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It’s not just physical: a qualitative study regarding the illness experiences of people with facioscapulohumeral muscular dystrophy

Minne Bakker, Karen Schipper, Alexander C. Geurts and Tineke A. Abma

ABSTRACT

Purpose: Little is known about the illness experiences of people with Facioscapulohumeral Muscular Dystrophy (FSHD). The aim of this study was to provide insight into the illness experiences of people with FSHD in order to tailor rehabilitation programs to individual needs and expectations.

Methods: Twenty-five semi-structured interviews were conducted with people with FSHD. The interviews were audiorecorded, transcribed and checked. Computerized (MAXqda) and manual techniques were used for thematic data analysis.

Results: Intra- as well as extra-individual aspects play a role in the illness experiences of people with FSHD. Integrating the consequences of the diagnosis and symptoms, coping with heredity and progeny, adjusting to a decreasing independence, and the accompanying changing relationship with one’s partner, are mentioned as intra-individual aspects. As extra-individual factors are the responses of the social environment, which was mentioned as well as used assistive devices, and maintaining or giving up work.

Conclusions: Better understanding of the individual illness experiences, cognitions, and social context of people with FSHD can give health professionals tools to improve their care and give researchers direction for future studies to evaluate healthcare improvements from a holistic, patient-centred perspective.

IMPLICATIONS FOR REHABILITATION

- FSHD has a major impact on people’s lives. Besides the physical consequences, issues such as heredity, progeny, changing (intimate) relationships, social interactions and work should be addressed by rehabilitation professionals.
- Depending on the timing of the diagnosis (early or later in life) people with FSHD could, in addition to medical consultation and physical therapy, profit from support by a social worker, occupational therapist and/or genetic counselor for the above-mentioned themes to be addressed more extensively.
- It is relevant for rehabilitation professionals to become familiar with the personal characteristics and social circumstances of the patient before communicating the diagnosis and prognosis in order to individually tailor the context of the communication.

Introduction

Chronic diseases obviously have a great impact on the quality of people’s lives.[1,2] This is particularly true for progressive diseases as people constantly have to adjust to increasing disabilities.[3,4] There are many progressive diseases, of which neuromuscular diseases (NMD) constitute a large group.[5] There are about 500 different types of NMDs that are all incurable, but which differ in the rate of progression, ranging from fast progressive (e.g. Amyotrophic Lateral Sclerosis (ALS)) to slowly progressive (e.g. FacioScapuloHumeral Muscular Dystrophy (FSHD)).

In the literature, many quantitative studies describe the quality of life of people with NMDs.[6-8] All these studies conclude that NMDs often lead to the loss of daily activities and a decreased general health.[6,9] These quantitative studies provide important insights into patients’ functioning. However, a limitation of such studies is that they are less suitable for exploring the individual meaning of living with a NMD.[10]

The course and consequences of a disease on the experienced quality of life depend not only on physical limitations, but also on everyday life in a social context.[11] Relatively few studies have addressed this broader context of the illness experiences of people with NMDs. The studies that have been conducted conclude that living with a chronic, deteriorative illness has great impact on people’s lives. This has to do with physical aspects, such as deterioration of the muscles, excessive fatigue and pain, as well as with social aspects, such as increased dependency, stigmatization and the need for constantly making (physical and psychological) adaptations.[12-14]

Recently, the number of qualitative studies addressing the illness experiences of people with NMDs is increasing.[15] Currently, the qualitative literature largely represents studies about individuals living with ALS, Duchenne Muscular Dystrophy (DMD) or Myotonic Dystrophy (MD).[13,15] Other diseases, such as FSHD,
have been studied to a much lesser extent.[15] This is remarkable, as different types of NMD have their own characteristics leading to unique problems and illness experiences. Duchenne Muscular Dystrophy, for example, is characterized by an early age of onset and a relatively fast progression, leading to a shortened life-span. ALS is known for an even faster disease progression leading to death within a few years after diagnosis. Therefore, illness experiences in these patients cannot be regarded as representative of people with a slowly progressive muscle disease. MD is, like FSHD, a hereditary muscular dystrophy with a slow to moderate progression, depending on the genetic defect size. However, unlike FSHD, MD is typically a systemic disease affecting sensory and cognitive functions in addition to skeletal muscles and muscles of internal systems such as the heart.[16] As a consequence, MD can also not serve as a representative model for studying the illness experiences of people with a slowly progressive muscle disease.

FSHD is the most common, slowly progressive, muscular dystrophy that does not have a significant impact on people’s life expectancy. Although extra-skeletal muscular symptoms (e.g. retinal vascular changes and hearing loss) can also be present in FSHD, these sensory deficits are rarely symptomatic or disabling.[17] Hence, FSHD can be regarded as the most frequent ‘pure’ form of slowly progressive muscular dystrophy. In Europe, FSHD has an estimated prevalence of 1:15,000 to 1:20,000. The disease is characterized by progressive weakness of the facial and shoulder blade stabilizing muscles. In later stages, humeral, trunk, pelvic and leg muscles often become involved as well.[18] Despite its high prevalence, still little is known about the illness experiences of people with FSHD. So far, only one qualitative study has addressed the illness experiences of people with FSHD. This study focused into people with muscular dystrophy in general (including FSHD) and showed that people with different types of muscular dystrophy address, in general, similar issues and problems, such as experiencing the diagnosis and the hereditary character of their disease as ‘traumatic’ and feeling insecure about the future.[19] As this study was focused on the comparison between different muscular dystrophies, an in-depth insight into the illness experiences of people living with FSHD is still lacking. Therefore, the aim of this study was to provide better insight in the illness experiences of people with FSHD. The obtained results can be used to tailor rehabilitation programs to the individual needs and expectations of these patients.

**Design and methods**

**Design**

The focus of this study was the illness experiences of people with FSHD. Semi-structured interviews were used for data collection, as these are preferred for gaining an understanding of the experiences of people.[20] The data were analyzed via a thematic analysis, because there was little former knowledge about the illness experiences of people with FSHD.[21]

**Participants**

Participants were purposively selected.[22] Patients had to be diagnosed with FSHD, they had to be at least 18-years old, able to understand and speak Dutch, and had to be under supervision of a rehabilitation specialist. This last criterion was chosen to make sure participants had symptoms for which they needed medical consultation. This raised the likelihood that they would be able to tell about their illness experiences. The participants were recruited via different university medical centers and rehabilitation centers in the Netherlands. In total, 25 participants were recruited for an interview. These participants were first approached by their rehabilitation specialist and after consent they were called by one of the researchers (KS or MB). During this phone call, patients were given further information about the study and were asked to participate in the interview. If willing to participate, an appointment was made for the interview.

**Data collection**

For this study, semi-structured interviews were held at people’s homes for comfort and to create an open atmosphere. The interviews had an open nature, starting with the grand tour question ‘Can you tell me something about your life with FSHD?’ Themes that were formulated in a topic guide and addressed in the interviews included personal experiences with the illness, possible difficulties, and the social environment (see the appendix for the entire topic guide). The topics were deliberately formulated broadly in order not to restrict participants. Follow-up questions were asked to get more detailed answers. The wording of the follow-up questions depended upon the comprehensiveness of the answers. The interviews were conducted in the form of conversations.

The process of data collection and analysis was iterative so that emerging themes could be further explored and validated over the course of the research.[23] This led to refinement of the topic guide. The first analysis showed that no more new themes emerged after 21 interviews which suggested data saturation.[24] To confirm the saturation, four additional interviews were held. Since the additional interviews did not give new insights compared with the other interviews, saturation was met. The interviews were conducted by the first and second author. Both were trained interviewers and took general quality criteria for the semi-structured interviews into account, such as asking open-ended questions, using different probes, and avoiding jargon.[25] The interviews lasted between 60 and 90 min and were, after participants’ consent, audio-recorded and transcribed.

**Data analysis**

Interviews were analyzed using inductive, thematic analysis.[26] The transcripts were separately read and re-read and coded by the first two authors during as well as after data collection. While reading the transcripts, phrases considering illness experiences (directly or indirectly) were coded with open coding. This means that phrases were coded with descriptive codes, such as ‘shame to use wheelchair’, ‘unwilling to cut back work’, ‘disappointed in reaction of friends’ et cetera. The codes were documented in a code list and new codes in the transcripts were each time added to the list. Codes and coded segments were then compared and grouped as main and sub-categories. Categories such as ‘responses of social environment’, ‘heredity’, ‘feelings considering the diagnosis’ were formulated as main categories, based on the code list. Sub-codes were for example ‘emotions regarding heredity’, ‘choices related to having children’ et cetera. All transcripts, including the ones that were already coded, were constantly checked and coded with use of the emerged categories. Then, the different categories of both researchers were compared and discussed in the research team until consensus was reached. This was done to increase the dependability by preventing distortions caused by the personal and professional background of the individual researcher.[27] The discussion was mainly focused on the categorization of themes. To categorize the main codes, we used the model of ‘The disablement process’ as described by Verbrugge et al.[28] In this model, a difference is made between extra- and intra-individual factors that influence the functional limitations and, thereby, the disability of
people. Relevant themes were agreed upon, and, for each theme, the most suitable quotes were selected for the final report. A mix of computerized (MAXqda) and manual techniques were used to facilitate data analysis.

Rigour
To enhance credibility, all participants received an interpretation of their interview to check the accuracy of the interviewer’s interpretation (member check).[23] Besides, two different investigators were involved in the analysis process (MB and KS) (investigator triangulation).[29] The investigators arrived at the same conclusions, which heightened our confidence in the findings.[30]

Results
Twenty-five interviews were held with 11 women (44%) and 14 men (56%) with FSHD, who were aged between 24 and 77 years (mean 56 years). The duration of the disease was within the range 1–49 years, with a mean of 16 years. Nine participants (36%) were in gainful employment, two of them full-time. Ten were on early retirement and four on retirement pension. Two (8%) were using a ventilator at night. Eight (32%) were dependent on a wheelchair; 6 of them (24%) used a wheelchair only for long distances and two of them (8%) were entirely dependent on a wheelchair. All participants were native Dutch and lived independently together with their spouses. Table 1 shows an overview of the patient characteristics.

Data analysis revealed different aspects of FSHD influencing the lives and therefore the illness experiences of the participants. These aspects can be divided into intra-individual and extra-individual factors. Intra-individual aspects originate from or operate within a person.[28] This includes aspects such as coping, lifestyle and activity accommodations. Extra-individual factors concern aspects introduced from outside the person, such as care, medicines or rehabilitation, supportive devices and the physical and social environment. The data analysis showed that the illness of people with FSHD relates to intra- as well as extra-individual factors. Intra-individual factors concern the feelings evoked when hearing the diagnosis, integrating the disabilities into one’s life, handling fatigue, wrestling with heredity and progeny, and the changing relationship with one’s partner. Extra-individual aspects include reaction of the social environment to the diagnosis, postponing the use of assistive devices, and giving up work. After 25 interviews, no new themes emerged from the data analysis, which suggested that all relevant themes concerning the intra- and extra-individual factors had been addressed. The intra- and extra-individual aspects that emerged from the data are presented in Figure 1 and will be discussed in this section.

Intra-individual factors
Feelings evoked when hearing the diagnosis
The interviews showed that participants experienced the diagnosis in different ways; some participants experienced the diagnosis as a shock, whereas for others it was more of a relief. The experiences were influenced by the process of the diagnosis. Particularly for those for whom the diagnosis had been a long search, the diagnosis was a relief.

I shook his [the doctor’s] hand and said “thank you for giving me clarity, I’m so glad to finally know what it is!” (R 20)

For other participants, the diagnosis was unexpected and overwhelming. Especially for participants who did not experience severe complaints, the diagnosis had a great impact. However, even for participants who had severe complaints, the diagnosis was sometimes startling and overwhelming.

It [the diagnosis] was a shock. Suddenly I had a muscle disease and nobody knew what my future would look like. (R 6)

Integrating the functional consequences of the diagnosis into one’s life
As mentioned above, most participants experienced the diagnosis as a positive influence on their illness legitimacy. However, acceptance of their illness was still difficult. The progression of the illness made the process of acceptance hard as the limits keep increasing.

I try to keep control over it [the disease], but you can’t control it. That’s what makes it so difficult, you know you’ll get worse, but when? That’s what makes this hard. (R 11)

Participants described the constant deterioration as very hard; the constant adjustments they had to make were a challenge and made acceptance difficult.

People with ALS have a healthy life for years and are sick for a short period of time. FSHD is lifelong with increasing impairments. I don’t know what’s worse… (R 18)

Although the diagnosis often led to more legitimacy and understanding of their limitations, some participants found that the diagnosis limited them; they described it as a pitfall to become overprotective of their body, resulting in avoiding physical activity.

I sometimes wish I didn’t know [the diagnosis]. Because I know I sometimes refuse to do things that I actually enjoy and would do if I didn’t know. I don’t do some things out of fear of wearing myself out. (R 1)

The fact that in FSHD the shoulder girdle is affected first, often in combination with weakness of the facial and neck muscles, was experienced as difficult by some participants. The participants experienced the loss of strength in their arms as very disabling.

Difficulties with walking are inconvenient, but then you can use a wheelchair. But you need your arms to take care of yourself. I can hardly...
eat soup with a spoon. You lose your independence, become more dependent. That's difficult. (R 13)

The fact that also facial muscles are affected is experienced as a major downside by some patients. They feel constantly confronted with their disease, as it is always visible in mirrors and pictures. Besides, it also negatively affects their non-verbal communication.

My face shows extreme expressions. Even if I'm serious it looks as if I'm smiling. You communicate a lot with your face. I lost half of my abilities to communicate. (R 6)

Handling fatigue

All participants mentioned fatigue as an important symptom. Although the participants had other physical symptoms as well, fatigue was experienced as the main cause of limitations.

The fatigue is more problematic than my bad way of walking or the fact that I can't kneel down anymore. (R 9)

All participants described difficulties with handling fatigue. This had to do with different aspects of their fatigue. One aspect was that it was difficult to predict when they were overusing their energy. The fact that fatigue made it difficult for them to plan in advance was experienced as very problematic.

I can be overwhelmed by fatigue. I have to find out how far I can go and plan in advance, but it is difficult to know your limits and to estimate what your energy level will be. (R 4)

Some participants recognized two different types of fatigue: physical fatigue and mental fatigue. They tried to distinguish which type of fatigue they were experiencing in order to decide how to react to it.

I have to listen to my body to find out whether my muscles are tired or whether it’s my mind that is tired. Those are two different kinds of fatigue. If my mind is tired I have to ignore it, but if my muscles are tired it is very important to give in. Because, if I want to be able to function well, I have to make sure that my body and muscles have enough energy to get me through the day. (R 5)

Although all participants felt it was best not to exhaust their body and muscles, to take the time to rest was often rather difficult for them.

Of course I feel the fatigue, but my need to perform is stronger (…) I feel the need to keep on going and I want to prove myself. I want to go for it and give 100%. (R 1)

Wrestling with heredity and progeny

The heredity of the disease was an important theme for most of the participants. Most of them were not aware of the diagnosis FSHD or its hereditary nature when they decided to have children. Nevertheless, they often felt guilty about passing on the disease to some of their children and grandchildren.

I keep thinking that this is my fault [that our children have FSHD]. (R21)

Some felt relieved that they did not have to make the choice of whether they wanted to take a risk for their children to have FSHD, as they were not aware of the diagnosis at the time they decided to have children. They felt it would have been a very difficult decision and were glad to have been spared in this regard.

I am just glad I got my daughter before I got diagnosed. Otherwise it would have been a very hard choice. (R 19)

Most of the participants who were already diagnosed before they had children chose to have children in spite of the diagnosis. They felt that they, despite their functional limitations, had a good life and, therefore, did not see FSHD as a reason not to have children.

I recently had a conversation with my daughter about having children. I said to her “if you decide to have an abortion you’re telling me my life is not worth living”. That’s what it would feel like, as if life with FSHD isn’t good enough. But let me stress that life with FSHD is worth living. (R 6)

Still, deciding whether or not to have children was a rational decision for some participants.

My wife would have liked to have two or three children, but we decided to only take the risk once. (R 6)
Changes in the relationship with a partner

Several participants pointed out that having FSHD had a great impact on their relationship with their partner or spouse. In a relationship where one of the partners is increasingly disabled, the patient as well as the partner have to make sacrifices. Activities such as trips, holidays and visits tend to become more difficult, taking things like accessibility and the amount of luggage into account. Some participants experienced this as being a burden on their partners.

I am disabled, but he [the husband] is limited now as well. He has to adjust to my tempo. Rationally I know that I don't have to feel guilty about this, but that takes time. (R 9)

The fact that the participants became increasingly dependent sometimes put pressure on their relationship. Numerous participants pointed out that they were aware of this role pattern and realized that this might influence their relationship in a negative way:

We need to be careful that it does not become a caregiver relationship. (R 18)

Although some participants had these negative feelings, most of them felt they were able to handle the relationship with their partner quite well. As progression in FSHD is relatively slow, they had time to figure out new ways, together with their partner, to handle the increasing muscle weakness, fatigue, and associated functional limitations.

Extra-individual factors

Responses of the social environment to the diagnosis

Participants described the broad impact of the diagnosis on a personal as well as on a social level. On the social level, the diagnosis often led to illness legitimacy. This often implied more understanding by their social environment regarding certain limitations or needs and less negative feelings of the participants such as guilt or shame. Many participants stated that, before the diagnosis, they were diagnosed with FSHD.

...talk to the one who is pushing the wheelchair. (R 6)

Apart from personal legitimacy, participants also experienced more understanding and support from family, friends and colleagues. Several participants described that friends or family members changed their attitudes towards them from the moment they were diagnosed with FSHD.

My dad and granddad always solved problems with their fists; they punched people when they got angry. But I knew I wouldn’t win a fight with someone, so I never let it come to that. He [my father] always thought of me as a wimp. But when I got diagnosed, he finally got it [that I couldn’t fight]. Since then, we have had a much better relationship. (R 6)

On the other hand, participants experienced a lack of understanding, especially from people who were less close. This has to do with the fact that FSHD is not a very familiar well-known disease,

FSHD is not really a big issue to others. Cancer and ALS are much more threatening. (R 18)

Postponing the use of assistive devices

Another aspect of having a muscle disease that was addressed by the participants was the use of assistive devices. In general, participants described it as very difficult to be designated to receive assistive devices. Several participants pointed out that they tried to postpone the use of devices as long as possible. The reason for this postponement was two-fold. Firstly, the use of a device meant the acknowledgement of new limitations. As described before, this decline in physical abilities was hard to accept for most participants, which made it difficult to acknowledge new limitations.

The fact that you need a new device makes you realize that it [the disease] is deteriorating. (R 24)

Secondly, some participants pointed out a more social aspect of the use of devices. They felt people would address them differently when using assistive devices such as a wheelchair or scooter.

I hate to use the wheelchair. People don’t talk to you any more, they only talk to the one who is pushing the wheelchair. (R 6)

The difficulty of (giving up) work

The impact of the disease on daily life was often related by the participants to their ability to work. Some had to make adjustments in their work, others had to change their jobs, and some had to give up work. The adjustments could be small things to help the participants through the day.

You have to be pragmatic sometimes. So the children in the class knew I had trouble lifting and carrying things, so they helped me out. Or I explained to them that I could not write high on the blackboard. (R 19)

Sooner or later, more radical adjustments often had to be made, such as changing job or working fewer hours. These were often difficult decisions. For some participants, it felt like they had to give up their ‘way of life’.

I used to work with clients (…), that was my job, and now I have to quit even though I don’t want to. I will no longer be the person the clients will come to when they have a problem. That does not feel right. (R 11)

Other participants could not adjust their job anymore and had to quit entirely. Most participants found the decision to quit working very difficult and experienced it as a great loss.

I was already weary from the journey when I arrived at work with the whole day ahead. I had to quit working. I had lost the battle against fatigue… (R 13)

Although the decision to quit working was often hard, once the decision was taken, some participants felt relieved.

I used to come home after a long day at work, and then I would quickly eat something before crashing (…). You do that year after year, thinking that’s normal, until you quit working. And then suddenly you get energy for other things, like visiting friends or family. (…) I missed out on those things for a very long time. (R 19)

Most participants started other activities after they stopped working, such as doing some charity work, taking courses at university, and visiting family and friends. The advantage of those activities is that they can plan these themselves, depending on their level of energy.

I thought that it [quitting my job] would be terrible, but it’s wonderful! (…) I don’t have a strict schedule anymore (…) I go to bed later at night, get up later in the morning and take my time to get started for the day, starting with some exercises. (R 21)
Discussion

As can be seen in Figure 1, FSHD affects many aspects of peoples’ lives, which concerns intra- as well as extra-individual aspects. Intra-individual aspects include lifestyle changes people make due to their disease, such as handling fatigue and integrating the functional consequences of the diagnosis into their lives. Besides, aspects of coping were mentioned in reference to the hereditary nature of the disease and to the changing relationship with a partner, which is influenced by the increased dependence. Extra-individual aspects concern the response of the social environment to the diagnosis, the use of assistive devices, and the difficulty of maintaining or giving up work. The results show that the impact of the disease depends on personal circumstances such as one’s familiarity with the diagnosis, the duration and severity of symptoms, and the adjustments people need to make to live with FSHD. People mainly struggle with the continuing progressiveness of the disease and the accompanying increase in disability and use of assistive devices, which have a great impact on their daily lives, e.g. on their ability to work and maintain relationships. The fact that the upper limbs and facial muscles are affected early in FSHD plays a noteworthy role in the experienced illness and disabilities.

Compared to the literature on illness experiences of people with NMDs, our results regarding people with FSHD show some similarities as well as differences. Various themes addressed by people with FSHD are addressed in other NMDs as well. A first aspect mentioned by people with FSHD as well as people with other NMDs are the deteriorating symptoms leading to a decreased quality of life.[19,31,32] Mainly the uncertainty about the future, regarding physical state and abilities, and the heredity of the disease are often mentioned in the literature about NMDs.[13,33,34]

Besides, becoming gradually more dependent was addressed in the context of the spousal relationship and with regard to the use of assistive devices. The continuous behavioral adaptation addressed by the participants of this study is recognized in other progressive diseases as well.[3,4] Research has shown that losing independence during an illness process often influences the illness experiences of people.[35,36] Loss of independence can undermine the equality of the patient in relationships, putting these under strain. The gradual transition to dependency was described as an important theme in a study about illness experiences of people with different types of muscular dystrophy (Becker muscular dystrophy, FSHD, limb-girdle muscular dystrophy, Emery-Dreifuss muscular dystrophy, myotonic dystrophy, and myopathy distalis tarda hereditaria), showing that patients make a great effort to manage their disabilities in order to not become dependent.[19] This effort can be recognized in the present study as well, as patients felt reluctant to start using an assistive device.

Besides becoming dependent, our study showed two additional reasons for patients to postpone the use of assistive devices: firstly, the feeling that an assistive device confirms their physical decline and, secondly, the idea that using an assistive device negatively influences their social standing. This shows that the prescription of assistive devices is not as straightforward as one might think. Some people felt reluctant to use such devices as they felt it changed their position in society. This problematic attitude toward devices is recognized in the literature. People can experience the use of prostheses as enabling as well as disabling; enabling because they are able to participate again in (social) activities with the device; disabling because the device accentuates their dependency.[37]

A lot of themes addressed by our participants were related to the concepts of ‘self-fulfilment’ and ‘identity’, for example, the acceptance of the diagnosis, whether or not to have children, and whether and when to adjust or give up working. Some participants pointed out that it was difficult to accept the diagnosis and integrate it into their lives. This so-called ‘chronic sorrow’ was identified in the study by Nätterlund et al. (2001) as well. They described that the constant loss of physical abilities and necessary adaptations influence one’s identity as a healthy person.[19,38,39]

This process was described by Boström et al. (2004) as an example of ‘loss of self’ as originally put forward by Charmaz.[40] This could explain why our participants described it as ‘very difficult’ having to lose their job due to fatigue or functional limitations caused by FSHD. Loss of work (capacity) can be experienced as a loss of identity or at least of a part of one’s identity. This loss of identity can be a difficult process, for which psychological support or counseling might be useful.

Besides these similarities between FSHD and other NMDs, some differences arose. Firstly, participants did not address care or rehabilitation as an important factor in our study. Usually, this is, according to the Disablment Process Model, one of the main extra-individual factors that influence the physical impairments and, thereby, the disability of people.[13] Other studies showed that care can play a vital role in the lives of people with NMDs, as people gradually become dependent on care as a result of the progressive nature of the NMD.[41,42] However, this often concerns people with rapidly progressive NMDs, such as ALS or DMD, who highly depend on healthcare.[13] For slowly progressive NMDs, care is, just as in our study, less explicitly discussed in the context of illness experiences.[15]

A second difference concerns the impact of the diagnosis. Although many studies show that the diagnosis is of great importance,[15,19,43] our results show a slightly different attitude towards the diagnosis than some other studies. Nätterlund et al described that patients with different types of muscular dystrophy all experienced the diagnosis as ‘somewhat traumatic’. This is, however, contrary to our results, as these showed that the diagnosis can be experienced as a relief by patients as well. This might be the result of the specific circumstances people were living in when receiving the diagnosis. For patients who experience a lot of complaints for a long period of time, the diagnosis is much more likely to be a relief than for patients without many complaints. Besides, the hereditary character of the disease could play a role: for patients with FSHD running in the family, the diagnosis might be less surprising than for people who are the first with (diagnosed) FSHD in the family. All these factors may play a role in the experience of the diagnosis. This shows that a diagnosis can have different meanings.[44] On the one hand, the diagnosis provides exemption from role obligations whereby people no longer feel blamed for their disability. On the other hand, the label can lead to a devaluation of the experience of the body. In that case, the diagnosis can lead to feelings of being overwhelmed and to unpredictability, as patients feel they cannot control their life.[35]

This was experienced by some participants in our study too.

A further difference is related to the nature of the symptoms of FSHD. Although the decreasing independence is mentioned in other NMDs, the nature of the disabilities in FSHD differs from most other NMDs. The fact that in FSHD the upper limbs are often affected first plays an important role in the increased dependence. The results of our study show that people with FSHD feel that their legs can be more easily ‘replaced’ by devices, such as a wheelchair, scooter or wheeled walker, than their arms. The loss of muscle strength in the arms makes people dependent on care from their partner or a professional. This may also apply to other types of muscular dystrophy that affect the upper limbs, such as...
of this study are transferable to similar contexts.[27] Besides, differ-
ences of people with FSHD, which implies that the findings
feel the results are a good reflection of the most important illness
considering the illness experiences in FSHD. However, data collec-
tion could mean that we have not addressed all relevant themes
variety of themes addressed by the participants. At the same time,
open character made it possible to gain information about a wide
openness of our study was a strength as well as a weakness. The
independence and the accompanying changing relationship with
hereditary nature of the disease and adjusting to a decreasing
impact on their illness experience due to the detrimental conse-
quinces for independency and communication. Currently rehabili-
tation is often focused on the physical consequences of the FSHD
with a coordinating role for a physiatrist and provision of addi-
tional support by a physiotherapist. The results of our study show
that, besides physical aspects, psychosocial aspects are important
to address in the rehabilitation care. To address these needs,
patients could profit from support and advice from a social worker,
occupational therapist and/or genetic Counselor as well.

Strengths and limitations
The results of this study should be interpreted with some caution.
Our sample consists of a rather homogeneous group of partici-
pants with respect to cultural background (all were native Dutch
people) and disease burden (all experienced some disease burden
as they were under supervision of a rehabilitation specialist). A
more varied sample might have yielded different results.[46] The
openness of our study was a strength as well as a weakness. The
open character made it possible to gain information about a wide
variety of themes addressed by the participants. At the same time,
this could mean that we have not addressed all relevant themes
considering the illness experiences in FSHD. However, data collect-
ion was stopped only after saturation was reached. Therefore, we
feel the results are a good reflection of the most important illness
experiences of people with FSHD, which implies that the findings
of this study are transferable to similar contexts.[27] Besides, differ-
ent procedures in the analysis, such as contextual description,
have a positive influence on the transferability of the findings
presented in this article. Qualitative research aims for theoretical (ver-
sus statistical) generalization, and there is reason to assume that
the themes and patterns presented here may be applicable to a
broader group of people with FSHD or even to persons with other
types of slowly progressive muscular dystrophy such as limb-girdle
muscular dystrophy and Emery-Dreifuss muscular dystrophy.[20]

Conclusion
The results of this study highlight the illness experiences of people
with FSHD. The findings can be divided into intra- and extra-indi-
vidual aspects. Intra-individual aspects relate to issues that origin-
ate from or operate within the person. In FSHD, integrating the
consequences of the diagnosis and symptoms, such as fatigue, are
examples of intra-individual aspects. Furthermore, coping with the
hereditary nature of the disease and adjusting to a decreasing
independence and the accompanying changing relationship with
one’s partner were found as intra-individual aspects. Besides, extra-
individual aspects also play a role in the illness experiences of
people with FSHD. This concerns factors that operate from outside
the person. In FSHD, the response of the social environment was
mentioned as well as the reluctance to use of assistive devices,
and problems with maintaining work. Although some major
themes, such as the uncertainty about the future regarding phys-
ical state and abilities, and the heredity of the disease were similar
to illness experiences reported by patients with different types of
NMD, other themes were more specific for people with FSHD. This
probably had to do with the specific characteristics of FSHD, such
as the onset in the upper limbs and facial muscles. These are
important factors to take into account in the care for people with
FSHD. Better understanding of the individual illness experiences,
cognitions, and social context of people with FSHD can give health
professionals tools to improve their care and give researchers dir-
rection for future studies to evaluate healthcare improvements
from a holistic, patient-centred perspective.[47]

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Appendix

**Topic guide**

**Opening**
- Introducing ourselves
- Aim of the project
- Aim and estimated duration of the interview
- Consent about audio tape and member check

**Grand tour question**
- Can you tell me something about your diagnosis (what is it, since when)
- Can you tell me something about what it is like to live with FSHD?

**Disabilities**
- What are your most important limitations or disabilities?
  - How do they affect your life?
Do you get help for your disabilities (professionally or non-professionally)?

**Social life**
- Is your social life affected by the FSHD? Could you elaborate on that?
- Can you tell me something about your family situation?
- Did you already know about the FSHD when you got children? Did it play a role? Please explain

**Work**
- Are you employed?
- Can you describe the impact of the FSHD on your (former) job?

**Rounding up**
- Do you have anything to add to the interview?
- Thank you very much for your time