most commonly five to six tests (n=11, 39.29%) (Median = 6.5, IQR = 3.25) (Table 3.5, Figure 3.5). The most common diagnostic tests were blood tests (n=23, 82.14%), electrocardiogram (n=18, 64.29%), and echocardiogram (n=16, 57.14%).

Time from symptoms to diagnosis

- Participants most commonly had more than a year between noticing symptoms and being diagnosed (n=11, 42.31%), followed by between 6 months and a year (n=7, 26.92%). There were five participants (19.23%) that had noticed symptoms between one and six months before getting diagnosed, and three participants (11.54%) that had less than one month.

Time from diagnostic tests to diagnosis

- The majority of participants waited between 2 and 3 weeks (n=8, 28.57%) or more than 4 weeks (n=8, 28.57%).

Diagnosis provider and location

- The diagnosis was given most commonly by the haematologist (n=9, 32.14%), followed by a cardiologist (n=7, 25.00%). The diagnosis was most commonly given at a specialist clinic (n=28, 67.86%).

Understanding of disease at diagnosis

- Participants were asked in the structured interview how much they knew about their condition at diagnosis and the reason for their level of knowledge. There were 15 participants (41.67%) that gave no specific reason for their level of knowledge. There were eight participants (22.22%) who said they came to understand their condition more over time and through lived experience, and four participants (11.11%) described knowing very little about their condition at diagnosis, but that they were aware of family history with the condition.

- Overall, there were 27 participants (75.00%) that described knowing nothing or very little at diagnosis and these were the most common themes. There were three participants (8.33%) who noted that they knew good amount about the condition at diagnosis.

Emotional support at diagnosis

- Almost half of participants (including carers) had enough support (n=17, 47.22%), 6 participants (16.67%) had no support, and 13 participants (36.11%) had some support but it wasn't enough.

Information provided at diagnosis

- The majority of participants had enough information (n=20, 71.43%) at diagnosis. There were eight participants (28.57%) that had some information but not enough, and there were no participants that had no information at all at diagnosis.

Costs at diagnosis

- There were 12 participants (42.86%) who could recall the out of pocket expenses at diagnosis. There were eight participants who had no out of pocket expenses at diagnosis (28.57%), two that spent between $100 and $500 (7.14%), four who spent between $500 and $1000 (14.29%), and two who spent more than $1000 (7.14%) in out of pocket expenses

- In the follow-up question about the burden of costs at diagnosis, for 12 participants (60.00%) the cost was either slightly significant or not significant at all. For 5 participants (25.00%) the out of pocket expenses were somewhat significant, and for 3 participants (15.00%), the burden of out of pocket expenses were moderately significant.

Genetic tests and biomarkers

- The majority of participants had no conversation about biomarker/genomic/gene testing that might be relevant to treatment (n=17, 60.71%). There were three participants who brought up the topic with their doctor (10.71%), and eight whose doctor brought up the topic (28.57%).

- Over half of the participants (not including carers) have not had any testing but would like to (n=15, 53.57%). There were a total of 10 participants that had the test, either paying for it themselves (n=5, 17.86%), or not paying out of pocket (n=5, 17.86%). Three participants did not have the test and had no interest in having one (10.71%).

- The majority of participants were not sure if they had specific biomarkers (n=15, 53.57%), there were five that stated they had no biomarkers (17.86%), and eight that were able to name specific markers that they had.

Understanding of prognosis

- Participants were asked in the structured interview to describe what their current understanding of their prognosis was. There were 15 participants (41.67%) that described that they had a discussion about prognosis, and there were 14 participants (38.89%) did not mention having discussions about prognosis.

- Overall, 18 participants (50.00%) described having a clear understanding of their prognosis and 11 described having an unclear understanding (30.56%).

- There were two main themes that were equally reported, including participants describing their prognosis in relation to the specific medical interventions they need to manage their condition (n=9, 25.00%) and relating their prognosis to a specific timeframe that they are expected to live (n=9, 25.00%). There were eight participants (22.22%) that described their prognosis in relation to poor outcomes or as a terminal condition and five participants (13.89%) that understood their prognosis as positive and their condition as manageable.

Section 4 Summary: Decision-making

Discussions about treatment

- Participants were asked to recall what treatment options they were presented with and how they felt about such options. The most common response from participants was that it was difficult to remember/other response (n=14, 38.89%) which was closely followed by multiple treatment options were discussed which was described by 13 participants (36.11%). Six participants described discussing one treatment option (16.67%) and three participants described no treatment options being discussed (8.33%).

- Among participant who discussed multiple treatment options, five described participating in decision-making (13.89%), four described not participating in the decision-making process (11.11%) and four described being told what to do without discussion (11.11%). Three participants described being presented with no options because no therapies were available (8.33%). Out of those who were presented with one option three participants described being told what to do without discussion (8.33%) and two participants described some but very little discussion (5.56%).
- Some participants described discussions of specific treatments. Six participants described discussing the option of a stem cell transplant (16.67%), while four participants described discussing the option of a liver transplant (11.11%). Other participants described being presented with the option of chemotherapy (n=3, 8.33%), Green tea extract (n=3, 8.33%), Velcade or dexamethasone (n=3, 8.33%) and Bone marrow transplant (n=2, 5.56%).

Decision-making

- Participants were asked in the structured interview what they considered when making decisions about treatment. The most reported consideration was quality of life as part of multiple aspects that they consider when making decisions about treatment and this was described by 13 participants (36.11%). This was followed by efficacy as part of multiple aspects they consider (n=9, 25.00%); side effects as part of multiple aspects they consider (n=9, 25.00%); the long term impact and side effects of treatment as part of multiple aspects they consider (n=7, 19.44%), taking the advice of their clinician as part of multiple aspects they consider (n=6, 16.67%), considering the potential impact on their family or dependents as part of multiple aspects they consider (n=5, 13.89%), survival benefit as part of multiple aspects they consider (n=5, 13.89%) and taking the advice of their clinician as the only aspect they consider (n=5, 13.89%).

Changes in decision-making

- Participants were asked if the way they made decisions had changed over time. There were 15 participants (41.67%) that felt the way they made decisions about treatment had not changed over time, and 12 participants (33.33%) that described decision-making changing. Nine participants (25.00%) were unsure/other or gave no response.

- Where participants had changed the way they make decisions, this was primarily in relation to becoming more informed and/or assertive (n=7, 19.44%). Three participants described their decision-making changing over time as they are more aware of their health, responsibilities and/or limitations (8.33%) Other participants described changing over time as they are more accepting of their condition and choices available (n=1, 2.78%), they are more focused on how treatment impacts their family and dependents (n=1, 2.78%), they are more cautious and considered (n=1, 2.78%) and they are more focused on quality of life or the impact of side effects (n=1, 2.78%).

- Among participants who described no change in the way they make decisions the most common response was that this was because they had always been informed/assertive (n=7, 19.44%) followed by those who did not mention any reason (n=4, 11.11%). Other responses were that there had been no change because they always took the advice of clinicians (n=2, 5.56%) and because they have had no treatment options to choose from (n=1, 2.78%).

Section 5 Summary: Treatment

Main provider of treatment

- The haematologist was the main provider of amyloidosis treatment for the majority of participants (n=19, 67.86%).

Access to healthcare professionals

- All participants had access to a general practitioner (n=28, 100.00%) and the majority had access to a cardiologist (n=26, 92.86%), and haematologist (n=24, 85.71%) for the treatment of their amyloidosis.

Respect shown

- The majority of participants indicated that they had been treated with respect throughout their experience (n=31, 86.11%), five participants (13.89%) participants felt they had been treated with respect with the exception of one or two occasions, there were no participants who felt they weren't treated with respect.

Healthcare system

- The majority of participants had private healthcare insurance (n=23, 82.14%), five participants (17.86%) asked if they want to be treated as a public or private patient. The majority of participants had not been asked if they had private health insurance (n=15, 53.57%). Throughout their treatment, equal numbers of participants were treated as a public patient (n=11, 39.29%), or private patient (n=11, 39.29%), and most commonly in the public hospital system (n=13, 46.43%) (Table 5.4).

Affordability of healthcare

- The majority of participants never cancelled their appointments due to cost (n=23, 82.14.00%), while four (14.29%) participants rarely had to cancel appointments. Almost all participants (n=27, 96.43%) never had any trouble paying for prescriptions.

Cost of amyloidosis

- Almost all participants never or rarely found it difficult to pay for basic necessities such as housing food and electricity (n=25, 89.29%). There were two participants (7.14%) had to pay for additional carers for themselves or their family. Participants spent between $0 and $1400 per month on amyloidosis. The amount spent was extremely significant or moderately significant burden for 4 participants (14.29%), five found it somewhat significant (17.86%), and 19 participants found costs slightly or not at all significant (67.86%).

Changes to employment status

- Half of the participants (n=18, 50.00%) of this PEEK study were retired at the time of the amyloidosis diagnosis. There were six participant (16.67%) that quit their job, and four (11.11%) reduced their work hours.

- There were 25 (89.29%) participants with a main partner or carer, 13 partners or main carers (46.43%) did not have a job or were retired at the time of diagnosis, seven (25.00%) had no change in employment status, and three (10.71%) quit their job.

Reduced income due to amyloidosis

- A third of participants (32.14%) had a reduced family income due to amyloidosis. Participants noted a drop in monthly income of between $100 to over $5,000 per month. For 18 of these participants (54.54%), the burden of this reduced income was extremely or moderately significant.

Treatment

- The most common drugs taken for *ATTR-cardiac* subgroup were loop-acting diuretics (n=8, 44.44%), followed by doxycycline (n=7, 38.89%), and Diffusional (n=5, 27.78%).

- The most common treatment for *AL-amyloidosis* subgroup was Melphalan and Dexamethasone (50.00%).

Surgery

- There were five participants that had surgery, four participants had a single surgery for amyloidosis, and one patient had four or more surgeries. The types of surgeries that participants had include pacemaker related surgeries, liver transplant, defibrillator fitting, and carpal tunnel surgery.

Lifestyle changes

- Nearly half of the participants made no lifestyle changes (n=13, 46.43%). The most common lifestyle changes were exercise (n=12, 42.86%), and diet (n=9, 32.14%).

Complementary therapies

- There were 24 participants (85.71%) that used some form of complementary therapies to manage their amyloidosis. The most common complementary therapies used were exercise (n=18, 64.29%) and dietary supplements (n=13, 46.43%), and for ATTR-cardiac participant, half weighed themselves daily (n=9, 50.00%).

Clinical trials

- There was a total of 26 participants (92.86%) that had discussions about clinical trials, either by bringing up the topic themselves (n=5, 17.86%) or their doctor bringing up the topic (n=21, 75.00%).

- There was a single participant (3.57%) who had taken part in a clinical trial, and 22 (78.57%) who would like to take part in a clinical trial if there was a suitable one.

Description of mild side effects

- In the structured interview, participants were asked how they would describe the term ‘mild side effects’. The most common description of ‘mild side effects’ was in relation to a specific symptom as an example (n=19, 52.78%). The most common specific side effects given as an example was fatigue and/or tiredness (n=7, 19.44%) followed by diarrhoea (n=4, 11.11%). Another description of ‘mild side effects’ was those that can be self-managed and do not interfere with daily life (n=15, 41.67%).

Description of severe side effects

- In the structured interview, participants were asked how they would describe the term ‘severe side effects’. The most common description of ‘severe side effects’ given was a specific side effect given as an example (n=17, 47.22%). The most common specific side effect given was pain (n=6, 16.67%), followed by neuropathy/sensory disturbance (n=4, 11.11%) and nausea or vomiting (n=4, 11.11%). Other descriptions

of ‘severe side effects’ included those that impact everyday life/ability to conduct activities of daily living (n=12, 33.33%). Four participants described coping with all side effects (11.11%).

Adherence to treatment

- Participants were asked in the structured interview what influences their decision to continue with a treatment regime. The most common theme described was adhering as per the advice of their specialist or as long as its prescribed (n=16, 44.44%). Participants also reported not giving up on any treatment (n=6, 16.67%) and adhering to treatment for a specific amount of time (n=5, 13.89%).

What needs to change to feel like treatment is effective

- Participants were asked to describe what needs to change to feel like treatment is effective. The most common response from 11 participants (30.56%) was needing to experience evidence of stable disease or no disease progression. This was followed by needing to experience an improvement in general wellbeing (n=9, 25.00%).

Information needed to be confident in new treatments

- Participants were asked to describe what information would be needed to be confident in a new treatment. The most common response from17 participants (47.22%) was needing the advice of their clinician followed by 14 participants (38.89%) was needing to know about side effects to feel confident about trying a new treatment. There were 11 participants (30.56%) that reported needing scientific evidence and this was followed by needing to conduct their own research (n=9, 25.00%); needing to know about efficacy (n=9, 25.00%) and needing to know the overall benefits (n=8, 22.22%).

Support needed for treatment at home

- Participants were asked to describe what support they would need if they were having treatment at home. The two most common responses were participants not needing support (n=8, 22.22%) and needing support from their friends or family (n=8, 22.22%). There were seven participants that reported needing regular check-ups with a GP or nurse (19.44%) and this was followed by needing someone to call if they have a question or issue (n=4, 11.11%). Four participants described needing training and education on how to administer treatment.

Section 6 Summary: Information and communication

Access to information

- In the structured interview, participants were asked what information they had been able to access since they were diagnosed. The most common type of information accessed by 20 participants (55.56%) was through the internet in general. This was followed by books, pamphlets and newsletters (n=15, 41.67%) and information from specific health charities (n=12, 33.33%). There were eight participants (22.22%) that described accessing information through their treating clinician and seven participants (19.44%) that described accessing information through Facebook and/or social media. Other types of information accessed included other patients' experiences (n=4, 11.11%) and primarily through journals or research articles (n=4, 11.11%).

Information that has been helpful

- In the structured interview, participants were asked to describe what information they had found to be *most* helpful. The most common type of information found to be helpful by 12 participants (33.33%) was information from reliable source, and this was followed by talking to their doctor or specialists (n=7, 19.44%). There were six participants (16.67%) that described health charities as being helpful and six (16.67%) that described information that's easy to understand as being helpful. Other types of information described as being helpful included information about what to expect (n=5, 13.89%), information specific to their condition (n=5, 13.89%) and other people's experiences (n=4, 11.11%).

Information that has not been helpful

- In the structured interview, participants were asked if there had been any information that they did not find to be helpful. The most common response by 18 participants (50.00%) was that no information was not helpful, and this was followed by GP and specialists as being not helpful (n=5, 13.89%).

Information preferences

- Participants were asked whether they had a preference for information online, talking to someone, in written (booklet) form or through a phone app. Overall, the most common theme was talking to someone (n=10, 27.78%). There were seven participants (19.44%) that described a preference for talking to someone plus online information. There were also seven participants (19.44%) that described online information as their main preference.

- There were 12 participants (33.33%) whose rationale for their preference was simply a personal preference or gave no strong rationale for their preference. Among those who gave a rationale for their preference, seven (19.44%) described it as due to being able to digest information at their own pace and six (16.67%) described it as due to being able to, or having time to, ask questions.

Timing of information

- Participants in the structured interview were asked to reflect on their experience and to describe when they felt they were most receptive to receiving information. The most common time that participants described being receptive to receiving information was from the beginning/diagnosis (n=12, 33.33%) and this was followed by participants describing being receptive to information a specific amount of time after (n=7, 19.44%). There were six participants (16.67%) that described being receptive to information after the shock of diagnosis.

Partners in health

- The Partners in Health questionnaire (PIH) measures an individual's knowledge and confidence for managing their own health. The Partners in Health comprises a global score, 4 scales; knowledge, coping, recognition and treatment of symptoms, adherence to treatment and total score.

- The **"Partners in health: knowledge"** scale measures the participants knowledge of their health condition, treatments, their participation in decision-making and taking action when they get symptoms. Participants in this study had excellent knowledge about their condition and treatments.

- The **"Partners in health: coping"** scale measures the participants ability to manage the effect of their health condition on their emotional well-being, social life and living a healthy life (diet, exercise, moderate alcohol and no smoking). Participants in this study had very good ability to manage the effects of their health condition on emotional well-being, social life and healthy behaviours.

- The **"Partners in health: treatment"** scale measures the participants ability to take medications and complete treatments as prescribed and communicate with healthcare professionals to get the services that are needed and that are appropriate. Participants in this study had an excellent ability to adhere to treatments and communicate with healthcare professionals.

- The **"Partners in health: recognition and management of symptoms"** scale measures how well the participant attends all healthcare appointments, keeps track of signs and symptoms, and physical activities. Participants in this study had excellent recognition and management of symptoms.

Information given by health professionals

- Participants were asked about what type of information they were given by healthcare professionals. Information about treatment options (n=27, 75.00%), disease management (n=26, 72.22%), and disease cause (n=22, 61.11%) were most frequently given to participants by healthcare professionals, and information about psychological/social support (n=8, 22.22%), and complementary therapies (n=4, 11.11%) were given least often.

Information searched independently

- Participants were then asked after receiving information from healthcare professionals, what information did they need to search for independently. Information about disease management (58.33%) disease cause (55.56%), and treatment options (55.56%) were most often searched for independently by participants. Psychological/social support (27.78%), and hereditary considerations (30.56%) were least searched for.

Information gaps

- The largest gaps in information, where information was neither given to patients nor searched for independently were for psychological/social support (n=21, 58.33%), hereditary considerations genes or genomic biomarker information (n=21, 58.33%), and complementary therapies (n=20, 55.56%). Participants were given most information either from healthcare professionals or independently for disease management (n=16, 44.44%), and treatment options (n=15, 41.67%). The topic that was most searched for independently following no information from health professionals was complementary therapies (n=12, 33.33%).

Most accessed information

- Participants were asked to rank which information source that they accessed most often, where 1 is the most trusted and 5 is the least trusted. Across all participants, information from the hospital or clinic where treated was most accessed, followed by information from non-profit or charities or patient organisations.

My Health Record

- My Health Record is an online summary of key health information, an initiative of the Australian Government. Eleven participants (39.29%) had accessed "My Health Record". There were 15 (53.57%) who had not, two participants did not know what it is (7.14%), and four participants (4.00%) were not sure. Of those that had accessed "My Health Record", five participants (45.45%) found it good or
- acceptable, six participants (54.54%) found it poor, or very poor.

Section 7 Summary: Care and support

Care coordination

- The **"Care coordination: communication"** scale measures communication with healthcare professionals, measuring knowledge about all aspects of care including treatment, services available for their condition, emotional aspects, practical considerations, and financial entitlements. On average, the participants in this study scored in the middle of the scale, indicating that participants had moderate communication with healthcare professionals.

- The **"Care coordination: navigation"** scale navigation of the healthcare system including knowing important contacts for management of condition, role of healthcare professional in management of condition, healthcare professional knowledge of patient history, ability to get appointments and financial aspects of treatments. On average, the participants in this study had good navigation of the healthcare system.

- The **"Care coordination: total score"** scale measures communication, navigation and overall experience of care coordination. On average, participants in this study had very good communication, navigation and overall experience of care coordination.

- The **"Care coordination: care coordination global measure"** scale measures the participants overall rating of the coordination of their care. On average, participants in this study rated their care coordination as very good.

- The **"Care coordination: Quality of care global measure"** scale measures the participants overall rating of the quality of their care. On average, participants in this study rated their quality of care as excellent.

Experience of care and support

- In the structured interview, participants were asked what care and support they had received since their diagnosis. This question aims to investigate what services patients consider to be support and care services. The most frequent description of care and support was family and friends (n=19, 52.78%). This was followed by receiving support through a hospital or clinical setting (n=14, 38.89%); through face-to-face peer support (n=10, 27.78%); through charities (n=7, 19.44%). There were seven participants that described finding or accessing support as challenging (19.44%).

Section 8 Summary: Quality of life

Experience of quality of life

- In the structured interview, participants were asked whether they felt that their condition had affected their quality of life. Overall, there were 19 participants (52.78%) that described a negative impact on quality of life and seven participants (19.44%) that felt that there had been minimal impact on their quality of life. The most common themes in relation to having a negative impact on quality of life included a reduced capacity for physical activity (n=15, 41.67%) and emotional strain on family or a change in relationship dynamics (n=13, 36.11%). There were also eight participants (22.22%) that described a negative impact as they are unable to travel or need to adapt significantly in order to travel. In addition, six participants (16.67%) described a negative impact as a result of fatigue, and another six (16.67%) noted a negative impact due to reduced social interaction. There were four participants (11.11%) that described a negative impact on their quality of life due to an inability to work or needing to make changes with their work.

Impact on mental health

- In the structured interview, participants were asked to share any impact on their emotional and mental health as a result of their condition. The most common theme that participants reported was experiencing at least some impact on their mental and emotional health (n=20, 55.56%). There were also seven participants (19.44%) that described experiencing no impact on their mental and emotional health overall.

Regular activities to maintain mental health

- In the structured interview, participants were asked what they needed to do to maintain their emotional and mental health. The most common way that participants reported managing their mental and emotional health was by using coping strategies such as remaining social, making lifestyle changes or having hobbies (n=10, 27.78%). There were nine participants (25.00%) that described the importance of physical exercise in maintaining their mental health and seven (19.44%) that described the importance of family and friends in this endeavour. Other common themes included consulting a mental health professional (n=6, 16.67%), experiencing an impact but not using any activities to maintain their mental health (n=5, 13.89%) and not doing any activities to maintain their mental health as they have experienced no impact (n=4, 11.11%).

Regular activities to maintain health

- In the structured interview, participants were asked to share some of the things they needed to do every day to maintain their health. The most common way that participants reported managing their health was by understanding their limitations (n=15, 41.67%). There were 10 participants (27.78%) that described staying physically active and nine (25.00%) that described the importance of complying with treatment. Other common themes included maintaining a healthy diet (n=7, 19.44%) and the importance of self-care, for example getting more rest or seeking support for housework (n=5, 13.89%).

Impact on relationships

- In the structured interview, participants were asked whether their condition had affected their personal relationships. The most common themes in relation to impact on relationships was participants describing their relationships with family being strengthened (n=6, 16.67%) and experiencing changing dynamics in their relationships due to added anxiety, exacerbations and/or physical limitations (n=6, 16.67%).

- Overall, there were nine participants (25.00%) that described a negative impact on relationships, eight participants (22.22%) that reported a positive impact on relationships and seven participants (19.44%) that

felt that relationships had not been impacted. There were also five participants (13.89%) who noted an impact on their relationships but did not feel it was positive or negative overall.

Burden on family

- In the structured interview, participants were asked whether they felt that their condition placed additional burden on their family. Where participants described there was no additional burden, this was primarily described in general terms, with no specific examples provided (n=11, 30.56%). On the other hand, where participants felt there was an additional burden, this was primarily described in relation to the additional mental or emotional strain placed on their family (n=7, 19.44%), the extra household duties and responsibilities their family needed to take on (n=6, 16.67%) and as a burden in general, with no specific examples (n=4, 11.11%).

- Overall, there were 16 participants (44.44%) that felt there was an additional burden and 11 participants (30.56%) that reported no additional burden.

Experience of anxiety related to disease progression

- The Fear of Progression questionnaire measures the level of anxiety people experience in relation to their conditions. The Fear of Progression questionnaire comprises a total score, between 12 and 60, with a higher score denoting increased anxiety. Overall the participants had a mean total score of 33.19 (SD = 9.92), which corresponds to moderate levels of anxiety.

Section 9 Summary: Expectations and messages to decision-makers

Expectations of future treatments

- In the structured interview, participants were asked what their expectations of future treatments are. The most common theme was participants expected treatments to be more affordable (n=18, 50.00%), followed by the expectation that future treatments would be more effective (n=8, 22.22%). There were six participants (16.67%) that recommended future treatments should have fewer or less intense side effects and four participants (11.11%) that called for future treatments to be less invasive.

Expectations of future information

- Participants were asked in the structured interview if there was anything that they would like to see changed in the way information is presented or topics that they felt needed more information. The most common theme was participants having no recommendations or feeling satisfied with the information currently available (n=7, 19.44%), and this was followed by the expectation that future information would be easier to understand (n=6, 16.67%). There were five participants (13.89%) that recommended more information to inform the community and decision-makers about the condition. There were also four participants (11.11%) who suggested future information provide more details about new treatments and trials and four participants (11.11%) that called for more details about the specific classification of their condition.

Expectations of future communication with healthcare professionals

- Participants were asked in the structured interview what they would like to see in relation to the way that healthcare professionals communicate with patients. The most common theme was the expectation that future communication will involve health professionals having a better knowledge of the condition (n=13, 36.11%), and this was followed by no recommendations or participants feeling they had experienced good communication (n=10, 27.78%).

Expectations of future care and support

- Participants were asked in the structured interview whether there was any additional care and support that they thought would be useful in the future, including support from local charities. The most common theme was more access to support services in future (n=8, 22.22%), and this was followed by participants having no recommendations or being satisfied with the care they have received (n=6, 16.67%). There were four participants (11.11%) that recommended future care and support involving more peer support such as support groups and four participants (11.11%) that called for care and support to include more long-term condition management or care planning.

What participants are grateful for in the health system

- Participants were asked in the structured interview what aspects of the health system that participants are grateful for. The most common theme was participants expressing feeling grateful for the entire healthcare system (n=13, 36.11%). This was followed by those who were grateful for healthcare staff (n=10, 27.78%), low cost or free medical care through the government (n=10, 27.78%), timely access to treatment (n=5, 13.89%) and access to private healthcare/insurance (n=4, 11.11%).

Symptoms and aspects of quality of life

- Participants were asked to rank which symptoms/aspects of quality of life would they want controlled in a treatment for them to consider taking it. The most important aspects reported for participants with

ATTR-cardiac were heart and lung symptoms (e.g. short of breath, palpitations, chest pain), and arm and leg symptoms (e.g. numbing, tingling, weakness, pain).

- The most important aspects reported for participants with AL amyloidosis were heart and lung symptoms (e.g. short of breath, palpitations, chest pain), and kidney symptoms (fatigue, loss of appetite and swelling in feet, ankles or legs).

Values for decision-making

- Participants were asked to rank what is important for them overall when they make decisions about treatment and care. The most important aspects were 'How safe the medication is and weighing up the risks and benefits', and 'The severity of the side effects'. The least important were 'The financial costs to me and my family'.

Values for decision-makers

- Participants were asked to rank what is important for decision-makers to consider when they make decisions that impact treatment and care. The two most important values were quality of life for patients, and access for all patients to all treatments and services; the least important was economic value to government.

Time taking medication to improve quality of life

- Participants were asked in the online questionnaire, how many months or years would you consider taking a treatment, provided it gave you a good quality of life, even if it didn't offer a cure. The majority of participants (n=19, 67.86%) would use a treatment for more than ten years for a good quality of life, even if it didn't offer a cure.

Message to decision-makers

- Participants were asked, 'If you were standing in front of the health minister, what would your message be in relation to your condition?'. The most common message was that treatments need to be affordable (n=10, 27.78%). This was followed by the message that there should be more clinical trials and/or new treatments (n=8, 22.22%), that there should be improved access to support and care (n=6, 16.67%), the need to take the condition seriously (n=5, 13.89%), the need to invest in professional development so that clinicians better understand the condition (n=5, 13.89%) and finally, to invest in research, including the effort to find new treatments (n=4, 11.11%).

Section 10 Summary: Advice to others in the future

Advice to other patients and families in the future

- In the structured interview, participants were asked what advice they would give to other patients and their families. Six themes emerged as a result, the most frequent of which was that newly diagnosed patients should seek peer support or join support groups (n=9, 25.00%), followed by advice to seek and accept support in general (n=8, 22.22%). Other themes that emerged were to do research and ask questions (n=6, 16.67%), to find the best medical support for you which may include seeking a second opinion (n=5, 13.89%), try to stay positive (n=4, 11.11%) and finally, to be aware of your own body and trust your instincts (n=4, 11.11%).

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