

## RESEARCH PAPER

# Young adults' experiences of living with recessive limb-girdle muscular dystrophy from a salutogenic orientation: an interview study

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### Abstract

**Purpose:** To describe young adults' experiences of living with recessive limb-girdle muscular dystrophy (LGMD2) from a salutogenic orientation. **Methods:** A qualitative explorative interview study, including 14 participants aged 20–30 years, was performed focusing on comprehensibility, manageability and meaningfulness in daily life. Content analysis was used for data analysis. **Result:** Living with LGMD2 not only implies learning to live with the disease and the variations between good and bad periods but also means trying to make sense of a progressive disease that brings uncertainty about future health, by striving to make the best of the situation. Disease progression involves practical and mental struggle, trying to maintain control over one's life despite vanished physical functions that require continual adjustments to the body. Restrictions in a double sense were described, not only due to the disease but also due to poor comprehension of the disease in society. Lack of knowledge about LGMD2 among professionals often results in having to fight for the support needed. **Conclusion:** In order to manage daily life, it is important to be seen and understood as an individual in contacts with professionals and in society in general, to have informal social support and meaningful activities as well as access to personal assistance if necessary.

### Keywords

Chronic diseases, LGMD2, muscular weakness, rare diseases

### History

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### ► Implications for Rehabilitation

- Recessive limb-girdle muscular dystrophy (LGMD2) is a group of progressive disorders, which manifest in physical and psychological consequences for the individual.
- According to the salutogenic orientation, people need to find life comprehensible, manageable and meaningful, i.e. to achieve a sense of coherence (SOC), but living with LGMD2 may recurrently challenge the individual's SOC.
- Through the holistic view of the individual's situation that the salutogenic orientation provides, professionals may support the individual to strengthen SOC and thereby facilitate the movement towards health.

### Introduction

Young adults diagnosed with recessive limb-girdle muscular dystrophy (LGMD2) [1] not only have to face the transition period from adolescence to adulthood [2] but also have to cope with a rare chronic disease, which is characterised by progressive muscle degeneration and weakness [3]. In a time when the young adults are supposed to strive for autonomy, their ability to perform activities of daily living may decline, with increased dependency on others. Articles about LGMD2 focus on genetic diagnosis [4] and therapeutic approaches [5–7] as well as prevalence [8,9], which is estimated at 8.1–69 per million and varies for different

forms of LGMD2 in different populations [3]. As the disease progresses, the individual will need support from a multidisciplinary healthcare team [1] and from staff in the municipality [10] in order to manage daily life. However, there is lack of knowledge about young adults' experiences of living with LGMD2, and the literature review revealed no study in this area.

LGMD2 is a group of muscular dystrophies (MDs) [11] in which the common feature is an involvement of the proximal musculature in the shoulder and pelvic girdles; it is otherwise characterised by broad genetic and clinical heterogeneity [12]. There are different subtypes of LGMD2, with the classification based on gene identification. The age of onset may vary from early childhood to adulthood, and rate of progression varies and may not be linear [12]. The muscle weakness generally begins with difficulties in running, climbing stairs and getting up from the floor, followed by weakness in the shoulder girdle. Eventually

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walking ability may vanish [3]. Deterioration of physical capacity could manifest as experiences of longing for missed activities and relationships and wishing to be seen as a person among adolescents with Duchenne MD [13], which is the most common childhood MD [14]. The change from being independent to being in need of assistive devices and support from others to manage activities is described as a psychologically trying process among adults with different forms of MD, but several persons experience better psychological adaptation over time [15]. There is no curative treatment for LGMD2, and although the benefit of steroids has been reported in some of the subtypes, treatment remains supportive. Physiotherapy to promote walking and prevention of contracture development through stretching and orthoses is important for maximising functional ability [6]. Complications due to cardiac involvement and respiratory muscle weakness may occur in a few of the subtypes of LGMD2. The prognosis for LGMD2 is not uniform and therefore early identification of potential complications may improve survival [12]. LGMD2 is a group of rare diseases [3], which according to the European definition is a life-threatening or chronically debilitating condition that affects no more than one of 2000 citizens [16]. Living with a rare diagnosis in itself is an additional burden to the real disability since the individuals often face difficulties when accessing support from healthcare and society due to lack of knowledge about the disease [17].

The late teens through the twenties have been described as the transitional period leading from adolescence to adulthood. It is a self-reflective period when the individual often explores different areas in life such as love, education, work and home-making. It is not only viewed as a time of opportunities and high expectations but also as a time of anxiety and uncertainty because the lives of the young individuals are often unsettled [2]. Living with chronic and disabling conditions at the end of formal schooling requires careful planning for further education or work, for moving from home to independent living and for changing from paediatric to adult health care [18]. In Sweden, personal assistance takes the form of a human aide who performs physical activities that a person is unable to do due to functional impairment, in order to provide independence and full participation in society [19].

Chronic disease threatens control [20] as well as meaning and coherence in life [21]. The salutogenic orientation focuses on what causes the individual's movement towards health on the health–disease continuum, rather than the aetiology of the disease [22]. Living with a chronic disease is a stressor that the individual needs to cope with and adapt to. By trying and receiving help to comprehend and manage the situation and make life meaningful, the individual can achieve a sense of coherence (SOC). The SOC consists of the concepts: comprehensibility, manageability and meaningfulness. Comprehensibility refers to whether or not the individual finds that inner or outer stimuli make sense in terms of being coherent, structured and clear. Manageability refers to the individual's degree of confidence that internal and external resources to cope are available. Meaningfulness refers to areas in life that are important for the individual and the perceptions of whether or not life's difficulties are worth an investment of energy and engagement. A strong SOC leads to improved health [22]. The salutogenic orientation does not disregard the fact that an individual has been diagnosed with a disease, but relates to all aspects of the individual when investigating how a person can be helped to move towards increased health on the health–disease continuum [23].

Multidisciplinary health care and support from staff in the municipality are important resources when it comes to facilitating the movement towards increased health for young adults with LGMD2, but to optimise the support given in daily life it is

essential to gain more knowledge about how the young adults experience everyday life.

The purpose of this study was to describe, from a salutogenic orientation, young adults' experiences of living with LGMD2.

## Methods

A qualitative explorative study was conducted using semi-structured interviews for data collection, which gave the participants the opportunity to respond in their own words and to express their own personal experiences but within a given frame [24]. The salutogenic orientation [22] was chosen as a theoretical framework because it provides a holistic view of the participants' experiences of living with LGMD2. Furthermore, it focuses on internal and external resources available for coping and the individual's movement towards health.

## Participants

Purposeful sampling was used [24]. Those invited to participate were individuals aged 18–30 years diagnosed with LGMD2. Individuals with cognitive impairment and those who did not speak Swedish were excluded.

Fourteen participants were included in this study from hospitals in different healthcare regions ( $n=10$ ), from the association Neuro Sweden ( $n=1$ ) and from a web-based association for people with disability ( $n=3$ ). The managers at the hospital clinics involved and the chairman of Neuro Sweden gave their written approval for this study. With the help of the development coordinator for habilitation centres in southern Sweden, a contact list was compiled of physicians and physiotherapists working with individuals diagnosed with LGMD2. The principal investigator (first author) contacted the physicians and the physiotherapists on the list, as well as members with key positions in Neuro Sweden, by e-mail or phone, with a request to forward a letter with information about this study to potential participants. An information letter about this study was also sent by the principal investigator by e-mail to members of the web-based association who could be included. Those who were willing to participate in this study sent contact information to the principal investigator, who then telephoned the participants to arrange a time and place for the interviews. Participants recruited through healthcare had confirmed LGMD2 diagnoses, based on analysis of muscle biopsy and genetic testing [5], but four of the participants were self-reporting confirmed diagnoses.

## Interviews

An interview guide with semi-structured questions, based on the content of the SOC-13 questionnaire [22], was developed to focus on the participants' comprehensibility, management and meaningfulness when living with LGMD2. Examples of main interview questions were: Could you describe what it means to live with LGMD2? Is there any support that you need that you do not have access to? If so, could you describe what it is? What do you perceive as the most meaningful in life? Follow-up questions were asked to reach a deeper understanding of the participants' experiences, e.g. How do you mean? and Could you give an example? The interview guide was pilot-tested with three respondents (included in the study) and no changes were made. The interviews were conducted between June 2012 and November 2013, at the participants' homes ( $n=12$ ), at the respondent's workplace ( $n=1$ ) and at a neutral place ( $n=1$ ). Each interview lasted about one hour and was conducted by a nurse (first author) without any professional or private relation to the participants. The interviewer is experienced in the care of individuals with MD, as an anaesthetic/intensive care nurse and being a next of kin

Table 1. Example of coding agenda according to Mayring (2000).

Category	Definition	Example	Coding rules
Comprehensibility	Slow muscular weakening Difficult when lost functions become obvious Continual adjustments	<i>It happens slowly... slowly but I don't notice it until I use my body for a special thing and then I think, "Damn, I could do this last year, now I can't do it". For every deterioration you have to reorganize your life... that's when it gets difficult... you adapt the whole time. (10)</i>	Cognitive category that refers to experiences of what it means to live with the diagnosis and whether implications of the disease make sense and are predictable or not
Manageability	Struggling to receive support Lack of knowledge about the disease among professionals	<i>Just to get them (professionals in health-care) to understand that I need things that they don't normally give... you have to nag a lot and really explain, and not so many people know about muscular diseases. (5)</i>	Behavioural category which includes experiences of whether internal and external resources to cope are available or not
Meaningfulness	Being engaged in meaningful activities	<i>An important part of it is having that job... it's fun and things happen and the interaction with people is important. (4)</i>	Motivational category which refers to experiences that bring joy and meaning in life

to a person with the diagnosis. The interviews were recorded and transcribed verbatim. Collection and analysis of data proceeded concurrently until no new data were added to the analysis [24].

### Data analysis

Qualitative content analysis according to Mayring [25] was used for data analysis. First, the interview text was read through several times in order to get a sense of the whole. The text was then read line by line and content units were identified and coded inductively. Codes were compared and contrasted and those with similar content were grouped into subcategories. Second, the subcategories were deductively organised into the main concepts that describe a person's SOC in the salutogenic orientation, namely comprehensibility, manageability and meaningfulness [22], using these concepts as deductive categories. Coding rules determining under what circumstances a text passage could be coded with a category were explicitly defined (Table 1) and thus new subcategories were deductively formed in a dynamic process. Throughout, the coding and categorisation were named as close as possible to the text [25]. Continuous discussions regarding analysis and content in the categories were held until coder agreement was achieved together with the two co-authors, who are experienced in qualitative research and particularly in using content analysis [24,25]. This was in order to increase trustworthiness of the data and reduce the influence of potential interviewer bias.

### Ethical considerations

Ethical approval for this study was obtained from the regional ethic board in Linköping, and this study was performed in accordance with the Declaration of Helsinki [26]. Written informed consent was obtained from the participants in connection with the interviews.

### Results

Fourteen respondents, eight females and six males, aged 20–30 years (median = 25), participated in this study (Table 2). The median age of symptom debut was 11 years (range 0.5–25). Physical functional abilities varied among the participants. Although some were physically active with no or minor limitations in everyday life due to the disease, others were in need of wheelchair for ambulation and had experienced increasing

Table 2. Demographic characteristics of the participants.

Participants	Numbers (N = 14)
Subtypes of LGMD2	
LGMD2A	5
LGMD2B	3
LGMD2E	1
LGMD2I	5
Living conditions	
Living on their own	4
Cohabiting	4
Living with parents	4
Living with other relatives	2
Siblings with LGMD2	6
Having children	1
Education	
Completed upper secondary school	10
Dropped out of upper secondary school	4
Finished a university education	3
Occupation	
Working full-time	3
Working part-time	1
Studying full-time	4
Not working/studying	6
Ambulation	
Walking with no assistive device	3
Walking with a stick	1
Walking/using wheelchair	3
Using wheelchair for ambulation	7
Access to personal assistants (from 98 h/week up to 24-h basis)	7
Using continuous positive airway pressure (CPAP) at night	2

difficulties in raising their arms and performing activities such as combing hair or lifting a cup to drink. The participants could be divided into three groups according to their descriptions of dependency on other people and assistive devices in everyday life: those who were independent managed daily life on their own ( $n = 2$ ); those who were in transition from being independent to become more dependent received informal support from next of kin due to increased physical impairment and limited walking ability ( $n = 5$ ); and those who were dependent were entitled to personal assistance and always used a wheelchair for ambulation ( $n = 7$ ). Experiences of reduced energy were described, and



Table 3. Overview of deductive categories and subcategories developed through data analysis in an interview study with young adults living with recessive limb-girdle muscular dystrophy.

Categories	Subcategories
Comprehensibility	Slow muscular weakening requires continual adjustments Having a rare disease with limited information available Uncertainty regarding future health
Manageability	Struggling in order to receive support needed from society The need for person-centred care Personal assistance in order to replace physical functions Important to have informal social support Making the best of the situation
Meaningfulness	Engagement in meaningful activities Increasing people's understanding of disability Setting and achieving goals

some of the participants reported having frequent pain in the back or in the whole body.

The results showed that the individual's SOC in everyday life may be recurrently challenged as the disease progresses. However, the participants were determined to make the best of the situation, relying on internal and external resources to cope. The categories and subcategories developed from the data analysis are listed in Table 3.

### Comprehensibility

Living with LGMD2 was not only described as being a normal part of life but also as a struggle to get through the day. The participants perceived that the disease is manifested in both physical and psychological consequences. There were periods described as physically and mentally stable and other periods experienced as more difficult to cope with.

#### *Slow muscular weakening requires continual adjustments*

Comprehensibility about the disease was very much based on the symptoms that had arisen. A majority of the participants described experiences of slow muscular weakening, resulting in vanished physical functions, which influenced and continually required adjustments in life. Sometimes, it was possible to feel and anticipate the coming deterioration and to be a step ahead, mentally as well as practically, but often muscular weakening did not become obvious until the person thought back or tried to use the body in a special way and realised that functions had vanished. Some of participants said that, despite knowing their diagnosis, they had not expected that physical functions would vanish the way they had, rather that the progression would have ended somewhere. Comprehending that it is a progressive disease, yet not really knowing what to expect, was described as mentally difficult and created many thoughts and emotions. Participants experienced psychological distress, which was exacerbated when physical functions had vanished, when complications became evident or when other misfortunes that did not have to do with the diagnosis occurred in life. Some of the participants had gone through periods of depression. The participants described continually having to adjust to changes in the body and to the surroundings, as the disease progressed.

I suppose that's what I think is the major thing about the diagnosis, that it's constant change . . . adapting all the time to the body and new situations. (1; dependent)

The transition from ambulation to becoming dependent on using a wheelchair was perceived to involve major emotional and practical adjustments. It was described as a psychologically distressing process, although the wheelchair was viewed as a great assistive device that provided freedom. Being dependent on other people could be mentally strenuous and require patience. Experiences of physical and social limitations due to the disease were described. The participants said that when physical prerequisites differ, it becomes important to think about things in daily life that other people might take for granted. Every activity has to be carefully planned, taking into account the time and energy required, as well as obstacles in society that could restrict the choice of activities, such as stairs and buildings that are not adapted. Experiences of not having equal rights to act in society were described, and although the perception was that it had become better over the years, incomprehension in society was felt at times to inhibit daily life. It could be people talking above one's head when one is using a wheelchair, or being viewed as a victim, which could in turn affect the way the individuals viewed themselves.

#### *Having a rare disease with limited information available*

The participants experienced the consequences of having a disease that is rare. The perception was that there is limited information available about the diagnosis and that it is difficult to find anything to associate with when searching for information on the Internet. Some of the participants were frustrated at not receiving answers to questions posed to professionals in healthcare, while others were content with the information they had or preferred not to seek further information. Some of the participants expressed a sense of having information about the disease within the body and thought that it could be more difficult for next of kin, standing alongside, to comprehend the disease. The disease was sometimes described as invisible since it does not become obvious to others until the person moves in a way that is affected by weak muscles, which could mislead people to expect that the person is capable of performing more physical activities than is possible. Limited information available about the diagnosis often left the individuals to provide the information themselves.

It's really difficult to find anything at all (information about the disease) that is not just medical terms . . . more information is needed for society so that you could say, 'Here, read this' . . . but there's nearly nothing and so you have to explain to the person. (14; Dependent)

It could be mentally difficult repeatedly having to inform and explain for people when coming into a new context. Some of the participants were open and gave information about the diagnosis when meeting new people, while others were more apprehensive or had tried to hide it by avoiding situations where the disease became obvious. Meeting people with the same or similar diagnosis could create a sense of fellowship and understanding for each other, although exchanging experiences was not always found to be as easy as first thought. Many of the participants had never met anybody with the same diagnosis outside their family and they not only wondered about how other people live their lives but also hesitated to meet anybody since it could create negative thoughts about the future if the other person was worse. There was also awareness about individual differences, and siblings within the same family, despite having the same diagnosis, experienced differences in how the disease had developed.

### *Uncertainty regarding future health*

The participants perceived that the diagnosis brings uncertainty regarding future health. There was a recognition that the disease had shaped who they are today and thus concern about who to become in the future. Some of the participants had reconciled themselves with the thought of slowly becoming weaker, while others described anxiety about what will happen and if support needed will be received. If negative thoughts about the future arose, there was a sense of trying to live in the present, one day at a time.

Now I'm alive and this is how I feel...if I could just think, "OK, it's fine now" and not think so much about the future, that it can get worse, then everything would be good, but there are these thoughts that keep spinning, "What will happen?"...you have to take one day at a time. (6; in transition)

Some of the participants did not expect any major progression of the disease within the near future. It was felt to be better not to know all the different scenarios that might occur, since it is not beneficial to worry about things that might never happen. At the same time, it was considered important to do what can be done in order to facilitate future health by finding and treating complications in time and through habilitation. There was a gleam of hope that researchers might find a cure but also a recognition that it is better not to wish too much in order not to be disappointed. When thinking about possibly having children, there were reflections about how to manage parenthood and the fact that it is a hereditary disease.

### **Manageability**

The participants described not only their need for but also work required in order to get access to external support from different professionals in the society in order to manage daily life. Informal social support was also perceived as important, as well as a sense of relying on internal resources within oneself to manage daily life.

### *Struggling in order to receive support needed from the society*

The participants expressed an appreciation for living in a time and in a country with high-standard health care and social welfare including personal assistance, but the perception was that it is often time-consuming and requires much work to access the support needed. As the disease progressed, there were practical and emotional issues that require information seeking and help in order to manage. The support most needed varied among the participants. In addition to medical concerns and the need for assistive devices, the participants described the need for, e.g. physiotherapy, personal assistance, financial support when not being able to work, adaptations in the home and car adaptation. Moving was perceived to require much planning. Thus, many of the participants had contact with several professionals from the multidisciplinary healthcare as well as from different authorities. Often they had to explain the consequences of the diagnosis and their needs or declare that there was no improvement and that granted support was still needed. Experiences of being met with scepticism, not being listened to and having to fight for support were described, which was perceived to bring additional difficulties when coping with the disease.

You have to struggle all day long to cope with having a disease and then have the energy to fight for your life as a person...sometimes you get angry because nobody listens...you have to struggle to get what you need. (14; dependent)

The participants perceived that having contact with professionals in healthcare who have previous knowledge of the diagnosis provides confidence, and there was a desire to be able to have direct contact with a specialist if needed. Despite growing physically weaker, the frequent visits to hospital and the contact with professionals had been reduced, and after being transferred to adult healthcare, it had become more difficult to get in contact with healthcare and to know who to turn to. Sometimes years could pass without being called for a doctor's appointment, which gave a feeling of being neglected. Experiences of not being seen and understood as an individual within healthcare were described.

because they don't see the whole...they don't see me as a person and individual but as the disease, so they can't try to suggest creative solutions for me...I think they ought to think a bit for themselves about how they behave and what they say, because we are well aware that we are sick...it gets very palpable and they don't understand because they can't put it in relation to different people so it's really difficult. (2; dependent)

When physical activities were limited, some of the participants found it difficult to avoid weight gain due to medication or when beginning to use the wheelchair more. The risk of a vicious circle with eating, bad conscience and weight gain was described by one participant. Another was frustrated and felt a lack of help and had bought slimming pills over the Internet, which resulted in severe heart palpitations after taking the pills.

### *The need for person-centred care*

The participants described how living with LGMD2 had given them experiences about how their body works and how to manage different situations in order to feel well. By listening to body signals, there was an ambition to do what can be done in order to maintain physical functions as long as possible, but it could be difficult to get a response when talking about personal experiences.

The most frustrating thing about healthcare is that I always get the answer "Everybody is different so it's not possible to say what it will be like", whereas I have a clearer picture of what it will be like and I want to be ahead of things...I notice the sneaking changes, while for them it's when there's a dramatic change...it's when the change arises and it's difficult health-wise...I want to do what I can do to feel well today but I also know that it will pay off in five years...I have always been told that you do exercise to get better, not to stay or to feel well...a new aspect has come in the last few years, that it's important to be able to feel well on an everyday basis, not that your Achilles tendons actually are getting longer. (1; dependent)

Several of the participants thought that information about physical activity had been vague or varied over the years and between different professionals, which could lead to uncertainty about what to do. Some of the participants said that they had found their own ways and had a sense of creating their own conditions; doing bikram yoga, for example, which is performed in warmth, had given physical and mental strength. It was described as important to listen to the body and to not overwork when performing physical activities.

The first thing they said was, "It's good if you don't take exercise"...I have gone on taking quite a bit of exercise...and then my counts rise when I exert myself so much

that I get brownish urine . . . that's not good so I cut back, of course, and I know how to prevent it as well as possible and drink a lot of fluid and warm up properly . . . it feels as if you've made your own conditions, and I wouldn't have been able to do these things if I had stopped taking exercise when they told me to. (4; independent)

Some of the participants had received water training and stretching programmes from a physiotherapist but wanted more organised habilitation. There was also a recognition that other things of importance in life at times are given priority over habilitation, e.g. when education requires all attention, and that it could be difficult to find the time, energy and motivation to train.

#### *Personal assistance in order to replace physical functions*

Personal assistance was described by some participants as being a major part of their lives. Assistance was viewed as arms and legs that perform physical activities that the individuals are not able to do by themselves. It was experienced as a privilege that brings independence with the help of another person and freedom to do activities, as well as confidence in knowing that help is available, but it also entailed administrative work and affected private life. It could also be difficult to find a role of one's own. Some of the participants described the importance of defining what personal assistance means to them and "being in charge", while others experienced more of a friendship or could plan their day according to different assistants. Having family members as assistants was viewed as a way to associate without the presence of an outside person. There were ambivalent feelings about having parents as assistants. On the one hand, it felt like a relief since they know all the needs, but on the other hand, it was difficult to be so dependent on them, and sometimes frustration was taken out on the parents. The participant who had children had assistance to look after them, which would not otherwise have been possible when being alone with them, and this was found to work well. The investigation conducted in order to receive or renew personal assistance was described as a process of self-disclosure, where every minute of help needed during a day is counted and summarised. Some experienced having to say that the plan with personal assistance was to be able to walk again within half a year, despite knowing that it would not be possible. Participants described being denied assistance, getting reduced hours when applying for more and how the hours granted were not enough to live the life they wanted, e.g. having to give up an education due to withdrawal of personal assistance.

Assistance is there so that you can have a life of full dignity, but that's not what they grant you hours for . . . often it's more to survive and it's unfortunate that you should have to wait till you're so much worse when you could have done so much more before, perhaps. (13; dependent)

#### *Important to have informal social support*

The participants felt that support from family and friends is important in order to manage practical as well as emotional issues in daily life. It was found important to have somebody to turn to and talk with and not keep everything inside. There was a recognition that parents are ordinary people and do not have the answer to everything, but many of the participants experienced great support from their parents.

I have managed very well because I have a really good mother . . . I wouldn't have managed any of this if it hadn't been

for her . . . she said, "This will work out fine . . . we'll manage . . . it's not the end of the world . . . there are more people who are sick . . . you can do it." She gave me very strong self-confidence. (10; in transition)

Participants often counted on their parents as backups in the contact with professionals or if something unexpected occurred. Some received help with cleaning, cooking, laundry and shopping. Positive next of kin and friends, who understand the situation without focusing on the hindrances and who could easily give a hand, were appreciated. There was a preference for realising one's own limitations instead of others saying what to do. Individuals in need of support, without personal assistance, had to lean on help given by next of kin and friends, which could be difficult at times.

#### *Making the best of the situation*

The perception was that mental well-being makes it easier to manage the situation. It was therefore considered important to try to think positively, to focus on possibilities and to have things to look forward to. There was a recognition that nothing can be taken for granted and a motivation to try to make the most out of life. Participants said that since they cannot do anything about having the disease, they somehow have to learn to accept living with it and try to make the best of the situation, although it could take time.

The most important thing is that you must accept it, and that's what takes such a long time . . . that's what it's like . . . you can't do anything, change anything . . . you simply have to try to make the best of the situation. (6; in transition)

Some of the participants described themselves as stubborn, which had helped them to fight and go through with things they wanted to do even though it required much effort. Participants found new creative ways to do things differently and felt that, given the right support, it was possible to manage more than expected.

#### **Meaningfulness**

The perception was that when energy is reduced, activities that are meaningful and bring energy should be prioritised. Activities that bring joy and make participants feel physically and mentally well, that they have a function and engagements of importance for personal development and/or of importance for other people, were described as meaningful.

#### *Engagement in meaningful activities*

The participants felt that being engaged in meaningful activities was important from a health perspective. The perception was that associating and having fun with other people contributes to well-being. Pets were a source of joy and comfort for some of the participants. Sometimes, things that were not reflected so much upon before were appreciated more, such as the freedom of driving a car and socialising with friends. Having a job was, besides a source of income, a way to form an identity, and the social interaction with people was considered important for well-being. Some of the participants have had the experience of being forced to give up a job due to physical deterioration. Leaving working life was described as mentally difficult. However, when strength was reduced, it was not possible or justifiable to work full-time or to work at all any longer since working was not worth its price, when it took so much time and energy that it was not possible to do other important things in life.



Healthy people work and mix with friends and do things, but I haven't got the strength to do both so I have to choose... on the days when I work I go home afterwards and take it easy but then I try to do fun things and take exercise on the days when I'm free. (2; dependent)

Many of the participants had been active in different sports when younger but were not able to perform them any longer. Sometimes, it was possible to change from being an active performer to become an onlooker or to participate at an organisational level, but sometimes it was not meaningful to stick to an interest. Finding new interests that really mean something was not always easy, but experiences of, e.g. finding the joy of becoming politically active or starting to dive after receiving the diagnosis were described. Some of the participants enjoyed the computer, using Skype to connect to other people. It was found easier to do meaningful activities during spring and summer when it is warm, compared to wintertime.

I notice the difference between winter and summer... then it's as if I can let out all the things spinning around in my head that I want to do because I feel up to it... I suppose it has to do with the light and the heat... when it's cold I'm always a bit too lightly dressed because otherwise I can't move... it's as if all the energy for half a year is used for shivering. (8; dependent)

#### *Increasing people's understanding of disability*

Participants were active in different ways to increase people's understanding of disability. This could be involvement in political issues concerning accessibility and attitudes in society, giving lectures, writing and presenting poetry about living with disability and being active in different associations. Experiences of being a mentor for younger people with neuromuscular disorders were also described. Being a mentor implies keeping in touch and doing activities together in order to support the younger person, who could express issues of concern that might be difficult to share with parents. The mentor is there to listen and contribute his or her own experiences.

I find all my energy in Young Mentors, where I'm active and support people in the same situations that I've been in, although younger... it's a joy you can spread... if you have a person who understands fully how it feels and what it's been like, that alone helps them. (11; in transition)

#### *Setting and achieving goals*

Having goals to look forward to and having the support needed to set and achieve the goals was described as meaningful, since it was then possible to see a future despite the disease. Goals could be: to complete an education, to find a job, to move from the parents' home and to travel. Many of the participants liked to travel, and destinations were often countries with a warm climate. Experience of swimming with dolphins and looking forward to doing that again were described. Travelling alone with personal assistance and realising that it was possible to be independent of next of kin gave a sense of freedom.

Now I have got a place on this education programme... when I can see a future despite everything... some kind of goal... that gives me a great deal of energy... now I'm starting to think about where I can travel this summer... and then, now that I can do these things with assistance... when it works it's

absolutely fantastic because then you feel that you can do anything... that you have freedom and no restrictions... that gives life some meaning. (9; dependent)

## **Discussion**

This study is unique as it describes young adults' experiences of living with LGMD2 with a salutogenic orientation. The main finding is the determination among the participants to try to make the best of the situation, despite having to cope with a rare progressive disease that requires continual adjustments to the body and brings uncertainty about future health, which could be mentally demanding. Simultaneously, experiences of having to cope with obstacles and lack of knowledge about the disease in society were described, as well as a sense of often having to struggle for the support needed. Informal social support was described as important in order to manage daily life, as well as engagement in meaningful activities and access to personal assistance when needed. The findings indicate that LGMD2 can negatively influence the individual's SOC as the disease progresses.

Comprehensibility as a cognitive category [22] shows that life with LGMD2 at times does not make sense or is not predictable. Comprehensibility regarding the disease was very much shaped by symptoms that had arisen over the years and the adjustments to the body and new situations that had followed. Charmaz [20] describes how people experience chronic illness as an interruption in life, as an intrusive illness or as immersion in illness. In this study, the disease could be viewed as an interruption, as long as the impairment did not affect daily routines, but just occasionally influenced the person. As the disease progressed and demanded allotted time and adjustments, it became intrusive and threatened control over oneself and situations, which could result in uncertainty about future health and who to become. Experiences of immersion in illness were described in this study when situations arose that required focusing all attention on the disease, for example, due to complications or hospitalisation. There were times when the disease was stable and everything was running well, whereas other times were experienced as practically and mentally more difficult. These findings could be compared with how people experience chronic illness in terms of good days, when having control over mind, body and actions and bad days, when one cannot be one's preferred self and valued pursuits are difficult or impossible to perform [20]. Life could be described as founded on the disease for some of the participants, meaning that although the disease organises life, it does not entirely define or fill it [20]. In order to achieve mental well-being despite a progressive disease that cannot be controlled and that brings uncertainty about future health, the participants tried to control stressful thoughts and emotions associated with the disease, e.g. by not thinking about future health. This could be referred to as emotionally focused coping, which means regulating emotions tied to the stress situation without changing the reality [27]. Some of the participants were frustrated at not receiving answers to questions about the disease, while others preferred not to seek further information. Therefore, information about the diagnosis must be individualised depending on the person's needs. Furthermore, the individual can be strengthened to gain better control in life through increased knowledge about entitled support from society when having functional impairment and information about where in society to turn regarding different issues [28].

Manageability as a behavioural category [22] shows the additional difficulties in managing daily life because it is a rare disease with limited information available. Problem-focused

coping was used by obtaining information about what to do and taking action in order to change the reality of the troubled person–environment relationship [27]. Participants found, however, that it is often time-consuming and requires much work to organise life when living with LGMD2, and that there is much bureaucracy associated with the provision of support by society. When living with a rare disease, there is risk of being treated arbitrarily when needing public service, due to lack of knowledge about the diagnosis [17]. This could create contradictions, as in this study when having to state that the plan for personal assistance is to be able to walk, knowing that it is not possible or having to declare that no improvement has been made and granted support is still needed despite a doctor's certificate confirming that it is a progressive disease. Procedures like this are not only a hindrance to the individual's manageability but they also demand time and administrative work for professionals. Therefore, suitable procedures to administer support for individuals with LGMD2 need to be developed, acknowledging that it is a progressive disease. Development of international guidelines for management of LGMD2, as there are for Duchenne MD [29,30] and congenital MD [31], could serve as an important tool for the individuals, their families and professionals. In order to facilitate manageability, the individuals also need the possibility to have direct contact with a specialist if needed.

In order to manage and make sense of the disease, participants relied on internal resources by listening to signals within the body and trying to learn to accept living with the disease. A hindrance to manageability was the perception of not always being seen and understood as an individual in society, rather being reduced to being the disease. The participants were in a self-reflective period of life, when they think about who they are and what they want out of life [2]. They were well aware of limitations and possibilities due to the disease, and when trying to make the best of the situation, different practical and emotional issues of concern arose that had to be addressed to professionals in healthcare and authorities in society. At the same time, through experiences of living with the disease in combination with a sense of having information about the disease within the body, the individuals had become experts on what the diagnosis implied for them and what their needs were, and there was frustration when they felt they were not being listened to. It is not possible, nor expected, that professionals in healthcare have knowledge about every rare diagnosis [16], but recipients of care need to be viewed from a holistic perspective as individuals [32] and professionals need to acknowledge the expert knowledge that the individual possesses. Empowerment means balancing the power relation between the individual and the professionals so that the focus to a greater extent is on problems and solutions formulated by the individual [28]. Person-centred care highlights how important it is for professionals to know the person behind the patient, and by initiating the person's narrative about experiences of living with the disease in an everyday context a partnership and shared decision making between the individual and professionals could be established [21].

Meaningfulness as a motivational category [22] shows that engagement in meaningful activities and being able to achieve goals was important for well-being. Simultaneously, experiences of being forced to quit a job or abandon an interest due to vanished physical functions were described, and it could be difficult to find new meaningful activities. According to Charmaz [33], people with chronic disease measure their pursuits by scrutinising involvements, accounting for illness and setting priorities and further adopting such measures as markers of who they are and are becoming. Being able to participate in meaningful activities thereby nourishes and validates the self. Increasing the understanding of disability in society was described as meaningful both for the individual's own sake and for the benefits it brings to other

people. The values of social interaction between adolescents with MD has been shown [34] and could be combined with young adults with their own experiences of the disease as mentors. Some of the participants were dependent on other people in order to manage daily life, which can be mentally difficult. Empowerment means strengthening the individual's own ability and opportunity to control one's life regarding areas that are important for quality of life, such as: circumstances that influence health, where and how to live, what education to attend, what to work with and what to do during leisure time [28], all of which are important aspects when it comes to striving for autonomy and being independent of parents in the transition to adulthood [2]. Access to personal assistance is a way to empower the individual to gain better control in life regarding these areas and to become independent with the help of another person.

All of the participants in this study had next of kin to support them, which was described as meaningful and important in order to manage everyday life. Informal social support could be regarded as a generalised resistance resource that facilitates effective stress management [22]. Positive aspects of caregiving for young individuals with MD has been shown to be more recognised by next of kin, who felt that they received a higher level of professional help and psychological social support [35]. Professionals therefore need to acknowledge and support not only the individual but also next of kin.

This study included participants belonging to different healthcare regions in Sweden, living in urban areas as well as in the countryside, and the variety of phenotypes described in the literature [12] were represented, which gave a range of different experiences of living with LGMD2. Data were first inductively coded in order to minimise the risk of overlooking data and to strengthen the credibility of this study [24]. Using the salutogenic orientation as a theoretical framework in the analysis was a strength considering the aim of the study, although the concepts of comprehensibility, manageability and meaningfulness are closely intertwined [22] and thereby difficult to separate into watertight categories. The two co-authors, who are trained in qualitative content analysis, evaluated the data analysis to confirm its relevance, which enhances the credibility of the study [24]. A limitation of qualitative research is that the result is contextual and cannot be generalised, but patterns in the findings might be transferable to similar groups in comparable contexts [24,36].

To conclude, living with LGMD2 was described both as being a normal part of life and as a struggle to get through the day, which refers not only to coping with the disease and its physical and psychological consequences for the individual but also having to face obstacles and incomprehension of the diagnosis in society. In order to optimise support given by professionals, it is essential not only to acknowledge the unique experiences the individual possesses by living with LGMD2 but also to view the person behind the disease [21] and in cooperation with the individual find out what actions are needed in order to strengthen the individual's SOC in daily life. Informal social support is important for individuals to manage everyday life and therefore the roles of next of kin need to be recognised by professionals. Personal assistance enables independence and engagement in meaningful activities as well as goal achievement. The salutogenic orientation provides a holistic view of the individual's situation and a coping strategy, in which the individual can be supported by professionals to strengthen SOC and thereby facilitate movement towards health on the health–disease continuum.

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### Declaration of interest

The authors report no conflicts of interest.

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