Living with recessive limb-girdle muscular dystrophy
affected young adults’ and parents’ perspectives, studied through a salutogenic framework

Anna Carin Aho
Living with recessive limb-girdle muscular dystrophy

affected young adults’ and parents’ perspectives, studied through a salutogenic framework
LIVING WITH RECESSIVE LIMB-GIRDLE MUSCULAR DYSTROPHY affected young adults' and parents' perspectives, studied through a salutogenic framework.

CARIN LINNAEUS UNIVERSITY PRESS
LIVING WITH RECESSIVE LIMB-GIRDLE MUSCULAR DYSTROPHY
affected young adults' and parents' perspectives, studied through a salutogenic framework

ANNA CARIN AHO

LINNAEUS UNIVERSITY PRESS
Abstract


Aim: The overall aim of this thesis, using a salutogenic framework, was to develop knowledge about experiences and perceptions of living with recessive limb-girdle muscular dystrophy and its influences on health, from the affected young adults’ and their parents’ perspectives.

Methods: A qualitative explorative and descriptive study design was used. Semi-structured interviews were held with 14 young adults diagnosed with recessive limb-girdle muscular dystrophy, aged 20 –30 years, and 19 parents. Data analyses were conducted using content analysis (I, II, III) and phenomenography (IV). In order to mirror the interview data, the participants also answered the 13-item sense of coherence questionnaire.

Findings: Recessive limb-girdle muscular dystrophy has a major impact on the affected young adults’ and their parents’ lives as the disease progresses. Health described in terms of well-being was thus perceived to be influenced, not only by physical, emotional and social consequences due to the disease and worry about disease progression but also by external factors, such as accessibility to support provided by society and other people’s attitudes. There was, however, a determination among the participants to try to make the best of the situation. The importance of being able to mobilize internal resources, having social support, meaningful daily activities, adapted environment, the young adult being seen as a person and having support from concerned professionals, including personal assistance when needed, was thereby described. Self-rated sense of coherence scores varied. Those who scored above or the same as median among the young adults (≥ 56) and the parents (≥ 68) expressed greater extent satisfaction regarding social relations, daily activities and external support than those who scored less than median.

Conclusion: This thesis highlights the importance of early identification of personal perceptions and needs to enable timely health-promoting interventions. Through dialogue, not only support needed for the person to comprehend, manage and find meaning in everyday life can be identified, but also internal and external resources available to enhance health and well-being, taking into account the person’s social context as well as medical aspects.

Keywords: LGMD2, parents, salutogenic, sense of coherence, young adults
Abstract


Aim: The overall aim of this thesis, using a salutogenic framework, was to develop knowledge about experiences and perceptions of living with recessive limb-girdle muscular dystrophy and its influences on health, from the affected young adults' and their parents' perspectives.

Methods: A qualitative explorative and descriptive study design was used. Semi-structured interviews were held with 14 young adults diagnosed with recessive limb-girdle muscular dystrophy, aged 20–30 years, and 19 parents. Data analyses were conducted using content analysis (I, II, III) and phenomenography (IV). In order to mirror the interview data, the participants also answered the 13-item sense of coherence questionnaire.

Findings: Recessive limb-girdle muscular dystrophy has a major impact on the affected young adults' and their parents' lives as the disease progresses. Health described in terms of well-being was thus perceived to be influenced, not only by physical, emotional and social consequences due to the disease and worry about disease progression but also by external factors, such as accessibility to support provided by society and other people’s attitudes. There was, however, a determination among the participants to try to make the best of the situation. The importance of being able to mobilize internal resources, having social support, meaningful daily activities, adapted environment, the young adult being seen as a person and having support from concerned professionals, including personal assistance when needed, was thereby described. Self-rated sense of coherence scores varied. Those who scored above or the same as median among the young adults (≥ 56) and the parents (≥ 68) expressed greater extent satisfaction regarding social relations, daily activities and external support than those who scored less than median.

Conclusion: This thesis highlights the importance of early identification of personal perceptions and needs to enable timely health-promoting interventions. Through dialogue, not only support needed for the person to comprehend, manage and find meaning in everyday life can be identified, but also internal and external resources available to enhance health and well-being, taking into account the person’s social context as well as medical aspects.

Keywords: LGMD2, parents, salutogenic, sense of coherence, young adults
We have to recognise that disablement (impairment) is not merely the physical state of a small minority of people. It is the normal condition of humanity. Sutherland, 1981
We have to recognise that disablement (impairment) is not merely the physical state of a small minority of people. It is the normal condition of humanity.

Sutherland, 1981
CONTENTS
Preface .................................................................................................................. 5
Abbreviations ....................................................................................................... 6
Original articles .................................................................................................... 7
Introduction .......................................................................................................... 8
  Background ....................................................................................................... 9
    Muscular dystrophy ................................................................................... 9
    Recessive limb-girdle muscular dystrophy ............................................ 9
    Experiences of living with muscular dystrophy ....................................... 11
    Young adults in transition to adulthood ................................................ 12
    Theories of health related to physical disability ...................................... 13
    The concept of well-being ....................................................................... 14
Theoretical framework ....................................................................................... 15
  The salutogenic theory ............................................................................. 15
  Person-centred care .................................................................................. 17
  Nursing .................................................................................................... 18
Rationale .......................................................................................................... 19
Aim ..................................................................................................................... 20
Method ............................................................................................................... 21
  Study design .............................................................................................. 21
  Participants ................................................................................................. 23
Data collection ................................................................................................. 26
  Semi-structured interviews ...................................................................... 26
  The self-administered SOC-13 questionnaire .......................................... 27
Data analysis .................................................................................................... 27
  Qualitative content analysis ..................................................................... 28
  Content analysis ....................................................................................... 30
  Phenomenography .................................................................................... 31
  Analysis of SOC scores ........................................................................... 31
Rigor ................................................................................................................. 31
Ethical considerations ..................................................................................... 33
Findings .............................................................................................................. 34
  Comprehensibility ...................................................................................... 34
  Manageability ............................................................................................ 38
  Meaningfulness .......................................................................................... 41
  Self-rated SOC ........................................................................................... 42
Discussion ........................................................................................................ 43
  Methodological discussion ....................................................................... 43
  Discussion of findings .............................................................................. 46
Conclusion ....................................................................................................... 51
Implications .................................................................................................... 53
Svensk sammanfattning .................................................................................... 55
Acknowledgements ............................................................................................ 58
References ......................................................................................................... 60
Twenty-five years ago, I made a choice to become a nurse because I wanted to support people who are ill to enhance their health. As an anesthetic and intensive care nurse I have cared over the years for many critically ill patients, including persons diagnosed with different forms of muscular dystrophies. I have tried to understand the patients' situation, sympathized with them, worried about them and done my best to support them. However, at the end of the day when my working shift was over, I closed the door to the patients and went on with my own life. Being a next of kin of a person living with muscular dystrophy I can never close that door. Instead, I have had to enter into a world I previously only had professional or theoretical knowledge about and I have found that the only choice of being a next of kin to a person diagnosed with muscular dystrophy is how to proceed. As I searched for more knowledge, first in the role of being a next of kin and then within the frame of the Master's programme in caring science, I realized the lack of information about experiences of living with the diagnosis recessive limb-girdle muscular dystrophy. Therefore, this thesis is my endeavour to contribute to an increased understanding about life with the disease from the perspectives of the affected persons and their parents.
Preface

Twenty-five years ago, I made a choice to become a nurse because I wanted to support people who are ill to enhance their health. As an anesthetic and intensive care nurse I have cared over the years for many critically ill patients, including persons diagnosed with different forms of muscular dystrophies. I have tried to understand the patients’ situation, sympathized with them, worried about them and done my best to support them. However, at the end of the day when my working shift was over, I closed the door to the patients and went on with my own life. Being a next of kin of a person living with muscular dystrophy I can never close that door. Instead, I have had to enter into a world I previously only had professional or theoretical knowledge about and I have found that the only choice of being a next of kin to a person diagnosed with muscular dystrophy is how to proceed. As I searched for more knowledge, first in the role of being a next of kin and then within the frame of the Master’s programme in caring science, I realized the lack of information about experiences of living with the diagnosis recessive limb-girdle muscular dystrophy. Therefore, this thesis is my endeavour to contribute to an increased understanding about life with the disease from the perspectives of the affected persons and their parents.
Original articles


IV. Aho A.C., Hultsjö S. & Hjelm K. Perceptions of the transition from receiving the diagnosis recessive limb-girdle muscular dystrophy to becoming in need of human support and using a wheelchair (submitted)

Permission to reprint the articles has been obtained from the respective journals.

Abbreviations

DMD Duchenne Muscular Dystrophy
GRR General Resistance Resources
GRR-RD Generalized Resistance Resources – Resistance Deficits
HRQOL Health-Related Quality of Life
LGMD Limb-Girdle Muscular Dystrophy
LGMD1 Dominant Limb-Girdle Muscular Dystrophy
LGMD2 Recessive Limb-Girdle Muscular Dystrophy
MD Muscular Dystrophy
QoL Quality of Life
RD Resistance Deficit
SFS Svensk Författningssamling
SOC Sense of Coherence
SOC-13 Sense of Coherence Questionnaire 13-item
SOC-29 Sense of Coherence Questionnaire 29-item
SRR Specific Resistance Resources
WHO World Health Organization
WMA World Medical Association
Original articles


IV. Aho A.C., Hultsjö S. & Hjelm K. Perceptions of the transition from receiving the diagnosis recessive limb-girdle muscular dystrophy to becoming in need of human support and using a wheelchair (submitted)

Permission to reprint the articles has been obtained from the respective journals.
Introduction

The muscular dystrophies (MD) are a group of disorders that involve progressive muscle weakness and increased need of human support to manage daily life, which affects not only the person but the whole family (Emery 2008). Recessive limb-girdle muscular dystrophies (LGMD2) refer to a group of MDs that are genetically and clinically heterogeneous but have in common an involvement of the proximal musculature in the shoulder and pelvic girdle (Rosales and Tsao 2012). LGMD2 can develop into severe physical impairment, negatively affecting health over time. Thereby, in a time of life when affected young adults are supposed to strive for autonomy, their ability to perform physical activities of daily living may decline. As the disease progresses the person will need support from an interdisciplinary healthcare team and from staff in the municipality in order to manage daily life. Recent research has increased the knowledge about genetic diagnosis and management of LGMD2 (Straub and Bertoli 2016, Narayanaswami et al. 2014, Mitsuhashi and Kang 2012, Rosales and Tsao 2012). There is, however, a lack of knowledge about experiences and perceptions of living with the disease, and the literature review revealed no study in the area. At present, there is no cure available and the focus of healthcare must be to support the affected person to optimize health and well-being. The salutogenic theory introduced by Antonovsky (1987) focuses on what causes 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 (SOC). In this thesis, the overall objective, using a salutogenic framework, was to develop knowledge about experiences and perceptions of living with LGMD2 and its influences on health, from the affected young adults’ and the parents’ perspectives. Increased knowledge and understanding among healthcare professionals may facilitate the dialogue and the cooperation with the person, including the parents, and support provided may thereby be optimized.
Background

Constantly coping with a progressive disease poses not only organizational but also existential difficulties for the person, and the threat of permanent dependency may strip away earlier self-definitions based upon independence (Charmaz, 1997). Young adults living with LGMD2 not only have to face the transition to adulthood but may also have to cope with declined physical abilities and increased need of assistive devices and human support to manage daily life. Increasing the knowledge about people’s experiences of living with a disease, in order to support them to optimize health and well-being, is a core in caring science (Meleis 2012) and this thesis is, to the best of our knowledge, the first that focuses on young adults living with LGMD2 and their parents.

Muscular dystrophy

The MDs are a group of inherited diseases in which various genes controlling muscle functions are defective and cause muscle wasting and weakness (Emery 2008). Persons living with various forms of MDs are found throughout the world. A precise genetic diagnosis is essential as it enables accurate follow-up controls, the prevention of known possible complications, and genetic counselling (Emery 2008). As the disease progresses, affected persons may need to be cared for, not only by an interdisciplinary healthcare team specializing in neuromuscular disorders, but also by healthcare professionals working, for instance, at medical, surgical and orthopaedic hospital wards or within home and primary healthcare.

Recessive limb-girdle muscular dystrophy

LGMD2 comprises a genetically and clinically heterogeneous group of muscular disorders that previously was diagnosed by exclusion (Mahmood and Xin Mei 2014). It was not until 1995 that the European Neuromuscular Centre Workshop established more precise criteria for the diagnosis and the classification of different subtypes of LGMD2, based on their genetic characteristics (Bushby 1995). Today, over 20 different forms of LGMD2 have been identified and although individually rare, all the different subtypes of LGMD2 together form an important group among the MDs (Straub and Bertoli 2016).

The LGMD2s are autosomal, which means that both males and females can be affected (Emery 2008). Hereditably and genetically, the LGMD2s differ from Duchenne MD (DMD) which is the most common childhood MD that affects boys with symptom debut at the age of 2–5 years (Manzur et al. 2008). The age of onset of LGMD2 may vary from early childhood to adulthood (Norwood et al. 2007). 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 walking ability may vanish and
a wheelchair will be needed for ambulation (Rosales and Tsao 2012). Intellect is, however, unaffected (Emery 2008). Disease severity and rate of progression varies, not only between different forms of LGMD2 but also within the same subtype of LGMD2. Cardiac and respiratory complications may also arise in some of the subtypes of LGMD2 (Rosales and Tsao 2012). Thus, some persons may be as severely affected as individuals living with DMD, and have reduced life expectancy, whereas others have late onset and mild progression (Norwood et al. 2007). This means that the prognosis for persons diagnosed with LGMD2 is not uniform. The importance of early identification and treatment of potential complications to improve well-being and survival is thereby emphasized (Norwood et al. 2007).

In this thesis, the subtypes 2A, 2B, 2E and 2I are represented. LGMD2A is the most common form of LGMD2 with a mean age of symptom debut in the early teens (Norwood et al. 2007). Usually loss of ambulation occurs 10–30 years after the onset of muscular weakness (Rosales and Tsao 2012). The mean age of onset for LGMD2B is 20 ± 5 years. The person usually has normal sporting ability until an abrupt onset of difficulties appears (Norwood et al. 2007). In LGMD2E and LGMD2I, there is variation in the age of the first symptoms and the severity of disease progression. Cardiac involvement may also arise (Rosales and Tsao 2012).

Depending on disease severity, there is a variation in support needed for the affected person to manage daily life. There is currently no curative treatment for LGMD2 (Straub and Bertoli 2016) and although the benefit of steroids has been reported in some of the subtypes (Nigro et al. 2011), treatment remains supportive. In order to provide care efficiently, the importance of genetic diagnosis of LGMD2 has been described (Narayanaswami et al. 2014) which can be referred to as personalized medicine (Josko 2014). Management of the disease involves emotional and physical support, such as assistive devices and surgical interventions, as well as identification and treatment of complications which may arise (Mahmood and Xin Mei 2014). In addition, physiotherapy and stretching are recommended to prevent contractions and promote walking (Nigro et al. 2011). As the disease progresses, healthcare professionals should also proactively anticipate and facilitate decision making regarding, for instance, the need for wheelchair and assistance with activities of daily living (Narayanaswami et al. 2014). Furthermore, the individual should be allowed to talk about feelings and experiences of living with the disease and the importance of enabling the person to have open discussions with relatives, friends and professionals involved in management of the disease is emphasized (Emery 2008). There is, however, a gap in knowledge about experiences of living with the disease.
Experiences of living with muscular dystrophy

Previous qualitative research on various aspects of living with MD is sparse and has predominantly focused on adolescents (Pehler and Craft-Rosenberg 2009, Parkyn and Coveney 2013), adults (Boström and Ahlström 2004, Rahbek et al. 2005) and young men living with DMD (Dreyer et al. 2010, Hamdani et al. 2015). However, studies focusing on young adults living with LGMD2 and their parents are lacking.

Experiences of deterioration of physical capacity over 10 years among adults diagnosed with various forms of MD have been described, as well as psychosocial consequences caused by the disease, such as changes in appearance, loss of key roles and experiences of stigma as physical impairment became more obvious (Boström and Ahlström 2004). Among adolescents living with DMD, deterioration of physical capacity may manifest as experiences of longing for missed activities and relationships and wishing to be seen as a person (Pehler and Craft-Rosenberg 2009) and the value of social interaction in a group for adolescent boys with MD has been shown (Parkyn and Coveney 2013). Experiences of pain and longing for love have been described among adult men living with DMD but also experiences of having good quality of life (QoL) in terms of not worrying about the disease or the future and positive assessment of income, hours of personal assistance, housing, years spend in school and ability to perform in desired activities (Rahbek et al. 2005). Life with home mechanical ventilation among young men diagnosed with DMD and the need for individualized tailored care have also been described (Dreyer et al. 2010) as well as the need for palliative care services in families of males living with DMD (Arias et al. 2011).

Living with MD does not only affect the person but also next of kin (Boström et al. 2006, Emery 2008). Parents of boys diagnosed with DMD have been described as perceiving the disease in three ways: as a severe loss; as a call to adapt; and as a way to rediscover the child. Also, parents may hope for cure, the child’s well-being and to see their child becoming a whole person (Samson et al. 2009). Parents of adult persons diagnosed with MD have been described as perceiving that they have more responsibilities to support their children and to provide for their needs than do parents with healthy children (Boström et al. 2006). The parents thereby may experience a substantial caregiver burden in terms of having to be available all the time, but they also value caregiving as important and rewarding (Pangalila et al. 2012). Positive aspects of caring for young people with MD have been more recognized by parents who perceived that they received a higher level of professional support and social support from friends and/or partners (Magliano et al. 2014). Among female caregivers, stress as well as life satisfaction have been associated with social support, resiliency, income and form of MD (Kenneson and Bobo 2010).
The transition to adulthood has been shown to be a challenging time for young men living with DMD and their parents (Abbott and Carpenter 2014) and the parents have been found to have an important role in supporting their sons’ transition (Yamaguchi and Suzuki 2015). Successful transition to adulthood, from the perspectives of young men diagnosed with DMD, have been described in terms of becoming as independent as possible, approximating normal life trajectories, and planning for future adulthood (Hamdani et al. 2015).

**Young adults in transition to adulthood**

Transition is a process that is triggered by a change and can be viewed as a passage from one state to another (Meleis 2010). The developmental transition to adulthood involves biological growth, physical as well as cognitive, and normatively governed psychosocial maturations. The late teens through the twenties have been described as the transitional period leading from adolescence to adulthood (Arnett 2004). It is viewed as a self-reflective period of time when the young adults think about who they are and what they want to get out of life. Having left formal school but not yet entered the enduring responsibilities that are normative in adulthood, such as stable occupation, marriage and parenthood, young adults often explore a variety of possible life directions in love, work and house making. It is described not only as a time of opportunities and high expectations but also as a time of anxiety and uncertainty because the lives of the young persons are often unsettled. The main criteria for gradual transition into adulthood are accepting responsibility for oneself, making independent decisions and becoming financially independent (Arnett 2004).

Young adults and their parents often view each other as persons and not merely as children and parents. These changing perceptions on both parties allow them to establish a new relationship, as friends and companions (Arnett 2004). Young adults may provide emotional and practical support to their parents (Cheng et al. 2015), who often have a chance to take a second look at their own lives and reassess, for instance, their work and dreams for the future (Arnett and Fishel 2013).

Young adults living with chronic conditions and their parents also have to face the young adults’ transition from paediatric to adult healthcare (Aldiss et al. 2015, Joly 2015). This transition can be viewed as a process that is influenced by the healthcare system and the social context of the affected person (Chu et al. 2015). Young adults’ experiences of this transition seem to be comparable across diagnosis. It involves, for instance, loss of familiar surroundings and relationships combined with feelings of insecurity but also experiences of achieving responsibility (Fegran et al. 2014) and wanting to be part of the process with support from providers who listen and are sensitive to their needs (Betz et al. 2013). Although the transfer forces movements towards independence, the young adults may in many ways still be dependent on their
parents (Fegran et al. 2014). Preparation for the transition should therefore start early and focus on strengthening the persons’ independence without undermining parental involvement (Chesshir et al. 2013, Aldiss et al. 2015). However, feelings of being abandoned by the healthcare team and experiences of loss, fear and uncertainty have been described by parents who felt that the transition process was facilitated by their own resourcefulness, family support and ability to establish new relationships within the adult healthcare setting (Davis et al. 2014). The transition to adult healthcare is hence a challenge, not only for the young adults and their parents but also for healthcare professionals, and it requires coordination between paediatric and adult healthcare as well as interdisciplinary teamwork (Nehring et al. 2015, Ciccarelli et al. 2015). Beyond the support from an interdisciplinary healthcare team, the young adults also need support from authorities in society that provide services to people living with chronic and disabling conditions (Joly 2015), who in general are less likely than their peers to get married, achieve higher education, be employed and have independent living, which may negatively affect health and well-being (Blum 2005).

Theories of health related to physical disability

The goal for health and medical services is good health and care on equal terms for the entire population according to the Swedish Health and Medical Services Act (Svensk författningssamling 1982:763). For people living with chronic diseases and disability, healthcare professionals need to concentrate on supporting the person to optimize health and well-being. In this thesis, the concept of disability refers to the International Classification of Functioning, Disability and Health, where a person’s functioning is conceived as a dynamic interaction between health conditions and contextual factors, which includes both personal and environmental factors (World Health Organization 2002). The central question that arises in connection with this thesis is; what does good health mean related to a person living with a chronic disease and physical disability? The answer to this question varies, however, depending on which theory of health is referred to. Among several different theories to define health, two main perspectives are evident: the biostatistical and the holistic perspectives.

The biostatistical perspective views health as the absence of disease (Boorse 1977). Thus, a healthy human body is one in which every organ makes at least its species-typical contribution to the goals of survival and reproduction. Having a disease, caused by pathological processes in the body that reduce normal physical functions and increase the risk of reduced longevity due to complications, is not compliant with having good health according to this perspective.

The holistic perspectives of health involve the whole person. The concept of well-being was introduced in WHO’s definition of health in 1948. Health was there viewed as “a state of complete physical, mental and social well-
being and not merely the absence of disease or infirmity” (WHO 2014 p.1)
This definition is often seen as wide and utopian, since the word “complete” makes it unlikely that anyone would be regarded as being healthy for a reasonable period of time, but it can still be viewed as a goal to strive for.

In caring science, the concept of health refers to objective as well as subjective dimensions and not merely to the absence of disease or disability (Eriksson 1996). The objective dimensions are in general measurable and involve integrated biological, psychological and social functions whereas the subjective dimension comprises the person’s own feelings of well-being.

There are also philosophical theories of health. The holistic theory of health presented by Nordenfelt (2007), for instance, claims that a person is healthy if the person is in a bodily and mental state which is such that the person has the ability to realize vital goals (Nordenfelt 2007). A person diagnosed with a chronic disease, who adjusted vital goals to ability, can thus be considered to have good health despite functional impairments. Criticism of this theory is that it is relativistic on the individual level and leads to counter-intuitive results. For instance, a person with low degree of ability and low ambition will be considered to have good health whereas a person with physical and mental abilities and high ambition might be regarded as having low health if his or her vital goals are not reached (Tengland 2007).

When discussing health and physical disability, the holistic two-dimensional theory of health presented by Tengland (2007) refers to abilities and health-related well-being. Health means having the abilities and the dispositions which people typically develop in their cultures, and being able to use those in acceptable circumstances. Health also means having a subjective experience of well-being whereas the opposite is suffering (Tengland 2007). People living with a chronic disease can thus experience having good health despite disability. However, they cannot be regarded as fully healthy due to reduced abilities, and this primarily has to do with the justification that society needs to support persons with disabilities to manage daily life (Tengland 2007). In the context of healthcare, this theory highlights the importance of focusing, not only on objectively measurable pathological processes in the body or physical abilities but also on the individual’s subjective feelings of well-being.

The concept of well-being
The ultimate goal of nursing is to support the person to maintain and promote health and well-being (Meleis 2012). The concept of well-being is, however, multidimensional and difficult to define (Dodge 2012). It can be viewed as a general term that constitutes a good life and involves all areas in life including physical, mental and social dimensions (WHO 2002). Well-being can thereby be viewed as a centre of physical, psychological and social equilibrium, which in turn can be affected by life events or challenges (Dodge 2012). Subjective well-being has also been described as a person’s cognitive and affective...
evaluations of his or her life. These evaluations comprise emotional reactions to events as well as cognitive judgements of satisfaction and fulfilment. Well-being thus means experiencing pleasant emotions, low level of negative moods, and high life satisfaction (Diener et al. 2002).

Philosophical discussions of well-being are often based on the distinction between three kinds of conceptions of the good life. It includes the hedonistic, the desire-fulfilment, and the objective pluralism theory (Brulde 2007a). The hedonistic theory means that a good life is identical with the pleasant life and what is best for the person is what makes the person’s life happiest. To have a good life is thus to feel good. The desire-fulfilment theory argues that a person has a good life if the person lives the life he or she wants to live. The only thing that has positive final value for a person is that his or her intrinsic desires are fulfilled, and the only negative final value for the person is that his or her intrinsic aversions are fulfilled. Being diagnosed with a chronic disease such as LGMD2 may, for instance, imply the fulfilment of an intrinsic aversion for the affected person and their next of kin. This in turn will negatively influence their well-being according to the desire-fulfilment theory. The objective pluralism theory means that there are several objective values, besides pleasure and happiness, which make a life good for a person, regardless of how the person views these values. Examples of objective values are knowledge, friendship, love, functioning well, personal development and having meaningful work. Therefore, having a good life and experiencing well-being means having these values present to a high degree (Brulde 2007a).

Well-being can also be regarded as being a mix of the above-mentioned theories (Brulde 2007a, Brulde 2007b). In this thesis, well-being is thus viewed as the person’s subjective experiences of feeling well, being happy and being satisfied with life as a whole, taking into account traditional objective values.

**Theoretical framework**

The salutogenic theory presented by Antonovsky (1987) was chosen in this thesis as a theoretical framework to describe how daily life can be comprehended, managed and found meaningful when living with LGMD2, from the affected young adults’ and their parents’ perspectives. The foundation on which this thesis is based is caring science with a focus on person-centred care and nursing.

**The salutogenic theory**

The salutogenic theory focuses on what causes a person’s movement towards health on the health ease/dis-ease continuum rather than what is the aetiology of the disease (Antonovsky 1987). Where on this continuum a person is located depends on the person’s SOC, which consists of the three components;
comprehensibility, manageability and meaningfulness. Comprehensibility refers to whether or not the person finds that inner and outer stimuli make sense in terms of being coherent, ordered, structured and clear. Manageability refers to the person’s belief that internal and external resources to cope with and handle a situation are available or not. Meaningfulness refers to areas in life that are important for the person and the perceptions of whether or not life’s difficulties are worth an investment of energy and engagement. In order to experience health, people thus need to understand their lives and they need to be understood by others, believe that they are able to manage the situation and perceive that it is meaningful enough to find the motivation to continue.

Life experiences that are characterized by lack of coherence, under- or overload and a sense of not being able to influence the situation can be viewed as stressors that may negatively affect the person’s SOC (Antonovsky 1987). Being diagnosed with a progressive disease such as LGMD2 can be regarded as a stressor that the person and their next of kin constantly need to cope with. Coping is the individual’s effort to manage life conditions that are stressful and it is influenced by the person’s goals, beliefs about the self and the world and personal resources (Lazarus 2006). The salutogenic theory does not disregard the fact that an individual has been diagnosed with a disease but relates to all aspects of the person when questioning how the person can be helped to maintain or to move towards enhanced health (Antonovsky 1996). Any characteristics of the person, the group or the environment that can facilitate effective stress management are viewed as Generalized Resistance Resources (GRR) according to Antonovsky (1987). It involves all potential resources that the person is able to mobilize and use in order to cope with difficulties, including, for instance, material resources such as money, knowledge and social support. The GRR contribute to provide the person with coherent and meaningful life experiences that are characterized by consistency, participation in decision making and balance in underload-overload. These experiences may in turn strengthen the person’s SOC (Antonovsky 1987).

All the GRR can be viewed on a continuum. Antonovsky (1987) thus merged the concept of GRR with the concept of stressors and combined them into one concept: Generalized Resistance Resources – Resistance Deficits (GRR-RD). A person who is high on the continuum tends to have consistent, balanced life experiences and participation in decision making whereas a person who is low on the continuum tends to have inconsistent, low-balanced life experiences and low participation in decision making. There is also a dynamic relationship between SOC and GRR-RD (Idan et al. 2017). This means that GRR-RD may contribute to a person’s level of SOC but the level of SOC may also contribute to mobilize GRR for enhancing of stress management. Thereby, when faced with a stressor, a person with a strong SOC will chose the specific coping strategy that seems to be the most suitable and is therefore more likely to embrace a healthy adaptive behaviour when
afflicted by disease, e.g. to search for care and follow prescriptions, compared with a person with weak SOC. Assessment of a person’s SOC can be made with the use of the self-administered SOC questionnaire (Antonovsky 1987).

In addition to GRR, there are Specific Resistance Resources (SRR) that contribute to enhance the person’s stress management (Antonovsky 1987). The SRR are instrumentalities whose meanings are defined in terms of the particular stressors they are invoked to manage (Mittelmark et al. 2017). Nursing can thereby be viewed as a GRR while the nurse providing help with a particular problem is a SRR (Sullivan 1989).

Person-centred care

The goal for health and medical services is good health based on the person’s needs (SSF 1982:763). Person-centred care includes three basic components: the patient’s narratives about his or her own life situation which shift the focus from what a patient is to who a person is; partnership between patients and caregivers which involves shared information and decision making; and documentation in patient records to safeguard partnership (Ekman et al. 2011).

The existential philosophy that forms the base in person-centred care is the foundation on which ontological and epistemological assumptions in this thesis rest. Thereby, human persons must be understood as unities of embodied souls or ensouled bodies (Smith 2010). Ontologically, this means that a human being exists both as material body and immaterial “soul” in singular unity. The human body is composed of a number of elements and it is from the relation or interaction of bodily parts that human causal capacities and personhood emerges.1 Epistemologically, monitoring and measurement of vital bodily elements and/or functions can provide important knowledge about what a human being is (Smith 2010). For instance, in personalized medicine the person’s genetic profile is used to guide decisions made in regard to the prevention, diagnosis, and treatment of the disease (Josko 2014). The importance of genetic diagnosis of LGMD2 is evident in order to optimize treatment, anticipate complications and determine the heredity of the disease (Narayanaswami et al. 2014). Furthermore, gene therapy may be used to develop novel therapies (Mitsuhashi and Kang 2012). Personalized medicine, as a base for healthcare professionals’ knowledge about the disease and treatment, is therefore essential in person-centred care for individuals diagnosed with LGMD2. However, while the physical body appears in the unique shape of the body and the sound of the voice, it is in acting and speaking that humans show who they are and their unique personal identity is revealed (Arendt 1989). Therefore, to gain understanding about who a person

1 Emergence refers, according to Smith (2010), to the process of constituting a new entity with its own specific characteristics through the interactive combination of other, different entities that are necessary to create the new entity but that do not contain the characteristics present in the new entity. Thus, the whole becomes more than the sum of its parts.
LGMD2 refers to a heterogeneous group of progressive muscular disorders that are presented worldwide and that may manifest in severe physical impairments and increased need of human aid to manage daily life (Rosales and Tsao 2012). The disease affects not only the person but the whole family and currently there is no cure available. The situation for young adults diagnosed with LGMD2 and their parents is complex considering the progressivity of the disease that coincides with the young adult’s transition to adulthood.

Through the years, increased knowledge about genetic diagnosis and management of LGMD2 has improved the care for persons living with LGMD2 (Straub and Bertoli 2016). The importance of emotional and practical support, assistive devices, physiotherapy, surgical interventions as well as early identification and treatment of complications has thereby been described (Mitsuhashi and Kang 2012). Healthcare professionals are thus important resources when it comes to supporting the persons’ management of the disease and its consequences. Nurses have an important role in identifying physical and psychological difficulties or needs that the person faces due to the disease in the context of their social and material environment (Meleis 2012). There is, however, a gap in knowledge about experiences of living with the disease. This thesis contributes new knowledge, not only by describing experiences and perceptions of living with LGMD2 from the perspectives of affected young adults and their parents but also by using the salutogenic theory developed by Antonovsky (1987) as a theoretical framework. Increased knowledge among healthcare professionals, based on the salutogenic theory, is valuable as it may enhance their understanding for the young adults’ and their parents’ situation. This in turn may not only facilitate dialogue and cooperation between healthcare professionals and the person, including the parents, but also enable support provided to promote health and well-being to be optimized. On a personal level, this thesis can benefit persons living with LGMD2 and their next of kin, who may be strengthened by the fact, although living with a rare disease, they are not alone in coping with LGMD2.

Nursing

The discipline of nursing deals with persons who are in constant interaction with their social and material environment and have unmet needs, are not able to care for themselves or do not adapt to the environment due to interruptions in health (Meleis 2012). Nursing focuses on support in meeting the needs of the person and enhancing adaptation capability, self-care ability, health and well-being.

The ongoing relationship with nurses who provide around-the-clock care at hospitals often prompts recipients of care to share their experiences in narrative dialogues that make their health and illness experiences more understandable in the context of their goals, social relationships and daily activities. This in turn enables congruent plans for action. Nurses thereby tend to get to know their recipient of care more profoundly than do other healthcare providers. They also often monitor and coordinate care provided by others in the healthcare team (Meleis 2012).

Nurses also support persons who are experiencing, anticipating or completing an act of transition (Meleis 2010). Transition denotes a change in health status, in role relationships, in expectations or in abilities and requires the person to incorporate new knowledge, to alter behaviour and to change the definition of self in a social context. In this thesis, the young adults were in transition to adulthood (Arnett 2004) which also affects the parents (Arnett and Fishel 2013).

Nurses thus support the person to manage their disease and their life transitions. The importance of utilizing available resources and creating new resources is thereby described (Meleis 2012), which can be viewed as GRR according to the salutogenic theory (Antonovsky 1987).
Rationale

LGMD2 refers to a heterogeneous group of progressive muscular disorders that are presented worldwide and that may manifest in severe physical impairments and increased need of human aid to manage daily life (Rosales and Tsao 2012). The disease affects not only the person but the whole family and currently there is no cure available. The situation for young adults diagnosed with LGMD2 and their parents is complex considering the progressivity of the disease that coincides with the young adult’s transition to adulthood.

Through the years, increased knowledge about genetic diagnosis and management of LGMD2 has improved the care for persons living with LGMD2 (Straub and Bertoli 2016). The importance of emotional and practical support, assistive devices, physiotherapy, surgical interventions as well as early identification and treatment of complications has thereby been described (Mitsuhashi and Kang 2012). Healthcare professionals are thus important resources when it comes to supporting the persons’ management of the disease and its consequences. Nurses have an important role in identifying physical and psychological difficulties or needs that the person faces due to the disease in the context of their social and material environment (Meleis 2012). There is, however, a gap in knowledge about experiences of living with the disease.

This thesis contributes new knowledge, not only by describing experiences and perceptions of living with LGMD2 from the perspectives of affected young adults and their parents but also by using the salutogenic theory developed by Antonovsky (1987) as a theoretical framework. Increased knowledge among healthcare professionals, based on the salutogenic theory, is valuable as it may enhance their understanding for the young adults’ and their parents’ situation. This in turn may not only facilitate dialogue and cooperation between healthcare professionals and the person, including the parents, but also enable support provided to promote health and well-being to be optimized. On a personal level, this thesis can benefit persons living with LGMD2 and their next of kin, who may be strengthened by the fact that, although living with a rare disease, they are not alone in coping with LGMD2.
Aim

The overall aim of this thesis, using a salutogenic framework, was to develop knowledge about experiences and perceptions of living with recessive limb-girdle muscular dystrophy and its influences on health, from the affected young adults’ and their parents’ perspectives. The specific aims, from a salutogenic perspective, were to describe:

- Young adults’ experiences of living with recessive limb-girdle muscular dystrophy (I).
- Health perceptions related to sense of coherence among young adults living with recessive limb-girdle muscular dystrophy (II).
- Experiences of being parents of young adults living with recessive limb-girdle muscular dystrophy (III).
- 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 (IV).
Method

Study design

In order to describe experiences and perceptions of living with LGMD2 from the affected young adults’ and their parents’ perspectives, this thesis has a qualitative explorative and descriptive study design. This study design was chosen considering the aims of the studies in this thesis and that it is an uncharted area of research that needs to be investigated (Patton 2015). The salutogenic theory (Antonovsky 1987) was used to obtain an overall view of the participants’ situation when living with LGMD2.

This thesis comprises four studies that are based on data collected from 14 young adults diagnosed with LGMD2 and 19 of their parents. It includes qualitative data from semi-structured interviews and quantitative data from the self-administered 13-item Sense of Coherence (SOC-13) questionnaire. Semi-structured interviews were chosen as this provides an opportunity, not only for the participants to express themselves within a given frame (Patton 2015) but also for the interviewer to acquire knowledge about the participants’ personal experiences and perceptions (Marton and Booth 1997). The SOC-13 questionnaire (Antonovsky 1987) was used to mirror qualitative data. Thereby, the combination of qualitative and quantitative data enabled a more complete view of the participants’ situation (Patton 2015). An overview of the four studies in this thesis is presented in Table 1.
Table 1. Overview of the four studies in this thesis. The participants were: young adults (n=14) aged 20–30 years and diagnosed with recessive limb-girdle muscular dystrophy (LGMD2) and their parents (n=19).

<table>
<thead>
<tr>
<th>Study</th>
<th>Aim From a salutogenic perspective to describe:</th>
<th>Sample</th>
<th>Methods for data collection</th>
<th>Methods for data analysis</th>
</tr>
</thead>
<tbody>
<tr>
<td>I</td>
<td>Young adults’ experiences of living with LGMD2</td>
<td>Young adults diagnosed with LGMD2</td>
<td>Semi-structured interviews</td>
<td>Qualitative content analysis (Mayring 2000)</td>
</tr>
<tr>
<td>II</td>
<td>Health perceptions related to sense of coherence among young adults living with LGMD2</td>
<td>Young adults diagnosed with LGMD2</td>
<td>Semi-structured interviews, The SOC-13 questionnaire</td>
<td>Qualitative content analysis (Mayring 2000), Descriptive statistics</td>
</tr>
<tr>
<td>III</td>
<td>Experiences of being parents of young adults living with LGMD2</td>
<td>Parents of young adults diagnosed with LGMD2</td>
<td>Semi-structured interviews, The SOC-13 questionnaire</td>
<td>Content analysis (Patton 2015), Descriptive statistics</td>
</tr>
<tr>
<td>IV</td>
<td>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</td>
<td>Young adults diagnosed with LGMD2 and their parents</td>
<td>Semi-structured interviews</td>
<td>Phenomenography (Sjöström and Dahlgren 2002)</td>
</tr>
</tbody>
</table>
Participants

A purposeful sampling procedure was used to ensure information-rich participants (Patton 2015) who could share their experiences and perceptions of living with LGMD2. Invited to participate were young adults, aged 18–30 years and diagnosed with LGMD2, and their parents. Persons who could not take part in the study due to cognitive impairment or inability to speak Swedish were excluded.

The young adults diagnosed with LGMD2 (n=14) were recruited from hospitals in three different healthcare regions (n=10), from the Swedish association of persons living with neurological diseases and their next of kin, Neuro Sweden (n=1), and from a web-based association for people living with various disabilities and their next of kin (n=3). The managers at the hospital clinics involved and the chairman of Neuro Sweden gave their written approval for the study. A contact list of healthcare professionals working with persons diagnosed with LGMD2 was first compiled with the help of the development coordinator for habilitation centres in southern Sweden. The principal investigator (first author) contacted the healthcare professionals on the list, as well as key members in Neuro Sweden, by e-mail or phone, with a request to forward a letter with information about the study to potential participants. The information letter about the study was also sent by the principal investigator by e-mail to members of the web-based association for people living with disability, who in their presentation stated age 18–30 and specified form of LGMD2.

The parents (n=19) were invited to participate through an information letter about the study that was forwarded to them by their young adults who had already agreed participation. All the young adults had at least one parent who accepted participation in the study.

The young adults and the parents who were willing to participate sent their contact information, by post or e-mail, to the principal investigator, who then telephoned them to arrange a time and place for the interviews.

Thirty-three participants, 14 young adults and 19 parents from 13 different families, took part in the studies that form the base in this thesis. Demographic characteristics of the participants are presented in Table 2. There was a variation in the young adults’ physical functioning and need of support. Based on their descriptions, three groups were found regarding dependency on human aid and a wheelchair to manage daily life: those who were independent were physically active and managed daily life on their own; those who were in transition from being as independent a person as same-aged peers to becoming more dependent were receiving informal support from next of kin with activities such as clothing, cooking, laundry, shopping and cleaning, and some...
of them occasionally used a wheelchair; and those who were dependent were entitled to personal assistance and always used a wheelchair for ambulation. Some of the parents worked, occasionally or full-time, as personal assistants for their young adults. In the families represented in this thesis, all of the young adults’ parents were married, except two who were cohabiting with another person or a single parent.

In the families, there were five siblings diagnosed with LGMD2 who did not take part in the studies in this thesis. Three of them declined participation and two of them were not invited to participate due to the age criterion set for the studies. In 10 of the families there were siblings without the disease.
Table 2. Demographic characteristics of the participants

<table>
<thead>
<tr>
<th>Characteristics</th>
<th>Numbers (N=14)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Young adults</strong></td>
<td></td>
</tr>
<tr>
<td>Characteristics</td>
<td></td>
</tr>
<tr>
<td>Females</td>
<td>8</td>
</tr>
<tr>
<td>Males</td>
<td>6</td>
</tr>
<tr>
<td>Age (years)¹</td>
<td>25 (20–30)</td>
</tr>
<tr>
<td>Age of symptom debut (years)¹</td>
<td>11 (0.5–25)</td>
</tr>
<tr>
<td>Subtypes of LGMD2</td>
<td></td>
</tr>
<tr>
<td>LGMD2A</td>
<td>5</td>
</tr>
<tr>
<td>LGMD2B</td>
<td>3</td>
</tr>
<tr>
<td>LGMD2E</td>
<td>1</td>
</tr>
<tr>
<td>LGMD2I</td>
<td>5</td>
</tr>
<tr>
<td>Dependency on other people/wheelchair in daily life</td>
<td></td>
</tr>
<tr>
<td>Independent</td>
<td>2</td>
</tr>
<tr>
<td>In transition to become more dependent</td>
<td>5</td>
</tr>
<tr>
<td>Dependent</td>
<td>7</td>
</tr>
<tr>
<td>Living conditions</td>
<td></td>
</tr>
<tr>
<td>Living on their own</td>
<td>4</td>
</tr>
<tr>
<td>Cohabiting</td>
<td>4</td>
</tr>
<tr>
<td>Living with parents/other relatives</td>
<td>6</td>
</tr>
<tr>
<td>Education</td>
<td></td>
</tr>
<tr>
<td>Completed upper secondary school</td>
<td>10</td>
</tr>
<tr>
<td>Dropped out of upper secondary school</td>
<td>4</td>
</tr>
<tr>
<td>Finished a university education</td>
<td>3</td>
</tr>
<tr>
<td>Occupation</td>
<td></td>
</tr>
<tr>
<td>Working (full-time/part-time)</td>
<td>4 (3/1)</td>
</tr>
<tr>
<td>Studying full-time</td>
<td>4</td>
</tr>
<tr>
<td>Not working/studying</td>
<td>6</td>
</tr>
<tr>
<td><strong>Parents</strong></td>
<td></td>
</tr>
<tr>
<td>Characteristics</td>
<td></td>
</tr>
<tr>
<td>Females</td>
<td>13</td>
</tr>
<tr>
<td>Males</td>
<td>6</td>
</tr>
<tr>
<td>Age (years)¹</td>
<td>50 (47–65)</td>
</tr>
<tr>
<td>Occupation</td>
<td></td>
</tr>
<tr>
<td>Working (full-time/part-time)</td>
<td>17 (13/4)</td>
</tr>
<tr>
<td>Being retired</td>
<td>2</td>
</tr>
</tbody>
</table>

¹) Median (range)
**Data collection**

Data were collected between June 2012 and November 2013 through semi-structured interviews and the self-administered SOC-13 questionnaire.

**Semi-structured interviews**

Semi-structured interviews were conducted by the principal investigator, who is a nurse, and were held individually or with parental couples at a place chosen by the participants. A majority of the interviews were performed at the participants’ homes but one was conducted at the participant’s workplace and three at a neutral place. In families where two parents participated, they all chose to be interviewed together and both came to speak throughout these interviews.

An interview guide with semi-structured questions, based on the salutogenic theory (Antonovsky 1987), was formulated to focus on the participants’ comprehensibility, manageability and meaningfulness when living with LGMD2 or being a parent of an affected young adult. Considering the aims of the studies in this thesis, questions about the participants’ perceptions of receiving the diagnosis and their perceptions of health were also elicited. The main interview questions were:

- Could you describe what it means to live with LGMD2/being a parent of a young adult living with LGMD2?
- What did it mean to you when you/your son or daughter received the diagnosis?
- 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?
- What do you experience as the most meaningful in life?
- Can you describe what health means to you?’

Follow-up questions were asked to reach a deeper understanding of the participants’ experiences and perceptions of living with LGMD2, e.g. How do you mean? What do you think about that? and Could you give an example? 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. At the end of the interviews, questions about the participants’ demographic characteristics (that had not been clarified during the interviews) were asked. These were structured questions about age, form of LGMD2, age of symptom debut, living conditions, education and whether or not the young adult used a wheelchair, was entitled to personal assistance and used any form of breathing support. Each interview lasted about an hour, with an average time of 75 minutes. All the participants, except one parent, also answered the self-administered SOC-13 questionnaire at the end of the interview.
The self-administered SOC-13 questionnaire

The concept of SOC has been operationalized in the self-administered SOC questionnaire developed by Antonovsky (1987). In this thesis, the shorter SOC-13 questionnaire was used which is a condensed version of the original 29-item questionnaire (SOC-29). The SOC-13 questionnaire was chosen in order not to tire the participants. In addition, it does not include questions about the past 10 years and the future that could be difficult to answer for young adults diagnosed with a progressive disease, who may live with reduced life expectancy, and their parents. The SOC-13 questionnaire contains questions aimed at measuring how a person rates SOC in terms of comprehensibility, manageability and meaningfulness in daily life (Antonovsky 1987). The instrument has been found to be reliable, valid and cross-culturally applicable (Eriksson and Lindström 2005). The Swedish translation of the instrument that was used in this thesis (Antonovsky 2005) has been shown among students to be reliable with a Cronbach’s alpha of 0.89 (Olsson et al. 2009). The SOC-13 questionnaire has previously been used in a Swedish context for people living with various chronic diseases such as chronic obstructive pulmonary disease (Delgado 2007), Parkinson’s disease (Caap-Ahlgren and Dehlin 2004), type 2 diabetes (Sandén-Eriksson 2000), and congenital heart failure (Falk et al. 2007). The person is asked to mark each answer on a 7-point Likert scale with an extreme pole at each side. The score range is between 13 and 91. A high score represents a strong SOC (Antonovsky 1987).

Permission to use the questionnaire was obtained from the copyright holder.

Data analysis

All the interviews were recorded and verbatim transcribed by the first author. The transcribed text comprised a total of 450 A4 pages, with single line spacing (219 versus 231 pages from the young adults and the parents). The interview text was first read through several times to get a sense of the whole and become familiarized with the text. Because of the richness of the interview data, a decision was taken together with the two co-authors to divide the data from the interviews with the young adults into three articles: Experiences of living with LGMD2 (I); Health perceptions (II); and Perceptions of the transition from receiving the diagnosis to becoming in need of human support and a wheelchair (IV). Similarly, the interview data from the parents were divided into two articles: Experiences of being parents of young adults living with LGMD2 (III); and Perceptions of the young adult’s transition from receiving the diagnosis to becoming in need of human support and a wheelchair (IV).
Interview data were analysed using: qualitative content analysis (Mayring 2000) in studies I and II; content analysis (Patton 2015) in study III; and phenomenography as described by Sjöström and Dahlgren (2002) in study IV. All the qualitative data analyses were made in a dynamic process, working back and forth between the interview texts and the preliminary groupings of data. Throughout the data analysis, the content of the categories, themes and outcome space was checked to confirm its relevance by the two co-authors and continuous discussions were held until coder agreement was achieved to ensure the credibility of the studies (Patton 2015, Mayring 2000, Sjöström and Dahlgren 2002).

**Qualitative content analysis**

Qualitative content analysis is a method to analyse interview texts step by step according to Mayring (2000).

In study I, after each interview, content units were identified and coded inductively (Mayring 2000) in order not to leave out any data regarding the young adults’ experiences of living with LGMD2. Data collection and analysis thus proceeded concurrently. Codes were then compared and contrasted and those with similar content were brought together into preliminary subcategories. The data analysis then proceeded by using the Step model of deductive category application presented by Mayring (2000). Deductive categories refer to preformulated categories that are theoretically derived. The deductive categories used in this study were the concepts that form the base when describing a person’s SOC, namely: comprehensibility, manageability and meaningfulness (Antonovsky 1987). In order to determine under what circumstances a text passage could be coded with a deductive category, coding rules were defined: comprehensibility was viewed as a 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 was defined as a behavioural category which includes experiences of whether internal and external resources to cope are available or not; and meaningfulness was regarded as a motivational category which refers to experiences that bring joy and meaning in life. By organizing the interview text into the deductive categories, new subcategories were developed and named as closely as possible to the text (Mayring 2000). An example of data analysis in study I is presented in Table 3.
Table 3. Young adults’ experiences of living with recessive limb-girdle muscular dystrophy: an example of data analysis (I).

<table>
<thead>
<tr>
<th>Category</th>
<th>Subcategories</th>
<th>Transcribed text</th>
</tr>
</thead>
<tbody>
<tr>
<td>Meaningfulness</td>
<td>Engagement in meaningful activities</td>
<td><em>I have to do something that’s meaningful</em></td>
</tr>
<tr>
<td></td>
<td>Increasing people’s understanding of disability</td>
<td><em>I am deeply involved in political issues of accessibility and society’s outlook and attitudes</em></td>
</tr>
<tr>
<td></td>
<td>Setting and achieving goals</td>
<td><em>I have goals that I try to achieve all the time</em></td>
</tr>
</tbody>
</table>

In study II, the Step model for inductive content analysis presented by Mayring (2000) was used to analyse the interview data regarding the young adults’ health perceptions. This is a method used to reduce the text and form categories based on the material. The interview text was thereby read line by line and content units were marked and coded with names close to the text. The codes with closely related meanings were grouped together into tentative subcategories. The tentative subcategories were then revised, compared and contrasted and those with similar content were abstracted into categories. An example of data analysis in study II is illustrated in Table 4.

Table 4. Health perceptions among young adults living with recessive limb-girdle muscular dystrophy: an example of data analysis (II).

<table>
<thead>
<tr>
<th>Category</th>
<th>Subcategories</th>
<th>Transcribed text</th>
</tr>
</thead>
<tbody>
<tr>
<td>Health viewed as well-being</td>
<td>Physical and mental well-being</td>
<td><em>it (health) is about feeling well both physically and mentally</em></td>
</tr>
<tr>
<td></td>
<td>Well-being with the disease as part of life</td>
<td><em>it (the disease) is so normal now... everything works all right so I’m OK</em></td>
</tr>
</tbody>
</table>
Content analysis

Content analysis according to Patton (2015) was used in study III to analyse the interview data regarding experiences of being parents of young adults living with LGMD2. This is a method used to reduce the data and make sense of it by identifying the core meanings in the text and bringing them together into themes (Patton 2015). The interview text was first coded by reading it line by line and labelling different text sections with a code that described the content of the text. Subthemes were developed by grouping codes with similar contents together. The subthemes were then organized into the main areas that form the base of a person’s SOC, i.e. comprehensibility, manageability and meaningfulness (Antonovsky 1987). Throughout the data analysis, comprehensibility was defined as a cognitive component that refers to experiences of what it means being parents of young adults diagnosed with LGMD2; manageability was defined as a behavioural component which includes experiences of whether or not internal and external resources to cope are available; and meaningfulness was defined as a motivational component that refers to areas in life which are important and bring joy for the person. Themes were then developed within the frame of each main area. An example of data analysis in study III is seen in Table 5.

Table 5. Experiences of being parents of young adults living with recessive limb-girdle muscular dystrophy: an example of data analysis (III).

<table>
<thead>
<tr>
<th>Main area: Comprehensibility</th>
<th>Subthemes</th>
<th>Transcribed text</th>
</tr>
</thead>
<tbody>
<tr>
<td>Being influenced by the young adult’s disease</td>
<td>Thoughts and emotions tied to the young adult’s disease</td>
<td><em>Mother (M): it’s more of a general sorrow</em></td>
</tr>
<tr>
<td></td>
<td>Experiences of being influenced by the young adult’s well-being</td>
<td><em>M: it pains me when he is in pain</em></td>
</tr>
<tr>
<td></td>
<td>Experiences of having a caregiving burden</td>
<td><em>M: my husband and I are very tied</em></td>
</tr>
<tr>
<td>Difficulty in fully grasping the young adult’s disease</td>
<td>Uncertainty about disease progression and worries about the future</td>
<td><em>M: the worst thing is the worry, how will the disease progress?</em></td>
</tr>
<tr>
<td></td>
<td>Information not always shared by the young adult</td>
<td><em>Father (F): he doesn’t inform us about everything</em></td>
</tr>
<tr>
<td></td>
<td>Difficulties in fully comprehending the young adult’s situation</td>
<td><em>F: even those of us who are involved in it don’t really understand it</em></td>
</tr>
</tbody>
</table>
Phenomenography
In study IV, phenomenography was used for data analysis following the steps described by Sjöström and Dahlgren (2002). This is a method that focuses on various ways in which different people perceive something (Marton 2014). In study IV, the participants’ perceptions of the transition, from receiving the diagnosis to the young adult becoming in need of human support and using a wheelchair, were in focus. The most significant statements from each participant regarding this transition were identified in the interview text and compiled (Sjöström and Dahlgren 2002). In order to find the central parts of the text, the statements were condensed and preliminary grouping of similar statements were made. The statements were then compared to find variations in the participants’ perceptions. Through revision of the preliminary groups, five descriptive categories distinct from each other were established and named close to the interview text to illustrate their content (Sjöström and Dahlgren 2002). The three 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 regarded as a process that is overshadowed by the descriptive category – Concern about disease progression – and influenced by the descriptive category – Factors facilitating everyday life (IV).

Analysis of SOC scores
Quantitative data from the items of the SOC-13 questionnaire were calculated (Antonovsky 1987, Bengel et al. 1999) and analysed with descriptive statistics in terms of median SOC scores and range (II, III). The individual SOC scores were also related to the interview texts, by reading each text and identifying various patterns of perceptions which may illustrate the different levels of SOC score among the young adults (II) and the parents (III).

Rigor
Rigor refers to methodological means to ensure the adequacy and trustworthiness throughout the whole process of qualitative research (Patton 2015, Tobin and Begley 2004). The terms used are credibility, transferability, dependability and confirmability.

Credibility refers to the relevance of the methods that are chosen to answer the research inquires (Patton 2015). In this thesis, data triangulation was obtained not only by combining qualitative data from semi-structured interviews with quantitative data from the self-administered SOC-13 questionnaire but also by including both the young adults’ and their parents’ perspectives (Patton 2015). There was a variation in demographic characteristics among the participants. The young adults also belonged to different healthcare regions in Sweden and there was a variation in their form
of LGMD2 and disease progression. The young adults, as well as their parents, thereby represented a range of different experiences which is important in order to find various perceptions within a group of people (Marton and Booth 1997). Credibility also refers to the fit between the participants’ views of their life situation and the researcher’s presentation of them (Patton 2015, Tobin and Begley 2004). Examples of transcribed text are thereby presented along with the categories and the themes that were developed through data analysis and named close to the text (I, II and III). The competence of the researchers is also important to ensure study credibility (Patton 2015). In qualitative research, the investigator is also an instrument and reflexivity means emphasizing the importance of self-awareness throughout the research process. In this thesis, all the interviews were conducted by the principal investigator who is experienced in meeting and caring for persons living with chronic conditions, including MD, as an anaesthetic/intensive care nurse and being a next of kin to a person diagnosed with MD (who is not in the same age as those being interviewed in this thesis). The interviewer did not, however, have any professional or personal relation to the participants in this thesis. Throughout the research process, continuous discussions to enhance the credibility of the studies were held with the two co-authors (Patton 2015) who are experienced nurses and researchers trained in qualitative methods using content analysis as well as phenomenography.

Transferability concerns the investigator’s responsibility to provide the readers with sufficient information in order to establish the degree of similarity to other groups of persons in comparable contexts (Patton 2015, Tobin and Begley 2004). In qualitative research, the person’s subjective experiences and perceptions are central and there is no single correct or “true” interpretation of the data. The findings can therefore not be generalized. In this thesis, the participants’ demographic characteristics are clearly described. The young adults were diagnosed with LGMD2 and they, as well as their parents, lived in a Swedish context. In a comparable context, patterns of the findings might be transferable to young adults living with other chronic and disabling diseases and their parents.

Dependability refers to the importance of the research process being logical, traceable and documented (Patton 2015, Tobin and Begley 2004). In this thesis, the methods for data collection and data analysis are clearly described to ensure dependability of the studies.

Confirmability focuses on ensuring that the data and interpretations of them are not figments of the investigator’s imagination but are clearly derived from the data (Patton 2015, Tobin and Begley 2004). In the four studies that form the base of this thesis, illuminative quotations were presented in the findings to establish the confirmability of the studies.
Ethical considerations

Ethical approval for the studies in this thesis was obtained from the regional ethics board in Linköping and the studies were performed in accordance with the Declaration of Helsinki (World Medical Association 2013). The participants received written and orally given information about the study, that it was voluntary to participate and that they could withdraw their participation without explanation at any time. All the participants gave their written informed consent to take part in the study in connection with the interviews. Collected data were anonymized and coded with numbers. Data analysis and presentation of data were made at a group level. All the data were stored in a locked safety box to which only the principal investigator had access to. The participants’ identity was thereby concealed and all the data were treated confidentially throughout the research process.
Findings

An overview of the findings is presented in Figure 1.

Comprehensibility

The time around diagnosis was described as being a difficult period of time by the young adults and their parents (IV): “I can hardly talk about that time because it was really difficult.” Some of the participants perceived that the diagnosis had come as a shock and was difficult to comprehend. They had viewed themselves, or their young adult, as being healthy and/or had believed that a cure would be available when the cause of the symptoms was found. Realizing that it is a progressive disease with currently no cure available was described as being emotionally difficult. Receiving the diagnosis had for some of the participants been so overwhelming that they perceived not having been able to take in all the information given by healthcare professionals at the time. Lack of information and professional support to cope with the disease at the time of diagnosis was also described.

Although being emotionally difficult, receiving the diagnosis was felt as being a relief 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 the disease and follow-up controls at the hospital. The diagnosis was also used as a tool for information, not only when searching for information but also when informing people about the disease. Knowing the exact diagnosis based on genetics did not mean much for some of the participants, while others thought that it was important considering future progress in research and if a cure should be found (IV).
Figure 1. Experiences and perceptions of living with recessive limb-girdle muscular dystrophy (LGMD2) from the affected young adults’ and their parents’ perspectives, studied through a salutogenic framework: an overview of the findings.
As the disease progressed, physical and emotional consequences of living with LGMD2 were described (I): “So it’s bloody trying both physically and mentally.” A majority of the young adults (I) had experienced slow muscular weakening, resulting in vanished physical functions, which required continual adjustments to the body and the surroundings. Comprehending that it is a progressive disease, yet not really knowing what to expect was described as being difficult and it evoked many thoughts and emotions. Experiences of psychological distress were expressed among the young adults and some of them had gone through periods of depression (I). The young adults’ transition from walking to becoming in need of using a wheelchair was perceived to be a psychologically distressing process that involved major emotional, social and practical adjustments (IV). Among the parents (III), it was perceived as mentally difficult to see the young adult’s gradual physical deterioration and inability to perform activities that previously caused no problems. Frustration over not being able to do anything to stop the disease from proceeding was also described. Thoughts and emotions that were connected, not only to the disease but also to the young adult’s well-being were expressed. Comprehending that it is a progressive muscular disease was described in terms of sadness that the parents always carried with them. Some of the parents also expressed feelings of being torn between different obligations, such as supporting the young adult and the rest of the family, work duties and household requirements (III).

The variation between good and bad periods was also described (I, III). Living with LGMD2 was thus described by the young adults (I) not only as a struggle to get through the day but also as being a normal part of life. There were periods expressed as more difficult to cope with and periods experienced as physically and mentally stable. The parents (III) also described the variation between bad periods, when the disease intruded on everyday life, requiring all the attention, and good periods when the disease was viewed as being stable. Regardless of the young adult’s physical abilities, however, concern about how the disease would develop was expressed.

Uncertainty and concern about disease progression was described by the participants (I, III and IV): “We don’t know how fast the disease develops.” Among the young adults (I) there was a recognition that the disease had shaped who they are today and thus concern about who to become in the future was mentioned. While some of them did not expect any major disease progression, others expressed anxiety about what will happen and if support needed from society will be granted in the future. Reconciliation with the thought of slowly becoming weaker was also expressed (I). Among the parents (III), uncertainty about disease progression and concerns about what will happen when not being able to give support as a parent any longer was described as creating worries about the future.
The young adults (I) as well as the parents (III) perceived that there is limited information available about the diagnosis. They also recognized that the disease may develop differently for each individual affected. Although some of the participants were content with the information they had regarding the diagnosis, or preferred not to receive any more realizing that it would become worse, others wanted further information but did not really know where to find it. Some of the young adults (I) 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 parents (III) expressed difficulties in fully grasping the disease and the young adult’s situation. Some of them also experienced that information about the disease was not always shared by the young adults and this could create a sense of not knowing what was going on and uncertainty about how to support the young adult to enhance their health (III).

Health was described in terms of well-being (II, III): “well-being ... that you feel good.” The young adults (II) viewed health as a subjective experience of intertwined physical and mental well-being that involved a sense of being satisfied with life. Health-promoting factors were described in terms of having a balanced lifestyle regarding physical activity and food habits, social relations and meaningful daily pursuits. Similarly, health was viewed by the parents (III) as physical, mental and social well-being. Although concern was expressed about the young adults taking responsibility for their bodies, health was also described in terms of being able to live a good life under the conditions that are given, which involved, for instance, being able to take part in activities that are found joyful and the absence of pain or other diseases. Some of the young adults (II), however, described experiences of pain and being exhausted as well as complications from the heart and/or the respiratory system. Two of them used continuous positive airway pressure (CPAP) as breathing support during the night.

Although living with a chronic disease, there was among the young adults a sense of not being ill (II). The importance of mental well-being, rather than physical abilities, was thereby emphasized and the perception was that a subjective experience of feeling healthy can be attained regardless of physical impairments. This view was shared by some of the parents (III) who thought that their young adults were in good health despite having a disease and physical disability. As the disease progressed, health was perceived by the young adults (II) as being influenced not only by physical impairments and mental strain caused by the disease but also by external factors such as entitlement to support, accessibility in society and how they were treated by other people. While some of the young adults described being constantly reminded and inhibited by the disease and its consequences, others seemed to have achieved well-being with the disease incorporated as part of who they are (II).
Manageability

The participants described their efforts to make the best of the situation (I, III): “I try to make the best of the situation.” The young adults (I) recognized that since they cannot do anything about having the disease, they somehow have to learn to accept living with it and the importance of trying to think positively, focusing on possibilities and having joyful things to look forward to was described. Living with LGMD2 had given them experiences of how their body works and how to manage in order to feel well. Experiences of having found new creative ways to perform activities in daily life differently were also expressed. Every activity, however, had to be planned for, taking into account physical abilities as well as the surroundings (I). The parents (III) tried their best to support their young adults, emotionally as well as practically, and experiences of having learned how to manage everyday life were described. Focusing on solutions rather than on problems, actions could be taken to solve difficulties in daily life. Sometimes, however, thoughts and emotions tied to unsolved problems had to be repressed to manage the situation. Recognizing that their children had become young adults and needed to have a life of their own, the parents described the balancing between being available to support but without being overprotective. Some of the parents found strength in God. Experiences of having an “internal power” that made it possible to manage more than first expected were also described (III). Several of the participants also hoped for researchers to find a cure (I, III).

Although disease progression was perceived to cause physical, emotional, practical and social difficulties, a sense of having learned to cope with and to live with the disease was also described (IV). New ways of living were found that involved personal assistance and the young adult always using a wheelchair for ambulation. Management was, however, also perceived to be affected by other people’s attitudes (IV).

Perceptions of being influenced by other people’s attitudes as the disease became more obvious were also described (IV): “it’s a physical limitation ... but a lot of the reason you’re affected by it is other people’s attitudes.” Some of the young adults felt that it was easier if people knew about the disease and they had chosen to be open about it. Others expressed feelings of shame over physical limitation. They described how they had tried to hide the disease from other people as long as possible by avoiding activities that would reveal physical limitation. The parents, who thought it was difficult to see their young adult’s struggle, tried to encourage them to tell people about how it is. Beginning to use a wheelchair was perceived to influence, not only how the young adults viewed themselves but also how they were viewed by other people. Although the participants thought that people’s attitudes in society towards persons using a wheelchair have become better through the years, experiences of the young adults being stigmatized were also described, for
instance, people talking above their head. It was also expressed as difficult when people felt sorry for them because it was perceived as reducing them and their situation. The wheelchair was thereby 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. Although beginning to use a wheelchair was perceived to be emotionally difficult, it also provided a new freedom. An electric wheelchair thus enabled independent ambulation and participation in activities. It was thus viewed as a great assistive device that enabled the young adult to live a fulfilled life (IV).

The importance of having informal social support in order to facilitate management was highlighted by the participants (I, II, III, and IV): “we have such a big social network that it works out ... it solves itself.” Practical as well as emotional support from family and friends was thereby perceived as essential in order to manage daily life. The participants also thought that being able to talk about feelings and to process different things together helped them to cope with their situation. The young adults experienced great support from their parents (I) who could feel strengthened and comforted by the young adults (III). Some of the parental couples felt that they were able to share many things together but the importance of giving each other space was also recognized. Other parents expressed lack of support from spouse and experiences of crying and screaming when being alone were described (III). Some of the participants were active in interest organizations and had found it valuable to meet and exchange experiences with persons who were in similar situations (I, III).

The need for external support from society in order to manage daily life was also described by the participants (I, III): “it (personal assistance) really is a way to be independent even if it’s with somebody else’s help.” The participants expressed an appreciation for living in a time and in a country that provide highly qualified healthcare and social welfare including personal assistance. They perceived, however, that they often had to struggle in order to receive support needed. There was a variation in the support most needed by the young adults (I). In addition to medical aspects, the need for e.g. assistive devices, physiotherapy, financial support, personal assistance and adaptation in the home and car adaptation was described. Many of the young adults (I) thereby had contact with several different professionals, not only from healthcare but also from different social authorities, and they often had to explain the disease and their needs. The consequences of living with a rare disease were thereby expressed. Experiences of not being seen and understood as a person, being met with skepticism or not being listened to were perceived to bring additional difficulties in coping with the disease (I). Some of the young adults thus perceived that their personal thoughts and experiences of
living with LGMD2 sometimes were neglected by the professionals (II). Among the parents (III) lack of confidence in professionals or a sense of not being listened to caused frustration and worry about how to manage. Lack of information about support available was also expressed (III).

After the young adult had been transferred to adult healthcare, the young adults (I) and their parents (III) perceived that it had become more difficult to get in contact with healthcare and to know where and who to turn to. Some of the young adults counted on their parents as backups in the contact with professionals (I). Their parents described how they took an active part but also how they tried to encourage their young adult to take personal responsibility (III). For the young adults (I) having contact with healthcare professionals who had previous knowledge of the diagnosis provided confidence, and there was a desire to be able to have direct contact with a specialist if needed. The parents (III) described their concern about their young adult having contact with professionals they could rely on and the importance of professionals being dedicated, listening and able to explain unclear things. Perceiving that the young adults were called to regular follow-up controls at the hospital and taken care of was thereby a relief for the parents (III).

Entitlement to personal assistance was perceived to enable the young adult to live an independent life (I) and to reduce the parents’ caregiving duties (III). Difficulties, however, in receiving personal assistance were described as well as experiences of being granted insufficient hours for the young adult to be able to complete an education, for instance, or move to independent living (I, III). Young adults in need of support but without personal assistance had to rely on help provided by next of kin and friends. This could be difficult at times (I) and feelings of being restricted in doing activities were described (II). Some of the parents (III) expressed not only a sense of being standby and ready to help out in various situation but also experiences of having caregiving burden, such as feelings of being stuck or being deprived as regards their own social life. In addition, some of the young adults did not want external support, which increased the demands on the parents. Times of difficulty combining work and caregiving were described as well as experiences of becoming burned-out as the demands had become overwhelming (III). The need to receive sufficient external support from society was thereby emphasized, not only to enable for the young adults (I) but also for their parents (III) to live fulfilled lives.
Meaningfulness

Several of the young adults (I) and the parents (III) said that the young adult’s disease had given them new perspectives in life and had helped them to focus on what is meaningful in everyday life, such as spending time with family and friends. Through the years, they had not only learned a lot, but their experiences had also enriched and strengthened them: “I’ve learnt a lot through the years too … you grow as a human being in a different way … which maybe you do when you have children anyway, but this is like a step further.” Among the parents there was also a sense of having grown into the situation and having learnt to live with the young adult’s disease (III). Experiences of becoming more humble and positive about life, having met people and making friends that one would not have done otherwise, finding the motivation from the disease to do things and becoming more creative in solving practical difficulties were described among the young adults (II).

The value of having meaningful social relations was described (I, III): “my family … we have such incredibly good contact … that’s what means the most to me.” Some of the parents perceived that their relationship with their young adult had become closer because of the disease and the importance of valuing each other in the family was described (III). While some of the participants expressed having a strong social network, others experienced having a limited social life outside the family (I, III). For some of the participants, pets were also a source for joy and comfort.

The importance of being engaged in meaningful activities was also stated (II, III): “to get to work and there’s just me, and to be able to work with what I think is fun … is also a good way of getting the strength to cope.” The young adults described activities that bring joy and contribute to physical as well as mental well-being as being meaningful (I). Engagements of importance for personal development and/or of importance for other people were also perceived as meaningful, such as trying to increase people’s understanding of disability in society and being a mentor for younger persons with neuromuscular disorders. Some of the young adults seemed content with their daily activities. There were those who worked or studied and/or had meaningful leisure activities. Others regretted the lack of meaningful activities. Reduced physical abilities and limited energy made it difficult to perform various activities, and some described experiences of being forced to give up a job or an interest due to physical deterioration. The importance of being able to set and achieve new goals was thereby emphasized by the young adults (I). It could be, for instance, to attend an education, to find a job, to move to independent living and to travel (I). The parents (III) tried their best to enable and encourage their young adults to reach their goals. Every effort perceived to contribute to the young adult being able to live a fulfilled life and
experience well-being despite the disease was described as meaningful. The parents thus expressed a wish for their young adults to be able to do things that are vital for them before it became physically more difficult. The need for the parents to have their own space and meaningful activities was also emphasized. Some of them found joy in their work or retirement but others were not satisfied with their working situation or felt that they did not have time or energy to engage in any leisure activities of their own. Nevertheless, among the parents there was a sense of trying to take care of oneself and promote one’s own health (III).

**Self-rated SOC**

There was a variation in self-rated SOC scores (II, III). The median scores were 56 (range 37–77, mean 56) for the young adults and 68 (range 53–86, mean 68) for the parents. Among the young adults (II), those who scored the highest lived independent lives and/or were working. Those who scored the lowest were females, dependent on human aid, who stated that they very often “have feelings inside they would rather not feel” when answering the SOC-13 questionnaire. Among the parents (III), the females scored lower than the men (median 66 versus 72). Those who scored the highest and the lowest were parents to young adults in need of human aid and a wheelchair. Whereas those who scored the highest occasionally worked as personal assistants for their young adults who had moved to independent living, those who scored the lowest worked full-time as personal assistants for their young adults living at home.

All the participants (II, III) described their striving to cope with the young adult’s disease and its consequences. Those who scored above or the same as median (≥ 56 among the young adults and ≥ 68 among the parents), however, expressed satisfaction regarding social life, daily activities and external support to a greater extent than those who scored lower than median. This was seen regardless of the young adult’s physical abilities, showing that, despite severe physical disability, a high score was achievable for affected young adults (II) and their parents (III).

A summary of factors perceived to facilitate the participants’ management in everyday life is: the young adult being seen as a person, not only by people in their immediate surroundings but also by people in society and by professionals; having a supportive social network; being able to mobilize internal resources, such as trying to think positively; having meaningful daily pursuits; the environment being adapted; and having contact with concerned professionals who support coping with the disease and its consequences, including entitlement to personal assistance when needed (IV).
Discussion

This thesis is unique as it describes experiences and perceptions of living with LGMD2, from the affected young adults’ and their parents’ perspectives, using a salutogenic framework. The findings show that health viewed as well-being was perceived to be influenced as the disease progressed, not only by physical, emotional and social consequences due to the young adult’s disease but also by external factors, such as accessibility to support provided by society and other people’s attitudes. There was also a variation between good and bad periods. The parents tried their best to support their young adult, emotionally as well as practically. Difficulty in fully comprehending the disease was described as well as uncertainty and worry about disease progression. There was, however, a determination among the participants to try to make the best of the situation. In order to facilitate management in everyday life, the need for informal social support as well as the need for external support provided from society and concerned healthcare professionals was emphasized. The importance of having social relationships and activities that are meaningful was also described. There was a variation in self-rated SOC scores, which was related to satisfaction regarding social network, daily pursuits and received external support.

Methodological discussion

In this thesis, young adults diagnosed with LGMD2 and their parents were included. In order to ensure information-rich participants, the sampling procedure was purposeful (Patton 2015). There was a variation in the participants’ experiences of living with LGMD2 which is important in order to identify different perceptions within a group of people (Marton and Booth 1997). The young adults, who were recruited through healthcare (n=10) had confirmed LGMD2 diagnoses, based on analysis of muscle biopsy and genetic testing (Guglieri and Bushby 2008). Those who were recruited through the interest organization, Neuro Sweden (n=1), or from the web-based association for people living with disability (n=3), were self-reporting confirmed
diagnoses. The limb-girdle muscular dystrophies (LGMDs) are, however, classified into two main groups depending on the inheritance pattern (Emery 2008). LGMD2 represents the recessive forms in which both genes of a pair, one gene from each of the parents who both are unaffected, have to be abnormal in order to cause the disease. Spontaneous mutations that cause the disease may also develop. LGMD1 represents the dominant forms in which only one gene of a pair, from an affected parent, is needed to be abnormal to cause the disease (Emery 2008). In this thesis, persons diagnosed with LGMD1 were not included. The main reasons were that LGMD1 only represents around 10% of all the LGMDs (Rosales and Tsao 2012), have an adult onset of symptoms and are milder (Nigro et al. 2011) and the focus in study III was not to be on parents being affected by the disease themselves.

In some of the families, the participants’ experiences and perceptions were influenced by siblings who were diagnosed with LGMD2 but who did not take part in the study and whose voices therefore not are heard in this thesis. This might be a limitation. According to the parents, the reasons for the young adults not wanting to participate were: unknown, lack of time and a too sensitive topic or the young adult feeling depressed.

In order to enable a calm and friendly atmosphere, the semi-structured interviews were conducted at secluded places chosen by the participants, mainly at their homes. The question about receiving the diagnosis required an answer based on the participants’ retrospective memories and there is a risk of recall bias (Polit and Beck 2013) which may have influenced the findings. The young adults’ and their parents’ answers, however, were consistent. The inquiries about whether or not the young adults used a wheelchair often generated longer and more reflective answers than first expected. Also, when filling in the SOC-13 questionnaire (Antonovsky 1987), some of the participants chose to explain single answers or were reminded about a situation that they wanted to share. There were also questions posed about how to fill in the questionnaire. The presence of the interviewer while answering the questionnaire thus did not seem to have negatively influenced the participants, rather the contrary.

In order to respond to the aims of the studies in this thesis, different methods for qualitative data analysis were used, i.e. qualitative content analysis (I, II), content analysis (III) and phenomenography (IV). Interview data was also related to the participants’ SOC scores (II, III) to provide a more complete picture in order to understand the participants’ situation.

In studies I and II, the step models for qualitative content analysis presented by Mayring (2000) were used to facilitate the data analysis. Using the salutogenic theory (Antonovsky 1987) in the qualitative data analysis (I, II, III) was a strength considering the aims of the studies. It also facilitated the organization of the interview data. The three concepts (comprehensibility,
manageability and meaningfulness) that were used as deductive categories in study I, however, are closely intertwined and therefore difficult to separate into categories distinctly separated from each other. In study II, the categories and subcategories developed through data analysis are partly overlapping. According to Patton (2015), categories should have external heterogeneity which means that they should be separable. Therefore, in study III, content analysis was used and the findings were presented in descriptive themes that may overlap (Patton 2015). In study IV, the participants’ various perceptions of the transition from receiving the diagnosis to the young adult becoming in need of human aid and a wheelchair were in focus and therefore a phenomenographic study was conducted. This is a method that initially was developed within educational research but it is also used in the context of healthcare to investigate various ways in which people make sense of their experiences (Sjöström and Dahlgren 2002).

In this thesis, there was a sense among the young adults of not being ill although living with a chronic disease (II). According to the salutogenic theory (Antonovsky 1987), all people are more or less ill or well all the time. At any point of time, a person can therefore be seen somewhere along a health ease/dis-ease continuum, from maximally ill (dis-ease pole) to maximally well (ease pole). Due to a misunderstanding, “dis-ease” was unfortunately turned into “disease” in already published articles (I, II and III). This is a mistake that is not uncommon (Vinje et al. 2017) but it is unfortunate since Antonovsky (1987) regarded health as a continuum and thereby rejected the classification into categories of sick or well.

The SOC-13 questionnaire is not psychometrically tested for persons living with MD or their parent in a Swedish context. Moreover, the level of a normal SOC has not been specified (Antonovsky 1987) and it is therefore difficult to know what a person’s SOC score at a given time really means in practice (Eriksson and Lindström 2005). In this thesis, however, the young adults’ and their parents’ median SOC scores were used to mirror the interview data (II, III), which strengthens the findings (Patton 2015).

The principal investigator is experienced in caring for persons diagnosed with MD, not only professionally as a nurse but also personally from being a next of kin to a person living with the disease. In this thesis, these experiences can be viewed in terms of being a strength but they can also be questioned as being a weakness (Patton 2015). The strength is that the interviews and the understanding for the participants’ situation were facilitated. The potential risk, however, that collection and analysis of data were guided by the principal investigator’s pre-understanding in the area of research, for instance, by posing leading questions during the interviews, is a weakness. The principal investigator was aware of this risk throughout the research process. Each interview was therefore an opportunity for self-reflection and learning, not
only during the interview but also in the process of transcription. Data analyses were conducted close to the interview texts. Continuous discussions were held with the two co-authors regarding data analyses and content in the groupings of data, until coder agreement was achieved, in order to increase the credibility of the studies in this thesis (Patton 2015).

Ethically, all research involving human participants must be preceded by assessment of predictable risks and burdens to the persons in comparison with foreseeable benefits to them or other persons affected by the condition under investigation (WMA 2013). During the semi-structured interviews, the participants expressed various feelings and there was laughter as well as sadness. Some of the participants became emotionally affected and began to cry. On those occasions, the interviewer asked if the participant would like to quit the interview or take a pause, but everybody wanted to continue the interview. If any of the participants had become inconsolable, the backup was to take contact with the young adult’s healthcare team for further professional support to cope with emotions evoked during the interviews. There was, however, no need to consider this during any of the interviews. Several of the participants rather expressed appreciation over the study and that somebody showed interest in their experiences of living with LGMD2. They also said that they were happy being able to take part in the study and share their experiences, and there was a sense of doing something good that could benefit others. Thus, despite some of the participants becoming emotionally affected during the interview, the positive aspects of taking part in the study seemed to dominate among all of the participants.

Discussion of findings

Health was described in this thesis in terms of being closely related to well-being (I, III) which is also seen in the holistic theories of health (WHO 2014, Nordenfelt 2007, Tengland 2007, Eriksson 1996). Well-being can here be viewed as the balance point between a person’s resources and faced challenges, where the resources as well as the challenges comprise physical, psychological and social dimensions (Dodge 2012). Related to a person’s SOC, the importance of having a balance between underload and overload in order to manage daily life is described (Antonovsky 1987). Examples of overload were seen in this thesis among some of the parents who not only found it difficult to combine work, household requirements and caregiving duties but also had experiences of becoming burned-out.\(^2\) Examples of underload were seen among some of the young adults, who experienced not

\(^2\) The term “burned-out” was used by some of the parents in this thesis (III). According to Cullberg (2006), this concept should be avoided as it leads to the wrong perception that something is definitely destroyed and that the person is a passive party in a destructive process.
only lack of meaningful activities or social relations outside the family but also lack of external resources needed to live a fulfilled life, such as access to personal assistance (I, II). This in turn can be regarded as RD and as a hindrance to management. Difficulty in fully comprehending the disease was also described as well as uncertainty about how the disease would develop (IV). These findings indicate that comprehensibility, manageability and meaning in everyday life may be negatively influenced when being diagnosed with LGMD2 or being a parent to a person diagnosed with the disease. The participants tried, however, to make the best of the situation (I, III).

Evaluation of findings

Emotion-focused coping as well as problem-focused coping (Lazarus 2006) was used by the participants in order to manage the disease and its consequences (I, III). Emotion-focused coping means regulation of emotions tied to the stressful situation without changing the reality, for instance, trying to live in the present and trying not to worry about disease progression. Problem-focused coping means that actions are taken to change the reality of the troubled person-environment relationship, such as seeking information and trying to solve problems in everyday life. In this thesis, the variation between good and bad periods was also described (I, III). Living with the disease was thereby described both in terms of being a normal part of life and as a struggle to get through the day (I). A sense of having learned how to cope with the disease was also expressed by the participants and not only negative but also positive aspects of living with the disease were described (I, III). Thus, when having an internal SOC, suffering from a disease can be viewed not only as pathogenic but also as a source of learning for both the affected person and the caregivers (Oliveira 2014). In order to preserve the view of the world as being coherent when facing difficulties, a person with a strong SOC may also, temporarily or permanently, narrow the limits of which areas of life are considered to be important (Antonovsky, 1987). A person can also adapt vital goals to abilities in order to maintain or enhance well-being (Nordenfelt 2007). This means that things that were not reflected so much upon before the diagnosis may be more appreciated, such as spending time with family and friends (I, III). The health/illness transition thereby involves changes, not only moving from feeling healthy to chronic illness but also moving from chronicity to a new sense of well-being that encompasses the chronicity (Meleis 2012).

As the disease progressed, new ways of living had to be found that involved support from other people and the young adult using a wheelchair for ambulation (IV). The paradox for young adults living with chronic and disabling conditions is that gaining independence necessitates being dependent on human aid and assistive devices (Cook et al. 2013). The need for services and resources provided by society to facilitate management of the disease and the young adults’ independence has also been described. It could be, for instance, economic funding and receiving support required to move to
independent living and to attend an education or taking a job (Joly 2015). According to the salutogenic theory (Antonovsky 1987), access to advanced healthcare and social welfare can be referred to as GRR. The specific support provided by single healthcare professionals and personal assistants in order to manage particular difficulties in everyday life can be viewed as SRR. It could be, for instance, to receive help to fill in various application forms for support needed or to receive support from concerned professionals to cope with the disease at the time of diagnosis. Provision of specific support that could facilitate everyday management can also be viewed as SRR, for instance, entitlement to assistive devices, transportation facilities and financial support. In this thesis, entitlement to personal assistance was perceived to bring independence, not only for the young adults (I) but also for the parents (III). Entitlement to personal assistance (SFS 1993:387) is thereby a way to empower the person to gain control over their life regarding, for instance: where and how to live, what education to attend, what to work with and what leisure activities to perform (Tengland 2008). These are all important areas when it comes to striving for autonomy and independence of parents in the transition to adulthood (Arnett 2004).

The parents (III) scored higher on SOC than their young adults (II), with a median score of 68 versus 56. Antonovsky (1987) assumed that a person’s SOC would be relatively stable throughout adult life. There are, however, studies indicating that SOC in a general population tends to increase with age (Eriksson and Lindström 2005, Nilsson et al. 2010, Feldt et al. 2011) or may decrease with age (Hendrikx et al. 2008). The decrease in median SOC scores during a year, from 74 to 47 using the SOC-13 questionnaire, among persons diagnosed with Parkinson’s disease in a Swedish context, also indicates that SOC is sensitive to change when living with a chronic disease (Caap-Ahlgren and Dehlin 2004). The relationship of SOC to stressful events, coping strategies, health status, and QoL has also been described among Swedish women who were newly diagnosed with breast cancer (Sarenmalm et al. 2013). The findings showed that the women who scored a high SOC (> 75) using the SOC-13 questionnaire, reported fewer stressful events and used more coping strategies than those who scored a lower SOC. They also reported better health status and QoL (Sarenmalm et al. 2013). The SOC has previously been shown to be related to perceived health, especially mental health, and QoL. The stronger the SOC, the better perceived health (Eriksson and Lindström 2006) and the better QoL (Eriksson and Lindström 2007) were found. Persons living with MD have been shown to have reduced QoL compared with controls (Graham et al. 2011, Burns et al. 2012). Parents’ proxy reports also indicate that having DMD negatively influences their son’s health-related quality of life (HRQOL) in several domains compared with a general population (Baiardiini et al. 2011). Parents tend, however, to underestimate their sons’ HRQOL (Lim et al. 2014, Bray et al. 2010, Opstal et
al. 2014). There are also measurement difficulties when using questionnaires that include physical functions for persons living with MD since the scores will decline as physical abilities deteriorate in the path of the progressive disease. An advantage with the SOC-13 questionnaire is that it reaches beyond disease severity and can be used whether or not the person has a disease. Regardless of physical abilities, a strong SOC is thereby achievable among affected young adults (II) and among their parents (III). Further research is, however, needed to explore factors that may be related to SOC among persons living with MD and their next of kin.

The salutogenic theory focuses on how people can manage to stay well despite being faced with stress and disease, which are viewed as part of life (Antonovsky 1987). Using the salutogenic theory within healthcare, there might be a risk that single healthcare professionals, in their eagerness to help and to cheer the person up, take precedence not only to try and identify the person’s GRR but also to explain them for the person. This probably only leads to the person having a bad conscience and feelings of guilt about not being able to appreciate the positive aspects of life at a difficult period of time. Also, something that might be viewed as a GRR from the outside, such as having a job, might in fact be a stressor for the single person. This highlights the importance of acknowledging the person’s own experiences and perceptions of living with the disease. Among the young adults, however, experiences of not being listened to and feelings of not being viewed as a person by healthcare professionals were described, as well as a sense of often having to struggle for support needed (I). In the context of healthcare, there is a risk that objective data dominate the foundation for care and little consideration is given to the affected person’s perceptions or resources for management (Ekman et al. 2011). Moreover, healthcare professionals, like people in general, want to live in a notion of being protected against misfortune and disaster (Cullberg 2006). Therefore, having to face another person’s vulnerability to a progressive disease and severe physical disability touches their own lives. Instead of listening to the person’s needs, immediate actions are often taken by healthcare professionals to address the problems, such as prescribing medicine or give advice about how the person should act. Although these actions might not be wrong, the problem is that in the absence of having a dialogue with the person, the actions might not have any relevance either for the affected person (Cullberg 2006). In addition, limited time and organizational factors may influence the communication between healthcare professionals and recipients of care.

The importance of medical advances cannot be emphasized enough. Some diseases, however, cannot be cured and the person has to live with a chronic condition. They are in need of continuous support from concerned healthcare professionals in order to manage daily life (I, III). At the same time, through
their experiences, they become experts on what it means for them to live with the disease (I). Routine treatment and management of the disease often occur at home rather than at a hospital clinic. Persons living with chronic diseases and their families thereby become active participants in the care rather than the passive recipients of unilateral directives (Charmaz 1997). They also have become more informed and vocal about what they need from their healthcare providers (Meleis 2012), and persons living with a rare disease can become knowledgeable providers (Cook et al. 2013). This knowledge needs to be acknowledged by healthcare professionals. Therefore, in a time when healthcare becomes more specialized and technologically advanced, the importance of listening to the affected persons’ perceptions and needs, rather than acting on assumptions about what is best for the person, must be highlighted. This could be achieved by applying the salutogenic theory to person-centred care for persons living with chronic diseases, with focus on the dialogue between the person and healthcare professionals.

In the context of healthcare, the salutogenic theory (Antonovsky 1987) as well as person-centred care (Ekman et al. 2011) involve interdisciplinary healthcare professionals and focus on the person’s own perceptions, which may be highlighted through the dialogue between healthcare professionals and recipients of care. The genuine dialogue occurs when each of the participants really has in mind the other or others and turns to them with the intention of establishing a mutual relation (Buber 2002). According to the salutogenic theory (Antonovsky 1987), comprehensibility involves the person’s experiences of predictability, manageability means having a balance between underload and overload and meaningfulness refers to experiences of participation in the outcome. Each meeting between healthcare professionals and the person therefore needs to bring a sense of predictability, balance and meaningful participation for the affected person. There is otherwise a risk that healthcare professionals create a situation and experience that is difficult for the person to comprehend and manage. This can cause the person harm, and despite not being permanent, the damage is done (Antonovsky 1987). Among the parents (III), for instance, situations were described when being left alone with thoughts and questions after receiving serious information about the young adult’s disease from healthcare professionals, which was perceived to negatively influence management.

Applying the salutogenic theory to person-centred care means that healthcare professionals should not only initiate the person’s narratives about their own life situation, but should also try to acquire an understanding of how the person comprehends, manages and finds meaning in their present situation. The dialogue would thereby become more structured and reduce the risk of leaving out important areas in life. The person’s own perceptions regarding, for instance, information needed to comprehend the disease and support required to manage daily and meaningful activities can thereby be highlighted.
Through dialogue, a mutual understanding may thus be reached and form the base for establishing a partnership and shared decision making between healthcare professionals and the person, including their next of kin (Ekman et al. 2011). This does not only mean that the person relies on an interdisciplinary healthcare team with knowledge about the diagnosis and treatment but also highlights the importance of acknowledging the person’s and their next of kin’s own perceptions of living with the disease, in order to identify and optimize support needed. The importance of having the availability of the right SRR at the right time is thereby emphasized.

**Conclusion**

This thesis, using a salutogenic framework, contributes new knowledge about experiences and perceptions of living with LGMD2 and its influences on health, from the affected young adults’ and their parents’ perspectives.

The young adults experienced that LGMD2 influenced physical, psychological and social dimensions of health as the disease progressed. The parents were also influenced, not only by thoughts and emotions tied to the disease or caregiving duties but also by the young adult’s well-being. There was also a variation between good periods when the disease was perceived to be stable and everyday life was running well and bad periods when the disease was intruding on everyday life, requiring all the attention. A summary of the findings is presented in Table 6.

Each young adult diagnosed with LGMD2 and each parent has a unique situation to cope with. The various experiences expressed in this thesis emphasize the importance of early identification of personal perceptions and needs in order to enable timely health-promoting interventions. The salutogenic theory enables an overall view of the person’s situation and it focuses on internal as well as external resources available to deal with the difficulties that the person experiences. By applying the salutogenic theory to person-centred care, with focus on the dialogue between the person and healthcare professionals, support needed for the person to comprehend, manage and find meaning in everyday life can be identified as well as interventions required to enhance health and well-being.
This thesis highlights the importance of early identification of personal perceptions and needs as well as timely interventions to support young adults living with LGMD2 and their parents to optimize health and well-being. In order to acquire an overall view of the persons' situation, the salutogenic theory should be applied to person-centred care, taking into account not only medical aspects but also the person's own experiences and perceptions of living with the disease. This requires dedicated healthcare professionals who are able to listen to the person's narratives and who are sensitive to the person's needs. In cooperation with the interdisciplinary healthcare team, nurses have a key role in identifying physical, psychological, practical and social challenges or needs that the person experiences in everyday life due to the disease. The parents' situation should also be recognized, especially the situation for those who provide daily support to a young adult living at home. Through dialogue, healthcare professionals should try to understand how the young adult and the parents comprehend, manage and find meaning in their present situation. Concrete questions that healthcare professionals can ask to open up for a dialogue are, for instance: Do you have any reflections or questions about the diagnosis? How does your daily life work? What is your main activity for spending your days (having meaningful work, leisure activities, social network or not)? Where do you find your energy? Is there any support that you would need to facilitate management in daily life that you do not have access to? If so, could you describe what it is? The person should also be encouraged to write down any reflections, questions and needs that turn up in everyday life so that it can be discussed at the next follow-up control at the hospital. Through dialogue, not only physical, psychological, practical and social difficulties and needs can be identified but also internal as well as external GRR and health-promoting interventions that can facilitate the person's coping with the disease and its consequences. The provision of the right SRR for the single person to solve specific difficulties in everyday life can thereby be optimized.

The young adults and their parents need to feel confident that external support to manage the disease and its consequences is available throughout disease progression, in order to reduce worry about future management. Therefore, besides regular follow-up controls, they need to know where and who to turn to within the interdisciplinary healthcare team regarding various concerns that may turn up. They should also be offered professional support to cope with the disease and its consequences, not only at the time of diagnosis but throughout disease progression. Beginning to use a wheelchair, for instance, can be a psychologically distressing process, which has to be acknowledged by healthcare professionals when introducing it. Information provided to enhance the person's comprehensibility about the diagnosis should be individualized.

Table 6. A summary of the findings.

| Health: Health viewed as intertwined physical and mental well-being was perceived to be influenced, not only by physical, practical, emotional and social consequences due to the disease but also by external factors, such as accessibility to support provided by society and other people’s attitudes. |
| Comprehensibility: Realizing that it is a progressive disease, yet not really knowing what to expect caused worry about how the disease would develop. The perception was that available information about the disease is limited. |
| Manageability: Trying to make the best of the situation, the importance of having social and professional support to cope with the disease and its consequences was expressed, including young adults being entitled to personal assistance when needed. |
| Meaningfulness: Experiences of acquiring new perspectives in life were described as well as the importance of having social relations and activities that are meaningful. |
| Generalized resistance resources: Being able to mobilize internal resources, having a strong social network, access to healthcare and social welfare and the availability of interest organizations. |
| Specific resistance resources: Support provided by single professionals or family and friends to solve particular difficulties in everyday life. It also involves provision of specific external support that can facilitate everyday management, such as entitlement to assistive devices, home and car adaptation, transportation facilities and financial support. |
Implications

This thesis highlights the importance of early identification of personal perceptions and needs as well as timely interventions to support young adults living with LGMD2 and their parents to optimize health and well-being. In order to acquire an overall view of the persons’ situation, the salutogenic theory should be applied to person-centred care, taking into account not only medical aspects but also the person’s own experiences and perceptions of living with the disease. This requires dedicated healthcare professionals who are able to listen to the person’s narratives and who are sensitive to the person’s needs. In cooperation with the interdisciplinary healthcare team, nurses have a key role in identifying physical, psychological, practical and social challenges or needs that the person experiences in everyday life due to the disease. The parents’ situation should also be recognized, especially the situation for those who provide daily support to a young adult living at home.

Through dialogue, healthcare professionals should try to understand how the young adult and the parents comprehend, manage and find meaning in their present situation. Concrete questions that healthcare professionals can ask to open up for a dialogue are, for instance: Do you have any reflections or questions about the diagnosis? How does your daily life work? What is your main activity for spending your days (having meaningful work, leisure activities, social network or not)? Where do you find your energy? Is there any support that you would need to facilitate management in daily life that you do not have access to? If so, could you describe what it is? The person should also be encouraged to write down any reflections, questions and needs that turn up in everyday life so that it can be discussed at the next follow-up control at the hospital. Through dialogue, not only physical, psychological, practical and social difficulties and needs can be identified but also internal as well as external GRR and health-promoting interventions that can facilitate the person’s coping with the disease and its consequences. The provision of the right SRR for the single person to solve specific difficulties in everyday life can thereby be optimized.

The young adults and their parents need to feel confident that external support to manage the disease and its consequences is available throughout disease progression, in order to reduce worry about future management. Therefore, besides regular follow-up controls, they need to know where and who to turn to within the interdisciplinary healthcare team regarding various concerns that may turn up. They should also be offered professional support to cope with the disease and its consequences, not only at the time of diagnosis but throughout disease progression. Beginning to use a wheelchair, for instance, can be a psychologically distressing process, which has to be acknowledged by healthcare professionals when introducing it. Information provided to enhance the person’s comprehensibility about the diagnosis should be individualized,

Progressionstakten varierar, såväl mellan som inom de olika formerna av sjukdomen, vilket innebär att den kan utvecklas individuellt olika för olika personer. Inom ett fåtal olika former av sjukdomen kan komplikationer från hjärta och lungor uppstå. Andra komplikationer som kan utvecklas är skolios och kontraktioner, exempelvis i hälsenor och armbågar.


Det saknas idag botande behandling och personer diagnostiserade med LGMD2 behöver kontinuerligt stöd från ett tvärprofessionellt hälso- och sjukvårdteam för att kunna hantera sjukdomen och dess konsekvenser.

Tidigare forskning har fokuserat på medicinska aspekter, såsom diagnostisering och klassificering av olika former av sjukdomen samt behandling som är symptomatisk, men det saknas kunskap om vad det innebär att leva med sjukdomen utifrån unga vuxnas och deras föräldrars perspektiv.

Based on the person’s needs, and repeated. Information should also be provided about external support available to facilitate everyday management. In order to support the young adult’s transition to adulthood, the importance of timely interventions must be emphasized to enable the young adult to move to independent living, to attend an education or have a job and have meaningful leisure activities. Timely entitlement to personal assistance can unload the young adult who does not have to use reduced energy for practical daily chores but instead can perform activities that are valuable, before it becomes physically more difficult. This in turn will also unload the parents who will have reduced caregiving duties and thereby will be able to spend time with their young adult as parents rather than as caregivers.

The situation for young adults living with LGMD2 and their parents is complex considering the progressivity of the disease, and the solutions for management often lie beyond the healthcare system. Healthcare professionals should therefore 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 relevant interest organizations that can support the young adult’s and the parents’ management of the disease and its consequences.
Svensk sammanfattning


Det saknas idag botande behandling och personer diagnostiserade med LGMD2 behöver kontinuerligt stöd från ett tvärprofessionellt hälso- och sjukvårds teamwork för att kunna hantera sjukdomen och dess konsekvenser. Tidigare forskning har fokuserat på medicinska aspekter, såsom diagnostisering och klassificering av olika former av sjukdomen samt behandling som är symptomatisk, men det saknas kunskap om vad det innebär att leva med sjukdomen utifrån unga vuxnas och deras föräldrars perspektiv.
Ökad kunskap och förståelse om vad det innebär att leva med LGMD2, kan bidra till att stöd som ges av hälso- och sjukvårdspersonal kan optimeras.

Det övergripande syftet var att bidra med kunskap om vad det innebär att leva med LGMD2 utifrån unga vuxnas och deras föräldrars perspektiv. Avhandlingens fyra delsyften (I–IV), utifrån den salutogena teorin, var att beskriva:

- Unga vuxnas upplevelser av att leva med LGMD2 (I).
- Hälsouppfattningar relaterat till känsla av sammanhang hos unga vuxna som lever med LGMD2 (II).
- Upplevelser av att vara föräldrar till ung vuxen som lever med LGMD2 (III).
- Uppfattningar om övergången, från att få diagnosen LGMD2 till att behöva stöd från annan person och använda rullstol för att kunna hantera vardagen, utifrån de unga vuxnas och deras föräldrars perspektiv (IV).


Resultatet visar att sjukdomen, allteftersom den framskrider, har en omfattande påverkan på de unga vuxnas och deras föräldrars liv. Såväl fysiska, emotionella och sociala konsekvenser av sjukdomen beskrevs men även påverkan av yttre faktorer såsom tillgång till stöd från samhället och andra människors bemötande. Det fanns dock en beslutsamhet att försöka att göra det bästa av situationen. Några av deltagarna uttryckte att de genom sjukdomen hade fått nya perspektiv på vad som är meningsfullt i livet och även om sjukdomen påverkade vardagen så behövde den inte alltid vara i fokus. Andra unga vuxna uttryckte att de ständigt blev påminda och hämmade av sjukdomen och dess konsekvenser. Osäkerhet om hur sjukdomen skulle utvecklas skapade även oro inför framtidens hälsa. Deltagarna beskrev också variationen mellan goda och dåliga perioder. Faktorer som ansågs underlättat att hantera sjukdomen och dess konsekvenser var: att bli sedd som person av människor i samhället och av personal inom vård och omsorg; att ha socialt stöd från närstående och vänner; att ha förmåga att mobilisera inre resurser såsom positivt tänkande; att ha meningsfulla dagliga aktiviteter, exempelvis arbete, studier och/eller fritidsaktiviteter; att den fysiska miljön är anpassad; och att ha stöd från engagerad personal, inklusive att bli berättigad personlig assistent när behovet uppstår.
Självskattad KASAM varierade bland deltagarna. Resultatet visar att de som skattade över eller samma som median bland de unga vuxna (≥ 56) och föräldrarna (≥ 68) i större utsträckning uttryckte att de var nöjda med sitt sociala nätverk, sina dagliga aktiviteter och erhållet stöd från samhället jämfört med de som skattade lägre än median.


Ökad kunskap och förståelse om vad det innebär att leva med LGMD2, kan bidra till att stöd som ges av hälso- och sjukvårdspersonal kan optimeras. Det övergripande syftet var att bidra med kunskap om vad det innebär att leva med LGMD2 utifrån unga vuxna och deras föräldrars perspektiv. Avhandlingens fyra delsyften (I–IV), utifrån den salutogena teorin, var att beskriva:

1. Unga vuxnas upplevelser av att leva med LGMD2 (I).
2. Hälsouppfattningar relaterat till känsla av sammanhang hos unga vuxna som lever med LGMD2 (II).
3. Upplevelser av att vara föräldrar till ung vuxen som lever med LGMD2 (III).
4. Uppfattningar om övergången, från att få diagnosen LGMD2 till att behöva stöd från annan person och använda rullstol för att hantera vardagen, utifrån de unga vuxnas och deras föräldrars perspektiv (IV).

Acknowledgements

I am grateful and feel privileged to have had the opportunity to attend the research education at the Department of Health and Caring Sciences at Linnaeus University in Växjö. It has been an enriching time and I would like to express my sincere gratitude to all of the persons who made it possible and who supported me in different ways throughout my PhD journey. I would like to give special thanks to the following persons:

The participants in this thesis – for sharing your experiences and perceptions of living with LGMD2. Your strength has encouraged and inspired me.

The key healthcare professionals and members of Neuro Sweden – for helping me to recruit the participants for this thesis. Your positive answer to my question about forwarding the information letter about the study to potential participants made me hopeful and enabled this thesis.

Katarina Hjelm, my main supervisor – for believing in me and my area of research. With your knowledge you have created a learning atmosphere in which you have not only continuously challenged me to reflect upon and to scientifically improve my work but also have encouraged me to work independently, secure in the confidence that you will be there to guide me right throughout the whole research process and this thesis. By enabling me to attend this PhD education and through your dedicated supervision, you have supported me to strengthen my sense of coherence.

Sally Hultsjö, my co-supervisor – for your positive encouragement and support throughout my PhD education. With your knowledge and constructive comments you have highlighted how the scientific quality of my work can be enhanced. Through your optimism and belief in me and my work, you have not only continuously reassured and motivated me but also challenged me to progress in my work.
All the staff involved in the research education – especially Ulrica Hörberg, Emina Hadziabdic, Catarina Gaunitz, Fateme Yazdi and Mikael Andersson. Your kind support in various situations has been valuable and important to me.

The group of PhD students at Linnaeus University – for the time we spent together in various seminars and courses, for valuable comments on my research work and for our discussions about scientific as well as everyday matters. I have found myself become enriched, not only with new colleagues but also with new friends.

Monica Eriksson – for valuable comments at my final seminar. By sharing your knowledge about the salutogenic theory, you gave me new insights and helped me to improve my work.

Alan Crozier – for language review of the articles and this thesis.

My dear and precious family – for your unconditional love and for always being there for me. I love you all very much. You are my strength throughout life.
References


eBook: https://link-springer-com.proxy.lnu.se/content/pdf/10.1007/978-3-319-04600-6.pdf.


