Problems of Adults with a Mitochondrial Disease – The Patients' Perspective: Focus on Loss

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Abstract *Objective:* This study aimed to identify problems as experienced by adults with a mitochondrial disease. We chose to describe these problems from the patients' perspective as we thought this would give optimal input for care improvement.

Design: A qualitative design using the grounded theory approach, involving single individual open interviews with 16 adults with a mitochondrial disease.

Results: This study revealed "loss" as the main concept, where one loss often preceded other losses. Respondents mentioned loss of energy as the most basic loss in having a mitochondrial disease, leading to multiple varied losses in all areas of daily life. Looking back, they mentioned psychosocial-spiritual problems and dealing with actual and expected future losses as most prominent and difficult. In our study, we determined seven comprehensive themes: "loss of energy", "loss of independence and autonomy", "loss of social participation", "loss of personal identity", "loss of dreams and future" as well as "lack of health care" and "coping and adjustment".

Conclusions: In this first study on this theme, we developed a theory about patient's losses related to mitochondrial disease. They experience a variety of problems in the physical, psychological, social and spiritual human domain. Patients experience a lack of health care and professional support as well. They use various strategies to manage their multiple losses and experience circumstances in which it is difficult to manage them effectively. The results can be used as a guide for improving health care, education and research and might result in an improved quality of life.

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Abbreviations

MD(s)	Mitochondrial disease(s)
VKS	Volwassenen en Kinderen met een Stofwisselingsziekte (adults and children with a metabolic disease).
VSN	Vereniging Spierziekten Nederland (Association of Muscular Diseases Netherlands)

Introduction

The term mitochondrial disease (MD) is used for complex genetic diseases affecting cellular energy metabolism which manifests in tissues and organs with high-energy requirements such as brain, muscles and heart (Smeitink et al. 2001). Typical signs and symptoms include muscle weakness, physical effort problems, spasticity, cramps, fatigue and exhaustion, heart failure, hanging eyelids, deafness, blindness, epilepsy, mental disabilities, kidney failure, liver failure, diabetes mellitus, migraines, behavioural problems and problems with sleep and nutrition (Smeitink et al. 2006; Dodds 2001). The manifestations, age of onset and progression of MD vary considerably, thus complicating its identification. In general, there are two types of MDs, those that manifest themselves at birth with rapid deterioration often resulting in an early death and those that manifest themselves later in life. Both types occur equally often (Dodds 2001). Among the different groups of inborn errors of metabolism, MD is the most frequent with an incidence of approximately 1:5,000 (Sanderson et al. 2006; Smeitink et al. 2006; Schaefer et al. 2004) but likely to be much higher due to under diagnosis (Edmond 2009). Currently, there is no effective pharmacological treatment. Management varies considerably and is limited to supplemental and supportive interventions of which the effectiveness varies and clear evidence is lacking (Koene and Smeitink 2009; Parikh et al. 2009; Chinnery et al. 2006). In recent years, substantial progress has been made in the clinical and biomedical understanding of MD. However, studies on consequences in daily life are lacking (Noorda et al. 2007), while we know from clinical practice that adults with an MD experience many problems in all areas of human functioning and in health care and that having an MD has a huge impact on their own life as well as on the lives of their family. To gain more insight into and better understanding of these problems, we carried out a qualitative study. The aim was to identify problems of adults in daily life in relation to their MD. We chose to describe these problems from the patients' perspective as we believed this would give optimal input for health care improvement and might result in an improved quality of life.

Method

Design

Because our main purpose was to explore and understand unknown experiences of persons with MD, we chose a qualitative design. Grounded theory, an inductive method, was applied, as a method suitable in situations where

little is known about a particular topic or problem area (Holloway and Wheeler 2002). The purpose was to conceptualize the essence of problems of patients with MD, to generate comprehensive explanations of phenomena that are "grounded in the patient's reality" and to develop theory (Charmaz 2009; Silverman 2006; Polit and Beck 2004; Holloway and Wheeler 2002).

Recruitment

To achieve optimal homogeneity in the group of MD characterized by their heterogeneity, only persons included were those diagnosed with one of the most prevalent and well-defined MDs, namely, MELAS (mitochondrial encephalopathy lactic acidosis and stroke), MERRF (myoclonic epilepsy associated with ragged-red fibres), NARP (neurogenic muscle weakness ataxia and retinitis pigmentosa) or LD (Leigh disease). Other inclusion criteria were 18 years or older, willing to consent, living in the Netherlands and mastering the Dutch language. Diversity in registration and classification of MD made a targeted recruitment difficult. Therefore, we broadly approached the patient group by sending a letter with information about the study, a general questionnaire and an invitation to participate in this study. Figure 1 presents the recruited groups and enrolled patients. Patients who decided to participate were asked to fill out a general questionnaire and to sign the consent form for participation in and tape recording of the interview.

Sample

The questionnaire was used to purposefully sample respondents who were eligible for our study and to obtain maximum variation between participants in relation to their illness and personal situation. A total of 43 persons reacted positively, from whom 20 met the inclusion criteria. Saturation, the collection of data to the point where a sense of completeness of data is attained because new data yield redundant information (Polit and Beck 2004), was reached after 14 interviews. Two additional interviews were conducted to ensure the themes were relevant and no new themes evolved. With that, totally 16 persons participated in the study. Most respondents were female ($n = 12$), had a mean age of 43 years, had low levels of education ($n = 11$) and were incapacitated for employment ($n = 11$). Eight respondents were single or divorced and nine had one or more children from whom three had children affected with MD. Seven respondents lived with a partner and/or children, two lived with parents, and four lived alone. Three respondents lived in an adapted house or care institute. Respondents' mean age of "first manifestation

Fig. 1 Recruitment and sample

Table 1 Medical diagnoses in adults with MD ($n = 16$)

Medical diagnoses	Frequency
Mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes (MELAS)	11
Myoclonic epilepsy with ragged-red fibres (MERRF)	4
Leigh syndrome (LS)	1

of symptoms” was 16.8 years ($n = 14$) and of “age of diagnosis” was 36.3 years ($n = 16$). Overviews of respondent’s diagnosis and symptoms are presented in Tables 1 and 2.

Data Collection

Data were collected by using open interviews of 60–120 min, which started with an open-ended question, to give the researcher the possibility to explore experiences: “What are the most important problems you experience in

daily life in relation to your MD?” The interviews were conducted by the first author (GN), who was trained in open interviewing. We used open interviews to facilitate in-depth information, not only about actual problems and needs, but also about the perception or the context in which they arise. The first three interviews served as try outs for the researcher and were evaluated by an expert in qualitative research (TvdH). Because only minor amendments were proposed, the three piloted interviews were included in the analysis. To increase *bracketing*, *objectivity* and *neutrality* in interviewing, there were different moments of reflection with the supervisor (TvA) and the research group (TvA, BvE, GN, LS, JS). Data collection took place in 2007–2008. All respondents were interviewed at home. We proposed private interviews, but six patients chose to have a third person with them for support during the interview (three partners and three mothers). All interviews were tape-recorded and transcribed verbatim. The quality of the data was enhanced by creating optimal conditions for participants’ real narratives, such as interviewing in the familiarity of their own house and showing

Table 2 Symptoms in adults with MD ($n = 16$)

Symptoms	Frequency
<i>Total symptoms</i>	
Muscle features: (chronic) fatigue, fatigability, weakness, cramp, ptosis	16
Pain (overall pain, muscle pain)	11
Hearing loss	9
Cardiac problems	8
Diabetes mellitus	7
Disabled in daily activities	7
Impaired temperature regulation	7
Respiratory problems	7
Cognitive problems (thinking, memory)	7
Extreme increase of symptoms during mild infections	6
Gastrointestinal problems, e.g. heartburn, constipation and loss of weight	5
Migraine	5
Visual loss	5
Epilepsy	4
Kidney problems	4
Deceased family members in first family degree	4
Problems with perspiration	3
Short stature	3
<i>Total number of symptoms per person</i>	
0 symptoms	0
1–5 symptoms	2
6–10 symptoms	2
11–15 symptoms	7
16–20 symptoms	5

empathy. *Member validation* was used by giving verbal summaries during and after the interview.

Data Analysis

Data analysis was based on the principles of constant comparison, the main method within grounded theory.

All relevant and meaningful fragments of the transcript were systematically coded, compared and categorized. The final stage concentrated on selecting central and main themes and systematically relating these to other themes to answer the research question. The first researcher (GN) coded the interviews. To increase the *theoretical sensitivity* of the researcher, five transcripts were analysed and independently coded by the researcher and supervisor (TvA), and the – minor – variations in interpretation were discussed until consensus was achieved. Results were presented to the members of the research group (TvA, BvE, GN, LS, JS) for critical reflection (*peer debriefing*).

Results

This study revealed “loss” as the central theme: All respondents reported experiences in the area of loss, where loss means losing one or more essential elements of a persons’ life and where each loss often contributes to other loss(es), which could lead to the hypothesis that one loss leads to another loss. All respondents seemed fully aware of these experiences. They mentioned loss of energy as the base on having an MD, leading to multiple losses in all areas of daily life. Looking back, they mentioned psychosocial problems and dealings with (future) losses as most prominent and difficult. As respondents got older, they experienced more and/or other problems. For example, young persons mentioned loss of their desire to have (more) children, whereas older persons experienced loss of participation because their child(ren) left home or significant others died. In our study, we determined seven comprehensive themes: “loss of energy”, “loss of independence and autonomy”, “loss of social participation”, “loss of personal identity”, “loss of dreams and future” as well as “lack of health care” and “coping and adjustment”. These themes and the relationships between them will be described successively. Table 3 reports the respondents’ quotes, supporting the validation of our study results.

Loss of Energy: Continuous and Overpowering

All respondents mentioned a variable loss of energy and had the experience that their energy was decreasing and their body was failing gradually.

Physically, all respondents suffered from overall weakness and mild or extreme/intense fatigue or exhaustion, frequently without a direct link to physical effort. Loss of energy manifested itself as loss of muscle power, muscle pain, stiffness, firmness and/or cramps, and problems with physical effort, condition, walking, moving and speaking. Recovery had a changing and uneven course.

Psychologically, energy problems reflected reduced concentration and memory. Loss of concentration established itself during reading, conversations, performing more activities at the same moment and “complex” activities. Loss of memory primarily concerned problems with short-term memory. Respondents forgot what they just had said or read, or did not remember what they just intended to say.

Respondents with a more stable energy pattern experienced similar symptoms every day. Other patients experienced unstable periods of changing levels of energy, which were usually unpredictable, of variable intensity, occurring without a specific reason and often without a pattern. All respondents mentioned progression and an increase in loss of energy over time.

Table 3 Respondent's quotes**Loss of energy: continuous and overpowering**

I'm dead and dead tired and it is increasingly difficult to start moving. It's just like a battery that quickly empties. And recharging takes a long time...

There are good days and bad days. One day I can walk a long way and the other day I can hardly walk a short distance.

Loss of independence and autonomy: being able to do less and less

After bathing, I have no more energy for drying myself. I'm first going to sit on the ground to recover. And when after a time of resting I still do not have enough energy, my husband needs to help me.

Loss of social participation: when belonging is lost

Normally I don't look sick. And when I am feeling sick or exhausted I stay home. Nobody sees me on bad days.

It is very difficult now I am not able to go to a theatre any longer. You must now fill your time with fewer things from a second or lesser choice.

Loss of personal identity: not being themselves anymore

And there goes my ego if I need to go home during dinner in a restaurant and have to ask my friends to bring me there...

Loss of dreams and future: not living the expected life

We are often busy with questions about life that most people do not have or even think about.

The first question after hearing the diagnosis was how long will my daughter and I live?

Coping and adjustment: crossing borders and searching for a new balance

When I heard the diagnosis it was the completion of the puzzle. It also gave certainty. Then I knew what was going on...

Nobody can imagine what a struggle it is, every day again... Why should I fight if I know it is only getting worse with me? Why should I... You are forced to give it a place in your life, but it will never happen because you do not agree with it...

Lack of health care

Doctors have no time and attention for questions, I am outside within 5 min, while I have several things I want to know. For them probably small things, for me things to make life easier.

Doctors never have attention for the whole but only for one part. The internist looks at diabetes, the cardiologist at the heart, the neurologist to my muscles. But nobody keeps in mind that the symptoms stem from MD. There is no communication between professionals. They do not know what other professionals are doing and prescribing. Consequence is that I have to remember who is prescribing what and why. Now you have to inform doctors. You are getting so tired and sick of that...

Loss of Independence and Autonomy: Being Able to Do Less and Less

In most respondents, the loss of energy resulted in an increasingly lower level of functioning which led to more restrictions in daily life activities necessary to live independently. All respondents mentioned loss of mobility such as difficulties with walking, cycling, driving a car, personal care, eating, speaking, writing, housekeeping, gardening, daily shopping and caring for their child(ren) and dogs. They increasingly depended on instruments such as a stick, wheelchair, scoot mobile and/or the devices of others and taught themselves a lot of tricks and other ingenious ways to cope. Three respondents could no longer live independently and were dependent on parents or professionals. Another three planned to move to a smaller ground-floor apartment without a garden or a house with custom-made facilities.

Increasing energy-related problems in speaking and concentration played an important role in communication with other people, deteriorating when combined with hearing and/or vision problems. When respondents' ability to think, communicate, decide and/or act independently was restricted, it was more and more difficult to accept help

from others and to live their life according to their own values and rules.

Loss of Social Participation: When Belonging Is Lost

Loss of energy and reduced ability to function independently seemed to restrict social participation in public and personal life. Many respondents had to reduce their job or study hours or lost their employment, leading to a lower income while expenses increased, loss of meaningful activities and social contacts with colleagues, often resulting in feelings of emptiness. Replaced voluntary work or study became problematic when expectations could no longer be met. Many respondents experienced problems with leisure activities, public life and agencies. Most respondents expressed much pressure, change and loss within the relationship with their own healthy partner, children, family and friends, especially when there were different emotions and perceptions, often leading to changed roles and exclusion. When they had children, they expressed their concerns about them and the importance of adequate attention and support for them, while bringing up, caring for and supporting them as well other family members were increasingly difficult. More complex was

living in a family with a strong family history of more than one MD-diagnosed relative. This strengthened the patients' own losses and problems and had a high impact on all family members. Some experienced this very complicated situation as being almost unbearable and difficult to handle. Most respondents mentioned lack of understanding and support from their social environment as one of the most important problems they faced. They indicated that their relatives did not acknowledge their problems or were increasingly withdrawing, whereas some others experienced over-pitying or overprotection, which respondents found difficult to handle. Underestimating having an MD, due to its frequently invisible character and lack of knowledge, influenced this as well as the own insufficient energy to enjoy a social life. All mentioned the importance of a qualitative supportive social network, but most of them experienced erosion of it while the need for support increased. This had a major impact and often resulted in withdrawal from life, frequently leading to feelings of not belonging to a group (anymore), struggling alone and (fear for) possible loneliness and social isolation. Some respondents mentioned the value of new "rich" and meaningful contacts with fellow patients because they truly understood their situation.

Loss of Personal Identity: Not Being Themselves Anymore

Many respondents indicated problems with alterations in their roles and level of self-esteem. They were always aware of "being different" and mentioned to be indecisive about their own role(s), e.g. between being a person with symptoms or a patient, being a first- or second-class person or being a participant or outsider in social networks. But they were also indecisive about others' roles: Is their partner still partner or caregiver? They were faced with questions about a changing "self", "who am I (now)" and "how do others see me (now)"? Respondents wished to confront others' perception of them, as being sick, pathetic, putting on airs, whining and lazy. Ideally, they liked to be normal and maintain their own role as before, even if that no longer exists.

Loss of Dreams and Future: Not Living the Expected Life

The loss(es) they experienced overwhelmed the most respondents and their expectations about life. They expressed their concerns about the future and mentioned unattainable expectations of life and life goals, mostly with respect to relationship(s) and social context such as the desire to have children. They could not live the expected life or the life they dreamt about. Respondents were faced with more and more spiritual and existential – and unanswerable – questions about why, why me, why so

much, why all at once and about suffering and mortality. Because of the unpredictable and fatal course of the disease, there were often many questions about the unknown uncertainties with regards to life and longevity. Most respondents were faced and struggling with difficult ethical questions about life and heredity, such as genetic testing and informing children and other family members. Two respondents talked about hope, specific for medical treatment.

Lack of Health Care: Need for Professional Support

In the professional context, respondents mentioned lack of health care as an important factor influencing coping with their loss(es). Firstly, respondents mentioned shortcomings in information provision while they needed clear and actual information about all aspects of having an MD, preferably tailored to their own needs, opportunities and circumstances. But also conflicting needs, such as "knowing" was just as hard as "not knowing". All mentioned to understand difficulties for professionals to predict the course of their MD, because "nobody knows". Secondly, many respondents were of the opinion that professional attention and treatment was mainly focussed on biomedical information, tests and sometimes physical symptoms while they experienced insufficient psychological, social and spiritual attention. Thirdly, most respondents mentioned a structural shortage of follow-up, professional care, sensitivity and support, especially when the doctor was telling there was nothing they could do. They mentioned to need an expert who "thinks along with them". Finally, lack of coordination and continuity in health care was mentioned as a major problem, especially when they saw multiple professionals for various symptoms and problems. Uncertainty and doubt increased when professionals had different or contrary approaches on treatment and support and/or had insufficient (new) knowledge about and clinical experience with MD. This made communication about their problems more difficult and stressful.

Coping and Adjustment: Crossing Borders and Searching for a New Balance

The process of coping and adjusting started with the discovery of having an MD. This period was preceded mostly by a long, uncertain and stressful period characterized by incorrect medical diagnoses, feelings of "not being heard or believed" and little recognition. Respondents were confronted with a complex, progressive and untreatable disease on which they reacted in different ways: worried, shocked, falling into a black hole and feeling exhausted and completely alone. Respondents from an affected family were more aware of what they could expect.

The initial impact was associated with awareness of consequences for daily life, actual and possible future losses and suffering, many questions about expectancy of life and future, and feelings of grief, fear, anger, sadness, emotional pain and powerlessness.

Moving towards the palliative phase, management focused on searching for a new balance. At the beginning on finding a new energy balance and maintaining physical functions, which are difficult experiences because they did not know their boundaries, did not recognize their limit in time and/or had no grip on balancing and anticipating on overload. Balancing required consistent discipline while they exhausted themselves deliberately to continue the things of life, stay with their social group and meet the expectations of others or to live a more pleasant, joyful and valuable life. They realized that this takes delay and/or days of recovery. Besides crossing borders, they adapted their lifestyle by taking time to rest if they needed to, scheduling activities precisely, listening to themselves and figuring out what works for them. Looking back, energy management was not the most difficult part of adjustment. Uncertainties about the unpredictable and progressive course of MD made life heavier while each loss should be dealt with. Most respondents expressed constant, daily stress while their lack of energy negatively influenced their capacity to cope effectively. Respondents used a wide variety of coping strategies, more or less effective. Problem-focused strategies included searching and finding information; learning new skills, tricks and devices and planning activities. Information gave not only more certainty and grip on their situation but also confrontation with unwanted information, confusion, uncertainty, threat and fear.

Respondents indicated that dealing with their increasing loss(es) was accompanied with emotions as powerlessness, despair, grief, rebellion, anger and/or negativity, mostly as reaction on feelings of uncertainty, vulnerability and (existential) anxiety of what might or might not come. Most respondents also experienced grief and anger towards professionals, agencies and their social environment because of their lack of understanding. They felt a continuous pressure of standing up for their situation and rights while they actually had no energy for it. There were feelings of doubt and guilt towards the partner or children also because of passing defective genes to their children and unfulfillable life expectations. This could lead to more emotional pressure on their relationship as well as to more solidarity. Some respondents emphasized feelings of shame in relation to limitations, accepting help or the use of devices and put off aid as long as possible.

Emotion-focused strategies included “grieving and mourning”, especially after regression and “denial” such as shutting out fatal consequences, not thinking about it or blocking it. For most respondents, it was “hard to talk about

their situation, feelings and emotions”, because they did not want to remind of their losses, complain about their situation or not finding the right moments or circumstances.

Benefits came not only from “positive attitudes to life”, “enjoying the good things in life”, “remembering positive memories”, “receiving support, sympathy, understanding and conformation from others”, “seeking peers” and “using humour, life mottos and maxims” as “living for today”, “making the best of the situation” and “there are people who are worse off than I am” but also from reading literature about fellow’s experiences, grief and poetry.

For adjusting to MD in daily life, respondents used several coping strategies simultaneously in different stages of life and disease, with varied effects. For the most of them, it was hard to resign themselves to their situation and loss. Adjustment took place gradually if it occurs; acceptance seemed to be hardly reachable or not.

Discussion

As similar studies were not reported, comparing with other studies of patients with MD is hardly possible. However, the results resemble those from studies in people with other loss-related neurodegenerative and severe chronic diseases (Issaksson and Ahlström 2008; Ahlström 2007; Issaksson et al. 2007; Pool et al. 2004; Pool and Stap 2004; Paterson et al. 2003; Lindgren 1996; Eakes et al. 1993) [Abma TA, Nierse CJ, Horemans AMC, Vos I, de Jonge R, van Engelen B, 2007, Over kwaliteit en gezondheid. Een dialoog over onderzoek tussen mensen met een spierziekte en onderzoekers (About quality and health. A dialogue about research between adults with a myopathie and researchers). Niet gepubliceerde uitgave van VSN, ISNO/UMC St Radboud, ZonMW, unpublished]. Yet, the great uncertainty about life and longevity, an unpredictable progressive course, the invisible character and multiple recurrent losses are probably more specific for having an MD.

The concept “loss” is described in literature mostly in related terms as sadness, stress, distress, grief, chronic sorrow, bereavement and mourning (Corr et al. 2000; Lowes and Lyne 2000). Loss in our study matches closely with the concept analysis of Robinson and McKenna (1998). They refer to a study of Peretz (1970), who described loss as “a part of self and including a number of physical, social and role-related losses”. Rando (1993) argues also that loss may be physical (tangible) as well as psychosocial (intangible, symbolic). In our study, respondents narrated mostly at first about their overwhelming tangible problems probably because these are more concrete and therefore easier to talk about. They narrated about their intangible problems in second instance, while they mentioned psychosocial-spiritual problems and

dealing with (future) losses as most prominent and difficult in their life.

For “multiple losses” Robinson and McKenna (1998) refer to Powel-Cope (1995), who concluded also that loss precipitated other losses as in other diagnostic groups such as diabetes mellitus (Manderson and Kokanovic 2009), multiple sclerosis (Issaksson and Ahlström 2008) and chronic pain (Harris et al. 2003). Again, our results are in the line of these studies. The more tangible losses have been mentioned in these studies, too, so we know already that fatigue is a typical symptom of neurological diseases (Chaudhuri and Behan 2004), which is present in more than 60% of patients with a neuromuscular disorder (Schillings et al. 2007; Kalkman et al. 2005). Remarkably, in our study, only two respondents talked about hope (for medical treatment). Yet, hope can be an important up-lifting emotion and in that way important for coping and adjustment (Ahlström 2007; Smith and Kirby 2009), resilience (Ho et al. 2010) and taking control of one’s life (Bauman 2004). Consequences of loss can be positive and negative. Although our study is focussed on “problems”, respondents narrated also about positive consequences as personal growth, deepened relationships, enjoying the usual things of life and changing perspectives on life goals. From literature, we know that positive consequences are focused on recovery (Mancini and Bonanno 2009), personal growth (Folkman and Moskowitz 2004), life satisfaction, positive affect, dyadic adjustment, better relationships, promoted well-being, new priorities, goals sustained (Pakenham 2005; Mohr et al. 1999) and increasing comfort (Issaksson and Ahlström 2008). This perspective requires effective management (Issaksson and Ahlström 2008) and a change of focus from pathology to positive psychology (Somerfield and McCrae 2000).

Losses require management methods, which refer to strategies used by the patient (internal management) and to interventions provided by relatives and professionals (external management) both (Issaksson and Ahlström 2008; Eakes et al. 1998). From the results of our study, we conclude that most respondents use different kind of methods managing their multiple losses both effectively and ineffectively. Effective internal management includes mastering with realistic awareness, allowing and sharing emotions, accepting and adjusting, using different strategies, “can-do” attitude, thinking positively and constructively, creating hope, taking care of oneself and obtaining and using support from the own social network. Effective external management refers to “endorsing management” and includes a caring and encouraging family, respectful acknowledgement from friends and confirmation from health care professionals (Issaksson and Ahlström 2008).

Finally, the strengths of this study are reflected in describing problems from the patients’ perspective and the

measures taken to improve reliability and validity (Tong et al. 2007; Russel and Gregory 2003; Giacomini and Cook 2000a, b; Pope and Mays 1995). Nevertheless, it has its limitations. Although the sample shows a great diversity in characteristics, it does not reflect all types of patients, e.g. it does not involve patients with a different cultural background. Because the number of respondents who met the inclusion criteria was limited, the use of purposive sampling was limited, too. However, taking account of the richness of the obtained data, we think the themes are also likely to be comparable to problems of other patients with MD. Involving patients as primary informants was very valuable because they are competent interpreters of their own world, which can provide a valid, meaningful description of their problems. Respondents were very willing to participate in this study contributing to a better health care and quality of life for themselves as well as for new diagnosed patients. More than 23 persons took the initiative to express that they wanted to be involved in the study, but did not meet the inclusion criteria. Finally, during six interviews, a third person was present on request of the respondent, which might have contributed to a more comfortable and safe situation but also might have influenced the results. Nevertheless, we suspect that respondents could speak freely and openly because they and the third persons showed to be very concerned.

Conclusion and Recommendations

We conclude that adults with MD experience a variety of problems in the physical, psychological, social and spiritual human domain. They experience a lack of health care and professional support as well. We also conclude that they use various strategies to manage their multiple losses both effectively and ineffectively. They experience circumstances in which it is difficult to manage their losses effectively.

We think that our results are relevant to improve quality of health care, education and research and might result in an improved quality of their lives. Understanding their experiences and problems might help professionals in supporting patients adequately and tailoring patient care. For internal management, interventions such as training and promotion of health, coping, empowerment, self-management and participation in clinical decision making could be considered. For external management methods, we think about interventions for significant relatives as education and promoting awareness, involving and supporting. For health care improvement, interventions in the field of integrative and transmutal care could be considered, including education and improvement of competences of professionals in patient and family centeredness, communication with

patients, shared decision making, information providing and presence. Also, opportunities in shared care could be considered, in which “patients and professionals” can communicate and exchange knowledge and experiences, including between the expertise centre, the general practitioner, the involved local professionals and rehabilitation centres. We see also digital opportunities as e-health, e-learning, e-consults and e-conferencing.

Further research is recommended, for instance, for inquiring the perspectives of professionals and patients’ significant relatives; for further deepening, defining and validating concepts as energy management, loss and quality of life in relation to MD and for inquiring and deepening effective coping strategies, family support strategies and other interventions, which can be used as input for effective management programmes.

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A Concise 2–3-Sentence Summary

A qualitative study identifying problems of adults with a mitochondrial disease presented “loss” as the main theme, where mostly one loss preceded other losses. “Loss of energy”, “loss of independence and autonomy”, “loss of social participation”, “loss of personal identity”, “loss of dreams and future” and “lack of health care” and “coping and adjustment” were identified as the most important themes related to mitochondrial diseases.

Authors’ Contributions

Study design and planning: GN, TvA and LS, Data collection and analysis: GN, TvA, TvdH, Manuscript preparation: GN, TvA, TvdH, JS, LS, BvE, Manuscript revision: GN, TvA, TvdH, LS, BvE.

Guarantor for the Article

Name of one author who serves as guarantor for the article, accepts full responsibility for the work and/or the conduct of the study, had access to the data and controlled the decision to publish: GN

Competing Interests

All authors declare that in answer to all questions on the JIMD competing interest form they have nothing to declare.

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Ethic Approvals

The local medical ethics committee (CMO – Nijmegen / Arnhem) approved the study.

Informed Consent

All respondents gave written informed consent before participating in the interview.

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