

# **Summary of results**

### Section 1: Introduction and methodology

- Mitochondrial disease is a heterogeneous 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 Health – 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 expectancy 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 next 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 were 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.