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RESEARCH PAPER



# Perceptions of the transition from receiving the diagnosis recessive limb-girdle muscular dystrophy to becoming in need of human support and using a wheelchair: an interview study

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## ABSTRACT

**Purpose:** To describe perceptions of the transition from receiving the diagnosis recessive limb-girdle muscular dystrophy to becoming in need of human support to manage daily life and using a wheelchair for ambulation, from the affected young adults' and their parents' perspectives.

**Method:** A qualitative and descriptive study design was used. Semi-structured interviews were held with 14 young adults diagnosed with recessive limb-girdle muscular dystrophy and 19 parents. Phenomenography was used for data analysis.

**Results:** The diagnosis was described as being a shock and difficult to comprehend but also as a relief and a tool for information. Beginning to use a wheelchair was perceived to be mentally difficult but it also provided freedom. New ways of living involved physical, emotional, practical, and social difficulties as well as experiences of learning to adapt to the disease. The transition was overshadowed by concern about disease progression and influenced by facilitating factors, which were young adult being seen as a person; supportive family and friends; mobilized internal resources; meaningful daily activities; adapted environment; and professional support.

**Conclusions:** The different perceptions expressed in this study highlight the importance of identifying personal perceptions and needs in order to optimize support provided by healthcare professionals.

## ARTICLE HISTORY

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## KEYWORDS

Disability; LGMD2; parents; phenomenography; transition; young adults

## ► IMPLICATIONS FOR REHABILITATION

- The different perceptions described in this study emphasize the need for person-centered care for young adults living with recessive limb-girdle muscular dystrophy and their parents.
- Regular controls and professional support to cope with the disease and its consequences should be offered, not only at the time of diagnosis but also throughout disease progression.
- Beginning to use a wheelchair can be a psychologically distressing process, which has to be acknowledged by healthcare professionals when introducing it.
- Healthcare professionals should not only recognize the importance of having social relations and activities that are meaningful but also be a link to authorities in society and to interest organizations that can help to facilitate the person's management of the disease.

## Introduction

The health/illness transition involves changes, such as moving from feeling healthy to chronic illness or from chronicity to a new sense of well-being that encompasses the chronicity [1]. People living with limb-girdle muscular dystrophy (LGMD2), which involves progressive muscular weakness predominantly in the shoulder and pelvic girdle [2], may also have to face the transition from being as independent a person as same aged peers to becoming in need of assistive devices and human aid to manage daily life. At present, there is no cure available for LGMD2 and the focus in healthcare must be to support the person to optimize health and well-being. The salutogenic theory introduced by Antonovsky [3] focuses on what causes a person's movements towards health and how people manage to stay well despite difficulties. Central in this theory is how a person comprehends,

manages, and finds meaning in everyday life, i.e., the person's sense of coherence. In previous interview studies, based on the salutogenic theory, experiences of living with LGMD2 from the affected young adults' [4] and their parents' [5] perspectives have been described as well as health perceptions of the young adults [6]. Variation was found in the young adults' need of human aid and assistive devices to manage daily life. This article focuses on how the transition from receiving the diagnosis LGMD2 to becoming in need of human support to manage daily life and using a wheelchair for ambulation is perceived by the affected young adults and their parents. This transition may be experienced in various ways and have different meanings for different people. Moreover, people make sense and act in relation to a situation as they perceive it [7]. This means that there are various ways of

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experiencing, influencing people's perceptions, and make them able to manage situations in more or less efficient ways.

LGMD2 refers to a genetically and clinically heterogeneous group of muscular dystrophies that may manifest in physical disability [8]. The age of the first symptoms varies from early childhood to adulthood. Progression of the disease also varies and may not be linear. In this study, the young adults and their parents also had to face the young adults' transition to adulthood. This transition has previously been found to be a challenging time for young men living with Duchenne muscular dystrophy (DMD) and their families [9]. The change from being independent to becoming in need of support from other persons and assistive devices to manage daily activities has previously been described as a psychologically trying process among adults diagnosed with various forms of muscular dystrophy (MD) [10]. Experiences of progressive functional decline that influenced identity, social status, and social relationships have also been shown in the transition from receiving a diagnosis and living with motor neurone disease [11]. Moreover, integrating a wheelchair has been described from the users' perspectives as a manifold process which involved practical, personal, and social dimensions [12]. Constantly coping with a progressive disease thus poses existential and organizational difficulties for the person, and the threat of permanent dependency may strip away earlier self-definitions based on independence and may also influence the way the person is viewed by others [13].

The situation for young adults living with LGMD2 and their parents is complex considering the progressivity of the disease. In order to optimize the support provided, healthcare professionals need to acknowledge different ways in which the transition, from receiving the diagnosis to becoming in need of support from another person and using a wheelchair, is perceived, but no study focusing on this has been found in the literature review.

The purpose of this study was to describe perceptions of the transition from receiving the diagnosis LGMD2 to becoming in need of human support to manage daily life and using a wheelchair for ambulation, from the affected young adults' and their parents' perspectives.

## Methods

This study has a qualitative descriptive design. Data were collected through semi-structured interviews to enable the participants to express themselves within a given frame [14] and provide an opportunity for the interviewer to acquire knowledge about the participants' personal experiences and perceptions [7].

### Participants

Purposive sampling was used [14]. Young adults aged 18–30 years and diagnosed with LGMD2 were invited to participate in the study through an information letter distributed by healthcare professionals and key members of the Swedish Association of Persons with Neurological Disabilities, Neuro Sweden. The information letter was also sent through email by the principle investigator (first author) to possible participants who were members of a web-based association for persons with disability and their next of kin. The young adults who agreed to participate forwarded a letter with information about the study to their parents and at least one parent from each family agreed to take part in the study. For further details, see previous publications [4,5].

Fourteen young adults, from 13 different families, and 19 of their parents agreed to participate in this study (Table 1). The young adults were recruited through healthcare ( $n=10$ ), from Neuro Sweden ( $n=1$ ) and from the web-based association for

Table 1. Characteristics of the participants.

Participants	Number ( $N=33$ )
Young adults	14
Females	8
Males	6
Median age (range)	25 (20–30)
Parents	19
Females	13
Males	6
Median age (range)	50 (47–65)

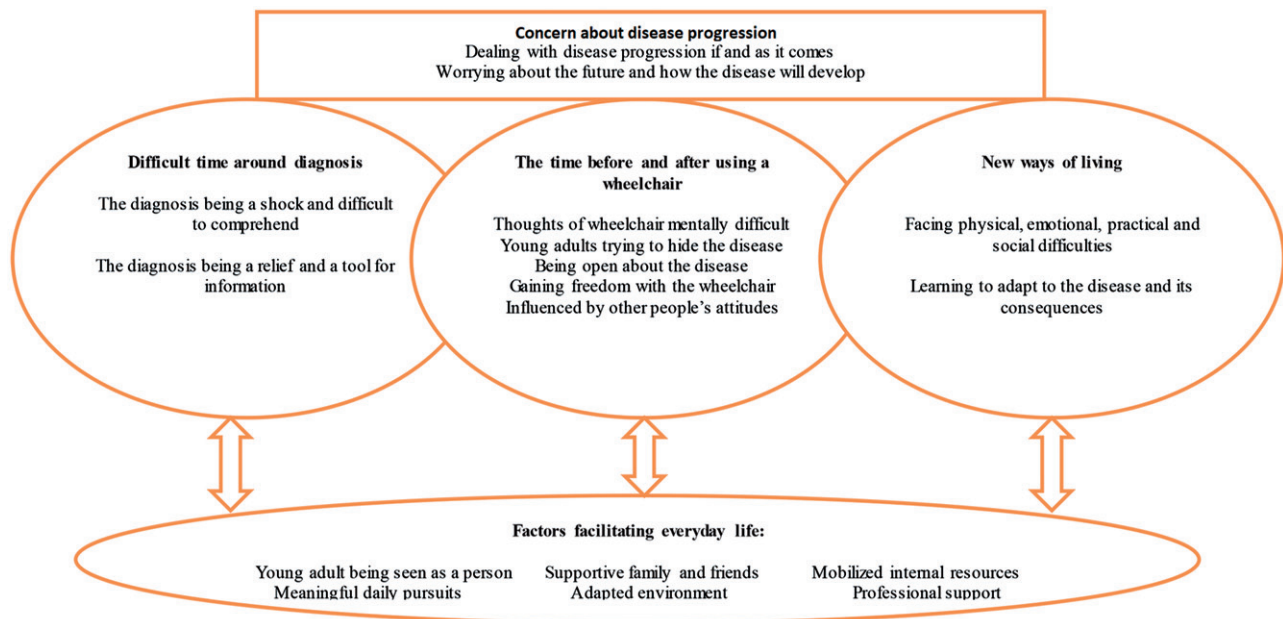
persons with disability ( $n=3$ ). The first symptoms of the disease had varied among the young adults, and the median age when symptoms first appeared was 11 years (range 0.5–25). There was also variation regarding the young adults' form of LGMD2 and disease progression. Two of the young adults managed daily life on their own. Five of them received informal support (e.g., help with clothing, shopping, cooking, and cleaning) from parents and/or other next of kin, had limited walking ability and some of them sometimes used a wheelchair. Seven of the young adults had personal assistance and always used a wheelchair for ambulation. Some of the participants lived in the countryside, while others lived in urban areas. The participants thereby represented a range of different experiences, which is important in order to capture various perceptions within a group of people [7].

### Interviews

The interviews were conducted by a nurse (first author) between June 2012 and November 2013. Based on the salutogenic theory and the content of the sense of coherence questionnaire developed by Antonovsky [3], an interview guide with semi-structured interview questions was formulated to capture the participants' comprehensibility, manageability, and meaningfulness when living with LGMD2 or being a parent of an affected person. The main interview questions focusing on the participants' comprehensibility were: Could you describe what it means to live with LGMD2/to be a parent of a young adult living with LGMD2? What did it mean to you when you/your son or daughter received the diagnosis? Interview questions focusing on the participants' perceptions of manageability were: Is there any support that you would need that you do not have access to/and that your son or daughter would need that he or she does not have access to? If so, could you describe what it is? Finally, the participants' perceptions of meaningfulness were focused on by asking: What do you experience as most meaningful in life? The interview guide was tested in the three first interviews with the young adults and with the parents (included in the study) and no changes were made. All the interviews were held at a place chosen by the participants, mainly at their homes. The young adults were interviewed individually whereas the parents were interviewed individually or in parental couples. A total of 27 interviews were thereby conducted and each interview lasted about an hour. During the interviews, different statements about the transition, from receiving the diagnosis LGMD2 to the young adult becoming in need of human support and using a wheelchair, frequently and spontaneously turned up. In this study, the participants' various perceptions of this transition are in focus. Follow-up questions to reach a deeper understanding of the participants' perceptions were asked, e.g., How do you mean? and What do you think about that? All the interviews were recorded and transcribed verbatim by the first author.

### Data analysis

Phenomenography was used for data analysis as it focuses on different ways in which people perceive something [15]. The seven



**Figure 1.** The outcome space: five descriptive categories illustrating perceptions of the transition from receiving the diagnosis recessive limb-girdle muscular dystrophy to becoming in need of human support to manage daily life and using a wheelchair for ambulation, from the affected young adults' and their parents' perspectives. In each descriptive category, various perceptions expressed by the participants are shown.

steps for analysis described by Sjöström and Dahlgren [16] were followed. First, the transcribed interview data were read through to become familiar with the text. Second, statements from each participant, regarding perceptions of the transition, from receiving the diagnosis LGMD2 to the young adult becoming in need of human support and using a wheelchair, were identified and compiled. Third, the statements were condensed to find the central parts of the text. Fourth, a preliminary grouping of similar statements was made. Fifth, the statements were compared to find variation in the participants' perceptions and through revision of the preliminary groups; descriptive categories distinct from each other were established in a dynamic process, working back and forth between the interview text, the preliminary groups and the descriptive categories. Sixth, the descriptive categories were named close to the text to illustrate their content. Seventh, a contrastive comparison of the descriptive categories was conducted, where the unique character of each descriptive category as well as the relationships between the descriptive categories were illustrated in the findings [16] as the outcome space (Figure 1) [7].

Data analysis was conducted close to the interview text to ensure the agreement between the participants' perceptions and the investigators' presentation of them [14]. The two coauthors, who are experienced in phenomenographic research, checked the content of the preliminary groupings and the descriptive categories throughout the analysis process to ensure study credibility [16]. Continuous discussions were held until coder agreement was achieved. The relevance of the descriptive categories is supported by illuminative quotations to establish the confirmability of the study [16]. Moreover, data triangulation was obtained by using a variety of data sources, i.e., including the young adults' as well as their parent's perceptions in the study [14].

#### **Ethical approval**

The study was approved by a regional ethics board and conducted in accordance with the Declaration of Helsinki [17]. All the participants gave their written informed consent to take part in the study.

#### **Results**

Different perceptions of the transition, from receiving the diagnosis LGMD2 to the young adult becoming in need of other people and using a wheelchair, are illustrated in the outcome space (Figure 1). The descriptive categories – difficult time around the diagnosis, the time before and after using a wheelchair, and new ways of living – have a horizontal course and can be viewed as a process that is overshadowed by concern about disease progression and influenced by factors facilitating everyday life.

#### **Difficult time around diagnosis**

The years before and after receiving the diagnosis were perceived to be a difficult period of time for several reasons, and feelings of having been depressed were described.

#### **The diagnosis being a shock and difficult to comprehend**

Some of the participants perceived that the diagnosis had come as a shock and that the meaning of the diagnosis had been difficult to comprehend when it was received. Sometimes the parents have had to seek emergency healthcare for their young children's symptoms. Other times, the symptoms had been vague and difficult to pinpoint, but having had a sense that "there was something wrong" was described. Before physical deterioration became obvious to others, some of the young adults remembered increased difficulties in performing physical activities and keeping up with peers, without knowing the reason for it. Efforts to suppress or explain the first symptoms were described and some of them had tried to exercise harder. Parents recalled how they had tried to encourage their children and sometimes had thought that they were lazy. These were memories that caused remorse among the parents. Eventually, seeking healthcare and realizing that it is a progressive disease with no cure currently available was perceived to be mentally difficult.

it (the diagnosis) came as a shock...the first two years were really difficult and then after that you began to get a bit more used to it. (Young adult in need of informal support, family 3)

Some of the participants thought that receiving the diagnosis was so overwhelming that they had not been able to take in all the information given by healthcare professionals at the time. Others felt that they had not been offered sufficient information or professional support to cope with the disease at the time of diagnosis. Young adults mentioned, for instance, how during that period they had slept most of the time, neglected school, and tried to suppress their thoughts about the disease. It was also perceived as difficult to inform other people about something that one did not understand oneself. For the parents, although being tough, some of them described how they had focused on managing everyday life and had found their energy in that life went on. Some of the young adults had siblings who had been diagnosed first and then they themselves were tested as well. Therefore, some of them had not experienced any symptoms at the time, and they described difficulties coping with and comprehending what it means to have a muscular disease.

Then my revolt started. That's when I started smoking, hanging out with the wrong people... I felt really bad, had nobody to talk to... I shut myself in a lot because I couldn't cope, for I knew somehow that I was different from my friends but not why or how... if you just find out that you have a muscle disease you don't know what it means...nobody explained that to me. (Young adult entitled to personal assistance, family 1)

#### ***The diagnosis being a relief and a tool for information***

Although mentally difficult, receiving the diagnosis felt like a relief and a tool for information for some of the participants. It was a way to make sense of the symptoms and something to relate to and cope with. It also meant contact with healthcare professionals with knowledge about neuromuscular diseases and regular controls at the hospital. The participants realized that it is a difficult disease to diagnose, that it has to be searched for in order to be found. Some of the young adults who had symptoms of the muscular disease as small children did not receive the diagnosis until they were teenagers. Not having known the cause of the symptoms had meant sometimes not receiving appropriate support and also not being able to inform other people about the diagnosis. Young adults' perceptions of having been bullied in school because of their symptoms were described and their parents had found it difficult to know how to support their children prior to the diagnosis. Comprehending that there was something wrong, the thoughts and feelings connected to the uncertainty were described as mentally difficult, and having waited for an answer, the diagnosis was perceived to facilitate coping.

it was a relief even though it was heavy, but it was so troublesome mentally before, and we understood that X was in really bad mental health... after X got the diagnosis he has now accepted it and that has made it easier for us too. (Mother of young adult living an independent life, family 2)

The diagnosis was also used when searching for information and informing people about the disease. Those who were younger when the first symptoms appeared felt that they had "always" lived with the disease. Being informed as a child that they had a muscular disease did not mean much for them at the time, and the perception was that it had been more important for their parents to get a diagnosis of their disease. For some of the participants, who already knew that they (or their children) had a muscular disease, receiving the exact diagnosis based on genetics did not mean much. Others thought that it was important to know considering future progress in research and whether a cure would be found. Having a diagnosis was also perceived to make it easier for other people to understand the disease and its consequences.

For me it didn't mean so much, but I noticed in people around me that their expectations or demands on me changed when they got a name for the diagnosis. They understood and were able to handle it and respond to it in a different way... I have always used it (the diagnosis) as a tool for information. (Young adult entitled to personal assistance, family 9)

#### ***The time before and after using a wheelchair***

The young adults' transition from walking to beginning to use a wheelchair was perceived to be a psychologically distressing process that involved major emotional, social, and practical adjustments.

#### ***Thoughts of wheelchair mentally difficult***

The wheelchair somehow symbolized physical deterioration and the seriousness of the disease and there was sadness over deterioration in walking ability. Being able to walk, some of the young adults said that they had not thought much about eventually having to use a wheelchair, while others had feared that since the day they received the diagnosis. Being informed that a wheelchair might be needed in the future was described as mentally difficult and how the information was given was perceived to influence management. One young adult referred to it as the "doomsday talk." Some of the young adults thought that they wanted to walk as long as possible, realizing that they would have to spend the rest of their time in a wheelchair. Fear of becoming worse if using a wheelchair was also described. Beginning to use a wheelchair was also perceived to influence how the young adults viewed themselves and concern about how people in society would view them was also expressed.

It's a really big step for me to handle a wheelchair... then I think, 'maybe I'm not the same person' ... I have this huge fear... 'how will people look at me if I have to use a wheelchair?' (Young adult in need of informal support, family 3)

#### ***Young adults trying to hide the disease***

As the disease progressed, some of the young adults described how they had tried not only to avoid the wheelchair but also to hide their disease from other people as long as possible. Feelings of being ashamed and embarrassed over physical limitations were expressed. If questions turned up, it was sometimes found easier to say that it was an injury or back pain than to tell about the disease. Activities that revealed physical limitations, such as rising from a chair or using stairs, were perceived as mentally difficult to perform among other people. Because of fear of exposing oneself to the glances of other people and the risk of falling and not being able to get up unaided, the young adults avoided doing things; for instance, some of them had not been in a store for several years. Bitterness was also expressed about missing activities that would have been possible if a wheelchair had been used earlier. If a wheelchair was used, it would be at places where there was low risk of being recognized, or they made efforts to conceal themselves.

I wouldn't be prepared to sit in it (the wheelchair) in the village... I didn't want people to know about it (the disease)... I suppose it's mostly because I feel shame... so I hid a lot. (Young adult in need of informal support, family 4)

Parents who thought that it would be easier for the young adults to tell about the disease and accept help found it difficult to see their young adult's struggle. Although having an

understanding of the young adult not wanting to bring the wheelchair everywhere when being able to walk, the parents also expressed worries that the young adults would fall or be left behind when going out with friends. The parents were also influenced by the young adult not wanting to tell about the disease.

he struggles an awful lot ...he doesn't want to show me or anybody ... not getting up... not showing people how he walks...it's terribly difficult (Mother providing informal support, family 3)

### ***Being open about the disease***

As the disease progressed, some of the young adults chose to be open about their disease, and parents described how they tried to encourage them to tell people about how it is. A wheelchair was sometimes used to save energy and to be able to take part in different activities. The perception was that it is easier if people know about the disease and there was an understanding that people may wonder what is wrong as physical limitations became obvious. When coming into a new context, some of the young adults preferred to gather and inform everybody at the same time. Others conveyed information about their disease when the right moment turned up or waited for people to ask. The importance of not being ashamed of oneself was emphasized.

You should never be ashamed of what you are ... I have never hidden my disease... I have been open about it...that I sit in a wheelchair ...it's nothing I'm ashamed of...no...feeling shame doesn't make it better, for then you'd never go out ... (Young adult in need of informal support, family 10)

### ***Gaining freedom with the wheelchair***

Although mentally difficult, beginning to use a wheelchair for ambulation was perceived as gaining freedom. As walking ability deteriorated, the young adults described their choice, either to struggle to walk as long as possible or beginning to use a wheelchair. There was also a choice whether or not to tell about the disease. Eventually, reaching the point when walking ability had vanished, sometimes because of a fall accident, there was no choice but to use a wheelchair, and physical impairment became obvious to other people. A manual wheelchair usually had to be pushed by another person because of arm weakness, but an electric wheelchair made independent ambulation and activities possible.

When I stopped being able to walk it was really difficult but at the same time it was so nice because then I had no choice... when everyone else thought, 'now she's really disabled', then I had greater freedom than I'd had since I was small... (Young adult entitled to personal assistance, family 7)

### ***Influenced by other people's attitudes***

As physical impairments became obvious to other people, the participants perceived that they were influenced by other people's attitudes towards them. Some of the participants expressed concern while others tried not to bother about it. In the time before using a wheelchair, the young adults thought that they were generally treated well by people in society but that people sometimes would stare at them. If they fell in public places, feelings of embarrassment and fear that people would think they were drunk were described. The parents were also influenced by their young adult's reactions, as described by one mother.

People look, make comments and think that you can't hear... I have to control myself... I get so angry...although it's quite nice because I

notice now that she... she's got used to it somehow or chooses not to see ... (Mother providing informal support, family 11)

The participants thought that people's attitudes in society towards persons using a wheelchair had become better over the years. Thus, smiling and helpful people were described. The wheelchair was, however, perceived to primarily symbolize disability for other people. It was described as difficult when people felt sorry for them because it was perceived as reducing them and their situation. Feelings of being stigmatized were also expressed. Some of the young adults also perceived that they were treated differently by some people when they began to use a wheelchair; for instance, being ignored or talked to in a childish way or being viewed as asexual and innocent. Situations when having felt discriminated were also mentioned, such as being told that the wheelchair takes too much space on the dance floor when going out with friends. The young adults perceived that people sometimes seemed afraid of talking to them. Situations when people talked above their head were also described, for instance, when being in a shop and the staff instead talked to the person who was with them. This was thought to be frustrating and sad. The young adults expressed various perceptions of why people acted in that way, for instance, that people: may think that they have cognitive disability and/or are not able to communicate; may be uncertain of how to act and afraid of doing wrong; may be ignorant; or may perceive that it is easier to talk with people at the same eye level. Perceptions were thus expressed that in society there are underlying views of people living with disability.

If there's somebody with a disability in the newspaper then it's either a great pity of that person...or...it's to show how strong the person is...even though she has these problems... there are a lot of stereotypes and it doesn't help anybody in making each other more human. (Young adult entitled to personal assistance, family 12)

### ***New ways of living***

As the disease progressed, the young adults as well as their parents had to find new ways of living that involved increased need of help from other people to manage daily life and the young adult always using a wheelchair for ambulation.

### ***Facing physical, emotional, practical, and social difficulties***

Disease progression was perceived to cause physical, emotional, practical, and social difficulties. It was therefore described as difficult to do anything spontaneously and every activity had to be planned. Frustration over everyday dependency on other people was mentioned by the young adults. Although resignation about vanished walking ability was expressed, new concerns had turned up that were perceived as mentally difficult to cope with, for instance, increased arm weakness. One of the young adults also had severe cardiac complications with reduced life expectancy. Reduced physical abilities and fatigue made it difficult to perform various activities. In addition, inaccessibility in society, for instance caused by stairs, made it difficult not only to enter certain public buildings, such as shops and restaurants, but also to visit friends' homes. Having an adapted bathroom at home, difficulties were encountered using other bathrooms. Therefore, some of the young adults described how they sometimes reduced fluid intake to avoid having to use the bathroom when being away from home. This was also something that the parents felt concerned about. Moreover, the wheelchair was perceived to sometimes be a barrier that made it difficult for people to approach the young adults and to view them as persons with other interests and

characteristics than their disease. Altogether, feelings of becoming more restricted from participating in various activities were expressed, which also influenced the parents.

when they were teenagers they hung around with (friends) doing a lot of things they really weren't able to manage... I suppose it worked out somehow... now X gets pains and it's so difficult and there have to be two assistants... it's sad that it's not worth the cost any more... taking part in things is bothersome now... (Mother of young adults entitled to personal assistance, family 9)

### **Learning to adapt to the disease and its consequences**

Because disease progression most often was perceived to be slow, a sense of having learned to cope and to live with the disease was expressed. Human aid in the shape of personal assistance and the young adults moving out of their parents' home was perceived to bring independence and facilitate management, not only for the young adults but also for the parents. Having become reconciled to the wheelchair, it was viewed as a great assistive device that enabled the young adult to live an as fulfilled life as possible. It was also found easier than before to tell others about the disease because now, "this is how it is" and there was no choice of trying to hide it anymore. Some of the young adults also perceived that they had become more mature through the years and they did not bother so much about how people in society would view them any longer. Instead, a sense of "this is how I am" was described and that people had to accept that or it was their loss. A sense of having to accept the disease and its consequences was thus described.

Since it's getting worse so slowly you have time to come to terms with it... I have learned now... I've begun to accept that I'm sick, that I have to reconcile myself with the idea that I'll get worse and worse... if you fight against it, everything just gets harder. (Young adult entitled to personal assistance, family 1)

### **Concern about disease progression**

Regardless of the young adult's physical ability and need of assistive devices, the participants expressed uncertainty and concern about how the disease would progress.

### **Dealing with disease progression if and as it comes**

As long as the disease did not influence daily life and everything was going well, it was perceived as difficult to know what to expect. Although being concerned, there was a sense of having to live in the present and to deal with disease progression if and as it comes.

to know that the children have muscular dystrophy... it's terrible when you think about it... it makes me a bit blinkered... that 'we'll take it when it comes'... it's terrible if... when that day comes... but we're not there yet at least. (Mother of young adults living independent lives, family 5)

### **Worrying about the future and how the disease will develop**

As the disease progressed, continuous adjustments to the young adults' physical deterioration had to be made which slowly revealed the complexity of the disease, and worries were expressed about further disease progression.

Although I knew about my diagnosis I didn't know that it would be as hard as this to get up from the sofa... you often think, 'how much worse... how bad can it get?'... you get really worried. (Young adult in need of informal support, family 3)

## **Factors facilitating everyday life**

Factors perceived to facilitate management in everyday life were: the young adult being seen as a person; supportive family and friends; mobilized internal resources; meaningful daily pursuits; adapted environment; and professional support.

### **Young adult being seen as a person**

The young adult being viewed as a person by other people, rather than being considered as sick and disabled, was perceived to facilitate management. Apart from having physical limitations, the young adults in general considered themselves to be like anybody else. Therefore, the importance of being seen as a person, not only by people in their immediate surroundings but also by people in the society and by professionals, was highlighted.

It's much more fun when people see the person in me... not the wheelchair... (Young adult entitled to personal assistance, family 13)

### **Supportive family and friends**

The participants described the importance of having a supportive family and friends to share thoughts and feelings with and to strengthen each other. It was expressed as a sense of being confident that no matter how the disease would develop, there would be people around who would do their best to support.

They help me and they are there on the way... there's nothing they wouldn't do for me, to make things easier for me or help me, and that's really nice... my family is absolutely the most important thing for me. (Young adult entitled to personal assistance, family 1)

### **Mobilized internal resources**

Trying to make the best of the situation, the value of being able to mobilize internal resources was described. It was therefore perceived as important to focus on positive thoughts and things that are essential in life and to have joyful matters to look forward to. The need to sometimes be alone to find the energy to cope with the disease within oneself was also expressed. Several of the participants felt that the disease had brought new perspectives to their lives and that they had discovered that it is possible to manage more than first thought.

However many restrictions came along, you adapt... that's a perspective I didn't have before... you can manage more than you think... despite the obstacles... you just carry on regardless... I have better self-confidence again... in the past I thought, 'what a bother it is' and I felt awful sad... the more time passes, the more I think, 'yet I manage awesomely well and I have succeeded in doing all these things even though I have a disease.' (Young adult in need of informal support, family 8)

### **Meaningful daily pursuits**

The participants thought that it was important to have daily pursuits that are meaningful and to have social interaction with other people. Among the parents, feelings of being strengthened by the young adult's disease to deal with their own working situation were described.

you dare to do things... 'what is important in life?'... I can't go to a job where I'm not happy... and so I have to do something about it... you deal with it in a different way. (Mother of young adult entitled to personal assistance, family 6)

### **Adapted environment**

As the disease progressed, the importance of the environment being adapted was described, which also involved the need to have various assistive devices that could facilitate everyday management. This included not only adaptations in the home and the car but also adaptations necessary in order for the young adults being able, for instance, to attend an education, work, participate in leisure activities and have access to public places. When going to unknown places, the importance of being proactive and planning ahead was described.

Think ahead... plan... there's a lot of things like 'why didn't I think about that step being there?' but I have portable ramps that I can take along. (Young adult entitled to personal assistance, family 6)

### **Professional support**

Entitlement to personal assistance when needed, in order to become independent with help from another person, and support from concerned professionals was perceived to facilitate management. Professional support comprised not only a multidisciplinary healthcare team and staff working in different municipal or community agencies but also employees when working and teachers when attending education. Just knowing where and who to turn to regarding different issues was perceived to provide a sense of security, and the importance of being listened to was emphasized.

it was a really good doctor we met the last time...he listened and understood what we said and then he gave very good explanations. (Mother of young adult entitled to personal assistance, family 13)

### **Summary of the findings**

Receiving the diagnosis LGMD2 was not only described as being a shock and difficult to understand but also as being a relief and a tool for searching and/or providing information about the disease. Some of the young adults were consequently open about the disease, whereas others tried to hide it as long as possible. Although the wheelchair was perceived as providing a new freedom, the transition from walking to becoming in need of using a wheelchair was described as a psychologically distressing process. Perceptions of being influenced by other people's attitudes were also expressed. As the disease progressed, new ways of living had to be found that involved personal assistance and the young adult always using a wheelchair for ambulation. Disease progression was perceived to cause physical, emotional, practical, and social difficulties, but a sense of having learned to live with the disease and its consequences was also described. Concern about how the disease would develop was expressed regardless of the young adult's physical ability and need of support. Factors perceived to facilitate management were: the young adult being viewed as a person, not only by people in the immediate surroundings but also by people in society and by professionals; having supportive family and friends; being able to mobilize internal resources, such as trying to think positively; having daily activities that are meaningful; the environment being adapted: and receiving support from concerned professionals, including entitlement to personal assistance when needed.

### **Discussion**

This study is unique as it describes perceptions of the transition from receiving the diagnosis LGMD2 to becoming in need of human support and using a wheelchair, from the affected young

adults' and their parents' perspectives. The findings show that the time around diagnosis was perceived to be difficult, although receiving the diagnosis was thought to be not only a shock and difficult to comprehend but also a relief and a tool for information. Beginning to use a wheelchair was thought to be mentally difficult but also a way to gain freedom. New ways of living were perceived to involve physical, emotional, practical, and social difficulties, but a sense of learning to adapt to the disease and its consequences was also described. The transition was overshadowed by the concern about how the disease will develop and influenced by factors perceived as facilitating management in everyday life.

Some of the participants in this study found that the diagnosis had come as a shock and was difficult to comprehend. Feelings of shock and efforts to make sense of the diagnosis of a progressive disorder among adults have been described before [11] and parents of boys diagnosed with DMD have been shown to experience the greatest emotional impact in the first years following diagnosis and around the time of loss of ambulation [18]. In order to experience a sense of coherence according to the salutogenic theory [3], a person needs not only to comprehend his or her life and be understood by others but also to perceive that resources to cope are available and be able to feel meaningfulness in daily life. Receiving a diagnosis of a progressive disease and the chain of events that the message induces or symbolizes may influence the person's and their next of kin's sense of coherence. It can be viewed as a crisis that the person needs to cope with. Cullberg [19] has described four different stages that a person has to go through when facing a crisis: the shock; the reaction; the processing; and the reorientation phase. These stages, however, are not distinctly separated from each other, just as disease progression and management of life situation for the affected persons and their parents in this study may vary over time. During the shock phase, it can be difficult for the person to take in information, as described in this study. This highlights the importance of repeated information, orally and in writing, and follow-up meetings with persons newly diagnosed with LGMD2 and their parents. Considering the progressivity of LGMD2, they should also be offered professional support to cope with the disease, not only at the time of diagnosis but also throughout the disease progression, in order to facilitate management. It could be needed, for instance, when complications turn up or walking ability vanishes. Previously, communication in medical consultations at the time of a cancer diagnosis has been found to be dominated by information-giving although the affected adolescents or young adults and their relatives often expressed emotional concerns [20]. Among family carers, emotional stress engendered by poor communication when receiving the news of a diagnosis of a progressive disease has been shown, but a majority of the participants were satisfied because they had longer consultation time, the neurologist was empathetic, the information and support provided was perceived to be relevant, and there was a plan for following up support [21]. It is therefore important, not only how the diagnosis but also how information about support available for future management of the disease is communicated by healthcare professionals. In the present study, some of the young adults did not receive the diagnosis until several years after the first symptoms of the disease appeared. The diagnosis was perceived to be a relief as it helped to make sense of the symptoms and meant contact with healthcare professionals with knowledge about neuromuscular diseases. LGMD2 has previously been diagnosed by exclusion [22] and it was not until 1995 that more precise criteria for the diagnosis and the classification of different forms of LGMD2 based on genetics were established [23]. This means that

some of the young adults in this study had their first symptoms before techniques to establish a precise diagnosis were available. Today, over 20 different forms of LGMD2 have been identified and a precise genetic diagnosis is essential as it allows more accurate follow-up controls, genetic counseling, and the prevention of known possible complications [24].

In this study, the young adults' transition from being able to walk to becoming in need of using a wheelchair was perceived to be a psychologically distressing process. Each person has a sense of themselves from which they perceive the world around them and their bodies [25]. Physical deterioration may force the person to reflect and reevaluate the perception of self. Chronic disease thus brings tension between body, self, and identity, followed by increased awareness of how one is viewed by other people [13]. According to Arendt [26], the human physical body appears in the unique shape of the body and the sound of the voice, but it is in acting and speaking that persons show who they are and their personal identity is revealed. On the other hand, "what" somebody is, such as characteristics, talents, and shortcomings, may either be disclosed or hidden. As seen in this study, some of the young adults were open and told people about the disease when establishing a new context, while others tried to hide it as long as possible by avoiding activities where muscular weakness would become obvious to others. Anxiety and shame connected to limited walking ability as well as uncertainty about how people will judge one's appearance has previously been described among adults living with MD [10]. Worry and anxiety when beginning to use a wheelchair have also been shown among persons living with DMD [27]. According to Charmaz [13], people living with chronic diseases may limit their activity and autonomy rather than affirm further physical deterioration that would threaten their self-perception. The process of resistance and acceptance of using a wheelchair is thereby related to an on-going process of changing self-identity [12]. In this study, some of the young adults perceived that they were sometimes treated differently by people in society when they began to use a wheelchair, i.e., initially being viewed for "what" they are (a person with disability) rather than "who" they are (a unique person). Awareness of the varying reactions of others to the wheelchair and fear of not being regarded as the person they are has previously been described [12]. How a person is regarded by others involves both the self that a person intends to project through speech and actions, and also the self that others interpret from the person's speech and actions [25]. In this study, some of the young adults with limited ability to perform physical actions, such as walking, perceived that sometimes people initially viewed them as not being able to communicate either. Interaction with other people thus means not only being an active person but also being the object of the actions of others [26]. Healthcare professionals therefore need not only to proactively anticipate and facilitate affected persons and their families in decision making regarding loss of mobility and beginning to use a wheelchair [28] but also need to acknowledge the complexity of the transition from being able to walk to becoming in need of using a wheelchair for ambulation. Thus, the importance of enabling the person to have open discussions with relatives, friends, and professionals involved in management of the disease is emphasized. Moreover, information about different interest organizations that can provide opportunities for social gatherings and sharing of experiences should be provided by healthcare professionals [29].

Disease progression was perceived to cause physical, emotional, practical, and social difficulties for the participants in this study. According to the International Classification of Functioning, Disability and Health [30], a person's functioning should be

regarded as a dynamic interaction between health conditions and contextual factors. From an adult user's perspective, challenges driving an electric wheelchair in the community and difficulties accessing public buildings have previously been described [31]. Social participation for people using a wheelchair is thereby related to the physical, social, and attitudinal environment, which should be taken into account by urban planners and service providers [32]. Furthermore, the importance of the young adults becoming as independent as possible in their transition to adulthood must be emphasized [33]. Some of the participants in this study perceived that they had learned to cope with the disease and had become reconciled to the wheelchair. Previously, adult persons living with progressive physical deterioration have been found to experience better psychological adaptation over time [10] and employ adaptive strategies to cope with their disease [11]. Experiences of having incorporated the wheelchair as part of the body have also been shown [12]. Acceptance of a chronic disease, however, may be a difficult ongoing process. Moreover, acceptance and denial appear intertwined in a way that can be adaptive for the person [34]. Experiences of having to take each day as it comes and addressing functional problems as they arise, but also difficulty coping with uncertainty about the future has also been described [11]. In this study, internal and external factors perceived to facilitate management were expressed. All resources, within the person or the environment, that facilitate stress management and help the person to cope with the situation, can be viewed as generalized resistance resources according to the salutogenic theory [3]. These resources need to be identified and encouraged, in dialog between the person and healthcare professionals, in order to promote the person's health.

When using a qualitative method, the findings are contextual and cannot be generalized [14]. The findings may, however, be transferable to similar groups. In this study, the interview data is part of longer interviews with young adults living with LGMD2 [4] and their parents [5]. Despite the richness of data, this can be considered as a limitation. The participants' awareness of the interviewer being a nurse may also have influenced their answers. Moreover, the findings in this study are partly based on the participants' retrospective memories, and there is a risk of recall bias [35], which means discrepancies between memory and experiences [36] that may have influenced the findings. By including both the young adults and their parents, however, data triangulation was obtained in the present study, showing that the young adults' and their parents' perceptions were compatible [14]. It can be questioned whether or not the young adults and their parents represent two different perspectives of the young adults' transition. However, considering that they had coped with the young adult's disease together for several years as a family, including both the young adults and their parents enriched the data as they represented a range of experiences and expressed various perceptions of the young adults' transition. The strength of phenomenographic studies is that they emphasize that people vary in their experiences and how they think about themselves and the surrounding world [16]. In the context of healthcare, the care has to be based on considerations of the vast differences between people, which excludes routine-based dialogs with and treatment of recipients of care. This highlights the need for person-centered care that focuses on the person's own perceptions of living with the disease, which can be expressed through the persons' and their parents' narratives about their life situation [37].

To conclude: the different perceptions expressed in this study emphasize the need for person-centered care. Healthcare professionals should initiate the affected persons' and their parents' narratives about their situation. Through dialog, not only support

needed to manage the disease but also factors that facilitate everyday life and bring meaningfulness to the person can be identified and encouraged, taking into account the social context in which the person lives as well as medical aspects. Information provided to comprehend the disease and its consequences should be individualized and repeated. Professional support to cope should be offered, not only at the time of diagnosis, when going through the stages of a crisis, but also later on when having to manage disease progression. Beginning to use a wheelchair can be a psychologically distressing process that is influenced not only by sadness about deteriorating walking ability and the young adult's self-perception but also by other people's attitudes, which have to be acknowledged by healthcare professionals when introducing the wheelchair. Healthcare professionals should also be a link, not only to other authorities in society that provide services to persons with physical disability but also to interest organizations where the young adults and their parents can have the opportunity to meet families in similar situations.

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