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PATIENT EXPERIENCE EXPECTATIONS KNOWLEDGE

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Mitochondrial Disease Australian Study

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INTERNATIONAL CENTRE FOR COMMUNITY-DRIVEN RESEARCH

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Summary of results

Section 1: Introduction and methodology

- Mitochondrial disease is a heterogenous group of diseases that have dysfunctional mitochondrial respiratory changes that are caused by mutations to nuclear or mitochondrial DNA. The disease may affect single organ or may affect multiple organs, and usually affect organs that have the highest energy needs such as muscles, brain, eyes and heart.
- The prevalence of mitochondrial disease is estimated at 11.5 per 100,000, however this may underestimate the prevalence with reports of one in 200 healthy births having a mitochondrial DNA mutation.
- Patient Experience, Expectations and Knowledge (PEEK) is a research program developed by the International Centre for Community-Driven Research (ICCDR). The aim of PEEK is to conduct patient experience studies across several disease areas using a protocol that will allow for comparisons over time (both quantitative and qualitative components). PEEK studies give us a clear picture and historical record of what it is like to be a patient at a given point in time, and by asking patients about their expectations, PEEK studies give us a way forward to support patients and their families with treatments, information and care.
- In this PEEK study, 50 people with mitochondrial disease or their carers, throughout Australia participated in the study that included a structured interview and quantitative questionnaire. This study in mitochondrial disease is therefore the largest mixed methodology study in Australia. 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: Demographics and study population characteristics

Demographics

- Fifty participants from Australia were recruited into the study, including 44 (88.00%) participants with mitochondrial disease and 6 (12.00%) carers of people with mitochondrial disease. There were an additional five participants that were both a patient and carer, however they responded to the questionnaire and interview as a patient rather than a carer.
- The majority of participants were from NSW (n=18, 36.00%), Victoria (n=12, n=24.00%), and Queensland (n=10, 20.00%), and most live in major cities (n=30, 60.00%).
- Thirty-seven females (74.00%) and 13 males (26.00%) participated.

Baseline Heath – SF36 score

The Short Form Health Survey 36 (SF36) measures baseline health, or the general health of an individual. A higher score indicates better baseline health.

- The overall scores for the cohort for emotional well-being were in the second highest quintile indicating very good baseline health.
- The overall scores for the cohort for pain were in the middle quintile indicating moderate baseline health.
- The overall scores for the cohort for physical functioning, role functioning/emotional, energy/fatigue, social functioning, general health, and health change were in the second lowest quintile indicating poor baseline health.
- The overall score for role functioning /physical were in the lowest quintile indicating very poor baseline health.

SF36 scores by general health

- Those with higher general health scored significantly better compared to lower general health for the physical functioning, emotional well-being, social functioning, role functioning/emotional, energy/fatigue, pain and health change scales.

SF36 scores by physical functioning

- Those with higher physical functioning scored significantly better compared to those with lower physical functioning for the SF36 role functioning/physical, energy/fatigue, social functioning, pain, general health and health change subscales.

SF36 scores by emotional well-being

- Those with higher emotional well-being scored significantly better compared to those with lower emotional well-being for the SF36 role functioning/physical, role functioning/emotional, social functioning, pain, general health and health change subscales.

SF36 scores by social functioning

- Those with higher social functioning scored significantly better compared to those with lower social functioning for the SF36 physical functioning, role functioning/physical, role functioning/emotional, emotional well-being, energy/fatigue, pain, general health and health change subscales.

SF36 scores by hearing problems

- No significant differences were observed between those with hearing problems and those with no hearing problems for any of the SF36 subscales

SF36 scores by eye problems

- No significant differences were observed between those with eye problems and those with no eye problems for any of the SF36 subscales

SF36 scores by location

- No significant differences were observed between those that live in metropolitan areas and those that live in regional or rural areas for any of the SF36 subscales

SF36 scores by education

- No significant differences were observed between those with a university qualification and those with high school or trade qualifications for any of the SF36 subscales.

SF36 scores by Socio-Economic Indexes For Areas (SEIFA)

- No significant differences were observed between those that live in an area with a higher SEIFA score (more advantaged) and those that live in an area with a lower SEIFA score for any of the SF36 subscales.

Section 3: Experience of symptoms and diagnosis

Symptoms at diagnosis

- The first question was in the online questionnaire and asked participants to recall all of the symptoms that they experienced and their quality of life while experiencing those symptoms. The most commonly reported symptoms were muscle symptoms by (such as muscle weakness, exercise intolerance, pain, fatigue, cramps and low muscle tone), noted by 47 (94.00%) participants, followed by fatigue (n=45, 90.00%), digestive tract symptoms (n=36, 72.00%), problems with eyes (n=34, 68.00%), central nervous system symptoms (n=32, 64.00%), and hearing problems (n=24, 48.00%). The symptoms that had the lowest average quality of life were central nervous symptoms (mean = 2.28; n=32, 64.00%), muscle symptoms (mean = 2.52; n=47, 94.00%), heart symptoms (mean = 2.53; n=15, 30%) and digestive tract symptoms (mean = 2.64; n=36, 72.00%).
- In the structured interview, participants were asked to describe the symptoms that actually led to their diagnosis, as opposed to all the symptoms that they could recall. There were 14 participants (28.00%) that described fatigues and/or a lack of stamina and 11 participants (22.00%) that described having gastrointestinal distress ranging from nausea, diarrhoea to constipation. The next most common symptoms leading to diagnosis were failing to thrive as an infant (n=8, 16.00%), weakness in the legs or not being able to use their legs (n=7, 14.00%) and migraines that were sometimes also described as being stroke-like (n=7, 14.00%).
- In relation to sub-group variations, participants from a low socio-economic area (26.09%) and those with a low general health (25.00%) reported having severe migraines more frequently compared to the general population (14.00%), while those with a high general health reported this less frequently (0.00%). In relation to gastrointestinal distress, participants who had a high school or trade education reported this less frequently (11.54%) while those with a university education (33.33%) and those that are hearing impaired (37.50%) reported this more frequently than the general population (22.00%). Participants with a university education (20.83%) and participants with hearing impairment (20.83%) reported diabetes being a condition that led to their diagnosis more frequently than the general population (1000%). Participants with high physical function (40.91%) reported experiencing fatigue and/or lack of stamina more frequently than the general population (28.00%) while those with low physical function reported this less frequently (17.86%). Participants with high social function (40.00%) also reported experiencing fatigue and/or lack of stamina more frequently than the general population (28.00%).

- As part of the structured interview analysis in relation to symptoms that lead to diagnosis, there were 13 participants (26.00%) that noted a hereditary component that led to their diagnosis. In some cases it was a known hereditary link while in others, the hereditary link was identified as part of the diagnostic process.

Support at diagnosis

- In the questionnaire, participants were asked whether they felt supported at the time of diagnosis. There were 36 participants (72.00%) that indicated that they had no support at diagnosis, while 3 participants (6.00%) noted that they had enough support. An additional 11 participants (22.00%) indicated that they had some support but that it was not enough.
- In relation to sub-group variations, participants with no eye problems reported having no support at diagnosis more frequently than the general cohort (81.25% compared to 72.00% in the general cohort),. Participants that had higher general health reported that they had no support at diagnosis, more frequently than the general cohort (86.36% compared to 72.00% in the general cohort), and reported less frequently than the general cohort that they had some support but it wasn't enough, (13.64% compared to 22.00% in the general cohort)

Genetic/biomarker tests

- Participants were asked whether they had ever had a discussion about genetic tests or tests to see if there were biomarkers that might be relevant to their condition or treatment. Six participants (12.00%) indicated that they had brought up the topic for discussion with their doctor, 15 participants (30.00%) reported that their doctor had brought up the topic for discussion, 29 participants (58.00%) had no discussion about genetic tests.
- In relation to sub-group variations, participants with higher social functioning indicated that their doctor brought up the topic of biomarker/genetic testing, more frequently than the general cohort and those with lower social functioning less frequently (higher social functioning 45.00%; lower social functioning 20.00%, compared to 30.00% in the general cohort). Participants with no eye problems indicated that no one brought up the topic of biomarker/genetic testing, more frequently than the general cohort (68.75%, compared to 58.00% in the general cohort).
- Participants were asked about their interest in this type of test if it was available, the majority noted that they had not had this test, but would like to (n=26, 52.00%), 8 participants (16.00%) reported having this test and not paying out of pocket for it, 8 had this test as part of a clinical trial (16.00%), and two paid for this test themselves (4.00%). There were 6 participants (12.00%) indicated that they had not had this test and were not interested in it.
- In relation to sub-group variations, participants that had hearing problems, no eye problems and that were university educated indicated that they had not had this test but would like to, less frequently than the general cohort (41.67%, 31.25% and 33.33% respectively compared to 54.00% in the general cohort), while participants that did not have hearing problems, had no eye problems and had high school or trade qualifications indicated that they had not had this test but would like to, more frequently than the general cohort (61.54%, 61.76%, and 69.33% respectively, compared to 54.00% in the general cohort).
- In the structured interview, participants were also asked to talk about their understanding of genetic or biomarker testing. Some of the descriptions included understanding that the test is used for diagnosis of mitochondrial disease; understanding that the test cannot help them but may help others in the future; and understanding that the test cannot target treatment as there are no treatments available or that there was no clinical indication following the test.

Understanding of condition at diagnosis

- Participants were asked how much they knew about mitochondrial disease at diagnosis. There were 31 participants (62.00%) that described knowing nothing about mitochondrial disease and this was the most common response. There were also eight participants (16.00%) that described knowing about mitochondrial disease by the time they were diagnosed because the time to diagnosis was relatively long, giving them time to educate themselves.

Understanding of prognosis

- Participants were asked whether anyone talked to them about prognosis. The most common theme noted by 26 participants (52.00%) was prognosis had not been clearly discussed. The next most common theme was that participants understood that mitochondrial disease came with a poor prognosis that was primarily related to physical decline and this was noted by 9 participants (18.00%). There were seven participants (14.00%) that described the need for ongoing management of their condition and this included the management of exacerbations. The final theme in relation to understanding of prognosis was that mitochondrial disease came with a poor prognosis, including reduced life expectance and/or a rapid disease progression. This was noted by six participants (12.00%).

Section 4: Experience of health professional communication

Conversations about treatments

- Participants were asked to describe the conversations they have had about mitochondrial disease treatment options. The most common treatments discussed were Coenzyme Q10 and ATP support (n=19, 38.00%). The next most common theme was that participants were told that there is no treatment for mitochondrial disease (n=16, 32.00%). Other themes included having no or little discussion about treatment options (n=9, 18.00%) and having discussions about lifestyle changes (diet, exercise etc.) (n=9, 18.00%).
- In relation to sub-group variations, participants with a university education (45.83%) reported being told that there were no treatments for mitochondrial disease, more frequently than the general population (32.00%)

Decision-making

- There were 16 participant (32.00%) that noted considering side effects, of which 10 participants noted a combination of both the benefits of the treatment as well as the side effects. The nest most common consideration was cost (n=9, 18.00%) followed by impact on lifestyle, including ability to work (n=6, 12.00%).
- In relation to sub-group variations, there were no participants from low socio-economic areas that reported considering the impact on their lifestyle (n=0, 0.00%) and there were no participants with high physical functioning (n=0, 0.00%) or high social functioning (n=0, 0.00%) that reported considering quality of life when making decisions about treatment.
- In the final question about decision-making, participants were asked whether they felt the way they made decisions had changed over time since they were diagnosed. Overall there were 26 participants (52.00%) that felt as though the way they make decisions has changed over time, while 20 participants (40.00%) felt that it had not changed.
- Where participants did feel as though the way they made decisions had changed, the most common reason for this was that they had become more informed (n=11, 22.00%) and that they consider quality of life more in the process of making treatment decisions (n=7, 14.00%).
- In relation to sub-group variations, participants from rural areas (25.00%) reported considering quality of life more frequently than the general population (14.00%).

Section 5: Experience of treatment

Discussions about Clinical Trials

- In this PEEK study, 64% of all participants (n=32) describe not being spoken to about clinical trials, seven participants brought up the topic with their doctor (14.00%) and the doctors of 11 participants brought up the topic (22.00%).

Participation in Clinical Trials

- Seven participants have taken part in a clinical trial (14.00%), and 33 participants have not taken part in a clinical trial would like if one was suitable for them (66.00%). Ten participants have not taken part and do not want to (20.00%)

Treatments experienced

- Participants were asked in the questionnaire to identify the treatments that they had experienced. most common treatments were Coenzyme Q10 (n=36, 72.00%), vitamins and supplements (n=32, 64.00%), followed by physical therapy (n=15, 30.00%), and diet (n=11, 22.00).
- Participants were asked to rate their quality of life on a scale of 1 to 7, while using each specific treatment (with 1 being 'Life was very distressing and 7 being 'Life was great'). Mean quality of life scores ranges from 3.34 to 4.33, that is, all quality of life scores were within the 'life was a little distressing' to 'Life was average' range. The treatment that scored the least impact on quality of life was speech therapy (mean score 4.33). All other treatments were in the 'Life was a little distressing' range (mean scores range 3.34 to 3.86).
- The treatments that had a mean effectiveness score of at least 3 (moderately effective) were respiratory therapy (average score 3.50), speech therapy (average score 3.33), and diet (average score 3.09). The remaining treatments scored had a mean effectiveness score of at least 2, that is in the somewhat effective range.
- Participants were asked in the structure interview to provide a description of mild side effects. The most common description of mild side effects were those that do not greatly impact activities of daily living (n=11, 22.00%). In relation to specific side effects that were considered to be mild, there were seven participants (14.00%) that described headaches, six participants (12.00%) that described gastrointestinal problems (diarrhoea and cramping) and five participants (10.00%) that described increased fatigue (and related irritability) as a mild side effect. There were also six participants that did not describe a mild side effect but talks about mitochondrial disease being part of everyday life (Particularly pain).
- In relation to sub-group variations, participants with high social functioning (40.00%) described mild side effects as those that do not greatly impact activities of daily living more frequently than the general population (22.00%).
- Participants were asked in the structure interview to provide a description of severe side effects. The most common description of severe side effects were those that limit daily activities for an extended period of time (n=19, 38.00%), seven participants (14.00%) described sever side effects as an effect requiring hospitalisation or medical attention/permanent damage, or a life threatening effect or inability to function. In relation to specific side effects that were considered severe, nine participants (18.00%) described severe fatigue, four participants (8.00%) described chronic headaches and four participants (8.00%) described loss of mobility or independence.
- In relation to sub-group variations, participants with a high school or trade education (15.38%), low physical function(25.00%) and low social functioning (26.67%) described severe side effects as effects limiting their daily activities for an extended period of time, less frequently than the general population (38.00%), while those with a university education (62.50%), high physical function (54.44%), high social functioning (55.00%), high general health (50.00%) and hearing impairment (50.00%) described this more frequently.

Adherence to medication

- Participants were asked in the online questionnaire if, in general, if they were good at taking medicine and sticking to it. The majority of participants were good at sticking to treatments all of the time (n=30, 60.00%) and the remaining were good at sticking to treatments most of the time (n=20, 40.00%). No participants felt they were never, rarely or sometimes good at sticking to treatments.
- Participants were also asked in the structured interview how long they stick with a therapy before they think it might not be working or give up on it. Close to half of all participants (n=24, 48.00%) describes using treatment for a period of one to three months before deciding if its working. The next most common theme was continuing a treatment indefinitely or as recommended by clinician/specialist (n=9, 18.00%) and there were six participants (12.00%) that described not trying new medications for mitochondrial disease and such not knowing how long they would continue a treatment.
- In relation to sub-group variations, participants from rural areas (30.00%) and participants with a hearing impairment (29.17%) reported continuing a treatment indefinitely or as recommended by clinician/specialist more frequently than the general population (18.00%). Participants with a hearing impairment (29.17%) reported using treatment for a period of one to three months before deciding if its working less frequently than the general population (48.00%), while those with high social functioning (60.00%) and high general health (59.09%) reported this more frequently.
- Participants were asked what needed to change for them to feel as though a treatment was working. The most common description was needing to feel more energetic, and increase in physical ability, to know a treatment is working (n=20, 40.00%). This was followed by needing to see improved symptoms by clinical measurement (test result) (n=13, 26.00%) and needing to generally feel better to know that a treatment is working (n=9, 13.00%). There were five participants (10.00%) that described needing to reduce pain to know a treatment is working and five participants (10.00%) needing to improve their quality of life to know a treatment is working.
- In relation to sub-group variations, participants from metropolitan areas (26.67%) and participants from high socio-economic areas (25.93%) reported needing to feel more energetic, and increase in physical ability, to know a treatment is working, less frequently than the general population (40.00%) while participants from rural areas (60.00%), participants from low socio-economic areas (56.52%). Participants with a hearing impairment (41.67%) reported improved symptoms by clinical measurement (test result) more frequently than the general population (26.00%). Participants with high physical functioning (31.82%) described needing to generally feel better to know that a treatment is working, more frequently than the general population (18.00%).

Complementary therapies

- Participants were asked whether they had used any complementary therapies. The most common therapies that were considered complementary and described by participants were vitamins, minerals and supplements (n=14, 28.00%) and allied health e.g. physiotherapy (including massage and hydrotherapy), speech therapy, occupational therapy (n=14, 14.00%). The next most frequent complementary therapies described were alternative medicine, e.g. osteopathy, acupuncture, chiropractor, Bowen therapy (n=12, 24.00%). There were also 11 participants (22.00%) that noted that they did not use any complementary therapies.

Service provision and affordability

- The main physician treating participants for mitochondrial disease were general practitioners (N=19, 38.00%), followed by neurologists (N=12, 24.00%) and mitochondrial specialists (N=11, 22.00%).
- Participants had access to a general practitioner (n=48, 96.00%), neurologist (n=43, 86.00%), mitochondrial specialist (n=29, 58.00%) and cardiologist (n=28, 56.00%) for the treatment of their mitochondrial disease.
- The majority of patients had private healthcare insurance (n=37, 74.00%), 29 (58.00%) participants were treated as public patients, 12 (24.00%) as private patients and 9 (18.00%) as equally public and private patients. The majority of participants were treated in the public hospital system (n=32, 64.00%).
- Almost half of participants have never missed medical appointments due to cost (n=24, 48.00%), and most have never been unable to afford prescription medications (n=34, 64.00%). Almost half of participants have found it somewhat to extremely difficult paying for basic needs due to their diagnosis with mitochondrial disease (n=24, 48.00%).

Changes to work status

- The work status for a number of participants changed due to their diagnosis with mitochondrial disease with about a quarter of participants reducing the number of hours worked (n=13, 26,00%), and 19 (38.00%) quitting their jobs.
- Of those that had a partner or carer, four carers/partners had to quit their job (23.53%), seven had to reduce the number of hours worked (41.18%), carers have had to take leave either with pay (n=2, 11.76%), or without pay (n=5, 29.41%).

Experience of respect during treatment

- Participants were asked if they felt they had been treated with respectfully throughout their treatment. Half of the participants felt that they had been treated respectfully with the exception of one or two occasions (n=25, 50.00%), 18 felt that they had been treated respectfully (36.00%) and seven felt they had not been treated respectfully (14.00%).

Section 6: Information and communication

Access to information

- The most common response from over half of all participants was accessing information from the Australian Mitochondrial Disease Foundation (n=32, 64.00%). The next most common theme was accessing information via the internet (n=25, 50.00%). There were 14 participants (28.00%) that described accessing information from medical journals and peer reviewed papers and 13 participants (26.00%) that described accessing information from online forums including Facebook.
- In relation to sub-group variations, participants from rural areas (75.00%), participants with a high school or trade education (76.92), participants with low physical functioning (75.00%) and low general health (75.00%) reported accessing information from the Australian Mitochondrial Disease Foundation more frequently than the general population (64.00%), while participants with a university education (50.00%) and high physical functioning reported this less frequently. Participants from rural areas (65.00%) and those with high physical function (68.18%) reported accessing information from the internet more frequently than the general population (50.00%), while those from low socio-economic areas (37.04%) and those with low physical functioning (35.71%) reported this less frequently. Participants from rural areas (15.00%) reported accessing medical journals less frequently than the general population (28.00%).

Information that was helpful

- There was a range of information that participants found particularly helpful including information from the AMDF (n=9, 18.00%) research papers (n=7, 14.00%), communicating with others with mitochondrial disease (n=7, 14.00%) and information from clinical teams (n=5, 10.00%).

Information that was not helpful

- The most common theme described by 22 participant (44.00%) was that no information was unhelpful. There were no other themes noted by more than five participants, however where participants made a comment about information that was not helpful, this included stories about other patients (n=3, 6.00%), lack of concise yet comprehensive information (n=3, 6.00%), and information that is too general (n=2, 4.00%) or too scientific (n=2, 4.00%).

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. The most common theme was talking to someone (n=25, 50.00%) of which, five participants specified a preference for talking to someone face-to-face. The next most common theme was a preference for information online (n=21, 42.00%) and a preference for information in a written format such as a booklet (n=7, 14.00%).

- In relation to sub-group variations, participants with a hearing impairment (29.12%) described a preference for online information less frequently than the general population (42.00%), while participants with low physical functioning (53.57%) and low general health (53.57%) reported this preference more frequently. Participants with a hearing impairment (25.00%) reported a preference for accessing written information more frequently than the general population (14.00%)

Timing of information

- The most common time that participants described being receptive to receiving information was at the time of diagnosis (n=18, 36.00%) and this was followed by participants describing that there was not a specific time that they were most receptive and that it is an ongoing process (n=10, 20.00%). There were also six participants (12.00%) that described there not being a specific time when they were most receptive - depends on their emotional state and level of interest.
- In relation to sub-group variations, participants with low general health (46.43%) described being most receptive to information at diagnosis, more frequently than the general population (36.00%)

Health professional communication

- Participants were asked to describe the communication that they had had with health professionals throughout their experience. The most common theme was that participants described most healthcare professionals not knowing about mitochondrial disease (n=11, 22.00%). This was followed by participants being satisfied with health professional communication (n=10, 20.00%). The next most common themes were participants describing excellent communication (n=7, 14.00%), having minimal communication with healthcare professionals (n=6, 12.00%) and mostly good experiences, however there is a general lack of understanding of mitochondrial disease (n=6, 12.00%).
- In relation to sub-group variations, participants from low socio-economic areas (34.78%) described being satisfied with health professional communication more frequently than the general population (20.00%). Participants with high physical function (9.09%) and high general health (4.55%) described most healthcare professionals not knowing about mitochondrial disease less frequently than the general population (22.00%) while those with low physical functioning (32.14%) and low general health (35.71%) described this more frequently. Participants with high social functioning (25.00%) described excellent communication with their specialists more frequently than the general population (14.00%).

Knowledge and confidence

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 sub scales; knowledge, coping, recognition and management of symptoms, and adherence to treatment. A higher score denotes a better understanding and knowledge of disease.

Partners in health – overall score

- Overall, the participants scored in the top quintile for adherence to treatment indicating very good adherence to treatment. The scores for knowledge, recognition and management of symptoms, and total score were in the second highest quintile indicating good understanding and knowledge of disease. The score for coping was in the middle of the range of scores for this scale.

Partners in health - by general health

- Participants with higher general health had a statistically significant, better outcome for the coping subscale compared those with lower general health.

Partners in health – by physical functioning

- Participants with higher physical functioning had a statistically significant, better outcomes for the coping, adherence to treatment, and total score compared those with lower physical functioning.

Partners in health – by emotional well-being

- Participants with higher emotional well-being had a statistically significant, better outcomes for the coping, adherence to treatment, and total score compared those with lower emotional well-being.

Partners in health – by social functioning

- Participants with higher social functioning had a statistically significant, better outcomes for the coping, and total score compared those with social functioning.

Partners in health – by hearing problems

- No differences were observed between those with no hearing problems and those with hearing problems for any PIH subscale.

Partners in health – by eye problems

- Participants with no eye problems had significantly higher scores for the PIH knowledge, adherence to treatment and total score compared to those with eye problems.

Partners in health – by location

- Participants living in regional or rural areas had had a statistically significant, worse outcomes for the total score subscales compared those living in metropolitan areas.

Partners in health – by education

- No differences were observed between those with university education and those with high school or trade qualifications for any PIH subscale.

Partners in health – by SEIFA

- No differences were observed between those that lived in a higher SEIFA area compared to those that lived in an area with lower SEIFA scores for any PIH subscale.

Information given by health care professionals

- Participants were asked about what type of information they were given by healthcare professionals and what type of information they searched for independently:
- Information about disease cause (50.00%), treatment options (38.00%), and disease management (38.00%) were most frequently given to participants by healthcare professionals.
- Information about clinical trials (14.00%), interpreting test results (14.00%) and complementary therapies (16.00%) were give least often.
- Eight participants (16.00%) indicated that they received no information at all from health professionals about mitochondrial disease.

Information searched for independently

- Participants were asked about what type of information they searched for after receiving information from healthcare professionals:
- Information about treatment options (63.27%), disease management (59.18%), and disease cause (57.14%) were most frequently given to searched for independently.
- Information about interpreting test results (28.57%), hereditary, genes and biomarkers (28.57%) and psychological support (30.61%) were give least often.

Gaps in Information obtained

- The largest gaps in information, where information was neither given to patients nor searched for independently were how to interpret test results (62.00%), and psychological/social support (56.00%).
- Participants were given most information either from healthcare professionals or independently for treatment options (78.00%) and disease cause (78.00%).
- Clinical trials (42.00%) was the topic that was most searched for independently following no information from health professionals.

Most trusted information sources

- Across all participants, information from the participants' hospital or clinic and from the non-profit or charitable organisations was near equal and was most trusted. Information from pharmaceutical companies was least trusted. This order of preference was the same for all sub-groups.

Section 7: Experience of care and support

Care coordination

- Overall the cohort had a care received score in the highest quintile, indicating very good care received. The scores for navigation and care coordination fell in the second highest quintile indicating good scores. The Total score and communication score were in the middle of the scale.

Care coordination – by general health

- There were no differences observed in any care coordination scales between those with higher general health and those with lower general health

Care coordination – by physical functioning

- There were no differences observed in any care coordination scales between those with higher physical functioning and those with lower physical functioning

Care coordination – by emotional well-being

- There were no differences observed in any care coordination scales between those with higher emotional well-being and those with lower emotional well-being

Care coordination– by social functioning

- Participants with higher social functioning had a significantly better outcome compared to those with lower social functioning for the Care coordination: Navigation scale. No other statistically significant differences were observed between these two groups for any Care Coordination scores

Care coordination – by hearing problems

- There were no differences observed in any care coordination scales between those with hearing problems and those with no hearing problems

Care coordination – by eye problems

- There were no differences observed in any care coordination scales between those with eye problems and those with no eye problems

Care coordination – by location

- There were no differences observed in any care coordination scales between participants that live in metropolitan areas and those that live in regional or rural areas.

Care coordination – by education

- There were no differences observed in any care coordination scales between participants with university qualifications and those with high school or trade qualifications

Care coordination – by SEIFA

- There were no differences observed in any care coordination scales between participants that live in areas with higher SEIFA scores and those that live in areas with lower SEIFA scores.

Care and support

- Participants were asked what care and support they had received throughout their experience. This question aims to investigate what services patients consider to be support and care services. The most common description of care and support was in the form of domestic and home care support from government services and NDIS (n=14, 28.00%), this was followed by participants describing that they did not receive any care and support in general (n =9, 18.00%) and not receiving significant support and care from the clinical setting (n=9, 18.00%). There were also seven participants (14.00%) that described receiving support from family and friends.
- In relation to sub-group variations, participants with high social functioning (30.00%) describes not receiving any care and support more frequently than the general population (18.00%).

Section 8: Experience of quality of life

Quality of life

- The most common impact on quality of life described by participants was poor mental health as a consequence of mitochondrial disease (n=19, 38.00%). There were also eight participants (16.00%) that noted poor mental health of family or friends (as carers) as a consequence of the disease. This was followed by a significant impact on family relationships and family dynamics (n=16, 32.00%) and withdrawing from activities with family and friends due to physical limitations (n=16, 32.00%). There were 13 participants (26.00%) that spoke about the need to access mental health services to maintain their quality of life, 12 participants (24.00%) that described that having days where physical limitations can be frustrating and eight participants (16.00%) that described limitations in travelling.
- In relation to sub-group variations, participants from metropolitan areas (26.67%) and participants with high physical function (27.27%) reported limitations of freedom to travel more frequently than the general population (16.00%), while participants from rural areas reported this less frequently (5.00%). Participants from rural areas (25.00%) described pleasure with maintaining hobbies and activities to overcome feelings of sadness or depression, more frequently than the general population (14.00%). Participants from low socio-economic areas (26.09%) and participants with a university degree (25.00%) described inability to participate in workforce to their level of expectation due to Mitochondrial disease, more frequently than the general population (14.00%). Participants with high school or trade education (34.62%) and those with high physical functioning (36.36%) reported having some days where physical limitations can be frustrating, more frequently than the general population (24.00%). Participants with high physical function (22.73%) reported little or no impact on family or friends' quality of life more frequently than the general population (12.00%).

Regular activities to maintain health

- The most common regular activity needed to maintain health reported by participants was having adequate rest to minimise fatigue (n=21, 42.00%). This was followed by having regular exercise (n=15, 30.00%) and eating a healthy/modified diet (n=10, 20.00%). There were seven participants (14.00%) that described taking prescription medication, six participants (12.00%) that considered taking supplements as an activity to maintain health and six participants (12.00%) that reported maintaining hobbies and activities in support of good mental health.
- In relation to sub-group variations, participants from low socio-economic areas (56.52%), those with high social function (60.00%) and low general health (57.14%) reported ensuring they have adequate rest to minimise fatigue, more frequently than the general population (42.00%). Participants with high physical function (50.00%) reported having regular exercise more frequently than the general population (30.00%) while those with low physical function (14.29%) and low general health (17.86%) reported this less frequently.

Impact on relationships

- The most common theme described by participants was a negative impact on personal relationships due to people withdrawing from relationships or not being able to understand (n=14, 28.00%) and this was followed by a negative impact on personal relationships due to social isolation (n=11, 22.00%). The next most common theme was a negative impact on personal relationships due to not being able to do all activities with family and friends (n=10, 20.00%). There were six participants (12.00%) that described a positive impact of strengthening relationships.
- In relation to sub-group variations, participants from low socio-economic areas (43.48%) and those with low social function (40.00%) reported a negative impact on personal relationships due to people withdrawing from relationships or not being able to understand, more frequently than the general population (28.00%), while those from high socio-economic areas (14.81) and high social function (10.00%) reported this less frequently. Participants from metropolitan areas (33.33%) and those with low emotional well-being (33.33%) reported a negative impact on personal relationships due to social isolation, more frequently than the general population (22.00%) while those from rural areas (10.00%) reported this less frequently. Participants from low socio-economic areas (30.43%) described a negative impact on personal relationships due to not being able to do all activities with family and friends, more frequently than the general population (20.00%). Participants with high physical function (27.27%) and high social function (35.00%) described no impact on personal relationships more frequently than the general population (16.00%), while those with low social function described this less frequently.
- Participants were also asked if their condition caused any additional burden on their family. The most common theme was there was an additional burden on family, but the participant did not articulate a specific reason why there was a burden (n=13, 26.00%). The next most common theme was there was a burden due to needing help with transport and driving due to vision impairment (n=8, 16.00%), followed by participants describing that there was no additional burden, that it is just part of their life as they know it (n=7, 14.00%).
- In relation to sub-group variations, participants with a university education (37.50%), those with low physical function (39.29%) and those with low social function (33.33%) reported there being an additional burden (no additional information) more frequently than the general population (26.00%), while those with high physical function (9.09%) and those with high social function (15.00%) reported this less frequently.

Anxiety and fear of progression

- The Fear of Progression questionnaire measures the level of anxiety people experience in relation to their conditions. The Fear of Progression questionnaire comprises a total score, with a higher score denoting increased anxiety. Overall the entire cohort had a median total score of 34.10, which is a score in the middle of the scale.

Fear of progression – by general health

- There was no difference observed in the fear of progression total score between participants that had higher general health compared to those with lower general health.

Fear of progression – by physical functioning

- There was no difference observed in the fear of progression total score between participants that had higher physical functioning compared to those with lower physical functioning.

Fear of progression – by emotional well-being

- There was no difference observed in the fear of progression total score between participants that had higher emotional well-being compared to those with lower emotional well-being.

Fear of progression – by social functioning

- There was no difference observed in the fear of progression total score between participants that had higher social functioning compared to those with lower social functioning.

Fear of progression – by hearing problems

- There was no difference observed in the fear of progression total score between participants that had hearing problems compared to those with no hearing problems.

Fear of progression – by eye problems

- There was no difference observed in the fear of progression total score between participants that had eye problems compared to those with no eye problems.

Fear of progression – by hearing problems

- There was no difference observed in the fear of progression total score between participants that had hearing problems compared to those with no hearing problems.

Fear of progression – by location

- There was no difference observed in the fear of progression total score between participants that live in metropolitan areas and those that live in regional or rural areas.

Fear of progression – by level of education

- There was no difference observed in the fear of progression total score between participants with university qualifications and those with high school or trade certificates.

Fear of progression – by SEIFA

- There was no difference observed in the fear of progression total score between participants that live in an area with a higher SEIFA score and those that live in a lower SEIFA score.

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

Expectations of future treatments

- The most common theme described by participants was that cost was an important consideration in relation to future treatments (n=18, 36.00%). This was followed by the need for effective treatments for mitochondrial disease, where participants may have also noted that there are no or limited treatments available (n=16. 36.00%). There were seven participants (14.00%) that described the need for clinical trials in mitochondrial disease and six participants (12.00%) that described the need for treatments that reduce muscle fatigue/improve muscle strength.
- In relation to sub-group variations, participants from low socio-economic areas (52.17%) described cost as a consideration more frequently than the general population (36.00%), while those from high socio-economic areas (25.93%) reported this less frequently. Participants from metropolitan areas (46.67%) and those with low emotional well-being (45.83%) reported the need for effective treatments for mitochondrial disease, more frequently than the general population (32.00%), while those from rural areas (15.00%) reported this less frequently.
- 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 were tiredness and fatigue, muscle symptoms and nervous system symptoms; the least important were underactive thyroid or parathyroid, and excess body hair.
- Participants were asked to rank what is important for them overall when they make decisions about treatment and care. The most important aspects were safety of treatment/weighing up risks and benefits, and severity of side effects. The least important were ability to stick to treatment, and including family in decision-making.
- 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 patient,s and access for all patients to all treatments and services; the least important was economic value to government.

Expectation of future information provision

- The most common theme was that participants described being satisfied with current information and therefore had no recommendation (n=11, 22.00%). There were nine participants (18.00%) that described the need for information about their specific type of mitochondrial disease, and nine participants (18.00%) that described the need for healthcare professionals to deliver accurate, comprehensive and honest information (including prognostic information. There were also six participants (12.00%) that described the need for centralised and reliable information.
- In relation to sub-group variations, participants with high general health (31.82%) described the need for healthcare professionals to deliver accurate, comprehensive and honest information (including prognostic information), more frequently than the general population (18.00%).

Expectation of future healthcare professional communication

- The most common theme was that participants recommend healthcare professional education in relation to mitochondrial disease and more understanding of the impact and implications of the condition (n=16, 32.00%). This was followed by the recommendation that healthcare professionals are more proactive and attentive (n=9, 18.00%). There were also nine participants (18.00%) that did not have a recommendation as they have been satisfied with communication. Where participants were satisfied with communication it was primarily because communication had been open communication. There were seven participants (14.00%) that recommended that healthcare professionals need to have more empathy.
- In relation to sub-group variations, participants from rural areas (45.00%) and those from low socio-economic areas (43.48%) recommended healthcare professional education in relation to mitochondrial disease and more understanding of the impact and implications of the condition, more frequently than the general population (32.00%).

Expectation of future care and support

- The most common recommendation was for centralised and coordinated care across specialists and allied health professionals (including more communication between doctors) (n=13, 26.00%). In a similar theme, there were also six participants (12.00%) that recommended caseworkers be employed to support patients navigate health, medical and emotional needs. This was followed by the recommendation for support groups to help patients noting that it is difficult due to the diversity within the patient population (n=7, 14.00%) and more equity in access to services and support for adults with rare disease (n=7, 14.00%).
- In relation to sub-group variations, participants with a university education (50.00%) and those with a hearing impairment (45.83%) recommended centralised and coordinated care across specialists and allied health professionals, more frequently than the general population (26.00%), while those with a high school or trade education (3.85%) recommended this less frequently.

What participants are grateful for in the Australian health system

- The most common theme was participants describing being grateful for Medicare in relation to access to specialists (n=17, 34.00%), followed by being grateful for the compassion and support shown by healthcare professionals (n=16, 32.00%). There were 10 participants (20.00%) that described being grateful for Medicare in relation to access to allied health professionals and seven participants (14.00%) described being grateful for their healthcare card and the financial relief it provides. Other aspects of the health system that participants spoke about being grateful for were subsidised diagnostic tests (n=6, 12.00%), government initiatives that support ongoing health and quality of life (for example NDIS, Better Start Program and At home nursing services) (n=6, 12.00%) and the quality of specialist expertise in Australia (n=5, 10.00%).
- In relation to sub-group variations, participants from rural areas (45.00%) described being grateful for Medicare (Access to specialists) more frequently than the general population (34.00%). Participants with a university education (45.83%), those with high physical function (54.55%), and those with high general health (50.00%) reported being grateful for the compassion and support shown by healthcare professionals more frequently than the general population (32.00%), while those with low physical function (17.86%) reported this less frequently. Participants from rural areas (25.00%), those with a hearing impairment (25.00%) and those with low physical function (25.00%) described being grateful for their healthcare card and the financial relief it provides, more frequently than the general population (14.00%), while there we no participants with high physical function (0.00%) that reported this.

Messages

- The most common message is to support more research (n=20, 40.00%), however this was a general statement with no specific area noted. The next most common theme was to provide more education to the healthcare professionals, particularly education about managing the condition (n=15, 30.00%), and this was followed by the message to increase awareness of mitochondrial disease among the community (n=12, 24.00%). There were 12 participants (24.00%) whose message is to provide more holistic and multidisciplinary/allied health care, and eight participants (16.00%) whose message is to improve treatments by following the example of other countries that have more advanced systems.
- In relation to sub-group variations, participants from rural areas (55.00%) called for more research more frequently than the general population (40.00%). Participants with a hearing impairment (41.67%) had the message to provide more education to the healthcare professionals, more frequently than the general population (30.00%). Participants with a university education (12.50%) called for more awareness less frequently than the general population (24.00%). Participants with a university education (33.33%) and those with a hearing impairment (37.50%) had the message to support more funding (in general), more frequently than the general population (22.00%), while those with a high school or trade education reported this less frequently (11.54%). Participants with high physical function (13.64%) had the message to provide more holistic and multidisciplinary/allied health care less frequently than the general population (24.00%).

Section 10: Advice to other patients and families

- Participants were asked what advice they would give to other people who are newly diagnosed with mitochondrial disease and their families. The most common advice is to ask questions and learn as much as you can (n=14, 28.00%). This was followed by the advice to talk to AMDF for information and support and to be part of the community (n=8, 16.00%), seek help (general) (n=8, 16.00%) and to find the right specialist as it is a rare disease and be comfortable with your healthcare team (n=8, 16.00%). There were seven participants (14.00%) whose advice is to seek help through psychological support, six participants (12.00%) whose advice is to share your story to help others and six participants (12.00%) whose advice is to be hopeful.

Section 1 Introduction and methods

Section 1: Introduction and methodology

Summary

- Mitochondrial disease is a heterogenous group of diseases that have dysfunctional mitochondrial respiratory changes that are caused by mutations to nuclear or mitochondrial DNA. The disease may affect single organ or may affect multiple organs, and usually affect organs that have the highest energy needs such as muscles, brain, eyes and heart.
- The prevalence of mitochondrial disease is estimated at 11.5 per 100,000, however this may underestimate the prevalence with reports of one in 200 healthy births having a mitochondrial DNA mutation.
- Patient Experience, Expectations and Knowledge (PEEK) is a research program developed by the International Centre for Community-Driven Research (ICCDR). The aim of PEEK is to conduct patient experience studies across several disease areas using a protocol that will allow for comparisons over time (both quantitative and qualitative components). PEEK studies give us a clear picture and historical record of what it is like to be a patient at a given point in time, and by asking patients about their expectations, PEEK studies give us a way forward to support patients and their families with treatments, information and care.
- In this PEEK study, 50 people with mitochondrial disease or their carers, throughout Australia participated in the study that included a structured interview and quantitative questionnaire. This study in mitochondrial disease is therefore the largest mixed methodology study in Australia. 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.

Introduction

Mitochondrial disease is a heterogenous group of diseases that have dysfunctional mitochondrial respiratory changes that are caused by mutations to nuclear or mitochondrial DNA[1]. The disease may affect single organ or may affect multiple organs[1], and usually affect organs that have the highest energy needs such as muscles, brain, eyes and heart[2]. More commonly described clinical subtypes of mitochondrial disease include:[2]

- Chronic progressive external ophthalmoplegia (CPEO) Infantile myopathy and lactic acidosis (fatal and non-fatal forms),
- Kearns-Sayre syndrome (KSS)
- Leber hereditary optic neuropathy (LHON)
- Leigh syndrome (LS)
- Mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes (MELAS)
- Myoclonic epilepsy with ragged-red fibres (MERRF)
- Neurogenic weakness with ataxia and retinitis pigmentosa (NARP)
- Pearson Syndome

The prevalence of mitochondrial disease is estimated at 11.5 per 100,000[1], however this may underestimate the prevalence with reports of one in 200 healthy births having a mitochondrial DNA mutation[3].

Patient Experience, Expectations and Knowledge (PEEK)

Patient Experience, Expectations and Knowledge (PEEK) is a research program developed by the International Centre for Community-Driven Research (ICCDR). The aim of PEEK is to conduct patient experience studies across several disease areas using a protocol that will allow for comparisons over time (both quantitative and qualitative components). PEEK studies give us a clear picture and historical record of what it is like to be a patient at a given point in time, and by asking patients about their expectations, PEEK studies give us a way forward to support patients and their families with treatments, information and care.

The research protocol used in PEEK studies is independently driven by ICCDR. PEEK studies include a quantitative and qualitative component. The quantitative component is based on a series of validated tools. The qualitative component is the result of two years of protocol testing by ICCDR to develop a structured interview that solicits patient experience data and provides patients with the opportunity to provide advice on what they would like to see in relation to future treatment, information and care. The structured interview has also been designed so that the outcomes of PEEK studies can inform policy, research, care, information, supportive care services and advocacy efforts.

Methodology

Participants

To be eligible for the study, participants needed to have been diagnosed with mitochondrial disease or be a carer to someone with mitochondrial disease, 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. Recruitment commenced on 19 April 2018 and the study closed for recruitment on 15 May 2018. Participants were recruited via email and social media through ICCDR and study partner the Australian Mitochondrial Disease Foundation, who sponsored the study and also sent information via electronic direct mail.

Ethics

Ethics approval for this study was granted (as a low or negligible risk research study) by the Centre for Community-Driven Research Ethics Committee (Reference CS_Q4_03).

Data collection

Data for the online questionnaire was collected using Zoho Survey (Zoho Corporation Pvt. Ltd. Pleasanton, California, USA, www.zoho.com/survey). Participants completed the survey between 19 April 2018 and 18 May 2018.

There were four researchers who conducted telephone interviews and used standardised prompts throughout the interview. The interviews were recorded and transcribed verbatim. Identifying names and locations were not included in the transcript. All transcripts were checked against the original recording for quality assurance.

Interview data was collected from 27 April 2018 to 23 May 2018.

Online questionnaire (quantitative)

The online questionnaire consisted of the 36-Item Short Form Health Survey (SF36) (RAND Health)[4], a modified Cancer Care Coordination Questionnaire for Patients (CCCQ) (Young et al 2011)[5], the Short Fear of Progression Questionnaire (FOP12) (Hinz et al)[6], and the Partners in Health version 2 (PIH) (Petov 2010)[7]. In addition investigator derived questions about demographics, diagnosis, treatment received and future treatment decisions making were included.

Structured Interview (qualitative)

Interviews were conducted via telephone by a registered nurse or researcher with a background in psychology, who were trained in qualitative research. The first set of interview questions guided the patient through their whole experience from when symptoms were noticed up to the present day.

The next set of questions allowed patients to reflect on what they would like to see in the future in relation to treatment and care, and asked them what their messages to decision-makers would be about the care and treatment patients with their condition receive. The interview also asks patients about the advice they would give to others recently diagnosed with their condition or disease. All interviews were recorded and transcribed verbatim.

Questionnaire analysis

Statistical analysis was conducted using R included in the packages "car", "dplyr" and "ggplot2" (R 3.3.3 GUI 1.69 Mavericks build (7328). The aim of the statistical analysis of the SF36, CCCQ, FOP12, and PIH responses was to identify variations by general health status (SF36 general health, SF36 physical functioning, SF36 social functioning, SF36 emotional well-being and SF36 social functioning), location, education status and Socio-economic Indexes for Areas (SEIFA). Global scales and sub scales were calculated according to reported instructions[4-7]. For all comparisons, a two-sample t-test was used when assumptions for normality and variance were met, or when assumptions were not met, a Wilcoxon rank sum test with continuity correction was used.

Questions where participants were asked to rank preferences were analysed using weighted averages. Weights were applied in reverse, the most preferred option was given the largest weight equal to the number of options, the least preferred option was given the lowest weight of 1.

Structured interviews analysis

A content analysis was conducted using conventional analysis to identify major themes from structured interviews. Text from the interviews were read line-by-line by the lead researcher and then imported into NVivo 8 (QSR International). Each question within the interview was individually analysed. Initial categories and definitions were identified and registered in NVivo. The minimum coded unit was a sentence however there were also paragraphs and phrases that were coded as a unit.

A second researcher verified the codes and definitions, and the text was coded until full agreement was reached using the process of consensual validation. Where a theme occurred less than 5 times it was not included in the study discussion, however these were reported in tables and graphs. A sub-group analysis was also conducted. Where there was a variation of more than 10 percent in any sub-group compared to the general population (cohort), these were reported.

Data analysis and final reporting was completed on 10 July 2018.

Position of this study

A search was conducted in Pubmed to identify mitochondrial disease quality of life or patient experience studies that had been conducted in the past ten years in developed countries (Table 1.1).

Ten studies were identified that included between six and 231 participants with mitochondrial disease or their carers. All of the studies used quantitative methods, three studies were part of clinical trials[8-10], three studies focused on parent and carer experience[11-13], two focused on physical activity[14,15], a single study of quality of life[16] and one of fatigue[15].

In this PEEK study, 39 people with mitochondrial disease and 11 parents or carers of people with mitochondrial disease throughout Australia participated in the study that included a structured interview and quantitative questionnaire. This study in mitochondrial disease is therefore the largest mixed methodology study in Australia. 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.

Table 1.1: Comparative studies

Author/Country/Year	Number participants	Participant type	Study type	QL	Function	Symptoms	Anxiety/Depression	Behaviour	Diagnosis	Burden	Resources	Study Focus
Glover et al/Canada/2010[9]	30	Individual	Quantitative	✓	✓							Clinical trial
Martinelli et al/Italy/2012[8]	10	Individual	Quantitative	✓	✓							Clinical trial
Eom & Lee/Korea/2017[11]	70	Parent/Carer	Quantitative		✓	✓	✓	✓	✓			Neurodevelopment and parent stress
Kim et al/Korea/2010[12]	33	Parent/Carer	Quantitative	✓			✓			✓		Caregiver burden
Verhaak et al/Netherlands/2016[16]	72	Individual	Quantitative	✓	✓	✓	✓					QL
Martens et al/Netherlands/2014[15]	6	Individual	Quantitative	✓				✓				Physical activity (function)
Gorman et al/UK/2015[17]	132	Individual	Quantitative			✓	✓					Fatigue
Bates et al/UK/2013[14]	10	Individual	Quantitative	✓		✓						Physical activity (intervention)
Senger et al/USA/2016[13]	231	Parent/Carer	Quantitative				✓				✓	Parent experience
Enns et al/USA/2012[10]	14	Individual	Quantitative	✓	✓							Clinical trial

Abbreviations

CCDR	Centre for Community-Driven Research
dF	Degrees of Freedom. The number of values in the final calculation of a statistic that are free to vary.
IQR	Interquartile range. A measure of statistical dispersion, being equal to the difference between 75th and 25th percentiles, or between upper and lower quartiles.
FOP	Fear of Progression. Tool to measure anxiety related to progression.
MS	Mean of Squares. Estimates of variance across groups
SD	Standard Deviation. A quantity expressing by how much the members of a group differ from the mean value for the group.
SF 36	Short Form Health Survey 36
t	t-Statistic. Size of the difference relative to the variation in your sample data.
PEEK	Patient Experience, Expectations and Knowledge
PIH	Partners in Health
p	Probability value. A small *p*-value (typically ≤ 0.05) indicates strong. A large *p*-value (> 0.05) indicates weak evidence.
QoL	Quality of LIfe
W	The W statistic is the test value from the Wilcoxon Rank sum test. The theoretical range of W is between 0 and (number in group one)x(number in group 2). When W=0, the two groups are exactly the same.

References

1. Chinnery PF. Mitochondrial Disorders Overview. In: Adam MP, Ardinger HH, Pagon RA, et al., eds. GeneReviews((R)). Seattle (WA); 1993 (Updated 2014).

2. Pfeffer G, Majamaa K, Turnbull DM, Thorburn D, Chinnery PF. Treatment for mitochondrial disorders. *Cochrane Database Syst Rev* 2012; (4): CD004426.

3. Elliott HR, Samuels DC, Eden JA, Relton CL, Chinnery PF. Pathogenic mitochondrial DNA mutations are common in the general population. *Am J Hum Genet* 2008; **83**(2): 254-60.

4. 36-Item Short Form Survey (SF-36) Scoring Instructions. n.d. https://www.rand.org/health/surveys_tools/mos/36-item-short-form/scoring.html (accessed 10 February 2017.

5. Young JM, Walsh J, Butow PN, Solomon MJ, Shaw J. Measuring cancer care coordination: development and validation of a questionnaire for patients. *BMC Cancer* 2011; **11**: 298.

6. Hinz A, Mehnert A, Ernst J, Herschbach P, Schulte T. Fear of progression in patients 6 months after cancer rehabilitation-a- validation study of the fear of progression questionnaire FoP-Q-12. *Support Care Cancer* 2015; **23**(6): 1579-87.

7. Petkov J, Harvey P, Battersby M. The internal consistency and construct validity of the partners in health scale: validation of a patient rated chronic condition self-management measure. *Qual Life Res* 2010; **19**(7): 1079-85.

8. Martinelli D, Catteruccia M, Piemonte F, et al. EPI-743 reverses the progression of the pediatric mitochondrial disease--genetically defined Leigh Syndrome. *Mol Genet Metab* 2012; **107**(3): 383-8.

9. Glover EI, Martin J, Maher A, Thornhill RE, Moran GR, Tarnopolsky MA. A randomized trial of coenzyme Q10 in mitochondrial disorders. *Muscle Nerve* 2010; **42**(5): 739-48.

10. Enns GM, Kinsman SL, Perlman SL, et al. Initial experience in the treatment of inherited mitochondrial disease with EPI-743. *Mol Genet Metab* 2012; **105**(1): 91-102.

11. Eom S, Lee YM. Preliminary Study of Neurodevelopmental Outcomes and Parenting Stress in Pediatric Mitochondrial Disease. *Pediatr Neurol* 2017; **71**: 43-9 e1.

12. Kim KR, Lee E, Namkoong K, Lee YM, Lee JS, Kim HD. Caregiver's burden and quality of life in mitochondrial disease. *Pediatr Neurol* 2010; **42**(4): 271-6.

13. Senger BA, Ward LD, Barbosa-Leiker C, Bindler RC. The Parent Experience of Caring for a Child with Mitochondrial Disease. *J Pediatr Nurs* 2016; **31**(1): 32-41.

14. Bates MG, Newman JH, Jakovljevic DG, et al. Defining cardiac adaptations and safety of endurance training in patients with m.3243A>G-related mitochondrial disease. *Int J Cardiol* 2013; **168**(4): 3599-608.

15. Martens AM, Gorter H, Wassink RG, Rietman H. Physical activity of children with a mitochondrial disease compared to children who are healthy. *Pediatr Phys Ther* 2014; **26**(1): 19-26.

16. Verhaak C, de Laat P, Koene S, et al. Quality of life, fatigue and mental health in patients with the m.3243A > G mutation and its correlates with genetic characteristics and disease manifestation. *Orphanet J Rare Dis* 2016; **11**: 25.

17. Gorman GS, Elson JL, Newman J, et al. Perceived fatigue is highly prevalent and debilitating in patients with mitochondrial disease. *Neuromuscul Disord* 2015; **25**(7): 563-6.

Section 2 Demographics

Section 2: Demographics and study population characteristics

Demographics

- Fifty participants from Australia were recruited into the study, including 44 (88.00%) participants with mitochondrial disease and 6 (12.00%) carers of people with mitochondrial disease. There were an additional five participants that were both a patient and carer, however they responded to the questionnaire and interview as a patient rather than a carer.
- The majority of participants were from NSW (n=18, 36.00%), Victoria (n=12, n=24.00%), and Queensland (n=10, 20.00%), and most live in major cities (n=30, 60.00%).
- Thirty-seven females (74.00%) and 13 males (26.00%) participated.

Baseline Heath – SF36 score

The Short Form Health Survey 36 (SF36) measures baseline health, or the general health of an individual. A higher score indicates better baseline health.

- The overall scores for the cohort for emotional well-being were in the second highest quintile indicating very good baseline health.
- The overall scores for the cohort for pain were in the middle quintile indicating moderate baseline health.
- The overall scores for the cohort for physical functioning, role functioning/emotional, energy/fatigue, social functioning, general health, and health change were in the second lowest quintile indicating poor baseline health.
- The overall score for role functioning /physical were in the lowest quintile indicating very poor baseline health.

SF36 scores by general health

- Those with higher general health scored significantly better compared to lower general health for the physical functioning, emotional well-being, social functioning, role functioning/emotional, energy/fatigue, pain and health change scales.

SF36 scores by physical functioning

- Those with higher physical functioning scored significantly better compared to those with lower physical functioning for the SF36 role functioning/physical, energy/fatigue, social functioning, pain, general health and health change subscales.

SF36 scores by emotional well-being

- Those with higher emotional well-being scored significantly better compared to those with lower emotional well-being for the SF36 role functioning/physical, role functioning/emotional, social functioning, pain, general health and health change subscales.

SF36 scores by social functioning

- Those with higher social functioning scored significantly better compared to those with lower social functioning for the SF36 physical functioning, role functioning/physical, role functioning/emotional, emotional well-being, energy/fatigue, pain, general health and health change subscales.

SF36 scores by hearing problems

- No significant differences were observed between those with hearing problems and those with no hearing problems for any of the SF36 subscales

SF36 scores by eye problems

- No significant differences were observed between those with eye problems and those with no eye problems for any of the SF36 subscales

SF36 scores by location

- No significant differences were observed between those that live in metropolitan areas and those that live in regional or rural areas for any of the SF36 subscales

SF36 scores by education

- No significant differences were observed between those with a university qualification and those with high school or trade qualifications for any of the SF36 subscales.

SF36 scores by Socio-Economic Indexes For Areas (SEIFA)

- No significant differences were observed between those that live in an area with a higher SEIFA score (more advantaged) and those that live in an area with a lower SEIFA score for any of the SF36 subscales.

Table 2.1: Demographics

Characteristic	n=	Percentage of participants
Participant type n=50		
Person with mitochondrial disease	44	88.00
Parent/care of someone with mitochondrial disease	6	12.00
Location: State n=50		
New South Wales	18	36.00
Victoria	12	24.00
Queensland	10	20.00
South Australia	5	10.00
Tasmania	3	6.00
Western Australia	2	4.00
Geographical location n=50		
Major City	30	60.00
Inner Regional	13	26.00
Outer Regional	6	12.00
Remote	1	2.00
Social Economic Indexes for Areas n=50 (1= most disadvantaged)		
1	3	6.00
2	2	4.00
3	4	8.00
4	5	10.00
5	3	6.00
6	6	12.00
7	3	6.00
8	8	16.00
9	13	26.00
10	3	6.00
Gender n=50		
Female	37	74.00
Male	13	26.00
Age of participant n=50		
25-34	6	12.00
35-44	8	16.00
45-54	13	26.00
55-64	13	26.00
65-74	5	10.00
75-84	5	10.00
Race n=49		
Caucasian/White	47	95.92
Australian	1	2.04
Portuguese	1	2.04

Table 2.1: Demographics (continued)

Characteristic	n=	Percentage of Participants
Highest level of education obtained n=50		
Less than High School degree	2	4.00
High school degree or equivalent	13	26.00
Some College but no degree	10	20.00
Trade	1	2.00
Associate degree	3	6.00
Bachelor Degree	9	18.00
Graduate degree	12	24.00
Employment status (can choose more than one category) n=50		
Currently receiving Centrelink support	11	22.00
Disabled, not able to work	17	34.00
Employed, working full time	10	20.00
Employed, working part time	6	12.00
Full/part time carer	4	8.00
Full/part time study	2	4.00
Not employed, looking for work	1	2.00
Retired	10	20.00
My health Record Access n=50		
No	26	52.00
Yes	5	10.00
I Don't know what "My health record" is	11	22.00
Not Sure	8	16.00
My health Record Use n=5		
Good	1	20.00
Acceptable	1	20.00
Poor	2	40.00
Very Poor	1	20.00
Carer status n=50		
Carer to children	14	28.00
Carer to spouse	2	4.00
Carer to parents	2	4.00
I am not a carer	32	64.00

Demographics

Fifty participants from Australia were recruited into the study, including 44 (88.00%) participants with mitochondrial disease and 6 (12.00%) carers of people with mitochondrial disease. There were an additional five participants that were both a patient and carer, however they responded to the questionnaire and interview as a patient rather than a carer. The majority of participants were from NSW (n=18, 36.00%), Victoria (n=12, n=24.00%), and Queensland (n=10, 20.00%), and most live in major cities (n=30, 60.00%). Thirty-seven females (74.00%) and 13 males (26.00%) participated. Demographics of participants are listed in Table 2.1

Disease description

Twenty-four (48.00%) participants described their disease as a syndrome with mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes (MELAS) being the most commonly described syndrome. Other participants described their mitochondrial disease by their main symptoms (n=11, 22.00%), five (n=10.00%) described a deficiency, two (4.00%) described a mutation, two (4.00%) had a mixed description and six (12.00%) described mitochondrial disease in general.

Table 2.2: Mitochondrial disease description

Disease description	Number	Percentage of Participants
Syndrome		
CPEO	2	4.00
KSS	3	6.00
Leigh's syndrome	2	4.00
LHON	3	6.00
MELAS	11	22.00
MELAS/NARP/Leigh like	1	2.00
MERRF	1	2.00
NARP/MERRF	1	2.00
Symptoms		
General mitochondrial disease diagnosis, described main symptoms	11	22.00
No description		
General mitochondrial disease diagnosis	6	12.00
Deficiency		
Alpha-methylacyl-CoA racemase deficiency	1	2.00
Complex I and IV deficiency	2	4.00
Complex IV deficiency	1	2.00
COX deficiency	1	2.00
Mutation		
m.3302 A>G	1	2.00
MT 3113 A-G	1	2.00
Mixed		
MELAS, m.3233 A>G	1	2.00
Complex IV deficiency/ Leigh's Disease	1	2.00

Subgroup analysis

Subgroup analysis are included throughout the study and the subgroups are listed in Table 2.2. The Short Form Health Survey 36 (SF36) measures baseline health, or the general health of an individual. Four of the nine subscales have been used in the subgroup analysis, **general health**, those with a higher than average score for the cohort in the SF36 general health scale (n=22, 44.00%) compared to those with an average or less score (n=28, 56.00%); **physical health**, those that scored above average for the cohort in the SF36 Physical functioning scale (n=22, 44.00%) compared to those that scored average or below (n=28, 56.00%); **emotional well-being**, those that scored above average for the cohort in the SF36 Emotional well-being scale (n=26, 52.00%) compared to those that scored average or below (n=24, 48.00%); **social functioning**, those that scored above average for the cohort in the SF36 Social functioning scale (n=20, 40.00%) compared to those that scored average or below (n=30, 60.00%). Those that had **hearing problems** (n=24, 48.00%) were compared to those that had no hearing problems (n=26, 52.00%), and those with **eye problems** (for example drooping eyelids, inability to move eyes and vision loss) (n=34, 68.00%) were compared to those with no eye problems) n=16, 32.00%). The **location** of participants was evaluated by

postcode using the Australian Statistical Geography Maps (ASGS) Remoteness areas accessed from DoctorConnect (doctorconnect.gov.au), those living in a metropolitan area (n=30, 60.00%) were compared to those living in regional/rural areas (n= 20, 40.00%). Comparisons were made by **education** status, those with university degree (n= 24, 48.00%) and those with high school or trade (n=26, 52.00%); and by Socio-economic Indexes for Areas (**SEIFA**) (www.abs.gov.au), a higher score denotes a higher level of advantage. Those with a higher SEIFA score of 7-10 (n=27, 54.00%) compared to those with a lower SEIFA score of 1-6 (n=23, 46.00%).

Table 2.3: Demographics used for sub-group analysis

Characteristic	n=	Percentage of participants
SF36 General health		
Higher general health	22	56.00
Lower general health	28	44.00
SF36 Physical functioning		
Higher physical functioning	22	56.00
Lower physical functioning	28	44.00
SF36 Emotional well-being		
Higher emotional well-being	26	52.00
Lower emotional well-being	24	48.00
SF36 Social functioning		
Higher social functioning	20	40.00
Lower social functioning	30	60.00
Hearing problems		
Hearing problems	24	48.00
No hearing problems	26	52.00
Eye problems		
Eye problems	34	68.00
No eye problems	16	32.00
Location		
Metropolitan	30	60.00
Regional/rural	20	40.00
Education		
Trade or high school	26	52.00
University	24	48.00
Socio-Economic Indexes for Areas (SEIFA)		
Higher SEIFA	27	54.00
Lower SEIFA	23	46.00

Co-morbidities

Participants noted other conditions they have, the most commonly reported conditions were chronic pain (n=27, 54.00%), followed by sleep problems (n=21, 42.00%), anxiety (n=21, 42.00%) and depression (n=20, 41.00%). Only one participant noted that they had no other condition.

Table 2.4: Co-morbidities

Co-morbidities	N=	Percentage of participants
Anxiety	21	42.00
Arrhythmias	6	12.00
Arthritis	10	20.00
Asthma	12	24.00
Cardiovascular problems	5	10.00
Chronic pain	27	54.00
CNS problems	6	12.00
COPD	4	8.00
Depression	20	40.00
Diabetes	10	20.00
Eye/vision problems	4	8.00
Gastrointestinal	4	8.00
Hypertension	10	20.00
Musculoskeletal problems	13	26.00
Sleep problems	21	42.00
Other	15	

Baseline health

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

Summary statistics for the entire cohort are displayed alongside the possible range of each scale in Table 2.5, where the scale has a normal distribution mean and SD are used as a central measure, otherwise the median and IQR are used.

The overall scores for the cohort were in the second highest quintile for emotional well-being (median = 68.00, IQR=20.00) indicating good scores for the cohort. The scores for pain were in the middle quintile, (Median = 45.00, IQR= 45.00) indicating moderate scores, the scores for physical functioning (Median =32.50, IQR = 35.75), role functioning/emotional (Median = 33.33, IQR = 100.00), energy/fatigue (Mean = 22.50, SD = 17.71), social functioning (Median = 37.50, IQR = 25.00), general health (Median = 25.00, IQR = 20.00), and health change (Median = 25.00, IQR =25.00) were in the second lowest quintile indicating poor baseline health. The median score for role functioning /physical (Median = 0.00, IQR = 0.00) were in the lowest quintile indicating very poor baseline health.

Comparisons of SF36 have been made based on general health (Figures 2.1 to 2.8, Tables 2.6 to 2.7), physical functioning (Figures 2.9 to 2.16, Tables 2.8 to 2.9), emotional well-being (Figures 2.17 to 2.24, Tables 2.10 to 2.11), social functioning, (Figures 2.25 to 2.32, Tables 2.12 to 2.13), hearing problems (Figures 2.33 to 2.41, Tables 2.14 to 2.15), eye problems (Figures 2.42 to 2.50, Tables 2.16 to 2.17), location (Figures 2.51 to 2.59, Tables 2.18 to 2.19), education (Figures 2.60 to 2.68, Tables 2.20 to 2.21), and SEIFA (Figures 2.69 to 2.77, Tables 2.22 to 2.23).

Table 2.5: SF36 summary statistics all participants

SF36 scale	Mean	SD	Median	IQR	Possible range
Physical functioning	35.70	30.76	32.50	43.75	0-100
Role functioning/physical	12.50	26.85	0.00	0.00	0-100
Role functioning/emotional	43.33	45.80	33.33	100.00	0-100
Energy/fatigue*	22.50	17.71	25.00	23.75	0-100
Emotional well-being	64.00	16.54	68.00	20.00	0-100
Social functioning	39.75	26.21	37.50	25.00	0-100
Pain	46.90	28.43	45.00	45.00	0-100
General health	28.00	18.82	25.00	20.00	0-100
Health change	35.50	24.79	25.00	25.00	0-100

*Normal distribution use mean and SD

Comparisons of SF36 sub scales by general health

Comparisons of SF36 sub scales were made between participants with higher general health and lower general health. Comparisons between higher general health and lower general health for the SF36 general health subscale were excluded due to selection bias. Boxplots of each SF36 scale by general health are displayed in Figures 2.1-2.8.

A two-sample t-test was used when assumptions for normality and variance were met (Table 2.6), or when assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 2.7). A two sample t-test indicated that the mean score for the SF36 emotional well-being scale was significantly higher for those with higher general health (Mean =72.36, SD = 11.83) compared to those with lower general health (Mean = 57.43, SD = 16.91) [t(48) = 3.52, p=0.0010], and the mean score for social functioning scale was significantly higher for those with higher general health (Mean =53.98, SD = 25.70) compared to those with lower general health (Mean = 28.57, SD = 20.93) [t(48) = 3.85, p=0.0003}.

A Wilcoxon rank sum test with continuity correction indicated a those with higher general health (Median = 52.50, IQR =57.50) had a significantly better outcome compared to those with lower general health (Median = 22.50, IQR = 32.50) for SF36 physical functioning scale [W=457.00, p=0.035]; those with higher general health (Median = 83.33, IQR =91.67) had a significantly better outcome compared to those with lower general health (Median = 0.00, IQR = 66.67) for SF36 functioning/emotional scale [W=425.50, p=0.0131]; those with higher general health (Median = 27.50, IQR =20.00) had a significantly better outcome compared to those with lower general health (Median = 10.00, IQR = 21.25) for SF36 energy/fatigue scale [W=440.00, p=0.0097]; those with higher general health (Median = 57.50, IQR =41.88) had a significantly better outcome compared to those with lower general health (Median = 32.50, IQR = 25.00) for SF36 pain scale [W=451.50, p=0.0049]; and those with higher general health (Median = 37.50, IQR =25.00) had a significantly better outcome compared to those with lower general health (Median = 25.00, IQR = 12.50) for SF36 health change scale [W=421.00, p=0.0179].

No significant differences were observed for physical functioning, role limitations/physical.

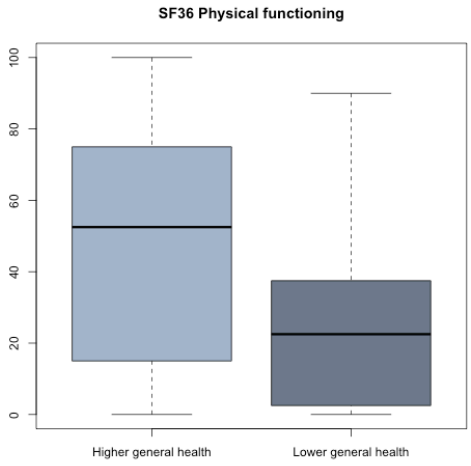


Figure 2.1: Boxplot of SF36 physical functioning by general health

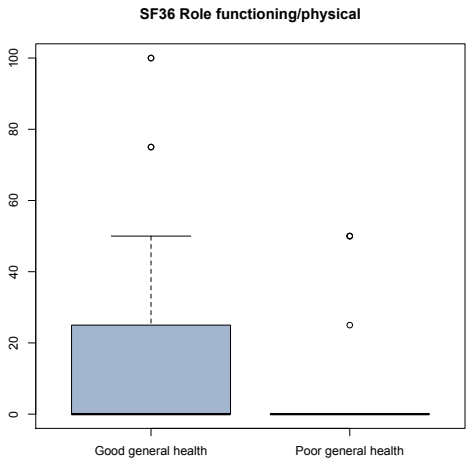


Figure 2.2: Boxplot of SF36 role limitations due to physical health by general health

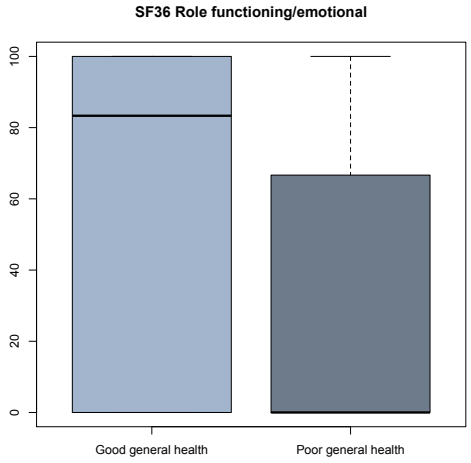


Figure 2.3: Boxplot of SF36 role limitations due to emotional problems by general health

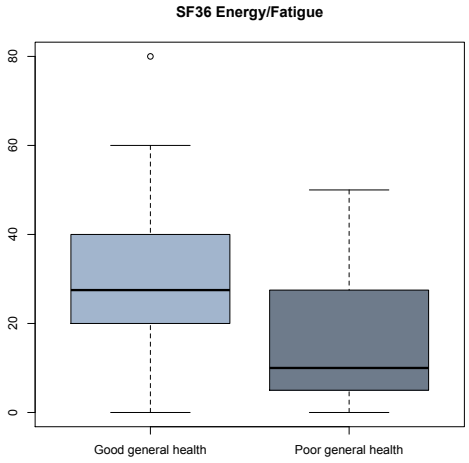
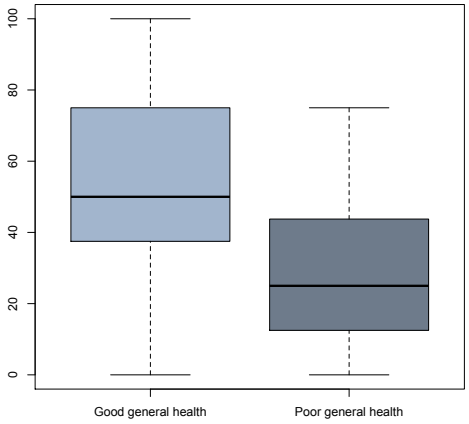


Figure 2.4: Boxplot of SF36 energy/fatigue by general health

Figure 2.5: Boxplot of SF36 emotional well-being by general health



2.6: Boxplot of SF36 social functioning by general health

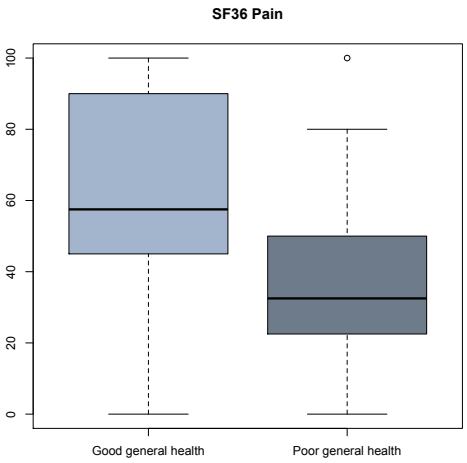


Figure 2.7: Boxplot of SF36 pain by general health

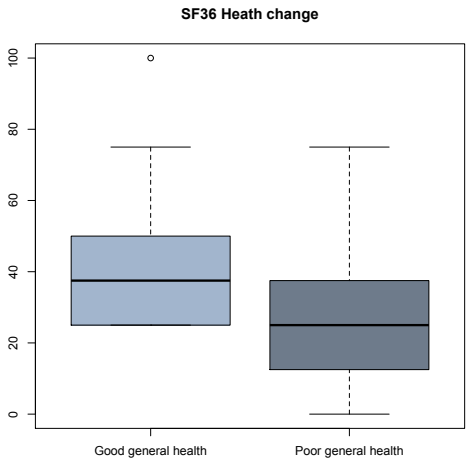


Figure 2.8: Boxplot of SF36 health change by general health

Table 2.6 Summary statistics and t-test SF36 scales by general health

SF36 by General Health	Group	Count	Mean	SD	t	dF	p
Emotional well-being	Higher general health	22	72.36	11.83	3.52	48	0.0010*
	Lower general health	28	57.43	16.91			
Social functioning	Higher general health	22	53.98	25.70	3.85	48	0.0003*
	Lower general health	28	28.57	20.93			

* Statistically significant at p<0.05

Table 2.7: Summary statistics and Wilcoxon rank sum test SF36 scales by general health

SF36 scale by general health	Group	Count	Median	IQR	W	p
Physical functioning	Higher general health	22	52.50	57.50	457.00	0.0035*
	Lower general health	28	22.50	32.50		
Role functioning/physical	Higher general health	22	0.00	25.00	367.50	0.1140
	Lower general health	28	0.00	0.00		
Role functioning/emotional	Higher general health	22	83.33	91.67	425.50	0.0131*
	Lower general health	28	0.00	66.67		
Energy/Fatigue	Higher general health	22	27.50	20.00	440.00	0.0097*
	Lower general health	28	10.00	21.25		
Pain	Higher general health	22	57.50	41.88	451.50	0.0049*
	Lower general health	28	32.50	25.00		
Health change	Higher general health	22	37.50	25.00	421.00	0.0179*
	Lower general health	28	25.00	12.50		

* Statistically significant at p<0.05

Comparisons of SF36 sub scales by physical functioning

Comparisons of SF36 subscales were made between those that had above average for the group SF36 physical functioning scores (higher physical functioning) compared to those with average or below scores (lower physical functioning). Comparisons between higher physical functioning and lower physical functioning for the SF36 physical functioning subscale were excluded due to selection bias. Boxplots of each SF36 scale by metastatic status are displayed in Figures 2.9-2.16. A two-sample t-test was used when assumptions for normality and variance were met (Table 2.8), or when assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 2.9).

A two sample t-test indicated that those with higher physical functioning (mean=35.68, SD=20.31) had significantly better baseline health compared to those with lower physical functioning (mean=21.96, sd=15.36) for the SF36 general health scale [t(48)=2.72, p=0.0090).

A Wilcoxon rank sum test with continuity correction indicated that those with higher physical functioning (Median =0.00, IQR = 43.75) had significantly better baseline health for role functioning/physical [W=410.00, p=0.0061] compared to those with lower physical functioning (Median =0.00, IQR = 0.00); and those with higher physical functioning (Median =25.00, IQR = 18.75) had significantly better baseline health for energy/fatigue [W=420.50, p=0.0276] compared to those with lower physical functioning (Median =12.50, 25.00); and those with higher physical functioning (Median =50.00, IQR = 25.00) had significantly better baseline health for social functioning [W=494.50, p=0.0002] compared to those with lower physical functioning (Median =25.00, IQR = 25.00); and those with higher physical functioning (Median =57.50, IQR = 22.50) had significantly better baseline health for pain [W=439.00, p=0.0102] compared to those with lower physical functioning (Median =32.50, IQR = 25.00); and those with higher physical functioning (Median =37.50, IQR = 50.00) had significantly better baseline health for general health [W=438.00, p=0.0064] compared to

those with lower physical functioning (Median =25.00, IQR = 12.50).

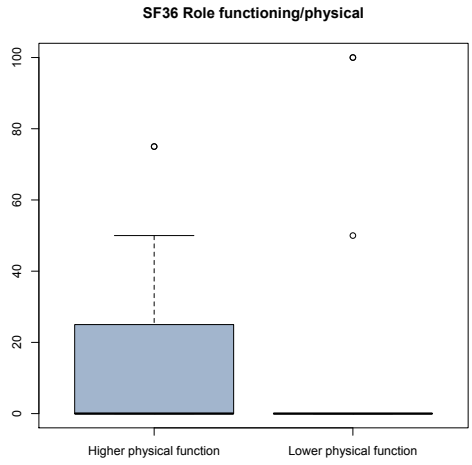


Figure 2.9: Boxplot of SF36 role limitations due to physical health by physical functioning

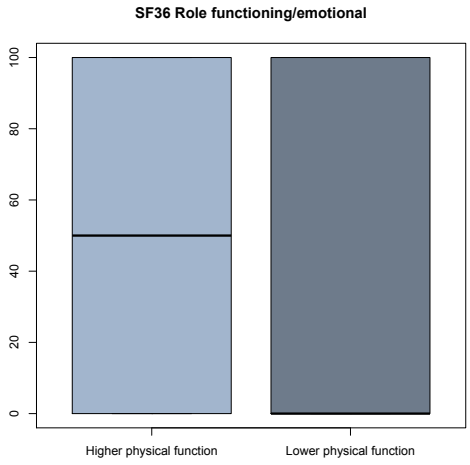


Figure 2.10: Boxplot of SF36 role limitations due to emotional problems by physical functioning

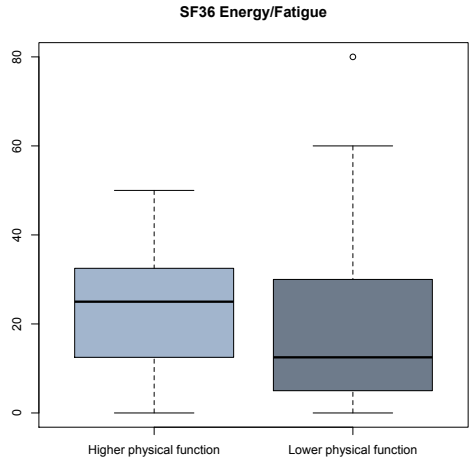


Figure 2.11: Boxplot of SF36 energy/fatigue by physical functioning

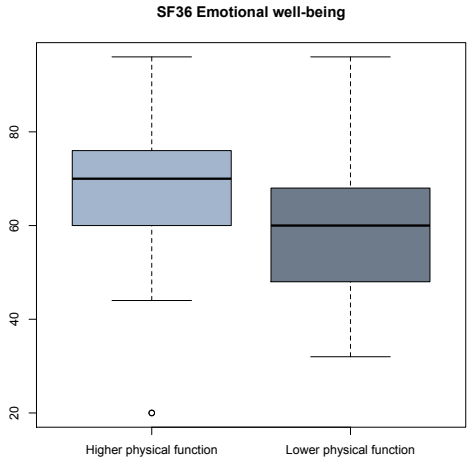


Figure 2.12: Boxplot of SF36 emotional well-being by physical functioning

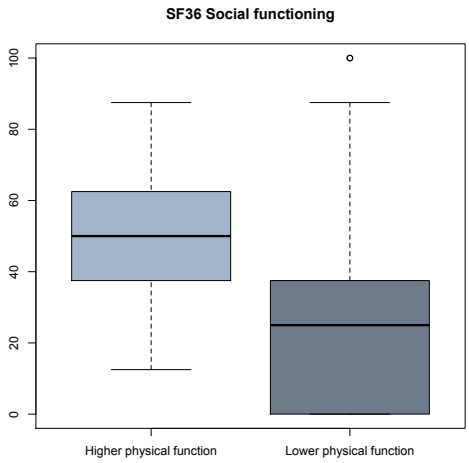


Figure 2.13: Boxplot of SF36 social functioning by physical functioning

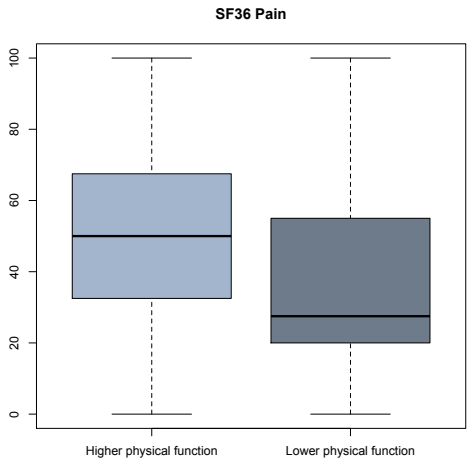


Figure 2.14: Boxplot of SF36 pain by physical functioning

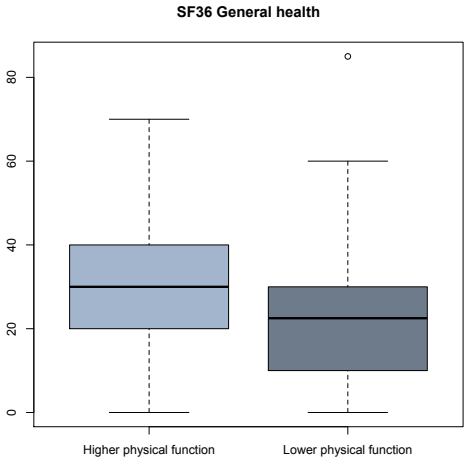


Figure 2.15: Boxplot of SF36 general health by physical functioning

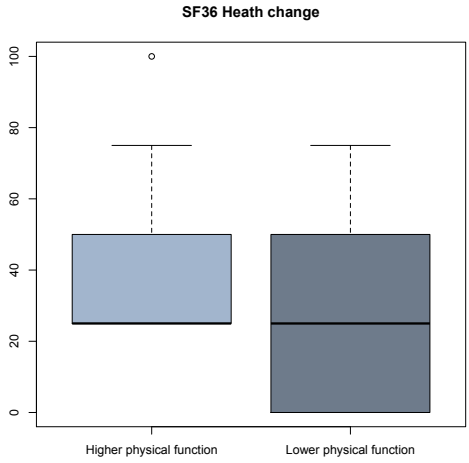


Figure 2.16: Boxplot of SF36 health change by physical functioning

Table 2.8: Summary statistics t-test SF36 subscales by physical functioning

SF36 by General Health	Group	Count	Mean	SD	t	dF	p
Emotional well-being	Higher physical functioning	22	65.09	19.64	0.41	48	0.6838
	Lower physical functioning	28	63.14	13.96			
General Health	Higher physical functioning	22	35.68	20.31	2.72	48	0.0090*
	Lower physical functioning	28	21.96	15.36			

Table 2.9: Summary statistics Wilcoxon rank sum test with continuity correction SF36 subscales by physical functioning

SF36 scale by physical functioning	Group	Count	Median	IQR	W	p
Role functioning/physical	Higher physical functioning	22	0.00	43.75	410.00	0.0061*
	Lower physical functioning	28	0.00	0.00		
Role functioning/emotional	Higher physical functioning	22	33.33	100.00	312.00	0.9408
	Lower physical functioning	28	16.67	100.00		
Energy/Fatigue	Higher physical functioning	22	25.00	18.75	420.50	0.0276*
	Lower physical functioning	28	12.50	25.00		
Social functioning	Higher physical functioning	22	50.00	25.00	494.50	0.0002*
	Lower physical functioning	28	25.00	25.00		
Pain	Higher physical functioning	22	57.50	22.50	439.00	0.0102*
	Lower physical functioning	28	32.50	25.00		
Health change	Higher physical functioning	22	37.50	50.00	438.00	0.0064*
	Lower physical functioning	28	25.00	12.50		

* Statistically significant at $p<0.05$

Comparisons of SF36 sub scales by emotional well-being

Comparisons of SF36 subscales were made between those that had above average for the group SF36 physical functioning scores (higher physical functioning) compared to those with average or below scores (lower physical functioning). Comparisons between higher emotional well-being and lower emotional well-being for the SF36 emotional well-being subscale were excluded due to selection bias. Boxplots of each SF36 scale by metastatic status are displayed in Figures 2.17-2.24. A two-sample t-test was used when assumptions for normality and variance were met (Table 2.10), or when assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 2.11).

A two sample t-test indicated that the mean score for the SF36 general health [t(48) = 2.48, p=0.0166] was significantly better for those with higher emotional well-being (Mean = 34.04, SD 20.45) compared to those with lower emotional well-being (Mean = 21.46, SD = 15.00).

A Wilcoxon rank sum test with continuity correction indicated that those with higher emotional well-being (Median =0.00, IQR = 43.75) had significantly better baseline health for role functioning/physical [W=398.50, p=0.0212] compared to those with lower emotional well-being (Median =0.00, IQR=0.00); those with higher emotional well-being (Median = 100.00, IQR = 8.33) had significantly better baseline health for role functioning/emotional [W=506.50, p<0.0001] compared to those with lower emotional well-being (Median =0.00, IQR=8.33); those with higher emotional well-being (Median = 50.00, IQR = 34.38) had significantly better baseline health for social functioning [W=467.00, p = 0.0024] compared to those with lower emotional well-being (Median =25.00, IQR=25.00); those with higher emotional well-being (Median = 50.00, IQR = 31.88) had significantly better baseline health for pain [W=417.00, p=0.0412] compared to those with lower emotional well-being (Median = 27.50, IQR=35.63); and those with higher emotional well-being (Median = 37.50, IQR = 50.00) had significantly better baseline health for health change [W= 462.50, p=0.0017] compared to those with lower emotional well-being (Median = 25.00, IQR=25.00);

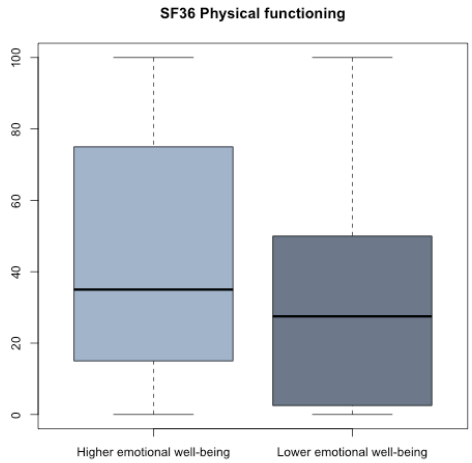


Figure 2.17: Boxplot of SF36 physical functioning by emotional well-being

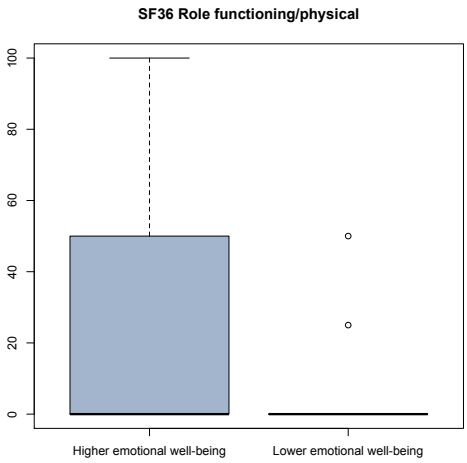


Figure 2.18: Boxplot of SF36 role limitations due to physical health by emotional well-being

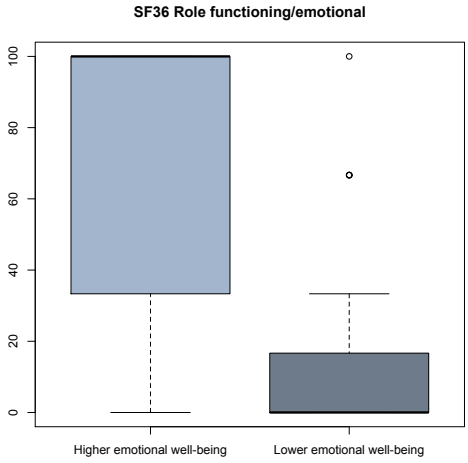


Figure 2.19: Boxplot of SF36 role limitations due to emotional problems by emotional well-being

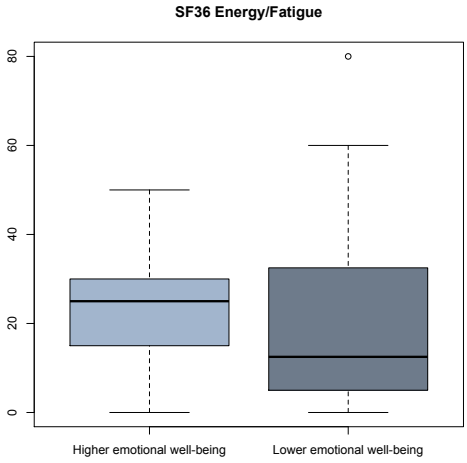


Figure 2.20: Boxplot of SF36 energy/fatigue by emotional well-being

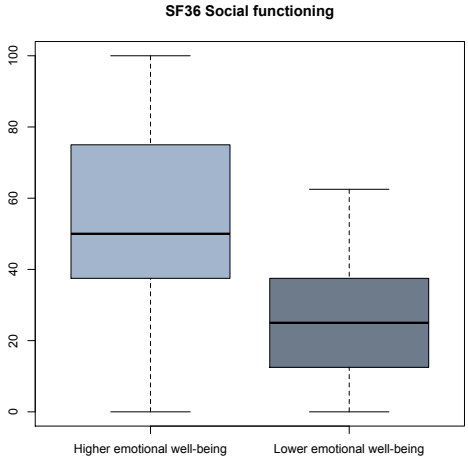


Figure 2.21: Boxplot of SF36 social functioning by emotional well-being

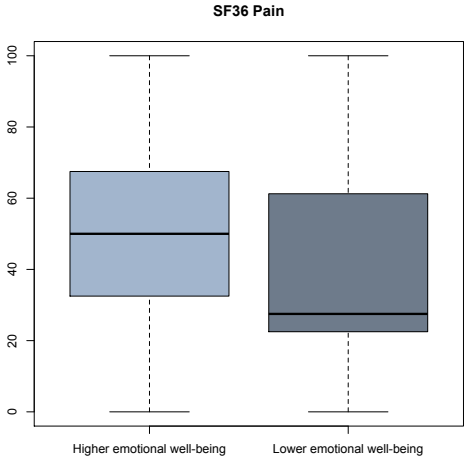


Figure 2.22: Boxplot of SF36 pain by emotional well-being

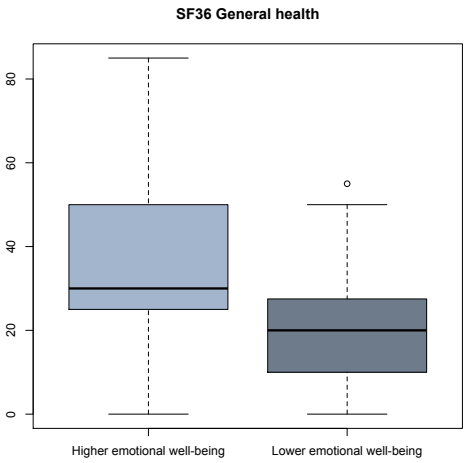


Figure 2.22: Boxplot of SF36 general health by emotional well-being

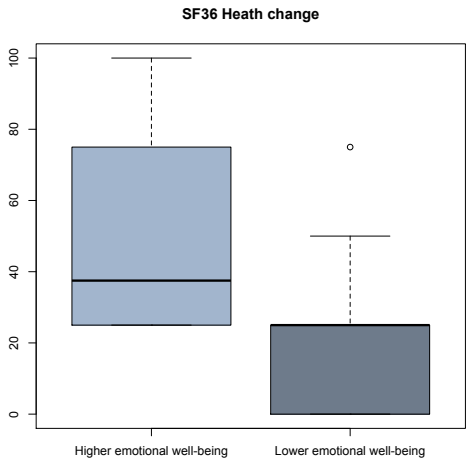


Figure 2.24: Boxplot of SF36 health change by emotional well-being

Table 2.10: Summary statistics and two sample t-test SF36 subscales by emotional well-being

SF36 scale by Emotional well-being	Group	Count	Mean	SD	t	dF	p
General health	Higher emotional well-being	26	34.04	20.45	2.48	48	0.0166*
	Lower emotional well-being	24	21.46	15.00			

Table 2.11: Summary statistics Wilcoxon rank sum test with continuity correction SF36 subscales by emotional well-being.

SF36 scale by Emotional well-being	Group	Count	Median	IQR	W	p
Physical functioning	Higher emotional well-being	26	35.00	56.25	380.50	0.1842
	Lower emotional well-being	24	27.50	46.25		
Role functioning/physical	Higher emotional well-being	26	0.00	43.75	398.50	0.0212*
	Lower emotional well-being	24	0.00	0.00		
Role functioning/emotional	Higher emotional well-being	26	100.00	66.67	506.50	<0.0001*
	Lower emotional well-being	24	0.00	8.33		
Energy/Fatigue	Higher emotional well-being	26	25.00	15.00	344.00	0.5383
	Lower emotional well-being	24	12.50	26.25		
Social functioning	Higher emotional well-being	26	50.00	34.38	467.00	0.0024*
	Lower emotional well-being	24	25.00	25.00		
Pain	Higher emotional well-being	26	50.00	31.88	417.00	0.0412*
	Lower emotional well-being	24	27.50	35.63		
Health change	Higher emotional well-being	26	37.50	50.00	462.50	0.0017*
	Lower emotional well-being	24	25.00	25.00		

Comparisons of SF36 sub scales by social functioning

Comparisons of SF36 subscales were made between those that had above average for the group SF36 social functioning scores (higher social functioning) compared to those with average or below scores (lower social functioning). Comparisons between higher social functioning and lower social functioning for the SF36 social functioning subscale were excluded due to selection bias. Boxplots of each SF36 scale by education status are displayed in Figures 2.25-2.32. A two-sample t-test was used when assumptions for normality and variance were met (Table 2.12), or when

assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 2.13).

A two sample t-test indicated that the mean score for the SF36 social functioning [t(48) = 4.09, p=0.0002] was significantly better for those with higher social functioning (Mean = 74.20, SD= 12.55) compared to those with lower emotional well-being (Mean = 57.20, SD = 15.00).

A Wilcoxon rank sum test with continuity correction indicated that those with higher social functioning (Median =50.00, IQR = 50.00) had significantly better baseline health for physical functioning [W=515.50, p<0.0001] compared to those with lower social functioning (Median =10.00, IQR=37.50); those with higher social functioning (Median =12.50, IQR = 50.00) had significantly better baseline health for role functioning/physical [W=444.00, p<0.0001] compared to those with lower social functioning (Median =0.00, IQR=0.00); those with higher social functioning (Median =100.00, IQR = 75.00) had significantly better baseline health for role functioning/emotional [W=429.50, p = 0.0056] compared to those with lower social functioning (Median =0.00, IQR=66.67); those with higher social functioning (Median =30.00, IQR = 15.00) had significantly better baseline health for energy/fatigue [W=464.50, p = 0.0011] compared to those with lower social functioning (Median =10.00, IQR=20.00); those with higher social functioning (Median =67.50, IQR = 35.63) had significantly better baseline health for pain [W=521.50, p < 0.0001] compared to those with lower emotional well-being (Median =27.50, IQR=22.50); those with higher emotional well-being (Median =32.50, IQR = 25.00) had significantly better baseline health for general health [W=468.00, p = 0.0008] compared to those with lower emotional well-being (Median =20.00, IQR=20.00); and those with higher emotional well-being (Median =50.00, IQR = 50.00) had significantly better baseline health for health change [W=475.00, p = 0.0002] compared to those with lower emotional well-being (Median =25.00, IQR=0.00).

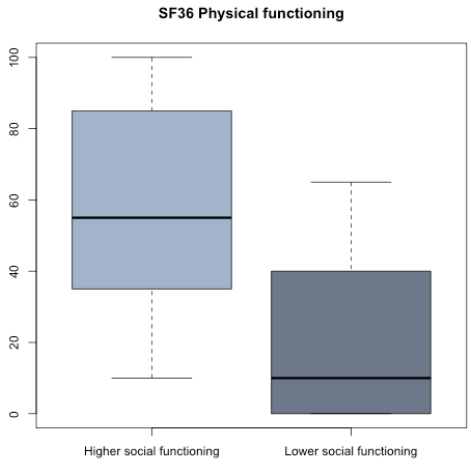


Figure 2.25: Boxplot of SF36 physical functioning by social functioning

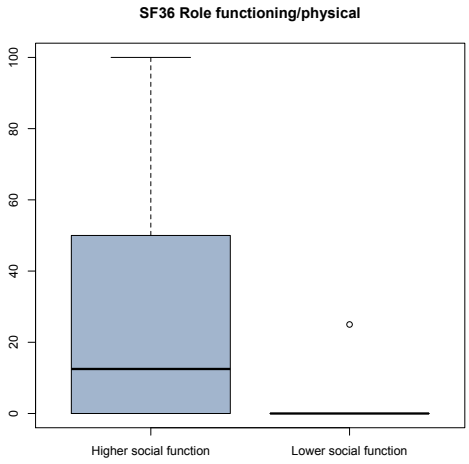


Figure 2.26: Boxplot of SF36 role limitations due to physical health by social functioning

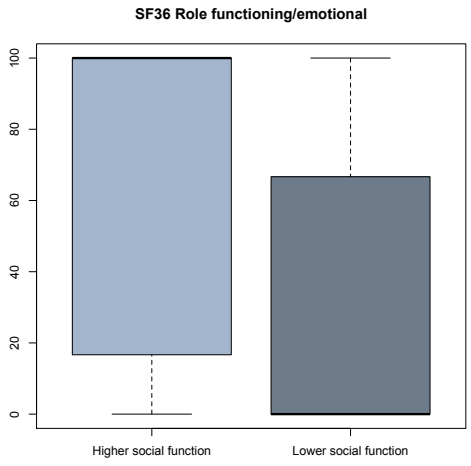


Figure 2.27: Boxplot of SF36 role limitations due to emotional problems by social functioning

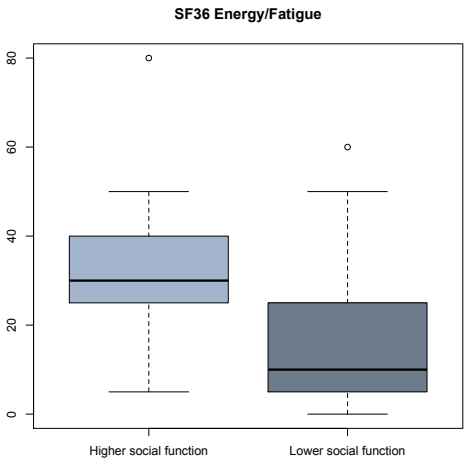


Figure 2.28: Boxplot of SF36 energy/fatigue by social functioning

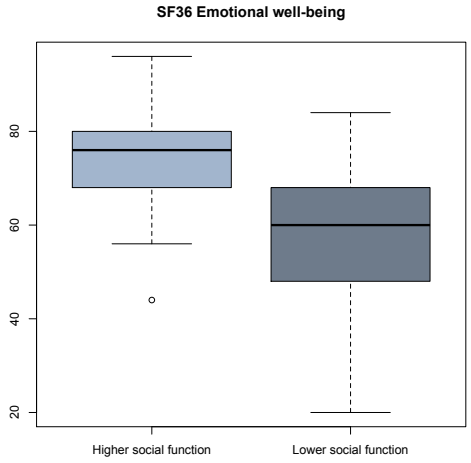


Figure 2.29: Boxplot of SF36 emotional well-being by social functioning

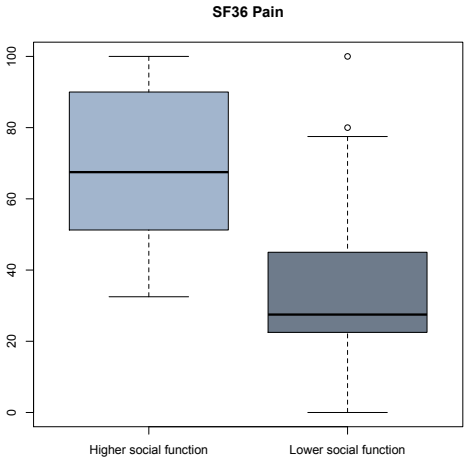
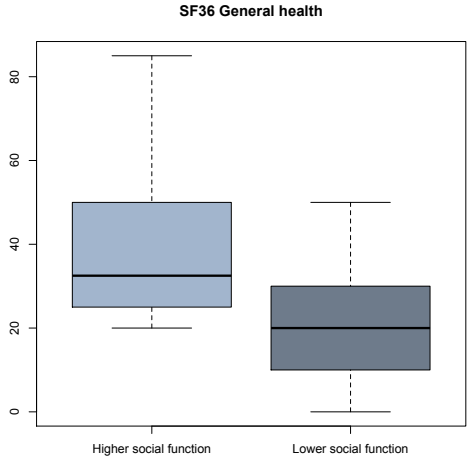


Figure 2.30: Boxplot of SF36 pain by social functioning



2.31: Boxplot of SF36 general health by social functioning

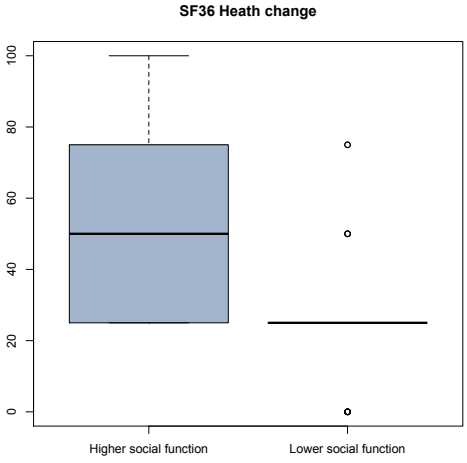


Figure 2.32: Boxplot of SF36 health change by social functioning

Table 2.12: Summary statistics and two sample t-test SF36 subscales by social functioning

SF36 scale by social functioning	Group	Count	Mean	SD	t	dF	p
Emotional well-being	Higher social functioning	20	74.20	12.55	4.09	48	0.0002*
	Lower social functioning	30	57.20	15.00			

Table 2.13: Summary statistics Wilcoxon rank sum test with continuity correction SF36 subscales by social functioning

SF36 scale by social functioning	Group	Count	Median	IQR	W	p
Physical functioning	Higher social functioning	20	50.00	50.00	515.50	<0.0001*
	Lower social functioning	30	10.00	37.50		
Role functioning/physical	Higher social functioning	20	12.50	50.00	444.00	<0.0001*
	Lower social functioning	30	0.00	0.00		
Role functioning/emotional	Higher social functioning	20	100.00	75.00	429.50	0.0056*
	Lower social functioning	30	0.00	66.67		
Energy/Fatigue	Higher social functioning	20	30.00	15.00	464.50	0.0011*
	Lower social functioning	30	10.00	20.00		
Pain	Higher social functioning	20	67.50	35.63	521.50	<0.0001*
	Lower social functioning	30	27.50	22.50		
General health	Higher social functioning	20	32.50	25.00	468.00	0.0008*
	Lower social functioning	30	20.00	20.00		
Health change	Higher social functioning	20	50.00	50.00	475.00	0.0002*
	Lower social functioning	30	25.00	0.00		

* Statistically significant at p<0.05

Comparisons of SF36 sub scales by hearing problems

Comparisons of SF36 subscales were made by hearing problems, comparing those with hearing problems with those that have no hearing problems. Boxplots of each SF36 scale by hearing problem status are displayed in Figures 2.33-2.41. A two-sample t-test was used when assumptions for normality and variance were met (Table 2.14), or when assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 2.15).

No significant differences were observed between those with hearing problems and those with no hearing problems for any of the SF36 subscales.

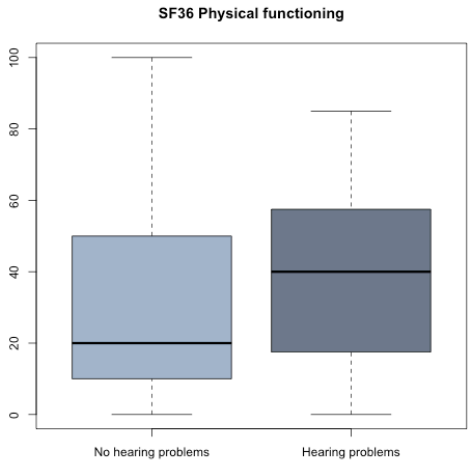


Figure 2.33: Boxplot of SF36 physical functioning by hearing problems

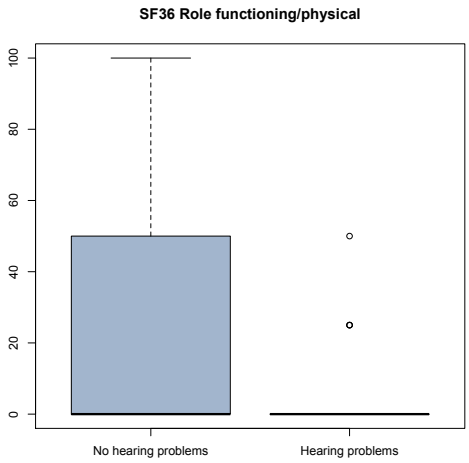


Figure 2.34: Boxplot of SF36 role limitations due to physical health by hearing problems

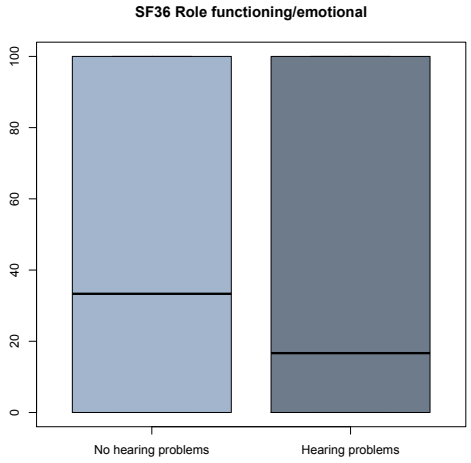


Figure 2.35: Boxplot of SF36 role limitations due to emotional problems by hearing problems

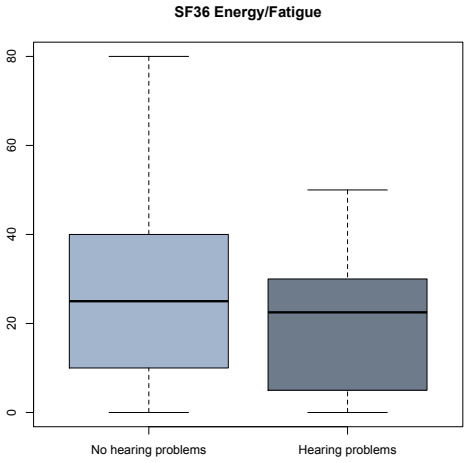


Figure 2.36: Boxplot of SF36 energy/fatigue by hearing problems

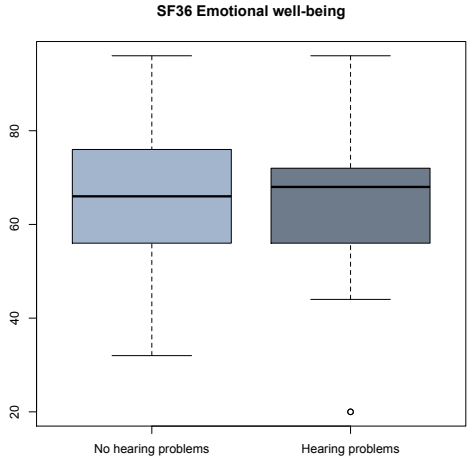
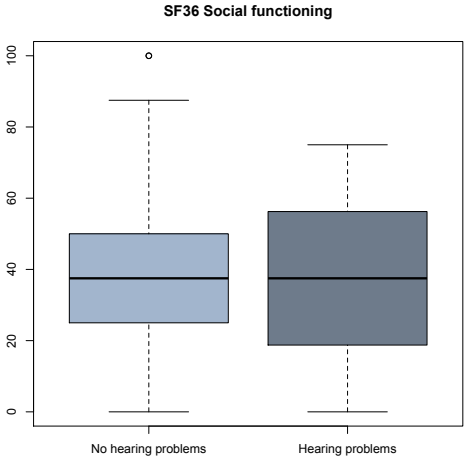


Figure 2.37: Boxplot of SF36 emotional well-being by hearing problems



2.38: Boxplot of SF36 social functioning by hearing problems

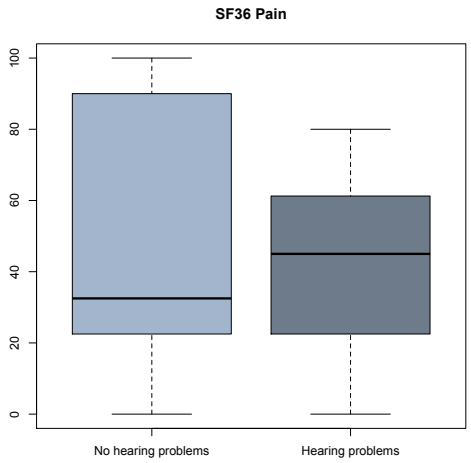
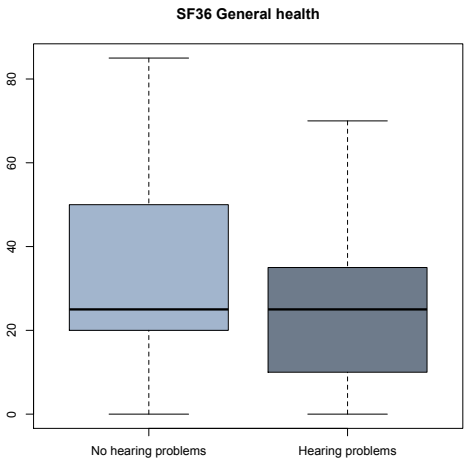


Figure 2.39: Boxplot of SF36 pain by hearing problems



2.40: Boxplot of SF36 general health by hearing problems

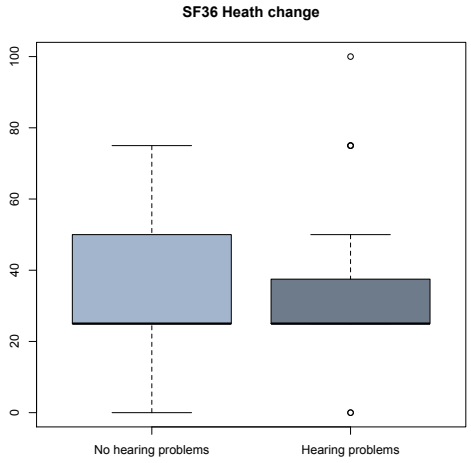


Figure 2.41: Boxplot of SF36 health change by hearing problems

Table 2.14: Summary statistics and two sample t-test SF36 subscales by hearing problems

SF36 scale by hearing problems	Group	Count	Mean	SD	t	dF	p
General health	No hearing problems	26	30.58	19.51	1.01	48	0.3185
	Hearing problems	24	25.21	18			

Table 2.15: Summary statistics Wilcoxon rank sum test with continuity correction SF36 subscales by hearing problems

SF36 scale by hearing problems	Group	Count	Median	IQR	W	p
Physical functioning	No hearing problems	26	20.00	38.75	267.00	0.3848
	Hearing problems	24	10.00	37.50		
Role functioning/physical	No hearing problems	26	0.00	37.50	356.50	0.2382
	Hearing problems	24	0.00	0.00		
Role functioning/emotional	No hearing problems	26	33.33	100.00	323.00	0.8249
	Hearing problems	24	16.67	100.00		
Energy/Fatigue	No hearing problems	26	25.00	28.75	357.00	0.3846
	Hearing problems	24	22.50	25.00		
Emotional well-being	No hearing problems	26	66.00	20.00	324.00	0.8226
	Hearing problems	24	68.00	14.00		
Social functioning	No hearing problems	26	37.50	25.00	331.00	0.7162
	Hearing problems	24	37.50	31.25		
Pain	No hearing problems	26	32.50	61.88	317.00	0.9299
	Hearing problems	24	45.00	36.88		
Health change	No hearing problems	26	25.00	25.00	335.50	0.6306
	Hearing problems	24	25.00	6.25		

* Statistically significant at $p<0.05$

Comparisons of SF36 sub scales by eye problems

Comparisons of SF36 subscales were made by eye problems, comparing those with eye problems with those that have no eye problems. Boxplots of each SF36 scale by eye problem status are displayed in Figures 2.42-2.50. A two-sample t-test was used when assumptions for normality and variance were met (Table 2.16), or when assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 2.17).

No significant differences were observed between those with eye problems and those with no eye problems for any of the SF36 subscales.

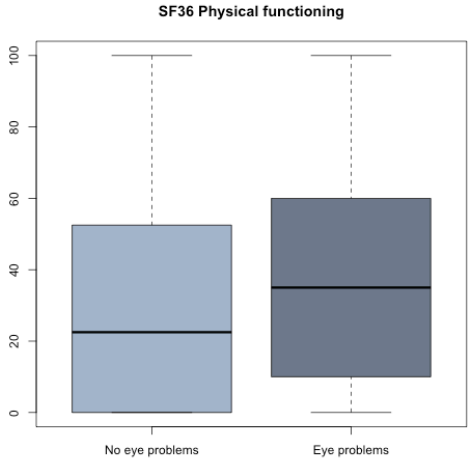


Figure 2.42: Boxplot of SF36 physical functioning by eye problems

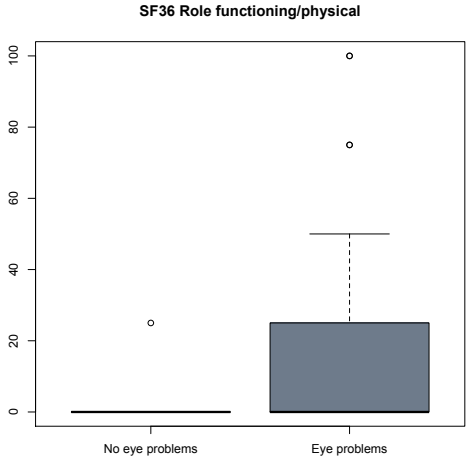


Figure 2.43: Boxplot of SF36 role limitations due to physical health by eye problems

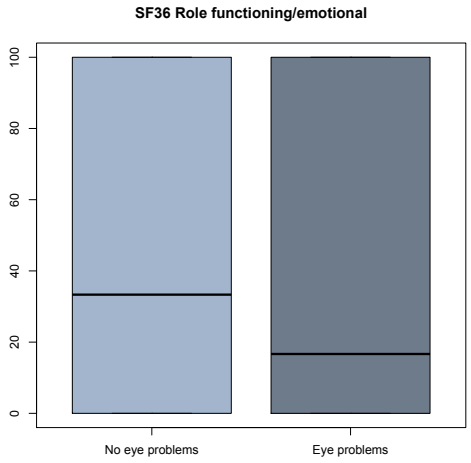


Figure 2.44: Boxplot of SF36 role limitations due to emotional problems by eye problems

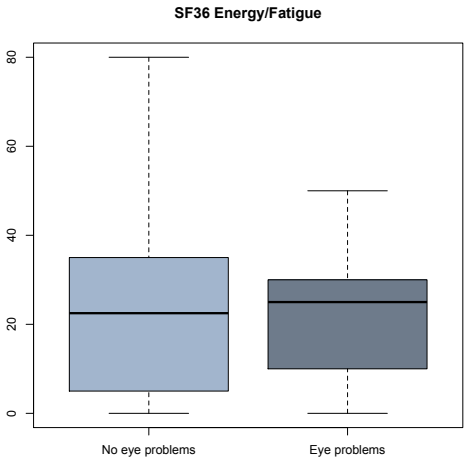


Figure 2.45: Boxplot of SF36 energy/fatigue by eye problems

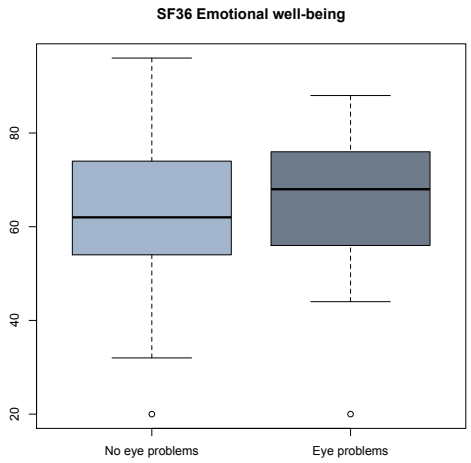
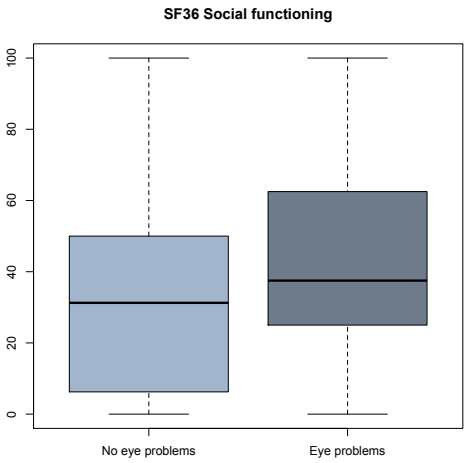


Figure 2.46: Boxplot of SF36 emotional well-being by eye problems



2.47: Boxplot of SF36 social functioning by eye problems

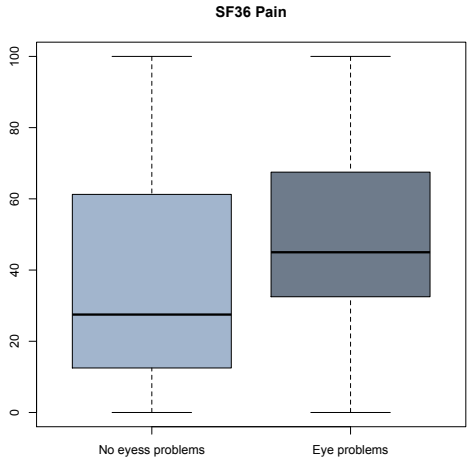
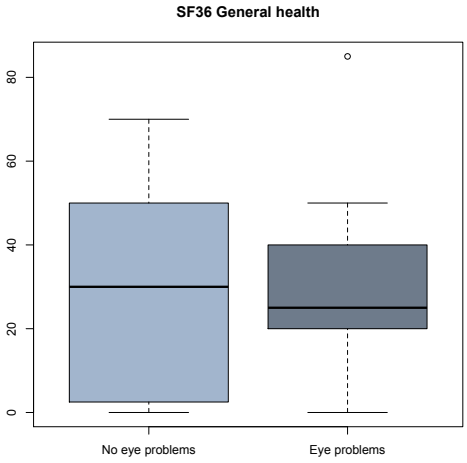


Figure 2.48: Boxplot of SF36 pain by eye problems



2.49: Boxplot of SF36 general health by eye problems

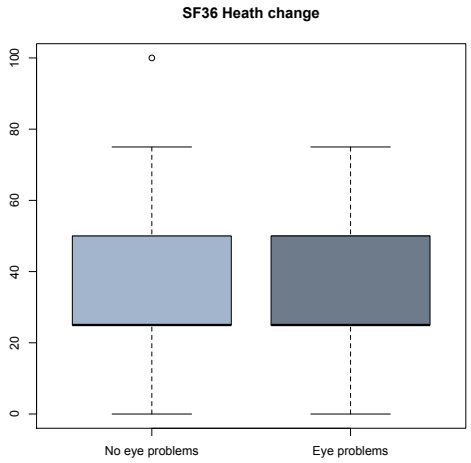


Figure 2.50: Boxplot of SF36 health change by eye problems

Table 2.16: Summary statistics and two sample t-test SF36 subscales by eye problems

SF36 scale by eye problems	Group	Count	Mean	SD	t	dF	p
Social functioning	No eye problems	16	31.25	28.87	-1.60	48	0.1166
	Eye problems	34	43.75	24			
Pain	No eye problems	16	39.06	32.82	-1.35	48	0.1839
	Eye problems	34	50.59	26			

Table 2.17: Summary statistics Wilcoxon rank sum test with continuity correction SF36 subscales by eye problems

SF36 scale by eye problems	Group	Count	Median	IQR	W	p
Physical functioning	No eye problems	16	22.50	51.25	233.50	0.4267
	Eye problems	34	18.51	35.00		
Role functioning/physical	No eye problems	16	0.00	0.00	205.00	0.0563
	Eye problems	34	0.00	25.00		
Role functioning/emotional	No eye problems	16	33.33	100.00	276.50	0.9281
	Eye problems	34	16.67	100.00		
Energy/Fatigue	No eye problems	16	22.50	27.50	256.00	0.7457
	Eye problems	34	25.00	20.00		
Emotional well-being	No eye problems	16	62.00	18.00	248.00	0.6236
	Eye problems	34	68.00	19.00		
General health	No eye problems	16	30.00	46.25	271.50	1.0000
	Eye problems	34	25.00	17.50		
Health change	No eye problems	16	25.00	25.00	259.50	0.7882
	Eye problems	34	25.00	25.00		

* Statistically significant at $p<0.05$

Comparisons of SF36 sub scales by location

Comparisons of SF36 subscales were made by location, comparing those that live in metropolitan areas with those that live in regional or rural areas. Boxplots of each SF36 scale by location are displayed in Figures 2.51-2.59. A two-sample t-test was used when assumptions for normality and variance were met (Table 2.18), or when assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 2.19).

No significant differences were observed between those that live in metropolitan areas and those that live in regional or rural areas for any of the SF36 subscales.

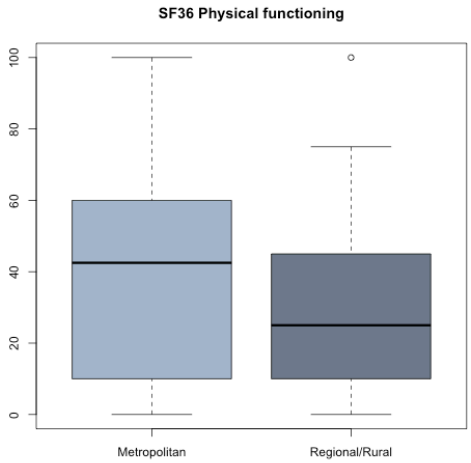


Figure 2.51: Boxplot of SF36 physical functioning by location

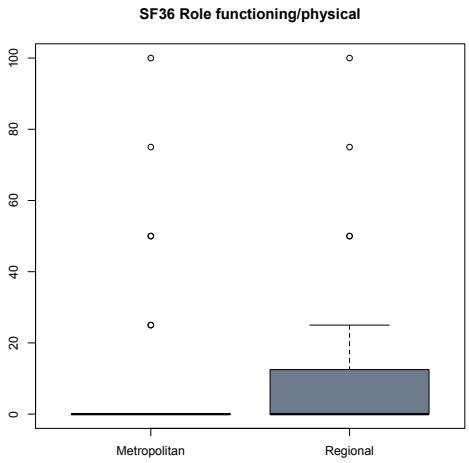


Figure 2.52: Boxplot of SF36 role limitations due to physical health by location

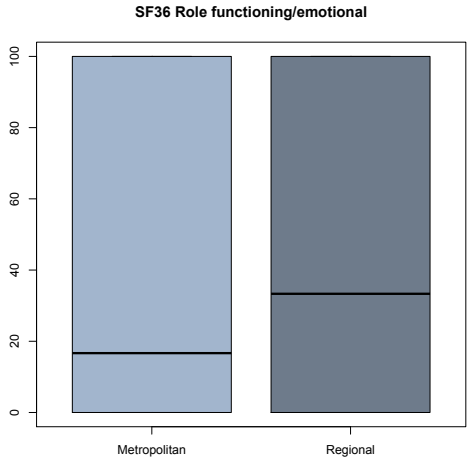


Figure 2.53: Boxplot of SF36 role limitations due to emotional problems by location

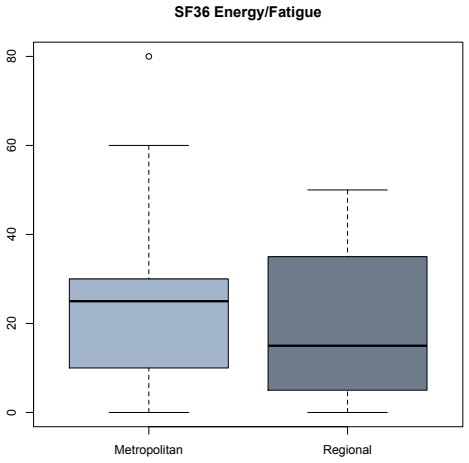


Figure 2.54: Boxplot of SF36 energy/fatigue by location

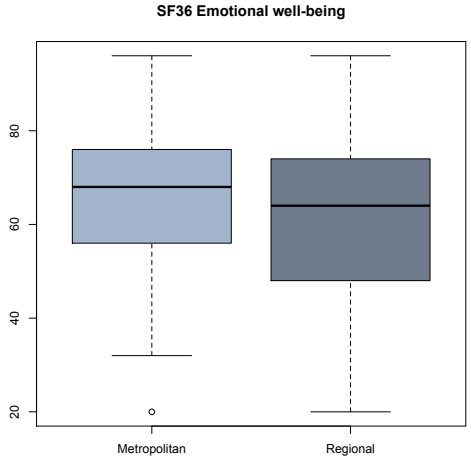
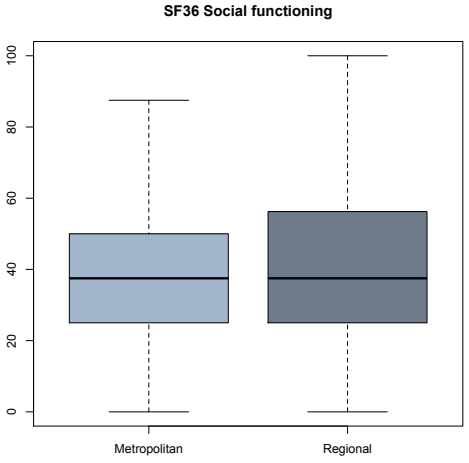


Figure 2.55: Boxplot of SF36 emotional well-being by location



2.56: Boxplot of SF36 social functioning by location

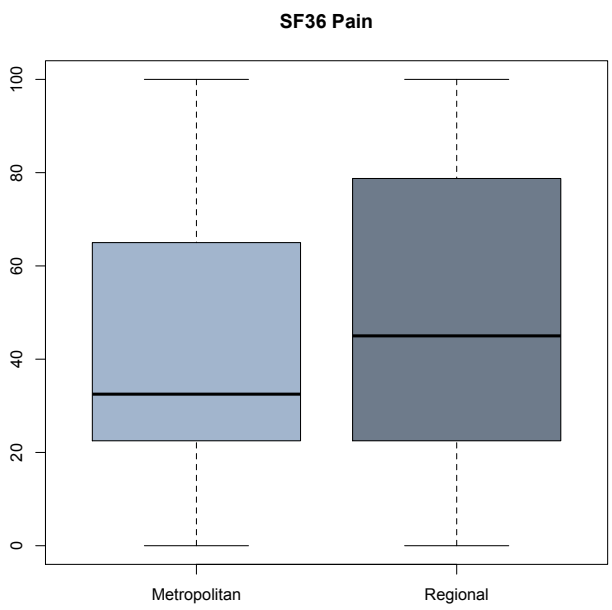
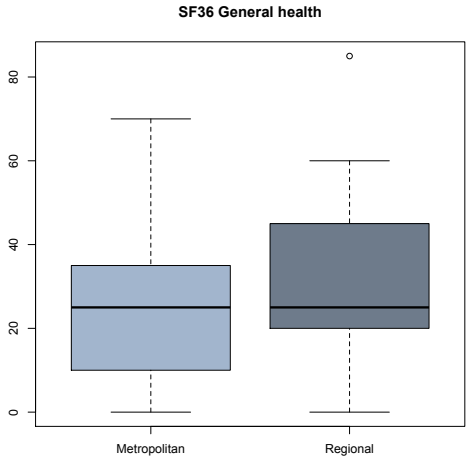


Figure 2.57: Boxplot of SF36 pain by location



2.58: Boxplot of SF36 general health by eye location

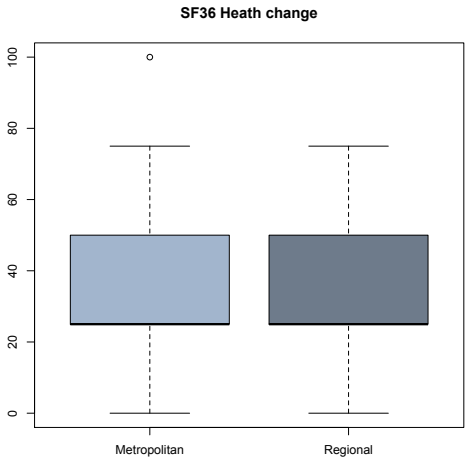


Figure 2.59: Boxplot of SF36 health change by location

Table 2.18: Summary statistics and two sample t-test SF36 subscales by location

SF36 scale by location	Group	Count	Mean	SD	t	dF	p
Emotional well-being	Metropolitan	30	64.80	16.46	0.42	48	0.6798
	Regional	20	62.80	17			
Pain	Metropolitan	30	43.08	27.56	-1.17	48	0.2490
	Regional	20	52.63	29			
General health	Metropolitan	30	25.83	17.96	-1.00	48	0.3237
	Regional	20	31.25	20			

Table 2.19: Summary statistics Wilcoxon rank sum test with continuity correction SF36 subscales by location

SF36 scale by location	Group	Count	Median	IQR	W	p
Physical functioning	Metropolitan	30	42.50	48.75	340.00	0.4315
	Regional	20	25.00	32.50		
Role functioning/physical	Metropolitan	30	0.00	0.00	283.00	0.6520
	Regional	20	0.00	6.25		
Role functioning/emotional	Metropolitan	30	16.67	100.00	288.50	0.8131
	Regional	20	33.33	100.00		
Energy/Fatigue	Metropolitan	30	25.00	20.00	333.00	0.5173
	Regional	20	15.00	27.50		
Social functioning	Metropolitan	30	37.50	25.00	308.50	0.8726
	Regional	20	37.50	28.13		
Health change	Metropolitan	30	25.00	25.00	330.50	0.5224
	Regional	20	25.00	25.00		

* Statistically significant at p<0.05

Comparisons of SF36 sub scales by education

Comparisons of SF36 subscales were made by education, those that had a university qualification were compared with those that high school or trade qualifications. Boxplots of each SF36 scale by education are displayed in Figures 2.60-2.68. A two-sample t-test was used when assumptions for normality and variance were met (Table 2.20), or when assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 2.21).

No significant differences were observed between those that with a university qualification and those with high school or trade qualifications for any of the SF36 subscales.

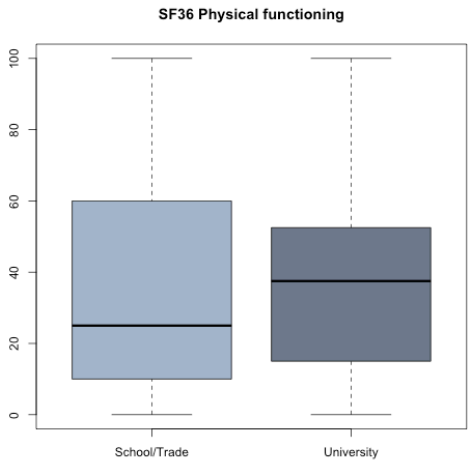


Figure 2.60: Boxplot of SF36 physical functioning by education

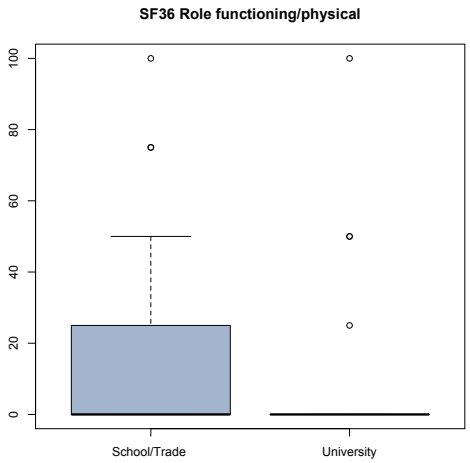


Figure 2.61: Boxplot of SF36 role limitations due to physical health by education

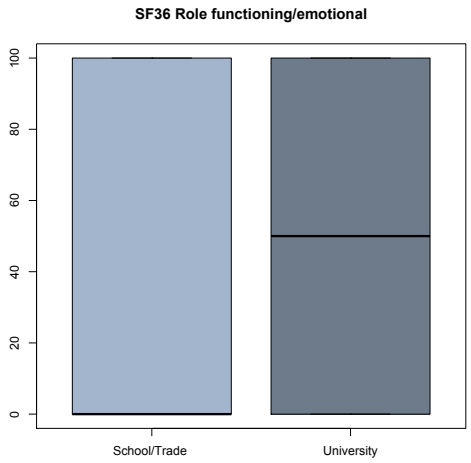


Figure 2.62: Boxplot of SF36 role limitations due to emotional problems by education

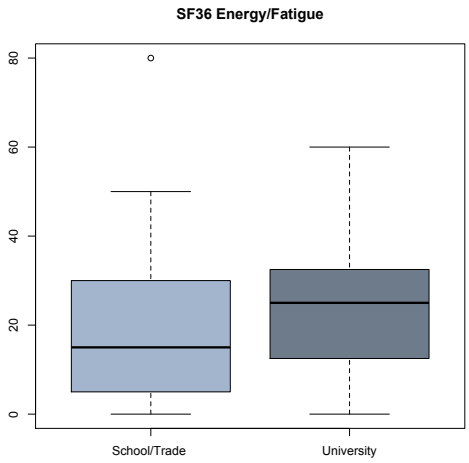


Figure 2.63: Boxplot of SF36 energy/fatigue by education

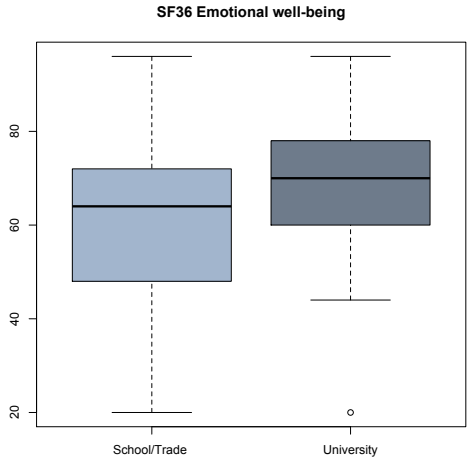
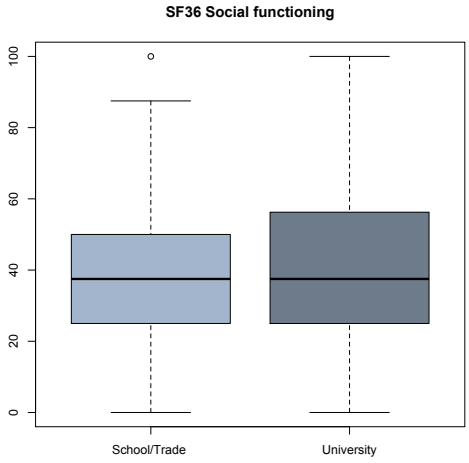


Figure 2.64: Boxplot of SF36 emotional well-being by education



2.65: Boxplot of SF36 social functioning by education

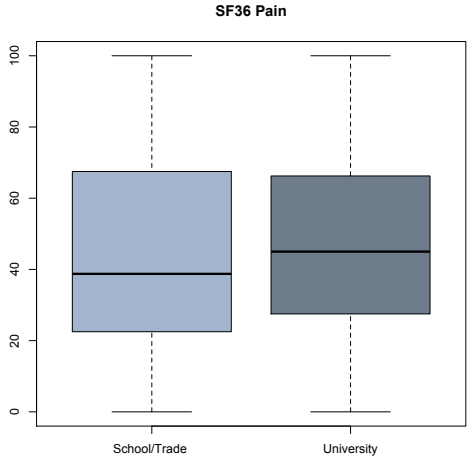
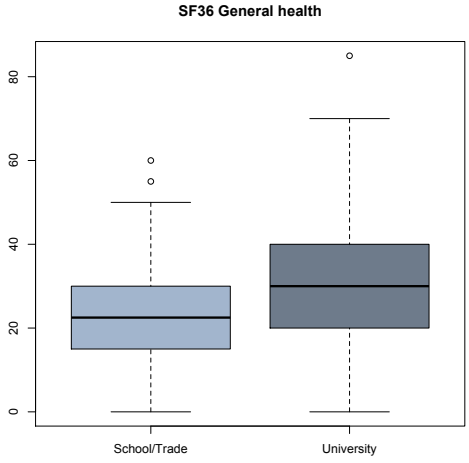


Figure 2.66: Boxplot of SF36 pain by education



2.67: Boxplot of SF36 general health by education

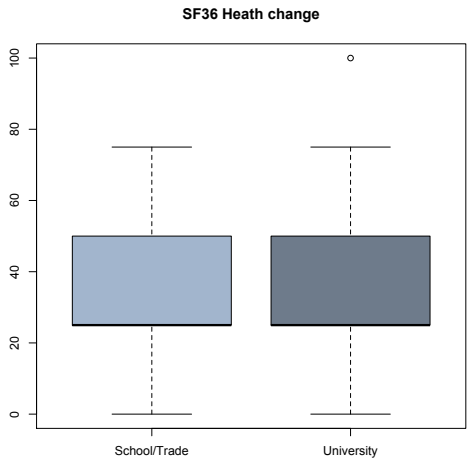


Figure 2.68: Boxplot of SF36 health change by education

Table 2.20: Summary statistics and two sample t-test SF36 subscales by education

SF36 scale by education	Group	Count	Mean	SD	t	dF	p
Emotional well-being	School/Trade	26	60.92	17.17	-1.38	48	0.1735
	University	24	67.33	15			
Social functioning	School/Trade	26	39.42	28.44	-0.09	48	0.9280
	University	24	40.10	24			
Pain	School/Trade	26	46.06	30.34	-0.22	48	0.8300
	University	24	47.81	27			
General health	School/Trade	26	25.00	17.03	-1.18	48	0.2446
	University	24	31.25	20			

Table 2.21: Summary statistics Wilcoxon rank sum test with continuity correction SF36 subscales by education

SF36 scale by education	Group	Count	Median	IQR	W	p
Physical functioning	School/Trade	26	25.00	47.50	277.50	0.5067
	University	24	37.50	36.25		
Role functioning/physical	School/Trade	26	0.00	12.50	344.50	0.3910
	University	24	0.00	0.00		
Role functioning/emotional	School/Trade	26	0.00	100.00	250.00	0.1949
	University	24	50.00	100.00		
Energy/Fatigue	School/Trade	26	15.00	20.00	250.00	0.2295
	University	24	25.00	17.50		
Health change	School/Trade	26	25.00	25.00	284.50	0.5724
	University	24	25.00	25.00		

Comparisons of SF36 sub scales by Socio-Economic Indexes For Areas (SEIFA)

Comparisons of SF36 subscales were made by SEIFA, those lived in an area with a higher SEIFA (more advantaged) were compared with those lived in an area with a lower SEIFA. Boxplots of each SF36 scale by SEIFA are displayed in Figures 2.69-2.77. A two-sample t-test was used when assumptions for normality and variance were met (Table 2.22), or when assumptions for normality and variance were not met,

a Wilcoxon rank sum test with continuity correction was used (Table 2.23).

No significant differences were observed between those lived in an area with a higher SEIFA (more advantaged) and with those lived in an area with a lower SEIFA.

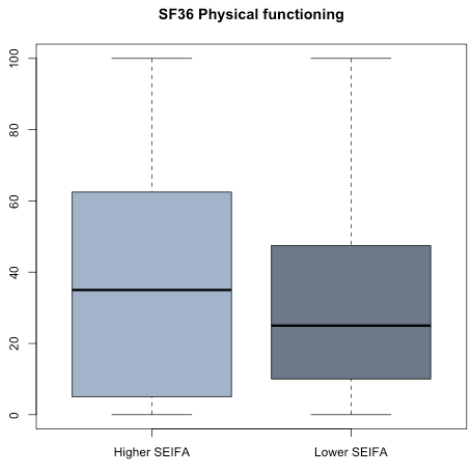


Figure 2.69: Boxplot of SF36 physical functioning by SEIFA

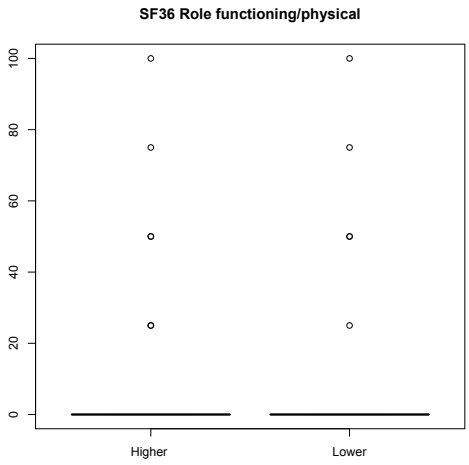


Figure 2.70: Boxplot of SF36 role limitations due to physical health by SEIFA

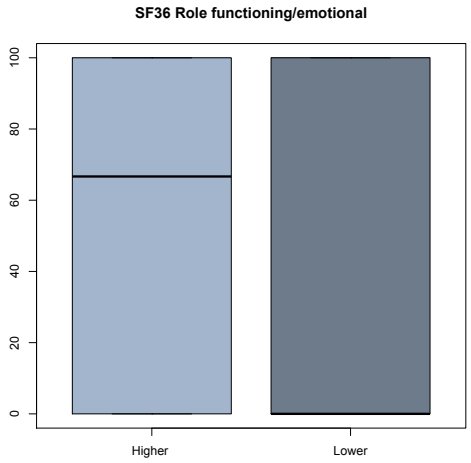


Figure 2.71: Boxplot of SF36 role limitations due to emotional problems by SEIFA

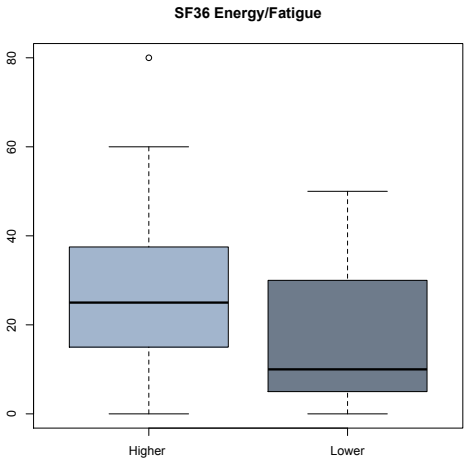


Figure 2.72: Boxplot of SF36 energy/fatigue by SEIFA

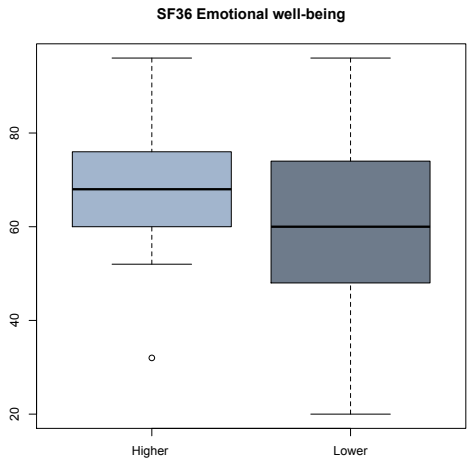
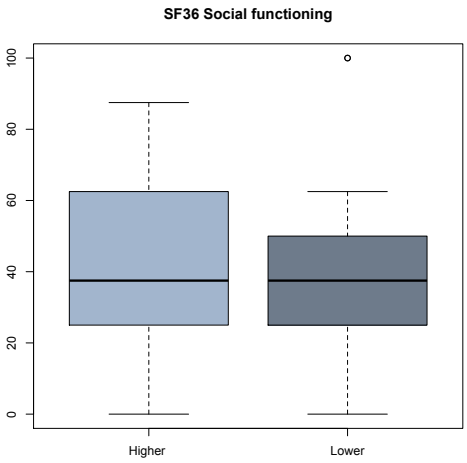


Figure 2.73: Boxplot of SF36 emotional well-being by SEIFA



2.74: Boxplot of SF36 social functioning by SEIFA

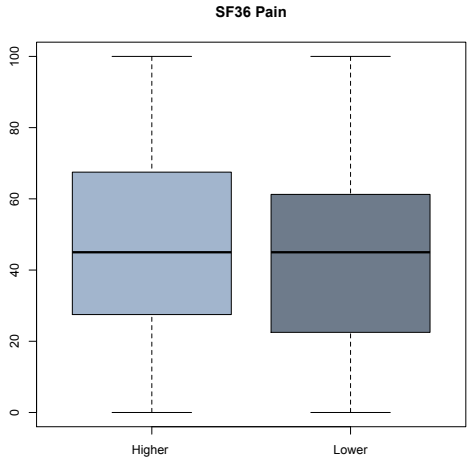
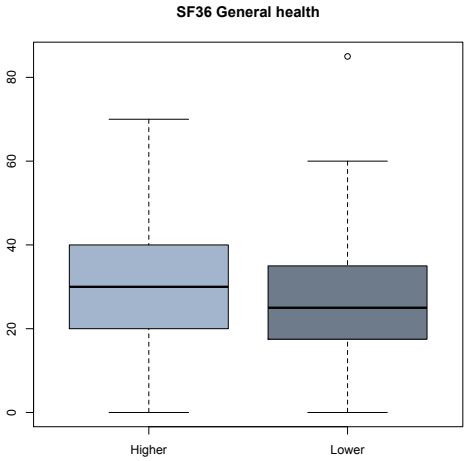


Figure 2.75: Boxplot of SF36 pain by SEIFA



2.76: Boxplot of SF36 general health by SEIFA

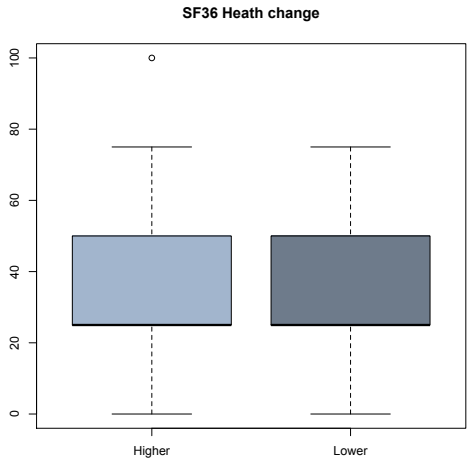


Figure 2.77: Boxplot of SF36 health change by SEIFA

Table 2.22: Summary statistics and two sample t-test SF36 subscales by SEIFA

SF36 scale by SEIFA	Group	Count	Mean	SD	t	dF	p
Role functioning/emotional	Higher SEIFA	27	42.22	46.27	1.22	48	0.2267
	Lower SEIFA	23	45.00	46			
General health	Higher SEIFA	27	25.83	17.96	0.21	48	0.8353
	Lower SEIFA	23	31.25	20			
Health change	Higher SEIFA	27	37.50	26.06			
	Lower SEIFA	23	32.50	23			

Table 2.23: Summary statistics Wilcoxon rank sum test with continuity correction SF36 subscales by SEIFA

SF36 scale by SEIFA	Group	Count	Median	IQR	W	p
Physical functioning	Higher SEIFA	27	35.00	57.50	344.00	0.5183
	Lower SEIFA	23	25.00	37.50		
Role functioning/physical	Higher SEIFA	27	0.00	0.00	310.00	1.0000
	Lower SEIFA	23	0.00	6.25		
Energy/Fatigue	Higher SEIFA	27	25.00	20.00	403.50	0.0701
	Lower SEIFA	23	15.00	27.50		
Emotional well-being	Higher SEIFA	27	68.00	20.00	389.50	0.1249
	Lower SEIFA	23	64.00	25.00		
Social functioning	Higher SEIFA	27	37.50	25.00	335.00	0.6364
	Lower SEIFA	23	37.50	28.13		
Pain	Higher SEIFA	27	32.50	40.63	311.50	0.9922
	Lower SEIFA	23	45.00	55.63		
Health change	Higher SEIFA	27	25.00	25.00	339.50	0.5503
	Lower SEIFA	23	25.00	25.00		

Section 3 Symptoms and diagnosis

Section 3: Experience of symptoms and diagnosis

Symptoms at diagnosis

- The first question was in the online questionnaire and asked participants to recall all of the symptoms that they experienced and their quality of life while experiencing those symptoms. The most commonly reported symptoms were muscle symptoms by (such as muscle weakness, exercise intolerance, pain, fatigue, cramps and low muscle tone), noted by 47 (94.00%) participants, followed by fatigue (n=45, 90.00%), digestive tract symptoms (n=36, 72.00%), problems with eyes (n=34, 68.00%), central nervous system symptoms (n=32, 64.00%), and hearing problems (n=24, 48.00%). The symptoms that had the lowest average quality of life were central nervous symptoms (mean = 2.28; n=32, 64.00%), muscle symptoms (mean = 2.52; n=47, 94.00%), heart symptoms (mean = 2.53; n=15, 30%) and digestive tract symptoms (mean = 2.64; n=36, 72.00%).
- In the structured interview, participants were asked to describe the symptoms that actually led to their diagnosis, as opposed to all the symptoms that they could recall. There were 14 participants (28.00%) that described fatigues and/or a lack of stamina and 11 participants (22.00%) that described having gastrointestinal distress ranging from nausea, diarrhoea to constipation. The next most common symptoms leading to diagnosis were failing to thrive as an infant (n=8, 16.00%), weakness in the legs or not being able to use their legs (n=7, 14.00%) and migraines that were sometimes also described as being stroke-like (n=7, 14.00%).
- In relation to sub-group variations, participants from a low socio-economic area (26.09%) and those with a low general health (25.00%) reported having severe migraines more frequently compared to the general population (14.00%), while those with a high general health reported this less frequently (0.00%). In relation to gastrointestinal distress, participants who had a high school or trade education reported this less frequently (11.54%) while those with a university education (33.33%) and those that are hearing impaired (37.50%) reported this more frequently than the general population (22.00%). Participants with a university education (20.83%) and participants with hearing impairment (20.83%) reported diabetes being a condition that led to their diagnosis more frequently than the general population (1000%). Participants with high physical function (40.91%) reported experiencing fatigue and/or lack of stamina more frequently than the general population (28.00%) while those with low physical function reported this less frequently (17.86%). Participants with high social function (40.00%) also reported experiencing fatigue and/or lack of stamina more frequently than the general population (28.00%).
- As part of the structured interview analysis in relation to symptoms that lead to diagnosis, there were 13 participants (26.00%) that noted a hereditary component that led to their diagnosis. In some cases it was a known hereditary link while in others, the hereditary link was identified as part of the diagnostic process.

Support at diagnosis

- In the questionnaire, participants were asked whether they felt supported at the time of diagnosis. There were 36 participants (72.00%) that indicated that they had no support at diagnosis, while 3 participants (6.00%) noted that they had enough support. An additional 11 participants (22.00%) indicated that they had some support but that it was not enough.
- In relation to sub-group variations, participants with no eye problems reported having no support at diagnosis more frequently than the general cohort (81.25% compared to 72.00% in the general cohort),. Participants that had higher general health reported that they had no support at diagnosis, more frequently than the general cohort (86.36% compared to 72.00% in the general cohort), and reported less frequently than the general cohort that they had some support but it wasn't enough, (13.64% compared to 22.00% in the general cohort)

Genetic/biomarker tests

- Participants were asked whether they had ever had a discussion about genetic tests or tests to see if there were biomarkers that might be relevant to their condition or treatment. Six participants (12.00%) indicated that they had brought up the topic for discussion with their doctor, 15 participants (30.00%) reported that their doctor had brought up the topic for discussion, 29 participants (58.00%) had no discussion about genetic tests.

- In relation to sub-group variations, participants with higher social functioning indicated that their doctor brought up the topic of biomarker/genetic testing, more frequently than the general cohort and those with lower social functioning less frequently (higher social functioning 45.00%; lower social functioning 20.00%, compared to 30.00% in the general cohort). Participants with no eye problems indicated that no one brought up the topic of biomarker/genetic testing, more frequently than the general cohort (68.75%, compared to 58.00% in the general cohort).
- Participants were asked about their interest in this type of test if it was available, the majority noted that they had not had this test, but would like to (n=26, 52.00%), 8 participants (16.00%) reported having this test and not paying out of pocket for it, 8 had this test as part of a clinical trial (16.00%), and two paid for this test themselves (4.00%). There were 6 participants (12.00%) indicated that they had not had this test and were not interested in it.
- In relation to sub-group variations, participants that had hearing problems, no eye problems and that were university educated indicated that they had not had this test but would like to, less frequently than the general cohort (41.67%, 31.25% and 33.33% respectively compared to 54.00% in the general cohort), while participants that did not have hearing problems, had no eye problems and had high school or trade qualifications indicated that they had not had this test but would like to, more frequently than the general cohort (61.54%, 61.76%, and 69.33% respectively, compared to 54.00% in the general cohort).
- In the structured interview, participants were also asked to talk about their understanding of genetic or biomarker testing. Some of the descriptions included understanding that the test is used for diagnosis of mitochondrial disease; understanding that the test cannot help them but may help others in the future; and understanding that the test cannot target treatment as there are no treatments available or that there was no clinical indication following the test.

Understanding of condition at diagnosis

- Participants were asked how much they knew about mitochondrial disease at diagnosis. There were 31 participants (62.00%) that described knowing nothing about mitochondrial disease and this was the most common response. There were also eight participants (16.00%) that described knowing about mitochondrial disease by the time they were diagnosed because the time to diagnosis was relatively long, giving them time to educate themselves.

Understanding of prognosis

- Participants were asked whether anyone talked to them about prognosis. The most common theme noted by 26 participants (52.00%) was prognosis had not been clearly discussed. The next most common theme was that participants understood that mitochondrial disease came with a poor prognosis that was primarily related to physical decline and this was noted by 9 participants (18.00%). There were seven participants (14.00%) that described the need for ongoing management of their condition and this included the management of exacerbations. The final theme in relation to understanding of prognosis was that mitochondrial disease came with a poor prognosis, including reduced life expectance and/or a rapid disease progression. This was noted by six participants (12.00%).

Experience of symptoms before diagnosis

Participants were asked to recall the symptoms that they noticed in themselves that led them to pursue further investigation with a clinician. This question was asked both in an online questionnaire and as part of the structured interview. Responses from both sources of information were cross-validated to compile these results.

The first question was in the online questionnaire and asked participants to recall all of the symptoms that they experienced and their quality of life while experiencing those 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". Table 3.1 describes symptoms and quality of life due to symptoms. Muscle symptoms include muscle weakness, exercise intolerance, pain fatigue, cramps and low muscle tone. Nervous system symptoms include developmental delays, mental retardation or regression, dementia, seizures, coma, neuro-psychiatric disturbances, atypical cerebral palsy, myoclonus, movement disorders, ataxia, migraine and strokes. Problems with eyes include drooping eyelids, inability to move eyes and vision loss. The most commonly reported symptom was muscle symptoms (such as muscle weakness, exercise intolerance, pain, fatigue, cramps and low muscle tone), these were experienced by 47 (94.00%) of participants. Other commonly experienced symptoms included fatigue (n=45, 90.00%), digestive tract symptoms (n=36, 72.00%), problems with eyes (n=34, 68.00%), central nervous system symptoms (n=32, 64.00%), and hearing problems (n=24, 48.00%). The symptoms that had the lowest average quality of life were central nervous symptoms (mean = 2.28; n=32, 64.00%), muscle symptoms (mean = 2.52; n=47, 94.00%), heart symptoms (mean = 2.53; n=15, 30%) and digestive tract symptoms (mean = 2.64; n=36, 72.00%).

Table 3.1: Symptoms experienced and mean QoL

Symptom	Symptom experienced	n=50	%	QOL mean	QOL SD
Muscle symptoms	Yes	47	94.00	2.52	0.96
	No	3	6.00		
Fatigue	Yes	45	90.00	2.96	1.17
	No	5	10.00		
Digestive tract symptoms	Yes	36	72.00	2.64	1.13
	No	14	28.00		
Problems with eyes	Yes	34	68.00	3.15	1.60
	No	16	32.00		
Central nervous system symptoms	Yes	32	64.00	2.28	1.11
	No	18	36.00		
Hearing problems	Yes	24	48.00	2.71	1.04
	No	26	52.00		
Heart symptoms	Yes	15	30.00	2.53	0.99
	No	35	70.00		
Fatty lumps in skin	Yes	11	22.00	3.73	0.79
	No	39	78.00		
Diabetes	Yes	10	20.00	3.50	0.85
	No	40	80.00		
Excess body hair	Yes	9	18.00	3.33	1.22
	No	41	82.00		
Kidney problems	Yes	7	14.00	2.67	1.37
	No	42	84.00		
Liver failure	Yes	2	4.00	3.00	
	No	48	96.00		
Underactive thyroid or parathyroid	Yes	2	4.00	3.50	
	No	48	96.00		

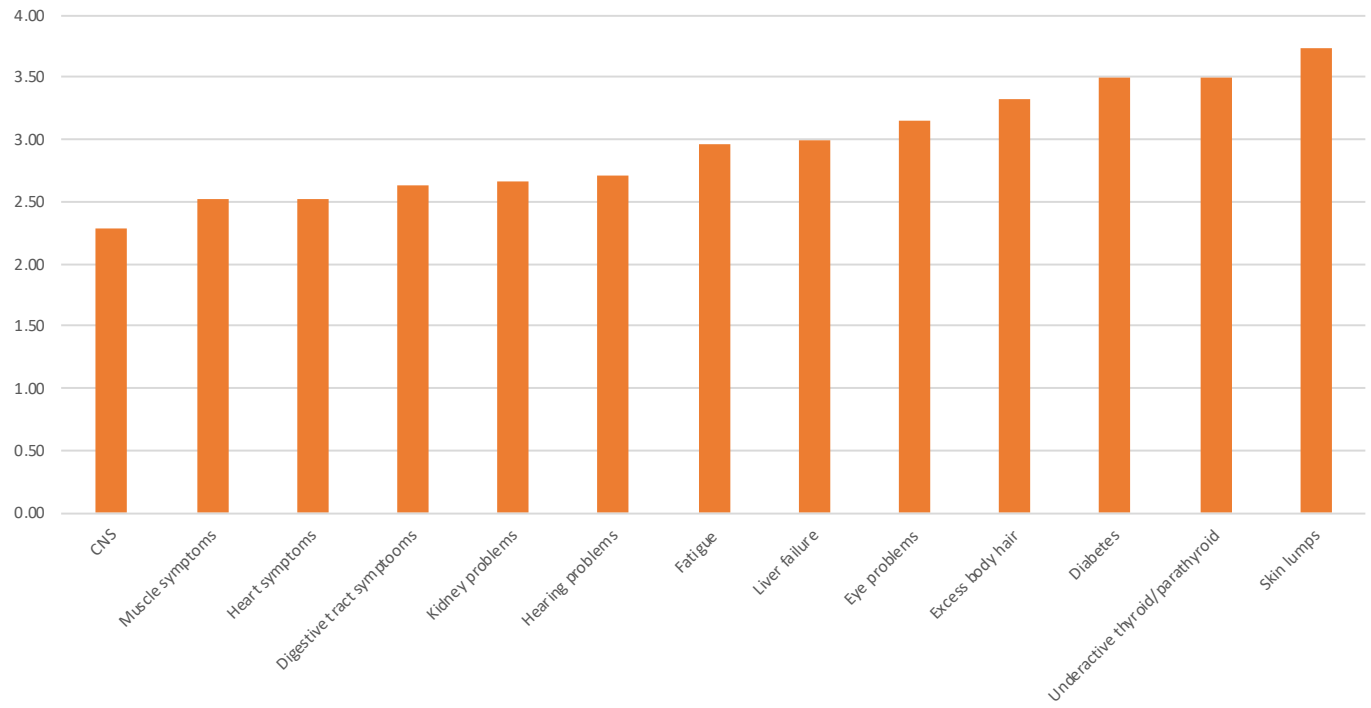


Figure 3.1 Symptoms and mean quality of life scores

Symptoms leading to diagnosis

In the structured interview, participants were asked to describe the symptoms that actually led to their diagnosis, as opposed to all the symptoms that they could recall. There were 14 participants (28.00%) that described fatigues and/or a lack of stamina and 11 participants (22.00%) that described having gastrointestinal distress ranging from nausea, diarrhoea to constipation. The next most common symptoms leading to diagnosis were failing to thrive as an infant (n=8, 16.00%), weakness in the legs or not being able to use their legs (n=7, 14.00%) and migraines that were sometimes also described as being stroke-like (n=7, 14.00%).

In relation to sub-group variations, participants from a low socio-economic area (26.09%) and those with a low general health (25.00%) reported having severe migraines more frequently compared to the general population (14.00%), while those with a high general health reported this less frequently (0.00%). In relation to gastrointestinal distress, participants who had a high school or trade education reported this less frequently (11.54%) while those with a university education (33.33%) and those that are hearing impaired (37.50%) reported this more frequently than the general population (22.00%). Participants with a university education (20.83%) and participants with hearing impairment (20.83%) reported diabetes being a condition that led to their diagnosis more frequently than the general population (10.00%). Participants with high physical function (40.91%) reported experiencing fatigue and/or lack of stamina more frequently than the general population (28.00%) while those with low physical function reported this less frequently (17.86%). Participants with high social function (40.00%) also reported experiencing fatigue and/or lack of stamina more frequently than the general population (28.00%).

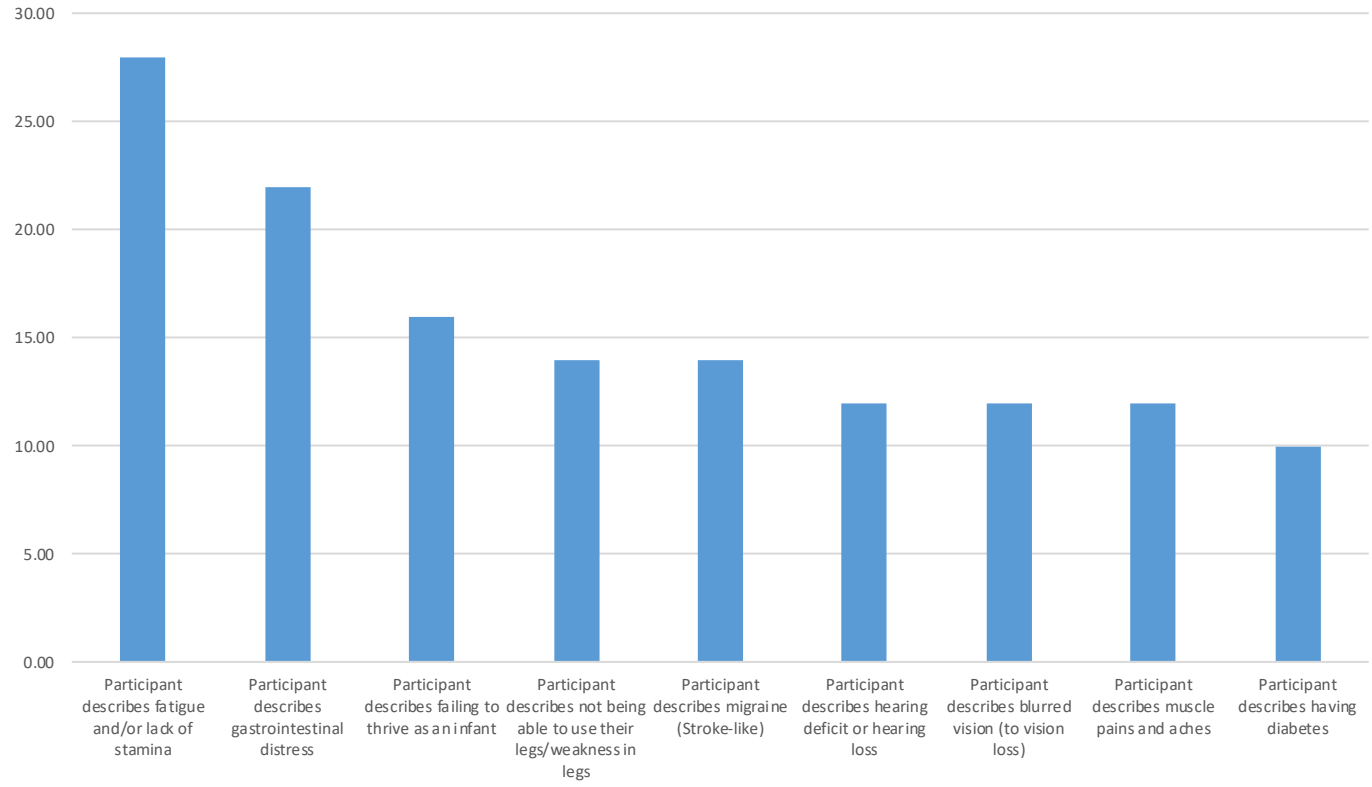


Figure 3.2: Symptoms leading to diagnosis (Percentage of all participants)

Participant describes fatigue and/or lack of stamina

So my earliest symptoms were two, then I had always problems with the cold, given my body temperature. Trouble exercising, I couldn't do what the other kids do. I just couldn't keep up, but I damn well tried. I just had to rest a lot, go to bed early. Which I didn't think anything of, and nobody else did then either. Participant 24

Okay. Well, I guess early on it was fatigue and also knowing that I was just hopeless at sports. Whereas, I've got five siblings who were all quite well coordinated and good at sport. I wasn't, which was quite embarrassing sometimes, but I was supposedly normal…My teachers in primary school and in secondary school, two different teachers at different times said to me, you need an earlier bed time, you must be staying up too late. I was embarrassed to tell them actually I went to bed at the same time as my younger sister. I wasn't staying up too late. That wasn't why I was tired. Participant 34

Yep. I was born with it. I was a little bit delayed in comparison to everybody else. Sorry. I always lacked the energy that everybody else had and that I noticed from around the time I was about 14, 15. And then, growing up, I just didn't have the stamina. Of course, I was working … I was always constantly … I would always need like ten hours sleep. Otherwise, I didn't feel well. Participant 40

Participant describes gastrointestinal distress

I thought the symptoms for those is celiac disease, because I was actually diagnosed with celiac disease. I suddenly got all these symptoms of gut problems. I was sure I've gotten rid of all the gluten out of my diet and I was having all these symptoms. Participant 21

I was also getting really fatigued, and then I also had this weird diarrhoea, like before my menopause, I went into menopause about 55, and I'm now 60. Before my menopause every time I had my period I would get diarrhoea, just a sudden cramping, and have to go to the toilet in a hurry and then within five years that diarrhoea became more and more problematic. Then before I retired, I had to retire my work because I couldn't maintain it, I was having diarrhoea maybe 10 times a week, unexpectedly. Now it's back to just maybe just once a month. Participant 36

Starting to see problems with bowel and bladder and so forth. She'd already started to get issues with her stomach, feeling sick in the mornings. Participant 47

Participant describes failing to thrive as an infant

It's my son with mitochondrial disease and it was around seven months of age, six, seven months of age, and he wasn't sitting up, and he wasn't responsive to a lot of ... I pretty much had noticed that other mothers, you know, mothers group and all the rest of it, children weren't doing the same things. He wasn't reaching the same milestones. At that point in time, and I think for my son, NAME, I had to express my concern with the community nurse. It was the GP and they thought it might have just been muscular dystrophy and I think NAME was around that time, around the six, seven-month mark he had his first cold and that's where he went down. He just slept, and slept, and slept and slept. Still, at that point, no alarm bells where kind of going off other than I remember the GP suggesting that perhaps it might be a muscular thing, the lack of sitting up and being to hold things. That it might be best to go and see the paediatrician. At that point, I had the referral. I made the appointment to the local paediatrician. ...I remember my husband arriving home and I was in tears thinking there is something wrong here. Anyhow, I went to our appointment armed with a list of questions, and feedback, and on that list was that NAME would do this strange hiccupping thing, and then just fold into himself, and I'd only witnessed him do that four times and for whatever. As soon as I mentioned that the paediatrician guy, I saw the look on his face and he kind of just went, can you tell me more about that? Then on queue for whatever reason, NAME proceeded to have what I now know was an infantile spasm. Participant 45

So, right from birth. So we didn't know that it was mito at the time, but it was poor feeding, failure to thrive, that sort of thing. Participant 46

She was really, really lethargic before she was born. This is my third child, so I noticed that she was significantly inactive and I kept having to go onto those monitors. This is 18 years ago so they were built. To try and trace movement, they traced very light movement only and then when she was born, she had problems with feeding; just no strength to suck. Then she was just lethargic, so I had to develop and then quite a long way behind. At 12 months, she still wasn't rolling over and things like that. She had no head control. There's a lot of those developmental muscle that's not ranged in that 12 months. Participant 49.

Participant describes not being able to use their legs/weakness in legs

Well, I wasn't born with it but I noticed it in early 2012. I was at lunch with my wife and young kids and I sat down at the table. As we finished our lunch, we went to get up and go and I was on my lunch break at work and I couldn't get out of the chair. I was very surprised with that. I thought, "What's the matter with my legs?" I can't push myself up and struggled when I got up. I had problems from there. Participant 6

I felt as though it came in cycles, that my fatigue would get a lot worse for a while and then I would have all these muscle problems in my legs, my thighs and that went on for 40 years, I suppose, almost. Participant 34

Well I've been OCCUPATION, and probably about seven or eight years ago I started to notice that I was having trouble doing some of the exercises that I normally do, particularly squats, getting up from the floor. It started off that I had to use my hands to push up from the floor, and then that gradually increased over a number of years to being having to put the right foot forward, and having these and particular arrangements, the left leg back and blah, blah, blah. Then I was up to the point where I have to get a chair, and push up from the chair rather than from the floor and stuff and- ... have my legs in a certain arrangement and ... Yeah, so the difficulty getting up from the floor or out of chairs has got increasingly worse. Participant 36

Participant describes migraine (Stroke-like)

My first noticeable symptoms that I remember are at aged 8 suffering from chronic migraines and cyclical vomiting. Participant 30

As far as I know I was born with it. I had a lot of different symptoms through my early childhood with constipation, migraine and things like that. But it wasn't until later and it wasn't until I had a stroke like migraine that it came out, that was in Christmas, December 2015. Through various doctors, neurologists and things like that and they did a muscle biopsy. I'm still in the process of being diagnosed of actually which mitochondrial disease it is. Participant 35

It wasn't until my late forties…I kept getting constant migraines, and I'd get vomiting episodes, and like, auras, and feeling like I had a stroke. So, they sent me up to get a brain MI. Participant 40

Participant describes hearing deficit or hearing loss

Yeah. I think probably the first thing is that there wasn't a stage of mito. What happened to me is I've got, just remembering, the A genes would be the G gene or whatever, so my hearing started going in my 20th decade, and now I've actually linked it to mito. What happened was, my hearing started to go in my 20th, about 23, 24, and no one could explain it, and then why that started to happen. Participant 15

I noticed that my hearing had really deteriorated. I had noticed before 2004 that I was asking people to repeat themselves a lot but had seen an ear, nose and throat specialist and seen a audiologist who said yes, your hearing's not great but it's not time for any intervention then. Participant 20

Well, what I noticed first was hearing loss, really. I guess that was the first symptom for me. Participant 26

Participant describes blurred vision (to vision loss)

Yes, sure. I was diagnosed about three years ago. In the preceding couple of years, I'd started to notice when I was reading, particularly when I was tired I would get a separation of the lines that I was reading, almost like a double vision just of that line. I thought it was bad contacts or bad reading glasses. Basically, I ended up going to a different optometrist who then went, "Something's not right". They sent me off to a specialist, and he said, "You got CPEO", which is obviously part of the mitochondrial thing, and then from there I've had other issues develop in that time since then. I was having reading issues. It was Dr. NAME at the LOCATION, and I don't know what the tests were called. There was a whole barrage of eye tests that they did. He also sent me off to have neurological. and they got me to do an MRI on my brain to make sure it wasn't a tumour that was causing it. I also went and saw another specialist who did tests on my legs and things like that. Participant 2

I lost my central vision in YEAR, so age eight. Participant 13

On and off and that was peculiar, what happened to me, but nobody ever put it down to anything in particular of course. It wasn't until I was query about my eyesight, which only happened 18 years ago. I was living in LOCATION. I was having trouble, I couldn't see in the dark. I found it hard finding things in a handbag and things like that, if anything was dark. When I went to get a new prescription for glasses, I was told, "Well, there's these strange pigments on your retinas and do you have trouble seeing in the dark?" and I said, "Yes." I knew that I had a nephew who'd been diagnosed with something to do with retinitis pigmentosa and so I said so, "Well I want to follow it up." I was sent to LOCATION Eye Hospital because we were living in LOCATION at the time. They said, "It's very rare what we're seeing in your eyes. We've only seen once or twice before and it's probably a mitochondrial disease." I said, "What's that?". Participant 34

Participant describes muscle pains and aches

Sorry, I was diagnosed as an adult. I guess my experience is a bit…There might have been times that I showed symptoms as a child, but who knows now, whether or not, because I have this random stuff. If looking back on my time, because I've written it down…it was a couple of times in 2014 and then in 2015, I have about these pains, so like I'd pain all through, like....all around my joints. I had in my hand, and then it just felt like I had pain in my elbows and my knees. It felt like it was in my joints at the time I rested and went to my doctor and he said like it might be a viral thing, so he gave me steroids. I stayed home for like a few weeks and have the steroids and it kind of went away after a while. After

trying maintaining parameters and stuff; and then it came back again, did the same thing. Then it came back in, November 2015. The pain, I could feel more in my legs, not just in my joints anymore. It really like spread and then really understood then it was muscular, not joints I had the pain come back again, but it never went away this time. I tried the steroids, I tried different drugs and nothing happened. I went to rheumatologist for a long time. Participant 5.

Yes. It was January in 2012. I just woke up one morning, I was fine, went to work the day before. A little bit cold not fluey, like I had head cold coming on but nothing else. I woke up the next morning and my feet were so swollen, I couldn't walk on them. My hands were swollen and wouldn't move. My whole body from head to toe just the pain that was crossing through my body was just unbelievable. Participant 18

I was sent to a specialist arthritis doctor, Dr. NAME down at LOCATION. After three visits to him, he looked at me and he said, "I'll see you in six months." I said, "No you won't doctor, I'm sorry. You're not listening to me, I said I have not got joint pain, I have got muscular pain. It's in my thighs mainly and my calves. When I hang washing out I hurt. It's just muscular." Anyway, we came back and they were just giving me painkillers and things like that…. When I look back to when I'm young, we lived at LOCATION for 21 years, I used to work at PLACE and I'd walk, which would be almost both half a mile. For all the walking, playing squash, doing exercise and everything that I did in my younger days, I always had very sore legs, sore shins and I just took that as life. Everyone must have be like that, I never talked about it, I didn't asked questions and other little odd things that I used to get pains, unexplained. I'd put up with them for a week and I'd go to the doctor and they'd send me to scans. Nothing would be wrong with me, you forget about it and the pain eventually would go, that still happens. Anyway, that's it. Participant 31

Hereditary conditions noted in relation to diagnosis

As part of the structured interview analysis in relation to symptoms that lead to diagnosis, there were 13 participants (26.00%) that noted a hereditary component that led to their diagnosis. In some cases it was a known hereditary link while in others, the hereditary link was identified as part of the diagnostic process.

No. A lot of different things had happened. What happened was I had a MRI done and the specialist that I was seeing initially, he actually was treating my sister. When he was just looking at my MRI he went, "Hang on, I've seen similar." He got my sisters in my eyes and put them together and there was all these white lesions on our brain and that kind of thought that's when we got in touch. Participant 1

My mum was there at that time and she probably had a memory flash up because my…I have found a file and that someone had been diagnosed with LHON which we were never actually told about. She didn't really know anything about it, but it just rang a bell for mom. She went home, looks for documentation and saw that her mother, my grandmother or my brother's grandmother had been diagnosed with this condition. I took it straight back to this ophthalmologist who said, "Well, really sorry to tell you but it's genetic. It's inherited on the maternal line. You need to get tested but it's pretty much 100% certain already. That's what this is or at least that you carry it and given the similarities, that would be it manifesting." Participant 8

Yes, I've inherited it from my mum but I didn't know I had it until I had the muscle biopsy and I didn't have any conditions then, I do now. Participant 16

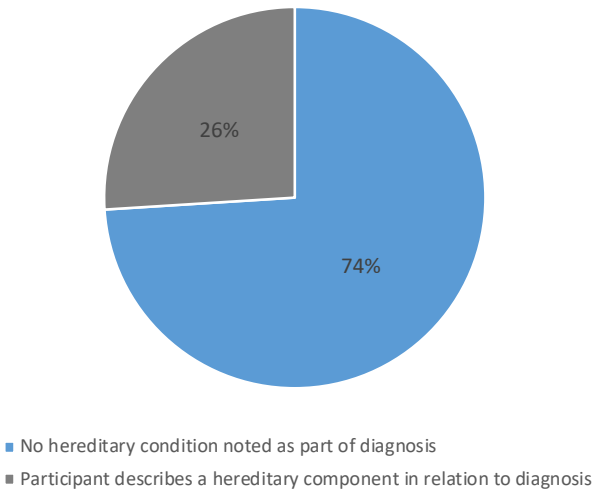


Figure 3.3: Hereditary condition noted as part of diagnosis

Misdiagnosis and other conditions suspected

During the description of their diagnosis, there were a number of participants that noted that they were misdiagnosed or another condition was suspected before their mitochondrial disease diagnosis including multiple sclerosis, muscular dystrophy, fibromyalgia and chronic fatigue.

Table 3.2: Conditions misdiagnosed or suspected at diagnosis.

Conditions misdiagnosed or suspected before diagnosis	All participants	
	n=50	%
Muscular dystrophy suspected or misdiagnosed	3	6.00
Rheumatoid arthritis suspected suspected or misdiagnosed	3	6.00
Diagnosed through other investigation or treatment/therapy regime	2	4.00
Multiple sclerosis suspected suspected or misdiagnosed	1	2.00
Fibromyalgia suspected or misdiagnosed	1	2.00
Chronic fatigue syndrome suspected or misdiagnosed	1	2.00

Multiple sclerosis suspected

I was reading a book one night and I was like, "Oh, I'm shutting one eye to read." I tried to not shut my eye to read and I realized that I couldn't because I was getting double vision. That's when I realized there must have been something wrong. I went and saw another neuro-ophthalmologist then. I was living in LOCATION at that point, went and saw another neuro-ophthalmologist who thought that I had MS. He sent me for testing for MS. Then when that came back all clear, he sent me for a blood test and I remember hearing the word mitochondrial, but he never explained what it was, never really said what he was doing or looking for or anything. Participant 10

Muscular dystrophy suspected

I pretty much had noticed that other mothers, you know, mothers group and all the rest of it, children weren't doing the same things. He wasn't reaching the same milestones. At that point in time, and I think for my son, NAME, I had to express my concern with the community nurse. It was the GP and they thought it might have just been muscular dystrophy and I think NAME was around that time, around the six, seven-month mark he had his first cold and that's where he went down. He just slept, and slept, and slept and slept. Participant 45

Fibromyalgia suspected

He tried to figure out what it was, almost diagnosed me with fibromyalgia, but then it didn't really add up. This is fatigue and everything else, and she referred me to a neurologist who did a muscle biopsy. Participant 5

Chronic fatigue suspected

Well, I think first, I was diagnosed as having chronic fatigue after I had a diagnosis of-- What's it called? (Glandular fever) Glandular fever. Yes. That's it. When I was 21 and well, I didn't get chronic fatigue diagnoses until about, I don't know about 15 years later or something. Participant 34

Diagnostic pathway

Participants noted in the questionnaire the approximate date when they first noticed symptoms and then the approximate date when they were diagnosed. From the information reported, from those participants were an approximate time lapse could be made (n=44), the range of time between symptoms and diagnosis was from 1 month to 58 years. Almost half of the participants were diagnosed within two years of noticing symptoms (n=19, 43.18), a quarter of participants were diagnosed between 3 and 5 years from noticing symptoms (n=11, 25.00%), and approximately a third of participants were diagnosed more than six years after experiencing symptoms (n=14, 31.81%).

Table 3.3: Time from symptoms to diagnosis

Approximate time between noticing symptoms and diagnosis (n=44)	N=	Percentage of participants
< 1 year	4	9.09
1-2 years	15	34.09
3-5 years	11	25.00
6-10 years	3	6.82
10-20 years	7	15.91
>20 years	4	9.09

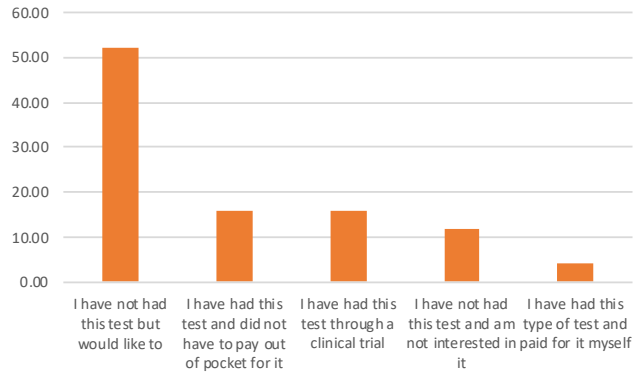


Figure 3.4: Time from symptoms to diagnosis (Percentage of all participants)

As noted, in the structured interview, participants were asked about symptoms leading to diagnosis and how the participant came to be diagnosed. As reflected in the results above, there were some participants that had a relatively straight forward diagnosis and others that had a long and complicated pathway to diagnosis. The following quotes are provided to demonstrate these variations:

Long, complicated diagnosis

I thought the symptoms for those is celiac disease, because I was actually diagnosed with celiac disease. I suddenly got all these symptoms of gut problems. I was sure I've gotten rid of all the gluten out of my diet and I was having all these symptoms and realized they were the symptoms my daughter was getting. My daughter and I compared symptoms and I went to a dietitian and she said, "Have you done this? Have you done this? Have you done this?" I've done all those things and yet I was still sick. I was going to a gastroenterologist who said, "It must be psychological." No, you're far out. This is exactly happened with the daughter. At the same time I was seeing the psychiatrist, I didn't get that far. I don't think I did. I was seeing the gastroenterologist and he said, "Well, if it was for your daughter having the same symptoms, I would have said it was psychiatric, because if some daughter being sick and da-da-da." He said, "But I've noticed this NAME in LOCATION." At the same time, my GP said, "I've heard of this NAME in LOCATION."...she's a professor at LOCATION.... Participant 21

Well it took a long time. I'd seen lots of different specialists. Finally, they picked up rheumatoid arthritis marker, so I was sent to a Rheumatologist, and when I went to him he had a ... What do you call it? A locum or somebody had come up from I think it was LOCATION, and he actually noticed how I was getting out of the chair in the waiting room, and asked me about it. He was the first one who actually put his hands on and tested my muscles and went, "Oh, yeah they are weak." Even though I'd been telling multiple- ... specialists the same story for a long time, I was fobbed off for a long time. Like, "Oh, you don't exercise enough." Which I knew was wrong. I was also getting really fatigued, and then I also had this weird diarrhoea, like before my menopause, I went into menopause about 55, and I'm now 60. Before my menopause every time I had my period I would get diarrhoea, just a sudden cramping, and have to go to the toilet in a hurry and then within five years that diarrhoea became more and more problematic. Then before I retired, I had to retire my work because I couldn't maintain it, I was having diarrhoea maybe 10 times a week, unexpectedly. Now it's back to just maybe just once a month….Then I got referred to a neurologist who did muscle biopsies in my leg and my shoulder. They had ragged red fibres, so he suspected it was mitochondrial and referred me to NAME in LOCATION. Participant 36

Well it took a long time. I'd seen lots of different specialists. Finally, they picked up rheumatoid They took a ... it was a very, very long process. Probably two and a half years. Was a muscle biopsy and blood test. And they went to the Netherlands, and it was basically a point mutation. Yeah so it was a.... I forgot what they call it, but yeah it was a mutation…But that was the very final one. Participant 46

Relatively straight forward diagnosis

Okay. So the major symptom was just ptosis. Pretty slight ptosis on the left side. Yes, I don't know if this is relevant. But I just recently started using Snapchat and I was taking a lot of selfies. Yes I just, I guess when you take a lot of selfies you sort of notice things that weren't really there before. That would be the first symptom, and I went to see an ophthalmologist. Just to ask what was up with the ptosis, and expecting just him to say that it is just a bit of a lazy eye. But no, then he did a couple of really simple tests, just test my bilateral eye movements. Stuff like that and that it was lacking- he ask, how long have I had double visions or stuff like that. He essentially diagnosed. It was a very quick diagnosis, mitochondrial disease. Then obviously I have to get like biopsy and stuff just to confirm. Participant 11

I took my mum for an appointment to see a neurologist NAME and he said, "Oh, looking at your daughter " Straight away just looking at her, said, "Oh, I feel you got Mitochondrial myopathy." She had muscle biopsy and then he said, "Looking at the daughter, I feel she's showing the signs of it too." That would have to do with the ptosis of the eyes. Participant 16.

I was diagnosed in 2003. It all came about because my mother who also had it was in hospital…. They couldn't work out why she wasn't getting better. Based on some other family information, they tested her for mitochondrial disease. It turned out that she had it. At the same time, I was having a lot of symptoms with fatigue and headaches, and just not being able to do as much as I used to. I got tested as well. That's how that came about. I had started with a muscle biopsy. I was officially diagnosed by a muscle biopsy. Probably what I've just described. There were some other family members that were suspected to have it. I went to my GP and she referred me to a specialist who organized a muscle biopsy. Participant 43

Costs at diagnosis

In the questionnaire, participants were asked to estimate the amount of out of pocket expenses they had for diagnostic tests and medical consultations. Twenty-one participants (42.00%) had no out of pocket expenses, 13 participants (26.00%) spent more than $1000, 8 participants spent $1000 or less (16.00%), the remaining 8 participants were unable to recall how much they spent (16.00%).

Table 3.4: Costs of diagnosis

Cost	N=50	Percent
0	21	42.00
$0-500	2	4.00
$501-1000	6	12.00
>$1000	13	26.00
Not known/can't recall	8	16.00

The participants were then asked on the online questionnaire if the amount they spent was a burden, for half of the participants (50.00%), it was no burden at all, 14 participants found it extremely or moderately significant (28%), and 11 participants found it with somewhat or slightly significant (22.00%).

Table 3.5 Cost of diagnosis – level of burden

Cost a significant burden	N=50	Percent
Extremely significant	6	12.00
Moderately significant	8	16.00
Somewhat significant	5	10.00
Slightly significant	6	12.00
Not at all significant	25	50.00

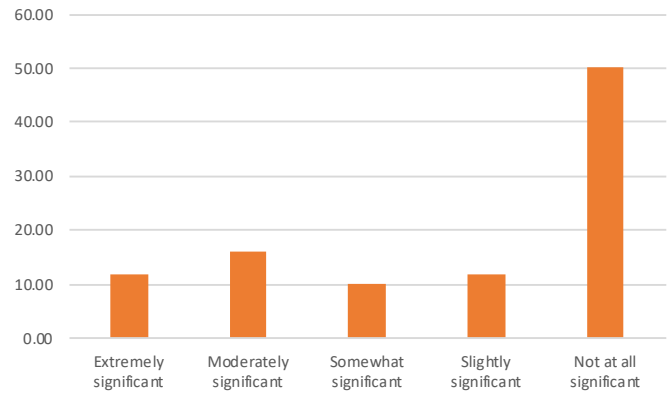


Figure 3.5 Cost of diagnosis – level of burden
(% of all participants)

Understanding of disease at diagnosis

Participants were asked how much they knew about mitochondrial disease at diagnosis. There were 31 participants (62.00%) that described knowing nothing about mitochondrial disease and this was the most common response.

Participant describes knowing nothing about mitochondrial disease at diagnosis

I didn't know anything about it before then. I've never heard of it. Completely new...shocked. I did hit the books so I did a lot of research. I helped inform my family a bit when we couldn't get to the doctors. I took that on myself a little bit. I ended up getting involved with AMDF. You've probably been in touch with them then. Participant 8

No, I never understood because I had my mum living with me for 10 years. I never understood why mom was always tired and basically, she'd look like she was sleeping all the time when she wasn't because of her. She had really bad ptosis over the eyes but she was always in bed by five o'clock, six o'clock in the evening. I never understood the condition where now, I understand the condition because I'm always ready for my bed at six o'clock. Participant 16

We didn't know anything, but it was good to get an answer about everything. Yes, because as I say, I was getting lots and lots of migraine headaches and just having lots of time off work, just getting so sick. Everything wasn't going down a normal pathway of diabetes treatment. It was good just to get an answer, but then there was nobody... It took us a while to find someone that actually knew what it was. Now, I'm under neurology. I see Dr. NAME down there. He's been helping us go through and somehow, we found out about the Mitochondrial Foundation and get the newsletters but no, we didn't know anything about it. It was very unknown and still is very unknown. Participant 19

There were also eight participants (16.00%) that described knowing about mitochondrial disease by the time they were diagnosed because the time to diagnosis was relatively long, giving them time to educate themselves.

Participant describes knowing about mitochondrial disease as the time to diagnosis was relatively long, giving them time to educate themselves

Well, by that point I did know a bit because there had been a bit of time between when I first had the word mentioned to me to when I got a diagnosis. I had a bit of time to get on to doctor Google and read up about it and because the neuro-ophthalmologist that I saw initially, he clinically diagnosed me with Kearns-Sayre syndrome from my vision loss and everything that I've got. Then I had a narrowed field, so then I could have a look and research that. By the time I got a diagnosis, I was pretty up there with what was happening and what it was. Participant 10

When I was diagnosed, I knew a little bit because I had spoken to the ophthalmologist about it. Well he said that mitochondrial disease so I did a research myself, so I guess I did not entirely know the cause or much about any treatment or anything like that but I would say I had like a decent amount of knowledge by the time I was diagnosed, because it was kind of ongoing process as well. Okay, right. So I was just going to say that because I think there was about an extended gap, like a two-year gap between when I first saw the ophthalmologist and when I got diagnosed, saying between that two years, I was sort of looking up more or less everything I could about it. To learn a little bit. Participant 11

Well, we actually bought it up with the doctors because I've been an inpatient for four or five months at that point. We were getting literally nowhere. I bought it up. Then, it took us another three months to get a consult with the metabolic team. Well, I guess that's why we've been really onto this because my symptoms fit very well....Everything else has been excluded at that point. We didn't really understand why all of these bodily systems were malfunctioning. It does makes sense....In actual fact, as I kept telling them at the time, "A...teenager sitting in bed, in your hospital, has pretty much diagnosed herself." Because she literally had every single symptom listed on the website. We couldn't explain it with anything else. It just seemed to be the most obvious. Participant 47

Table 3.6: Understanding of disease at diagnosis

Understanding of disease at diagnosis	All participants	
	n=50	%
Participant describes knowing nothing about mitochondrial disease at diagnosis	31	62.00
Participant describes knowing about mitochondrial disease as the time to diagnosis was relatively long, giving them time to educate themselves	8	16.00
Participant describes knowing very little about mitochondrial disease at diagnosis	7	14.00
Participant describes knowing about mitochondrial disease before diagnosis (scientific background)	2	4.00
Participant describes no-one knowing much about mitochondrial disease and the uncertainty of the diagnosis	2	4.00

Support at diagnosis

In the questionnaire, participants were asked whether they felt supported at the time of diagnosis. There were 36 participants (72.00%) that indicated that they had no support at diagnosis, while 3 participants (6.00%) noted that they had enough support. An additional 11 participants (22.00%) indicated that they had some support but that it was not enough.

In relation to sub-group variations, participants with no eye problems reported having no support at diagnosis more frequently than the general cohort (81.25% compared to 72.00% in the general cohort),. Participants that had higher general health reported that they had no support at diagnosis, more frequently than the general cohort (86.36% compared to 72.00% in the general cohort), and reported less frequently than the general cohort that they had some support, but it wasn't enough, (13.64% compared to 22.00% in the general cohort).

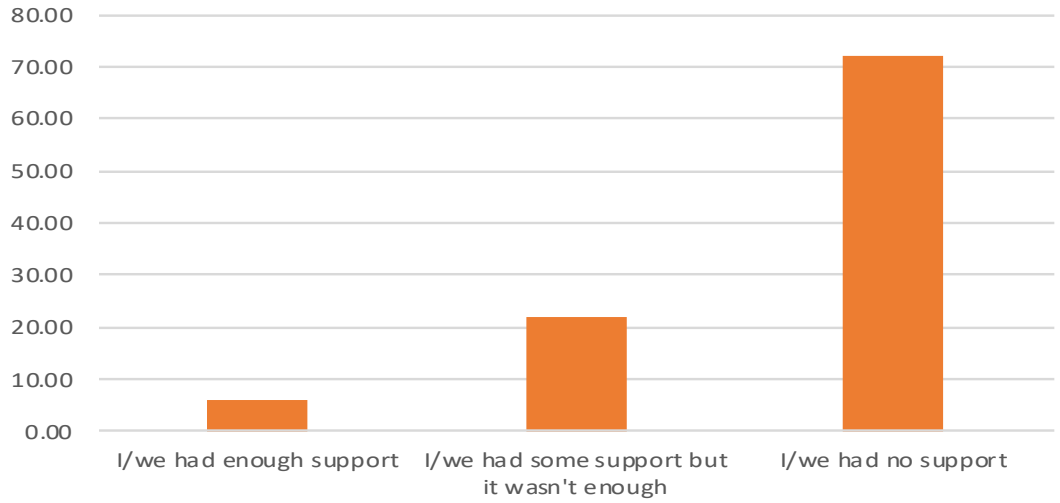


Figure 3.6: Support at diagnosis (% of all participants)

Table 3.7 Support at diagnosis

			General Health		Physical function		Emotional well-being	
Support at diagnosis	N=50	Percent	Higher N=22	Lower N=28	Higher N=22	Lower N=28	Higher N=26	Lower N=24
I/we had enough support	3	6.00	1 (5.00%)	2 (7.14%)	1 (4.45%)	2 (7.14%)	1 (4.55%)	2 (6.67%)
I/we had some support, but it wasn't enough	11	22.00	4 (20.00%)	17 (25.00%)	4 (18.18%)	7 (25.00%)	4 (18.18%)	7 (23.33%)
I/we had no support	36	72.00	15 (75.00%)	19 (67.86%)	17 (77.27%)	19 (67.86%)	17 (77.27%)	21 (70.00%)

	Social functioning		Hearing impairment		Eye/visual impairment	
Support at diagnosis	Higher N=20	Lower N=30	No hearing problems N=26	Hearing problems N=24	No eye problems N=16	Eye problems N=34
I/we had enough support	0 (0.00%)	3 (10.71%)	1 (3.85%)	2 (8.33%)	0 (0.00%)	3 (8.82%)
I/we had some support, but it wasn't enough	3 (13.64%)	8 (28.57%)	5 (19.23%)	6 (25.00%)	3 (18.75%)	8 (23.53%)
I/we had no support	19 (86.36%)	17 (60.71%)	20 (76.92 %)	16 (66.67%)	13 (81.25%)	23 (67.65%)

	Location		Education		SEIFA	
Support at diagnosis	Metropolitan N=30	Regional N=20	School/ Trade N=26	University N=24	Higher N=27	Lower N=23
I/we had enough support	1 (3.33%)	2 (10.00%)	2 (7.69%)	1 (4.17%)	1 (3.70%)	2 (8.70%)
I/we had some support, but it wasn't enough	6 (20.00%)	5 (25.00%)	6 (23.08%)	5 (20.83%)	5 (18.52%)	6 (26.09%)
I/we had no support	23 (76.67%)	13 (65.00%)	18 (69.23%)	18 (75.00%)	21 (77.78%)	15 (65.22%)

Diagnostic tests

Participants were asked on the online questionnaire what tests that they had received leading up to their diagnosis with mitochondrial disease. The majority of participants had blood tests (n=43, 86.00%), medical history (32, n=64.00%), genetic tests (n=31, 62.00%), muscle or tissue biopsies (n=31, 62.00%), eye tests (n=30, 60.00%), urine tests (n=27, 54.00%) and family history (n=26, 52.00%).

Table 3.8: Diagnostic tests

Diagnostic test	N=50	Percent
Blood tests	43	86.00
Medical history	32	64.00
Genetic tests	31	62.00
Muscle/Tissue biopsy	31	62.00
Eye tests	30	60.00
Urine tests	27	54.00
Family history	26	52.00
Hearing test	24	48.00
Hair tests	9	18.00
Imaging(Ultrasound, MRI, CT, X-ray)	5	10.00
Nerve conduction tests	5	10.00
Skin cell tests	5	10.00
Lumber puncture	2	4.00
Electroencephalography	1	2.00
Endoscope	1	2.00
Exercise testing	1	2.00

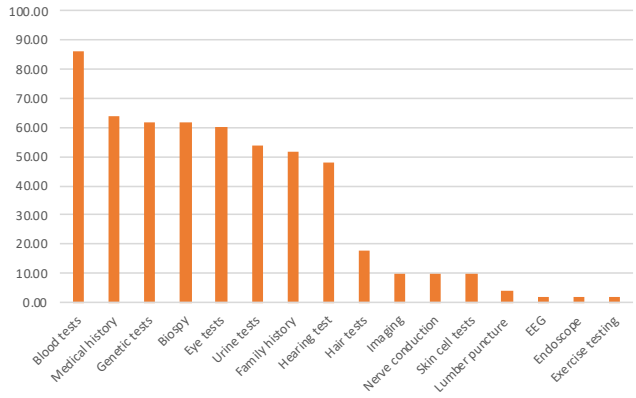


Figure 3.7: Diagnostic tests (% of all participants)

Time from diagnostic tests to diagnosis

Participants were asked on the online questionnaire about the amount of time from diagnostic test until they received a diagnosis. The time ranged from less than one week to 27 years. The majority of participants described the time in months (n=28, 56.00%), others described the time in weeks (n=11, 22.00%), years (n=8, 16.00%) or not known/still waiting for diagnosis (n=3, 6.00%).

Table 3.9: Time to diagnosis

Time from tests to diagnosis	n=50	Percent
< 1week	2	4.00
1-2 weeks	1	2.00
2-3 weeks	7	14.00
3-4 weeks	1	2.00
>4 weeks	36	72.00
Don't know	3	6.00

Diagnosis delivery

Participants were asked who gave them their diagnosis and where the diagnosis was given. The majority were diagnosed by a neurologist (N=23, 46.94%), followed by a geneticist (n= 9, 18.37%) and mitochondrial specialist (n= 7, 14.00%).

Table 3.10 Diagnosis provider

Health professional who gave diagnosis	N=49	Percent
Neurologist	23	46.94
Geneticist	9	18.37
Mitochondrial specialist	7	14.29
Eye specialist	4	8.16
Ophthalmologist	2	4.08
Functional medicine specialist	1	2.04
Gastroenterologist/Digestive system specialist	1	2.04
Neuro-ophthalmologist	1	2.04
ophthalmologist + ENT specialist	1	2.04

Most participants received their diagnosis at a specialist clinic (n=24, 48.98%), followed by the hospital (n=18, 36.73%).

Table 3.11 Diagnosis location

Where was diagnosis given	n=49	Percent
Specialist clinic	24	48.98
Hospital	18	36.73
General practice	5	10.20
By letter	1	2.04
By phone	1	2.04

Table 3.12 Genetic and biomarker tests

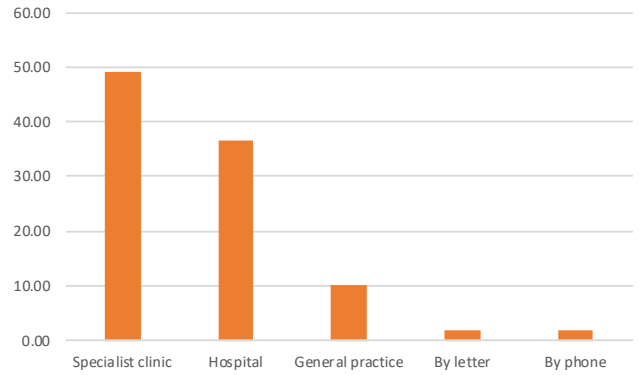


Figure 3.8 Diagnosis location

Genetic and biomarker tests

Participants were asked whether they had ever had a discussion about genetic tests or tests to see if there were biomarkers that might be relevant to their condition or treatment. There were 6 participants (12.00%) that indicated that they had brought up the topic for discussion with their doctor and 15 participants (30.00%) that reported that their doctor had brought up the topic for discussion. There were also 29 participants (58.00%) that indicated that no one had ever spoken to them about this.

In relation to sub-group variations, participants with higher social functioning indicated that their doctor brought up the topic of biomarker/genetic testing, more frequently than the general cohort and those with lower social functioning less frequently (higher social functioning 45.00%; lower social functioning 20.00%, compared to 30.00% in the general cohort). Participants with higher social functioning indicated that no one brought up the topic of biomarker/genetic testing, less frequently than the general cohort and those with lower social functioning more frequently (higher social functioning 40.00%; lower social functioning 70.00%, compared to 58.00% in the general cohort). Participants with no eye problems indicated that no one brought up the topic of biomarker/genetic testing, more frequently than the general cohort (68.75%, compared to 58.00% in the general cohort).

Genetic testing	All participant	% of all participants	General Health		Physical function		Emotional well-being	
	N=50	Percent	Higher N=22	Lower N=28	Higher N=22	Lower N=28	Higher N=26	Lower N=24
I brought up the topic with my doctor for discussion	6	12.00	1 (4.55%)	5 (17.86%)	2 (9.09%)	4 (14.29%)	3 (11.54%)	3 (12.50%)
My doctor brought up the topic with me for discussion	15	30.00	8 (36.36%)	7 (25.00%)	9 (36.36%)	7 (25.00%)	9 (34.62%)	6 (25.00%)
No one has ever spoken to me about this type of test	29	58.00	13 (59.09%)	16 (57.14%)	12 (54.54%)	17 (60.70%)	14 (53.85%)	15 (62.50%)

	Social functioning		Hearing impairment		Visual or eye impairment	
	Higher N=20	Lower N=30	No hearing problems N=26	Hearing problems N=24	No eye problems N=16	Eye problems N=34
I brought up the topic with my doctor for discussion	3 (15.00%)	3 (10.00%)	3 (11.54%)	3 (12.50%)	1 (6.25.00%)	5 (14.71%)
My doctor brought up the topic with me for discussion	9 (45.00%)	6 (20.00%)	7 (26.92%)	8 (33.33%)	4 (25.00%)	11 (32.35%)
No one has ever spoken to me about this type of test	8 (40.00%)	21 (70.00%)	16 (61.45%)	13 (54.17%)	11 (68.75%)	18 (52.94%)

	Location		Education		SEIFA	
	Metropolitan N=30	Regional N=20	School/ Trade N=26	University N=24	Higher N=27	Lower N=23
I brought up the topic with my doctor for discussion	2 (6.67%)	4 (20.00%)	3 (11.54%)	3 (12.50%)	2 (7.41%)	4 (17.39%)
My doctor brought up the topic with me for discussion	10 (33.33%)	5 (25.00%)	7 (26.92%)	8 (33.33%)	9 (33.33%)	6 (26.09%)
No one has ever spoken to me about this type of test	18 (60.00%)	11 (55.00%)	16 (61.45%)	13 (54.17%)	16 (59.26%)	13 (56.52%)

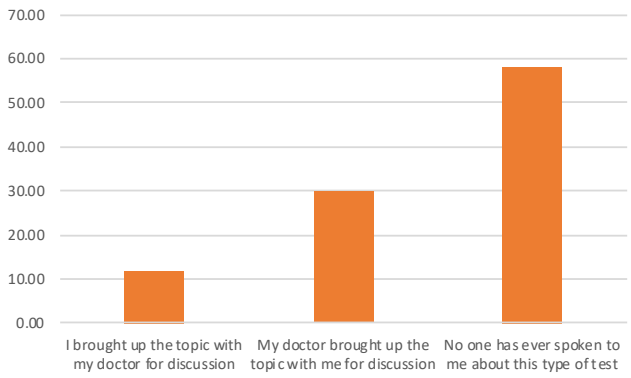


Figure 3.9: Genetic and biomarker tests (% of all participants)

Participants were also asked about their interest in this type of test if it was available. The majority of participants noted that they had not had this test, but would like to (n=26, 52.00%). There were 8 participants (16.00%) that reported having this test and not paying out of pocket for it, 8 had this test as part of a clinical trial (16.00%), and two paid for this test themselves (4.00%). There were 6 participants (12.00%) indicated that they had not had this test and were not interested in it.

In relation to sub-group variations, participants that had hearing problems, no eye problems and that were university educated indicated that they had not had this test but would like to, less frequently than the general cohort (41.67%, 31.25% and 33.33% respectively compared to 54.00% in the general cohort), while participants that did not have hearing problems, had no eye problems and had high school or trade qualifications indicated that they had not had this test but would like to, more frequently than the general cohort (61.54%, 61.76%, and 69.33% respectively, compared to 54.00% in the general cohort).

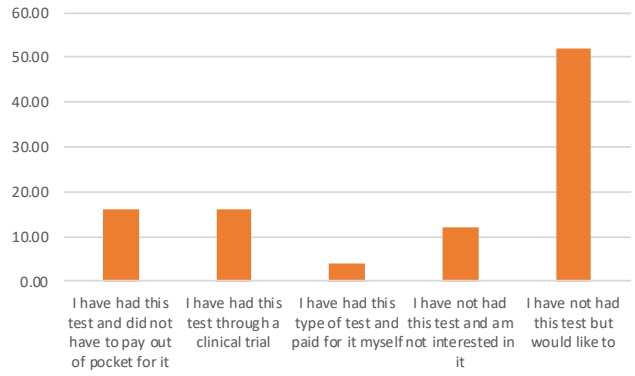


Figure 3.10: Interest in genetic and biomarker test (% of all participants)

Table 3.13: Interest in genetic and biomarker test

Genetic testing	All participants	All participants	General Health		Physical function		Emotional well-being	
	N=50	Percent	Higher N=22	Lower N=28	Higher N=22	Lower N=28	Higher N=26	Lower N=24
I have had this test and did not have to pay out of pocket for it	8	16.00	6 (27.27%)	2 (7.14%)	3 (13.64%)	5 (17.86%)	5 (19.23%)	3 (12.50%)
I have had this test through a clinical trial	8	16.00	3 (13.64%)	5 (17.86%)	5 (22.73%)	3 (10.71%)	5 (19.23%)	3 (12.50%)
I have had this type of test and paid for it myself	2	4.00	1 (4.55%)	1 (3.57%)	2 (9.09%)	0 (0.00%)	1 (3.85%)	1 (4.17%)
I have not had this test and am not interested in it	6	12.00	2 (9.09%)	4 (14.29%)	0 (0.00%)	6 (21.43%)	3 (11.54%)	3 (12.50%)
I have not had this test but would like to	26	52.00	10 (45.45%)	16 (57.14%)	12 (54.54%)	14 (50.00%)	12 (46.15%)	14 (58.33%)

	Social functioning		impairment		Eye or visual impairments	
	Higher N=20	Lower N=30	No hearing problems N=26	Hearing problems N=24	No eye problems N=16	Eye problems N=34
I have had this test and did not have to pay out of pocket for it	3 (15.00%)	5 (16.67%)	4 (15.38%)	4 (16.67%)	2 (12.5%)	6 (17.65%)
I have had this test through a clinical trial	5 (25.00%)	3 (10.00%)	3 (11.54%)	5 (20.83%)	1 (6.25%)	7 (20.59%)
I have had this type of test and paid for it myself	1 (5.00%)	1 (3.33%)	0 (0.00%)	2 (8.33%)	2 (12.50%)	0 (0.00%)
I have not had this test and am not interested in it	1 (5.00%)	5 (16.67%)	3 (11.54%)	3 (12.50%)	6 (37.50%)	0 (0.00%)
I have not had this test but would like to	10 (50.50%)	16 (53.33%)	16 (61.54%)	10 (41.67%)	5 (31.25%)	21 (61.76%)

	Location		Education		SEIFA	
	Metropolitan N=30	Regional N=20	School/ Trade N=26	University N=24	Higher N=27	Lower N=23
I have had this test and did not have to pay out of pocket for it	4 (13.33%)	4 (20.00%)	4 (15.38%)	4 (16.67%)	4 (14.81%)	4 (17.39%)
I have had this test through a clinical trial	5 (16.67%)	3 (15.00%)	2 (7.69%)	6 (25.00%)	4 (14.81%)	4 (17.39%)
I have had this type of test and paid for it myself	1 (3.33%)	1 (5.00%)	0 (0.00%)	2 (8.33%)	1 (3.70%)	1 (4.35%)
I have not had this test and am not interested in it	5 (16.67%)	1 (5.00%)	2 (7.69%)	4 (16.67%)	4 (14.81%)	2 (8.70%)
I have not had this test but would like to	15 (50.00%)	11 (55.00%)	18 (69.23%)	8 (33.33%)	14 (51.85%)	12 (52.17%)

Participants were asked if they had any particular mitochondrial disease biomarkers, the majority of participants (n=39, 78.00%) were not sure.

Table 3.14 Biomarkers

Biomarkers	n=50	Percent
I'm not sure	39	78.00
m.3232A>G	5	10.00
m 3243A>G	2	4.00
11778	1	2.00
FGF21,GDF15,m.3232A>G,	1	2.00
m.3302A>G	1	2.00
m 3113 A>G	1	2.00

In the structured interview, participants were also asked to talk about their understanding of genetic or biomarker testing. Some of the descriptions are provided below and include understanding that the test is used for diagnosis of mitochondrial disease; understanding that the test cannot help them but may help others in the future; and understanding that the test cannot target treatment as there are no treatments available or that there was no clinical indication following the test:

Participant understands that the test is used for diagnosis of mitochondrial disease

That it would just give me a name to at least put to and allow me to understand the cause of the vision loss, which was a good thing. Participant 13

The genetic treatment was, again, under NAME. I'm not too sure of the exact results, genetically speaking, but whatever it was, it was enough to confirm the diagnosis that they had been making through the years. Participant 23

It was partly for our peace of mind to know what was actually wrong with us. We weren't just fit or lazy, but there was actually a diagnosable illness. It was probably for our peace of mind. Participant 43

Participant understands that the test can not help them but may help others in the future

I didn't believe that there was anything that could help me because they keep saying there's no treatment, so basically, I was of the opinion that it was more for potential of assisting in the future for other people. Participant 2

I don't know whether they were going to help me. We were under the understanding they might help other people. Participant 41

Participant understands that the test can not target treatment as there are no treatments available or that there was no clinical indication following the test

Well, actually, they've got it wrong because there isn't anything they can do with it, there is only so…there is no treatment or I've had as much treatment as I can have. Participant 9

….so it wasn't until after his diagnosis that we sort of talked more about their use, which was kind of like a moot point kind of thing, really. Participant 46

Communication and understanding of prognosis

Participants were asked whether anyone talked to them about prognosis. The most common theme noted by 26 participants (52.00%) was prognosis had not been clearly discussed:

Participant describes prognosis not being discussed

No, nothing. All we've received is what I've said about Dr. NAME a couple of years ago. The name and I received some information from AMDF by Googling and NAME at AMDF. Also we went down recently to NAME Hospital and went to a clinic there, run by NAME. They took 15 vials of blood and 5 urine jars, and I've got to do further testing with them at the LOCATION Hospital. I did an Echocardiogram recently about two weeks ago. They got to do further tests and they're going to contact me in about six months, they said. Participant 6

No I can't because it's such a fickle thing…I think my neurologist who I have a great deal of respect for, would say it's very difficult to make a prognosis as indefinite things, such and such will happen at such a time, or even what organs might be affected. Participant 20

No, no. I mean, that's the nature of the condition is that it is very ... the fact that he's holding on there. I mean, he has absolutely shocked everyone. No one would have thought that the child ... generally, when a baby presents as he did, they generally don't make it through and he's low abnormal mitochondrial cell load is 95 to 100% effective. He's quite the miracle. That's why we just keep on going. Participant 45

The next most common theme was that participants understood that mitochondrial disease came with a poor prognosis that was primarily related to physical decline. This was noted by 9 participants (18.00%)

Participant describes poor prognosis - decline in physical function

I'm already experiencing a bit of what I've been told is a drop foot. I've also get a lactic acid build up very quickly when I walk. If I walk up two flights of stairs, I feel like I'm weak in my legs. I have periods where I become very lethargic. I'm going to lose the ability to like I get out in the garden on a Saturday and get everything done and then Sunday I would go and do family stuff. Now, it takes me a weekend what I used to do in three-quarters of a day, simply because of the lethargy of losing strength. I've definitely noticed that in my arms and legs. I suppose the drawback there, if you feel that when you're told you need to get into the gym but instead of being sore for one or two days after, you're sore for a week or seven days after, which means to go start doing it, you're going to be constantly sore. It's a bit of a mental thing to keep overcome. Participant 2

My main issues which really troubles me is my calf muscles. I'm finding it difficult to walk around. Difficulty getting up off a chair, that's hard. I've got to struggle. I'm finding it difficult to walk around. Simply get up off a chair is hard. I can't get up without using the arms on the chair to push myself up. Uneven ground is difficult for me to walk on. Going upstairs, I can still use stairs by using a handrail or else I'll fall over. Participant 6

In the last three years, it seems to have got worse. I'm just more tired and I have no energy and very...I've had to quit my job. It seems very unknown, but it seems to be getting worse. Participant 19

There were seven participants (14.00%) that described the need for ongoing management of their condition and this included the management of exacerbations:

Participant describes a ongoing management of their condition, often with exacerbations

The current prognosis is it remains the same. As I said, I see Dr. NAME, the neurologist, every six months. If I do have a really bad three or four days, I'll give her a call and she gets me into an earlier session, but there's no basic cure, there's no...except pain medication which tends to..at the...in the pain management center. Participant 23

It's mixed. There are some days that are really quite frightening and I guess you sort of reflect on what your family's gone through and the symptoms they had, then you assume that you are going to get the same thing which isn't an easy thing to go through. I guess it makes it more challenging in terms of starting a family as well. That was always something that we had assumed would just happen. That's now presented a whole lot of challenges as well and also how is my health going to be in a few years and will I be a productive mother if we do have a child. There's a lot of ups and downs. Participant 26

I'm supposed to be on managing the diabetes side because part of it I've got eye problems. That's being monitored annually by an Ophthalmologist. That's slightly deteriorating. I have diabetes. That's been monitored. I'm supposed to control that. I know I should better than I am. Exercising and the medication. I've had one medication to start with to control seizures. Then, after 12 months or so, they changed that to my current medication that I've been taking for about eight years now. Participant 29

The final theme in relation to understanding of prognosis was that mitochondrial disease came with a poor prognosis, including reduced life expectance and/or a rapid disease progression. This was noted by six participants (12.00%):

Participant describes poor prognosis - reduced life expectancy and/or rapid progression of disease

When I was diagnosed, NAME said, "It'll probably shorten your life." et cetera. He said we would just go along because as I said earlier he explained that there was no medication he could give me, only painkillers and things like that. He didn't seem to know a real lot about it. When I went to him, he only had one other patient that had been diagnosed with it. He's an MS specialist actually, I think that's what he really is noted for. That's about it. Participant 31

Not really, they just said it's ...they're thinking at the moment which is good because the fast progression means it will be terminal quickly....hoping that it's not going to be that. Participant 35

I've been told there is no cure. It's uncertain, I don't know, because it seems to have progressed more rapidly in the last five years. I don't know whether I'll stay as I am now for another 10 to 15 years, or whether it will continue to deteriorate at that rapid rate, I have no idea. Participant 36

Table 3.15: Understanding of prognosis

Understanding of prognosis	All participants	
	n=50	%
Participant describes prognosis not being discussed	26	52.00
Participant describes poor prognosis - decline in physical function	9	18.00
Participant describes a relatively stable disease/controlled (may have some exacerbations)	7	14.00
Participant describes poor prognosis - reduced life expectancy and/or rapid progression of disease	6	12.00

Section 4 Decision-making

Section 4: Experience of health professional communication

Conversations about treatments

- Participants were asked to describe the conversations they have had about mitochondrial disease treatment options. The most common treatments discussed were Coenzyme Q10 and ATP support (n=19, 38.00%). The next most common theme was that participants were told that there is no treatment for mitochondrial disease (n=16, 32.00%). Other themes included having no or little discussion about treatment options (n=9, 18.00%) and having discussions about lifestyle changes (diet, exercise etc.) (n=9, 18.00%).
- In relation to sub-group variations, participants with a university education (45.83%) reported being told that there were no treatments for mitochondrial disease, more frequently than the general population (32.00%)

Decision-making

- There were 16 participant (32.00%) that noted considering side effects, of which 10 participants noted a combination of both the benefits of the treatment as well as the side effects. The nest most common consideration was cost (n=9, 18.00%) followed by impact on lifestyle, including ability to work (n=6, 12.00%).
- In relation to sub-group variations, there were no participants from low socio-economic areas that reported considering the impact on their lifestyle (n=0, 0.00%) and there were no participants with high physical functioning (n=0, 0.00%) or high social functioning (n=0, 0.00%) that reported considering quality of life when making decisions about treatment.
- In the final question about decision-making, participants were asked whether they felt the way they made decisions had changed over time since they were diagnosed. Overall there were 26 participants (52.00%) that felt as though the way they make decisions has changed over time, while 20 participants (40.00%) felt that it had not changed.
- Where participants did feel as though the way they made decisions had changed, the most common reason for this was that they had become more informed (n=11, 22.00%) and that they consider quality of life more in the process of making treatment decisions (n=7, 14.00%).
- In relation to sub-group variations, participants from rural areas (25.00%) reported considering quality of life more frequently than the general population (14.00%).

Discussions about treatment

Participants were asked to describe the conversations they have had about mitochondrial disease treatment options. The most common treatments discussed were Coenzyme Q10 and ATP support (n=19, 38.00%).

Participant describes being told about supplements, such as Coenzyme Q10 and ATP support

Take CQ10 and, you know, don't do heavy exercise, and that was it. Yeah, so that was all that I ...There was no discussion about, you know, you can do this or have this done, or anything. Participant 15

I asked what could one do? And I was told that there was nothing. It was suggested that I take Magnesium Orotate, particularly the Orotate. Which I did. But at that point, there was nothing else. Subsequently Professor NAME and her team as medical scientists did develop ATP support. Which the recipe they gave to bioceuticals to compound the process. And it's now sold commercially. Participant 24

I remember they suggested when they were little, to go on this Coenzyme Q10, but it was just unaffordable back then. You were looking at $30 a bottle. Participant 44

The next most common theme was that participants were told that there is no treatment for mitochondrial disease (n=16, 32.00%).

Participant describes being told that they is no treatment for mitochondrial disease

He said there is no treatment and that it will probably, his initial words, just the CPEO part of it, he said, "It's probably not going to kill you, but it's going to become an inconvenience. You wouldn't be able to drive. You'll lose your license eventually", things like that. That there was no treatment…."There is no definitive treatment because everybody presents differently". There's no if I take it, this is going to slow it down, or take this, it's going to stop it. It's just basically roll with the punches. Very little for funding, don't expect a miracle cure. That was pretty well it. Participant 2

No treatments were offered to me in the early years, it was merely about the diagnosis. Later came attempts to treat individual symptoms…with varying degrees of success. I am generally the one who researches information and approaches my medical team about trying something. Participant 30

They basically told me that there was no treatment. They told me I could go on something called ATP Support, the bioceuticals. That's the only thing that I was offered. Participant 40

Other themes included having no or little discussion about treatment options (n=9, 18.00%) and having discussions about lifestyle changes (diet, exercise etc.) (n=9, 18.00%).

Participant describes having little to no discussion about treatment options during diagnosis

Okay. I had a bit of a conversation with a geneticist too. They did not really say much. The geneticist only said it was very tentative. This is what some people are doing and sometimes it works for them. But for the most part, I don't think I really had a thorough conversation with any healthcare provider about the treatment. They said to look after yourself when you're there. There wasn't really much out there neither….So they said play it by ear essentially. If so get tests on every year and if something becomes debilitating then we'll treat that. Participant 11

…they gave us a little bit of information and explained that it would be the fatigue again, the headaches. Now, he's just finally….on the migraine headache to try and get rid of pain. There wasn't really much discussion about anything at the start. Participant 19

Nothing about treatment at all, just given painkillers. Participant 31

Participant describes having a discussion about lifestyle changes, e.g. diet and exercise

They totally suggested things like a diet, so metabolic diet. I was on that, which was going to help with your energy and stuff like that. I was on that for a while, but then when I got sick this time, that's when they said no, that diet wasn't actually good for me because the food I got to eat was a big part of that diet. I stopped that, use the same diet. Participant 1

He said, "There is no real treatment, except that it would be useful to get some exercise going, to keep things in a stable condition." That's about all I got. Participant 17

And they did say diet. We always had a dietician with us. They changed his diet up a little bit, and said, "He can't fast, he has to have regular food intake. Try and get this type of food into him, and also exercise." They also really put exercise at the forefront, and they also put early intervention. So we actually got a pretty good ... given that there's no treatment as such that they know is effective, we actually got a pretty….They did, they really did, and I think that's part of the reason he's done so well. Not ... I mean, for some reason he has done well, but I think that's also been a major factor. Participant 46

In relation to sub-group variations, participants with a university education (45.83%) reported being told that there were no treatments for mitochondrial disease, more frequently than the general population (32.00%)

Table 4.1: Conversations about treatment

Conversations about treatment	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant describes being told about supplements, such as Coenzyme Q10 and ATP support	19	38.00	11	36.67	8	40.00	9	33.33	10	43.48
Participant describes being told that there is no treatment for mitochondrial disease	16	32.00	10	33.33	6	30.00	9	33.33	7	30.43
Participant describes having little to no discussion about treatment options during diagnosis	9	18.00	5	16.67	4	20.00	5	18.52	4	17.39
Participant describes having a discussion about lifestyle changes, e.g. diet and exercise	9	18.00	6	20.00	3	15.00	5	18.52	4	17.39

Conversations about treatment	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant describes being told about supplements, such as Coenzyme Q10 and ATP support	19	38.00	11	42.31	8	33.33	9	37.50	15	44.12
Participant describes being told that there is no treatment for mitochondrial disease	16	32.00	5	19.23	11	45.83	10	41.67	10	29.41
Participant describes having little to no discussion about treatment options during diagnosis	9	18.00	5	19.23	4	16.67	3	12.50	5	14.71
Participant describes having a discussion about lifestyle changes, e.g. diet and exercise	9	18.00	4	15.38	5	20.83	3	12.50	7	20.59

Conversations about treatment	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant describes being told about supplements, such as Coenzyme Q10 and ATP support	19	38.00	9	40.91	10	35.71	10	38.46	9	37.50
Participant describes being told that there is no treatment for mitochondrial disease	16	32.00	7	31.82	9	32.14	8	30.77	8	33.33
Participant describes having little to no discussion about treatment options during diagnosis	9	18.00	4	18.18	5	17.86	5	19.23	4	16.67
Participant describes having a discussion about lifestyle changes, e.g. diet and exercise	9	18.00	3	13.64	6	21.43	6	23.08	3	12.50

Conversations about treatment	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant describes being told about supplements, such as Coenzyme Q10 and ATP support	19	38.00	9	45.00	10	33.33	9	40.91	10	35.71
Participant describes being told that there is no treatment for mitochondrial disease	16	32.00	6	30.00	10	33.33	8	36.36	8	28.57
Participant describes having little to no discussion about treatment options during diagnosis	9	18.00	5	25.00	4	13.33	4	18.18	5	17.86
Participant describes having a discussion about lifestyle changes, e.g. diet and exercise	9	18.00	4	20.00	5	16.67	4	18.18	5	17.86

Conversations about treatment	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant describes being told about supplements, such as Coenzyme Q10 and ATP support	**19**	38.00	5	83.33	4	28.57	2	22.22	5	45.45	3	30.00
Participant describes being told that there is no treatment for mitochondrial disease	**16**	32.00	0	0.00	2	14.29	5	55.56	6	54.55	3	30.00
Participant describes having little to no discussion about treatment options during diagnosis	**9**	18.00	0	0.00	5	35.71	1	11.11	1	9.09	2	20.00
Participant describes having a discussion about lifestyle changes, e.g. diet and exercise	**9**	18.00	2	33.33	2	14.29	1	11.11	2	18.18	2	20.00

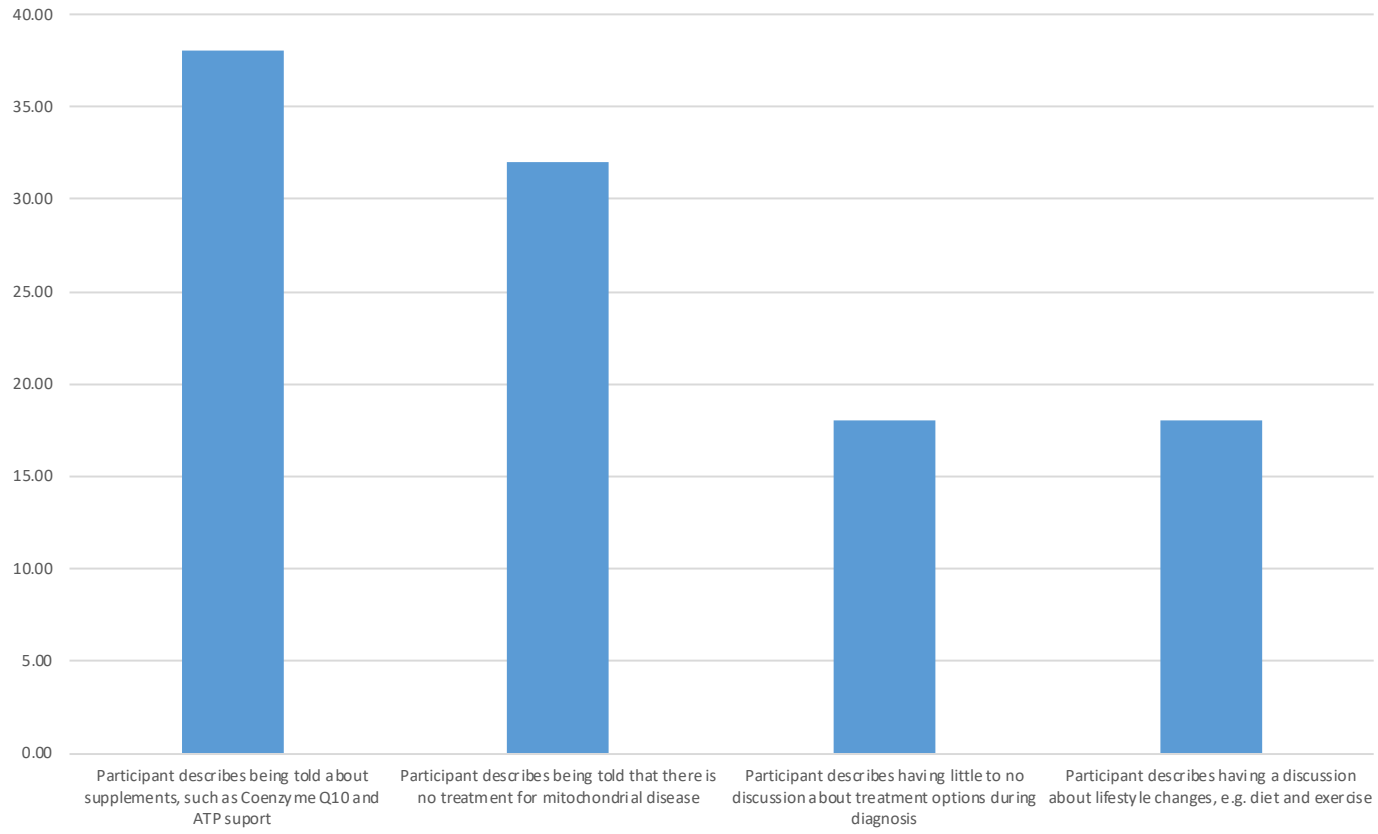


Figure 4.1: Conversations about treatment

Decision-making

What is considered when making decisions

Participants were asked about the things that they take into consideration when making decisions about treatment. There were 16 participant (32.00%) that noted considering side effects, of which 10 participants noted a combination of both the benefits of the treatment as well as the side effects.

Participant describes their main consideration as the side effects

Toxic. Like if it's adverse side effects. I do tend to ask the support groups, just joined a mitochondrial group on Facebook. And also, the United one, the one that's overseas, with the world group. Participant 18

More like how it will affect me long term and that's pretty much it really. How it will affect me personally like my health that kind of stuff really. Participant 38

Being aware of the medical aspects and medication and the contraindication with the condition. That's really important that doctors monitor that. I'm unaware of what could actually make this condition worse. Participant 42

Participant describes considering a combination of the benefits and side effects

The benefits, side effects. That'd be the two big things. What are the benefits going to be? Participant 2

What are the benefits? What are the negatives? If it's going to be…The benefit have to weigh out the negatives. Participant 8

Side effects mainly. Always check them out to see whether it's worth going through, that's all, nothing else. If they suggested something, you try to see if it helped. I always took a course of it or maybe two courses, that was it then if it wasn't doing any good, I wouldn't take it anymore. Participant 10

I would definitely be looking at how risky is something and whether it's worth the risk or not. Well, I'm not sure what the benefits would be if it helped. The risk, benefits, that's really. Participant 25

The nest most common consideration was cost (n=9, 18.00%) followed by impact on lifestyle, including ability to work (n=6, 12.00%).

Participant describes considering the impact on their lifestyle, including work

Obviously there's ramifications of it in terms of my lifestyle, because I live alone. That's the biggest one. Otherwise, I'm prepared to try anything. Participant 19

A second part will be, you know, I'm working full-time, so what effect that will have on me in my work life. They're the probably two considerations that I would have. Participant 21

Oh yes. Also like how it impacts my daily routine, like how long it takes. That's a lot to take into consideration. Participant 45

In relation to sub-group variations, there were no participants from low socio-economic areas that reported considering the impact on their lifestyle (n=0, 0.00%) and there were no participants with high physical functioning (n=0, 0.00%) or high social functioning (n=0, 0.00%) that reported considering quality of life when making decisions about treatment.

Table 4.2: Considerations when making decisions

Considerations when making decisions about treatment	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant describes considering a combination of the benefits and side effects	10	20.00	5	16.67	5	25.00	4	14.81	6	26.09
Participant describes their main consideration as the side effects	6	12.00	5	16.67	1	5.00	4	14.81	2	8.70
Participant describes their main consideration as the evidence to support the use of the treatment (Efficacy)	10	20.00	5	16.67	5	25.00	5	18.52	5	21.74
Participant describes their main consideration as the cost of the treatment	9	18.00	7	23.33	2	10.00	5	18.52	4	17.39
Participant describes considering the impact on their lifestyle, including work	6	12.00	5	16.67	1	5.00	6	22.22	0	0.00
Participant describes taking the advice from the specialist with no specific considerations	5	10.00	4	13.33	1	5.00	3	11.11	2	8.70
Participant describes considering their quality of life when deciding on a treatment	5	10.00	2	6.67	3	15.00	2	7.41	3	13.04

Considerations when making decisions about treatment	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant describes considering a combination of the benefits and side effects	10	20.00	7	26.92	3	12.50	5	20.83	7	20.59
Participant describes their main consideration as the side effects	6	12.00	2	7.69	4	16.67	4	16.67	3	8.82
Participant describes their main consideration as the evidence to support the use of the treatment (Efficacy)	10	20.00	3	11.54	7	29.17	6	25.00	6	17.65
Participant describes their main consideration as the cost of the treatment	9	18.00	5	19.23	4	16.67	4	16.67	7	20.59
Participant describes considering the impact on their lifestyle, including work	6	12.00	4	15.38	2	8.33	3	12.50	4	11.76
Participant describes taking the advice from the specialist with no specific considerations	5	10.00	3	11.54	2	8.33	2	8.33	4	11.76
Participant describes considering their quality of life when deciding on a treatment	5	10.00	2	7.69	3	12.50	3	12.50	2	5.88

Considerations when making decisions about treatment	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant describes considering a combination of the benefits and side effects	10	20.00	4	18.18	6	21.43	5	19.23	5	20.83
Participant describes their main consideration as the side effects	6	12.00	2	9.09	4	14.29	5	19.23	1	4.17
Participant describes their main consideration as the evidence to support the use of the treatment (Efficacy)	10	20.00	5	22.73	5	17.86	6	23.08	4	16.67
Participant describes their main consideration as the cost of the treatment	9	18.00	2	9.09	7	25.00	6	23.08	3	12.50
Participant describes considering the impact on their lifestyle, including work	6	12.00	3	13.64	3	10.71	4	15.38	2	8.33
Participant describes taking the advice from the specialist with no specific considerations	5	10.00	3	13.64	2	7.14	1	3.85	4	16.67
Participant describes considering their quality of life when deciding on a treatment	5	10.00	0	0.00	5	17.86	2	7.69	3	12.50

Considerations when making decisions about treatment	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant describes considering a combination of the benefits and side effects	10	20.00	4	20.00	6	20.00	5	22.73	5	17.86
Participant describes their main consideration as the side effects	6	12.00	3	15.00	3	10.00	1	4.55	5	17.86
Participant describes their main consideration as the evidence to support the use of the treatment (Efficacy)	10	20.00	4	20.00	6	20.00	3	13.64	7	25.00
Participant describes their main consideration as the cost of the treatment	9	18.00	4	20.00	5	16.67	2	9.09	7	25.00
Participant describes considering the impact on their lifestyle, including work	6	12.00	3	15.00	3	10.00	2	9.09	4	14.29
Participant describes taking the advice from the specialist with no specific considerations	5	10.00	2	10.00	3	10.00	2	9.09	3	10.71
Participant describes considering their quality of life when deciding on a treatment	5	10.00	0	0.00	5	16.67	1	4.55	4	14.29

Considerations when making decisions about treatment	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant describes considering a combination of the benefits and side effects	10	20.00	3	50.00	4	28.57	1	11.11	1	9.09	1	10.00
Participant describes their main consideration as the side effects	6	12.00	1	16.67	4	28.57	1	11.11	0	0.00	0	0.00
Participant describes their main consideration as the evidence to support the use of the treatment (Efficacy)	10	20.00	1	16.67	4	28.57	1	11.11	3	27.27	1	10.00
Participant describes their main consideration as the cost of the treatment	9	18.00	2	33.33	4	28.57	1	11.11	1	9.09	1	10.00
Participant describes considering the impact on their lifestyle, including work	6	12.00	1	16.67	0	0.00	1	11.11	2	18.18	2	20.00
Participant describes taking the advice from the specialist with no specific considerations	5	10.00	0	0.00	1	7.14	1	11.11	2	18.18	1	10.00
Participant describes considering their quality of life when deciding on a treatment	5	10.00	1	16.67	2	14.29	0	0.00	1	9.09	1	10.00

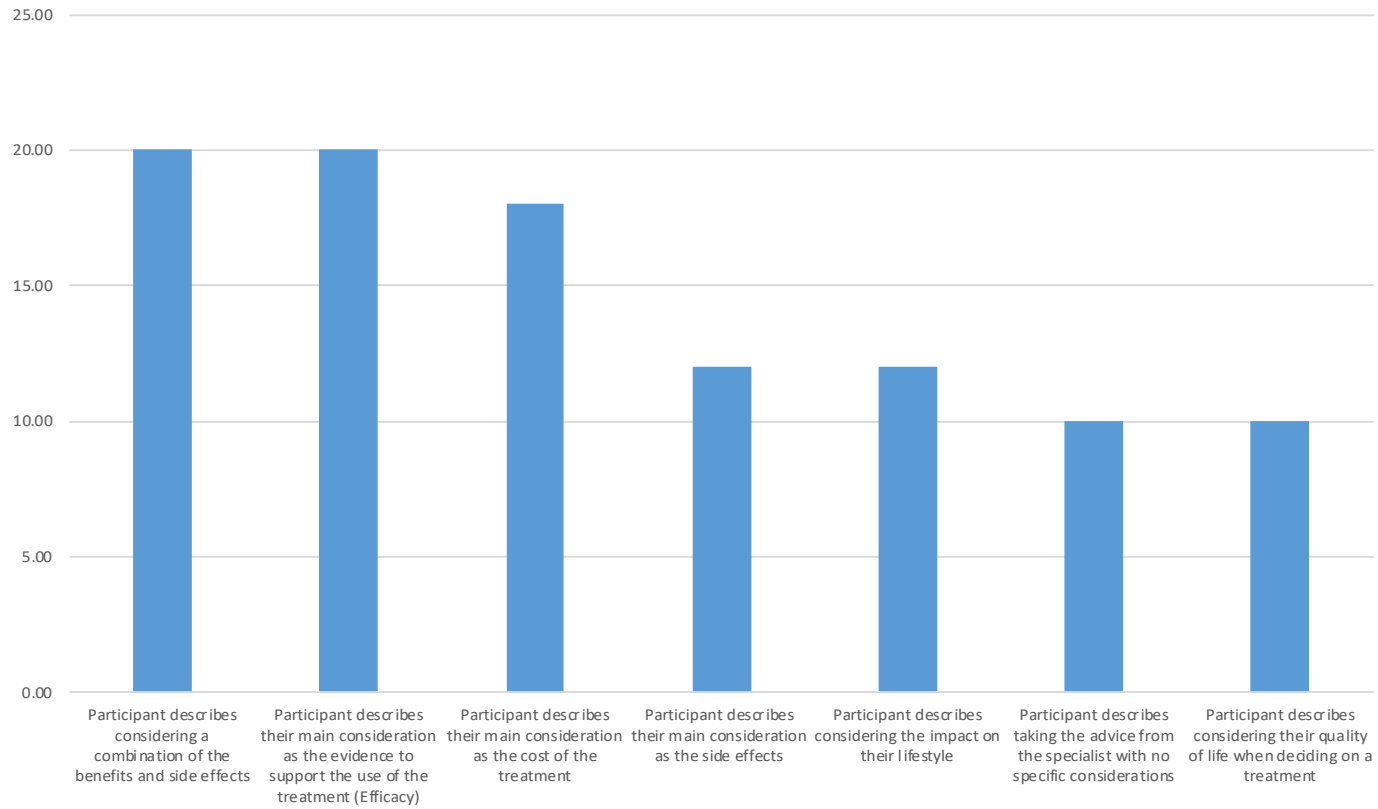


Figure 4.2: Considerations when making decisions

Changes in decision-making

In the final question about decision-making, participants were asked whether they felt the way they made decisions had changed over time since they were diagnosed. Overall there were 26 participants (52.00%) that felt as though the way they make decisions has changed over time, while 20 participants (40.00%) felt that it had not changed.

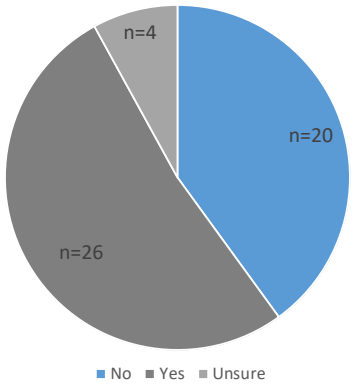


Figure 4.3: Changes in decision-making

Where participants did feel as though the way they made decisions had changed, the most common reason for this was that they had become more informed (n=11, 22.00%) and that they consider quality of life more in the process of making treatment decisions (n=7, 14.00%):

Participant describes decision-making changing as they have become more informed

It probably has because I'm a bit more informed now. When they talk about having the bone marrow transfusion and they talk about that sort of thing I understand. Whereas at first I probably wouldn't have gotten what they were really on about. Participant 6

No, it probably has changed. I'd say it probably has changed because I've got a little bit more knowledge but trying to find a local doctor really is very hard. Participant 12

Most changed over time. Initially, I probably looked at anything, grabbed onto anything that might be a treatment and might have possible benefits. Over time, I've become someone who definitely looks at the science base to it and the evidence base to it. Participant 34

Participant describes decision-making changing as they consider quality of life more

No. Yeah, I can see that if it dragged on for 10 years that I'd get to the point where I'd say, "Look I just can't go backwards and forwards to see them all the time. I'm not going to do it anymore." Participant 7

No, it's definitely changed. It changes all the time anyway through your life. I think your priorities change quite a lot in life. When you have a diagnosis like this where you're unsure of if you can get any assistance or if anything is ever going to get better or change. It's just prioritizing, what's important, what makes you happy, what keeps the family running. Participant 28

My decisions now are much more informed than when I was first diagnosed – then we were looking for the "magic bullet". Now my choices are based around quality of life treatments. Participant 32

In relation to sub-group variations, participants from rural areas (25.00%) reported considering quality of life more frequently than the general population (14.00%).

Table 4.3: Decision-making over time

Does decision-making change over time?	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant describes decision-making changing as they have become more informed	11	22.00	5	16.67	6	30.00	5	18.52	6	26.09
Participant describes decision-making changing as they consider quality of life more	7	14.00	2	6.67	5	25.00	2	7.41	5	21.74
Participant describes decision-making not changing as they have not been given any treatment options	5	10.00	3	10.00	2	10.00	2	7.41	3	13.04

Does decision-making change over time?	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant describes decision-making changing as they have become more informed	11	22.00	5	19.23	6	25.00	5	20.83	8	23.53
Participant describes decision-making changing as they consider quality of life more	7	14.00	3	11.54	4	16.67	5	20.83	5	14.71
Participant describes decision-making not changing as they have not been given any treatment options	5	10.00	3	11.54	2	8.33	3	12.50	5	14.71

Does decision-making change over time?	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant describes decision-making changing as they have become more informed	11	22.00	7	31.82	4	14.29	4	15.38	7	29.17
Participant describes decision-making changing as they consider quality of life more	7	14.00	1	4.55	6	21.43	3	11.54	4	16.67
Participant describes decision-making not changing as they have not been given any treatment options	5	10.00	3	13.64	2	7.14	2	7.69	3	12.50

Does decision-making change over time?	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant describes decision-making changing as they have become more informed	11	22.00	6	30.00	5	16.67	3	13.64	8	28.57
Participant describes decision-making changing as they consider quality of life more	7	14.00	1	5.00	6	20.00	3	13.64	4	14.29
Participant describes decision-making not changing as they have not been given any treatment options	5	10.00	2	10.00	3	10.00	3	13.64	2	7.14

Does decision-making change over time?	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant describes decision-making changing as they have become more informed	11	22.00	1	16.67	2	14.29	5	55.56	3	27.27	0	0.00
Participant describes decision-making changing as they consider quality of life more	7	14.00	1	16.67	2	14.29	0	0.00	3	27.27	1	10.00
Participant describes decision-making not changing as they have not been given any treatment options	5	10.00	0	0.00	3	21.43	1	11.11	1	9.09	0	0.00

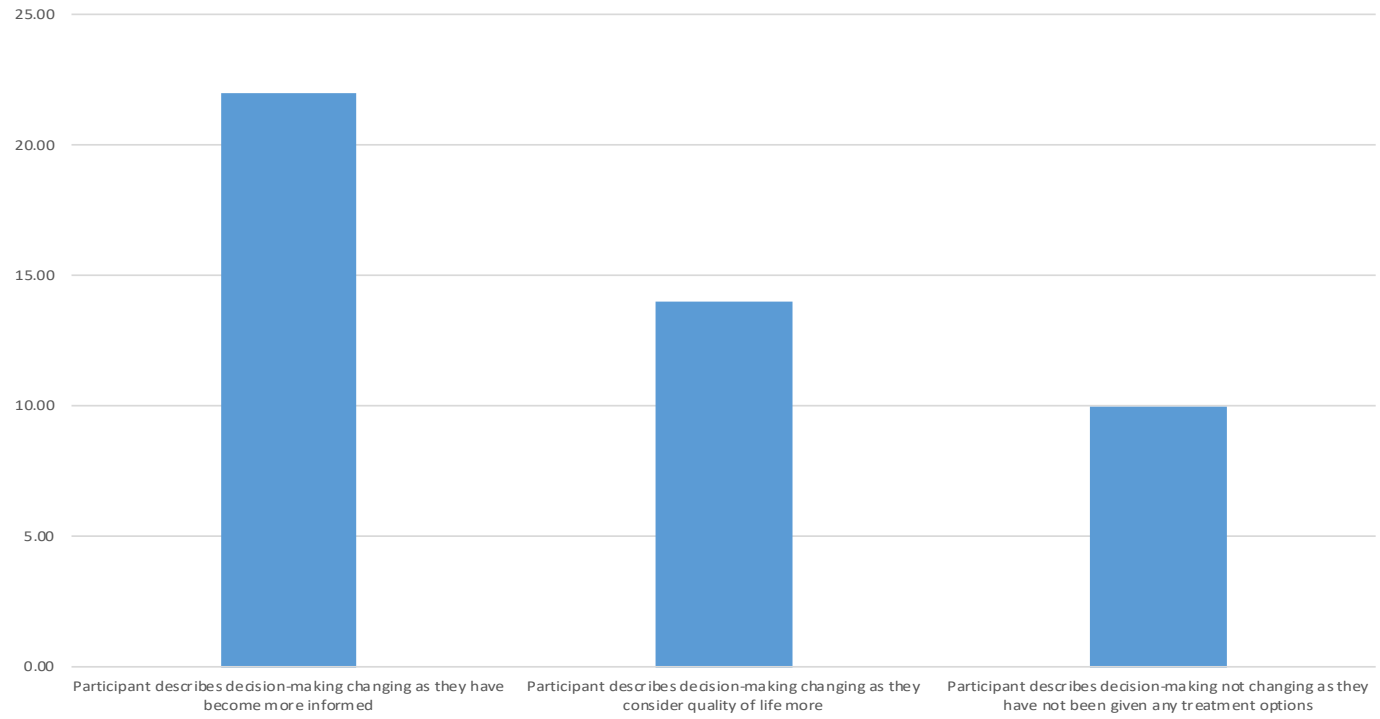


Figure 4.4: Decision-making over time

Section 5 Treatment and health service provision

Section 5: Experience of treatment

Discussions about Clinical Trials

- In this PEEK study, 64% of all participants (n=32) describe not being spoken to about clinical trials, seven participants brought up the topic with their doctor (14.00%) and the doctors of 11 participants brought up the topic (22.00%).

Participation in Clinical Trials

- Seven participants have taken part in a clinical trial (14.00%), and 33 participants have not taken part in a clinical trial would like if one was suitable for them (66.00%). Ten participants have not taken part and do not want to (20.00%)

Treatments experienced

- Participants were asked in the questionnaire to identify the treatments that they had experienced. most common treatments were Coenzyme Q10 (n=36, 72.00%), vitamins and supplements (n=32, 64.00%), followed by physical therapy (n=15, 30.00%), and diet (n=11, 22.00).
- Participants were asked to rate their quality of life on a scale of 1 to 7, while using each specific treatment (with 1 being 'Life was very distressing and 7 being 'Life was great'). Mean quality of life scores ranges from 3.34 to 4.33, that is, all quality of life scores were within the 'life was a little distressing' to 'Life was average' range. The treatment that scored the least impact on quality of life was speech therapy (mean score 4.33). All other treatments were in the 'Life was a little distressing' range (mean scores range 3.34 to 3.86).
- The treatments that had a mean effectiveness score of at least 3 (moderately effective) were respiratory therapy (average score 3.50), speech therapy (average score 3.33), and diet (average score 3.09). The remaining treatments scored had a mean effectiveness score of at least 2, that is in the somewhat effective range.
- Participants were asked in the structure interview to provide a description of mild side effects. The most common description of mild side effects were those that do not greatly impact activities of daily living (n=11, 22.00%). In relation to specific side effects that were considered to be mild, there were seven participants (14.00%) that described headaches, six participants (12.00%) that described gastrointestinal problems (diarrhoea and cramping) and five participants (10.00%) that described increased fatigue (and related irritability) as a mild side effect. There were also six participants that did not describe a mild side effect but talks about mitochondrial disease being part of everyday life (Particularly pain).
- In relation to sub-group variations, participants with high social functioning (40.00%) described mild side effects as those that do not greatly impact activities of daily living more frequently than the general population (22.00%).
- Participants were asked in the structure interview to provide a description of severe side effects. The most common description of severe side effects were those that limit daily activities for an extended period of time (n=19, 38.00%), seven participants (14.00%) described sever side effects as an effect requiring hospitalisation or medical attention/permanent damage, or a life threatening effect or inability to function. In relation to specific side effects that were considered severe, nine participants (18.00%) described severe fatigue, four participants (8.00%) described chronic headaches and four participants (8.00%) described loss of mobility or independence.
- In relation to sub-group variations, participants with a high school or trade education (15.38%), low physical function(25.00%) and low social functioning (26.67%) described severe side effects as effects limiting their daily activities for an extended period of time, less frequently than the general population (38.00%), while those with a university education (62.50%), high physical function (54.44%), high social functioning (55.00%), high general health (50.00%) and hearing impairment (50.00%) described this more frequently.

Adherence to medication

- Participants were asked in the online questionnaire if, in general, if they were good at taking medicine and sticking to it. The majority of participants were good at sticking to treatments all of the time (n=30, 60.00%) and the remaining were good at sticking to treatments most of the time (n=20, 40.00%). No participants felt they were never, rarely or sometimes good at sticking to treatments.
- Participants were also asked in the structured interview how long they stick with a therapy before they think it might not be working or give up on it. Close to half of all participants (n=24, 48.00%) describes using treatment for a period of one to three months before deciding if its working. The next most common theme was continuing a treatment indefinitely or as recommended by clinician/specialist (n=9, 18.00%) and there were six participants (12.00%) that described not trying new medications for mitochondrial disease and such not knowing how long they would continue a treatment.
- In relation to sub-group variations, participants from rural areas (30.00%) and participants with a hearing impairment (29.17%) reported continuing a treatment indefinitely or as recommended by clinician/specialist more frequently than the general population (18.00%). Participants with a hearing impairment (29.17%) reported using treatment for a period of one to three months before deciding if its working less frequently than the general population (48.00%), while those with high social functioning (60.00%) and high general health (59.09%) reported this more frequently.
- Participants were asked what needed to change for them to feel as though a treatment was working. The most common description was needing to feel more energetic, and increase in physical ability, to know a treatment is working (n=20, 40.00%). This was followed by needing to see improved symptoms by clinical measurement (test result) (n=13, 26.00%) and needing to generally feel better to know that a treatment is working (n=9, 13.00%). There were five participants (10.00%) that described needing to reduce pain to know a treatment is working and five participants (10.00%) needing to improve their quality of life to know a treatment is working.
- In relation to sub-group variations, participants from metropolitan areas (26.67%) and participants from high socio-economic areas (25.93%) reported needing to feel more energetic, and increase in physical ability, to know a treatment is working, less frequently than the general population (40.00%) while participants from rural areas (60.00%), participants from low socio-economic areas (56.52%). Participants with a hearing impairment (41.67%) reported improved symptoms by clinical measurement (test result) more frequently than the general population (26.00%). Participants with high physical functioning (31.82%) described needing to generally feel better to know that a treatment is working, more frequently than the general population (18.00%).

Complementary therapies

- Participants were asked whether they had used any complementary therapies. The most common therapies that were considered complementary and described by participants were vitamins, minerals and supplements (n=14, 28.00%) and allied health e.g. physiotherapy (including massage and hydrotherapy), speech therapy, occupational therapy (n=14, 14.00%). The next most frequent complementary therapies described were alternative medicine, e.g. osteopathy, acupuncture, chiropractor, Bowen therapy (n=12, 24.00%). There were also 11 participants (22.00%) that noted that they did not use any complementary therapies.

Service provision and affordability

- The main physician treating participants for mitochondrial disease were general practitioners (N=19, 38.00%), followed by neurologists (N=12, 24.00%) and mitochondrial specialists (N=11, 22.00%).
- Participants had access to a general practitioner (n=48, 96.00%), neurologist (n=43, 86.00%), mitochondrial specialist (n=29, 58.00%) and cardiologist (n=28, 56.00%) for the treatment of their mitochondrial disease.
- The majority of patients had private healthcare insurance (n=37, 74.00%), 29 (58.00%) participants were treated as public patients, 12 (24.00%) as private patients and 9 (18.00%) as equally public and private patients. The majority of participants were treated in the public hospital system (n=32, 64.00%).
- Almost half of participants have never missed medical appointments due to cost (n=24, 48.00%), and most have never been unable to afford prescription medications (n=34, 64.00%). Almost half of participants have found it somewhat to extremely difficult paying for basic needs due to their diagnosis with mitochondrial disease (n=24, 48.00%).

Changes to work status

- The work status for a number of participants changed due to their diagnosis with mitochondrial disease with about a quarter of participants reducing the number of hours worked (n=13, 26,00%), and 19 (38.00%) quitting their jobs.
- Of those that had a partner or carer, four carers/partners had to quit their job (23.53%), seven had to reduce the number of hours worked (41.18%), carers have had to take leave either with pay (n=2, 11.76%), or without pay (n=5, 29.41%).

Experience of respect during treatment

- Participants were asked if they felt they had been treated with respectfully throughout their treatment. Half of the participants felt that they had been treated respectfully with the exception of one or two occasions (n=25, 50.00%), 18 felt that they had been treated respectfully (36.00%) and seven felt they had not been treated respectfully (14.00%).

Clinical Trials

Discussions about Clinical Trials

In this PEEK study, 64% of all participants (n=32) describe not being spoken to about clinical trials, seven participants brought up the topic with their doctor (14.00%) and the doctors of 11 participants brought up the topic (22.00%).

Table 5.1: Discussions about clinical trials

Have you discussed clinical trials with your doctor?	N=50	Percentage of participants
I brought up the topic of clinical trials with my doctor for discussion	7	14.00
My doctor brought up the topic of clinical trials for discussion	11	22.00
No one has ever spoken to me about clinical trials	32	64.00

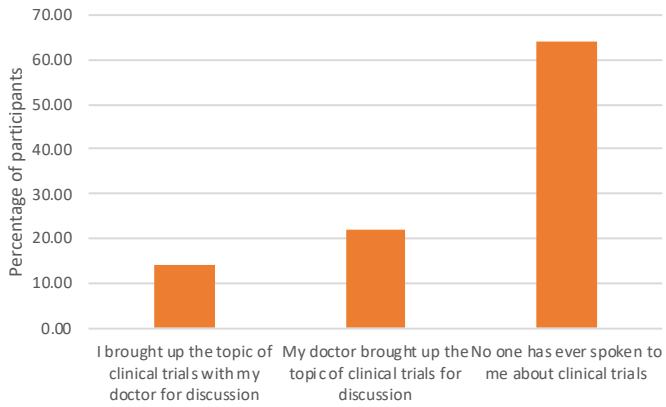


Figure 5.1: Discussions about clinical trials

Seven participants have taken part in a clinical trial (14.00%), and 33 participants have not taken part in a clinical trial would like if one was suitable for them (66.00%). Ten participants have not taken part and do not want to (20.00%).

Table 5.2: Participation in clinical trials

Participation in clinical trials	N=50	Percentage of participants
Have not participated in a clinical trial and do not want to	10	20.00
Have not participated in a clinical trial but would like to if there is one for me	33	66.00
Have participated in a clinical trial	7	14.00

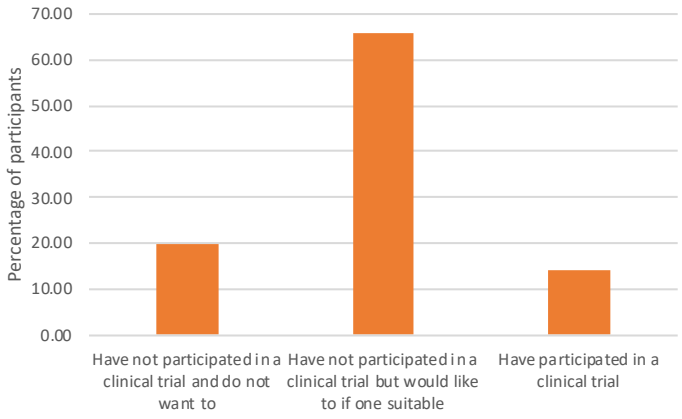


Figure 5.2: Participation in clinical trials

Treatments experienced

Participants were asked in the questionnaire to identify the treatments that they had experienced. most common treatments were Coenzyme Q10 (n=36, 72.00%) and Vitamins and supplements (n=32, 64.00%). This was followed by physical therapy (n=15, 30.00%), diet (n=11, 22.00%), speech therapy (n=9, 18.00%), and respiratory therapy (n=4, 8.00%).

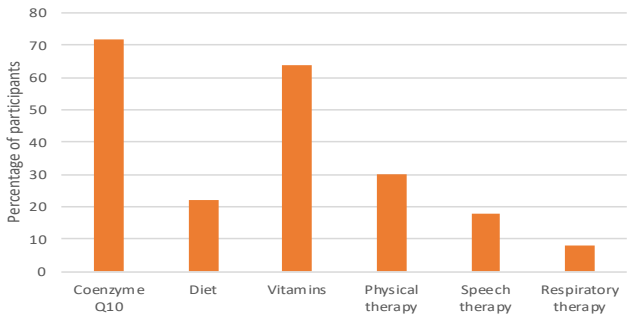


Figure 5.3: Treatments experienced (% of all participants)

Table 5.3: Treatments experienced

Treatment	N=50	Percent	Mean QOL score	Range (Worst QOL = 1, Best QOL = 7)	Mean effectiveness score	Range (Ineffective = 1, Very Effective = 5)
Coenzyme Q10	36	72.00	3.86	1-7	2.56	1-5
Diet	11	22.00	3.82	1-6	3.09	1-5
Vitamins	32	64.00	3.34	1-6	2.44	1-5
Physical therapy	15	30.00	3.60	1-6	2.80	1-5
Speech therapy	9	18.00	4.33	1-7	3.33	1-4
Respiratory therapy	4	8.00	3.75	1-5	3.50	1-4

As a follow-up question (within the questionnaire), participants were asked to rate their quality of life on a scale of 1 to 7, while using each specific treatment (with 1 being 'Life was very distressing and 7 being 'Life was great'). Mean quality of life scores ranges from 3.34 to 4.33, that is, all quality of life scores were with the 'life was a little distressing' to 'Life was average' range. The treatment that scored the least impact on quality of life was speech therapy (mean score 4.33). All other treatments were in the 'Life was a little distressing' range (mean scores range 3.34 to 3.86).

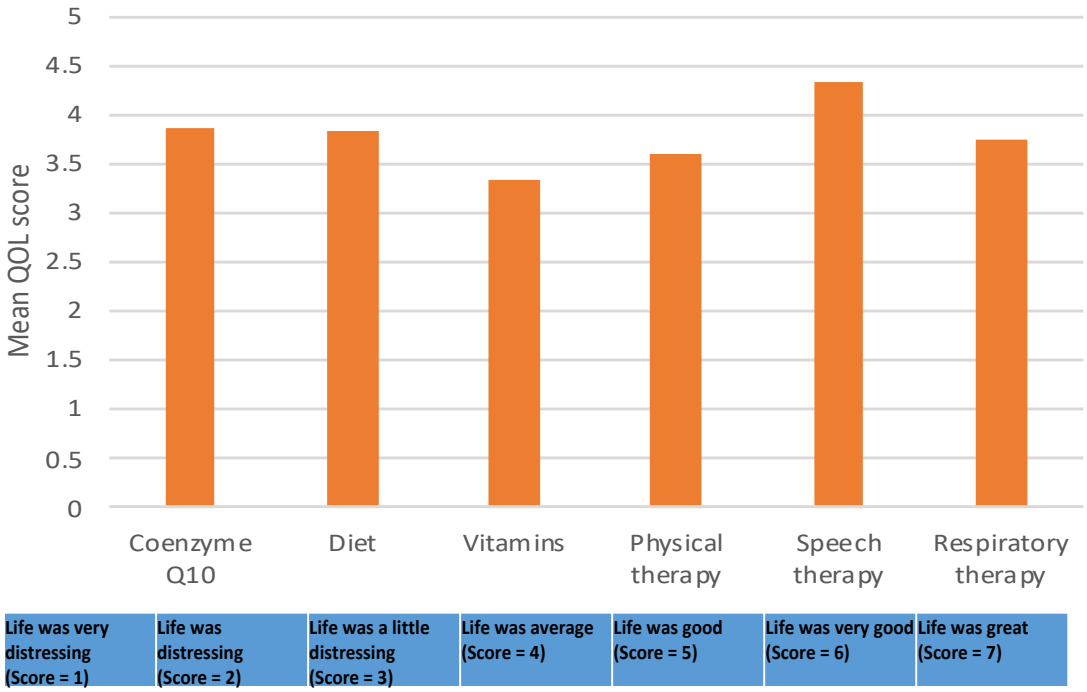


Figure 5.4: Treatments experienced and mean quality of life

A second follow-up question was asked in relation to how effective the participant felt the treatment was on a scale of 1 to 5 (with 1 being ineffective and 5 being very effective). The treatments that had a mean effectiveness score of at least 3 (moderately effective) were respiratory therapy (average score 3.50), speech therapy (average score 3.33), and diet (average score 3.09). The remaining treatments scored had a mean effectiveness score of at least 2, that is in the somewhat effective range.

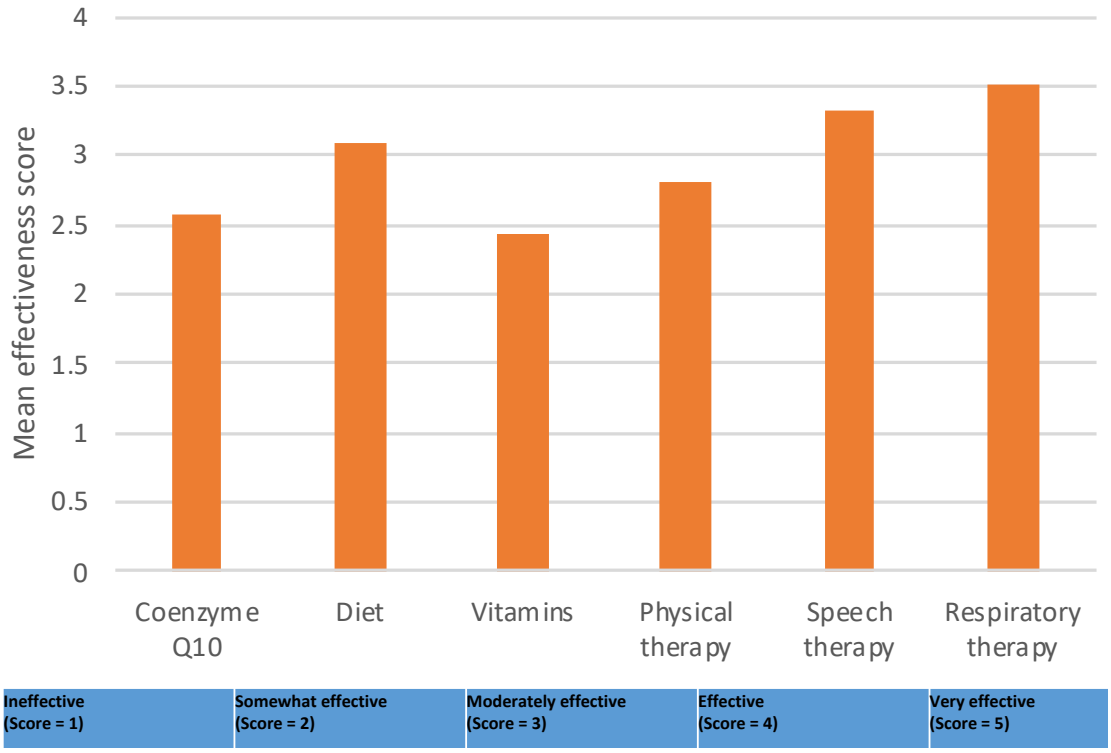


Figure 5.5: Treatments experienced and effectiveness

Side effects of treatment

Mild side effects

Participants were asked in the structure interview to provide a description of mild side effects. The most common description of mild side effects were those that do not greatly impact activities of daily living (n=11, 22.00%).

Participant describes mild side effects as those that do not greatly impact activities of daily living

Stuff that didn't impact on my day to day activities. Things that were potentially intermittent come and go. Something that you had to go through for a period of time but the benefit was that it did not stopping is that there'd be a benefit having gone through it I'd say. That's all mild to me. Participant 2

To me, probably mild side effects are ones that don't have any severe impact on your life and your lifestyle. It's like you can cope with it without making big alterations to what you can do and what you can't do, and probably live, what I call a normal life. That's what I call mild side effect. Participant 15

Mild side effects I suppose would be a little bit annoying, but don't affect what you do in the day. Mild side effects, I would say would be a bit annoying, don't have an effect on your day to day activities. Participant 43

In relation to specific side effects that were considered to be mild, there were seven participants (14.00%) that described headaches, six participants (12.00%) that described gastrointestinal problems (diarrhoea and cramping) and five participants (10.00%) that described increased fatigue (and related irritability) as a mild side effect. There were also six participants that did not describe a mild side effect specifically but spoke about mitochondrial disease being part of everyday life (Particularly pain).

Participant does not describe a mild side effect but talks about mitochondrial disease being part of everyday life (Particularly pain)

I do sort of but I cope with it all. Yes, I just got to. I've got no choice. I get out of bed and I'm in agony every morning. I wait for the painkillers to kick in. Participant 18

In a scientific sense it's very difficult. I don't know how you would describe it. I don't have any factual measurement as such. I've always taken the attitude as nothing can be done about it. I've just got to press on and keep doing things. If you've got a cut finger, you put a band aid on it and get on with it. Participant 27

Mild side effects mean that I first deserve to get up in the day and just to do daily chores that I can handle, and that would be mild side effects. I'll have a rest in the afternoon, that's around half an hour nap. That gets me up and going for dinner time or watching television which I write that past that would be the mild side effects. Mild side effect is maybe just to say almost 100% pain relieving, it's if I would get pain in some of the muscle groups and not the others, and that gives me a warning then to be careful. The main problem I will is I could be sitting where I'm sitting now and I could go just like you message if you would like to be entertained, and then sometimes I'm sitting down that then develops into a pain and a headache. If I'm walking around, of course, I could result in a fall, which I has happened quite a few. Then I use the pain just to give me balance into walking. That would be a mild side effect. Participant 23

In relation to sub-group variations, participants with high social functioning (40.00%) described mild side effects as those that do not greatly impact activities of daily living more frequently than the general population (22.00%).

Table 5.4: Description of mild side effects

Description of mild side effects	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant describes mild side effects as those that do not greatly impact activities of daily living	**11**	22.00	8	26.67	3	15.00	7	25.93	4	17.39
Participant describes mild side effects as headaches	**7**	14.00	4	13.33	3	15.00	3	11.11	4	17.39
Participant describes gastrointestinal problems (diarrhoea and cramping) as mild side effects	**6**	12.00	4	13.33	2	10.00	4	14.81	2	8.70
Participant does not describe a mild side effect but talks about mitochondrial disease being part of everyday life (Particularly pain)	**6**	12.00	4	13.33	2	10.00	4	14.81	2	8.70
Participant had not had any mild side effects and could not answer (N/A)	**6**	12.00	3	10.00	3	15.00	2	7.41	4	17.39
Participant describes mild side effects as something temporary, you can overcome in a short time period	**5**	10.00	4	13.33	1	5.00	3	11.11	2	8.70
Participant describes increased fatigue (and related irritability) as a mild side effect	**5**	10.00	4	13.33	1	5.00	4	14.81	1	4.35

Description of mild side effects	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant describes mild side effects as those that do not greatly impact activities of daily living	11	22.00	5	19.23	6	25.00	3	12.50	8	23.53
Participant describes mild side effects as headaches	7	14.00	4	15.38	3	12.50	5	20.83	4	11.76
Participant describes gastrointestinal problems (diarrhoea and cramping) as mild side effects	6	12.00	4	15.38	2	8.33	3	12.50	4	11.76
Participant does not describe a mild side effect but talks about mitochondrial disease being part of everyday life (Particularly pain)	6	12.00	4	15.38	2	8.33	1	4.17	5	14.71
Participant had not had any mild side effects and could not answer (N/A)	6	12.00	4	15.38	2	8.33	3	12.50	5	14.71
Participant describes mild side effects as something temporary, you can overcome in a short time period	5	10.00	2	7.69	3	12.50	3	12.50	2	5.88
Participant describes increased fatigue (and related irritability) as a mild side effect	5	10.00	4	15.38	1	4.17	2	8.33	3	8.82

Description of mild side effects	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant describes mild side effects as those that do not greatly impact activities of daily living	11	22.00	6	27.27	5	17.86	8	30.77	3	12.50
Participant describes mild side effects as headaches	7	14.00	3	13.64	4	14.29	4	15.38	3	12.50
Participant describes gastrointestinal problems (diarrhoea and cramping) as mild side effects	6	12.00	1	4.55	5	17.86	4	15.38	2	8.33
Participant does not describe a mild side effect but talks about mitochondrial disease being part of everyday life (Particularly pain)	6	12.00	2	9.09	4	14.29	2	7.69	4	16.67
Participant had not had any mild side effects and could not answer (N/A)	6	12.00	2	9.09	4	14.29	3	11.54	3	12.50
Participant describes mild side effects as something temporary, you can overcome in a short time period	5	10.00	1	4.55	4	14.29	3	11.54	2	8.33
Participant describes increased fatigue (and related irritability) as a mild side effect	5	10.00	3	13.64	2	7.14	3	11.54	2	8.33

Description of mild side effects	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant describes mild side effects as those that do not greatly impact activities of daily living	11	22.00	8	40.00	3	10.00	7	31.82	4	14.29
Participant describes mild side effects as headaches	7	14.00	2	10.00	5	16.67	2	9.09	5	17.86
Participant describes gastrointestinal problems (diarrhoea and cramping) as mild side effects	6	12.00	1	5.00	5	16.67	2	9.09	4	14.29
Participant does not describe a mild side effect but talks about mitochondrial disease being part of everyday life (Particularly pain)	6	12.00	0	0.00	6	20.00	2	9.09	4	14.29
Participant had not had any mild side effects and could not answer (N/A)	6	12.00	2	10.00	4	13.33	3	13.64	3	10.71
Participant describes mild side effects as something temporary, you can overcome in a short time period	5	10.00	2	10.00	3	10.00	3	13.64	2	7.14
Participant describes increased fatigue (and related irritability) as a mild side effect	5	10.00	3	15.00	2	6.67	3	13.64	2	7.14

Description of mild side effects	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant describes mild side effects as those that do not greatly impact activities of daily living	11	22.00	2	33.33	2	14.29	5	55.56	1	9.09	1	10.00
Participant describes mild side effects as headaches	7	14.00	1	16.67	3	21.43	0	0.00	2	18.18	1	10.00
Participant describes gastrointestinal problems (diarrhoea and cramping) as mild side effects	6	12.00	2	33.33	4	28.57	0	0.00	0	0.00	0	0.00
Participant does not describe a mild side effect but talks about mitochondrial disease being part of everyday life (Particularly pain)	6	12.00	0	0.00	0	0.00	0	0.00	4	36.36	2	20.00
Participant had not had any mild side effects and could not answer (N/A)	6	12.00	1	16.67	1	7.14	1	11.11	1	9.09	2	20.00
Participant describes mild side effects as something temporary, you can overcome in a short time period	5	10.00	0	0.00	2	14.29	2	22.22	0	0.00	1	10.00
Participant describes increased fatigue (and related irritability) as a mild side effect	5	10.00	2	33.33	1	7.14	0	0.00	0	0.00	2	20.00

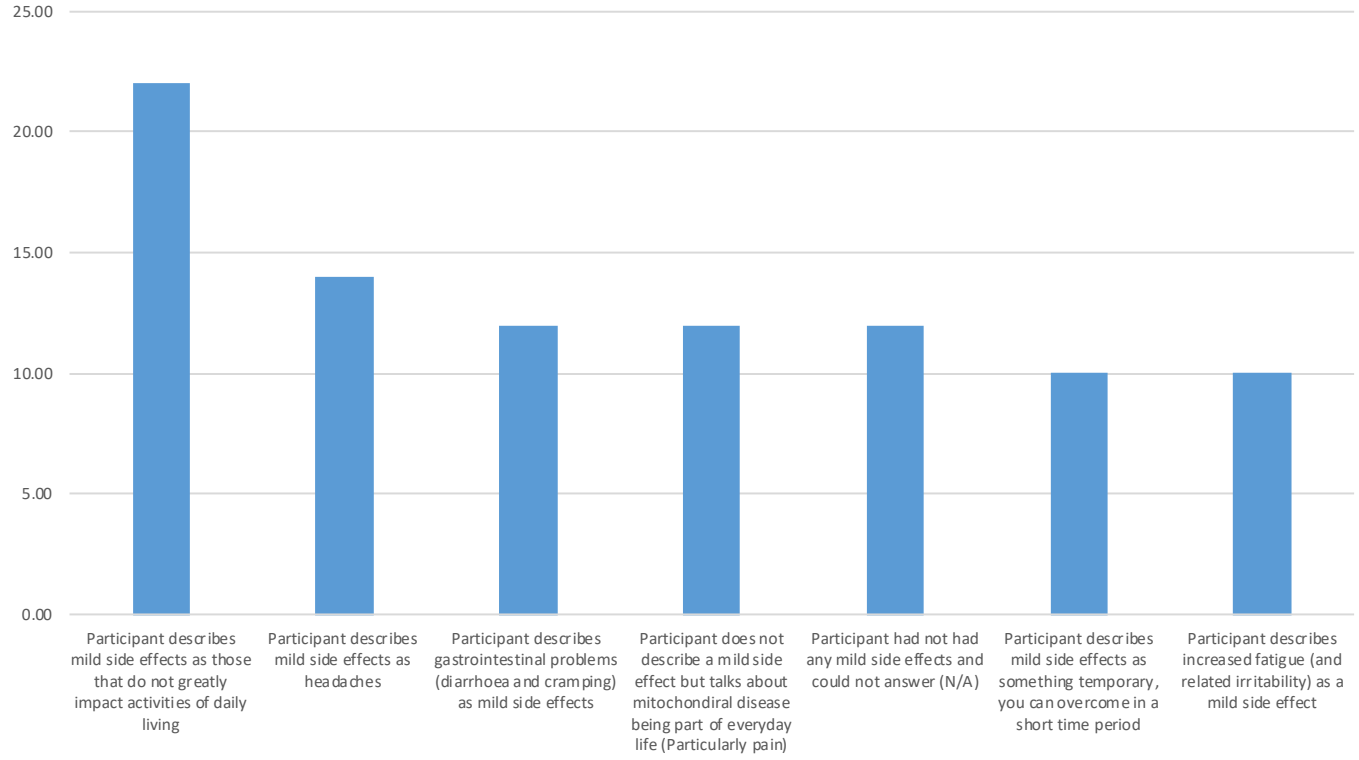


Figure 5.6: Description of severe side effects (% of all participants)

Severe side effects

Participants were asked in the structure interview to provide a description of severe side effects. The most common description of severe side effects were those that limit daily activities for an extended period of time (n=19, 38.00%), seven participants (14.00%) described sever side effects as an effect requiring hospitalisation or medical attention/permanent damage, or a life threatening effect or inability to function.

Participant describes severe side effects as effects limiting their daily activities (may be for an extended period of time).

Inability to perform daily tasks for an extended period. Inability to go to work, prolonged pain. Things that potentially stop me from being able to drive. Participant 2

It would be anything severe enough to stop me from doing my daily activities. Participant 13

Severe side effects make day to day activities hard, generally make me feel emotionally drained and physically tired. I usually will persist with what I'm doing for as long as I am able. Coping with daily activities is generally hard and usually means by the end you are drained of all resources. Participant 38

Participant describes severe side effects as an effect requiring hospitalisation or medical attention or permanent damage

Something, for me, that would be severe would be something that required hospital treatment, or something that severely impacted on my capacity to perform basic daily tasks like getting a meal, having a shower, or being able to be mobile. Participant 7

Severe side affect. Being given medication that have severe reaction to. Going into hospital and fasting and being put on a drip and become very lethargic. Participant 16

Severe side effects are ones that place me in bed or needing medical treatment which is frustrating as medical teams that don't understand the disease brush you off as though your overreacting. Participant 40

In relation to specific side effects that were considered severe, nine participants (18.00%) described severe fatigue, four participants (8.00%) described chronic headaches and four participants (8.00%) described loss of mobility or independence.

Participant describes severe side effects as loss of mobility or independence

The severe problems are the progression so that it is easy to fall over, Getting out of breath easily, difficulty walking with a walker, getting worse, pain in head where hair falling out, sometimes incontinent which makes going out, especially early in the morning, difficult. Participant 14

Severe side effects may include sudden muscle weakness in my legs, which prevents me from doing anything in the day and brings about anxiety and paranoia that I am getting worse. Having a combination of muscle fatigue, impaired hearing and reduced energy can bring about depression and thinking that my body will not recover. Participant 26

Mobility limitations requiring aids. All Myopathic weakness and developing disability including curvature of the spine and deformity and weakness in joints leading to all sorts more problems Aspiration causing pneumonias and inability to communicate via voice . Participant 49

In relation to sub-group variations, participants with a high school or trade education (15.38%), low physical function(25.00%) and low social functioning (26.67%) described severe side effects as effects limiting their daily activities for an extended period of time, less frequently than the general population (38.00%), while those with a university education (62.50%), high physical function (54.44%), high social functioning (55.00%), high general health (50.00%) and hearing impairment (50.00%) described this more frequently.

Table 5.5: Description of severe side effects

Description of severe side effects	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant describes severe side effects as effects limiting their daily activities for an extended period of time	19	38.00	11	36.67	8	40.00	9	33.33	10	43.48
Participant describes severe side effects as severe fatigue	9	18.00	4	13.33	5	25.00	5	18.52	4	17.39
Participant describes severe side effects as an effect requiring hospitalisation or medical attention/permanent damage, or a life threatening effect or inability to function	7	14.00	5	16.67	2	10.00	4	14.81	3	13.04
Participant describes severe side effects as chronic headaches	4	8.00	2	6.67	2	10.00	2	7.41	2	8.70
Participant describes severe side effects as loss of mobility or independence	4	8.00	1	3.33	3	15.00	2	7.41	2	8.70
Participant describes severe side effects as diarrhoea or nausea that affects the whole body	3	6.00	1	3.33	2	10.00	1	3.70	2	8.70
Participant describes severe side effects as cardiovascular issues such as shortness of breath and irregular heart rhythm	3	6.00	1	3.33	2	10.00	1	3.70	2	8.70

Description of severe side effects	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant describes severe side effects as effects limiting their daily activities for an extended period of time	19	38.00	4	15.38	15	62.50	12	50.00	11	32.35
Participant describes severe side effects as severe fatigue	9	18.00	4	15.38	5	20.83	4	16.67	7	20.59
Participant describes severe side effects as an effect requiring hospitalisation or medical attention/permanent damage, or a life threatening effect or inability to function	7	14.00	4	15.38	3	12.50	5	20.83	3	8.82
Participant describes severe side effects as chronic headaches	4	8.00	3	11.54	1	4.17	2	8.33	3	8.82
Participant describes severe side effects as loss of mobility or independence	4	8.00	2	7.69	2	8.33	1	4.17	3	8.82
Participant describes severe side effects as diarrhoea or nausea that affects the whole body	3	6.00	3	11.54	0	0.00	1	4.17	3	8.82
Participant describes severe side effects as cardiovascular issues such as shortness of breath and irregular heart rhythm	3	6.00	3	11.54	0	0.00	0	0.00	3	8.82

Description of severe side effects	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant describes severe side effects as effects limiting their daily activities for an extended period of time	19	38.00	12	54.55	7	25.00	11	42.31	8	33.33
Participant describes severe side effects as severe fatigue	9	18.00	5	22.73	4	14.29	5	19.23	4	16.67
Participant describes severe side effects as an effect requiring hospitalisation or medical attention/permanent damage, or a life threatening effect or inability to function	7	14.00	1	4.55	6	21.43	3	11.54	4	16.67
Participant describes severe side effects as chronic headaches	4	8.00	2	9.09	2	7.14	1	3.85	3	12.50
Participant describes severe side effects as loss of mobility or independence	4	8.00	0	0.00	4	14.29	2	7.69	2	8.33
Participant describes severe side effects as diarrhoea or nausea that affects the whole body	3	6.00	0	0.00	3	10.71	1	3.85	2	8.33
Participant describes severe side effects as cardiovascular issues such as shortness of breath and irregular heart rhythm	3	6.00	1	4.55	2	7.14	2	7.69	1	4.17

Description of severe side effects	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant describes severe side effects as effects limiting their daily activities for an extended period of time	**19**	38.00	11	**55.00**	8	**26.67**	11	**50.00**	8	28.57
Participant describes severe side effects as severe fatigue	**9**	18.00	5	25.00	4	13.33	5	22.73	4	14.29
Participant describes severe side effects as an effect requiring hospitalisation or medical attention/permanent damage, or a life threatening effect or inability to function	**7**	14.00	1	5.00	6	20.00	0	0.00	7	25.00
Participant describes severe side effects as chronic headaches	**4**	8.00	2	10.00	2	6.67	0	0.00	4	14.29
Participant describes severe side effects as loss of mobility or independence	**4**	8.00	1	5.00	3	10.00	2	9.09	2	7.14
Participant describes severe side effects as diarrhoea or nausea that affects the whole body	**3**	6.00	1	5.00	2	6.67	1	4.55	2	7.14
Participant describes severe side effects as cardiovascular issues such as shortness of breath and irregular heart rhythm	**3**	6.00	1	5.00	2	6.67	2	9.09	1	3.57

Description of severe side effects	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant describes severe side effects as effects limiting their daily activities for an extended period of time	**19**	38.00	1	16.67	7	50.00	5	55.56	3	27.27	3	30.00
Participant describes severe side effects as severe fatigue	**9**	18.00	1	16.67	2	14.29	1	11.11	4	36.36	1	10.00
Participant describes severe side effects as an effect requiring hospitalisation or medical attention/permanent damage, or a life threatening effect or inability to function	**7**	14.00	1	16.67	3	21.43	1	11.11	1	9.09	1	10.00
Participant describes severe side effects as chronic headaches	**4**	8.00	0	0.00	1	7.14	1	11.11	1	9.09	1	10.00
Participant describes severe side effects as loss of mobility or independence	**4**	8.00	1	16.67	1	7.14	0	0.00	1	9.09	1	10.00
Participant describes severe side effects as diarrhoea or nausea that affects the whole body	**3**	6.00	0	0.00	1	7.14	1	11.11	0	0.00	1	10.00
Participant describes severe side effects as cardiovascular issues such as shortness of breath and irregular heart rhythm	**3**	6.00	1	16.67	1	7.14	0	0.00	1	9.09	0	0.00

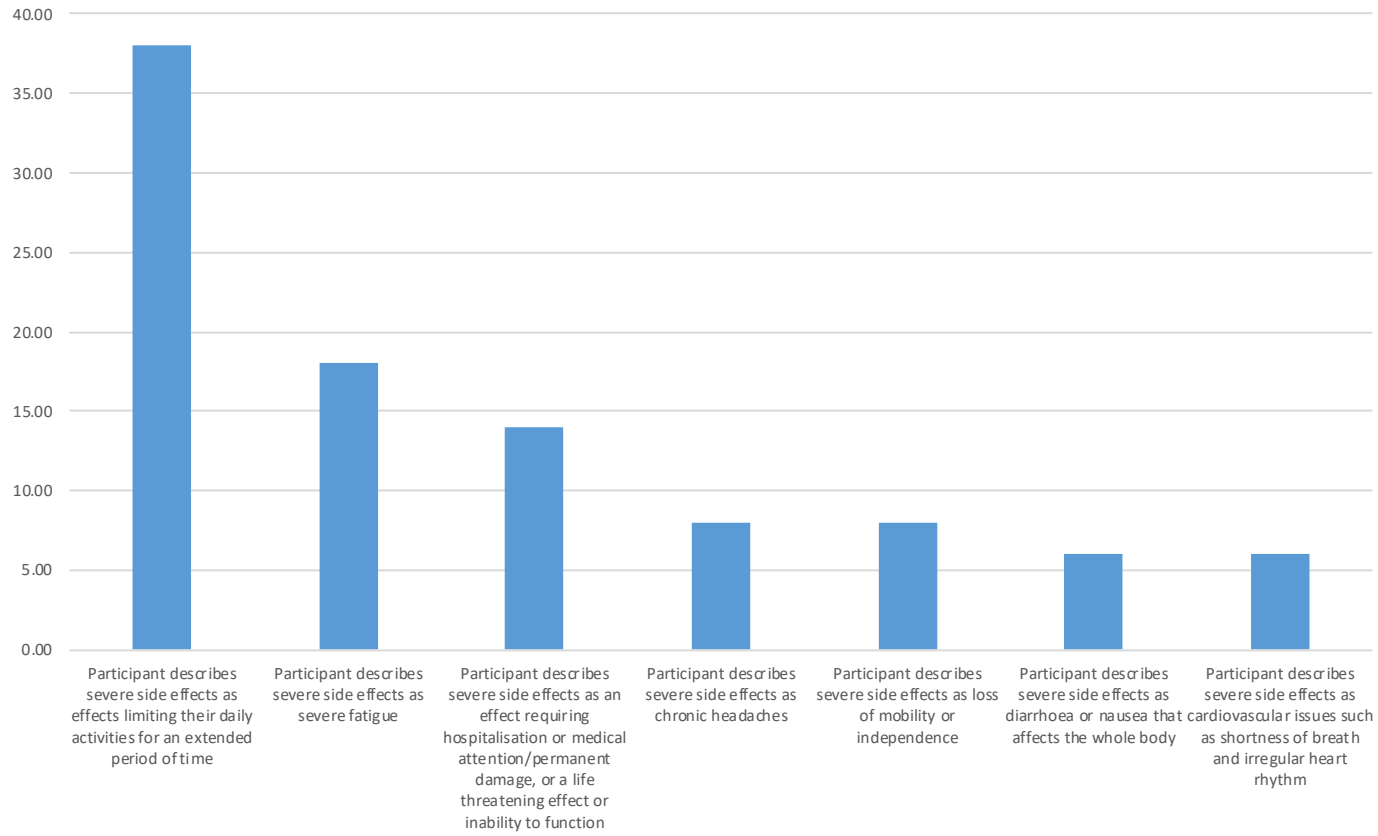


Figure 5.7: Description of severe side effects (% of all participants)

Adherence to medications

Participants were asked in the online questionnaire, if in general, if they were good at taking medicine and sticking to it. The majority of participants were good at sticking to treatments all of the time (n=30, 60.00%) and the remaining were good at sticking to treatments most of the time (n=20, 40.00%). No participants felt they were never, rarely or sometimes good at sticking to treatments.

Table 5.6: Participants ability to stick with treatments

Participant very good at sticking to taking medicine	N=50	Percentage of participants
Never	0	0.00
Rarely	0	0.00
Sometimes	0	0.00
Most of the time	20	40.00
All of the time	30	60.00

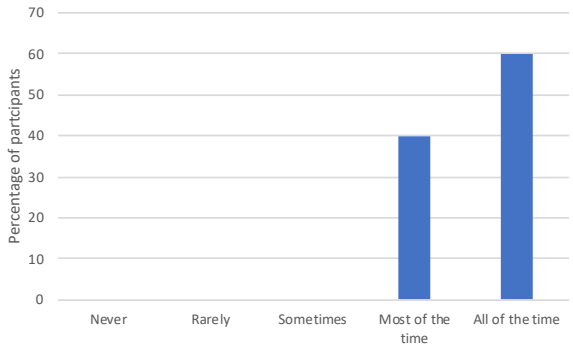


Figure 5.8: Participants ability to stick with treatments

Participants were also asked in the structured interview how long they stick with a therapy before they think it might not be working or give up on it. Close to half of all participants (n=24, 48.00%) describes using treatment for a period of one to three months before deciding if its working.

Participant describes continuing a treatment indefinitely or as recommended by clinician

It would depend on the outcome, the benefits. If somebody said this one stick with all these terrible side effects for six months because we know at the end it will all be good, then I'll stick with it. Participant 3

I always keep doing it unless the doctors tell me otherwise. Participant 9

I'd actually persevere with it for as long as ... I mean, you're right to say I'm looking to the future with this one because we have really experienced that sort of chance but I guess an example if I'm to look back to the concoctions that he's on, I just ... as long as I know that something can't give him any hurt, like be harmful to him I always continue on with it. Participant 45

The next most common theme was continuing a treatment indefinitely or as recommended by clinician/specialist (n=9, 18.00%) and there were six participants (12.00%) that described not trying new medications for mitochondrial disease and such not knowing how long they would continue a treatment.

Participant describes continuing a treatment indefinitely or as recommended by clinician

It would depend on the outcome, the benefits. If somebody said this one stick with all these terrible side effects for six months because we know at the end it will all be good, then I'll stick with it. Participant 3

I always keep doing it unless the doctors tell me otherwise. Participant 9

I'd actually persevere with it for as long as ... I mean, you're right to say I'm looking to the future with this one because we have really experienced that sort of chance but I guess an example if I'm to look back to the concoctions that he's on, I just ... as long as I know that something can't give him any hurt, like be harmful to him I always continue on with it. Participant 45

Participant describes not trying new medications for mitochondrial disease and such not knowing how long they would continue a treatment

I haven't really. Nothing new has been tried since 2012. Like when Neurontin and Lyrica and the other one didn't work, they started on the Oxycontin. It went up to increments and down. They weren't doing anything. They were but I was finding by about 10...11 in the morning that the extreme pain has started to creep back in. I normally have two Panadeine for around that time now. But that's it. My medication hasn't increased now in three years, I've been on the same dose. Participant 18

I don't. It's not applicable for me because I've never had treatments for mito. Generally, as a person, I'm very good at sticking at things and making decisions about where I should go, especially on things like, mental, I don't know what you call them, states. Participant 20

I haven't tried any other medication. Everything else has been supplements. Participant 24

In relation to sub-group variations, participants from rural areas (30.00%) and participants with a hearing impairment (29.17%) reported continuing a treatment indefinitely or as recommended by clinician/specialist more frequently than the general population (18.00%). Participants with a hearing impairment (29.17%) reported using treatment for a period of one to three months before deciding if its working less frequently than the general population (48.00%), while those with high social functioning (60.00%) and high general health (59.09%) reported this more frequently.

Table 5.7: How long patients stick to a therapy

Adherence to treatment	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant describes using treatment for a period of one to three months before deciding if its working	24	48.00	15	50.00	9	45.00	11	40.74	13	56.52
Participant describes continuing a treatment indefinitely or as recommended by clinician/specialist	9	18.00	3	10.00	6	30.00	5	18.52	4	17.39
Participant describes not trying new medications for mitochondrial disease and such not knowing how long they would continue a treatment	6	12.00	5	16.67	1	5.00	5	18.52	1	4.35
Participant describes the time period varying depending on what the treatment is, however if it is causing severe discomfort then they would cease	4	8.00	4	13.33	0	0.00	3	11.11	1	4.35
Participant describes continuing a treatment indefinitely unless there is a severe reaction	2	4.00	1	3.33	1	5.00	2	7.41	0	0.00

Adherence to treatment	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant describes using treatment for a period of one to three months before deciding if its working	24	48.00	13	50.00	11	45.83	7	29.17	18	52.94
Participant describes continuing a treatment indefinitely or as recommended by clinician/specialist	9	18.00	3	11.54	6	25.00	7	29.17	7	20.59
Participant describes not trying new medications for mitochondrial disease and such not knowing how long they would continue a treatment	6	12.00	3	11.54	3	12.50	3	12.50	4	11.76
Participant describes the time period varying depending on what the treatment is, however if it is causing severe discomfort then they would cease	4	8.00	3	11.54	1	4.17	2	8.33	2	5.88
Participant describes continuing a treatment indefinitely unless there is a severe reaction	2	4.00	1	3.85	1	4.17	2	8.33	1	2.94

Adherence to treatment	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant describes using treatment for a period of one to three months before deciding if its working	24	48.00	10	45.45	14	50.00	15	57.69	9	37.50
Participant describes continuing a treatment indefinitely or as recommended by clinician/specialist	9	18.00	6	27.27	3	10.71	6	23.08	3	12.50
Participant describes not trying new medications for mitochondrial disease and such not knowing how long they would continue a treatment	6	12.00	2	9.09	4	14.29	3	11.54	3	12.50
Participant describes the time period varying depending on what the treatment is, however if it is causing severe discomfort then they would cease	4	8.00	2	9.09	2	7.14	1	3.85	3	12.50
Participant describes continuing a treatment indefinitely unless there is a severe reaction	2	4.00	2	9.09	0	0.00	1	3.85	1	4.17

Adherence to treatment	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant describes using treatment for a period of one to three months before deciding if its working	24	48.00	12	60.00	12	40.00	13	59.09	11	39.29
Participant describes continuing a treatment indefinitely or as recommended by clinician/specialist	9	18.00	5	25.00	4	13.33	5	22.73	4	14.29
Participant describes not trying new medications for mitochondrial disease and such not knowing how long they would continue a treatment	6	12.00	2	10.00	4	13.33	2	9.09	4	14.29
Participant describes the time period varying depending on what the treatment is, however if it is causing severe discomfort then they would cease	4	8.00	0	0.00	4	13.33	1	4.55	3	10.71
Participant describes continuing a treatment indefinitely unless there is a severe reaction	2	4.00	2	10.00	0	0.00	1	4.55	1	3.57

Adherence to treatment	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant describes using treatment for a period of one to three months before deciding if its working	**24**	48.00	3	50.00	7	50.00	4	44.44	5	45.45	5	50.00
Participant describes continuing a treatment indefinitely or as recommended by clinician/specialist	**9**	18.00	1	16.67	4	28.57	2	22.22	1	9.09	1	10.00
Participant describes not trying new medications for mitochondrial disease and such not knowing how long they would continue a treatment	**6**	12.00	0	0.00	1	7.14	1	11.11	3	27.27	1	10.00
Participant describes the time period varying depending on what the treatment is, however if it is causing severe discomfort then they would cease	**4**	8.00	2	33.33	1	7.14	0	0.00	0	0.00	1	10.00
Participant describes continuing a treatment indefinitely unless there is a severe reaction	**2**	4.00	0	0.00	0	0.00	0	0.00	1	9.09	1	10.00

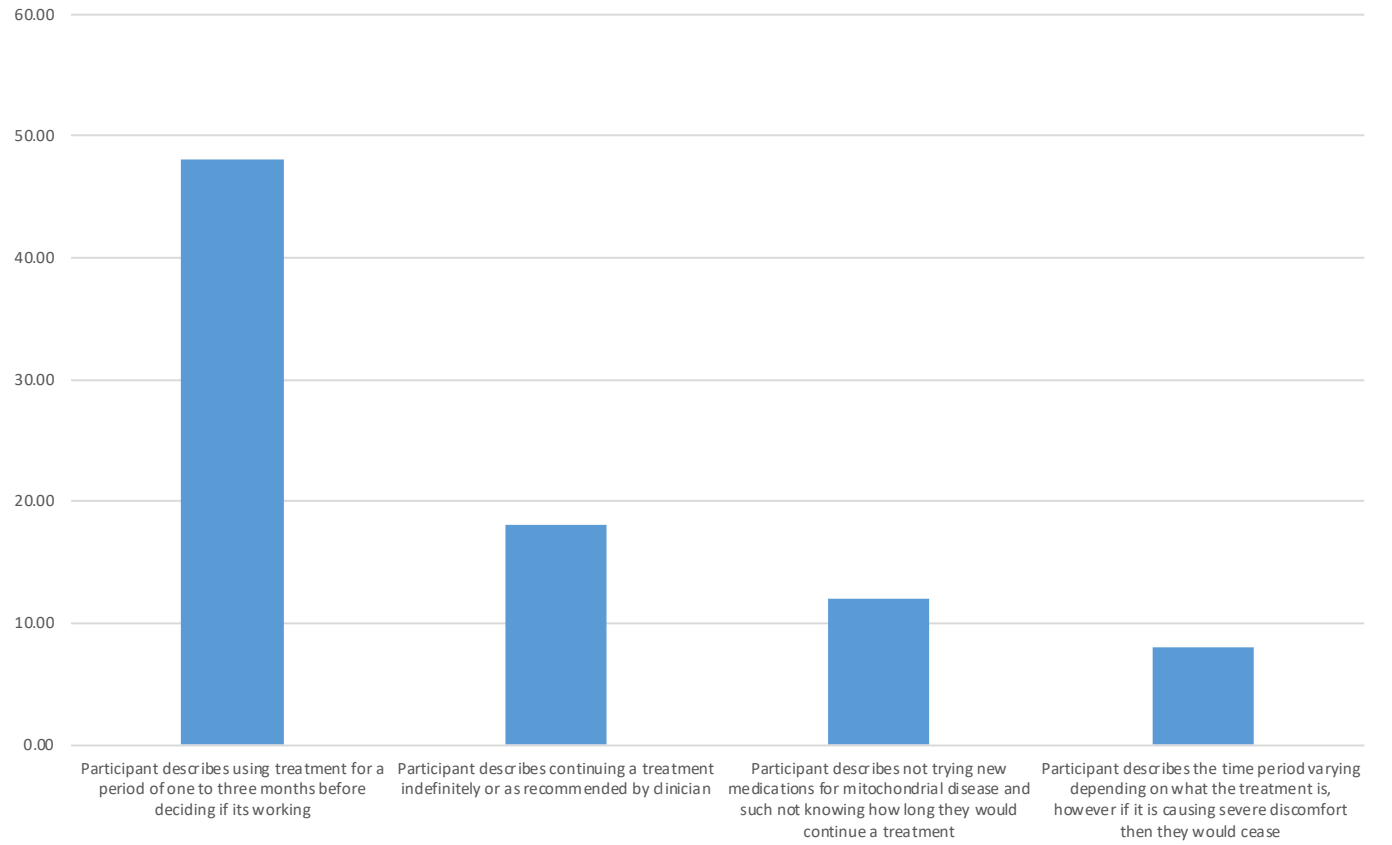


Figure 5.9: How long patients stick to a therapy (% of all participants)

Impact of treatment

Participants were asked what needed to change for them to feel as though a treatment was working. The most common description was needing to feel more energetic, and increase in physical ability, to know a treatment is working (n=20, 40.00%).

Participant describes needing to feel more energetic, and increase in physical ability, to know a treatment is working

Well, changing my energy levels and fatigue level. That is a big benefit to me. I'd also walk long distances. All I can do at present is walking the pool, which I was doing, and a bit of therapy. Walking unaided down the street, when I walk about 15 meters and I would need a rest, whatever. (An improvement of that would mean it has improved?) Yes, and get up stairs, too. That would be a big benefit. I used to get up the stairs using my hands on the armrest, can't do that now. Participant 6

If I felt better. [chuckles] If I wasn't so bloody tired all the time. Participant 10

I would hope that I have more energy. I think that's the first thing, it's just to feel like I've got more energy in me and be able to do things. I guess that's the main thing because that's where it all falls apart. Just being able to get through the day without feeling really tired and exhausted, being able to just get home and do things longer but yes, it's the energy factor I think for me, I'm looking for. Participant 26

My energy levels would be one thing. The biggest…I'd like to be able to not be so tired….We went back to school and within about 10 minutes of being there, I thought I hadn't been on holidays at all. I spent one day doing some gardening at home and it took me about three weeks to get over the fact that I've done that. Participant 38

This was followed by needing to see improved symptoms by clinical measurement (test result) (n=13, 26.00%) and needing to generally feel better to know that a treatment is working (n=9, 13.00%).

Participant describes improved symptoms by clinical measurement (test result)

I suppose, I would usually have the blood test, definitely. If I feel a lot better, then that's a good thing. Now I'm just taking the tablets I told you, Q10. Participant 1

Everything is got to have a benefit. If it's not working, if it's not getting a result whether that be something that can be measured by way of blood tests or whether it's a feeling of not feeling right on it, get off it. It's not working. Participant 2

When they review it and ask me how it's gone. "Yes, that's good. I haven't had an episode for so and so." Participant 29

Participant describes needing to generally feel better to know that a treatment is working

Anything that could improve how you feel and, you know, reduce the lactic acidosis. Anything that you actually feel … Health. Health is, of course, you know, in long term, you know? Participant 15

Normally energy. My mood will change so I'll either feel happy or better. I can just feel a difference within. I know it sounds stupid but I know when I don't have my antidepressant I feel more grumpy and Mum's like, "You only haven't had it for one day." I'm like, "I just feel it." Participant 26

Well, I guess, I have had some improvement in energy and in general, like just every day like sleep and I look better and I generally feel better. Participant 47

There were five participants (10.00%) that described needing to reduce pain to know a treatment is working and five participants (10.00%) needing to improve their quality of life to know a treatment is working.

Participant describes needing to reduce pain to know a treatment is working

The extreme soreness. I don't know how to explain it, but I'm so sore to touch. If the grandchildren grab me on the arm or something, and they are only little, if they do, oh goodness. It sends me through the roof. Just so sore. Participant 18

That would be dropping the level of pain, or the pain might steers to go well. Participant 23

To be without chronic pain, chronic fatigue is a big part of it, to be without those two things as much as possible. Participant 35

Participant describes needing to improve their quality of life to know a treatment is working

My quality of life, so my capacity to perhaps have increased mobility, better finer motor skills. Participant 7

I will continue with a treatment when I can see a measurable reduction in symptoms and/or improvement in a quality of life symptom. Participant 30

I guess the quality of life. Just the fact that she doesn't crash as often or she has more energy. She can function, she can think without being so distressed because she gets brain fog and fatigue. Participant 49

In relation to sub-group variations, participants from metropolitan areas (26.67%) and participants from high socio-economic areas (25.93%) reported needing to feel more energetic, and increase in physical ability, to know a treatment is working, less frequently than the general population (40.00%) while participants from rural areas (60.00%), participants from low socio-economic areas (56.52%). Participants with a hearing impairment (41.67%) reported improved symptoms by clinical measurement (test result) more frequently than the general population (26.00%). Participants with high physical functioning (31.82%) described needing to generally feel better to know that a treatment is working, more frequently than the general population (18.00%).

Table 5.8: What needs to change to know a treatment has worked

What needs to change to feel like treatment is effective	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant describes needing to feel more energetic, and increase in physical ability, to know a treatment is working	**20**	40.00	8	**26.67**	12	**60.00**	7	**25.93**	13	**56.52**
Participant describes improved symptoms by clinical measurement (test result)	**13**	26.00	9	30.00	4	20.00	9	33.33	4	17.39
Participant describes needing to generally feel better to know that a treatment is working	**9**	18.00	7	23.33	2	10.00	6	22.22	3	13.04
Participant describes needing to reduce pain to know a treatment is working	**5**	10.00	1	3.33	4	20.00	1	3.70	4	17.39
Participant describes needing to improve their quality of life to know a treatment is working	**5**	10.00	3	10.00	2	10.00	3	11.11	2	8.70

What needs to change to feel like treatment is effective	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant describes needing to feel more energetic, and increase in physical ability, to know a treatment is working	**20**	40.00	10	38.46	10	41.67	8	33.33	17	50.00
Participant describes improved symptoms by clinical measurement (test result)	**13**	26.00	5	19.23	8	33.33	10	**41.67**	6	17.65
Participant describes needing to generally feel better to know that a treatment is working	**9**	18.00	5	19.23	4	16.67	4	16.67	7	20.59
Participant describes needing to reduce pain to know a treatment is working	**5**	10.00	3	11.54	2	8.33	3	12.50	2	5.88
Participant describes needing to improve their quality of life to know a treatment is working	**5**	10.00	2	7.69	3	12.50	3	12.50	2	5.88

What needs to change to feel like treatment is effective	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant describes needing to feel more energetic, and increase in physical ability, to know a treatment is working	**20**	40.00	7	31.82	13	46.43	12	46.15	8	33.33
Participant describes improved symptoms by clinical measurement (test result)	**13**	26.00	7	31.82	6	21.43	9	34.62	4	16.67
Participant describes needing to generally feel better to know that a treatment is working	**9**	18.00	7	**31.82**	2	7.14	7	26.92	2	8.33
Participant describes needing to reduce pain to know a treatment is working	**5**	10.00	3	13.64	2	7.14	1	3.85	4	16.67
Participant describes needing to improve their quality of life to know a treatment is working	**5**	10.00	0	0.00	5	17.86	2	7.69	3	12.50

What needs to change to feel like treatment is effective	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant describes needing to feel more energetic, and increase in physical ability, to know a treatment is working	**20**	40.00	10	50.00	10	33.33	7	31.82	13	46.43
Participant describes improved symptoms by clinical measurement (test result)	**13**	26.00	6	30.00	7	23.33	6	27.27	7	25.00
Participant describes needing to generally feel better to know that a treatment is working	**9**	18.00	5	25.00	4	13.33	4	18.18	5	17.86
Participant describes needing to reduce pain to know a treatment is working	**5**	10.00	1	5.00	4	13.33	2	9.09	3	10.71
Participant describes needing to improve their quality of life to know a treatment is working	**5**	10.00	0	0.00	5	16.67	1	4.55	4	14.29

What needs to change to feel like treatment is effective	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant describes needing to feel more energetic, and increase in physical ability, to know a treatment is working	**20**	40.00	3	50.00	5	35.71	5	55.56	4	36.36	3	30.00
Participant describes improved symptoms by clinical measurement (test result)	**13**	26.00	0	0.00	3	21.43	5	55.56	3	27.27	2	20.00
Participant describes needing to generally feel better to know that a treatment is working	**9**	18.00	2	33.33	4	28.57	2	22.22	1	9.09	0	0.00
Participant describes needing to reduce pain to know a treatment is working	**5**	10.00	0	0.00	1	7.14	1	11.11	1	9.09	2	20.00
Participant describes needing to improve their quality of life to know a treatment is working	**5**	10.00	1	16.67	3	21.43	0	0.00	1	9.09	0	0.00

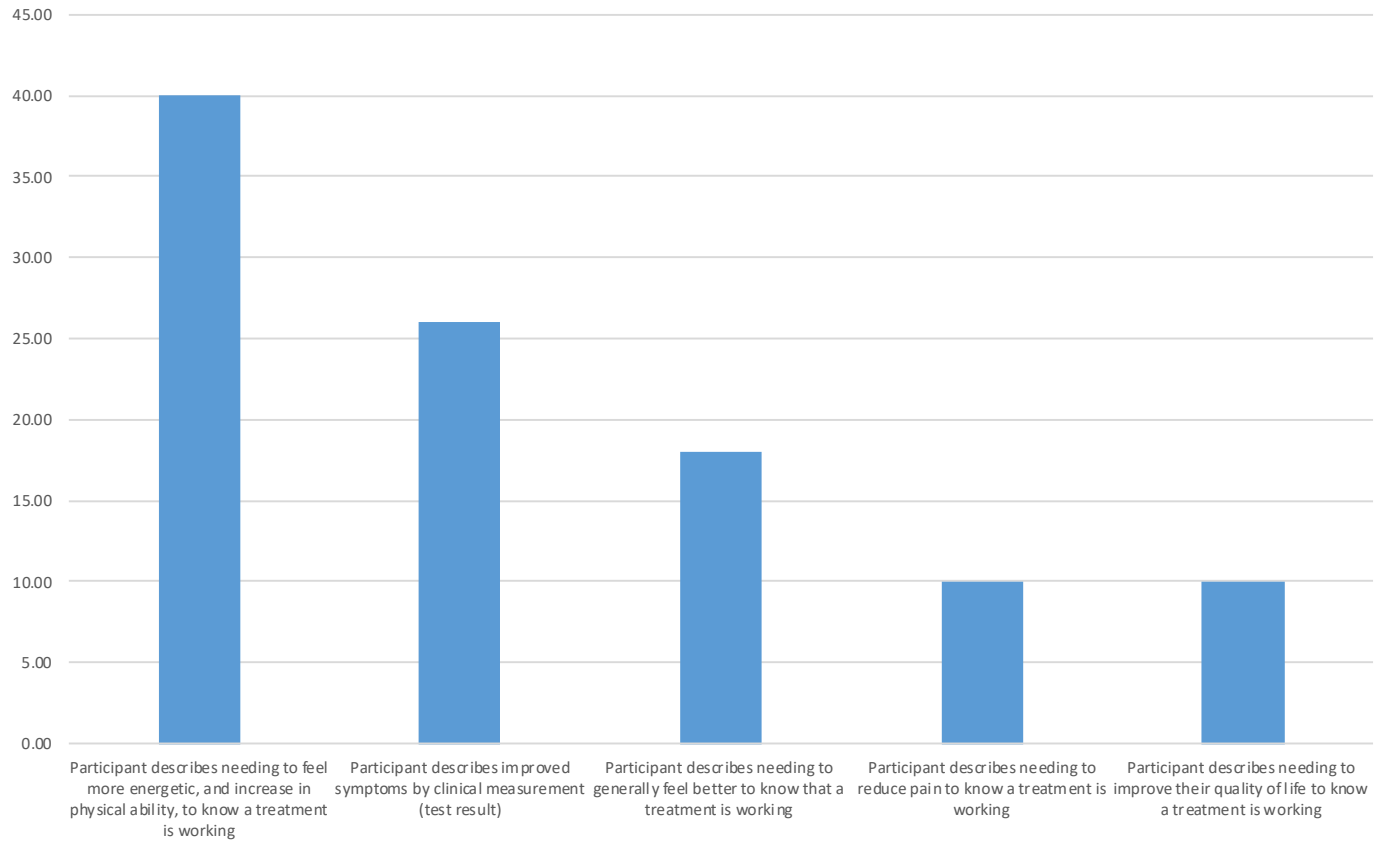


Figure 5.10: What needs to change to know a treatment has worked

Use of complementary therapies

Participants were asked whether they had used any complementary therapies. The most common therapies that were considered complementary and described by participants were vitamins, minerals and supplements (n=14, 28.00%) and allied health e.g. physiotherapy (including massage and hydrotherapy), speech therapy, occupational therapy (n=14, 14.00%).

Participant describes taking vitamin, mineral and various supplements

Yes. I've taken magnesium tablets from time to time in the hope that that'll make me feel better. [laughs] They don't really work very well, though. They may sometimes though. No, not really. Participant 10

I tried creatine. I was told creatine helps. Folic acid as well. There are few studies on folic acid. I had like a sort of cocktail going on for a while. Creatine, folic acid, CoQ10 and L-carnitine. That was about it. Participant 11

I'm just trialling some turmeric. I know it's not proven or anything, but I thought I'd ... I don't go back to my Rheumatologist until July, so I'm hoping if I tried turmeric daily, and it makes any difference I'd be able to say to him, "I don't like your choices, I'll stick with the turmeric for a while." But ask me that at the end of July. Participant 36

Participant describes having a form of allied health e.g. physiotherapy (including massage and hydrotherapy), speech therapy, occupational therapy

I did do physio for a bit. Then that wasn't really helping in the sense that I wasn't getting any pain relief from that because I did get a lot of muscle pain. Participant 1

I gave massage a go but my massage therapist was honest with me and said that I had no muscle left, and that massage wasn't going to work for me. Participant 12

I tried all sorts of things to get rid of the migraines and also physiotherapy but perhaps, the physiotherapy wasn't exactly targeted to the muscles. I'm looking at starting physiotherapy for that to maintain my muscle strength. Participant 26

Right. Yes. Massages, I find helpful because I get a lot of muscle soreness and sore neck and shoulders and things like that. I do find that it just gives you a general feeling of well being, but also it just reduces the discomfort. Yes, I do find that good. Participant 34

The next most frequent complementary therapies described were alternative medicine, e.g. osteopathy, acupuncture, chiropractor, Bowen therapy (n=12, 24.00%).

Participant describes having a form of alternative medicine, e.g. osteopathy, acupuncture, chiropractor, Bowen therapy

I have a lot of osteopath appointments and she tries to alter my migraines and fix through soft manipulation on my head and on my back. Participant 19

Yes, because I initially had headaches or migraines. Something around the time that I lost my hearing but again, I'm not 100% sure that it's related but I think it is. I tried acupuncture for that. Participant 26

All we've ever used for her was chiropractic. That was good at the times that we used it. It was good. We're reluctant to because we don't have anyone to help us assess her at the moment and her muscles and bones have worsened quite a lot. The chiropractors and everyone said that they'd be happy to work with someone that can be guided. Even they notice the difference. Participant 49

There were also 11 participants (22.00%) that noted that they did not use any complementary therapies.

Table 5.9: Use of complementary therapies

Use of complementary therapies	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant describes taking vitamin, mineral and various supplements	14	28.00	7	23.33	7	35.00	9	33.33	5	21.74
Participant describes having a form of allied health e.g. physiotherapy (including massage and hydrotherapy), speech therapy, occupational therapy	14	28.00	9	30.00	5	25.00	7	25.93	7	30.43
Participant describes having a form of alternative medicine, e.g. osteopathy, acupuncture, chiropractor, Bowen therapy	12	24.00	7	23.33	5	25.00	5	18.52	7	30.43
Participant describes not trying any complementary medicines	11	22.00	8	26.67	3	15.00	7	25.93	4	17.39

Use of complementary therapies	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant describes taking vitamin, mineral and various supplements	14	28.00	8	30.77	6	25.00	5	20.83	9	26.47
Participant describes having a form of allied health e.g. physiotherapy (including massage and hydrotherapy), speech therapy, occupational therapy	14	28.00	6	23.08	8	33.33	6	25.00	12	35.29
Participant describes having a form of alternative medicine, e.g. osteopathy, acupuncture, chiropractor, Bowen therapy	12	24.00	6	23.08	6	25.00	6	25.00	9	26.47
Participant describes not trying any complementary medicines	11	22.00	5	19.23	6	25.00	6	25.00	6	17.65

Use of complementary therapies	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant describes taking vitamin, mineral and various supplements	14	28.00	5	22.73	9	32.14	9	34.62	5	20.83
Participant describes having a form of allied health e.g. physiotherapy (including massage and hydrotherapy), speech therapy, occupational therapy	14	28.00	5	22.73	9	32.14	7	26.92	7	29.17
Participant describes having a form of alternative medicine, e.g. osteopathy, acupuncture, chiropractor, Bowen therapy	12	24.00	7	31.82	5	17.86	6	23.08	6	25.00
Participant describes not trying any complementary medicines	11	22.00	5	22.73	6	21.43	5	19.23	6	25.00

Use of complementary therapies	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant describes taking vitamin, mineral and various supplements	14	28.00	5	25.00	9	30.00	7	31.82	7	25.00
Participant describes having a form of allied health e.g. physiotherapy (including massage and hydrotherapy), speech therapy, occupational therapy	14	28.00	7	35.00	7	23.33	4	18.18	10	35.71
Participant describes having a form of alternative medicine, e.g. osteopathy, acupuncture, chiropractor, Bowen therapy	12	24.00	5	25.00	7	23.33	6	27.27	6	21.43
Participant describes not trying any complementary medicines	11	22.00	4	20.00	7	23.33	6	27.27	5	17.86

Use of complementary therapies	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant describes taking vitamin, mineral and various supplements	**14**	28.00	2	33.33	4	28.57	0	0.00	5	45.45	3	30.00
Participant describes having a form of allied health e.g. physiotherapy (including massage and hydrotherapy), speech therapy, occupational therapy	**14**	28.00	2	33.33	6	42.86	2	22.22	2	18.18	2	20.00
Participant describes having a form of alternative medicine, e.g. osteopathy, acupuncture, chiropractor, Bowen therapy	**12**	24.00	3	50.00	4	28.57	1	11.11	1	9.09	3	30.00
Participant describes not trying any complementary medicines	**11**	22.00	1	16.67	1	7.14	4	44.44	2	18.18	3	30.00

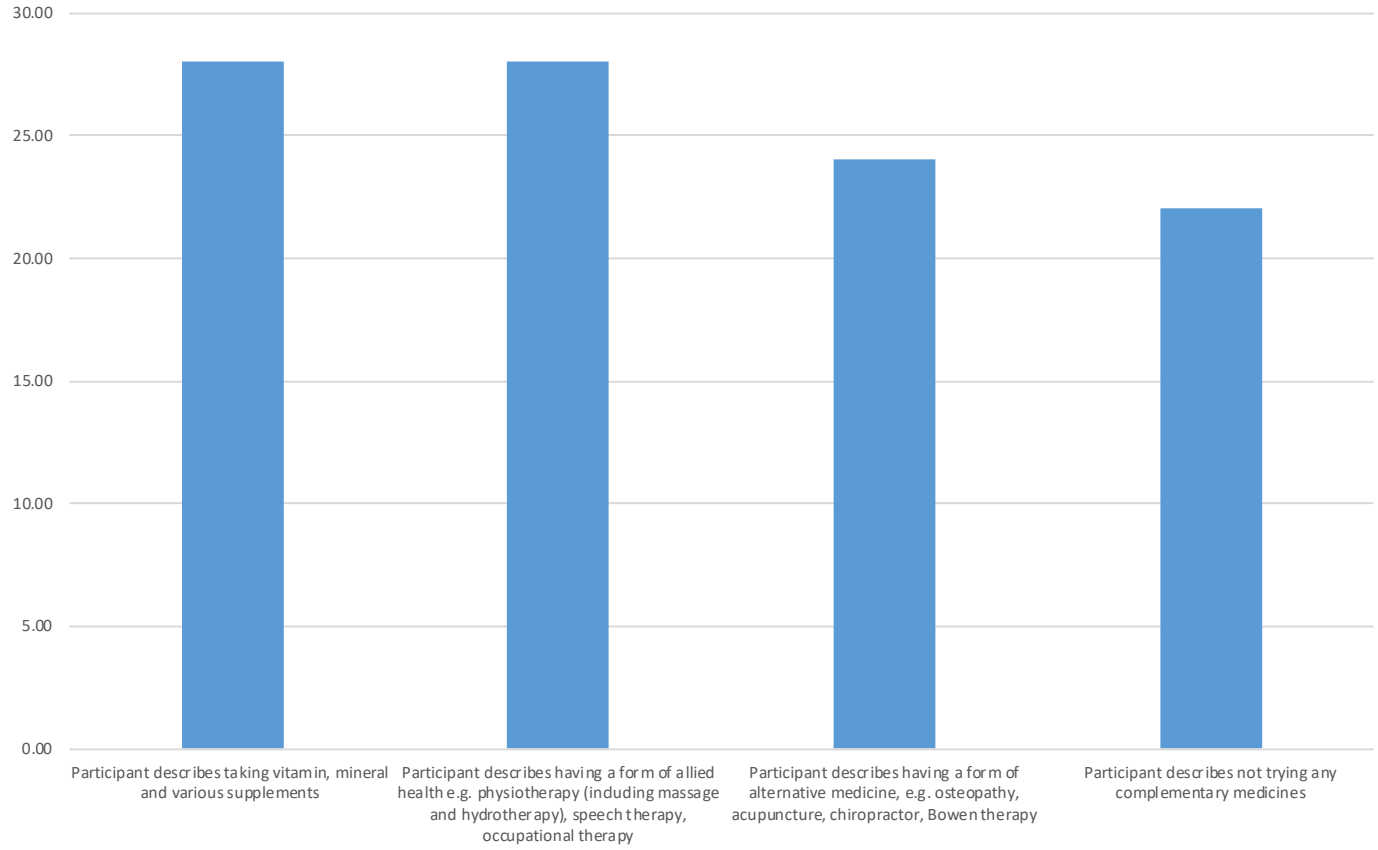


Figure 5.11: Use of complementary therapies (% of all participants)

Service provision and affordability

Details about the healthcare system, where participants were treated for mitochondrial disease and financial implications from treatment are listed in Tables 5.10 to 5.12.

The main physician treating participants for mitochondrial disease were general practitioners (N=19, 38.00%), followed by neurologists (N=12, 24.00%) and mitochondrial specialists (N=11, 22.00%).

Participants had access to a general practitioner (n=48, 96.00%), neurologist (n=43, 86.00%), mitochondrial specialist (n=29, 58.00%) and cardiologist (n=28, 56.00%) for the treatment of their mitochondrial disease.

The majority of patients had private healthcare insurance (n=37, 74.00%), 29 (58.00%) participants were treated as public patients, 12 (24.00%) as private patients and 9 (18.00%) as equally public and private patients. The majority of participants were treated in the public hospital system (n=32, 64.00%).

Almost half of participants have never missed medical appointments due to cost (n=24, 48.00%), and most have never been unable to afford prescription medications (n=34, 64.00%). Almost half of participants have found it somewhat to extremely difficult paying for basic needs due to their diagnosis with mitochondrial disease (n=24, 48.00%).

The work status for a number of participants changed due to their diagnosis with mitochondrial disease with about a quarter of participants reducing the number of hours worked (n=13, 26,00%), and 19 (38.00%) quitting their jobs (Table 5.12). Of those that had a partner or carer, four carers/partners had to quit their job (23.53%), seven had to reduce the number of hours worked (41.18%), carers have had to take leave either with pay (n=2, 11.76%), or without pay (n=5, 29.41%).

Table 5.10: Provider of treatment

Health professional	N=50	Percent
General practitioner	19	38.00
Neurologist	12	24.00
Mitochondrial specialist	11	22.00
Geneticist	2	4.00
Metabolic Medicine	2	4.00
Endocrinologist	1	2.00
Functional medicine specialist	1	2.00
neuro-ophthalmologist	1	2.00
Not currently receiving treatment or management	1	2.00

Table 5.11: Access to health services

Access to health professionals	N=50	%
General Practitioner	48	96.00
Neurologist	43	86.00
Mitochondrial specialist	29	58.00
Cardiologist/Heart specialist	28	56.00
Gastroenterologist/Digestive system specialist	25	50.00
Physiotherapy	25	50.00
Dietitian	24	48.00
Geneticist	23	46.00
Counselling or psychological support	19	38.00
Occupational therapy	17	34.00
Registered Nurse	17	34.00
Endocrinologist/diabetes,hormone, thyroid specialist	14	28.00
Psychiatrist	13	26.00
Respiratory physician	12	24.00
Genetic counsellor	11	22.00
Nephrologist/Kidney specialist	9	18.00
Ophthalmologist	8	16.00
Paediatrician	8	16.00
Hepatologist/Liver specialist	4	8.00
Audiologist	3	6.00
Pain Specialist	2	4.00
Chiro and myotherapy	1	2.00
Dentist	1	2.00
ENT	1	2.00
Gynaecologist	1	2.00
Neuro-ophthalmologist	1	2.00
Orthopaedics	1	2.00
Rheumatologist	1	2.00
Sexual health physician	1	2.00
Social Worker	1	2.00

Table 5.12: Service provision and affordability

		N=50	Percent
Private health insurance	Yes	37	74.00
	No	13	26.00
Treated as public or private patient	Equally public and private	9	18.00
	Private	12	24.00
	Public	29	58.00
Primary hospital system treated in	Both public and private	10	20.00
	Private	8	16.00
	Public	32	64.00
Had to delay or cancel healthcare appointments due to affordability	Never	24	48.00
	Rarely	9	18.00
	Sometimes	13	26.00
	Often	3	6.00
	Very often	1	2.00
Unable to fill prescription due to cost	Never	34	68.00
	Rarely	9	18.00
	Sometimes	7	14.00
	Often	0	
	Very often	0	
Difficult to pay for basic needs as a result of mitochondrial disease diagnosis	Not at all difficult	19	38.00
	Slightly difficult	7	14.00
	Somewhat difficult	12	24.00
	Moderately difficult	8	16.00
	Extremely difficult	4	8.00
Change in employment due to mitochondrial disease (can choose more than one option)	I have accessed my Superannuation early due to my mitochondrial disease	6	12.00
	I have had to quit my job	19	38.00
	I have reduced the number of hours that I work	13	26.00
	I have taken leave from work with pay	4	8.00
	I have taken leave from work without pay	3	6.00
	I was retired or did not have a job when I was diagnosed with mitochondrial disease	13	26.00
	My work status has not changed since I was diagnosed with mitochondrial disease	10	20.00
Change in carer job status (can choose more than one option) (N=34)	My partner/main carer/other carer had to quit their job	4	23.53
	My partner/main carer/other carer reduced the number of hours that they work	7	41.18
	My partner/main carer/other carer took leave from work without pay	5	29.41
	My partner/main carer/other carer was retired or did not have a job	7	41.12
	My partner/main carer/other carer took leave from work with pay	2	11.76
	The employment status of my partner/main carer/other carer status has not changed	14	41.18
	I do not have a partner or other carer	16	

Monthly out of pocket expenses

Participants were asked what their monthly spend was on mitochondrial disease, four participants had no expenses (8.89%), 10 (22.22%) spent less than $100, 17 (37.78%) spent more than $100 but less than $500, three (6.69%) spent between $501 and $1000 per month, and 11 (24.44%) spent more than $1000 per month.

Table 5.13: Approximate monthly spend on mitochondrial disease

Monthly expenses (n=45)	N=	Percentage of participants
$0	4	8.89
$1 - $100	10	22.22
$101 - $500	17	37.78
$501-$1000	3	6.69
>$1000	11	24.44

Participants were then asked if their monthly spend due to mitochondrial disease was a significant burden, 20 participants found it to be extremely or moderately significant (40.00%), 20 participants found it somewhat or slightly significant (40.00%) and ten found not significant at all (20.00%).

Table 5.14: Burden of monthly spend on mitochondrial disease

Cost of monthly treatment a significant burden	N=50	Percent
Extremely significant	14	12.00
Moderately significant	6	16.00
Somewhat significant	16	10.00
Slightly significant	4	12.00
Not at all significant	10	50.00

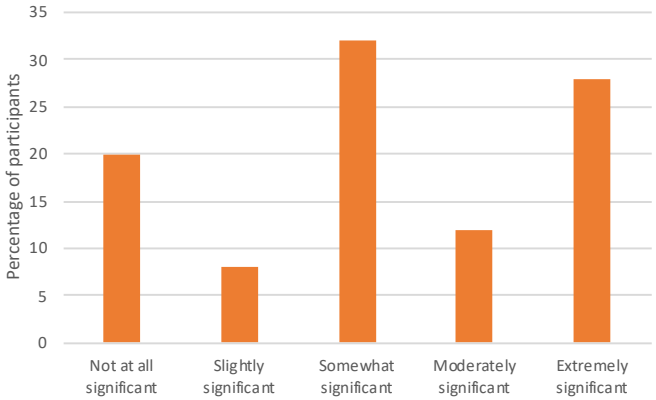


Figure 5.12: Burden of monthly spend on mitochondrial disease

Experience of respect during treatment

Participants were asked if they felt they had been treated with respectfully throughout their treatment. Half of the participants felt that they had been treated respectfully with the exception of one or two occasions (n=25, 50.00%), 18 felt that they had been treated respectfully (36.00%) and seven felt they had not been treated respectfully (14.00%).

Table 5.15: Participant feels that they have been treated respectfully throughout treatment

Have you been treated respectfully throughout your experience?	n=50	Percentage of participants
No	7	14.00
Yes	18	36.00
Yes, with the exception of one or two occasions	25	50.00

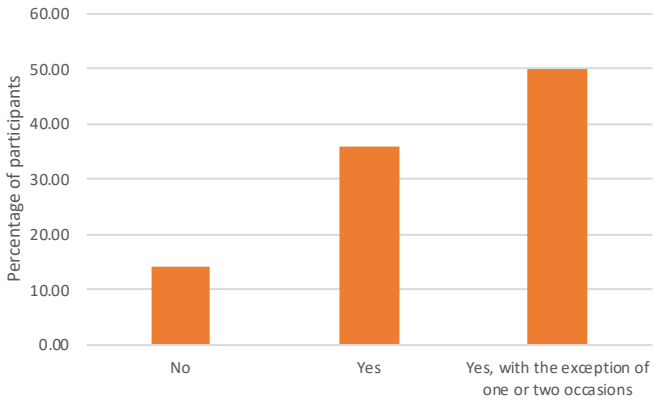


Figure 5.13: Participant feels that they have been treated respectfully throughout treatment

Section 6 Information and communication

Section 6: Information and communication

Access to information

- The most common response from over half of all participants was accessing information from the Australian Mitochondrial Disease Foundation (n=32, 64.00%). The next most common theme was accessing information via the internet (n=25, 50.00%). There were 14 participants (28.00%) that described accessing information from medical journals and peer reviewed papers and 13 participants (26.00%) that described accessing information from online forums including Facebook.
- In relation to sub-group variations, participants from rural areas (75.00%), participants with a high school or trade education (76.92), participants with low physical functioning (75.00%) and low general health (75.00%) reported accessing information from the Australian Mitochondrial Disease Foundation more frequently than the general population (64.00%), while participants with a university education (50.00%) and high physical functioning reported this less frequently. Participants from rural areas (65.00%) and those with high physical function (68.18%) reported accessing information from the internet more frequently than the general population (50.00%), while those from low socio-economic areas (37.04%) and those with low physical functioning (35.71%) reported this less frequently. Participants from rural areas (15.00%) reported accessing medical journals less frequently than the general population (28.00%).

Information that was helpful

- There was a range of information that participants found particularly helpful including information from the AMDF (n=9, 18.00%) research papers (n=7, 14.00%), communicating with others with mitochondrial disease (n=7, 14.00%) and information from clinical teams (n=5, 10.00%).

Information that was not helpful

- The most common theme described by 22 participant (44.00%) was that no information was unhelpful. There were no other themes noted by more than five participants, however where participants made a comment about information that was not helpful, this included stories about other patients (n=3, 6.00%), lack of concise yet comprehensive information (n=3, 6.00%), and information that is too general (n=2, 4.00%) or too scientific (n=2, 4.00%).

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. The most common theme was talking to someone (n=25, 50.00%) of which, five participants specified a preference for talking to someone face-to-face. The next most common theme was a preference for information online (n=21, 42.00%) and a preference for information in a written format such as a booklet (n=7, 14.00%).
- In relation to sub-group variations, participants with a hearing impairment (29.12%) described a preference for online information less frequently than the general population (42.00%), while participants with low physical functioning (53.57%) and low general health (53.57%) reported this preference more frequently. Participants with a hearing impairment (25.00%) reported a preference for accessing written information more frequently than the general population (14.00%)

Timing of information

- The most common time that participants described being receptive to receiving information was at the time of diagnosis (n=18, 36.00%) and this was followed by participants describing that there was not a specific time that they were most receptive and that it is an ongoing process (n=10, 20.00%). There were also six participants (12.00%) that described there not being a specific time when they were most receptive - depends on their emotional state and level of interest.
- In relation to sub-group variations, participants with low general health (46.43%) described being most receptive to information at diagnosis, more frequently than the general population (36.00%)

Health professional communication

- Participants were asked to describe the communication that they had had with health professionals throughout their experience. The most common theme was that participants described most healthcare professionals not knowing about mitochondrial disease (n=11, 22.00%). This was followed by participants being satisfied with health professional communication (n=10, 20.00%). The next most common themes were participants describing excellent communication (n=7, 14.00%), having minimal communication with healthcare professionals (n=6, 12.00%) and mostly good experiences, however there is a general lack of understanding of mitochondrial disease (n=6, 12.00%).
- In relation to sub-group variations, participants from low socio-economic areas (34.78%) described being satisfied with health professional communication more frequently than the general population (20.00%). Participants with high physical function (9.09%) and high general health (4.55%) described most healthcare professionals not knowing about mitochondrial disease less frequently than the general population (22.00%) while those with low physical functioning (32.14%) and low general health (35.71%) described this more frequently. Participants with high social functioning (25.00%) described excellent communication with their specialists more frequently than the general population (14.00%).

Knowledge and confidence

- 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 sub scales; knowledge, coping, recognition and management of symptoms, and adherence to treatment. A higher score denotes a better understanding and knowledge of disease.

Partners in health – overall score

- Overall, the participants scored in the top quintile for adherence to treatment indicating very good adherence to treatment. The scores for knowledge, recognition and management of symptoms, and total score were in the second highest quintile indicating good understanding and knowledge of disease. The score for coping was in the middle of the range of scores for this scale.

Partners in health - by general health

- Participants with higher general health had a statistically significant, better outcome for the coping subscale compared those with lower general health.

Partners in health – by physical functioning

- Participants with higher physical functioning had a statistically significant, better outcomes for the coping, adherence to treatment, and total score compared those with lower physical functioning.

Partners in health – by emotional well-being

- Participants with higher emotional well-being had a statistically significant, better outcomes for the coping, adherence to treatment, and total score compared those with lower emotional well-being.

Partners in health – by social functioning

- Participants with higher social functioning had a statistically significant, better outcomes for the coping, and total score compared those with social functioning.

Partners in health – by hearing problems

- No differences were observed between those with no hearing problems and those with hearing problems for any PIH subscale.

Partners in health – by eye problems

- Participants with no eye problems had significantly higher scores for the PIH knowledge, adherence to treatment and total score compared to those with eye problems.

Partners in health – by location

- Participants living in regional or rural areas had had a statistically significant, worse outcomes for the total score subscales compared those living in metropolitan areas.

Partners in health – by education

- No differences were observed between those with university education and those with high school or trade qualifications for any PIH subscale.

Partners in health – by SEIFA

- No differences were observed between those that lived in a higher SEIFA area compared to those that lived in an area with lower SEIFA scores for any PIH subscale.

Information given by health care professionals

- Participants were asked about what type of information they were given by healthcare professionals and what type of information they searched for independently:
- Information about disease cause (50.00%), treatment options (38.00%), and disease management (38.00%) were most frequently given to participants by healthcare professionals.
- Information about clinical trials (14.00%), interpreting test results (14.00%) and complementary therapies (16.00%) were give least often.
- Eight participants (16.00%) indicated that they received no information at all from health professionals about mitochondrial disease.

Information searched for independently

- Participants were asked about what type of information they searched for after receiving information from healthcare professionals:
- Information about treatment options (63.27%), disease management (59.18%), and disease cause (57.14%) were most frequently given to searched for independently.
- Information about interpreting test results (28.57%), hereditary, genes and biomarkers (28.57%) and psychological support (30.61%) were give least often.

Gaps in Information obtained

- The largest gaps in information, where information was neither given to patients nor searched for independently were how to interpret test results (62.00%), and psychological/social support (56.00%).
- Participants were given most information either from healthcare professionals or independently for treatment options (78.00%) and disease cause (78.00%).
- Clinical trials (42.00%) was the topic that was most searched for independently following no information from health professionals.

Most trusted information sources

- Across all participants, information from the participants' hospital or clinic and from the non-profit or charitable organisations was near equal and was most trusted. Information from pharmaceutical companies was least trusted. This order of preference was the same for all sub-groups.

Access to information

Participants were asked what information they had accessed in relation to their condition. The most common response from over half of all participants was accessing information from the Australian Mitochondrial Disease Foundation (n=32, 64.00%). The next most common theme was accessing information via the internet (n=25, 50.00%).

Participant describes accessing information from the Australian Mitochondrial Disease Foundation

Most of it's been on the AMDF website or phoning them. They've been very beneficial to me. Participant 6

Well, so once again, the foundation website is amazing. Basically I've used that foundation website, and then I've breadcrumbed. Participant 7

The AMDF has been very good. They've produce booklets, and of course there's what's online as well. Participant 34

The Australian Mitochondrial Disease Foundation actually sponsored me to go to an information day where I learned quite a bit, and spoke to other people that had similar or worse situations, and children with the disease, and became more aware of all the different forms that it can take. Participant 36

…the AMDF have had a lot more information on their website. They do an information session once a year and they publish a booklet that you can give to your GP. That's quite informative. Probably the Mitochondrial Disease Foundation provide the most information. Participant 43

Participant describes accessing information from the internet (general searching e.g. google)

So far as information that I've been able to get them myself is basically Dr. Google who's been the other source. It makes a severely dire reading. Participant 2

Internet's wonderful, just Mr. Google. Participant 4

Well, just most of it's been through the Internet. I'd say just about all of it's been through the Internet really. Participant 13

What I read on Google and the books they were sending out to me when I was first diagnosed, doing a lot of researching mitochondria. Participant 17

Really, what I can find on Google. Participant 18

I would say, most of it, we've actually done ourselves through Google. Participant 50

There were 14 participants (28.00%) that described accessing information from medical journals and peer reviewed papers and 13 participants (26.00%) that described accessing information from online forums including Facebook.

Participant describes accessing medical journals, peer reviewed papers

I have access to a lot of information. I've done a lot of research online looking at various published journal articles, looking at resources from the NPS website. Participant 8

If something interesting pops up or something new pops up or every now and then, I'll just look through the databases about some new research that's going on with mitochondrial disease. I have like quite a lot of information. Participant 11

Written public scientific publications. Anyone who's done things in that area, I have tried to keep up to date with…I also get articles from Pubmed and a few other places out to me most days. It's mainly literature and speakers in the area. Participant 27

Medical journals, medical textbooks, internet – I look for high quality published materials. Participant 30

I tried through Elsevier that you can get. I had the Lancet coming as email every week. Participant 32

So it's quite a information out then there's a number research papers on mitochondria and you can get in and do research on mitochondrial disease and other things and there's a lot of research papers you can read up on. Participant 42

Participant describes accessing information from online forums for mitochondrial disease (including Facebook)

…one of the most useful things is the Facebook groups with people who are -- There's one called Mito Café and just one for adults with Mitochondrial disease. There's a lot of people on that one, but often you'll post about something or ask a question and people will have their own experience that they can contribute. Participant 5

There are online forums which I've taken part of and also try and speak to other people who have been affected by this. Participant 8

I think the most information I've ever been able to find has come through a lady that I found out about on the internet via Facebook. Participant 28

In relation to sub-group variations, participants from rural areas (75.00%), participants with a high school or trade education (76.92), participants with low physical functioning (75.00%) and low general health (75.00%) reported accessing information from the Australian Mitochondrial Disease Foundation more frequently than the general population (64.00%), while participants with a university education (50.00%) and high physical functioning reported this less frequently. Participants from rural areas (65.00%) and those with high physical function (68.18%) reported accessing information from the internet more frequently than the general population (50.00%), while those from low socio-economic areas (37.04%) and those with low physical functioning (35.71%) reported this less frequently. Participants from rural areas (15.00%) reported accessing medical journals less frequently than the general population (28.00%).

Table 6.1: Access to information

Information accessed	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant describes accessing information from the Australian Mitochondrial Disease Foundation	**32**	64.00	17	56.67	15	**75.00**	16	59.26	16	69.57
Participant describes accessing information from the internet (general searching e.g. google)	**25**	50.00	12	40.00	13	**65.00**	10	**37.04**	15	65.22
Participant describes accessing medical journals, peer reviewed papers	**14**	28.00	11	36.67	3	**15.00**	9	33.33	5	21.74
Participant describes accessing information from their clinician	**13**	26.00	9	30.00	4	20.00	9	33.33	4	17.39
Participant describes accessing information from online forums for mitochondrial disease (including Facebook)	**7**	14.00	4	13.33	3	15.00	1	**3.70**	6	26.09
Participant describes accessing information from mitochondrial foundations websites	**6**	12.00	4	13.33	2	10.00	3	11.11	3	13.04

Information accessed	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant describes accessing information from the Australian Mitochondrial Disease Foundation	**32**	64.00	20	**76.92**	12	**50.00**	16	66.67	24	70.59
Participant describes accessing information from the internet (general searching e.g. google)	**25**	50.00	14	53.85	11	45.83	13	54.17	20	58.82
Participant describes accessing medical journals, peer reviewed papers	**14**	28.00	6	23.08	8	33.33	6	25.00	7	20.59
Participant describes accessing information from their clinician	**13**	26.00	6	23.08	7	29.17	6	25.00	7	20.59
Participant describes accessing information from online forums for mitochondrial disease (including Facebook)	**7**	14.00	3	11.54	4	16.67	3	12.50	6	17.65
Participant describes accessing information from mitochondrial foundations websites	**6**	12.00	3	11.54	3	12.50	2	8.33	5	14.71

Information accessed	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant describes accessing information from the Australian Mitochondrial Disease Foundation	**32**	64.00	11	**50.00**	21	**75.00**	14	53.85	18	**75.00**
Participant describes accessing information from the internet (general searching e.g. google)	**25**	50.00	15	**68.18**	10	**35.71**	12	46.15	13	54.17
Participant describes accessing medical journals, peer reviewed papers	**14**	28.00	4	18.18	10	35.71	6	23.08	8	33.33
Participant describes accessing information from their clinician	**13**	26.00	5	22.73	8	28.57	8	30.77	5	20.83
Participant describes accessing information from online forums for mitochondrial disease (including Facebook)	**7**	14.00	5	22.73	2	7.14	4	15.38	3	12.50
Participant describes accessing information from mitochondrial foundations websites	**6**	12.00	2	9.09	4	14.29	2	7.69	4	16.67

Information accessed	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant describes accessing information from the Australian Mitochondrial Disease Foundation	**32**	64.00	12	60.00	20	66.67	11	50.00	21	**75.00**
Participant describes accessing information from the internet (general searching e.g. google)	**25**	50.00	11	55.00	14	46.67	13	59.09	12	42.86
Participant describes accessing medical journals, peer reviewed papers	**14**	28.00	4	20.00	10	33.33	4	18.18	10	35.71
Participant describes accessing information from their clinician	**13**	26.00	6	30.00	7	23.33	7	31.82	6	21.43
Participant describes accessing information from online forums for mitochondrial disease (including Facebook)	**7**	14.00	4	20.00	3	10.00	3	13.64	4	14.29
Participant describes accessing information from mitochondrial foundations websites	**6**	12.00	4	20.00	2	6.67	1	4.55	5	17.86

Information accessed	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant describes accessing information from the Australian Mitochondrial Disease Foundation	**32**	64.00	5	83.33	5	35.71	7	77.78	6	54.55	9	90.00
Participant describes accessing information from the internet (general searching e.g. google)	**25**	50.00	1	16.67	8	57.14	5	55.56	8	72.73	3	30.00
Participant describes accessing medical journals, peer reviewed papers	**14**	28.00	2	33.33	6	42.86	1	11.11	3	27.27	2	20.00
Participant describes accessing information from their clinician	**13**	26.00	2	33.33	4	28.57	2	22.22	2	18.18	3	30.00
Participant describes accessing information from online forums for mitochondrial disease (including Facebook)	**7**	14.00	0	0.00	5	35.71	1	11.11	1	9.09	0	0.00
Participant describes accessing information from mitochondrial foundations websites	**6**	12.00	0	0.00	1	7.14	2	22.22	1	9.09	2	20.00

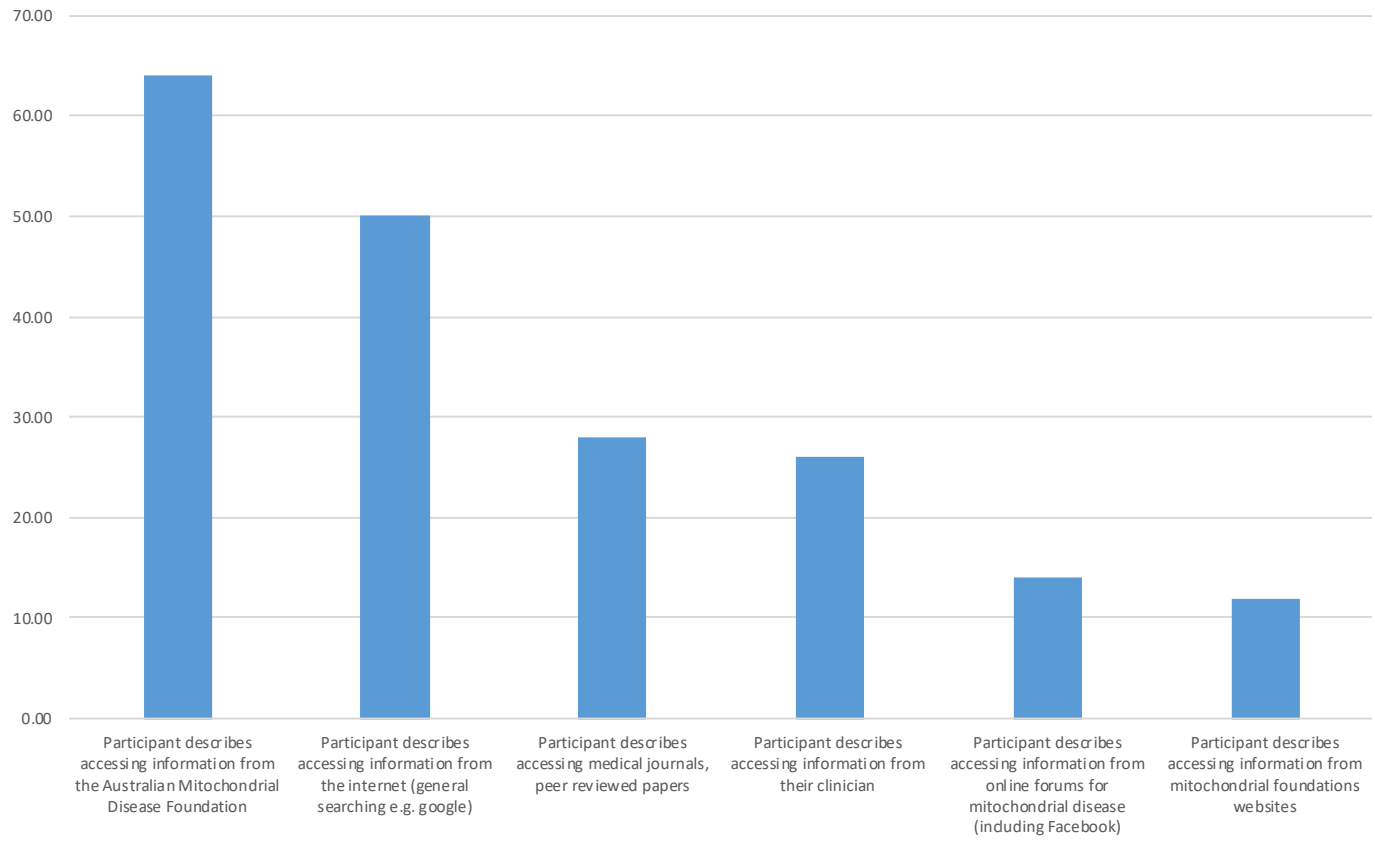


Figure 6.1: Access to information (% of all participants)

Information that was helpful

After talking about all of the information that participants had received, they were then asked what information had been helpful. There was a range of information that participants found particularly helpful including information from the AMDF (n=9, 18.00%) research papers (n=7, 14.00%), communicating with others with mitochondrial disease (n=7, 14.00%) and information from clinical teams (n=5, 10.00%).

Participant describes the AMDF as being helpful

The AMDF is really great for information. Participant 10

I have to say the AMDF focus has been possibly the most useful document, not just for me. I've been able to give that to my GP who herself, she didn't know what ALS was until I came along. I don't know whether she's read it or not. [Interviewer: you've given it to her?] I've given it to her and various other physicians that I was dealing with or friends, my mother. That was a really useful, eye-opening document for me. Also for me, being able to tell other people or show other people or let them know what it was. Participant 20

One lady, NAME from the AMDF was exceptionally useful. She was really helpful on a personal level. I had contacted her a couple times which was really, really good. Participant 26

Definitely, the mitochondrial disease foundation . Participant 43

Participant describes no information being specifically helpful

There's no information that's really helpful. A lot of it's interesting. There's nothing helpful. Participant 21

It's hard for me because I've already studied that. It's the same stuff as what I've already studied. I find it a bit repetitive also when you go through one website and then you go through another. Even one in America or something, they're all sort of similar in one sense or another. That's what I find. Participant 22

Well, there's none. None has been helpful at all really because it'd be the way you read if you would know if you've had anything to do with it. It's always CoQ_{10} is always mentioned which I have taken since 2000. None. Nothing. No. Participant 31

Participant describes current research papers as being helpful

There's a fantastic published article which talks about the different variations of LHON and percentage of prevalence between males and females, percentages of affectation within people, and also percentage of recovery because some types actually lead towards some recovery. It's a little bit tricky. I tend to look for different types of research at different times so it's hard to say which one is the best. Participant 8

I guess information that has some science base to it, some information where there is some-- they make it clear how many people they have looked at to make this out in their conclusions or something. Because often, you'll read an article and find out it was only one person, there were two people that we're talking about, just the sample size really isn't big enough to be sure that's right. Participant 13

Probably the written published articles as well if there's a specialist who has something that said something that's new to me then I would certainly listen to them and then go and try and research it. Participant 27

I don't know. I'd have to say more in the trials, new research, that kind of stuff probably. Participant 45

Participant describes communicating with others with mitochondrial disease as helpful

There's Facebook. It's very good because people will share resources and experiences. And there are international Facebook pages and that's better than Australia. Australia is just so far behind everybody else from what I can see. In terms of trials and things like that and the mitochondrial association networks. Those ones are probably the main ones and my own in the...When people put mitochondrial news bulletin in published recent studies and then I follow the studies and take it from there. Participant 3

Just individual people's stories is quite reassure-- It's very upsetting, but it can be reassuring too that you think, "Well, I'm not mad." This is what's been happening to me", the sort of thing. Participant 34

The most helpful one was actually finding that group, which my doctor absolutely had no idea. He didn't even know ... He knows what mitochondrial is, but very vague and basic to the point that he even prescribed me something that I went, "Hey, is that from this or that group of drug?" Participant 40

Participant describes information form clinical team as being helpful

I think, first off, when I got it, I actually talked to a GP that specialised in it just to have an idea about maybe some of the things you can do.... One of the things, you know, you see other, what do you call it, I call it peptides, but probably other, you know, you think, oh, should I take it? I wouldn't take it without...well, without a specialist, ... Telling me, "Yeah, that's okay.",or "That's not okay.", as far as you don't know, whether it's, you know, bogus information or not. Participant 15

Probably the written published articles as well if there's a specialist who has something that said something that's new to me then I would certainly listen to them and then go and try and research it. Participant 27

Receiving the information, what was the most helpful? I suppose some of the things that the neurologist just said to us where she has actually probably had some clients that she has seen that are in their teens and early 20s. Well, I guess helpful probably would be more that it's being more easing on our minds, that's positive information from her that she's had clients that are older than NAME, in their late teens and doing really well and they've continued to thrive regardless of having Leigh's disease. Participant 50

Table 6.2: Information that was helpful

Information that has been helpful	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant describes the AMDF as being helpful	9	18.00	5	16.67	4	20.00	4	14.81	5	21.74
Participant describes no information being specifically helpful	8	16.00	4	13.33	4	20.00	3	11.11	5	21.74
Participant describes current research papers as being helpful	7	14.00	5	16.67	2	10.00	5	18.52	2	8.70
Participant describes communicating with others with mitochondrial disease as most useful	7	14.00	4	13.33	3	15.00	3	11.11	4	17.39
Participant describes information form clinical team as being helpful	5	10.00	4	13.33	1	5.00	4	14.81	1	4.35

Information that has been helpful	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant describes the AMDF as being helpful	9	18.00	3	11.54	6	25.00	5	20.83	7	20.59
Participant describes no information being specifically helpful	8	16.00	6	23.08	2	8.33	4	16.67	5	14.71
Participant describes current research papers as being helpful	7	14.00	3	11.54	4	16.67	2	8.33	5	14.71
Participant describes communicating with others with mitochondrial disease as most useful	7	14.00	4	15.38	3	12.50	2	8.33	5	14.71
Participant describes information form clinical team as being helpful	5	10.00	2	7.69	3	12.50	2	8.33	2	5.88

Information that has been helpful	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant describes the AMDF as being helpful	9	18.00	5	22.73	4	14.29	5	19.23	4	16.67
Participant describes no information being specifically helpful	8	16.00	4	18.18	4	14.29	3	11.54	5	20.83
Participant describes current research papers as being helpful	7	14.00	3	13.64	4	14.29	6	23.08	1	4.17
Participant describes communicating with others with mitochondrial disease as most useful	7	14.00	3	13.64	4	14.29	4	15.38	3	12.50
Participant describes information form clinical team as being helpful	5	10.00	4	18.18	1	3.57	3	11.54	2	8.33

Information that has been helpful	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant describes the AMDF as being helpful	9	18.00	5	25.00	4	13.33	4	18.18	5	17.86
Participant describes no information being specifically helpful	8	16.00	3	15.00	5	16.67	4	18.18	4	14.29
Participant describes current research papers as being helpful	7	14.00	3	15.00	4	13.33	3	13.64	4	14.29
Participant describes communicating with others with mitochondrial disease as most useful	7	14.00	3	15.00	4	13.33	2	9.09	5	17.86
Participant describes information form clinical team as being helpful	5	10.00	3	15.00	2	6.67	4	18.18	1	3.57

Information that has been helpful	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant describes the AMDF as being helpful	9	18.00	0	0.00	5	35.71	1	11.11	2	18.18	1	10.00
Participant describes no information being specifically helpful	8	16.00	1	16.67	1	7.14	2	22.22	2	18.18	2	20.00
Participant describes current research papers as being helpful	7	14.00	1	16.67	3	21.43	2	22.22	1	9.09	0	0.00
Participant describes communicating with others with mitochondrial disease as most useful	7	14.00	2	33.33	2	14.29	1	11.11	2	18.18	0	0.00
Participant describes information form clinical team as being helpful	5	10.00	1	16.67	0	0.00	1	11.11	1	9.09	2	20.00

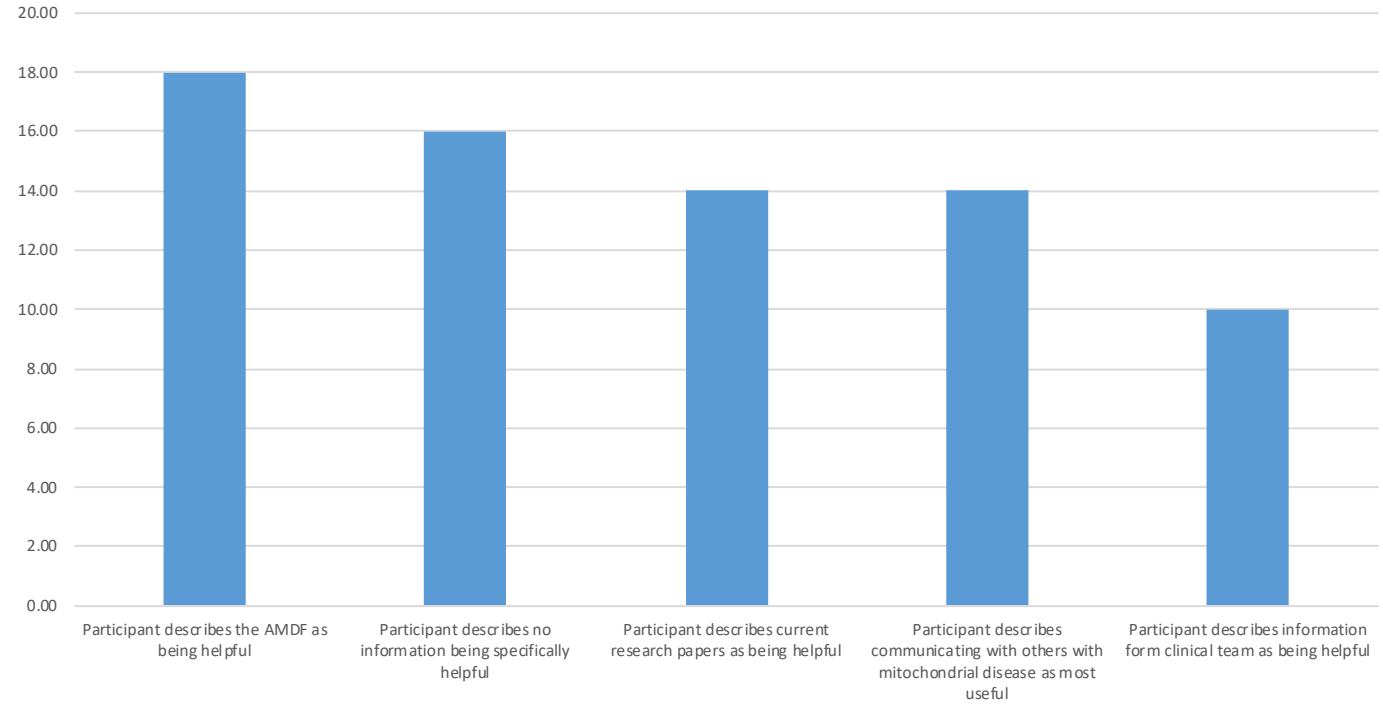


Figure 6.2: Information that was helpful

Information that was not helpful

Participants were asked whether there was any information they had come across that was not helpful. The most common theme described by 22 participant s(44.00%) was that no information was unhelpful:

Participant describes no information as being unhelpful

Not really because I find that altogether it paints a picture. I think it would be quite useful to have it all in one spot if possible and I know that that's something that the foundation has been working towards…but it is also tricky because there are so many different types of mito. It's hard to have a definitive resource library on each. Participant 8

No, it's all relevant to what's going on and it's helpful in a way that it takes you up with what I'm doing now with HOSPITAL that came from AMDF. I wouldn't have gone down that track if I didn't have any of the literature or anything from the AMDF. Participant 35

No, I haven't come across anything that's a bit weird or whacky or anything like that. Participant 42

There were no other themes noted by more than five participants, however where participants made a comment about information that was not helpful, this included stories about other patients (n=3, 6.00%), lack of concise yet comprehensive information (n=3, 6.00%), and information that is too general (n=2, 4.00%) or too scientific (n=2, 4.00%).

Table 6.3: Information that was not helpful

Information that has not been helpful	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant describes no information as being unhelpful	22	44.00	13	43.33	9	45.00	10	37.04	12	52.17
Participant describes the stories about other patients as unhelpful	3	6.00	1	3.33	2	10.00	2	7.41	1	4.35
Participant describes the lack of concise and comprehensive information as unhelpful	3	6.00	1	3.33	2	10.00	1	3.70	2	8.70
Participant describes not knowing if information is helpful or unhelpful	2	4.00	2	6.67	0	0.00	2	7.41	0	0.00
Participant describes information that is too general (not specific to their type of disease) as unhelpful	2	4.00	1	3.33	1	5.00	1	3.70	1	4.35
Participant describes information that is too scientific as unhelpful	2	4.00	0	0.00	2	10.00	1	3.70	1	4.35
Participant describes health professionals that do not take a holistic approach as unhelpful	2	4.00	2	6.67	0	0.00	1	3.70	1	4.35

Information that has not been helpful	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant describes no information as being unhelpful	22	44.00	14	53.85	8	33.33	10	41.67	16	47.06
Participant describes the stories about other patients as unhelpful	3	6.00	2	7.69	1	4.17	2	8.33	1	2.94
Participant describes the lack of concise and comprehensive information as unhelpful	3	6.00	0	0.00	3	12.50	2	8.33	3	8.82
Participant describes not knowing if information is helpful or unhelpful	2	4.00	2	7.69	0	0.00	1	4.17	2	5.88
Participant describes information that is too general (not specific to their type of disease) as unhelpful	2	4.00	0	0.00	2	8.33	2	8.33	0	0.00
Participant describes information that is too scientific as unhelpful	2	4.00	1	3.85	1	4.17	2	8.33	2	5.88
Participant describes health professionals that do not take a holistic approach as unhelpful	2	4.00	0	0.00	2	8.33	2	8.33	0	0.00

Information that has not been helpful	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant describes no information as being unhelpful	22	44.00	10	45.45	12	42.86	12	46.15	10	41.67
Participant describes the stories about other patients as unhelpful	3	6.00	1	4.55	2	7.14	3	11.54	0	0.00
Participant describes the lack of concise and comprehensive information as unhelpful	3	6.00	1	4.55	2	7.14	1	3.85	2	8.33
Participant describes not knowing if information is helpful or unhelpful	2	4.00	1	4.55	1	3.57	1	3.85	1	4.17
Participant describes information that is too general (not specific to their type of disease) as unhelpful	2	4.00	2	9.09	0	0.00	1	3.85	1	4.17
Participant describes information that is too scientific as unhelpful	2	4.00	1	4.55	1	3.57	1	3.85	1	4.17
Participant describes health professionals that do not take a holistic approach as unhelpful	2	4.00	0	0.00	2	7.14	1	3.85	1	4.17

Information that has not been helpful	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant describes no information as being unhelpful	22	44.00	8	40.00	14	46.67	10	45.45	12	42.86
Participant describes the stories about other patients as unhelpful	3	6.00	1	5.00	2	6.67	2	9.09	1	3.57
Participant describes the lack of concise and comprehensive information as unhelpful	3	6.00	2	10.00	1	3.33	1	4.55	2	7.14
Participant describes not knowing if information is helpful or unhelpful	2	4.00	1	5.00	1	3.33	1	4.55	1	3.57
Participant describes information that is too general (not specific to their type of disease) as unhelpful	2	4.00	0	0.00	2	6.67	1	4.55	1	3.57
Participant describes information that is too scientific as unhelpful	2	4.00	1	5.00	1	3.33	0	0.00	2	7.14
Participant describes health professionals that do not take a holistic approach as unhelpful	2	4.00	0	0.00	2	6.67	0	0.00	2	7.14

Information that has not been helpful	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant describes no information as being unhelpful	22	44.00	4	66.67	5	35.71	6	66.67	2	18.18	5	50.00
Participant describes the stories about other patients as unhelpful	3	6.00	1	16.67	0	0.00	0	0.00	1	9.09	1	10.00
Participant describes the lack of concise and comprehensive information as unhelpful	3	6.00	0	0.00	2	14.29	0	0.00	0	0.00	1	10.00
Participant describes not knowing if information is helpful or unhelpful	2	4.00	0	0.00	1	7.14	1	11.11	0	0.00	0	0.00
Participant describes information that is too general (not specific to their type of disease) as unhelpful	2	4.00	0	0.00	1	7.14	0	0.00	1	9.09	0	0.00
Participant describes information that is too scientific as unhelpful	2	4.00	0	0.00	0	0.00	0	0.00	2	18.18	0	0.00
Participant describes health professionals that do not take a holistic approach as unhelpful	2	4.00	0	0.00	2	14.29	0	0.00	0	0.00	0	0.00

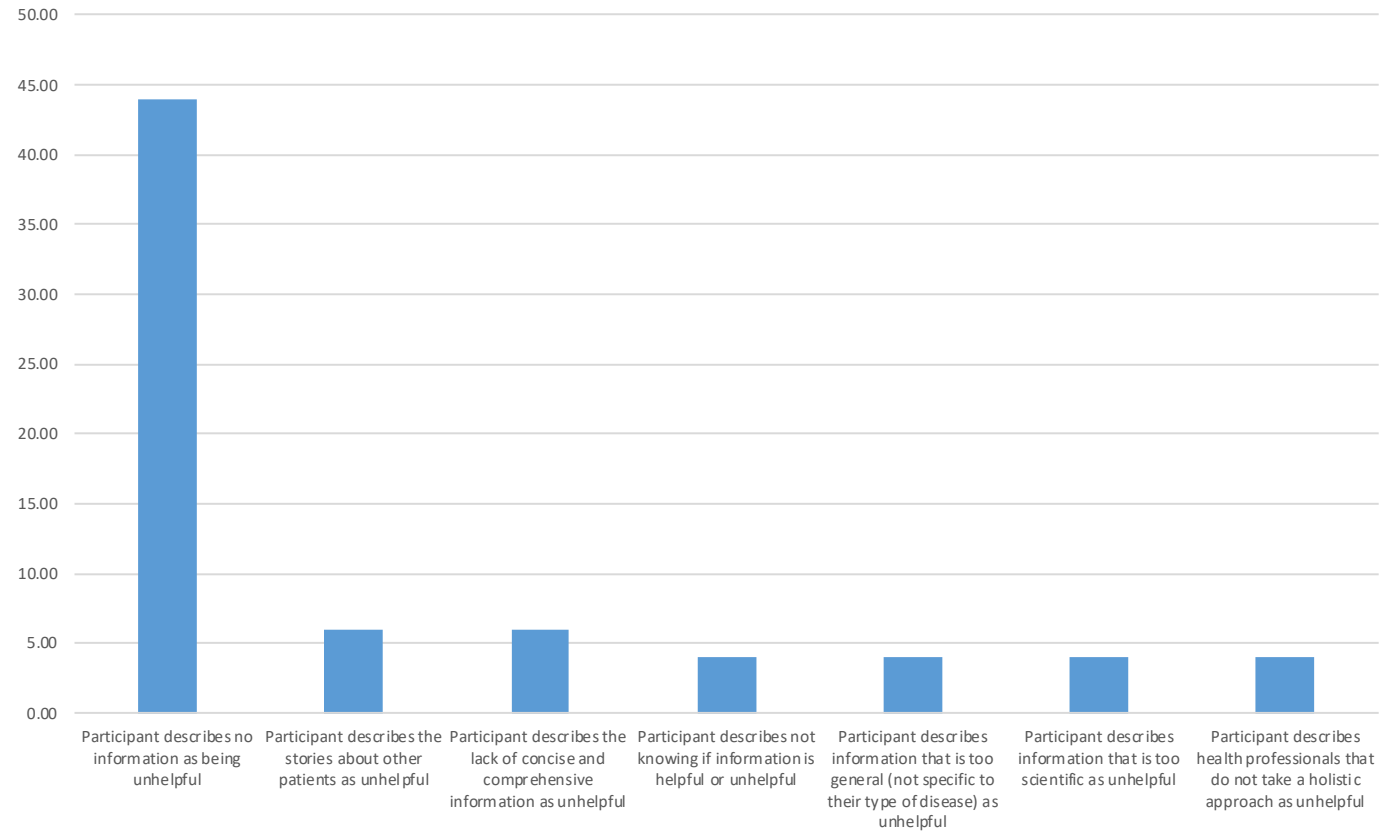


Figure 6.3: Information that was not helpful (% of all participants)

Information preferences (Format of information)

Participants were asked whether they had a preference for information online, talking to someone, in written (booklet) form or through a phone App. The most common theme was talking to someone (n=25, 50.00%) of which, five participants specified a preference for talking to someone face-to-face.

Participant describes preferring to talk to someone

I suppose my preference, it would be getting the information and then talking through it. Yes, that's sort of. Then, that's a good thing that my daughter comes to the appointment because she understands it a lot more, having a medical background. Then we can talk about through that. Participant 1

I tend to prefer, I'm fortunately not a millennial, but I tend to prefer to talk to someone because then generally the information that's given then become very specific to you rather then the online thing where it's just, sort of, it can become specific, but it's, what's the word, yeah, so I prefer to talk to someone about it rather because I figure rightly or wrongly you get a better understanding…Yes. Tell it to you rather than, sort of, this is general. Participant 15

Participant describes preferring to talk to someone (face-to-face)

I prefer to speak to someone face to face …Because I got cataracts in my eyes from my immune-suppressants hormone they've developed. I can't read too well. I'm using a magnifying glass at present [laughs]. Participant 6

Probably because of the way I learn, so I like to hear something, I like to have a discussion, and I like to read, so having something that's audio-visual and face-to-face would be my preferred option. Participant 7

Most of my questions, every time I go, it's quite a long consultation with Doctor NAME, she's quite thorough and everything, she just goes over anything that's happened to me with all my medical conditions. Participant 16

The next most common theme was a preference for information online (n=21, 42.00%) and a preference for information in a written format such as a booklet (n=7, 14.00%).

Participant describes preferring information online

I prefer it online because I like to be able to read it and digest it on my own time. I think one of the hard parts about having something like Mito is, the doctor will will speak at you, but you have no record, you can't go, what was it that they said and what was that word that they used again. Although I like people usually talking something through with me. I think reading it online is the most useful, then you can Google all the words if you don't know them or something. It just means you can digest it in your own time. Participant 5

I think with my experiences so far, online information has been more effective than talking to people. Mainly because a lot of people that I talk to don't really know how to help. Also, a lot of people that I talk to have dozens of other patients as well. I feel like sometimes they don't spend enough time with all their patients. Participant 11

I think online is the most accessible. You can always review it over and over again. If you have a conversation with someone, you sometimes miss some of that information or sometimes it's just too much for you. I think having it online is really useful. Participant 26

Participant describes preferring information in a written format like a booklet

Also, just sometimes it's a quickness and for each information can be a lot easier in terms of booklet from…No, sorry. I just thought I think know in terms of booklet, that one is quite handy. I like being able to hold onto something and can look at it but if I move quite a bit, it's a bit tricky to always access them or keep them. Participant 8

I like to try a more…it's just by an invitation in reading and thinking, I suppose. I was very pleased to get the booklets from AMDF. Participant 17

On paper is good, so I can keep it and think about it. Participant 24

In relation to sub-group variations, participants with a hearing impairment (29.12%) described a preference for online information less frequently than the general population (42.00%), while participants with low physical functioning (53.57%) and low general health (53.57%) reported this preference more frequently. Participants with a hearing impairment (25.00%) reported a preference for accessing written information more frequently than the general population (14.00%).

Table 6.4: Information preferences (Format)

Information preferences	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant describes preferring to talk to someone	20	40.00	12	40.00	8	40.00	12	44.44	8	34.78
Participant describes preferring to talk to someone (face-to-face)	5	10.00	2	6.67	3	15.00	2	7.41	3	13.04
Participant describes preferring information online	21	42.00	13	43.33	9	45.00	10	37.04	12	52.17
Participant describes preferring information in a written format like a booklet	7	14.00	5	16.67	2	10.00	4	14.81	3	13.04
Participant describes not having a preferred information format and/or various modes are acceptable	6	12.00	5	16.67	1	5.00	6	22.22	0	0.00
Participant describes preferring information from their specialist	5	10.00	4	13.33	1	5.00	2	7.41	3	13.04

Information preferences	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant describes preferring to talk to someone	20	40.00	11	42.31	9	37.50	13	54.17	11	32.35
Participant describes preferring to talk to someone (face-to-face)	5	10.00	2	7.69	3	12.50	2	8.33	3	8.82
Participant describes preferring information online	21	42.00	12	46.15	10	41.67	7	29.17	16	47.06
Participant describes preferring information in a written format like a booklet	7	14.00	2	7.69	5	20.83	6	25.00	3	8.82
Participant describes not having a preferred information format and/or various modes are acceptable	6	12.00	4	15.38	2	8.33	4	16.67	3	8.82
Participant describes preferring information from their specialist	5	10.00	4	15.38	1	4.17	1	4.17	5	14.71

Information preferences	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant describes preferring to talk to someone	20	40.00	11	50.00	9	32.14	12	46.15	8	33.33
Participant describes preferring to talk to someone (face-to-face)	5	10.00	2	9.09	3	10.71	2	7.69	3	12.50
Participant describes preferring information online	21	42.00	7	31.82	15	53.57	12	46.15	10	41.67
Participant describes preferring information in a written format like a booklet	7	14.00	3	13.64	4	14.29	5	19.23	2	8.33
Participant describes not having a preferred information format and/or various modes are acceptable	6	12.00	3	13.64	3	10.71	3	11.54	3	12.50
Participant describes preferring information from their specialist	5	10.00	2	9.09	3	10.71	3	11.54	2	8.33

Information preferences	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant describes preferring to talk to someone	20	40.00	6	30.00	14	46.67	10	45.45	10	35.71
Participant describes preferring to talk to someone (face-to-face)	5	10.00	2	10.00	3	10.00	3	13.64	2	7.14
Participant describes preferring information online	21	42.00	9	45.00	13	43.33	7	31.82	15	53.57
Participant describes preferring information in a written format like a booklet	7	14.00	2	10.00	5	16.67	5	22.73	2	7.14
Participant describes not having a preferred information format and/or various modes are acceptable	6	12.00	3	15.00	3	10.00	3	13.64	3	10.71
Participant describes preferring information from their specialist	5	10.00	2	10.00	3	10.00	2	9.09	3	10.71

Information preferences	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant describes preferring to talk to someone	**20**	40.00	2	33.33	5	35.71	4	44.44	4	36.36	5	50.00
Participant describes preferring to talk to someone (face-to-face)	**5**	10.00	0	0.00	0	0.00	2	22.22	1	9.09	2	20.00
Participant describes preferring information online	**21**	42.00	4	66.67	9	64.29	2	22.22	4	36.36	3	30.00
Participant describes preferring information in a written format like a booklet	**7**	14.00	1	16.67	2	14.29	0	0.00	2	18.18	2	20.00
Participant describes not having a preferred information format and/or various modes are acceptable	**6**	12.00	0	0.00	1	7.14	1	11.11	2	18.18	2	20.00
Participant describes preferring information from their specialist	**5**	10.00	1	16.67	1	7.14	1	11.11	1	9.09	1	10.00

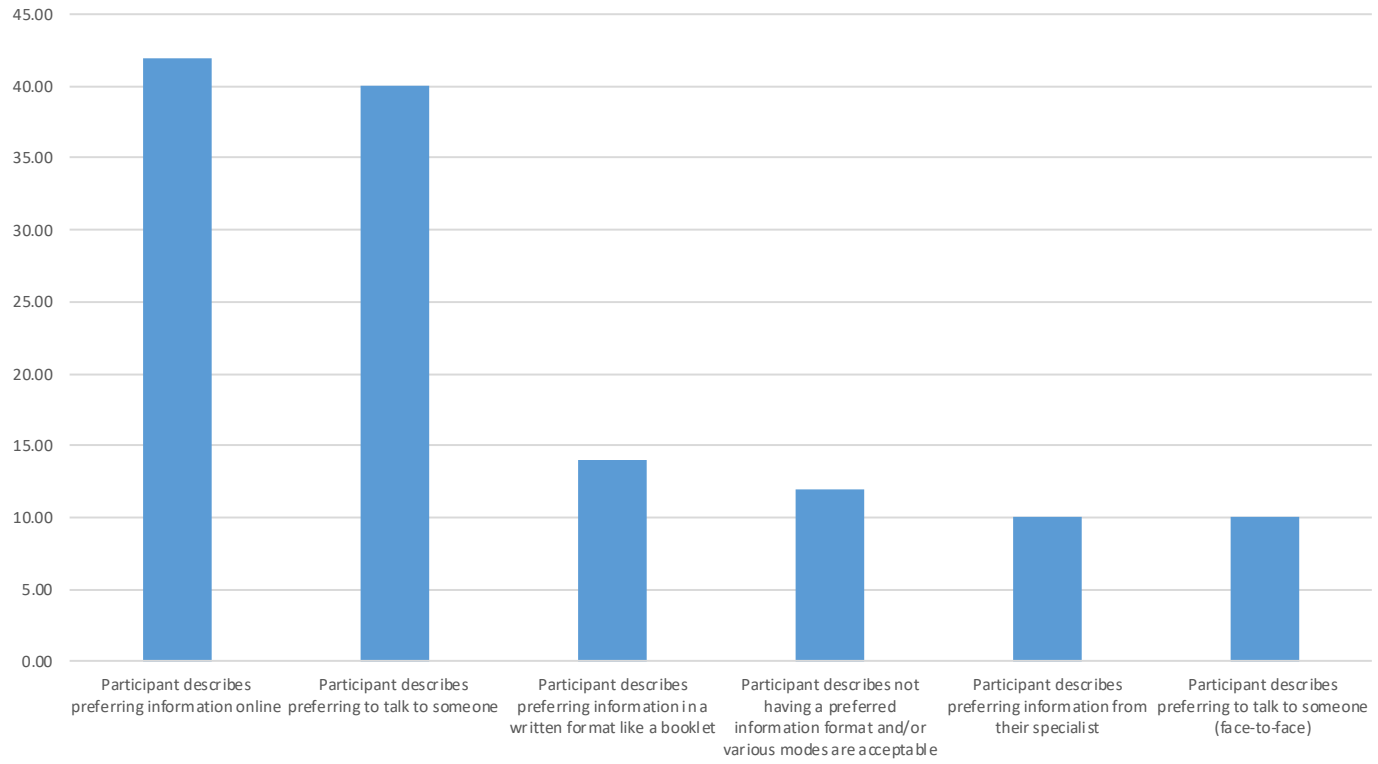


Figure 6.4: Information preferences – Format
(% of all participants)

Information preferences (Timing of information)

Participants were asked to reflect on their experience and think about when they were most receptive to receiving information, not when they actually received the information, but when they felt they could take it all in. The most common time that participants described being receptive to receiving information was at the time of diagnosis (n=18, 36.00%).

Participant describes being most receptive when they were first diagnosed (beginning)

I think the most, because I wanted to know, was at the very beginning when I had never heard of it and just wasn't being talked about. I wanted to be aware and informed and that's why I actually sorted out a lot of information so I could find it out for myself and help my family with that. Participant 8

Yes, at first because I knew about the mitochondrial. In humans, in all cases, I wanted to learn more about it and to understand it a bit more. I knew what's going on. It's like if you say to someone, "I've got cancer," they understand. They know what's going to happen. Where will I say, "Okay. We've got mitochondrial disease." "MELAS huh, what's that?" They still don't grasp it. I'd like to teach that person as much as I can as well to pass that information on. Participant 22

Yes. I think around the diagnosing process and around when we're doing the muscle biopsies and things like that. I was pretty keen on trying to understand what I could do and now I sort of got an understanding that I'm very limited in what I can do. Participant 49

The next most common theme was participants describing that there was not a specific time that they were most receptive and that it is an ongoing process (n=10, 20.00%).

Participant describes there not being a specific time when they were most receptive - an ongoing process

Not really. I think when you're…I think that's the time when you're like, "Tell me everything. I want to know. I want to know. I want to know", but you don't take it in. It's later on that you can have time to sit back and go through things a little bit more then it all starts to sink in. There's not really a time as such. It's on-going. Participant 10

That's overwhelming, anything after that doesn't matter. Is there a time that's most receptive to receiving information? No, I would say to people if you've had the tests and you're now going to get the results for potentially a diagnosis. Take someone with you that can take notes. Because your brain becomes paralysed. And you're overwhelmed with information that you don't understand, in most cases. And even though the doctor's trying to explain it to you, you kind of stop thinking. It's hard to explain. Yeah, your brain stops listening. The brain stops listening and it starts thinking about all sorts of possibility. So you don't absorb. So you need somebody else there with you. Well to take notes preferably, yeah to take notes. Participant 24

That's a hard one. Yeah, we always knew it wasn't going to be what we wanted to hear, so yeah. No, there would have never been a better time. Participant 46

There were also six participants (12.00%) that described there not being a specific time when they were most receptive - depends on their emotional state and level of interest.

Participant describes there not being a specific time when they were most receptive - depends on their emotional state and level of interest

Look, I think at any time, because I think if I receive the information, and I'm not up to looking at it, or reading it, or dealing with it, I'll just put it into a folder and come back to it later, so at any time, really. Participant 7

You really needed somebody to guide you then, and there wasn't anyone. You just have to do it, you can't drop your bundle. Participant 28

When will I be most receptive? Probably when I'm in a good mood. [laughs] Does that make sense, if I'm not depressed and feeling blue. Participant 42

In relation to sub-group variations, participants with low general health (46.43%) described being most receptive to information at diagnosis, more frequently than the general population (36.00%)

Table 6.5: Information preferences (Timing)

Timing of information	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant describes being most receptive when they were first diagnosed (beginning)	**18**	36.00	13	43.33	5	25.00	11	40.74	7	30.43
Participant describes there not being a specific time when they were most receptive - an ongoing process	**10**	20.00	5	16.67	5	25.00	7	25.93	3	13.04
Participant describes there not being a specific time when they were most receptive - depends on their emotional state and level of interest	**6**	12.00	3	10.00	3	15.00	2	7.41	4	17.39
Participant describes not being receptive during diagnosis but being more receptive post diagnosis	**3**	6.00	1	3.33	2	10.00	1	3.70	2	8.70
Participant describes being always receptive to receiving information	**2**	4.00	1	3.33	1	5.00	1	3.70	1	4.35
Participant describes being most receptive a year(s) after diagnosis	**2**	4.00	1	3.33	1	5.00	0	0.00	2	8.70
Participant describes being more receptive now once learning more about the disease, compared to the beginning	**2**	4.00	1	3.33	1	5.00	1	3.70	1	4.35

Timing of information	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant describes being most receptive when they were first diagnosed (beginning)	**18**	36.00	11	42.31	7	29.17	7	29.17	13	38.24
Participant describes there not being a specific time when they were most receptive - an ongoing process	**10**	20.00	6	23.08	4	16.67	5	20.83	7	20.59
Participant describes there not being a specific time when they were most receptive - depends on their emotional state and level of interest	**6**	12.00	2	7.69	4	16.67	5	20.83	5	14.71
Participant describes not being receptive during diagnosis but being more receptive post diagnosis	**3**	6.00	3	11.54	0	0.00	1	4.17	3	8.82
Participant describes being always receptive to receiving information	**2**	4.00	1	3.85	1	4.17	1	4.17	1	2.94
Participant describes being most receptive a year(s) after diagnosis	**2**	4.00	0	0.00	2	8.33	1	4.17	1	2.94
Participant describes being more receptive now once learning more about the disease, compared to the beginning	**2**	4.00	1	3.85	1	4.17	1	4.17	2	5.88

Timing of information	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant describes being most receptive when they were first diagnosed (beginning)	**18**	36.00	8	36.36	10	35.71	10	38.46	8	33.33
Participant describes there not being a specific time when they were most receptive - an ongoing process	**10**	20.00	3	13.64	7	25.00	7	26.92	3	12.50
Participant describes there not being a specific time when they were most receptive - depends on their emotional state and level of interest	**6**	12.00	3	13.64	3	10.71	3	11.54	3	12.50
Participant describes not being receptive during diagnosis but being more receptive post diagnosis	**3**	6.00	1	4.55	2	7.14	1	3.85	2	8.33
Participant describes being always receptive to receiving information	**2**	4.00	0	0.00	2	7.14	0	0.00	2	8.33
Participant describes being most receptive a year(s) after diagnosis	**2**	4.00	1	4.55	1	3.57	1	3.85	1	4.17
Participant describes being more receptive now once learning more about the disease, compared to the beginning	**2**	4.00	1	4.55	1	3.57	2	7.69	0	0.00

Timing of information	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant describes being most receptive when they were first diagnosed (beginning)	**18**	36.00	5	25.00	13	43.33	5	22.73	13	**46.43**
Participant describes there not being a specific time when they were most receptive - an ongoing process	**10**	20.00	6	30.00	4	13.33	5	22.73	5	17.86
Participant describes there not being a specific time when they were most receptive - depends on their emotional state and level of interest	**6**	12.00	3	15.00	3	10.00	3	13.64	3	10.71
Participant describes not being receptive during diagnosis but being more receptive post diagnosis	**3**	6.00	1	5.00	2	6.67	1	4.55	2	7.14
Participant describes being always receptive to receiving information	**2**	4.00	0	0.00	2	6.67	1	4.55	1	3.57
Participant describes being most receptive a year(s) after diagnosis	**2**	4.00	1	5.00	1	3.33	1	4.55	1	3.57
Participant describes being more receptive now once learning more about the disease, compared to the beginning	**2**	4.00	1	5.00	1	3.33	1	4.55	1	3.57

Timing of information	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant describes being most receptive when they were first diagnosed (beginning)	**18**	36.00	2	33.33	5	35.71	5	55.56	4	36.36	2	20.00
Participant describes there not being a specific time when they were most receptive - an ongoing process	**10**	20.00	2	33.33	2	14.29	0	0.00	3	27.27	3	30.00
Participant describes there not being a specific time when they were most receptive - depends on their emotional state and level of interest	**6**	12.00	0	0.00	3	21.43	1	11.11	1	9.09	1	10.00
Participant describes not being receptive during diagnosis but being more receptive post diagnosis	**3**	6.00	0	0.00	0	0.00	1	11.11	1	9.09	1	10.00
Participant describes being always receptive to receiving information	**2**	4.00	1	16.67	0	0.00	0	0.00	0	0.00	1	10.00
Participant describes being most receptive a year(s) after diagnosis	**2**	4.00	0	0.00	1	7.14	1	11.11	0	0.00	0	0.00
Participant describes being more receptive now once learning more about the disease, compared to the beginning	**2**	4.00	0	0.00	1	7.14	0	0.00	1	9.09	0	0.00

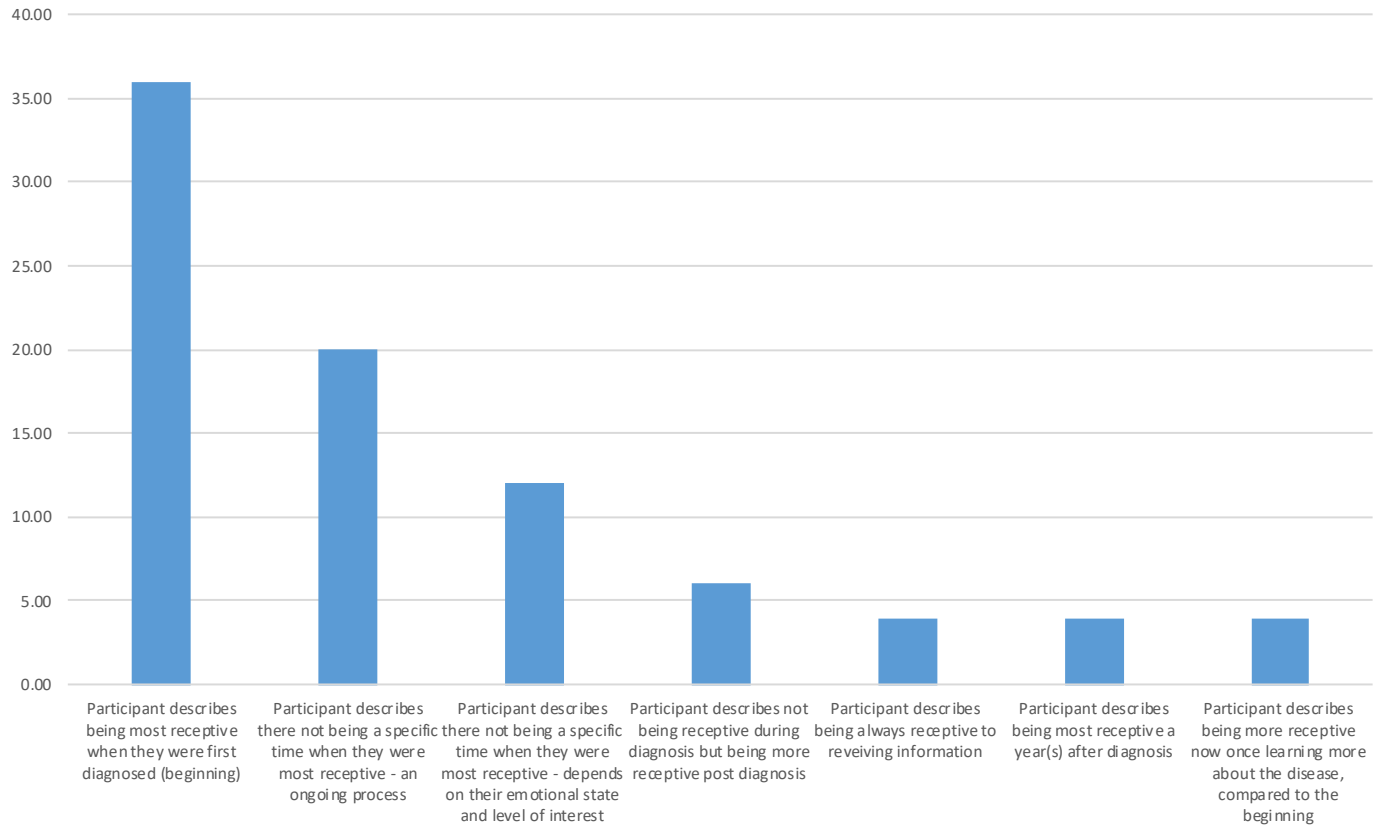


Figure 6.5: Information preferences – Timing
(% of all participants)

Communication with health professionals

Participants were asked to describe the communication that they had had with health professionals throughout their experience. The most common theme was that participants described most healthcare professionals not knowing about mitochondrial disease (n=11, 22.00%). This was followed by participants being satisfied with health professional communication (n=10, 20.00%).

Participant describes most healthcare professionals not knowing about mitochondrial disease

I'm always educating people wherever I go like the other day I had to go and see a urologist and they want to know about the Mitochondrial myopathy. Again, I just seem to forever educating people about it…No, it's not a common thing like the cold. Participant 16

My GP is useless. [laughs] Upon saying that, she's lovely. She knows nothing about it and has no interest even though she has a patient with it, knowing just in finding anything out. Even when I had to have the colonoscopy, they hadn't heard of it. None of these medical professionals that I've dealt with seems to have even heard of it. As I said, because I don't look like I'm ill, unless I'm having a really bad day and got a really bad link which does happen, I think it's all taken with a grain of salt by the medical profession. Participant 18

Difficult. I am usually the Mito educator, explaining the disease process to them and why certain treatments are not suitable or contraindicated. They will also not speak to each other, or read each other's notes, so I have to give a "potted history" of everything that has happened since I last saw them. Participant 30

Participant describes being satisfied with health professional communication

My GP is excellent. Of course, he's young. He's done courses or whatever you study. He's studied on it as well, so he's very good with it. I might go to him with a problem and he will say, "Okay. Maybe it's from the MELAS, maybe it's not, so we'll go and get it tested." Participant 22

It's been fine. They've all been on top on top of everything. Participant 35

I've mostly found my doctors to be to be very, informed and helpful. I would say there's not enough information, but I don't think that's the doctors' fault. Participant 43

The next most common themes were participants describing excellent communication (n=7, 14.00%), having minimal communication with healthcare professionals (n=6, 12.00%) and mostly good experiences, however there is a general lack of understanding of mitochondrial disease (n=6, 12.00%).

Participant describes excellent communication with their specialists

My GP is excellent. Of course, he's young. He's done courses or whatever you study. He's studied on it as well, so he's very good with it. I might go to him with a problem and he will say, "Okay. Maybe it's from the MELAS, maybe it's not, so we'll go and get it tested." Participant 22

Excellent. I'm very fortunate, not everybody has the same experience in this country or in others. I have a friend in Boston who's Facebooked me this morning. She's having major problems, and she can't get a doctor to tell her what's wrong. She has Mito, but she's also having these other problems where she falls, literally just drops. And they won't tell her. So yes, I'm very fortunate. Participant 24

So the metabolic specialist is brilliant. When you go outside of that into other areas of the hospital, you find that you inform them, more than the other way around, for sure. Participant 46

Participant describes having minimal communication with healthcare professionals

Virtually nil. Not many know or really…the hospital know nothing and now none of them know much about it. Participant 6

Pretty appalling. Look, I think it would be best described as non-existent. Because I don't think it's an appropriate response from a medical practitioner to say, "I don't know anything about it," and just basically leave it at that. Participant 7

Zero. Literally, zero. My doctor just looks at me like ... I think when I started having the B12 shots, and I started getting better, and he'd go, "Oh, that's great." But no questions, no "Oh, hang on a sec. Maybe the B12 has played a role here." Just basically push everything aside. "That's good. It's good. You sure you want to go back to work? Okay. That's good." … And I did ask my doctor, "What can I expect with this?" And he goes, "You'll probably lose your eyesight. Probably lose your hearing." That's what he told me on the phone, when I talked to him. And I literally went, "What the hell?" Is there something wrong with my brain? I'm going to lose my hearing. And I'd do something, I'm scared. Participant 40

In relation to sub-group variations, participants from low socio-economic areas (34.78%) described being satisfied with health professional communication more frequently than the general population (20.00%). Participants with high physical function (9.09%) and high general health (4.55%) described most healthcare professionals not knowing about mitochondrial disease less frequently than the general population (22.00%) while those with low physical functioning (32.14%) and low general health (35.71%) described this more frequently. Participants with high social functioning (25.00%) described excellent communication with their specialists more frequently than the general population (14.00%).

Table 6.6: Communication with health professionals

Health professional communication	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant describes most healthcare professionals not knowing about mitochondrial disease	11	22.00	5	16.67	6	30.00	5	18.52	6	26.09
Participant describes being satisfied with health professional communication	10	20.00	5	16.67	5	25.00	2	7.41	8	34.78
Participant describes excellent communication with their specialists	7	14.00	4	13.33	3	15.00	4	14.81	3	13.04
Participant describes having minimal communication with healthcare professionals	6	12.00	5	16.67	2	10.00	5	18.52	2	8.70
Participant describes mostly good experiences, however there is a general lack of understanding of mitochondrial disease	6	12.00	5	16.67	1	5.00	5	18.52	1	4.35
Participant describes a few poor experiences with general practitioners	4	8.00	3	10.00	1	5.00	3	11.11	1	4.35
Participant describes feeling as though time with specialists is too short (rushed)	4	8.00	3	10.00	1	5.00	3	11.11	1	4.35

Health professional communication	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant describes most healthcare professionals not knowing about mitochondrial disease	11	22.00	5	16.67	6	30.00	5	18.52	6	26.09
Participant describes being satisfied with health professional communication	10	20.00	5	16.67	5	25.00	2	7.41	8	34.78
Participant describes excellent communication with their specialists	7	14.00	4	13.33	3	15.00	4	14.81	3	13.04
Participant describes having minimal communication with healthcare professionals	6	12.00	5	16.67	2	10.00	5	18.52	2	8.70
Participant describes mostly good experiences, however there is a general lack of understanding of mitochondrial disease	6	12.00	5	16.67	1	5.00	5	18.52	1	4.35
Participant describes a few poor experiences with general practitioners	4	8.00	3	10.00	1	5.00	3	11.11	1	4.35
Participant describes feeling as though time with specialists is too short (rushed)	4	8.00	3	10.00	1	5.00	3	11.11	1	4.35

Health professional communication	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant describes most healthcare professionals not knowing about mitochondrial disease	11	22.00	2	9.09	9	32.14	5	19.23	6	25.00
Participant describes being satisfied with health professional communication	10	20.00	5	22.73	5	17.86	4	15.38	6	25.00
Participant describes excellent communication with their specialists	7	14.00	3	13.64	4	14.29	6	23.08	1	4.17
Participant describes having minimal communication with healthcare professionals	6	12.00	3	13.64	4	14.29	2	7.69	5	20.83
Participant describes mostly good experiences, however there is a general lack of understanding of mitochondrial disease	6	12.00	2	9.09	4	14.29	4	15.38	2	8.33
Participant describes a few poor experiences with general practitioners	4	8.00	3	13.64	1	3.57	4	15.38	0	0.00
Participant describes feeling as though time with specialists is too short (rushed)	4	8.00	3	13.64	1	3.57	2	7.69	2	8.33

Health professional communication	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant describes most healthcare professionals not knowing about mitochondrial disease	**11**	22.00	2	9.09	9	32.14	5	19.23	6	25.00
Participant describes being satisfied with health professional communication	**10**	20.00	5	22.73	5	17.86	4	15.38	6	25.00
Participant describes excellent communication with their specialists	**7**	14.00	3	13.64	4	14.29	6	23.08	1	4.17
Participant describes having minimal communication with healthcare professionals	**6**	12.00	3	13.64	4	14.29	2	7.69	5	20.83
Participant describes mostly good experiences, however there is a general lack of understanding of mitochondrial disease	**6**	12.00	2	9.09	4	14.29	4	15.38	2	8.33
Participant describes a few poor experiences with general practitioners	**4**	8.00	3	13.64	1	3.57	4	15.38	0	0.00
Participant describes feeling as though time with specialists is too short (rushed)	**4**	8.00	3	13.64	1	3.57	2	7.69	2	8.33

Health professional communication	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant describes most healthcare professionals not knowing about mitochondrial disease	**11**	22.00	1	16.67	3	21.43	1	11.11	2	18.18	4	40.00
Participant describes being satisfied with health professional communication	**10**	20.00	0	0.00	3	21.43	3	33.33	2	18.18	2	20.00
Participant describes excellent communication with their specialists	**7**	14.00	1	16.67	1	7.14	1	11.11	1	9.09	3	30.00
Participant describes having minimal communication with healthcare professionals	**6**	12.00	1	16.67	1	7.14	3	33.33	2	18.18	0	0.00
Participant describes mostly good experiences, however there is a general lack of understanding of mitochondrial disease	**6**	12.00	1	16.67	3	21.43	1	11.11	0	0.00	1	10.00
Participant describes a few poor experiences with general practitioners	**4**	8.00	0	0.00	2	14.29	0	0.00	2	18.18	0	0.00
Participant describes feeling as though time with specialists is too short (rushed)	**4**	8.00	0	0.00	1	7.14	0	0.00	2	18.18	1	10.00

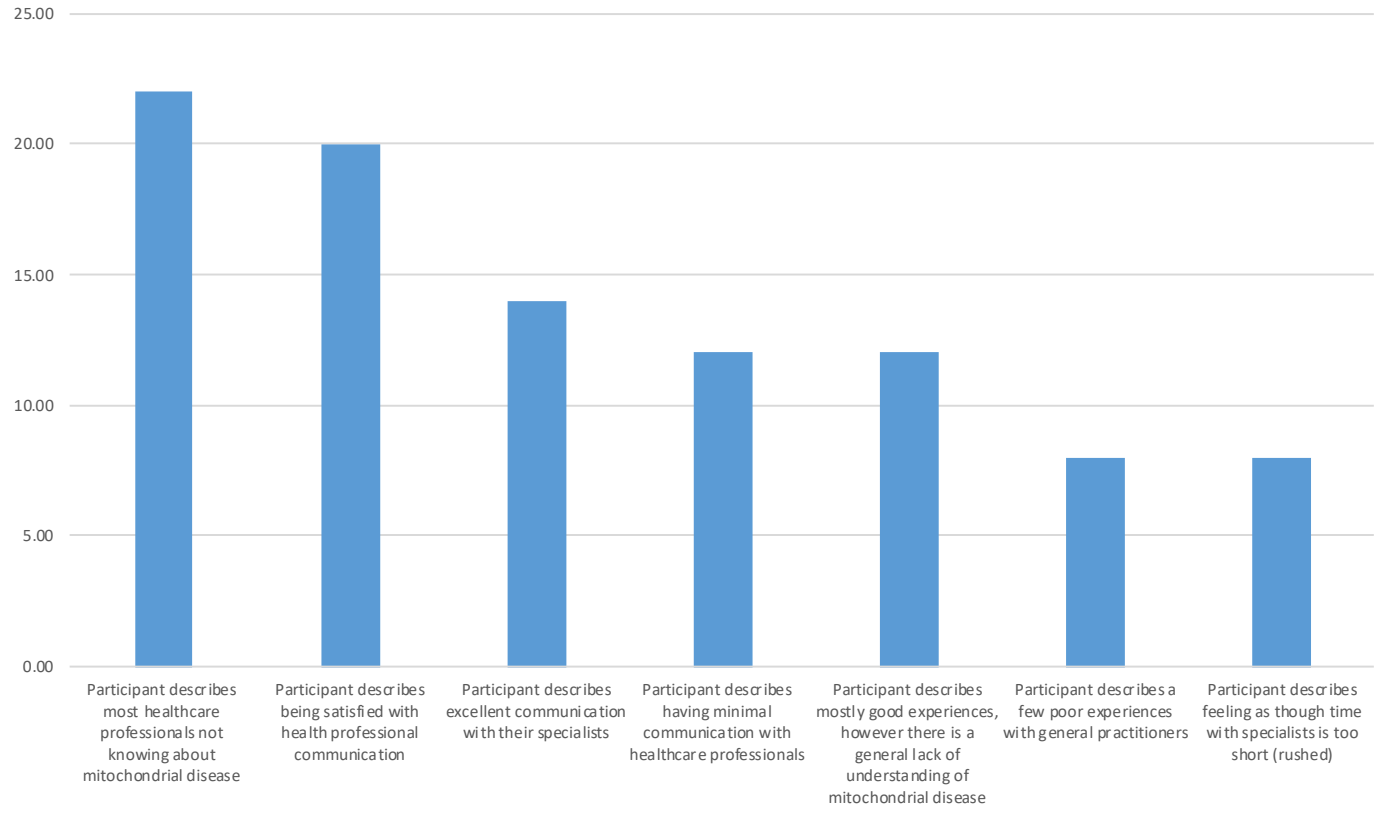


Figure 6.6: Communication with health professionals (% of all participants)

Knowledge and confidence

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 sub scales; knowledge, coping, recognition and treatment of symptoms, adherence to treatment and total score. A higher score denotes a better understanding and knowledge of disease. Summary statistics for the entire cohort are displayed alongside the possible range of each scale in Table 6.7. Overall, the participants scored in the top quintile for adherence to treatment (Median=14.00, IQR = 1.00) indicating very good adherence to treatment. The scores for knowledge (Median = 24.00, IQR = 3.00), recognition and management of symptoms (Mean = 18.76, SD = 2.89) and total score (Median=71.50, IQR = 12.75) were in the second highest quintile indicating good outcomes. The score for coping (Mean = 13.40, SD = 4.73),was in the middle of the range of scores for this scale.

Box plots display each of the Partners in Health sub-scales by general health, physical functioning, emotional well-being, social functioning, hearing problems status, eye problem status, location, education and SEIFA (Figures 6.7 – 6.51).

Comparisons of PIH global and sub scales have been made based on general health (Figures 6.7 to 6.11, Tables 6.8 to 6.9), physical functioning (Figures 6.12 to 6.16, Tables 6.10 to 6.11), emotional well-being (Figures 6.17 to 6.21, Tables 6.12 to 6.13), social functioning (Figures 6.22 to 6.26, Table 6.14 to 6.15), hearing problem status (Figures 6.27 to 6.31, Tables 6.16 to 6.17), eye problem status (Figures 6.32 to 6.36, Tables 6.18 to 6.19), location (Figures 6.37 to 6.41, Tables 6.20 to 6.21), education status (Figures 6.42 to 6.46, Tables 6.22 to 6.23), and SEIFA, (Figures 6.47 to 6.51, Tables 6.24 to 6.25).

Table 6.7: Summary statistics all participants Partners in Health

	Mean	SD	Median	IQR	Possible range
Knowledge	23.32	6.05	24.00	3.00	0-32
Coping*	13.40	4.73	13.00	3.00	0-24
Recognition and management of symptoms*	18.76	2.89	19.00	1.75	0-24
Adherence to treatment	13.18	3.26	14.00	1.00	0-16
Total score	68.66	12.75	71.50	4.25	0-96

*** Normal distribution use Mean and SD**

Comparisons of PIH sub scales by general health

Comparisons of PIH subscales were made general health, those that had a SF36 general health score above average for the group (Higher general health) were compared with those that had an average or lower score (Lower general health). Summary statistics are listed in Tables 6.11 and 6.12.

Two sample t-test was used when assumptions for normality and variance were met (Table 6.16). When assumptions normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 6.17).

A two sample t-test indicated no significant difference in the recognition and management of symptoms scale [t(48)= 0.41, p-=0.0.4786] with those with higher general health (Mean = 19.09, SD = 3.13) scoring similarlt to those with lower general health (Mean = 18.50, SD = 2.71).

A Wilcoxon rank sum test with continuity correction indicated a significant difference in the coping score [W=442.00, p=0.0088], those with higher general health (Median = 15.00, IQR = 5.75) scoring higher than those with lower general health (Median = 12.00, IQR = 4.25). No other statistically significant differences were observed between these two groups

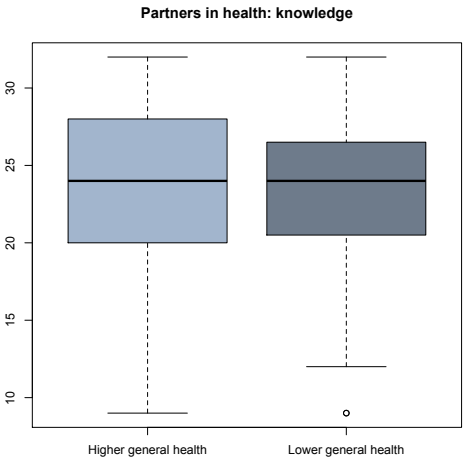


Figure 6.7: Boxplot of PIH knowledge by general health

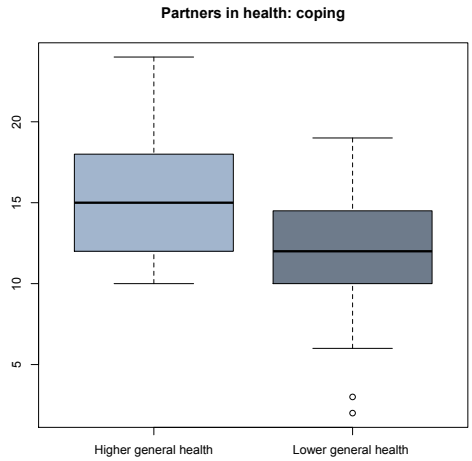


Figure 6.8: Boxplot of PIH coping by general health

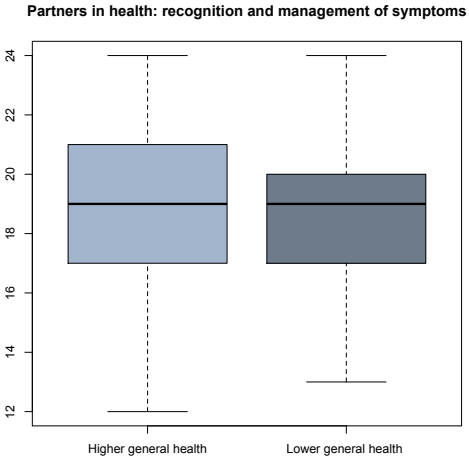


Figure 6.9: Boxplot of PIH recognition and management of symptoms by general health

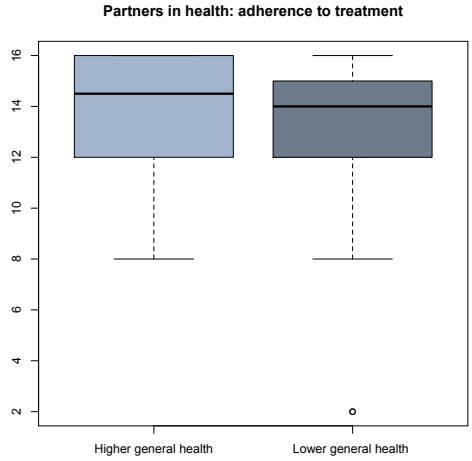


Figure 6.10: Boxplot of PIH adherence to treatment by general health

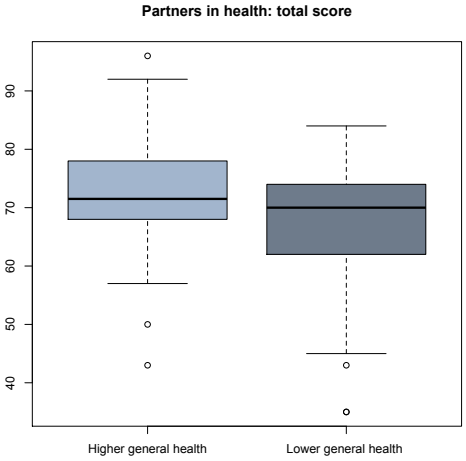


Figure 6.11: Boxplot of PIH total score by general health

Table 6.8: Summary statistics and t-test PIH scales by general health

Partners in health scale by general health	Group	Count	Mean	SD	Median	IQR	t	dF	p
Recognition and management of symptoms	Higher general health	22	19.09	3.13	19.00	3.75	0.71	48	0.4786
	Lower general health	28	18.50	2.71	19.00	3.00			

Table 6.9: Summary statistics and Wilcoxon rank sum test PIH scales by general health

Partners in health scale by general health	Group	Count	Mean	SD	Median	IQR	W	p
Knowledge	Higher general health	22	23.82	6.33	24.00	7.75	332.50	0.6371
	Lower general health	28	22.93	5.91	24.00	5.50		
Coping	Higher general health	22	15.59	4.39	15.00	5.75	442.00	0.0088*
	Lower general health	28	11.68	4.30	12.00	4.25		
Adherence to treatment	Higher general health	22	13.59	2.68	14.50	4.00	346.50	0.4504
	Lower general health	28	12.86	3.67	14.00	3.00		
Total score	Higher general health	22	72.09	12.28	71.50	9.75	384.00	0.1392
	Lower general health	28	65.96	12.68	70.00	11.50		

*Statistically significant at p<0.05

Comparisons of PIH sub scales by Physical function

Comparisons of PIH subscales were made physical functioning, those that had a SF36 physical functioning score above average for the group (Higher physical functioning) were compared with those that had an average or lower score (Lower physical functioning) . Summary statistics are listed in Tables 6.11 and 6.14.

Two sample t-test was used when assumptions for normality and variance were met (Table 6.16). When assumptions normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 6.17). A two sample t-test indicated a significant difference in the coping scale [t(48)= 2.27, p=0.275] those with a higher emotional well-being score (Mean = 15.05, SD = 4.21) scoring higher than those with a lower emotional well-being score (Mean = 12.11, SD = 4.77).

No other statistically significant differences were observed between these two groups for any other PIH sub scale (Tables 6.18).

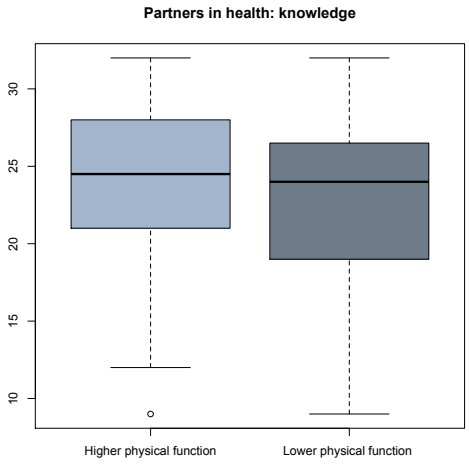


Figure 6.12: Boxplot of PIH knowledge by physical functioning

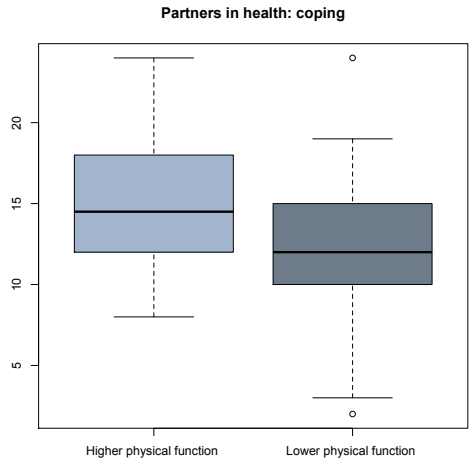


Figure 6.13: Boxplot of PIH coping by physical functioning

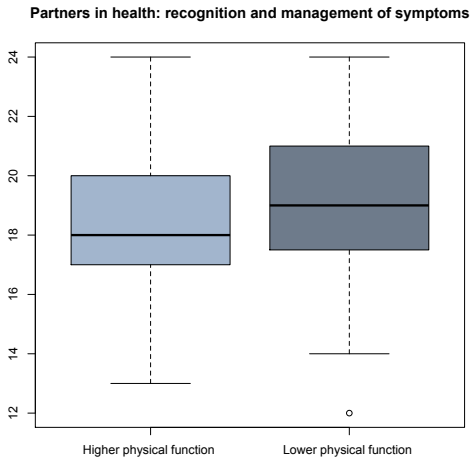


Figure 6.14: Boxplot of PIH recognition and management of symptoms by physical functioning

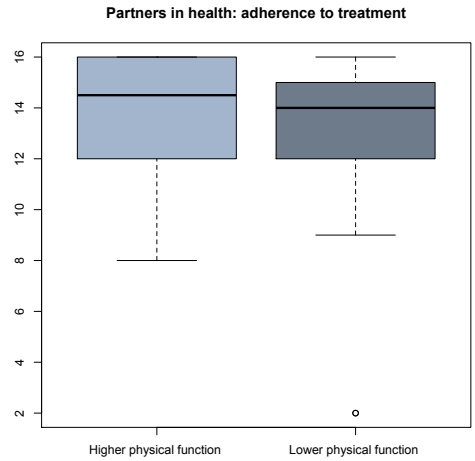


Figure 6.15: Boxplot of PIH aherence to treatment by physical functioning

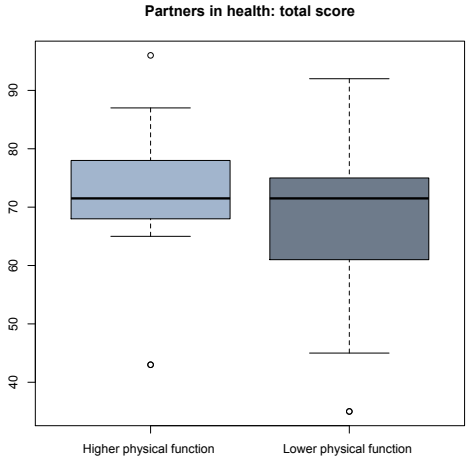


Figure 6.16: Boxplot of PIH total score by physical functioning

Table 6.10: Summary statistics and t-test PIH subscales by physical functioning

Partners in health scales by physical function	Group	Count	Mean	SD	t	dF	p
Coping	Higher physical function	22	15.05	4.21	2.27	48	0.0275*
	Lower physical function	28	12.11	4.77			
Recognition and management of symptoms	Higher physical function	22	18.45	2.67	-0.66	48	0.5132
	Lower physical function	28	19.00	3.08			

Table 6.11: Summary statistics Wilcoxon rank sum test with continuity correction PIH subscales by physical functioning

Partners in health scales by physical function	Group	Count	Median	IQR	W	p
Knowledge	Higher physical function	22	24.50	6.75	334.00	0.6162
	Lower physical function	28	24.00	6.75		
Adherence to treatment	Higher physical function	22	14.50	4.00	342.50	0.4995
	Lower physical function	28	14.00	3.00		
Total score	Higher physical function	22	71.50	4.25	347.50	0.4450
	Lower physical function	28	71.50	13.50		

Comparisons of PIH sub scales by emotional well-being

Comparisons of PIH subscales were made by emotional well-being, those that had a SF36 emotional well-being score above average for the group (Higher emotional well-being) were compared with those that had an average or lower score (Lower emotional well-being) . Summary statistics are listed in Tables 6.11 and 6.14.

Two sample t-test was used when assumptions for normality and variance were met (Table 6.16). When assumptions normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 6.17). A two sample t-test indicated a significant difference in the coping scale [t(48)= 4.50, p<0.0001] those with a higher emotional well-being score (Mean = 15.85, SD = 4.12) scoring higher than those with a lower emotional well-being score (Mean = 10.75, SD = 3.87).

A Wilcoxon rank sum test with continuity correction indicated a significant difference in the adherence to treatment score [W=412.50, p=0.0485], those with higher emotional well-being (Median = 15.00, IQR = 2.75) scoring higher than those with lower emotional well-being (Median = 14.00, IQR = 5.50); and for the total score [W= 455.50, p = 0.0054], those with higher emotional well-being (Median = 72.50, IQR = 8.50) scoring higher than those with lower emotional well-being (Median = 68.00, IQR =18.75).

No other statistically significant differences were observed between these two groups

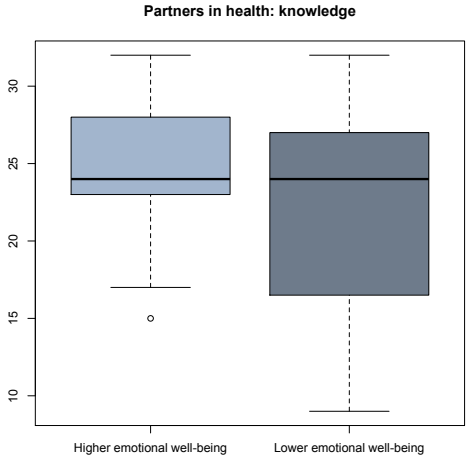


Figure 6.17: Boxplot of PIH knowledge by emotional well-being

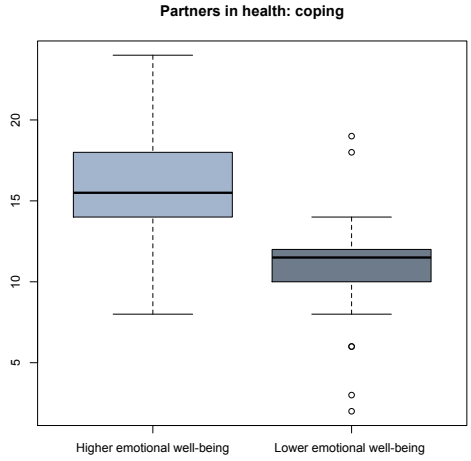


Figure 6.18: Boxplot of PIH coping by emotional well-being

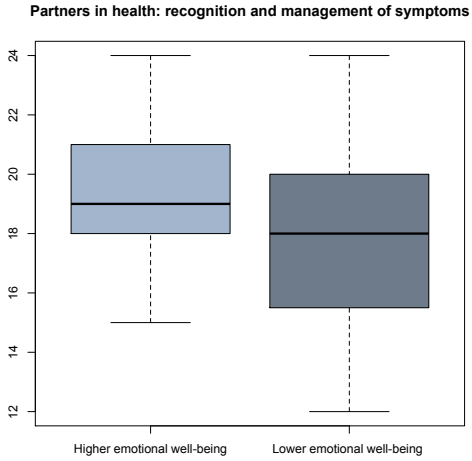


Figure 6.19: Boxplot of PIH recognition and management of symptoms by emotional well-being

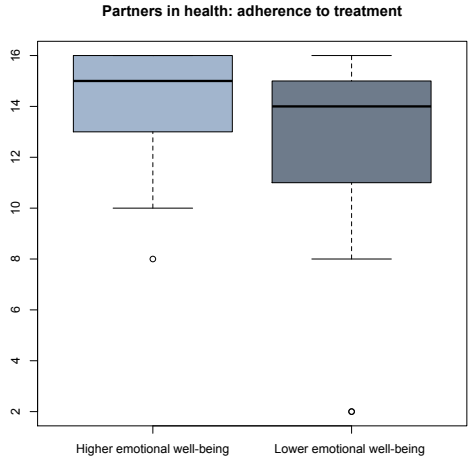


Figure 6.20: Boxplot of PIH adherence to treatment by emotional well-being

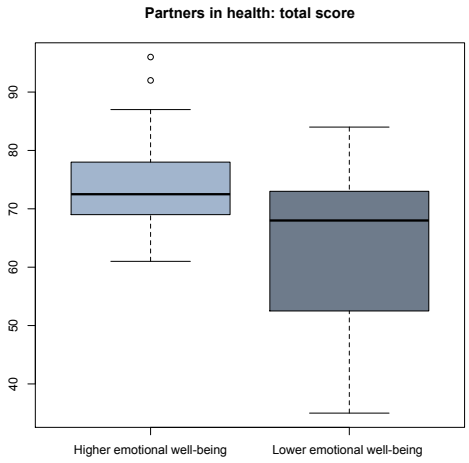


Figure 6.21: Boxplot of PIH total score by emotional well-being

Table 6.12: Summary statistics and two sample t-test PIH subscale by emotional well-being

Partners in health scales by emotional well-being	Group	Count	Mean	SD	Median	IQR	T	dF	p
Coping	Higher emotional well-being	26	15.85	4.12	15.50	3.75	4.50	48	<0.0001*
	Lower emotional well-being	24	10.75	3.87	11.50	2.00			
Recognition and management of symptoms	Higher emotional well-being	26	19.50	2.23	19.00	3.00	1.94	48	0.0586
	Lower emotional well-being	24	17.96	3.33	18.00	2.00			

* Statistically significant at p<0.05

Table 6.13: Summary statistics Wilcoxon rank sum test with continuity correction PIH subscales by emotional well-being

Partners in health scales by emotional well-being	Group	Count	Mean	SD	Median	IQR	W	p
Knowledge	Higher emotional well-being	26	24.65	4.35	24.00	4.50	361.50	0.3386
	Lower emotional well-being	24	21.88	7.29	24.00	10.25		
Adherence to treatment	Higher emotional well-being	26	14.12	2.14	15.00	2.75	412.50	0.0485*
	Lower emotional well-being	24	12.17	3.95	14.00	3.50		
Total score	Higher emotional well-being	26	74.12	8.25	72.50	8.50	455.50	0.0054*
	Lower emotional well-being	24	62.75	14.23	68.00	18.75		

* Statistically significant at p<0.05

Comparisons of PIH sub scales by social functioning

Comparisons of PIH subscales were made by social functioning, those that had a social functioning score above average for the group (Higher social functioning) were compared with those that had an average or lower score (Lower social functioning) . Summary statistics are listed in Tables 6.11 and 6.14.

Two sample t-test was used when assumptions for normality and variance were met (Table 6.16). When assumptions normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 6.17). A two sample t-test indicated a significant difference in the coping scale [t(48)= 5.47, p<0.0001] those with a higher social functiong (Mean = 16.95, SD = 3.86) scoring higher than those with a lower social functioning score (Mean = 11.03, SD = 3.67).

A Wilcoxon rank sum test with continuity correction indicated a significant difference in the total score [W=426.50, p=0.0124], those with higher social functioning (Median = 74.00, IQR = 10.50) scoring higher than those with lower social functioning (Median = 70.00, IQR = 11.00).

No other statistically significant differences were observed between these two groups

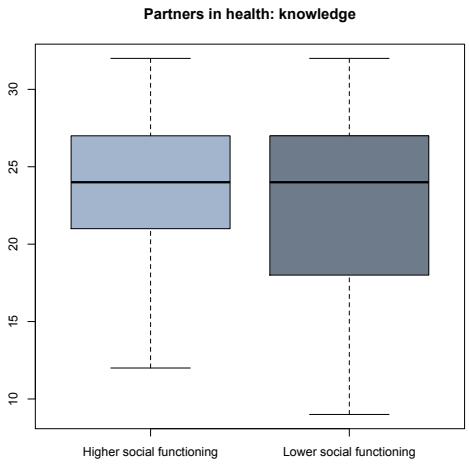


Figure 6.22: Boxplot of PIH knowledge by social functioning

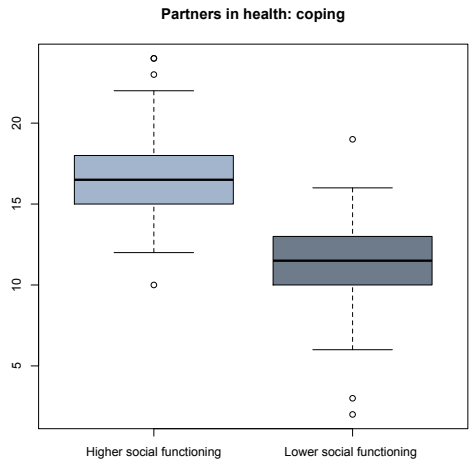


Figure 6.23: Boxplot of PIH coping by social functioning

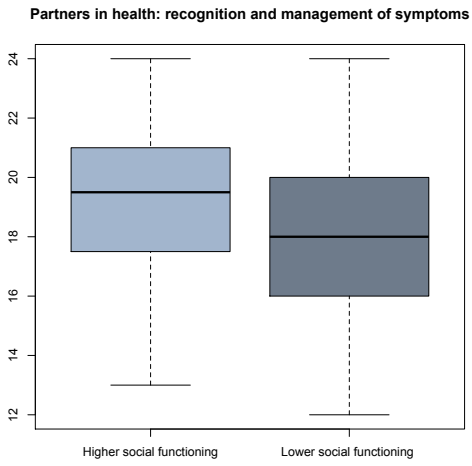


Figure 6.24: Boxplot of PIH recognition and management of symptoms by social functioning

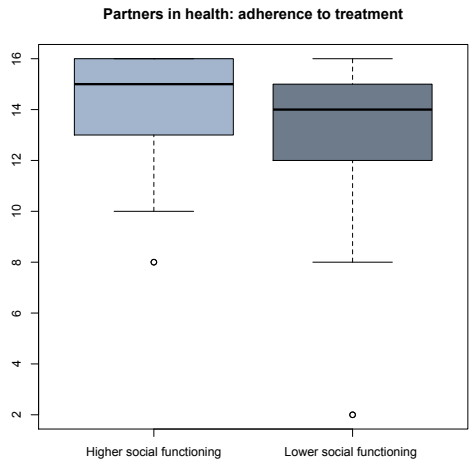


Figure 6.25: Boxplot of PIH adherence to treatment by social functioning

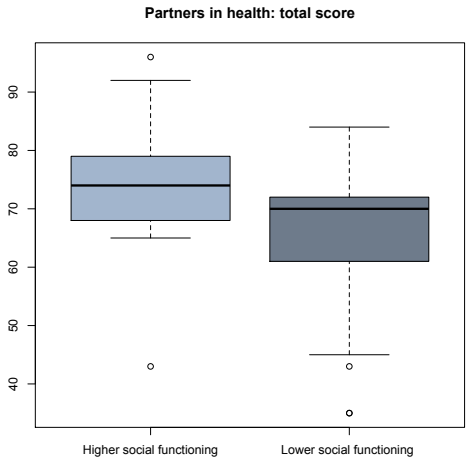


Figure 6.26: Boxplot of PIH total score by social functioning

Table 6.14: Summary statistics and two sample t-test PIH subscale by social functioning

Partners in health scales by social functioning	Group	Count	Mean	SD	Median	IQR	t	dF	p
Coping	Higher	20	16.95	3.86	16.50	3.00	5.47	48	<0.0001*
	Lower	30	11.03	3.67	11.50	2.75			
Recognition and management of symptoms	Higher	20	19.60	2.84	19.50	3.25	1.71	48	0.0935
	Lower	30	18.20	2.83	18.00	2.75			

*Statistically significant at p<0.05

Table 6.15: Summary statistics Wilcoxon rank sum test with continuity correction PIH subscales by emotional well-being

Partners in health scales by social functioning	Group	Count	Mean	SD	Median	IQR	W	p
Knowledge	Higher	20	23.80	5.29	24.00	5.50	303.00	0.9603
	Lower	30	23.00	6.58	24.00	8.50		
Adherence to treatment	Higher	20	13.85	2.54	15.00	3.00	359.00	0.2391
	Lower	30	12.73	3.64	14.00	3.00		
Total score	Higher	20	74.20	11.30	74.00	10.50	426.50	0.0124*
	Lower	30	64.97	12.48	70.00	11.00		

*Statistically significant at p<0.05

Comparisons of PIH sub scales by hearing problem status

Comparisons of PIH subscales were made by those that had no hearing problems compared with those with hearing problems. Summary statistics are listed in Tables 6.11 and 6.14.

Two sample t-test was used when assumptions for normality and variance were met (Table 6.16). When assumptions normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 6.17).

No statistically significant differences were observed between these two groups

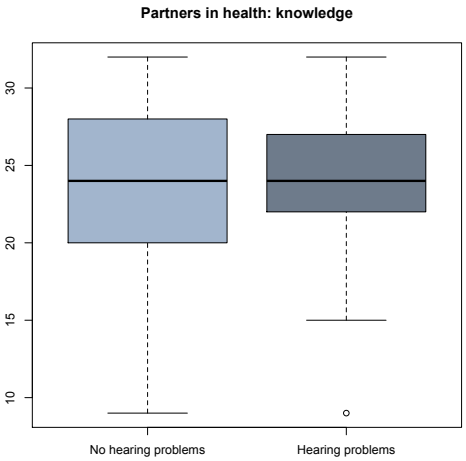


Figure 6.27: Boxplot of PIH knowledge by hearing problems

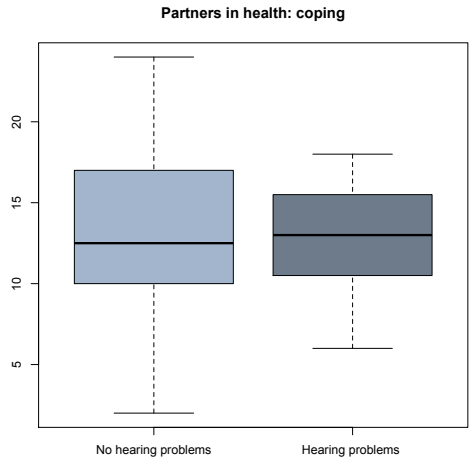


Figure 6.28: Boxplot of PIH coping by h

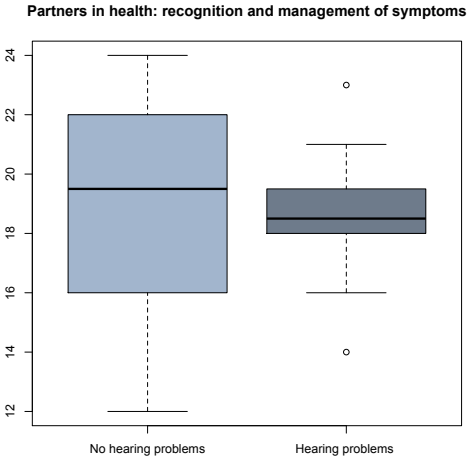


Figure 6.29: Boxplot of PIH recognition and management of symptoms by hearing problems

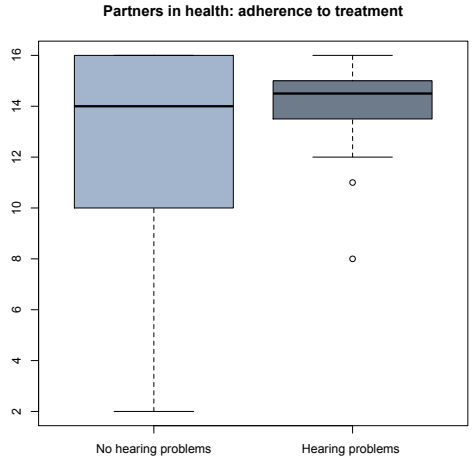


Figure 6.30: Boxplot of PIH adherence to treatment by h

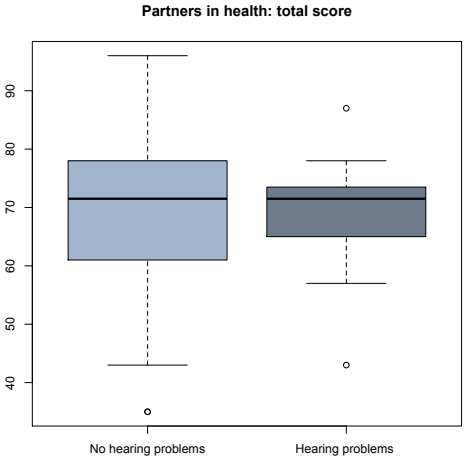


Figure 6.31: Boxplot of PIH total score by hearing problems

Table 6.16: Summary statistics and two sample t-test PIH subscale by hearing problems

Partners in health scales by hearing problems	Group	Count	Mean	SD	Median	IQR	t	dF	p
Knowledge	No hearing problems	26	23.08	6.52	24.00	7.75	-0.29	48	0.7708
	Hearing problems	24	23.58	5.63	24.00	4.50			

Table 6.17: Summary statistics Wilcoxon rank sum test with continuity correction PIH subscales by hearing problems

Partners in health scales by hearing problems	Group	Count	Mean	SD	Median	IQR	W	p
Coping	No hearing problems	26	13.92	5.59	12.50	6.50	339.50	0.5983
	Hearing problems	24	12.83	3.61	13.00	4.50		
Recognition and management of symptoms	No hearing problems	26	18.88	3.63	19.50	5.75		
	Hearing problems	24	18.63	1.86	18.50	4.50	347.00	0.4999
Adherence to treatment	No hearing problems	26	12.38	4.01	14.00	5.50		
	Hearing problems	24	14.04	1.92	14.50	1.25	254.50	0.2607
Total score	No hearing problems	26	68.27	15.92	71.50	15.00	340.50	0.5859
	Hearing problems	24	69.08	8.40	71.50	8.25		

Comparisons of PIH sub scales by eye problem status

Comparisons of PIH subscales were made by eye problem status, those that had no eye problems were compared with those that eye problems. Summary statistics are listed in Tables 6.11 and 6.14.

Two sample t-test was used when assumptions for normality and variance were met (Table 6.16). When assumptions normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 6.17).

A Wilcoxon rank sum test with continuity correction indicated a significant difference in the knowledge sub scale [W=399.50, p=0.0079], those with no eye problems (Median = 27.50, IQR = 7.00) scoring higher than those with eye problems (Median = 24.00, IQR = 7.50); a significant difference in the adherence to treatment sub scale [W=367.50, p=0.0447], those with no eye problems (Median = 15.00, IQR = 2.00) scoring higher than those with eye problems (Median = 14.00, IQR = 3.75); and a significant difference in the total score [W=383.50,
p=0.0207], those with no eye problems (Median = 74.00, IQR = 9.00) scoring higher than those with eye problems (Median = 68.50, IQR = 10.50).

No other statistically significant differences were observed between these two groups

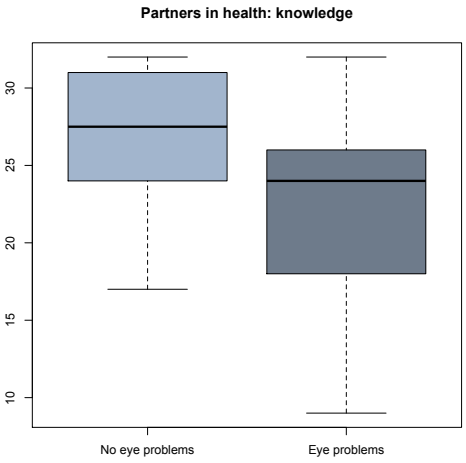


Figure 6.32: Boxplot of PIH knowledge by eye problems

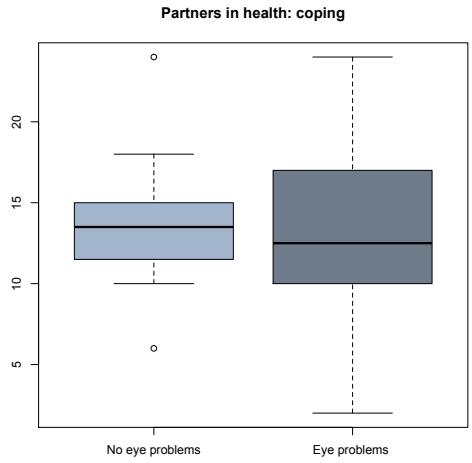


Figure 6.33: Boxplot of PIH coping by eye problems

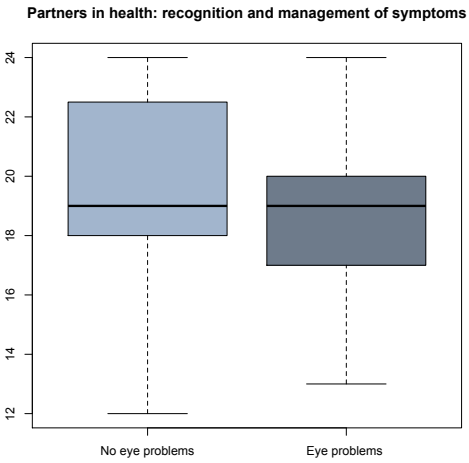


Figure 6.34: Boxplot of PIH recognition and management of symptoms by eye problems

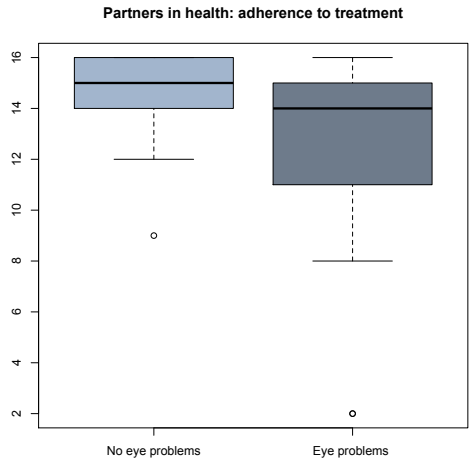


Figure 6.35: Boxplot of PIH adherence to treatment by eye problems

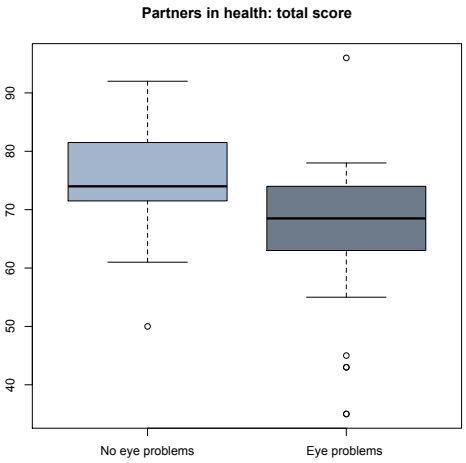


Figure 6.36: Boxplot of PIH total score by eye problems

Table 6.18: Summary statistics and two sample t-test PIH subscale by eye problems

Partners in health scales by eye problems	Group	Count	Mean	SD	Median	IQR	t	dF	p
Coping	No eye problems	16	13.56	3.92	13.50	3.25	0.17	48	0.8695
	Eye problems	34	13.32	5.12	12.50	6.75			
Recognition and management of symptoms	No eye problems	16	19.75	3.36	19.00	4.25	1.69	48	0.0969
	Eye problems	34	18.29	2.56	19.00	6.75			

Table 6.19: Summary statistics Wilcoxon rank sum test with continuity correction PIH subscales by eye problems

Partners in health scales by eye problems	Group	Count	Mean	SD	Median	IQR	W	p
Knowledge	No eye problems	16	26.44	4.95	27.50	7.00	399.50	0.0079*
	Eye problems	34	21.85	6.02	24.00	7.50		
Adherence to treatment	No eye problems	16	14.50	1.83	15.00	2.00	367.50	0.0447*
	Eye problems	34	12.56	3.61	14.00	3.75		
Total score	No eye problems	16	74.25	10.20	74.00	9.00	383.50	0.0207*
	Eye problems	34	66.03	13.11	68.50	10.50		

*Statistically significant at p<0.05

Comparisons of PIH sub scales by location

Comparisons of PIH subscales were made by location, those that lived in a metropolitan area were compared with those that lived in a regional or rural location. Summary statistics are listed in Tables 6.11 and 6.14.

Two sample t-test was used when assumptions for normality and variance were met (Table 6.16). When assumptions normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 6.17).

A Wilcoxon rank sum test with continuity correction indicated a significant difference in the total score [W=410.00, p=0.0298], those that lived in a metropolitan area (Median = 72.50, IQR = 8.75) scoring higher than those that lived in a regional or rural area (Median = 68.00, IQR = 12.00).

No other statistically significant differences were observed between these two groups

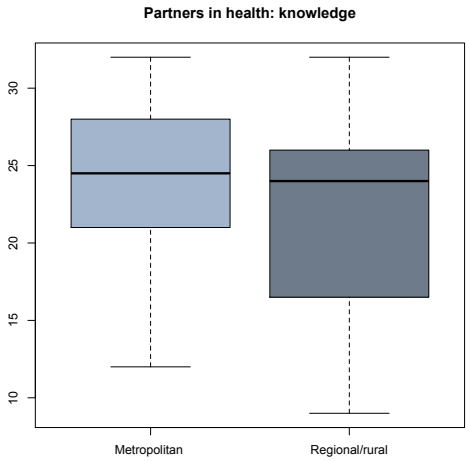


Figure 6.37: Boxplot of PIH knowledge by location

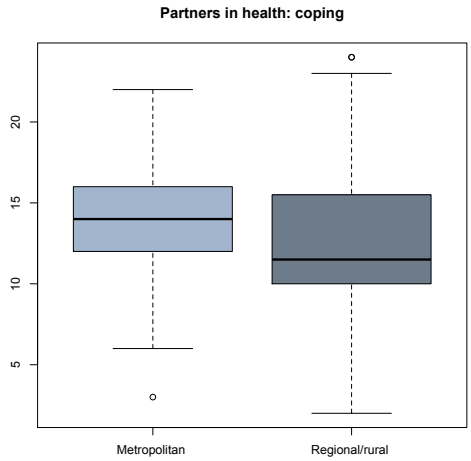


Figure 6.38: Boxplot of PIH coping by location

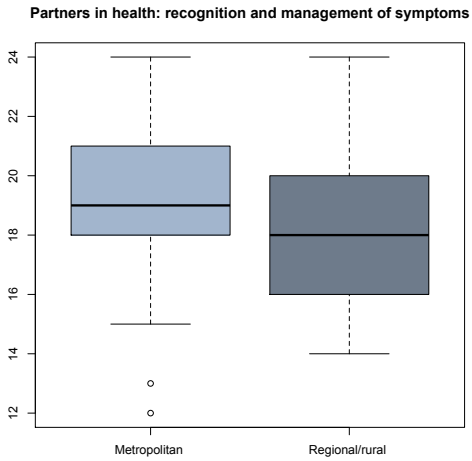


Figure 6.39: Boxplot of PIH recognition and management of symptoms by location

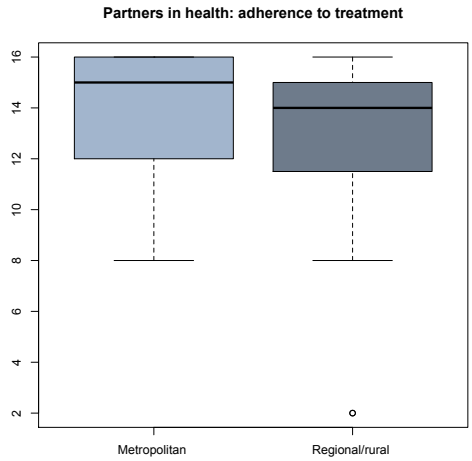


Figure 6.40: Boxplot of PIH adherence to treatment by location

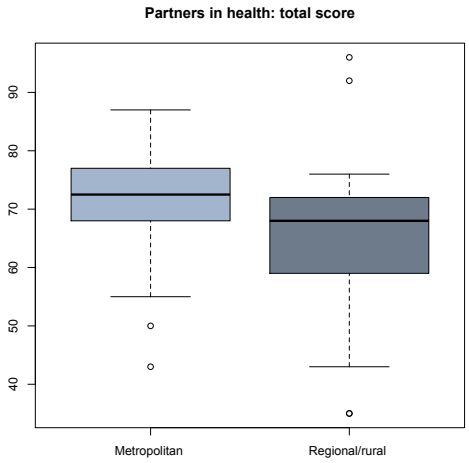


Figure 6.41: Boxplot of PIH total score by location

Table 6.20: Summary statistics and two sample t-test PIH subscale by social functioning

Partners in health scales by location	Group	Count	Mean	SD	Median	IQR	t	dF	P
Recognition and management of symptoms	Metropolitan	30	19.13	2.83	19.00	3.00	1.12	48	0.2675
	Regional/Rural	20	18.20	2.97	18.00	5.25			

Table 6.21: Summary statistics Wilcoxon rank sum test with continuity correction PIH subscales by emotional well-being

Partners in health scales by location	Group	Count	Mean	SD	Median	IQR	W	p
Knowledge	Metropolitan	30	24.43	5.32	24.50	7.00	365.00	0.1989
	Regional/Rural	20	21.65	6.81	24.00	9.25		
Coping	Metropolitan	30	13.87	3.84	14.00	4.00	381.00	0.1092
	Regional/Rural	20	12.70	5.85	11.50	5.25		
Adherence to treatment	Metropolitan	30	13.83	2.31	15.00	3.50	369.00	0.1681
	Regional/Rural	20	12.20	4.20	14.00	3.25		
Total score	Metropolitan	30	71.27	9.50	72.50	8.75	410.00	0.0298*
	Regional/Rural	20	64.75	15.97	68.00	12.00		

*Statistically significant at p<0.05

Comparisons of PIH sub scales by education status
Comparisons of PIH subscales were made education, those that had a university qualification were compared with those that high school or trade qualifications. Summary statistics are listed in Tables 6.11 and 6.14.

Two sample t-test was used when assumptions for normality and variance were met (Table 6.16). When assumptions normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 6.17).

No statistically significant differences were observed between these two groups

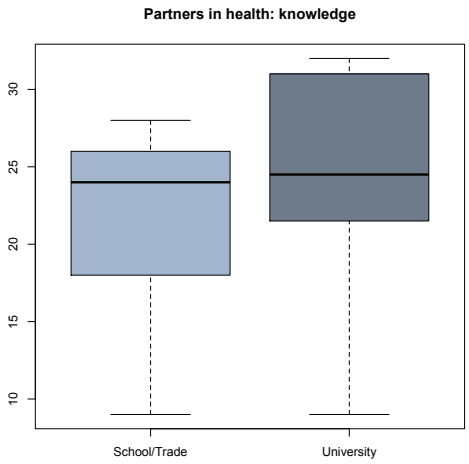


Figure 6.42: Boxplot of PIH knowledge by education

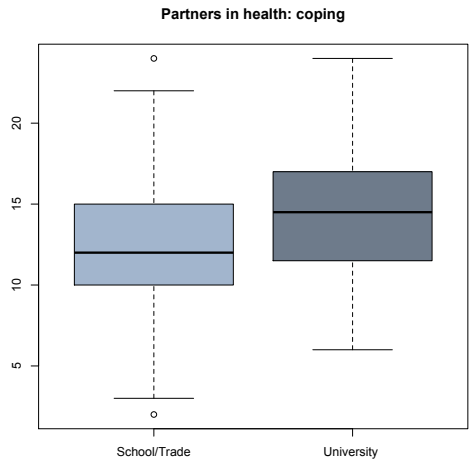


Figure 6.43: Boxplot of PIH coping by education

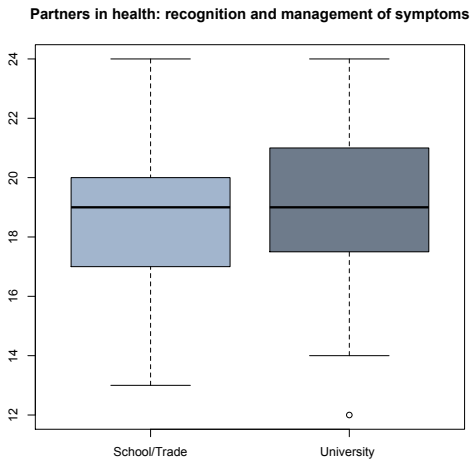


Figure 6.44: Boxplot of PIH recognition and management of symptoms by education

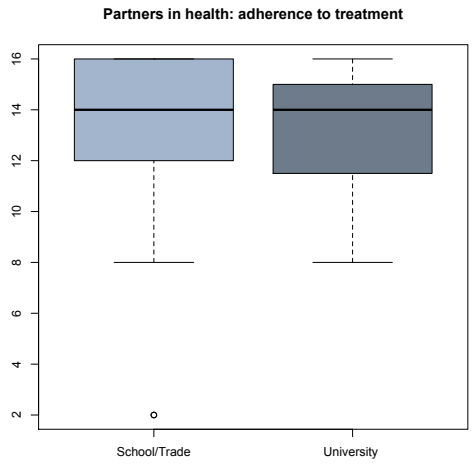


Figure 6.45: Boxplot of PIH adherence to treatment by education

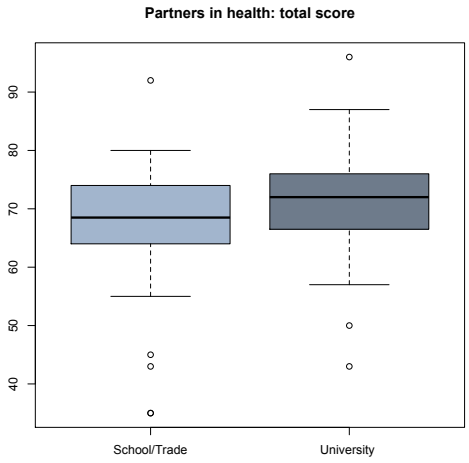


Figure 6.46: Boxplot of PIH total score by education

Table 6.22: Summary statistics and two sample t-test PIH subscale by education

Partners in health scales by education	Group	Count	Mean	SD	Median	IQR	T	dF	p
Coping	School/Trade	26	12.54	5.05	12.00	6.00	-1.35	48	0.1823
	University	24	14.33	4.25	14.50	5.25			
Recognition and management of symptoms	School/Trade	26	18.62	2.94	19.00	4.25	-0.37	48	0.7167
	University	24	18.92	2.89	19.00	5.25			

Table 6.23: Summary statistics Wilcoxon rank sum test with continuity correction PIH subscales by education

Partners in health scales by education	Group	Count	Mean	SD	Median	IQR	W	p
Knowledge	School/Trade	26	22.08	5.86	24.00	7.75	249.00	0.2222
	University	24	24.67	6.08	24.50	9.25		
Adherence to treatment	School/Trade	26	13.12	3.84	14.00	3.25	337.50	0.6218
	University	24	13.25	2.57	14.00	3.25		
Total score	School/Trade	26	66.35	13.72	68.50	13.00	250.00	0.2314
	University	24	71.17	11.37	72.00	8.75		

Comparisons of PIH sub scales by SEIFA

Comparisons of PIH subscales were made by SEIFA, those that lived in an area with a SEIFA score of 7-10 (Higher SEIFA) were compared with those that lived in an area with a SEIFA score of 1-6 (Lower SEIFA) . Summary statistics are listed in Tables 6.11 and 6.14.

Two sample t-test was used when assumptions for normality and variance were met (Table 6.16). When assumptions normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 6.17).

No statistically significant differences were observed between these two groups

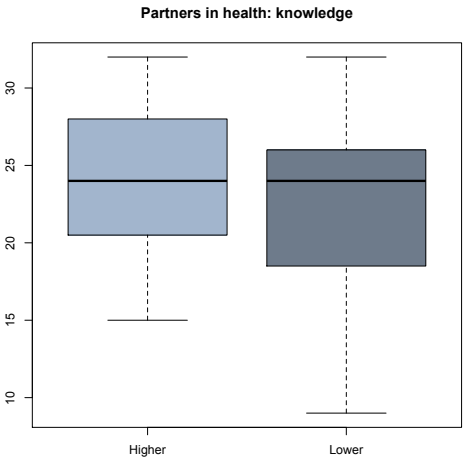


Figure 6.47: Boxplot of PIH knowledge by SEIFA

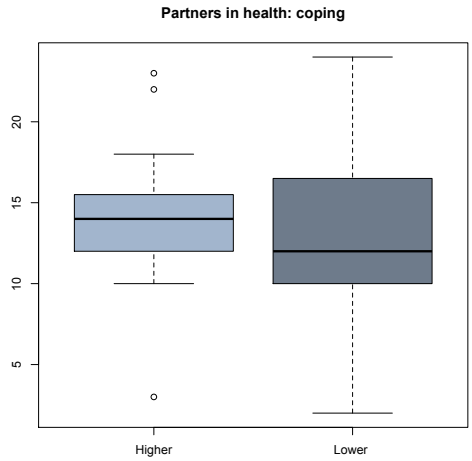


Figure 6.48: Boxplot of PIH coping by SEIFA

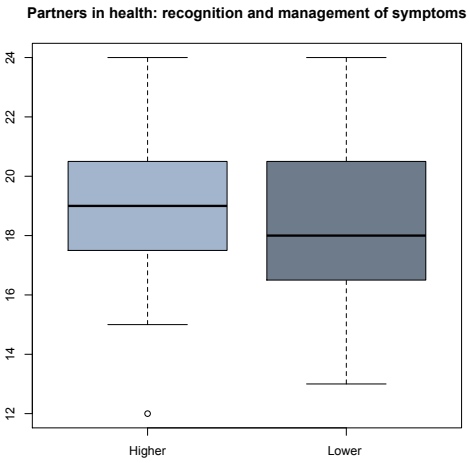


Figure 6.49: Boxplot of PIH recognition and management of symptoms by SEIFA

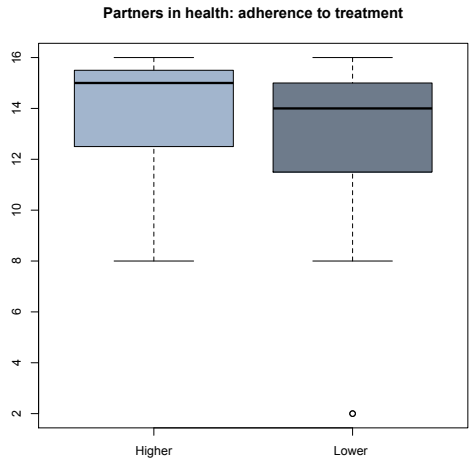


Figure 6.50: Boxplot of PIH adherence to treatment by SEIFA

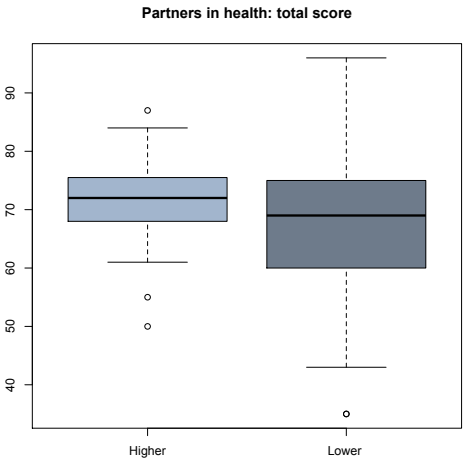


Figure 6.51: Boxplot of PIH total score by SEIFA

Table 6.24: Summary statistics and two sample t-test PIH subscale by SEIFA

Partners in Health scales by SEIFA	Group	Count	Mean	SD	Median	IQR	t	dF	p
Coping	Higher SEIFA	27	13.85	3.97	14.00	3.50	0.73	48	0.4695
	Lower SEIFA	23	12.87	5.53	12.00	6.50			
Recognition and management of symptoms	Higher SEIFA	27	19.04	2.68	19.00	3.00	0.73	48	0.4683
	Lower SEIFA	23	18.43	3.15	18.00	6.50			

Table 6.25: Summary statistics Wilcoxon rank sum test with continuity correction PIH subscales by SEIFA

Partners in Health scales by SEIFA	Group	Count	Mean	SD	Median	IQR	W	p
Knowledge	Higher SEIFA	27	24.30	5.04	24.00	7.50	345.50	0.4994
	Lower SEIFA	23	22.17	7.00	24.00	7.50		
Adherence to treatment	Higher SEIFA	27	13.85	2.27	15.00	3.00	370.50	0.2392
	Lower SEIFA	23	12.39	4.05	14.00	3.50		
Total score	Higher SEIFA	27	71.04	8.31	72.00	7.50	363.50	0.3058
	Lower SEIFA	23	65.87	16.30	69.00	15.00		

*Statistically significant at $p<0.05$

Information given by healthcare professionals and searched for independently.

Participants were asked about what type of information they were given by healthcare professionals and what type of information they searched for independently. Information about disease cause (50.00%), treatment options (38.00%), and disease management (38.00%) were most frequently given to participants by healthcare professionals, and information about clinical trials (14.00%), interpreting test results (14.00%) and complementary therapies (16.00%) were give least often (Figure 6.30). Eight participants (16.00%) indicated that they received no information at all from health professionals about mitochondrial disease.

Within subgroups, the types of information given differed, the most notable differences were observed for information: those with higher general health received more information about disease cause (68.18% compared to 37.17% for lower general health) and those with lower general health received more information about diet (39.29% compared to 13.64% for higher general health); those with higher physical functioning received more information disease cause (68.18%%) compared to those with lower physical functioning (35.71%) and hereditary, genes and biomarkers (45.5% compared to 25.00% for lower physical functioning); those with higher emotional well-being (46.15%) received information more often about disease management compared to those with lower emotional well-being (29.17%); those with no hearing problems (38.46%) received information about diet more often than those with hearing problems (16.67%); those that lived in regional or remote areas received more information about treatment options (55.00% compared with metropolitan 30%) and disease management (50.00% compared with metropolitan 30.00%) and hereditary, genes and biomarkers (50.00% compared with metropolitan 30.00%) those with a university education (62.50%) received information more often about disease cause compared to those with a high school or trade qualification (38.46%) and those that lived in an area with a higher SEIFA score (37.04%) received information about diet more often than those that lived in an area with a lower SEIFA score (17.39%).

Participants were asked about what type of information they searched for after receiving information from healthcare professionals. Information about treatment options (63.27%), disease management (59.18%), and disease cause (57.14%) were most frequently given to searched for independently, and information about interpreting test results (28.57%), hereditary, genes and biomarkers (28.57%) and psychological support (30.61%) were give least often (Figure 6.30). Two participants (4.08%) indicated that they did not search for any information.

Within subgroups, the types of information searched for differed, the most notable differences were observed for information: those with lower physical functioning health searched for more information about clinical trials (53.57% compared to 31.82% for higher physical functioning); those with lower social functioning searched for information more often about disease cause (68.97% compared to those with higher social functioning 40.00%), and those with information about disease management (72.41% compared to those with higher social functioning 40.00%); those with hearing problems searched for more often for information about disease cause (69.57% compared to those with no hearing problems 46.15%) and more often for clinical trials (61.54% compared to those with no hearing problems 26.09%); those with no eye problems searched for more often for information about disease management (81.25% compared to those with eye problems 48.48%) and those with eye problems searched for information more often about clinical trials (51.52%% compared to those with no eye problems 31.25%); those living in regional or rural locations searched for more information about clinical trials (63.16% compared to metropolitan 33.33%), interpreting test results (42.11% compared to metropolitan 20.00%), physical activity (57.89% compared to metropolitan 26.67%), and psychological/social support (47.37% compared to metropolitan 20.00%); those that lived in an area with a higher SEIFA score searched for more information about disease cause (66.67% compared to lower SEIFA 45.45%).

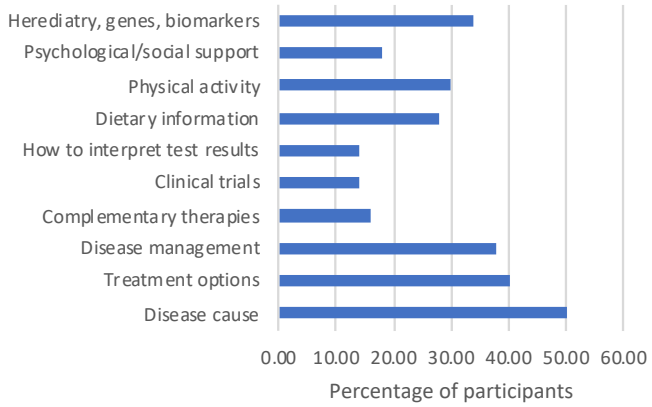


Figure 6.52: Information given by healthcare professionals: all participants

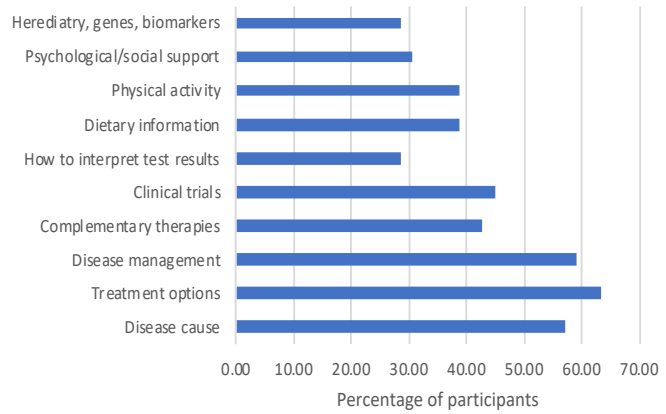


Figure 6.53: Information searched for independently: all participants

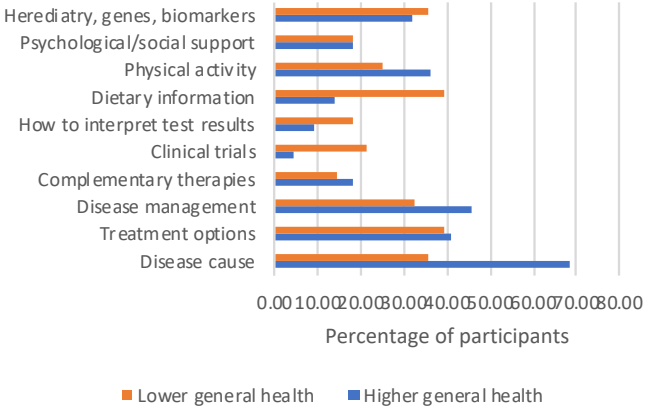


Figure 6.54: Information given by healthcare professionals by general health

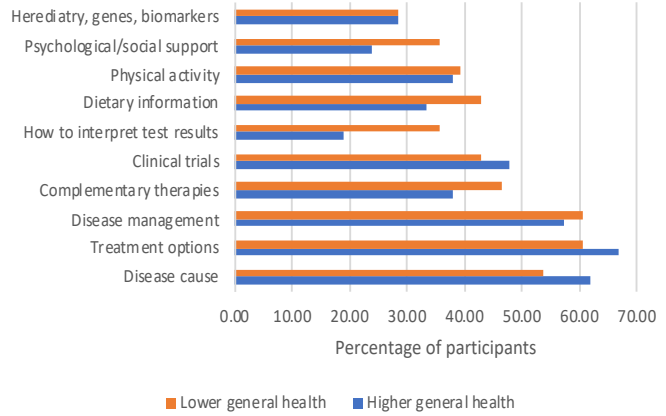


Figure 6.55: Information searched for independently by general health

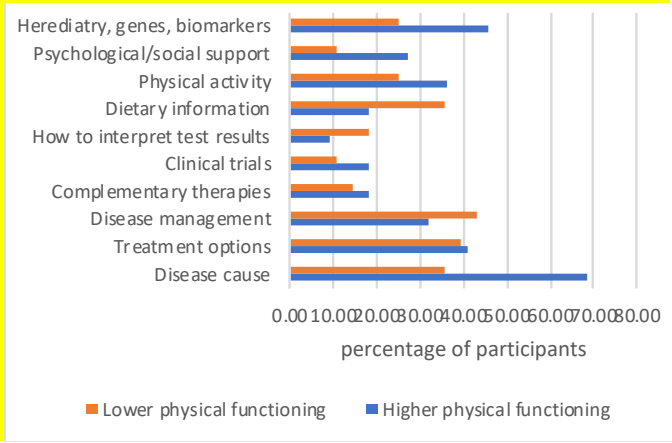


Figure 6.56: Information given by healthcare professionals by physical functioning

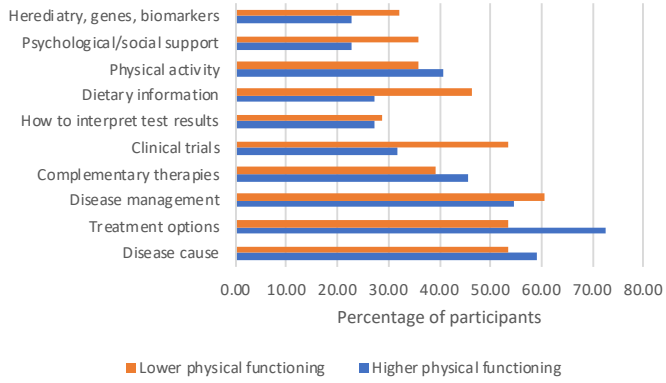


Figure 6.57: Information searched for independently by physical functioning

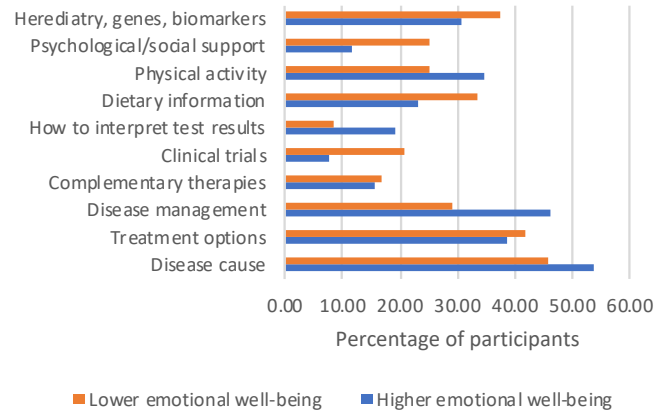


Figure 6.58: Information given by healthcare professionals by emotional well-being

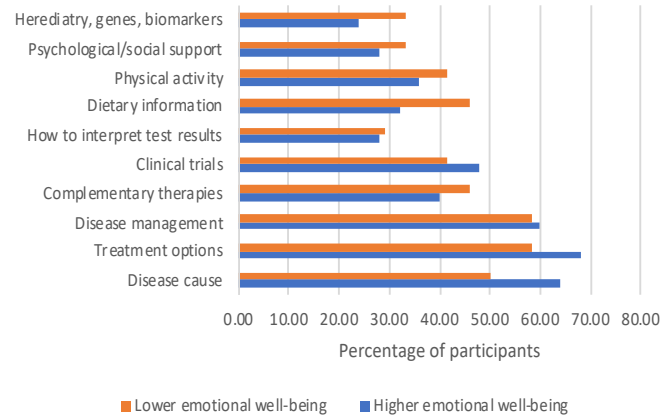


Figure 6.59: Information searched for independently by emotional well-being

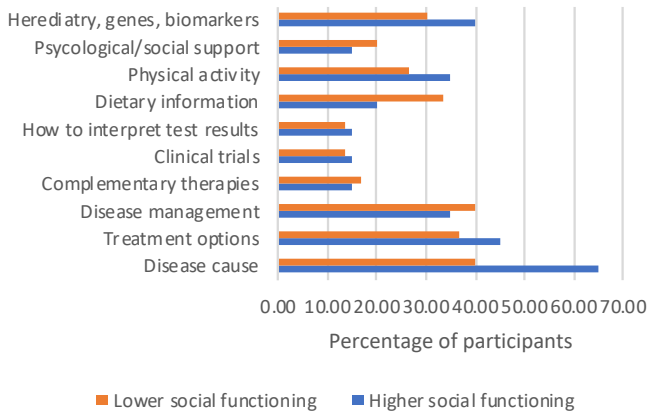


Figure 6.60: Information given by healthcare professionals by social functioning

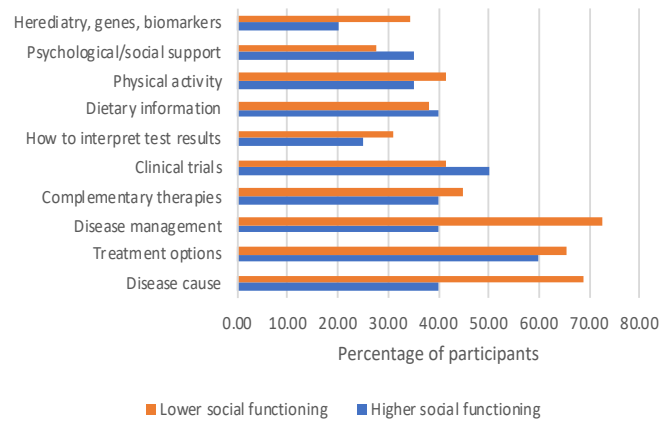


Figure 6.61: Information searched for independently by social functioning

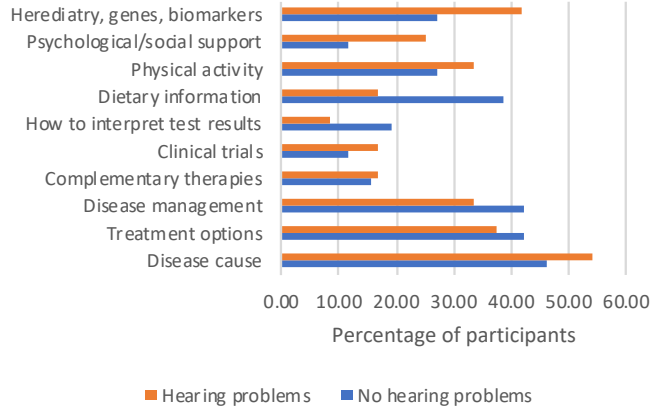


Figure 6.62: Information given by healthcare professionals by hearing problems

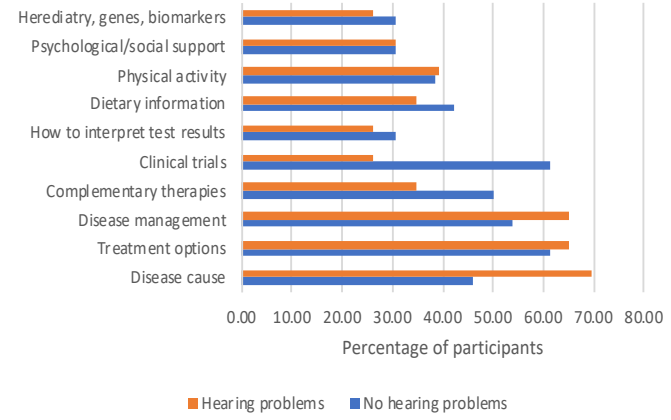


Figure 6.63: Information searched for independently by hearing problems

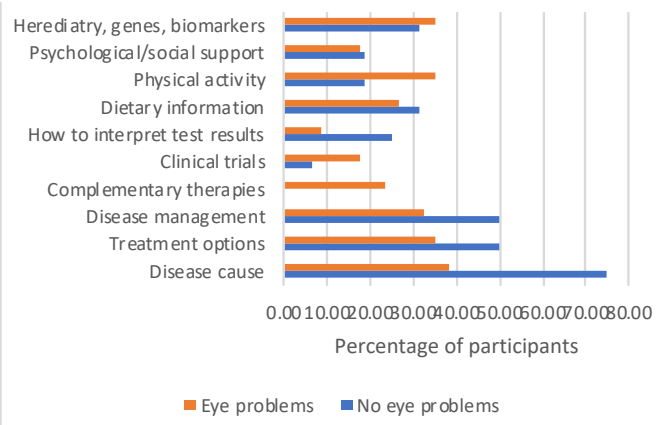


Figure 6.64: Information given by healthcare professionals by eye problems

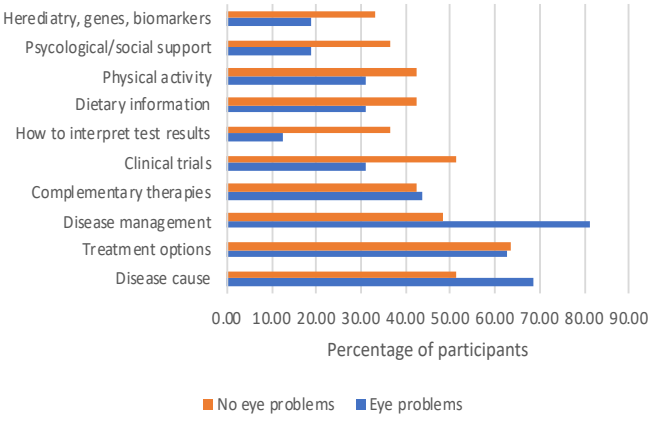
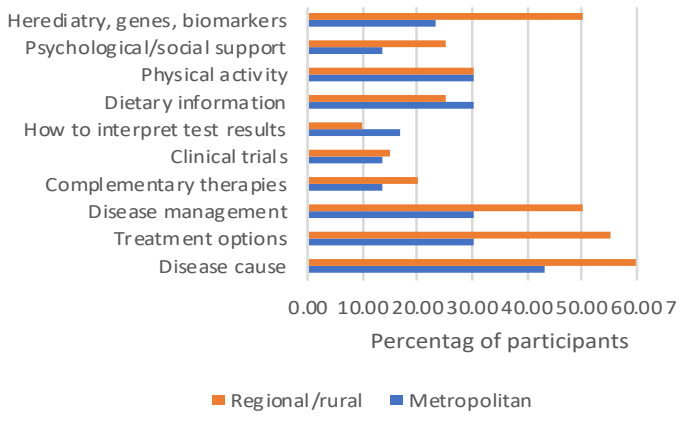


Figure 6.65: Information searched for independently by eye problems



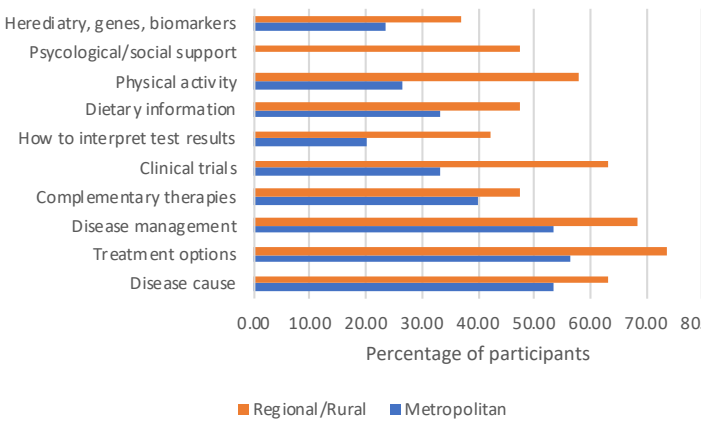


Figure 6.66: Information given by healthcare professionals by location

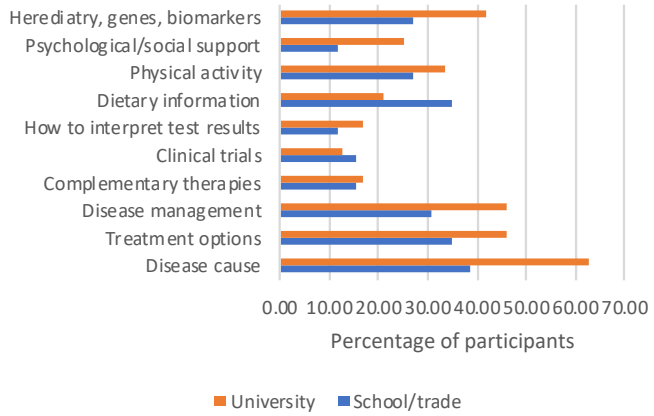


Figure 6.67: Information searched for independently by location

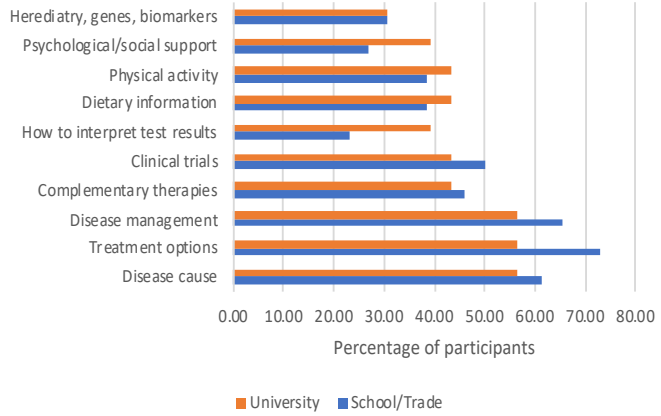


Figure 6.68: Information given by healthcare professionals by education

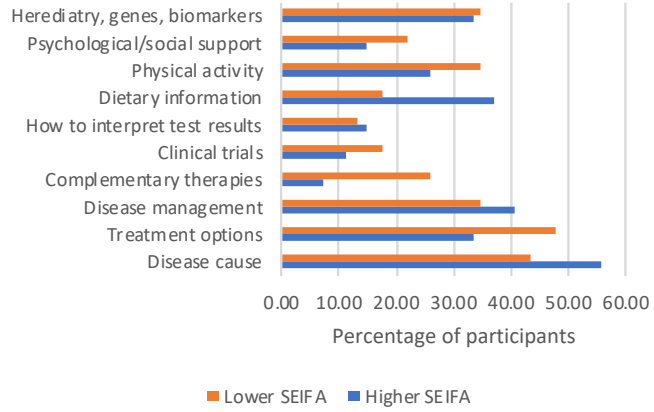


Figure 6.69: Information searched for independently by education

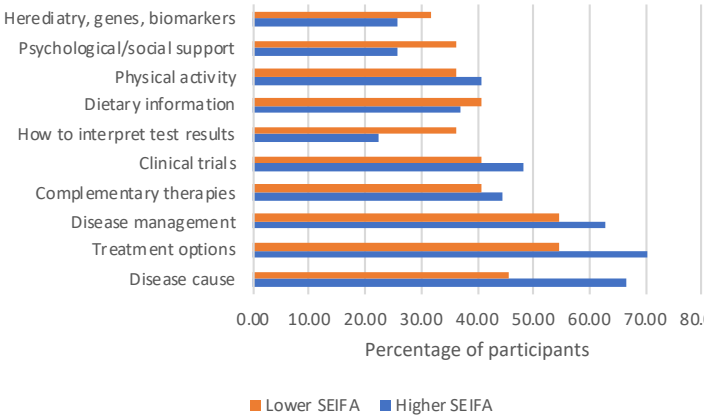


Figure 6.70: Information given by healthcare professionals by SEIFA

Figure 6.71: Information searched for independently by SEIFA

Information gaps

The largest gaps in information, where information was neither given to patients nor searched for independently were how to interpret test results (62.00%), and psychological/social support (56.00%) (Figure 6.72). Participants were given most information either from healthcare professionals or independently for treatment options (78.00%) and disease cause (78.00%) (Figure 6.72). Clinical trials (42.00%) was the topic that was most searched for independently following no information from health professionals (Figure 6.72).

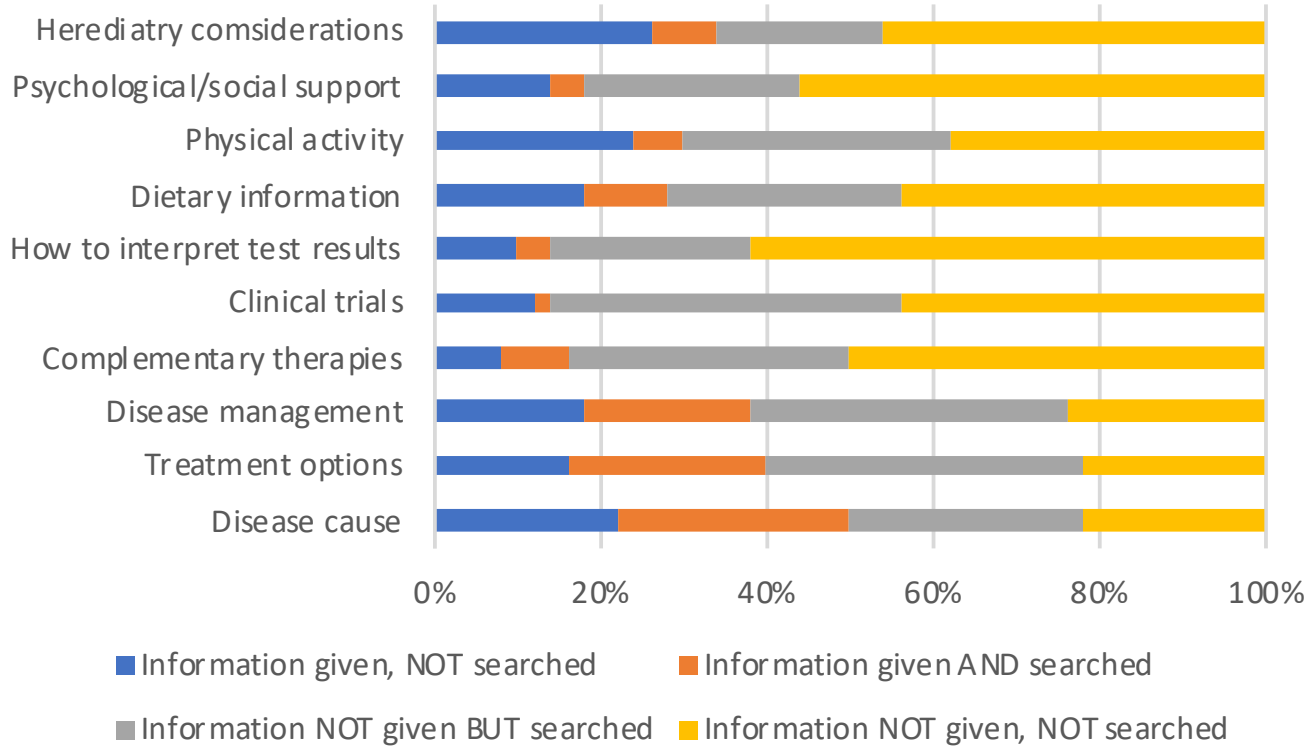


Figure 6.72: Proportion of information given by health care professionals and searched for independently.

Most trusted information sources

Participants were asked to rank which information source that they most trusted, where 1 is the most trusted and 4 is the least trusted. A weighted average is presented in Figure 6.41. With a weighted ranking, the higher the score, the more trusted the source of information to the participant. Across all participants, information from the participants' hospital or clinic and from the non-profit or charitable organisations was near equal and was most trusted. Information from pharmaceutical companies was least trusted. (Figure 6.73). This order of preference was the same for all sub-groups (Figures 6.74 – 6.82).

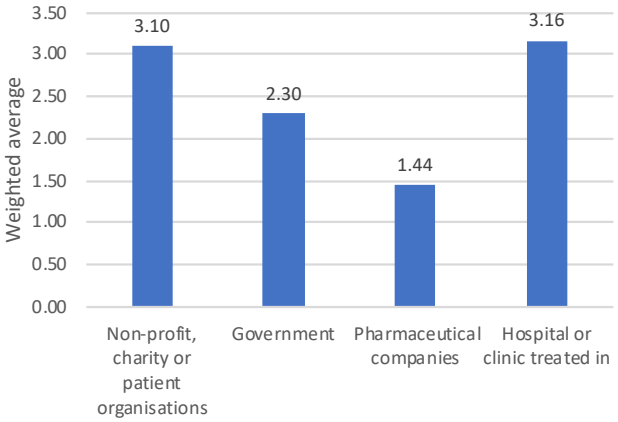


Figure 6.73: Most trusted information sources

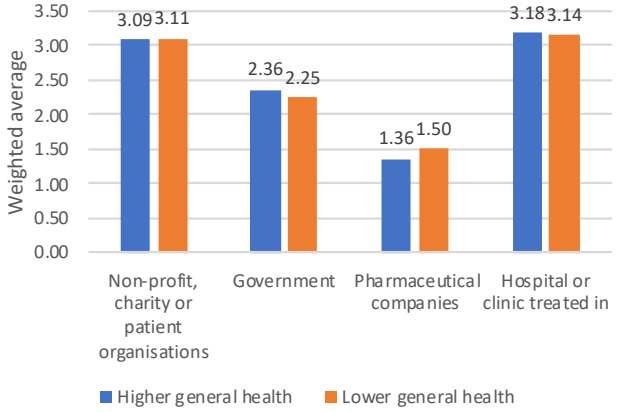


Figure 6.74: Most trusted information sources by general health

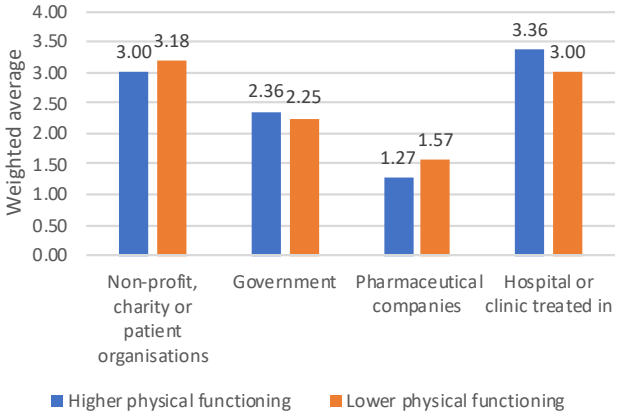


Figure 6.75: Most trusted information sources by physical functioning

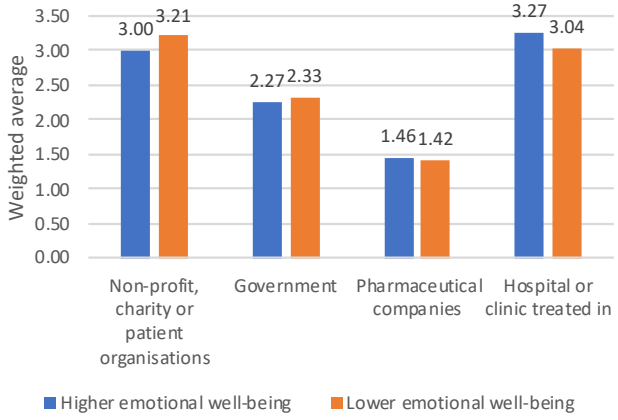


Figure 6.76: Most trusted information sources by emotional well-being

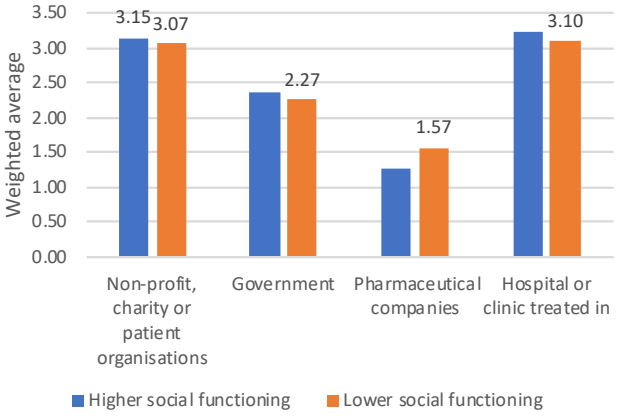


Figure 6.77: Most trusted information sources by social functioning

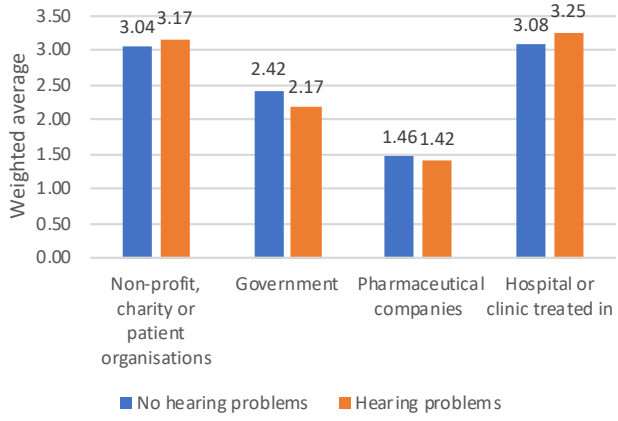


Figure 6.78: Most trusted information sources by hearing problems

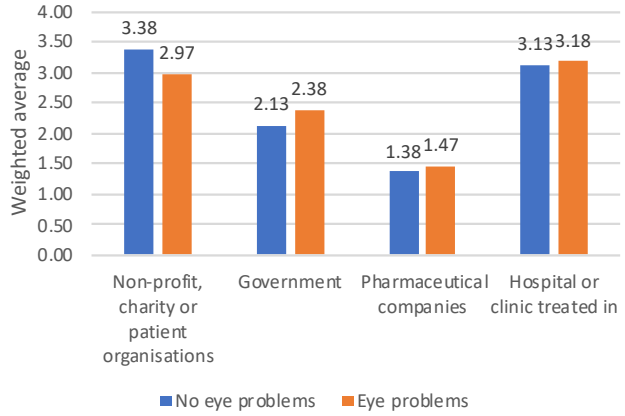


Figure 6.79: Most trusted information sources by eye problems

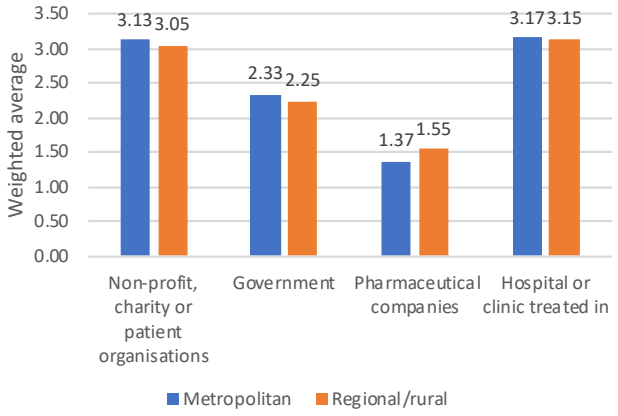


Figure 6.80: Most trusted information sources by location

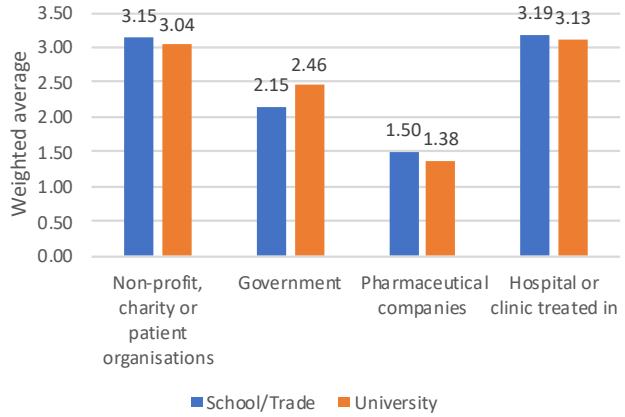


Figure 6.81: Most trusted information sources by education

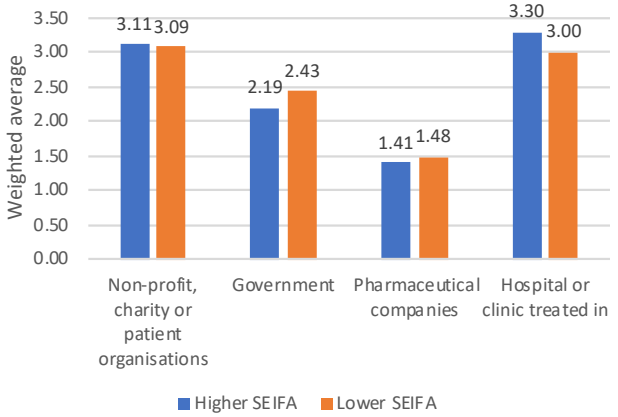


Figure 6.82: Most trusted information sources by SEIFA

Section 7 Care and support

Section 7: Experience of care and support

Care coordination

- The care coordination scores for the entire cohort for navigation, total score, care coordination global measure and quality of care global measure were all in the middle of the scale, indicating moderate outcomes. The communication score was in the second lowest quintile indicating poor communication.

Care coordination – by general health

- There were no differences observed in any care coordination scales between those with higher general health and those with lower general health

Care coordination – by physical functioning

- There were no differences observed in any care coordination scales between those with higher physical functioning and those with lower physical functioning

Care coordination – by emotional well-being

- There were no differences observed in any care coordination scales between those with higher emotional well-being and those with lower emotional well-being

Care coordination– by social functioning

- Participants with higher social functioning had a significantly better outcome compared to those with lower social functioning for the Care coordination: Navigation scale. No other statistically significant differences were observed between these two groups for any Care Coordination scores

Care coordination – by hearing problems

- There were no differences observed in any care coordination scales between those with hearing problems and those with no hearing problems

Care coordination – by eye problems

- There were no differences observed in any care coordination scales between those with eye problems and those with no eye problems

Care coordination – by location

- There were no differences observed in any care coordination scales between participants that live in metropolitan areas and those that live in regional or rural areas.

Care coordination – by education

- There were no differences observed in any care coordination scales between participants with university qualifications and those with high school or trade qualifications

Care coordination – by SEIFA

- There were no differences observed in any care coordination scales between participants that live in areas with higher SEIFA scores and those that live in areas with lower SEIFA scores.

Care and support

- Participants were asked what care and support they had received throughout their experience. This question aims to investigate what services patients consider to be support and care services. The most common description of care and support was in the form of domestic and home care support from government services and NDIS (n=14, 28.00%), this was followed by participants describing that they did not receive any care and support in general (n =9, 18.00%) and not receiving significant support and care from the clinical setting (n=9, 18.00%). There were also seven participants (14.00%) that described receiving support from family and friends.
- In relation to sub-group variations, participants with high social functioning (30.00%) describes not receiving any care and support more frequently than the general population (18.00%)

Experience of coordination of care

A Care Coordination questionnaire was completed. The Care Coordination questionnaire comprises a total score, 2 sub scales (communication and navigation), and a single question for each relating to care-coordination and care received. A higher score denotes better care outcome. Summary statistics for the entire cohort are displayed alongside the possible range of each scale in Table 7.1. Overall the entire cohort had a median care received score of 9.0, which is in the highest quintile, indicating very good care received. The scores for navigation (mean = 22.28, sd = 5.27), total score (mean = 55.68, sd=13.52), care coordination global measure (median = 5.00, IQR = 2) and quality of care global measure (median = 6.0, IQR =1.00) were in the middle of the scale. The communication scale (median = 33.40, IQR = 9.77) was in the second lowest quintile indicating poor communication.

Comparisons of care coordination have been made based on general health (Figures 7.1 to 7.5, Tables 7.2 to 7.3), physical functioning (Figures 7.6 to 7.10, Tables 7.4 to 7.5), emotional well-being (Figures 7.11 to 7.15, Tables 7.6 to 7.7), social functioning, (Figures 7.16 to 7.20, Tables 7.8 to 7.9), hearing problems (Figures 7.21 to 7.25, Tables 7.10 to 7.11), eye problems (Figures 7.26 to 7.30, Tables 7.12 to 7.13), location (Figures 7.31 to 7.35, Tables 7.14 to 7.15), education (Figures 7.36 to 7.40, Table 7.16), and SEIFA (Figures 7.41 to 7.45, Tables 7.17 to 7.18).

Table 7.1: Summary statistics Total score - Communication and Navigation

Care coordination scale	Mean	SD	Median	IQR	Possible range
Communication*	33.40	9.77	36.00	4.00	13-65
Navigation*	22.28	5.27	21.50	4.50	7-35
Total score*	55.68	13.52	57.00	7.75	20-100
Care coordination global measure	4.76	2.36	5.00	2.00	1-10
Quality of care global measure	5.52	2.34	6.00	1.00	1-10

*Normal distribution, use mean and sd as central measure

Comparisons of Care Coordination scores by general health

Comparisons of Care Coordination scores were made between those that have higher general health and those that have lower general health. Boxplots for each of the care coordination scales are displayed in Figures 7.1 to 7.5. A two-sample t-test was used when assumptions for normality and variance were met (Table 7.2), or when assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 7.3). There were no statistically significant differences observed between groups for any Care Coordination scores.

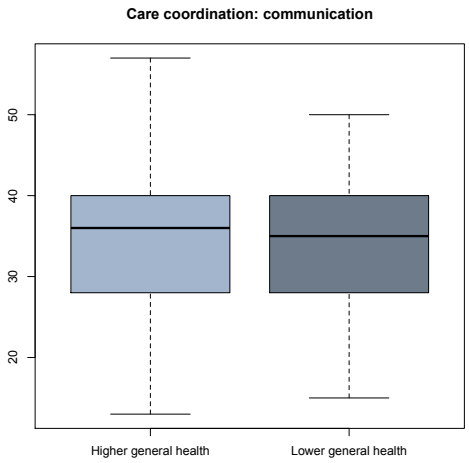


Figure 7.1: Boxplot of Care coordination: communication by general health

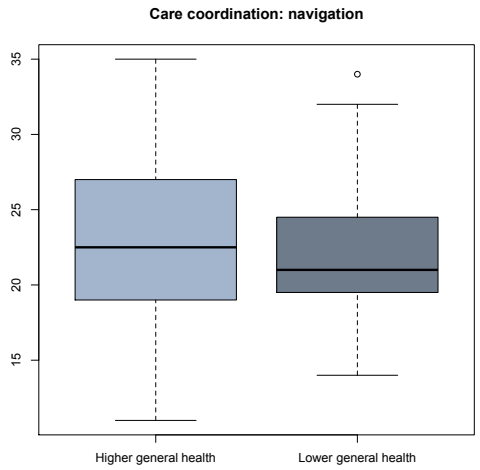


Figure 7.2: Boxplot of Care coordination: navigation by general health

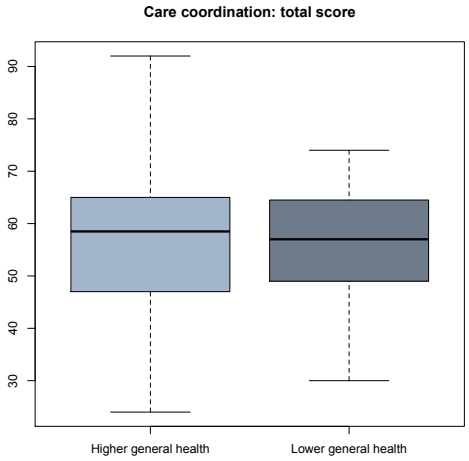


Figure 7.3: Boxplot of Care coordination: total score by general health

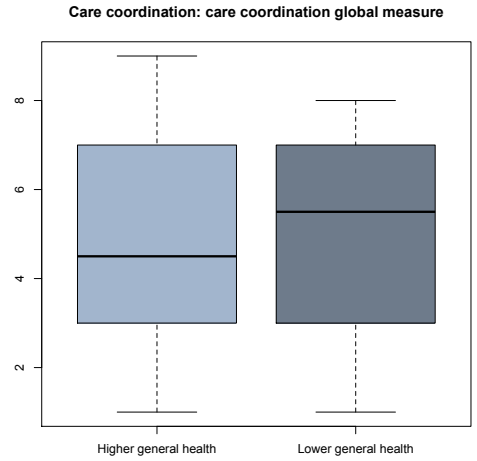


Figure 7.4: Boxplot of Care coordination: care coordination global measure by general health

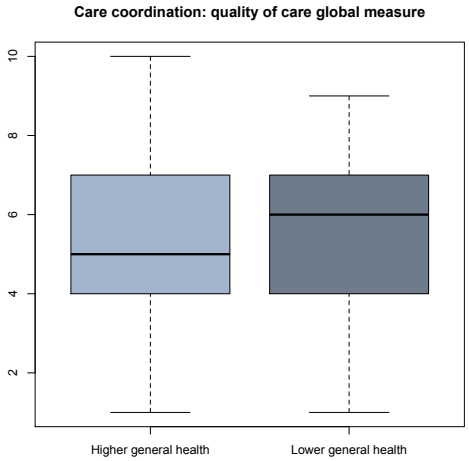


Figure 7.5: Boxplot of Care coordination: quality of care global measure by general health

Table 7.2: Summary statistics and Two sample t test by general health

Care coordination scale by general health	Group	Count	Mean	SD	t	dF	p
Communication	Higher general health	22	33.36	10.93	-0.02	48	0.9817
	Lower general health	28	33.43	8.96			
Navigation	Higher general health	22	22.86	6.13	0.69	48	0.4931
	Lower general health	28	21.82	4.55			
Total score	Higher general health	22	56.23	15.84	0.25	48	0.8026
	Lower general health	28	55.25	11.66			

Table 7.3: Summary statistics Wilcoxon rank sum test with continuity correction by general health

Care coordination scale by general health	Group	Count	Median	IQR	W	p
Care coordination global score	Higher general health	22	4.50	3.75	293	0.7746
	Lower general health	28	5.50	4.00		
Quality of care global score	Higher general health	22	5.00	3.00	290	0.7295
	Lower general health	28	6.00	2.50		

Comparisons of Care Coordination scores by physical functioning

Comparisons of Care Coordination scores were made between participants with higher physical functioning and those with lower physical functioning. Boxplots for each of the care coordination scales are displayed in Figures 7.6 to 7.10. A two-sample t-test was used when assumptions for normality and variance were met (Table 7.4), or when assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 7.5). There were no statistically significant differences observed between groups for any Care Coordination scores

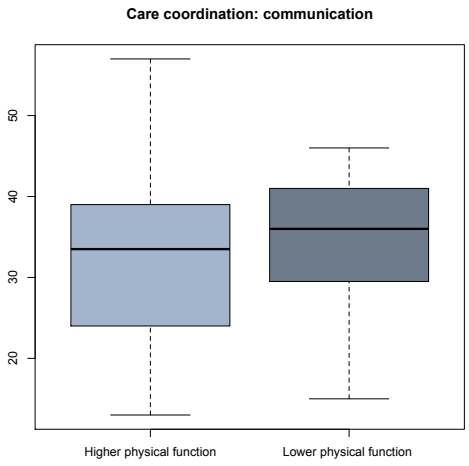


Figure 7.6: Boxplot of Care coordination: communication by physical functioning

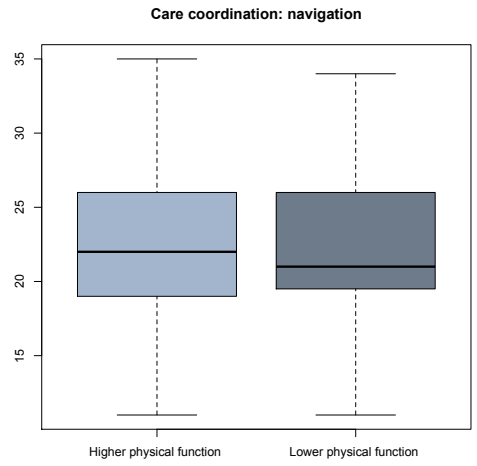


Figure 7.7: Boxplot of Care coordination: navigation by physical functioning

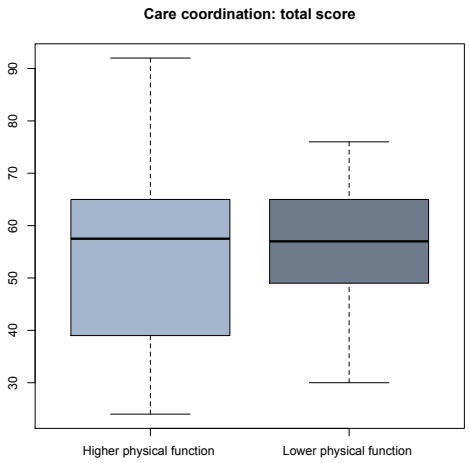


Figure 7.8: Boxplot of Care coordination: total score by physical functioning

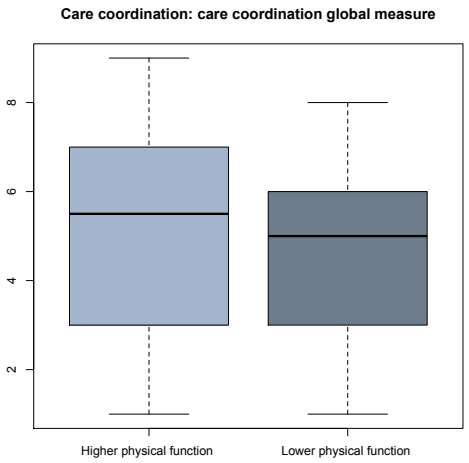


Figure 7.9: Boxplot of Care coordination: care coordination global measure by physical functioning

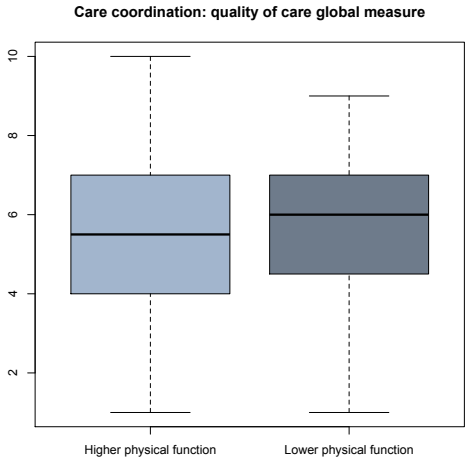


Figure 7.10: Boxplot of Care coordination: quality of care global measure by physical functioning

Table 7.4: Summary statistics and Two sample t test by physical functioning

Care coordination scale by physical functioning	Group	Count	Mean	SD	t	dF	p
Communication	Higher physical functioning	22	32.64	11.31	-0.49	48	0.6291
	Lower physical functioning	28	34.00	8.53			
Navigation	Higher physical functioning	22	22.27	5.19	-0.01	48	0.9932
	Lower physical functioning	28	22.29	5.42			
Total Score	Higher physical functioning	22	54.91	15.51	-0.35	48	0.7247
	Lower physical functioning	28	56.29	11.98			

Table 7.5: Summary statistics Wilcoxon rank sum test with continuity correction by physical functioning

Care coordination scale by physical functioning	Group	Count	Median	IQR	W	p
Care coordination global measure	Higher physical functioning	22	5.50	4.00	337.50	0.5668
	Lower physical functioning	28	5.00	3.00		
Care coordination quality of care global measure	Higher physical functioning	22	5.50	3.00	330.00	0.6709
	Lower physical functioning	28	6.00	2.25		

Comparisons of Care Coordination scores by emotional well-being

Comparisons of Care Coordination scores were made between those that have higher emotional well-being compared to those with lower emotional well-being. Boxplots for each of the care coordination scales are displayed in Figures 7.11 to 7.15. A two-sample t-test was used when assumptions for normality and variance were met (Table 7.6), or when assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 7.7). No statistically significant differences were observed between these two groups for any Care Coordination scores (Tables 7.11 and 7.12).

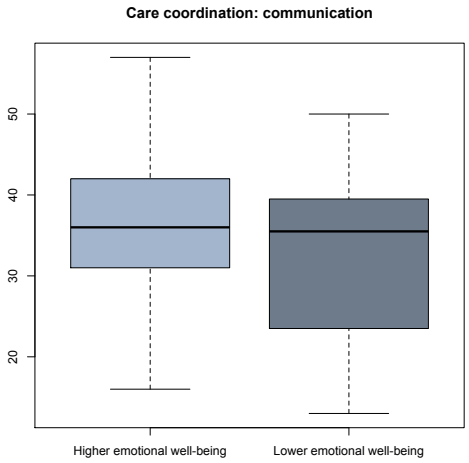


Figure 7.11: Boxplot of Care coordination: communication by social functioning

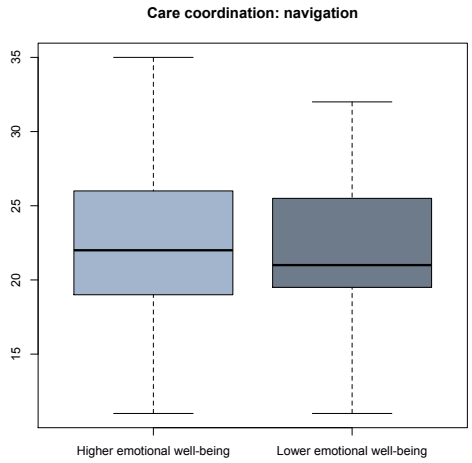


Figure 7.12: Boxplot of Care coordination: navigation by social functioning

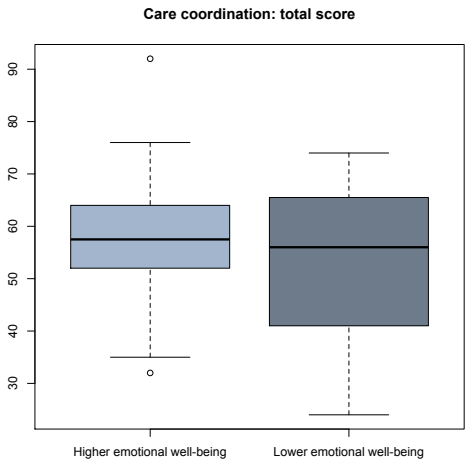


Figure 7.13: Boxplot of Care coordination: total score by emotional well-being

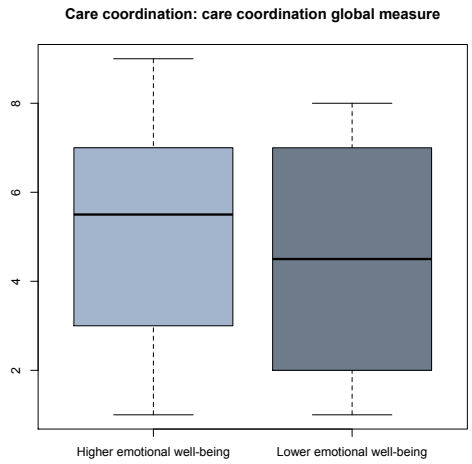


Figure 7.14: Boxplot of Care coordination: care coordination global measure by emotional well-being

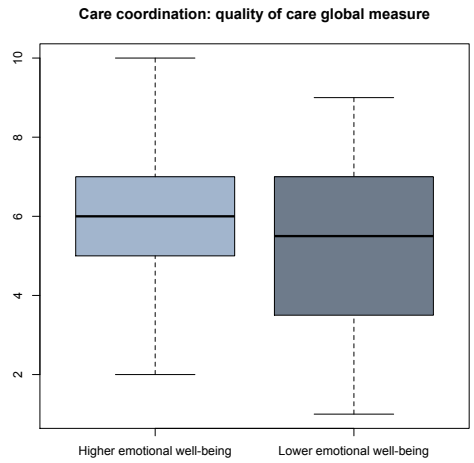


Figure 7.15: Boxplot of Care coordination: quality of care global measure by emotional well-being

Table 7.6: Summary statistics and Two sample t test by emotional well-being

Care coordination scale by emotional well-being	Group	Count	Mean	SD	t	dF	p
Communication	Higher emotional well-being	26	35.08	9.15	1.27	48	0.2097
	Lower emotional well-being	24	31.58	10.28			
Navigation	Higher emotional well-being	26	22.88	5.50	0.84	48	0.4039
	Lower emotional well-being	24	21.63	5.04			
Total score	Higher emotional well-being	26	57.96	12.54	1.25	48	0.2175
	Lower emotional well-being	24	53.21	14.36			

* Statistically significant at p<0.05

Table 7.7: Summary statistics Wilcoxon rank sum test with continuity correction by emotional well-being

Care coordination scale by emotional well-being	Group	Count	Median	IQR	W	p
Care coordination global measure	Higher emotional well-being	26	5.50	3.50	360.50	0.3462
	Lower emotional well-being	24	4.50	4.50		
Quality of care global measure	Higher emotional well-being	26	6.00	2.00	343.00	0.5493
	Lower emotional well-being	24	5.50	3.25		

Comparisons of Care Coordination scores by social functioning

Comparisons of Care Coordination scores were made between those that have higher social functioning compared to those with lower social functioning. Boxplots for each of the care coordination scales are displayed in Figures 7.16 to 7.20. A two-sample t-test was used when assumptions for normality and variance were met (Table 7.8), or when assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 7.9). A Wilcoxon rank sum test with continuity correction indicated a those with higher social functioning (Median = 25.00, IQR =5.25) had a significantly better outcome compared to those with lower social functioning (Median = 20.00, IQR = 3.00) for the Care coordination: Navigation scale [W=438.00, p=0.0063]. No other statistically significant differences were observed between these two groups for any Care Coordination scores.

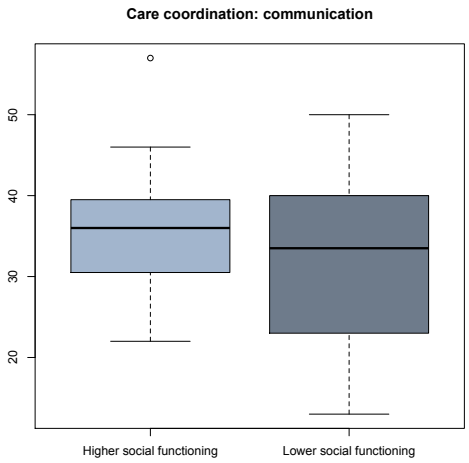


Figure 7.16: Boxplot of Care coordination: communication by social functioning

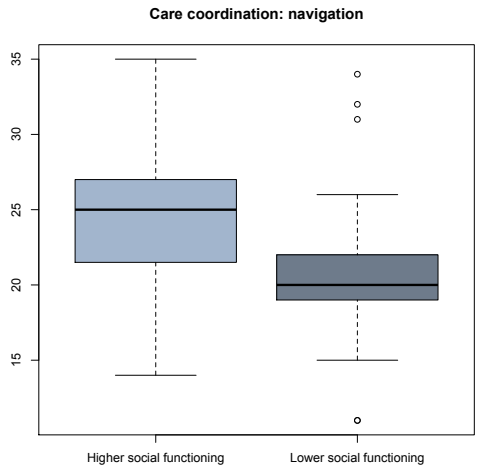


Figure 7.17: Boxplot of Care coordination: navigation by social functioning

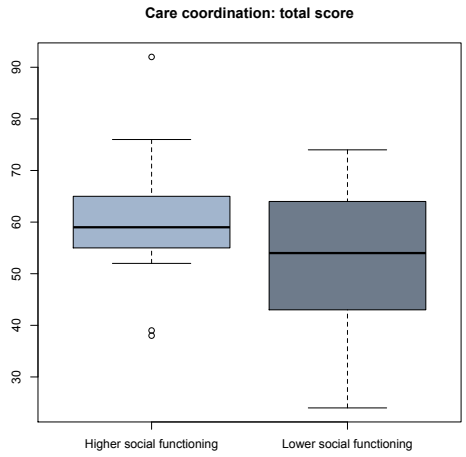


Figure 7.18: Boxplot of Care coordination: total score by social functioning

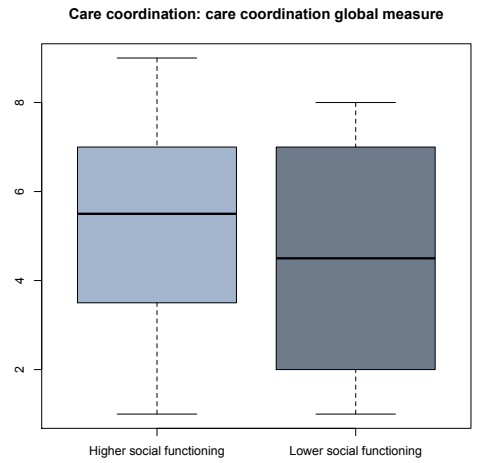


Figure 7.19: Boxplot of Care coordination: care coordination global measure by social functioning

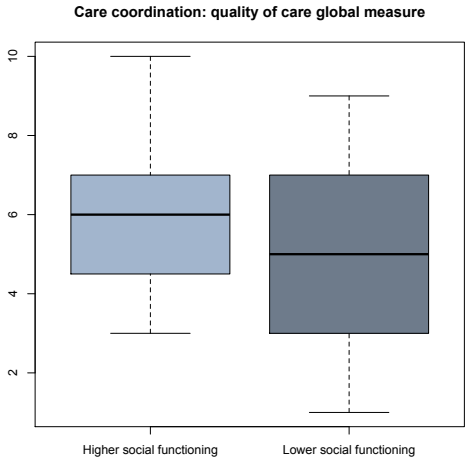


Figure 7.20: Boxplot of Care coordination: quality of care global measure by social functioning

Table 7.8: Summary statistics and Two sample t test by social functioning

Care coordination scale by emotional well-being	Group	Count	Median	IQR	W	p
Care coordination global measure	Higher emotional well-being	26	5.50	3.50	360.50	0.3462
	Lower emotional well-being	24	4.50	4.50		
Quality of care global measure	Higher emotional well-being	26	6.00	2.00	343.00	0.5493
	Lower emotional well-being	24	5.50	3.25		

Table 7.9: Summary statistics Wilcoxon rank sum test with continuity correction by metastatic status

Care coordination scale by social functioning	Group	Count	Median	IQR	W	p
Communication	Higher social functioning	20	36.00	8.50	349.00	0.3363
	Lower social functioning	30	33.50	16.25		
Navigation	Higher social functioning	20	25.00	5.25	438.00	0.0063*
	Lower social functioning	30	20.00	3.00		
Care coordination global measure	Higher social functioning	20	5.50	3.25	350.50	0.3170
	Lower social functioning	30	4.50	4.50		
Quality of care global measure	Higher social functioning	20	6.00	2.25	351.50	0.3072
	Lower social functioning	30	5.00	3.75		

* Statistically significant at $p<0.05$

Comparisons of Care Coordination scores by hearing problems

Comparisons of Care Coordination scores were made between those that have hearing problems and those that do not. Boxplots for each of the care coordination scales are displayed in Figures 7.21 to 7.25 A two-sample t-test was used when assumptions for normality and variance were met (Table 7.10), or when assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 7.11). No statistically significant differences were observed between these two groups for any Care Coordination scores.

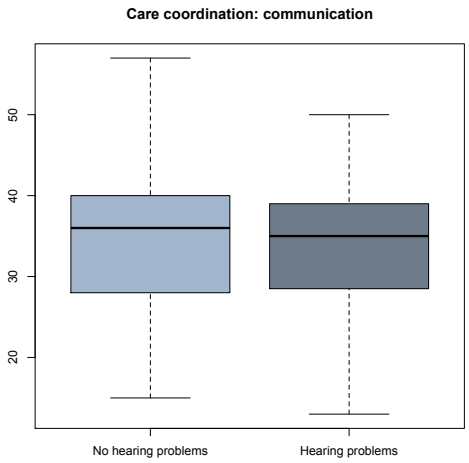


Figure 7.21: Boxplot of Care coordination: communication by hearing problems

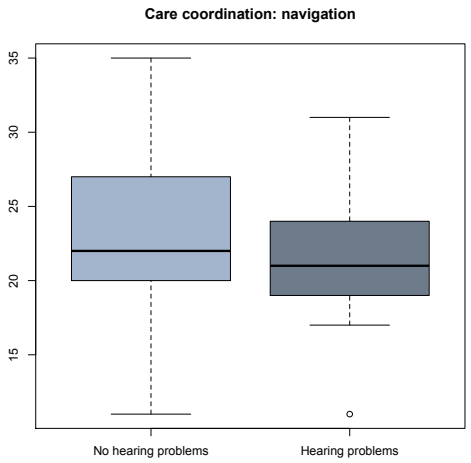


Figure 7.22: Boxplot of Care coordination: navigation by hearing problems

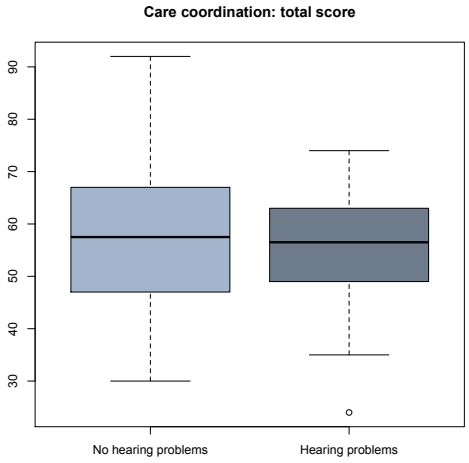


Figure 7.23: Boxplot of Care coordination: total score by hearing problems

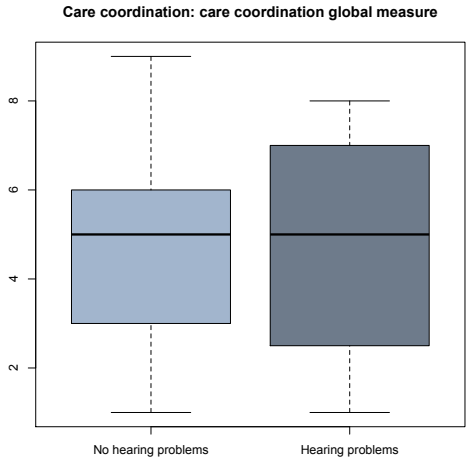


Figure 7.24: Boxplot of Care coordination: care coordination global measure by hearing problems

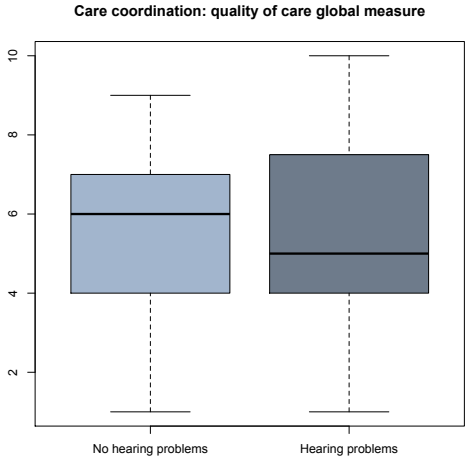


Figure 7.25: Boxplot of Care coordination: quality of care global measure by hearing problems

Table 7.10: Summary statistics and Two sample t test by hearing problems

Care coordination scales by hearing problems	Group	Count	Mean	SD	t	dF	p
Communication	No hearing problems	26	34.00	10.28	0.45	48	0.6559
	Hearing problems	24	32.75	9.36			
Total score	No hearing problems	26	56.88	14.79	0.65	48	0.5174
	Hearing problems	24	54.38	12.17			
Quality of care global measure	No hearing problems	26	5.50	2.21	-0.06	48	0.9506
	Hearing problems	24	5.54	2.52			

Table 7.11: Summary statistics Wilcoxon rank sum test with continuity correction by hearing problems

Care coordination scales by hearing problems	Group	Count	Median	IQR	W	p
Navigation	No hearing problems	26	22.00	6.75	354.00	0.4184
	Hearing problems	24	21.00	5.00		
Care coordination global measure	No hearing problems	26	5.00	2.75	333.50	0.6803
	Hearing problems	24	5.00	4.25		

Comparisons of Care Coordination scores by eye problems

Comparisons of Care Coordination scores were made between those that have eye problems and those that do not. Boxplots for each of the care coordination scales are displayed in Figures 7.26 to 7.30 A two-sample t-test was used when assumptions for normality and variance were met (Table 7.12), or when assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 7.13). No statistically significant differences were observed between these two groups for any Care Coordination scores.

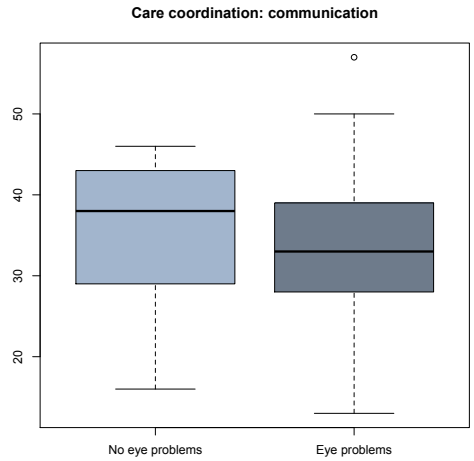


Figure 7.26: Boxplot of Care coordination: communication by eye problems

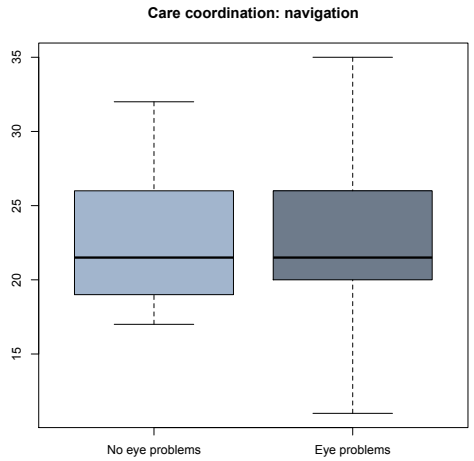


Figure 7.27: Boxplot of Care coordination: navigation by eye problems

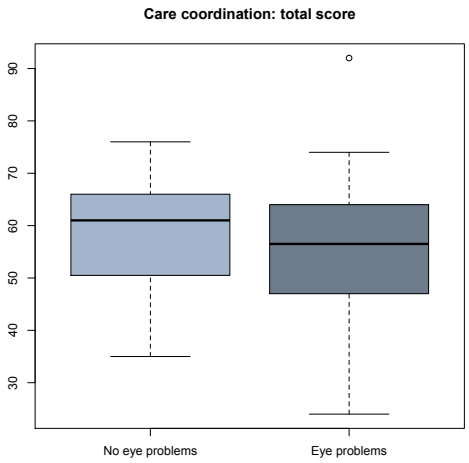


Figure 7.28: Boxplot of Care coordination: total score by eye problems

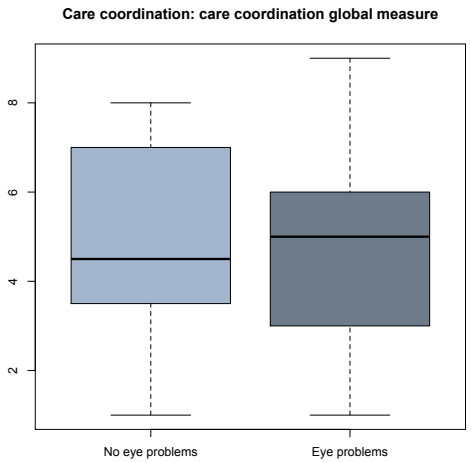


Figure 7.29: Boxplot of Care coordination: care coordination global measure by eye problems

Figure 7.30: Boxplot of Care coordination: quality of care global measure by eye problems

Table 7.12: Summary statistics and Two sample t test by eye problems

Care coordination scales by eye problems	Group	Count	Mean	SD	t	dF	p
Total score	No eye problems	16	57.94	12.34	0.81	48	0.4235
	Eye problems	34	54.62	14.08			
Quality of care global measure	No eye problems	16	5.88	2.16	0.73	48	0.4676
	Eye problems	34	5.35	2.44			

Table 7.3: Summary statistics Wilcoxon rank sum test with continuity correction by eye problems

Care coordination scales by eye problems	Group	Count	Median	IQR	W	p
Communication	No eye problems	16	38.00	13.50	338.00	0.1727
	Eye problems	34	33.00	10.50		
Navigation	No eye problems	16	21.50	7.00	277.50	0.9168
	Eye problems	34	21.50	6.00		
Care coordination global measure	No eye problems	16	4.50	3.25	290.50	0.7052
	Eye problems	34	5.00	3.00		

* Statistically significant at p<0.05

Comparisons of Care Coordination scores by location

Comparisons of Care Coordination scores were made between those that live in metropolitan areas compared to those that live in regional or rural areas. Boxplots for each of the care coordination scales are displayed in Figures 7.31 to 7.35 A two-sample t-test was used when assumptions for normality and variance were met (Table 7.14), or when assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 7.15). No statistically significant differences were observed between these two groups for any Care Coordination scores.

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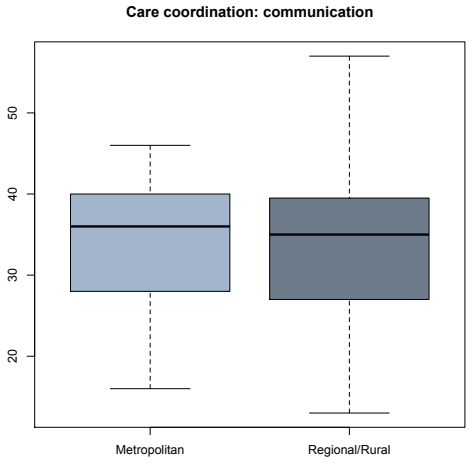


Figure 7.31: Boxplot of Care coordination: communication by location

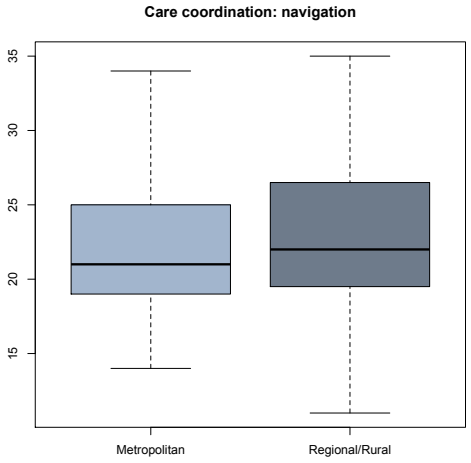


Figure 7.32: Boxplot of Care coordination: navigation by location

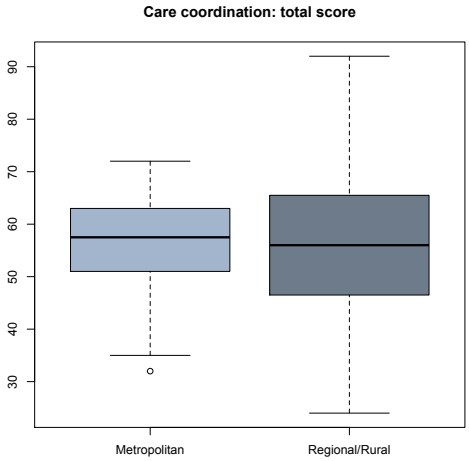


Figure 7.33: Boxplot of Care coordination: total score by location

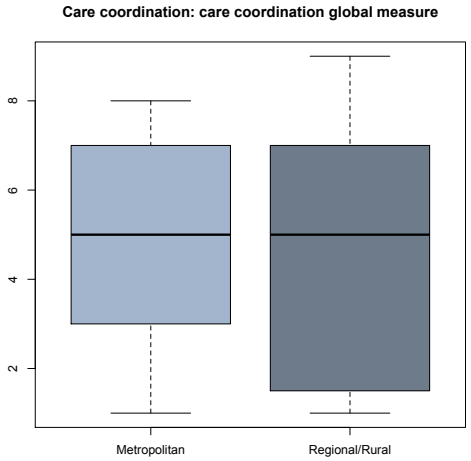


Figure 7.34: Boxplot of Care coordination: care coordination global measure by location

Figure 7.35: Boxplot of Care coordination: quality of care global measure by location

Table 7.14: Summary statistics and Two sample t test by location

Care coordination scales by location	Group	Count	Mean	SD	t	dF	p
Communication	Metropolitan	30	33.57	8.52	0.1463	48	0.8843
	Regional/Remote	20	33.15	11.63			
Navigation	Metropolitan	30	22.10	4.37	-0.293	48	0.7707
	Regional/Remote	20	22.55	6.51			

Table 7.15: Summary statistics Wilcoxon rank sum test with continuity correction by location

Care coordination scales by location	Group	Count	Median	IQR	W	p
Total score	Metropolitan	30	57.50	11.50	303.00	0.9605
	Regional/Remote	20	56.00	18.50		
Care coordination global measure	Metropolitan	30	5.00	3.50	333.00	0.5154
	Regional/Remote	20	5.00	5.25		
Quality of care global measure	Metropolitan	30	5.50	3.00	303.00	0.9601
	Regional/Remote	20	6.00	4.75		

Comparisons of Care Coordination scores by education

Comparisons of Care Coordination scores were made between those that have with university qualifications and those with high school or trade. Boxplots for each of the care coordination scales are displayed in Figures 7.36 to 7.40.

Assumptions for normality and variance were met (Table 7.16), a two-sample t-test was used to compare mean scores. No statistically significant differences were observed between these two groups for any Care Coordination scores.

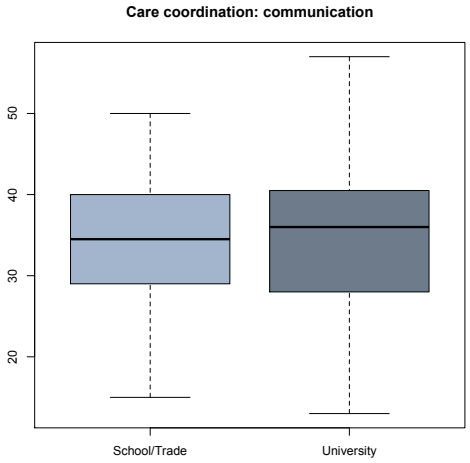


Figure 7.36: Boxplot of Care coordination: communication by education

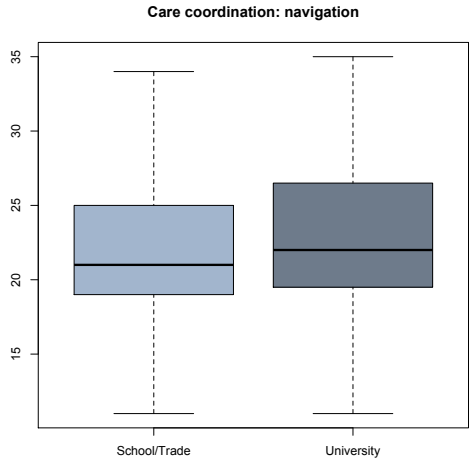
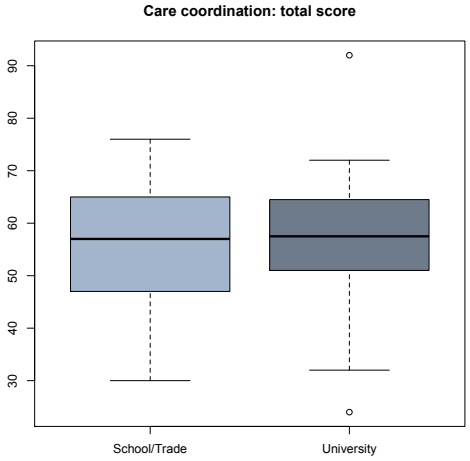


Figure 7.37: Boxplot of Care coordination: navigation by education



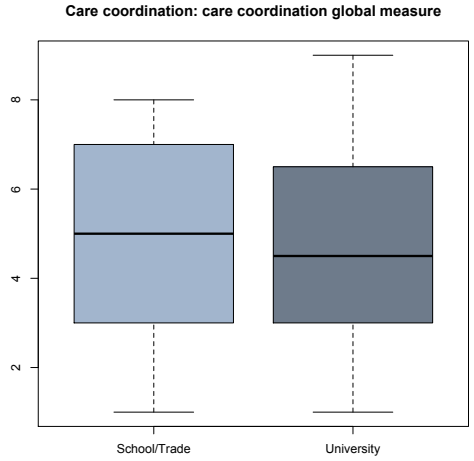


Figure 7.38: Boxplot of Care coordination: total score by education

Figure 7.39: Boxplot of Care coordination: care coordination global measure by education

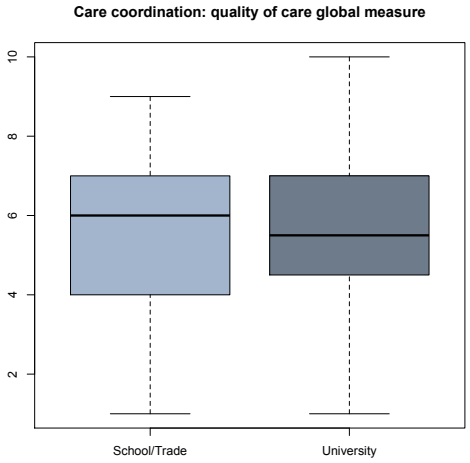


Figure 7.40: Boxplot of Care coordination: quality of care global measure by education

Table 7.16: Summary statistics and Two sample t test by education

Care coordination scales by education	Group	Count	Mean	SD	t	dF	p
Communication	School/Trade	14	33.38	9.14	-0.01	48	0.9909
	University	12	33.42	10.61			
Navigation	School	10	21.65	5.15	-0.87	48	0.3872
	University	8	22.96	5.42			
Total score	School	18	55.04	12.44	-0.35	48	0.7307
	University	16	56.38	14.83			
Care coordination global measure	School	26	4.92	2.17	0.50	48	0.6162
	University	24	4.58	2.59			
Quality of care global measure	School	22	5.38	2.40	-0.42	48	0.6748
	University	20	5.67	2.32			

Comparisons of Care Coordination scores by SEIFA

Comparisons of Care Coordination scores were made between those that have with higher SEIFA and those with lower SEIFA Boxplots for each of the care coordination scales are displayed in Figures 7.41 to 7.45. A two-sample t-test was used when assumptions for normality and variance were met (Table 7.17), or when assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 7.18). No statistically significant differences were observed between these two groups for any Care Coordination scores.

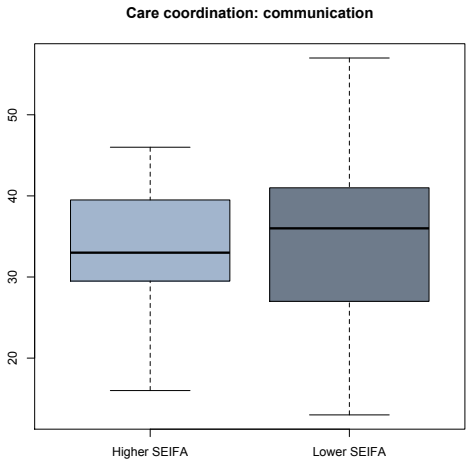


Figure 7.41: Boxplot of Care coordination: communication by SEIFA

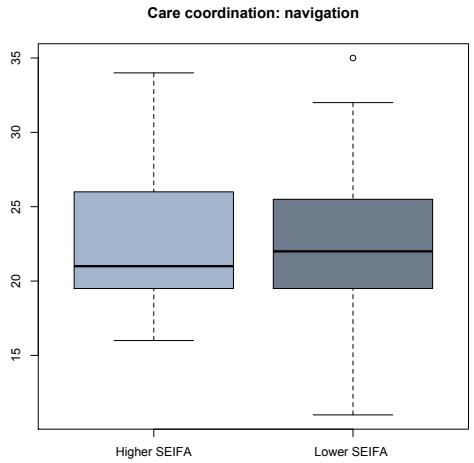


Figure 7.42: Boxplot of Care coordination: navigation by SEIFA

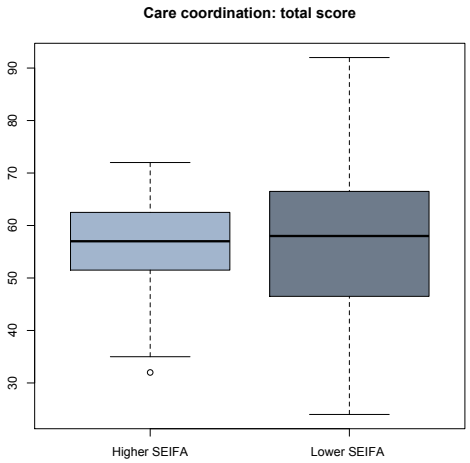


Figure 7.43: Boxplot of Care coordination: total score by SEIFA

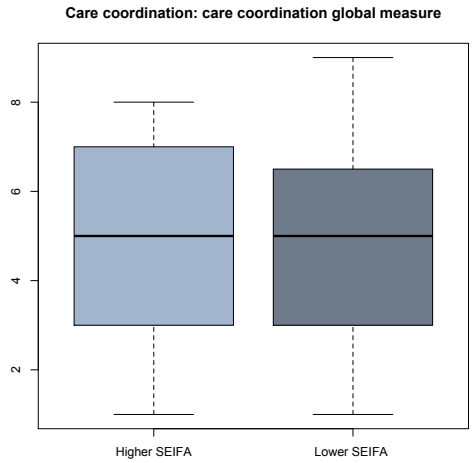


Figure 7.44: Boxplot of Care coordination: care coordination global measure by SEIFA

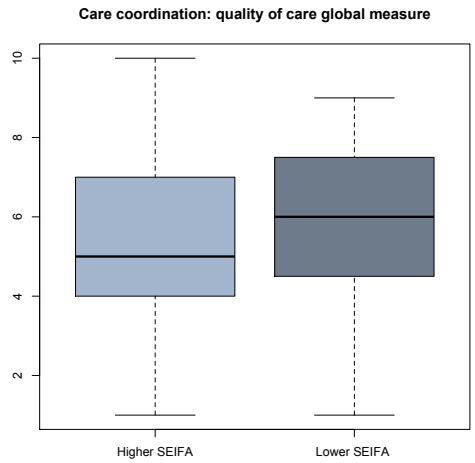


Figure 7.45: Boxplot of Care coordination: quality of care global measure by SEIFA

Table 7.17: Summary statistics and Two sample t test by SEIFA

Care coordination scales by SEIFA	Group	Count	Mean	SD	t	dF	P
Communication	Higher SEIFA	27	33.00	8.28	-0.31	48	0.7573
	Lower SEIFA	23	33.87	11.45			

Table 7.18: Summary statistics Wilcoxon rank sum test with continuity correction by SEIFA

Care coordination scales by SEIFA	Group	Count	Median	IQR	W	p
Navigation	Higher SEIFA	27	21.00	6.50	308	0.9688
	Lower SEIFA	23	22.00	6.00		
Total score	Higher SEIFA	27	57.00	11.00	297.5	0.8076
	Lower SEIFA	23	58.00	20.00		
Care coordination global measure	Higher SEIFA	27	5.00	4.00	326.5	0.7604
	Lower SEIFA	23	5.00	3.50		
Quality of care global measure	Higher SEIFA	27	5.00	3.00	261.5	0.3398
	Lower SEIFA	23	6.00	3.00		

Experience of care and support

Participants were asked what care and support they had received throughout their experience. This question aims to investigate what services patients consider to be support and care services. The most common description of care and support was in the form of domestic and home care support from government services and NDIS (n=14, 28.00%), this was followed by participants describing that they did not receive any care and support in general (n =9, 18.00%) and not receiving significant support and care from the clinical setting (n=9, 18.00%). There were also seven participants (14.00%) that described receiving support from family and friends.

Participant describes support and care in the form of domestic and home care support from government services and NDIS

Other than that, because of my actual condition or my condition, I have NDIS, so I have somebody who comes in twice a week, so it's things like going grocery shopping, because I find that quite difficult, so that to me is care and support. Pretty much that's it, but I think that covers me for what I need. Participant 7

We started a while ago having paying someone to clean house. Just recently, I've gone on to the NDIS which there are a few hiccups, but I'm being given, supposedly given funding for a lot more of that sort of thing. It's still just starting but it's not really happened yet, but it's progressing hopefully, so that's good. Participant 34

I've got NDIS funding which I've been trying to utilize. They tell you off for not using enough money then when you may need to use it, they won't let you use it. Participant 38

Participant describes not receiving any care and support

Not a bit, nothing. Participant 3

No. None. Participant 10

Nothing. Not that I can think. I can't really think of the time. Participant 11

Participant describes not receiving significant support and care from the clinical setting

I'm supposed to have, what do you call it? Occupational therapy. It had never happened since I've been out of hospital. I'm supposed to do that. A lot of things supposed to happen didn't happen. Participant 1

I just see the doctor on a regular basis but just there are no other real support. Participant 19

Sure, easy. Zero…I do think that the system could be a little better when it does come to remote people. When we do head to LOCATION or somewhere maybe they could be, I don't know, maybe there could be a better way of dealing with particularly young adolescent people transferring to the adult system. That they don't fall through the holes that we have, that their appointments are possibly grouped together with the number of specialists that are not available to us in this area. Because they don't see them all the time. NAME can present very well in the half hour or hour appointment that she's there for by getting through there and getting from there and then at the end of that day, oh my gosh, she doesn't do well at all. Participant 49

In relation to sub-group variations, participants with high social functioning (30.00%) describes not receiving any care and support more frequently than the general population (18.00%)

Table 7.19: Perceptions of care and support received

Care and support received	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant describes support and care in the form of domestic and home care support from government services and NDIS	14	28.00	8	26.67	6	30.00	8	29.63	6	26.09
Participant describes not receiving any care and support	9	18.00	7	23.33	2	10.00	6	22.22	3	13.04
Participant describes not receiving significant support and care from the clinical setting	9	18.00	4	13.33	5	25.00	4	14.81	5	21.74
Participant describes support and care from family friends (general)	7	14.00	4	13.33	3	15.00	3	11.11	4	17.39
Participant describes support and care in the form of community health services (Neurological service)	6	12.00	4	13.33	2	10.00	3	11.11	3	13.04
Participant describes receiving additional care and support for allied health services	6	12.00	5	16.67	2	10.00	4	14.81	3	13.04
Participant describes support and care in the form of family or friends to help with transport(to appointments and everyday activties)	5	10.00	4	13.33	1	5.00	3	11.11	2	8.70
Participant describes support and care in the form of help with visual impairment (Vision Australia)	5	10.00	2	6.67	3	15.00	2	7.41	3	13.04
Participant describes satisfaction with accessing support and assistance from the AMDF	5	10.00	3	10.00	2	10.00	4	14.81	1	4.35
Participant describes support and care in the form of connecting with other patients and sharing their experience	4	8.00	3	10.00	1	5.00	2	7.41	2	8.70
Participant describes support and care in the form of family and friends helping with domestic help	4	8.00	2	6.67	3	15.00	2	7.41	3	13.04

Care and support received	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant describes support and care in the form of domestic and home care support from government services and NDIS	14	28.00	7	26.92	7	29.17	7	29.17	9	26.47
Participant describes not receiving any care and support	9	18.00	7	26.92	2	8.33	3	12.50	9	26.47
Participant describes not receiving significant support and care from the clinical setting	9	18.00	5	19.23	4	16.67	6	25.00	7	20.59
Participant describes support and care from family friends (general)	7	14.00	3	11.54	4	16.67	3	12.50	5	14.71
Participant describes support and care in the form of community health services (Neurological service)	6	12.00	3	11.54	3	12.50	4	16.67	4	11.76
Participant describes receiving additional care and support for allied health services	6	12.00	3	11.54	4	16.67	3	12.50	4	11.76
Participant describes support and care in the form of family or friends to help with transport(to appointments and everyday activties)	5	10.00	1	3.85	4	16.67	3	12.50	2	5.88
Participant describes support and care in the form of help with visual impairment (Vision Australia)	5	10.00	2	7.69	3	12.50	3	12.50	3	8.82
Participant describes satisfaction with accessing support and assistance from the AMDF	5	10.00	2	7.69	3	12.50	4	16.67	4	11.76
Participant describes support and care in the form of connecting with other patients and sharing their experience	4	8.00	2	7.69	2	8.33	1	4.17	4	11.76
Participant describes support and care in the form of family and friends helping with domestic help	4	8.00	3	11.54	2	8.33	2	8.33	2	5.88

Care and support received	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant describes support and care in the form of domestic and home care support from government services and NDIS	14	28.00	4	18.18	10	35.71	7	26.92	7	29.17
Participant describes not receiving any care and support	9	18.00	6	27.27	3	10.71	4	15.38	5	20.83
Participant describes not receiving significant support and care from the clinical setting	9	18.00	6	27.27	3	10.71	5	19.23	4	16.67
Participant describes support and care from family friends (general)	7	14.00	4	18.18	3	10.71	2	7.69	5	20.83
Participant describes support and care in the form of community health services (Neurological service)	6	12.00	3	13.64	3	10.71	3	11.54	3	12.50
Participant describes receiving additional care and support for allied health services	6	12.00	2	9.09	5	17.86	4	15.38	3	12.50
Participant describes support and care in the form of family or friends to help with transport(to appointments and everyday activties)	5	10.00	1	4.55	4	14.29	2	7.69	3	12.50
Participant describes support and care in the form of help with visual impairment (Vision Australia)	5	10.00	2	9.09	3	10.71	3	11.54	2	8.33
Participant describes satisfaction with accessing support and assistance from the AMDF	5	10.00	4	18.18	1	3.57	2	7.69	3	12.50
Participant describes support and care in the form of connecting with other patients and sharing their experience	4	8.00	2	9.09	2	7.14	4	15.38	0	0.00
Participant describes support and care in the form of family and friends helping with domestic help	4	8.00	2	9.09	3	10.71	1	3.85	4	16.67

Care and support received	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant describes support and care in the form of domestic and home care support from government services and NDIS	14	28.00	5	25.00	9	30.00	5	22.73	9	32.14
Participant describes not receiving any care and support	9	18.00	6	30.00	3	10.00	3	13.64	6	21.43
Participant describes not receiving significant support and care from the clinical setting	9	18.00	3	15.00	6	20.00	6	27.27	3	10.71
Participant describes support and care from family friends (general)	7	14.00	2	10.00	5	16.67	3	13.64	4	14.29
Participant describes support and care in the form of community health services (Neurological service)	6	12.00	1	5.00	5	16.67	3	13.64	3	10.71
Participant describes receiving additional care and support for allied health services	6	12.00	3	15.00	4	13.33	3	13.64	4	14.29
Participant describes support and care in the form of family or friends to help with transport(to appointments and everyday activties)	5	10.00	0	0.00	5	16.67	3	13.64	2	7.14
Participant describes support and care in the form of help with visual impairment (Vision Australia)	5	10.00	1	5.00	4	13.33	1	4.55	4	14.29
Participant describes satisfaction with accessing support and assistance from the AMDF	5	10.00	4	20.00	1	3.33	2	9.09	3	10.71
Participant describes support and care in the form of connecting with other patients and sharing their experience	4	8.00	3	15.00	1	3.33	2	9.09	2	7.14
Participant describes support and care in the form of family and friends helping with domestic help	4	8.00	2	10.00	3	10.00	2	9.09	3	10.71

Care and support received	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant describes support and care in the form of domestic and home care support from government services and NDIS	14	28.00	0	0.00	2	14.29	3	33.33	4	36.36	5	50.00
Participant describes not receiving any care and support	9	18.00	0	0.00	3	21.43	2	22.22	3	27.27	1	10.00
Participant describes not receiving significant support and care from the clinical setting	9	18.00	1	16.67	2	14.29	3	33.33	1	9.09	2	20.00
Participant describes support and care from family friends (general)	7	14.00	1	16.67	1	7.14	1	11.11	2	18.18	2	20.00
Participant describes support and care in the form of community health services (Neurological service)	6	12.00	2	33.33	1	7.14	1	11.11	1	9.09	1	10.00
Participant describes receiving additional care and support for allied health services	6	12.00	3	50.00	2	14.29	0	0.00	1	9.09	1	10.00
Participant describes support and care in the form of family or friends to help with transport(to appointments and everyday activties)	5	10.00	0	0.00	2	14.29	0	0.00	1	9.09	2	20.00
Participant describes support and care in the form of help with visual impairment (Vision Australia)	5	10.00	0	0.00	1	7.14	3	33.33	1	9.09	0	0.00
Participant describes satisfaction with accessing support and assistance from the AMDF	5	10.00	0	0.00	2	14.29	1	11.11	1	9.09	1	10.00
Participant describes support and care in the form of connecting with other patients and sharing their experience	4	8.00	1	16.67	1	7.14	2	22.22	0	0.00	0	0.00
Participant describes support and care in the form of family and friends helping with domestic help	4	8.00	1	16.67	1	7.14	1	11.11	1	9.09	1	10.00

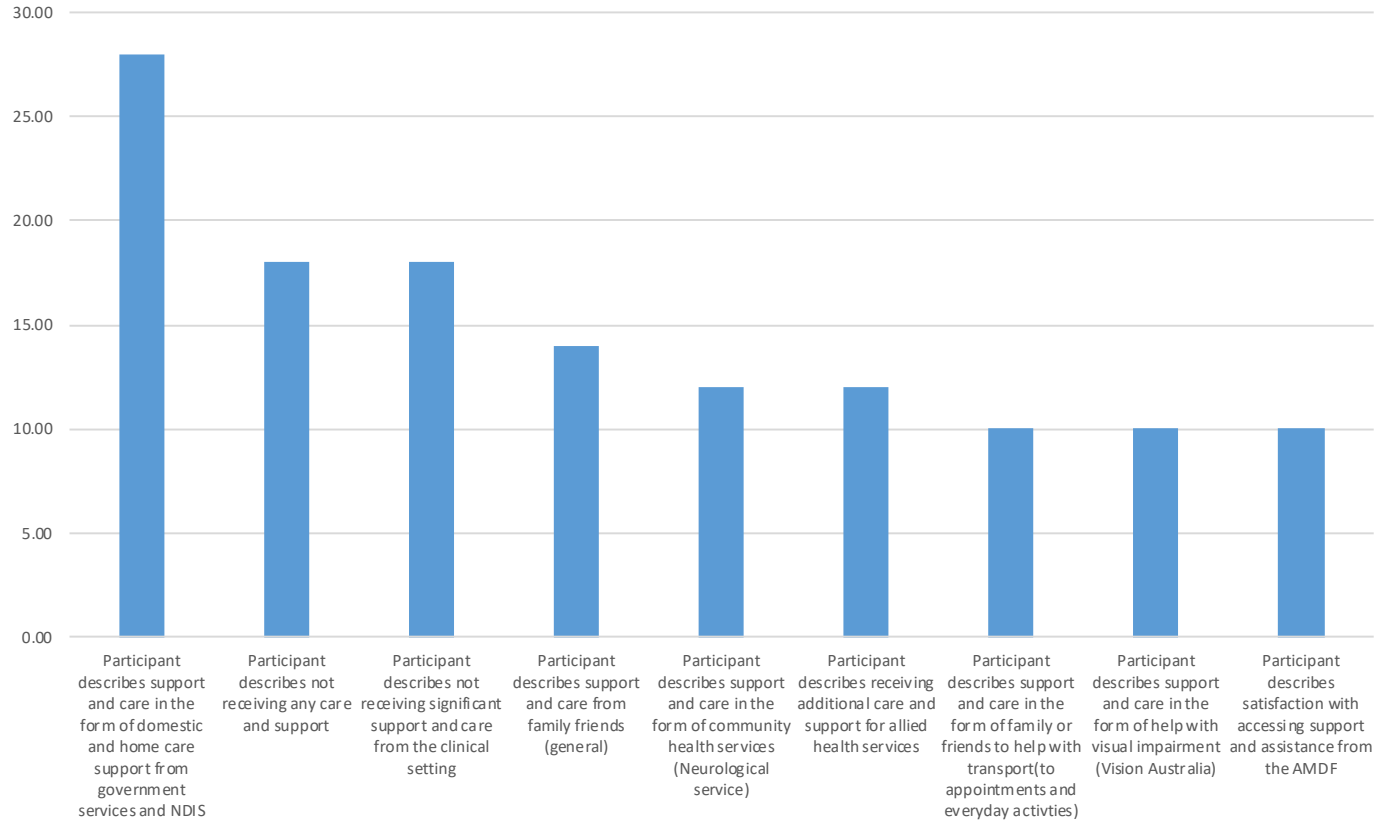


Figure 7.46: Perceptions of care and support received (% of all participants)

Section 8 Quality of life

Section 8: Experience of quality of life

Quality of life

- The most common impact on quality of life described by participants was poor mental health as a consequence of mitochondrial disease (n=19, 38.00%). There were also eight participants (16.00%) that noted poor mental health of family or friends (as carers) as a consequence of the disease. This was followed by a significant impact on family relationships and family dynamics (n=16, 32.00%) and withdrawing from activities with family and friends due to physical limitations (n=16, 32.00%). There were 13 participants (26.00%) that spoke about the need to access mental health services to maintain their quality of life, 12 participants (24.00%) that described that having days where physical limitations can be frustrating and eight participants (16.00%) that described limitations in travelling.
- In relation to sub-group variations, participants from metropolitan areas (26.67%) and participants with high physical function (27.27%) reported limitations of freedom to travel more frequently than the general population (16.00%), while participants from rural areas reported this less frequently (5.00%). Participants from rural areas (25.00%) described pleasure with maintaining hobbies and activities to overcome feelings of sadness or depression, more frequently than the general population (14.00%). Participants from low socio-economic areas (26.09%) and participants with a university degree (25.00%) described inability to participate in workforce to their level of expectation due to Mitochondrial disease, more frequently than the general population (14.00%). Participants with high school or trade education (34.62%) and those with high physical functioning (36.36%) reported having some days where physical limitations can be frustrating, more frequently than the general population (24.00%). Participants with high physical function (22.73%) reported little or no impact on family or friends' quality of life more frequently than the general population (12.00%).

Regular activities to maintain health

- The most common regular activity needed to maintain health reported by participants was having adequate rest to minimise fatigue (n=21, 42.00%). This was followed by having regular exercise (n=15, 30.00%) and eating a healthy/modified diet (n=10, 20.00%). There were seven participants (14.00%) that described taking prescription medication, six participants (12.00%) that considered taking supplements as an activity to maintain health and six participants (12.00%) that reported maintaining hobbies and activities in support of good mental health.
- In relation to sub-group variations, participants from low socio-economic areas (56.52%), those with high social function (60.00%) and low general health (57.14%) reported ensuring they have adequate rest to minimise fatigue, more frequently than the general population (42.00%). Participants with high physical function (50.00%) reported having regular exercise more frequently than the general population (30.00%) while those with low physical function (14.29%) and low general health (17.86%) reported this less frequently.

Impact on relationships

- The most common theme described by participants was a negative impact on personal relationships due to people withdrawing from relationships or not being able to understand (n=14, 28.00%) and this was followed by a negative impact on personal relationships due to social isolation (n=11, 22.00%). The next most common theme was a negative impact on personal relationships due to not being able to do all activities with family and friends (n=10, 20.00%). There were six participants (12.00%) that described a positive impact of strengthening relationships.
- In relation to sub-group variations, participants from low socio-economic areas (43.48%) and those with low social function (40.00%) reported a negative impact on personal relationships due to people withdrawing from relationships or not being able to understand, more frequently than the general population (28.00%), while those from high socio-economic areas (14.81) and high social function (10.00%) reported this less frequently. Participants from metropolitan areas (33.33%) and those with low emotional well-being (33.33%) reported a negative impact on personal relationships due to social isolation, more frequently than the general population (22.00%) while those from rural areas (10.00%) reported this less

frequently. Participants from low socio-economic areas (30.43%) described a negative impact on personal relationships due to not being able to do all activities with family and friends, more frequently than the general population (20.00%). Participants with high physical function (27.27%) and high social function (35.00%) described no impact on personal relationships more frequently than the general population (16.00%), while those with low social function described this less frequently.

- Participants were also asked if their condition caused any additional burden on their family. The most common theme was there was an additional burden on family, but the participant did not articulate a specific reason why there was a burden (n=13, 26.00%). The next most common theme was there was a burden due to needing help with transport and driving due to vision impairment (n=8, 16.00%), followed by participants describing that there was no additional burden, that it is just part of their life as they know it (n=7, 14.00%).
- In relation to sub-group variations, participants with a university education (37.50%), those with low physical function (39.29%) and those with low social function (33.33%) reported there being an additional burden (no additional information) more frequently than the general population (26.00%), while those with high physical function (9.09%) and those with high social function (15.00%) reported this less frequently.

Anxiety and fear of progression

- The Fear of Progression questionnaire measures the level of anxiety people experience in relation to their conditions. The Fear of Progression questionnaire comprises a total score, with a higher score denoting increased anxiety. Overall the entire cohort had a median total score of 34.10, which is a score in the middle of the scale.

Fear of progression – by general health

- There was no difference observed in the fear of progression total score between participants that had higher general health compared to those with lower general health.

Fear of progression – by physical functioning

- There was no difference observed in the fear of progression total score between participants that had higher physical functioning compared to those with lower physical functioning.

Fear of progression – by emotional well-being

- There was no difference observed in the fear of progression total score between participants that had higher emotional well-being compared to those with lower emotional well-being.

Fear of progression – by social functioning

- There was no difference observed in the fear of progression total score between participants that had higher social functioning compared to those with lower social functioning.

Fear of progression – by hearing problems

- There was no difference observed in the fear of progression total score between participants that had hearing problems compared to those with no hearing problems.

Fear of progression – by eye problems

- There was no difference observed in the fear of progression total score between participants that had eye problems compared to those with no eye problems.

Fear of progression – by hearing problems

- There was no difference observed in the fear of progression total score between participants that had hearing problems compared to those with no hearing problems.

Fear of progression – by location

- There was no difference observed in the fear of progression total score between participants that live in metropolitan areas and those that live in regional or rural areas.

Fear of progression – by level of education

- There was no difference observed in the fear of progression total score between participants with university qualifications and those with high school or trade certificates.

Fear of progression – by SEIFA

- There was no difference observed in the fear of progression total score between participants that live in an area with a higher SEIFA score and those that live in a lower SEIFA score.

Experience of quality of life

Participants were asked whether their condition has had an impact on their quality of life. The most common impact on quality of life described by participants was poor mental health as a consequence of mitochondrial disease (n=19, 38.00%). There were also eight participants (16.00%) that noted poor mental health of family or friends (as carers) as a consequence of the disease.

Participant describes poor mental health of patient as a consequence of the disease

Then there's also just a constant feeling in the back of my head that I'm being held down by this and held back a little bit. Sometimes it gets difficult. I feel a lot lower these days. Probably the worst thing is thinking that or knowing that it's going to get worse. It's never really going to get better unless some miracle treatment comes out but overall, quality of life is still very good. Participant 11

At first, in my head, it was devastating but not in my husband's head. I suffered gradually from depression. Do occasionally but I've got a really good therapist, who I still to talk to. That has made things better. It's really important that people with a disease have a therapist. Participant 12

It does affect my mental health. I feel restricted sometimes and I feel like people have to look after me a bit. I see a psychiatrist regularly and I take antidepressant medication. It's frustrating because sometimes I would like to be out and about doing more interesting things, but I need to stay home and be quiet. The times when I'm particularly tired and I just need to rest, those have a fairly depressing quality. Participant 43

This was followed by a significant impact on family relationships and family dynamics (n=16, 32.00%) and withdrawing from activities with family and friends due to physical limitations (n=16, 32.00%).

Participant describes a significant impact on family relationships and family dynamics

Well, the kids are always saying, "You don't come around and play with us anymore, Dad." Basically, family. I can't do that. I used to run around and play around with my other son and pass balls to him and kick. Participant 6

It impacts upon us 24/7. We always have to have back-up plans and think ahead to ensure that we are covered should something happen or change suddenly. My whole family is affected. Participant 30

Yes, I think it's affected everything. My relationship with my husband and with my children, really, really serious. Participant 34

Participant describes withdrawing from activities with family and friends due to physical limitations and the subsequent emotional response

For me, it's affected it terribly. My life stopped stopped….. I had to stop work then. I was studying. I was working in the pharmacy and studying to be a dispense tech. I just got so unreliable that I just had to quit. I don't even go out anymore. It's because I never know how I'm going to be. Obviously, if it's a wedding or something, I'll push through regardless of how I feel, but just to go the ball club or something on a Friday night for the raffles, I normally don't go. Because I can be fine when I leave home and half an hour after I get there, feel like absolute hell, so I just normally don't go. Participant 18

Sometimes, I wonder about those things because I really do have a huge problem with being in groups. I can't process language, group language. I've never been great. I've been able to do it and major part of my job was being able to do it. I don't mind that I don't do those things anymore. A lot of people think it's become pretty odd, I think, because I don't like gatherings or drinks or parties or whatever. I most certainly do family gatherings, and they're usually very big, but they don't de-stress me quite as much in that people will know that basically-- Well, I don't know if they know. That answer was slightly to the left of it, if you know what I mean. Participant 20

Social life is absolutely non-existent. I don't have the energy to do anything. I don't go out or I go out with a few friends once a week especially to a church meeting type of thing. I've got to get on till three until I'm pumped for feeding at night, it puts down me or anything else. It just stops everything, anything you wanted to do. You just don't have the energy to do anything. Participant 38

Massively. She lost all her friends having to move from school to home-schooling. It's created more anxiety around social interaction now because it was the wrong age to have to be taken out of school and stuff like that. Participant 49

There were 13 participants (26.00%) that spoke about the need to access mental health services to maintain their quality of life, 12 participants (24.00%) that described that having days where physical limitations can be frustrating and eight participants (16.00%) that described limitations in travelling.

Participant describes accessing mental health services to maintain high quality of life

In terms of taking care of my mental and emotional health, recently I've started to see a psychologist again, just to make sure that I have all the tools to ensure my wellbeing, my mental health, is kept as healthy as possible, given the current situation. I meditate, and I do things that relax me, but I also think, for my mental health, given my personality, I'm a social justice activist, so something that I'm passionate about actually helps improve my mental health. Participant 7

Again, over life it's been different times, but it certainly has affected my mental health severely, probably 15 years ago. Over quite a few years, or really 10 years, working with a psychologist and other healthcare professionals at times and also-- it was a psychiatrist as well at times on things, I'm working with my GP, we've addressed those mental health issues which are kind of separate to the mitochondrial disease. I guess once they're addressed then you can do a lot. Participant 13

I have no one to talk to so much about it. I now go to a counsellor once a month which is absolutely essential. That's what I do to keep sane, I go and see the counsellor. He had three changes of business, but it's totally understood when she goes into business that whenever she leaves she takes me with her. That's an understanding with her and her bosses that I go wherever she goes, because of having found someone I can talk to and who helps me keep sane, I stay with her. That's what I do. Participant 21

Participant describes having some days where physical limitations can be frustrating

Pretty telling question that one, isn't it? I try not to think about it. Like I said before, I get lactic acid build up very quickly in my legs or what feels like lactic acid in my legs very, very quickly, two flights of stairs. My office is on the second floor. When the lift aren't working, it's like, "Do I really have to do this?" You're constantly looking-- If I'm walking from one part of the city to the other, I'll try and walk the way that's the most flat even if it's a bit longer, so I don't have to walk up any hills because it's uncomfortable. The doctor at the LOCATION the other week when I was there said it was time to get back into the gym. I have got an issue with my shoulder at the moment that I'm getting some treatment on, and once that's fixed up I'll get back into the gym. I always have to psych myself up for that because I know that once I start I'm going to be in constant pain again because as I said before, in my past life when I've been body building and the like, you know you're going to do arms, back, chest and legs. Whatever you're doing, it'll hurt for a couple of days. but the next four or five days you going to do another part of your body, so it's not hurting for a few days by the time you on to the next one. I'm dreading the fact that whatever I do the pain may now last for five, six, seven, eight days, which means there's going to be no respite because by the time it starts feel better, I'm already on to that part body. That's an impact on my being. Participant 2

Okay. Probably two aspects. It stopped me doing some of the things that I love, as all the side effects from it, which is part of it, I guess. It's more the things that I haven't because of the mito. I can't play golf anymore, I'm too tired after nine rounds, and I love that. At least some of the things that you love, so it's like my sport, you know, seems like with the hearing loss that I stopped playing hockey. I can't walk long distances. The effect with the hearing loss has also affected my balance, especially at night. I'm in trouble walking at night, or just falling over, you know? That's ridiculous. Participant 15

I realize I can't work as much as what I used to. If I clean the whole house for a day, I need to rest, do nothing for the next one, two days to get my energy back. It's hard for me to get up early in the morning. I do have insomnia as well sometimes. Sometimes I'll sleep to ten o'clock, eleven o'clock. I just can't get up. I want to get up earlier, but I can't. I'll set the alarm early. I do get energy-less very easy. Participant 22

Every day is a challenge for NAME as far as he can't dress himself, toilet himself, feed himself, he needs to have assistance with that. He needs to get help getting into his wheelchair, I put him into the car and I him to school. I collect him daily, he needs assistance with showering every night. He needs help with day to day tasks, things that everybody just takes basically for granted that you do on your own. It's like having a toddler basically for NAME. Participant 50

In relation to sub-group variations, participants from metropolitan areas (26.67%) and participants with high physical function (27.27%) reported limitations of freedom to travel more frequently than the general population (16.00%), while participants from rural areas reported this less frequently (5.00%). Participants from rural areas (25.00%) described pleasure with maintaining hobbies and activities to overcome feelings of sadness or depression, more frequently than the general population (14.00%). Participants from low socio-economic areas (26.09%) and participants with a university degree (25.00%) described inability to participate in workforce to their level of expectation due to Mitochondrial disease, more frequently than the general population (14.00%).

Participants with high school or trade education (34.62%) and those with high physical functioning (36.36%) reported having some days where physical limitations can be frustrating, more frequently than the general population (24.00%). Participants with high physical function (22.73%) reported little or no impact on family or friends' quality of life more frequently than the general population (12.00%).

Table 8.1: Quality of life

Impact on quality of life	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant describes poor mental health of patient as a consequence of the disease	19	38.00	11	36.67	8	40.00	10	37.04	9	39.13
Participant describes a significant impact on family relationships and family dynamics	16	32.00	10	33.33	7	35.00	10	37.04	7	30.43
Participant describes withdrawing from activities with family and friends due to physical limitations	16	32.00	10	33.33	6	30.00	8	29.63	8	34.78
Participant describes accessing mental health services to maintain high QOL	13	26.00	6	20.00	7	35.00	6	22.22	7	30.43
Participant describes having some days where physical limitations can be frustrating	12	24.00	9	30.00	4	20.00	6	22.22	7	30.43
Participant describes the limitations of freedom to travel	8	16.00	8	26.67	1	5.00	6	22.22	3	13.04
Participant describes poor mental health of family or friends as carers as a consequence of the disease	8	16.00	4	13.33	4	20.00	4	14.81	4	17.39
Participant describes pleasure with maintaining hobbies and activities to overcome feelings of sadness or depression	7	14.00	2	6.67	5	25.00	4	14.81	3	13.04
Participant describes inability to participate in workforce to their level of expectation due to Mitochondrial disease	7	14.00	4	13.33	4	20.00	2	7.41	6	26.09
Participant describes little or no impact for family or friends in relation to quality of life	6	12.00	5	16.67	1	5.00	3	11.11	3	13.04
Participant describes no real affect on quality of life of patient	5	10.00	2	6.67	3	15.00	3	11.11	2	8.70
Participant describes feelings of sadness and/or frustration with experiencing episodes of illness or injury	5	10.00	3	10.00	2	10.00	3	11.11	2	8.70

Impact on quality of life	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant describes poor mental health of patient as a consequence of the disease	19	38.00	8	30.77	11	45.83	11	45.83	13	38.24
Participant describes a significant impact on family relationships and family dynamics	16	32.00	10	38.46	7	29.17	8	33.33	10	29.41
Participant describes withdrawing from activities with family and friends due to physical limitations	16	32.00	10	38.46	6	25.00	8	33.33	14	41.18
Participant describes accessing mental health services to maintain high QOL	13	26.00	5	19.23	8	33.33	7	29.17	7	20.59
Participant describes having some days where physical limitations can be frustrating	12	24.00	9	34.62	4	16.67	7	29.17	10	29.41
Participant describes the limitations of freedom to travel	8	16.00	5	19.23	4	16.67	4	16.67	6	17.65
Participant describes poor mental health of family or friends as carers as a consequence of the disease	8	16.00	4	15.38	4	16.67	5	20.83	6	17.65
Participant describes pleasure with maintaining hobbies and activities to overcome feelings of sadness or depression	7	14.00	5	19.23	2	8.33	4	16.67	5	14.71
Participant describes inability to participate in workforce to their level of expectation due to Mitochondrial disease	7	14.00	2	7.69	6	25.00	4	16.67	5	14.71
Participant describes little or no impact for family or friends in relation to quality of life	6	12.00	3	11.54	3	12.50	1	4.17	5	14.71
Participant describes no real affect on quality of life of patient	5	10.00	4	15.38	1	4.17	2	8.33	4	11.76
Participant describes feelings of sadness and/or frustration with experiencing episodes of illness or injury	5	10.00	2	7.69	3	12.50	1	4.17	3	8.82

Impact on quality of life	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant describes poor mental health of patient as a consequence of the disease	19	38.00	6	27.27	13	46.43	11	42.31	8	33.33
Participant describes a significant impact on family relationships and family dynamics	16	32.00	6	27.27	11	39.29	7	26.92	10	41.67
Participant describes withdrawing from activities with family and friends due to physical limitations	16	32.00	7	31.82	9	32.14	10	38.46	6	25.00
Participant describes accessing mental health services to maintain high QOL	13	26.00	5	22.73	8	28.57	4	15.38	9	37.50
Participant describes having some days where physical limitations can be frustrating	12	24.00	8	36.36	5	17.86	6	23.08	7	29.17
Participant describes the limitations of freedom to travel	8	16.00	6	27.27	3	10.71	6	23.08	3	12.50
Participant describes poor mental health of family or friends as carers as a consequence of the disease	8	16.00	4	18.18	4	14.29	4	15.38	4	16.67
Participant describes pleasure with maintaining hobbies and activities to overcome feelings of sadness or depression	7	14.00	1	4.55	6	21.43	5	19.23	2	8.33
Participant describes inability to participate in workforce to their level of expectation due to Mitochondrial disease	7	14.00	4	18.18	4	14.29	3	11.54	5	20.83
Participant describes little or no impact for family or friends in relation to quality of life	6	12.00	5	22.73	1	3.57	4	15.38	2	8.33
Participant describes no real affect on quality of life of patient	5	10.00	3	13.64	2	7.14	4	15.38	1	4.17
Participant describes feelings of sadness and/or frustration with experiencing episodes of illness or injury	5	10.00	1	4.55	4	14.29	1	3.85	4	16.67

Impact on quality of life	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant describes poor mental health of patient as a consequence of the disease	19	38.00	5	25.00	14	46.67	7	31.82	12	42.86
Participant describes a significant impact on family relationships and family dynamics	16	32.00	6	30.00	11	36.67	8	36.36	9	32.14
Participant describes withdrawing from activities with family and friends due to physical limitations	16	32.00	5	25.00	11	36.67	7	31.82	9	32.14
Participant describes accessing mental health services to maintain high QOL	13	26.00	4	20.00	9	30.00	3	13.64	10	35.71
Participant describes having some days where physical limitations can be frustrating	12	24.00	6	30.00	7	23.33	6	27.27	7	25.00
Participant describes the limitations of freedom to travel	8	16.00	5	25.00	4	13.33	5	22.73	4	14.29
Participant describes poor mental health of family or friends as carers as a consequence of the disease	8	16.00	3	15.00	5	16.67	5	22.73	3	10.71
Participant describes pleasure with maintaining hobbies and activities to overcome feelings of sadness or depression	7	14.00	3	15.00	4	13.33	3	13.64	4	14.29
Participant describes inability to participate in workforce to their level of expectation due to Mitochondrial disease	7	14.00	3	15.00	5	16.67	3	13.64	5	17.86
Participant describes little or no impact for family or friends in relation to quality of life	6	12.00	3	15.00	3	10.00	4	18.18	2	7.14
Participant describes no real affect on quality of life of patient	5	10.00	3	15.00	2	6.67	3	13.64	2	7.14
Participant describes feelings of sadness and/or frustration with experiencing episodes of illness or injury	5	10.00	0	0.00	5	16.67	2	9.09	3	10.71

Impact on quality of life	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant describes poor mental health of patient as a consequence of the disease	19	38.00	2	33.33	9	64.29	1	11.11	4	36.36	3	30.00
Participant describes a significant impact on family relationships and family dynamics	16	32.00	3	50.00	3	21.43	2	22.22	5	45.45	4	40.00
Participant describes withdrawing from activities with family and friends due to physical limitations	16	32.00	2	33.33	2	14.29	2	22.22	7	63.64	3	30.00
Participant describes accessing mental health services to maintain high QOL	13	26.00	2	33.33	2	14.29	3	33.33	4	36.36	2	20.00
Participant describes having some days where physical limitations can be frustrating	12	24.00	2	33.33	3	21.43	5	55.56	1	9.09	2	20.00
Participant describes the limitations of freedom to travel	8	16.00	2	33.33	3	21.43	2	22.22	2	18.18	0	0.00
Participant describes poor mental health of family or friends as carers as a consequence of the disease	8	16.00	2	33.33	2	14.29	1	11.11	2	18.18	1	10.00
Participant describes pleasure with maintaining hobbies and activities to overcome feelings of sadness or depression	7	14.00	1	16.67	1	7.14	2	22.22	2	18.18	1	10.00
Participant describes inability to participate in workforce to their level of expectation due to Mitochondrial disease	7	14.00	1	16.67	3	21.43	0	0.00	4	36.36	0	0.00
Participant describes little or no impact for family or friends in relation to quality of life	6	12.00	0	0.00	2	14.29	1	11.11	1	9.09	2	20.00
Participant describes no real affect on quality of life of patient	5	10.00	1	16.67	2	14.29	0	0.00	1	9.09	1	10.00
Participant describes feelings of sadness and/or frustration with experiencing episodes of illness or injury	5	10.00	1	16.67	1	7.14	1	11.11	1	9.09	1	10.00

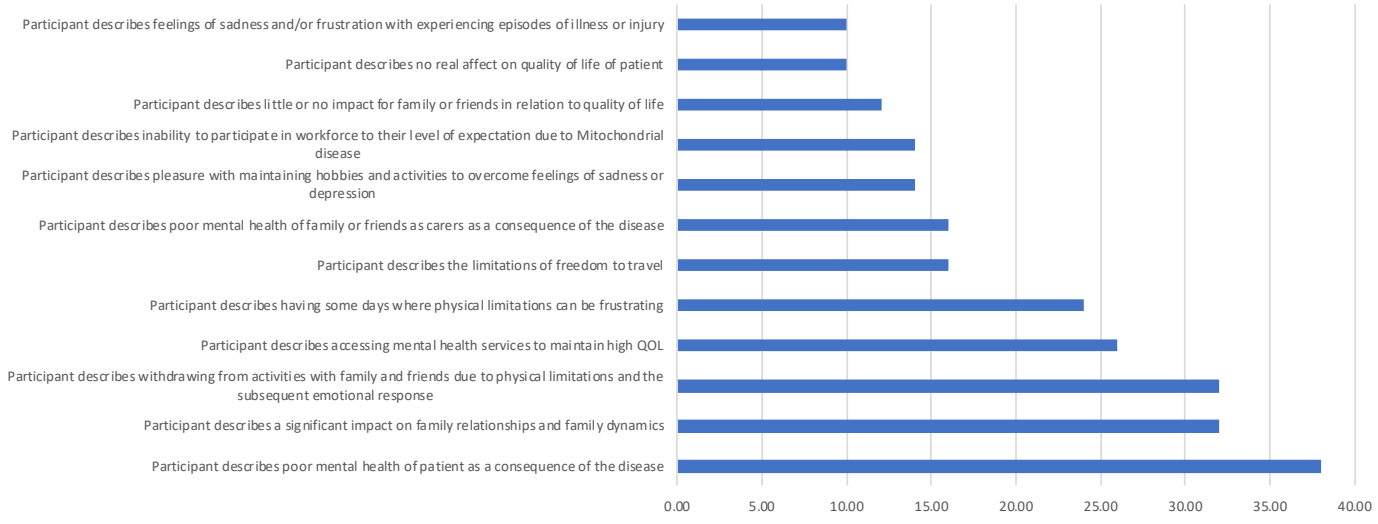


Figure 8.1: Quality of life (% of all participants)

Everyday activities to manage health

Participants were asked what some of the things are that they needed to do regularly to maintain their health. The most common regular activity needed to maintain health reported by participants was having adequate rest to minimise fatigue (n=21, 42.00%).

Participants reports ensuring they have adequate rest to minimise fatigue

Sleep. [laughs] Sleep a lot, rest whenever I need to rest, go to chiro and masseuse monthly. Be flexible, in terms of how I might be social. If I need someone to come to me then I do that, if I need to take an afternoon nap in order to spend time with my friends, I'll do that. Participant 5

Come this time of the afternoon, where I normally am, I'll lay down for two hours. I don't sleep because I won't during the night, but I'll take time out to watch a bit of TV and catch up on a bit of paperwork and all that sort of stuff, regenerate. Participant 16

I've just got to stop. I've got to rest. As I said, if I'm walking, I've got to stop and really only long enough for the body to build a bit more energy again then I'll take off again. No, I don't think I do anything special for that. Participant 31

This was followed by having regular exercise (n=15, 30.00%) and eating a healthy/modified diet (n=10, 20.00%).

Participant reports completing regular exercise to increase energy levels

Diet, exercise has been incredibly important. Being more vigorous about exercise now than I probably was. I was doing the fair bit but really making sure that I keep up with this even if I don't want to. Diet and getting enough rest if I need to, being vigilant about that yes. Participant 3

I also try to do a bit of cardio every now and then because I noticed that after doing cardio something like consecutive few days after that, I feel like I have a bit more energy than usual. I try to keep the fitness up. Participant 11

Exercise. Mild exercise. Walking with my dog. Participant 20

I have to be meticulously doing stretches generally after coffee in the morning before I try and walk because of balance and muscles working it's really important for me. Participant 27

Participant describes eating a healthy and/or modified diet

I'm on a very strict diet. I've lost a huge number of teeth so I cannot chew things. It's a very soft-based diet. It has to be very soft-based anyway because I can't swallow properly because the muscles are all gone in my throat. It's very limited what I can do. Participant 12

Whether it's just taking time out, not doing housework for the day or making sure that I'm monitoring my blood sugar if I feel a bit out of it or making sure that I know what's going on whether it's because by inch in a level, whether it's because I haven't eaten properly. Just to try to make minimal changes and making sure I eat frequently, don't fast, make sure I get a regular sleep pattern. Just little things that I can do in a day, make sure I take medication. Participant 26

Trying to eat healthy, is also a little bit of a challenge. And yeah, that's about it. Participant 40

There were seven participants (14.00%) that described taking prescription medication, six participants (12.00%) that considered taking supplements as an activity to maintain health and six participants (12.00%) that reported maintaining hobbies and activities in support of good mental health.

Participant describes taking prescription medication on a daily basis

My body's completely changed because I have no muscle around my intestines. I constantly have to take very large amounts of laxatives being under a gastroenterologist who treats me like a paraplegic. Participant 12

Just little things that I can do in a day, make sure I take medication. Participant 26

Rest regularly, eat well, drink a lot, take medications, use an electric wheelchair to conserve energy, constantly monitor my health status...dysautonomia, epilepsy, dysphagia, etcetera. Participant 30

Participant describes taking supplements on a daily basis

CoQ10. Like I said that really helps with energy levels. Participant 11

Trying to have maybe some vitamins to help that regard. I guess that would be something that would help. Participant 29

It's basically just taking the medication and taking the vitamins to get me through the day, from day to day and trying to have enough sleep. (exercise) I don't exercise enough. [laughs]. Participant 35

Participant reports maintain hobbies and activities in support on good mental health

Eat chocolate. [chuckles] Totally, totally against all the rules, but it's something that helps keep me centred sane and totally needed. I also involved in a quilting club, a walking club. I need that distraction. My daughter quilt so it's something that we do in common. I don't know. It's just you've got to keep your mind away from disease. You can't live with disease all the time so do other things, keep your mind off it. Yes. I think that's it. Participant 21

I only do voluntary work so it's not like it's paid work or anything. It's just otherwise I'd go mad if I was at home by myself all the time. Participant 38

Also, she loves to study, so she's doing a Uni course and she's…I mean, that course is limited to doing two units, which she should be doing one because she's getting exhausted all the time. It's having that outward focus and living your life that way. So always having something that you're working towards that's really important. Participant 49

In relation to sub-group variations, participants from low socio-economic areas (56.52%), those with high social function (60.00%) and low general health (57.14%) reported ensuring they have adequate rest to minimise fatigue, more frequently than the general population (42.00%). Participants with high physical function (50.00%) reported having regular exercise more frequently than the general population (30.00%) while those with low physical function (14.29%) and low general health (17.86%) reported this less frequently.

Table 8.2: Everyday activities to manage health

Regular activities to maintain health	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participants reports ensuring they have adequate rest to minimise fatigue	**21**	42.00	13	43.33	9	45.00	9	33.33	13	**56.52**
Participant reports having regular exercise to increase energy levels	**15**	30.00	10	33.33	5	25.00	10	37.04	5	21.74
Participant describes eating a healthy and/or modified diet	**10**	20.00	6	20.00	4	20.00	6	22.22	4	17.39
Participant describes taking prescribed medication on a daily basis	**7**	14.00	3	10.00	4	20.00	3	11.11	4	17.39
Participant describes taking supplements on a daily basis	**6**	12.00	4	13.33	2	10.00	5	18.52	1	4.35
Participant reports maintaining hobbies and activities in support of good mental health	**6**	12.00	5	16.67	1	5.00	3	11.11	3	13.04
Participants reports no modification to behaviour or diet- just takes each day as it comes	**5**	10.00	2	6.67	3	15.00	2	7.41	3	13.04
Participant reports requiring home care or family providing care 24/7	**5**	10.00	4	13.33	1	5.00	3	11.11	2	8.70
Participant recommends minimising daily activities or using the spoon theory to accomplish all necessary requirements of a daily or weekly basis	**5**	10.00	3	10.00	2	10.00	3	11.11	2	8.70
Participant reports that monitoring health to ensure continuous good health	**5**	10.00	2	6.67	3	15.00	2	7.41	3	13.04
Participant reports maintaining a positive frame of mind	**4**	8.00	4	13.33	0	0.00	2	7.41	2	8.70

Regular activities to maintain health	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participants reports ensuring they have adequate rest to minimise fatigue	**21**	42.00	12	46.15	10	41.67	10	41.67	14	41.18
Participant reports having regular exercise to increase energy levels	**15**	30.00	6	23.08	9	37.50	9	37.50	9	26.47
Participant describes eating a healthy and/or modified diet	**10**	20.00	4	15.38	6	25.00	4	16.67	7	20.59
Participant describes taking prescribed medication on a daily basis	**7**	14.00	2	7.69	5	20.83	5	20.83	5	14.71
Participant describes taking supplements on a daily basis	**6**	12.00	4	15.38	2	8.33	4	16.67	4	11.76
Participant reports maintaining hobbies and activities in support of good mental health	**6**	12.00	4	15.38	2	8.33	3	12.50	4	11.76
Participants reports no modification to behaviour or diet- just takes each day as it comes	**5**	10.00	5	19.23	0	0.00	0	0.00	5	14.71
Participant reports requiring home care or family providing care 24/7	**5**	10.00	2	7.69	3	12.50	2	8.33	4	11.76
Participant recommends minimising daily activities or using the spoon theory to accomplish all necessary requirements of a daily or weekly basis	**5**	10.00	0	0.00	5	20.83	3	12.50	4	11.76
Participant reports that monitoring health to ensure continuous good health	**5**	10.00	2	7.69	3	12.50	3	12.50	4	11.76
Participant reports maintaining a positive frame of mind	**4**	8.00	2	7.69	2	8.33	3	12.50	3	8.82

Regular activities to maintain health	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participants reports ensuring they have adequate rest to minimise fatigue	**21**	42.00	10	45.45	12	42.86	11	42.31	11	45.83
Participant reports having regular exercise to increase energy levels	**15**	30.00	11	50.00	4	14.29	10	38.46	5	20.83
Participant describes eating a healthy and/or modified diet	**10**	20.00	6	27.27	4	14.29	5	19.23	5	20.83
Participant describes taking prescribed medication on a daily basis	**7**	14.00	2	9.09	5	17.86	3	11.54	4	16.67
Participant describes taking supplements on a daily basis	**6**	12.00	4	18.18	2	7.14	4	15.38	2	8.33
Participant reports maintaining hobbies and activities in support of good mental health	**6**	12.00	2	9.09	4	14.29	4	15.38	2	8.33
Participants reports no modification to behaviour or diet- just takes each day as it comes	**5**	10.00	0	0.00	5	17.86	0	0.00	5	20.83
Participant reports requiring home care or family providing care 24/7	**5**	10.00	2	9.09	3	10.71	1	3.85	4	16.67
Participant recommends minimising daily activities or using the spoon theory to accomplish all necessary requirements of a daily or weekly basis	**5**	10.00	1	4.55	4	14.29	4	15.38	1	4.17
Participant reports that monitoring health to ensure continuous good health	**5**	10.00	1	4.55	4	14.29	3	11.54	2	8.33
Participant reports maintaining a positive frame of mind	**4**	8.00	3	13.64	1	3.57	2	7.69	2	8.33

Regular activities to maintain health	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participants reports ensuring they have adequate rest to minimise fatigue	**21**	42.00	12	60.00	10	33.33	6	27.27	16	57.14
Participant reports having regular exercise to increase energy levels	**15**	30.00	6	30.00	9	30.00	10	45.45	5	17.86
Participant describes eating a healthy and/or modified diet	**10**	20.00	5	25.00	5	16.67	4	18.18	6	21.43
Participant describes taking prescribed medication on a daily basis	**7**	14.00	1	5.00	6	20.00	1	4.55	6	21.43
Participant describes taking supplements on a daily basis	**6**	12.00	3	15.00	3	10.00	2	9.09	4	14.29
Participant reports maintaining hobbies and activities in support of good mental health	**6**	12.00	2	10.00	4	13.33	2	9.09	4	14.29
Participants reports no modification to behaviour or diet- just takes each day as it comes	**5**	10.00	0	0.00	5	16.67	0	0.00	5	17.86
Participant reports requiring home care or family providing care 24/7	**5**	10.00	1	5.00	4	13.33	2	9.09	3	10.71
Participant recommends minimising daily activities or using the spoon theory to accomplish all necessary requirements of a daily or weekly basis	**5**	10.00	2	10.00	3	10.00	1	4.55	4	14.29
Participant reports that monitoring health to ensure continuous good health	**5**	10.00	1	5.00	4	13.33	2	9.09	3	10.71
Participant reports maintaining a positive frame of mind	**4**	8.00	2	10.00	2	6.67	1	4.55	3	10.71

Regular activities to maintain health	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participants reports ensuring they have adequate rest to minimise fatigue	**21**	42.00	3	50.00	8	57.14	3	33.33	3	27.27	5	50.00
Participant reports completing regular exercise Participant reports having regular exercise to increase energy levels	**15**	30.00	2	33.33	5	35.71	1	11.11	4	36.36	3	30.00
Participant describes eating a healthy and/or modified diet	**10**	20.00	0	0.00	4	28.57	3	33.33	2	18.18	1	10.00
Participant describes taking prescribed medication on a daily basis	**7**	14.00	1	16.67	3	21.43	2	22.22	1	9.09	0	0.00
Participant describes taking supplements on a daily basis	**6**	12.00	1	16.67	2	14.29	1	11.11	1	9.09	1	10.00
Participant reports maintaining hobbies and activities in support of good mental health	**6**	12.00	1	16.67	2	14.29	1	11.11	1	9.09	1	10.00
Participants reports no modification to behaviour or diet- just takes each day as it comes	**5**	10.00	0	0.00	1	7.14	0	0.00	3	27.27	1	10.00
Participant reports requiring home care or family providing care 24/7	**5**	10.00	0	0.00	3	21.43	1	11.11	1	9.09	0	0.00
Participant recommends minimising daily activities or using the spoon theory to accomplish all necessary requirements of a daily or weekly basis	**5**	10.00	0	0.00	1	7.14	1	11.11	3	27.27	0	0.00
Participant reports that monitoring health to ensure continuous good health	**5**	10.00	2	33.33	3	21.43	0	0.00	0	0.00	0	0.00
Participant reports maintaining a positive frame of mind	**4**	8.00	0	0.00	1	7.14	2	22.22	1	9.09	0	0.00

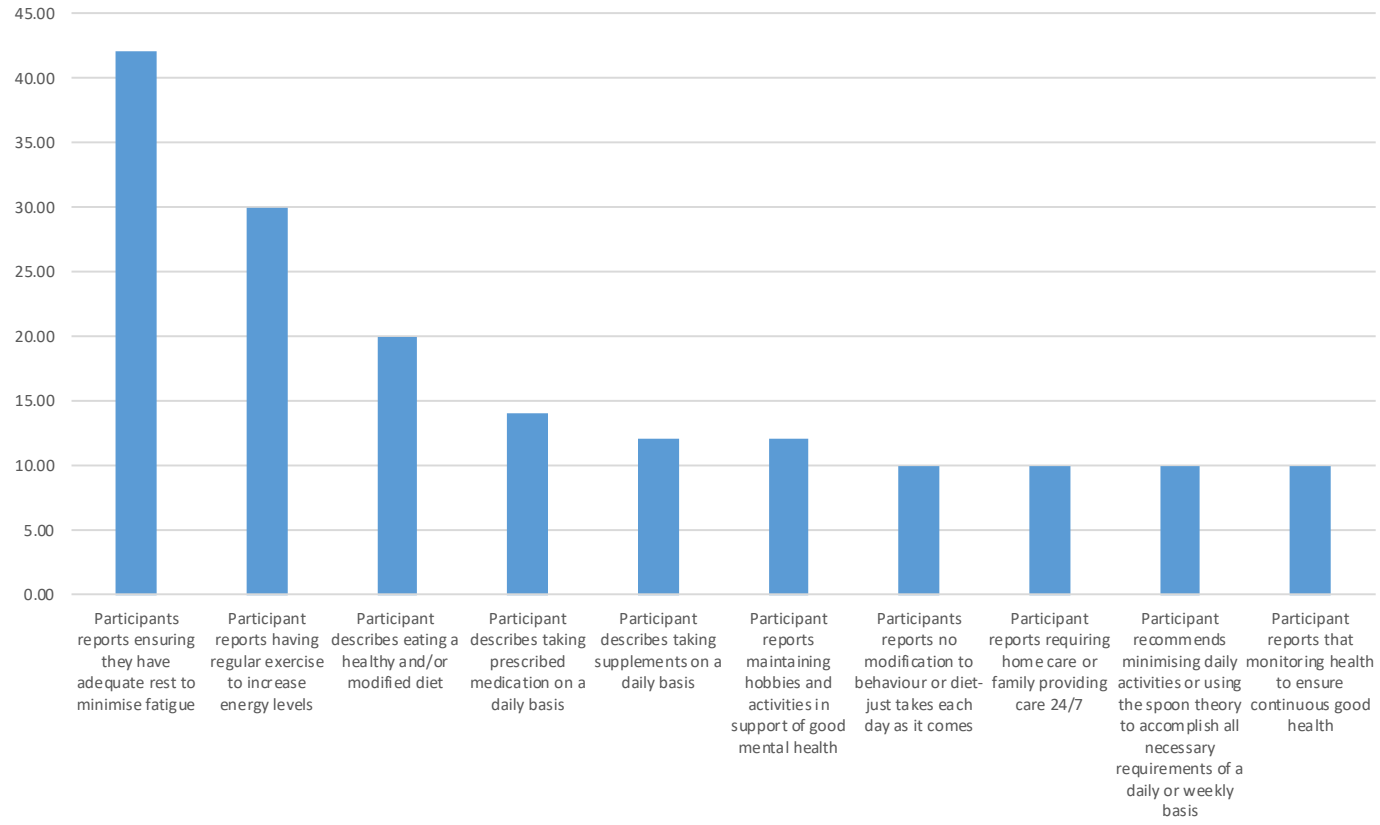


Figure 8.2: Everyday activities to manage health (% of all participants)

Impact on relationships with family and friends

Participants were asked whether having mitochondrial disease has had an impact on their relationships with family and friends. The most common theme described by participants was a negative impact on personal relationships due to people withdrawing from relationships or not being able to understand (n=14, 28.00%) and this was followed by a negative impact on personal relationships due to social isolation (n=11, 22.00%).

Participant describes a negative impact on personal relationships due to people withdrawing from relationships or not being able to understand

The relationship I was in when I was diagnosed which was a four year relationship, it wasn't a short term one. He left. I was diagnosed…he walked out Christmas eve. Couldn't deal with it. The poor fellow. Participant 18

Yes, probably with my friends because it's hard to explain it because I look all right, and I don't think they see that I'm really tired. I've probably lost a few friends. Especially at uni when I went to uni and that because there was no real diagnosis and I was tired and sick and couldn't really do the things I was doing. They were like, "Why aren't you doing those things?" Participant 19

Definitely. Yes, I think my sisters particularly don't really want to know about it, and I think they think I'm just making a mountain out of a molehill kind of thing. I suppose I'm fortunate that, in a way, I've just accepted the way I was, but they don't understand what it's like to be left behind and all that sort of stuff always. I did manage to get two of them to come with me to a genetic counselling appointment that I had once. That was very helpful really for me just for them to be convinced that, yes, this is a real thing, and something we should all know about, so it's good. I think it's really frustrating for my husband, but he's quiet. He tries to be understanding and he does a lot of practical things that are helpful... Participant 34

Participant describes a negative impact on personal relationships due to social isolation

Unfortunately, I think so, but not in any intentional way like no one's…I think it's just hard because you have less time when you've got Mitochondrial disease, you need to have so much rest. Relationships that were built on going out to dinner just can't…It's hard to sustain anything like that anymore. Participant 5

Absolutely. I have no friends, as I cannot predictably leave the house and people feel uncomfortable being around me. Participant 30

Definitely, yes. He doesn't have close friends. He's never had anyone ask him over for a play date. He doesn't have little buddies apart from kids that are at school. His best friend is his dog that we got eight, nine months ago. Participant 50

The next most common theme was a negative impact on personal relationships due to not being able to do all activities with family and friends (n=10, 20.00%).

Participant describes a negative impact on personal relationships due to not being able to do all activities with family and friends

Yes, from time to time…I've been trying to come up with a plan medication-wise, and the family have said to me that when I'm on some of those…We don't know which ones they were because I was trying a myriad of them, but they said there was periods there when I was a grumpy ass. Soon as I got off it, went back to being my normal self, so that sort of impacted on them. Knocked down for a walk with my missis with the dog in the afternoon, that's impacted on her. They'd be periods where your family wants to do something that might involve a bit of walking and stuff like that, and I'll make up an excuse…no not an excuse. I'll come up with something else to do that means that I have to participate in that just to avoid the pain that comes with walking those distances and things. Next time I'm playing golf forget about playing a round of golf and walking there. Then if you've got to be in a buggy you will forget about it. Participant 2

Yes. My husband worries about me and these things, but we obviously don't do bush walking, we live on a farm and I can't do the stuff with him that he'd like us to do, we haven't had children, that's the biggest. When I was first diagnosed I didn't know what it was or how it would be transmitted, so I chose not to have children because I didn't want to transmit something that I didn't know what I was going to be transmitting. The worry for people, I think they worry. Participant 3

Yes, because I can't do the things they do. My family, they think there's nothing wrong with me, I'm just putting it all on. Old age, some said to me, "It's just old age dad." Participant 42

There were six participants (12.00%) that described a positive impact of strengthening relationships.

Participant describes a positive impact of strengthening relationships

In a lot of ways it's brought me a lot closer to a lot of other people. I have a friend who has got a very different chronic illness to me, but we share a lot of the same frustrations. In some ways, it's brought me closer to other people. Participant 5

Now, I've got a lovely fellow I see. He's just gorgeous. He seems to deal with it. I think he deals with it better than I do. He's a bit of a rock for me actually. [Interviewer: That's wonderful] It is. It is because I'd given up. [laughs] I just thought this was going to be it. Participant 18

Yeah, it's actually quite ... it's really lovely to see…seeing that a little bit more as he gets older but he's got some lovely school friends. There are some lovely, little mother hens he's had over the years. He's had that right through since preschool, actually. He's a lovable child and of course, the fact that he is wheelchair bound means that you have some beautiful little things. They just want to fuss over him and there's this one little girl, NAME at school. They've gone through an intervention together and they just have a really, very beautiful connection but in saying that I'm being told that nine-minute school that the kids in his class. He's in a support class of eight children and they fight as to who's going to sit next to NAME. It's really, very, very special…It's really lovely to see and know about. Participant 45

In relation to sub-group variations, participants from low socio-economic areas (43.48%) and those with low social function (40.00%) reported a negative impact on personal relationships due to people withdrawing from relationships or not being able to understand, more frequently than the general population (28.00%), while those from high socio-economic areas (14.81) and high social function (10.00%) reported this less frequently. Participants from metropolitan areas (33.33%) and those with low emotional well-being (33.33%) reported a negative impact on personal relationships due to social isolation, more frequently than the general population (22.00%) while those from rural areas (10.00%) reported this less frequently. Participants from low socio-economic areas (30.43%) described a negative impact on personal relationships due to not being able to do all activities with family and friends,

more frequently than the general population (20.00%). Participants with high physical function (27.27%) and high social function (35.00%) described no impact on personal relationships more frequently than the general population (16.00%), while those with low social function described this less frequently.

Table 8.3: Impact on relationships

Impact on relationships	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant describes a negative impact on personal relationships due to people withdrawing from relationships or not being able to understand	**14**	28.00	7	23.33	7	35.00	4	**14.81**	10	**43.48**
Participant describes a negative impact on personal relationships due to social isolation	**11**	22.00	10	**33.33**	2	**10.00**	7	25.93	5	21.74
Participant describes a negative impact on personal relationships due to not being able to do all activities with family and friends	**10**	20.00	5	16.67	5	25.00	3	11.11	7	**30.43**
Participant describes no impact on personal relationships (No specific reason)	**8**	16.00	6	20.00	2	10.00	6	22.22	2	8.70
Participant describes a positive impact of strengthening relationships	**6**	12.00	3	10.00	3	15.00	4	14.81	2	8.70
Participant describes impact on personal relationships (No specific reason)	**5**	10.00	4	13.33	1	5.00	3	11.11	2	8.70
Participant describes a negative impact of personal relationships due to communication problems (associated with hearing loss)	**5**	10.00	3	10.00	2	10.00	2	7.41	3	13.04

Impact on relationships	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant describes a negative impact on personal relationships due to people withdrawing from relationships or not being able to understand	**14**	28.00	8	30.77	6	25.00	8	33.33	11	32.35
Participant describes a negative impact on personal relationships due to social isolation	**11**	22.00	6	23.08	6	25.00	3	12.50	8	23.53
Participant describes a negative impact on personal relationships due to not being able to do all activities with family and friends	**10**	20.00	5	19.23	5	20.83	5	20.83	8	23.53
Participant describes no impact on personal relationships (No specific reason)	**8**	16.00	6	23.08	2	8.33	3	12.50	5	14.71
Participant describes a positive impact of strengthening relationships	**6**	12.00	5	19.23	1	4.17	3	12.50	5	14.71
Participant describes impact on personal relationships (No specific reason)	**5**	10.00	2	7.69	3	12.50	1	4.17	1	2.94
Participant describes a negative impact of personal relationships due to communication problems (associated with hearing loss)	**5**	10.00	4	15.38	1	4.17	3	12.50	5	14.71

Impact on relationships	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant describes a negative impact on personal relationships due to people withdrawing from relationships or not being able to understand	**14**	28.00	7	31.82	7	25.00	5	19.23	9	37.50
Participant describes a negative impact on personal relationships due to social isolation	**11**	22.00	4	18.18	8	28.57	4	15.38	8	**33.33**
Participant describes a negative impact on personal relationships due to not being able to do all activities with family and friends	**10**	20.00	4	18.18	6	21.43	5	19.23	5	20.83
Participant describes no impact on personal relationships (No specific reason)	**8**	16.00	6	**27.27**	2	7.14	6	23.08	2	8.33
Participant describes a positive impact of strengthening relationships	**6**	12.00	2	9.09	4	14.29	2	7.69	4	16.67
Participant describes impact on personal relationships (No specific reason)	**5**	10.00	2	9.09	3	10.71	4	15.38	1	4.17
Participant describes a negative impact of personal relationships due to communication problems (associated with hearing loss)	**5**	10.00	3	13.64	2	7.14	2	7.69	3	12.50

Impact on relationships	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant describes a negative impact on personal relationships due to people withdrawing from relationships or not being able to understand	**14**	28.00	2	**10.00**	12	**40.00**	5	22.73	9	32.14
Participant describes a negative impact on personal relationships due to social isolation	**11**	22.00	3	15.00	9	30.00	4	18.18	8	28.57
Participant describes a negative impact on personal relationships due to not being able to do all activities with family and friends	**10**	20.00	5	25.00	5	16.67	4	18.18	6	21.43
Participant describes no impact on personal relationships (No specific reason)	**8**	16.00	7	**35.00**	1	**3.33**	5	22.73	3	10.71
Participant describes a positive impact of strengthening relationships	**6**	12.00	2	10.00	4	13.33	0	0.00	6	21.43
Participant describes impact on personal relationships (No specific reason)	**5**	10.00	4	20.00	1	3.33	3	13.64	2	7.14
Participant describes a negative impact of personal relationships due to communication problems (associated with hearing loss)	**5**	10.00	2	10.00	3	10.00	3	13.64	2	7.14

Impact on relationships	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant describes a negative impact on personal relationships due to people withdrawing from relationships or not being able to understand	**14**	28.00	1	16.67	5	35.71	2	22.22	4	36.36	2	20.00
Participant describes a negative impact on personal relationships due to social isolation	**11**	22.00	3	50.00	4	28.57	0	0.00	3	27.27	2	20.00
Participant describes a negative impact on personal relationships due to not being able to do all activities with family and friends	**10**	20.00	1	16.67	3	21.43	3	33.33	1	9.09	2	20.00
Participant describes no impact on personal relationships (No specific reason)	**8**	16.00	0	0.00	2	14.29	2	22.22	1	9.09	3	30.00
Participant describes a positive impact of strengthening relationships	**6**	12.00	1	16.67	3	21.43	0	0.00	2	18.18	0	0.00
Participant describes impact on personal relationships (No specific reason)	**5**	10.00	1	16.67	1	7.14	2	22.22	0	0.00	1	10.00
Participant describes a negative impact of personal relationships due to communication problems (associated with hearing loss)	**5**	10.00	0	0.00	2	14.29	1	11.11	0	0.00	2	20.00

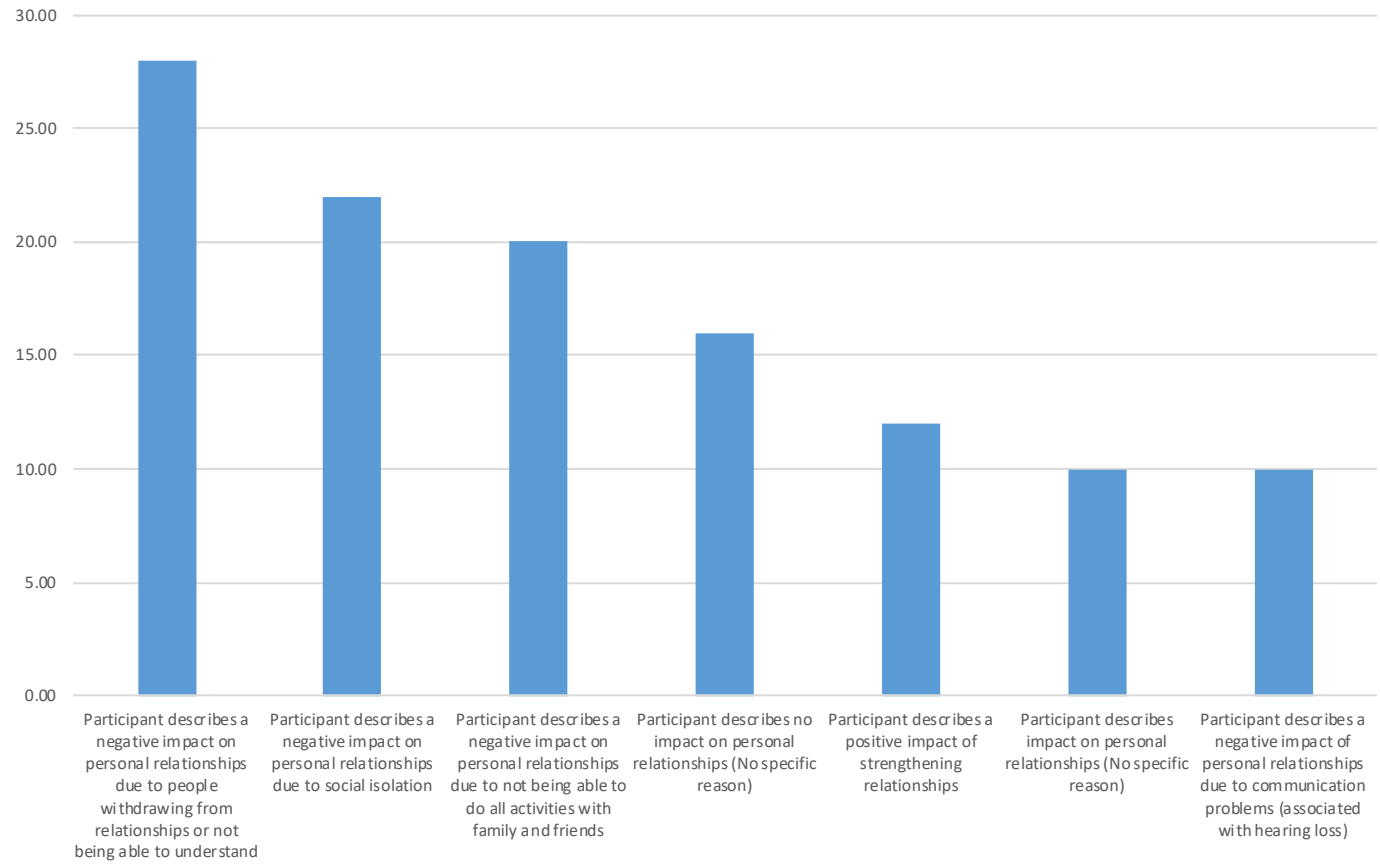


Figure 8.3: Impact on relationships (% of all participants)

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, with a higher score denoting increased anxiety. Summary statistics for the entire cohort are displayed alongside the possible range of the scale in Table 8.4. Overall the entire cohort had a mean total score of 34.10, which is a score in the middle of the scale.

Boxplots of Fear of Progression Total Score (FOPTS) by disease stage, metastatic status, location, and education status are displayed in Figures 8.5 to 8.13.

Comparisons of FOPTS have been made based on general health (Figure 8.5), physical functioning (Figure 8.6), emotional well-being (Figure 8.7), social functioning, (Figures 8.8), hearing problems (Figure 8.9), eye problems (Figure 8.10), location (Figure 8.11), education (Figure 8.12), and SEIFA (Figure 8.13).

Comparisons were made by a two-sample t-test was used when assumptions for normality and variance were met (Table 8.5), or when assumptions for normality and variance were not met, a Wilcoxon rank sum test with continuity correction was used (Table 8.6).

There were no statistically significant differences between any of the subgroups for FOPTS.

In addition to the fear of progression questionnaire, participants were asked if they become anxious if they did not experience side effects of treatment as it makes them feel that the treatment is not working. The majority of participants never (n=28, 56.00%), seldom (n=5, 10.00%), sometimes (n=12, 24.00%), often (n=3, 6.00%) and very often (n=3, 6.00%).

Table 8.4: Fear of Progression Total Score

	Mean*	SD	Median	IQR	Possible range
FOPTS	34.10	8.09	34.00	4	12-60

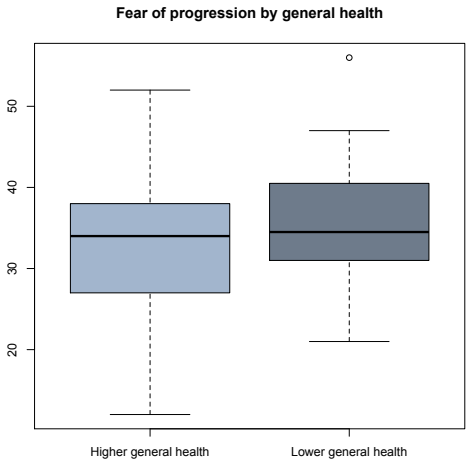


Figure 8.5: Boxplot of FOPTS by general health

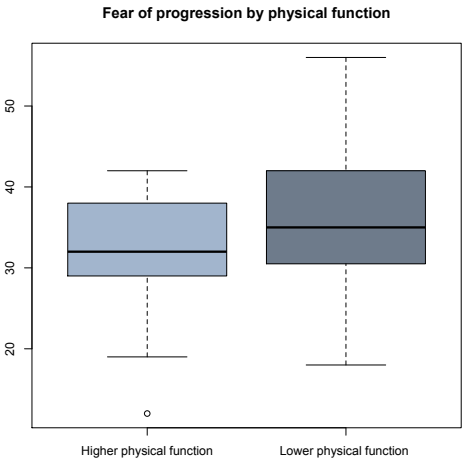


Figure 8.6: Boxplot of of FOPTS by physical functioning

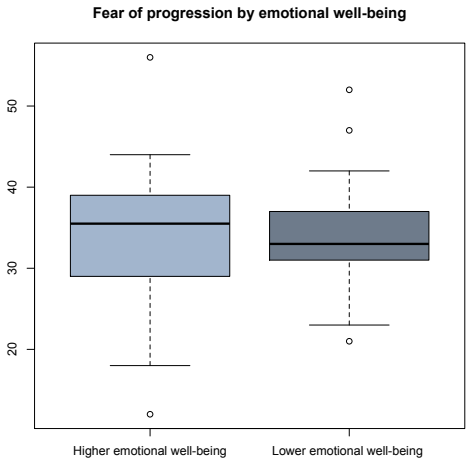


Figure 8.7: Boxplot of FOPTS by emotional well-being

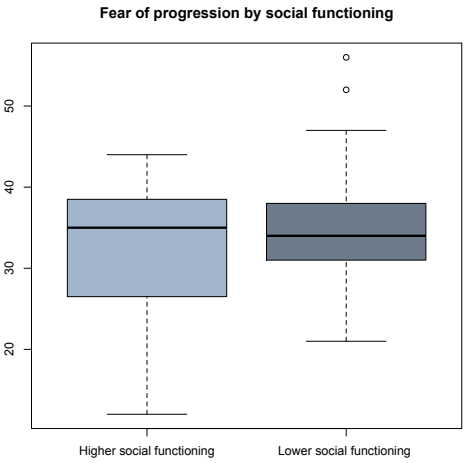


Figure 8.8: Boxplot of FOPTS by social functioning

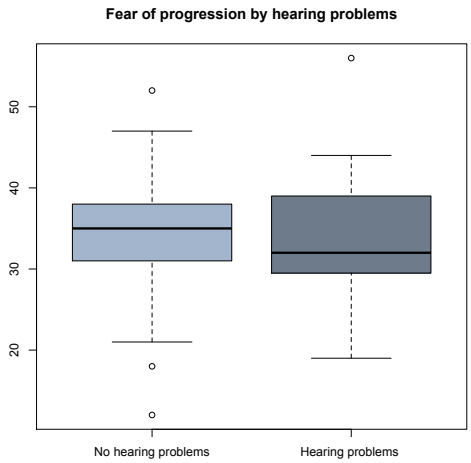


Figure 8.9: Boxplot of FOPTS by hearing problems

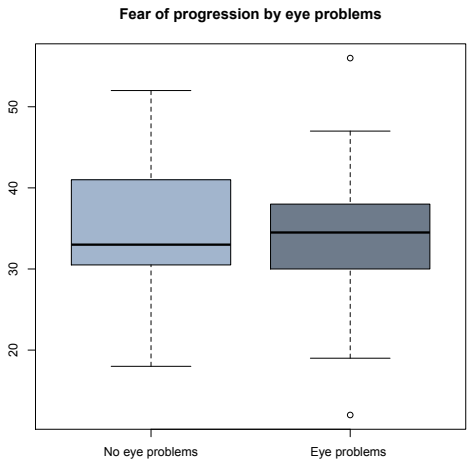


Figure 8.10: Boxplot of of FOPTS by eye problems

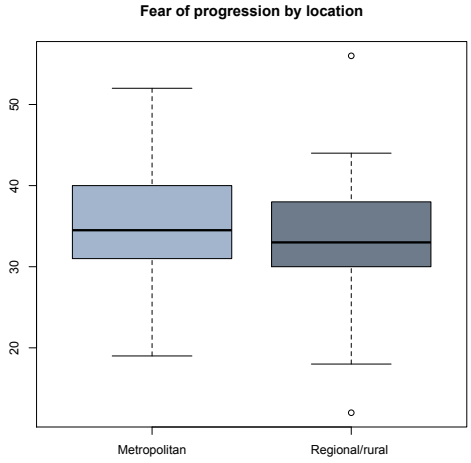


Figure 8.11: Boxplot of FOPTS by location

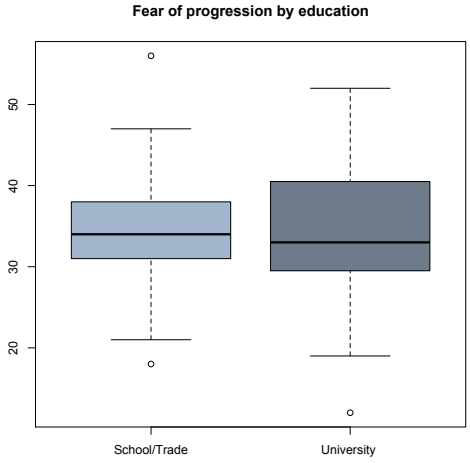


Figure 8.12: Boxplot of FOPTS by education

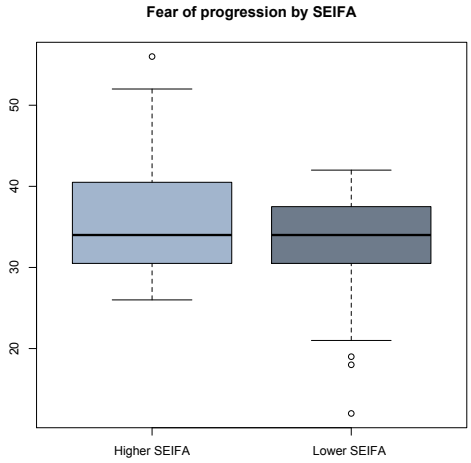


Figure 8.13: Boxplot of FOPTS by SEIFA

Table 8.5: Summary statistics and two sample t-test FOPTS

FOPTS by subgroups	Groups	Count	Mean	SD	t	dF	P
General health	Higher general health	2	32.36	9.17	-1.36	48	0.1812
	Lower general health	28	35.46	7.00			
Emotional well-being	Higher emotional well-being	26	33.85	9.32	-0.23	48	0.8201
	Lower emotional well-being	24	34.38	6.69			
Hearing problems	No hearing problems	26	34.42	8.40	0.29	48	0.7722
	Hearing problems	24	33.75	7.90			
Eye problems	No eye problems	26	34.69	7.80	0.35	48	0.7285
	Eye problems	34	33.82	8.32			
Location	Metropolitan	30	35.00	6.94	0.96	48	0.3405
	Regional/rural	20	32.75	9.60			
Education	School/Trade	26	34.35	7.60	0.22	48	0.8254
	University	24	33.83	8.74			

Table 8.6: Summary statistics Wilcoxon rank sum test with continuity correction of FOPTS

FOPTS by subgroups	Groups	Count	Median	IQR	W	P
Physical functioning	Higher physical functioning	22	32.00	8.00	248.00	0.2439
	Lower physical functioning	28	35.00	11.25		
Social functioning	Higher social functioning	20	35.00	11.50	274.00	0.6129
	Lower social functioning	30	34.00	6.50		
SEIFA	Higher SEIFA	27	34.00	10.00	363.50	0.3058
	Lower SEIFA	23	34.00	7.00		

Section 9 Expectations and messages

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

Expectations of future treatments

- The most common theme described by participants was that cost was an important consideration in relation to future treatments (n=18, 36.00%). This was followed by the need for effective treatments for mitochondrial disease, where participants may have also noted that there are no or limited treatments available (n=16. 36.00%). There were seven participants (14.00%) that described the need for clinical trials in mitochondrial disease and six participants (12.00%) that described the need for treatments that reduce muscle fatigue/improve muscle strength.
- In relation to sub-group variations, participants from low socio-economic areas (52.17%) described cost as a consideration more frequently than the general population (36.00%), while those from high socio-economic areas (25.93%) reported this less frequently. Participants from metropolitan areas (46.67%) and those with low emotional well-being (45.83%) reported the need for effective treatments for mitochondrial disease, more frequently than the general population (32.00%), while those from rural areas (15.00%) reported this less frequently.
- 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 were tiredness and fatigue, muscle symptoms and nervous system symptoms; the least important were underactive thyroid or parathyroid, and excess body hair.
- Participants were asked to rank what is important for them overall when they make decisions about treatment and care. The most important aspects were safety of treatment/weighing up risks and benefits, and severity of side effects. The least important were ability to stick to treatment, and including family in decision-making.
- 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 patient,s and access for all patients to all treatments and services; the least important was economic value to government.

Expectation of future information provision

- The most common theme was that participants described being satisfied with current information and therefore had no recommendation (n=11, 22.00%). There were nine participants (18.00%) that described the need for information about their specific type of mitochondrial disease, and nine participants (18.00%) that described the need for healthcare professionals to deliver accurate, comprehensive and honest information (including prognostic information. There were also six participants (12.00%) that described the need for centralised and reliable information.
- In relation to sub-group variations, participants with high general health (31.82%) described the need for healthcare professionals to deliver accurate, comprehensive and honest information (including prognostic information), more frequently than the general population (18.00%).

Expectation of future healthcare professional communication

- The most common theme was that participants recommend healthcare professional education in relation to mitochondrial disease and more understanding of the impact and implications of the condition (n=16, 32.00%). This was followed by the recommendation that healthcare professionals are more proactive and attentive (n=9, 18.00%). There were also nine participants (18.00%) that did not have a recommendation as they have been satisfied with communication. Where participants were satisfied with communication it was primarily because communication had been open communication. There were seven participants (14.00%) that recommended that healthcare professionals need to have more empathy.
- In relation to sub-group variations, participants from rural areas (45.00%) and those from low socio-economic areas (43.48%) recommended healthcare professional education in relation to mitochondrial disease and more understanding of the impact and implications of the condition, more frequently than the general population (32.00%).

Expectation of future care and support

- The most common recommendation was for centralised and coordinated care across specialists and allied health professionals (including more communication between doctors) (n=13, 26.00%). In a similar theme, there were also six participants (12.00%) that recommended caseworkers be employed to support patients navigate health, medical and emotional needs. This was followed by the recommendation for support groups to help patients noting that it is difficult due to the diversity within the patient population (n=7, 14.00%) and more equity in access to services and support for adults with rare disease (n=7, 14.00%).
- In relation to sub-group variations, participants with a university education (50.00%) and those with a hearing impairment (45.83%) recommended centralised and coordinated care across specialists and allied health professionals, more frequently than the general population (26.00%), while those with a high school or trade education (3.85%) recommended this less frequently.

What participants are grateful for in the Australian health system

- The most common theme was participants describing being grateful for Medicare in relation to access to specialists (n=17, 34.00%), followed by being grateful for the compassion and support shown by healthcare professionals (n=16, 32.00%). There were 10 participants (20.00%) that described being grateful for Medicare in relation to access to allied health professionals and seven participants (14.00%) described being grateful for their healthcare card and the financial relief it provides. Other aspects of the health system that participants spoke about being grateful for were subsidised diagnostic tests (n=6, 12.00%), government initiatives that support ongoing health and quality of life (for example NDIS, Better Start Program and At home nursing services) (n=6, 12.00%) and the quality of specialist expertise in Australia (n=5, 10.00%).
- In relation to sub-group variations, participants from rural areas (45.00%) described being grateful for Medicare (Access to specialists) more frequently than the general population (34.00%). Participants with a university education (45.83%), those with high physical function (54.55%), and those with high general health (50.00%) reported being grateful for the compassion and support shown by healthcare professionals more frequently than the general population (32.00%), while those with low physical function (17.86%) reported this less frequently. Participants from rural areas (25.00%), those with a hearing impairment (25.00%) and those with low physical function (25.00%) described being grateful for their healthcare card and the financial relief it provides, more frequently than the general population (14.00%), while there we no participants with high physical function (0.00%) that reported this.

Messages

- The most common message is to support more research (n=20, 40.00%), however this was a general statement with no specific area noted. The next most common theme was to provide more education to the healthcare professionals, particularly education about managing the condition (n=15, 30.00%), and this was followed by the message to increase awareness of mitochondrial disease among the community (n=12, 24.00%). There were 12 participants (24.00%) whose message is to provide more holistic and multidisciplinary/allied health care, and eight participants (16.00%) whose message is to improve treatments by following the example of other countries that have more advanced systems.
- In relation to sub-group variations, participants from rural areas (55.00%) called for more research more frequently than the general population (40.00%). Participants with a hearing impairment (41.67%) had the message to provide more education to the healthcare professionals, more frequently than the general population (30.00%). Participants with a university education (12.50%) called for more awareness less frequently than the general population (24.00%). Participants with a university education (33.33%) and those with a hearing impairment (37.50%) had the message to support more funding (in general), more frequently than the general population (22.00%), while those with a high school or trade education reported this less frequently (11.54%). Participants with high physical function (13.64%) had the message to provide more holistic and multidisciplinary/allied health care less frequently than the general population (24.00%).

Expectations of future treatment

Participants were asked a series of questions about their expectations for future treatments, information, health professional communication, and care and support.

When asked about their expectations of future treatment, there were a number of themes that emerged. The most common theme described by participants was that cost was an important consideration in relation to future treatments (n=18, 36.00%).

Participant describes cost as a consideration in access to treatments

Lending more information of what treatment help people and cost is a big one because some of my medication is not on the PBS so it's expensive. Participant 19

Cost is a huge one. Recently, I had to go to the bank and through the financial assistance because I'd just hit a wall. The amount that I get in remuneration is about a third of what my salary was. I've never been good with money and I'd hit a wall.. Participant 20

Hopefully, some extra treatments. Hopefully, they're not too expensive for us. That chronic comes into play pretty much. Participant 22

Well, the cost is very steep. We're outlaid few thousand dollars already. We try not to outlay any more money. [laughs] Participant 35

It would be good if all treatments were on the PBS. I take a very high dose coenzyme Q10 and it does cost. If I could get it through PBS, it would be cheaper and for my son as well. Participant 43

This was followed by the need for effective treatments for mitochondrial disease, where participants may have also noted that there are no or limited treatments available (n=16. 36.00%).

Participant describes the need for effective treatments for mitochondrial disease (may also note that there are no or limited treatments available)

Any treatment for mito would be great. For LHON, there are two main types of treatment, both of which are still in clinical trials. There's the idebenone program (vitamins), which is available on a very limited basis in Australia, and gene therapy trials, available only overseas. The latter may be promising but it's too expensive and laborious to take part in. Bringing trials to Aus would be incredible, as would getting past trial stage. Participant 8

I'll just like to see that new treatments are actually...have got good evidence behind them and not being rushed through. Seems to be a lot of new treatments suddenly emerging on the market, which just the evidence could be quite questionable and really the effectiveness is quite questionable too, and even the way the trials are run are quite questionable. I'd like to see the same standard applied to trials and stuff for and treatments for mitochondrial disease as would be set for general population. Participant 13

There were seven participants (14.00%) that described the need for clinical trials in mitochondrial disease and six participants (12.00%) that described the need for treatments that reduce muscle fatigue/improve muscle strength.

Participant describes the need for clinical trials in mitochondrial disease

I'll just like to see that new treatments are actually...have got good evidence behind them and not being rushed through. Seems to be a lot of new treatments suddenly emerging on the market, which just the evidence could be quite questionable and really the effectiveness is quite questionable too, and even the way the trials are run are quite questionable. I'd like to see the same standard applied to trials and stuff for and treatments for mitochondrial disease as would be set for general population. Participant 13

Well, I'd like to see more research done into it. I realize there is some research but it all seems to be more for the younger people, not when you're being diagnosed as an adult. I don't know. Just don't know what to say there. Participant 31

Participant describes the need for treatments that reduce muscle fatigue/improve muscle strength

That is...My biggest is moving again. Not so unbalanced and we used to play fully for a long time. They're doing that now. Kind of run. Participant 6

If they had some treatment that would make my muscles not so tired then that would be the main thing. Participant 10

For me personally, if there was a medication to help me get over my fatigue and reduce that lactic acid so that I could live a bit better and do the things that I'd like to do, yeah. Anything that can do stuff like that would be fantastic. Participant 15

In relation to sub-group variations, participants from low socio-economic areas (52.17%) described cost as a consideration more frequently than the general population (36.00%), while those from high socio-economic areas (25.93%) reported this less frequently. Participants from metropolitan areas (46.67%) and

those with low emotional well-being (45.83%) reported the need for effective treatments for mitochondrial disease, more frequently than the general population (32.00%), while those from rural areas (15.00%) reported this less frequently.

Table 9.1: Expectations of future treatment

Expectations of future treatments	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant describes cost as a consideration in access to treatments	18	36.00	11	36.67	8	40.00	7	25.93	12	52.17
Participant describes the need for effective treatments for mitochondrial disease (may also note that there are no or limited treatments available)	16	32.00	14	46.67	3	15.00	9	33.33	8	34.78
Participant describes the need for clinical trials in mitochondrial disease	7	14.00	4	13.33	3	15.00	4	14.81	3	13.04
Participant describes the need for treatments that reduce muscle fatigue/improve muscle strength	6	12.00	4	13.33	2	10.00	3	11.11	3	13.04
Participant describes not being sure because they haven't had any treatments to compare to	5	10.00	2	6.67	3	15.00	4	14.81	1	4.35
Participant describes 'a cure' but also acknowledging this is not likely (for example, participant laughs when saying this)	5	10.00	2	6.67	3	15.00	2	7.41	3	13.04
Participant describes the need for treatments to reduce general exhaustion caused by mitochondrial disease	5	10.00	3	10.00	2	10.00	2	7.41	3	13.04
Participant describes the need to evaluate the effectiveness of the use of vitamins and supplements to treat symptoms	5	10.00	5	16.67	0	0.00	4	14.81	1	4.35

Expectations of future treatments	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant describes cost as a consideration in access to treatments	18	36.00	11	42.31	8	33.33	9	37.50	13	38.24
Participant describes the need for effective treatments for mitochondrial disease (may also note that there are no or limited treatments available)	16	32.00	7	26.92	10	41.67	9	37.50	9	26.47
Participant describes the need for clinical trials in mitochondrial disease	7	14.00	5	19.23	2	8.33	3	12.50	4	11.76
Participant describes the need for treatments that reduce muscle fatigue/improve muscle strength	6	12.00	3	11.54	3	12.50	2	8.33	5	14.71
Participant describes not being sure because they haven't had any treatments to compare to	5	10.00	3	11.54	2	8.33	1	4.17	4	11.76
Participant describes 'a cure' but also acknowledging this is not likely (for example, participant laughs when saying this)	5	10.00	4	15.38	1	4.17	2	8.33	4	11.76
Participant describes the need for treatments to reduce general exhaustion caused by mitochondrial disease	5	10.00	3	11.54	2	8.33	2	8.33	5	14.71
Participant describes the need to evaluate the effectiveness of the use of vitamins and supplements to treat symptoms	5	10.00	2	7.69	3	12.50	2	8.33	2	5.88

Expectations of future treatments	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant describes cost as a consideration in access to treatments	18	36.00	10	45.45	9	32.14	10	38.46	9	37.50
Participant describes the need for effective treatments for mitochondrial disease (may also note that there are no or limited treatments available)	16	32.00	9	40.91	8	28.57	6	23.08	11	45.83
Participant describes the need for clinical trials in mitochondrial disease	7	14.00	4	18.18	3	10.71	5	19.23	2	8.33
Participant describes the need for treatments that reduce muscle fatigue/improve muscle strength	6	12.00	3	13.64	3	10.71	4	15.38	2	8.33
Participant describes not being sure because they haven't had any treatments to compare to	5	10.00	1	4.55	4	14.29	1	3.85	4	16.67
Participant describes 'a cure' but also acknowledging this is not likely (for example, participant laughs when saying this)	5	10.00	2	9.09	3	10.71	3	11.54	2	8.33
Participant describes the need for treatments to reduce general exhaustion caused by mitochondrial disease	5	10.00	3	13.64	2	7.14	3	11.54	2	8.33
Participant describes the need to evaluate the effectiveness of the use of vitamins and supplements to treat symptoms	5	10.00	2	9.09	3	10.71	3	11.54	2	8.33

Expectations of future treatments	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant describes cost as a consideration in access to treatments	18	36.00	6	30.00	13	43.33	8	36.36	11	39.29
Participant describes the need for effective treatments for mitochondrial disease (may also note that there are no or limited treatments available)	16	32.00	6	30.00	11	36.67	9	40.91	8	28.57
Participant describes the need for clinical trials in mitochondrial disease	7	14.00	4	20.00	3	10.00	5	22.73	2	7.14
Participant describes the need for treatments that reduce muscle fatigue/improve muscle strength	6	12.00	4	20.00	2	6.67	4	18.18	2	7.14
Participant describes not being sure because they haven't had any treatments to compare to	5	10.00	1	5.00	4	13.33	1	4.55	4	14.29
Participant describes 'a cure' but also acknowledging this is not likely (for example, participant laughs when saying this)	5	10.00	2	10.00	3	10.00	3	13.64	2	7.14
Participant describes the need for treatments to reduce general exhaustion caused by mitochondrial disease	5	10.00	2	10.00	3	10.00	3	13.64	2	7.14
Participant describes the need to evaluate the effectiveness of the use of vitamins and supplements to treat symptoms	5	10.00	3	15.00	2	6.67	4	18.18	1	3.57

Expectations of future treatments	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant describes cost as a consideration in access to treatments	18	36.00	4	66.67	7	50.00	1	11.11	4	36.36	3	30.00
Participant describes the need for effective treatments for mitochondrial disease (may also note that there are no or limited treatments available)	16	32.00	4	66.67	6	42.86	3	33.33	1	9.09	3	30.00
Participant describes the need for clinical trials in mitochondrial disease	7	14.00	0	0.00	2	14.29	2	22.22	2	18.18	1	10.00
Participant describes the need for treatments that reduce muscle fatigue/improve muscle strength	6	12.00	0	0.00	2	14.29	2	22.22	0	0.00	2	20.00
Participant describes not being sure because they haven't had any treatments to compare to	5	10.00	0	0.00	1	7.14	0	0.00	4	36.36	0	0.00
Participant describes 'a cure' but also acknowledging this is not likely (for example, participant laughs when saying this)	5	10.00	0	0.00	0	0.00	2	22.22	2	18.18	1	10.00
Participant describes the need for treatments to reduce general exhaustion caused by mitochondrial disease	5	10.00	0	0.00	3	21.43	0	0.00	1	9.09	1	10.00
Participant describes the need to evaluate the effectiveness of the use of vitamins and supplements to treat symptoms	5	10.00	1	16.67	2	14.29	1	11.11	0	0.00	1	10.00

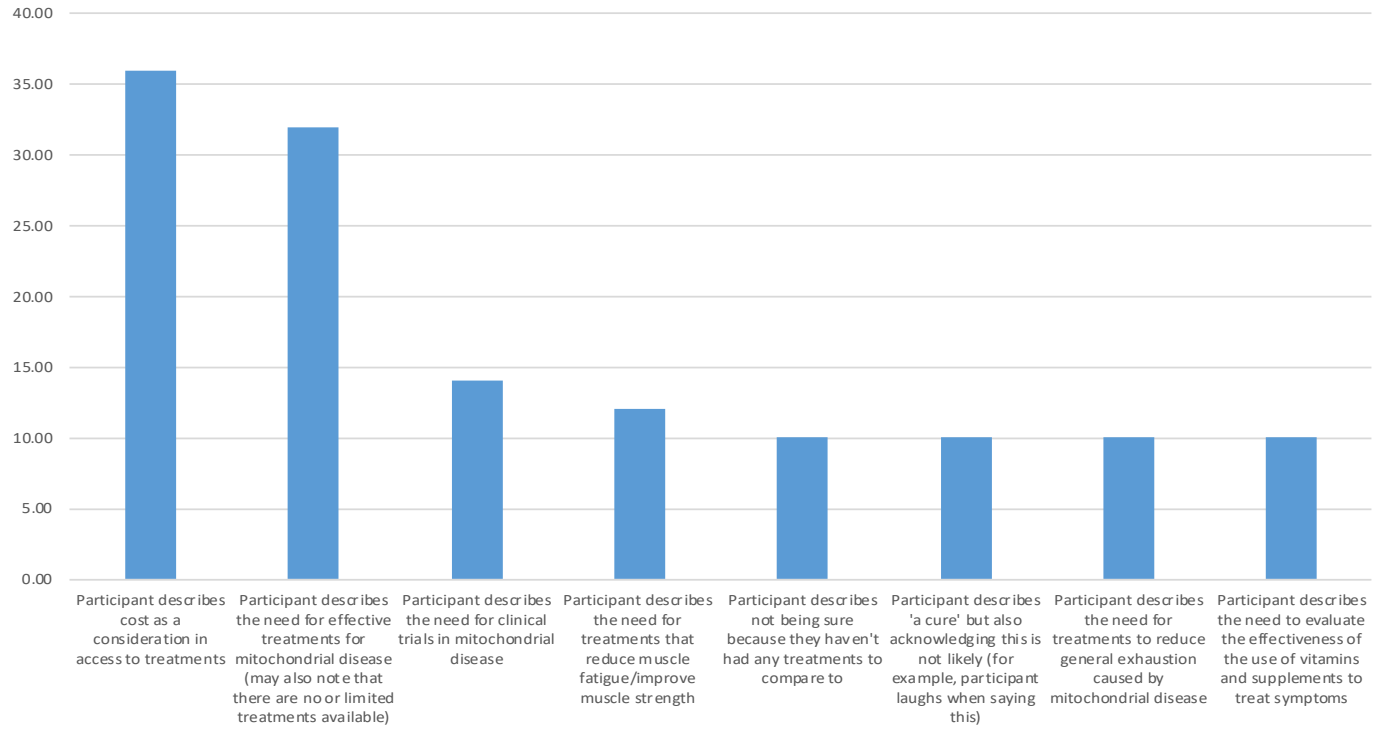


Figure 9.1: Expectations of future treatment (% of all participants)

Values when making decisions about treatment

Symptoms/aspects of quality of life important for treatments

Participants were asked about the value of access to treatments that reduce symptoms and improve quality of life even if they do not offer a cure. The majority of participants thought that it would be of very significant or significant value (n=44, 88.00%),five participants (10.00%) felt it would be of moderate or some value, and 1 participants (2.00%) felt it would be of or no value.

Table 9.2: Value of treatment to improve symptoms and quality of life

Value of access to treatment that reduces symptoms, improves quality of life but may not offer cure	N=50	Percentage of participants
No value at all to me	1	2.00
Some value to me	2	4.00
Moderate value to me	3	6.00
Significant value to me	7	14.00
Very significant value to me	37	74.00

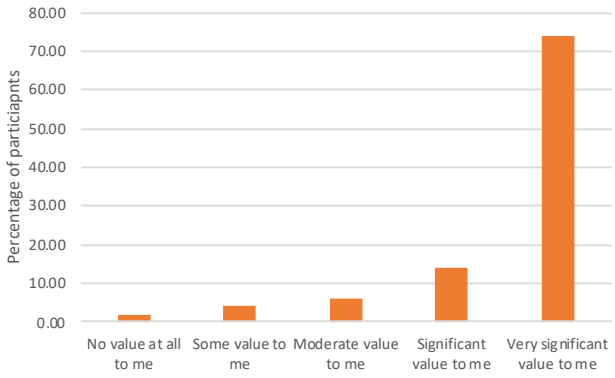


Figure 9.2: Value of treatment to improve symptoms and quality of life

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, were 1 is the most important and 12 is the least important. A weighted average is presented in Figure 9.3. With a weighted ranking, the higher the score, the greater value it is to participants. The most important aspects reported were tiredness and fatigue, muscle symptoms, and nervous system symptoms; the least important were underactive thyroid or parathyroid, and excess body hair. Figures 9.4 to 9.12 show the weighted rank by general health, physical functioning, emotional well-being, social functioning, hearing problems, eye problems, location, education and SEIFA, the symptoms and aspects of quality of life are similar within sub groups and follow much the same pattern as the entire cohort.

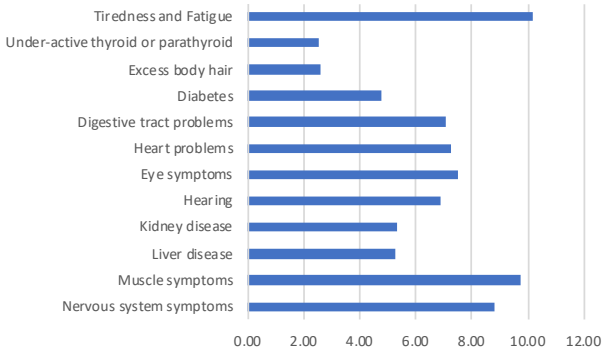


Figure 9.3: Symptoms/aspects of quality of life important for treatments all participants

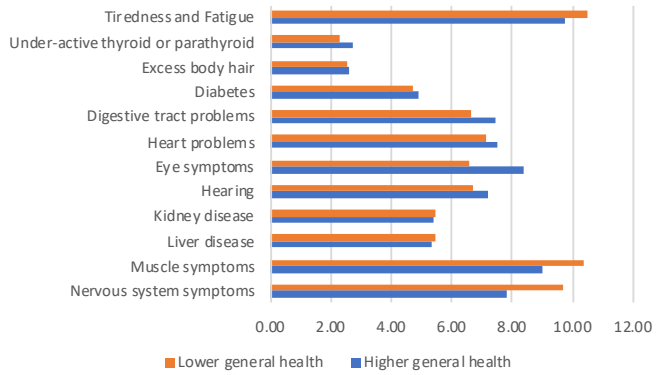


Figure 9.4: Symptoms/aspects of quality of life important for treatments by general health

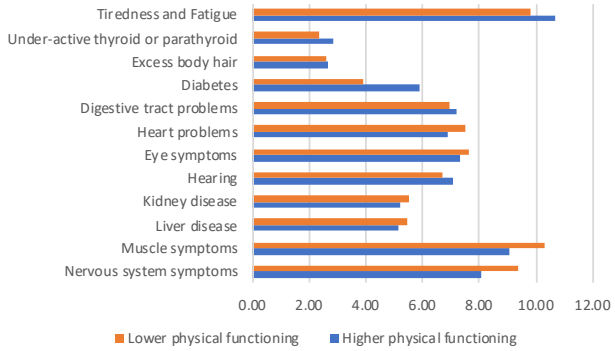


Figure 9.5: Symptoms/aspects of quality of life important for treatments by physical functioning

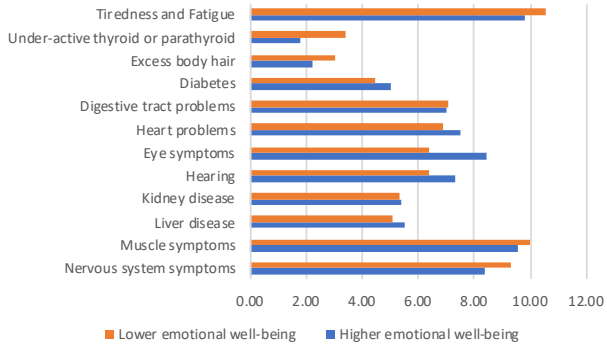


Figure 9.6: Symptoms/aspects of quality of life important for treatments by emotional well-being

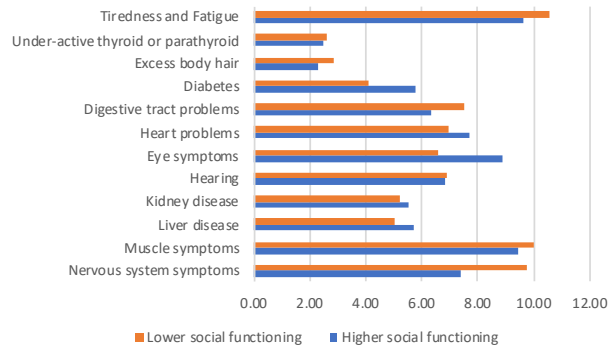


Figure 9.7: Symptoms/aspects of quality of life important for treatments by social functioning

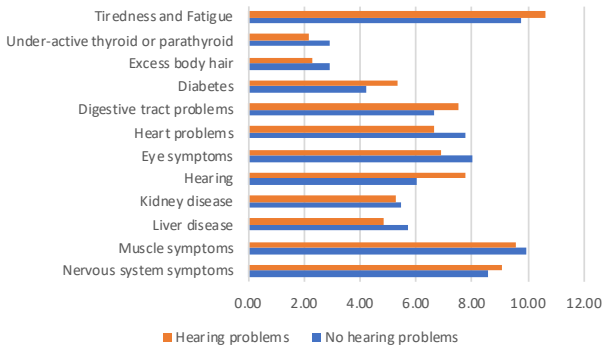


Figure 9.8: Symptoms/aspects of quality of life important for treatments by hearing problems

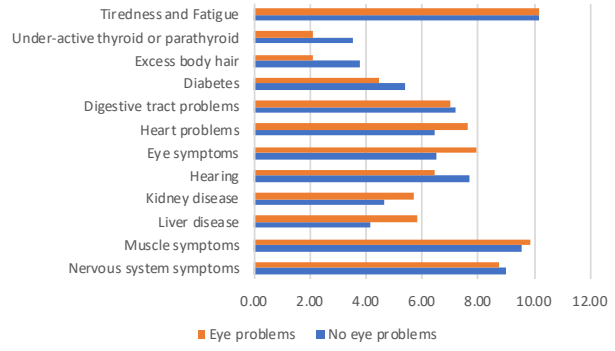


Figure 9.9: Symptoms/aspects of quality of life important for treatments by eye problems

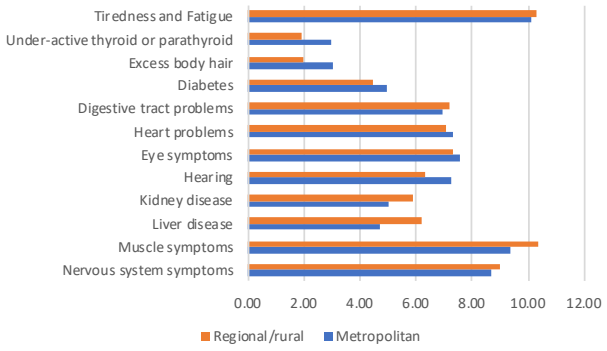


Figure 9.10: Symptoms/aspects of quality of life important for treatments by location

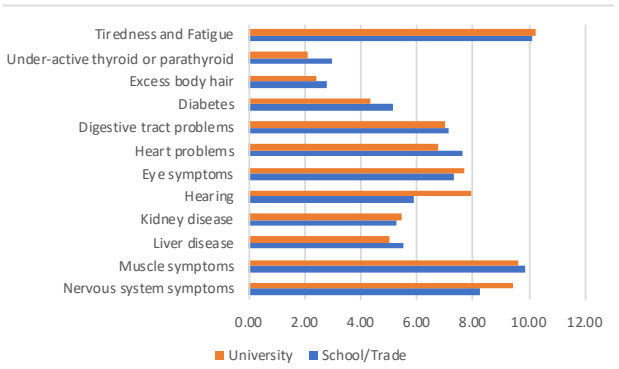


Figure 9.11: Symptoms/aspects of quality of life important for treatments by education

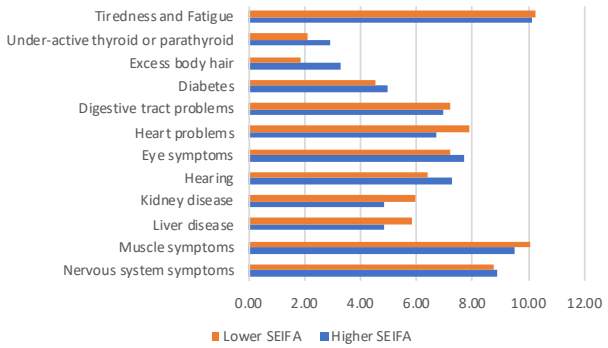


Figure 9.12: Symptoms/aspects of quality of life important for treatments by SEIFA

Values that are important to patients when making decisions

Participants were asked to rank what is important for them overall when they make decisions about treatment and care, where 1 is the most important and 8 is the least important. A weighted average is presented in Figure 9.13. With a weighted ranking, the higher the score, the greater value it is to participants. The most important aspects were safety of treatment/weighing up risks and benefits, and severity of side effects. The least important were ability to follow and stick to a treatment, and including family in decision-making. Figures 9.14 to 9.22 show the weighted rank by general health, physical functioning, emotional well-being, social functioning, hearing problems, eye problems, location, education and SEIFA, the values for making treatment decisions are similar within sub groups and are similar to the overall cohort.

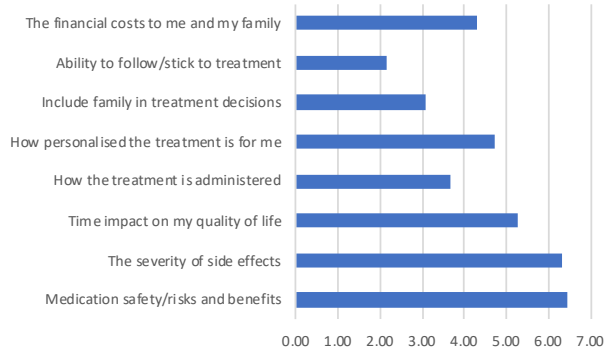


Figure 9.13: Values important when making decisions all participants

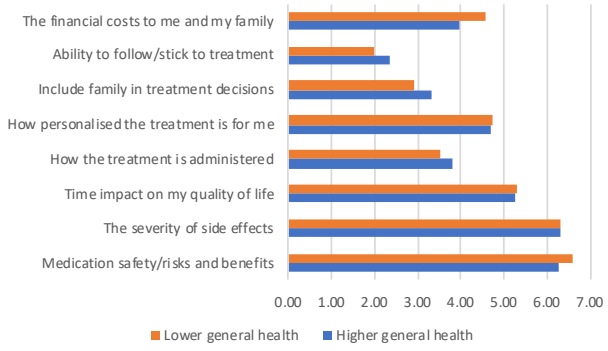


Figure 9.14: Values important when making decisions by general health

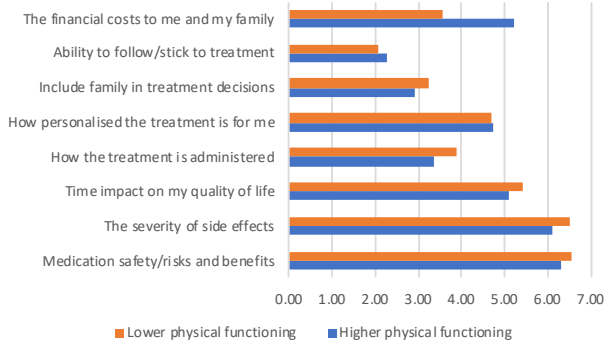


Figure 9.15: Values important when making decisions by physical functioning

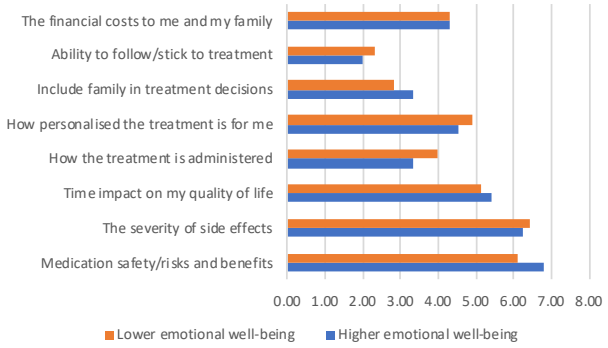


Figure 9.16: Values important when making decisions by emotional well-being

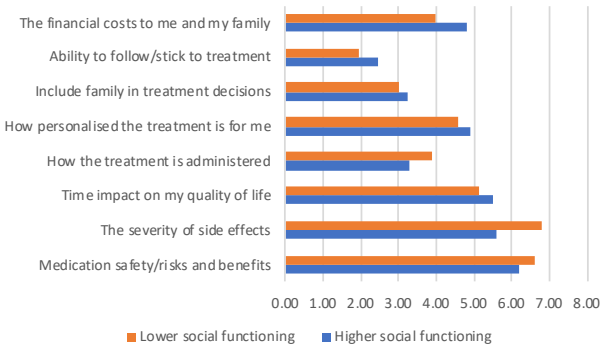


Figure 9.17: Values important when making decisions by social functioning

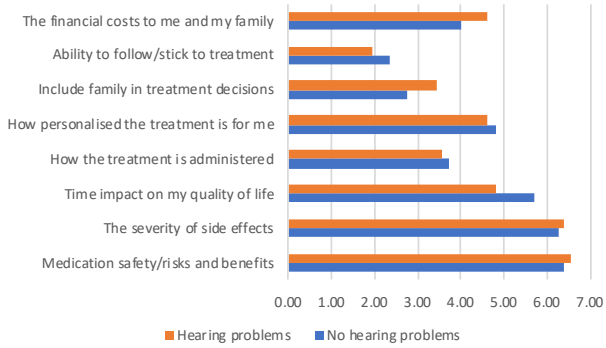


Figure 9.18: Values important when making decisions by hearing problems

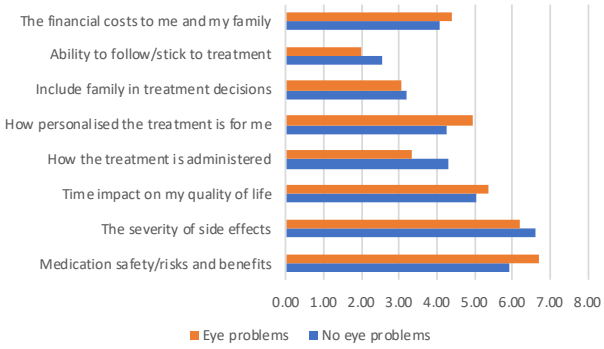


Figure 9.19: Values important when making decisions by eye problems

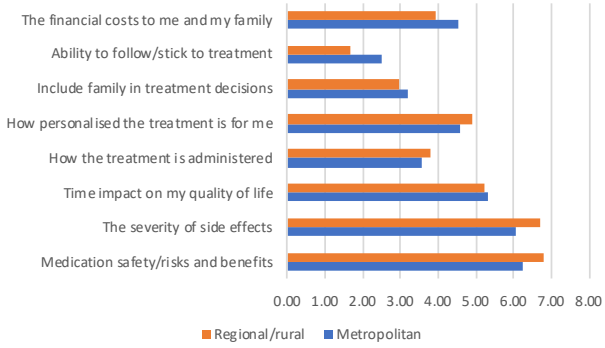


Figure 9.20: Values important when making decisions by location

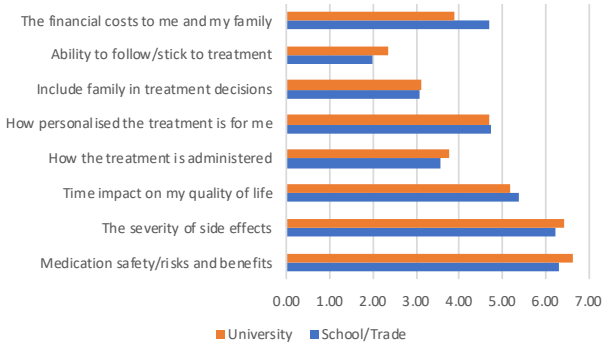


Figure 9.21: Values important when making decisions by education

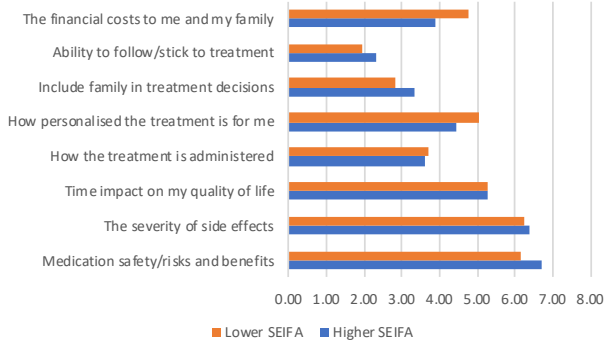


Figure 9.22: Values important when making decisions by SEIFA

Values that are important to patients when others are making decisions on their behalf

Participants were asked to rank what is important for decision-makers to consider when they make decisions that impact treatment and care, where 1 is the most important and 5 is the least important. A weighted average is presented in Figure 9.23. With a weighted ranking, the higher the score, the greater value it is to participants. 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. Figures 9.24 to 9.32 show the weighted rank by general health, physical functioning, emotional well-being, social functioning, hearing problems, eye problems, location, education and SEIFA, the values for making decisions on their behalf are similar within sub groups and are similar to the overall cohort.

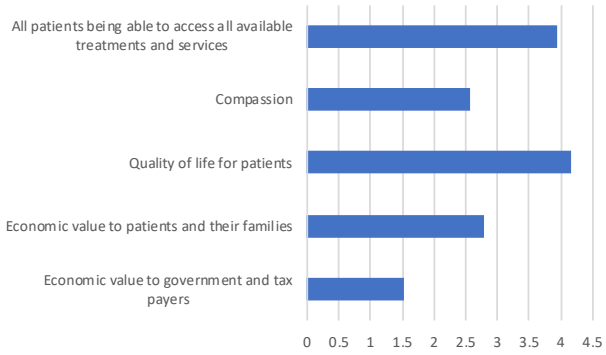


Figure 9.23: Values to consider on behalf of patients/families (weighted rank all participants)

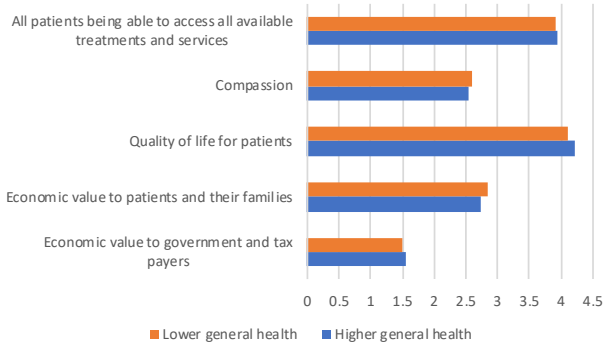


Figure 9.24: Values to consider on behalf of patients/families (weighted rank by general health)

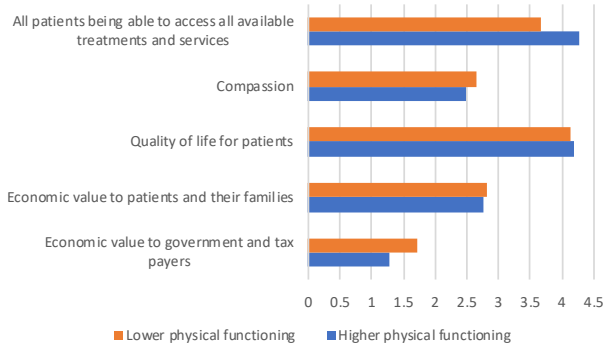


Figure 9.25: Values to consider on behalf of patients/families (weighted rank by physical functioning)

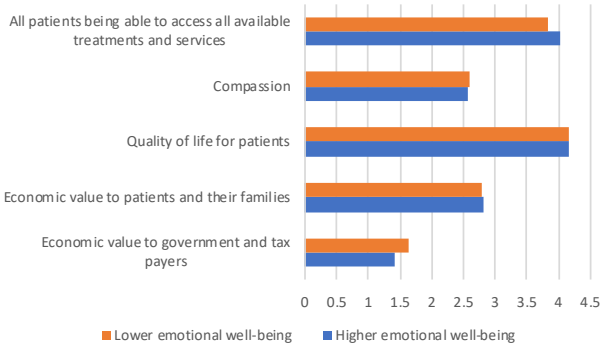


Figure 9.26: Values to consider on behalf of patients/families (weighted rank by emotional well-being)

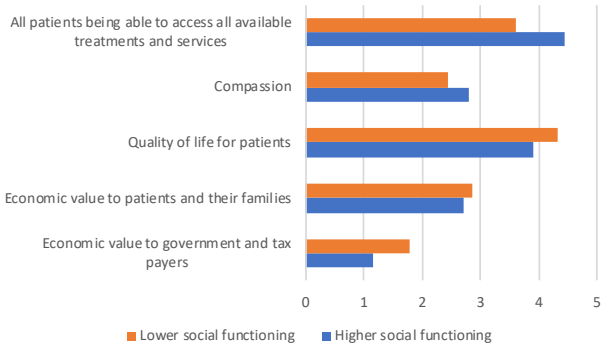


Figure 9.27: Values to consider on behalf of patients/families (weighted rank by social functioning)

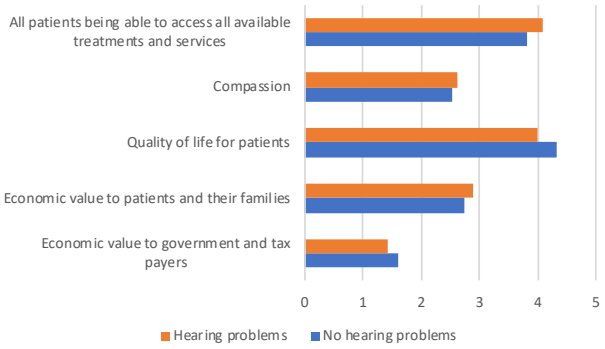


Figure 9.28: Values to consider on behalf of patients/families (weighted rank by hearing problems)

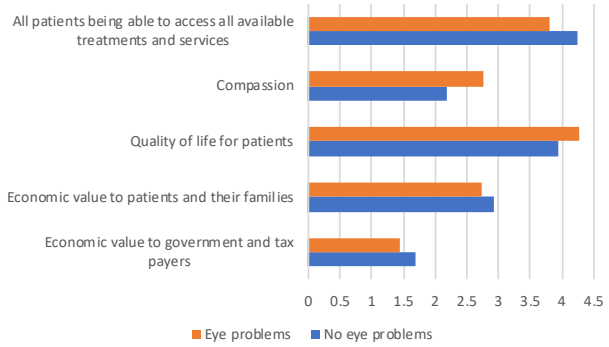


Figure 9.29: Values to consider on behalf of patients/families (weighted rank by eye problems)

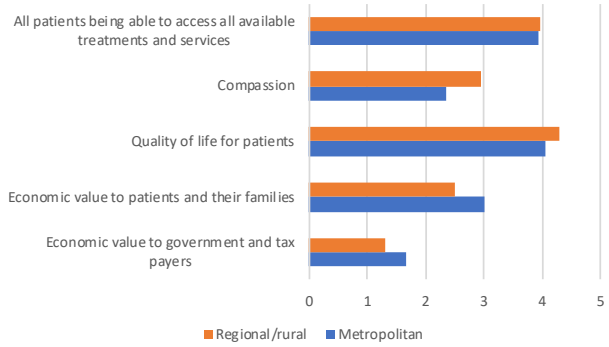


Figure 9.30: Values to consider on behalf of patients/families (weighted rank by location)

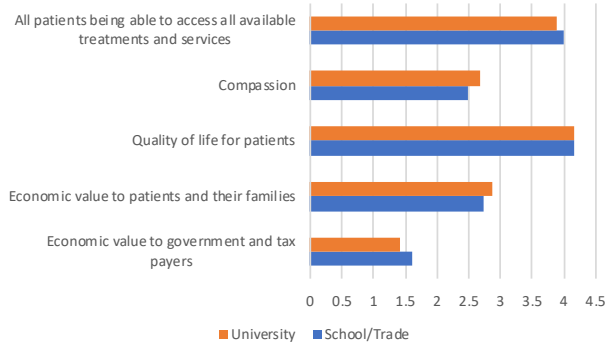


Figure 9.31: Values to consider on behalf of patients/families (weighted rank by education)

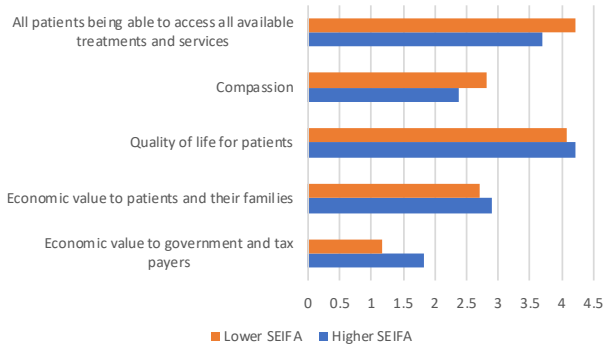


Figure 9.32: Values to consider on behalf of patients/families (weighted rank by SEIFA)

Expectation of information provision

Participants were asked what they would like to see in the future in relation to information provision. The most common theme was that participants described being satisfied with current information and therefore had no recommendation (n=11, 22.00%).

Participant describes being satisfied with current information. No recommendation.

No, I think that AMDF was one of the best examples of explaining a medical condition or a physical condition about the same. I don't think I need more than that. Participant 20

Well, seeing most of ours comes from the internet anyway or from, well, it's still on the emails from the Mitochondrial Foundation. No. Where we're, I don't think….What we've got's suitable. Participant 31

I know everything was...The internet was a great help. Then the AMDF, Australian Mitochondrial Federation has been a great help. They send all the literature in paper form so that was fine. I preferred that than an email. Participant 35

There were nine participants (18.00%) that described the need for information about their specific type of mitochondrial disease, and nine participants (18.00%) that described the need for healthcare professionals to deliver accurate, comprehensive and honest information (including prognostic information. There were also six participants (12.00%) that described the need for centralised and reliable information.

Participant describes the need for information about their specific type of mitochondrial disease

I think that map thing I was talking about before would be really useful. Some decision tree type thing online where you go like, "Have you just been diagnosed with mitochondrial disease?" "Yes." Then this is the basic information that you'll need to know. Then breaking it down further, instead of like, "Are you an adult or are you a parent? We'll read this information." Participant 5

I just like to see more on MELAS. There seemed to be lots…I don't know whether this is general, but I know that the Murdoch Institute when they have found stuff they were working on Leigh's disease, which I understand is a huge major thing. Never seems to be anything on MELAS as much. Maybe they've done all there is to know about MELAS. I don't know, but there's nothing ever seem to come up about MELAS. I have the vested interest in. Participant 21

Well, I suppose having a concise…Mitochondrial is so very…some in different strains, but perhaps some streamlining each different one so that you don't have to find out, get all the information about everything about mitochondria, just your specific strain would be the most helpful. Participant 48

Participant describes the need for healthcare professionals to deliver accurate, comprehensive and honest information (including prognostic information)

No, not really. Probably something that if it had interested me at the time or I didn't get any information on was having children. I've never wanted to have kids, which turned out to be a really good thing because I wouldn't be able to now anyway seeing that I would pass it on. If I was one of these people that had a raging maternal instinct and really wanted to have babies, that would have been something that should have been discussed when I was being diagnosed. Although, maybe somebody that's not me would have asked. Definitely, I was never asked what my thoughts were about having a family. That's probably something that needs to be talked about a little bit more and about testing, and stuff that maybe can be done before hand to say whether you're going to be passing stuff or not. Participant 10

Well, really and fairly, we don't get a lot of information from the health people or not at all. it's mainly what I read on the internet and whatnot so basically that hasn't changed much. I would like to, like in LOCATION, I'd probably like him to have got back to us like he said he was going to do and things like that and see what actually was in his study, any findings or whatever about that at all. That hasn't been followed up. I haven't heard anything. I have tried to look something up on the internet and haven't found anything. Participant 14

It's not a nice diagnosis. But it's a label that people can see, oh you've got something real. And then there's doctors that diagnose Mitochondrial Disease when it's not. And in fact, I know a patient, she was in contact with me. And she was told by a GP she had Mito. Anyway, she was recommended and referred to the LOCATION hospital neurologist there. And NAME had to tell her that she didn't have it, she was so angry. She was angry. Instead of being relieved and saying, "All right, well what do I do now? Who do I see to find out what I have." Anyway, so that's my thing. Education, widely and broadly. Participant 24

Yes, obviously if the patients had tests that come back and show this organ system or that organ system has been affected and it's obviously not working, it's not really sneakily work, that should be conveyed to the patient with a far more honest picture of where they likely to end up. What the process is likely to be so that when the patient or some patients would just wouldn't accept that they wouldn't keep it in the context of as it is, "I'm trying to help you. If you get this symptom it just says that a few more of your cells are not doing so well." Gives people realistic ...likely outcome that if something goes wrong with one of the organ systems, the patient isn't left thinking, "What on earth is wrong with me?" compared to a tiny that that's part of the course and not panic. It's not something else it's likely to be this. Participant 27

In relation to sub-group variations, participants with high general health (31.82%) described the need for healthcare professionals to deliver accurate, comprehensive and honest information (including prognostic information), more frequently than the general population (18.00%).

Table 9.3: Expectations of information provision

Expectations of future information	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant describes being satisfied with current information. No recommendation.	11	22.00	5	16.67	6	30.00	7	25.93	4	17.39
Participant describes the need for information about their specific type of mitochondrial disease	9	18.00	6	20.00	3	15.00	5	18.52	4	17.39
Participant describes the need for healthcare professionals to deliver accurate, comprehensive and honest information (including prognostic information)	9	18.00	6	20.00	4	20.00	6	22.22	4	17.39
Participant describes the need for centralised and reliable information	6	12.00	3	10.00	3	15.00	4	14.81	2	8.70
Participant describes the need for information in relation to genetic predisposition and/or hereditary factors	4	8.00	2	6.67	2	10.00	3	11.11	1	4.35
Participant describes the need to increase public awareness with more information available on the condition	4	8.00	3	10.00	2	10.00	5	18.52	0	0.00
Participants describes the need to provide information about research programs for treatments	4	8.00	2	6.67	2	10.00	3	11.11	1	4.35
Participant recommends providing education programs for front line healthcare professionals to be aware of mitochondrial disease	4	8.00	4	13.33	0	0.00	2	7.41	2	8.70

Expectations of future information	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant describes being satisfied with current information. No recommendation.	11	22.00	5	19.23	6	25.00	4	16.67	6	17.65
Participant describes the need for information about their specific type of mitochondrial disease	9	18.00	5	19.23	4	16.67	2	8.33	6	17.65
Participant describes the need for healthcare professionals to deliver accurate, comprehensive and honest information (including prognostic information)	9	18.00	5	19.23	5	20.83	4	16.67	7	20.59
Participant describes the need for centralised and reliable information	6	12.00	4	15.38	2	8.33	5	20.83	4	11.76
Participant describes the need for information in relation to genetic predisposition and/or hereditary factors	4	8.00	2	7.69	2	8.33	2	8.33	4	11.76
Participant describes the need to increase public awareness with more information available on the condition	4	8.00	3	11.54	2	8.33	3	12.50	3	8.82
Participants describes the need to provide information about research programs for treatments	4	8.00	4	15.38	0	0.00	3	12.50	4	11.76
Participant recommends providing education programs for front line healthcare professionals to be aware of mitochondrial disease	4	8.00	1	3.85	3	12.50	3	12.50	3	8.82

Expectations of future information	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant describes being satisfied with current information. No recommendation.	11	22.00	4	18.18	7	25.00	5	19.23	6	25.00
Participant describes the need for information about their specific type of mitochondrial disease	9	18.00	3	13.64	6	21.43	4	15.38	5	20.83
Participant describes the need for healthcare professionals to deliver accurate, comprehensive and honest information (including prognostic information)	9	18.00	4	18.18	6	21.43	7	26.92	3	12.50
Participant describes the need for centralised and reliable information	6	12.00	4	18.18	2	7.14	2	7.69	4	16.67
Participant describes the need for information in relation to genetic predisposition and/or hereditary factors	4	8.00	1	4.55	3	10.71	4	15.38	0	0.00
Participant describes the need to increase public awareness with more information available on the condition	4	8.00	2	9.09	3	10.71	4	15.38	1	4.17
Participants describes the need to provide information about research programs for treatments	4	8.00	3	13.64	1	3.57	2	7.69	2	8.33
Participant recommends providing education programs for front line healthcare professionals to be aware of mitochondrial disease	4	8.00	1	4.55	3	10.71	3	11.54	1	4.17

Expectations of future information	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant describes being satisfied with current information. No recommendation.	11	22.00	3	15.00	8	26.67	5	22.73	6	21.43
Participant describes the need for information about their specific type of mitochondrial disease	9	18.00	4	20.00	5	16.67	3	13.64	6	21.43
Participant describes the need for healthcare professionals to deliver accurate, comprehensive and honest information (including prognostic information)	9	18.00	3	15.00	7	23.33	7	31.82	3	10.71
Participant describes the need for centralised and reliable information	6	12.00	2	10.00	4	13.33	2	9.09	4	14.29
Participant describes the need for information in relation to genetic predisposition and/or hereditary factors	4	8.00	2	10.00	2	6.67	2	9.09	2	7.14
Participant describes the need to increase public awareness with more information available on the condition	4	8.00	3	15.00	2	6.67	2	9.09	3	10.71
Participants describes the need to provide information about research programs for treatments	4	8.00	3	15.00	1	3.33	2	9.09	2	7.14
Participant recommends providing education programs for front line healthcare professionals to be aware of mitochondrial disease	4	8.00	2	10.00	2	6.67	1	4.55	3	10.71

Expectations of future information	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant describes being satisfied with current information. No recommendation.	11	22.00	0	0.00	2	14.29	2	22.22	4	36.36	3	30.00
Participant describes the need for information about their specific type of mitochondrial disease	9	18.00	2	33.33	3	21.43	2	22.22	2	18.18	0	0.00
Participant describes the need for healthcare professionals to deliver accurate, comprehensive and honest information (including prognostic information)	9	18.00	3	50.00	2	14.29	0	0.00	3	27.27	2	20.00
Participant describes the need for centralised and reliable information	6	12.00	1	16.67	1	7.14	1	11.11	1	9.09	2	20.00
Participant describes the need for information in relation to genetic predisposition and/or hereditary factors	4	8.00	0	0.00	2	14.29	0	0.00	1	9.09	1	10.00
Participant describes the need to increase public awareness with more information available on the condition	4	8.00	2	33.33	1	7.14	0	0.00	0	0.00	2	20.00
Participants describes the need to provide information about research programs for treatments	4	8.00	0	0.00	0	0.00	2	22.22	1	9.09	1	10.00
Participant recommends providing education programs for front line healthcare professionals to be aware of mitochondrial disease	4	8.00	1	16.67	2	14.29	1	11.11	0	0.00	0	0.00

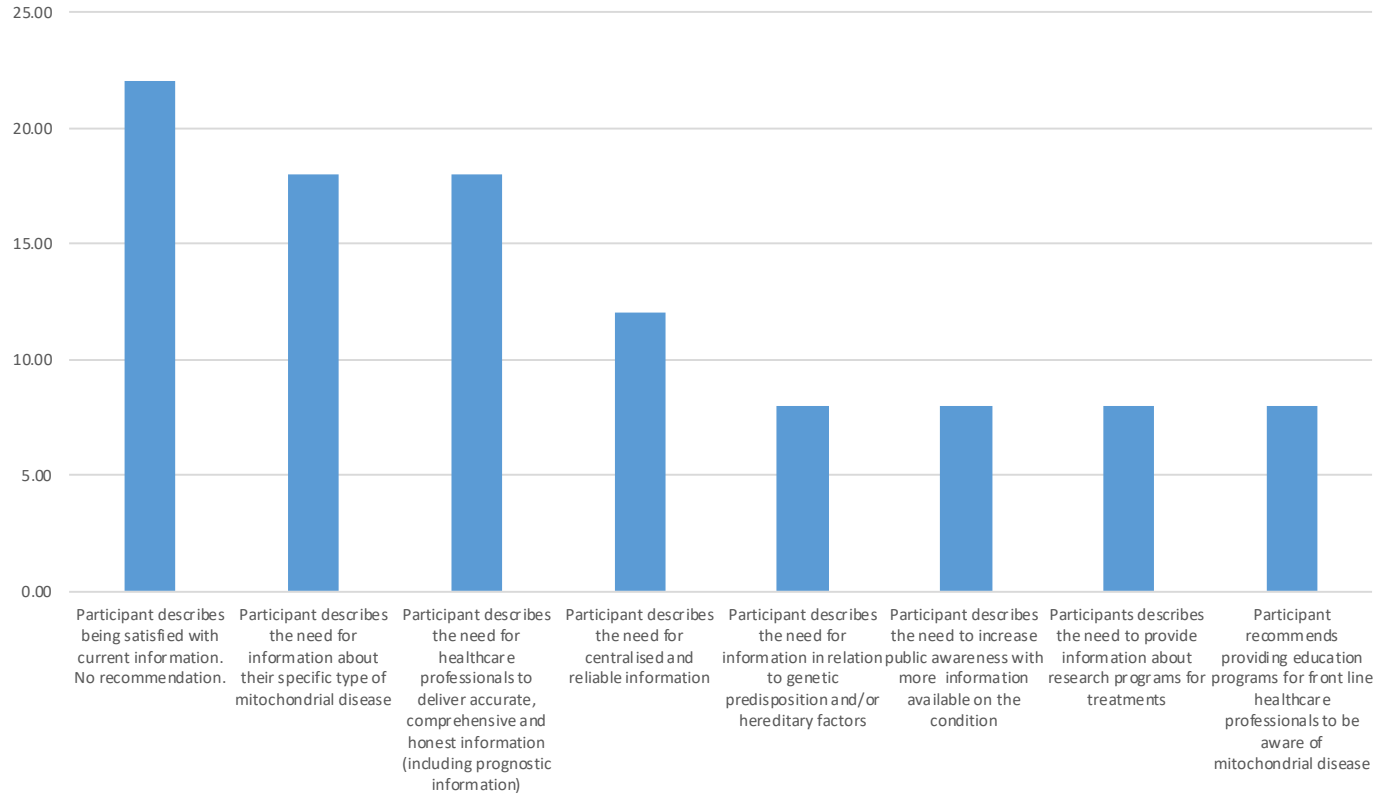


Figure 9.33: Expectations of information provision (% of all participants)

Expectation of health professional communication

Participants were asked whether there was anything they would like to see improved in the future in relation to the way that health professionals communicate with patients. The most common theme was that participants recommend healthcare professional education in relation to mitochondrial disease and more understanding of the impact and implications of the condition (n=16, 32.00%).

Participant recommends healthcare professional education in relation to mitochondrial disease and more understanding of the impact and implications of the condition

Well, obviously GPs and other health specialists have limited to negligible knowledge about the disease. If there was a brochure that was distributed to every other doctor or was put in the GP's magazine, a summary update on it regularly. Because I'm sick of telling doctors what it is, or what I can't take and what I can take … I'm sick of having that fight with doctors, and then they think they'll tell me this that and the rest and they don't anymore. Participant 3

Yes. Definitely. I think, for example, and I think it comes back to educate and inform. For example, the GP who just said, "I don't know anything about it. I don't have a booklet. Well, I don't know what to do." I thought, well, you know, that's really not good enough, and I think that problem is GPs are overburdened already. It's that whole vicious circle that is often associated with medical professionals, so they need to educate general practitioners, educate specialists. Participant 7

Well I'd like all the Doctors that I see to understand more. A lot of them don't understand what my limitations and stuff are. I feel a bit alone. I had to instigate to get my mobility sticker, I had to ask. "Oh, well what do you need that for?" Then I had to ask how do I get a key for disabled toilet access. "Oh, they don't have keys do they?" "Yes, they do. How do I do it?" Everything's been a struggle and I've had to instigate it. Participant 36

This was followed by the recommendation that healthcare professionals are more proactive and attentive (n=9, 18.00%).

Participant recommends healthcare professionals being more proactive and attentive

I think you just get told you have an appointment at a day at a particular date, but knowing what to expect out of that appointment, it would also be useful. Sometimes, you get really high expectations over something and it's not helpful at all. I know they're specialists, but it would be good sometimes if they did think about or at least bothered to ask like, "Are you getting support here or there?" Who else is going to ask. Participant 5

Yes. That they understand it. That they are able to be, basically ... speak to their clients and understand what their clients want, what their patients need from them. Participant 40

[laughs] One thing, they never ask questions about what your symptoms are and how it's affecting you. You have to go in there with a problem and they try and scratch their head and see what they can do about it. They should..they don't come up with a plan. I actually ask the GP after a bit of management and she just shook her head and said, "I don't know." Participant 42

There were also nine participants (18.00%) that did not have a recommendation as they have been satisfied with communication. Where participants were satisfied with communication it was primarily because communication had been open communication.

Participant does not have a recommendation as they have been satisfied with communication (Open communication)

No because my professor had been really good. He's been really open with me. He's never given up on me. He never gave up looking for what I had. He is always been open with me. Participant 12

I'd say most healthcare professionals I've dealt with have been pretty good. I think more and more these days they are more patient centered than they once were, so they actually do listen to the patient more than they once might have. Generally, I think the help has been in the right direction. Participant 13

No, I think I've had pretty good communication really. My GP always gives me a print out of any tests I've had done. My, what do you call it? Everything to-date that's on my file, I know there are a lot of problems with, what's the health record that you're supposed to be able to access online, and all that sort of stuff? Participant 34

There were seven participants (14.00%) that recommended that healthcare professionals need to have more empathy.

Participant recommends healthcare professionals need to have more empathy

Just that the health professionals…I don't know is a bit of a understanding of what we go through. Where there's condition. I reckon it also needs to be more info with the general community to be aware of what we go through and then. Participant 6

Just a little bit more understanding when you say something that they actually believe you rather than not believing that what you said is true, but I guess that's just a matter of finding the right doctor. Participant 19

Yes, no. I definitely would I think and I've had run ins over the years with various ones and thankfully, well, they've basically got to the point ... there was on health care professional I refused to see again because I think some times and this happens in some of these younger people that are very knowledgeable. They know the ins and the outs of the condition but they forget that they're talking to a parent and that your child is just a not a case study. It's actually your child. Participant 45

In relation to sub-group variations, participants from rural areas (45.00%) and those from low socio-economic areas (43.48%) recommended healthcare professional education in relation to mitochondrial disease and more understanding of the impact and implications of the condition, more frequently than the general population (32.00%).

Table 9.4: Expectations of health professional communication

Expectations of future health professional communication	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant recommends healthcare professional education in relation to mitochondrial disease and more understanding of the impact and implications of the condition	16	32.00	8	26.67	9	45.00	7	25.93	10	43.48
Participant recommends healthcare professionals being more proactive and attentive	9	18.00	6	20.00	3	15.00	5	18.52	4	17.39
Participant does not have a recommendation as they have been satisfied with communication (Open communication)	9	18.00	5	16.67	4	20.00	4	14.81	5	21.74
Participant recommends healthcare professionals need to have more empathy	7	14.00	3	10.00	4	20.00	4	14.81	3	13.04
Participant recommends centralised and coordinated care across specialists and allied health professionals (including more communication between doctors)	6	12.00	3	10.00	3	15.00	3	11.11	3	13.04
Participant recommends that healthcare professionals ensure information is easily accessible	6	12.00	4	13.33	2	10.00	2	7.41	4	17.39

Expectations of future health professional communication	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant recommends healthcare professional education in relation to mitochondrial disease and more understanding of the impact and implications of the condition	16	32.00	10	38.46	7	29.17	9	37.50	10	29.41
Participant recommends healthcare professionals being more proactive and attentive	9	18.00	4	15.38	5	20.83	6	25.00	7	20.59
Participant does not have a recommendation as they have been satisfied with communication (Open communication)	9	18.00	4	15.38	5	20.83	3	12.50	7	20.59
Participant recommends healthcare professionals need to have more empathy	7	14.00	3	11.54	4	16.67	4	16.67	6	17.65
Participant recommends centralised and coordinated care across specialists and allied health professionals (including more communication between doctors)	6	12.00	3	11.54	3	12.50	3	12.50	4	11.76
Participant recommends that healthcare professionals ensure information is easily accessible	6	12.00	1	3.85	5	20.83	2	8.33	5	14.71

Expectations of future health professional communication	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant recommends healthcare professional education in relation to mitochondrial disease and more understanding of the impact and implications of the condition	16	32.00	8	36.36	9	32.14	7	26.92	10	41.67
Participant recommends healthcare professionals being more proactive and attentive	9	18.00	3	13.64	6	21.43	5	19.23	4	16.67
Participant does not have a recommendation as they have been satisfied with communication (Open communication)	9	18.00	4	18.18	5	17.86	6	23.08	3	12.50
Participant recommends healthcare professionals need to have more empathy	7	14.00	4	18.18	3	10.71	5	19.23	2	8.33
Participant recommends centralised and coordinated care across specialists and allied health professionals (including more communication between doctors)	6	12.00	1	4.55	5	17.86	3	11.54	3	12.50
Participant recommends that healthcare professionals ensure information is easily accessible	6	12.00	3	13.64	3	10.71	2	7.69	4	16.67

Expectations of future health professional communication	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant recommends healthcare professional education in relation to mitochondrial disease and more understanding of the impact and implications of the condition	16	32.00	7	35.00	10	33.33	8	36.36	9	32.14
Participant recommends healthcare professionals being more proactive and attentive	9	18.00	4	20.00	5	16.67	2	9.09	7	25.00
Participant does not have a recommendation as they have been satisfied with communication (Open communication)	9	18.00	5	25.00	4	13.33	2	9.09	7	25.00
Participant recommends healthcare professionals need to have more empathy	7	14.00	3	15.00	4	13.33	3	13.64	4	14.29
Participant recommends centralised and coordinated care across specialists and allied health professionals (including more communication between doctors)	6	12.00	2	10.00	4	13.33	2	9.09	4	14.29
Participant recommends that healthcare professionals ensure information is easily accessible	6	12.00	2	10.00	4	13.33	4	18.18	2	7.14

Expectations of future health professional communication	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant recommends healthcare professional education in relation to mitochondrial disease and more understanding of the impact and implications of the condition	16	32.00	2	33.33	4	28.57	3	33.33	4	36.36	4	40.00
Participant recommends healthcare professionals being more proactive and attentive	9	18.00	1	16.67	2	14.29	3	33.33	2	18.18	1	10.00
Participant does not have a recommendation as they have been satisfied with communication (Open communication)	9	18.00	0	0.00	2	14.29	2	22.22	3	27.27	2	20.00
Participant recommends healthcare professionals need to have more empathy	7	14.00	1	16.67	2	14.29	2	22.22	2	18.18	0	0.00
Participant recommends centralised and coordinated care across specialists and allied health professionals (including more communication between doctors)	6	12.00	1	16.67	3	21.43	1	11.11	0	0.00	1	10.00
Participant recommends that healthcare professionals ensure information is easily accessible	6	12.00	0	0.00	2	14.29	0	0.00	1	9.09	3	30.00

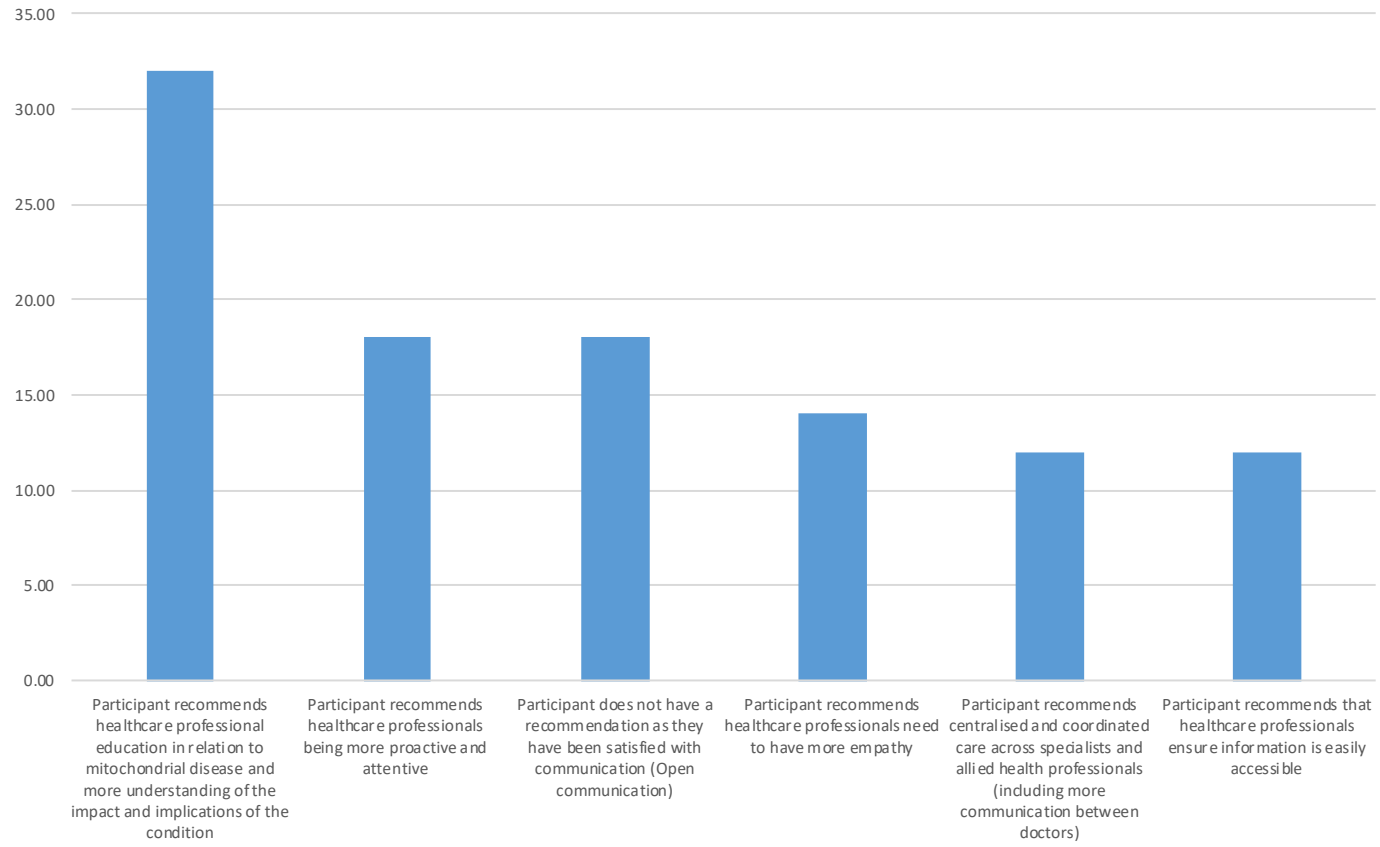


Figure 9.34: Expectations of health professional communication (% of all participants)

Expectation of care and support

Participants were asked whether there was anything they would like to see in relation to the care and support they receive. The most common recommendation was for centralised and coordinated care across specialists and allied health professionals (including more communication between doctors) (n=13, 26.00%). In a similar theme, there were also six participants (12.00%) that recommended caseworkers be employed to support patients navigate health, medical and emotional needs.

Participant recommends centralised and coordinated care across specialists and allied health professionals (including more communication between doctors)

Yes. I think a network in LOCATION METROPOLITAN would be useful and having some sort of specialist. Somebody, even if it's in the north or northwest of the state, somebody who is trained in this area would be really useful. Participant 26

I would love to see specialist educators set up that anyone can access. For example, they have contact staff who can give specific advice and help you understand things when you have cancer, diabetes, Chronic pain, mental health, etc. I would like to see the same for Mito, with different ones for paediatrics and others for adults. Participant 30

Yes. Well, I think there is already the one, the LOCATION Hospital which has a statewide service. I think it's pretty good. It's very difficult having so many different people to deal with. It is a bit conky sometimes and there's a lot of wasting time in between things. I've been on a wait for a motorized scooter. It's just never appeared. How long do you think, "Well, what's really going on here?" Because of my situation, I'm okay. I don't know what it would be like if you were really desperate. Participant 34

Participant recommends caseworkers be employed to support patients navigate health, medical and emotional needs

Yeah, just someone to, some health, somewhere you can actually go to, and how can I improve my condition, etc., sort of like being diagnosed and then you're left to your own, I guess is it…Yeah, so someone that, okay, well, even if that person can put you onto other things. Sometimes, also, you know, it would have been great to be able to talk to people that have certainly my type of condition, because, as you know, there's a lot of variances. People with my hearing and diabetes being able to have a, sort of, group

discussion about how they cope with it, etc. those sort of things. Like subcategories, especially mito, area support groups. Participant 15

There should also be a social work help line which you can contact in your state to get support for sourcing equipment, funding, medical specialists, etc. Participant 30

Yeah I think social ... I haven't seen a social worker. I haven't had any assistance with setting up all these things that I've had to set up. I know I'm supposed to have seen social workers and speech pathologists and all the rest of it, and genetic counsellors, but that just hasn't happened. I know that there are things out there, and people out there that I could be seeing that might know more about treatment options, but I really haven't got there yet. Participant 36

This was followed by the recommendation for support groups to help patients noting that it is difficult due to the diversity within the patient population (n=7, 14.00%) and more equity in access to services and support for adults with rare disease (n=7, 14.00%).

Participant recommends support groups to help patients noting that it is difficult due to the diversity within the patient population

The AMDF started a support groups, that would have been really beneficial to me in the early days, but because of the AMDF I only started the same year that I was diagnosed, really there. That stuff is more readily available now, which I would have found helpful back in the day. Participant 10

Sometimes, also, you know, it would have been great to be able to talk to people that have certainly my type of condition, because, as you know, there's a lot of variances. People with my hearing and diabetes being able to have a, sort of, group discussion about how they cope with it, etc. those sort of things. Like subcategories, especially mito, area support groups. Participant 15

Probably, when we first…when I was first diagnosed to understand it, perhaps a support group where you could go and have a big whinge with other people that are going through the same thing as you. Participant 41

Participant recommends more equity in access to services and support for adults with rare disease

Well, I think we have to move away from the charity system because that's the same with the problems that they're finding the funding between what was the state government and now the federal with NDIS. People are still locked into…how do I explain? Someone else nation the cost of things with like the NDIS won't they're selective and what they fund and so people are still being advised and works for children, children's charities raised lots of money that charities are provided and therefore you have to be grateful or you have to be in need and it's not a good system. I think as a society we could do a lot better than that. It's such ad hoc like that it's not equitable. You can have two people living next door to each other and one tubal child will get all that extra bit of funding and thing because they're newsworthy and people feel good about doing things which is great charities good in that sense but an adult with the same disease doesn't get anything because then they're not the cute child but they're still impacted by the disorder or disease. So, yes, I have lots of issues with charities and things like that. Mitochondrial disease, I don't know, analytics change because we don't have access like to it with NAME. There's nothing in the public health system for it. For individual's, like a body part, like you can just take a gastrointestinal system along with it, there's nothing. There's not a holistic thing. I don't know if there is purely for people that might just have something, that's a different thing. I don't know. Participant 4

I guess it's really just having a well-funded NDIS really would solve it. Making sure that all services--anything that someone needs, they have access to. I think it still takes…I still know few people who're just…it would just be fantastic if I they had a laptop computer in school for them, but it would take a year or two of applications here and applications there before they actually get it. I guess it's just that old people…whether it's me or young people at school with a disability or the mitochondrial disease, get the services that they require whenever they may be. Participant 13

In relation to sub-group variations, participants with a university education (50.00%) and those with a hearing impairment (45.83%) recommended centralised and coordinated care across specialists and allied health professionals, more frequently than the general population (26.00%), while those with a high school or trade education (3.85%) recommended this less frequently.

Table 9.5: Expectations of care and support

Expectations of future care and support	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant recommends centralised and coordinated care across specialists and allied health professionals (including more communication between doctors)	13	26.00	7	23.33	6	30.00	9	33.33	4	17.39
Participant recommends support groups to help patients noting that it is difficult due to the diversity within the patient population	7	14.00	5	16.67	2	10.00	6	22.22	1	4.35
Participant recommends more equity in access to services and support for adults with rare disease	7	14.00	5	16.67	2	10.00	4	14.81	3	13.04
Participant recommends caseworkers be employed to support patients navigate health, medical and emotional needs	6	12.00	3	10.00	3	15.00	1	3.70	5	21.74
Participant recommends greater and/or more access to home care/support at home	4	8.00	3	10.00	1	5.00	3	11.11	1	4.35
Participant recommends having reliable information available (via patient organisations)	4	8.00	2	6.67	2	10.00	2	7.41	2	8.70

Expectations of future care and support	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant recommends centralised and coordinated care across specialists and allied health professionals (including more communication between doctors)	13	26.00	1	3.85	12	50.00	11	45.83	7	20.59
Participant recommends support groups to help patients noting that it is difficult due to the diversity within the patient population	7	14.00	5	19.23	2	8.33	3	12.50	5	14.71
Participant recommends more equity in access to services and support for adults with rare disease	7	14.00	3	11.54	4	16.67	4	16.67	5	14.71
Participant recommends caseworkers be employed to support patients navigate health, medical and emotional needs	6	12.00	1	3.85	5	20.83	5	20.83	4	11.76
Participant recommends greater and/or more access to home care/support at home	4	8.00	3	11.54	1	4.17	2	8.33	2	5.88
Participant recommends having reliable information available (via patient organisations)	4	8.00	2	7.69	2	8.33	2	8.33	4	11.76

Expectations of future care and support	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant recommends centralised and coordinated care across specialists and allied health professionals (including more communication between doctors)	13	26.00	6	27.27	7	25.00	8	30.77	5	20.83
Participant recommends support groups to help patients noting that it is difficult due to the diversity within the patient population	7	14.00	4	18.18	3	10.71	5	19.23	2	8.33
Participant recommends more equity in access to services and support for adults with rare disease	7	14.00	3	13.64	4	14.29	4	15.38	3	12.50
Participant recommends caseworkers be employed to support patients navigate health, medical and emotional needs	6	12.00	3	13.64	3	10.71	4	15.38	2	8.33
Participant recommends greater and/or more access to home care/support at home	4	8.00	1	4.55	3	10.71	1	3.85	3	12.50
Participant recommends having reliable information available (via patient organisations)	4	8.00	3	13.64	1	3.57	1	3.85	3	12.50

Expectations of future care and support	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant recommends centralised and coordinated care across specialists and allied health professionals (including more communication between doctors)	13	26.00	4	20.00	9	30.00	6	27.27	7	25.00
Participant recommends support groups to help patients noting that it is difficult due to the diversity within the patient population	7	14.00	5	25.00	2	6.67	4	18.18	3	10.71
Participant recommends more equity in access to services and support for adults with rare disease	7	14.00	2	10.00	5	16.67	2	9.09	5	17.86
Participant recommends caseworkers be employed to support patients navigate health, medical and emotional needs	6	12.00	2	10.00	4	13.33	5	22.73	1	3.57
Participant recommends greater and/or more access to home care/support at home	4	8.00	0	0.00	4	13.33	1	4.55	3	10.71
Participant recommends having reliable information available (via patient organisations)	4	8.00	3	15.00	1	3.33	2	9.09	2	7.14

Expectations of future care and support	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant recommends centralised and coordinated care across specialists and allied health professionals (including more communication between doctors)	13	26.00	0	0.00	3	21.43	3	33.33	6	54.55	1	10.00
Participant recommends support groups to help patients noting that it is difficult due to the diversity within the patient population	7	14.00	1	16.67	1	7.14	2	22.22	1	9.09	2	20.00
Participant recommends more equity in access to services and support for adults with rare disease	7	14.00	1	16.67	2	14.29	3	33.33	1	9.09	0	0.00
Participant recommends caseworkers be employed to support patients navigate health, medical and emotional needs	6	12.00	1	16.67	3	21.43	1	11.11	1	9.09	0	0.00
Participant recommends greater and/or more access to home care/support at home	4	8.00	1	16.67	0	0.00	0	0.00	2	18.18	1	10.00
Participant recommends having reliable information available (via patient organisations)	4	8.00	0	0.00	2	14.29	0	0.00	1	9.09	1	10.00

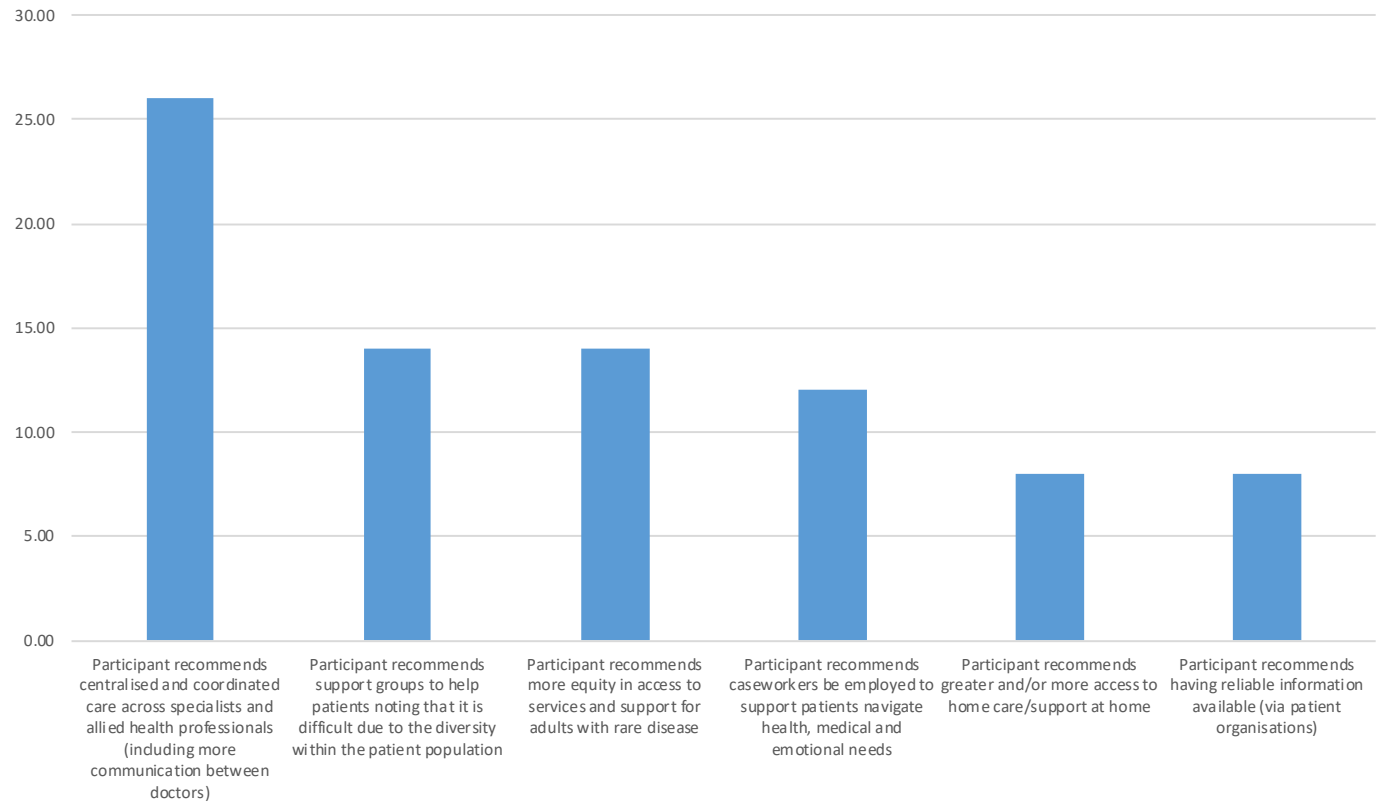


Figure 9.35: Expectations of care and support (% of all participants)

What participants are grateful for in the Australian health system

Participants were asked what they were grateful for in relation to the Australian health system. The most common theme was participants describing being grateful for Medicare in relation to access to specialists (n=17, 34.00%), followed by being grateful for the compassion and support shown by healthcare professionals (n=16, 32.00%).

Participant describes being grateful for Medicare (Access to specialists)

I think some of the small things have actually been the most useful like before I was diagnosed, when my rheumatologist said, "I don't know what you have. I'm going to refer you to a neurologist." That referral system, that made a very huge difference to my life, if she just said, "Oh, no. I think you've got fibromyalgia." I'd feel wonderful I have fibromyalgia, but wondering in the back of my head whether or not I need a second opinion. I think having very qualified people, but also refer to other very qualified people is really…I think the quality generally is very good. My doctor is amazing, I love her. She's great. Participant 5

Yes, it's easy to access to a specialist. That's a big plus and it is as…though this is become as known and treatment becomes clear, you find, technically, more and more specialists. But after I fight this disease, but speaking for myself, I can't speak loudly enough of the medical people at the hospital. Participant 23

The doctors I've seen have been excellent and it's affordable. I can go and see my neurologist and I can afford it. Participant 43

Opportunities, new technologies and stuff, and surgeries that we've had the opportunity to consider through our specialist. That's been especially good. Participant 47

Participant describes being grateful for the compassion and support shown by healthcare professionals

I think that being grateful, and it cost me a lot of money to get to the endpoint, I think some of the ability of the doctors, etc., continuing to try to get to the bottom of things rather than just, "Oh, well, I'm not quite sure how to go." Participant 15

I'm very pleased with the attitude of the workers in the health system. They're very kind and considerate. Thinking now, they've been rather generous with the benefits that I get, like a wheelchair…Participant 17

Yes. I have been very grateful for the expertise of the medical staff and their compassion. We understand they are trying to do whatever they can. There's nothing else really that they can do at this point in time. Participant 48

There were 10 participants (20.00%) that described being grateful for Medicare in relation to access to allied health professionals and seven participants (14.00%) described being grateful for their healthcare card and the financial relief it provides.

Participant describes being grateful for Medicare (Access to allied health professionals)

Early intervention. Early intervention. Absolutely that would be the number one thing. The physical, the OT, and the speech therapy. [Interviewer: He's improved?] Yeah, absolutely. We've had that since 11 months old, and I think that's been the biggest. Participant 46

It's really good being a pensioner and getting medication fairly cheaply, and it's really good being bulk-billed so I can go to the doctor any time. It's good having the Medicare plan to see psychologists at reduced rates. That's all been really good. Participant 47

I think the way they do things in the children's hospital where they gathered a team together, I think it was under the heading of Adolescent Medicine. It was, I think, a brilliant approach because this affects all body systems in every body system. I think that having a team around all of those systems accessible, people who are willing to be involved in mitochondrial care and understanding approach is invaluable. Participant 49

Participant describes being grateful for their healthcare card and the financial relief it provides

DVA have been wonderful. They sent the occupational therapy who happens to be the same lady who used to come to NAME. It's all very, very good. Their help for me has been invaluable. Participant 32

Yes, I've been very grateful that I was put onto disability support pension. Say in England, the National Health Service there is so overstretched. I know that they're doing some fantastic research over there, but to actually get assistance at home, I don't think that would have happened. Participant 34

Oh, I think Medicare's fantastic. Having a Health Care Card, having ... I've seen a lot of specialists, and they're hundreds of dollars, and it's all added up to thousands of dollars, and I just don't have any more

now that I'm not working. It's fantastic that- ... Medicare covers a lot of things, and Health Care Card helps with the travel costs and stuff like that. Participant 36

Other aspects of the health system that participants spoke about being grateful for were subsidised diagnostic tests (n=6, 12.00%), government initiatives that support ongoing health and quality of life (for example NDIS, Better Start Program and At home nursing services) (n=6, 12.00%) and the quality of specialist expertise in Australia (n=5, 10.00%).

In relation to sub-group variations, participants from rural areas (45.00%) described being grateful for Medicare (Access to specialists) more frequently than the general population (34.00%). Participants with a university education (45.83%), those with high physical function (54.55%), and those with high general health (50.00%) reported being grateful for the compassion and support shown by healthcare professionals more frequently than the general population (32.00%), while those with low physical function (17.86%) reported this less frequently. Participants from rural areas (25.00%), those with a hearing impairment (25.00%) and those with low physical function (25.00%) described being grateful for their healthcare card and the financial relief it provides, more frequently than the general population (14.00%), while there we no participants with high physical function (0.00%) that reported this.

Table 9.6: Aspects of the Australian health system that participants are grateful for

Aspects of the Australian health system that patients are grateful for	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant describes being grateful for Medicare (Access to specialists)	17	34.00	8	26.67	9	45.00	7	25.93	10	43.48
Participant describes being grateful for the compassion and support shown by healthcare professionals	16	32.00	11	36.67	6	30.00	9	33.33	8	34.78
Participant describes being grateful for Medicare (Access to allied health professionals)	10	20.00	4	13.33	6	30.00	3	11.11	7	30.43
Participant describes being grateful for their healthcare card and the financial relief it provides	7	14.00	2	6.67	5	25.00	3	11.11	4	17.39
Participant describes being grateful for subsidised diagnostic tests	6	12.00	4	13.33	2	10.00	5	18.52	1	4.35
Participant describes being grateful for government initiatives that support ongoing health and quality of life (for example NDIS, Better Start Program and At home nursing services)	6	12.00	3	10.00	3	15.00	5	18.52	1	4.35
Participant describes frustration at the lack of specialists and specialised services in regional areas and the financial cost incurred when travelling to metropolitan areas for care	5	10.00	2	6.67	3	15.00	2	7.41	3	13.04
Participant describes frustration at the lack of services tailored towards mitochondria disease	5	10.00	4	13.33	1	5.00	4	14.81	1	4.35
Participant describes being grateful for the quality of specialist expertise in Australia	5	10.00	5	16.67	0	0.00	3	11.11	2	8.70

Aspects of the Australian health system that patients are grateful for	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant describes being grateful for Medicare (Access to specialists)	17	34.00	7	26.92	10	41.67	10	41.67	10	29.41
Participant describes being grateful for the compassion and support shown by healthcare professionals	16	32.00	6	23.08	11	45.83	9	37.50	10	29.41
Participant describes being grateful for Medicare (Access to allied health professionals)	10	20.00	4	15.38	6	25.00	5	20.83	8	23.53
Participant describes being grateful for their healthcare card and the financial relief it provides	7	14.00	2	7.69	5	20.83	6	25.00	4	11.76
Participant describes being grateful for subsidised diagnostic tests	6	12.00	4	15.38	2	8.33	3	12.50	4	11.76
Participant describes being grateful for government initiatives that support ongoing health and quality of life (for example NDIS, Better Start Program and At home nursing services)	6	12.00	5	19.23	1	4.17	4	16.67	6	17.65
Participant describes frustration at the lack of specialists and specialised services in regional areas and the financial cost incurred when travelling to metropolitan areas for care	5	10.00	3	11.54	2	8.33	1	4.17	2	5.88
Participant describes frustration at the lack of services tailored towards mitochondria disease	5	10.00	2	7.69	3	12.50	3	12.50	2	5.88
Participant describes being grateful for the quality of specialist expertise in Australia	5	10.00	1	3.85	4	16.67	1	4.17	3	8.82

Aspects of the Australian health system that patients are grateful for	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant describes being grateful for Medicare (Access to specialists)	17	34.00	6	27.27	11	39.29	10	38.46	7	29.17
Participant describes being grateful for the compassion and support shown by healthcare professionals	16	32.00	12	54.55	5	17.86	8	30.77	9	37.50
Participant describes being grateful for Medicare (Access to allied health professionals)	10	20.00	3	13.64	7	25.00	6	23.08	4	16.67
Participant describes being grateful for their healthcare card and the financial relief it provides	7	14.00	0	0.00	7	25.00	3	11.54	4	16.67
Participant describes being grateful for subsidised diagnostic tests	6	12.00	5	22.73	1	3.57	5	19.23	1	4.17
Participant describes being grateful for government initiatives that support ongoing health and quality of life (for example NDIS, Better Start Program and At home nursing services)	6	12.00	2	9.09	4	14.29	4	15.38	2	8.33
Participant describes frustration at the lack of specialists and specialised services in regional areas and the financial cost incurred when travelling to metropolitan areas for care	5	10.00	0	0.00	5	17.86	1	3.85	4	16.67
Participant describes frustration at the lack of services tailored towards mitochondria disease	5	10.00	3	13.64	2	7.14	2	7.69	3	12.50
Participant describes being grateful for the quality of specialist expertise in Australia	5	10.00	3	13.64	2	7.14	3	11.54	2	8.33

Aspects of the Australian health system that patients are grateful for	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant describes being grateful for Medicare (Access to specialists)	17	34.00	6	30.00	11	36.67	6	27.27	11	39.29
Participant describes being grateful for the compassion and support shown by healthcare professionals	16	32.00	6	30.00	11	36.67	11	50.00	6	21.43
Participant describes being grateful for Medicare (Access to allied health professionals)	10	20.00	5	25.00	5	16.67	4	18.18	6	21.43
Participant describes being grateful for their healthcare card and the financial relief it provides	7	14.00	0	0.00	7	23.33	2	9.09	5	17.86
Participant describes being grateful for subsidised diagnostic tests	6	12.00	5	25.00	1	3.33	4	18.18	2	7.14
Participant describes being grateful for government initiatives that support ongoing health and quality of life (for example NDIS, Better Start Program and At home nursing services)	6	12.00	3	15.00	3	10.00	1	4.55	5	17.86
Participant describes frustration at the lack of specialists and specialised services in regional areas and the financial cost incurred when travelling to metropolitan areas for care	5	10.00	1	5.00	4	13.33	2	9.09	3	10.71
Participant describes frustration at the lack of services tailored towards mitochondria disease	5	10.00	2	10.00	3	10.00	4	18.18	1	3.57
Participant describes being grateful for the quality of specialist expertise in Australia	5	10.00	2	10.00	3	10.00	2	9.09	3	10.71

Aspects of the Australian health system that patients are grateful for	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant describes being grateful for Medicare (Access to specialists)	17	34.00	2	33.33	7	50.00	1	11.11	4	36.36	3	30.00
Participant describes being grateful for the compassion and support shown by healthcare professionals	16	32.00	4	66.67	5	35.71	1	11.11	4	36.36	3	30.00
Participant describes being grateful for Medicare (Access to allied health professionals)	10	20.00	2	33.33	3	21.43	1	11.11	2	18.18	2	20.00
Participant describes being grateful for their healthcare card and the financial relief it provides	7	14.00	1	16.67	1	7.14	0	0.00	3	27.27	2	20.00
Participant describes being grateful for subsidised diagnostic tests	6	12.00	0	0.00	3	21.43	1	11.11	1	9.09	1	10.00
Participant describes being grateful for government initiatives that support ongoing health and quality of life (for example NDIS, Better Start Program and At home nursing services)	6	12.00	2	33.33	0	0.00	2	22.22	1	9.09	1	10.00
Participant describes frustration at the lack of specialists and specialised services in regional areas and the financial cost incurred when travelling to metropolitan areas for care	5	10.00	0	0.00	0	0.00	0	0.00	1	9.09	4	40.00
Participant describes frustration at the lack of services tailored towards mitochondria disease	5	10.00	0	0.00	0	0.00	2	22.22	1	9.09	2	20.00
Participant describes being grateful for the quality of specialist expertise in Australia	5	10.00	1	16.67	2	14.29	0	0.00	2	18.18	0	0.00

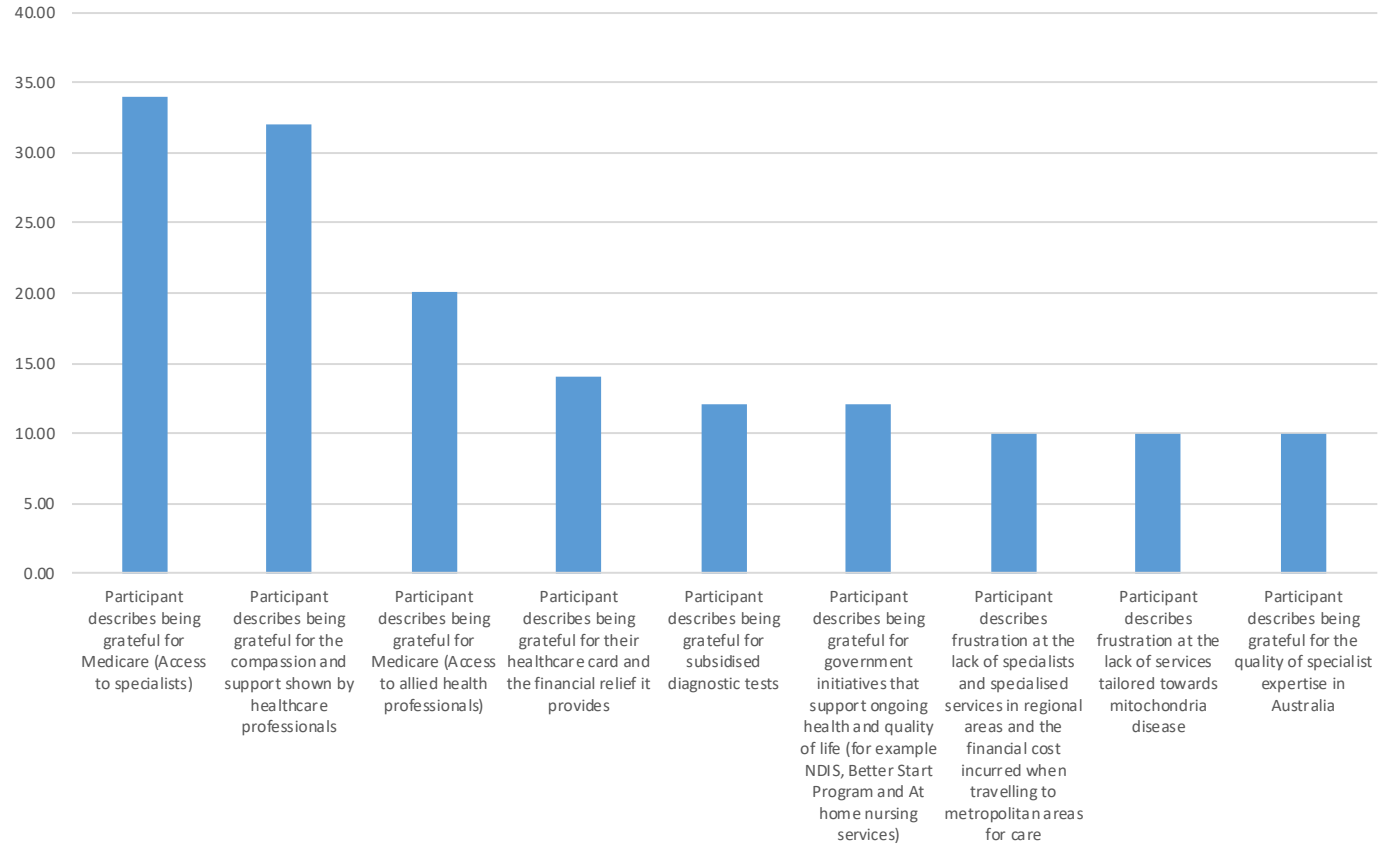


Figure 9.36: Aspects of the Australian health system that participants are grateful for

Messages to decision-makers about the treatment and care that people affected by mitochondrial disease

Participants were asked what their message to people who make decisions about their condition would be. The most common message is to support more research (n=20, 40.00%), however this was a general statement with no specific area noted.

Participant's message is to support more research (General - no specific area)

Just in general, give us some research money. That's the biggest thing. Research and awareness. We're not going to get anywhere without research. Participant 10

More money into research and more involvement in the people with mitochondria in the research. What are the subjects, what are the standards. Give us confidence that they will be able to solve the problems as essential to the treatment, so that they have better treatment and more success. Participant 17

Well, I'd say it's too scarce. They'd need to put money into research but then, how many things do the poor people have to research? Must be millions. Well, you just take the mitochondrial, the different kinds there are and how different everybody is affected. I'd just probably say to him, "Just throw a bit more money into research to try and help." I'm at the end of my life. As I said, it just doesn't bother me but you can imagine a child with something like this if it is diagnosed when they're a child. It would be pretty devastating and the research is needed. Participant 31

The next most common theme was to provide more education to the healthcare professionals, particularly education about managing the condition (n=15, 30.00%), and this was followed by the message to increase awareness of mitochondrial disease among the community (n=12, 24.00%).

Participant's message is to provide more education to the healthcare professionals (Particularly education about managing the condition)

Definitely educating doctors, GPs and things like that. If they knew more about it…I know when aren't convincing. I've seen my main GP, but I also see it, medical things could have been easier. Sometimes you can't get an appointment. When they ask you know what…they want my background and I told them what I have got, a couple of them were like oh, my God. I'm going to research this. That's good for them, to research it, but it's like a general knowledge of mitochondria would be better too. I think a lot of it--they don't know if. Participant 1

Health minister, while we acknowledge the work of you, minister, in improving hospital care, there is certainly room for improvement. Employing education programmes for GPs and healthcare workers, and particularly implementing a full-time position for a counsellor as a wraparound service to help improve the emotional and physical needs of these people would be extremely economically beneficial. Participant 7

Further education for Doctors, put mitochondrial on the Doctor's training curriculum. Something about social workers or genetic counsellors, or some other arm needs to be attached to just the physical diagnosis, because there's so many other bits that need like all the things I've talked about. The equipment, the test forms, the continence aids, the social ... putting a will in place, advanced are directives, all that sort of stuff I know about because I'm a nurse, and I've dealt with palliative health and I know all of those things. But for somebody who doesn't know anything about what's wrong with them, it would be a really hard thing to deal with, and certainly nobody's done any of the things that have helped me, or set my place up. I've had to do it myself, so there's a gap somewhere between the diagnosis, and your investigations, and somebody acting on what you need. Participant 36

Participant's message is increase awareness of mitochondrial disease among the community

Good question. To conduct more research for the cure mito. To make the community more aware of the condition. Participant 6

Maybe raise awareness of it because I would say a good 90% of the people that I talked to have never heard about it before. Which makes sense because it is a fairly rare disease but it might make it easier on a lot of people if everyone knew even just a little bit about it. Participant 11

More babies die of Mitochondrial conditions than die of all the childhood cancers combined. Did you know that? And yet people have never heard about Mitochondrial conditions. So again, it comes back to education. Participant 24

My message would be educate people. Because it's a minority, and it's not diabetes, it's just as bad. Participant 40

To get it out in the public system of what it is and I know people with it are a lot worse than I am, to helping any way they can. Participant 41

There were 12 participants (24.00%) whose message is to provide more holistic and multidisciplinary/allied health care, and eight participants (16.00%) whose message is to improve treatments by following the example of other countries that have more advanced systems.

Participant's message is to provide more holistic and multidisciplinary/allied health care

Then I'd also say there's got to be a support function after you've been diagnosed, like where people can go and get proper information, but also being updated with what can help and also where you can actually have health professionals or specialists in the area given more research to try and combat it. Also, to set up a proper after-care support department, not a department, but somewhere that ... Yeah, somewhere that you can go and talk to and you've got all the right information rather than, you know, trying to find the information off the internet or that sort of thing. That would be my main thing. Participant 15

I think we need a large central clinic with a lot of multidiscipline things that it should be lodged five days a week, but it should also be able to move around the country. It should move. The people in the country should be able to access it and in the outer suburbs so that it could move around. That'll be one week in the city, because everyone should be able to access it no matter where they live without having to do multiple large amount of traveling. Sounds like Eutopia, doesn't it? Participant 21

Yeah, it's not really covered. I go to somebody a long way away, and they've referred me back to somebody else, but nothing happens. [Interviewer: You feel it's up to you to follow it up, and if you don't do it, then no ones going to be checking that you're okay?] Yeah, well they change it too, like I was referred from LOCATION A to particular ... I think it was a physiotherapist, or a rehab clinic at LOCATION B, but then my GP changed it, and just stuck me in LOCATION C for a couple of days, and people looked at me, but nobody really ... I didn't see one Doctor in the whole time I was there. I think the rehab people came once and stuck their head in, but I never saw ... I didn't really see a social worker or anybody. Participant 36

People with mitochondrial disease don't just need medication. They also need physical therapies. I would like occupational therapy and physiotherapy. I would really love more affordable access to that. [pause] If you get a healthcare plan, you get five free visits with the physio in a year. Five isn't enough. Yes, more affordable physical therapies please. Participant 4

Participant's message is to improve treatments by following the example of other countries that have more advanced systems

...we can be investing more money into preventions and treatments; we can be helping people with mito have a better quality of life. Australia is behind in its approach to mitochondrial disease. The UK has legalised the first known preventative measure and yet we're still fighting for clinical trials, yet alone for the disease to be heard of. We can do better. Participant 8

There is absolutely nothing as far as treatment goes for my son and hundreds of other people in Australia for mitochondrial disease. All we have is vitamin supplements to treat a whole complicated area of health conditions associated with mitochondrial disease and that something needs to be done about it. Participant 50

In relation to sub-group variations, participants from rural areas (55.00%) called for more research more frequently than the general population (40.00%). Participants with a hearing impairment (41.67%) had the message to provide more education to the healthcare professionals, more frequently than the general population (30.00%). Participants with a university education (12.50%) called for more awareness less frequently than the general population (24.00%). Participants with a university education (33.33%) and those with a hearing impairment (37.50%) had the message to support more funding (in general), more frequently than the general population (22.00%), while those with a high school or trade education reported this less frequently (11.54%). Participants with high physical function (13.64%) had the message to provide more holistic and multidisciplinary/allied health care less frequently than the general population (24.00%).

Table 9.7: Messages to decision-makers

Aspects of the Australian health system that	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant's message is to support more research (General - no specific area)	20	40.00	9	30.00	11	55.00	11	40.74	9	39.13
Participant's message is to provide more education to the healthcare professionals (Particularly education about managing the condition)	15	30.00	8	26.67	7	35.00	7	25.93	8	34.78
Participant's message is increase awareness of mitochondrial disease among the community	12	24.00	8	26.67	4	20.00	6	22.22	6	26.09
Participant's message is to provide more holistic and multidisciplinary/allied health care	12	24.00	7	23.33	5	25.00	5	18.52	7	30.43
Participant's message is to support more funding (General - no specific area)	11	22.00	8	26.67	3	15.00	7	25.93	4	17.39
Participant's message is to improve treatments by following the example of other countries that have more advanced systems	8	16.00	5	16.67	4	20.00	6	22.22	3	13.04

	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant's message is to support more research (General - no specific area)	20	40.00	9	34.62	11	45.83	9	37.50	12	35.29
Participant's message is to provide more education to the healthcare professionals (Particularly education about managing the condition)	15	30.00	8	30.77	7	29.17	10	41.67	9	26.47
Participant's message is increase awareness of mitochondrial disease among the community	12	24.00	9	34.62	3	12.50	7	29.17	10	29.41
Participant's message is to provide more holistic and multidisciplinary/allied health care	12	24.00	5	19.23	7	29.17	6	25.00	9	26.47
Participant's message is to support more funding (General - no specific area)	11	22.00	3	11.54	8	33.33	9	37.50	7	20.59
Participant's message is to improve treatments by following the example of other countries that have more advanced systems	8	16.00	4	15.38	5	20.83	4	16.67	5	14.71

	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant's message is to support more research (General - no specific area)	20	40.00	9	40.91	11	39.29	11	42.31	9	37.50
Participant's message is to provide more education to the healthcare professionals (Particularly education about managing the condition)	15	30.00	5	22.73	10	35.71	6	23.08	9	37.50
Participant's message is increase awareness of mitochondrial disease among the community	12	24.00	7	31.82	5	17.86	8	30.77	4	16.67
Participant's message is to provide more holistic and multidisciplinary/allied health care	12	24.00	3	13.64	9	32.14	5	19.23	7	29.17
Participant's message is to support more funding (General - no specific area)	11	22.00	6	27.27	5	17.86	6	23.08	5	20.83
Participant's message is to improve treatments by following the example of other countries that have more advanced systems	8	16.00	4	18.18	5	17.86	4	15.38	5	20.83

	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant's message is to support more research (General - no specific area)	20	40.00	10	50.00	10	33.33	10	45.45	10	35.71
Participant's message is to provide more education to the healthcare professionals (Particularly education about managing the condition)	15	30.00	4	20.00	11	36.67	4	18.18	11	39.29
Participant's message is increase awareness of mitochondrial disease among the community	12	24.00	5	25.00	7	23.33	5	22.73	7	25.00
Participant's message is to provide more holistic and multidisciplinary/allied health care	12	24.00	4	20.00	8	26.67	4	18.18	8	28.57
Participant's message is to support more funding (General - no specific area)	11	22.00	6	30.00	5	16.67	5	22.73	6	21.43
Participant's message is to improve treatments by following the example of other countries that have more advanced systems	8	16.00	2	10.00	7	23.33	5	22.73	4	14.29

	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant's message is to support more research (General - no specific area)	20	40.00	1	16.67	5	35.71	5	55.56	5	45.45	4	40.00
Participant's message is to provide more education to the healthcare professionals (Particularly education about managing the condition)	15	30.00	1	16.67	3	21.43	5	55.56	3	27.27	3	30.00
Participant's message is increase awareness of mitochondrial disease among the community	12	24.00	2	33.33	5	35.71	3	33.33	0	0.00	2	20.00
Participant's message is to provide more holistic and multidisciplinary/allied health care	12	24.00	1	16.67	3	21.43	2	22.22	4	36.36	2	20.00
Participant's message is to support more funding (General - no specific area)	11	22.00	1	16.67	4	28.57	2	22.22	3	27.27	1	10.00
Participant's message is to improve treatments by following the example of other countries that have more advanced systems	8	16.00	2	33.33	1	7.14	1	11.11	2	18.18	3	30.00

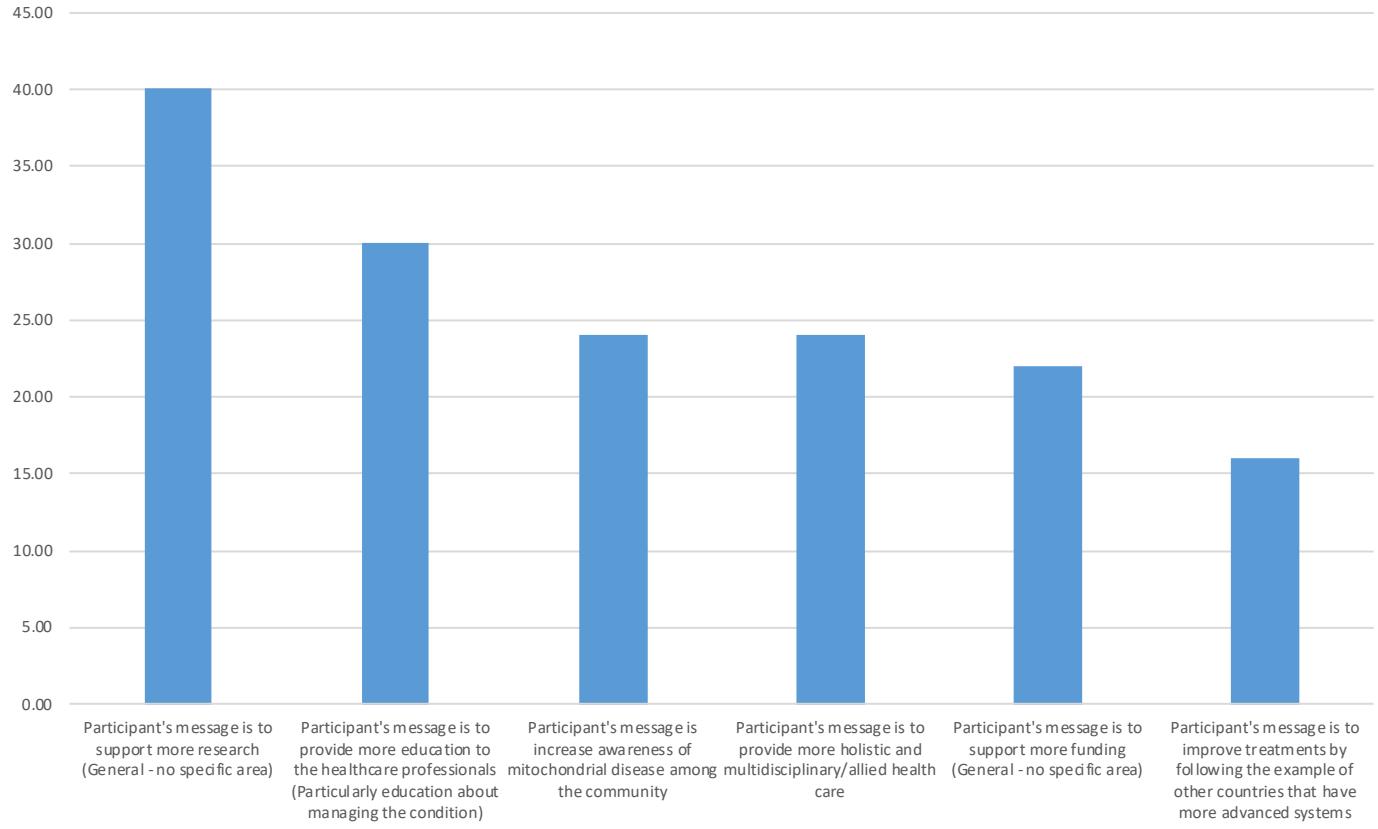


Figure 9.37: Messages to decision-makers (% of all participants)

Section 10 Advice to other patients and families

Section 10: Advice to other patients and families

- Participants were asked what advice they would give to other people who are newly diagnosed with mitochondrial disease and their families. The most common advice is to ask questions and learn as much as you can (n=14, 28.00%). This was followed by the advice to talk to AMDF for information and support and to be part of the community (n=8, 16.00%), seek help (general) (n=8, 16.00%) and to find the right specialist as it is a rare disease and be comfortable with your healthcare team (n=8, 16.00%). There were seven participants (14.00%) whose advice is to seek help through psychological support, six participants (12.00%) whose advice is to share your story to help others and six participants (12.00%) whose advice is to be hopeful.

Advice to others diagnosed with mitochondrial disease

Participants were asked what advice they would give to other people who are newly diagnosed with mitochondrial disease and their families. The most common advice is to ask questions and learn as much as you can (n=14, 28.00%). This was followed by the advice to talk to AMDF for information and support and to be part of the community (n=8, 16.00%), seek help (general) (n=8, 16.00%) and to find the right specialist as it is a rare disease and be comfortable with your healthcare team (n=8, 16.00%). There were seven participants (14.00%) whose advice is to seek help through psychological support, six participants (12.00%) whose advice is to share your story to help others and six participants (12.00%) whose advice is to be hopeful.

Participant's advice is to ask questions and learn as much as you can

I think when you're in with the doctor ask lots of questions and keep asking questions till you get answers. Whether the answers can't give any you more information, this is it, this is all we've got, that's okay to be told that. It's annoying, they told us asks questions. I think a lot of people don't ask enough questions. I know when I was working in with pathologies like that, I had a lot of patient asking me what they have been diagnosed for. I said I'm not a person to be asking this. You've got to talk to your doctor. They said, "They're not telling me anything." I said, "You've got to just keep asking questions. Don't leave until you feel satisfied." If you're not- I think my answer would be telling people ask, just keep asking. Definitely it would help. Participant 1

General advice, take your time and it's okay, and it sucks. I think other advice that I would give is like be your own advocate. Don't let the medical professional just because they've got lots of degrees and experience doesn't mean you still can't advocate for yourself and ask lots of questions and take a proactive approach. Participant 5

Go and get your information, go and see a psychologist who is probably very familiar with family therapy and with illness and disease. Participant 7

Participant's advice is to talk to AMDF for information and support and to be part of the community

There are services out there like the AMDF. You don't have to suffer in silence, you can ring them. There's some really great people that work there, they would have a chat to you on the phone if you're stressing out about stuff. Participant 10

Link into organisations like the AMDF. Participant 30

I would highly recommend they join, register with The Mitochondrial Disease Foundation. That's probably the best source of information. They have support groups and information days, and conference phone calls. If I met someone who had just been diagnosed, that's what I would suggest they do. Participant 43

Participant's advice is to seek help (General)

Unfortunately, all I can describe is my condition. It has developed into a…Get as much potential help. That will be to accepting help. Participant 17

Just let them know that they're not alone and there are people out there that are suffering the same as them, and that, if they need guidance or help, that we're always here for them. Participant 25

I could remember ringing up NAME, somebody or another, NAME I think it was. I rang up just because I don't know where to turn, I don't know what to do, I don't know who to see. Someone tried suggesting ultraviolet light. Somebody else is suggesting this at me. I said, "Things, they cost the earth, I can't afford that." There needs to be someone that you can actually talk to who understands what you're saying. Participant 38

Participant's advice is to seek help - find the right specialist as it is a rare disease and be comfortable with your healthcare team

Information is powerful and to learn everything that you can and if you're not happy with the medical care getting or the doctor that you're seeing, if you don't have a relationship, find someone else. You don't have to say, "Oh, we didn't like him, or he is useless. We just didn't get on, could I have a referral to someone else?" I think sometimes people persist with relationships that are not working. If it's not working and they're not on the same page or they're not seeing the person or yourself how that you think they should be. Just get a referral to someone else. Participant 4

If they're not getting anywhere in finding someone who's listening to what's going on with their body and the tests they've had done, persisting to find someone. Whether that's by asking other people who have got it. Participant 27

Find the best medical team for you they may not be Mito specialists, but people willing to work with you. Participant 30

Participant's advice is to seek help (Psychological support)

Go and get your information, go and see a psychologist who is probably very familiar with family therapy and with illness and disease. Participant 6

I think also considering the support of…from a psychological perspective, not just the physical symptoms and how that affects people and being able to access supports for that too. Participant 26

I feel younger people would need…I know my daughter had a lot of trouble coming to terms with it. I think having me to bounce things off helped with her. I do feel that especially people who are from the country like we're, they would need some health-someone to talk to, someone to unload to mainly and I know I don't know whether that's what it is. This is, of course, by the mitochondrial whatever they've got. Apart from social workers, really…on that at all. For their mental health, I know, it hit my daughter very hard when she had to give up work because she's just so unpredictable. She'll have two or three days where she feels quite good and then she'll spend the next few days on the bed on and off and she says no employer would put up with her. Participant 31

Table 10.1: Advice from patients

Advice to patients in the future	All participants		Metropolitan		Rural		SEIFA (High)		SEIFA (Low)	
	n=50	%	n=30	%	n=20	%	n=27	%	n=23	%
Participant's advice is to ask questions and learn as much as you can	14	28.00	9	30.00	6	30.00	7	25.93	8	34.78
Participant's advice is to talk to AMDF for information and support and to be part of the community	8	16.00	6	20.00	3	15.00	3	11.11	6	26.09
Participant's advice is to seek help (General)	8	16.00	6	20.00	2	10.00	4	14.81	4	17.39
Participant's advice is to seek help - find the right specialist as it is a rare disease and be comfortable with your healthcare team	8	16.00	7	23.33	1	5.00	4	14.81	4	17.39
Participant's advice is to seek help (Psychological support)	7	14.00	4	13.33	3	15.00	3	11.11	4	17.39
Participant's advice is to share your story to help others and help you find your path	6	12.00	4	13.33	3	15.00	2	7.41	5	21.74
Participant's advice is to be hopeful	6	12.00	5	16.67	2	10.00	5	18.52	2	8.70
Participant's advice is to take one day at a time and appreciate each day	5	10.00	3	10.00	2	10.00	4	14.81	1	4.35
Participant's advice is to be a strong advocate for your or your child's health and wellbeing	5	10.00	3	10.00	2	10.00	1	3.70	4	17.39

Advice to patients in the future	All participants		High school or trade		University		Hearing impairment		Eye or visual impairment	
	n=50	%	n=26	%	n=24	%	n=24	%	n=34	%
Participant's advice is to ask questions and learn as much as you can	14	28.00	9	34.62	6	25.00	6	25.00	12	35.29
Participant's advice is to talk to AMDF for information and support and to be part of the community	8	16.00	6	23.08	3	12.50	3	12.50	7	20.59
Participant's advice is to seek help (General)	8	16.00	4	15.38	4	16.67	6	25.00	6	17.65
Participant's advice is to seek help - find the right specialist as it is a rare disease and be comfortable with your healthcare team	8	16.00	4	15.38	4	16.67	3	12.50	7	20.59
Participant's advice is to seek help (Psychological support)	7	14.00	2	7.69	5	20.83	3	12.50	3	8.82
Participant's advice is to share your story to help others and help you find your path	6	12.00	6	23.08	1	4.17	3	12.50	4	11.76
Participant's advice is to be hopeful	6	12.00	2	7.69	5	20.83	5	20.83	3	8.82
Participant's advice is to take one day at a time and appreciate each day	5	10.00	2	7.69	3	12.50	3	12.50	3	8.82
Participant's advice is to be a strong advocate for your or your child's health and wellbeing	5	10.00	0	0.00	5	20.83	4	16.67	4	11.76

Advice to patients in the future	All participants		Physical function (High)		Physical function (Low)		Emotional well-being (High)		Emotional well-being (Low)	
	n=50	%	n=22	%	n=28	%	n=26	%	n=24	%
Participant's advice is to ask questions and learn as much as you can	14	28.00	8	36.36	7	25.00	6	23.08	9	37.50
Participant's advice is to talk to AMDF for information and support and to be part of the community	8	16.00	4	18.18	5	17.86	5	19.23	4	16.67
Participant's advice is to seek help (General)	8	16.00	4	18.18	4	14.29	4	15.38	4	16.67
Participant's advice is to seek help - find the right specialist as it is a rare disease and be comfortable with your healthcare team	8	16.00	5	22.73	3	10.71	3	11.54	5	20.83
Participant's advice is to seek help (Psychological support)	7	14.00	1	4.55	6	21.43	5	19.23	2	8.33
Participant's advice is to share your story to help others and help you find your path	6	12.00	3	13.64	4	14.29	2	7.69	5	20.83
Participant's advice is to be hopeful	6	12.00	2	9.09	5	17.86	4	15.38	3	12.50
Participant's advice is to take one day at a time and appreciate each day	5	10.00	0	0.00	5	17.86	4	15.38	1	4.17
Participant's advice is to be a strong advocate for your or your child's health and wellbeing	5	10.00	4	18.18	1	3.57	3	11.54	2	8.33

Advice to patients in the future	All participants		Social functioning (High)		Social functioning (Low)		General health (High)		General health (Low)	
	n=50	%	n=20	%	n=30	%	n=22	%	n=28	%
Participant's advice is to ask questions and learn as much as you can	14	28.00	7	35.00	8	26.67	6	27.27	9	32.14
Participant's advice is to talk to AMDF for information and support and to be part of the community	8	16.00	6	30.00	3	10.00	5	22.73	4	14.29
Participant's advice is to seek help (General)	8	16.00	5	25.00	3	10.00	2	9.09	6	21.43
Participant's advice is to seek help - find the right specialist as it is a rare disease and be comfortable with your healthcare team	8	16.00	4	20.00	4	13.33	2	9.09	6	21.43
Participant's advice is to seek help (Psychological support)	7	14.00	5	25.00	2	6.67	2	9.09	5	17.86
Participant's advice is to share your story to help others and help you find your path	6	12.00	2	10.00	5	16.67	2	9.09	5	17.86
Participant's advice is to be hopeful	6	12.00	3	15.00	4	13.33	2	9.09	5	17.86
Participant's advice is to take one day at a time and appreciate each day	5	10.00	0	0.00	5	16.67	1	4.55	4	14.29
Participant's advice is to be a strong advocate for your or your child's health and wellbeing	5	10.00	2	10.00	3	10.00	3	13.64	2	7.14

Advice to patients in the future	All participants		Under 18		24-44		45-54		55-64		65-74+	
	n=50	%	n=6	%	n=14	%	n=9	%	n=11	%	n=10	%
Participant's advice is to ask questions and learn as much as you can	14	28.00	2	33.33	6	42.86	2	22.22	3	27.27	2	20.00
Participant's advice is to talk to AMDF for information and support and to be part of the community	8	16.00	1	16.67	3	21.43	3	33.33	2	18.18	0	0.00
Participant's advice is to seek help (General)	8	16.00	0	0.00	4	28.57	2	22.22	1	9.09	1	10.00
Participant's advice is to seek help - find the right specialist as it is a rare disease and be comfortable with your healthcare team	8	16.00	0	0.00	4	28.57	3	33.33	1	9.09	0	0.00
Participant's advice is to seek help (Psychological support)	7	14.00	1	16.67	4	28.57	0	0.00	1	9.09	1	10.00
Participant's advice is to share your story to help others and help you find your path	6	12.00	1	16.67	3	21.43	1	11.11	1	9.09	1	10.00
Participant's advice is to be hopeful	6	12.00	2	33.33	1	7.14	3	33.33	1	9.09	0	0.00
Participant's advice is to take one day at a time and appreciate each day	5	10.00	1	16.67	1	7.14	1	11.11	1	9.09	1	10.00
Participant's advice is to be a strong advocate for your or your child's health and wellbeing	5	10.00	0	0.00	3	21.43	0	0.00	2	18.18	0	0.00

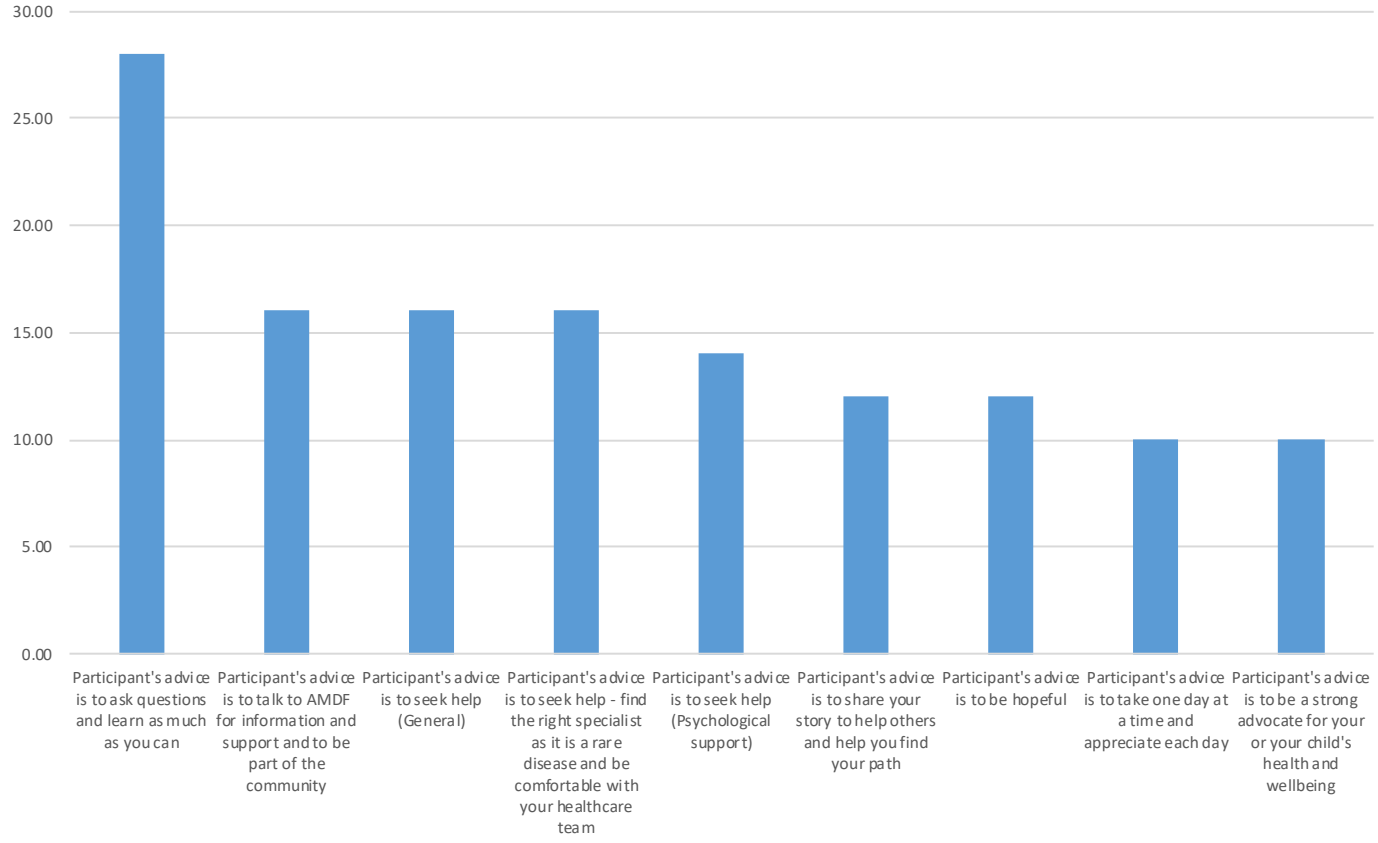


Figure 10.1: Advice from patients (% of all participants)

Section 11 Discussion

Symptoms, risk and diagnosis

Mitochondrial disease is one of the most common groups of genetic diseases, is caused by mutations or deletions of mitochondrial or nuclear DNA. The disease may affect single organs, multiple organs or systems. Presentation can occur from birth to old age[1], paediatric onset is associated with more severe multi-systemic symptoms, increased progression and poorer prognosis compared to adult onset[2].

Symptoms

Symptoms may present in single or multiple organs or systems. Symptoms of the skeletal muscles include muscle weakness, exercise intolerance and fatigue[3]. Ophthalmologic (eye) symptoms include cataract, cortical blindness (total or partial vision loss due to damage in brain) and homonymous hemianopsia (visual field loss). Common central nervous system symptoms include developmental delays, seizures, stroke like episodes, coma, lethargy and autism spectrum symptoms[4-6]. Gastrointestinal symptoms include cachexia (weakness and wasting of body), peripheral neuropathy (weakness and numbness usually in hands and feet), ophthalmoplegia (paralysis of eye muscles) and diarrhea[1]. Symptoms of the endocrine system include diabetes mellitus, short stature, hypothyroidism (insufficient thyroid hormones), hypoparathyroidism (insufficient hypothyroid hormones), adrenal insufficiency, hypogonadism (insufficient hormones for gonad activity)[1]. Heart symptoms include cardiomyopathy (heart unable to pump sufficient blood supply to body), arrhythmias, conduction defects, pulmonary hypertension[1]. Other symptoms include hearing loss and deafness, kidney dysfunction and liver problems[7].

The results of this PEEK study are consistent with the literature with the most commonly reported symptoms being muscle symptoms by (such as muscle weakness, exercise intolerance, pain, fatigue, cramps and low muscle tone), fatigue, digestive tract symptoms, problems with vision and eyes, central nervous system symptoms, and hearing problems. It is important to note however that the way patients describe symptoms and the way symptoms are reported in the literature can vary in the language used. Where this PEEK study adds to the literature is in the quality of life while experiencing those symptoms, with symptoms that had the lowest average quality of life being central nervous symptoms, muscle symptoms, heart symptoms and digestive tract symptoms.

Clusters of symptoms are defined as phenotypic (observable characteristics) mitochondrial syndromes, not all diagnosed with mitochondrial disease will have a phenotypic mitochondrial syndrome diagnosis. Some of the most common phenotypic mitochondrial symptoms are listed in Table 11.1.

Diagnosis

Mitochondrial disease presents with a number of different phenotypes, and there is a lack of specific biomarkers to confirm diagnosis[16]. Diagnosis of mitochondrial disease should be considered for muscle or central nervous system disease with disease of two or more organ systems, or diseases in three or more organ systems[1]. A detailed medical and family history can help diagnose they type of inheritance pattern, and mapping of clinical symptoms to define extent of disease and help with management[17]. Clinical investigations should include neurological, cardiac, ophthalmological, hearing, growth and psychomotor development. Imaging studies including CT and MRI may be used in patients that have seizures or stroke like episodes[18,19]. The tests available include biochemical studies of blood and urine, biopsies of muscle, skin, and liver, and DNA testing, however it is not well established how much testing is needed to confirm or exclude a diagnosis[20]. More than 200 genes have been identified in the development of mitochondrial disease[21]. A survey of North American mitochondrial clinics reported that clinicians make a diagnosis based on a combination of the clinical phenotype, biochemical abnormalities and their professional opinion[16].

The DNA tests may look for mutations or deletions in mitochondrial DNA or in nuclear DNA. The mitochondrial DNA is smaller with fewer genes than nuclear DNA, as such point mutations or sequencing entire genome may be employed[16]. Testing of nuclear DNA may involve a selective panel of known mutations, testing with a 100 or more genes or whole genome sequencing[16].

Table 11.1: Common phenotypic mitochondrial syndromes.

Syndrome	Description
Leigh syndrome	Brain lesions with developmental regression, respiratory abnormalities, feeding problems, often with eye problems. Usually presents in first year of life[1,8].
Alpers disease	Neurodegeneration, seizures and liver dysfunction[9].
Mitochondrial Recessive Ataxia Syndrome (MIRAS)	Ataxia (lack of muscle control/coordination), neuropathy (nerve damage), encephalopathy (brain disease) with seizures[10].
Myopathy, Encephalopathy, Lactic Acidosis and Stroke-like episodes (MELAS)	Myopathy (muscle fibre dysfunction), encephalopathy, lactic acidosis and stroke-like episodes, other clinical symptoms include cognitive decline, deafness, sort stature, ataxia, and diabetes[11].
Myoclonic Epilepsy and Ragged Red Fibres (MERRF)	Myoclonic epilepsy (muscle jerk) and ragged red fibres seen in muscle biopsy, some cases have visual, audial and cardiac involvement[5].
Pearson syndrome	Infant onset, sideroblastic anaemia (produces dysfunctional red blood cells), lactic acidosis, pancreatic dysfunction, many do not survive infancy[12].
Kearns-Sayre Syndrome (KSS)	Symptoms include ptosis (drooping eyelids), pigmentary retinopathy (eye disorder) and progressive external ophthalmoplegia, cardiac conduction defects, cerebellar ataxia and cerebral folate deficiency, onset before 20 years of age[13,14].
Progressive External Ophthalmoplegia (PEO)	Progressive External Ophthalmoplegia occurs in a number of phenotypes and can also occur in isolation Myopathy, Lactic Acidosis, and Sideroblastic Anaemia[1].
Leber hereditary optic neuropathy (LHON)	Painless vision loss affecting both eyes, occurs usually in adolescence or in adults[1].
Neuropathy, Ataxia, Retinitis Pigmentosa (NARP)	Neuropathy, ataxia, retinitis pigmentosa (eye disease with damage to retina) often presents with Leigh syndrome[1]
Myopathy, Lactic Acidosis, and Sideroblastic Anaemia (MLASA)	Myopathy, lactic acidosis, and sideroblastic Anaemia, affects mostly skeletal muscle and bone marrow[15].
Reversible Disorders	Include infantile reversible myopathy, reversible infantile liver disease[1].

Prenatal screening

The purpose of prenatal screening is to determine the risk of the foetus developing mitochondrial disease. A study from a UK mitochondrial clinic observed an increasing demand for this service over a five-year period[22]. The majority of people requesting testing (>90%) had another child with mitochondrial disease, other reasons included asymptomatic known carriers and having an affected sibling[22]. Suggest that this service is important for couples with known risk factors and should be offered with pre and post-test genetic counselliing[22].

Experience of diagnostic pathways

The reported diagnostic pathways of people with mitochondrial disease describe a multitude of symptoms, physicians, and incorrect diagnoses. A North American study reports the first discussion of MD was commonly with a general practitioner, followed by a specialist doctor[23]. The average number of doctors seen from the time of symptoms to a mitochondrial disease diagnosis was 8.19, and the diagnosis was most frequently from a neurologist, clinical geneticist or metabolic specialist[23]. More than half had at least one alternative diagnosis before receiving MD diagnosis; the most common other diagnoses given were psychiatric disorders, fibromyalgia, chronic fatigue syndrome, Multiple Sclerosis, gastrointestinal disease and seizure[23]. The most common symptoms that lead to seeking a diagnosis were weakness, fatigue, difficulty walking, droopy eyelids and impaired coordination[23], this is in contrast with a Korean study that reports the most common symptoms as seizure and delayed development[24]. The most common tests were blood tests, muscle biopsy, MRI, urine organic acids and DNA testing[23], this is consistent with a UK study where the most common investigations were blood or cerebral spinal fluid, muscle biopsy, DNA testing, skin biopsies, histological studies of either muscle or liver and imaging studies[25].

It is clear that the diagnostic pathway for mitochondrial disease is complex. In this PEEK study, participants were asked whether they felt supported at the time of diagnosis. The majority of participants indicated that

they had no support at diagnosis, indicating that this is an area of support that needs additional attention. This is particularly important as more than half of the participants in this PEEK study reported that they had not had a discussion about genetic tests and over half of the participants noted that they had not had a discussion about clinical trials.

Diagnostic challenges

The frustrations that caregivers experience in during the diagnostic period of their child can be attributed to the emotional experience of coming to terms with their child's health status and with obstacles experienced with their interactions with healthcare professionals and support networks[26]. The ability to discuss and understand prognosis is clearly an important aspect of decision-making. In this PEEK study, prognosis had not been discussed with a little over half of all participants. was prognosis had not been clearly discussed. Caregivers are driven to find a diagnosis to have a better understanding of their child's prognosis and the hope for better treatment options, however, with mitochondrial disease due to the number of clinical phenotypes and lack of information about prognosis, caregiver uncertainty remains after diagnosis[27-29].

Treatment

There is no cure for mitochondrial disease with best practice guidelines based on treating symptoms and complications of the disease, and due to the variation in symptoms and affected organs, it is individual to each patient[30]. A coordinated clinical team of healthcare professionals treat the symptoms of mitochondrial disease including neurologists, cardiologists, metabolic physicians, endocrinologists, nephrologists, gastroenterologists, ophthalmologists, audiological physicians, paediatricians, psychologists, nurses, physiotherapy, speech and language and occupational therapy.[1] [31] Below is an overview of supportive care, allied health and common treatments for conditions associated with mitochondrial disease. This is consistent with the results of this PEEK study where participants the most common treatments were Coenzyme Q10, vitamins and supplements, followed by physical therapy and diet.

Clinics and specialist care **-** A survey of North American mitochondrial specialist, reported that the majority of specialists are either neurologists, geneticists or both[16]. Most of the mitochondrial clinics are not specific to mitochondrial disease and while most specialists have trained in paediatrics, they have both adult and child patients[16]. The consultations are lengthy and require extensive work outside of the consultation including coordination of testing, reviewing records and consultation with a range of specialists. [16]

Nutritional supplements (vitamin cocktails) - A Cochrane review of randomized clinical trials found no significant clinical outcome in the use of nutritional supplements such as coenzyme Q10, creatine, carnitine, dichloroacetate or vitamin cocktails[30]. However a non-randomized trial of coenzyme Q10 showed clinical improvement across phenotypes [34]. EPI-473, an anti-oxidant has shown clinical improvement in Leigh Syndrome[35] and LHON[36].

Occupational/speech/language/physio therapy (Allied health) - Occupational therapy for daily activities at home, school and work, speech therapy for problems with oral motor skills in particular swallowing, in addition some syndromes may benefit from learning sign language, physiotherapy for strengthening, posture and stretching to maintain mobility and function and educational support depending on cognitive and physical function[18].

Lactic acidosis - Lactic acidosis is one of the main symptoms, especially in children. Treatment of cute cases of lactic acidosis with sodium bicarbonate, dichloroacetate may also be used but long term however the long-time use and affects are not well known[1,30,37].

Diet - Therapies that promote growth of mitochondria include drugs such as bezafibrate and resveratrol[38], in addition to following a ketogenic diet[39]

Preventative - Reproductive options include antenatal testing, pre-implantation genetic screening[40], and mitochondrial donation[41].

Hearing - Monitor hearing, some may benefit from hearing aids or cochlear implants[42]

Vision - Correction for ptosis by prosthetic inserts in spectacles or by surgical intervention, also monitor for conditions such as cataracts, optic atrophy and retinopathy[31]

CNS - Seizures are treated with anti-convulsant drugs and involuntary spasms are treated with anti-dystonia medications or botox[31]

Stroke - Arginine therapy for prevention of stroke like episodes in MELAS[11]

Endocrine - Screening for diabetes is important, usually responds to hypoglycaemics or low dose of insulin, metformin should not be given due to risk of lactic acidoisis[43]

Respiratory - respiratory muscles should be monitored for weakness, this may be especially problematic following anaesthesia, formal respiratory support may be needed for sleep apnoea [31].

GI - Gastrointestinal problems may include swallowing difficulties, failure to thrive, weight loss, constipation, pseudo-obstruction, nausea and vomiting. Speech and language assessments are important for swallowing assessments, percutaneous gastronomy may be needed[31].

Cardiac - Heart screening important, implantable devices such as pacemakers and defibrillators may be needed. Ace inhibitors for left ventricular hypertrophy[31].

Electrolyte disturbances - Low calcium and potassium levels are common in children with renal problems, these should be monitored and treated[19,44]

Biomarkers

Mutations of the mitochondrial DNA (primary) or genes of nuclear DNA (secondary) that impact the mitochondria[47]. Mitochondrial DNA mutations are most commonly inherited maternally, whereas mitochondrial DNA deletions occur *de novo* during embryonic development[47]. There are 37 genes in mitochondrial DNA, and for each gene mutations have been reported that result in MD[47]. Mothers with defective mitochondrial DNA may be asymptomatic, the copy numbers may be below a threshold needed for the dysfunction, the copy numbers in subsequent pregnancies is not predictable, however pre-natal testing can give an accurate measure of dysfunction[22,48]

Heterogeneity of mitochondrial disease

There are over 1300 mitochondrial proteins described generated from nuclear DNA, mutations have been reported in over 250 of these[49]. Inheritance of these defects can occur de novo or from either parents[47].

Sequencing of the mtDNA is often conducted to exclude or confirm primary disease, this can be achieved due to the relatively small size of mtDNA[47]. Next generation sequencing (NGS) based techniques can be used to examine panels of candidate genes, and other techniques which sequence the whole genome are being implemented[47,50].

Complementary therapies

Use of complementary therapies

The reported use of complementary therapies is high in the mitochondrial disease community, with a number of studies reporting usage between 70 and 90%[45,51,52]. The most commonly described complementary therapies are nutritional supplements, other therapies including homeopathy preparations and self-help techniques including reiki and yoga have also been described[45,51,52]. This is consistent with this PEEK study where the most common therapies described by participants were vitamins, minerals and supplements, and access to allied health professionals, while a little over one fifth of all participants noted that they did not use any complementary therapies.

Diet

People with mitochondrial disease benefit from an adequate diet to cope with symptoms such gastrointestinal problems, metabolic problems, muscle weakness, fatigue, dysphagia and diabetes[51]. A study of Dutch people with mitochondrial disease reported inadequate protein, calcium, fibre and fluid intake[51].

Vitamins and vitamin cocktails

Despite the lack of evidence about the use of supplements for MD, "mito cocktails' are recommended by physician, often under pressure from patients and advocacy groups[53-55]. However, nutritional supplements are frequently used, most commonly coenzyme q10, multivitamins, carnitine, riboflavin, vitamin D and vitamin C[45,51]. A North American study reported that over three quarters of participants took more than four supplements and combinations almost unique to each participant[45]. Perceived benefits from nutritional side effects took between two weeks and three months to achieve, more than half participants felt that their most difficult symptoms were relieved by using supplements, these included fatigue, exercise intolerance, muscle pains and weakness. Gastrointestinal and neurological symptoms were less responsive[45]. About one third experienced side effects including nausea, diarrhea

and unpleasant smell and a further 10% stopped taking supplements due to side effects[45]

Cost of complementary therapies

The out of pocket expenses of supplements has been reported as inconvenient, and many would like to see cost reduction by insurance coverage[45]. The amount spent reported varies, with a North American study reporting that almost a third of participants spend more than AUD$268 per month[45], and a Dutch study reporting that adults spend AUD$568 per annum and children AUD$774 per annum[52].

Quality of life

Quality of life in adults with mitochondrial disease has been reported to be affected by the losses of energy, independence, social participation, identity and future[56]. While quality of life is often attributed to physical impact, in this PEEK study, the most common impact on quality of life described by participants was poor mental health as a consequence of mitochondrial disease, with some noting poor mental health of family or friends. Likewise, the Fear of Progression questionnaire used in this PEEK study measures the level of anxiety people experience in relation to their conditions. The Fear of Progression questionnaire comprises a total score, with a higher score denoting increased anxiety. Overall the entire cohort had a median total score of 34.10, which is a score in the middle of the scale.

Caregivers to those with mitochondrial disease experience significant burden, anxiety, and depression, compared to caregivers of other chronic childhood conditions, in addition, caregivers have poorer quality of life particularly in the role limitations, vitality and mental health domains[24,57]. Anxiety is the greatest contributor to caregiver burden, though income, age of child, number of hospitalisations and medical visits, number of involved organs also contribute to caregiver stress[46,57]. Stress is reduced with improved family integration, social support and greater healthcare knowledge[46].

Health professional communication, support and education

Caregiver stress reduced my being informed

Caregiver stress is reduced with greater healthcare knowledge, a healthcare professional as a point of contact is important for optimal communication and may reduce stress during diagnosis and as new information is available[58].

Communication with health professionals about complementary therapies

A Dutch study about the use of complementary therapies reported on communication between patient and physician regarding complementary therapy use was conducted by about a third of patients and was almost always initiated by the patient[52]. Physician reaction to use of complementary therapies was generally positive. Advice from physicians about complementary therapies was rated as important by most of the participants. Future research about complementary therapies was rated as important or very important by most of the cohort, with about half of the children and 80% of the adults willing to take part in clinical trials[52].

Genetic counselling to educate and inform

The role of the genetic counsellor is to explain the complexities of the disease and the complexities of obtaining a diagnosis, inheritance and reproductive options[59]. A detailed family history has the benefit of aiding diagnosis and can also be used as a risk assessment tool[59], however, genetic counselling is difficult with mitochondrial disease due to the number of possible mechanisms of inheritance[58].

Coordination of care

Coordination of care is essential and challenging for mitochondrial disease as care is across all levels of the health system and involves many healthcare professionals[18]. Liaison with local services should be facilitated soon after diagnosis to facilitate needs such as home adaptations, equipment, therapy, education support for children and support for carers[18]. Specialist nurses and community nurses can play a central role in family support[18]. It is important to consider the support needs of family and carers including emotional support, changes to employment status and loss of income, increased travel between home, school and hospital and the care of other siblings[18]. Joining a support group may be useful, with reports that the majority of those who join a support group had found it beneficial[23].

In this PEEK study, the absence of care coordination and multidisciplinary care was highlighted by the expectation of future care and support in the form of centralised and coordinated care across specialists and allied health professionals (including more

communication between doctors), and the recommendation for caseworkers be employed to support patients navigate health, medical and emotional needs.

Summary: Characterisation of the study population

People that receive a diagnosis of mitochondrial disease often endure a long and complicated diagnosis, which is often experienced without adequate support. Once diagnosed, there are no direct treatment options available with management of the disease centred around multi-disciplinary care, diet and exercise management. However, as the disease presents in various ways as clusters of symptoms that are defined as phenotypic (observable characteristics) mitochondrial syndromes, not all diagnosed with mitochondrial disease will fit into a specific group and there is a great need for individualised case management, which is also a key recommendation from this PEEK study population.

Some of the frustrations experienced by people diagnosed with mitochondrial disease and their families is the lack of understanding about the disease by health professionals resulting in the need for mechanisms to support health professional education.

This patient population is well informed, as evidenced by this PEEK study where the scores for knowledge, recognition and management of symptoms, and total score were in the second highest quintile indicating good understanding and knowledge of disease. The score for coping with their condition was in the middle of the range of scores for this scale and participants in this PEEK study reported psychological stress and anxiety caused by the disease as key impacts on their quality of life. As some of the key activities that were reported in relation to maintaining general health included having adequate rest to minimise fatigue, regular exercise and eating a healthy/modified diet, rather than supporting mental health, this may suggest that more support is needed to help this patient population access psychological health services.

This is a patient population that is grateful for the support and services that are available to them, particularly Medicare (in relation to access to specialists and allied health professionals in particular) and the compassion and support shown by healthcare professionals. However, as there are no treatments available for this patient population, their key message is to support more research, and to provide more education to the healthcare professionals, particularly education about managing the condition.

References

1. Davison JE, Rahman S. Recognition, investigation and management of mitochondrial disease. *Arch Dis Child* 2017; **102**(11): 1082-90.

2. Horvath R, Kemp JP, Tuppen HA, et al. Molecular basis of infantile reversible cytochrome c oxidase deficiency myopathy. *Brain* 2009; **132**(Pt 11): 3165-74.

3. Giordano C, Pichiorri F, Blakely EL, et al. Isolated distal myopathy of the upper limbs associated with mitochondrial DNA depletion and polymerase gamma mutations. *Arch Neurol* 2010; **67**(9): 1144-6.

4. Pinto M, Pickrell AM, Moraes CT. Regional susceptibilities to mitochondrial dysfunctions in the CNS. *Biol Chem* 2012; **393**(4): 275-81.

5. Rahman S. Mitochondrial disease and epilepsy. *Dev Med Child Neurol* 2012; **54**(5): 397-406.

6. Palmieri L, Persico AM. Mitochondrial dysfunction in autism spectrum disorders: cause or effect? *Biochim Biophys Acta* 2010; **1797**(6-7): 1130-7.

7. Cormier-Daire V, Chretien D, Rustin P, et al. Neonatal and delayed-onset liver involvement in disorders of oxidative phosphorylation. *J Pediatr* 1997; **130**(5): 817-22.

8. Lake NJ, Compton AG, Rahman S, Thorburn DR. Leigh syndrome: One disorder, more than 75 monogenic causes. *Ann Neurol* 2016; **79**(2): 190-203.

9. Young MJ, Copeland WC. Human mitochondrial DNA replication machinery and disease. *Curr Opin Genet Dev* 2016; **38**: 52-62.

10. Wong LJ, Naviaux RK, Brunetti-Pierri N, et al. Molecular and clinical genetics of mitochondrial diseases due to POLG mutations. *Hum Mutat* 2008; **29**(9): E150-72.

11. Koenig MK, Emrick L, Karaa A, et al. Recommendations for the Management of Strokelike Episodes in Patients With Mitochondrial Encephalomyopathy, Lactic Acidosis, and Strokelike Episodes. *JAMA Neurol* 2016; **73**(5): 591-4.

12. Rotig A, Cormier V, Blanche S, et al. Pearson's marrow-pancreas syndrome. A multisystem mitochondrial disorder in infancy. *J Clin Invest* 1990; **86**(5): 1601-8.

13. Broomfield A, Sweeney MG, Woodward CE, et al. Paediatric single mitochondrial DNA deletion disorders: an overlapping spectrum of disease. *J Inherit Metab Dis* 2015; **38**(3): 445-57.

14. Maceluch JA, Niedziela M. The clinical diagnosis and molecular genetics of kearns-sayre syndrome: a complex mitochondrial encephalomyopathy. *Pediatr Endocrinol Rev* 2006; **4**(2): 117-37.

15. Shahni R, Wedatilake Y, Cleary MA, Lindley KJ, Sibson KR, Rahman S. A distinct mitochondrial myopathy, lactic acidosis and sideroblastic anemia (MLASA) phenotype associates with YARS2 mutations. *Am J Med Genet A* 2013; **161A**(9): 2334-8.

16. Parikh S, Goldstein A, Koenig MK, et al. Practice patterns of mitochondrial disease physicians in North America. Part 1: diagnostic and clinical challenges. *Mitochondrion* 2014; **14**(1): 26-33.

17. Koopman WJ, Beyrath J, Fung CW, et al. Mitochondrial disorders in children: toward development of small-molecule treatment strategies. *EMBO Mol Med* 2016; **8**(4): 311-27.

18. Kisler JE, Whittaker RG, McFarland R. Mitochondrial diseases in childhood: a clinical approach to investigation and management. *Dev Med Child Neurol* 2010; **52**(5): 422-33.

19. Moggio M, Colombo I, Peverelli L, et al. Mitochondrial disease heterogeneity: a prognostic challenge. *Acta Myol* 2014; **33**(2): 86-93.

20. Mitochondrial Medicine Society's Committee on D, Haas RH, Parikh S, et al. The in-depth evaluation of suspected mitochondrial disease. *Mol Genet Metab* 2008; **94**(1): 16-37.

21. Koopman WJ, Willems PH, Smeitink JA. Monogenic mitochondrial disorders. *N Engl J Med* 2012; **366**(12): 1132-41.

22. Nesbitt V, Alston CL, Blakely EL, et al. A national perspective on prenatal testing for mitochondrial disease. *Eur J Hum Genet* 2014; **22**(11): 1255-9.

23. Grier J, Hirano M, Karaa A, Shepard E, Thompson JLP. Diagnostic odyssey of patients with mitochondrial disease: Results of a survey. *Neurol Genet* 2018; **4**(2): e230.

24. Eom S, Lee YM. Preliminary Study of Neurodevelopmental Outcomes and Parenting Stress in Pediatric Mitochondrial Disease. *Pediatr Neurol* 2017; **71**: 43-9 e1.

25. Verity CM, Winstone AM, Stellitano L, Krishnakumar D, Will R, McFarland R. The clinical presentation of mitochondrial diseases in children with progressive intellectual and neurological deterioration: a national, prospective, population-based study. *Dev Med Child Neurol* 2010; **52**(5): 434-40.

26. Lewis C, Skirton H, Jones R. Living without a diagnosis: the parental experience. *Genet Test Mol Biomarkers* 2010; **14**(6): 807-15.

27. Stewart JL, Mishel MH. Uncertainty in childhood illness: a synthesis of the parent and child literature. *Sch Inq Nurs Pract* 2000; **14**(4): 299-319; discussion 21-6.

28. Graungaard AH, Skov L. Why do we need a diagnosis? A qualitative study of parents' experiences, coping and needs, when the newborn child is severely disabled. *Child Care Health Dev* 2007; **33**(3): 296-307.

29. Rosenthal ET, Biesecker LG, Biesecker BB. Parental attitudes toward a diagnosis in children with unidentified multiple congenital anomaly syndromes. *Am J Med Genet* 2001; **103**(2): 106-14.

30. Pfeffer G, Majamaa K, Turnbull DM, Thorburn D, Chinnery PF. Treatment for mitochondrial disorders. *Cochrane Database Syst Rev* 2012; (4): CD004426.

31. Rahman S, Hanna MG. Diagnosis and therapy in neuromuscular disorders: diagnosis and new treatments in mitochondrial diseases. *J Neurol Neurosurg Psychiatry* 2009; **80**(9): 943-53.

32. Phoenix C, Schaefer AM, Elson JL, et al. A scale to monitor progression and treatment of mitochondrial disease in children. *Neuromuscul Disord* 2006; **16**(12): 814-20.

33. Schaefer AM, Walker M, Turnbull DM, Taylor RW. Endocrine disorders in mitochondrial disease. *Mol Cell Endocrinol* 2013; **379**(1-2): 2-11.

34. Emmanuele V, Lopez LC, Berardo A, et al. Heterogeneity of coenzyme Q10 deficiency: patient study and literature review. *Arch Neurol* 2012; **69**(8): 978-83.

35. Martinelli D, Catteruccia M, Piemonte F, et al. EPI-743 reverses the progression of the pediatric mitochondrial disease--genetically defined Leigh Syndrome. *Mol Genet Metab* 2012; **107**(3): 383-8.

36. Sadun AA, Chicani CF, Ross-Cisneros FN, et al. Effect of EPI-743 on the clinical course of the mitochondrial disease Leber hereditary optic neuropathy. *Arch Neurol* 2012; **69**(3): 331-8.

37. Kaufmann P, Engelstad K, Wei Y, et al. Dichloroacetate causes toxic neuropathy in MELAS: a randomized, controlled clinical trial. *Neurology* 2006; **66**(3): 324-30.

38. Kanabus M, Heales SJ, Rahman S. Development of pharmacological strategies for mitochondrial disorders. *Br J Pharmacol* 2014; **171**(8): 1798-817.

39. Hughes SD, Kanabus M, Anderson G, et al. The ketogenic diet component decanoic acid increases mitochondrial citrate synthase and complex I activity in neuronal cells. *J Neurochem* 2014; **129**(3): 426-33.

40. Smeets HJ, Sallevelt SC, Dreesen JC, de Die-Smulders CE, de Coo IF. Preventing the transmission of mitochondrial DNA disorders using prenatal or preimplantation genetic diagnosis. *Ann N Y Acad Sci* 2015; **1350**: 29-36.

41. Craven L, Herbert M, Murdoch A, Murphy J, Lawford Davies J, Turnbull DM. Research into Policy: A Brief History of Mitochondrial Donation. *Stem Cells* 2016; **34**(2): 265-7.

42. Sinnathuray AR, Raut V, Awa A, Magee A, Toner JG. A review of cochlear implantation in mitochondrial sensorineural hearing loss. *Otol Neurotol* 2003; **24**(3): 418-26.

43. Murphy R, Turnbull DM, Walker M, Hattersley AT. Clinical features, diagnosis and management of maternally inherited diabetes and deafness (MIDD) associated with the 3243A>G mitochondrial point mutation. *Diabet Med* 2008; **25**(4): 383-99.

44. Martin-Hernandez E, Garcia-Silva MT, Vara J, et al. Renal pathology in children with mitochondrial diseases. *Pediatr Nephrol* 2005; **20**(9): 1299-305.

45. Karaa A, Kriger J, Grier J, et al. Mitochondrial disease patients' perception of dietary supplements' use. *Mol Genet Metab* 2016; **119**(1-2): 100-8.

46. Senger BA, Ward LD, Barbosa-Leiker C, Bindler RC. The Parent Experience of Caring for a Child with Mitochondrial Disease. *J Pediatr Nurs* 2016; **31**(1): 32-41.

47. Alston CL, Rocha MC, Lax NZ, Turnbull DM, Taylor RW. The genetics and pathology of mitochondrial disease. *J Pathol* 2017; **241**(2): 236-50.

48. Mancuso M, Orsucci D, Angelini C, et al. Redefining phenotypes associated with mitochondrial DNA single deletion. *J Neurol* 2015; **262**(5): 1301-9.

49. Mayr JA, Haack TB, Freisinger P, et al. Spectrum of combined respiratory chain defects. *J Inherit Metab Dis* 2015; **38**(4): 629-40.

50. Haack TB, Danhauser K, Haberberger B, et al. Exome sequencing identifies ACAD9 mutations as a cause of complex I deficiency. *Nat Genet* 2010; **42**(12): 1131-4.

51. Zweers H, Janssen MC, Leij S, Wanten G. Patients With Mitochondrial Disease Have an Inadequate Nutritional Intake. *JPEN J Parenter Enteral Nutr* 2017: 148607117699792.

52. Franik S, Huidekoper HH, Visser G, et al. High prevalence of complementary and alternative medicine use in patients with genetically proven mitochondrial disorders. *J Inherit Metab Dis* 2015; **38**(3): 477-82.

53. Enns GM, Kinsman SL, Perlman SL, et al. Initial experience in the treatment of inherited mitochondrial disease with EPI-743. *Mol Genet Metab* 2012; **105**(1): 91-102.

54. Avula S, Parikh S, Demarest S, Kurz J, Gropman A. Treatment of mitochondrial disorders. *Curr Treat Options Neurol* 2014; **16**(6): 292.

55. Parikh S, Goldstein A, Koenig MK, et al. Practice patterns of mitochondrial disease physicians in North America. Part 2: treatment, care and management. *Mitochondrion* 2013; **13**(6): 681-7.

56. Noorda G, van Achterberg T, van der Hooft T, Smeitink J, Schoonhoven L, van Engelen B. Problems of adults with a mitochondrial disease - the patients' perspective: focus on loss. *JIMD Rep* 2012; **6**: 85-94.

57. Kim KR, Lee E, Namkoong K, Lee YM, Lee JS, Kim HD. Caregiver's burden and quality of life in mitochondrial disease. *Pediatr Neurol* 2010; **42**(4): 271-6.

58. Sofou K. Mitochondrial disease: a challenge for the caregiver, the family, and society. *J Child Neurol* 2013; **28**(5): 663-7.

59. Vento JM, Pappa B. Genetic counseling in mitochondrial disease. *Neurotherapeutics* 2013; **10**(2): 243-50.

60. Pelentsov LJ, Laws TA, Esterman AJ. The supportive care needs of parents caring for a child with a rare disease: A scoping review. *Disabil Health J* 2015; **8**(4): 475-91.

Section 12 Next steps

Next steps

At the end of each PEEK study, CCDR identifies three key areas that, if improved, would significantly increase the quality of life and/or the ability for individuals to better manage their own health.

In relation to the mitochondrial disease community, these three areas are:

1. Mechanisms to support health professional education, including those that support patients in explaining their condition to new health professionals that they may encounter
2. Case management services to provide holistic management of mitochondrial disease to patient and their families and ensure continuity of care across health services
3. Information that empowers patients, particularly at diagnosis; and ensure that all information is available in formats that are appropriate to those with visual and hearing impairments

2018 Mitochondrial disease metrics

Data collected in this PEEK study also provides a basis on which future interventions and public health initiatives can be based. Some of the 2018 mitochondrial disease metrics that the sector can work together to improve upon are provided in Table 12.1

Table 12.1: Mitochondrial disease 2018 Metrics

Area of evaluation	2018 data	
Baseline health	**Mean**	**Median**
Physical functioning	**32.50**	**43.75**
Role functioning/physical	12.50	0.00
Role functioning/emotional	**43.33**	**33.33**
Energy/fatigue	22.50*	25.00
Emotional well-being	**64.00**	**68.00**
Social functioning	39.75	37.50
Pain	**46.90**	**45.00**
General health	28.00	25.00
Health change	**35.50**	**25.00**
Percentage of participants that have accessed My Health Record	10.00%	
Percentage of participants that have a discussion about biomarkers/genetic tests	42.00%	
Knowledge of condition and treatments (Partners in Health)	**Mean**	**Median**
Knowledge	**23.32**	**24.00**
Coping	13.40*	13.00
Recognition and management of symptoms	**18.76***	**19.00**
Adherence to treatment	13.18	14.00
Total score	**68.66**	**71.50**
Care Coordination and care received	**Mean**	**Median**
Communication	**33.40***	**36.00**
Navigation	22.28*	21.50
Total Score	**55.68***	**57.00**
Care coordination global measure	4.76	5.00
Quality of care global measure	**5.52**	**6.00**
Fear of progression	**Mean**	**Median**
Total Score	**34.10***	**34.00**

*Normal distribution, use mean as measure of central tendency