Living in a Field of Tension

Between Development and Degeneration:

A Grounded Theory Study of Family Transitions and Neuromuscular Disease

by

Veronika Waldboth

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Florence Nightingale Faculty of Nursing and Midwifery

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Abstract

Background

Neuromuscular diseases are rare genetic conditions that present with progressive muscular weakness. As affected children develop, they become more physically dependent on their families for support and care. Previous research has shown the disease impact on the young person and their maturity into adulthood. However, there is little evidence about the effects of the young person's transition into adulthood on their family, which this study explored.

Methods

Informed by family systems theory, a constructivist Grounded Theory study was conducted. Data were collected by interviews with affected young people and their family members. The interviews were audio-recorded and transcribed followed by analysis comprising coding, memo writing, theoretical sampling and constant comparison techniques.

Findings

The analysis of 31 interviews from 12 families resulted in an interpretive theory constituted by four categories describing the transition experience of families as "a life in a field of tension between development and degeneration". The family transition experience was characterised by their 1) living and coping with physical dependence, while striving for independence; 2) balancing proximity within a family situation of interdependence, 3) conforming and challenging social standards and expectations and 4) grieving and fearing for loss, while joining forces for life. As a consequence, family functioning and wellbeing was threatened by recurring tension from stress, physical and emotional difficulties,

strained relationships and conflict. The families' strategies of coping and adaptation and the ways they related and communicated with each other, shaped their transition experience.

Conclusions

Families that experience threats to their wellbeing and functioning would benefit from professional support. Nurses can offer assistance by assessing and monitoring the families' needs and planning appropriate family interventions. Such interventions can focus on supporting family relationships and communication, factors that are associated with positive outcomes including effective coping and mastery, and positive adaptation to change.

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Statement of Originality

I hereby confirm that the work presented in this thesis is my own and that no part of it has been published or submitted for publication elsewhere, apart from the publication specified.

All data were collected and analysed by myself and the study was conceptualised with the support of my supervisors Professor Dr. Alison Metcalfe, Dr. Christine Patch and Professor Dr. Romy Mahrer-Imhof.

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List of Abbreviations

AHV	Federal Old Age and Survivors' Insurance
BMD	Becker Muscular Dystrophy
CEC	Cantonal Ethics Committee
CF	Cystic Fibrosis
DMD	Duchenne Muscular Dystrophy
EURORDIS	European Organisation for Rare Diseases
FSGI	Family Systems and Genetic Illness Model
FSI	Family Systems - Illness Model
GCP	Good Clinical Practice
HFG	Swiss Legal Requirements for Human Research
ICH	International Conference on Harmonisation of Technical Requirements for Registration of Pharmaceuticals for Human Use
IQ	Intelligence Quotient
IV	Federal Disability Insurance
LGMD	Limb-Girdle Muscular Dystrophy
MEB	Muscle-Eye-Brain disease
MD	Muscular Dystrophy
MDC	Congenital Muscular Dystrophy
NMD	Neuromuscular Disease
OKP	Compulsory General Health Insurance
QoL	Quality of Life
RNA	Ribonucleic Acid
SCD	Sickle Cell Disease
SMA	Spinal Muscular Atrophy
SMN	Survival Motor Neuron
UK	United Kingdom
WHO	World Health Organization
ZHAW	Zurich University of Applied Sciences

1. Chapter 1: Introduction

This study is investigating family transitions when a young family member is living with childhood neuromuscular disease (NMD) and transitions into adulthood. The transition into adulthood is a developmental stage of the human life cycle that every young person is expected to go through. A chronically debilitating NMD, however, can disrupt this expected transition, which impacts on the young person's life and as a consequence alters the family life cycle. Whilst the effects of NMD have been explored quite widely for the young person and their carers who is usually their mother, there has been little exploration of the implications for the whole family. Therefore, this study aims to investigate the impact of NMD for the family system and its functioning when the affected young person transitions into adulthood. To understand the effects on the family, it is important to have a deeper understanding of disease related processes and developmental tasks, and also to consider the context in which families are living, which is the focus of this chapter.

1.1. Neuromuscular Conditions

NMDs are a group of rare conditions that are either inherited or acquired and affect any part of a nerve and muscle (Hilton-Jones & Turner 2014, Muscular Dystrophy Canada 2009). There are many different types of NMDs that vary in cause and pattern of inheritance, time of onset and rate of progression, as well as in symptoms and prognosis. The prognosis depends on the specific type of NMD varying from mild and very slowly progressive forms with a normal life expectancy to severe muscle weakness with progressive functional disability and death at a young age (Bleck & Robb 2011, Bushby et al. 2010a, Kolb & Kissel 2015, Wang et al. 2007). Among the NMDs that affect children and

young people who will be transitioning into adulthood, are Muscular Dystrophies (MDs) and Spinal Muscular Atrophies (SMAs) (Amato & Russell 2008, Bäumer & Talbot 2014, Flanigan 2014).

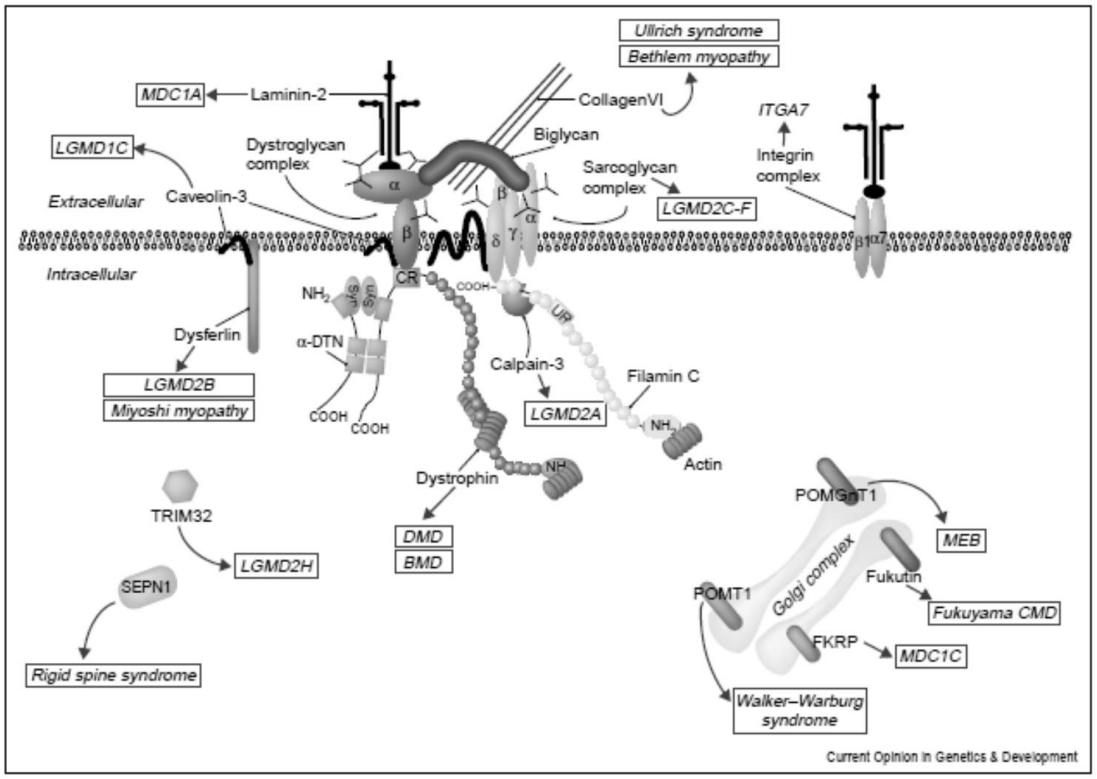
MDs and SMAs are rare conditions; the most common childhood MD, Duchenne MD, affects one out of every 3,600 – 6,000 of all male newborns, while SMAs affect one in every 11,000 newborns (Bushby et al. 2010a, Kolb & Kissel 2015, Papazian & Alfonso 2002). The Swiss national register for NMDs reported a prevalence of 353 affected individuals of all ages including different types of MDs and SMAs and 21 asymptomatic and symptomatic carriers in 2015 (Klein et al. 2016). Rare conditions affect few individuals, but they are collectively common. Around 30 million Europeans and 25 million North Americans are affected by one out of 5,000 different rare diseases including NMDs (Commission of the European Communities 2008, Schieppati et al. 2008). Estimates of the international prevalence of rare diseases were recently published in the Orphanet Report Series (2016), but the heterogeneous nature of NMDs makes it impossible to report a single estimate of incidence or prevalence, which would depend on the choice of sub-types that are categorised.

1.1.1. Muscular Dystrophies

MDs are the most prevalent group of NMDs and they are characterised by a genetic defect responsible for loss of muscle protein expression causing progressive wasting and fibrosis of the muscles (Brandsema & Darras 2015, Falzarano et al. 2015, Papazian & Alfonso 2002). There are over thirty different types of MDs resulting from genetic mutations that affect proteins in the

nucleus, membrane (sarcolemma) and basic unit (sarcomere) of striated muscle tissue, or in the basement membrane and extracellular matrix surrounding muscle fibres (Figure 1) (Amato & Russell 2008, Flanigan 2014). The dystrophin protein which is absent or altered in MDs referred to as dystrophinopathies is localisable on the subsarcolemmal surface where it links intracellular to extracellular structures via dystroglycan complex. The gene responsible for this alteration causing Duchenne and its milder version Becker MD is located on the X chromosome at Xp21 (Amato & Russell 2008). Dystrophin proteins are involved in cell signalling pathways and they have a stabilising role for the muscle membrane. Absence of dystrophin causes failure of muscle fibre integrity and necrosis of muscle tissue which is gradually replaced by connective and fatty tissue, thereby reducing their function as a muscle (Amato & Russell 2008, Flanigan 2014). The loss of muscle has repercussions for the functioning of major organs such as the heart, lungs and movement of the skeleton, for instance.

Figure 1: MDs and related proteins



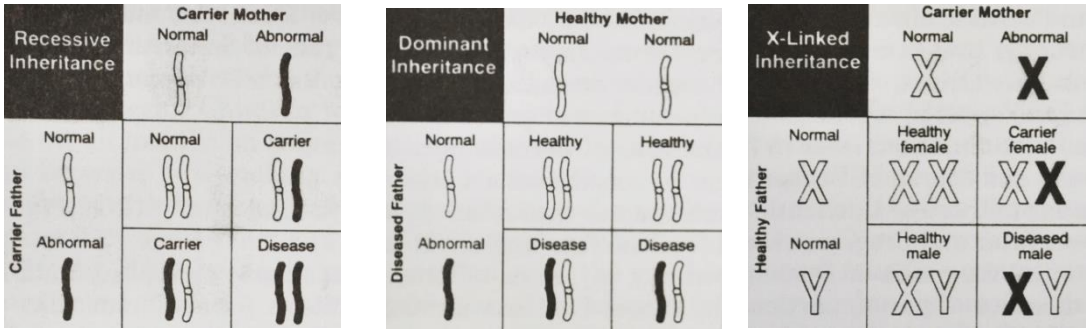
DMD: Duchenne MD; BMD: Becker MD, LGMD: Limb-girdle MD, MDC: Congenital MD; MEB: Muscle-Eye-Brain disease

Figure from: Dalkilic I. & Kunkel L.M. (2003) Muscular dystrophies: genes to pathogenesis. Current Opinion in Genetics & Development 13(3), p.233, illus. (Republished with permission from Elsevier; permission conveyed through Copyright Clearance Center, Inc.)

The inheritance pattern of Duchenne and Becker MD is X-linked recessive (Amato & Russell 2008, Flanigan 2014). As females have two and males have one X chromosome, every son born to a female carrier who has the defect on one chromosome has a 50% chance of inheriting the gene and having the condition, whereas daughters have the same chance of being a carrier (Figure 2). Although running in families, in one third of individuals affected by Duchenne MD the genetic defect results from a spontaneous mutation (Amato & Russell 2008, Flanigan 2014). In addition, most female carriers of Duchenne MD do not have any symptoms and if there were no prior phenotypes in their family, they

are not aware of their carrier status. Only between 2.5% and 7% of affected females are manifesting carriers showing signs of mild weakness and cardiomyopathy, severe muscle wasting is very rare (Amato & Russell 2008, Flanigan 2014).

Figure 2: Pattern of inheritance



Figures from: Siegel I.M. (1999) *Muscular Dystrophy in Children: a Guide for Families.* Demos Medical Publishing, New York, p.7, illus. (Republished with permission of Demos Medical Publishing; permission conveyed through Copyright Clearance Center, Inc.)

Male newborns who have inherited the genetic defect develop Duchenne MD (Bushby et al. 2010a, Papazian & Alfonso 2002). First symptoms usually present in early childhood and diagnosis is made between four and five years of age. By the time the child is diagnosed, families may already have other offspring. Therefore, there is the risk that more than one child is affected. Diagnosis is made late because most affected children appear normal at birth and reach expected milestones such as sitting and standing. Delayed walking and abnormal gait including waddling and toe walking frequently appear between 3 and 8 years of age (Amato & Russell 2008, Flanigan 2014). Weaknesses in the muscles of the lower limbs lead to the characteristic Gower sign, where affected children rise up from a squatting position by using their hands and arms to climb up their own bodies (Amato & Russell 2008, Siegel 1999). Using stairs and ambulation in general becomes more and more difficult

with time, and by the age of 13 years most individuals living with Duchenne MD are confined to wheelchairs and dependent on others for their physical care. They further develop kyphoscoliosis (spinal curvature) and muscle and connective tissue contractures which impair their body posture. In addition, smooth muscle tissue such as the gastrointestinal tract and the central nervous system may also be involved, with some individuals with Duchenne MD having an average intelligence quotient (IQ) below the mainstream mean (Amato & Russell 2008, Busby et al. 2010a, Busby et al. 2010b). In addition to the wasting of striated muscles, respiratory and cardiac functions worsen as both organs are also affected by muscle wasting. As a consequence of cardiac and respiratory failure the life of individuals with Duchenne MD is limited with a survival to the second, third or fourth decade of life (Brandsema & Darras 2015, Kohler et al. 2009).

Becker MD is one third as frequent as Duchenne MD and it is the milder version (Amato & Russell 2008, Flanigan 2014). It can be distinguished from Duchenne MD by its slower rate of progression and by genetic analysis, but it is caused by mutations in the same gene, dystrophin. Symptoms in affected individuals vary widely, but they are usually able to walk past the age of 15 years. Limb-girdle MDs are a group of NMDs that affect males and females equally by being either autosomal dominant (types 1) or recessively inherited (type 2) (Amato & Russell 2008, Norwood & Bushby 2014). They show similar phenotypes to dystrophinopathies.

1.1.2. Spinal Muscular Atrophies

SMAs are mostly autosomal recessively inherited NMDs that affect male and female newborns (Kolb & Kissel 2015). These conditions present with progressive muscular weakness and atrophy caused by degeneration of the anterior horn cells of the spinal cord (Amato & Russell 2008). The severity of the phenotype depends on the genetic defect of the survival motor neuron (SMN) genes I and II on chromosome 5. Both genes produce similar proteins that are responsible for messenger ribonucleic acid (RNA) synthesis (Amato & Russell 2008, Bäumer & Talbot 2014). Although the SMN protein is found in many different cells, due to unknown reasons spinal motor neurons are selectively vulnerable.

There are different types of SMA that can be distinguished according to their phenotype including age of onset and course of the disease, with mild and intermediate types II and III showing similar clinical manifestations as Duchenne MD (Amato & Russell 2008, Kolb & Kissel 2015, Schaaf & Zschocke 2013). SMAs type I manifest clinically in newborns or within the first six months of life and are associated with death in the first year in 80% of the cases, whereas type IV is an adult onset SMA. Young people with types II and III experience childhood onset and most of them transition into adulthood. Approximately two thirds of the individuals living with SMA type II, for instance, survive until their mid-twenties (Amato & Russell 2008). Most individuals living with type II sit, but never walk, whereas if affected by SMA type III, they develop the ability to stand and walk. Young people affected by either of these middle types experience progressive muscular wasting, develop contractures and scoliosis, and they also have a varying degree of bulbar involvement which contributes to

respiratory complications (Bäumer & Talbot 2014). Due to their health situation, affected individuals become dependent on assistive technology and tools such as on wheelchairs and ventilators and they need assistance with performing activities of daily life.

1.1.3. The Effects of NMD

Considering the physiological effects of the disease, the health situation of affected young people is serious. To date there is still no primary treatment, but in the previous decades there have been promising developments so that life expectancy of individuals with severe childhood NMD has risen because of progress in health care and medicine (Amato & Russell 2008, Brandsema & Darras 2015, Bushby et al. 2005, Kohler et al. 2009). Crucial to these children's survival were improvements in prevention and treatment of respiratory and cardiac complications, through the introduction of mechanical ventilation and preventive cardiac therapy. Patients now also benefit from a multidisciplinary approach to alleviate symptoms and slow progression, including treatment for health maintenance, continuous monitoring of disease progression and prevention of complications (Busby et al. 2010a, 2010b, Papazian & Alfonso 2002, Wang et al. 2007). Due to these improvements in treatment, affected individuals now live longer than ever before, and many more transition into adulthood. The median survival for patients with Duchenne MD without treatment with assisted ventilation was reported at 14.4 – 20.5 years, while the current median age of survival for affected young people who live in Switzerland and receive respiratory treatment is 35 years (Kohler et al. 2009).

With prolongation of life and due to the progressive nature of NMD, families are confronted with an increase in the affected young person's caregiving needs. Parents who cared for their child described their caregiver experience as important and rewarding, but also related with substantial caregiver burden (Pangalila et al. 2012). The child's need for continuous support was highlighted as problematic, especially when the affected child had a tracheostomy as they needed constant monitoring to prevent complications. Caring for a chronically ill child was also associated with a substantial economic burden (Armstrong et al. 2016, Klug et al. 2016, Landfeldt et al. 2014, Schreiber-Katz et al. 2014). An international study by Landfeldt et al. (2014) reported estimates of the household burden for families living with Duchenne MD in Germany, Italy, UK and the United States between $58,440 and $71,900 per patient per year (PPPY). Another study of families living with Duchenne MD in Germany estimated the mean economic burden including direct and indirect annual costs per patient at €78,913 (Schreiber-Katz et al. 2014). A substantial economic burden was also described for the German SMA population, with an estimated overall mean of direct and indirect costs at €70,566 PPPY (Klug et al. 2016). Armstrong et al. (2016) reported remarkably higher annual costs for total healthcare expenditures for individuals with SMA compared to healthy children, with $47'862 (± 88'607) and $1'861 (± 6'374), respectively ($p<0.001$).

In addition to the reported financial burden, affected individuals and their family members are confronted with psychosocial and emotional difficulties. Individuals with Duchenne MD have been described to have an increased risk for emotional and psychosocial difficulties as a result of their health situation, among which behavioural problems, social withdrawal, isolation and depression

(Polakoff et al. 1998). Most rare genetic conditions affect physical and mental abilities and are consequently associated with a reduced quality of life (QoL) (Drotar 1981, Elliot 2015, Schieppati et al. 2008). Cohen and Biesecker (2010) reviewed evidence on QoL of individuals living with various rare genetic conditions. They revealed that the overall QoL of individuals affected by MD was significantly poorer in affected individuals compared to their healthy counterparts. Surprisingly, QoL in the physical functioning domain was significantly higher in the affected than in the unaffected group as reported by one study included in the review (Grootenhuis et al. 2007). This rather unexpected result considering the physical limitations resulting from MDs was explained as related to the individuals change in values because of their condition and a reframing strategy in order to adjust expectations (Grootenhuis et al. 2007, Cohen & Biesecker 2010). Affected young people may have downplayed their physical limitations and difficulties and internalised this strategy in order to cope better with their life situations in the longer term.

Levels of anxiety and depression in parents associated with their caregiver experience were reported, but these levels were comparable to the general population according to Pangalila et al. (2012). Landfeldt et al. (2016), on the contrary, described moderate to extreme anxiety or depression levels in half of the caregivers who participated in their study (383 of 770). These discrepancies between the two studies can be explained by sample size and patient demographics, with a mean age of the patients in the study of Pangalila et al. (2012) of 27 (±6.1) years and in Landfeldt et al. (2016) of 14 years. At a younger age or during adolescence, families may be more vulnerable to mental difficulties than in emerging adulthood or adult life. During this developmental

time, parents may also not only be caring for their affected child, but for their other children and possibly also for their own elderly parents, and were therefore having higher socio-economic pressures on them.

1.2. The Transition into Adulthood

Today most individuals living with childhood NMD will be transitioning into adulthood as a result of improvements in disease management. Transition is understood as the *"passage from one life phase, condition, or status to another"* where it includes processes such as phases, disruptions and responses aimed at the creation of new stability (Chick & Meleis 1986, p. 239-240). The transition into adulthood is a developmental stage in the human life cycle which is defined as the transitional period of a young person between childhood and adulthood which involves various changes for the young person on a physical, cognitive, psychological and socio-emotional level (Santrock 2013). Individuals need to master certain milestones, but the developmental experience for each person is unique with the age of achievement of specific milestones being variable (Bill & Knight 2007). Depending on the young person's characteristics and on external factors such as the family and social context, the development experience differs. Becoming an adult is a time of transition and personal development, which can be different for individuals affected by a chronic childhood disease as compared to their healthy peers. For young persons with NMD, for example, physical limitations and dependence on others increases at the same time as they are expected to break away from their parental homes and live an independent life (Parkyn & Coveney 2011).

1.2.1. Individual and Family Development

To gauge the full extent of implications of chronic illness on individuals and families, it is important to contrast major processes and overall characteristics of the expected adolescent development. During the transition into adulthood young people are expected to experience various changes on different levels. They are expected to complete physical growth and develop sexual characteristics (Christie & Viner 2005, Santrock 2013). On a cognitive level, they are building their own attitudes and values, developing new skills including abstract and logical thinking and they form their personal as well as their sexual identities. It is also a time when young people start to identify with their peers, develop interests in intimate relationships and are anticipated to become more and more independent from their families (Christie & Viner 2005, Santrock 2013). In light of the above, the transition into adulthood is complex and multidimensional, where changes occur in many spheres of a young person's life.

Different developmental theorists of the last century defined sets of tasks or objectives that young people have to reach during their transition into adulthood (Christy & Viner 2005). Most theorists focused on single aspects on which they then elaborated. Stanley Hall (1844-1924) proposed that adolescent development was primarily based on biological changes (Santrock 2013), whereas Sigmund Freud (1856–1939) concentrated on the psychosexual development viewing adolescence as a recurrence of the sexual development of childhood (Bill & Knight 2007, Christy & Viner 2005). The Swiss biologist Joan Piaget (1896–1980) was a developmental scientist who studied cognitive development and stressed the importance of abstract thinking for adolescence,

while Lev Vygotsky's (1896-1834) elaborated on his sociocultural cognitive theory which emphasised that culture and social interactions shape cognitive thinking (Santrock 2013). Furthermore, the development of a personal identity was viewed as crucially important for the transition into adulthood by Erik Homburger Erikson (1902–1994) (Bill & Knight 2007, Christie & Viner 2005, Santrock 2013). All of these theories contribute to our understanding of the transition into adulthood, and demonstrate that the rich complexity of adolescent development cannot be portrayed by one single model (Santrock 2013).

The developmental theories introduced above focus on the development of the person as an individual, but miss the contribution of the young person's experience embedded into a larger social context such as a family group. Humans are social beings and every person is part of a social group or family situated in a specific socio-historical context with existing mutual influences between family members as well as external influencing factors (Christie & Viner 2005, McGoldrick et al. 2013b, Rolland 1987, Rolland & Williams 2005). A chronically ill young person is particularly connected with his or her family, because of their physical dependence on others which are usually family members including parents and siblings. As these individuals age, they become even more dependent on those around them.

While young people transition into adulthood, other family members experience this transition in a different way. For example, parents may see their child's transition into adulthood as a launching of their children, which may result in experiences of new found independence or feelings of loss for parents (McGoldrick et al. 2013b). The previously introduced developmental theories

are therefore only valuable to a certain extent, because this study adopts a more comprehensive approach where individual development is viewed within the context of family relationships. By contrast, family developmental theorists argue that the individual's development and transition needs to be viewed within the context of their family and the progressions of individuals' relationships towards a maturity of interdependence, rather than seeing the person only as an individual, uninfluenced and totally independent of others. The theoretical perspective of this study is therefore a family developmental lens, comprising human life cycle theory (Carter & McGoldrick 2005, McGoldrick et al. 2013b).

1.2.2. Chronic Illness in Adolescence

During their transition into adulthood, young people who live with NMD are confronted with developmental tasks, but they also have to deal with the impact of the chronic condition on their lives (Bill & Knight 2007). There are many practical implications of the chronic illness including the treatment, management and therapies, the integration of special nutrition and mobility regimen, as well as psychological challenges such as coping with having physical limitations and facing uncertainty or death at an early age (Bill & Knight 2007).

Today, state of the art procedures for disease management have been agreed upon in various guidelines. Bushby and colleagues (2010a, 2010b) developed a toolkit for the management of Duchenne MD including assessment and interventions of the muscular-skeletal system, pharmacological and psychosocial management, as well as management of respiratory, cardiac and gastrointestinal complications. At least six-monthly young people's strength, function (e.g. respiratory and cardiac function), and range of movement should

be assessed and pharmacological treatment and interventions adjusted (e.g. treatment with corticosteroids and respiratory interventions) (Busby et al. 2010a, Bushby et al. 2010b). Besides interventions to support respiratory and cardiac function, management of contractures and nutritional assessments are indicated to prevent over- or underweight and manage dysphagia. As part of the psychological management, learning behaviour, coping strategies and social integration need to be monitored and dealt with if problematic, and the transfer to adult care services needs to be planned and carried out (Bushby et al. 2010a, Bushby et al. 2010b).

Similarly, a consensus paper for the treatment of SMA was developed by Wang et al. (2007) including recommendations on family education and counselling, pulmonary, gastrointestinal, nutritional and orthopaedic care and rehabilitation, as well as palliative care. Families need to be informed and counselled about SMA, the disease process, genetic topics such as carrier testing and available treatment options as well as about resources for further information (e.g. through a main contact person) (Wang et al. 2007). Respiratory management includes periodic assessments and monitoring every three to six months and interventions adjusted to the individual patient's need comprise airway clearance and secretion management, respiratory support through ventilation, routine immunisations and a low threshold for starting antibiotics. Additionally, treatment focuses on nutrition and contracture management as well as on regular exercises and determination of the need for assistive technology and adaptive aids (Wang et al. 2007). The consensus statement also gives recommendations on preoperative and acute care management for this high risk group for complications, as well as for anticipatory palliative care planning.

Affected families live in specific socio-historical contexts and they are confronted with reciprocal effects of chronic illness and development (Table 1) (Christie & Viner 2005, World Health Organization 2007). The interplay between the individual, their chronic illness, their development and their life within a specific family and healthcare context adds to the complexity of the transition experience by affecting the physical and psychosocial aspects of their development. Physical and psychosocial developments are viewed as interrelated, the timing of one can have an impact on the other (Christie & Viner 2005, World Health Organization 2007). Chronic illness can cause delay or small growth in puberty, which affects the young person's physical appearance and is also associated with their psychological and social development (Christie & Viner 2005). As a consequence, young people may have low self-esteem or develop feelings of depression and are treated differently by others as they look less mature than their healthy peers. Young people with physical disabilities have been observed to be at high risk for developing a mental disorder (British Medical Association 2003).

Table 1: Reciprocal effects of chronic illness and development

Effects	Biological	Psychological	Social
Chronic illness on development	Delayed puberty; Short stature; Reduced bone mass accretion.	Infantilisation; Adoption of sick role as personal identifier; Egocentricity persists into late adolescence; Impaired development of sense of sexual or attractive self.	Reduced independence at a time of when independence is normally developing; Failure of peer relationships then intimate (couple) relationships; Social isolation; Educational failure & then vocational failure; Failure of development of independent living ability.
Development on chronic illness	Increased caloric requirement for growth may negatively impact on disease parameters; Pubertal hormones may impact upon disease parameters.	Poor adherence and poor disease control due to: - poorly developed abstract thinking and planning; - difficulty in imagining the future; self-concept as being "bullet-proof"; - rejection of medical professionals as part of separation from parents; - exploratory (risk-taking) behaviours.	Associated health risk behaviours: - chaotic eating habits may result in poor nutrition; - smoking, alcohol & drug use often in excess of normal population rates; - sexual risk-taking, possibly in view of realisation of limited life span.

Table adapted from: World Health Organization (2007) *The Adolescent with a Chronic Condition. Epidemiology, Developmental Issues and Health Care Provision.* (Michaud P.-A., Suris J.C., Viner R.) WHO Press, Geneva, p.32 (Adapted version published with permission of WHO Press)

1.2.3. Transfer to Adult Services

The chronically ill young person's transition into adulthood is also a time when they have to transfer from paediatric to adult health care services. This has been described as an additional challenge for affected young people and their families, besides the developmental and disease related tasks confronting them (Abbott et al. 2012, Schrans et al. 2013, World Health Organization 2007). The Swiss law defines age limits for paediatric care and promotes the affected young person's transfer to adult services at the age of 16 years, despite benefits of a more gradual or later transfer recommended for young people living with chronic illness (Schweizerische Akademie der Medizinischen Wissenschaften 2013). The Swiss Association for the Health of Adolescents declares necessary requirements for high quality care for adolescents and

criticises the upper age limit when adolescents are allowed to be cared for in paediatric hospitals (Schweizerische Gesellschaft für die Gesundheit Adoleszenter 2014).

According to an investigation of preferences of Swiss adolescents living with chronic illness, half of the participants would prefer the age of 18 to 20 years or older for transferring from paediatric to adult services (Rutishauser et al. 2011). This can be explained by the fact that during adolescence chronically ill young people already have to deal with multiple changes and difficulties, which may delay their development. They are then confronted with the transfer to adult care, which is an additional task created by the health care system for whom they may not be ready yet. In the paediatric setting affected young people feel secure as family involvement is well established and also because they know their treating health professionals. By contrast, in adult health care services the adolescent is seen as an individual person that acts and decides independently which might exceed the young person's and the family's level of development at this point in time. Nurses, treating physicians and other health professionals are required to ensure medical treatment, while they are also considering the family's developmental needs.

Currently, however, health care services fail to meet the needs of affected individuals and their families. Affected young people, parents and experts, for instance, described a lack of involvement of the young person in the planning of the transfer of service, a lack of knowledge about care plans and the absence of key professionals at important meetings (Abbott et al. 2012, Schrans et al. 2013, World Health Organization 2007). These problems may be explained by

the fact that health professionals often have a lack of knowledge and understanding of family needs. When needs are not known they cannot be considered adequately. In order to ensure the quality of care and improve the transition experience of families living with a young person affected by NMD, further insights into the family experience have to be gained from which conclusions for improvement of the future health care provision can be drawn. This is particularly important, as in Switzerland family members are recognised as a crucial resource to manage current and future care needs (Bundesrat 2014).

In addition, unstandardised processes such as deficient information sharing and communication strategies as well as lack of family involvement in planning might have negative effects on the quality of care, and therefore need to be considered. Some Swiss hospitals have standardised transitional programmes in place aimed at improving the young persons' transfer experience (Becher 2013, Wille & Klein 2015), but there are no national policies or comprehensive transitional care plans available. Regardless of the existence of such policies, there may be room for improvement of health care services. In the example of the UK, for instance, where there are national policy plans for transfer to adult services in place, a comprehensive investigation on the transition experience of families living with Duchenne MD resulted in few examples of adherence to these plans (Abbott et al. 2012).

Besides the quality of the transfer to adult care, the general availability and quality of professional care was rated as insufficient by some families living with rare disease including families affected by NMD. Evidence suggests that these

families feel generally under supported and face difficulty in getting the right diagnosis, accessing appropriate health care services and they reported a lack of treatment options (Elliot 2015, Schieppati et al. 2008). There is also great disparity in resources as treating health professionals may lack specialist knowledge and patchy distribution of specialist services causes wide variation in quality of care and clinical outcomes (Wang et al. 2007). Only recently, in major Swiss hospitals specialised centres for patients affected by muscular diseases were established, which today contribute to high quality care. Nevertheless, a national report described a persistent lack of professional care services and gaps in the support chain for caregiving families, especially for the ones with a migrant background or for working caregivers (Eidgenössisches Büro für die Gleichstellung von Menschen mit Behinderungen 2016, Bundesrat 2014). Among the gaps were a lack of respite services including the availability of third party care during the absence of the family. Recently, therefore, the Swiss Federal government defined an action plan to improve the situation of family caregivers and to empower them to care for their relatives for a more prolonged time (Bundesrat 2014). The study of the families' illness experience has been identified as one of the fields of action.

1.3. Chapter Summary and Outlook

This chapter introduced physiological processes of childhood NMD, outlined the effects of the condition on the lives of affected individuals and their families and described the expected developmental path of young people who transition into adulthood. It was found that the affected individuals' transition needs to be viewed within the context of their family, because reciprocal effects between illness, development and family life can only be understood when adopting a

family perspective. Further research is needed to explore the family transition experience, by taking a close look at how the life situation of the affected young person including his or her growing dependence affects the family and their functioning as a unit, at a time when they would be expected to mature into independence. These insights are valuable for future improvements of health care services, which currently do not focus on family developmental needs and lack awareness of chronically ill young persons' being embedded within a larger social context where there are mutual influences. Therefore, a literature review was conducted that aimed at exploring the existing evidence on the family's transition experience more comprehensively. This literature review will be the focus of the next chapter.

2. Chapter 2: Literature Review

A better understanding of the impact of a chronic condition on family life is necessary. Therefore, in this chapter existing studies were critiqued in order to examine the evidence to identify the needs and challenges confronting affected families when a chronically ill young person is transitioning into adulthood. Being aware of the families' needs and expected challenges can empower health professionals to improve their services and to provide support in order to facilitate positive outcomes such as the families' mastery and effective coping with challenges. By doing this, potential burdens that might have significant consequences for the health and psychological wellbeing of all family members involved can be alleviated.

2.1. Introduction to the Literature Review

In order to further explore the experiences of families where a young family member has a chronic childhood disease and transitions into adulthood a comprehensive review and critique of existing literature was conducted prior to commencing empirical research. At the outset of a research study the conduct of a literature review is relevant to find out what work has been carried out previously in the research area to ensure that a consequent investigation is innovative and not repeating research unnecessarily (Creswell 2014, Silverman 2011). Unnecessary research implies that there is already established evidence where further insights would not add to the current understanding. Conducting the research would be unfair and unethical as it would waste resources such as the participants' time and create avoidable risks of negative effects, especially if the topic might raise sensitive issues for participants.

In addition, a literature review is useful to learn more about the subject of interest and to relate a consequently planned research study to its larger context, by creating the knowledge base for guiding methodological decisions and for contrasting research findings (Creswell 2014). Knowing the literature, for instance, makes aware of the essential starting points and potential components that require inclusion in data collection and interpretation of results to ensure that the most relevant topics are covered and discussed.

2.1.1. Scope of the Literature Review

An initial scoping of the literature in preparing the consequent research showed that there were few studies on the effect of NMDs on the family experience and their functioning across the life cycle focusing on the point in time where the affected young person was transitioning to adulthood. As a result of this preliminary search, the focus of a consequent literature review was extended from only including NMDs to the inclusion of additional chronic childhood conditions that share some essential features. Evidence included in the literature review was not on families living with NMDs only, but also on several other conditions that are genetic in origin, have their onset in childhood, are progressive in nature, have no known cure available and are chronically debilitating leading to physical impairments during adolescence (Schaaf & Zschocke 2013, World Health Organization 2007). This broad approach to the review of evidence allowed the comparison of family experiences across genetic childhood conditions and it informed the development of the consequent study, by considering and integrating information from related populations. When the topic of interest is under researched, information from related populations can be used to guide further methodological decisions. Therefore,

the aim of this literature review was to conduct a systematic review and critique existing literature about the experiences of families where a young person is living with a genetic childhood disease and transitions into adulthood. This study of the literature focused on progressive genetic childhood conditions including NMDs, Cystic Fibrosis (CF), Haemophilia and Sickle Cell Disease (SCD). The following research question guided the literature review:

What are the experiences of young people living with a genetic and chronic childhood condition and their families when the young person is transitioning into adulthood?

2.1.2. Disseminating Research Findings

There is national and international interest on this systematic review's findings and insights that were already made available to a wider audience comprising not only scholars and lecturers, but also clinical nurses and medical lay people such as affected families. This systematic literature review resulted in:

- A scientific presentation at an international conference in August 2015; *Waldboth V., Patch C., Mahrer-Imhof R. & Metcalfe A. (2015) Becoming an adult: Experiences of individuals affected by chronic childhood disease and their families during transition into adulthood: A systematic literature review. 12th International Family Nursing Conference, Odense, Denmark.*
- A peer-reviewed publication in an international Journal in August 2016; *Waldboth V., Patch C., Mahrer-Imhof R. & Metcalfe A. (2016) Living a normal life in an extraordinary way: A systematic review investigating experiences of families of young people's transition into adulthood when*

affected by a genetic and chronic childhood condition. International Journal of Nursing Studies 62, 44-59.

- A presentation at the Family Care Day for families living with muscular disease at the Swiss Paraplegic Centre in Nottwil;
- A presentation for researchers and lecturers at Florence Nightingale Faculty of Nursing and Midwifery, King's College London, UK, and ZHAW Institute of Nursing, Switzerland;
- and a lecture on the family transition experience when living with NMD for the Master of Advanced Studies in Paediatrics at ZHAW in December 2015.

The peer-reviewed article by Waldboth et al. (2016) (Appendix 1) is the most detailed report about the findings of this literature review, which also includes information on methods comprising a detailed description of the search strategy, literature selection processes, inclusion and exclusion criteria and data extraction and analysis. While this chapter does not want to repeat information unnecessarily, which can already be retrieved elsewhere, the emphasis here is to give an overview of methods and findings from the systematic literature with the focus on insights that informed and guided this subsequent Grounded Theory study that focused on families' experiences with NMD.

2.2. Methods

The study design chosen, a systematic literature review, is suitable to this study as it answers precise research questions and provides systematic procedures for identification, evaluation and synthesis of the findings of all relevant studies in an organised and reproducible way (Norman & Griffiths 2014). This

systematic review adopted an integrative approach to identify relevant articles and review information of qualitative, quantitative and mixed studies. By using an integrative approach information was considered valuable independent of the research methodology of the study, but it was selected on its merits to give better insights into an under researched area. According to Torraco (2005) and Whittemore and Knafl (2005) it is an approach suited to learning more about an emerging topic that would benefit from a holistic perspective, which is the family transition experience that is under study here.

Databases were searched from June to August 2014 among which PubMed, Cochrane Library, PsychINFO, CINAHL and AMED. Search terms used were 1) family, caregivers, young adult, adolescent; 2) adolescent development, transitional programs, transition to adult care; 3) muscular dystrophy, spinal muscular atrophy, cystic fibrosis, haemophilia and sickle cell disease. All identified articles were reviewed against their eligibility for inclusion and critically appraised using the “Qualsyst” standard quality assessment criteria (Kmet et al. 2004). Figure 3 represents the study identification and selection processes as well as the reasons for article exclusion.

Data from the articles that remained following the removal of duplicates, papers unrelated to genetic childhood conditions and families’ experiences of the transition were extracted and summarised in a table. Following data extraction, findings from the articles were analysed and synthesised using an interpretive approach to evidence synthesis, a variant of meta-ethnography called critical interpretive synthesis (Dixon-Woods et al. 2006, Pope et al. 2007). The benefits of combining qualitative and quantitative evidence in building a cumulative

knowledge base using interpretive synthesis has recently been reported (Barley et al. 2011, Metcalfe et al. 2008).

Figure 3: Literature selection process

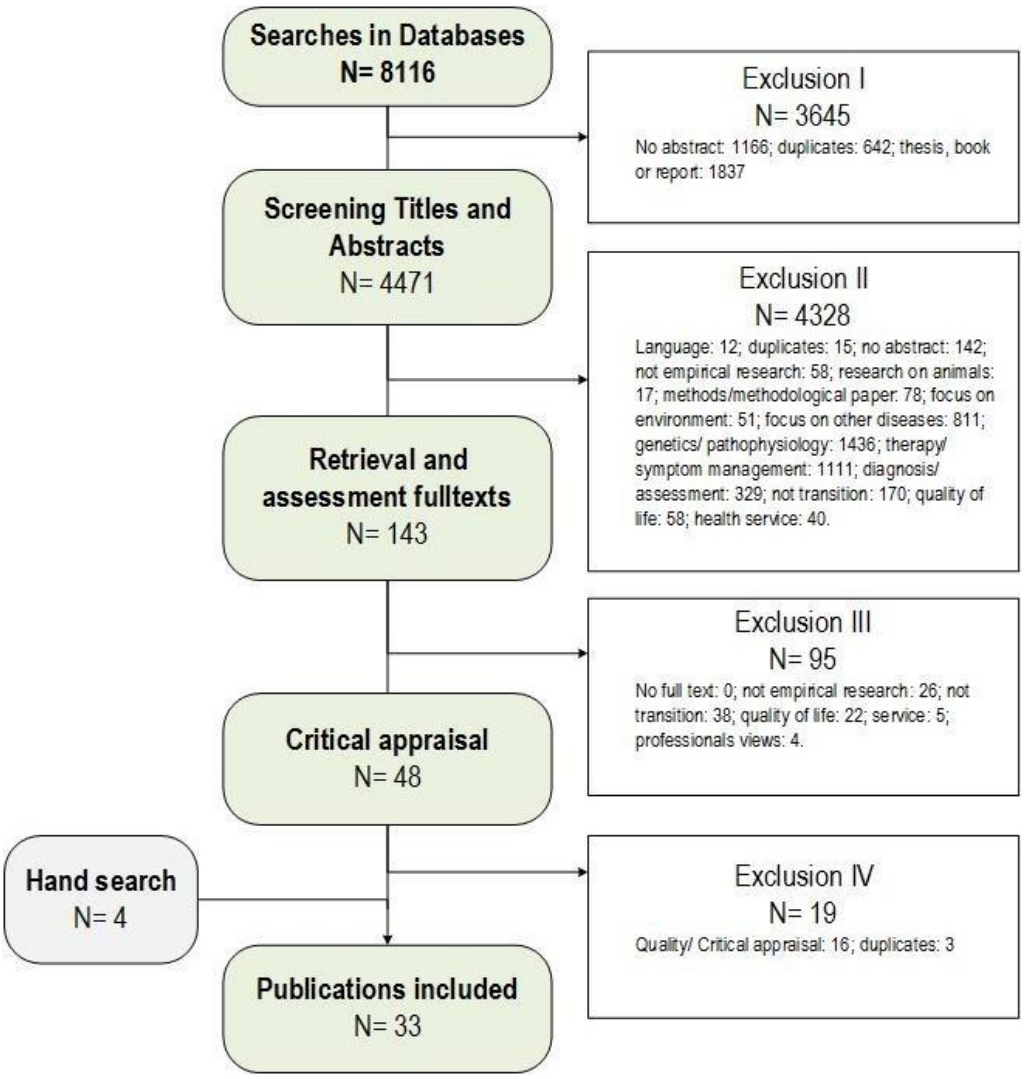


Figure from: Waldboth V., Patch C., Mahrer-Imhof R. & Metcalfe A. (2016) Living a normal life in an extraordinary way: A systematic review investigating experiences of families of young people's transition into adulthood when affected by a genetic and chronic childhood condition. *International Journal of Nursing Studies* **62**, p.47, illus.

2.3. Findings

In this literature review, a total of 8116 articles were retrieved from which 33 studies remained after reviewing them against the inclusion and exclusion criteria. 16 of these 33 articles involved individuals living with CF, ten with SCD,

seven with MD including Duchenne MD, and two with Haemophilia. 26 articles focused on one condition only and 7 articles were investigating two or more conditions. Studies have mostly been conducted in North America and Europe (18 in North America and Oceania, 14 in Europe, 1 Brazil). While only two of the articles were more than 20 years old, more than two thirds were published within the last decade (22). Three studies used a quantitative methodology (cross-sectional study designs), whereas the majority of the articles employed qualitative research designs (29). By and large, the quality of the studies was rated as good. For a more detailed description of methodological strengths and limitations and a tabular summary of the articles, consultation of Appendix 1 is recommended. A comprehensive tabular overview of the included articles and a more detailed description of this review's findings can also be found in Appendix 1, while in the following only a selection of themes will be presented. Table 2 portrays key themes and concepts which were extracted from the articles and characterise the affected young persons' and their families' experiences when a chronically ill family member transitions into adulthood.

Table 2: Family experiences: key themes and concepts

Reference	Family experiences: key themes and concepts
Admi (1996)	Family life, wider society, creation of identity, physical health, emotional and psychological health, self-management
Al-Yateem (2012)	Transfer to adult care
Antle et al. (2008)	Complexity of parenting a chronically ill child, concerns about the child's future
Atkin & Ahmad (2001)	Friends and peers, family life, wider society, education and career, creation of identity, physical health, emotional and psychological health, self-management, transfer to adult care, coping and adaptation strategies
Badlan (2006)	Education and career, creation of identity, physical health, emotional and psychological health, self-management, transfer to adult care, coping and adaptation strategies
Berge et al. (2007)	Family life, risky behaviour, independent living, creation of identity, physical health, emotional and psychological health, self-management, transfer to adult care, coping and adaptation strategies
Bregnballe et al. (2011)	Family life
Cappelli et al. (1989)	Family life, physical health, emotional and psychological health

Christian & D'Auria	Friends and peers, romantic relationships, creation of identity, self-management
Dreyer et al. (2010)	Friends and peers, romantic relationships, physical health, emotional and psychological health, coping and adaptation strategies
Dupuis et al. (2011)	Coping and adaptation, complexity of parenting a chronically ill child, concerns about the child's future
Erskine (2012)	Friends and peers, family life, wider society, creation of identity, physical health, emotional and psychological health, self-management, transfer to adult care
Gibson et al. (2007)	Friends and peers, romantic relationships, education and career, creation of identity, coping and adaptation strategies
Gibson et al. (2014)	Romantic relationships, wider society, independent living, creation of identity
Gjengedal et al. (2003)	Education and career, physical health, emotional and psychological health, self-management, complexity of parenting a chronically ill child, concerns about the child's future
Hauser & Dorn(1999)	Transfer to adult care, complexity of parenting a chronically ill child
Higham et al. (2013)	Romantic relationships, education and career, creation of identity, self-management
Johannesson et al. (1998)	Friends and peers, romantic relationships, creation of identity, physical health, emotional and psychological health
Moola & Norman (2011)	Romantic relationships, education and career, creation of identity, physical health, emotional and psychological health, self-management, complexity of parenting a chronically ill child, concerns about the child's future
Müller-Kägi et al. (2014)	Creation of identity, physical health, emotional and psychological health, self-management
Palmer & Boisen (2002)	Family, education and career, risky behaviour, creation of identity, self-management, coping and adaptation strategies
Parkyn & Coveney (2011)	Friends and peers, wider society, creation of identity, complexity of parenting a chronically ill child, concerns about the child's future
Pehler & Craft-Rosenberg (2009)	Friends and peers, creation of identity, physical health, emotional and psychological health, coping and adaptation strategies
Pizzignacco & de Lima (2006)	Romantic relationships, creation of identity, self-management, coping and adaptation strategies
Porter et al. (2014)	Transfer to adult care, complexity of parenting a chronically ill child, concerns about the siblings future
Schmitt (Germany)	Friends and peers, creation of identity, coping and adaptation strategies
Telfair et al. (1994)	Transfer to adult care, concerns about the child's future
Telfair et al. (2004)	Transfer to adult care
Tuchman et al. (2008)	Transfer to adult care
Valenzuela et al. (2013)	Friends and peers, family life, creation of identity
van Staa et al. (2011)	Transfer to adult care, complexity of parenting a chronically ill child, concerns about the child's future
Williams et al. (2009)	Creation of identity
Witte (1984)	Friends and peers, family life, wider society, concerns about the child's future

Table adapted from: Waldboth V., Patch C., Mahrer-Imhof R. & Metcalfe A. (2016) Living a normal life in an extraordinary way: A systematic review investigating experiences of families of young people's transition into adulthood when affected by a genetic and chronic childhood condition. *International Journal of Nursing Studies* **62**, p.49

Findings provided insights into three perspectives: 1) the young person's perspective on how to "live a normal life in an extraordinary way" and "manage a chronic and life threatening disease"; 2) the parent perspective on the "complexity of being a parent of a chronically ill adolescent" and "concerns about the child's future" and 3) the sibling perspective on "concerns about the sibling's future" (Figure 4). Overall, the perspective of the affected young person has received great research attention, while there were far fewer insights into the parents' and siblings' experience, or the experiences of the family as a whole.

Figure 4: The three perspectives

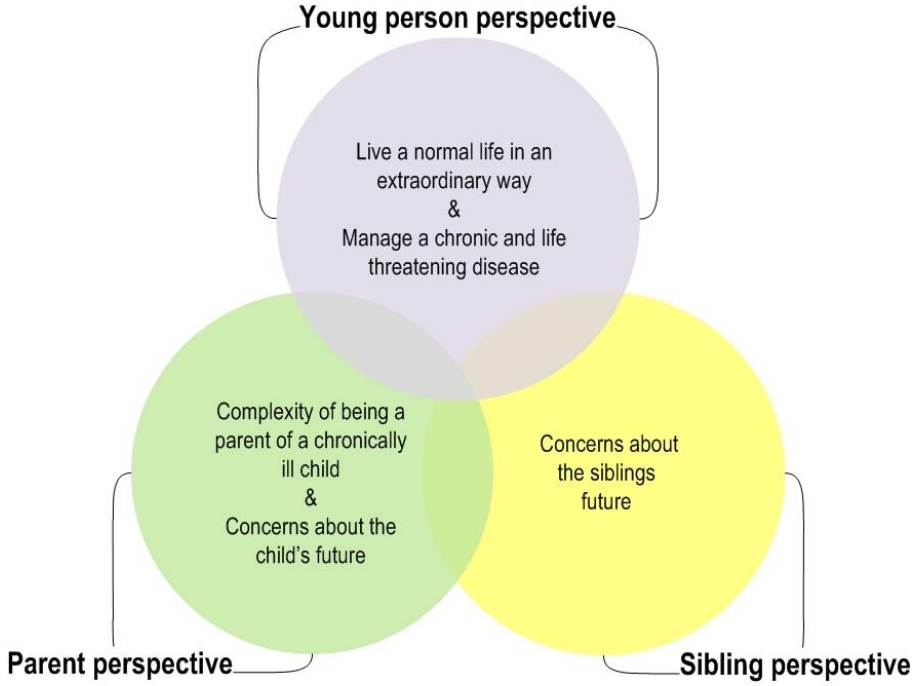


Figure from: Waldboth V., Patch C., Mahrer-Imhof R. & Metcalfe A. (2016) Living a normal life in an extraordinary way: A systematic review investigating experiences of families of young people's transition into adulthood when affected by a genetic and chronic childhood condition. *International Journal of Nursing Studies* **62**, p.53, illus.

The transition into adulthood was a time when affected young people wanted to take part in major life pursuits similar to their peers including contacts with friends and peers, engagement in romantic relationships and focus on education and career choices (Atkin & Ahmad 2001, Badlan 2006, Christian & D'Auria 1997, Dreyer et al. 2010, Dupuis et al. 2011, Gibson et al. 2007, 2014, Gjengedal et al. 2003, Higham et al. 2013, Moola & Norman 2011, Müller-Kägi et al. 2014, Palmer & Boisen 2002, Pizzignacco & de Lima 2006, Schmitt 1997). In addition, adolescence was characterised by the affected young persons' formation of their own identities as well as their growing independence from their families (Bregnballe et al. 2011, Valenzuela et al. 2013, Williams et al. 2009, Witte 1985).

Considering the affected young person's physical health situation, accomplishing some of these goals and related activities was more difficult for them than for their healthy counterparts. To give an example, findings from this literature review indicate that many of the young people were struggling with confusion about their own identities and experienced difficulties in social integration. Young people, for instance, had negative social experiences at school, work or through discrimination by wider society, which led to feelings of isolation and social withdrawal (Pehler & Craft-Rosenberg 2009, Valenzuela et al. 2013, Witte 1985).

A factor that influenced the affected young persons' behaviour was the visibility of the condition. Young people with less visible impairments tried not to disclose their disease, which led to problematic relationships in the longer term (Christian & D'Auria 1997, Johannesson et al. 1998, Pizzignacco & de Lima

2006). When searching for a romantic partner, for example, some young people living with CF kept their illness a secret to increase their chances of finding a partner. If regular contacts or romantic relationships developed, however, they could not maintain the concealment of their condition. Depending on how young people dealt with these situations, attempts to keep secrets had the potential to create interpersonal tensions, loss of trust once the truth was out and separation (Christian & D'Auria 1997, Johannesson et al. 1998, Pizzignacco & de Lima 2006). The visibility of the condition also influenced the young person's identity. Behaving authentically was less problematic when the disability was more visible, whereas when the condition could be hidden young people denied part of themselves and struggled to integrate the disability into their lives.

While affected young people were developing and maturing into adults, they had to manage a chronic and life threatening illness. Living with a chronic illness imposed many challenges on their and their families' lives; Coping with physical, emotional and psychological difficulties and mastering a complex disease management were among the tasks they had to accomplish (Admi 1996, Al-Yateem 2012, Atkin & Ahmad 2001, Berge et al. 2007, Capelli et al. 1989, Dreyer et al. 2010, Erskine 2012, Gjengedal et al. 2003, Johannesson et al. 1998, Pehler & Craft-Rosenberg 2009, Telfair et al. 1994, Telfair et al. 2004, Tuchmann et al. 2008).

Parents of affected adolescents engaged in traditional parenting activities, while they were also assisting with disease management and worrying for their children's future (Antle et al. 2008, Hauser & Dorn 1999, Parkyn & Coveney 2011, van Staa et al. 2011). Most parents found it difficult to detach and launch

their affected children into independence. Overprotective parenting behaviour resulted in strained family relationships, disagreements and ambivalent feelings towards each other and affected young people wished for a more open parenting style that would allow for more freedom. Healthy siblings adopted a caring attitude for their chronically ill siblings, and were also concerned about their future and ability to care for themselves (Porter et al. 2014).

2.4. Conclusions

This literature review allowed for the identification of main themes and concepts that gave valuable insights into the individual family members' transition experience when living with chronic illness. This knowledge informed further methodological decisions, such as the choice and planning of data collection strategies. However, findings from this review resulted in few insights into the parents and siblings' perspective, which makes it difficult to gauge the full extent of negative outcomes they face, despite being clearly present.

In addition, there is a lack of information on family members' relationships and interactions, as well as on their mutual influences on each other, including experiences of the parental dyad and on siblings' relationships. The quality of family relationships and communication shapes how families cope with chronic illness, how they provide the care that is required due to the physical dependence of the affected young person and how they mutually support each other to reach developmental tasks, without limiting each other's opportunities and choices.

Therefore, a more comprehensive understanding of the family transition experience, including an exploration of effects on family relationships and interactions is required, as we need to learn more about their functioning as a group within their specific contexts. In the following chapter, family theories will be discussed in order to frame a research study that allows for the exploration of the family transition experience when living with NMD.

3. Chapter 3: The Theoretical Framework

A theoretical framework can give deeper insights into family functioning and family processes and set out what is already known and established. It also reveals where gaps in the evidence base are that need to be explored by a consequent study. Moreover, theories can function as guidance for methodological decisions and for the interpretation of study findings, by putting choices and claims into context. To illuminate major theoretical assumptions that framed this research, theories on the functioning of family systems, their coping and adaptation to chronic illness and family development over the course of life will be introduced and discussed.

3.1. Considering Different Theoretical Frameworks

Theories that have been considered as theoretical frameworks for this research on the family transition experience when living with NMD were:

- family social science theories,
- family therapy theories and
- nursing conceptual frameworks.

Family social science theories are the most developed among these theories and they are also very informative (Kaakinen & Hanson 2010). They are informative as they have a broad focus on the family within its context, view individual family members as connected with each other, and explain how families relate to other systems (e.g. the society, the health and social care system). Social science theories are well suited to investigate certain family phenomena including family functioning and interaction, family members' reactions to health and illness, families' change over time and interchange with their larger environment (Kaakinen & Hanson 2010).

Family therapy theories and nursing conceptual frameworks, on the contrary, are newer and therefore also less evolved than social science frameworks. They can give fewer insights than social science theories, also because they have a slightly different and narrower focus. Family therapy theories, for instance, emerged from a practice discipline of family therapy where they were developed to work with families in problematic situations (Kaakinen & Hanson 2010). They focused on family dysfunction and described and explained observations and suggested interventions to inform clinical practice. Although family therapy theories can give some valuable insights into family dynamics and pattern of behaviour related to the illness experience, they are less comprehensive than social science theories and therefore more limited in considering the family within its larger context and development over time.

Among the above introduced theories, nursing conceptual frameworks (e.g. Parse's Human Becoming Theory, Orem's Self-Care Deficit Theory, Watson's Theory of Caring) are the least developed. They evolved during the 1960s and 1970s where emphasis was placed on developing discipline-specific models for nursing (Kaakinen & Hanson 2010). They therefore represent an important part of the nursing tradition, but have been widely criticised as conceptual frameworks for research since nursing conceptual frameworks follow a rather deductive approach and have not been further developed since their introduction. Therefore, nursing conceptual frameworks can give some insights, but they offer limited guidance for family research.

It is an imperative that family research that informs future scholars and health care professionals builds on a sound body of knowledge which stems from the best theory available (Kaakinen & Hanson 2010). What this study requires is a robust theory that guides methodological decisions and interpretation of results by providing information on family functioning and family development. This theory needs to be compatible with inductive knowledge generation to give new insights into a still under researched area. As family social science theories are the most developed and comprehensive theories that focus on family function and relationships, they are the best fit for this research on the family transition experience when a young family member is living with NMD.

3.2. Family Systems Theories

Family social science theories that informed and guided this study and will be presented in the next section were: Family systems theories and symbolic interactionist thoughts, different theoretical perspectives on families and chronic illness, family development and family-environment interchanges (Kaakinen & Hanson 2010). Despite their differences in thought, all of these theories build around key assumptions of systems theory. In other words, systems theory is a widely used theoretical framework whose assumptions are inherent to many other social science theories. Family systems theory is therefore well suited as major theoretical framework for guiding the following empirical work and it will be complemented by other social science theories to create a more holistic picture.

3.2.1. General Systems Theory

Family systems theory derived from general systems theory in the beginning of the 20th century (McDaniel et al. 2006, Segrin & Flora 2011). General systems theory is an overarching line of work that is thought to cut across traditional academic boundaries with the aim of developing a framework that organises general theories from all disciplines enabling the discussion of the relationships between these disciplines and the whole empirical world (Boulding 1956). In other words, it explains how systems function and how they relate to other systems. Because of this comprehensiveness, general systems theory's level of abstraction lies somewhere *"between the specific that has no meaning and the general that has no content"*, but searches for an *"optimum degree of generality"* (Boulding 1956, p. 197-198). To give a concrete idea of general systems theory, Figure 5 depicts some of its principles which have been applied to a medical patient situation by the internist Georg Engel (1980) aiming to describe the interrelation between systems. In this figure the effects of a medical event on the organismic (organs and cells) and social hierarchies (family and community) are illustrated. While the family has to cope with the threat of loss by death due to the cardiac arrest, on an organ systems level cells and tissues experience anoxic damage.

Figure 5: Interrelation between systems in a biomedical model

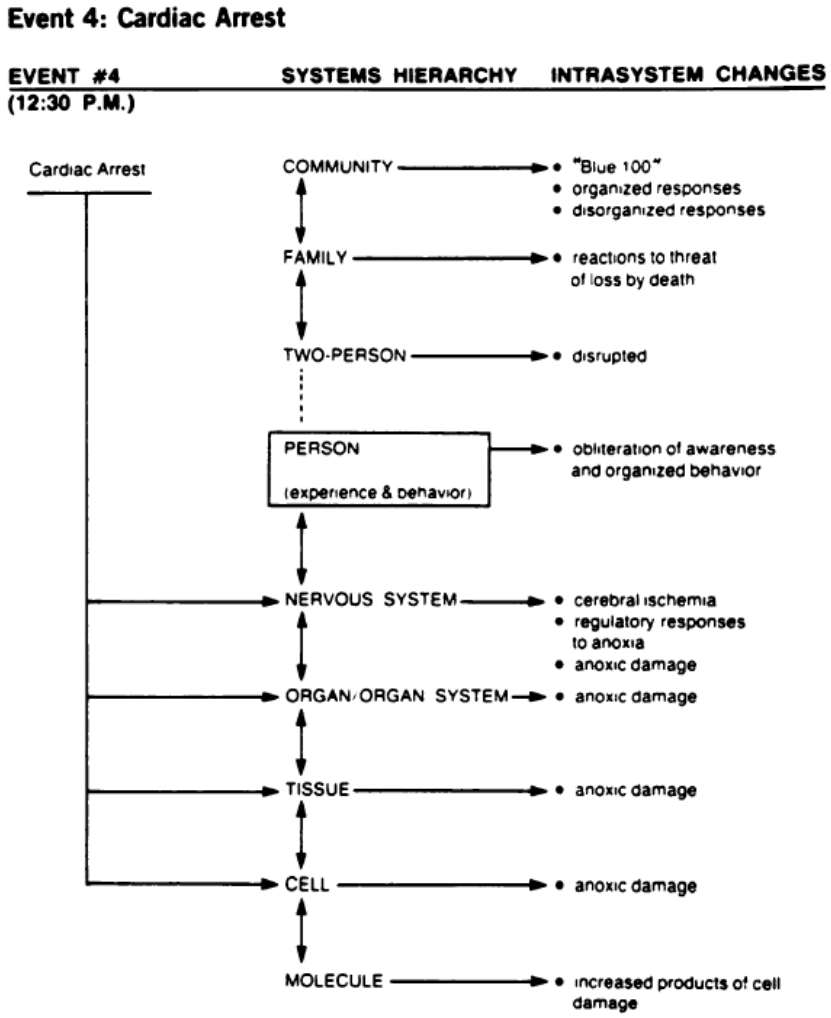


Figure from: Engel G.L. (1980) The clinical application of the biopsychosocial model. *The American Journal of Psychiatry* **137**(5), p.537, illus. (Republished with permission of The American Journal of Psychiatry; permission conveyed through Copyright Clearance Center, Inc.)

Major contributors to general systems theory in their disciplines were the biologist Ludwig von Bertalanffy (1968), the mathematician and engineer Norbert Weiner with his work in cybernetics (1948), and the sociologists Talcott Parson (1949) and Niklas Luhmann (1927-1998) (Broderick 1971, Kean & Gehring 2001, Runkel 2012, Segrin & Flora). Luhmann's work is very informative for gaining a better understanding of family functioning by viewing families as social systems. He is a well-known representative of sociological systems theory who developed Parson's action-based structural functional

theory further, by reversing it into a functional structural theory based on communication. According to the latter, structures develop from functions and social systems are oriented towards meaning by use of communication (Runkel 2012).

The structural functional approach assumes that an entity is a system, when it distinguishes itself from the environment (Luhmann 2006). Boundaries are needed that separate the system from its environment. These boundaries are called the “form” of the system and are regarded as the central characteristic that defines the system and constitutes its identity (Luhmann 2006). A system is therefore not made by its essential parts or constituting elements, but it is made by the form that distinguishes the “inside” from the “outside”. There must be a balance between the system and the environment which keeps the system working or alive, respectively (Luhmann 2006). This balance needs to be kept even if there are structural changes happening. The identity of a system requires continuity in sense of maintenance of the form, not the structure.

Luhmann (2006) describes communication as a main feature of social systems. Social systems are in need of communication, which is viewed as an operator that is able to reproduce a social system’s essential difference from its environment. Communication has three characteristics that predispose it to this special role: 1) it can function on its own, 2) it always remains what it is and 3) it possesses connectivity. In other words, communication develops from communication and it has the special property of being able to connect things (Luhmann 2006). At the same time as communication transcends the system it remains an internal feature. Communication may refer to itself, the external

world or usually both at the same time, but it always remains part of the system (Luhmann 2006). A system must be able to control its own conditions of connectivity.

3.2.2. Characteristics of Family Systems

Theoretical explanations inherent to family systems were abstracted from general system's theory (McDaniel et al. 2006, Segrin & Flora). Among the first who applied systems theory principles to families were Gregory Bateson (1956) and Don D. Jackson (1965) with their study of patients with schizophrenia and the patients' relationships with their families (Bateson et al. 1956, Broderick 1971). Other famous contributors were Nathan Ward Ackerman (1959), Salvador Minuchin (1974) and Murray Bowen (1978).

General systems theory claims that all parts of a system, for example, the planetary system are connected through invisible ties and that the single parts of this system influence each other. Consequently, family systems theory suggests that families are systems where individual family members relate, interact and mutually influence each other (Feetham & Thomson 2006). Families are constituted by individual family members (structure), but characterised by holism (function), which implies that they must be understood as a whole rather than as a sum of parts (Whitchurch & Constantine 1993). To clarify, families have qualities that emerge out of the relationships and interactions between family members, but do not derive from the component parts themselves. As a consequence, families cannot simply be comprehended by examining individual family members (Broderick 1993, Whitchurch & Constantine 1993).

Furthermore, families have boundaries, but these boundaries are not closed, but open to influences from the outside (Kean & Gehring 2001). A family system takes inputs from the environment and vice versa, but different families vary to the extent of their openness depending on their specific situation such as their pattern of interaction and family rules (Broderick 1993). Family members regulate the flow back and forth across the border via internal rules. If the family, for example, has an open pattern of interaction with the environment, outsiders are welcome to enter the system. However, if the family is more closed this will be made clear to anyone who wants to exit or enter. The only way into the family system, however, is through birth, adoption or marriage, and the only way out is through death (Carter & McGoldrick 2005).

Besides being open, a family is ongoing (Broderick 1993). Families are never static, but they change over time as they constantly engage with a dynamic environment and incorporate inputs and produce outputs (external contacts). Furthermore, they are connected with each other and with previous and future generations and therefore have a past, present and future (internal contacts) (Segrin & Flora 2011). In order to survive, family members have to regulate the relationships among each other and their relations with their larger environment, and they do so by applying rules that regulate external and internal contacts. They intend to balance energies between bridging to the outside, while protecting the family boundaries and by being bonded to each other, while also maintaining each family member's integrity (Broderick 1993).

In addition to being open and ongoing and therefore dynamic and changing over time, there is another characteristic inherent to social systems, which makes them distinctive from other systems; their ability for self-reflexivity (Whitchurch & Constantine 1993). Being self-reflexive implies having the ability to transcend humanness and to look at oneself or a person from the outside, by examining and explaining behaviour. Self-reflexivity is only possible because of communication, which facilitates the creation of meaning and symbolic interaction. Communication is defined as *"the synthesis of information, uttering and understanding"* and happens *"when information that has been uttered is understood"* (Luhmann 2006, p. 47). Communication is bound to comprehension; it occurs when there is somebody else who understands the messages of symbolic content (Whitchurch & Constantine 1993). Besides utterances, also non-verbal expressions and behaviour can have informative value (Bavelas 1990). Family members, for instance, share common experiences and reach a level of intersubjectivity or shared meaning that allows them to understand what the other person is thinking or referring to even if no words are used (Segrin & Flora 2011). Symbolic interaction and family meaning are best illuminated by a symbolic interactionist perspective, a line of thought that also assumes a systems perspective.

3.2.3. Symbolic Interactionist Assumptions

Symbolic interactionism is based on works of Georg Herbert Mead and his student Herbert Blumer (1969), whose ideas further developed pragmatist thoughts on human interaction and symbolic meaning (Blumer 1969, Hill & Hanson 1960, Segrin & Flora 2011). They described symbolic interactionism, a theory that influenced many other theorists such as, for instance, Grounded

Theorist's (Charmaz 2014). Mead and Blumer were interested in the way how people interact by use of symbolic communication and how meanings influenced their actions and were redefined and depended on the individuals' perception and their social environment. According to symbolic interactionism, people act and interact because of meaning and they construct shared meanings during symbolic interactions (Blumer 1969, Segrin & Flora 2011). Family meanings are defined as *"the interpretations, images, and views that have been collectively constructed by family members as they interact with each other; as they share time, space and life experience; as they talk to each other and dialogue their experiences"* (Patterson & Garwick 1994, p. 1). As a prerequisite, there are shared signs, symbols and interpretations that convey meanings which are culturally and historically sensitive. Through an interpretive process, which is in contrast to a social process internalised, these meanings are modified (Blumer 1969). During the process of interpretation a person points out the things that are meaningful to him or her and selects and revises these meanings according to the specific situation he or she is in.

Family meanings are not just socially constructed through symbolic interaction, they also inform action. Individuals act towards each other on behalf of the meanings others have for them (Blumer 1969). These others might be other individuals, other living beings (e.g. animals, plants), situations or institutions (Runkel 2012). If somebody has an affinity for animals this person's interaction with animals may be characterised by friendly behaviour. In contrast, if an individual thinks that a specific animal might be a threat to his or her life, there might be a completely different interaction taking place such as attack or escape. Meaning and as a consequence behaviour of individuals confronted

with the same situation will probably differ most of the time. It is not only human action and interaction that is influenced by meaning, but also how we perceive ourselves (Blumer 1969, Segrin & Flora 2011). Self-concepts develop during interactions with others and are strong motivators for action. In light of the above, symbolic interactionist assumptions are relevant to a better understanding of how families make meaning of different life situations and how they communicate verbally and non-verbally by use of shared signs, symbols and interpretations (Blumer 1969, Segrin & Flora 2011). The way they communicate and make meaning influences how they are coping and making adjustments when confronted with chronic illness and developmental tasks.

3.3. Theoretical Perspectives on Families and Chronic Illness

As families move through the life cycle they have to adjust to life cycle transitions and developments and need to cope with upcoming stressors (McGoldrick et al. 2013b). Chronic illness is such an unpredictable stressor that affects the individual and his or her family system. When families are confronted with chronic illness, Patterson (1991) argues for a circular sequence of effects between an affected child and the family system (Figure 6). When disability occurs, it has an impact on the health and development of the child, but families also face extra challenges that depend on the type of condition, such as the course of disease, prognosis and type and degree of physical or psychological limitations. Depending on the specific family situation and their response, there may be negative consequences for the family including increasing caretaking demands, emotional challenges and psychosocial problems (Patterson 1991).

Figure 6: Circular sequence of effects

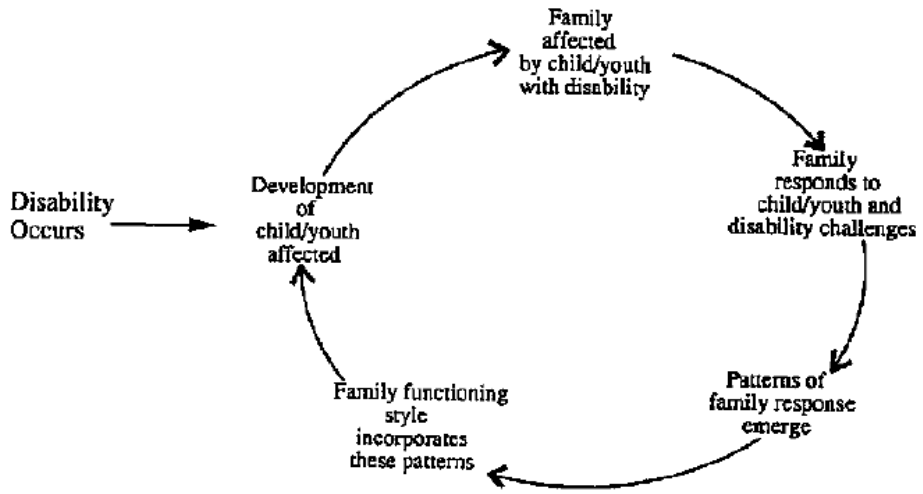


Figure from Patterson J.M. (1991) A family systems perspective for working with youth with disability. *Pediatrician* **18**, p.131, illus. (Republished with permission of S. Karger AG, Basel, Switzerland)

In contrast, families confronted with chronic illness may also experience positive impacts. Family units are reported to be more cohesive and to have a greater sense of what is important in life (Patterson 1991). Family members living with a chronically ill relative are also described as being more socially competent, having greater tolerance and empathy and becoming stronger and more resilient with time. According to Patterson's sequence of effects, family response is patterned and family members develop a coping style, which shapes how the family functions and how they deal with life cycle transitions such as the affected child's transition into adulthood.

3.3.1. Family Resilience and Functioning

Family resilience is the quality of bouncing back and moving on in life after experiencing health problems or normative life transitions such as developmental tasks (Earvolino-Ramirez 2007, Kaakinen et al. 2010, McCubbin & McCubbin 1988). Key processes involved in family resilience and related

coping and adaptation processes are personal strengths, family resistance and support by others such as the community. Among the influencing factors on family resiliency are also family beliefs, pattern of communication and family organisation when managing everyday life (Kaakinen et al. 2010, McCubbin & McCubbin 1988). Positive consequences of high family resilience are effective coping, mastery, and positive adaptation to change (Earvolino-Ramirez 2007).

An adequate family response allows healthy family functioning which, in turn, may have a positive impact on the individual family member's development and on disease management (Patterson 1991). Family functioning has to do with the purposes that the family serves in relation to the individual and the family group (Kaakinen et al. 2010, Segrin & Flora 2011). General functional aspects include family tasks such as nurturing (e.g. basic care, emotional and financial support) and socialising, comprising the ways a family reproduces offspring, interacts to socialise its young, cooperates to meet economic needs, and relates to the larger society. Specific family demands, however, may vary according to the specific family situation and developmental stage and they are defined by equifinality. Equifinality means that the same goal or the same state can be reached in different ways and that there is no single way how a family can function (Kean & Gehring 2001, Segrin & Flora 2011, Whitchurch & Constantine 1993).

The family systems theorist David Olson developed a theoretical model of family functioning; the Olson Circumplex Model (Figure 7) which suggests that there are three dimensions that are crucial to family functioning: family cohesion (the degree of emotional bonding and individual autonomy), family flexibility (the

family's ability to change and adapt) and family communication (Olson & Gorall 2003).

Figure 7: Circumplex model

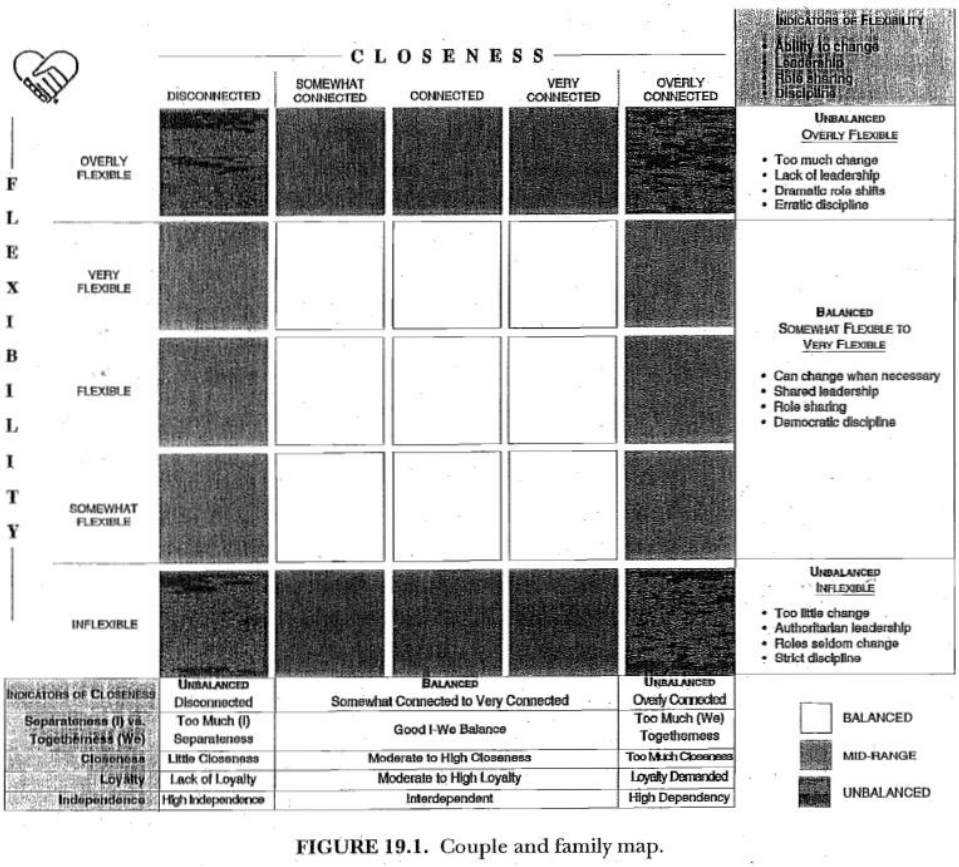


FIGURE 19.1. Couple and family map.

Figure from: Olson D.H. & Gorall D.M. (2003) Circumplex model of marital and family systems. In Normal Family Processes (Walsh F. ed.) Guilford, New York, pp.517. illus. (Republished with permission of Guilford Press)

Family flexibility is viewed on a continuum from overly flexible to inflexible, whereas family cohesion types can be disconnected, somewhat connected, connected, very connected or enmeshed (Olson & Gorall 2003). As a result, two types of family functioning can be distinguished: The three middle levels are each considered balanced, whereas the two extremes represent unbalanced family functioning types. Communication as the third dimension allows families to move on the flexibility and cohesion dimensions, in the face of situational or developmental stresses (Olson & Gorall 2003).

3.3.2. Family Relationships and Pattern of Interaction

The family is the primary social group which we are born into and where there are lifelong bonds. According to John Bowlby's work (1944), a child's relationship with its mother and father is of immediate importance to the child due to evolutionary pressures and it is shaping the child's later functioning (Cassidy 2008, Vetere 2008). It is assumed that attachment patterns are learned in childhood and persist throughout life, but opposite developments in adolescence can alter young people's behaviour. Attachment styles are inner working models that can tell us something about an individual's response to stress (Bretherton & Munholland 2008, Vetere 2008). There are secure attachment patterns that are characterised by the expression of positive and negative feelings by acknowledgement, reflection and negotiation and insecure attachment styles including avoidant, ambivalent or preoccupied behaviour.

Family members relate and communicate with each other and process shared information in order to comprehend it (Segrin & Flora 2011). Family members also communicate nonverbally by observing each other and they correct each other's behaviour through positive and negative feedbacks that are important communication tactics for the longer term, for instance, in order to reach a common goal. Families use stories, which are verbal accounts of important personal experiences that provide information on relationships, rules of interaction, and family beliefs (Pratt & Fiese 2004, Segrin & Flora 2011). Families naturally tell stories related to their meaning making, by remembering and interpreting past events and socialising or connecting with other

generations. However, stories can both give or hide information (e.g. through family secrets). The latter is a family strategy to control the flow of information.

Family members develop patterns of interaction, a set of communication strategies, and they have a patterned response when coping with challenges to find some degree of stability in their everyday lives. These patterns make a family unique, even if family structure is similar (Denham 2005, Kaakinen et al. 2010). Patterns of interaction are not stable, but are evolving over time and influenced, for example, by life cycle transitions such as the transition into adulthood and by events like chronic illness. Families need to evolve and change over time as part of a natural evolution process where relationships change and patterns of interaction alternate.

Family rituals have been described as patterns of interaction which have a stabilising impact on family life (McCubbin & McCubbin 1988). Family rituals and routines give organisation and meaning to family life and have also a potential impact on family health (Kaakinen et al. 2010). Family routines are defined as continuous behaviours related to everyday life such as mealtimes, whereas rituals are associated with symbolic communications during celebrations and as part of traditions. Rituals are a means to establish boundaries, signalise transitions and develop identity (Patterson & Garwick 1994, Segrin & Flora 2011). They have a facilitating factor for family stability and balance.

Everyday family life requires decision making (Kaakinen et al. 2010). Decisions are made by raising the issue, talking about the issue, supporting action, giving

importance to options and responding to it. Various models explain decision making within families. Turner (1970), for example, describes three decision making styles that are termed consensus (all family members agree before a decision is made), accommodation (adapting to the decision of another person) or de facto (when a decision resolves itself or the situation decides); Settles' (1999) model of decision making, on the other hand, explores the degree of choice that families have when making decisions (Segrin & Flora 2010). Factors that have an impact on the area of choice according to this model are manifold: the degree to which families are aware of different decision options, the number of family members with different roles involved, the families' resources, their mobility and flexibility, the individuals personality and skills, the constraint by families rules and societal norms and other events that families have no influence over.

Decision making is a joint family process that is influenced by power. Power is defined as the ability or potential to change the behaviour of another person (Levine & Boster 2001, Segrin & Flora 2011). Power occurs when two or more individuals interact. Processes related to power are influenced by perception, culture, past experiences and dependence. Children, for instance, are dependent on their parents, while parents hold power over them. Family rules are part of power processes and are classified into overarching paradigms, midlevel policies, meta- and concrete rules (Broderick 1993). The level of abstraction is highest in overarching rules and lowest in concrete rules, while meta-rules are rules about rules. A concrete rule may be to open the door when somebody knocks, whereas a meta-rule may relate to specific situations that lead to not opening a door when someone knocks (Broderick 1993).

Power relations and difficult decision making can lead to family conflicts (Segrin & Flora 2011). Whether the conflict is about a topic or a relationship is most of the time not definable. A moderate amount of conflict usually occurs in all families, while the conflict frequency is not a predictor for family dysfunction as some family members thrive from conflict but it is dependent on whether everybody knows the rules (Segrin & Flora 2011). In fact, the balance between conflicts and positive interactions and conflict strategy is more important for family function than the frequency of conflicts only. In case of family interaction conflicts usually go beyond two persons. A conflict strategy used in these kinds of situations that is not considered very effective is triangulation (Buehler & Welsh 2009). In these situations family members, who are in conflict, draw another person into it and this person may experience tensions as a consequence of being made part of the conflict. Triangulation of children into parental conflict, for instance, may have a severe negative impact on the children and can cause psychosocial difficulties.

3.4. Family Systems and Genetic Illness Model

Another comprehensive way of thinking about families and genetic illness is offered by the Family Systems and Genetic Illness Model (FSGI) (Rolland 1987, Rolland 1994, Rolland 2006a, Rolland & Williams 2005, Rolland & Williams 2006). The more recent FSGI-model integrates Rolland's current and former Family Systems Illness Model (FSI). The models are in line with family systems theory and comprise a categorisation for genetic disorders considering biopsychosocial factors and emphasising psychosocial impact. Besides the focus on genetic disease and the family perspective, a third important

dimension of this model is time, which is revealed by the inclusion of the course of the disease and related developmental tasks (Rolland 1987, Rolland 1994, Rolland 2006a, Rolland & Williams 2005, Rolland & Williams 2006). The FSGI-model was not primarily created for traditional medical treatment, but for examining the relationships between a genetic disease and individual and family variables aiming to understand influences on coping and adaptation and implications on the family. It is therefore, well suited as a theoretical framework for this study.

3.4.1. Psychosocial Illness Types

The FSGI-model is a categorisation with 36 potential psychosocial types of genomic disorders (Rolland & Williams 2005, Rolland & Williams 2006). This typology differentiates among the following four major variables: 1) Likelihood of development of a condition; 2) Overall clinical severity of the disease; 3) Clinical onset and 4) existence of effective treatment options (Table 3). The likelihood of development is based on the genetic mutation and can be distinguished in high, variable and low likelihood. This variable is influenced by the penetrance of a genetic mutation and the interaction of complex and multiple genes and other environmental factors (Rolland & Williams 2005, Rolland & Williams 2006).

Overall clinical severity of the disease can be categorised into high and low severity of clinical symptoms (Rolland & Williams 2005, Rolland & Williams 2006). This single new variable of the FSGI-model gives a quick overview of the disease burden for individuals and their families. Genetic conditions can be classified by the timing of clinical onset within the life cycle (Rolland & Williams 2005, Rolland & Williams 2006). There are three possible points: onset in

childhood and adolescence (0-20 years), onset in middle adulthood and childrearing years (20-60 years) and onset in old age (60+). Finally, the model includes a dualistic variable that considers if an effective treatment or intervention exists that can alter the onset of the disease or the disease progression or not (Rolland & Williams 2005, Rolland & Williams 2006).

Table 3: Typology of genetic conditions

TABLE 1
Psychosocial Typology of Genomic Disorders

		Timing of Clinical Onset					
		Child/Adolescent 0-20		Early/Mid Adulthood Childrearing 20-60		Later Life >60	
Treatment Can Alter Onset or Progression		Yes	No	Yes	No	Yes	No
	Clinical Severity						
Likelihood of Development High	High	Hemophilia	Tay Sachs Disease		Huntington Disease Alzheimer Disease (early onset)		
	Low	Hemophilia					
Variable	High			Breast Cancer (HBOC linked) Hemochromatosis		Breast Cancer (HBOC linked)	
	Low			Hemochromatosis			
Lower	High						Alzheimer Disease (APOE e4allele linked)
	Low						

Table from: Rolland J.S. & Williams J.K. (2005) Toward a biopsychosocial model for 21st-century genetics. *Family Process* **44**(1), p.8, illus. (Republished with permission of John Wiley and Sons, Inc; permission conveyed through Copyright Clearance Center, Inc.)

3.4.2. Phases of the Illness

According to the older FSI-model, chronic illness is characterised as an ongoing process with landmarks, transitions and changing demands including three major phases of post-clinical onset: Crisis, chronic and terminal (Rolland 1987, Rolland & Williams 2006). In addition to the FSI-model, the FSGI-model expands the time phases of genetic disease and includes four non-symptomatic phases as well as the original post-clinical onset phases (Rolland & Williams 2005, Rolland & Williams 2006, Rolland 2006a).

The non-symptomatic phases include the awareness, pretesting, testing and post-testing and the long-term adaptation phase. During the awareness phase there is some knowledge of a possible genetic risk (Rolland & Williams 2005, Rolland 2006a). Within many families, multigenerational stories about the genetic risk and concerns about vulnerability evolve. Genetic testing is a method to clarify eventual uncertainties about genetic risk. The family or individual family member considers testing during the first crisis phase: pretesting. Decision making processes, communication and family dynamics can affect the experience during this stage, which is also dependant on the type of genetic condition (Rolland 2006a). Testing and the result of the test can cause a second crisis. There are several developmental challenges including acceptance of the new health situation and its consequences, grieving about loss and identity change, making meaning and developing mastery and competency in disease management and flexibility in the face of uncertainty (Rolland 2006a). The long term adaption phase lasts from the positive test until the condition manifests. Challenges of this phase are recognition of the impact of the disease on family members' development, probable future loss, finding

meaning in the situation and remaining current regarding new information about genetic risk and advances in preventive or symptomatic treatment (Rolland 2006a). There are several transition periods that link these four non-symptomatic phases and finally link with the post-clinical onset phases.

The post-clinical onset phase is referred to as crises and it comprises pre-diagnosis with symptoms, the diagnosis and the initial adjustment period that is following it (Rolland 1987). Learning how to deal with symptoms and care team, creating meaning of the illness and acceptance of the situation as well as grieving for loss are among the major challenges of this period. The time between the crisis and terminal phase is called chronic phase and it is characterised through stability, progression or changes (Rolland 1987). Maintaining the idea of living a normal life and preserving autonomy for all family members while increasing mutual dependence and caretaking are the major challenges for families during this stage. The following terminal stage comprises the pre-terminal phase, the death and the mourning and resolution of loss after the death of a family member (Rolland 1987)

3.4.3. Family and Illness Concepts

Major concepts used to describe family and illness developments in the FSGI-model are structure building or maintaining and structure changing periods (transitions) and centripetal versus centrifugal family styles. While during structure building periods following a transition families try to reform a life structure and enrich life according to previous experiences, during transitions they are vulnerable and face challenges and change (Rolland 1987, Rolland & Williams 2005). Family styles can be centripetal, characterised by family

closeness and cohesion or centrifugal, characterised by family disengagement. Structure building or changing periods can have either centripetal or centrifugal effects (Rolland 1987). Other family components that are considered in the FSGI-model are the transgenerational history of illness which brings light into the families present behaviour by considering past key events, pattern of coping and adaptation and family beliefs and expectations (Rolland 1987, Rolland & Williams 2005). The transgenerational history of illness also puts the family in the context of time where developments are viewed as part of the natural life cycle.

3.5. Family Development over the Course of Life

The temporal evolvement of a family system is best explained by the family life cycle theory, a family developmental perspective. The life cycle is the natural context within which individuals and families develop and where effects of an illness on a systems level are accounted for (Carter & McGoldrick 2005, McGoldrick et al. 2013b, Kaakinen & Hanson 2010). A family developmental perspective combines individual family member's developmental tasks and acknowledges the families progression towards a maturity of interdependence, where individual family members are experiencing different transitions and mutually influence each other.

3.5.1. Family Developmental Tasks

The traditional developmental perspective on families was described in the work of Duvall (1977) and Duvall and Miller (1985) (Kaakinen & Hanson 2010). Their original work was based on nuclear families, with two heterosexual parents and one or more children, where they identified overall tasks that families need to

achieve in each life phase. Families with adolescents, for instance, have to allow for the young persons to establish their own identities and still remain part of the family, while adolescents are expected to focus on their future, education and jobs (Kaakinen & Hanson 2010). By contrast, families with young adults are expected to launch their children and after the young person has moved out, they reallocate roles, space and power and adapt relationships and communication.

Contemporarily, family structure is increasingly diverse (Kaakinen et al. 2010, Kaakinen & Hanson 2010, Segrin & Flora 2011). The traditional pathway is no longer the norm and therefore not always applicable. Carter and McGoldrick (2005) developed the life cycle model further to include variations in families. They replaced the concept of nuclear family with “immediate family,” which takes into account different family structures including single parent families, stepfamilies and gay or lesbian families. Consequently, this research defines family very broadly as *“any group of people related either biologically, emotionally, or legally”* (McDaniel et al. 2005, p. 2).

Besides a more broad family definition, McGoldrick et al. (2013b) also developed a more flexible concept of predictable stages with appropriate emotional tasks for individuals and families. They described different stages of the family life cycle which start with the life stage of young adult family members. These stages are as follows: 1) Young adults leave home; 2) they build new couples and enter into marriage; 3) Families have young children; 4) Families have children that grow into adolescents; 5) Families are launching their children and move on in life and 6) families live in their later life.

Generations have to adjust to life cycle transitions simultaneously. As young adults differentiate form their families, they establish new romantic relationships and their independence. Parents, on the contrary, need to evolve their relationship with their children to make family boundaries more permeable, and they need to refocus on their life and on being a midlife couple (McGoldrick et al. 2013b). When expected development takes place, families find balance between being a family and being an individual, by balancing connectedness and separation, belonging and individuation and accommodation and autonomy (Carter & McGoldrick 2005, McGoldrick et al. 2013b).

3.5.2. Life Cycle Stressors

All families move through the life cycle and they have to adapt to new situations where they are likely to experience stress and consequently anxiety (Carter & McGoldrick 2005, McGoldrick et al. 2013b). Stress is often greatest at transition points when family members have to rebalance, redefine and realign relationships. Among the major influencing factors on what families experience and how families cope with change, besides place and time in history, are world views and attitudes, culture and societal norms as well as intergenerational influences (Carter & McGoldrick 2005). Stress can also be transferred from past generations, for example, if two dramatic experiences happen close to each other and emotional reactions are delayed.

Families are confronted with potential stressors on all levels of the human system (Figure 8). Stressors on the individual level, for instance, are the genetic constitution and emotional, cognitive, interpersonal, as well as physical

development (McGoldrick et al. 2013b). The family may experience stressors related to their family history, pattern of interaction, attitudes, expectations, taboos and loaded issues.

Figure 8: Stressors and system levels

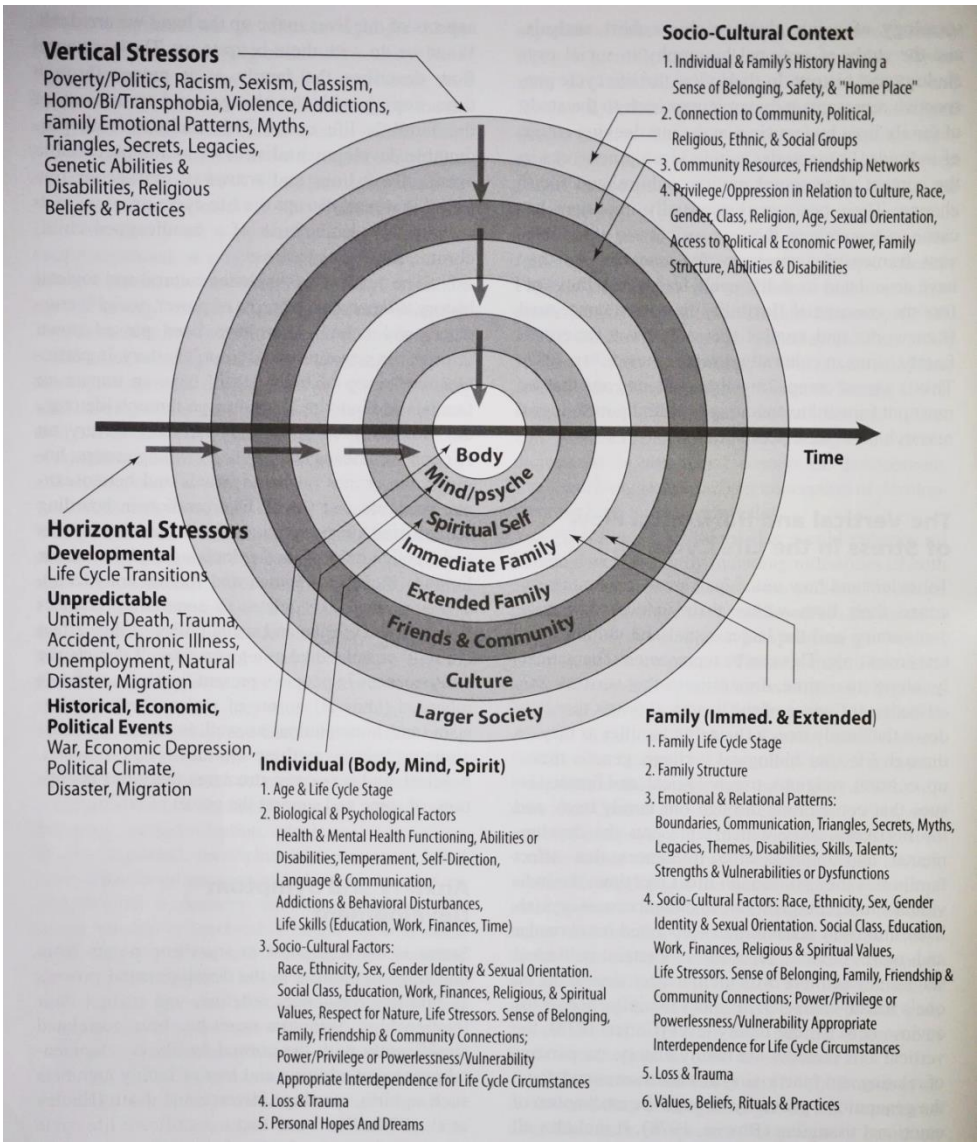


Figure from: McGoldrick M., Carter B. & Garcia-Preto N. (2013b) Overview: The life cycle in its changing context: Individual, family, and social perspectives. In *The Expanded Family Life Cycle: Individual, Family, and Social Perspectives* (McGoldrick M., Carter B. & Garcia-Preto N. eds.) Allyn & Bacon, Boston, p.8, illus.*

*(MCGOLDRICK, MONICA; CARTER, BETTY A.; GARCIA PRETO, NYDIA A.; CHAPTER, CONTRIBUTORS, EXPANDED FAMILY LIFE CYCLE, THE: INDIVIDUAL, FAMILY, AND SOCIAL PERSPECTIVES: INTERNATIONAL EDITION, 4th, ©2011. Reprinted by permission of Pearson Education, Inc., New York, New York.)

Stress occurs if they have to cope with changes or predictable and unpredictable events (McGoldrick et al. 2013b). There have been described

stressors also on a sociocultural level among which culture and socialisation, power and hierarchies, as well as beliefs and norms.

Families living with chronic illness may struggle to follow expected developments and expectations that are dictated from their own culture and society (Carter & McGoldrick 2005, Rolland 2012). A rigid application of norms and expectations as well as an overemphasis of the unique nature of chronically ill individuals may cause difficulties, such as an increased risk for mental illness. A more flexible application and a broader view of the human life cycle lessens stresses to families living with chronic illness, who have fewer models to guide them through transitions and life's stages (Carter & McGoldrick 2005).

3.6. The Family-Environment Interchange

The family system is naturally integrated into other systems that can provide support or put pressures on the family system. These systems include the extended family and friends, other resources for support such as networks and not least the Swiss health care and social care systems. A theory that adopts a family systems perspective and offers insights into the family and its environment is the bioecological theory by Urie Bronfenbrenner (1917-2005) (Kaakinen & Hanson 2010, Sandrock 2013). This theory describes the interactions and influences on the family from other systems at different levels of engagement; the micro-, meso-, exo-, macro- and chronosystems (Figure 9). The microsystem is the closest level of family everyday life including daily activities such as work, school and leisure, as well as direct interactions (Kaakinen & Hanson 2010).

Figure 9: Bioecological system theory

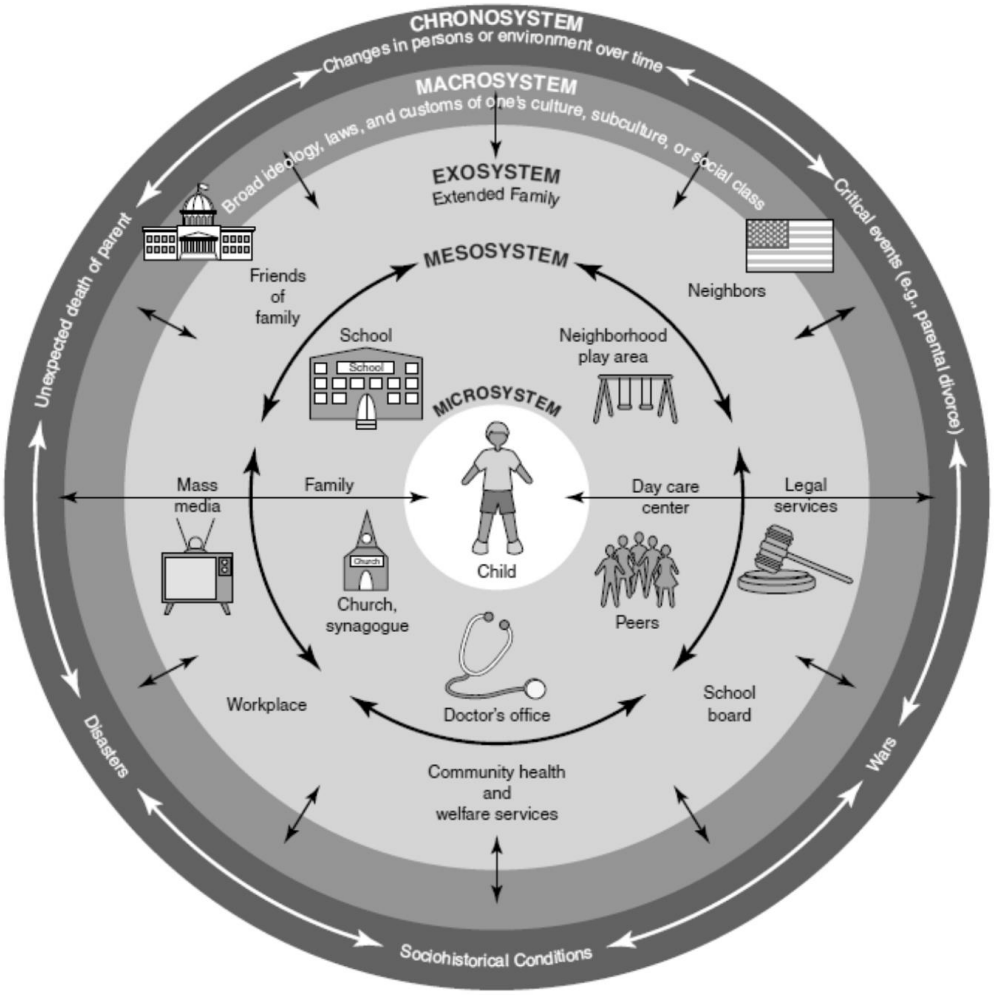


FIGURE 3-6 Bioecological systems theory model.

Figure from: Kaakinen J.R. & Hanson S.M.H. (2010) Theoretical foundations for the nursing of families. In *Family Health Care Nursing. Theory, Practice and Research* (Kaakinen J.R., Gedaly-Duff V., Coehlo D P. & Hanson S.M.H. eds.) F.A. Davies Company, Philadelphia, p.82, illus. (Republished with permission of F.A. Davis Company; permission conveyed through Copyright Clearance Center, Inc.)

The mesosystem accounts for effects on a first systems level such as the effects and relationships between the family and the school or the religious community (Kaakinen & Hanson 2010). The exosystem influences families indirectly, by not being actively involved, but nevertheless affected by factors such as governmental funding for schools or health care. Meso- and exosystems are part of a broader macrosystem, a level that comprises cultural

and societal attitudes and worldviews. The families' temporal context is referred to by their chronosystem, a level influenced by sociohistorical conditions occurring over time (e.g. environmental events and transitions) (Kaakinen & Hanson 2010). Therefore, to understand the exosystem of the families involved in this present study that are living in Switzerland, it is essential that the Swiss context is provided.

3.6.1. The Swiss Health Care and Social Systems

The Swiss health care system is historically shaped and can be dated back to the 19th century. With the creation of the Swiss Federal state in 1848 the Federal government took over responsibility for the monitoring of serious epidemics (Bundesamt für Gesundheit 2006). The first law on epidemics and the foundation of the Health Department followed in 1886, which were among the first actions of the Swiss Federal government. Five Federal laws emerged at the turn of the century including the management of epidemics, regulation of professional duties and education of physicians, vets and pharmacists. Furthermore, these laws regulated the movement of food, the work in factories and created the basis for the future law for health insurance (Bundesamt für Gesundheit 2006). In the 20th century, the Federal government invested more and more in health care as there were the secularisation of nursing and a change in the financial system, which in many regions had been based on charity until this point in time.

Switzerland is a democratic Federal state, which consists of 26 administrative divisions called "cantons" that can be further divided into 2249 municipalities (Schweizerische Bundeskanzlei 2016). While the Federal state is responsible

for matters like the national security, foreign relations and the national road network, cantons play a major role in implementing Federal laws according to their individual needs and they have semi-autonomy in sectors like culture, education and hospitals. Therefore, each canton has its own parliament, government, justice and constitution, which is in concordance with the Federal constitution (Schweizerische Bundeskanzlei 2016).

A major characteristic of the Swiss health care system which evolved historically and is still predominant today is the subsidiary role of the Federal government and the dominance of the cantons in regard to health care (Bundesamt für Gesundheit 2006). In other words, the general responsibility for the delivery of health care services is cantonal, leading to federalism and decentralisation. In addition, there is no single health care system or health policy as the Swiss landscape of health care systems comprises 26 cantonal health policies for each of the cantons. How competencies and responsibilities of the Federal government, the cantons and the municipalities are negotiated and divided is illustrated in Table 4.

Table 4: Selection of competencies and tasks

Competencies and tasks of the cantons	Competencies and tasks of the Federal government
• Implementation of Federal laws • Monitoring of professions and institutions • Health care provision (e.g. hospitals, nursing homes, community services, paramedics, …) • Education of professionals • Health protection • Prevention and health promotion • Financing of cantonal institutions, social welfare • Authorisation of contracts and rates	• Social security (e.g. health insurance, invalidity insurance) • Monitoring of pharmaceuticals and medical products • Monitoring of private insurances • Immunisation and transplantation • Management of alcohol and tobacco • Health protection (e.g. human research) • Work safety • Environmental and civic protection • Statistical information

Table adapted from: Bundesamt für Gesundheit (2006) *Gesundheitspolitiken in der Schweiz-Potential für eine nationale Gesundheitspolitik*. Volume. 1. Bundesamt für Bauten und Logistik, Bern, p.34-35 (Adapted version published with permission of Bundesamt für Gesundheit)

Besides federalism and decentralisation, the Swiss health care system has other characteristics that are intertwined with its historical development: the dominance of curative medicine, the liberalism that favours the private sector and the concept of solidarity of the social system (Bundesamt für Gesundheit 2006). Swiss social security is grounded in the principle of solidarity among all members of society. This means that despite physical and psychosocial diversity, as well as differences in gender, generations, income groups or other differences, members of Swiss society have an equal right for basic social services.

The social system of Switzerland is based on solidarity, but the many players of this system are fragmented, individuals are therefore confronted with great complexity. First of all, the social system comprises the compulsory general health insurance (OKP) which paid for 35% of health care costs in 2008, while the state and private households bore the rest (Bundesamt für Gesundheit et al. 2008, Widmer 2011). A general health insurance is required for every Swiss

resident and covers services for in- and outpatient settings for medication, maternity and birth, and for interventions in prevention and rehabilitation. In addition to the OKP, Swiss residents can apply for private supplementary insurances. These insurances for additional services, for instance, for complementary medicine, are granted by insurance companies and they vary in the services and premium policies included, whereas the general health insurance is the same for everybody (Bundesamt für Gesundheit et al. 2008, Widmer 2011). In total, there are approximately 80-90 different health insurance companies and the premiums must be paid monthly.

In addition to the health insurance, every Swiss resident has a Federal old age and survivors' insurance (AHV), an accident insurance and a disability insurance (IV), which are all secured by employment and deducted from wages (Bundesamt für Gesundheit et al. 2008, Kühne 2015). The AHV allows for financial support at old age and in combination with a pension fund, whereas the IV is directed to interventions to support working capacity and disability. Moreover, there are specific regulations and additional services such as supplementary benefits which add to the complexity of the Swiss social system.

This complexity can be illustrated in the example of neuromuscular conditions: The Swiss law defines birth defects among which there are classified genetic conditions such as NMDs (Schweizerische Akademie der Medizinischen Wissenschaften 2013). Where a birth defect is diagnosed financial support for medical treatment and for nursing care at home or in an institution is provided by the IV up to the age of 20 years (Kühne 2015). The compulsory health insurance pays for the remaining costs and there are additional support

services granted for individuals with NMD including supplements for intensive care services and allowance for helpless persons (Schweizerische Akademie der Medizinischen Wissenschaften 2013). When reaching adult age, the primary financial support source for individuals affected by NMD alters from the IV to the compulsory health insurance, while the IV remains responsible only for costs related to the person's earning capacity. Since 2012, in addition to the services described above, adults living with NMD can apply for financial support if they want to live independently and employ their own care assistants round the clock. This new service is referred to as "Assistenzbeitrag".

3.6.2. National and International Health Policy

In the last decades, different efforts on a national and international level have been described that were directed to improve the situation of families living with NMD. Among those efforts was the assurance of equal opportunities for disabled people through the UN-Convention on the rights of persons with disabilities and the Swiss Federal law on equal opportunities for the disabled. In June 2016, a first report about the international situation was published by the United Nations, which showed an overall extensive consideration of the rights for people with disability in Switzerland (Eidgenössisches Büro für die Gleichstellung von Menschen mit Behinderungen 2016, Eidgenössisches Departement des Inneren 2016).

On a European level, the European Council established a strategy for meeting the challenges of rare diseases including the formation of an expert group (Commission of the European Communities 2008, European Commission 2013). The Swiss Federal Council also launched a national strategy for orphan

diseases as part of their policy programme “Health 2020” (Bundesamt für Gesundheit 2014). Moreover, the European organisation for rare diseases (EURORDIS), a non-governmental group of patient organisations, was founded to represent more than 700 single patient organisations from 60 countries. The aim of this pan-European organisation is to empower and advocate for patient groups, to inform and raise awareness about rare diseases and to promote the quality of services and other improvements through research (European Organisation for Rare Diseases 2014).

Furthermore, in 2007, an international network for research on NMD has been established, which is called Translational Research in Europe for the Assessment and Treatment of Neuromuscular Disease (TREAT-NMD) (Bladen et al. 2013). As part of this network, national patient registers were implemented, including a Swiss register, to facilitate clinical research and recruitment of participants for clinical trials as well as for the assessment of effectiveness of treatment strategies internationally. Conducting clinical research, however, remains difficult (Bladen et al. 2013, Commission of the European Communities 2008, Wästfeld et al. 2006). Drug development for the treatment of NMD, for instance, is still at an early stage, because of the complexity of underlying disease mechanisms, unavailability of subjects for trials, and general poor market potential for these drugs with high development costs.

3.7. Research Need

Families are situated within a specific socio-historical context which they cannot be isolated from. They develop a patterned reaction to change and stress and

as NMDs have a severe impact on the affected young person's life, it is highly likely that not only affected individuals, but also their families experience major implications. Therefore, when we try to explore and gain a better understanding of their transition experience, a family perspective to research is crucial. This chapter introduced family systems theory, a theoretical perspective on families that is well suited for the study of the family transition experience when affected by NMD.

Previous research on the family transition experience when affected by NMD, however, focused mainly on the experiences of individual family members, resulting in limited insights into how the family system was affected. There is little knowledge about how family relationships and patterns of interaction influenced the family transition experience when living with a young family member with NMD who transitioned into adulthood. Therefore, there is a need to further explore the family transition experience. This knowledge is necessary to understand what the repercussion of NMD are, how families function, how they experience developmental tasks and which family characteristics, strategies and patterns of interaction are in favour of their wellbeing. On the basis of this evidence, health care professionals can better support families that struggle with their life situation in coping with NMD and developmental needs during the affected young person's transition into adulthood.

3.7.1. Study Aim and Research Question

This study aims to explore the experiences of families living with NMD during the transition of the affected young person into adulthood by applying a family systems approach. Moreover, it aims to identify major illness and development

related challenges and strategies by which families cope with their life situations in their specific context. By studying life experiences of these families their explanations and expectations of events can be identified and a better understanding of their challenges and coping strategies gained, which is valuable information that allows to inform and improve clinical practice. The following overarching research question was in the focus of the consequent study:

Where a young person is affected by NMD, what are theirs and their family members' experiences of the young persons' transition into adulthood?

Secondary questions included:

I. What challenges do young persons affected by NMD and their family members' face during the young persons' transition into adulthood?
II. How do young people affected by NMD and their family members cope with new life situations and what are facilitating factors for mastery and positive adaptation to change?
III. What are the young persons' and their family members' life experiences with the Swiss health care system during the young persons' transition into adulthood?
IV. Is it possible to identify a theory that describes the family transition experience when young persons affected by NMD transition into adulthood and if yes, what are the characteristics of this theory?

4. Chapter 4: Methodology

To answer the research question that was posed, the most appropriate research methodology has to be adopted. A research methodology connects philosophical assumptions about reality and knowledge with their more practical implications and research procedures. The careful choice and transparent description of a study's research methodology ensures the study findings are understood in their specific context and gives clarity about the research procedures undertaken. In this chapter, different research paradigms will be introduced and their suitability for the consequent empirical study discussed by contrasting them with each other and by delineating their philosophical assumptions in a socio-historical context. Then, the philosophical underpinning, social constructionism, and its methodological implications will be described and discussed. From this discussion constructivist Grounded Theory emerges as the chosen study design for this study.

4.1. Research Paradigms and Philosophical Assumptions

Research paradigms are characterised by their underlying philosophical assumptions of what the nature of reality is (ontology) and how knowledge is developed and generated (epistemology) using a specific type of inquiry (methodology) (Creswell 2013, Denzin & Lincoln 2011). Whether reality exists outside of the human mind and how knowledge is generated is subject of great debate and, depending on the philosophical stance taken, the suitability of a specific type of inquiry varies. It is therefore necessary to understand the differences between major research paradigms and their ontological and epistemological assumptions, which lay the ground for an informed choice and allow for a transparent and detailed description of the methodology. Such

information can provide the reader with insights into how the perspective chosen might affect the outcomes and interpretation of the findings.

4.1.1. Definition of Ontology and Epistemology

Ontology is the philosophical strand concerned with the nature of knowledge and reality which asks: *"What is the nature of reality? What kind of being is the human being?"* (Denzin & Lincoln 2011, p. 12). Today, a great divide persists between realist and relativist ontological assumptions, while there is also a blurring of the genres (Burr 1998, Burr 2003, Burr 2015, Lincoln & Guba 2000, Lincoln et al. 2011). Realists believe that there is a real world out there that exists independent of the observer, whereas relativists assume the existence of multiple realities that are co-constructed by people and are culturally and historically dependent (Burr 1998, Burr 2003, Burr 2015, Denzin & Lincoln 2011). Even if realist and relativist ontological strands seem to diverge, there is a growing variety of acceptance of some assumptions alongside each other as new philosophical standpoints emerge over time (Lincoln & Guba 2000, Lincoln et al. 2011).

Besides ontology, epistemological premises are crucial for their influence on the research paradigm chosen and the type of inquiry used. As Charmaz (2014, p. 6) depicts the relation between epistemology and research: *"Every way of knowing rests on a theory of how people develop knowledge".* Epistemology is linked with ontological belief and concerned with the nature of knowledge and truth and how knowledge is developed or generated (Creswell 2013, Denzin & Lincoln 2011, Reed 2011). To give an example, realists assume that an objectivist epistemology is what should be aimed for, which implies that reality

and meaning exist and can be comprehended apart from a human beings' awareness of their existence (Crotty 1998, Lincoln et al. 2011). Meaning is inherent to objects and not created or constructed by a human mind or through human interaction, which would be the case in an interpretivist-constructivist epistemological understanding.

When knowledge is created through research, a research project is linked with an explicit or implicit research paradigm that serves as philosophical underpinning and lens to look at data (Silverman 2011). Study planning and realisation, including data analysis and interpretation of findings are therefore highly influenced by the ontological and epistemological position taken, even if not made explicit. A research paradigm indicates not only analytical preferences and techniques for data collection, but it also predetermines the language used (Silverman 2011). Table 5 gives an overview of different research paradigms and their associated ontological and epistemological assumptions as well as forms of inquiry used.

Qualitative, quantitative as well as action based methodologies and major types of inquiry can be distinguished according to the role that the human mind and social interaction play. In quantitative research knowledge is generated deductively through theory-testing with experiments or surveys, where generated knowledge is considered to be independent of the subject (Creswell 2013, 2014, Denzin & Lincoln 2011, Silverman 2011). On the contrary, qualitative or inductive research is theory-generating by means of qualitative interviews or ethnographic observations. These combinations, however, are not exclusively linked. A particular research question may use quantitative or

qualitative data or a combination of both, for instance, as part of collaborative action research (Creswell 2013, 2014, Denzin & Lincoln 2011, Silverman 2011).

Table 5: Research paradigms and basic beliefs

Paradigm	Ontology	Epistemology	Methodology
Positivism	Naïve realism - assumes an apprehendable reality	Dualist – objectivist with true findings	Quantitative methods, experiment, verification of hypothesis
Postpositivism	Critical realism - imperfectly apprehendable reality assumed	Modified dualist – objectivist where findings are probably true	Quantitative inquiry, modified experimental, falsification of hypothesis, may include qualitative methods
Critical Theory et al.	Historical realism - virtual reality is shaped by different values (social, cultural,…) over time	Transactional – subjectivist where findings are value-mediated	Dialogic – dialectic inquiry
Constructivism	Relativism – local and co-constructed reality	Transactional – subjectivist, where findings are co-created	Hermeneutic – dialectic inquiry
Participatory	Participative reality - co-created by mind and given cosmos	Critical subjectivity in participatory transaction with cosmos	Political participation in collaborative action inquiry

Table adapted from: Lincoln Y.S., Lynham S.A. & Guba E.G. (2011) Paradigmatic controversies, contradictions, and emerging confluences, revisited. In *The SAGE Handbook of Qualitative Research* (Denzin N.K. & Lincoln Y.S. eds.) SAGE Publications, Thousand Oaks, pp. 100.*

4.1.2. Research Paradigms and Philosophical Assumptions in their Historical Context

In the following sections research paradigms and differences in ontologies, epistemologies and methodologies will be examined from a historical perspective as their chronological order is logically linked with their socio-historical development within which they can be best put in context and understood. For example, modern beliefs can only be understood when also considering precedent times, as modernism not only followed, but also emerged

and critiqued medieval beliefs (Burr 2015, Lincoln & Guba 2000). Thinkers of one era were influenced by their past and influenced their followers.

4.1.2.1. The Modern Era: Positivism and Postpositivism

Modernism represents the ontological assumptions of the time of Enlightenment from the mid-eighteenth century where the search for truth was guided by reason (Burr 2003). Modernists such as René Descartes' (1596-1650) and Emmanuel Kant (1724-1804) (Kenny 2010, Russell 2004) assumed the existence of a reality independent of the human mind and they superseded the dominant worldview of the mediaeval times, where the church was the only entity responsible for deciding about truth (Burr 2015, Lincoln & Guba 2000). The predominant ontology of the modern era was realism, and most realists, who believed in the existence of a real world that could be discovered, were also foundationalists. Foundationalism is the *"intellectual tradition that has sought to devise logically or scientifically irrefutable ground rules for the production of all valid knowledge"* (Weinberg 2008, p. 14). Not surprisingly there is a strong association between realist assumptions of modern times with their claim for objectivist knowledge generated through the scientific method, which clearly differentiates modernist traditions from the religious indoctrination of medieval times.

A research paradigm associated with the modern era that adopts "naïve realist" ontology is positivism, a research paradigm that assumes that there is an ultimate truth which is fully apprehensible and linked with an objectivist epistemology (Denzin & Lincoln 2011, Lincoln & Guba 2000). Today, naïve realist ontologies are becoming more and more extinct, as few people challenge

the idea that humans are limited in apprehending the real world or that social reality is a human construct anymore. Newer versions of realism developed such as a critical realist stance which is adopted, for instance, in some post-positivist research paradigms.

Roy Bhaskar's critical realism (1975) critiques both positivists and relativists. Critical realists assume that there is a world of events out there independent of the human mind, but that knowledge about this world is socially constructed (Denzin & Lincoln 2011, Elder-Vass 2012). Critical realists further assume an arrangement of reality in levels and that there are causal processes that drive the social and natural world. Events are caused by multiple interacting causal processes, such as the power of structures (Bhaskar 1975, Elder-Vass 2012). There is a strong association between positivist and post-positivist research paradigms and quantitative research methods. In quantitative research, knowledge is deductively generated and not actively constructed by humans, for instance, through statistical testing of a priori developed hypotheses (Weaver & Olson 2006).

4.1.2.2. The Breakdown of the Modern Era: Critical Theories

In the middle of the twentieth century an intellectual movement developed as a critique of modernism and foundationalism: postmodernism (Burr 2015, Lincoln & Guba 2000). Karl Popper (1992), Ludwig Wittgenstein (1889-1951) and Thomas Kuhn (1962) were among the philosophies that contributed to the breakdown of the modern era (Weinberg 2008). Another pioneer who inspired many of his postmodern successors was Georg Wilhelm Friedrich Hegel (1770-1831). Hegel's legacy comprised, roughly summarised, the introduction and

insistence of the human historical development and its dialectic - the mutual influence between the mind and the world (Russell 2004, Weinberg 2008). One of the most known of Hegel's scholars was Karl Marx (1818-1883) who adopted Hegel's understanding of history, but put emphasis on the superiority of materiality over consciousness. Marxian interpretations and other structuralist ideas were among the thoughts that influenced and were further developed within critical traditions (Burr 2003, Lincoln & Guba 2000). Structuralism is characterised by the search for underlying structures and rules that explain the world. For Marx the underlying structures to his theory were class relations.

Critical traditions, including critical theory as well as feminist and post-structural theories are associated with historical realism (Lincoln et al. 2011). These paradigms focus generally speaking on constraints placed on individuals or groups by specific factors such as gender, race, class or the like. They have a strong historical foundation where they challenge problematic life situations that result from disparate power distribution within societies and aim for the empowerment of individuals or groups of their concern (Creswell 2013). These paradigms are critical of socio-historical realities and values and in support of actual change. They are well suited for, but not exclusively linked with qualitative research methodologies (Flick 2014).

4.1.2.3. Postmodernism: Interpretivist and Constructionist Paradigms

Postmodernism evolved from, but rejected modern beliefs including the belief in human reason, the scientific method and the idea of an ultimate truth (Charmaz 2014, Weinberg 2008). Postmodernists deny the existence of a single reality, while proposing the co-existence of multiple realities and a historical and

cultural relativity (Burr 2003). Relativity implies that realities depend on different constructions and human experiences and are influenced by social interaction (Charmaz 2008, Denzin & Lincoln 2011). A relativist ontological stance can be interpreted more or less strictly (Lincoln et al. 2011, Schwandt 2000, Silverman 2011). There are, therefore, representatives of weak and strong relativism. The latter have been criticised for implying the principle of constant flux, which undermines the purpose of systematic research as all knowledge seems to be relative (Silverman 2011). To clarify, if there are multiple realities and endless alternatives, but no ultimate truth, there is no base to advocate for one view over the other (Burr 1998). In this case, it is argued that it would make no sense to engage in research activities at all as every perspective is equally valid and no one better than the other or more in need for further investigation.

The rise of the postmodern hermeneutic tradition followed after Hegel with the German philosopher Wilhelm Dilthey (1958). The ideas of Dilthey and the sociologist Max Weber (1949) challenged existing beliefs about knowledge by stating that the social world cannot be explained by natural laws, but only by interpretive understanding (Erickson 2011, Schwandt 2000, Weinberg 2008). They influenced younger generations of philosophers and sociologists such as early phenomenologists. By drawing on the works of Dilthey, for instance, the hermeneutic phenomenologist Martin Heidegger (1889-1976) developed his ontology called “being in the world” implying that humans are always connected with the world prior to our consciousness (Kenny 2010). According to Heidegger, proving the existence of an external world is absurd, as we are beings among other beings in the world.

The philosophies introduced above then paved the way for constructionist traditions. Constructionism argues that there is no meaning without a mind, and that meaning is nothing to be discovered, but it is socially constructed (Crotty 1998). Even though objects may be pregnant with potential meanings, there must be an active mind to attribute them. Constructionism seems to be well removed from an objectivist stance, but the distinction between constructionism and other postmodern epistemologies is crucial. Crotty (1998) distinguished between subjectivism and constructionism. In subjectivism meaning is created and imposed on the object by a subject, whereas in constructionism meaning is constructed and develops in interplay between subjects and objects in the world. To clarify, constructionism is defined as follows:

> *"It is the view that all knowledge, and therefore all meaningful reality as such, is contingent upon human practices, being constructed in and out of interaction between human beings and their world, and developed and transmitted within an essentially social context"* (Crotty 1998, p. 42)

Schwandt (2000) further distinguished between interpretivism, hermeneutics and social constructionism - three epistemologies that pertain to the same paradigm, but differ in the way they try to understand the human experience and as a consequence how they influence specific methodological decisions. A common characteristic of these three approaches is that they consider human action as meaningful (Schwandt 2000). In order to make this meaning explicit researchers try to interpret what they learn and to gain a better understanding of the participants' experience and situation.

What distinguishes interpretivism, hermeneutics and constructionism from each other is more subtle. Interpretivism and social constructionism are both hermeneutic in the sense that they study experiences within their contexts (Schwandt 2000). In other words, to understand parts, they try to make sense of the whole. When doing so, however, interpretivists argue for subjectivity without sacrificing objectivity, by doing research as an uninvolved observer and stepping outside of the own historical frame (Schwandt 2000). Social constructionism and hermeneutics, on the contrary, challenge this practice and a person's ability to step outside the self and the own tradition. They view traditions as a "power" that conditions a person's understanding and interpretation and always biases it to some extent. Therefore, it is not possible to free oneself from one's own standpoints, but it is recommended to engage with them by altering those traditions or prejudices that are disabling (Schwandt 2000). Hermeneutics has many commonalities with social constructionism, but they are also distinct from each other. The former considers meaning as negotiated, whereas constructionists assume that meaning is constructed against a backdrop of shared understandings and traditions within a culture or society (Schwandt 2000).

Social constructionists were the poststructuralists Michel Foucault (1977) and Jacques Derrida (1962), the psychologist Kenneth J. Gergen and the sociologists Peter L. Berger and Thomas Luckmann (Burr 2015, Crotty 1998). Poststructuralists were postmodern philosophers that were interested in language as a site of meaning (Burr 2015, Weinberg 2008). Among the most known poststructuralists were the above introduced philosophers Foucault with his discourse analysis and Derrida with his thoughts on deconstruction.

Foucault and Derrida emphasise the role of language for social interaction and stress how language is changing over time and as a site of meaning (Burr 2015, Kenny 2010). Foucault assumes that our traditions of speech are imbedded into larger institutions. There are specific claims of knowledge made by these institutions that are imposed on individuals and result in their classification (Gergen 2015). Foucault calls these institutions “disciplinary regimes”. An example of such a classification in the field of medicine is being healthy and unhealthy or normal and abnormal.

Gergen emphasised the importance of language for social life and stated that the only feature of psychological and social life is that it is in continuous change (Burr 2015, Gergen 2015). All knowledge is therefore historically and culturally specific and investigations need to go beyond the individual into the social, political and economic spheres surrounding it. For Schwandt (2000) Gergen’s interpretation of social constructionism is radical and readily leading to epistemological relativism, whereas at the same time it allows for critical reflection of human practices. Gergen offers a sound description of how knowledge is socially constructed by falling back on traditions of constructions. For example, he describes different constructions of the human body; for Plato the body was a tomb, for Descartes the body was a machine, for contemporary young people the body is used as a cultural sign of one’s identity and for a physician, a body may be something that has to be repaired (Gergen 2015). These constructions influence and explain the behaviour of one who holds them.

The sociologists Berger and Luckmann define their sociology of knowledge and delineate how human beings construct and sustain social phenomena through social practices (Berger & Luckmann 1991). In their well-known work “the social construction of reality” (1966), they describe the sociology of knowledge as purely epistemological, without having any ontological claim. Therefore, the purpose of their sociology of knowledge is to analyse social reality and the social construction of everyday life, but not to state what is true or not.

Berger and Luckmann view everyday life as a reality interpreted by people and meaningful to them as a coherent world (Berger & Luckmann 1991, Burr 2015). The central sphere of this reality is shaped around the “here and now” and characterised by routines and pattern and the use of language. As individuals live together in social groups, everyday life and as a consequence pattern and routines are shared among people (Berger & Luckmann 1991, Burr 2015). Language is viewed as a system of signs which is shared and establishes common sense knowledge. Language, however, has also a special characteristic: It is “objectified”, which means it can be detached from the here and now and used to reflect about the past or think about the future (Berger & Luckmann 1991, Burr 2015)

According to Berger and Luckmann (1991) social interactions during everyday encounters are characterised by typifications and subjectivity. Typifications are schemes that individuals apply when apprehending and dealing with others. They shape our actions by the use of rigid pattern. Subjectivity, in contrast, makes interactions and relations highly flexible as rigid patterns of typificatory schemes are modified by subjectivity during personal interaction, such as during

face-to face contacts (Berger & Luckmann 1991, Burr 2015). Other influencing factors on social interactions are the degree of intimacy and the personal interest towards the individuals involved. To conclude, social interactions are a complex and dynamic interplay between two or more people, where there are patterns as well as factors influencing and altering their realisation.

In this research it is assumed that humans cannot step outside their own cultural and historical context, as judgements about reality are always made from within a human mind (Burr 1998). We apprehend everything, even a reality that lies behind our constructions of reality, such as a “physical world”, from within our value system. For the consequent study it is important to adopt a research paradigm that is in accordance with constructionist and moderate relativist assumptions. Consideration was given to the use of critical realism. While a critical realist paradigm could also offer guidance on which constructions and views of reality to favour over others, it simultaneously undermines the central importance of the social construction of reality and diversity of social life (Burr 1998).

This study investigates social realities and adopts a moderate relativist stance to put constructions in the forefront, but it does not accept an “anything goes” philosophy of which relativism has been accused. To defend choices, this study makes a pragmatist argument, where the basis for judgement is to support those choices that work, create more possibilities for freedom and quality of life (Burr 1998). On the basis of social constructionism a critical and reflective stance will be adopted and decisions will be reflected within their context, including the current political and scientific knowledge base. It is assumed that

the philosophical debate will be ongoing and evolving anyway with time and in different places, which makes it of secondary importance.

4.2. The Chosen Philosophical Underpinning: Social Constructionism

A constructionist approach proves to be a suitable philosophical underpinning for the focus of this study. This study aims to explore life experiences of families where a young family member is living with NMD and transitions into adulthood. The overarching research question, “Where a young person is affected by NMD, what are theirs and their family members’ experiences of the young persons’ transition into adulthood?”, was posed as prior knowledge about the family transition experience is limited (Waldboth et al. 2016). Further investigations are needed to enable a better understanding of these families’ experiences, their meanings and associated behaviours. To illuminate human experience and family meaning when aiming for better understanding of new and multi-facetted life situations an inductive approach needs to be favoured over a more deductive method (Erickson 2011, Patton 2015).

Another reason for the aptness of a constructionist paradigm is that it matches with the theoretical underpinning of this investigation, a family systems perspective and a family developmental approach to the human life cycle (McGoldrick et al. 2013b, Rolland & Williams 2005). These family theories have not only emerged during the post-modern era, but are also influenced by a constructionist turn. Family meanings and understandings are viewed as socially co-constructed and not negotiated, imposed or discovered. A constructionist stance assumes that families co-construct different realities and

live in varying socio-historical contexts where their experiences are influenced by actions and interactions (Denzin & Lincoln 2011). When trying to gain a better understanding of the meaning of these families' experiences over time and when viewing family life as constructed and relative to the cultural and historical context, no other paradigm is better suited than constructionism.

Berger and Luckmann's (1991) sociology of knowledge and their analysis of the social construction of reality seem to be compatible with the nature of this research project. Their notions on the experience of time - where the centre is in the here and now and the connection with the future and the past happens through language - reflect some family systems and life cycle theoretical assumptions. Moreover, their description of a shared reality with the development of a shared language, practices and patterns offers a framework that can be linked with the study's aim of investigating the families' experience and pattern of interaction in the pursuit to understand what happens to the family system at the point of the young person's transition into adulthood.

Furthermore, constructionism is not only suited from a logical line of reasoning, but it is also widely accepted and compatible with my assumptions as a researcher. Jasper & Goodwin (2005) wrote in their opening statement: *"We are all social constructionist today, almost".* Philosophical assumptions do not only underpin a work and guide the research process by serving as a research paradigm. They in turn also reflect the researcher's beliefs and give an explanation for actions and interactions. In other words, researchers always bring a belief system and philosophical assumptions to their research, no matter if they are aware of it or not (Creswell 2013, Denzin & Lincoln 2011). These

philosophical assumptions are influenced by the researcher's discipline and the tradition he or she belongs to, they are historically inherited, but may be adopted without being reflected upon (Schwandt 2000). Despite their awareness, these beliefs guide the thinking and acting of researchers by influencing the questions asked, the theoretical framework chosen and the methods adopted (Creswell 2013, Denzin & Lincoln 2011). Therefore, awareness about philosophical assumptions, reflection upon how they influence actions and interactions and disclosure of these assumptions are crucial for the quality and credibility of qualitative research.

4.2.1. Characteristics of Social Constructionism

When fathoming the depths of constructionism to work out methodological implications a more sound discussion from a theoretical perspective is indicated. First of all, terminology and definitions need more clarification. In the literature, the terms constructionism, social constructionism and constructivism are often used interchangeably, but lack consistency. Charmaz (2014), for instance, refers to constructivism when describing her advancement of the Grounded Theory, while she acknowledges that her understanding of constructivism approximates with the current forms of social constructionism. A useful distinction for clarification and consistency is needed. Crotty (1998, p. 58) defines constructivism as *"the meaning-making activity of the individual mind"*, whereas constructionism refers to *"the collective generation of meaning"*. The social characteristic is captured by constructionism. The more precise expression social constructionism derived from works of the sociologists Karl Mannheim (1883-1947) and Berger and Luckmann, and the term simply means their version of constructionism. For reasons of clarity and consistency, from

this point onwards this study uses the term social constructionism to describe its constructionist philosophical underpinning.

Social constructionists challenge conventional views of the social world such as the views of common sense knowledge, power and knowledge relations (Burr 2015, Gergen 2015). They foster a change of perspective and ask, for example: Why do we categorise humans into seemingly natural groups of people distinct by gender (men and women) and not into short and tall people? How does this distinction into males and females shape our everyday life? (Burr 2015) This critical stance towards knowledge implies that knowledge is constructed and could easily be constructed differently. Therefore, this study adopts a critical and reflective stance to social constructions.

This assumption is supported by beliefs that ways of understanding social life are historically, locally and culturally relative and none of the ways is better or nearer to the truth than the other (Burr 2015). What is natural for children and what parents are expected to do changes significantly over time and differs according to the society lived in. Adolescence, for instance, is a developmental stage introduced quite recently into our understanding. The term adolescence derived from “adolescere” which is Latin and means to grow to maturity (Bill & Knight 2007). Different cultures have different markers for defining the beginning and end of life stages. In addition, over the last decades adolescence experienced a time lag. What a time lag is can be explained by giving an example: the age of marriage. The median age of marriage in the 1970’s was 21 for woman and 23 for men, whereas approximately fifteen years later it had already risen to 25 and 27, respectively (Arnett 2000).

Social constructionism assumes knowledge is constructed by social interaction and negotiation of meaning within a specific group of people (Burr 2015). Language, as the medium for interaction, plays a key role in this process of social construction of knowledge. Language is even considered a precondition of human thought, as it informs our mind and provides us with necessary categories and concepts (Burr 2015). With time, within a certain culture, an accepted way of knowledge develops which is then “true” for the whole group. Knowledge is not inherited by things themselves, but co-constructed by social interaction, but there develops a certain impression of stability of knowledge, which makes it more difficult to challenge the common sense. Knowledge and human action are assumed to go together (Burr 2015). This explains why the social life of the West is different from the social life of an indigenous population living in South America. In other words, depending on the social constructions, human behaviour and everyday actions and interactions vary. The analysis of social constructions of a group of people can therefore give insights into the motives for human behaviour.

4.2.2. Methodological Implications

A social constructionist methodology aims to emphasise the voice of participants and their socially and historically constructed perspectives of their situation (Creswell 2013). These principles are inherent to inductive research which is used in qualitative designs such as phenomenology and Grounded Theory. Over recent decades, qualitative approaches have developed and spread as they could differentiate and grasp differences among life worlds by moving from specific observations to broader abstractions and theories (Denzin

& Lincoln 2011). Qualitative research, in contrast to more deductive approaches, is better suited to capture diversity of the social reality. It is not only appropriate to capture diversity, but also to portray individualisation. By being sensitive to different ways of life and biographical patterns it locates individuals' experiences within their specific contexts (Flick 2014). Qualitative methodologies allow for the illumination of complex realities including their diversity and individuality.

Denzin and Lincoln (2011), leading scientists in the field of qualitative research, published the following definition from which some characteristics of qualitative methods can be deduced:

> *"Qualitative research is a situated activity that locates the observer in the world. Qualitative research consists of a set of interpretive, material practices that make the world visible. These practices transform the world. They turn the world into a series of representations, including fieldnotes, interviews, conversations, photographs, recordings, and memos to the self. At this level, qualitative research involves an interpretive, naturalistic approach to the world. This means that qualitative researchers study things in their natural settings, attempting to make sense of, or interpret phenomena in terms of the meanings people bring to them."* (Denzin & Lincoln 2011, p. 3)

This definition by Denzin and Lincoln (2011) illuminates the complex nature of qualitative research by being naturalistic and interpretive and comprising a set of methodological practices. Creswell (2013) agrees with this definition, but puts

more emphasis on the research process, from the philosophical assumption to the interpretive lens, and the research procedures. He delineates distinct qualitative study designs and defines a set of common elements where the researcher appears to be the key instrument throughout the qualitative research process (Creswell 2013). During data collection through interviews and observations, for instance, the researcher closely interacts with the participants. This interaction is emphasised as data collection takes place in the natural setting of the participant, where contextual information is sought and the attention is directed towards the person's experience and the meaning attributed to situations (Creswell 2013). Besides, qualitative research requires the researcher's interpretation and reflexivity. Researchers need to interpret data, apply complex reasoning through inductive and deductive logic when analysing and they reveal information about their own history and philosophical stance as a means of establishing credibility (Creswell 2013).

Another characteristic of qualitative research is its flexible and dynamic nature. The qualitative study design emerges throughout the research process and multiple methods can be used including data from interviews, observations or documents (Creswell 2013). The final account of a qualitative investigation is a complex and holistic report of multiple perspectives which takes into account many influencing factors on the participants' situation.

A factor to success for a social constructionist methodology and qualitative study design, which has often been criticised as lacking in its implementation and therefore inhibiting success, is contextual sensitivity (Silverman 2011). If a researcher is sensitive to the situation and setting it does not only foster his or

her understanding, but it also positively influences the quality and trustworthiness of the findings. Contextual sensitivity is therefore serving as a guiding principle to the methodological decisions of this study.

4.3. Study Design: A Constructivist Approach to Grounded Theory

The practical implications of a qualitative methodology and a social constructionist philosophy will be introduced by discussing available methods and study designs. This discussion is not exhaustive, as a preselection has been made according to the methods fit for this study's focus, their compatibility with previously presented philosophical assumptions and their general potential. The following relevant and commonly used study designs will be shortly introduced: Ethnography, Phenomenology and Grounded Theory.

4.3.1. Critique of Relevant Study Designs

4.3.1.1. Ethnography

Ethnography is the earliest distinct mode of inquiry within the qualitative research paradigm (Patton 2015). It has its origins in the 19th century with the exploration of exotic cultures usually located in foreign countries and rather remote settings. From being the main research approach of Anthropology, Ethnography has spread to other disciplines where its use today extended to investigations of areas of social life of contemporary societies (Flick 2014, Hammersley& Atkinson 2007, Patton 2015). Ethnographers focus on human action or institutional practices and how they are linked with their larger cultural context (Hammersley & Atkinson 2007). An ethnographer asks:

> *"What is the culture of this group of people? How does culture explain their perspectives and behaviours?"* (Patton 2015, p. 100)

In Ethnography, culture, which stands for ways of life, is crucial. It is through the study of cultural and social settings, by committing to first hand experiences and exploration of the natural setting of participants, that culture can be known (Mason 2011). Ethnographers assume that every group of people that interacts will evolve a culture comprising beliefs and behaviour patterns that are, as a consequence, determining human action (Patton 2015). As culture affects behaviour, it is also a key factor for change.

A main feature of ethnography is to study actions and accounts in their everyday context by observation and informal interviewing (Flick 2014, Hammersley & Atkinson 2007). For an extended period of time the researcher is participating in the everyday life of its participants and gathers all data available on the topic of interest. Ethnography does not want to develop a theory, but its product is a detailed and careful description of the area of concern which gives interpretations of meanings, functions and consequences of human action (Hammersley & Atkinson 2007).

4.3.1.2. Grounded Theory

Founders of Grounded Theory were the sociologists Barney Glaser and Anselm Strauss (Charmaz 2011, 2014, Glaser & Strauss 1999). In 1967 they released their book "discovery of Grounded Theory: strategies for qualitative research" where they firstly offered a systematic but flexible approach for collecting and analysing qualitative data. The release of this book took place at a time of

tension between quantitative and qualitative methods in social research where quantitative research and positivist philosophical assumptions dominated (Charmaz 2014). The launch of Grounded Theory, however, resulted in a growing interest and appreciation of qualitative methods. According to Bryant and Charmaz a factor for the success of the "Discovery of Grounded Theory" was its positivist idea, methodological consensus and provision of systematic strategies (Bryant & Charmaz 2007, Charmaz 2014). Over time, Grounded Theory has been further developed by others including Corbin & Strauss, Charmaz and Bryant and Clarke (Bryant & Charmaz 2007, Charmaz 2014, Corbin & Strauss 2015). Today its use has spread internationally and across disciplines, currently being considered the most influential qualitative research strategy in the social sciences (Bryant & Charmaz 2007, Charmaz 2014, Patton 2015).

Grounded Theory Method is a systematic, comparative method aiming for theory development grounded on data. Grounded Theory does not only refer to a research method, but also to a research product. Grounded theorists ask:

> *"What theory, grounded in fieldwork, emerges from systematic, comparative analysis as to explain what has been observed?"* (Patton 2015, p. 109)

Grounded theorists are interested in the study of pattern and relationships and the development of an explanatory theory of social processes (Charmaz 2014, Creswell 2013, Starks & Trinidad 2007). Researchers study these processes in the field where they engage in simultaneous data collection and analysis and

use constant comparison techniques and theoretical sampling strategies (Charmaz 2011, Charmaz 2014, Corbin & Strauss 2015, Glaser & Strauss 1999). The historical development of Grounded Theory will be described more fully later.

4.3.1.3. Phenomenology

Phenomenology is an approach to qualitative research rooted in the 20th century aiming at a deeper understanding of the lived experience (Creswell 2013, Patton 2015, Starks & Trinidad 2007). The method focuses on the underlying meaning, structure and essence of an experience or event and studies participants' experiences as follows:

> *"How they perceive it, describe it, feel about it, judge it, remember it, make sense of it, and talk about it with others".* (Patton 2015, p. 115)

According to phenomenologists the core meaning of an experience can be isolated. This core meaning is salient to the phenomenon, it makes a phenomenon what it is and without which it would not be what it is (Patton 2015). The phenomenon of interest may be an emotion such as fear or anger, a relationship such as marriage, or an experience related to a programme, organisation or culture (Creswell 2013, Patton 2015). Phenomenologists assume that the essence to shared experience is mutually understood by people who experienced the same phenomenon. The major methodological implication drawn from this assumption is to intensively interview those who have lived the experience and are reflecting about it retrospectively (Creswell

2013, Patton 2015). Within a phenomenological account the lived human experience is thoroughly captured and thickly described. Phenomenology has a strong philosophical component including works of Edmund Husserl, Martin Heidegger, Jean-Paul Sartre, Alfred Schutz and Maurice Merleau-Ponty and the research procedures employed depend on the specific type of methodology adopted (Creswell 2013, Denzin & Lincoln 2011).

4.3.2. Discussion of Study Design

The foci of the above presented qualitative research designs, while all addressing questions of meaning and understanding of the human experience, are distinct from each other (Creswell 2013, Denzin & Lincoln 2011, Patton 2015, Starks & Trinidad 2007). Ethnography, generally speaking, is studying social life and culture of a group of people while highly valuing the natural context of the experience (Hammersley & Atkinson 2007). Phenomenologists are focusing on the essence of shared experiences and Grounded Theory is targeted at social processes and theory development (Patton 2015). A study design well suited to capture the nature of the topic of interest, family experiences of living with a chronic condition during transition into adulthood within a family systems framework, is Grounded Theory. Social processes such as family interaction and communication and human development are central to the topic of interest. It is assumed that family members construct a shared understanding that evolves in time and is sensitive to their social and cultural context (Burr 2015). Social processes such as family interaction and communication can be investigated by the use of Grounded Theory methods (Charmaz 2014, Creswell 2013, Patton 2015).

Grounded Theory Method is a research approach that has been used to study family experiences before. Vandall-Walker and Clark (2011) studied family members' experiences with relatives' that were affected by critical illness. They developed the theory "working to get through" where they explain how family members work at gaining access to the bedside, to information and how they interact with the inpatient setting. Another study by Purcell et al. (2015) focused on parents' use of faith following a child's diagnosis with a rare disease. Their theoretical conception describes five ways of coping and making meaning of their families' situation. The investigation of relationship between adults with cerebral palsy and their siblings is an additional example of a recent Grounded theory study in family research (Dew et al. 2014). Sharing childhood experiences, contact in adulthood, diminishing parental role and increasing support needs are the major themes described by this group of researchers.

Choosing a Grounded Theory study design is reinforced, as the scope of this study is to develop a theoretical explanation of the families' transition experience when living with NMD. This theoretical work includes moving beyond description and to conceptualise the studied topic in order to better understand it in abstract terms. A Grounded Theory approach aims to generate a general interpretation or theory grounded in data (Charmaz 2014, Creswell 2013, Patton 2015), which adds to the existing body of knowledge. Ethnography and Phenomenology, in contrast, produce detailed descriptions of the human experience, but don't go beyond that (Denzin & Lincoln 2011, Patton 2015, Starks & Trinidad 2007). Ethnographic studies, however, can be theoretically further developed or combined with Grounded Theory Methods (Charmaz 2011, 2014). The study of ethical problems in nursing by Daniel Chambliss (1996) is

an example of a Grounded Theory Ethnography, where merging the two approaches enhanced the work (Timmermans & Tavory 2007).

Each of these approaches involve the use of purposive sampling techniques, but the number of participants, the frequency of the researcher-participant interactions and the unit of analysis depends on the purpose and research objectives of the project (Patton 2014, Starks & Trinidad 2007). Ethnographic studies have been used to study everything from small groups from a specific setting to nations. It is an open ended approach that starts with a "foreshadowed problem", a problem or topic of interest, in an area of social life and where refinement of the topic is expected with time (Hammersley & Atkinson 2007, Patton 2015). Typical sample sizes for phenomenological studies, on the other hand, range from one to ten participants, as a detailed account of a few people who lived through the experience can suffice to uncover the essence of a phenomenon (Starks & Trinidad 2007). Grounded Theory relies on theoretical sampling where sampling is used to elaborate and refine theoretical categories (Charmaz 2014, Glaser & Strauss 1999). It is a method that facilitates the study of complex phenomena by involving participants with differing experiences and therefore allowing the integration of multiple perspectives (Charmaz 2014, Starks & Trinidad 2007).

Another influencing factor for the choice of study design is language. This study is cross-language research. Cross-language research includes any qualitative research where translations are necessary during the research process (Squires 2008, Squires 2009, Temple 2002). In this study, data collection takes place in German speaking regions, but the working language of the project is

English. As translations have the potential to negatively influence the research process, they can also affect the credibility of a study, for instance, by inappropriate translation techniques or lack of transparency in methodological description. There is a risk that meaning gets lost, especially when there is no emphasis on the study context. Cross-language research is therefore not similarly compatible with all qualitative approaches (Squires 2008, Squires 2009, Twinn 1997). In phenomenological research, for instance, the exactness of language is considered to be essential to capture the essence of the lived experience (Patton 2015). Therefore, the use of phenomenological approaches is not recommended when conducting cross-language research.

In addition, this study assumes that language is culturally and historically influenced and that different cultures or societies have different words for expressing their experiences (Burr 2015). As both, ethnography and phenomenology, aim to develop detailed descriptions there is the risk of bias, when words do not translate easily or when they do not translate at all. A Grounded Theory study design, in contrast, seems to be more suitable for cross-language research as it operates on a higher level of abstraction and takes into account the setting and other contextual information (Charmaz 2014, Patton 2015). Nevertheless, also in Grounded Theory studies, translations can have a potential negative impact on the quality of the research and this has to be borne in mind by the researcher as part of the reflexivity process in analysing the data.

4.3.3. Grounded Theory and the Epistemological Shift

In light of the above, Grounded Theory is a well suited study design for this research, but it needs some further introduction and methodological discussion. As introduced above, original Grounded Theory by Glaser and Strauss arose at a time where quantitative thinking and the scientific method dominated and their version was ground-breaking for qualitative research, but must be located within a positivist tradition (Charmaz 2014, Denzin & Lincoln). The following years, Glaser and Strauss developed in distinct methodological directions and finally separated. While Glaser kept with his objectivist version, Strauss started to collaborate with Juliet Corbin and they achieved a good reputation of their advancement of technical procedures of Grounded Theory (Charmaz 2014, Corbin & Strauss 2015). In the 1990s, Grounded Theory Method moved fully away from the positivist paradigm during the so called “constructivist turn”. Scholars known for their work in this field are Kathy Charmaz, Kevin Bryant and Adele Clark (Bryant & Charmaz 2007, Charmaz 2014, Clarke & Friese 2007).

Philosophically, objectivist Grounded Theory views the world as an external reality that can be discovered and rejects the researcher’s impact on the research process, while constructivist Grounded Theory applies a social constructionist epistemology where multiple realities co-exist and are co-constructed between researcher and participants (Charmaz 2014, Creswell 2013). Between the original objectivist Grounded Theory by Glaser and Strauss, and the constructivist approaches of Charmaz, Clarke and Bryant as well as in the later works of Corbin and Strauss a clear epistemological shift is identifiable.

This shift in thinking manifests, for instance, in their approach as to whether to perform a literature review or use a theoretical framework in order to frame and inform a study. In original Grounded Theory and also in later works of Corbin and Strauss conducting a literature review prior to data collection and analysis, as well as using a theoretical framework is discouraged (Corbin & Strauss 2015). It is advocated that the purpose of Grounded Theory is to generate a new theory and not to be constrained by already existing evidence as is the case when applying existing concepts from empirical or theoretical work.

The consequent research, by contrast, is meant to design a new theory that is grounded in data by use of constructivist Grounded Theory methods, where it is acknowledged that the researcher and participant co-construct interview data. It is not possible to eliminate influences from prior knowledge completely, but more important is a reflection about the researcher's own background and assumptions that might bias analysis and interpretation (Charmaz 2014, Creswell 2013). At the outset of this research, a systematic literature review was conducted, which was used to identify a gap in the evidence base, an important step when aiming to avoid unnecessary replication of research. The evidence retrieved from this literature review informed further methodological decisions, such as the development of interview questions. According to Corbin and Strauss (2015) and Charmaz (2014), this use is appropriate providing consequent theory development is not influenced by existing concepts and theoretical insights generated by the literature review.

Furthermore, the theoretical framework of this study, family systems theory, provided direction during development of the research question and the

planning of this study. During data analysis, the research applied inductive strategies to theory development by remaining open to new concepts and insights emerging from the data. The theoretical framework will be used to extend upon the emerging theory and to interpret the data once data analysis has been concluded, a use that is in accordance with principles of Grounded Theory (Corbin & Strauss 2015). Whilst Grounded Theory is meant to design new theory, it is important to place that into a context of what is already known or has a shared perspective.

Despite the differences in theory of knowledge and indifferent to the question whether to use a theoretical framework or perform a literature review prior to empirical work, constructivist Grounded Theorists argue that basic grounded theory strategies that have been developed within the objectivist tradition, are transferrable among different epistemological paradigms (Charmaz 2014). Among these strategies are constant comparison techniques, coding of data, writing of notes in form of memos and theoretically informed sampling strategies. The aim is to develop an interpretive theory using abductive reasoning. Abduction starts with the examination of the data, but also involves searching for all possible theoretical explanations, until the researcher arrives at the most plausible interpretation of the information (Charmaz 2014).

Symbolic interactionist assumptions influenced constructivist Grounded Theory (Charmaz 2014). Interactionist philosophical perspectives on family meaning and interaction are in a sense constructionist, as it is assumed that family meaning and reality are socially constructed among family members (Charmaz 2008) Grounded Theory is a study design that benefits from this interactionist

and social constructionist lens, as it encourages innovation, new understandings and novel interpretations of social reality. This is particularly important for this study's focus. To clarify, this research investigates families' experiences of the transition into adulthood when a young family member is affected by NMD. It is assumed, that these families live within a specific socio-historical context and that they develop their own family meaning through family interaction. At the same time, however, they are confronted with inherited social constructions and social norms and standards, which might diverge from their experience of living with a chronic illness.

From this discussion of methodologies and study designs I conclude that a constructivist Grounded Theory study is the most suitable for this research project. Therefore, this study follows the strategies of inquiry described in "Constructing Grounded Theory" by Charmaz (2014) and uses complementarily Corbin and Strauss's work "Basics of Qualitative Research" (2015) to inform basic Grounded Theory strategies for data collection and analysis.

4.4. Chapter Summary and Outlook

A Grounded Theory study adopting a social constructionist underpinning has been considered well suited to this study's focus, as it allows grasping the plurality of family experiences by integrating various perspectives and acknowledging the complexity of social reality. A constructivist Grounded Theory is a qualitative study design, which offers systematic procedures for data collection and analysis, while at the same time being flexible and dynamic in nature and allowing for innovation through an epistemological stance that challenges taken for granted knowledge. This methodological approach

matches the researcher's worldview and fits with the theoretical underpinning of this study, so that an emerging theory can be contextualised with family systems and family developmental theories.

5. Chapter 5: Methods

The following chapter will describe methodological processes that are based on principles of constructivist Grounded Theory and that are aligned with social constructionist and family systems assumptions. The qualities which influenced further practical implications and methods are as follows:

1) A focus on family processes including action and interaction as well as pattern of behaviour and language, 2) sensitivity to the situational context of the family experience when co-constructing data, for instance, through interviewing or observing; 3) a naturalist and interactive practice of co-creation of knowledge between researcher and participant; 4) an interpretive way of looking at the data while trying to make sense of family members' experiences and interpreting their meanings and behaviours; 5) a critical reflective stance towards participants' accounts and my own assumptions as a researcher, with special attention to potential biases; 6) the implementation of Grounded Theory strategies including constant comparison techniques, coding, abductive reasoning, memo writing and theoretical sampling and 7) a data collection and analysis process that results in a complex report of an interpretive theory grounded in data illuminating family members' experiences from different perspectives and within their contexts. How these principles were applied within this specific study will be described in the following sections.

5.1. The Study Design

The chosen study design for this investigation is a constructivist approach to Grounded Theory (Charmaz 2014). The constructivist Grounded Theory method is characterised by recruitment and sampling of participants, data collection and simultaneous start of data analysis and theory building (Figure 10) (Charmaz

2014). Throughout these research processes, notes in form of memos are collected which foster innovative thinking and theoretical developments. In addition, constant comparison techniques and theoretical sampling are applied while collecting additional data and analysing it, both activities addressing emergence and development of theoretical insights in form of saturated categories and an interpretive theory. In light of the above, Grounded Theory is both a method and a result. This Grounded Theory method consists of a systematic guideline for collecting and analysing data, whereas a Grounded Theory is an interpretive theory that aims to provide an explanation and understanding of meaning and human action (Charmaz 2000, Charmaz 2014).

Figure 10: A visual representation of a grounded theory

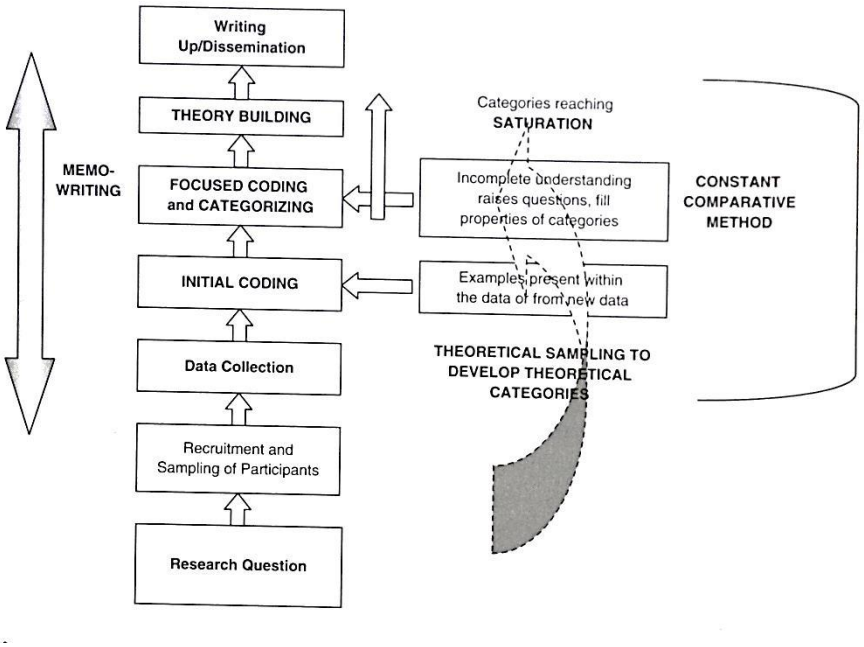


Figure from: Charmaz C. (2014) *Constructing Grounded Theory*. 2nd ed. SAGE Publications, London, p.18, illus. (Republished with permission of SAGE-Publications)*

*Original figure by Alison Tweed. An earlier version of this figure originally appeared in Tweed A., & Charmaz K. (2011). Grounded Theory methods for mental health practitioners. In *Qualitative Research Methods in Mental Health and Psychotherapy: A Guide for Students and Practitioners (*Harper D. & Thompson A.R. eds.) John Wiley and Sons, Oxford, p. 133, illus. (Republished with permission of John Wiley and Sons; permission conveyed through Copyright Clearance Center, Inc.)

5.2. Participants and Setting

This study uses a family systems framework to investigate life experiences of families where a young family member is living with NMD and transitions into adulthood. The unit of analysis is the family, which was earlier defined very broadly as a group held together by blood, emotional or legal ties (McDaniel et al. 2005). The aim was to include the view of more than one family member through interviews with the young person affected and their family members with whom they had a physical, emotional or socio-psychological tie. Potential participants were therefore asked to identify other possible family members who might be willing to participate. However, a family could participate, even if in the end just one family member (the affected young person, parent or sibling) was willing to take part.

Family members with different family compositions and from multiple sites were considered potential participants, as variety is useful for theory development in Grounded Theory studies (Creswell 2013). Nuclear families were able to participate as well as stepfamilies or families with other family compositions. The affected young person, their parents and younger and older siblings were participants of first choice. All next of kin who were considered part of the family by these family members were also considered for participation. The spouse or life partner of the affected young person, other members of the extended family and close friends could take part if they wished. In addition to referring to themselves as members of the family, family members were expected to have close contact with each other and to share or have shared experiences of daily living with the affected young person during transition into adulthood. This circumstance was considered important as it ensures that participants are able

to give rich information about the topic of interest providing rich data with thick descriptions, which is indicated for Grounded Theory studies (Charmaz 2014, Charmaz 2000).

5.2.1. Sampling and Criteria for Eligibility

Sampling strategies and predefined inclusion and exclusion criteria for individuals and families served as guiding principles for inclusion of participants. Initially, sampling was done purposefully. Purposeful sampling is indicated for qualitative research, as it is a means to inform understanding of the research problem while aiming for gathering rich data (Charmaz 2014, Creswell 2013). To further develop theoretical categories at a more advanced stage of analysis and theory building, purposive sampling was replaced by theoretical sampling, which will be described in section 5.4.3.2.

Inclusion criteria for the young person affected by neuromuscular disease were age range between 14 and 30 years, resident in Switzerland, first symptom onset of NMD in childhood (from 0 to 10 years) and moderate to severe physical impairment according to the Duchenne MD physical impairment and dependency score (DID) (Kohler et al. 2005).

There are different definitions of adolescence and norms of age ranges available in the literature. This study, however, considers childhood and adolescence as social constructions and overlapping phases that are not fully definable and depend on the individual development and the cultural context of the young person. The Swiss Law for Human Research (HFG) defines adolescents as children from the age of 14 (Bundesversammlung der

Schweizerischen Eidgenossenschaft 2014). Therefore, in this study the lower age limit for inclusion of young people living with NMD was set according to the HFG and in accordance with the recommendations of the local Ethics Committee, respectively, at the age of 14.

The upper age limit, in contrast, was set at 25 years at the beginning of the data collection, but had to be revised upwards during the data collection phase because of requests for participation by young adults between 25 and 30 years of age. Amending the upper age limit to 30 years was necessary to allow these potential participants to take part. The young adults said they were able to reflect back on their experiences during their transition into adulthood and believed they had something important to contribute now, with the benefit of hindsight.

Parents were included, if the young person was within the above defined age range and affected by NMD (disease onset in childhood and moderate to severe physical impairment). Younger siblings (with a lower age limit of 8 years) and older siblings of a young person were included under the same conditions, as well as other next of kin. The lower age limit for siblings at 8 years was recommended by the local Ethics Committee. In addition to the criteria outlined above, participants needed to have their residence in Switzerland and were excluded if they were non German-speaking or cognitively impaired and not able to give informed consent.

Theoretical saturation served as guiding principle for the total number of families included in this study. Theoretical saturation is the moment when there

is no new information resulting from data collection that adds to additional understanding or property development of the categories (Charmaz 2014, Glaser & Strauss 1999). Charmaz (2014) renounces predefined sample sizes, but gives recommendations when to increase the number of participants such as when interviews are the only data source, when constructing complex conceptual analyses or when attempting to raise credibility of the study. Giving estimates on sample sizes in Grounded Theory studies are discussed controversially, but often these estimates are required by review boards or funding bodies. Compared to other qualitative designs, Creswell (2013) claims that Grounded Theory studies are likely to include a larger number of participants in order to achieve detail in theory including between 20-60 participants. Despite these recommendations, in this study theoretical saturation was the measure of choice for when to stop sampling, because it is a measure to ensure the quality of the study's result. Additionally, it is considered as unfair to put families through an event that is not necessary, therefore, data collection processes stopped, when theoretical saturation of the emerging categories was reached.

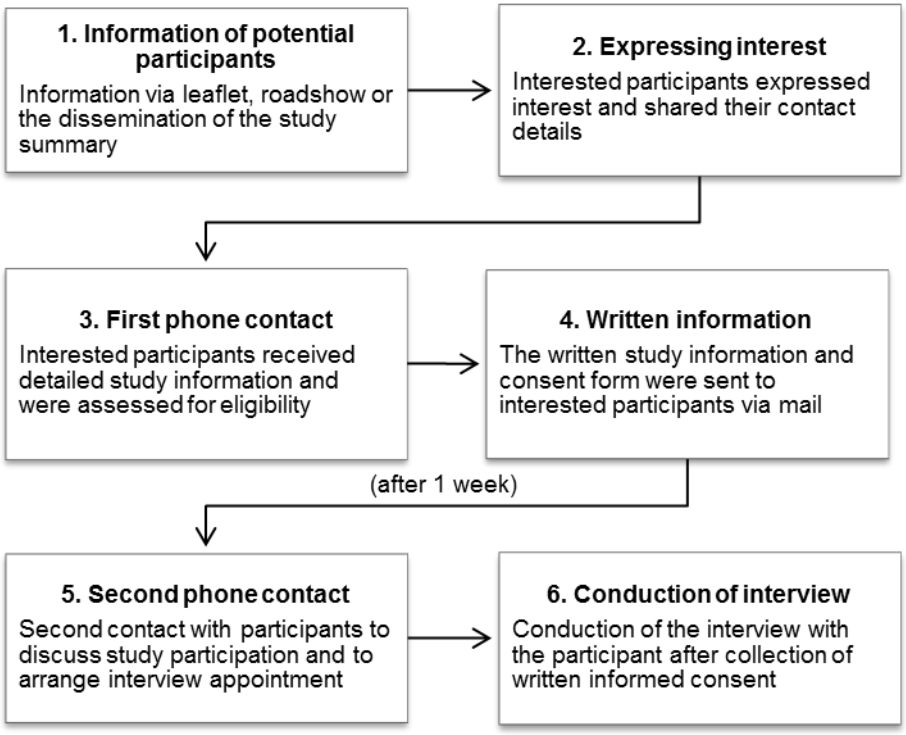
5.2.2. Recruitment Strategy

Different approaches were taken for recruiting participants in line with the Grounded Theory methodology. Different recruitment sites were targeted as an apparently small number of potential participants and a variety of contexts and potential life situations were expected. For example, younger affected individuals were expected to be living at their family's home, whereas older individuals may live independently or in a long term care institution.

Family members were recruited in the canton Zurich, a gathering point for young people with rare muscular disease (Klein et al. 2014, Klein et al. 2016), but families included in data collection were also living in other German speaking regions of Switzerland. The first step of recruitment included the identification of organisations that agreed to collaborate and give access to potential participants. Collaborating organisations in recruitment were care facilities specialised in the care of patients with physical disability, children's and an adult tertiary hospitals, an association for muscular diseases and self-help groups for individuals with NMD and their families. All of these organisations were either formally or informally linked with each other and had contacts with potential participants in varying life situations. The children's tertiary hospital, for instance, has a shared consultation office with the adult tertiary hospital where they collaborate during the transfer of the young person from paediatric to adult services. Depending on the age of the young person they are either cared for in the children's or adult's setting.

There were two processes of recruitment of families. Either the young person affected by NMD was recruited first and provided access to other family members or a family member (for example a parent or a sibling) was recruited first and provided access to the young person with NMD or other family members. The recruitment strategy comprised six steps (Figure 11). In the first step participants were informed about the study by collaborating organisations using study leaflets, road shows or the dissemination of a study summary (Appendix 2.1).

Figure 11: Recruitment of participants



Participants who expressed their interest in study participation and shared their contact details were then contacted by phone. The purpose of this phone call was to give detailed information about the study and what was involved, to answer any questions the potential participant had, to assess if inclusion and exclusion criteria were met and to identify other family members and potential study participants. After this initial contact, when participants were considered eligible and met all criteria written study information, a consent form and information leaflet was sent to the participant's home via mail. If the individual interested in participation was younger than 14 years, he or she was asked to inform his or her parents or legal guardian about their interest in study participation and to ask for their written consent. In this case, study information for the parent or legal guardian was added to the mail. Enough time was given to the participants to decide whether to participate or not. The minimum time frame was one week after provision of the written participant information via

mailing. After the expiration of this term, the researcher contacted the study participant for a second time via phone, to confirm study participation and to arrange an interview appointment. Those willing to participate were asked to sign an informed consent form. A more detailed description of the ethical considerations inherent to these processes is presented in the next section.

5.3. Ethical Considerations

This study was approved by the cantonal Ethics Committee (CEC) of the canton Zurich (KEK-ZH-Nr.2014-0225) (Appendix 2.2). The study protocol, the proposed patient information and consent form as well as other study-specific documents were designed according to the Swiss legal requirements for Human Research (HFG) and have been submitted to and approved by CEC (Appendix 2.3 and 2.4). In addition, the study was carried out in accordance to the Convention on Human Rights and Biomedicine (Council of Europe 1997), the Nuremberg Code (Schweizerische Akademie der Medizinischen Wissenschaften 2015), the Declaration of Helsinki (World Medical Association 2013), the guidelines of Good Clinical Practice (GCP) issued by the International Conference on Harmonisation of Technical Requirements for Registration of Pharmaceuticals for Human Use (ICH) as well as the Swiss Law and Swiss regulatory authority's requirements.

5.3.1. Study Information, Informed Consent and Informed Assent

Participant autonomy was among the guiding ethical principles of this research (Schweizerische Akademie der Medizinischen Wissenschaften 2015). Ensuring participants' autonomy comprises their complete information, voluntary participation and the special protection of participants who are particularly

vulnerable. Children and adolescents as well as individuals living with physical disability who are dependent on care are particularly vulnerable (Schweizerische Akademie der Medizinischen Wissenschaften 2015). This research with children and adolescents living with NMD is therefore only indicated, because the desired insights of this study can only be gained with their participation. When vulnerable groups are not included in research due to ethical concerns, there are fewer insights into their experiences and a consequent lack in the evidence base may increase their vulnerability in the longer term (Schweizerische Akademie der Medizinischen Wissenschaften 2015).

In this study, participants were informed about the study, its purpose, the procedures involved and its benefits and potential risks. Moreover, they were informed that the participation in the study would be voluntary and that they could withdraw from the study at any time and that withdrawal of consent would not affect their subsequent medical or nursing care. All participants received a participant information sheet and a consent form describing the study and providing sufficient information for the participants to make an informed decision about their participation in the study. As the written consent of a participant was obtained before the participant took part in the interview, participants had time to read the document and consider their statement before signing. In addition, they received a copy of the signed document that was also signed and dated by me and will be retained as part of the study records. Subsequently, a detailed description of participant information and consent form for all participant groups is given which was developed in accordance with the Swiss legal requirements (HFG).

Adults (18 years and older) and young people between 14 and 17 years of age received oral and written study information and gave oral and written consent to participation. If the person affected by NMD was not able to give written consent, they gave oral consent on tape. The verbal informed consent was recorded on a separate audio file, because the interview file was deleted after transcription.

Children between 11 and 13 years of age received age appropriate oral and written information and gave oral consent to participation, whereas children between 8 and 10 years of age received oral information and gave oral consent. Written informed consent of the parent or legal guardian was collected in each case as required by the Swiss law (HFG). Children have the right to receive information and participate in decision making processes, even if they need the informed consent of their parent or legal guardian for study participation (Schweizerische Akademie der Medizinischen Wissenschaften 2015). Despite their particular vulnerability, they are able to take responsibility for their bodies and give an informed assent. The information for children was developed after consultation of available templates from CEC Zurich and relevant literature and pretested by children of the relevant age groups. The developmental maturity of the child and the appropriateness of the study information was assessed and adapted during encounters with the children.

5.3.2. Participants' Privacy and Confidentiality

There was no relationship of dependence between myself and the participant families and no information was returned to a treating or referring physician or

nurse. Participant data obtained as a result of this qualitative study were considered confidential and were not disclosed to third parties. Interview transcripts and demographic information were encoded to assure participants' privacy. After transcription interview audio files were deleted. The password protected file containing information about solution for data encryption was saved on a secure computer at ZHAW and password protected. My supervisors had no access to this information and they received encoded data only. In order to assure traceability after the study ends, data will be stored in a secure archive at ZHAW for ten years after study conclusion. Anonymity of the participants will be guaranteed when presenting the data at scientific meetings or publishing in scientific journals. All participants will be assigned pseudonyms or codes.

5.3.3. Risks, Benefits and Dealing with Ethically Challenging Situations

Besides autonomy, two additional ethical principles guided this research: beneficence and justice (Schweizerische Akademie der Medizinischen Wissenschaften 2015). The consideration of risks and benefits for study participants stands for the principle of beneficence, while the principle of justice aims for their equal distribution among different individuals and groups (Schweizerische Akademie der Medizinischen Wissenschaften 2015). Study participation did not represent an increased risk for participants. They took part in an interview that lasted approximately one hour where they talked about their experiences and gave demographic information. The CEC Zurich classified this study as low risk (risk group A). Moreover, participants did not benefit directly from study participation and they received oral and written information about this fact prior to participation. However, they contributed to planned future

improvements in the care of other families affected by NMD and were given an opportunity to share their experiences.

Despite the low risk, during and after the interview different emotions were triggered. In order to minimise negative effects I planned enough time at the end of each interview to discuss with the participants where they might seek further support if they were distressed. In case of emerging distress or questions that I was not competent to meet prior defined indicators for action were available and contact details of counselling services were at hand and could be offered to the participants. Defined indicators for action were when the participant asked for referral; when the participant asked for information or assistance that was not within the scope of the research project or when the participant showed signs of emotional distress. Emotional distress can be expressed by crying, expressions of anxiety and strong feelings of anger and guilt. However, crying is also a normal emotional reaction, so prior to taking action the researcher had to check with the participant to find out if it was because of distress or part of the expressed emotion and lasting for a short period of time. If participants reported bad practice or a crime the plan to act was to assess the situation and the potential risk for the people involved. In case of increased risk I first informed the participant about my concerns and my need to take action. Then I discussed the situation and possible actions with my supervisors and took action in order to protect participants' confidence and to introduce safety measures for the persons at risk.

5.3.4. Involving Patients and Professionals

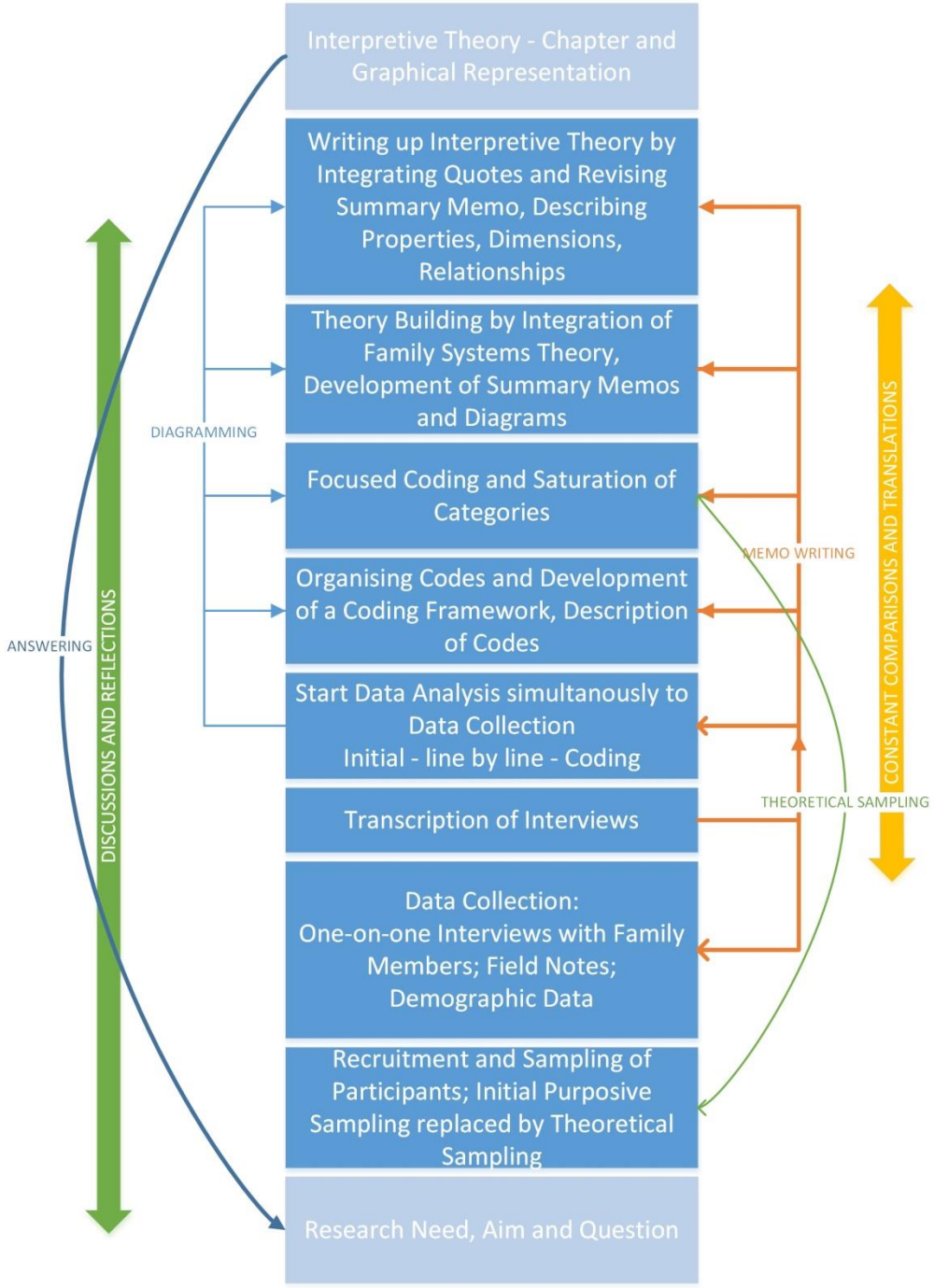
The consultation or involvement of patients, professionals or the public in research planning and realisation is not required by the Swiss law for human research (HFG), but it is considered that it may facilitate improved health outcomes and research quality and is therefore recommended (Schweizerische Akademie der Medizinischen Wissenschaften 2016). When consulting patients and professionals, for instance, we receive valuable insights into the relevance of a topic (Schweizerische Akademie der Medizinischen Wissenschaften 2015). Moreover, the involvement of patients fosters patient participation in decision making and it empowers them to express their needs and ideas which may benefit them by improving services (Schweizerische Akademie der Medizinischen Wissenschaften 2016). This study did not involve patients in the planning stage, but through the chosen methodology family members were engaged in the data collection and analysis processes. To clarify, data were gathered through interviews with family members. In addition, their feedback on first interpretations of findings was sought. The methodological usefulness of this type of respondent validation is under debate, but patient involvement is a proper ethical goal and patients' comments can provide important new insights (Silverman 2011). I was invited to give a presentation at an event for patients, where I presented some of my preliminary findings and received important feedback that supported me in advancing my theory development.

5.4. Data Collection and Analysis

As introduced before, in Grounded Theory studies, data collection and analysis start simultaneously by use of different techniques (Charmaz 2014, Corbin & Strauss 2015, Glaser & Strauss 1999). While in the following sections data

collection strategies and principles of data analysis will be described in more detail, figure 12 outlines how different techniques influenced and informed each other.

Figure 12: Research process (data collection and analysis)



5.4.1. Gathering Data using Interviews

Grounded Theory Methods are open to multiple sources of data (Glaser & Strauss 1999), but interviewing and observing are among the most frequently used techniques that allow for the generation of first-hand data (Corbin & Strauss 2015, Charmaz 2014). This research aimed to explore an area that has not yet been thoroughly researched where collecting first-hand information was indicated and interviewing is considered a suitable and effective method to do so. Data were collected from one-on-one interviews with family members and field notes of these encounters from January 2015 until January 2016.

Every participant was asked to take part in a one-on-one interview, including children and adolescents. If possible, interviews were conducted face to face, but the researcher was open to telephone interviews, if preferred by the participant. One-on-one interviews are better suited for the generation of thick descriptions than focus groups, as family members might be less hesitant to speak and freer to dominate the conversation (Creswell 2013). In family focus groups, by contrast, individual experiences and narratives can be confounded and disturbed by group dynamics and power structures, related to the families' often hierarchical structure because of their multi-generational nature (Flick 2014). In addition, when planning to collect data using family focus groups, some families might feel reluctant to take part, as family relationships are difficult or individual experiences would not be embraced by the whole group. Despite the fact that a family focus group would reveal information on family interactions, it was thought more might emerge from individuals being able to speak more candidly. Discussions about personal feelings and sensitive topics, for example, were expected to be facilitated during one-on-one interviews

compared to focus groups (Flick 2014). Sexuality or family conflicts are taboo issues that young people were expected to discuss more openly with a neutral external person, than with a close family member. Nevertheless, to allow for getting a grasp on family interactions, contextual factors and family members' behaviour were observed and noted in field notes. The interviews took place at the participants' homes or if they wished were arranged in a neutral and quiet place of their preference. Qualitative research adopts a naturalistic approach to data collection and the participant's natural setting is suited for doing interviews as well as for collecting contextual information (Denzin & Lincoln 2011).

Intensive interviewing techniques were applied. Charmaz (2014) describes these techniques as a gently-guided, one-sided interaction that explores research participants' perspectives where the participant talks and the interviewer encourages, listens and learns. The role of the researcher is to be open and not let nervousness or embarrassment inhibit the flow of information (Corbin & Strauss 2015). In this study, participants were asked broad and open ended questions. These question types are suited to encourage conversation and avoid forcing data into preconceived ideas (Charmaz 2014). An example of an initial question was: Could you describe an ordinary day of your daily life? These open ended questions were then followed by questions that invited a more detailed discussion of the topics introduced. Overall, main themes that were covered during the interview were family life, social life and friendships, living and work situation, health situation and growing up and coping with the effects of NMD either as the affected person, or as a family member.

A semi-structured interview guide (Appendix 2.5) comprising these questions and appropriate to the participants' age was developed. The topic guides for young persons and adults differed only in the form of address. Children under the age of 12, however, had the opportunity to engage in warm up activities and to do drawings and tell stories about their drawing as part of the interview. Beginning an interview with an activity that calms and reassures the child is advised (Zwiers & Morrissette 1999). Despite the offer, some children close to the age of 12 preferred not to do drawings, but to be interviewed in ways that were more similar to how young people were interviewed. The researcher adjusted the interview style to fit with the child's preferences, age and cognitive development and abilities and tried to cover the contents but at the same time to be flexible enough to be responsive to the child's individual needs (Zwiers & Morrissette 1999).

Warm up activities for children consisted of a card game (Appendix 2.6) where the child and the interviewer alternated in choosing from a deck of question cards, then playing the role of the interviewer by asking each other the questions on the cards (Teachman & Gibson 2013). This warm up activity aimed to increase the comfort of the child and to diminish the power differentials by giving explicit permission for the child to question the interviewer. After this warm up activity the children were asked to do a drawing of their family and to talk about the drawing. Drawings are considered a method of facilitating children's reports of their experiences (Gross & Hayne 1998, Wesson & Salmon 2001). The focus of this interview technique was on what the child said rather than on the interpretation of the child's drawing. During the interview the researcher asked open ended questions to guide the child's report.

Before the first interview, interview guides were piloted by individuals in the relevant age groups. Initial questions were clear to these individuals and no major changes were necessary after the pilot. The interview guides were slightly modified as data collection proceeded. This approach is consistent with Grounded Theory methods and allows refinement of questions in order to gather information necessary for theory development (Charmaz 2014, Corbin & Strauss 2015). Topic guides are viewed as flexible tools that can be revised or fine-tuned during the process of data collection. Revisions of the topic guide of this study comprised a switch in the order of interview questions. Initially, the last thematic block focused on participant's expectations and life goals and was expected to lead to a pleasant end of the interview. Participants, however, felt rather distressed, as these questions related to their future which they were rather concerned about. In order to end the interview with a less distressing topic, I decided to introduce the subject earlier and follow the participants' flow of speech to gain a conversational level before ending (Charmaz 2014).

All interviews were recorded and then transcribed word by word. A detailed discussion of issues of cross-language research and the management of differences in language related to data processing are described in the following subsection. The transcripts were encoded to assure participants' privacy. Data cleansing was assured by having all interviews proofread by a second person different from the transcribing person. Then, all transcripts were stored, converted into compatible files and added into an Atlas.ti file. Data analysis and management was supported by the data analysis software program ATLAS.ti. 7.2.

In addition to the interviews, after each interview, demographic data were gathered from all participants including gender, age, diagnosis of the affected person, relationships, marital status, living situation, employment, school leaving qualifications and overall health status by means of a demographic questionnaire (Appendix 2.7). Demographic data were used to provide a succinct overview of all the participants in the study and considered to give valuable contextual information. Demographic data were encoded and stored in SPSS files.

Shortly after the interview, the researcher made field notes about her observations, feelings and difficulties that she experienced before, during and after the interview. These field notes were included in the data analysis, and were further developed into reflective notes and informed analytical memos. Reflective field notes and memos were used to think about potential biases and the roles of the researcher being not only a researcher, but also being a health professional by background and having personal experiences by being a family member of an adult living with NMD.

Interviewing has been criticised as being inaccurate if not viewed as a construction or reconstruction between interviewer and participant (Atkinson & Silverman 1997, Charmaz 2014, Fontana & Frey 2000, Silverman 2011). This study takes a constructivist stance and assumes that interviews are not neutral as they always occur under specific circumstances and must be viewed as retrospective narratives and performances that participants give, where what people reveal cannot be directly linked to events they experienced. Family

members, for example, talk about their experiences in a way that fits it by highlighting some features while downplaying others (Silverman 2011). Nevertheless, interviews are a means to provide access to subjectivity and they give fleeting insights into participants' experiences (Atkinson & Silverman 1997). In order to ensure accuracy, it is viewed as lying within the researchers' responsibility to be aware of the nature of interviews and to be reflexive about background assumptions, disciplinary perspectives and other factors that influence how the interviews are shaped. The participant-researcher interactions as well as the individuals' background, their professional status, gender, race and age, have an impact on the narrative told (Charmaz 2014).

In order to foster shared understanding during the interview, researchers are advised to introduce themselves, to gain trust and to establish rapport with the person opposite (Fontana & Frey 2000). Before each interview, I did present myself as a researcher with a background in nursing and as a family member of an adult living with NMD. My insider experience turned out to be beneficial for gaining the families' trust, but it also came along with potential downsides, such as my worries of unwittingly forcing assumptions on data. Throughout data collection and analysis it was crucial to test my own assumptions, learn about participant's worlds, maintain curiosity to what participants shared, and interpret carefully what people said and did not say (Charmaz 2014).

5.4.2. Managing Cross-language Research

Data collection took place in German-speaking Swiss cantons. The spoken language in these Swiss cantons is "Swiss German" (Schwiizerdütsch) and the written language is "Standard German" (Hochdeutsch). In some Swiss regions

there are a high percentages of families who have migrated from other countries, with more than a fourth of the population in the canton of Zurich (statistics from 2015) (Migrationsamt 2016). I am an Italian migrant myself. I grew up in South Tyrol in the north of Italy where I graduated from the "Claudiana Landesfachhochschule für Gesundsheitsberufe" in Bozen and received my Bachelor of Science in Nursing from the collaborating University of Verona. The Bachelor Programme in Bozen is bilingual, Italian and German, and I do have good command of both languages. My first language, however, is a German dialect similar to the Austrian German, called "South Tyrolean" (Südtirolerisch) which exists only as a spoken language. The major written language in South Tyrol is Standard German, the same as in Switzerland. My spoken mother language is South Tyrolean and my written mother language is Standard German. To clarify, South Tyrol is a trilingual region with a large German speaking minority and an Italian and Dolomitic Ladin language group. In 2009 I moved to Switzerland for professional reasons. I have good command of spoken Swiss German as I have now worked and lived in the canton Zurich for over eight years as a nurse and research associate. After completing my Master of Science in Nursing at ZHAW, I chose to pursue my professional career by attending a PhD programme at Florence Nightingale Faculty of Nursing and Midwifery at King's College in London. The language used in postgraduate research training, supervision meetings and thesis writing is British English. I have good command of spoken and written English and have continuously employed strategies to improve the quality of my writing and speaking.

In light of the above, I am confronted with a diversity of languages during everyday life. My research project, specifically, is considered cross-language research, as data are translated during the research process. Each language involved in this process is viewed as an individual sign-system that is culturally and historically shaped (Burr 2015). Languages are changing and different languages influence each other, while translations are viewed as a means of transmitting information across languages. Conserving meaning while translating, however, is more complex. Meaning is influenced by the language used within the specific context of the interaction (Burr 2003). Even if individuals speak the same language and develop a shared understanding, it is difficult to know if they share the exact same understanding of the words they use and the constructions they make. In addition, by not knowing contextual factors or if consistent translations are lacking, there is a risk for incorrect translations and loss of meaning (Squires 2008). It might be impossible to translate exactly, which needs to be viewed as a methodological limitation. Not addressing these linguistic challenges threatens the quality of cross-language research (Squires 2008).

The following paragraphs will delineate a detailed methodological description of the translation processes that have been carried out in this study. Interviews were conducted in the participant's original spoken language, Swiss German. This method is appropriate to obtain understanding of participant's experiences and to minimise loss of meaning (Smith et al. 2008, Twinn 1997). Participants who have migrated from other countries that had another native language than German were required to have good command of oral Swiss or Standard German in order to qualify for participation.

Following recommendations (Chen & Boore 2009; van Nes et al. 2010), the verbatim transcription of the records and the data analysis of the transcripts was performed in the original written language of the informants and the researcher; Standard German. This approach is advantageous as time and costs can be saved. Moreover, potential limitations of the analysis can be prevented when the researcher analyses in her mother language (van Nes et al. 2010).

In order to allow for discussions with my English speaking supervisors, codes, definitions of codes and relevant quotations were translated into English and reviewed by a bilingual Swiss academic mentor from ZHAW. The researcher and the Swiss academic mentor discussed potential translations and used fluid descriptions of meanings rather than single words in their translations (van Nes et al. 2010). This was considered to be important to provide technically and conceptually accurate translations.

In qualitative research quotations are used to confirm results and to increase the trustworthiness of the study (van Nes et al. 2010). In order to avoid misinterpretations while quoting, the thesis therefore comprises quotes in the original and the target language, German and English. The translation of the quotes was reviewed by a bilingual proof-reader to ensure their accuracy. In publications where space is more limited quotes will be written in the journal's language only.

5.4.3. The Process of Data Analysis

5.4.3.1. Coding and Constant Comparison

As previously introduced, data analysis began shortly after the first data was collected. Grounded Theory data analysis is a very dynamic process where the researcher organises data, asks questions and makes comparisons while moving between the abstract and the concrete and trying to identify patterns and relationships (Corbin & Strauss 2015). Grounded Theorists apply principles of coding, constant comparison, memo writing and theoretical sampling, but they are also advised to develop their own set of strategies that fit the purpose of the specific study (Charmaz 2014, Corbin & Strauss 2015, Glaser & Strauss 1999). Examples of the process of analysis (e.g. coding with Atlas.ti., diagramming, memo writing) of this study are appended (Appendix 2.8).

According to Charmaz (2014) coding is the first step within the Grounded Theory process of analysis that links data collection with theory development. Coding is described as the process of breaking up data into components and organising them by indexing or denoting concepts (Corbin & Strauss 2015, Mason 2011). Charmaz describes the scope of coding as follows:

> *"Codes sort, synthesize and, most significantly analyze data. Codes connect raw data with the Grounded Theorist's conceptualization of them"* (Charmaz 2014, p. 341)

In this study coding was performed with the aid of the qualitative analysis software Atlas.ti.7.2 as well as on paper. Both types of coding had their advantages. The coding using the software allowed for the management of

much larger sets of data more effectively, whereas the coding on paper better promoted the generation of new ideas and insights on quotes. Throughout the analysis both types of coding have been used in an alternating manner, while the coding using the Software predominated.

Although there are other types of coding available, in this study only initial and focused coding were performed. Initial coding makes the researcher familiar with the data and reveals first conceptual ideas (Charmaz 2014, Corbin & Strauss 2015). Throughout initial coding the researcher reads the data, labels words, lines or segments of data using codes by remaining open to all possible theoretical directions. The researcher is advised to perform initial coding spontaneously, moving quickly through the data and using simple codes preferably in the form of gerunds or in the language of action (Charmaz 2014).

I started with coding a couple of interviews using the software and applying line by line coding which facilitated keeping open-minded and close to the data. After completion of a first round of coding, codes were then organised, defined and further developed into a coding framework. Then, the coding framework was used to code some more interviews on paper, which facilitated comparisons between interviews of family members of the same family. At every level of analysis, data, codes and concepts were compared to each other (constant comparison) to find similarities and differences and to facilitate analytical thinking (Charmaz 2014, Corbin & Strauss 2015). Classic questions that guided the analysis were: What is happening here? What are the basic social and psychological processes (Charmaz 2014)?

In a second stage, initial codes were further developed into an advanced coding framework and first theoretical thoughts were tested by use of diagramming techniques and memo writing. The advanced coding framework was then used to analyse larger amounts of data in a more targeted manner, a process referred to as focused coding (Charmaz 2014). This dynamic process of coding raw data, comparing it constantly with each other and theorising about it was repeated and alternated multiple times and required from the researcher being open, letting go of preconceived ideas and keeping close to the data by looking at it again and again (Charmaz 2014, Corbin & Strauss 2015).

According to Strauss and Corbin there is an additional coding type, axial coding, which specifies the relations, properties and dimensions of a category with the aim to *"sort, synthesize and organise large amounts of data and reassemble them in new ways after open coding"* (Charmaz 2014, p. 147) This study did not use axial coding by labelling text passages, but while further developing the categories specific questions were asked to make links and connections across family members and between the emerging categories visible. These questions included: What is the condition influencing the family's situation? What is the family's context? What are the strategies of coping used? How did families react? What are the consequences?

5.4.3.2. Memo Writing, Diagramming and Theoretical Sampling

Memo writing, diagramming and theoretical sampling are strategies for analysis that are interrelated and linked with data collection, analysis and writing about the results, while advancing the theoretical development of the analysis (Appendix 2.8). Memo writing, to begin with, is an intermediate step between

coding and writing when researchers "*stop and analyze their ideas about their codes and emerging categories"* (Charmaz 2014, p. 343). Corbin and Strauss (2015) compare memo writing with brainstorming and letting loose with thoughts. By thinking and writing about ideas the analysis moved forward and initial theoretical writings emerged. Different methods of memo writing were applied including writing electronic memos directly into Word- and Atlas.ti-files, but also on paper (analysis journal), to allow for spontaneity. Memos were first managed chronologically and then thematically and revisited as well as summarised as the analysis moved further.

In addition, diagramming was used combined with memo writing, to explore and depict coding frameworks, relationships between codes and emerging categories (Corbin & Strauss 2015). Diagrams are conceptual visualisations of data that are helpful to support moving beyond mere descriptions into a more advanced theoretical sphere. In this study diagrams were used to organise data and to make sense of them, as well as to advance theory building and allow for theoretical discussions with peers and supervisors. For each family, for instance, I developed a diagram as an intermediate step to allow for comparison of experiences among family members and subsequently across families.

Beyond storing and advancing analysis, memos and diagrams informed data collection by guiding theoretical sampling. Theoretical sampling is a method of data collection that replaces initial purposive sampling once analysis moves further, and it differs by being more strategic, specific and systematic (Corbin & Strauss 2015, Charmaz 2014). Theoretical sampling aims at theoretical saturation by collecting data from places, people and events for the purpose of

maximising theoretical insights and to further develop emerging theoretical categories. As this study progressed, for instance, I included and interviewed families where a young family member had died during the transition stage, as it gave further insights into a developing category, whereas interviewing health professionals which was initially on my mind, was no longer relevant for theory construction as the study proceeded.

5.4.3.3. Building and Writing up the Grounded Theory

All strategies involved in data collection and analysis including coding, constant comparison, memo writing and theoretical sampling finally led into the theory building stage (Charmaz 2014). This constructivist Grounded Theory aimed for the development of an interpretive theory assuming that the findings consist of an interpretation of the participants as well as of the researcher. Different researchers can arrive at different conclusions when studying the same data, as well as the same researcher can interpret data differently, after some time has passed (Corbin & Strauss 2015). Interpretive theories can therefore not be isolated from their context of development (Charmaz 2014).

In this study, initial strategies involved in theory building were writing a descriptive summary memo that integrates all previous memos, using integrative diagrams to sort out relationships between categories and discussions throughout the research and in particular for theoretical developments with peers and supervisors (Corbin & Strauss 2015). Advanced strategies directed at finalising the Grounded Theory comprised putting renewed emphasis on processes while revising the summary memo, developing a line of reasoning and checking for logic and variation as well as testing for

grounding in data. The major categories, their properties and dimensions as well as relevant quotes in support of the interpretive theory were identified and described.

5.4.4. Reflexivity and Criteria for Evaluation

A researcher's background, perspectives and assumptions have an impact on the research he or she carries out and these influences cannot be completely eliminated (Charmaz 2014, Corbin & Strauss 2015). Though the full extent of potential biases can never be known, it is important to be aware and reflexive about perspectives, biases and assumptions. Throughout this research, I kept a reflective journal where I reflected about my professional and personal background, my values and beliefs and about my experiences during the different stages of this research; the planning of the study, the data collection and analysis and the theory development. Every time when I experienced something distressing or irritating I described the situation in my journal and reflected about my reactions and emotions.

Another strategy that fostered reflexivity included regular discussion with my supervisors and peers, who pointed out some of my hidden assumptions and potential biases and inspired my thinking throughout this whole journey. An example for a hidden assumption that my supervisors pointed out during a discussion was my preconception that "being independent" was a high value for families that had to be more important than others such as "protection". Once aware of this assumption, that being independent was important for me personally, my horizon broadened and I was overall more aware of what I

brought to the table as well as more eager to challenge taken for granted knowledge.

Major quality criteria used to reflect the strengths and limitations of this study were credibility, originality, resonance and usefulness (Charmaz 2014). Corbin acknowledged that these criteria developed by Charmaz are the most comprehensive and therefore well suited to evaluate constructivist Grounded Theory studies as they comprise both the scientific and creative features of qualitative research (Corbin and Strauss 2015).

The criteria will be shortly introduced: The credibility of results is concerned with having sufficient evidence to justify the claims and to allow the reader to gain understanding. Charmaz (2014, p. 337) asks inter alia: *"Do the categories cover a wide range of empirical observations"?* Originality is evaluated by describing the social and theoretical significance of the study and if it gives new insights by challenging, extending and refining evidence. Resonance means the fullness of the portrayed experiences and can be evaluated by asking: *"Does your Grounded Theory make sense to the participants or people who share the circumstances?"* (Charmaz 2014, p. 338) To conclude, usefulness is rated by describing the work's contribution to the advancement of the knowledge base and improvements of everyday worlds.

After completion of analysis and writing up the findings the criteria outlined above were used to reflect about the quality of the study and the developed interpretive theory. The results of this evaluation are presented in the discussion chapter.

5.5. Chapter Summary and Outlook

This methods chapter provided a detailed and transparent description of the methods applied with the focus of how data were collected and analysed. In this study family members provided insights into the family experience of transitioning into adulthood when living with NMD by participating in one-on-one interviews. Data were collected in Switzerland and the recruitment strategy comprised collaborations with various clinical sites and the use of different approaches to identify potential participants. Intensive interviews with adults, young people and children were semi-structured and gave insights on the topic of interest from a family perspective. Recorded interview data were then transcribed and analysed using principles of Grounded Theory comprising coding, constant comparison techniques and memo writing. By critically reflecting about the research process and result and by evaluating it with the outlined quality criteria, the quality of this study was reinforced. In the now following findings chapter the resulting Grounded Theory will be presented.

6. Chapter 6: Findings

The previous chapter discussed issues related to the research methodology and procedures involved in data collection and analysis. The following sections report my findings, which are based on family members' experiences of the affected young person's transition into adulthood when living with NMD. First some contextual information is introduced by describing the study participants and setting and then the description of the categories and interpretive theory follows.

6.1. Description of Participants and Setting

In total 31 family members from 12 different families participated in this study. Ten affected young people and 21 family members took part in one-to-one interviews. Interviews were conducted from January 2015 until January 2016. The average length of the one-to-one interviews was 68 minutes. In three cases interviews were conducted with one family member only, the affected young person, while in nine other families more than one family member participated. An anonymised family genogram of each family can be found in Appendix 3. Ages of participants have been reported categorically to ensure privacy. All participants were living in a German speaking region of Switzerland at the time of data collection. Family members were mainly, but not exclusively living in the same canton as the affected young person.

Table 6 gives an overview of the demographic information of the affected young people who participated in this study. Six out of ten of them were males. The mean age of affected young people was 21.7 years, ranging from 15 to 28 years. The majority of them were single, while two affected young people were

in a relationship with their partner. Four affected young people each were living with their families or in a care institution. Half of the young people living with NMD were employed part-time, while the rest were still going to school / university or reported to be self-employed. All affected young people were asked to rate their physical and mental / emotional health, which they referred to as ranging from good to very good and excellent.

Table 6: Demographic data of affected young people

Demographics of affected young persons		**N**	**M (±SD) Min-Max**
Sex	*male*	6	
	female	4	
Age	*mean (years)*		21.7 (±4.6)
	range (years)		15-28
Condition	*DMD*	4	
	SMA	3	
	other	3	
Marital status	*single*	8	
	in partnership	2	
Living situation	*with family*	4	
	with friends	1	
	care institution	4	
	with partner	1	
Number of people in the household	*mean*		2.5 (±1.8)
	range		1-6
Employment / school situation	*pupil*	3	
	student	1	
	employed	5	
	self-employed	1	
Employment rate of those employed	*part time (<80%)*	5	
School leaving qualification	*primary school*	3	
	apprenticeship	6	
	higher education	1	
Physical health*	*good*	6	
	very good	2	
	excellent	2	
Mental / emotional health*	*very good*	7	
	excellent	3	

* Participants rated their physical and mental / emotional health on a five point Likert scale from poor, fair, good, very good to excellent. (Appendix 2.7)

The 21 family members were composed of 13 parents, seven siblings and one partner (Table 7). The majority of the family members who participated were female (n=14). The average age of the parents was 49.9 (±6.4) years, while that of the siblings was 17.6 (±7.1) years, respectively. Almost all parents had a partner and they were married or registered / living in a partnership. The majority of those family members that were employed (n=12) worked part-time and parents' highest educational qualifications were apprenticeship and higher education. Family members rated their physical and mental / emotional health from poor to excellent with more than half of the family members reporting being in a very good physical and mental / emotional health situation. However, two family members each rated their health as less than good in both categories.

Table 7: Demographic data of family members

Demographics of family members		**n**	**M (±SD) Min-Max**
Sex	*male*	7	
	female	14	
Relationship with the affected person	*parents*	13	
	mother	9	
	father	4	
	siblings	7	
	sister	5	
	brother	2	
	partner	1	
Age	*parents range (years)*		49.9(±6.4) 37-56
	siblings range (years)		17.6(±7.1) 8-27
Condition of the related young person	*DMD*	11	
	SMA	7	
	other	3	
Marital status of all	*single*	6	
	in partnership	4	
	married / registered partnership	10	
	divorced	1	

Marital status of parents	*single*	0	
	in partnership	2	
	married / registered partnership	10	
	divorced	1	
Living situation	*with family*	17	
	alone	1	
	with partner	3	
Number of people in the household	*mean*		3.9(±1.7)
	range		1-6
Employment / school situation	*pupil*	*4*	
	student	*1*	
	unemployed	*2*	
	permanently unable to work	*1*	
	housewife / househusband	*1*	
	employed	*11*	
	student and employed	*1*	
Employment rate of those employed	*part time (<80%)*	7	
	full time (≥80%)	5	
School leaving qualifications of all	*no primary school*	*1*	
	primary school	*3*	
	apprenticeship	*12*	
	gymnasium	*1*	
	higher education	*4*	
School leaving qualifications of parents	*no primary school*	*0*	
	primary school	*1*	
	apprenticeship	*9*	
	gymnasium	*0*	
	higher education	3	
Physical health*	poor	1	
	fair	1	
	good	5	
	very good	11	
	excellent	3	
Mental / emotional health*	poor	1	
	fair	1	
	good	6	
	very good	11	
	excellent	2	

* Participants rated their physical and mental / emotional health on a five point Likert scale from poor, fair, good, very good to excellent. (Appendix 2.7)

6.2. Description of the Categories

Data analysis resulted in four categories which describe the transition experience of families that live with an affected young person during his or her transition into adulthood. The four categories illuminate different areas of the family transition in coping with and adapting to the effects of the NMD relating to the functional, relational, social and emotional domains of family life. Each category puts an emphasis on a different life domain but they are naturally intertwined: 1) “Dynamics of life” - the functional domain; 2) “Balance of proximity” – the relational domain; 3) “Social ties” – the social domain and 4) “intense innermost” – the emotional domain. From the categories an interpretive theory emerged that fosters a deeper understanding of the family transition experience.

6.2.1. Category Dynamics of Life: The Functional Domain of Family Life

With most young people the transition from adolescence to adulthood is marked by growing independence from their families, physically and psychologically. In contrast, the transition into adulthood of young people living with NMD was marked by a time of continuing loss of muscular strength and loss of physical abilities leading to a growing physical dependence at a time of development that usually aims for the opposite - the young person’s growing independence and development as a person. This situation required the young person and their family to make significant adjustments in their expectations of independence and in how they were functioning as a family to manage everyday life while coping with a progressive disease and allowing personal growth. On a functional level, families had to shift their priorities and efforts by integrating

both - efforts to support the affected child's expected growth towards an independent person and efforts to cope with his or her increasing physical dependence due to the degeneration of the body.

6.2.1.1. Striving for Independence

Parents wanted to assist their children, but also to promote their child's growth towards independence, despite their physical limitations. It was clear to all family members, that adolescence was associated with the affected young person's need to become more independent and develop as a person. However, achieving a degree of independence from their families was experienced as a challenging process. Some affected individuals reported being afraid of becoming more independent and doing things on their own, as they were not used to it yet. They preferred the presence and assistance of a family member, despite being aware of the eventual need and wish for more independence. The physical limitation made young people more fragile and fearful, while family members assumed responsibility and were willing to help and give comfort even if not needed.

Affected young person:	*"I have never been able to do something on my own, I am always, I have always needed somebody… also with homework, I have never been able to do homework on my own, really never, I by myself in my room? I have never been able to focus or go somewhere on my own, no. Nowadays I will take my sister along, hey will you join me? I cannot go alone, except if it is a few metres to the bus stop. And I am still afraid sometimes, alone. It comes, it comes with age and with time, over time it gets easier. And then I will be able to do it."* *"Ich hab nie etwas selber können, ich bin immer, ich habe immer jemanden gebraucht… auch mit den Hausaufgaben, ich habe Hausaufgaben nie selber machen können, wirklich nie, ich alleine in meinem Zimmer? Ich hab mich nie selber konzentrieren können oder selber irgendwo hingehen, nein. Heutzutage nehme ich manchmal meine Schwester mit, hey kommst du mit mir mit? Ich kann nicht alleine, nur wenn's ein paar Meter sind zur Bushaltestelle. Und ich hab jetzt ein paarmal noch Angst, selbst. Es kommt, es kommt einfach mit dem Alter oder mit der Zeit, je mehr man Zeit lässt, desto besser wird's. Und dann, mach ich das dann schon.* *25:82, Family 2*

Assessment of the Situation

Some parents tried to assess the affected young person's remaining abilities to make sure that they received the right amount of support, while not pampering or doing too much for them which might inhibit their growth towards independence. To do so, they discussed and negotiated the amount of assistance required with the other members of the family to make sure that everybody shared the same understanding. Faced with the young persons' growth and physical degeneration, families reported difficulties in factually assessing the situation. Some affected young persons were described as taking advantage by asking for help they were not perceived as needing. Some reasons which led to the affected young person taking advantage have been introduced above; fear of doing things alone and being used to assistance. Differences in power and development among family members influenced how family members assessed the situation. Some younger siblings did not yet fully understand their families' situation due to their cognitive abilities which still had

to develop. Some parents had difficulties to coming to terms with their child's situation and attempted to postpone their child's loss of abilities. They were not able or willing to factually assess the situation and pushed the affected young person to his or her limits or discussed interventions such as how to conserve strengths. Strengths could be conserved by using tools or technology such as electric wheelchairs and if activities were performed by a family member.

By postponing the loss of abilities through the interventions introduced above, young people were at risk for either being pushed too hard even if their abilities were exhausted, or being spared. Being spared implies that young people would still have the ability to act, but that movements would be associated with much more difficulty for them than it would be for a healthy person. In some cases parents who postponed the young person's loss of abilities were potentially giving painful or burdensome directives. With the passing of time and increase of the young person's physical dependence and psychological independence these situations relaxed. Family members accepted their family situation better and young people were more empowered to express and negotiate their own needs.

Parent:	*"Once he said: Do you know mum, you now have to accept that I am not able to walk that far anymore. In the past he skied, he could, he did everything. He sat in the snow (laughs) and said: I don't want to ski anymore, I DON'T want to ski anymore. And I kept saying for a long time, try it again, try it again."* *„Er hatte auch mal gesagt: Weisst du Mama, du musst es jetzt akzeptieren, dass ich nicht mehr so weit gehen kann. Früher konnte er Ski fahren, er ist, alles hat er gemacht. Er ist im Schnee gesessen (lacht) und sagte: Ich will nicht mehr Ski fahren, ich will NICHT mehr Ski fahren. Und ich sagte lange versuche es nochmals, versuche es nochmals."* *3:174, Family 4*

Becoming Psychologically Independent

Eventually, during adolescence and young adulthood most affected young people became more and more independent from their families. Being independent meant for them being responsible for their own lives and making their own decisions on a psychological, emotional and cognitive level. A young person described his attitude with *"what I can do or organise on my own with my mind, that I then do that by myself"* (24:21, Family 1). Being independent meant that the young person gave instructions, while other people assisted with physical things in the sense of a more functional support. To clarify, functional support implies that the affected young person needed a helping hand on an executive level, where somebody else assisted with physical activities that he or she was not able to perform.

Through their independence, affected young people also reached a sense of normality and a certain degree of freedom, by feeling equal to their peers and not bound by their condition. Family members also rated their affected family members' life as comparable to that of a healthy person, if the young person living with NMD reached a degree of independence that allowed him or her to pursue and engage in common social and professional activities. The meaning that affected young people and their families attributed to their independence and to the limitations caused by NMD - if they felt limited by the disease or not - influenced the level of independence that young people achieved and how family members rated their family situation.

Affected young person:	*„What I actually think when I look at it like this, is that I have reached an absolutely normal level, so… well easy because I am totally… totally independent and somehow never… dependent on support. Not physical support, but more logistical / executive support. The physical part is something that I cannot control in a sense. There I always have to be as well organised as possible, so that if I need somebody, someone is there, or otherwise that I am clear on what I need so that I can do it by myself, right.“* *„Was ich eigentlich finde, wenn ich es so anschaue, dann habe ich es auf ein völlig normales Niveau geschafft, also… Also gerade einfach, weil ich es völlig… völlig selbständig mache und irgendwie gar nie… auf Hilfe angewiesen bin. Also nicht auf physische Hilfe, sondern mehr auf geistige Unterstützung. Was physisch ist, ist nochmals ein Teil, den ich nicht steuern kann in dem Sinn. Dort muss ich mich auch in dem Sinn immer möglichst gut organisieren, dass, wenn ich jemanden brauche, jemand dabei ist und sonst einfach gut abgeklärt das es auch alleine geht, oder.“* *24:21, Family 1*

6.2.1.2. Coping with Physical Dependence

Families were confronted with their child's maturation, while they also took over caring activities or arranged for external help to support the physically dependent young person. Families were used to the affected young person's physical dependence and need for assistance since childhood, but during early adolescence, the amount of assistance required increased due to the progression of the disease. During early adolescence most affected young individuals had already experienced great loss of strength and abilities such as the loss of their ability to walk which led to them being dependent on wheelchairs to allow for mobility. Despite differences in disease progression among affected individuals as well as some more stable phases during the course of NMD, the loss of muscular strength continued during adolescence eventually leading to total loss of physical abilities to perform daily activities and to maintain life sustaining processes. The amount of remaining strengths and the degree of physical limitations varied from young person to young person even if two children in the same family were affected by the same genetic condition. In some more severe cases or in more advanced stages, the degree

of the young person's physical dependence was extreme and family members compared it to that of a baby or toddler, which implies total dependence requiring constant attention by someone else. The care of an affected young person was therefore marked by an increasing need for support in all activities of daily life and around the clock. Families assisted their affected child or children with personal care, but also with nutrition, mobility and transfers and supervised therapies and treatments. Parents also organised support from other family members or professionals if they were not available to take over the care.

Parent:	*"In the morning I assist with getting up, transferring to the toilet, washing, getting dressed, making breakfast… but then he has a small table that can be attached there [to the wheelchair] and then he can eat independently. (…) If the hand falls from the armrest I have to put it back, if he wants his legs… well, if he wants to lift up his legs, I have to do that. Um, of course, if we go outside, I assist with getting dressed depending on the weather, or if he needs money I have to operate the cash machine, um, (pause), and the reverse in the evening, getting undressed and helping him to bed. And during the night, depending on how well he sleeps, repositioning him once, twice, three times."* *"Am Morgen aufnehmen, aufs WC setzen, waschen, anziehen, Frühstück machen… und dann hat er aber so ein kleines Tischchen, das man da [am Rollstuhl] drauf tun kann, dann kann er selber essen. (…) Wenn die Hand runterfällt von der Armlehne muss ich sie wieder hoch tun, wenn er die Beine will… also das Bein so hochnehmen will, das machen. Ähm, ja logisch, wenn wir rausgehen anziehen, je nach Wetter, oder wenn er Geld haben muss den Bancomat bedienen, ähm (Pause), und abends halt wieder umgekehrt, ausziehen und ins Bett bringen. Und nachts, je nachdem wie gut dass er schläft, ein-, zwei-, dreimal umlagern."* *30:34, Family 1*

Characteristics of the Care

Characterising the personal care during this developmental stage was that the affected young person needed more assistance as their abilities decreased and physical dependence increased. In addition, young people underwent physical changes including physical growth and sexual maturation, which in turn influenced caring activities and required specific family adaptations. Physical

growth came along with weight gain and increasing immobility and inability to assist with transfers of position due to loss of muscular strength. Basic care became more difficult and parents needed to find new ways of lifting and transferring the affected young person from a lying to a sitting position. They organised and used assistive technology such as hoists or they cared for the person in pairs in order to be able to do what was required.

The physically demanding work was experienced as stressful and put assisting family members at risk from back pain. Assistance with transfers and repositioning during the night was also associated with disturbed sleep and consequent tiredness. The effects of the young person's growth and weight gain on the family can be illuminated by the experience of a single parent who was the only responsible person for the care of her child. This single parent described that the weight gain was among the contributing factors for being overwhelmed which then led to the placement of the young person in a care institution.

Besides physical growth, affected young people grew sexually and formed physical signs of sexual development during transition into adulthood. A simultaneous need for more privacy emerged and family members were expected to become less likely to see each other undressed or to carry out personal care and hygiene routines. The affected young persons need for assistance, however, violated this expected family boundary, as he or she required assistance with personal care. Parents reported increasing difficulties when having to perform intimate activities such as assistance with personal hygiene and toileting. Young people living with NMD also reported feelings of

embarrassment when parents carried out personal care. Particularly delicate was the management of signs of sexual maturation including the care during menstruation and ejaculation.

If the family caregiver was of the opposite sex, some caregiving activities were more challenging, because they violated the expectations of who is allowed to give care. From early adolescence onwards established family routines of care needed to be adapted and caring activities such as using the restroom and getting dressed were preferably conducted by family members of the same sex. Caring for a female was more within the responsibility of the mother or sister, while caring for a male within that of male relative, respectively. A family involved the younger sister of an affected female in care and she assisted her father when the mother was unavailable in order to avoid a situation when the father would see his teenage daughter naked.

Sibling:	*"I have to help get her undressed when showering, and I need to wrap the towel around her so that her father, my father, can transfer her onto the lift, so that she can shower and then I help her."* *„Beim Duschen muss ich ihr helfen sich auszuziehen, und muss ihr das Tuch herumwickeln dass ihr Vater, mein Vater, sie auf den Lift tun kann, damit sie dann duscht, und dann helfe ich."* *26:47, Family 2*

Consequences on Family Life

When describing assistive tasks, parents highlighted, by and large, that they wanted to assist and support each other and did not experience that as challenging or burdensome. However, family caregivers also described many caregiving activities, which were stressful as they required a lot of time and

patience. In order to get up and get ready for the day usually a lot of time was needed, because the affected young person required a lot of assistance and experienced fatigue. In the mornings, family members had to plan for enough time in order not to get stressed. They also had to get used to be ready to assist at any time if they were at home. At home family members were “on call” and they could be interrupted for help at any minute. This prevented the family members from concentrating on an activity, because there was always the risk of them being disturbed by the needs of the young person. While some family members did not experience that as a burden, others described the need to assist as stressful and frustrating.

Sibling:	*“Um, sometimes it is simply frustrating, when they [affected siblings] are always asking for you, then you get started with something, for example, I am studying and exactly then I want to start to read or something, and then it simply comes: Sister! (laughs) Then I have to leave everything and go to him. It is maybe just ten seconds, but still the frustration that you feel, it is just huge.”* *„Ähm, manchmal ist es einfach frustrierend, eben wenn sie [betroffene Geschwister] immer nachfragen, da will man etwas anfangen, zum Beispiel ich bin am Studieren und dann möchte ich genau dann etwas anfangen zu lesen oder so, und dann kommt es einfach: Schwester! (lacht) Dann muss ich einfach alles weglassen und dann wieder zu ihm. Es ist vielleicht nur zehn Sekunden, aber trotzdem den Frust den man dann spürt ist, dieser ist einfach riesig.“* *12:76, Family 5*

Because of the need to care for their child, parents reported that they were not able to pursue their own life goals, which they needed to adjust. Depending on the family situation and the amount of external support, parents or partners experienced restrictions in their careers, as they were only able to work part time or needed a flexible job which allowed them to stay home if required. While talking about assisting, some parents reflected about their future family situation and the fact that they themselves would become older with time. They doubted

that their abilities to care for their affected children would last forever and worried about what the care would look like when they themselves were too frail to assist and would also be in need of help by others. Some parents questioned their current situation and the suitability of the support model they had chosen for the families' future.

Parent:	*"At the same time I am aging as well, I am an adult and I also get older, it is… and then the children still expect help and we have to stay with them during the night and then… Then I will also need help, because that is hard on my body. Practically… it is impossible."* *„Und gleichzeitig mein Alter geht, ich bin auch noch erwachsen und es geht weiter, ist… und dann aber die Kinder erwarten noch Hilfe oder wir müssen auch in der Nacht bleiben und dann… da brauch ich auch eine Hilfe, weil das geht auch an die Physik. Also praktisch… das ist unmöglich."* *16:62, Family 5*

6.2.1.3. Negotiating Roles and Distributing Tasks

When adapting to the young person's needs, family members negotiated their role of a caregiver with their role as a family member. Family members were naturally attached to each other and wanted to assist. Parents viewed it as their role and responsibility to care for their affected children. Usually the mother or both parents took over more caring activities as the disease progressed and physical abilities vanished. Parents were in the natural position to do so, as they were the responsible adults, had the necessary skills and were holding powerful positions within the family. Grandparents and more extended family members were rarely involved in everyday caregiving. Simultaneously to taking over more caring activities, family members did not want to be exploited or reduced to sole caregivers as they felt they had their own lives and aims and wanted to be

respected for that. In some families, roles were negotiated and role conflicts discussed to reach a situation, where all family members were able to function.

***Parent**:*	*"I tell him [affected person], what you can do by yourself, you do by yourself. I am neither your maid, nor your nurse or anything else. Um, what you can do, you do. And where you need help, I am very happy to help you."* *„Ich sage, was du [betroffene Person] selber machen kannst, machst du selber. Ich bin weder dein Dienstmädchen noch deine Pflegefachfrau noch irgendetwas. Ähm, das was du selber kannst machst du selber. Und dort wo du Hilfe brauchst, helfe ich dir sehr gerne."* *3:187, Family 4*

In other situations the distribution of tasks and the management of role conflict were more challenging such as when expectations differed and discussions and negotiations were not strategies employed by all family members. To resolve role conflicts and distribute caring activities, family members needed to communicate with each other and coordinate. It resulted in a higher risk of caregiver burden for the person experiencing a role conflict as well as in potentially difficult family interactions and strained relationships, if this need was not satisfied.

Sibling:	*„She [mother] then probably also said to my father: You have to do something and similar, you are not doing anything. Because my father was definitely also someone, who was… who was rather focused on his own comfort. Um, yes, because my mother just really did a lot, she did really a lot, right? (…) And yes, and then my mum probably often felt frustrated, when he did not do what he actually could have done."* *"Die [Mutter] hat dann vermutlich schon mal zu meinem Vater gesagt: Mach du mal etwas und so, du machst ja gar nichts. Weil mein Vater ist halt sicher auch einer gewesen, der sich eher… der eher bequem gewesen ist. Ähm, ja weil meine Mutter halt einfach wirklich sehr, sehr viel gemacht hat oder? (…) Und ja, und dann hat sich meine Mutter vermutlich auch oft gestört, wenn er noch nicht gemacht hat, was er eigentlich hätte machen können."* *46:58, Family 12*

Involvement of Healthy Children

Parents of more than one child decided on the degree of their healthy children's involvement in caring activities depending on the actual need and their values and principles for parenting. Some parents tried to minimise their healthy children's involvement and allowed the sibling to have time on their own. They planned activities when the parent's attention was fully on the healthy sibling only. These families' intention was to protect the healthy siblings' childhood and the parent - child as well as the sibling's relationship with each other from harm. Although siblings were used to be involved in family life and to assist their affected brother or sister, they reported to be thankful if their parents gave them the possibility of just being a child or a sibling and not having to take over the role of a family caregiver.

At the same time as parents worried about the effects of assisting on their healthy children's lives, the families' centre of attention was clearly drawn towards the affected young person because of his or her special needs. It was clear that in some situations the siblings' help was welcomed and allowed for the family to function better. The siblings' support comprised not only an important resource for disease management, but also for the parents' respite. Therefore, some parents involved their healthy children to a high degree in assisting the affected person as well as in doing chores.

Effects on Siblings

Taking over responsibilities, despite the degree of involvement in care, influenced siblings' lives including their perception of themselves, their level of maturity and their decisions and actions. A young sibling revealed her idea of

being similar to her mother despite her own young age, because she switched caregiving and household tasks with her. Some siblings reported career choices that were either related to their caregiving experience within their family of origin (e.g. in the social sector) or the clear opposite. They either wanted to do something they knew from their homes and which they rated as meaningful to them, or they wanted to do something totally different from their current family reality.

Sibling:	*"Yes, right, whether vacuuming, making the bed, dusting, I can do everything, um, when my mother is busy with my little brother. Because now we sometimes take turns, if, I am nearly the same as her [mother], and then we can take turns: I do that, you do that, you do that, I do that."* *„Ja, oder, entweder staubsaugen, Betten machen, Staub wischen, kann ich alles machen, ähm, wenn meine Mutter sich mit meinem kleinen Bruder beschäftigt. Weil wir wechseln jetzt ein paarmal ab, wenn, ich bin ja fast gleich wie sie [Mutter], und dann, also, kann man sich immer abwechseln: ich mache das, du machst das, du machst das, ich das."* *26:64, Family 2*

Moreover, most siblings identified with their family of origin and felt very close. A healthy sibling described such a close attachment to her family that she reported not wanting to leave them to start her own family. When thinking about their future lives, some siblings also reflected about the genetic nature of their affected siblings' disease and the potential risks for passing on the disease to a new generation. Their family experience contributed to their own future plans of having a family and offspring. This can be explained by the fact that siblings knew what it meant to live with a family member affected by a chronic progressive illness and they could not imagine living a similar life as their parents.

Sibling: *„I don't know if the fact that I don't necessarily want to have children has something to do with my sister and how she lives. Yes I, yes I… I don't know. It is maybe a bad thing; I have a very good relationship with my sister, but now imagining myself with children? I don't see it, don't see that at all. And what I also think, I don't see myself with a normal child, let alone a disabled child, right."*

"Ich weiss nicht ob das, dass ich nicht unbedingt Kinder haben will etwas damit zu tun hat, wie meine Schwester ist und lebt. Ja ich, ja ich… ich weiss es nicht. Es ist vielleicht schlimm; ich habe es sehr gut mit meiner Schwester, aber jetzt mich mit Kindern? Sehe ich, sehe ich gar nicht. Und was ich auch glaube, ich sehe mich nicht mit einem normalen Kind, geschweige denn wenn das Kind behindert wäre, oder."

47:87, Family 7

6.2.1.4. Integrating Assistance by External Caregivers

While some families tried to keep the external support and its effects on family life to a minimum, the majority of families were supported by professionals or informal caregivers in order to master everyday life. The presence of an external caregiver influenced family life and was sometimes described as intrusive. This was the case if the external person was staying overnight, spent a prolonged time at the families' home or interfered in private matters. Young people and family members described that they usually behaved differently when somebody else was around. They tried to control their emotions such as mood swings more often and were friendlier, even if they did not feel like it. Some external caregivers, with whom they had good relationships and more continuous contacts, became part of the family and these encounters were described as positive.

Living at the Family's Home

Some young people were living at their family's home where they were supported by their family members or received additional support by the Swiss home care service which is called "Spitex". Young people had regular visits by

home care nurses which performed basic care and treatment for a certain amount of time a day, while the family was responsible for assisting the rest of the time. The home care service was described as supportive and as home care nurses were present, family members found time to rest and recover from their everyday life stress.

Parents described the care of the young person living with NMD as complex and requiring continuity of the home care service delivery. It was not helpful, if there were too many changes as it took the community nurses a while to get to know the special needs of the affected young persons. Continuity of staff was not only in favour of the quality of care, but also beneficial for the young persons' development as it favoured the development of the young persons' social skills and gave them a sense of belonging. When growing older some young people took over more responsibilities for their care by organising the home care service and discussing their schedules and needs.

Parent:	*"Above all it is the continuity of service that is very important… that there are not too many changes, which is important. Because such changes, even with EVERY very experienced nurse that cares for Tina [names have been anonymised], they need a certain amount of time to really get to know Tina and learn all the little tricks that she has developed herself or have emerged. And um, yes, they practically all, all became friends."* *„Da ist vor allem die Konstanz von der Betreuung wichtig… dass es nicht zu viele Wechsel gibt, was wichtig ist. Weil so Änderungen, JEDE auch erfahrene Krankenschwester welche Tina [Namen wurden anonymisiert] übernimmt, sie benötigen ein gewisse Zeit bis sie wirklich Tina kennen und alle kleinen Tricks kennen, welche sie selber entwickelt hat oder entstanden sind. Und ähm, ja, die sind praktisch alle, alle Freundinnen geworden."* *18:39, Family 7*

The financial reimbursement for some of the home care professionals' tasks was described as problematic. Monitoring because of the risk for respiratory

problems in situations of invasive ventilation, for instance, was not an intervention fully reimbursed by the disability insurance. The disability insurance distinguishes between professionals caring activities that comprise active interventions and care activities that were more passive such as monitoring. Families had to negotiate their needs and fight for receiving financial support to ensure the care, as monitoring was clearly required for the affected young persons' safety.

Living at Home with Care Assistants

Some older young people employed non-professional care assistants that were available around the clock and assisted with basic care, chores and if necessary also with writing notes at school or university. Young people clearly favoured care assistants over other services, because they could be chosen deliberately, allowed them to live at home and to have their own daily structure, which was described as very valuable. Some young people trained lay people of the same age such as students. Being cared for by students and spending time with them promoted the young persons' social integration as well as positively influenced their sense of freedom and normality. Young family members reported that friendships developed out of their caregiving relationships with students, which allowed for more meaningful social interactions with others and diminished the young persons' feeling of being different.

Young people living with the support of care assistants reached a high level of independence from their families. They took over responsibility for the organisation of their own care, which can be rated as a sign of maturation.

Young people were the care assistants' employers and they were responsible for the organisation and administrative management of their own care, while they received financial support for the expenses from disability insurance. The employment and management of care assistants was described as challenging and demanding as it included recruitment, interviewing and training the personnel as well as handling work schedules, sick notes and substitutes. Despite these duties, young people wanted to make the effort in order to become more independent.

Affected young person	*„Since (…) [year] I have been doing it by myself, I look for my people on my own and employ them and make payroll, yes exactly. It takes a lot of effort, but of course I find the financial contribution that we receive for care assistants good and positive, but it is also a lot of work and I can simply say, it gives me a kind of independence and a lot of freedom that I am simply willing to make the effort."* *„Seit (…) [Jahr] mache ich das alleine, ich suche meine Leute alleine und stelle sie an und mache Lohnabrechnungen, ja genau. Es ist recht aufwändig, aber ich finde klar ist der Assistenzbeitrag gut und positiv, aber es ist auch viel Arbeit und ich kann einfach sagen, mir gibt die Art von Selbständigkeit so viel Freiheit, dass ich einfach bereit bin den Aufwand auf mich zu nehmen."* *14:46, Family 7*

Living in a Long Term Care Institution

Other young people were living in care institutions or stayed there during school days where they were supported by professionals. Living in a long term care institution had positive and negative implications for these young people and their families. The long term care institution allowed for the affected young person to become more independent from the family so that strained family relationships calmed. Young people also lived in close proximity with other affected young people who shared a similar situation. By spending their

everyday life with peers who all had a physical or intellectual limitation, feelings of acceptance and belonging were fostered.

Young people who were living in long term care institutions were more independent from their families as these were not their primary caregivers anymore. Care institutions offered care services, yet they were highly organised and functioning because of standardised processes and routines.
While young people gained independence from their families, they were governed by institutional standards. To give an example, when moving into the long term care institution young people were allocated to groups, but were not able to decide about the group constellations. They shared their everyday life with other chronically ill people and with professional caregivers whom they were not able to choose. Difficulties such as strained relationships emerged if they did not agree with each other.

Some young people moved to the care institution by choice, which made settling in easier. If they had to leave their family home due to the escalation of the family situation or lack of financial support, getting used to a new living situation and external caregivers was described as challenging. Some young people felt lonely and isolated, when separated from their family. Moreover, the type and quality of care by professionals and family members differed. Family members were more flexible and had a greater emotional involvement, while care by professionals was more standardised and differed in type of relationship and intimacy, the conduct of individual caregiving tasks and the availability of support. Professional caregivers had to schedule caregiving activities as they

cared for more than one affected person. Young people had to wait for their turn, whereas parents' focus was on one person only.

Sibling: *„And there it is of course… you are dependent on the caregiver, you probably also have, yes, fear of intimacy would perhaps be hard to say, but it is another type of relationship, right? You don't always have, um, you cannot talk about the same things presumably or you are simply, yes, I don't know if you are somewhat more distant. But yeah it is just different in a sense."*

„Und dort ist es natürlich… du bist auf Betreuer angewiesen, hast vermutlich auch, ja, Berührungsängste ist jetzt vielleicht etwas schwer zu sagen, aber ein anderes Bezugsverhältnis, oder? Man hat nicht immer, ähm, man kann nicht über das Gleiche sprechen vermutlich oder man ist einfach etwas, ich weiss nicht ob es distanzierter ist oder so. Aber anders halt in dem Sinn, ja."

46:42, Family 12

What was also described as different was that young people had to express their needs more often if living in a care institution. At home there were established routines and rituals with the primary family caregiver, so that not everything had to be asked for. For example, a mother performed caregiving activities, without being asked by the affected person first and this initiative was described as disburdening by an affected young person who had moved into a care institution. The affected young person described how he was relieved of the burden to ask for support through the family members' automatic assistance.

Being a Back-up Carer

Despite achieving independence from their families through living in a care institution or with care assistants, families remained back-up carers. Even if parents were not responsible for the daily care anymore, they remained in the background to step in and assist if they were needed. During weekends or on

holidays when the affected young people spent time at home or when there was nobody else available to assist, parents or siblings filled in and helped out. Parents believed that the young persons' independence would not remain forever as they were always aware of the young persons' persistent vulnerability and a potentially rapid change of the health situation.

Parent: *„In times of need, she can ask me anytime, if I can come to look after her. And that is really… I can stop working earlier in the afternoon or do a night shift or similar… that happens. But she already knows that she can call me, if there were really no one there."*

"Wenn es Not am Mann ist, dann kann sie mich jeder Zeit fragen, ob ich sie hüten komme. Also das ist eigentlich… da kann ich auch einmal früher an einem Nachmittag aufhören oder abends Nachtwache oder so… das gibt es schon. Aber das weiss sie schon, dass sie mich anrufen kann, wenn wirklich niemand da wäre."

22:48, Family 6

6.2.1.5. Using Assistive Technology and Tools

To make everyday life feasible and care easier, families described the importance of developing and using creative interventions and assistive technology and tools as supportive measures. Among the most important devices were electric wheelchairs that allowed for mobility and respirators that ensured vital processes. Families also developed and used smaller tools during their everyday life. They had to be very creative and develop interventions which were necessary, as the life situation and the needs of the affected young person were special and required adapted solutions. During the transition into adulthood, young people felt an increased need for being like their peers and to do what they do, while managing the implications of the disease. A parent described her creative solution of buying a specific type of clothes in a bigger size for her daughter, which would not reveal itself when the affected young person was sitting in a wheelchair. These large trousers without buttons were in

favour of the young persons' independence as they enabled her to get dressed on her own by pulling them up after using the restroom.

Some families were very creative and they made adapted tools themselves. One family member created a bottle holder that could be attached to the wheelchair for when the affected young person was going out and wanted to have a drink. Most young people living with NMD were not able to hold their drinks, because of their lack of strength. A customised tool like a bottle holder empowered them to accept when somebody was offering a drink, alleviating potential awkwardness of the interaction and fostering participation in social activities.

Parent:	*"And he [partner] made the lift outside. And this table that can be attached [to the wheelchair] and in the bathroom (…) the edge of the basin that was a little too broad, he was able to remove that, just because… And Matt just now got a drink holder that is attached to the headrest which has two joints and is just a metal container, where he can place a PET-bottle or a cup for when he goes to a concert, right."* *„Und eben, er hat den Lift draussen, hat er gemacht. Und dieses Tischchen, zum dranmachen, und im Badezimmer (…) Und der Rand bis zum Lavabo war ein bisschen zu breit, das konnte er auch wegnehmen, einfach so... Und Matt hat jetzt gerade so eine Getränkehalterung am Rollstuhl, wo an der Kopfstütze angemacht ist, wo so zwei Gelenke hat, und einfach so ein Metallgefäss, wo er eine PET-Flasche oder einen Becher reinstellen kann, für an Konzerte, oder."* *30:41, Family 1*

The use of assistive technology and tools was rated as very helpful and it made everyday life of the young person and caregiving in general much easier. It gave the family a sense of freedom, as they could do things that otherwise would have been impossible. Automatic doors and controllable devices allowed the affected young person to open doors and turn on the lights. By making available assistive technology and tools, family members were able to make an

impact, which diminished their feelings of helplessness in a situation where they had no control over disease progression.

The organisation of more expensive assistive technology and tools, such as electric wheelchairs and architectural adaptions were described as time-consuming and exhausting as it involved contacts with the disability insurance services. In order to get financial support, family members had to fill in applications and argue for their needs. Despite their efforts some of the applications did not result in positive outcomes. What was described as helpful was to involve experts who knew how to argue more effectively, so that applications would have a better chance to get accepted. Families had to learn how to write good applications in order to get the tool or assistive technology they needed.

Parent:	*"And the IV [disability insurance] too, they make cuts where they can. And um, yes you have to provide good justification and most of the time it is better when you ask an expert for help, who can tell you. We, for instance, needed to adjust this passageway around the house, to raise it with tiles, because there was a gap. And Marvin could not get over the gap. And I wrote an application on my own and then, DENIED (pause). And then somebody came from the organisation W. and said: Listen Mrs Norton, write it like this and so on and so forth. Then I rewrote it and it was accepted."* *„Und die IV [Invalidenversicherung] auch, also eben die streichen was sie können. Und ähm, ja da muss man alles immer gut begründen und meistens ist es am besten wenn man irgendeinen Experten dazu holt, welcher einem sagen kann. Wir haben zum Beispiel hier den Durchgang, hier da ums Haus herum mit diesen Platten eben erhöhen müssen, weil da so ein Absatz war. Und Marvin konnte nicht über den Absatz (…) Ich habe das der IV geschrieben von mir aus und so, ABGELEHNT (Pause). (…) Und dann ist einer gekommen von der Organisation W. und sagte: Hören Sie Frau Norton, formulieren sie das so und so. Dann habe ich das neu formuliert und es ist angenommen worden."* *3:142, Family 4*

6.2.2. Category Balance of Proximity: The Relational Domain of Family Life

The young family member's condition created a situation of physical dependence where the family or an external person needed to be physically close and provide assistance with performing tasks. This physical dependence and the families need for protection resulted in close proximity and high family involvement at a time when they were expected to develop more flexible boundaries where children could develop and differentiate, reach a degree of freedom and distance to their families and become more independent. Close proximity and family involvement in each other's private lives challenged this developmental need and had impacts on the family transition experience, especially in the relational domain of family life.

6.2.2.1. Being Involved in Private Matters

During the transition into adulthood the affected young person developed a need for more freedom and independence which comprised an own private life distinct from their family. Their sexuality awakened and they developed interests in romantic relationships and a need for more privacy, where family members were not expected to be deeply involved. Assistance with personal care or some family practices which were beyond basic caregiving activities violated individuals' privacy and integrity, as well as challenged social expectations and family relationships. Among these practices were parents sleeping in the same room with their affected teenager or assisting the affected teenager with sexual acts such as masturbation, by positioning the hands if the young person wanted so. Activities related to the affected young person's sexuality were described as

taboo subjects. Families had few discussions about how affected young people could have a fulfilled sex life, which influenced the family's involvement.

Parents reported being aware of the high sensitivity required by these situations and related family interactions and they described that they and the affected young person had to mutually overcome some family boundaries which did not feel natural at first. Natural implies here that they handled these situations differently with their healthy children, where they were not involved in private matters of this kind. The affected young people's intellectual integrity allowed them to decide if they wanted their family members' involvement in personal spheres and vice versa. To some degree, however, they were in a relationship of dependency which made the situation more complicated, as even if young people reached some degree of independence and freedom of choice, they were still dependants.

Significance of Involvement

Parents experienced their involvement in each other's private lives as meaningful, despite related challenges and compromises. Sharing the bedroom was described as important by parents as it made assistance during the night easier. They were able to reposition the affected young person with short interruptions of their sleep, while they also ensured the young person's safety in case of emergency. Family members shared a bedroom since childhood and they described it as custom and did not want to change this practice during adolescence. Therefore, parental needs were side lined, despite the potential impacts on the parental relationship and privacy. Assistance with sexual acts was judged as important, as masturbation was viewed as natural. The physical

limitation, however, limited the affected young person in performing such a natural sexual act on their own. They needed assistance with these activities just as with other domains of life.

Parent:	*"Someone told me that her son masturbates sometimes … uh, and, that is actually also something natural to do and she tells me how… then she puts his hand, right, if he cannot do it, then she has to put his hand on his penis and I told David at some point that if he feels the need, then he can do that, which he then did, but that was a strange situation. And then I just placed a towel, a towel on top and then I, you have to wash him, right, you have to be able to do that. I mean, these are things that you have to get used to. Mutually, right? Then he said to me, mummy, can you put my hand down there, for example, right."* *"Da hat mir eine gesagt ihr Sohn macht manchmal Selbstbefriedigung… äh, und ist ja eigentlich auch etwas Natürliches und da hat sie erzählt… dann tut sie ihm die Hand, oder, wenn er es nicht kann, dann muss sie ihm irgendwie die Hand dort hinlegen auf das Glied und ich habe dem David auch irgendwann mal gesagt, wenn er mal das Bedürfnis hat, dann darf er das machen und das hat er ja dann auch gemacht, aber das ist doch schon ein wenig komisch gewesen. Und dann habe ich dann einfach mal so ein Tuch hingetan, ein Frotteetuch oben hin und dann habe ich ihn, muss man ihn einfach waschen, oder, das muss man auch können, ich meine das sind ja schon auch Sachen, an die man sich schon daran gewöhnen muss. Gegenseitig, oder? Dann hat er gesagt, du Mami, tust du mir die Hand runter, zum Beispiel, oder."* *44:111, Family 12*

In light of the above, these family members acted like they did, because it was meaningful to them and they also viewed it as valuable for their child's health and development. For these reasons, family boundaries became blurred and family members made sacrifices to each other's privacy and adapted family relationships. The intimacy that emerged between family caregiver and the affected young person during these interactions differed from other family relationships, such as from parent-child relationships with their healthy children. Sharing intimate situations required very close and trustful connections, but also increased the risk for enmeshment, rigidness and high interdependence. A natural degree of family interdependence emerges between family caregivers

and their physically dependent child, but it becomes more extreme, when family members are highly involved and when supporting and protecting is the family caregivers' life task or defining role and this view is shared or accepted by the affected child.

6.2.2.2. Balancing between Freedom and Protection

Family members described being torn between their need for freedom of development comprising detachment, while they also wanted to be close and protect each other. Mothers reported that they wanted to protect their children from any possible harm and that they would prefer to be ill if that meant that their affected children could be healthy. Some families were very concerned and anxious about the affected young person reaching more independence and they tried to protect their children from difficult experiences at the expense of their freedom. To prevent their affected child from potentially harmful situations they took measures to control and prevent situations from happening.

An example of such a situation was that parents monitored an affected teenager's social media account. The supervision of social media accounts empowered parents to keep track of the young persons' social interactions with others and assess their social skills and media competence. They were also in control of the contents of interactions and knew with whom their child was chatting. Families did not only monitor, but also correct the child's behaviour if necessary and had a say in the child's choice of contacts and therefore their choice of friends. This gave the parents a sense of security as they ensured that the affected teenage daughter was protected from any harmful social

experiences, which they expected to be more difficult for her to deal with than it would be for her healthy siblings.

Parent:	*"And everything that I tell you here, everything that I say is really only, because I care for her, because I, um, with all these clear rules, maybe they wouldn't be so strict if she were a normal child, but precisely because she has this handicap, and so that she will not be disappointed too often in her life, which I have the feeling can be really dangerous among such individuals. That they already have something [the disability] and if somebody then disappoints them, that that then would have more severe consequences."* *"Und alles was ich hier erzähle, alles was ich sage ist wirklich nur, weil ich mich um sie sorge, weil ich mich um, ähm, all die klaren Regeln, vielleicht wären die nicht so streng wenn sie ein normales Kind wäre, aber genau weil sie das Handicap hat, und damit sie im Leben nicht zu viel enttäuscht wird, was wirklich gefährlich ist, bei solchen Menschen, habe ich das Gefühl. Das sie schon irgendetwas [die körperliche Einschränkung] haben und wenn man dann enttäuscht wird von irgendjemandem, dass das dann schlimmere Auswirkungen hat."* *27:61, Family 2*

Some families were less anxious and allowed for more freedom or they lost their influence over the young person as part of their transition into adulthood. These parents worried and suffered when their child's decisions led to uncertain outcomes or had negative consequences such as if the affected young person's actions comprised unhealthy exploits. This was the case when the affected young person tested out his or her limits and engaged in activities that threatened their health. If the young person chose to drink alcohol and smoke cigarettes or marihuana, family members struggled to deal with these situations, as they wanted the best for their child and to preserve their health. The health situation of these young people was already severely compromised by NMD and activities such as drinking or smoking were more harmful for them than for healthy teenagers. Families described it as particularly difficult to deal with the

fact that their affected child was smoking while using a ventilator for respiratory support.

Parent:	*„And he then, I was glad, he also quit smoking pot. And that was rather difficult for him and afterwards he said, good then I will simply smoke cigarettes and I mean when I saw that or… him on the balcony outside, him with the cigarette, he wasn't able to light them himself, and someone put a cigarette into his mouth and, well, that was terrible for me to see and the crazy thing was, that you could see in the breathing tube, well that it created tar deposits there."* *„Und er hat dann, ich bin froh, er hat auch aufgehört zu kiffen. Und das ist für ihn ziemlich schwierig gewesen und nachher hat er gesagt gut, dann rauche ich halt Zigarette und ich meine als ich das mal gesehen habe oder er auf dem Balkon draussen, er mit der Zigarette, er hat die ja nicht selbst anzünden können, dann hat ihm eine da die Zigarette in den Mund getan und, also, das ist für mich furchtbar gewesen zum Sehen und das verrückte war, das man an dem Beatmungsschlauch gesehen hat, also dort hat es Ablagerungen vom Teer gegeben."* *44:37, Family 12*

Effects of Physical Distance

Physical distance from the family was described as being in favour of the young person's freedom of development. When young people spent time outside of home, it gave them freedom to make their own experiences and grow more mature by making mistakes, taking risks and learning from them. Changes in the living situation by moving out from home or receiving care provided by external caregivers was described as beneficial for family detachment and gaining more independence, and it had positive effects on previously constrained family relationships. In other words, after moving out, some parents and siblings were free of their caring duties and there was more time for other activities, which led to less stress and better relationships. Physical separation was not described as exclusively positive, but also as a worrying idea and a very painful process for both families and the affected young person. Distance

was associated with fear, feelings of failure and guilt, feelings of isolation and loneliness and fewer contacts among family members.

6.2.2.3. Experiencing Effects on Family Relationships and Interaction

Effects of Close Proximity

Most family members developed close relationships with each other as a result of the close proximity required by their family situation. Family members spend a lot of time together during everyday life and also during leisure time and holidays. They were together a lot and developed a strong emotional bond between each other. Affected young people and healthy siblings described their relationships with a parent and an affected person, respectively, as the person is *"my best friend"* (25:83, Family 2). Close relationships and regular contacts consolidated during adolescence and persisted, even if the young person became older and eventually moved out from their family home. When the young person was living with friends or the partner and care assistants or in an institution, families kept in touch and visited each other. Even if they moved out, most young people retained their children's room at their family home where they spent their holidays and weekends.

Close proximity was also associated with challenges. Parents were very much involved in their child's life since childhood with the consequence that they had little privacy and few time for themselves and for their romantic relationship with each other. Among the contributing factors to these situations were the child's dependence and need for assistance, the difficulty to organise substitutes and the parents' concerns about leaving the young person alone. Parents decided

not to spend time without the child or they brought him or her along if they spent time outside of the family home such as with friends.

The growing young persons developed their own interests and attitudes and if families kept their childhood practices, this led to disagreements and tensions. Young people did not share the same interests with their parents anymore and they revealed feeling frustrated when they wanted to spend time without the family, but were bound to each their family because of their need for giving or receiving care. Furthermore, after a disagreement between teenager and parent it was not possible for them to just get out of each other's way. Before bedtime, family members had to find a way to reconcile, as the young person needed support to get to bed.

Parent:	*"Yes just simply because in addition to being disabled, he was also going through normal puberty and totally annoyed me and um, was kind of not able to distance himself, like it would have been healthy for his age, because at the same time he simply was totally dependent on me. And that was sometimes really a bit much."* *"Ja halt einfach dass er zusätzlich zu der Behinderung auch noch ganz normal in der Pubertät gewesen ist und mich extrem genervt hat und ähm, sich auf eine Art nicht so abgrenzen konnte, wie es gesund wäre in dem Alter, weil er einfach gleichzeitig völlig auf mich angewiesen war. Und das war manchmal schon ein bisschen happig."* *30:25, Family 1*

Shared values and understandings influenced the family experience. If family members shared similar experiences, they described having a better understanding of each other's reality implying knowing and understanding each other's situation better. Understanding the other person better was related to positive parent-child relationships, more empathetic interactions and facilitated the development of a strong emotional connection. Adolescence was described

as a time when young people benefitted from their parents' understanding and support, in particular because they were experiencing changes and challenges.

Affected young person:	*"Yes, she had in a sense an awareness, that she just… (…) she was (…) [different from others] and I… well, am, physically disabled… and because of that for both of us… it was something that connected or joined us, where she then showed a lot understanding for me during puberty."* *„Ja, sie hat in dem Sinn die Baustelle gehabt, dass sie halt… (…) [anders] war [wie andere] und ich… also körperlich behindert bin… und durch das ist es gerade für beide… irgendetwas, das verbindet oder verbunden hat, wo sie dann sehr viel Verständnis für mich aufgebracht hat in der Pubertät."* *24:139, Family 1*

Tensions and Family Conflict

While some families were connected with each other, other family members felt emotionally more distant. A less strong emotional bond and fewer contacts with each other were described when there were unresolved emotional states including feelings of guilt and anger and when the young person's condition was experienced as overwhelming or burdensome. Some younger siblings did not feel comfortable with the affected siblings' condition. In order to not face these situations too often, they decided not to disclose the situation in front of others so that these others would not ask any inconvenient questions. They also decided to have fewer contacts with the affected sibling. Spending time with the affected person required additional efforts and was related with caregiving activities, which made it even more difficult for these siblings to be engaged.

Sibling:	*"I didn't really spend a lot of time with my brother, I also have to say that. Um, yes, it has not always been so easy for me… to do something with him, right? It is also because, you have, you have to deal with his care and the whole with kit and caboodle. And it probably would have been possible, but I somehow did… it was just still an additional effort to some extent."* *„Ich habe jetzt nicht wirklich viel mit meinem Bruder gemacht, das muss ich auch sagen. Ähm, ja, es ist für mich halt auch nicht immer so einfach gewesen… mit ihm etwas zu machen, oder? Es ist halt auch, man muss, man muss schauen mit der Pflege und allem Drum und Dran. Und es wäre vermutlich schon möglich gewesen, aber ich habe mich mit dem irgendwie… es ist halt zum Teil auch noch ein Aufwand."* *46:28, Family 12*

Among the reasons for being overwhelmed with the family situation were family communication and constellation. If parents of a single child were having a conflict with each other, the dependent person often felt involved or had to mediate. In turn, problems between the affected person and one parent potentially led to difficulties within the parental relationship and the other parent-child connection. In the worst case, physical violence resulted from triangulation of conflicts which were combined with psychological or emotional difficulties and excessive care demands. If family situations escalated they potentially resulted in harmful experiences for those who were less able to defend themselves from psychological or physical abuse.

Affected young person:	*"I am simply the person who suffers the most. I am the person, who has no money, but… you two have to figure out in a sense how you want to deal with each other, a little. In the past I have always been on my mother's side and that led to a lot, during puberty… well it created a lot of conflict with my father. He also used to hit me sometimes … simply because he was overwhelmed with it all… and…yes, in the evenings he often used to drink and then he hit me and that was a little like a… yes, like a vicious circle."* *"Ich bin einfach der Leidtragende. Ich bin der, der kein Geld hat, aber… ihr zwei müsst in dem Sinn selber schauen, wie ihr miteinander klar kommt, ein wenig. Vorher hatte ich immer so die Position meiner Mutter bezogen und das hat halt auch viel, in der Pubertät viel… also zu vielen Konflikten geführt mit meinem Vater. Er hat mich teilweise auch geschlagen… einfach weil er halt überfordert war mit dem Ganzen… und… ja, er trank halt oft abends und schlug mich dann und das war so ein wenig… ja, wie ein Teufelskreis."* *24:128, Family 1*

Tensions among family members emerged, because of the one-sided distribution of caregiving activities and if a family member did not do what was expected. If help was needed but unavailable, the other family members were left to deal with the situation. This caused tensions and potential conflicts. Tensions and conflicts among parents due to one-sided distribution of tasks were potentially leading to separation. During transition into adulthood, parents who participated in this study did not report any new parental conflicts that led to separation or divorce. Most single parents that participated had already separated around the time the child had been diagnosed. The death of the affected child was then again described as a potential difficult situation for parental relationships leading to separation or divorce, especially if one family member did not manage to come to terms with the loss of the loved one.

The Challenges of Parenting

Parents stated they had similar challenges for parenting with respect to their healthy and chronically ill children during their transition into adulthood. Some parenting tasks, however, differed among healthy and affected children, as parents assessed their needs and abilities differently. Affected young people were not expected to complete an education or have a career. It was more important that they were happy, whereas in contrast, healthy siblings were expected to be more successful. In addition, some of the family rules were applied to all of the children, while others were specifically valid for the affected young persons. The affected child was not allowed to stay up as long as the healthy children because parents wanted them to rest and maintain their strength. Children did not report feelings of envy, but they described it as challenging to accept that treatment and degree of attention among the healthy and affected siblings differed.

When parenting more than one child, parents faced the dilemma of deciding whom to give their attention to and to what extent. In all families, the parents' attention focused on the affected young person, because of his or her special needs. Concurrently, they were conscious about their healthy children's needs which they also tried to satisfy. They were torn between the needs of their children and even though they wanted they could not *"be in more than one place"* (44:60, Family 12). Parents established family rules allowing them to be in control of their family life and relationships. One family decided to spend a certain amount of time each week with the healthy sibling alone. Parents also involved themselves in sibling interactions, as they wanted to prevent future intra-familial difficulties by regulating the time the affected young person and the

sibling spend together, to prevent straining their relationship. The healthy sibling was allowed to do some things by him or herself, without having to take the affected sibling along.

Parent:	*"And sometimes I get involved and make it really clear to Nick [healthy sibling]: You can go on your own without Marvin [affected young person]. And Marvin: You have to accept that now. (…) I think it is important that Nick is not going to say some day that he always had to bring his disabled son, uh, brother along. I don't want that."* *„Und da mische ich mich manchmal ein und sage wirklich Nick (gesundes Geschwister) ganz klar: Du darfst alleine gehen ohne Marvin [Betroffener]. And Marvin: Du akzeptiert das jetzt. (…) Ich finde das wichtig, dass der Nick nicht einmal sagt, dass ich immer meinen behinderten Sohn, äh, Bruder habe mitnehmen müssen. Das möchte ich nicht."* *3:88; 3:93, Family 4*

6.2.3. Category Social Ties: The Social Domain of Family Life

Families shared values, beliefs and expectations with their former generations and they were also confronted with standards and expectations of their community, society and culture. Some families who participated in this study had migrated from other countries and in addition to Swiss culture, they also brought along cultural expectations from their countries of origin. When families living with a young person affected by NMD tried to conform to these standards and expectations difficulties emerged. The family situation clearly challenged some of what was expected and family members experienced difficulties and had to develop a more flexible understanding of social standards and expectations.

6.2.3.1. Being a Member of a Group

Being a member of a social group such as a family, a local community or a society, was of high importance to most families. Conformity with the standards

and expectations of a group was significant and beneficial. Living as part of a group was described as following their norms and expectations, which enhanced the family members coming to terms with their family situation. Being religious, for example, facilitated the family members' meaning making and fostered their coming to terms with NMD by explaining that the illness is wanted by a higher authority. Being a member of a group was also related to a sense of belonging and a subsequent commitment for mutual support. An affected young person described her family's commitment as follows: "*They promised me, that they always will be there for me" (25:97, Family 2*).

What members of most families expected from each other was conformity with family rules and expectations and actions towards the benefit of the group. During adolescence young people developed their own worldviews and opinions, while adopting some of the values and beliefs of their family and social group and challenging others. Young people were also more interested in social contacts outside of their families than before and as friendships and romantic relationships emerged, they got to know different ways of life. These experiences and consequent shifts in thinking had impacts on family life, such as on family relationships when living with NMD.

Families experienced fewer interpersonal tensions when they shared similar values and beliefs, which was the case for families with younger children. During adolescence, tensions emerged because of a change in worldviews and attitudes, which resulted in conflicts if not mutually respected. To give an example, a growing young person did not share the religious beliefs of the rest of the family. Parents were religious and visited religious sites regularly. The

physical dependence of the affected teenager did not allow for the family to spend much time separated from each other so that the young person had to participate in religious activities, but rebelled against this practice.

Sibling:	*"My parents go quite often to visit a religious site and that creates some conflicts because my younger [brother], Barney does not really believe in God and then he hates going to the religious site. But he just has to come along; he has to come along, because we can't leave him alone. Precisely, yes, he is almost forced to join us, and sometimes they [brothers] also have to… thus they have to get used to it and let go and do what the parents want."* *„Meine Eltern gehen noch recht oft zur religiösen Stätte und da gibt es auch ein wenig Konfliktsituationen weil mein jüngerer [Bruder], Barney glaubt nicht so an Gott, und dann hasst er es sehr zur religiösen Stätte zu gehen. Aber da muss er halt mit; er muss halt mitkommen weil wir ihn nicht alleine lassen können. Eben, ja, er ist fast gezwungen, dass er mitkommt, und manchmal müssen sie [Brüder] auch… also sich daran gewöhnen und loslassen und das machen was die Eltern wollen."* *12:37, Family 5*

6.2.3.2. Being Responsible for Each Other

Members of a family were expected to take responsibility for each other. Parents were not only emotionally bound to their children, but they were also expected to be accountable for their wellbeing. In some of the families, family members felt obliged to care for the young person at the families' home because of cultural or religious expectations. An affected young person reports: *"[They] view it as their obligation, so to say, to help us"* (11:45, Family 5). It was therefore not socially expected to place the affected young person into institutional care. If parents sent their child to an institution because of parental overload and excessive demands, this violated their personal expectation of being a good parent. Despite having positive effects on the wellbeing of all family members in the longer term, it was described to be extremely challenging and left parents with a feeling of failure, guilt and emotional pain.

Parent:	*"I had the feeling that I am just chickening out and am sending this boy away, and…"* *„Ich hatte das Gefühl ich kneife jetzt einfach und schiebe diesen Jungen ab, und...* *30:8, Family 1*

Besides parents, healthy siblings also had some degree of responsibility for their chronically ill family members. Most of them did not have the power to challenge what was expected nor were they able to choose what their everyday life looked like. Even if healthy siblings questioned some of the families' standards and expectations, such as their being involved in the care, in the end they had no other choice than to conform, which increased the risk for intra-familial tensions.

Sibling:	***Interviewer**: "What were situations that have led to conflicts within the family?"* ***Participant**: "(…) in the past I have always… I never wanted to help her, and after that… a few times I also needed time to recover, and after she went to country L… I had some respite and now I always want to help her, no matter what, I am always ready to help."* ***Interviewer**: "Was sind so Sachen, wo in der Familie zum Streit geführt haben?"* ***Teilnehmende**: „(…) früher bin ich immer… habe ich immer ihr nicht gewollt helfen, und nachdem… ich habe ein paarmal auch Erholung gebraucht, und nachdem sie in Land L gewesen ist… habe ich ein bisschen Erholung gehabt und jetzt will ich ihr immer helfen, egal was ist, bin ich immer hilfsbereit."* *26:83, Family 2*

Families described cultural expectations about how caring activities were distributed among family members. Mothers and other female caregivers such as grandmothers and sisters were more often incorporating the role of a family caregiver. Because of cultural reasons, not all family members were allowed to take over the same types of caring activities. If the affected young person was

male, then the female family members were not expected to assist with personal care, while the father took over a more prominent role.

Parent:	*"Perhaps in specific cultures it is like that, there are certain things that only the father is allowed to do, like undressing. Or that I then find out that he dreams something, dreamed something and the mother then stays in the background. (…) taking care of intimate areas… it can happen that somebody… preferably not the mother, but when I am not around she does, otherwise I do it. As a father somehow… the brother who is two years older preferably not, and definitely not the older sister!* *„Wahrscheinlich in gewissen Kulturen ist es so, gewisse Dinge darf nur der Papa machen, also eben die Kleider ausziehen. Oder dass ich dann erfahre dass er träumt, etwas geträumt hat und die Mama bleibt da etwas im Hintergrund. (…) im intimen Bereich die Pflege… es kann schon mal vorkommen dass jemand… Mama lieber nicht, aber wenn ich nicht anwesend bin oder, aber sonst mache ich es. Und als Vater irgendwie… der zwei Jahre ältere Bruder lieber nicht, und die ältere Schwester auf keinen Fall!"* *2:128 / 2:131, Family 3*

Sometimes everyday life was unfeasible and families felt unable to fulfil what was expected from them. During the transition into adulthood parental tasks included being responsible for the care and making sure that the children attended school and that they themselves were going to work to contribute to the family's maintenance. In sum, the specific family situation required realisation of various tasks, which sometimes resulted in stressful situations.

Punctuality and not letting other people wait was described as an important value by some families. For example, parents described mornings as very stressful, when they had to assist their affected children to get up and ready for the school bus. In the morning they felt it particularly difficult to care for their child who was still dormant or tired and moving slowly. Family members had to be organised and to get up early, but as it was challenging to have the time and patience, many mornings remained stressful and created interpersonal

tensions. Some parents hurried and pushed the affected child who sometimes left without having breakfast. When reflecting about these situations families described feelings of inadequacy as they were not able to fulfil what was expected; being punctual. They dealt with these situations by controlling their emotions and trying harder to stay calm.

Parent:	*"These are contentious issues, when I rush him and get angry and inpatient. And later I also raise my voice: So, now get going, the others are waiting outside and after that there are other children and they will have to wait, too, because Marvin is the first one who gets picked up. (…) Yes and he replies: Yes I am doing it and I am coming. And later we end up screaming at each other."* *„Das sind Streitpunkte, dass ich ihn hetze und verärgert werde und ungeduldig werde. Und nachher auch lauter werde: So und jetzt mach mal, und sie warten draussen, und nachher kommen andere Kinder und nachher müssen sie auch warten, weil Marvin ist der erste welcher abgeholt wird. (…) Ja und er sagt: Ja ich mache ja und ich komme ja. Und dann nachher am Ende schreien wir uns gegenseitig an."* *3:262, Family 4*

6.2.3.3. Being Different is Normal

The effects of NMD on family life marked the families' difference from a healthy family. During transition into adulthood, the young persons' awareness of being different intensified, as they developed cognitively and had more frequent contacts with others outside of the family with whom they compared themselves.

Affected young person:	*"When I now imagine going to school, where everybody has a moped or everybody has a partner and is talking about the first time and you are simply nowhere and can't participate. You cannot ski and you cannot tinker on your moped (laughs). You can maybe tinker the wheelchair, but not the moped! (Laughs) Right! You know… maybe such things where you more CONSCIOUSLY notice: I am different!"* *„Wenn ich mir jetzt die Schulzeit vorstelle, in der jeder ein Mofa hat oder jeder einen Partner und vom ersten Mal erzählt und du einfach noch nirgends bist oder gar nicht teilhaben kannst. Du kannst nicht skifahren gehen oder du kannst kein „Töffli" (Mofa) frisieren. (lacht) Du kannst vielleicht den Rolli frisieren, aber nicht das Töffli! (lacht) Oder nein! Du weisst… vielleicht solche Sachen, bei denen du BEWUSSTER merkst: Ich bin anders!"* *21:53, Family 6*

Being in familiar surroundings, spending time with family and friends and engaging in activities they were used to, gave young people a sense of normality. Growing up within a specific family situation influenced family members' view of what normality is. Healthy siblings were used to their family situation. Even if they recognised differences between their and other families' lives, what they knew was "normal" for them. A sibling describes caring for her ill sibling: "*I am used to it, how can I say, I grew up with it and for me it is a normal task*" (12:08, Family 5). This implies that family members got used to their specific situation and it became their normality.

Affected young person:	*"I would never have felt differently somehow. I think that even through my social environment… that they sometimes even forget that I am in a wheelchair. At some point I am simply normal, right. Sometimes it happens to my boyfriend that he says: Come on, let's go to this club or bar! And then I say: Is it wheelchair accessible? Ah! I don't even know. Because he is not thinking about it and that is nice on the one side, because then it is not always the disability in the forefront, but you as a person."* *„Ich hätte mich jetzt auch nie irgendwie anderswertig gefühlt. Ich glaube, dass selbst durch mein Umfeld… dass sie manchmal sogar vergessen, dass ich im Rollstuhl bin. Irgendwann bin ich einfach normal, nicht wahr. Meinem Freund passiert manchmal auch, dass er sagt: Komm wir gehen in diesen Club oder in diese Bar! Und dann sage ich: Ist es dort rollstuhlgängig? Ah! Das weiss ich gar nicht. Weil er das gar nicht einmal denkt und das ist einerseits auch schön, weil dann ist nicht immer die Behinderung im Vordergrund, sondern du als Mensch!"* *21:41, Family 6*

Families varied to the extent how much they stressed their normality or their difference. Some families viewed the physical disability as a single element of their lives that did not essentially describe who they were. These families felt more "normal", but some situations, challenged their attitude. If they had to apply for disability benefits such as for financial support when buying assistive tools or technology or in order to get invalidity pension, they were required to list all their limitations and stress their deficits. A deficit oriented view of their "self", however, was not how these individuals perceived themselves. In order to get the necessary financial support they had to conform to the categorisations and requirements imposed by society. These situations were described as frustrating, as they required them to make diverging efforts, being and living normally, but reporting their difference.

Affected young person:	*"Sometimes it is a little ridiculous, because you have to give evidence that you are disabled and at the same time you have to try to do everything that is possible and that somehow your wishes are recognised. For example, with the financial support for assistive living, because you have to fill out a self-declaration and I mean I am a person who believes, hey, I am cooking, I go shopping, I do everything on my own and with them I simply have to say: No, I cannot cook, somebody has to take the things out of the fridge for me and I know what I want to buy if somebody is opening the fridge for me, because I can't. And that is sometimes really not that fun... actually you really have to say what you cannot do."* *"Manchmal ist es ein wenig lächerlich, weil du beweisen musst, dass du behindert bist und gleichzeitig auch versuchen alles Mögliche zu machen und deine Wünsche irgendwie so ein bisschen anerkannt werden. Z.B. bei den Assistenzbeträgen, weil du musst immer wieder so eine Selbstdeklaration ausfüllen, und ich meine ich bin ein Mensch der findet, hey ich koche, ich gehe einkaufen, ich mache alles alleine und bei denen musst du einfach sagen, nein ich kann nicht selbst kochen, jemand muss mir die Dinge aus dem Kühlschrank nehmen und ich weiss schon was ich einkaufen will wenn mir jemand den Kühlschrank aufmacht weil ich nicht kann. Und das ist manchmal nicht so lustig… eigentlich musst du wirklich sagen was du nicht kannst."* *14:94, Family 7*

During transition into adulthood, young people experienced confusion about their own identity and ambivalence of feelings and thoughts. They described a strong desire to be normal and to live a normal life. Living a normal life comprised having a normal job in the free economy and not in a protected workplace and being well integrated in society. Young people wanted to be treated normally, also because they found that it would better prepare them for their future lives. One affected participant recommended: *"By treating them normally not wrapping them in cotton wool… that is maybe beneficial and nice and good as long as you are a child, but it will not be useful for later life, as you will not be wrapped in cotton wool at the work place."* (21:69, Family 6).

At the same time as they wanted to be normal, young people knew that they had different needs and they expected others to treat them differently, according to their specific needs. Wanting to be normal and being treated

normally conflicted with their difference and their need to be treated in a special way. Young people were confronted with ambivalent needs and family members and external people varied in how they coped with these needs. Some treated the affected person rather "normally" while others gave them special treatment. Some of these situations resulted in confusion, tense emotional states and stressful interactions.

Affected young person:	*"Then I said in a loud voice: Damn it! I do not just go to place M. for fun. I must also go to work, just the same as you! And if you let me leave [the train] first, then I can go down the ramp and am gone and you can… you can run! (…) In part I have the feeling that I had to show these people somehow, I don't know, maybe almost show them somehow that although I am in wheelchair, that otherwise I am totally… well, am completely normal."* *"Dann sagte ich mal ganz laut irgendwie: Verdammt! Ich gehe auch nicht nur zum Spass nach Ort M.! Ich muss dann auch arbeiten gehen, genau gleich wie ihr anderen auch! Und wenn ihr mich zuerst rausgehen lasst, dann bin ich die Rampe runter und weg und dann könnt ihr… könnt ihr rennen! (…) Teilweise habe ich da irgendwie einfach das Gefühl muss ich diesen Leuten auch mal zeigen - ich weiss nicht - vielleicht fast irgendwie auch mal zeigen muss, dass ich zwar im Rollstuhl bin, aber dass ich eigentlich sonst voll… also, ganz normal bin."* *23:26, Family 10*

6.2.3.4. Having a Social Life

Affected young people who transitioned into adulthood wished for social interactions outside of their families. Families reported different barriers to the young person's social participation imposed by expectations of members of society. Making new contacts was particularly difficult for young people. Their health situation and the related different physical appearance influenced how they were perceived and accepted by others. The first impression that one generates is usually marked by a scan of the others' physical appearance, which clearly placed the young persons' physical limitations in the foreground.

The visibility of the condition and maybe also a concomitant lack of physical attraction, as the affected young person did not meet the socially accepted beauty norms, contributed to barriers to making contact. Most people hesitated to make contact, because they were not used to these situations. They felt intimidated or overwhelmed and did not know how to approach the affected young person. Some also questioned the young person's intellectual integrity, as they viewed it as associated with the physical limitation. Once the first contact was made, the situation changed and the affected young person was not judged by his or her physical appearance anymore, but also by their personality.

Affected young person:	*"I notice that from the outside a person sees, she is simply sitting, she has a tube into her throat and that is somehow the picture of the disability where distance between me and others is created. The moment they let themselves get to know me, then it changes quickly, then people also see what's behind the disability, but the first contact is always difficult."* *"Ich merke, dass von aussen eigentlich eine Person merkt, sie sitzt einfach, ist eine, die einen Schlauch im Hals hat und das ist irgendwie das Bild von der Behinderung wo wie ein Abstand halten ist zwischen mir und anderen Personen, sobald man sich darauf einlässt mich kennenzulernen, dann wechselt es schnell, dann sehen die Leute auch was hinter der Behinderung steckt aber der erste Kontakt ist immer schwierig."* *14:68, Family 7*

Another barrier to social interaction and integration was if the affected young person needed round the clock presence of a caregiver due to their mechanical ventilation. Since most caregivers were adults, as a consequence of their presence, healthy peers refused to bring the affected young person along, as they wanted to be among their peers. In addition, some social interactions with their school mates were difficult or emotionally harmful, as affected young people were teased, felt excluded or were treated differently because of their

special needs. Spending time with peers, making new friends and finding a romantic partner were therefore described as rather frustrating endeavours. Nevertheless, most young people became more confident with time, grew emotionally more mature and were able to meet new people with openness and not take insults too personally.

Contacts with other Affected Families

Contacts with other affected families were described as positive and negative. Positive was that families did not have to explain their situation and felt understood and accepted: *"They know what you are talking about, right. I can tell somebody who is not affected and the person has no clue, how that really feels, right"* (*44:112, Family 12).* Family members could focus on what really mattered to them, while not having to explain their life situation. By contrast, meeting other affected families was experienced as unhelpful, as it was burdensome to hear about other families' difficulties and created a negative spiral of stress. Therefore, some family members and affected young people stressed the importance of having contacts with other people that shared the same reality, while others felt that their natural social environment was what they needed and they were not looking for contacts with other disabled.

Spending time with other young people who were living with a chronic condition was described as positive. E-Hockey, for instance, was referred to as a sport that allowed for the young person to spend time with other affected young people. They were able to make new contacts with people outside of their family environment, whereas family members could be actively involved in trainings or as spectators. An additional positive factor about E-Hockey was that besides

fostering social integration and being a fun and meaningful activity, it was also associated with aiming for a shared goal and experiencing success and self-efficacy.

Affected young person:	*"Thursday, that is the only day that is more special, because I have training from six until eight. That is such a… yes, always one of the highlights of the week! (laughs) (…) Yes, because I simply enjoy very much playing hockey, I mean I was already fascinated by all kinds of hockey as a young boy. So whether floorball, ice hockey or field hockey and it is simply the only sport that I can play with my disability. So… and it is just… I can play with a really young team. (…) They are quite exciting. So if you are otherwise just sitting at home a lot and not doing a lot, it is then just… yes… the training is a very good diversion. So… and we keep having goals that we are working towards."* *„Donnerstag, das ist der einzige Tag, der spezieller ist, weil ich abends Training habe von sechs bis acht. Das ist so ein… ja, eines der Wochenhighlights jeweils! (lacht) (…) Ja, weil ich halt sehr gerne Hockey spiele, also ich war schon als kleiner Junge sehr fasziniert von allen Hockeyarten. Also ob Unihockey, Eishockey oder Landhockey und es ist halt der einzige Sport, den ich mit meiner Behinderung spielen kann. Also… und ist halt… ich kann bei einem recht jungen Team mitspielen. (…) Also… sie sind noch recht belebend! Also, wenn man sonst halt recht viel zu Hause sitzt und nicht viel macht, ist es halt dann - ja - das Training eine sehr gute Abwechslung. Also… und wir haben auch immer wieder Ziele, auf die wir hinarbeiten."* *24:101, Family 1*

Living in a Romantic Relationship

Finding a romantic partner was difficult. Most individuals living with NMD who participated in this study were single. When looking for a potential partner or when making new contacts it was facilitating if the person had previous experiences with a handicapped person. If they were familiar, making the first contact and engaging in deeper relationships was more likely. Besides being sensitised to the other's reality, the partner also had to be more open and ready for getting to know the other person and engage in a different type of relationship that was less physical, but more platonic in nature. A major difficulty for couples where one partner was affected by NMD was their inability for

physical closeness including sexual activity. Even small gestures of affection like kissing or hugging the partner, were not possible because of the young persons' lack of strength and immobility. They were dependent on the healthy partner for physical contact.

Affected young person:	*"What is the most difficult for me or when I see other "pedestrian couples", is well the physical proximity, just simply that you in a sense… yes that… I mean, pedestrians do that and many pedestrians… they don't even notice anymore, when they are close to each other, instead it is rather… there are many small hand movements that they are able to make. For example, just putting the hand on the shoulder or holding the hand of the partner and um… Yes, or simply just hugging or kissing the partner spontaneously or just… the whole physical aspect depends on the fact that the partner is able to come up to you and give that to you."* *"Was ich am schwierigsten erlebe oder halt auch wenn ich gerade die „Fussgängerpärchen" sehe, ist halt die körperliche Nähe, halt einfach dass man in dem Sinn... Ja, dass… ich meine, die Fussgänger machen das so und viele Fussgänger… merken gar nicht mehr, wenn sie sich körperlich nahekommen, sondern es ist einfach… es sind ganz viele Handbewegungen, die einfach gehen. Zum Beispiel mal die Hand auf die Schulter legen oder die Hand des Partners halten oder ähm… Ja, oder halt einfach den Partner spontan umarmen oder zu küssen oder einfach… der ganze physische Aspekt lebt halt sehr davon, dass der Partner halt auf dich zukommt oder das halt dir auch geben kann."* *24:56, Family 1*

When the partner was affected by NMD, the healthy partner had to balance and negotiate his or her role as a partner with that of a caregiver and experienced role conflicts similar to parents and siblings. If the care was provided by caregivers external to the family it relieved the potential for role conflicts for the partner. Due to the physical dependence of the affected partner, there was also the risk that the affected person did not prioritise his or her own needs in order to lessen the healthy partner's burden, and these situations were among the reasons for breakups.

Affected young person:	*"I have seen many relationships fall apart, because for the "pedestrians" [those able to walk] it was simply too much… and they were not able to really say why, but in the end it was always about the caregiving, well in that sense the care and the lack of physicality, well in that sense, that is the biggest gap or the biggest difficulty."* *„Ich sah schon viele Beziehungen auseinander gehen, weil es halt für die Fussgänger dann einfach zu viel war… und sie konnten es auch gar nie richtig begründen, aber schlussendlich lief es immer darauf hinaus, dass halt einfach die Betreuung, also in dem Sinn die Pflege und die fehlende Physis, also in dem Sinn, dass das das grosse Manko ist oder die grosse Schwierigkeit."* *24:62, Family 1*

Some reactions of the wider society towards couples where one partner was living with NMD were described as hurtful. Living in a partnership with a disabled person was viewed as an act of mercy of the healthy person, instead of an equal partnership, which was expressed in front of the couples.

Having a Sex Life

During adolescence affected young people developed their sexual identity. Despite being part of the young persons' lives, sexuality was described as a taboo subject by families and professionals. An affected young person who was living in a romantic relationship described that she engaged in regular sexual intercourse and took contraceptives to prevent pregnancy. When she met with a new gynaecologist or physician, the professional was more or less surprised to learn about her being sexually active. Some young people lived their sexuality by contacts with professional sex workers, while still others were single and felt that they wanted to be sexually active only in a romantic relationship.

Affected young person:	*"There are people who sometimes order a woman, but I could never do that, that is totally against my principles. I don't know why, it doesn't cross my mind to do something like that, only with a woman that I love or like or I don't know, yes."* *„Es gibt auch Leute die manchmal eine Frau bestellen, aber das könnte ich nie machen, das spricht ganz gegen meine Prinzipien, ich weiss auch nicht, es kommt mir nicht in den Sinn das zu machen, nur mit einer Frau die ich liebe oder gern habe oder ich weiss auch nicht, ja."* *20:41, Family 11*

6.2.4. Category Intense Innermost: The Emotional Domain of Family Life

The time when young people affected by NMD transitioned into adulthood was associated with intense emotional states. Among the greatest hardships for families were being exposed to multiple losses and having to come to terms with the expected death of the young person at an early age. Despite these prospects, most families accepted their situation and tried to make the best of their lives. Despite the many challenges they faced, adolescence was also described as a time when they were full of hope and joined their forces to fight for a better life.

6.2.4.1. Grieving for Loss and a Future

The transition into adulthood was characterised by the families' grief for the losses they had experienced and expected to experience in the future. Families grieved for the young persons' physical decline and loss of abilities, which were rightly viewed as symptoms of the deterioration of health while drawing closer to death. They also experienced anticipatory grief by losing trust in the idea of a future life they initially had for themselves, but which now seemed unreachable and reflected their limited options.

Affected young people, parents and siblings reported feelings of sadness, fear and emotional pain as part of their grief. For some families these feelings were more intense than for others. A sibling described: *"You cannot really even not be sad"* (7:56, Family 3). For other family members fear and sadness were more intermittent and manifested from time to time. When family members were confronted with a deterioration of health or when they compared their own lives to healthy others, they remembered their losses, which revived grief.

Understanding the Consequences of NMD

While parents remembered the time of diagnosis when being informed about their child's life limiting condition as shocking, adolescence was particularly difficult for affected young people, because they developed the cognitive and intellectual abilities to comprehend and fully understand the consequences of the disease and its meaning for their lives. Adolescence was the time when affected young people started to realise that their condition was not only permanent, but also progressive leading to death at an early age. Affected young people had to learn to deal with their own finiteness at a young age, also by witnessing the death of an affected sibling or other affected young people they knew. These situations were not only difficult for affected young people, but also for other family members as it reminded them about their family situation and the potential rapid change of the young person's health situation.

Adolescence was a time when young people started to orient themselves and when they framed a plan of what they wanted to aim for in life. However, never before have they been fully aware of their own situation and coping with this truth and its implications during a time of development and aspiration was

overwhelming. In addition to the intense emotions that a young person goes through when striving for a vivid social life and starting to define and pursue major goals in life, these young people were also confronted with the highly destructive information of having a life limiting condition and facing death at a young age. During this time, most affected young persons' went through an emotional rollercoaster including feelings of anger, sadness and confusion and they were nervous and were being difficult. Family members struggled with their intense emotional state and found it very difficult to find the right measures to support the young person's coping.

Parent: *"For a time, I think at the beginning of puberty it was difficult, he was rather nervous then. (…) He got angry because of small things. Often he went to his room and used the reverse gear and with his wheelchair he slammed the door shut with such a power that you had the feeling… the wheelchair has an incredible power, the electric wheelchair (coughs)."*

„Eine Zeit lang, ich denke anfangs Pubertätszeiten war es schwierig, da war er recht nervös gewesen. (…) Wegen Kleinigkeiten war er wütend. Oft ging er in sein Zimmer und nahm den Rückwärtsgang und mit seinem Rollstuhl schlug er so fest die Türe zu, dass man das Gefühl bekam… der Rollstuhl, der hat unvorstellbare Kraft, also der Elektrorollstuhl (hustet)."

2:194, Family 3

Receiving health education and information about the disease, was highlighted as facilitating the young person's understanding and acceptance. Talking about disease-related issues, however, was experienced as emotionally stressful. Most family members avoided talking about the disease and its implications during their everyday family life or they limited these conversations to a minimum. By doing so they were able to put negative thoughts and feelings aside, focus on the more positive aspects of life or simply living from one day to the next. As a consequence of this practice, some young family members were

left alone to deal with their questions and uncertainty. Others were informed by externals, as young people received health education by professionals of the health care systems or at school.

Parent: *First of all he is in a school where there are others with disabilities. Secondly, he had a physiotherapist (…) who talked with him a lot about it. He has two friends, two friends who have the same condition (…) and then he asked the physiotherapist and why does he [friend] have oxygen, and why did he [friend] have to stiffen his back? And they discussed it."*

"Erstens mal ist er in einer Schule wo es auch andere Behinderte gibt. Zweitens hat er eine Physiotherapeutin gehabt (…) welche sehr viel mit ihm darüber geredet hat. Er hat Kollegen, zwei Kollegen gehabt wo das gleiche Bild haben (…) und dann fragte er die Physiotherapeutin und warum hat er [Kollege] Sauerstoff, und warum musste er [Kollege] den Rücken versteifen und so weiter. Und die haben das zusammen diskutiert."

3:169, Family 4

6.2.4.2. Experiencing Recurring Health Crises

Adolescence was a time when families experienced situations related to their child's vulnerability including emergencies and crises of health. Emergency situations or crises were terrifying and had an immediate impact on the family's life. At the same time as they fully focused the families' attention on the health situation of the ill family member, families remembered it in the longer term as a life-changing experience. They not only experienced the immediate and direct physical threat, but it also remained in their memory, causing uncertainty and fear of a recurring situation.

Parent: *„It was a horror scene then at our home. We were all (clears his throat) eating lunch and then he had… a piece of meat simply got stuck in his throat. And, uh, in a few seconds he turned red. He could not cough, could not breathe. And then they started, the others, the rest of the family, to scream and make noise."*

„Es war eine Horrorszene da bei uns Zuhause. Wir waren alle am (räuspert sich) am Mittagessen und dann er hat… es ist ihm einfach ein Stück Fleisch im Hals stecken geblieben. Und, ähh, in ein paar Sekunden ist er rot geworden. Er konnte nicht husten, nicht atmen. Und dann haben sie angefangen, die anderen, der Rest der Familie, zu schreien und zu lärmen."

2:206, Family 3

Families were confronted with recurring crises, uncertainty about when the next crises would happen and a resulting fear of the future. They feared the deterioration of health and the death of the ill young person. The need for final life-sustaining measures such as ventilators, tracheotomies or a feeding tube, were among the situations that families were concerned about the most, even if they allowed for the young persons' prolonged life. Healthy siblings worried not only about the affected young persons' health, but they were also concerned about their parents' wellbeing as these were highly involved in the young persons' life and care and at risk for physical and emotional hardships.

Hospitalisations

As a consequence of emergencies or crises of health, as well as for planned interventions such as for stabilisations of the back, affected young people were hospitalised. Most families described hospital stays as challenging, because they experienced professionals' lack of competency and expertise in dealing with their family members' situation. Families described the affected persons' needs as special and different from the needs of ordinary patients. The young persons' ability to communicate, for instance, was most of the time

compromised when they were hospitalised. In addition, their fear of death related to respiratory distress made caring for them even more difficult.

Parents who were experts in the care of the affected young person and able to communicate with them, stayed at the patients' bedside to give reassurance and to make sure everything went well. Parents had managed hospital stays like this since childhood and most kept this practice also during the child's transition into adulthood. A parent described negative effects of this practice; when a family caregiver spent a prolonged time at the hospital, they neglected their duties at the families' home, which resulted in interpersonal tension and also limited the affected young person's growth towards independence.

Contacts with Professionals

Encounters with health professionals that were experienced with patients living with NMD were described as great resources. They knew the affected young persons' special needs and were able to bring the necessary time and patience for dealing with a complex care situation. The affected individuals' immobility was associated with the need for assistance in all activities of daily life and the young person demanded a lot of support which was time-consuming and stressful for hospital staff. If the professional caregiver planned the care accordingly, it resulted in a more relaxed care situation and better relationships.

Some young people had good contacts with health care professionals with whom they had their routine checks at regular intervals at specialised centres for muscle diseases. These services were highly specialised and most young people knew and trusted their treating physicians. The transfer from the

paediatric to adult care services was described as a necessary step within the health care system, but few participants mentioned it as major concern.

Some family members' relationships with health care professionals were more strained and lacked trust, which was also reflected by the young person's refusal to have routine checks or the family's fear and presence at the hospital. Most family caregivers developed routines of care in order to manage everyday life and these routines were associated with a certain rigidity and inflexibility, which contributed to the difficulties with health care professionals. Some young people felt that the focus of the medical consultation was more on the disease and not on them as a person. They reported not feeling understood or listened to by their health professional, which resulted in difficulties in negotiating and accepting recommended treatments and interventions.

What added to the complexity was that adolescence was a time when young people were not interested in treatments and not able to see their future benefits. They refused treatments and therapies despite their potential. A young person described that he never wore splints and had now developed contractures in his hands, which caused him to regret refusing to wear splints. Overall, family members wished for more open conversations with their health care professionals, for the recognition of the family caregivers' expertise and for more competent professionals who were trained to care for patients with special needs.

Affected young person:	*„I know my body so well and… yes. I know exactly, how the wind blows. What it is like to be in hospital Q. Which pill I have to take at what dosage! And the health professionals do know a great deal, but not in my case, right. And it is still so… difficult… sometimes. Yes, almost a little scary, so the thought of being hospitalised in Q after a severe injury. I would not know exactly, if they would do the right thing, just because … because they are simply not trained for special cases, but instead for the routine cases. Yes."* *„Ich kenne meinen Körper so gut und… ja. Ich weiss genau, wie der Karren läuft. Wie es im Spital Q. ist. Welches Medikament ich wie dosiert nehmen muss! Und das Fachpersonal weiss zwar sehr gut Bescheid, aber nicht in meinem Fall, oder. Und es ist schon noch so… schwierig… manchmal. Ja, fast schon ein wenig beängstigend, so dass die Vorstellung bei einer schweren Verletzung ins Q zu kommen. Ich wüsste nicht genau, ob da richtig gehandelt würde, weil es einfach… weil man einfach nicht auf die Spezialfälle trainiert wird, sondern auf die Normalfälle. Ja."* *21:25, Family 6*

6.2.4.3. Strategies of Coping and Adaptation

In order to cope with difficulties and challenging emotional states, family members employed strategies that facilitated their wellbeing and functioning as a family. While some family members shared similar pattern of coping, as they influenced each other and inherited strategies from former generations, there were also differences. Some people avoided talking about the disease, while others coped better with difficulties by sharing their experiences. If the latter family members had nobody within the family to talk to, he or she either suffered in silence or turned to externals. Externals were professionals, friends or people they met at self-help groups.

Parent:	*„But he is one… one that really bottles everything up inside. And I am one that talks openly about it and I can somehow… well I feel that it is easier for me than for him. When I talk about it, then somebody else knows about it, and um, it somehow feels supportive when they listen to me. But he is not able to talk about it."* *„Aber er ist der... einer der das wirklich alles in sich reinfrisst. Und ich bin eine, die offen darüber redet und kann es wie… also ich fühle es so, dass ich es leichter habe als er. Wenn ich darüber rede, dann weiss noch jemand davon und ähm, so tut es mich irgendwie so wie unterstützen, dass man mir zuhört. Aber er kann nicht darüber reden."* *27:92, Family 2*

Families explained that the degree of negative emotion associated with the young person's condition was not bearable for a prolonged time. In order to be able to live with it, they had to think more positively. To see their own situation in a better light or to give it a more positive meaning, family members adapted their own perspective. To give an example, an affected young person, who was clearly limited in his everyday life, described not experiencing any challenges. He did not view his life as challenging, even though his health and abilities were severely compromised compared to a healthy person. It seems that he accepted his situation and made the best of it, by downplaying his challenges or reframing some of the hardships.

What was highlighted as helpful for coping and adaptation was to focus on the positive aspects of life and having hope for positive change. Families, for instance, hoped for a long life for the affected child and for future improvements in health care and medicine. They also managed to be positive by trying to live in the here and now and to deal with eventual difficulties day by day. One affected young person described that he simply tried to accept whatever happened and to make the best of each situation. These coping strategies empowered family members to come to terms with their families' situation better. They managed not to worry about future difficulties, but to deal with daily challenges to which they could respond.

Affected young person:	*"Well until now I have dealt with it that I have always accepted what has happened and tried to make the best of it. (…) I think that actually, so to speak, the best solution is to just take it the way it is, because I think, in my opinion, it doesn't help to get depressed and to only see the negative and such. I think it is better if you so to speak live in the moment and try to focus on the positive, yes."* *„Also bis anhin bin ich so damit umgegangen, dass ich es immer so angenommen habe wie es gekommen ist und versucht habe das Beste daraus zu machen. (…) ich finde das es eigentlich sozusagen die beste Lösung ist es einfach grad so zu nehmen wie es kommt, weil ich meine, meiner Meinung nach bringt es nichts sozusagen depressiv zu werden und nur das Schlechte zu sehen und so. Ich finde es ist besser sozusagen wenn man im Moment lebt und versucht die positiven Dinge herauszunehmen, ja."* *4:120, Family 4*

Being part of a family and receiving support, love and affection from each other was a facilitating factor that assisted and gave family members emotional strength, feelings of belonging and security even during more difficult times. An affected young person reflected: *"The goal is simply that we stick together and together we are able to do it, yes"* (4:23, Family 4).

Being religious also contributed to the family members' coming to terms with their situation, as it had the potential to give meaning to the families' life as well as emotional strength to cope with difficulties. It was described as relieving to believe that there is a higher authority which is responsible for a health situation and that illness is a challenge not a punishment or burden.

Affected young person:	*"Religion…. that it has been send by God, that it is actually not a punishment, but actually there is…. as a human… he is giving it to you, he simply wants to see how you deal with it, how you live with. And that you do not simply say, you now have this disease, that it is from Him and done. But rather, what you make of it, how you make the best of it."* *"Religion… dass es von Gott kommt, dass es eigentlich keine Strafe ist, sondern eigentlich da ist… als Mensch… er gibt dir das, er will einfach sehen, wie man damit umgeht, wie man damit lebt. Und, dass man nicht einfach sagt, du hast jetzt die Krankheit, das ist von ihm und fertig. Sondern, dass man was daraus macht, das Beste daraus macht."* *25:108, Family 2*

When families were confronted with a change in the health situation they needed to get used to the new circumstances. Parents who cared for the affected young person at home worried when they had to handle new medical aids as they were afraid of making mistakes. With the passing of time, they got used to the new situation and felt empowered and competent to deal with the management of assistive technology such as with tracheotomies and respirators. Waiting to get used to a new situation was not only described as a coping strategy used by parents, but also by affected young people.

Parent:	*"And we have qualms as lay people, we have qualms, about pressing [the tube] like so, making it tighter, that is also normal I think. But when I now see, how we do it now, or I, how I do it now, or how I did it few years ago… Now it is NOTHING, it is nothing. And I think that these people that find themselves in such a situation have to, they have to keep in mind, that it is nothing, that it is simple. (…) Because at first you feel unsettled and you think, you think about what can happen. You have to take it easy, as easy as possible, because in the end it all works."* *"Und wir haben Hemmungen als Laien, wir haben Hemmungen, so [die Kanüle] zu drücken, so fester zu machen, das ist auch normal denke ich. Und aber wenn ich jetzt sehe, wie wir das jetzt machen, oder ich, wie ich es jetzt mache, oder vor ein paar Jahren. Das ist jetzt NICHTS, das ist nichts. Und ich denke, dass diese Leute, die sich in einer solchen Situation befinden, die müssen das im Bewusstsein haben, dass das nichts ist, einfach dass das einfach ist.(…) Weil man, am Anfang ist man sehr beunruhigt und denkt, denkt, was kann jetzt passieren. Man muss es ein bisschen so nehmen, möglichst locker, weil alles geht am Schluss."* *19:52, Family 7*

Other strategies that family members used to cope with their situations were suppressing negative emotions and distracting themselves with more pleasant activities such as with sports or music and in the worst case with alcohol or narcotics. In case of ineffective coping with intense emotions, families experienced hazards to their wellbeing and mental health. Family members described social withdrawal, challenging behaviour and tense family

relationships resulting from family members' mood swings, nervousness and aggressiveness. Furthermore, they reported an increased risk for mental illness including depressive symptoms and suicidal thoughts. During adolescence, many young people living with NMD, but also some parents and siblings needed and received psychological support by a psychologist or psychiatrist in order to be able to better deal with their emotional lives.

Parent:	*"I know how it is like to withdraw from the family or from society, what types of feelings there are. You live in the dark and you want to stay there, to not hear or see anything. (…) But right during this adolescence it became obvious that he [affected young person] was moving towards a kind… towards depression, so three quarters of a year ago he started with the therapy."* *„Ich weiss wie es ist, wenn man sich zurückzieht von der Familie oder sonst von der Gesellschaft, was für Gefühle das sind. Du lebst im Dunkeln und willst dort bleiben, nichts hören und nichts sehen. (…) Aber eben genau diese Pubertätszeit zeigte, dass er in eine Art… Richtung Depressionen hineinläuft, deswegen seit einem dreiviertel Jahr hat er mit der Therapie begonnen."* *2:144, Family 3*

6.2.4.4. Joining Forces for Life

The transition into adulthood was described as a time when families joined their forces for life and fought for their rights and a better future. During this time, they bundled their strengths and put efforts into supporting their children's health, education and career, as well as their social integration. If the health of the young person living with NMD deteriorated, the families' centre of attention was on the young person and his or her getting better soon. Family members set their own needs aside and parents specifically spent a lot of time with the care of the young person. Families assisted with basic care, applied therapies and treatment and organised assistive technology and tools to manage the disease, make everyday life feasible and home accessible. Not surprisingly,

they became experts for disease management as they managed the complex life situation of their children.

Some families put much effort into supporting their affected child when it came to their social integration, education and career. The physical limitation did not allow for the young persons' participation in all activities and their physical appearance was experienced as a barrier to social interactions and making new contacts. Parents were aware of their children's needs and they tried hard to support their children in having an active social life. They spent leisure time with them, arranged social encounters and encouraged their children's social integration. But despite their efforts they had to bear the fact of not being able to fully tackle all the underlying problems. Activities with their families were not the things that affected young people wanted to do during their transition into adulthood. Young people wanted to spend time with other young people and were longing for a romantic partner and some of the adolescents refused when their parents offered to spend time with them.

Parent: *„Well that is something that actually makes me sad. That is something now when your hands are tied, when you are not able to help your own child that more likely makes me sad. And of course it doesn't help at all… I then try to say: Well come Marvin, let's go to the cinema together or something. But of course it is not the same for him to go to the cinema with his mother. But I cannot do anything more, or offer or say more. Yes, that actually makes me sad, and I strongly hope for Marvin that he can find a girlfriend or a wife someday. Because his sexuality is obviously taking place equally, it is just really much more difficult."*

„Also mich macht das eigentlich traurig. Das ist jetzt etwas wenn einem die Hände gebunden sind, wenn man dem eigenen Kind nicht helfen kann, mich macht das eher traurig. Und das bringt natürlich auch nichts… Ich probiere dann zu sagen: Ja komm Marvin, gehen wir zusammen ins Kino oder irgendetwas. Aber es ist natürlich für ihn nicht das Gleiche wenn er mit der Mutter ins Kino geht. Aber ich kann ja nichts mehr machen, oder mehr anbieten oder sagen. Ja, mich macht das eigentlich traurig, und ich hoffe ganz fest für Marvin, dass er auch einmal eine Freundin oder eine Frau finden kann. Weil die Sexualität findet natürlich genau gleich statt, aber es ist halt einfach wirklich sehr viel schwieriger."

3:255, Family 4

Parents supported their children by promoting their education and careers. Families whose children attended regular schools highlighted the positive effects it had on their children's lives - better social integration, better education and good prospects for a job. While most families were sooner or later advised to transfer their children to a school for children with special needs, which were better equipped to deal with chronically ill children, some fought for their children's' rights to attend a regular school. Attendance of a regular school was usually more complicated as there were architectural, institutional and personal barriers. Not the intellectual abilities of the young person, but their physical needs were impeding factors. The attendance of a regular school was made possible by parents and because of efforts of single individuals such as teachers.

Affected young person:	*“My parents supported me a lot and fought for my attendance in a regular classroom and they all somehow resisted it, both services even, social services and the health care system said that no, they could not offer support. And these are the type of things where I think: Hey, I already have to fight for other things, so why do I now have to fight for normal treatment? That sometimes seems a little pointless to me. (Pause) I think you should invest in finding a collaborative solution and not the opposite.“* *“Meine Eltern haben mich sehr unterstützt und dafür gekämpft, dass ich in eine Regelklasse komme und alle haben sich irgendwie dagegen gewehrt, sogar beide, Sozial- und Gesundheitswesen haben gefunden nein, sie können die Betreuung nicht bieten. And das sind so Sachen wo ich finde hey, also ich muss für andere Sachen kämpfen, also warum muss ich jetzt kämpfen, das ich normal behandelt werde? Das ist für mich manchmal ein wenig sinnlos. (Pause) Ich finde man sollte investieren, um eine Lösung zu finden wie man integrativ arbeiten kann und nicht das Gegenteil.“* *14:97, Family 7*

Families whose children attended a school for children with special needs or transferred into such a school during their school career described that fewer efforts were required from them as a family as the school took over many responsibilities. The young person’s support in activities of daily life such as in using the restroom and positioning was guaranteed by professionals during school hours. There were treatments and therapies available and schools offered support in the organisation of assistive technology and tools. In addition, educational contents and aims were adapted to the specific needs of the affected young person. A young person reported that the major benefit for him of transferring to a specialised school was that he was able to participate again.

Affected young person:	*"And there [school for children with special needs] I had the support from the therapists and that I simply had therapy and that they looked after my physical wellbeing and then I also got a wheelchair and then I could keep up with the others again and I could participate a lot more again, yes."* *"Und dort [Sonderschule] hatte ich auch die Unterstützung durch die Therapeuten und das ich einfach Therapie gehabt habe und so und auch zu meinem körperlichen Wohl geschaut worden ist und dann hatte ich auch einen Rollstuhl bekommen und dann bin ich den anderen auch wieder hinterher gekommen und dann konnte ich wieder besser mitmachen, ja."* *4:23, Family 4*

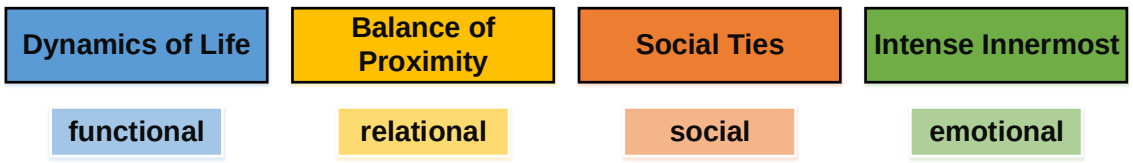
After completion of their basic education, most affected young people experienced difficulties in finding places for vocational training and jobs. The available options for disabled and the vocational guidance they received from institutions were described as unsatisfactory. This stage of their career was for most of the affected young persons and their families a time where they needed patience and endurance. Finding an internship or job in the free economy was clearly preferred as it made young people feel more normal. Job offers in protected workplaces were accepted with more hesitation, but if young people started to work there, it was described positively. Towards young adulthood, most young people who participated in this study worked part time. Having a job was meaningful for them, but not necessary in order to earn money for their living, as they received financial support from social services. In fact, even if young people wanted to work more hours, this was associated with a cut in financial support due to the regulations of disability pension.

6.2.5. Summary of the Categories

Families described their experience of living with NMD during the affected young persons' transition into adulthood as a time associated with change. Changes and associated challenges occurred as part of the young persons'

development and their physical decline and were bundled into four different fields of experience (Figure 13).

Figure 13: Categories depicting four fields of experience



The category “Dynamics of Life” comprised the functional domain of family life. Functional is understood as involving family members’ more practical actions including taking over caregiving activities, while also promoting the young persons’ development. Adolescence was described as a time when families were confronted with the progression of the disease, the young person’s physical growth, and their striving for more independence. Parents felt torn between the need to care for their children, while also releasing them into independence, whereas young people expressed worries and hesitated to do things on their own. Despite these difficulties, young people described how they reached a high level of independence, also because of receiving care by externals or living independently from their families. During adolescence, most family members took over more caring activities and adapted to the young person’s changed needs resulting from their growth and also from their sexual maturation. They negotiated and distributed tasks and used creative interventions, assistive technology and tools and received support by external caregivers to manage their everyday life. Family members revealed that caring for the affected person was not burdensome, but it increased family members’

stress and generated risks for secondary health problems, role conflicts and interpersonal tensions.

The relational field of experience which is the focus of the category “Balance of Proximity” describes the families’ difficulties to find the right degree of closeness and distance when a young person living with NMD transitioned into adulthood. During this time, young people were clearly dependent on the families’ support, but family members developed a degree of interdependence among each other. Family life was characterised by a strong emotional bond and high family involvement in each other’s lives leading to close relationships, but also having the potential for violation of privacy. During the transition into adulthood affected young people needed a certain degree of distance from their families and freedom to make their own experiences. Parents were aware of these diverging needs, but they described finding the right balance of proximity and parenting in general as challenging, because they wanted to protect their children as well as allow their freedom.

“Social Ties” is the category that described families’ social experiences as they were members of a family, society and culture. Being a member of a social group was associated with the need to conform to the standards and expectations of this group. Even if family members wanted to conform, their life situation clearly challenged some of what was expected. During the young person’s transition into adulthood, affected young people developed their own worldview and opinions, while adopting some of their parents’ views and challenging others. These discrepancies and lack of respect for diversity increased intrapersonal tension, also because family members were required to

be in close proximity to each other, due to the young person's need for assistance. In addition, family members were expected to take over responsibility for the care of their chronically ill children. If this "social tie" was violated because a parent placed the affected child in institutional care due to excessive demands, this left them with a feeling of inadequacy and was experienced as personal failure. Social integration and acceptance was challenging, as the young person's different physical appearance and physical limitation was a barrier to social contacts and activities. While family members wanted to be normal and conform to social standards and expectations, their life situation challenged some of what was expected and they were required to develop a more flexible interpretation of what normality is and experienced feelings of ambivalence, inadequacy and confusion about their identities.

Family members were confronted with an intense emotional life which was presented as part of the category "Intense Innermost". During the affected young person's transition from childhood into adulthood young people and family members were confronted with challenging emotional states as a consequence of their grief for loss and uncertainty regarding their future. Recurring negative emotions comprised sadness and fear and put them at risk for a compromised emotional wellbeing and mental difficulties. Adolescents were particularly vulnerable during this time as they developed the cognitive and intellectual skills to be fully aware of their own life situation. They experienced an emotional roller coaster which resulted in difficult behaviour and tense relationships. Families struggled with how to best support the affected young person in their process of acceptance and were also confronted with recurring health crises. Most families were coming to terms with their life

situation by using effective coping strategies such as being optimistic, living in the here and now and making the best of life. They described to be full of hope and explained how they joined their forces to fight for a better life. Parents put much effort to support the young person's health, social integration, education and career.

6.3. Description of the Interpretive Theory

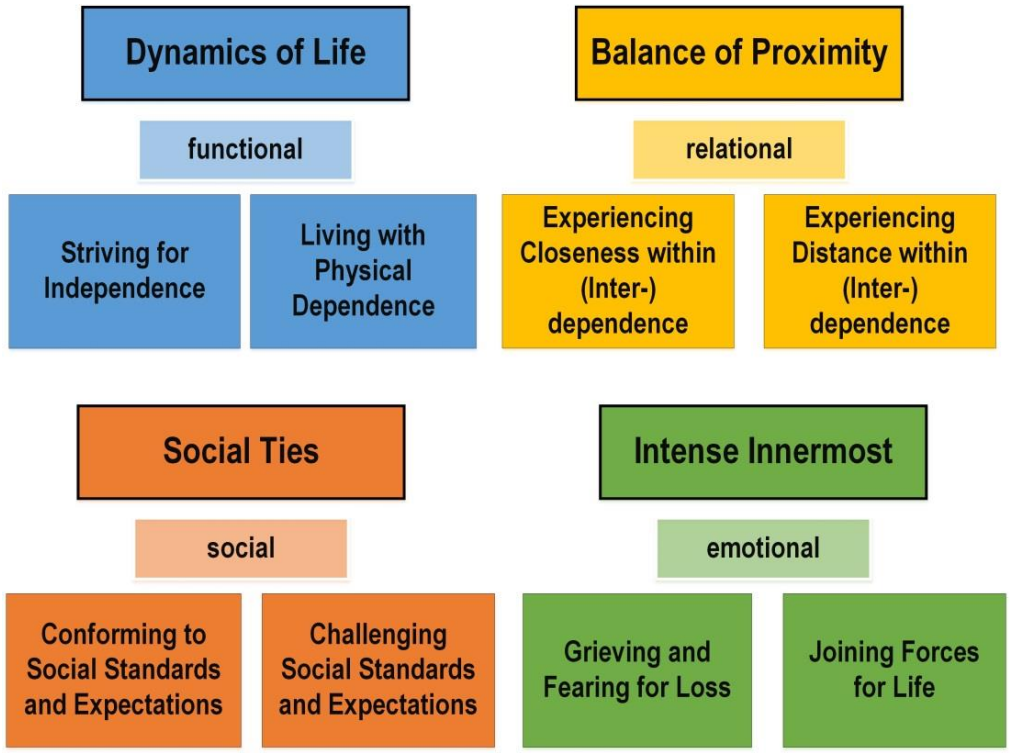
A more theoretical analysis of the four categories "Dynamics of Life", "Balance of Proximity", "Social Ties" and "Intense Innermost" generated an interpretive theory. This interpretive theory can be portrayed by its major components, which have been described and empirically supported by quotes to illuminate their grounding in family experiences. Another way to present the interpretive theory is by describing its more abstract characteristics.

6.3.1. The Characteristics of the Interpretive Theory

The analysis of the data resulted in an interpretive theory entitled "Living in a Field of Tension Between Development and Degeneration" which fosters a deeper understanding of the family transition experience. A characteristic of this interpretive theory is its dualism. Each of the four categories presented above are dualistic in nature. They are all characterised by the same dualism which is also present in the two conditions that shaped the families' life experience. These two conditions were the family development, because of the young person's transition into adulthood, and the physical degeneration of the young person's body due to his or her NMD. A conclusion that can be drawn from these life conditions is that families were living a life between two opposing life processes, development and degeneration. In addition, they also experienced

these diverging trends in different life domains (Figure 14). They were striving for independence, while living and coping with physical dependence; they experienced difficulty in balancing between closeness and distance within a family situation of (inter-) dependence; they conformed to and challenged social standards and expectations and grieved and feared for loss, while joining their forces for life.

Figure 14: Diverging trends in fields of experiences



In each of these domains of family life, family members were confronted with changes and associated challenges. Depending on the family members' strategies in coping with these changes and challenges, they potentially experience difficult consequences among which were stress and secondary health problems, as well as interpersonal tensions and family conflict. While some families who participated in this study adopted effective coping strategies

which allowed family functioning despite recurring difficulties and tension, others experienced threats to their personal integrity and physical and emotional wellbeing as a consequence of their life situation.

In addition to being dualistic, across all the domains the two extremes were also related to each other by belonging to the same field of experience, but within which they were extremely diverging and therefore potentially conflicting. Family members were not only confronted with two extremes which both required their efforts and were potentially challenging when singled out, but they also had an additional effect. Families were torn between the two opposing extremes, which resulted in feelings of ambivalence and inadequacy.

6.3.2. Consequences for Family Life

Among the consequences of these multiple and diverging needs for family life were stress and secondary health problems as well as strained relationships and family conflict. Stress was described as resulting from physical demands and intense but not necessarily negative emotional states. Family members experienced stress during caregiving activities, when they dealt with role conflicts or strained relationships, and if they were confronted with standards and expectations which they could not meet. Secondary health problems that have been described were hazards to physical and mental wellbeing, including back pain and tiredness as well as depressive symptoms and suicidal thoughts.

Family communication and pattern of interaction influenced their coping with challenges and adaptation to change and vice versa. Family conflicts and strained relationships, for instance, emerged as part of family situations, among

which ineffective coping with role conflicts, violation of family boundaries and privacy, diverging needs and expectations and unresolved emotional states.

Feelings of inadequacy resulted from situations where families were pulled between two sites like a robe in a tug of war. An example of such a situation is the families attempts to balance between closeness and distance within a family situation of interdependence. To clarify, families who transitioned into adulthood juggled between two relational states that can be viewed as two opposing ends on a continuum: closeness and distance. Family members tried to find a balance, but were continuously torn between one site and the other. They struggled to gauge for the right amount of closeness and distance required by the situation they were in and constantly had to reassess and recalibrate.

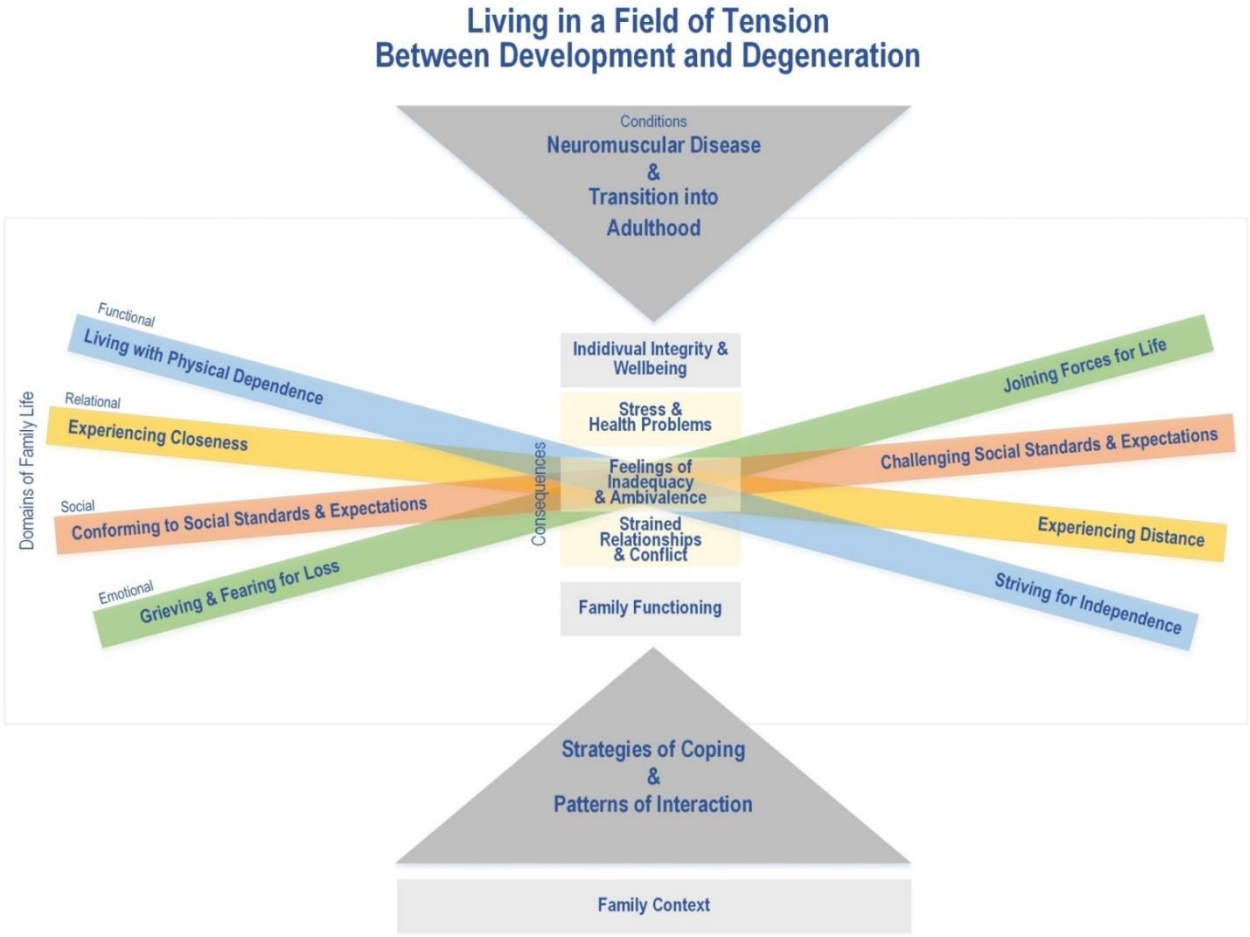
Furthermore, being expected and wanting to participate and integrate, but being prevented from integration or participation due to physical limitation created a sense of deficiency. Family members were excluded from certain activities and experienced limited availability of offers and services. Among the factors for exclusion there were architectural barriers, dependence on care, difficulties in social interactions because of overstrained others and special treatment by social or public institutions. Despite wanting to be part of society, they experienced several difficulties and were at risk of feelings of inadequacy.

Ambivalence is another consequence on family life which comprises the family members' inner conflicts in dealing with conflicting thoughts, feelings and values which result in difficult emotions and behaviour. During the young person's transition into adulthood family members experienced several changes and

challenges that caused feelings of ambivalence. For example, the affected young person's identity formation showed ambivalences. Family members described feeling different and normal at the same time. Feeling different and feeling normal, however, are two opposing and conflicting states that cause an inner conflict and can lead to confusion and difficult behaviour. Another example of ambivalence can be seen in the families' emotional experiences. Family members were exposed to extreme emotional states with intense and opposing feelings; they had to grieve and fear for losses at the same time as they were full of hope and had a strong will for life.

What adds to the complexity of this Grounded Theory is the fact that family experiences vary according to family pattern of interaction and strategies of coping, individual family members' characteristics and the family context. The interpretive theory must therefore be viewed as a theoretical abstraction outlining the shared conditions, fields of experience and consequences for family life (Figure 15).

Figure 15: Interpretive Theory



6.4. Chapter Summary and Outlook

Family members from 12 different families comprising 10 affected young people and 21 family members participated in this study. Four categories emerged from the analysis of the interviews describing experiences in four different fields of life: the functional, emotional, relational and social life domains. Each category is dualistic in nature and includes two diverging experiences that are opposing and at the same time connected and therefore resulting in feelings of inadequacy and ambivalence. Families were living and coping with physical dependence, while striving for independence. They balanced proximity within a

family situation of (inter-) dependence. Families conformed to social standards and expectations as well as challenged some of them and they were grieving and fearing for loss, at the same time as joining their forces for life. Among the consequences for family life resulting from these experiences were stress, secondary health problems, strained relationships and inner conflicts. As a result of recurring challenges, families described the family life during the affected young person's transition into adulthood as a life in a field of tension between development and degeneration. The following chapter discusses this study's findings and their contribution to the body of knowledge, in particular their contribution to our understanding of how families experience life cycle transitions when living with childhood NMD.

7. Chapter 7: Discussion

While the previous chapter described how the study findings have been organised into four categories forming an interpretive theory that describes the family transition experience, in the following sections study findings and processes will be discussed. As part of this, the emergence, significance and fit of the interpretive theory with existing literature is examined with the aim of further illuminating how the understanding of the family transition experience has advanced as an outcome of the study. By contrasting relevant findings with existing literature in the context of the methodological approach, it is possible to evaluate research processes and identify where findings are supported and where they provide new knowledge. These insights are relevant when drawing conclusions and implications for clinical practice and making suggestions for future research.

7.1. Emergence and Significance of the Interpretive Theory

In this study, data analysis resulted in the emergence of four categories that formed the interpretive theory “Living in a Field of Tension Between Development and Degeneration”. Constructivist Grounded Theory methods (Charmaz 2000, Charmaz 2014) allowed the collection of rich descriptions and insights into family members’ experiences. The categories that construct the theory emerged at an advanced stage of theory development and they organise family transition experiences into four life domains (functional, relational, social and emotional). As the four life domains emerged from developing summary memos and diagrams and integrating them with the theoretical framework (see chapter 5, figure 12), also their characteristics and connections with each other became evident. Every life domain has its own focus, but experiences are

intertwined and not analytically separable. The relational field of family life, for example, has the primary focus on family relationships and interactions, but relational aspects are not exclusively linked with this category, but may also play a subordinate role in other categories. This way, the four developed categories that describe family life experiences make sense when they are connected and they naturally form a greater whole, the theory “Living in a Field of Tension Between Development and Degeneration”. Charmaz confirms that an interpretive theory needs to make sense and follow logic (Charmaz 2014). Another shared characteristic of the interpretive theory and of each of the four constituting categories that became evident and confirms their connectedness is the diverging trends that create dualism, a contributing factor to the families’ experiences of recurring tension during the affected young persons’ transition into adulthood. Families reported that they felt torn between two opposites (e.g. closeness and distance in the relational life domain), a situation that added to their stress level.

These insights are significant. The developed interpretive theory fosters a deeper understanding of the family transition experience when living with a young person affected by NMD who transitions into adulthood. It illuminates the repercussion of illness and development on family life and explores how family functioning and wellbeing is influenced by the families’ specific circumstances. As the review of the literature revealed, most past investigations of the family transition experience when living with a chronically ill young person focused on individual family members’ perspectives (Waldboth et al. 2016). This study, in contrast, provides new insights into the effects of NMD on family life from a family perspective. The interpretive theory portrays how families experienced

the transition into adulthood of the affected young person, a time they described as filled with recurring tension and stress. The theory further reveals how family members' related and interacted with each other and how family meaning and context, strategies of coping and patterns of interaction influenced their illness behaviour and development.

By and large, it can be said that the findings of this study give answers to all research questions that were posed. An interpretive theory was developed and major illness and development related challenges and strategies by which families coped and adapted to their life situations in their specific contexts were identified. Therefore, the developed interpretive theory adds to the body of knowledge, but is not only scientifically significant as insights are also relevant for clinical practice and future improvements of health care services. Health care professionals need more knowledge about how they can better support families that struggle with their life situation in coping with NMD and developmental needs that are associated with life cycle transitions. Before drawing conclusions for clinical practice and making suggestions for future research, however, insights from this study will be examined in more detail in context of the wider body of knowledge. Therefore, in the following sections relevant findings will be contrasted with the wider literature to examine where they provide original contributions and where they are in support of or in conflict with existing theory and knowledge.

7.2. Discussion of the Families' Transition Experience

Findings showed that the affected young person's transition into adulthood and the deterioration of health due to NMD affected the families' transition experience. Effects were described for four different life domains. On a functional level, for instance, families coped with the young person's physical dependence by negotiating roles, distributing caregiving tasks and integrating external assistance and assistive technology and tools. Effects on family relationships were difficulties balancing between closeness and distance and the need for freedom and protection, while family members were generally highly involved in each other's lives. Families further tried to conform to the standards and expectations of their social group and to live accordingly, but their life situation challenged some of what was expected. In addition, in the emotional field of experience, families were confronted with intense feelings that resulted from their grief for losses and coping with uncertainty and health crises. While these findings are original to this study, the review of the literature confirmed that families living with a chronically ill young person affected by various chronic and genetic conditions also experienced multiple challenges when the affected individual transitioned into adulthood (Waldboth et al. 2016). In contrast to the evidence from this review, however, this study contributed more in depth insights as it explored the family transition experience when they were living with a young person that was affected by a specific chronic and genetic condition, namely NMD. By doing so and also by applying a family systems approach rather than an individualistic lens to data collection and analysis, new insights into the specific family transition experience and family situation when living with NMD were gathered. The characteristics of the families' transition experience will be further explored in the following sections.

7.2.1. The Families' Particular Vulnerability

Families living with NMD were confronted with a particular vulnerability. While all families are expected to experience transitions and are confronted with unpredictable stressors such as chronic illness during the course of life (Chick & Meleis 1986, McGoldrick et al. 2013b), the family transition experience when living with a young person which is affected by childhood NMD is more extreme. Firstly, families are confronted with a genetic disorder that has very severe implications on family life (Amato & Russell 2008, Miller 1991, Rolland 2006b). The severity and biopsychosocial impact of NMD on the lives of affected individuals and their families can be illustrated by consulting the FSGI-model: NMDs are a group of genetic conditions with a high likelihood of development, high overall clinical severity, onset in childhood and the unavailability of an effective treatment to alter or considerably decelerate disease progression (Rolland 2006b). While these types of genetic disorders often have the most severe implications, conditions with adult onset, lower severity and greater response to treatment are expected to be less disruptive.

Evidence confirms that while young people living with CF, SCD or Haemophilia experienced health problems and were affected by various physical symptoms during their transition into adulthood, individuals living with NMD were the most strongly impaired among them (Waldboth et al. 2016). This study, however, focused on NMD only and does not allow for comparisons across diseases. In addition, the severity of the illness and the degree of the associated physical limitations are not the only factors that need to be considered when trying to explain or anticipate an affected individuals' situation and a families' illness

behaviour. The visibility of a condition, for instance, was described in the review by Waldboth et al. (2016) to have a rather surprising effect on individuals' social lives. Individuals with milder limitations compared to those with more severe handicaps, showed more conspicuous behaviour that potentially led to more problematic social interactions in the long term (Leyendecker & Gebhard 2005, Waldboth et al. 2016). This suggests that while some of the pathophysiological implications on families are clearly linked with the severity of the condition, there are aspects that cannot be explained by only considering illness factors. The family transition experience is more complex and besides the severity of the condition other contributions such as the family life cycle need to be considered.

The affected young person's transition into adulthood when living with NMD was described as a particularly challenging time for families due to the simultaneous progression of the illness and need for development. Miller (1991) shares that families living with NMD experience specific moments of crises and move through stages before they come to terms with their life situation. From clinical experience she concludes that there are four time periods that are the most challenging for families: diagnosis, cessation of ambulation, adolescence and the late stages of the disease (Miller 1991). These stages are all periods of change that are influenced by illness and human development. While during diagnosis and cessation of ambulance the impact of the illness is clearly in the foreground, findings from this study reveal that during the affected young person's transition into adulthood the physical degeneration due to chronic illness coincides with the affected young person's developmental needs of becoming an adult person. This situation manifests in a particular dynamic and results in a life stage where families are confronted with multiple challenges and

diverse needs. The life of affected families is not only characterised by the effort to satisfy developmental needs and to cope with a severe and progressive illness, but there is also recurring tension, ambivalence and feelings of inadequacy resulting from extreme situations and diverging needs in each domain of family life. In other words, during adolescence families face multiple effects of NMD that further intensify as they are confronted with two opposing processes and feel torn between extremely diverging and potentially conflicting trends (dependence and independence, closeness and distance, conformity and non-conformity, grief for loss and hope and will for life).

Not an intensification of family experiences due to diverging needs, but a similar duality of social life was described by family systems theorists and relational dialectics. Carter and McGoldrick (2005) and McGoldrick and Carter (2013b) described a duality of expected family development over the course of life where, generally speaking, families are expected to find balance between connectedness and separation, belonging and individuation and accommodation and autonomy. Baxter and colleagues, on the contrary, based their research on the premises of Mikhail Bakhtin's (1895–1975) work on relational dialectics where social life is viewed as the result of "a contradiction-ridden, tension-filled unity of two embattled tendencies" (Bakhtin 1981, p. 272), and interplay between centripetal and centrifugal forces (Baxter 2004). They argue that relationships are knots of contradictory interplays (e.g. between integration-separation, certainty-uncertainty, and expression-non expression) that need to be examined in their concrete and situated particularities to understand how these opposites can "complete, enhance, and enable" at the same time as they "limit or constrain" individuals (Baxter 2004, p. 8). In this

study each of the four categories revealed such interplays where families living with NMD, for instance, described how they felt torn between two extremes, dependence and independence or how they coped with their situation by talking and not talking about their situation throughout the young person's transition into adulthood. While these findings are in support of relational dialectics (Baxter 2004, Bakhtin 1981) the question of whether it is the aim to find equilibrium or balance contradictory interplays (family systems assumption), or to enter into discourse and assume constant flux (relational dialectics assumption) remains unanswered. What this study offers is an explanation of how families experienced and coped with these diverging needs and how they related and interacted with each other within their specific contexts which shaped the quality of their transition experience.

7.2.2. Extra Challenges, Coping and Adaptation

During the affected young person's transition into adulthood families faced various illness and development related changes and challenges that added to their vulnerability and they needed to master and adjust to these new situations to ensure family functioning and each family member's wellbeing. Evidence suggests that all families develop a patterned response to change that characterises family functioning when they are confronted with chronic illness, but depending on the type of condition and on the specific family situation, there may be extra challenges (Kaakinen et al. 2010, Patterson 1991). As findings from this study illuminated the transition experience of families living with NMD, the complexity of their family situation and the extra challenges they were confronted with will be further discussed in the following sections.

7.2.2.1. Being a Family Caregiver: Stress and Role Conflicts

Findings suggest that an extra challenge of these families resulted from role conflicts and related caregiver stress. Being a family member of a young person who is affected by NMD also meant being a family caregiver. Family members experienced stress as a consequence of these dual roles. They described how they negotiated their role of a caregiver with their role as a family member or partner. As mothers were the primary caregivers in most families, they in particular struggled with being reduced to caregivers and being responsible for most caregiving activities, especially if there was little support from other family members or external parties. The availability of support was further influenced by family attitudes and their ideas of what accepted behaviour is. For instance, findings indicate that some families wanted as little external help as possible and hesitated to accept external support or involve help from members of the extended family in care as this was not considered appropriate.

Families described different situations related to their caregiving experience that were particularly stressful. For example, it was stressful and frustrating for them to care for the affected young person and to be available for assistance around the clock, with little time for respite. Affected young persons needed assistance with every daily activity, and most families developed routines of care in order to function, by distributing caregiving tasks and involving external help. They acknowledged the high demands that were associated with the young persons' physical disability and care, among which caregiving activities that were related with an increased risk for developing secondary physical problems (e.g. back pain and tiredness), a need to constantly plan and organise everyday life and

care and a lack of time for themselves and for activities with others including other family members.

Surprisingly, despite these demands and the efforts required for the management of everyday life, families did not depict caring for the affected young person as a burden. They rather described the negative and positive implications of NMD on their lives. Existing evidence suggests that chronic illness can have both, negative and positive effects (Ayres 2000, Patterson 1991). It increases the stress level and family members' vulnerability, which can make them more disorganised or even dysfunctional. Some families, however, become stronger and more resilient with time if confronted with chronic illness and families would benefit from appropriate interventions that focus on family resilience aiming to increase such positive effects. In light of the above, it is crucial not to decontextualise the family caregiving experience and reduce it to negative outcomes often conceptualised as caregiver stress or burden, but to relate the caregiver experience with the meaning families attribute to particular tasks or events (Ayres 2000). In this sense, caregiving stress lies in the eye of the viewer where negative outcomes or responses can be diminished by positive meaning making. Health professionals can have a positive role in focusing on the specific family situation and support families' in their positive meaning making, which will facilitate a greater resilience and sense of coping.

Parents of more than one child also described difficulties in having time for the rest of the family and concerns about the effects on their healthy children's lives. They further described how they retreated from many activities in order to function, including putting their personal aims aside, and withdrawing from

careers and social activities. These experiences can be described as role overload or role conflict. Role overload is defined as the inability to meet demands that are associated with a role and role conflicts may appear when expectations about different roles are incompatible (Kaakinen et al. 2010). As a consequence of role overload or conflict, two related concepts, individuals may withdraw from some activities which seem to be insignificant at present, but would have positive effects in the longer term. When family members are confronted with difficulties that result from role overload or role conflict, but lack resources and strategies how to confront those, professionals can offer support (Kaakinen et al. 2010).

7.2.2.2. Sexual Maturation and Family Boundaries

Another influencing factor on the families' caregiving experience that resulted in extra challenges was the affected young person's sexual maturation. As part of their transition into adulthood, young people living with NMD underwent physical changes including growth and the development of sexual characteristics. They experienced a need for more privacy and distance from their families and wanted to have an active social life, comprising romantic relationships and contacts with people outside of their family. These changes and needs are typical for the maturation of all young people and characterising of the formation of a sexual identity (Christie & Viner 2005, Santrock 2013). Due to the nature of NMD, however, some of these needs were more difficult to satisfy, which had specific implications on family life. For example, while affected young people became more dependent, also their need for more privacy increased, which made caregiving more complex. To clarify, during the affected young person's transition into adulthood their need for assistance increased, but young people

and their family caregivers felt more embarrassed when receiving or performing care that exposed or involved intimate body parts and they expressed the need for protection of private spheres. As this was not possible due to the young person's need for assistance, adaptions were necessary and cultural expectations of who is allowed to give care became drivers for organising care. The sex of the family caregiver, for instance, was a reason for such decisions. If the mother or sister gave care to the affected female and the brother or father assisted the affected male it was better accepted, whereas if family caregivers of the other sex performed intimate caregiving tasks this was described as stressful.

At the same time as affected young people matured sexually, they had the desire to spend time with their friends and peers, but experienced barriers to social participation such as limitations to making contacts and finding a romantic partner. They were therefore not able to participate in some of the experiences that would have assisted their identity formation and social integration. Evidence from the review of the literature indicates that because of their physical limitations chronically ill young people were unable to participate in some more physical activities with their peers and young people with NMD disclosed that for them it was difficult to find a partner and have a love life (Dreyer et al. 2010, Gibson et al. 2014, Gibson et al. 2007, Waldboth et al. 2016). Findings of this Grounded Theory study further illuminated that family members put efforts in supporting their affected child's social integration, but meeting some of their children's needs was beyond their sphere of influence or violated family boundaries and expectations. For instance, some parents adopted practices that aimed at allowing their child to have a fulfilled social and

sex life. Family involvement in private matters such as going out together and assistance with sexual practices, however, did not feel natural to families as it violated family boundaries and social expectations. Family boundaries are crucial for family functioning and for the preservation of the integrity of individual family members (Luhmann 2006). Family boundaries help to balance energies and relationships and they regulate the internal and external flow of information by the use of rules (Broderick 1993). Therefore, the violation of family boundaries threatens family functioning and wellbeing and possible violations and their consequences need to be assessed and treated with caution.

Furthermore, findings suggest that it did not feel natural to families to trespass boundaries and that it required efforts to develop a more flexible attitude towards them and towards what was expected and what was meaningful. These efforts comprised negotiation of needs, talking about taboo subjects (e.g. sexuality) and being sensitive to how family involvement in private matters affected family relationships and individual family member's integrity in the longer term. Families would benefit from professional support, but according to the findings of this study, sexuality is a taboo subject during contacts with health professionals, too. Health professionals might have a lack of information about the effects of NMD on the affected person's sexual development or they neglect or are not comfortable with talking about it. In addition, the situation can be explained by the fact that affected young people might not be viewed as sexual beings (World Health Organization 2007). Notwithstanding the above, the findings of this study revealed that affected young people are interested in romantic relationships and that some of them have an active and regular sex life with their partner or with professional sex workers. Adolescents with chronic

illness are in fact not less likely than their healthy peers to be sexually active and have been described as having an even higher rate of sexual intercourse (World Health Organization 2007). Considering the fact that they are less likely to benefit from the sexual education and the guidance their peers get, as well as their higher risk for sexual abuse due to their physical dependence and the reported family involvement in private matters, it is very important for professionals to reflect on the meaning of sexuality and to be prepared to offer appropriate support to parents and affected young people.

7.2.2.3. Balancing Proximity and Launching Children

Most families living with a young person with NMD described having a strong emotional bond and being highly involved in each other's lives. Strong and supportive family relationships and family involvement were equally reported by families living with other chronic and genetic conditions (Admi 1996, Atkin & Ahmad 2001, Erskine 2012, Waldboth et al. 2016). These parents explained how highly they were involved by illuminating the tasks they had to perform including normal parenting activities as well as assisting with disease management and care. During the affected young persons' transition into adulthood, however, young people with NMD and individuals living with other chronic conditions wished for more distance from their families and freedom to develop as a person. Parents were aware of these diverging needs, but struggled with caring and protecting their children and at the same time allowing for their freedom of development (Waldboth et al. 2016). This study revealed how they struggled to balance proximity and experienced a tension between the need to hold on and let their children go. Wider literature illuminates what adds to the situation: parents and siblings of chronically ill young people reported

feeling distressed and being constantly aware of the progressive nature of the disease, as well as having concerns about the affected young person's ability to live more independently (Antle et al. 2008, Cappelli et al. 1989, Dupuis et al. 2011, Gjengedal et al. 2003, Moola & Norman 2011, Parkyn & Coveney 2013, Porter et al. 2014, Telfair et al. 1994, van Staa et al. 2011, Waldboth et al. 2016). These factors resulted in family member's protective behaviour towards their chronically ill relatives and a simultaneous increase in difficulty to step back and let go.

The transition into adulthood and the launching of children, however, is a stage of the family life cycle where family relationships change and parents are expected to help their adolescents to move on in life, irrespective of the presence of a chronic illness (Branje et al. 2013, McGoldrick et al. 2013b). During the course of adolescence, families need to increase flexibility of family boundaries in order to function and accept their children's increasing exits and entries into the systems, despite concerns and associated emotional difficulties. Balancing flexibility and family cohesion are processes that aim for family functioning in the longer term by preventing extremes; inflexibility or being overly flexible and being disconnected or overly connected, respectively (Carter & McGoldrick 2005, McGoldrick et al. 2013b, Olson & Gorall 2003). Evidence suggests that depending on how families communicate with each other, they can balance between their needs more effectively and overcome eventual difficulties which influence the whole family's transition experience. By effectively talking to each other, families can find balance through adapting their flexibility style and their degree of cohesion (Olson & Gorall 2003). Furthermore, strained family relationships can benefit from more open parenting styles and

effective communication, changes in pattern of interaction that young people living with chronic illness wished for (Atkin & Ahmad 2001, Bregnballe et al. 2011, Erskine 2012, Waldboth et al. 2016).

The transition experience of families living with NMD can also be considered as an example of the centripetal tendency of chronic illness, during a centrifugal stage of the family course of life (Rolland 1987, Whitchurch & Constantine 1993). Affected young people are born with NMD and develop symptoms and are diagnosed during childhood, but the progression of the disease and the increase in dependence on support has a peak during adolescence, a time when families are expected to be more flexible and experience centrifugal forces. Most families that are confronted with chronic illness, however, develop a centripetal tendency, which means that they move or tend to move closer together, while adolescence has the opposite effect. According to Rolland (1987), the autonomy and individuation of each family member is at risk if they are in a life situation which has both centripetal and centrifugal effects at the same time. Different effects are described among which limited autonomy of single family members. Findings of this study illuminate situations where individual family members' neglected their own needs due to high involvement in each other's lives. For example, parents and siblings described having less time for themselves and their own life goals as they needed to assist the affected person. Furthermore, affected young people were constantly supervised by a family caregiver or external party and had little time alone.

In light of the above, the pressures that families are confronted with when balancing proximity become clear, but it needs to be considered that launching

children is not true for all families. Physical and mental disabilities, culture, gender, religious beliefs and other factors influence what young people actually want and how parents support and guide them through each developmental phase (Garcia-Preto & Blacker 2013). In the case of NMD, this implies that families may prefer that their affected adolescents continue to live at home, instead of supporting them moving out and grow more independent. Reasons for this behaviour may be that young people are insecure and afraid of leaving home or that parents want to ensure that the child's care needs are met as best as possible, namely by themselves (Garcia-Preto & Blacker 2013). Parents are experts in the care of their children and may want to continue to care for them, even if other options are available. They may also depend on the support of healthy siblings and their contribution to caregiving and household activities, which shapes how they interact and guide them, too (Garcia-Preto & Blacker 2013).

Parental overprotection or excessive protective behaviour by parents is common in families where a family member is affected by chronic illness (Hullmann et al. 2010, Patterson 1991, Waldboth et al. 2016). While it is described as a behaviour that is beneficial for acute crises, protecting a chronically ill young person from being more independent by sheltering him or her from experiences during their transition into adulthood affects the person's development and psychosocial adjustment. A particularly difficult situation for chronically ill adolescents is a mother's overprotectiveness, an often observed behaviour, combined with the father being under-involved (Patterson 1991). Then, parents may send different signals, when they actually should work together to set clear rules and limits that are applied with consistency and

directed at the affected young person's achievement of more independence. In these cases, family interventions can focus on parents to help them work together in setting clear limits and expectations (Patterson 1991) and to help family members understand each other's perspectives and reaching agreements on how to move forward.

7.2.2.4. Emotional Wellbeing and Family Communication

While a review of the literature was not able to give deep insights into how illness and development affected the emotional life of chronically ill young people and their families (Waldboth et al. 2016), this study resulted in more profound information about the family's inner life when living with NMD. During the young persons' transition into adulthood, families faced threats to their emotional wellbeing. Among the greatest hardships were grieving about losses such as the loss of physical abilities and a future, uncertainty and associated feelings of sadness and fear. Rolland (2006b) distinguishes between anticipatory grief and anticipatory loss, and defines the former as more narrowly related to the terminal phase of an illness. According to this definition, what families living with NMD experienced during transition into adulthood was anticipatory loss, as they were confronted with *"possible, probable, or inevitable future loss"* by the death of the affected child at a young age (Rolland 2006b, p. 140). The findings of this study revealed that the families' transition experience was shaped by anticipatory loss and the family members' grief and fear of the future which posed the risk for mental problems such as depression and suicidal thoughts.

These intense emotional states were influenced by the affected young person's stage of development. The development of cognitive abilities affected the families' illness experience as it allowed adolescents to fully understand their life situation for the first time. Adolescence is considered as the time when young people develop new skills including abstract and logical thinking, when they develop their views of the world and opinions that shape their personal identity (Christie & Viner 2005, Santrock 2013). While during adolescence affected young people and healthy siblings experienced these developmental changes, parents coped with and adapted to their children's changing needs in order to find balance and function as a family. At the same time, all family members were grieving for loss and trying to create meaning of the illness experience and accept their situation. These experiences resemble the first clinical onset-phase of chronic illness, which was described by Rolland (1987). Affected adolescents and their siblings experienced these "clinical-onset" effects of NMD during their transition into adulthood, much later than at the actual time of clinical onset of the condition, which in most of the cases can be dated back to the time of diagnosis in early childhood. This implies that in the case of chronic childhood illness, some of the expected psychosocial implications of genetic illness are delayed due to the children's stage of development. From this can be concluded that it is not only the illness itself that is the cause for the family members' emotional wellbeing, but the psychosocial consequences of their life situation that has impact and is often intensified at a key transition point.

Coming to terms with the idea of being affected by a chronic and life limiting illness and coping with anticipatory loss or death of a child at an early age was

described by the findings of this study as an ongoing and very difficult process. It was particularly difficult for adolescents as they were confronted with this truth at a time when they realised that their physical limitation was permanent and possibilities to find expression of their personality and the pursuit of life goals were constrained. This particular situation had effects on the whole family as young people experienced negative emotions and showed difficult behaviour. Findings of this Grounded Theory revealed that families were confronted with helplessness in dealing with their affected child's feelings and behaviour and their own emotional states.

Families described having difficulties in communicating with each other about their inner life, as talking about the disease and its effects was described as emotionally too overwhelming and was therefore avoided. At the same time, however, they identified information and communication about the disease as facilitating their coming to terms with NMD. Being informed and being able to communicate about feelings has been described to be in support of family coping (Metcalfe et al. 2008). By communicating with each other, families can express how they are feeling and develop a shared meaning which allows them to better understand each other's perspective. Through open family communication, family conflicts can be resolved and coping and coming to terms with their life situation can be facilitated (McGoldrick & Walsh 2013, Metcalfe et al. 2008, Metcalfe et al. 2011).

Families make meaning of situations by talking to each other and dialoguing their experiences (Blumer 1969, Patterson & Garwick 1994). If they have difficulty communicating with each other, health professionals can offer support

and empower families to communicate more effectively. Enhanced family communication is not only valuable for a successful management of anticipatory loss or transition into adulthood, but it also prepares them for the inevitable loss they are facing in the future and allows them to develop coping strategies that are helpful when they are confronted with the eventual death of a family member (McGoldrick & Walsh 2013). Interventions that aim to improve family communication are particularly valuable for families living with NMD also because grief was identified as a factor that inhibits open family communication (Metcalfe et al. 2008). Therefore, there is a need to enhance health professionals' awareness about which family members are at risk, to give them access to appropriate interventions and facilitate family communication.

According to the findings of this study sooner or later most families had contacts with professionals who functioned as a source of support for dealing with emotional difficulties. For example, professionals such as physiotherapists or psychologists gave access to information and functioned as conversation partners for affected young people, their parents and siblings. However, findings also revealed that some family members did not get the support they needed and as a consequence their emotional wellbeing was threatened. Evidence further confirms that affected young people and their families are at risk for socio-emotional difficulties and developing mental illness such as depression (British Medical Association 2003, Polakoff et al. 1998, Waldboth et al. 2016).

Finiteness of life and being confronted with the death of a young family member was described as major emotional difficulty by families, a fact that needs to be considered by health services. McGoldrick and Walsh (2013) described four

tasks for dealing with death and loss that are relevant: 1) facilitation of direct contact and clear and open family communication as part of a shared acknowledgement of death and loss; 2) a shared experience of loss comprising family rituals (e.g. funeral rituals and visits to memorial sites) and meaning making in order to reach acceptance and a sense of continuity, 3) reorganisation or family relationships and distribution of roles and functions by promotion of adaptive cohesion and family flexibility and 4) revisiting of other relationships as well as refocus on new life plans and goals (McGoldrick & Walsh 2013).

7.2.2.5. The Social Environment: Pressures and Expectations

Other extra challenges for families living with NMD were associated with the pressures and difficulties to conform to social standards and expectations that originated from the family's larger social environment. During the affected young person's transition into adulthood, family members were confronted with situations, where their expectations conflicted with expectations of their social group and where social expectations put them under external pressure. Independent of the families' situation and their resources and specific contexts, deviation from an expected path can result in difficulties and confrontation (Garcia-Preto & Blacker 2013), which in the case of families living with NMD resulted in stress, inner conflicts and interpersonal tension. Existing evidence supports the insight that families that are living with chronic illness experience difficulties meeting expectations that are dictated from their own culture and society (Carter & McGoldrick 2005, Rolland 2012). It is further assumed that a rigid application of social standards and expectations as well as an overemphasis on attempts to being normal (conformity) and being different

(non-conformity) causes difficulties, such as an increased risk for inner conflicts and threats to mental wellbeing. In order to lessen negative impacts, a more flexible application of expectations and a broader view of the human life cycle are advised (Carter & McGoldrick 2005).

Every family brings along their cultural and social expectations that contribute to how families approach life's tasks such as a child's care and transition into adulthood (Garcia-Preto & Blacker 2013). For example, even if most family members would agree that the expectation is to launch a late adolescent family member, families may have different beliefs and values that influence what launching their children means and how this is done. Families with specific cultural backgrounds such as families that have migrated to Switzerland from other countries, for instance, may expect their children to become less independent or are more strongly connected than locals because of cultural reasons and regardless of their health situation. The specific family context therefore needs to be assessed.

A social expectation that created pressure was the family members' feeling of being responsible for each other. Some parents and siblings felt responsible for the care of the affected young person and they experienced ambivalence and feelings of inadequacy if they were not able to meet this expectation. White & O'Brien (2010) described that exchanges of help and support are family phenomena that are motivated by affection, as well as by a sense of obligation. Caring for a family member is therefore associated with the experience of having both positive and negative feelings towards one another which can be subsumed as ambivalence. While ambivalence is a normal part of family life,

the more ambivalence there is the more a person is deviating from the norm and therefore confronted with non-conformity which implies difficulty (White & O'Brien 2010). Findings from the review of the literature illuminated how feelings of ambivalence were part of the transition experience of chronically ill individuals during their identity formation, where mixed feelings resulted in confusion (Waldboth et al. 2016). Their confusion was linked with feeling average and extraordinary at the same time (Admi 1996, Berge et al. 2007, Christian & D'Auria 1997, Dupuis et al. 2011, Gibson et al. 2007, Gibson et al. 2014, Müller-Kägi et al. 2014). Young people felt average, because they wanted to live a normal life and extraordinary as their lives were different and life's tasks required additional efforts compared to others (Waldboth et al. 2016).

Some of these additional efforts were related to school and working life. Adolescence was a time when young people made their choice of career, applied for internships and jobs, and worked for the first time. Mastering these tasks was associated with their strong wishes to live an as "normal" life as possible. Evidence from wider literature on chronic illness confirms that chronically ill young people perceived it as important to have an education and career, but they described various difficulties in attending schools and getting a traditional employment including their need for assistance in the school and workplace (Badlan 2006, Gibson et al. 2007, Gibson et al. 2014, Waldboth et al. 2016). Attending a regular school and finding an internship or job was not without difficulties also for young people living with NMD. They reported practical as well as social barriers. They, for instance, had to overcome architectural barriers such as inaccessible schools and class rooms and had to find supportive employees such as teachers or employers that were willing to

support them. Participating in school and working life therefore required many additional efforts by affected young people and their family members compared to their healthy peers.

Besides education and career, families were also confronted with additional efforts related to the Swiss social and health care systems. Findings described that during their transition into adulthood, some young people were moving out from their homes, changed their support model into institutional care or assisted living, and took over more responsibility for their own care, which comprised increasing contacts with health professionals. Taking over more responsibility for their own care, however, was not described as a straightforward process by all affected young people. Some reported being used to the assistance of their family and therefore they were afraid of becoming more independent even if that was what they desired. Parents and siblings attitudes also played an important role in this dynamic. Depending on how much effort they put in supporting the affected young person's independence or, on the contrary, in protecting them and inhibiting their development, they influenced the affected individuals choices and possibilities to make their own experiences. Findings from the literature review revealed that parents and siblings experienced doubts and had concerns that their chronically ill family member would be able to care for themselves in future (Porter et al. 2014, Telfair et al. 1994, van Staa et al. 2011, Waldboth et al. 2016), a factor that gives an explanation for family members' protective behaviour.

Moving out from home and living independently was associated with additional efforts as it meant loss of care by the family and the need to adapt to a new

environment. In turn, however, physical distance positively influenced the young person's independence and improved previously constrained family relationships as it released families from some of their caregiving tasks. Separation and individuation are transformation processes inherent to the family system when a young person transitions into adulthood: processes which allow for the young person to become more autonomous and independent (Garcia-Preto 2013). In the case of chronic illness as well as in unaffected families, this transformation comprises continuous renegotiation of relationships, as even though adolescents want to become more independent from their families, there is always a part of them that needs or wants to be nurtured and cared for by the parent.

The effects of physical distance on family relationships may be explained by the fact that physical distance affects family cohesion. Family cohesion is the families' degree of emotional bonding and individual autonomy (Olson & Gorall 2003). Disconnected and overly strong connected families have an unbalanced family functioning style. Physical closeness because of the need to give care and protect the affected child may cause strong bonds, but also result in overly close connections or enmeshment, which are related with negative consequences. While findings suggest that physical distance has a potential to positively influence family interactions and individual family members' development in the long term, there are many factors that have an influencing factor on the consequences that physical distance has for the individuals' and families' wellbeing. Race, class and ethnicity, for instance, influence family members' expectations and consequently shape their connections (Garcia-Preto 2013). In some ethnic groups promotion of early separation of

adolescents is the case, while others prolong adolescence and keep close boundaries around the family including a mutual obligation for caretaking. Therefore, it is advised that health professionals carefully assess the individual family situation, outweigh benefits and negative consequences of physical separation on each family member involved and support families in offering validation and the connection the adolescent needs to make a safe transition into adulthood (Garcia-Preto 2013).

Another expectation that families and affected individuals were confronted with during the young person's transition into adulthood was the transfer from paediatric to adult services that was scheduled for this developmental stage as required by the Swiss law (Schweizerische Akademie der Medizinischen Wissenschaften 2013). While the review of the literature described the transfer experience from paediatric to adult care services of chronically ill young people as a difficult process involving mixed feelings and experiences (Porter et al. 2014, Telfair et al. 1994, Telfair et al. 2004), this study resulted in little insights into the inner institutional transfer experience of families. This can be explained by the fact that besides the many other challenges and stressors they experienced during adolescence, the transfer was simply not a major concern or not a process that families experienced very consciously. Moreover, families may not view the service transition as a major concern because the quality of the transfer experience is adequate. What also needs to be considered is that this study's focus was very comprehensive, which adds value to a family systems theoretical lens compared to an individual approach in contextualising human experiences.

7.2.2.6. Concluding Remarks on the Discussion

Findings from this study give new insights into the specific vulnerability and complexity of the transition experience of families living with a young person affected by NMD by describing how they were confronted with diverging needs and extra challenges. The fact that these findings add to the body of knowledge was supported by a review of the literature from Waldboth et al. (2016), which investigated the transition experience of families living with a chronically ill young family member and resulted in little insights into the family transition experience. These findings were related to the fact that the studies included in the review focused on the individual family members' perspectives (Waldboth et al. 2016), while this Grounded Theory study had a broader family focus. This family focus comprised integration of the affected young persons', the siblings, parents and other next of kin's perspectives on the basis that the family is a whole. This way, it was possible to illuminate how families functioned and how they tried to ensure each other's wellbeing in context of NMD, and how family relationships, communication and strategies of coping shaped their transition experience. By and large, it can therefore be concluded that these new insights into the family transition experience fit and expand upon current family systems theory. Before drawing conclusions for clinical practice and making suggestions for future research, however, study strengths and weaknesses need to be examined to evaluate the quality of research processes that were involved in this study and to ensure the credibility of the study findings.

7.3. Discussion of Study Strengths and Limitations

To systematically evaluate the quality of this study's findings and related research processes quality criteria for constructivist Grounded Theory were

used (Charmaz 2014). These quality criteria comprised credibility, resonance, originality and usefulness of the study and its findings.

7.3.1. Credibility

The credibility of this study's results is supported by the quality of the data that have been collected, by the contextual sensitivity that was attained and by a transparent and systematic description of methodological procedures (Charmaz 2014, Silverman 2011). Data from 31 interviews with an average length of 68 minutes comprised rich descriptions of family members' experiences. The recruitment involved a well-balanced mix of female and male participants, as well as an adequate quantity of participating parents, siblings and young people living with NMD. However, no members of the extended family and only one partner contributed, which can be explained by the fact that most affected young people were single and that the families' considerations of who is part of the family and able to contribute meaningful insights restricted extended family members' participation.

The generation of rich data was favoured by use of one-to-one interviews instead of family group interviews, which allowed participants to contribute their personal experiences, without having to share them with the whole family, which may have inhibited what individuals might share. In addition, my previous experience and training in intense interviewing techniques and in Grounded Theory Methods that I gathered during my education, my work as a research associate and during a summer school with Kathy Charmaz in 2013 facilitated data gathering through enhanced techniques. Through critical reflections and

the supervision of my interview technique by means of reflective field notes and through discussions with my supervisors, my techniques continuously improved.

Furthermore, my professional background and my personal involvement facilitated interactions with participants. Being a nurse and a family member of a relative living with NMD allowed for a shared language and understanding, as well as for a timely establishment of a trustful relationship. However, it needs to be acknowledged that personal experiences influence social interactions and that what the researcher brings to the table needs to be reflected and considered as potentially influencing the outcome of an interview (Charmaz 2014, Creswell 2013). In addition, during an interview a co-construction of knowledge takes place, where there is always a certain degree of subjectivity that modifies the outcome, even if the focus is on gathering data on the family members' experiences and letting them tell their stories. When two individuals engage in social interactions a construction of a shared understanding and development of a shared meaning takes place (Blumer 1969) even if the researcher uses intensive interviewing techniques and adopts the passive role of the listener. Therefore, the researcher tried to actively listen, observe, and communicate openly and non-judgmentally and to be sensitive to underlying nuances and reflexive about preconceptions.

Family members described feeling understood and being enabled to openly share their experiences. My personal experience also made me sensitive to the families' context, which was valuable during data analysis and interpretation. To better understand more unknown territory, I engaged in activities such as a work shadowing in a long term care institution and at a school for children with

special needs. I also participated in meetings and discussions with professionals from acute care settings, including nurses from an intensive care unit and I met with affected families at different informal occasions such as at presentations. During these encounters I took field notes about my insights, and I also noted my observations before and after every interview.

The interpretive theory that emerged comprises four categories and gives detailed insights into four different fields of family experience: the functional, relational, social and emotional fields. This theory developed inductively and through thorough and constant comparison of interview data so that insights are grounded in data and can be supported by family members' quotes. The researcher acknowledges the fact that prior knowledge about a topic gained by personal experiences, consultation of the literature or related to insights gained from wider theoretical or philosophical evidence may influence theory construction (Charmaz 2014, Corbin & Strauss 2015). However, during the analysis the researcher engaged in inductive practices and tried to be open to insights from the data, while critically reflecting any potential influences of prior knowledge.

As previously introduced, conducting a literature review is not always regarded as best practice by some Grounded Theorists (Corbin & Strauss 2015), but as the families' transition experience when living with NMD is a very difficult and emotional topic it would have been unfair to repeat a study unnecessarily that might have previously been done (Creswell 2013, Silverman 2011). Therefore, a review of the literature was indicated to identify what evidence is available, but when collecting and analysing the data I tried to uncover hidden assumptions by

critical reflection and to put any preconceptions related to my prior knowledge or my views and experiences as a nurse or as a family member to one side.

As data were collected in Switzerland in the participants' mother tongue, the quotes have been translated into English by a bilingual academic and are presented in both languages as part of the findings chapter to ensure trustworthiness. However, exact translations are impossible, also because languages are different systems of signs that are historically and culturally shaped and where there are different words, meanings and definitions to the same terms (Berger & Luckmann 1991, Burr 2015), which needs to be acknowledged as a methodological limitation of cross-language research. Despite these limitations, this study adopted methods to monitor and control for these potential weaknesses and covered a wide range of empirical observations, so that there is sufficient evidence to support the claims that have been made in the emerging interpretive theory.

7.3.2. Resonance

In order to evaluate the fullness of the portrayed experiences which is referred to as resonance (Charmaz 2014), feedbacks from clinicians, researchers and affected families have been gathered. According to professionals, the developed model is understandable and represents the findings of this study in a concise way. The theory as a whole makes sense and gives a complete picture of the families' transition experience. Preliminary results were presented to family members as part of an oral presentation. Following this presentation, family members' validated the results by comparing them with their own experiences. Their feedback yielded some further insights into the relevance of

some topics over others for certain families. The findings generated by this study are considered situational and cultural and historically relative and therefore no confirmation of the findings was pursued by member checking (Silverman 2011). None the less, the estimates of family members and professionals described above can be viewed in favour of the quality of this Grounded Theory.

7.3.3. Originality

The findings of this study give new insights and allow a better understanding of the family experience when transitioning into adulthood with NMD, an area of interest that has not yet been extensively researched. During the young person's transition into adulthood, families were exposed to multiple changes and potential challenges in their functional, emotional, relational and social domains of life. Inner conflicts such as feelings of inadequacy and ambivalence resulted from diverging needs in each domain of life, as families were living a life between two opposing life processes, development and degeneration. Depending on their family context and resources and how families related and interacted with each other shaped how they adapted and coped with illness and development-related tasks. In case of ineffective coping they were confronted with negative effects on family functioning and family members' integrity and wellbeing. These findings are original to this study and have not been described previously.

7.3.4. Usefulness

In addition to the scientific significance, the generated knowledge is clinically important. Families' needs as well as their challenges and strategies of coping

were studied. This knowledge and understanding of the illness experience of families living with NMD during transition into adulthood may inform and help future health professionals to better assist affected families to anticipate and react to difficult and stressful situations. There is great potential for health professional to support families in coping and adapting to developmental and illness-related difficulties. In order to do so, they need to be aware of the complexity of the family situation and perform interventions that are targeted at the wellbeing of all family members and at family functioning, including psycho-educative and relationship-oriented interventions aiming to improve family relationships and pattern of interaction.

In addition, health care institutions and organisations have already shown interest in this study's findings, which will be disseminated for professionals and lay people. The investigation of the illness experience of living with a rare condition and the dissemination of the studies' results has the potential to increase public awareness and as a consequence acceptance of NMD by society. The family experience, in turn, might be positively influenced by a better informed society and more openness towards chronic illness and diversity.

8. Chapter 8: Conclusions

The developed interpretive theory depicts the families' transition experience as a time of recurring tensions between development and degeneration. This recurring tension resulted from the effects of NMD on family life and from the concurrent developmental needs families had to satisfy. Depending on the families' resources and context and on the ways how they adapted to change and coped with the multiple challenges and diverging needs they were confronted with, their transition experience differed. Effective coping and adaptation was associated with mastery of their life situation and positive outcomes, but most families experienced hazards to their wellbeing and functioning during the course of the young person's transition into adulthood. Stress related to caregiving, the risk of developing secondary physical and emotional problems as a consequence of stress and intense emotions, role conflicts and strained relationships as well as inner conflicts were among the factors that threatened family life.

These families would benefit from a family centred approach to care, including a family centred approach to researching their needs. Professional support needs to focus on the assessment of the needs and expectations of all family members in order to plan family interventions that assist them to more effectively cope and adapt. They need to offer interventions that support how families communicate with each other, how they manage role conflicts and role overload and how they support each other while ensuring individual family members' development and integrity as well as functioning and wellbeing of the whole family system. A detailed description of implications for clinical practice follows in the next section.

8.1. Implications for Clinical Practice

The interpretive theory that emerged from this study portrays the high level of vulnerability that family members are exposed to. During the affected young persons' transition into adulthood, all family members were confronted with changes, challenges and diverging needs related to the young persons' illness and developmental experience. Effects on family life were described for the whole family group on a functional, relational, social and emotional level, which had the potential to threaten family functioning, and individual family members' wellbeing and integrity. Appropriate interventions can reduce their vulnerability and ensure family functioning and each family member's wellbeing and integrity during the affected young person's transition into adulthood.

8.1.1. A Family Perspective to Care

Professionals can offer interventions that support families to better cope with challenges and adapt to change, by viewing them as a group and not focusing only on the chronically ill individual as if their experiences were not connected. A reductionist individualistic approach does not account for the families' transition experience and family members' illness behaviour, which manifests and has meaning only in the context of the family group and its larger socio-historical and cultural situation (Patterson 1991, Rolland 1987, Rolland & Williams 2005). Therefore, the focus of clinical interventions needs to shift from viewing the chronically ill individual in isolation, a paradigm inherent to the biomedical model which is today still prominent in many adult care services, to a family systems perspective. A family systems approach to care is useful, as it takes into consideration how family members relate, interact and mutually influence each other and which family qualities account for their illness

behaviour and their dealing with life cycle transitions (Feetham & Thomson 2006, Patterson 1991, Whitechurch & Constantine 1993). When health professionals view families in their wholeness and are aware about their needs and resources, they are equipped to better understand and support family functioning and individual family members' wellbeing by supporting their coping and adaptation.

8.1.2. Family Assessment

The interpretive theory "Living in a Field of Tension between Development and Degeneration" and its four constituting categories can function as theoretical framework for clinical practice and give deeper insights into the families' functional, relational, social and emotional life domains. Health professionals such as advanced practice nurses can use this interpretive theory to assess and monitor the family situation over the course of the young person's transition into adulthood, identify their needs and resources, and screen for difficulties or areas of concern where families would benefit from professional support.

Mutigenerational Assessment

An area of concern may be inadequate family coping and adaptation, key processes related to family resilience (Kaakinen et al. 2010). Family coping and adaptation cannot be understood apart from the families' past responses to illness-related events (Rolland 2006b). Health professionals need to assess the families' multigenerational history which can be done by the use of family genograms. Different authors offer instructions on how to use genograms and comprehensively assess the families' situation (McGoldrick et al. 2013a, Schober & Affara 2001, Wright & Leahey 2009). Through a multigenerational

assessment health professionals can identify key aspects of the families' illness experience and gain a better understanding of their coping strategies (Rolland 2006b). These insights are needed in order to explain family processes such as meaning making, coping and adaptation and to identify the families' resources and particular vulnerability.

Assessing Family Members' Expectations

Deviation from an expected path can result in difficulties and confrontation (Garcia-Preto & Blacker 2013). When helping families to better cope with difficulties and adapt to change, an assessment of the family situation and their context in regard to expectations and standards that guide their actions and interactions is crucial. Only by considering the behaviour of parents, siblings and affected young people in relation to their social and cultural expectations and beliefs can professionals offer adequate support (Garcia-Preto & Blacker 2013). For instance, through the assessment outdated information related to the illness may be uncovered, which influenced the families' expectations negatively. While it is advised to anticipate future caregiving expectations and family traditions of meeting care demands such as gender role scripts that may overburden female family members, overemphasis on anticipatory loss can have negative consequences. Asking families about their hopes and fears and about the course and outcome of the illness they anticipate may be emotionally disabling (Rolland 2006b). Health professionals can support affected families to achieve a healthy balance and face uncertainty, for instance, by acknowledging the possibility of future loss, but sustaining hope, living in the present and creating family meaning and a sense of mastery and competence.

8.1.3. Family Interventions

Health professionals can use the interpretive theory as a framework to assess the family situation and to identify appropriate interventions and make an advanced care plan. An advanced care plan considers interventions for families that aim to better prepare them for what expects them during the young person's transition into adulthood comprising coping with illness and development-related challenges and adapting to change. Family interventions can involve different approaches that are directed to improve their health and wellbeing when living with chronic illness, among which are psycho-educative and relationship-oriented interventions (Chesla 2010, Mahrer-Imhof & Bruylands 2014). The effectiveness of such family interventions has been described, but according to Chesla (2010) reviews of intervention research in children with chronic illness are largely narrative and less comprehensive than evidence on family interventions for chronically ill adults. Nevertheless, evidence suggests that complex family situations require multimodal approaches, which combine family education with family support and skill building as well as psychosocial interventions with the focus on communication, problem solving and conflict management (Chesla 2010).

When making an advanced care plan, the timing of interventions is a factor that needs to be considered. Findings of this study suggest that trustful relationships emerge from continuous contacts with professionals and may positively influence the acceptance and outcomes of interventions. While patients affected by NMD generally benefit from trustful relationships with health professionals that evolve with time (Bartalos 1991), a benefit of early interventions has been described for psychosocial and relationship-centred family interventions

(Metcalfe et al. 2011, Schwartz et al. 1991). For example, a qualitative study on family communication about genetic risk revealed that children and young people were aware of the condition from a young age and wanted to receive gradual information and be able to discuss and understand what was happening in their families (Metcalfe et al. 2011). Most parents, however, chose to delay discussions about genetic risk until particular events happened which made ignoring it inevitable. Discussions about genetic issues were very distressing for them and they would benefit from health professionals' advice and assistance in providing developmentally appropriate information to their children (Metcalfe et al. 2011). Evidence further indicates that in moments of crises, families are particularly open to interventions from health professionals that could aim to help them restructure their internal organisation and to develop new pattern of response to challenges related to illness and development (Patterson 1991).

Supporting Family Communication and Interaction

Interventions that are fundamental to facilitating families coping and adaptation may be directed at supporting family communication (Kaakinen et al. 2010, Olson & Gorall 2003, Patterson 1991). How families relate and communicate with each other and how they negotiate and distribute caregiving tasks can give insights into how they manage situational, developmental and illness-related tasks. Inadequate family communication is a factor that inhibits families' coping and adaptation. Patterson (1991) describes two kinds of family communication; instrumental and affective communication. In order to function, family members need to apply both types of communication as they need to focus on both, tasks and emotions, as well as to disclose and listen. The role of advanced practitioners such as advanced practice nurses can be to facilitate family

communication at times when families are stressed by change and challenges, so that they can achieve more positive outcomes and effectively cope and adapt (Kaakinen et al 2010). They can help families to better express and negotiate their needs within the family and assist them viewing their experiences and behaviours in relation to their families' social and cultural context (White & O'Brien 2010). Currently, the SPRinG collaborative works with families and genetic counsellors to co-design and test a psycho-educational intervention that aims to facilitate family communication and promote better coping and adaptation when living with an inherited genetic condition for parents and their children through multi-family discussion groups (The Socio-Psychological Research in Genomics 2016).

Advanced practitioners need to develop skills in facilitating better family communication, coping and adaptation to living with a chronic condition, to prevent the family from becoming dysfunctional and unduly distressed at the position they find themselves within. Professional must be sensitive to underlying tensions or conflicts and able to communicate in non-judgmental ways, considering the family situation and their embedment in a lifetime of relationships and actions (White & O'Brien 2010). In addition, professionals should also be able to recognise that if a family has too many difficulties they might require referral to family therapists. Family therapy or psychotherapy and family nursing are distinct disciplines, even if family therapy has had its influences on family nursing and there certainly are intersections (Kean 2001).

Furthermore, findings revealed that family members experience intense emotional states during crises of health and because of anticipatory loss.

Crucial to effective interventions that aim to support the family's grieving process is the health professionals' reflection about their own experiences with loss (Rolland 2006b). Clinicians' ability to work effectively with families can be influenced by their own family history of loss. By knowing about one's unresolved personal losses and fear of death, health professionals are sensitised to the families' dilemmas, they are able to prevent excessive optimism by aligning with hopes and beliefs of the family which can lead to neglecting the inevitability of future loss (Rolland 2006b). Therefore, there is the risk of professionals becoming overinvolved or emotionally distant, two situations that are associated with avoidance of engaging in necessary discussions about anticipatory loss.

Management of Role Overload or Conflicts

If family members are confronted with difficulties that are related to their roles, including role overload or conflict, health professional can offer support by providing information and suggesting strategies how to better negotiate roles within the family and distribute tasks in order to find balance and discover meaningful solutions (Kaakinen et al. 2010). Such a solution can be planning and spending time away from the responsibilities of a role and to engage in energy producing activities. For example, another family member or friend could take over the caregiving tasks for a limited period of time, or professional help could be organised in order to disburden the family caregiver (Kaakinen et al. 2010). Another possibility would be if the affected family member is temporarily cared for in an institution, while the other family members go on a vacation. When making these recommendations, health professionals need to carefully consider the family situation, as some of the interventions may violate family

values and beliefs of what is adequate and can only be fruitful if appropriate and accepted by all family members (Kaakinen et al. 2010). Involving all family members in a round table discussion and open communication may facilitate their acceptance and flexibility.

Sexual Maturation: Supporting Individual and Family Integrity

The sexual maturation of affected young people was identified as area of concern, as it was a taboo within the family and at professional contacts. Considering the fact that physically dependent individuals are less likely to benefit from the sexual education and the guidance their peers get, as well as their higher risk for sexual abuse due to their physical dependence (World Health Organization 2007) professional support is advised. It is important that professionals that offer assistance to parents and affected young people have an understanding of the families' developmental needs, are aware about taboo subjects and issues related to privacy and family boundaries and have an interest in and feel comfortable with working with them (Bill & Knight 2007). They also need to reflect on the meaning that sexuality and disability has for themselves, as critical reflection enables them to overcome their own limits such as social taboos. If sexual maturation negatively affects family relationships or individuals family members' integrity by violation of individuals' privacy and family boundaries, professionals may support family members in reflecting about their practices and building more healthy relationships with each other, for instance, by outsourcing critical caregiving tasks to externals. The preservation of family boundaries allows for the energy that individual family members need to master developmental tasks and to develop healthy

adult relationships with their family members as a result of their transition into adulthood (Coelho & Manoogian 2010).

8.2. Implications for Research

8.2.1. The Suitability of a Family Systems Framework

This study generated important new insights into the family transition experience, by adopting a family systems theoretical framework and a family developmental perspective (Carter & McGoldrick 2005, McGoldrick et al. 2013b, Rolland 1987, Rolland 1994, Rolland 2006a, Rolland & Williams 2005, Rolland & Williams 2006). A family systems framework demonstrated its suitability for the exploration of the family transition experience when living with NMD in combination with a qualitative methodology, as it allowed identifying an interpretive theory which describes different domains that are affected by chronic illness and developmental needs. Moreover, family systems theory was appropriate to illuminate the reciprocal effects among the illness experience, human development and family life, three factors that have been described to be associated with coping and adaption with genetic illness (Rolland 1987, Rolland 1994, Rolland 2006a, Rolland & Williams 2005, Rolland & Williams 2006).

Other family theories, such as family therapy theories or nursing conceptual frameworks may have their values, but they are less comprehensive than family systems theory. Family therapy theories, for instance, focus on family dysfunction and are therefore less well suited to discover both negative and positive illness experiences (Kaakinen & Hanson 2010). A more holistic systems perspective, by contrast, considers both positive and negative factors,

e.g. factors related to caregiver burden and family resiliency, and it views the family experience as related to a larger context and over the course of life (McGoldrick et al. 2013b, Kaakinen & Hanson 2010). Therefore, future research on the family experience of chronic illness and on life cycle transitions would benefit from a family systems theoretical framework, which could possibly inform the development of future, more family focused interventions.

However, it needs to be stated that while thinking about the family as a whole is highly relevant, it is also an extremely difficult process, as it involves great complexity (McGoldrick et al. 2013b). Many factors need to be considered including how individuals' and families' experiences and characteristics as well as their socio-cultural context are linked with each other and what their relations and influences with former and future generations are over the course of life. What also needs to be considered is that while in non-familial systems functions and roles can be carried out in a more stable way by simply replacing people that leave, family systems differ (McGoldrick et al. 2013b). Families are very restrictive in the ways how they incorporate and leave members (e.g. by birth, marriage and death) and these constraints as well as the high importance of family relationships which are irreplaceable, make "thinking family" a complex endeavour.

8.2.2. The Need for Further Research

As a result of this study, the need for further research emerged. Further research is required to identify evidence-based family interventions that aim to support individual family members' wellbeing and integrity and family functioning. Interventions may be psycho-educative in nature or relationship-

focused and targeting family communication and pattern of interaction, which are crucial for the families' coping and adaptation (Chesla 2010, Mahrer-Imhof & Bruylands 2014). Moreover, interventions may focus on how families can deal with anticipatory loss, role overload and conflicts and needs related to a chronically ill family members' sexual and cognitive maturation. A review on family interventions for children and adults revealed that interventions that most benefitted family members were relationship-focused, but the effectiveness of these interventions may be different for affected individuals (Chesla 2010). To clarify, family interventions may operate differently for unaffected family members than they do for individuals living with chronic illness which is an idea that was considered a new line of inquiry (Chesla 2010).

The first step of further research may include the identification of family interventions, which can be done by searching for and consulting a systematic review (Craig et al. 2008). In case there is no recent, high quality systematic review, it is advised to conduct one or to explore what is already known about similar interventions. Consequently, further development work is required to develop, pilot, evaluate, report and implement a complex intervention which supports the families' transition experience when living with NMD. In order to do so effectively, it is recommended to involve affected families and experienced health care professionals in the process of planning, developing and implementing complex interventions (Schweizerische Akademie der Medizinischen Wissenschaften 2016). Guidance for developing and evaluating complex interventions are offered by the Medical Research Council (Craig et al. 2008).

These complex interventions may be directed at supporting families to cope with the psychosocial and emotional difficulties of their transition experience. Interventions of this type are particularly valuable, as today there is still no primary treatment for NMD (Amato & Russell 2008, Brandsema & Darras 2015, Bushby et al. 2005, Kohler et al. 2009), also because drug development is complex and clinical research difficult (Bladen et al. 2013, Commission of the European Communities 2008, Wästfeld et al. 2006). Complex interventions that focus on management of psychosocial and emotional difficulties can support family coping and adaptation processes and complement symptom management and treatment to slow disease progression. These interventions need to be considered as they have the potential to have significant impacts on individual integrity and wellbeing and family functioning, and as a consequence they lead to an improved experience and quality of life.

8.3. Personal Reflection and Closing Remarks

My motivation for conducting a PhD sparked during my work as a research associate, where I managed research projects and gained knowledge and skills with different research methodologies. This work experience in the Institute of Nursing of ZHAW fostered my critical thinking, encouraged my professional development, and satisfied my curiosity by giving me the opportunity to learn or create something new. I always valued working with patients and their families as a bedside nurse, but while working in nursing research I gained the knowledge and confidence to challenge long-established routines and taken for granted knowledge, while aiming for improvement of chronically ill patients and their families' situation.

Reflecting upon my motivation to work in nursing research I will always remember an exit interview at a former work place, where I have been asked what I would have wished for in order to stay longer. My response was: "I would have wished for opportunities to develop professionally". I have always been curious to explore the world and people around me and enjoyed studying new subjects. Therefore, I welcomed my employer's offer to do a PhD. A doctoral study is a unique way to learn more about a topic of interest and contribute to existing knowledge. Furthermore, it is an opportunity for personal development, by allowing personal growth and intellectual development. It is a way to invest in my research career, strengthen my international network and familiarise myself with the responsibilities of a principle investigator.

While my motivation for a PhD was professional in nature, my passion for studying this particular topic, the family transition experience when affected by

NMD, had a strong personal connection. From the moment I decided to pursue a PhD it was clear that I wanted to study families living with childhood NMD. Early on in my career I was advised to work with this population, as I am a sibling of an adult living with NMD, but until that point I had not had the courage to study a - for me - very interesting but also highly emotional topic: The NMD illness experience. As a consequence of choosing this particular topic for my dissertation, I had to accept the fact that some experiences of being a doctoral student may be hurtful and emotionally tiring or overwhelming. However, with the support of my family, friends and supervisors, I managed to overcome these challenges. The benefits of focusing on this personal and emotional topic outweighed and with my personal commitment I have never struggled with my motivation to finalise this project.

It is important to mention that I was too young to remember details about how my brother transitioned into adulthood, but nevertheless I compared my own life experience with that of the affected families I interviewed. I reflected about my own role within my family and as a nurse and researcher. During my critical reflections it became obvious to me that being a researcher, a nurse and a family member came along with differing interest, which were all reflected by the meaning that I contributed to the research project and the pressure I put myself under. I wanted to conduct a research study that meets current scientific standards and allows me to complete my PhD. I also aimed to create something meaningful, which acknowledges what these families manage to accomplish, which is helpful to improve their future care by informing clinical practice, but most importantly, with my research I did not want to offend or hurt these families and in specific my family. I believe that my expectations are legitimate, but they

created a lot of stress. With passing of time I learned to deal with stress, by acknowledging my situation and making the best of it, similar to how my participants coped and adapted. It needs to be said that I have learnt a lot and I would like to conclude this reflection with the words of a person living with an adult onset NMD, Stephen Hawking, which in my opinion, reflect some of my personal experiences, acknowledge the importance of diversity and humanity and help to create meaning:

"We are all different, there is no such thing as a standard or run-of-the-mill human being, but we share the same human spirit. What is important is that we have the ability to create. This creativity can take many forms, from physical achievement to theoretical physics. However difficult life may seem, there is always something you can do and succeed at." (Hawking 2012)

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Appendix

Appendix 1: Peer-reviewed article

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Review

Living a normal life in an extraordinary way: A systematic review investigating experiences of families of young people's transition into adulthood when affected by a genetic and chronic childhood condition

Veronika Waldboth [a,b,*], Christine Patch [b,c], Romy Mahrer-Imhof [a], Alison Metcalfe [b]

[a] Institute of Nursing, Zurich University of Applied Sciences, Winterthur, Switzerland
[b] Florence Nightingale Faculty of Nursing and Midwifery, King's College London, London, UK
[c] Department of Clinical Genetics, Guys & St Thomas' NHS Foundation Trust, London, UK

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ABSTRACT

Introduction: The transition into adulthood is a developmental stage within the life cycle. A chronic childhood condition can disrupt this transition and create major challenges for both the young person and his or her family. Little is known about families' experiences when living with a rare genetic disease. Therefore, the purpose of this literature review was to understand experiences of families living with a chronic childhood disease during transition into adulthood by integrating evidence.

Method: A systematic review using an integrative approach to data inclusion and analysis comprising qualitative, quantitative and other methodological studies about a range of genetic and chronic childhood diseases was undertaken to identify relevant information. Databases searched were PubMed, Cochrane Library, PsychINFO, CINAHL, and AMED, using the search terms (1) family, caregivers, young adult, adolescent; (2) adolescent development, transitional programs, transition to adult care; (3) muscular dystrophy, spinal muscular atrophy, cystic fibrosis, haemophilia and sickle cell disease. Study findings were critically appraised and analyzed using critical interpretive synthesis.

Results: A total of 8116 citations were retrieved. 33 studies remained following the removal of duplicates, papers unrelated to genetic childhood conditions and families' experiences of the transition into adulthood. Findings provided three perspectives: (1) the young person's perspective on how to "live a normal life in an extraordinary way" and "manage a chronic and life threatening disease"; (2) the parent perspective on the "complexity of being a parent of a chronically ill child" and "concerns about the child's future" and (3) the sibling perspective on "concerns about the siblings future".

As a consequence of the genetic childhood condition, during the ill family members' transition into adulthood all family members were at risk for psychosocial difficulties as they mutually influenced each other. Previous research focused predominately on the individual illness experience, and less emphasis was put on the family perspective.

* Corresponding author at: Institute of Nursing, Zurich University of Applied Sciences, School of Health Professions, Technikumstrasse 81, 8401 Winterthur, Switzerland.
E-mail addresses: veronika.waldboth@zhaw.ch (V. Waldboth), Christine.Patch@gstt.nhs.uk (C. Patch), romy.mahrer@zhaw.ch (R. Mahrer-Imhof), alison.metcalfe@kcl.ac.uk (A. Metcalfe).

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Conclusions: Young people and their family members experienced multiple challenges and not only for the ill individual but also there were consequences and health risks for the whole family system. Therefore, a family systems perspective to research and care is indicated to assist affected families to cope with their complex life and health situation.

What is already known about this topic?

- Individuals affected by genetic conditions with an onset in childhood now live longer than ever before and an increasing number of young people reach adulthood.
- Chronic childhood conditions have an impact on the young persons' health and life situation and, as a consequence, influence quality of life of the whole family system.

What this paper adds

- Young people affected by chronic childhood disease and their family members experience multiple challenges imposed by the chronic condition and the developmental stage that the young person is in.
- The affected young person and his or her parents and siblings are at risk for psychosocial and emotional difficulties which may lead to developmental difficulties and health problems.
- A family systems approach to research and care is indicated to assist families to cope with major life transitions and their complex health and life situation.

1. Background

Diseases are defined as rare, when they affect less than 5 out of 10,000 individuals (Commission of the European Communities, 2008). Although individual diseases are defined as rare, rare diseases are collectively common (Elliot, 2015) and the total impact should not be underestimated. Approximately 30 million European and 25 million North American patients are currently affected by one out of 5000–6000 different rare or orphan diseases (Schieppati et al., 2008; Wästfelt et al., 2006). More than two thirds of these diseases involve children or adolescents (Boycott et al., 2014). The majority are genetic in origin and frequently they are life-threatening, life-limiting or chronically debilitating. Their impact on the quality of life of the affected young individual and his or her family is significant (Drotar, 1981; Neill, 2010; Rolland and Williams, 2005; Samson et al., 2009; Sobralske, 2013).

Families may suffer from psychosocial, emotional and physical difficulties (Pangalila et al., 2012). Moreover, chronic childhood diseases are associated with a substantial economic burden including direct, indirect and informal costs. Landfeldt et al. (2014), for instance, estimated a conservative amount of societal and household financial burden per patient per annum for rare genetic neuromuscular conditions at between $80,120 and $120,910 and $58,440 and $71,900, respectively. Thus, families may face economic hardship, feel stressed by inadequate services and are often under-supported (Elliot, 2015).

The delineated socio-psychological and economic burden increases with disease progression. This implies that the longer that children with progressive conditions live, the greater their physical dependence and need for assistance will be. Families face new challenges as chronically ill children are now living longer than ever before because of progress in health care and medicine (Bushby et al., 2005; Drotar, 1981; Kohler et al., 2009). Today, most individuals with rare chronic and genetic conditions such as cystic fibrosis or severe neuromuscular disease survive adolescence and reach adulthood. Thus, families have to deal with their children's increasing need for care and potential challenges, which are largely unexplored.

The transition into adulthood is a time of intense and focused transition and personal development for all young people but the process can be even more challenging for individuals affected by a chronic childhood disease (Arnett, 2000; Bill and Knight, 2007; Christie and Viner, 2005; Mietzel, 2002; Monteith, 2004). The challenges are from multiple perspectives and family members play an important role during this developmental stage, as the whole family needs to reorganize (McGoldrick et al., 2013; Miller et al., 2006). Young individuals need sufficient freedom to develop their skills in decision making, in taking responsibility for themselves and becoming more independent. Parents, on the contrary, need to be more flexible and to permit their child's independence. For the child's development it is essential that families find a balance between single family members' needs and family functioning (McGoldrick et al., 2013; Patterson, 1991; Rolland, 1994). For families affected by a genetic childhood condition, they have to also balance management of their child's illness and this might affect their functioning which has potential repercussions for the young person, their siblings and the parents.

Families have to assist the young person in managing the transition to adulthood whilst also managing a serious disease, requiring a family centred approach to care (Miller et al., 2006; Rolland and Williams, 2005; Rolland, 2012; Segrin and Flora, 2011).

The transition into adulthood was identified as a particularly sensitive and challenging time for young individuals living with chronic childhood condition and their families. This is influenced by the fact that the young person is expected to grow and probably wants to be more independent at the same time as their physical disability and dependence on their families' support increases. By assisting these families in their adjustment, young people and their families are prevented from becoming isolated and left to cope with the disease and the effects on family relationships (Miller et al., 2006; Rolland and Williams, 2005).

Despite knowing some of the challenges the young person faces, little is known about other family members' experiences when a young person is affected by a chronic childhood disease (Abbott et al., 2012; Michaud et al., 2007). In order to be able to assist these families, we need to understand, what young people, their parents and their siblings experience and the challenges confronting them. By investigating and integrating evidence about the experiences of these families we can identify their explanations and expectations of events and understand how families cope with the young person's transition into adulthood. This knowledge will help health professionals to better assist families to anticipate and react to difficult and stressful situations associated with long-term genetic conditions at an important life stage.

Therefore, the aim of this study was to conduct a systematic literature review and critique existing literature about the experiences of young individuals and their families where the young individual is affected by a genetic childhood disease and transitions into adulthood. The emphasis of this study was placed on progressive genetic childhood conditions including neuromuscular diseases, cystic fibrosis, haemophilia and sickle cell disease. These diseases are thought to have commonalities in the illness experience, as they all are genetic in origin, have their onset in childhood, and with physical impairments. The following research question was the focus for the literature review: What are the experiences of young people living with a genetic and chronic childhood condition and their families when the young person is transitioning into adulthood?

2. Methods

A systematic review was selected as the appropriate study design for this study as it answers precise research questions and allows identification, evaluation and synthesis of the findings of all relevant studies in an organized and reproducible way (Norman and Griffiths, 2014). This systematic review adopted an integrated approach and included empirical research from different research paradigms. This meant that identification of papers relevant to the question was not limited by methodological approaches. Integrating qualitative and quantitative evidence is an approach suited to learning more about an emerging topic that would benefit from a holistic conceptualisation and allows for the generation of new perspectives (Torraco, 2005; Whittemore and Knafl, 2005).

The theoretical framework underpinning this investigation was a family systems approach within in a biopsychosocial model for genetic diseases and the family life cycle (McGoldrick et al., 2013; Miller et al., 2006; Patterson, 1991; Rolland, 1994; Rolland and Williams, 2005; Segrin and Flora, 2011). These theories guided the research process (planning, study selection, data extraction, data analysis and -synthesis phases) and provided a framework for the interpretation of the findings.

2.1. Study planning and search strategy

At the planning stage variables of interest that would function as search terms were derived from the research question. To identify relevant literature key nursing, medical and psychosocial databases were searched from June to August 2014 including PubMed, Cochrane Library, PsychINFO, CINAHL and AMED. The search was conducted by two researchers using keywords, truncated keywords and subject headings. Search terms were combined with Boolean operators OR and AND and no limits were used. In addition to the search in electronic databases, other information sources were searched including a hand search in reference lists of identified articles, selected journals, websites and through contact with experts in the field. Search results were organized and managed using the reference manager EndNoteX4. Table 1 represents search terms and their application within specific electronic databases.

2.2. Literature selection process

Identified studies were then reviewed against all following inclusion and exclusion criteria for their eligibility and either included or excluded at four different stages of the study selection process (Fig. 1).

- Empirical peer-reviewed research articles including qualitative, quantitative, mixed and participatory approaches.
- Research on family experiences including family members' views, beliefs, experiences, actions, expectations, explanations and strategies, comprising single and multiple perspectives of young individuals living with

Table 1
Search terms.

Keywords and truncations	Subject headings or Mesh terms
family; families; caregive*; young adult*; adolescen*; transition*; muscular dystrop*; spinal, muscular atrop*; cystic fibro*; haemophil*; hemophil*; sickle cell dis*; sickle cell anemi*	**PubMed**: Family; Caregivers, Young Adult; Adolescent; Adolescent Development; Transition to Adult Care; Muscular Dystrophies; Muscular Atrophy, Spinal; Cystic Fibrosis; Hemophilia A; Hemophilia B; Anemia, Sickle Cell **PsychINFO**: Family; Caregivers; Adolescent Development; Muscular Dystrophy; Muscular Atrophy; Cystic Fibrosis; Hemophilia; Sickle Cell Disease **CINAHL**: Family; Caregivers; Young Adult; Adolescence; Transitional Programs; Muscular Dystrophy; Muscular Atrophy, Spinal; Cystic Fibrosis; Hemophilia; Anemia, Sickle Cell **AMED**: Family; Caregivers; Adolescent; Adolescence; Transitional Programs; Muscular Dystrophy; Muscular Atrophy; Cystic Fibrosis; Hemophilia; Anemia, Sickle Cell **Cochrane Library**: Family; Caregivers, Young Adult; Adolescent; Transition to Adult Care; Muscular Dystrophies; Muscular Atrophy, Spinal; Cystic Fibrosis; Hemophilia A; Hemophilia B; Anemia, Sickle Cell

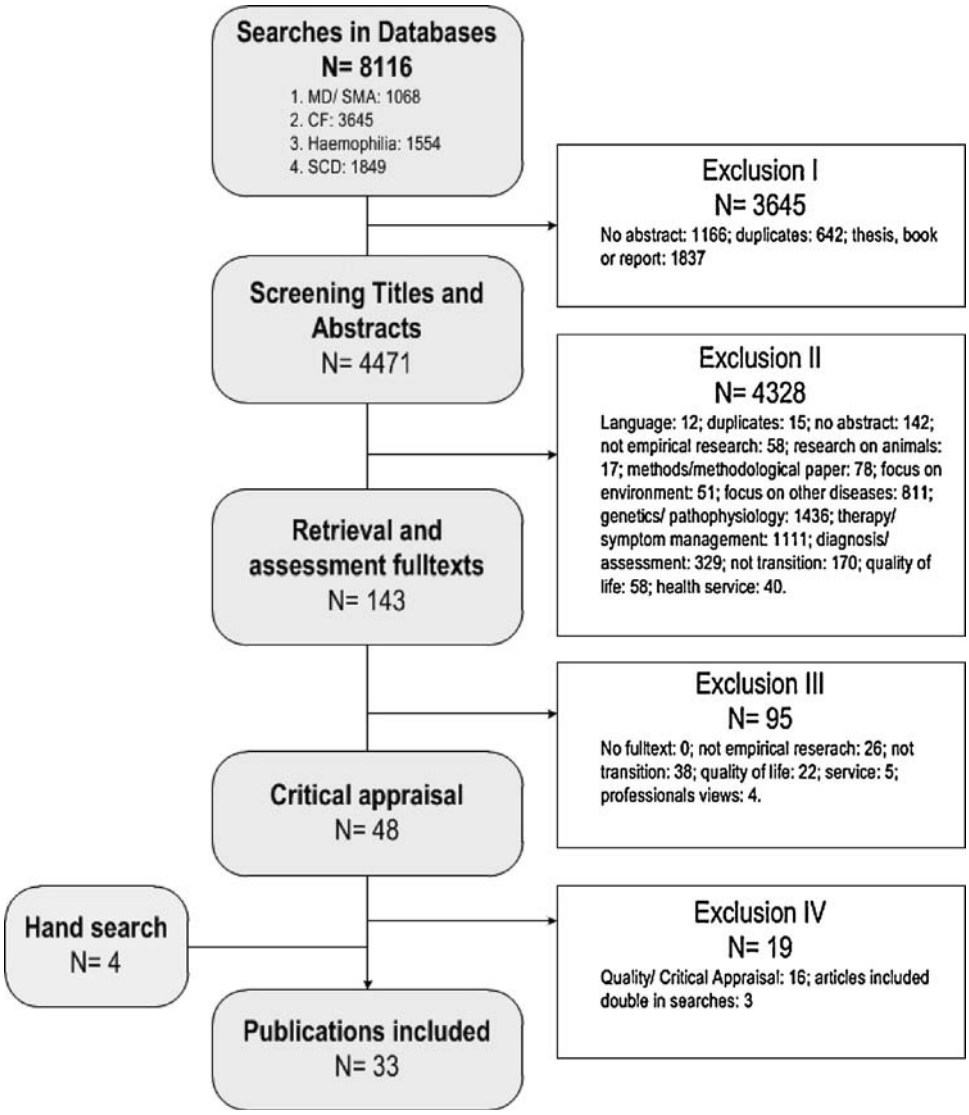

Fig. 1. Study selection process.

the condition, their parents, younger and older siblings and other next of kin.

- Research focusing on the transition into adulthood; Experiences of the young individual and his or her family, when the young person living with the genetic disease was 14–30 years old or where the reported mean age of the young participants was within that age limit.
- Research on genetic childhood conditions: Genetic conditions with symptom onset in childhood and progressive in nature, with moderate to severe physical impairment, no major cognitive impairment, with emphasis on neuromuscular diseases including muscular dystrophies (MD) and spinal muscular atrophies, cystic fibrosis (CF), haemophilia and sickle cell diseases (SCD).

Criteria for exclusion of articles were:

- Language of the articles other than English, German or Italian;
- Inability to obtain the full text of the article;
- Articles without abstract;
- Methodological papers and none peer-reviewed articles like reports, books and dissertations/thesis;
- Research including professional's perspectives only (description or evaluation of transition programmes or services only, health professional's views only);
- Quantitative measurements of Quality of Life only (these studies have been excluded, as a recent systematic review on Quality of Life in rare genetic conditions has been conducted by Cohen and Biesecker (2010)).

After an initial review of titles and abstracts identified through the search, full texts of articles that seemed potentially relevant were obtained for further assessment of eligibility. Studies meeting all inclusion and no exclusion criteria were then critically appraised using the "QualSyst" standard quality assessment criteria (Kmet et al., 2004), a pragmatic tool appropriate for the assessment of both qualitative and quantitative evidence.

Table 2
QualSyst assessment criteria.

Criteria for quantitative studies	Criteria for qualitative studies
1. Question/objective sufficiently described?	1. Question/objective sufficiently described?
2. Study design evident and appropriate?	2. Study design evident and appropriate?
3. Method of subject/comparison group selection or source of information/input variables described and appropriate?	3. Context for the study clear?
4. Subject/comparison group characteristics sufficiently described?	4. Connection to a theoretical framework/wider body of knowledge?
5. Interventional and random allocation described?	5. Sampling strategy described, relevant and justified?
6. Interventional and blinding of investigators reported?	6. Data collection methods clearly described and systematic?
7. Interventional and blinding of subjects reported?	7. Data analysis clearly described and systematic?
8. Outcome/exposure measure(s) well defined and robust to measurement/misclassification bias? Means of assessment reported?	8. Use of verification procedure(s) to establish credibility?
9. Sample size appropriate?	9. Conclusions supported by the results?
10. Analytic methods described/justified and appropriate?	10. Reflexivity of the account?
11. Some estimate of variance is reported for the main results?	
12. Controlled for confounding?	
13. Results reported in sufficient detail?	
14. Conclusions supported by the results?	

The QualSyst checklist for quantitative studies contains 14 items that rate criterion as 'met' (2 points), 'partially met' (1 point) or 'not met' and finally 'not applicable' (Table 2). A quantitative paper meeting all criteria could score up to a maximum of 28 points. The QualSyst checklist for qualitative studies consisted of 10 items using the same rating scale. The total possible sum score for qualitative articles was 20 points. Quality scores were calculated by dividing the sum scores by the total possible scores (Kmet et al., 2004). In this literature review the quality scores defined a minimum threshold for inclusion of articles and the cut-off point selected was relatively low, at 55%, to maximize the inclusion of a variety of papers with different conceptual levels (Dixon-Woods et al., 2006; Kmet et al., 2004).

To assure quality, the critical appraisal was conducted by two researchers. There were few discrepancies between the two raters with the maximum difference in sum scores being two and both raters agreed on the inclusion of the studies in the analysis. Therefore, the interrater agreement was evaluated as sufficient.

2.3. Data extraction process and synthesis of results

Information from selected studies was extracted according to the studies aims and purposes, methodology used, population and type of condition and key themes of the individual and family experiences, which were relevant to this reviews' aim.

Then, evidence was synthesized following an interpretive approach to evidence synthesis using a variant of meta-ethnography, called critical interpretive synthesis (Dixon-Woods et al., 2006; Pope et al., 2007). Potential benefits of building a cumulative knowledge base by combining qualitative and quantitative evidence in interpretive synthesis have recently been reported (Barley et al., 2011; Dixon-Woods et al., 2006; Metcalfe et al., 2008; Pope et al., 2007).

Qualitative and quantitative articles were analyzed and synthesized by two authors using techniques of critical interpretive synthesis; thematic analysis, graphical mapping and constant comparison (Dixon-Woods et al., 2006; Pope et al., 2007). Initially, key concepts and themes from the articles were extracted and coded. Building on these original results and explanations and interpretations of studies codes were transformed into a new conceptual form. At different stages new codes and concepts were thematically organized and further developed into categories. The emergent categories and the relationship between them were used to explain the phenomenon of interest.

Credibility, quality, rootedness of the evidence within the body of literature and contextual factors were considered and reflected throughout the process to critically appraise the existing evidence (Dixon-Woods et al., 2006).

Two checklists guided the reporting of this study where applicable: (1) The MOOSE – Meta-analysis of Observational Studies checklist (Stroup et al., 2000) and the PRISMA checklist (Moher et al., 2009).

3. Results

A total of 8116 citations were retrieved. 33 studies remained following the removal of duplicates and papers unrelated to families' experiences of the transition into adulthood and living with genetic childhood conditions. Details of the study selection process can be found in Fig. 1 and findings from individual studies are presented in Table 3.

3.1. Study characteristics

The majority of the articles employed qualitative research designs (29). Only three studies used a quantitative, cross-sectional study design. The conditions were distributed as follows: 26 articles focusing on one condition and 7 articles investigating two or more conditions. 16 articles included young individuals and families living with CF; 10 SCD, 7 MD including Duchenne MD, and 2 Haemophilia. Included studies were predominantly conducted in North America and Europe (18 in North America and Oceania, 14 in Europe, 1 Brazil). More than two thirds of the articles were published within the last decade (22); only two articles were more than 20 years old.

Table 3
Results of individual articles.

	Reference/ country	Purpose/aim	Methodology	Sample	Genetic condition	Families' experiences (key themes/concepts)	Quality-score (%)[a]
1	Admi (1996) USA	Generate a descriptive theory of the process of growing up	Qualitative design: Life history method, interviews, data analysis (Glaser and Strauss; Strauss and Corbin)	Young people (16–25 years), parents, spouses *N* = 21	CF	Family life, wider society, creation of identity, physical health, emotional and psychological health, self-management	90
2	Al-Yateem (2012) Ireland	Explore the experiences of young adults during the transfer to adult care	Qualitative design: Phenomenology (van Manen)	Young people (age range not specified) *N* = 15	CF	Transfer to adult care	80
3	Antle et al. (2008) Canada	Examine parents' health promotion efforts within the family context	Qualitative design: Long Interview Method (McCracken), combination of Phenomenology and Grounded Theory	Parents of adolescents (11–16 years), young people *N* = 15	MD and other conditions	Complexity of parenting a chronically ill child, concerns about the child's future	85
4	Atkin and Ahmad (2001) UK	Explore how young people live and cope with the disease	Qualitative design: Interviews, qualitative data analysis (Gubrium and Silverman)	Young people (aged 10–19 years) *N* = 51	SCD and thalassemia major	Friends and peers, family life, wider society, education and career, creation of identity, physical health, emotional and psychological health, self-management, transfer to adult care, coping and adaptation strategies	75
5	Badlan (2006) UK	Explore the subjective perspective of young people living with the condition	Qualitative design: Hermeneutic phenomenology (Moustakas), group and single interviews	Young people (17–31 years) *N* = 31	CF	Education and career, creation of identity, physical health, emotional and psychological health, self-management, transfer to adult care, coping and adaptation strategies	80
6	Berge et al. (2007) USA	Explore gender differences in the transition to adulthood in order to understand the psychosocial impact of the disease	Qualitative design: Focus group interviews, hermeneutic analysis (Addison; Miller; Crabtree)	Young people (16–21 years) *N* = 17	CF	Family life, risky behaviour, independent living, creation of identity, physical health, emotional and psychological health, self-management, transfer to adult care, coping and adaptation strategies	75
7	Bregnballe et al. (2011) Denmark	Determine the kind of parental support adolescents with the disease prefer	Qualitative design: Secondary analysis of interviews, interpretive description (Thorne)	Young people (14–25 years) *N* = 16	CF	Family life	75
8	Cappelli et al. (1989) Canada	Understand the impact of the disease on the adolescents perspective on psychological and social functioning	Quantitative Design: Cross sectional study with three groups, analysis of variance	Young people (mean age 14.5–14.7) *N* = 93	CF and diabetes, healthy control group	Family life, physical health, emotional and psychological health	77
9	Christian and D'Auria (1997) USA	Explore adolescents conceptualization of their illness experience and related life events of growing up with the condition	Qualitative design: Retrospective interviews (Strauss and Corbin), content analysis (Miles and Huberman)	Young people (12–18 years) *N* = 20	CF	Friends and peers, romantic relationships, creation of identity, self-management	85

Table 3 (*Continued*)

	Reference/ country	Purpose/aim	Methodology	Sample	Genetic condition	Families' experiences (key themes/concepts)	Quality-score (%)[a]
10	Dreyer et al. (2010) Denmark	Examine experience of living with physical impairment, with home mechanical ventilation and physical impairment	Qualitative design: Hermeneutic phenomenology (Ricoeur)	Young people and adults (21–40 years, the study focuses on teenage experiences) *N* = 19	Duchenne MD	Friends and peers, romantic relationships, physical health, emotional and psychological health, coping and adaptation strategies	85
11	Dupuis et al. (2011) Canada	Explore the experience of families as they attempted to prepare for the transfer from paediatric to adult care	Qualitative design: Interviews, content analysis (Miles and Huberman)	Young people (16–18 years), parents, professionals *N* = 26	CF	Coping and adaptation, complexity of parenting a chronically ill child, concerns about the child's future	75
12	Erskine (2012) UK	Examine young men's experiences with the disease	Qualitative design: Interpretive Phenomenological Analysis (Smith)	Young people (13–17 years) *N* = 8	SCD	Friends and peers, family life, wider society, creation of identity, physical health, emotional and psychological health, self-management, transfer to adult care	70
13	Gibson et al. (2007) Canada	Explore the identities and social positionings of men with the condition by examining how they responded to dominant discourses of disability, masculinity and assistive technologies	Qualitative designs: Ethnographic case studies, interviews, videos, content analysis (Miles and Huberman)	Young adults (22–36 years, results focus on young adolescence, young adulthood) *N* = 10	Duchenne MD	Friends and peers, romantic relationships, education and career, creation of identity, coping and adaptation strategies	75
14	Gibson et al. (2014) Canada	Investigate the intersectionality of gender, disability, and emerging adulthood	Qualitative design: Interviews, photo elicitation, narrative diaries, content analysis (Miles and Huberman)	Young people (16–27 years) *N* = 11	Duchenne MD	Romantic relationships, wider society, independent living, creation of identity	65
15	Gjengedal et al. (2003) Norway	Examine the experience of growing up with and living with the condition; explore the patients' and their families' encounters with the health care system	Qualitative design: Focus group interviews, data analysis (Knodel)	Adults (20–47 years, focus on experiences during adolescence, young adulthood), parents of children aged (2–7 years) *N* = 22	CF	Education and career, physical health, emotional and psychological health, self-management, complexity of parenting a chronically ill child, concerns about the child's future	60
16	Hauser and Dorn (1999) USA	Understand concerns, expectations and preparation needs regarding transfer, describe differences of services, barriers, experiences and common practices; identify natural points of transition, generate a framework for transition	Qualitative design: Focus group interviews, content analysis (not specified)	Young people, parents (mean age 16.2 years), caregivers, professionals *N* = 52	SCD	Transfer to adult care, complexity of parenting a chronically ill child	75

Table 3 (*Continued*)

	Reference/ country	Purpose/aim	Methodology	Sample	Genetic condition	Families' experiences (key themes/concepts)	Quality-score (%)[a]
17	Higham et al. (2013) UK	Explore the hopes and fears for the future of young adults with the condition	Qualitative design: Interviews, Grounded Theory (Strauss and Corbin)	Young people (18–29 years) *N* = 15	SCD	Romantic relationships, education and career, creation of identity, self-management	80
18	Johannesson et al. (1998) Sweden	Investigate how the disease was connected with psychosocial issues concerning puberty and motherhood	Qualitative design: Interviews, data analysis (not specified)	Young adults (22–34 years; focus on experiences during adolescence) *N* = 14	CF	Friends and peers, romantic relationships, creation of identity, physical health, emotional and psychological health	70
19	Moola and Norman (2011) Canada	Explore how young people and their parents understand their health in the future and the perspectives they bring towards the concept of time	Qualitative design: Interviews, thematic analysis (Braun & Clarke; Boyatzis)	Young people (11–17 years), parents, professionals *N* = 85	CF and other conditions	Romantic relationships, education and career, creation of identity, physical health, emotional and psychological health, self-management, complexity of parenting a chronically ill child, concerns about the child's future	70
20	Müller-Kägi et al. (2014) Switzerland	Investigate the process of adherence to treatment in adolescents	Qualitative design: Interviews, constructivist Grounded Theory (Charmaz)	Young people (15–22 years) *N* = 13	Haemophilia	Creation of identity, physical health, emotional and psychological health, self-management	75
21	Palmer and Boisen (2002) USA	Examine the perceptions of the process and experience of becoming an adult with the disease	Qualitative design: Interviews, content analysis	Young adults (20–26 years) *N* = 7	CF	Family, education and career, risky behaviour, creation of identity, self-management, coping and adaptation strategies	80
22	Parkyn and Coveney (2013) Australia	Explore the meaning of a group experience	Qualitative design: Discussion groups, online data collection, qualitative analysis (Coveney)	Young people (14–17 years), parents *N* = 11	MD	Friends and peers, wider society, creation of identity, complexity of parenting a chronically ill child, concerns about the child's future	85
23	Pehler and Craft-Rosenberg (2009) USA	Describe the lived experience of spirituality	Qualitative design: Phenomenology (van Manen)	Young people (12–17 years) *N* = 9	Duchenne MD	Friends and peers, creation of identity, physical health, emotional and psychological health, coping and adaptation strategies	85
24	Pizzignacco and de Lima (2006) Brasil	Investigate how the socialization process of children and adolescents with the condition based on their own experiences	Qualitative design: Interviews, qualitative analysis (Bogdan)	Children and young people (7–18 years) *N* = 8	CF	Romantic relationships, creation of identity, self-management, coping and adaptation strategies	60
25	Porter et al. (2014) USA	Describe the perspective of adolescents, siblings and caregivers regarding transfer and identify recommendations for improvement	Qualitative design: Focus group interviews, qualitative analysis (Saldana)	Young people (12–18 years), parents, caregivers, siblings *N* = 34	SCD	Transfer to adult care, complexity of parenting a chronically ill child, concerns about the siblings future	80

Table 3 (*Continued*)

	Reference/ country	Purpose/aim	Methodology	Sample	Genetic condition	Families' experiences (key themes/concepts)	Quality-score (%)[a]
26	Schmitt (1997) Germany	Describe values and goal orientation in chronically ill adolescents and young people	Qualitative design: Interviews, content analysis	Young people (13–18 years) *N* = 164	Haemophilia and other conditions	Friends and peers, creation of identity, coping and adaptation strategies	75
27	Telfair et al. (1994) USA	Identify concerns, feelings regarding transfer, feelings if programme would be necessary and what it should offer	Quantitative Design: Cross sectional study, simple bivariate statistics	Young people (13–30 years), parents, caregivers *N* = 36	SCD	Transfer to adult care, concerns about the child's future	73
28	Telfair et al. (2004) USA	Present the voice of the adolescent, if a service for transfer should exist and concerns and expectations	Quantitative Design: Cross sectional study, simple bivariate statistics	Young people (mean age 14 +) *N* = 172	SCD	Transfer to adult care	91
29	Tuchman et al. (2008) USA	Describe expectations and concerns about the transition	Qualitative design: Longitudinal, interviews, content and thematic analysis	Young people *N* = 22	CF, SCD and other conditions	Transfer to adult care	60
30	Valenzuela et al. (2013) USA	Understand how children and adolescents with condition perceived their lives and the disease	Participatory approach adopting a qualitative design: photo voice, group discussions, Grounded Theory analysis (Glaser and Strauss)	Children and young people (8–17 years) *N* = 16	SCD	Friends and peers, family life, creation of identity	70
31	van Staa et al. (2011) Netherlands	Map experiences with the recent transfer to adult care and to identify recommendations to improve the transition process	Qualitative design: Interviews, thematic analysis (Braun & Clarke)	Young people (15–22 years), parents, professionals *N* = 24	Haemophilia, SCD, CF and other conditions	Transfer to adult care, complexity of parenting a chronically ill child, concerns about the child's future	80
32	Williams et al. (2009) UK	Explore children's and young people's implicit and explicit accounts of the ways in which they defined, produced, and maintained a sense of non-difference and the ways in which this non-difference was threatened by their illness	Qualitative design: Interviews, framework analysis (Ritchie and Spencer)	Children and young people (7–17 years) *N* = 32	CF	Creation of identity	90
33	Witte (1985) UK	Investigate family experiences, psychological dynamics, communication processes and coping strategies within families	Qualitative design: Interviews, projective assessment and psychodrama, analysis (not specified)	Young people (13–16 years), parents *N* = unknown	MD	Friends and peers, family life, wider society, concerns about the child's future	55

[a] Quality score in % according to the QualSyst critical appraisal checklists (Kmet et al., 2004).

3.2. Methodological quality of included studies

The quality of the included studies was assessed using the QualSyst checklist (Kmet et al., 2004) which revealed a quality score giving an overall estimate of the quality of the study. The range of quality scores of the included studies varied from 55 to 91% (min 55% and max 100%). By and large, the quality of the studies can be rated as good with 23 out of 33 studies scoring 75% or above. Only four studies scored 60% or lower. The majority of qualitative studies showed methodological limitations or weaknesses in describing their connection to a theoretical framework

Table 4
Study quality and weaknesses.

Study quality and weaknesses	References (# see Table 3)
Question or objective not or partially described	9, 12, 33
Research context not or partially clear	10, 11, 12, 15, 22, 26
Sampling strategy not described/or deficient	5, 7, 14, 15, 19, 20, 24, 26, 29, 33
Study design not appropriate/data collection or analysis not described	5, 13, 14, 16, 18, 20, 24, 25, 29, 30, 33
No or partial connection to theoretical framework	2, 3, 4, 6, 12, 16, 17, 18, 20, 24, 29, 31
Credibility not or partially described	2, 4, 6, 7, 11, 13, 14, 15, 17, 19, 21, 24, 26, 29, 30, 33
Reflexivity not or partially described	1, 3, 4, 5, 6, 7, 9, 10, 11, 12, 13, 14, 15, 16, 17, 18, 20, 19, 21, 22, 23, 24, 25, 26, 29, 30, 31, 32, 33
Conclusions partially supported by results	12, 16, 18, 23, 30, 33
Questionnaire content not or partially discussed	27, 28
Sample size not or partially discussed	8
Variance not or partially discussed	8, 27, 28
Confounding variables not or partially discussed	8, 27, 28
Partial report of results	8, 27, 28

(12), partial or no account of credibility (16) or reflexivity (29). Table 4 lists all assessed weaknesses and the corresponding references.

3.3. Synthesis of results

Three perspectives emerged from the findings, focusing on the transition into adulthood: (1) the young person's perspective on how to live a normal life in an extraordinary way and manage a chronic and life threatening disease; (2) the parents' perspective on the complexity of being a parent of a chronically ill child and concerns about the child's future and (3) the sibling's perspective on concerns about the sibling's future (Fig. 2).

(1) The young person's perspective

The transition into adulthood was described as a time when young people affected by chronic childhood disease wanted to be and live like other young people. They wanted to engage in the same life activities as their healthy peers (Atkin and Ahmad, 2001; Badlan, 2006; Christian and D'Auria, 1997; Dreyer et al., 2010; Dupuis et al., 2011; Gibson et al., 2014; Gjengedal et al., 2003; Higham et al., 2013; Moola and Norman, 2011; Müller-Kägi et al., 2014; Palmer and Boisen, 2002; Pizzignacco and de Lima, 2006; Schmitt, 1997; Valenzuela et al., 2013; Williams et al., 2009). This desire to take part in major life pursuits that were considered "normal" for young people included increasing contacts with friends and engagement in social life, focus on education and career choices and a growing independence from their families. At the same time, however, affected individuals had to manage a chronic and life threatening disease which imposed many challenges to their lives. Mastering their complex disease management and everyday life required considerable efforts.

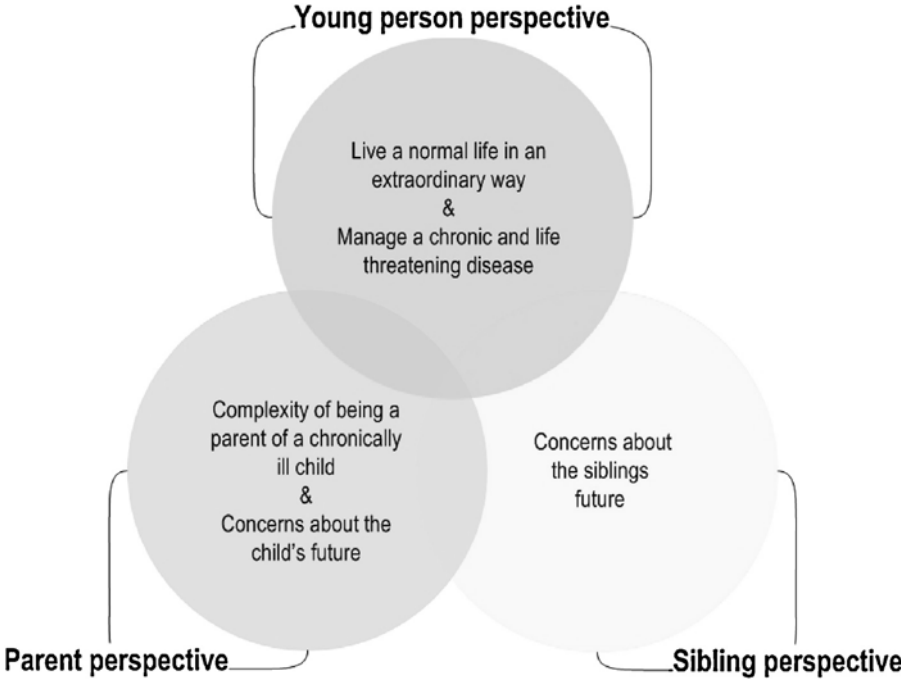

Fig. 2. Three perspectives.

3.4. Friendships and social life

At adolescence, young people with chronic diseases wanted to spend more time with their friends than with their families (Dreyer et al., 2010; Gibson et al., 2007; Parkyn and Coveney, 2013; Schmitt, 1997). Getting together with friends was highly important because it evoked positive feelings of being normal. Close friends, in particular, were described as very supportive during this developmental phase (Christian and D'Auria, 1997; Pehler and Craft-Rosenberg, 2009; Valenzuela et al., 2013). Not all diseases, however, allowed for the same amount of engagement in peer group activities. Wheelchair bound individuals, for instance, experienced environmental barriers and needed technical and personal assistance (Dreyer et al., 2010). Therefore, they were unable to participate in many teenage activities.

Not all contacts with peers or wider society were experienced positively. Some young people with MD and SCD reported being teased and discriminated against by others because of their physical appearance (Pehler and Craft-Rosenberg, 2009; Valenzuela et al., 2013; Witte, 1985). This caused feelings of isolation and led to social withdrawal. In contrast, young people with CF predominantly felt accepted by their peers (Johannesson et al., 1998; Palmer and Boisen, 2002). This may be due to the fact, that their disease was rather "invisible" for others. In addition to that, individuals with CF tried to hide symptoms and signs to prevent any negative social consequences (Christian and D'Auria, 1997; Johannesson et al., 1998; Pizzignacco and de Lima, 2006). For instance, young people with CF adjusted their cough to the situation they were in and developed a different private and a public coughing behaviour. Keeping secrets, however, contributed to an increased lack of interpersonal closeness. Despite these challenges, individuals with CF had a strong hope for intimate relationships, marriage and even parenthood (Higham et al., 2013; Johannesson et al., 1998). Some young people with MD also reported having romantic experiences, while others felt it was very difficult to have a love life due to their physical disabilities and an associated fear of rejection (Gibson et al., 2014; Gibson et al., 2007). They felt they had nothing to offer to their potential partners which resulted in a lack of openness to make new contacts. Therefore, teenage years for individuals with MD were described as lonely and frustrating (Dreyer et al., 2010).

3.5. Education and career

Most young people with chronic disease attended regular schools and perceived work as an important part of their lives (Badlan, 2006; Gibson et al., 2014). Young people with MD, for instance, were often interested in computer related fields. They expressed difficulties in traditional employment related to various factors such as symptoms of fatigue, architectural barriers and the need for assistance at the workplace (Gibson et al., 2007, 2014). For young people with CF, education and career had a high importance. Most of them succeeded in finding a job, but they needed to put in additional effort to make their work life possible (Gjengedal et al., 2003). Integrating the demanding treatment into their everyday routines was described as most challenging. These therapies included medical, physical and respiratory therapies. At a later point in time many individuals with CF experienced occupational restrictions of some kind and needed to re-evaluate their career choices due to ill health (Higham et al., 2013; Moola and Norman, 2011; Palmer and Boisen, 2002). Young people with SCD, on the contrary, experienced many absences from school because of their recurring hospitalisations due to painful crises (Atkin and Ahmad, 2001). They reported discrimination in education and workplace because of their race and their physical impairments; the majority of individuals with SCD originated from black African or Caribbean families.

"Disability", as a categorization imposed by society, contributed to ambivalent experiences. Young people affected by chronic diseases wanted to be seen as a person and not as a disability (Admi, 1996; Gibson et al., 2007; Pehler and Craft-Rosenberg, 2009). Therefore, they clearly distanced themselves from a disability-identity, wanted more public awareness for their situation and tried to adopt strategies to minimize their disability status (Admi, 1996; Gibson et al., 2007, 2014; Gjengedal et al., 2003; Pizzignacco and de Lima, 2006; Schmitt, 1997).

3.6. Family life

Young people with chronic disease had ambivalent feelings towards their families. On one hand their parents were their best allies and supported them to make everyday life possible. Family relationships were described as strong and supportive (Admi, 1996; Atkin and Ahmad, 2001; Erskine, 2012). On the other hand, there were many tensions between family members. Young people with SCD and CF, for instance, reported that their parents' constant focus on the disease frustrated them and led to strained relationships and disagreements (Atkin and Ahmad, 2001; Bregnballe et al., 2011; Erskine, 2012). Young people wished for a more open parenting style including discussions, trusting relationships, fewer controls and more responsibility.

Living independently was an issue for some of these young people. For individuals with MD moving away from parents was connected with the loss of care by their families and the need to accept care by others (Gibson et al., 2014). The majority of young people with CF left home, but they needed to adapt to the new situation and to create new support strategies (Berge et al., 2007).

3.7. Health situation

The health situation of young people with MD, CF, Haemophilia and SCD was characterized by various physical symptoms and an increased risk for emotional or psychosocial difficulties. Young people with CF suffered from slim physical appearance, frequent coughing and bowel movement problems (Admi, 1996). Furthermore they were at increased risk for infections and respiratory problems (Gjengedal et al., 2003). Puberty was delayed which led to feelings of shame due to an infantile body

(Johannesson et al., 1998). Individuals with SCD suffered from late onset of puberty, too, coming along with small growth and slow body development (Atkin and Ahmad, 2001; Erskine, 2012). Moreover, they suffered from unpredictable painful crisis. Young people affected by MD, in contrast, were most strongly impaired and needed technical assistance such as ventilators and wheelchairs from their early teenage years (Dreyer et al., 2010). As part of the disease progression they lost most of their abilities, not being able to participate in physical activities anymore (Pehler and Craft-Rosenberg, 2009). At some point in time the majority of young people with MD needed assistance in performing daily activities like bathing, dressing and eating (Dreyer et al., 2010). Adolescents with haemophilia were aware of their physical impairments imposed by the disease (Müller-Kägi et al., 2014). They were at risk for bleeding events after injury, surgery or spontaneously that may lead to pain and chronic joint disorders.

Most young people with MD, CF, Haemophilia and SCD experienced mixed or negative feelings such as uncertainty and they worried that their health situation would deteriorate (Cappelli et al., 1989; Gjengedal et al., 2003; Higham et al., 2013). Moreover, adolescence was particularly challenging for these young people as it was characterized by feelings of ambivalence and crisis regarding their identities. This ambivalence was linked to feeling average and extraordinary at the same time (Admi, 1996; Berge et al., 2007; Christian and D'Auria, 1997; Dupuis et al., 2011; Gibson et al., 2007, 2014; Müller-Kägi et al., 2014). Average, because they had the same interests, pursuits and challenges as their healthy peers and extraordinary, because it required additional effort to live their lives.

3.8. Self-management

Chronic diseases come along with lifelong needs for treatment. During transition into adulthood affected young people gradually needed to gain more skills and competence in managing their conditions. Some adolescents reported having ambivalent feelings towards their self-care which led to alternating phases of adherence and non-adherence (Admi, 1996; Higham et al., 2013; Müller-Kägi et al., 2014). A major contributing factor for non-adherence was using a therapy that marked out their difference from their peers. Therefore, they preferred flexible adherence to treatment as opposed to strict adherence or non-adherence. From testing out their boundaries and making mistakes themselves, young people learned what was good for them. Although they knew it might not be good for their health, some young people engaged in substance abuse (alcohol, nicotine) for a period of time during adolescence (Berge et al., 2007; Palmer and Boisen, 2002).

3.9. Transfer to adult care

The transfer from paediatric to adult care services that was generally scheduled for late adolescence caused mixed feelings (Porter et al., 2014; Telfair et al., 1994, 2004). Young people did not feel prepared for the transfer and reported a lack of information (Al-Yateem, 2012). In addition, they were afraid of losing health professionals whom they trusted and envisioned professionals used to caring for adults as less competent in managing their disease. Some young people were reluctant to meet the health professional alone as they were used to being accompanied by their relatives (Tuchman et al., 2008). However, after the transfer to adult care they felt empowered to separate from their families, having greater control over and being more involved in decision making.

(2) The parent perspective

Parents were strongly involved in the life and care of their chronically ill children. In addition to normal parenting activities they assisted in carrying out activities of daily life (e.g. body care) and disease management (Antle et al., 2008; Dupuis et al., 2011). Some parents reported assisting their children with physical activities that they could still do independently. However, the transition into adulthood and the transfer from paediatric to adult care, respectively, implied a parental role change. As children were growing older their needs changed. Some parents felt it was challenging to step back and let their children become more independent (Bregnballe et al., 2011; Hauser and Dorn, 1999; van Staa et al., 2011). They felt it was difficult to give them more responsibility and the opportunity to make their own decisions regarding life and health. Parents reported wanting the best for their children and being highly concerned about their future (Antle et al., 2008; Cappelli et al., 1989; Dupuis et al., 2011; Gjengedal et al., 2003; Moola and Norman, 2011). Most parents were constantly aware of the progressive nature of the disease which led to continuous feelings of distress. They worried about their children's health situation and their worst fear was the deterioration of health before death. Parents worried about their child's ability to engage in a social life, have an education and career and to handle finances (Antle et al., 2008; Parkyn and Coveney, 2013). Parents were concerned about their children's ability to adhere to treatment and manage their disease themselves as required in adult life (Porter et al., 2014; Telfair et al., 1994; van Staa et al., 2011).

(3) The sibling perspective

The information about siblings experiences in the literature selected for this review was limited. The investigation nevertheless revealed that siblings were also concerned about their ill sister's and brother's skills in managing their disease. They worried about their siblings' ability to take care for themselves in future (Porter et al., 2014). Furthermore, they were concerned about the transfer to adult care, because they believed there would be a less supportive atmosphere as in paediatric services.

4. Discussion

All studies reported that the transition into adulthood was perceived as a challenging developmental stage by the affected young person and by his or her family. How families coped and adapted to these challenges were illustrated from three perspectives, the affected young

person, the parents and the siblings, providing new insights into the individual family members experience.

However, the studies were limited in revealing detailed information at a family systems level, comprising family beliefs, effects on relationships and pattern of interaction. Thus, it is still unclear, how values and beliefs and the family members' life situation influenced family interaction and functioning during the affected young person's transition into adulthood. Findings suggest that in the course of adolescence families need to renegotiate their relationships (McGoldrick et al., 2013). Parents, for instance, need to develop an adult to adult relationship with their children. According to the findings of this review overprotective parenting behaviour was found to result in strained relationships, disagreements and ambivalent feelings towards each other. Nevertheless, other effects on the family systems, such as the parental dyad experience and siblings' relationships have not been the focus of previous research. Therefore, a more detailed investigation of the family experience, belief systems, family relationships and interaction pattern, and a description of major challenges and coping strategies at a family level is needed.

In the case of genetic childhood illness, individual family members experience specific challenges when an ill family member is transitioning into adulthood (Miller et al., 2006; Rolland and Williams, 2005; Segrin and Flora, 2011). As this review showed, parents' and sibling's psychological and emotional wellbeing was challenged by the young persons' condition, as they were concerned about their ill relatives' health and lives and as a consequence experienced negative emotions. In addition, young people living with the disease described several areas of challenging or difficult situations which were likely to have a psychosocial and emotional impact on their lives. Differentiation and establishment of a personal identity in relation to the family of origin, community and larger society, however, are essential steps that allow identity formation and enable a successful transition into adulthood and adult life (McGoldrick et al., 2013). Yet many of the young people were threatened by recurring feelings of ambivalence and confusion about their own identity and difficulties in having social relations and in becoming more independent from their families, which has been observed for other chronic childhood conditions (Drotar, 1981; Michaud et al., 2007; Sobralske, 2013; Taylor et al., 2008).

In addition, this study revealed that the development of the young individual's social skills and socializing in general were challenged by negative experiences at school, work place or through discrimination by wider society. Existing theoretical and empirical knowledge about the impact of negative social experiences on the lives of individuals living with chronic childhood conditions support this finding (Michaud et al., 2007; Mietzel, 2002; Sawyer et al., 2007).

The current review adds to the literature by considering the visibility of the condition as an influencing factor on the social experience. Young people with less visible impairments tried to hide and not disclose their disease which led to problematic social relationships. This indicates that the external visibility of the disorder affects how the disability is integrated into the person's own identity, adapting authentic behaviour within social interactions is less problematic when the disability is more visible. Evidence from the wider literature confirms that children with mild disabilities show a higher degree of conspicuous behaviour compared to more severely handicapped individuals with more evident disabilities (Leyendecker and Gebhard, 2005). However, the findings from the current review could not support a relationship between the extent of physical disability due to disease and social functioning.

Furthermore, parents who managed their children's health and assisted in activities of daily life since their child's birth, experienced difficulties with detachment and were at risk of overprotecting their growing children. In parents of children with cancer, asthma, diabetes or cystic fibrosis parental overprotection of children has been observed with no differences across disease groups (Hullmann et al., 2010). It is suggested that families with adolescents or emerging young adults need to increase the flexibility of family boundaries to permit development of children's independence and parents need to develop new adult relationships with their children (McGoldrick et al., 2013). The development of new family relationships and communication patterns influences individual personal development and family functioning. Families put much effort into meeting the developmental needs that allow their affected children to become independent with age, but their situation means that they have to allow for the development of psychological independence, while the young person is becoming more physically dependent. The struggles parents have in changing their behaviour may be understood by the social, psychological and physical challenges in having to adjust to an altered developmental trajectory different from that usually anticipated for children and parents.

Health management including transfer to adult care and development of self- management skills is described as unsatisfactory and problematic. Adherence to treatment, for example, came along with the disclosure of the disease and marked out young people's difference from their peers. Therefore, individuals with CF and haemophilia negotiated their need to be similar to their peers with their need for adhering to treatment in order to maintain their well-being (Müller-Kägi et al., 2014; Admi, 1996; Higham et al., 2013). As a consequence, poor self-management led to poor disease control. The transfer to adult care and adherence to treatment were described as particularly difficult for chronically ill young individuals living with many other chronic illnesses, too (Elliot, 2015; Sobralske, 2013; Taddeo et al., 2008). Greater socio-psychological support to facilitate the transition of young people into adulthood for the young person affected and their family is therefore essential if they are to manage their genetic condition effectively.

Overall, the young person's perspective has received more research attention than that of his or her parents or siblings. Most studies concerned individuals living with CF or SCD whereas families affected by neuromuscular diseases and haemophilia have not been researched as intensively until more recently.

This literature review revealed little information on the family systems and sibling's perspective. A recent review investigating the experiences of siblings of children with various chronic illnesses confirmed the lack of knowledge on the sibling's perspective (Knecht et al., 2015).

4.1. Implications for practice

Several areas emerged from this systematic review, which require careful attention by health professionals. To begin, the young persons' and his or her family's situation includes multiple challenges that require a multifaceted understanding of human development, family interaction and a complex disease management to allow setting the right priorities at the right time.

For this reason, young people transitioning into adulthood and their families would benefit from continuous support to assess how their needs and coping strategies change over time. However, family centred transitional care is still at an early stage. Family centred transitional care needs to enable the family system to be more flexible and to adapt to developmental changes and new health situations. Moreover, psychological health needs to be improved to prevent mental illness and young people need to be supported to be more independent if they wish so. Advanced practice nurses or general practitioners are in a good position to monitor both the young person's and his or her families' health and life situation, to coordinate specialist and interdisciplinary interventions and to help these families to prepare for the transition allowing them to live their lives with less focus on the disease and its management.

Thus, to improve the situation of these families, family centred, continuous and coordinated care is recommended, delivered by a multidisciplinary team consisting of trained and experienced health professionals regarding rare diseases.

4.2. Implications for research

Young people's successful transition into adulthood with a chronic childhood disease is challenging. Parents and siblings are at risk for negative consequences themselves. It is difficult to gauge the full extent of distress and socio-psychological difficulties that families experience with less information on the parents and on the siblings' perspective, although they are clearly present. Furthermore, the focus on the individual experience and lack of a family focus prevents us knowing more about the effect of family communication or interaction, any relationship struggles and the family's adaptability and cohesion. Thereby this literature review demonstrates the need for further research into the family's experiences to explore what the challenges are and how family members can be assisted in coping and adapting to the complex situation when a child or sibling is affected by a specific chronic condition. This knowledge is pivotal to promote families functioning, health and well-being, minimize caregiver burden, and as a consequence, reducing the poor socio-psychological situation that families report whilst limiting health related financial cost increase.

4.3. Strengths and limitations

The strengths of this literature review lie in the systematic and methodological approach that included a broad range of evidence integrating qualitative and quantitative research. Moreover, the search, the study selection process, the quality assessment and the data extraction and analysis of articles have been conducted by two independent and experienced researchers which contribute to the quality of this systematic literature review. This study limited its scope to chronic childhood disease with physical disability and excluded conditions that come along with cognitive impairment. Although experiences may be different according to intellectual abilities, this can be seen as a limitation.

5. Conclusions

Young people and their family members experience multiple challenges during the young persons' transition into adulthood. Failure to successfully negotiate this developmental stage may have consequences not only for the ill individual but also for the whole family. Thus, a family systems perspective to research and care is indicated to ensure that these individuals receive the best support available to cope with their complex life and health situation.

Authorship

All authors of this article assure to fulfil the criteria of authorship and confirm that the manuscript has not been published or considered elsewhere. There is no one else who fulfils the criteria but has not been included as an author.

Conflict of interests

There are no competing interests.

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Contributions of the authors

Conducting the search (VW and AM); Critical appraisal of the studies (VW and CP); Data analysis (VW and RM).

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Appendix 2.1: Study leaflet and study summary

Zürcher Hochschule für Angewandte Wissenschaften
Institut für Pflege

Florence Nightingale School of Nursing and Midwifery
King's College London

Erwachsenwerden mit einer neuromuskulären Erkrankung

Eine Untersuchung von Erfahrungen junger Menschen, die mit einer neuromuskulären Erkrankung leben und deren Familien

Erwachsenwerden: Erfahrungen von Menschen mit neuromuskulären Erkrankungen und deren Familien

Doktorarbeit von
Veronika Waldboth, MScN, RN; Zürcher Hochschule für Angewandte Wissenschaften

Prüferin der Studie
Prof Dr. Romy Mahrer-Imhof, PhD, RN; Zürcher Hochschule für Angewandte Wissenschaften

Teilnehmerinnen und Teilnehmer für wissenschaftliche Studie gesucht

Für eine wissenschaftliche Studie suchen wir junge Menschen, die mit einer neuromuskulären Erkrankung leben und ihre Familien. Eingeschlossen werden Familien und Einzelpersonen, die folgende Einschlusskriterien erfüllen:

- Junge Menschen im Alter von 14 bis 30 Jahren, die mit einer neuromuskulären Erkrankung leben, die bereits im Kindesalter aufgetreten ist und mit einer starken körperlichen Einschränkung einhergeht (z.B. Muskeldystrophie oder Spinale Muskelatrophie);
- Eltern, Geschwister und nahestehende Angehörige von jungen Menschen, wenn diese 14 bis 30 Jahre alt sind und mit einer neuromuskulären Erkrankung leben, die im Kindesalter aufgetreten ist und mit einer körperlichen Einschränkung einhergeht (z.B. Muskeldystrophie oder Spinale Muskelatrophie).

Ziel der Studie

Wir möchten Erfahrungen von jungen Menschen und deren Familien erfassen, die spezifisch für den Übergang vom Kindes- zum Erwachsenenalter sind, um

- die Herausforderungen zu verstehen, mit denen betroffene Familien während dieser Entwicklungsphase konfrontiert sind;
- Strategien zu identifizieren, mit denen sich betroffene Familien erfolgreich an veränderte Situationen anpassen;
- Faktoren zu identifizieren, die die Anpassung an veränderte Situationen beeinflussen und
- Erfahrungen zu verstehen, die betroffene Familien mit dem Schweizerischen Gesundheitssystem machen.

Ablauf und Dauer der Studie

Die Studie beinhaltet ein gemeinsames Gespräch mit einer wissenschaftlichen Mitarbeiterin. Jeder Teilnehmende wird zu einem Einzelgespräch eingeladen, das ungefähr eine Stunde dauert. Der Ort des Interviews kann vom Teilnehmenden bestimmt werden und im familiären Umfeld oder an einem neutralen Ort stattfinden. Die Teilnahme ist freiwillig und alle Daten werden vertraulich behandelt. Für die Teilnehmenden ergibt sich kein direkter medizinischer Nutzen.

Falls Sie an einer Studienteilnahme interessiert sind und oben genannte Kriterien auf Sie zutreffen, nehmen Sie bitte Kontakt mit der Studienleiterin, Frau Veronika Waldboth, auf. Dazu benutzen Sie nachfolgende Kontaktdaten (Website, Email oder Telefon) oder senden den beigelegten Tallon.

Kontakt:

Veronika Waldboth
Zürcher Hochschule für Angewandte Wissenschaften
Institut für Pflege
Technikumstrasse 71, Postfach
8401 Winterthur, Schweiz

Information und Anmeldung über die Projektwebsite:

www.gesundheit.zhaw.ch/transition

Tel. direkt: +41 58 934 6499
Email: veronika.waldboth@zhaw.ch

Bitte nehmen Sie zur Kenntnis, dass Ihre Daten bei Zustandekommen eines Kontakts mit Frau Waldboth registriert werden. Sollten Sie an einer Studienteilnahme nicht interessiert sein, teilen Sie uns dies mit und Ihre Daten werden unverzüglich gelöscht.
Wünschen Sie weitere Informationen, wenden Sie sich bitte ebenfalls an Frau Waldboth.

Winterthur, 29.06.2015

Erwachsenwerden mit einer neuromuskulären Erkrankung (Teilnahme-Tallon)

Bitte kreuzen Sie zutreffendes an:

Ich bin:

□ ein Jugendlicher oder junger Erwachsener, der mit einer neuromuskulären Erkrankung lebt und zwischen 14 bis 30 Jahre alt ist;

□ ein Vater eines betroffenen Jugendlichen oder jungen Erwachsenen;

□ eine Mutter eines betroffenen Jugendlichen oder jungen Erwachsenen;

□ ein Geschwister eines betroffenen Jugendlichen oder jungen Erwachsenen;

□ Andere Bezugsperson: _____.

Ich erwäge eine Teilnahme an der Studie und erkläre mich damit einverstanden, dass ich kontaktiert werde, um nähere Informationen zur Studie „Erwachsenwerden mit einer neuromuskulären Erkrankung“ zu erhalten, sowie eine mögliche Teilnahme an der Studie zu besprechen.

Kontaktdaten zu meiner Person:

Name	
Vorname	
Telefonnummer	
Adresse	
Emailadresse	
An diesem Tag / Uhrzeit bin ich gut erreichbar	

Datum	
Unterschrift	

Winterthur, 29.06.2015

Zürcher Hochschule für Angewandte Wissenschaften
Institut für Pflege

Florence Nightingale School of Nursing and Midwifery
King's College London

Erwachsenwerden mit einer neuromuskulären Erkrankung

Eine Untersuchung von Erfahrungen junger Menschen, die mit einer neuromuskulären Erkrankung leben und deren Familien

Diese Studie untersucht Erfahrungen von jungen Menschen, die mit einer neuromuskulären Erkrankung leben und ihrer Familien während des Übergangs vom Kind zum Erwachsenen. Eine chronische Krankheit, die sich im Kindesalter manifestiert und mit starken körperlichen Einschränkungen einhergeht kann diese Entwicklungsphase stören und sich auf die betroffenen Jugendlichen und jungen Erwachsenen, sowie auf deren Familien auswirken. Daher möchte diese Studie die Erfahrungen der betroffenen Familien untersuchen und verstehen, mit welchen Herausforderungen sie in dieser Lebensphase umgehen und welche Strategien sie anwenden, um mit veränderten Situationen zurecht zu kommen.

Studienziele

Das Ziel dieser Studie ist es, die Erfahrungen von Jugendlichen und jungen Erwachsenen und ihrer Familien zu verstehen.
Zu den Feinzielen gehören:

- Die Herausforderungen zu verstehen, mit denen betroffene Familien während dieser Entwicklungsphase konfrontiert sind;
- Strategien zu identifizieren, mit denen sich betroffenen Familien erfolgreich an veränderte Situationen anpassen;
- Faktoren zu identifizieren, die die Anpassung an veränderte Situationen beeinflussen;
- Erfahrungen der betroffenen Familien mit dem Schweizerischen Gesundheitssystem zu verstehen.

Methodisches Vorgehen

Diese pflegewissenschaftliche Studie hat einen qualitativen Ansatz. Daten werden durch Einzelgespräche mit den Jugendlichen und jungen Erwachsenen, deren Eltern, jüngeren und älteren Geschwistern und anderen nahestehenden Bezugspersonen erhoben. Die Anzahl der Teilnehmenden wird vorab auf ungefähr 8 bis 15 betroffene Familien geschätzt.

Eingeschlossen werden Familien und Einzelpersonen, die folgende Einschlusskriterien erfüllen:

- Junge Menschen im Alter von 14 bis 30 Jahren, die mit einer neuromuskulären Erkrankung leben, die bereits im Kindesalter aufgetreten ist (0-10 Jahre) und mit einer mässigen bis starken körperlichen Einschränkung einhergeht;
- Eltern, jüngere und ältere Geschwister (ab 8 Jahren) und nahestehende Angehörige von jungen Menschen, wenn diese 14 bis 30 Jahre alt sind und mit einer neuromuskulären Erkrankung leben, die im Kindesalter aufgetreten ist und mit einer körperlichen Einschränkung einhergeht;
- Teilnehmende, die in der Schweiz ansässig sind.

Nicht teilnehmen hingegen dürfen Personen

- die kein Deutsch sprechen
- oder die mit einer ausgeprägten geistigen Einschränkung leben, die es der Person nicht ermöglicht zur Teilnahme an der Studie einzuwilligen.

Die Studie beinhaltet ein gemeinsames Gespräch mit einer wissenschaftlichen Mitarbeiterin, das ungefähr eine Stunde dauert. Der Ort des Interviews kann vom Teilnehmenden bestimmt werden und Interviews können im familiären Umfeld oder an einem neutralen Ort stattfinden. Die Gespräche werden auf Tonband aufgenommen, abgeschrieben und Namen von Personen werden verschlüsselt. Daraufhin werden die Tonbandaufnahmen gelöscht. Die Teilnahme an einem Interview ist freiwillig und alle Daten werden vertraulich behandelt.
Mit der Durchführung des Interviews endet die Studienteilnahme des Teilnehmenden. Die Durchführung der Gespräche mit den Betroffenen und deren Familien oder nahestehenden Angehörigen ist für den Zeitraum von November 2014 bis Dezember 2015 geplant.

Nutzen und Risiken
Für die Versuchspersonen ergibt sich kein direkter medizinischer Nutzen. Die Teilnahme an dieser Studie sollte für die Teilnehmenden kein Risiko darstellen.

Persönlicher Hintergrund
Diese Studie ist Veronika Waldboth's Doktorarbeit in Pflege, die sie in Zusammenarbeit mit der Zürcher Hochschule für Angewandte Wissenschaften (ZHAW) und der Florence Nightingale School of Nursing and Midwifery am King's College London umsetzt. Die Supervisoren dieser Studie sind Prof Dr. Alison Metcalfe und Dr. Christine Patch vom King's College in London und Prof Dr. Romy Mahrer-Imhof von der ZHAW in Winterthur. Frau Waldboth ist Pflegefachfrau und hat Berufserfahrung im akutmedizinischen Bereich. Seit Abschluss ihres Masterstudiums in Pflegewissenschaft im Jahr 2012 ist sie als wissenschaftliche Mitarbeiterin am Institut für Pflege an der ZHAW angestellt.

Frau Waldboth's Interesse an dieser Thematik gründet auf persönlichen Erfahrungen mit einem Familienmitglied, das mit einer Muskeldystrophie lebt. Genetisch bedingte neuromuskuläre Erkrankungen, die sich im Kindesalter manifestieren, betreffen die ganze Familie. Der Einbezug der Eltern, Geschwister und anderer Bezugspersonen ist ihr daher ein zentrales Anliegen.

Falls sie an einer Studienteilnahme interessiert sind und oben genannte Kriterien auf Sie zutreffen, nehmen Sie bitte Kontakt mit Frau Waldboth, auf. Dazu benutzen Sie folgende Kontaktdaten:

Kontakt
Veronika Waldboth
Zürcher Hochschule für Angewandte Wissenschaften
Institut für Pflege
Technikumstrasse 71, Postfach
8401 Winterthur, Schweiz

Projektwebsite**:**

www.gesundheit.zhaw.ch/transition

Tel. direkt: +41 58 934 6499
Email: veronika.waldboth@zhaw.ch

Appendix 2.2: Approval by the Ethics Committee and confirmation letter

Zurich University
of Applied Sciences

**School of
Health Professions**

Nursing

Prof
Alison Matcalfe
Florence Nightingale School of Nursing and Midwifery
King's College London
James Clerk Maxwell Building
57 Waterloo Road
London SE1 8WA

Prof. Dr. Romy Mahrer Imhof
PhD, RN
Program Director MScN

romy.mahrer@zhaw.ch

TN03.13
Technikumstrasse 71
Postfach
CH-8401 Winterthur

Phone +41 58 934 63 44
Fax +41 58 935 63 44

Phone Main office +41 58 934 71 71
Fax Main office +41 58 935 71 71

www.gesundheit.zhaw.ch

Winterthur, November 3rd, 2014
Our reference: mahr

Ethic committee approval for the PhD study of Mrs. Veronica Waldboth "Becoming an adult: Experiences of individuals affected by neuromuscular disease and their families"

Dear Alison,

I hereby confirm that the Ethics Committee of the county of Zurich has approved the study of Veronica Waldboth "Becoming an adult: Experiences of individuals affected by neuromuscular disease and their families". The study registered unter KEK-ZH 2014-0225 received the Nihil Obstat September 25th, 2014.

Sincerely

Zurich University of Applied Sciences

Prof. Dr. Romy Mahrer Imhof

Kanton Zürich
Kantonale Ethikkommission

Prof. Dr. med. Dr. phil. Paul Hoff
Präsident Abteilung D

Niklaus Herzog, lic. iur. et theol.
Juristischer Sekretär
Stampfenbachstrasse 121
Postfach
8090 Zürich
Telefon +41 44 384 33 64
Fax +41 44 384 25 06
katharina.baertschi@puk.zh.ch
www.kek.zh.ch

ZHAW
Institut für Pflege
Frau Prof. Dr. R. Mahrer-Imhof
TN 03.13
Technikumstr. 71
8401 Winterthur

14. Januar 2015

Erfüllung der Auflagen
KEK-ZH-Nr: 2014-0225
Becoming an adult: Experiences of individuals affected by neuromuscular disease and their families

Sehr geehrte Frau Prof. Mahrer-Imhof

Wir beziehen uns auf unsere Beschlussmitteilung mit Auflagen vom 25.09.2014 und Ihr E-Mail/Schreiben vom 12.01.2015 und die darin aufgelisteten Dokumente zur obgenannten Studie.

Wir teilen Ihnen mit, dass die in der Beschlussmitteilung vom 25.09.2014 formulierten Auflagen mit Ihrem E-Mail/Schreiben vom 12.01.2015 und den beigefügten Dokumenten erfüllt sind.

Freundliche Grüsse

Paul Hoff

Niklaus Herzog

Kanton Zürich
Kantonale Ethikkommission

Prof. Dr. med. Dr. phil. Paul Hoff
Präsident Abteilung D

Niklaus Herzog, lic. iur. et theol.
Juristischer Sekretär
Stampfenbachstrasse 121
Postfach
8090 Zürich
Telefon +41 44 384 33 64
Fax +41 44 384 25 06
katharina.baertschi@puk.zh.ch
www.kek.zh.ch

Einschreiben
ZHAW
Institut für Pflege
Frau Prof. Dr. R. Mahrer-Imhof
TN 03.13
Technikumstr. 71
8401 Winterthur

25. September 2014

Beschlussmitteilung der Kantonalen Ethikkommission Zürich

Gesuch KEK-ZH-Nr. 2014-0225
Becoming an adult: Experiences of individuals affected by neuromuscular disease and their families

Gesuchsteller Prof. Dr. Romy Mahrer-Imhof

Zentren

I. Verfahren

☐ ordentliches Verfahren ☒ vereinfachtes Verfahren ☐ präsidiales Verfahren

II. Entscheid

☐ **Die Bewilligung wird erteilt**

Bedeutet: Das Vorhaben gemäss bewilligtem Forschungsplan kann gestartet und im Rahmen der anwendbaren rechtlichen Bestimmungen durchgeführt werden.

Bewilligungen für **klinische Versuche der Kategorie B und C** stehen unter dem **Vorbehalt**, dass

1. allfällig durch die zuständige eidgenössische Zulassungsbehörde (Swissmedic/BAG) festgestellte Mängel keine Änderungen der von der Ethikkommission evaluierten Unterlagen erfordern, und dass
2. die Bewilligung der eidgenössischen Zulassungsbehörde (Swissmedic/BAG) vorliegt.

☒ **Die Bewilligung wird mit Auflagen erteilt**

Bedeutet: Das Vorhaben gemäss bewilligtem Forschungsplan kann gestartet und im Rahmen der anwendbaren rechtlichen Bestimmungen durchgeführt werden. Die Auflagen sind innert angemessener Frist zu erfüllen. Die revidierten Dokumente werden nach Einreichung im präsidialen Verfahren geprüft.

Folgende Auflagen müssen erfüllt werden:

- Das Studienprotokoll ist bei der nächsten Überarbeitung noch wie folgt zu korrigieren: Unter 5.4, Abschnitt 2, sind die Altersangaben für die 3 Teilnehmergruppen in Entsprechung von Art. 3 lit. j und k HFG anzupassen („young persons **14**-17 years, children < **14** years, children under the age of **14**"). Bitte reichen Sie diese angepasste Protokollversion für unser Dossier bei Gelegenheit nach. Anmerkung: Dies ist keine (kostenpflichtige) substantielle Änderung an der Studie.

☐ **Gegenwärtig kann die Bewilligung noch nicht erteilt werden**

Bedeutet: Das Vorhaben kann **noch nicht** gestartet werden. Die nachfolgenden Bedingungen sind zu erfüllen. Die revidierten Dokumente werden nach Einreichung von der Ethikkommission geprüft.

Folgende Bedingungen müssen erfüllt werden:

☐ **Die Bewilligung wird nicht erteilt**

Bedeutet: Das Vorhaben kann in der vorliegenden Form nicht durchgeführt werden. Eine Neueinreichung ist möglich.

☐ **Auf das Gesuch wird nicht eingetreten**

Bedeutet: Die Ethikkommission ist für die Beurteilung rechtlich nicht zuständig. Entweder ist eine andere Stelle für die Bewilligung zuständig, oder das Vorhaben kann ohne Bewilligung durchgeführt werden.

☐ **Das Verfahren wird infolge Gegenstandslosigkeit abgeschrieben**

Bedeutet: Das Verfahren wird wegen Rückzugs des Gesuchs oder anderen Gründen gegenstandslos.

☐ **Das Verfahren wird sistiert**

☐ **Die Bewilligung wird entzogen**

III. Einteilung

☐ **Das Vorhaben gilt als klinischer Versuch gemäss KlinV**

☐ Kategorie ☐ A ☐ B ☐ C
☐ mit Arzneimitteln
☐ mit Medizinprodukten
☐ mit Transplantatprodukten
☐ der Gentherapie
☐ mit gentechnisch veränderten oder pathogenen Organismen
☐ der Transplantation
☐ anderer klinischer Versuch gemäss 4. Kapitel KlinV
☐ Umkategorisierung gemäss Art. 71 Abs. 3 KlinV, Kategorie ☐ A ☐ B ☐ C
☐ mit Strahlenquellen

☒ **Das Vorhaben gilt als Forschungsprojekt gemäss HFV**

☒ Forschung mit Personen, Kategorie ☒ A ☐ B

☐ Umkategorisierung gemäss Art. 48 Abs. 2 HFV, Risiko-Kategorie ☐ A ☐ B

☐ mit Strahlenquellen

☐ Weiterverwendung biologischen Materials und/oder gesundheitsbezogener Personendaten

☐ Forschung mit verstorbenen Personen

☐ Forschung an Embryonen und Föten einschliesslich Totgeburten

☐ **Weiterverwendung ohne vorbestehende Einwilligung (Art. 34 HFG, Art. 37-40 HFV)**

a. Verwendungszweck

b. Bezeichnung des biologischen Materials/Personendaten

c. zur Weitergabe berechtigter Personenkreis

d. zur Entgegennahme berechtigter Personenkreis

☐ **Multizentrisches Forschungsprojekt**

☐ BE ☐ NZ ☐ GE ☐ SG ☐ TI ☐ VD ☐ ZH ☐ VS ☐ TG

IV. Begründung

Die Ethikkommission stützt ihre Begründung auf die Unterlagen, wie sie aufgeführt sind:

☒ in der/den beiliegenden Checkliste/n unterschrieben am 25.09.2014

☐ in der /den Stellungnahme/n der Kantonalen Ethikkommission/en:

☒ im Beschluss der Kantonalen Ethikkommission Zürich vom 08.09.2014

☐ sowie auf

V. Kosten

Die Gebühren betragen CHF 500.00 (Diss.)[1].

[1] Art. 3 Gebührenreglement swissethics 2014

VI. Rechtsmittelbelehrung

Gegen diesen Beschluss kann innert 30 Tagen, von der Mitteilung an gerechnet, beim Regierungsrat des Kantons Zürich schriftlich Rekurs eingereicht werden. Die Rekursschrift muss einen Antrag und dessen Begründung enthalten. Der angefochtene Entscheid ist beizulegen oder genau zu bezeichnen. Die angerufenen Beweismittel sind genau zu bezeichnen und soweit möglich beizulegen.

VII. Mitteilung an den Gesuchsteller

und in Kopie an:

- ☐ **Sponsor**
- ☐ **Swissmedic**
- ☐ **BAG**
- ☐ **beteiligte, lokale EKs (multizentrische Studien)**
- ☐ **andere:**

VIII. Zusammensetzung der am Entscheid beteiligten Kommission

					am Beschluss beteiligt		
						nein	
	Name, Vorname	Berufliche Stellung / Titel	m	f	ja	abwesend	In Ausstand
Vorsitz	Hoff, Paul	Prof. Dr. med. Dr. phil.	☒	☐	☐	☐	☐
Mitglieder	Geschwindner, Heike	Pflegewissenschaftlerin, PhD,Msc	☐	☒	☒	☐	☐
	Jokeit, Henric*	Prof. Dr. rer. nat. Dipl.-Psych.	☒	☐	☐	☐	☐
	Keller, Emanuela	Prof. Dr. med.	☐	☒	☐	☐	☐
	Künig, Gabriella	PD Dr. med.	☐	☒	☐	☐	☐
Stv. Vorsitz	Möhler, Hanns	Pharmakologe, Prof. Dr.	☒	☐	☐	☐	☐
	Schmid Büchi, Silvia	Pflegewissenschaftlerin, Dr.	☐	☒	☐	☐	☐
	Schneebeli, Margrit	Pflegefachfrau	☐	☒	☒	☐	☐
	Schweizer, Esther	Pfarrerin	☐	☒	☒	☐	☐
	Siegrist, Michael	Prof. Dr. phil.	☒	☐	☐	☐	☐
für Biometrie zuständiges Mitglied *	Vischer, Daniel	Rechtsanwalt, lic. iur.	☒	☐	☐	☐	☐
	Züst, Barbara	lic. iur., Anästhesiepflegefachfrau	☐	☒	☐	☐	☐

Für die Ethikkommission:

H. Möhler

Hanns Möhler

Niklaus Herzog

Niklaus Herzog

Bemerkungen

Registrierungspflicht

Nach der Erteilung der Bewilligung durch die Ethikkommission muss der Sponsor den klinischen Versuch in einem von der Weltgesundheitsorganisation (WHO) anerkannten Primär-Register (www.who.int) oder im Register der Nationalen Medizinbibliothek der Vereinigten Staaten von Amerika (www.clinicaltrials.gov) registrieren. Mit dieser Erst-Registrierung erhält der Sponsor für den klinischen Versuch eine Identifikationsnummer, die er bei der anschliessenden Zweit-Registrierung im Schweizer Register „Swiss National Clinical Trials Portal" (SNCTP, vgl. www. kofam.ch) angeben muss. Im Schweizer Register sind die Rubriken „Ergänzende Datenbank" sowie „Organisationen" auszufüllen. Die Informationen über den klinischen Versuch sind in beiden Registern öffentlich zugänglich.

Die Kantonale Ethikkommission Zürich bestätigt, dass sie nach ICH-GCP arbeitet.

Vorgehen zur Einreichung revidierter Dokumente

- Revidierte Unterlagen sind der Ethikkommission digital zusammen mit der aktualisierten Checkliste zuzustellen (sowohl bereinigte als auch nicht beanstandete Dokumente sind auf der Checkliste aufzulisten).
- Die Änderungen sind in den revidierten Dokumenten zu markieren.
- Die revidierten Dokumente sind auch weiteren involvierten Zulassungsbehörden zuzustellen, sofern sie von diesen für die Bewilligung benötigt werden.

Meldungen und Berichterstattung an die Ethikkommission siehe Anhang 1 und Anhang 2

Anhang 1
Meldungen und Berichterstattung an die Ethikkommission ab 1. Januar 2014 für klinische Versuche (KlinV)

Meldung von Sicherheits- und Schutzmassnahmen
siehe Art. 37 KlinV:
Meldung an die EK innerhalb von 7 Tagen
Versuche mit Medizinprodukten: innerhalb von 2 Tagen

Abschluss, Abbruch oder Unterbruch des klinischen Versuchs
siehe Art. 38 KlinV:
Abschlussmeldung an die EK innerhalb von 90 Tagen
Abbruch- oder Unterbruchmeldung an die EK innerhalb von 15 Tagen
Schlussbericht an die EK: innerhalb 1 Jahres nach Abschluss/Abbruch

Schwerwiegende unerwünschte Ereignisse (Serious Adverse Events, SAE) bei klinischen Versuchen mit Arzneimitteln
Siehe Art. 40 KlinV:
Falls gemäss Protokoll nicht anders vorgesehen SAE mit Todesfolge innerhalb von 7 Tagen (an lokale EK nur lokale Ereignisse, an Leit-EK alle Ereignisse in der CH).

Verdacht auf eine unerwartete schwerwiegende Arzneimittelwirkung (Suspected Unexpected Serious Adverse Reaction, SUSAR)
Siehe Art. 41 KlinV:
SUSAR mit Todesfolge innerhalb von 7 Tagen, sonstige SUSARs innerhalb von 15 Tagen (an lokale EK nur lokale Ereignisse, an Leit-EK alle Ereignisse in der CH).

Schwerwiegende unerwünschte Ereignisse (Serious Adverse Events, SAE) bei klinischen Versuchen mit Medizinprodukten
Siehe Art. 42 KlinV:
Bei Versuchen der Kategorie C SAE bei Verdacht auf Zusammenhang mit Prüfprodukt oder erfolgtem Eingriff innerhalb von 7 Tagen (an lokale EK nur lokale Ereignisse, an Leit-EK alle Ereignisse in der CH).

Schwerwiegende unerwünschte Ereignisse (Serious Adverse Events, SAE) mit möglichem Zusammenhang zu untersuchter Intervention bei übrigen klinischen Versuchen
Siehe Art. 63 KlinV:
Meldung an EK innerhalb von 15 Tagen.

Berichterstattung über die Sicherheit der teilnehmenden Personen
Siehe Art. 43 KlinV:
1 mal jährlich Auflistung der Ereignisse weltweit (Annual Safety Report)

Mit dem jährlichen Sicherheitsbericht sind der EK auch alle Änderungen zu melden, die nicht bewilligungspflichtig sind (d.h. alle Änderungen, die gemäss Art. 29 KlinV nicht als wesentliche gelten).

Anhang 2
Meldungen und Berichterstattung an die Ethikkommission ab 1. Januar 2014 für Forschungsprojekte mit Ausnahme der klinischen Versuche (HFV)

Forschung mit Personen, die mit Massnahmen zur Entnahme biologischen Materials oder zur Erhebung gesundheitsbezogener Personendaten verbunden

Sicherheits- und Schutzmassnahmen siehe Art. 20 HFV
Meldung an die EK innerhalb von 7 Tagen

Schwerwiegende Ereignisse siehe Art. 21 HFV
Meldung innerhalb von 7 Tagen (an lokale EK nur lokale Ereignisse, an Leit-EK alle Ereignisse in der CH) und Unterbruch des Forschungsprojektes.

Abschluss und Abbruch des Forschungsprojekts siehe Art. 22 HFV
Meldung an die EK innerhalb von 90 Tagen

Weiterverwendung biologischen Materials und gesundheitsbezogener Personendaten für die Forschung

Siehe Art. 36 HFV:
Wechsel Projektleitung: Meldung an die EK: vorgängig

Abschluss und Abbruch des Forschungsprojekts
Meldung an die EK innerhalb von 90 Tagen

Weiterverwendung biologischen Materials und gesundheitsbezogener Personendaten für die Forschung bei fehlender Einwilligung und Information nach Artikel 34 HFG

Siehe Art. 40 HFV:
Änderungen der in der Bewilligung genannten Angaben
Meldung an die EK (vorgängig)

Abschluss oder Abbruch des Forschungsprojekts
Meldung an die EK innerhalb von 90 Tagen

Forschung an verstorbenen Personen (Art. 43 HFV)

Siehe Art. 43 HFV:
Wechsel der Projektleitung: Meldung an die EK (vorgängig)

Bei Forschungsprojekten mit verstorbenen Personen, die künstlich beatmet werden
Wesentliche Änderungen des Forschungsplans
Meldung an die EK (vorgängig)

Abschluss oder Abbruch des Forschungsprojekts
Meldung an die EK innerhalb von 90 Tagen

Checkliste ab Januar 2014 HFV Anhang 2 Punkt 1 bis 3

Gesuchsunterlagen für Forschungsprojekte mit Personen, die mit der Entnahme von biologischem Material oder der Erhebung von gesundheitsbezogenen Personendaten verbunden sind

Vorlagen für die Dokumente sind auf www.swissethics.ch abrufbar.

Nr.	Dokumentbezeichnung	Datum/ Versions-nummer	Allfälliger Verweis auf anderes Dokument	KEK: Bemerkung (freilassen)
0	**Begleitschreiben** – Rechnungsadresse muss vermerkt sein – muss vom Gesuchsteller (Projektleitung oder Sponsor) signiert sein	17.09.2014		+ 21.5.2014
1a	**Basisformular, einschliesslich der Laien-Zusammenfassung des Forschungsplans für die Patienten in der(n) jeweiligen Landessprache(n) am Durchführungsort** – muss von Projektleitung und falls zutreffend vom Sponsor signiert sein	21.05.2014		✓
1b	**Zusammenfassung des Forschungsplans (Synopsis)für KEK-Mitglieder** in der Landessprache der prüfenden KEK	17.09.2014 Version 2		✓
2	**Forschungsplan / Protocol** – muss von Projektleitung und falls zutreffend vom Sponsor signiert sein	17.09.2014 Version 2		✓
3a	**Aufklärungsbogen/Information und Einwilligungserklärung** – in der(den) jeweiligen Landessprache(n) am jeweiligen Durchführungsort, den hier verantwortlichen Personen und Kontakten – falls zutreffend auch Information für urteilsunfähige (z.B. Notfallpatienten, Demente), unmündige Personen, vertretungsberechtigte Personen (z.B. Eltern), oder für die schwangere PartnerIn des Studienteilnehmers – für Sub-Studien separate Information (z.B. Zusatz-MRI-Untersuchung, pharmakokinetische Untersuchung) – Information für die Weiterverwendung von Daten und Proben für zukünftige Forschungszwecke)	3a Information Kinder 3a Information und IC für Sorgeberechtigte von Kindern, Jugendlichen, Erwachsenen und Eltern von Betroffenen jeweils 17.09.2014 Version 2 (insgesamt 5 Dokumente)		Version 1 17.9.14

Nr.	Dokumentbezeichnung	Datum/ Versions-nummer	Allfälliger Ver-weis auf anderes Dokument	KEK: Bemerkung (freilassen)
3b	**Unterlagen betreffend Rekrutierung** – namentlich Anzeige, Inseratetexte oder Rekrutierungsschreiben an den Patienten oder Hausarzt	3b Rekrutierung 22.04.2014 Version 1 3b Unterstützungsschreiben (3 Dokumente)		✓ 6.3.14 NEH 7.3.14 NG 14.3.14 SHG
4	**Weitere Unterlagen, die der teilnehmenden Person abgegeben werden** – Patientenausweis, Tagebücher, Fragebogen in der jeweiligen Landessprache, **oder andere Unterlagen, die im Rahmen der Studie verwendet werden** – z.B. Interviewleitfaden, Scores, Fragebogen	Interviewleitfaden 22.04.2014 Version 1 Demografische Daten 22.04.2014 Version 1		✓ ✓
5	**Angaben über Art und Ausmass/Wert der Entschädigung der teilnehmenden Personen**	/		Teilnehmerinfo 12
6	**Bei Forschungsprojekten der Kategorie B:** – Versicherungsnachweis; oder – anderer Nachweis der Sicherstellung für allfällige Schäden	Versicherungsbestätigung 07.04.2014		✓
7	**Nachweis über sicheren Umgang mit biologischem Material und Personendaten** – namentlich dessen, beziehungsweise deren Aufbewahrung	Umgang mit Personendaten Version 1 22.04.2014		✓
8a	**Lebenslauf der Projektleitung und Nachweis der fachlichen Qualifikation (gemäss Art. 4 HFV)** – signiert und datiert	CV R. Mahrer-Imhof 08.05.2014 CV V. Waldboth 20.05.2014 CV and GCP A. Metcalfe 04.2014 CV and GCP C. Patch 05.2014		✓
8b	**Auflistung der am Forschungsprojekt beteiligten Personen** – einschliesslich ihrer Funktion und der entsprechenden fachlichen Kenntnisse	Staff list 21.05.2014		✓

Nr.	Dokumentbezeichnung	Datum/ Versions-nummer	Allfälliger Verweis auf anderes Dokument	KEK: Bemerkung (freilassen)
9	**Nachweis über die Eignung und Verfügbarkeit der Infrastruktur am Durchführungsort** – z.B.: Anzahl gleichzeitig durchgeführter Studien, Anzahl konkurrierender Studien, Vertretbarkeit der Geräteauslastung für das Forschungsprojekt etc.	Infrastruktur am Durch-führungsort Version 1 22.04.2014		✓
10	**Vereinbarung zwischen der Projektleitung und dem Sponsor oder weiteren Dritten** – namentlich bezüglich der Finanzierung des Forschungsprojektes, der Zuteilung von Aufgaben, der Entschädigung der Projektleitung sowie bezüglich der Publikation – muss von allen Parteien signiert sein	/		
11	**ZHAW (G) interne Vorprüfung von Gesuchsunterlagen zuhanden der EK**	01.05.2014		

Zusätzliche Gesuchsunterlagen für Forschungsprojekte, welche Untersuchungen mit Strahlenquellen umfassen (z.B. studienbegleitende Untersuchungen mit Röntgen, CT, Radiopharmazeutika für PET-Untersuchungen)

Einzureichen an die Ethikkommission:

Nr.	Dokumentbezeichnung	Datum/ Versions-nummer	Allfälliger Verweis auf anderes Dokument	KEK: Bemerkung (freilassen)
1	**Angaben zu wesentlichen Strahlenschutzaspekten, insbesondere eine Berechnung beziehungsweise Abschätzung der effektiven Strahlendosis, der Organdosis und allfälliger Tumordosen**	/		
2	**Die erforderliche Bewilligung für den Umgang mit Strahlenquellen oder radioaktiven Stoffen gemäss Artikel 28 des Strahlenschutzgesetzes vom 22. März 1991**[1] Die einzuhaltenden Dosisgrenzwerte richten sich nach Art. 28 Abs. 3-5 nach der Strahlenschutzverordnung vom 22. Juni 1994[2]	/		

Zusätzliche Gesuchsunterlagen für Forschungsprojekte, welche Untersuchungen mit offenen oder geschlossenen radioaktiven Strahlenquellen umfassen und eine Stellungnahme des BAG nach Artikel 19 Absatz 2 erfordern

(Gilt ab einer Dosis von ≥ 5 mSV pro Person und Jahr beim Einsatz i) von in der Schweiz nicht zugelassenen Radiopharmazeutika. ii) von Radiopharmazeutika, welche zwar zugelassen sind, aber ausserhalb einer nuklearmedizinischen Routineuntersuchung verwendet werden iii)oder von anderen offenen oder geschlossenen radioaktiven Strahlenquellen. In allen anderen Fällen insbesondere bei Röntgenuntersuchungen oder CT ist die Stellungnahme des BAG nicht erforderlich).

Unter oben genannten Voraussetzungen zusätzlich ans BAG einzureichen:
(Der zuständigen Ethikkommission ist gleichzeitig mitzuteilen, dass diese Einreichung erfolgt ist)

Nr.	Dokumentbezeichnung	Datum/ Versions-nummer	Allfälliger Verweis auf anderes Dokument	KEK: Bemerkung (freilassen)
1	**Angaben zu den Eigenschaften des Radiopharmazeutikums, namentlich zur Pharmakokinetik, Qualität, Stabilität, radiochemische Reinheit und Radionuklidreinheit**	/		
2	**Bei zugelassenen Radiopharmazeutika die Fachinformation**	/		
3	**Bei nicht zugelassenen Radiopharmazeutika die Angaben zum Herstellungsverfahren und zur Qualitätskontrolle des Radiopharmazeutikums, die Namen der hierfür verantwortliche Personen sowie Angaben zu deren fachlichen Qualifikation**	/		

[1] SR **814.50**
[2] SR **814.501**

Nr.	Dokumentbezeichnung	Datum/ Versions-nummer	Allfälliger Ver-weis auf anderes Dokument	KEK: Bemerkung (freilassen)
4	**Die Namen der für die Anwendung des Radiopharma-zeutikums am Menschen verantwortlichen Personen sowie Angaben zu deren fachlichen Qualifikation**	/		
5	**Angaben gemäss Formular des BAG für Forschungspro-jekte mit Radiopharmazeutika oder mit radioaktiv markierten Stoffen[3]**	/		

Ethikkommission

Ort/Datum:

Zürich 25.09.2019

Wissenschaftliches Sekretariat

[3] Dieses Formular kann beim Bundesamt für Gesundheit, Abteilung Strahlenschutz, 3003 Bern, bezogen oder der Internetadresse www.bag.admin.ch >Themen>Strahlung, Radioaktivität und Schall>Nuklearmedizin und Forschung>Radiopharmazeutika>Gesuchsformular abgerufen werden.

Zürcher Hochschule für Angewandte Wissenschaften
Institut für Pflege

Florence Nightingale School of Nursing and Midwifery
King's College London

Study Protocol

Study title
Becoming an adult: Experiences of individuals affected by neuromuscular disease and their families

Short title
Becoming and adult with neuromuscular disease

Study description
A qualitative study of experiences of young people suffering from neuromuscular disease during transition into adulthood and their families

Study Type	Dissertation – Qualitative Study
Study Categorisation	HFV, risk A
Sponsor	Zurich University of Applied Sciences, Winterthur, Switzerland Florence Nightingale School of Nursing and Midwifery, King's College London, United Kingdom
Principal Investigator	Prof Dr. Romy Mahrer-Imhof Zurich University of Applied Sciences (ZHAW) School of Health Professions Institute of Nursing Technikumstrasse 71 8401 Winterthur Tel. +41 58 934 63 44 Email: romy.mahrer@zhaw.ch
Project manager / PhD-student	Veronika Waldboth, MScN, RN Zurich University of Applied Sciences (ZHAW) School of Health Professions Institute of Nursing Technikumstrasse 71 8401 Winterthur Tel.+41 58 934 64 99 Email: veronika.waldboth@zhaw.ch
Protocol Version and Date	Version 4, 25.03.2015

Confidential

Signature Page

Study Title: Becoming an adult: Experiences of individuals affected by neuromuscular disease and their families

The Principal Investigator has approved the protocol version 4, dated 25.03.2015, and confirms hereby to conduct the study according to the protocol, current version of the World Medical Association Declaration of Helsinki, ICH-GCP guidelines and the local legally applicable requirements.

Principal Investigator: Prof Dr. Romy Mahrer-Imhof, PhD, RN

Winterthur, 30.3.15 — Place/Date

Signature

Project Manager / PhD- student

I have read and understood this protocol (Version 4, 25.03.2015) and agree to conduct the study as set out in this study protocol, the current version of the World Medical Association Declaration of Helsinki, ICH-GCP guidelines and the local legally applicable requirements.

Project Manager / PhD- student: Veronika Waldboth, MScN, RN

Winterthur 24.3.15 — Place/Date

Waldboth Veronika — Signature

Inhalt

Abstract

This study will explore experiences of young persons affected by neuromuscular disease and their families during transition into adulthood. Becoming an adult is considered to be a normal developmental stage within the life cycle. However, a chronic childhood condition can disrupt that transition and may have impact on the young persons' and their families' lives. Therefore, this study aims to understand experiences of families suffering from neuromuscular disease during transition of the affected young person into adulthood and to identify major challenges and strategies by which individuals and families successfully adapt to new situations.

The study design is a constructivist approach to Grounded Theory. Data are collected from single interviews with young people affected by neuromuscular disease and their families. Inclusion criteria for the young person affected by neuromuscular disease are age range between 14 to 30 years, resident in Switzerland, first symptom onset of the genetic neuromuscular disease in childhood and moderate to severe physical impairment. Moreover, parents, younger and older siblings and other next of kin of such a young person are asked to participate. Excluded are non-German-speaking individuals and persons with cognitive impairment who are not able to give informed consent. The sample size is estimated to be 40 participants of approximately 8 to 15 families. Interviews will be recorded and transcribed verbally. Collected data will be analysed using coding, memo writing and theoretical sampling aiming at data based theory development. The intent of this study is to move beyond description and to generate a general interpretation of the transition from childhood into adulthood which young people and their families experience. Data collection takes place in Switzerland. Study duration is three years and data collection and analysis will take place simultaneously and start in September 2014. The study ends in September 2016.

Study summary in local language

Diese Studie untersucht Erfahrungen von jungen Menschen, die an neuromuskulären Erkrankungen leiden und ihrer Familien während des Übergangs vom Kind zum Erwachsenen. Eine chronische Krankheit, die sich im Kindesalter manifestiert und mit starken körperlichen Einschränkungen einhergeht kann diese Entwicklungsphase stören und sich auf die betroffenen Jugendliche und jungen Erwachsenen, sowie auf deren Familien auswirken. Daher möchte diese Studie die Erfahrungen der betroffenen Familien untersuchen und verstehen, mit welchen Herausforderungen sie umgehen müssen und welche Strategien sie anwenden, um mit veränderten Situationen zurecht zu kommen.

Studienziel

Das Ziel dieser Studie ist es, die Erfahrungen von Jugendlichen und jungen Erwachsenen und ihrer Familien zu verstehen. Die Feinziele sind:

- Die Herausforderungen zu verstehen, mit denen betroffene Familien während dieser Entwicklungsphase konfrontiert sind;
- Strategien zu identifizieren, mit denen sich betroffenen Familien erfolgreich an veränderte Situationen anpassen;
- Faktoren zu identifizieren, die die Anpassung an veränderte Situationen beeinflussen;
- Erfahrungen der betroffenen Familien mit dem Schweizerischen Gesundheitssystem zu verstehen.

Methodisches Vorgehen

Diese pflegewissenschaftliche Studie hat einen qualitativen Ansatz. Daten werden durch Einzelgespräche mit den Jugendlichen und jungen Erwachsenen, deren Eltern, jüngeren und älteren Geschwistern und anderen nahestehenden Bezugspersonen erhoben. Die Anzahl der Teilnehmenden wird vorab auf ungefähr 8 bis 15 betroffene Familien geschätzt. Eingeschlossen werden Familien und Einzelpersonen, die folgende Einschlusskriterien erfüllen:

- Junge Menschen im Alter von 14 bis 30 Jahren, die an einer neuromuskulären Erkrankung leiden, die bereits im Kindesalter aufgetreten ist (0-10 Jahre) und mit einer mässigen bis starken körperlichen Einschränkung einhergeht;
- Eltern, jüngere und ältere Geschwister (ab 8 Jahren) und nahestehende Angehörige von jungen Menschen, wenn diese 14 bis 30 Jahre alt sind und an einer neuromuskulären Erkrankungen leiden, die im Kindesalter aufgetreten ist und mit einer körperlichen Einschränkung einhergeht;

- Teilnehmende, die in der Schweiz ansässig sind.

Nicht teilnehmen hingegen dürfen Personen

- die kein Deutsch sprechen
- oder die an einer ausgeprägten geistigen Einschränkung leiden, die es der Person nicht ermöglicht zur Teilnahme an der Studie einzuwilligen.

Die Studie beinhaltet ein gemeinsames Gespräch mit einer wissenschaftlichen Mitarbeiterin, das ungefähr eine Stunde dauert. Der Ort des Interviews kann vom Teilnehmenden bestimmt werden und Interviews können im familiären Umfeld oder an einem neutralen Ort stattfinden. Die Gespräche werden auf Tonband aufgenommen, abgeschrieben und Namen von Personen werden verschlüsselt. Daraufhin werden die Tonbandaufnahmen gelöscht. Die Teilnahme an einem Interview ist freiwillig und alle Daten werden vertraulich behandelt.
Mit der Durchführung des Interviews endet die Studienteilnahme des Teilnehmers. Die Durchführung der Gespräche mit den Betroffenen und deren Familien oder nahestehenden Angehörigen ist für den Zeitraum von September 2014 bis Oktober 2015 geplant.

Nutzen und Risiken

Die Teilnahme an dieser Studie sollte für die Teilnehmenden kein Risiko darstellen. Sie erklären sich zu einem Gespräch bereit, das ungefähr eine Stunde dauert. Dieses Gespräch kann verschiedene Emotionen auslösen. Insgesamt ist jedoch davon auszugehen, dass es die Teilnehmenden schätzen, sich in einem persönlichen Gespräch über ihre Erfahrungen, Lebenssituation und Befindlichkeit auszudrücken. Um diesen Umständen Rechnung zu tragen wird bei den Interviews genügend Zeit für den Abschluss des Gesprächs eingeplant. Beim Auftreten von negativen Emotionen während oder nach dem Interview wird bei Bedarf Kontakt mit einer entsprechenden Fachperson hergestellt.
Die Teilnahme an dieser Studie nützt den teilnehmenden Personen nicht direkt. Sie tragen aber dazu bei, dass in Zukunft andere Familien mit Muskelerkrankungen unterstützt werden können. Es fallen keine Kosten für die Studienteilnehmenden an. Allfällige Reisespesen werden nicht übernommen.

1. Study administrative structure

1.1. Sponsors

Zurich University of Applied Sciences
School of Health Professionals
Institute of Nursing
Technikumstr. 71
CH-8401 Winterthur

Florence Nightingale School of Nursing and Midwifery
King's College London
James Clerk Maxwell Building
57 Waterloo Road
London SE1 8WA
United Kingdom

1.2. Principal Investigator

Prof Dr. Romy Mahrer-Imhof, PhD, RN
Head of Master of Science in Nursing
Zurich University of Applied Sciences
School of Health Professionals
Institute of Nursing
Technikumstr. 71
CH-8401 Winterthur
Tel. +41 58 934 63 44
Email: romy.mahrer@zhaw.ch

1.3. Project Manager / PhD- student

Veronika Waldboth, MScN, RN
Research Associate
Zurich University of Applied Sciences

School of Health Professionals
Institute of Nursing
Technikumstr. 71
CH-8401 Winterthur
Tel. +41 58 934 63 99
Email: veronika.waldboth@zhaw.ch

1.4. Supervisors of doctoral study

Prof Dr. Alison Metcalfe, PhD, RN
Professor of Health Care Research & Associate Dean for Research
Florence Nightingale School of Nursing and Midwifery
King's College London
James Clerk Maxwell Building
57 Waterloo Road
London SE1 8WA
United Kingdom
Tel. +44 (0)207 848 3828
Email: alison.metcalfe@kcl.ac.uk

Dr. Christine Patch, PhD, registered genetic counsellor, RN
Consultant Genetic Counsellor/Manager & Reader in Clinical Genetics
Department of Clinical Genetics
Guys Hospital
Great Maze Pond
London SE1 9RT
United Kingdom
Tel. +44 (0)207 188 1364
Email: Christine.patch@gstt.nhs.uk

2. Ethical and regulatory aspects

Before the study will be conducted, the protocol, the proposed patient information and consent form as well as other study-specific documents have been submitted to the Ethics Committee Zurich (CEC ZH).

2.1. Categorisation of the study

This study is considered nursing research and rated as non- experimental research project involving persons (HFV) with risk category A. This study has a qualitative design and data are collected by single interviews with families affected by neuromuscular disease. The planning and preparation phase started in September 2013 and the planned study end is in September 2016 (study duration: 3 years). No changes are made to the protocol without prior Sponsor and CEC ZH approval, except where necessary to eliminate apparent immediate hazards to study participants. Premature study end or interruption of the study is reported within 15 days. The regular end of the study is reported to the CEC ZH within 90 days, the final study report will be submitted within one year after study end. Amendments are reported according to chapter 2.7.

2.2. Ethical conduct of the study

The study will be carried out in accordance to the protocol and with principles enunciated in the current version of the Declaration of Helsinki, the guidelines of Good Clinical Practice (GCP) issued by ICH, the Swiss Law and Swiss regulatory authority's requirements. The CEC ZH and regulatory authorities will receive annual safety and interim reports and be informed about study stop/end in agreement with local requirements.

2.3. Declaration of interest

The Principal Investigator and the Project Manager declare to have no financial, proprietary or other conflict of interest.

2.4. Patient information and Informed Consent

The investigators will explain to each participant the nature of the study, its purpose, the procedures involved, the expected duration, the potential risks and benefits and any discomfort it may entail. Each participant will be informed that the participation in the study is voluntary and that they may withdraw from the study at any time and that withdrawal of consent will not affect their subsequent medical assistance and treatment.

All participants for the study will be provided a participant information sheet and a consent form describing the study and providing sufficient information for the participant to make an informed decision about their participation in the study. Enough time will be given to the participant to decide whether to participate or not (time frame of minimum one week after provision of participant information).

Detailed description of participant information and consent for families with a young person suffering from neuromuscular disease:

- Adults (18+) will receive oral and written study information and need to give oral and written consent to participation. If the person affected by MD is not able to give written consent, they can give oral consent which is testified via record. The verbal informed consent is recorded on a separate audiofile, because the interview file will be deleted after transcription.
- Young persons between 14 and 17 years of age receive oral and written information and need to give oral and written consent to study participation. If the person affected by MD is not able to give written consent, they can give oral consent which is testified via record. The verbal informed consent is recorded on a separate audio file, because the interview file will be deleted after transcription.
- Children between 11 and 13 years of age receive age appropriate oral and written information and need to give oral consent to participation. Additionally, written informed consent of the parent or legal guardian is needed.
- Children between 8 and 10 years of age receive oral information and need to give oral consent. Written informed consent of the parent or legal guardian is needed.

The patient information sheet and the consent form have been submitted to the CEC ZH to be reviewed and approved. The formal consent of a participant, using the approved consent form, will be obtained before the participant participates at the interview. The participant is given time to read and consider their statement before signing and dating the informed consent form, and will be given a copy of the signed document. The consent form will also be

signed and dated by the investigator (or his designee) and will be retained as part of the study records.

2.5. Participant privacy and confidentiality

The investigator affirms and upholds the principle of the participant's right to privacy and that they will comply with applicable privacy laws. Anonymity of the participants will be guaranteed when presenting the data at scientific meetings or publishing them in scientific journals. Individual data obtained as a result of this study is considered confidential and disclosure to third parties is prohibited. Subject confidentiality will be further ensured by utilising subject identification code numbers to correspond to data in the computer files. For data verification purposes, authorised representatives of the Sponsor or an ethics committee may require direct access to parts of the data relevant to the study.

2.6. Early termination of the study

The Sponsor-Investigators may terminate the study prematurely according to ethical concerns, insufficient participant recruitment and when the safety of the participants is doubtful or at risk, respectively.

2.7. Protocol amendments

The Principal Investigator, the Project Manager and the Sponsors are allowed to amend the protocol or to provide suggestions for a protocol amendment. Substantial amendments are communicated to the CEC ZH and only implemented after approval of the CEC ZH and the Sponsor. All Non-substantial amendments are communicated to the CEC ZH within the Annual Safety Report (ASR).

3. Background and rational

The proposed study is looking at life experiences of adolescents and young adults suffering from genetic neuromuscular disease and of their families during transition into adulthood. There are different types of genetic neuromuscular disease and the most common are Muscular Dystrophy (MD) and Spinal Muscular Atrophy (SMA) (Amato & Russell, 2008; Strehle, 2009; Wirth, 2000). MD is a group of muscle diseases characterized by a genetic defect in muscle proteins that causes progressive wasting and fibrosis of muscles from all body parts including heart and lungs (Papazian & Alfonso, 2002; Schweizerische Muskelgesellschaft, 2011; Wirth, 2000). There are more than thirty types of MD that can differ in the genetic defect, inheritance pattern, age of first symptom onset or course of the disease. At the moment there is no cure to MD and prognosis depends on the specific type varying from mild and very slowly progressive forms with a normal life expectancy to severe muscle weakness with progressive functional disability. The major type, Duchenne MD affects 1 in every 3.500 of all male newborns and first symptoms usually present in early childhood (Papazian & Alfonso, 2002; Strehle, 2009; Wirth, 2000). SMA, in contrast, affects 1 in every 10.000 newborns and presents with progressive muscle weakness and atrophy caused by degeneration of the anterior horn cells of the spinal cord (Amato & Russell, 2008; Wirth, 2000). Four types of SMA are distinguished according to the age of onset and the course of the disease, with types II and III showing similar clinical manifestations as Duchenne MD (Schaaf & Zschocke, 2013).

Previous research on life experiences of individuals affected by MD have focused mainly on their experiences during childhood or adulthood. A contributing factor is that some forms have their onset in adulthood and have a mild disease course (Amato & Russell, 2008; Schweizerische Muskelgesellschaft, 2011). Whereas by contrast, there are more severe forms that have their first symptom onset in early childhood and which progress rapidly with that young person dying before reaching adulthood. In the last two decades, life expectancy of individuals with severe childhood MD rose because of progress in health care and medicine (Bushby et al., 2005; Kohler et al., 2009). Crucial to these children's survival were improvements in prevention and treatment of respiratory and cardiac complications, maintenance of muscular strength and function and in the management of spinal deformities. Today most individuals with severe MD go through adolescence and reach adulthood (Gibson, Young, Upshur, & McKeever, 2007). Becoming an adult is a time of transition and personal development but this can be different for individuals affected by a chronic childhood illness as compared to their healthy peers. For young persons with MD, for example, physical dependence increases at the same time as they are expected to break away from their

parental homes and live an independent life (Parkyn & Coveney, 2013). This can have an impact on many aspects of the young person's life such as their psychosocial and emotional wellbeing (Polakoff, Morton, Koch, & Rios, 1998). Individuals with Duchenne MD, for instance, have an increased risk for social isolation and the development of depression (Polakoff, et al., 1998).

Life experiences of adolescents suffering from MD have to date had little empirical investigation (Abbott, Carpenter, & Bushby, 2012; Gibson, et al., 2007). Moreover, the phenomenon of interest has mostly been explored from a single or disjointed perspective and did not include a family system approach (Loukas, Twitchell, Piejak, Fitzgerald, & Zucker, 1998; Peterson, 2005; Rolland & Williams, 2005). Genetic and chronic childhood illnesses have a high impact not only on the individual but on the whole family (Drotar, 1981; Neill, 2010; Rolland & Williams, 2005; Samson et al., 2009). Providing care to a family member with disability can be rewarding, but at the same time burdensome (Pangalila et al., 2012). It can lead to physical, psychological, emotional, social and financial problems for the caregiver. In the case of Duchenne MD increased levels of family distress have been reported (Polakoff, et al., 1998).

In Switzerland patients with MD are generally cared for by their relatives (Kohler et al., 2005). Therefore a family centred approach to care is indicated to prevent young people and their families from being left alone with their potential suffering which is linked to a genetic chronic disease (Rolland & Williams, 2005). By studying life experiences of these families we can identify their explanations and expectations of events and understand how families cope with neuromuscular disease during the young person's transition into adulthood. This knowledge may help health professionals to better assist affected families to anticipate and react to difficult and stressful situations. Furthermore, advanced knowledge and understanding gives a solid foundation for the development of tailored and family centred nursing interventions which have the potential to improve quality of care during transition from paediatric to adult health care services.

4. Study purpose and aims

This study will explore experiences of young persons affected by neuromuscular disease and their families during transition into adulthood. Becoming an adult is considered to be a normal developmental stage within the life cycle (Black, Holditch-Davis, & Miles, 2009; Macmillan & Copher, 2005; Rolland & Williams, 2005) and is usually defined from the late teens to the early twenties (Arnett, 2000). However, a genetic or chronic childhood condition can disrupt that transition and may have impact on the young persons' and their families' life experiences. Therefore, the study aim is to understand experiences of families suffering from neuromuscular disease during transition of the affected young person into adulthood and to identify major challenges and strategies by which individuals and families successfully adapt to new situations.

The following overarching research question is the focus for the investigation:

Where a young person is affected by neuromuscular disease, what are theirs and their family members' experiences of their transition into adulthood?

Secondary questions include:

I. What challenges do young persons affected by neuromuscular disease and their family members' face during transition into adulthood?
II. How do young persons affected by neuromuscular disease and their family members successfully adapt to new situations and what facilitates individuals and families adaption?
III. What are the young persons' and their family members' life experiences with the Swiss health care system during transition into adulthood?
IV. Is it possible to identify a process that describes the transition into adulthood for young persons affected by neuromuscular disease and if yes, what are properties of this process?

5. Methods

5.1. Study design

The proposed study design for the empirical investigation is a constructivist approach to Grounded Theory (GT) (Charmaz, 2006, 2008). Constructivist GT is a qualitative research design derived from sociology that aims to investigate human experiences within their social contexts (Burns & Grove, 2005; Creswell, 2007, 2014). In contrast to the objectivist GT method initially developed by Glaser and Strauss, it refuses to see the world as an external reality that can be discovered through research (Charmaz, 2006, 2008). Instead, it applies a social constructivist perspective, where multiple realities co-exist and are co-constructed between researcher and participants. The researcher views human interactions, actions and experiences embedded in their larger context and through an interpretive lens (Creswell, 2014). Moreover, the intent of this GT study is to move beyond description and to generate a general interpretation of the transition from childhood into adulthood that young people affected from neuromuscular disease and their families experience.

5.2. Participants and setting

Young persons affected by neuromuscular disease and their parents, younger and older siblings and other next of kin are considered family members. The term family is understood as a “group of people, either living together or remaining in close contact with each other” (Gudmundsdottir & Chesla, 2006) including nuclear and patchwork compositions and young people living in institutions or at home. Parents, siblings and other next of kin need to have close contact and share experiences of daily living with the young person affected by neuromuscular disease in order to be able to give rich information about the investigated subject. Participants can be individuals and families, who fulfil the following inclusion and exclusion criteria:

Inclusion criteria for young people affected by neuromuscular disease

Inclusion criteria for the young person affected by neuromuscular disease are:

- age range between 14 to 30 years,
- resident in Switzerland,
- first symptom onset of the genetic neuromuscular disease in childhood (age range from 0 to 10 years)

- and moderate to severe physical impairment according to the Duchenne muscular dystrophy physical Impairment and Dependency score (DID) (Kohler, et al., 2005).

Excluded are young persons affected by neuromuscular disease that are:

- non German-speaking
- and have a cognitive impairment so that they are not able to give informed consent.

Inclusion criteria for family members

Inclusion criteria for family members are:

- Parents of a young person, if the young person is affected by neuromuscular disease (disease onset in childhood, moderate to severe physical impairment) and is 14 to 30 years old,
- older siblings of a young person, if the young person is affected by neuromuscular disease (disease onset in childhood, moderate to severe physical impairment) and is 14 to 30 years old
- younger siblings (with a lower age limit of 8 years) of a young person, if the young person is affected by neuromuscular disease (disease onset in childhood, moderate to severe physical impairment) and is 14 to 30 years old
- other next of kin of a young person, if the young person is affected by neuromuscular disease (disease onset in childhood, moderate to severe physical impairment) and is 14 to 30 years old.
- Family members resident in Switzerland.

Excluded are:

- non German-speaking family members
- and individuals with cognitive impairment who are not able to give informed consent.

The sample size is estimated to be approximately 8 to 15 families. Data saturation will serve as a guiding principle during sampling for the total number of families recruited. Data saturation is the moment when there is no new information resulting from data collection that adds to additional understanding of developed categories (Creswell, 2007). For GT studies the inclusion of a large number of participants is recommended in order to achieve detail in theory (Creswell, 2013). In the qualitative investigation of experiences of next of kin's of patients with MD the sample size was 36 (Bostrom, Ahlstrom, & Sunvisson, 2006). Given the aim of this study and the heterogeneity of the sample including young people and their families the sample needed for saturation is estimated to be 40 participants from approximately 8 to 15 families.

5.3. Recruitment of participants

The recruitment strategy is purposive and starts with the identification of potential participants by collaborating organizations. Collaborating organizations in recruitment are the Mathilde-Escher-Heim, a facility in the city of Zurich specialised in the care of patients with neuromuscular disease, the Neuromuscular Centre for children and adolescents of the Children's University Hospital Zurich, the Swiss Association of Muscular Diseases and self-help groups for individuals with neuromuscular diseases and their families in the canton Zurich. These organizations have agreed to collaborate and to give access to potential participants (see documents 3b Unterstützungsschreiben).

There are two processes of recruitment of families:

1) The young person affected by neuromuscular disease is recruited first and may provide access to other family members;

2) A family member (for example a parent or an adult sibling) is recruited first and may provide access to the young person affected by neuromuscular disease or other family members.

The researcher aims to investigate the transition from childhood into adulthood from a family perspective and wants to interview more than one family member. Therefore the recruited participant is asked to identify other possible family members. However, a family can participate, even if just one family member (young person affected by MD or SMA, parent or sibling) is willing to participate. This may give a different insight into the investigated subject, because family function and structure may differ in families, where just one family member is willing to participate.

Recruitment strategy

Recruitment methods will include:

a) Information to potential study participants

   Collaborating organizations will inform potential participants via leaflet (leaflet text see 3b Rekrutierung Version 1 22.04.2014) about the study. Interested participants are asked to share their contact details (name, address, phone number) with the researcher via enclosed reply slip and envelope if they are willing to participate.

b) Organization of road shows

   Collaborating organizations will organise road shows for potential participants where the researcher presents the study (study aims, methods, benefits and risks). Interested participants receive the study leaflet and are asked to share their contact details.

c) Advertisement of the study

Collaborating organizations will disseminate a summary of the study including the contact details of the researcher via their websites and newsletters (abstract see 3b Rekrutierung Version 1 22.04.2014).

Study participation

Potential participants who shared their contact details are then contacted by phone. The purpose of this phone call is:

- to give detailed information about the study and what is involved,
- to answer any questions the potential participant has,
- to assess if inclusion and exclusion criteria are met and
- to identify other family members and potential study participants.

After this initial contact, a written study information, a consent form and a study leaflet for the information of other potential participants will be sent to the participant's home via mail. If the individual interested in participation is a minor, she/he is asked to inform her/his parent or legal guardian about their interest in study participation and to ask for their oral and written consent. In this case, a study information for the parent or legal guardian is added to the mail. For a graphical representation of study participation see figure 1.

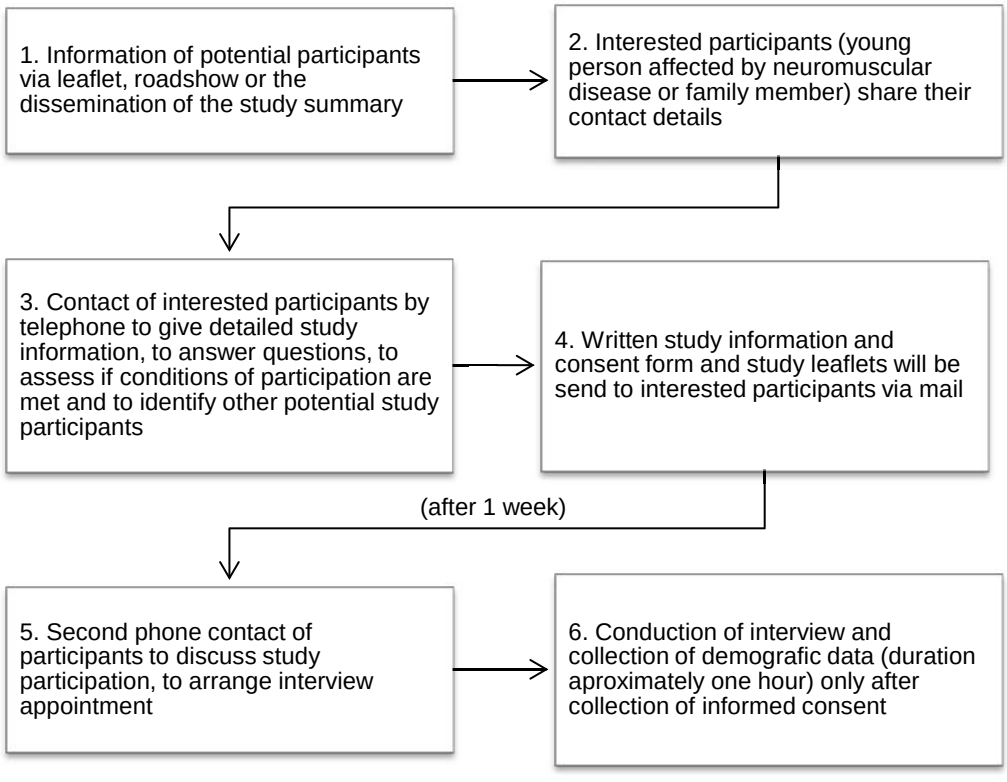

Figure 1: Study participation

Enough time will be given to the participant to decide whether to participate or not. The minimum time frame is one week after provision of the written participant information via mail. After the expiration of this term, the researcher contacts the study participant for a second time, to confirm study participation and to arrange an interview appointment. Participants are asked to bring the signed consent form to the interview appointment. No data will be collected before study participants receive study information and give their consent to participation.

These are the requirements for the study information and informed consent for this study according to the CEC ZH:

- Adults (e.g. individuals affected by MD from 18 to 30 years, parents and older siblings of full age (18+)) receive oral and written study information and need to give oral and written consent to participation. If the person affected by MD is not able to give written consent, they can give oral consent which is testified via record. The verbal informed consent will be recorded on an audio file separate from the interview, because the interview will be deleted after transcription.
- Young persons between 14 and 17 years of age (e.g. individuals affected by MD between 14 and 17 years, siblings between 14 and 17 years) receive oral and written information and need to give oral and written consent to study participation.
- Children between 11 and 13 years of age (younger siblings) receive age appropriate oral and written information and need to give oral consent to participation. Additionally, written informed consent of the parent or legal guardian is needed.
- Children between 8 and 10 years of age (younger siblings) receive oral information and need to give oral consent. Written informed consent of the parent or legal guardian is needed.

5.4. Data collection

Data are collected from single interviews with interview participants in their natural setting. Interview participants are young people affected by neuromuscular disease, their parents, younger and older siblings and other next of kin. Every participant takes part in one single interview. All participants including children and adolescents are interviewed alone. In the interviews the participants will be asked broad, open ended questions. An example of an initial question is: What does a normal day in your everyday life look like?
Main themes that will be covered by the interview are family life, social life and friends, living and work situation, health situation and growing up and coping with a neuromuscular disease either as the affected person, a parent or as the sibling. For every participant group an interview guide appropriate to the participants' age was developed. The topic guides for adolescents and adults differ only in the form of address (see 4 Interviewleitfaden Version 1 22.04.2014). Children under the age of 12 will have the opportunity to engage in warm up activities and to do drawings and tell stories about their drawing as part of the interview. Evidence suggests that the age of twelve would be the upper limit for children doing drawings (Coad, Plumridge, & Metcalfe, 2009; Gross, Hayne, & Drury, 2009; Woolford, Patterson, Macleod, Hobbs, & Hayne, 2013). Child focused interview techniques will include:

- Warm up activity: At the beginning of the interview the child is engaged in a warm up activity. This warm up activity consists of a card game where the child and the interviewer alternate in choosing from a deck of question cards, then playing the role of the interviewer by asking the question on the card. This warm up activity aims to increase comfort of the child and to diminish the power differentials by giving explicit permission for the child to question the interviewer (Teachman & Gibson, 2013).
- Drawings: After the warm up activity and initial questions the children will be asked to do a drawing of their family and to talk about the drawing. This is a powerful method of building rapport and facilitating children's report (Coad, Plumridge, & Metcalfe, 2009; Gross, Hayne, & Drury, 2009; Woolford, Patterson, Macleod, Hobbs, & Hayne, 2013). The focus is on what the child says rather than on an interpretation of the child's drawing. The researcher will also ask open ended questions to guide the child's report. Drawings are suited for data collection for participants that are not able to articulate their beliefs and emotions using spoken words, such as young children (Gross, et al., 2009; Guillemin, 2004).

Interview guides were pretested by interviewing individuals in the relevant age groups. Questions were clear to these test persons and no major changes were necessary after the pre-test. The interview guides can be slightly modified as data collection proceeds. This

approach is consistent with GT methods and allows refinement of questions in order to gather information necessary for theory development (Creswell, 2013).
Interviews will be recorded, transcribed word by word and then encoded to assure privacy. After transcription audio files will be deleted. The password protected file containing information about solution for data encryption is saved on a secured computer at ZHAW and the password is only known to the project manager. The principal investigator and the two supervisors of the study will not have access to this information and they will receive encoded data only. In order to assure traceability data will be stored in a secure archive at ZHAW for ten years after study conclusion.

Demographic data

After the interview of all participants demographic data are gathered including gender, age, diagnosis of the affected person, relationships, marital status, living arrangement, employment, school leaving qualifications and overall health status (see 4 Demografische Daten Version 1 22.04.2014). Demografic data will be encoded.

Field notes

Observations and reflections of the researcher during and after the interviews will be written down and included in the analysis as field notes.
Data collection is planned to take place in German-speaking Swiss cantons. Data collection and analysis will take place simultaneously and start approximately in September 2014 and end a year after.

5.5. Data analysis

Collected data from the interviews will be analysed using coding, memo writing and theoretical sampling with the aim of data based theory development (Charmaz, 2006). The analysis process is being supported by the data analysis software program ATLAS.ti. Only interview data will be analysed. Drawings from children are not considered data but a method of facilitating children's report. According to Charmaz (2006) two types of coding are performed, initial line-by-line coding and focused coding.

Coding interview data

- Initial coding is the first coding phase that makes the researcher familiar with the data and reveals first conceptual ideas. The researcher reads rich transcripts line by line and identifies themes and patterns. In a second step codes are allocated to these themes and

patterns. Codes in form of verbs are preferred. After coding of a few rich interviews, a coding scheme with code groups and allocated codes develops.

- The following phase, focused coding, is then used to analyse larger amounts of data using the coding scheme and looking for explanations in a more systematic way. While focused coding, constant comparison takes place and is used to make comparisons between data at every level of analysis. Thus, the researcher goes back to previously coded interviews and repeats coding. As a result of the coding phases categories emerge and are then linked with each other in order to develop a grounded theory (Creswell, 2013).

Memo writing and theoretical sampling

Memo writing is used as an assistant tool for the whole analysis process where codes are taken apart and analysed rigorously which helps to increase the level of abstraction of ideas (Charmaz, 2006).

Initial sampling can be followed by theoretical sampling. Theoretical sampling is purposive and aims to gather additional data to develop emerging theoretical categories (Charmaz, 2006). This stage may include additional recruitment of individuals suffering from neuromuscular disease and their families.

Descriptive Statistics

Demographic data will be analysed using the Predictive Analytics Software SPSS. Descriptive analysis will be carried out including frequency and distribution of single variables, such as central tendency (mean) and dispersion (range or standard deviation). Results will be presented in graphics or tables.

6. Translation methods

The delineated project is the project managers PhD study. The researcher attends the PhD-programme in nursing at the Florence Nightingale School of Nursing & Midwifery at King's College London, United Kingdom, in collaboration with the Zurich University of Applied Sciences in Winterthur, Switzerland. Data collection takes place in the German-speaking cantons of Switzerland. The spoken language in these Swiss cantons is Swiss German and the written language is Standard German. The researchers first language is a German dialect similar to the Austrian German, called the South Tyrolean dialect which exists only spoken; the written language is Standard German. The researcher has good command of spoken Swiss German and spoken and written Standard German, Italian and English. As the PhD-programme takes place in the United Kingdom the language used in postgraduate research training, supervision meetings and thesis writing is English. This implicates that at some point in time some data and results need to be translated into English. The following translation methods are used during this research process:

a) Interviews are planned to be conducted in the participant's original spoken language, Swiss German. This method is appropriate to obtain understanding of participant's experiences and to minimize loss of meaning (Smith, Chen, & Liu, 2008; Twinn, 1997).
b) The verbatim transcription of the records and the analysis of the data takes place in the original written language of the informants and the researcher, Standard German (Chen & Boore, 2009; van Nes, Abma, Jonsson, & Deeg, 2010). This approach is advantageous: firstly, time and costs can be saved, if not all interviews need to be translated into English. And secondly, potential limitations of the analysis process can be prevented that can happen, if analysis is not conducted in the researchers' first language.
c) In order to allow discussions with the English speaking supervisors Codes, definitions of Codes and relevant quotations are then translated into English by the PhD student and by a bilingual Swiss Supervisor from ZHAW. The researcher and the Swiss supervisor discuss potential translations and use fluid descriptions of meanings rather than single words in their translations (van Nes, et al., 2010). This is considered to be important to provide a technically and conceptually accurate translation. As the analysis phase is considered to be dynamic this procedure is repeated until the end of the analysis.
d) Quotations are planned to be included in the doctoral thesis and in publications. In qualitative research quotations are used to confirm results and to increase the trustworthiness of the study (van Nes, et al., 2010). In order to avoid misinterpretations while quoting, in the thesis quotes are presented in the original and the target language.

In publications the space is more limited and therefore the quotes will be written in the journal's language of choice.

e) Not addressing linguistic challenges "threatens the credibility, transferability, dependability and confirmability of cross-language qualitative nursing research" (Squires, 2008). Therefore, a detailed methodological description of the translation process is carried out during the study.

7. Ethical considerations

The study will seek approval of the CEC ZH prior to investigation. Participants will be informed that study participation is voluntary and that they can withdraw from the study at any time without any consequences. Information about study aims, methods, benefits and potential risks are given to all participants in an age-appropriate manner and according to the requirements of the CEC ZH prior to participation. All participants need to give their informed consent before taking part in data collection. If a minor participant wants to take part in the study, the person who is in custody of the child or adolescent (parent or legal guardian) receives study information and is asked to give their written consent. Collected data are treated confidentially and transcripts will be encoded and stored in a secure place. Details on storage of data are described on page 22.

Participants do not benefit directly from study participation and they are informed about this fact. However, they may contribute to future improvements in the care of other families affected by neuromuscular diseases.

Study participation does not represent an increased risk for the participants. They take part in an interview that lasts approximately one hour where they talk about their experiences and give demographic information. However, during and after the interview different emotions can be triggered. In order to minimize negative effects the researcher plans enough time for the final phase of the interview. In general, it can be expected that the participant appreciate having the opportunity to share their experiences and thoughts in a single interview. In case of emerging distress or questions that the researcher is not competent to meet indicators for action are defined and contact details of a counselling service of the University Children's Hospital of Zurich are offered to the participant. Indicators for action are: 1) if the participant shows signs of emotional distress such as strong feelings of anger and guilt, 2) if the participant asks for referral, 3) if the participant asks for information or assistance, that is not within the scope of the research project.

If participants report bad practice or a crime the researcher will assess the situation and the potential risk for involved people. In case of increased risk the researcher will 1) inform the

participant about her concerns and her need to take action, 2) discuss the situation and possible actions with her supervisors 3) and take action in order to protect confidence and to introduce safety measures for the persons at risk.
The researcher assures that there is no relationship of dependence between researcher and the participant family and that no information goes back to a treating or referring physician or nurse. Study participation generates no costs for the participants and possible travel expenses will not be covered.

8. Role of the researcher and reflexivity

The doctoral student's interest in family centred care of chronically ill patients is based on various professional and personal experiences in the field of nursing. First of all, her clinical nursing experience highlighted the importance and the current lack of systemic approaches to the care of chronically ill patients in Switzerland. Secondly, collaboration in different research projects consolidated her interests in family centred approaches to children and adult nursing care. For instance, the integration of parents and siblings of ill children into nursing care practice was described as significant in having positive effects by health professionals in paediatric care (Waldboth, Schluer, & Muller-Staub, 2013). Moreover, the researcher's personal experiences as a family caregiver caring for relatives with chronic illnesses and for a sibling suffering from limb-girdle MD contributed to her interest in this research area.
As a doctoral student and a family caregiver she brings possibilities and constraints to this investigation. There is a substantial need for emotional, intellectual and practical involvement with the families of concern. In contrast, it can be challenging not to be reserved or judgemental as a consequence of personal backgrounds. Working with experienced supervisors in study planning, data collection and analysis and regular and thorough reflection of the role of the researcher will be helpful to manage potentially difficult research phases. Detailed field notes are taken in order to make personal assumptions visible and transparent.

9. Quality criteria

Rigor in methods is planned to be ensured through in depth description of data collection and analysis processes throughout the research. Data analysis requires critical examination of the researcher's role by experienced researchers in order to strengthen the credibility of the study and respondent validation is assured using member checking after data analysis. Criteria for the evaluation of constructivist Grounded Theory research are presented in Charmaz (2006) and will be used to reflect strengths and limitations of the study throughout the research process. These criteria are credibility, originality, resonance and usefulness.

10. Time scale

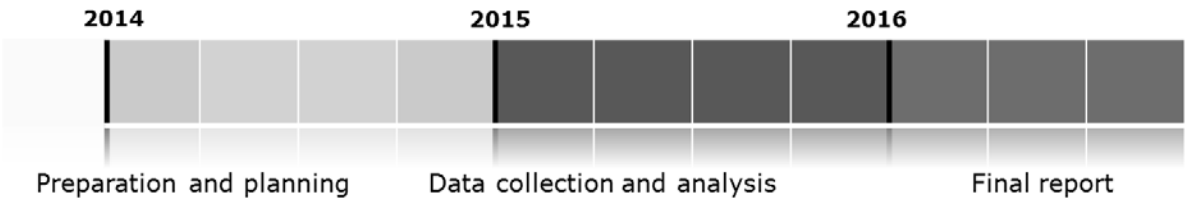

Figure 2: project barometer

Figure 2 and table 1 show preliminary planning and important milestones of the research project. The first milestone is the ethical approval from Ethics Committee Zurich in September 2014. Data collection and analysis then start approximately in September 2014 and take part simultaneously. After the end of data collection, analysis and theory construction in April 2016, the thesis will be written up. It is planned that the thesis will be submitted in September 2016.

Time scale	2014												2015												2016								
	1	2	3	4	5	6	7	8	9	10	11	12	1	2	3	4	5	6	7	8	9	10	11	12	1	2	3	4	5	6	7	8	9
1. preparation phase																																	
proposal writing	x	x	x	x	x																												
contact clinical site / identification of stakeholders	x	x	x	x	x		x		x		x		x		x		x				x				x				x				x
preparation of application for ethical approval		x	x	x	x																												
application for ethical approval					x	x	x	x	x																								
ehical approval									**x**	**x**																							
2. data collection																																	
analysis planning					x		x		x		x		x																				
participant recruitment									x	x	x	x	x	x	x	x	x	x	x	x	x	x											
interviews									x	x	x	x	x	x	x	x	x	x	x	x	x	x	x										
transcription									x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x									
quality assurance transcription										x	x	x	x	x	x	x	x	x	x	x	x	x	x	x									
completion data collection																								**x**									
3. data analysis																																	
data analysis										x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x					
memo writing										x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x					
theory development and theoretical sampling																				x	x	x	x	x	x	x	x						
interpretation of results																				x		x		x	x	x	x						
completion data analysis																												**x**					
4. dissemination process																																	
thesis writing									x		x		x		x		x		x		x		x	x	x	x	x	x	x	x	x	x	x
writing article																														x	x	x	x
presentation																															x	x	x
study conclusion																																	**x**

Table 1: Time scale

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Studieninformation für Kinder von 11 bis 13 Jahren

Erwachsen werden mit einer neuromuskulären Erkrankung

Studie: „Erwachsen werden: Erfahrungen von Menschen mit neuromuskulären Erkrankungen und deren Familien"

Du bist eingeladen, um an einer Studie teilzunehmen.

Bevor du Dich entscheidest an dieser Studie teilzunehmen, ist es wichtig, dass Du verstehst, warum die Studie gemacht wird und was auf Dich zukommt.

Dieses Informationsblatt soll Dich dabei unterstützen zu entscheiden, ob Du an der Studie teilnehmen möchtest oder nicht.

Bitte nimm Dir Zeit, um die nachfolgenden Informationen zu lesen und wenn Du Fragen hast, kannst Du sie mit Deiner Familie besprechen.

Was wird gemacht und warum?

Ich möchte mehr darüber wissen, wie es ist eine Muskelerkrankung zu haben. Dazu befrage ich Jugendliche, die an einer solchen Erkrankung leiden, und ihre Familien.

Warum gerade Du?

Du hast einen Bruder oder eine Schwester, die an einer Muskelkrankheit leiden und kannst uns davon erzählen. Wir haben mit Deinen Eltern gesprochen und die haben es erlaubt, dass auch Du an der Studie mitmachst.

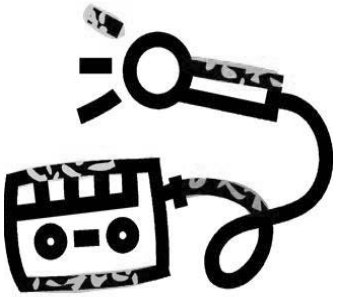
Was kommt auf Dich zu?

Wenn Du an der Studie teilnehmen möchtest, sag es Deinen Eltern. Dann werden Deine Eltern einen Termin mit mir vereinbaren, und wir treffen uns für ein Gespräch.

Was musst Du beim Gespräch machen?

Beim Gespräch werde ich Dir Fragen stellen wie zum Beispiel: „Wie sieht ein normaler Tag in Deinem Leben aus? Du darfst ein Bild von Dir und Deiner Familie zeichnen. Dann sprechen wir über deine Zeichnung.

Unser Gespräch dauert ungefähr eine Stunde. Gut wäre, wenn Du alleine mit mir sprichst.

Das Gespräch wird auf Tonband aufgenommen. Wir werden es dann anhören und abschreiben. Dein Name wird durch Zahlen ersetzt, sodass niemand weiss, dass Du das gesagt hast. Wir werden niemandem davon erzählen, was Du gesagt hast. Auch nicht deinen Eltern oder Geschwistern.	

Musst Du mitmachen?
Nein, Du allein entscheidest, ob Du mitmachen willst oder nicht.

Was nützt es Dir, wenn Du an der Studie teilnimmst?
Die Teilnahme an dieser Studie nützt Dir und Deiner Familie nicht direkt. Du trägst aber dazu bei, dass in Zukunft andere Personen mit Muskelerkrankungen unterstützt werden können. Es ist wichtig zu wissen, wie es ist mit einer Muskelerkrankung zu leben. Dann können Fachpersonen aus dem Gesundheitswesen (zum Beispiel Pflegefachfrauen wie ich) Personen, die an dieser Krankheit leiden und deren Familien besser unterstützen.

An wen kannst Du dich wenden?
Wenn du Fragen hast, wende Dich an deinen Eltern. Diese können Deine Fragen vielleicht beantworten oder sich dann an mich wenden:

Veronika Waldboth
Zürcher Hochschule für Angewandte Wissenschaften
Institut für Pflege
Technikumstrasse 71
CH- 8401 Winterthur

Tel. direkt: +41 58 934 6499
Email: veronika.waldboth@zhaw.ch

Zürcher Hochschule für Angewandte Wissenschaften
Institut für Pflege

Florence Nightingale School of Nursing and Midwifery
King's College London

Studieninformation für Eltern von betroffenen Personen

Erwachsen werden mit einer neuromuskulären Erkrankung

Studie: „Erwachsen werden: Erfahrungen von Menschen mit neuromuskulären Erkrankungen und deren Familien"

Sponsoren:
Institut für Pflege an der Zürcher Hochschule für Angewandte Wissenschaften (ZHAW) in Winterthur (CH) und Florence Nightingale School of Nursing and Midwifery am King's College in London (UK)

Inhaltsverzeichnis

Sehr geehrte Damen und Herren

Wir sind vom Institut für Pflege der Zürcher Hochschule für Angewandte Wissenschaften in Winterthur. Diese pflegewissenschaftliche Studie ist die Doktorarbeit in Pflege von Frau Veronika Waldboth und wird in Zusammenarbeit mit dem Institut für Pflege der Zürcher Hochschule für Angewandte Wissenschaften (ZHAW) in Winterthur und der Florence Nightingale School of Nursing and Midwifery am King's College in London durchgeführt. Frau Waldboth arbeitet als wissenschaftliche Mitarbeiterin an der ZHAW und wird das Projekt in der Schweiz umsetzen.

1. Auswahl der Personen, die an der Studie teilnehmen können

Es können alle in der Schweiz ansässigen Personen an dieser Studie teilnehmen, die an einer neuromuskulären Erkrankung leiden oder Angehörige (Eltern, Geschwister und andere nahestehende Angehörige) einer Person sind, die an einer neuromuskulären Erkrankung leidet. Die von der neuromuskulären Erkrankung betroffene Person muss zwischen 14 bis 30 Jahre alt sein und die ersten Symptome der Erkrankung müssen bereits im Kindesalter aufgetreten sein (zwischen 0 bis zehn Jahren). Derzeit soll eine mässige bis starke körperliche Einschränkung vorliegen.
Nicht teilnehmen dürfen Personen, die kein deutsch sprechen und die an einer ausgeprägten geistigen Einschränkung leiden, die es der Person nicht ermöglicht zur Teilnahme an der Studie einzuwilligen.

2. Ziel der Studie

Ziel dieser Studie ist es, die Erfahrungen von Menschen mit genetisch bedingten neuromuskulären Erkrankungen zu erfassen, die spezifisch für den Übergang vom Kindes- zum Erwachsenenalter sind. Feinziele dieser Studie sind es, die Herausforderungen zu verstehen, mit denen betroffene Familien während dieser Entwicklungsphase konfrontiert sind; Strategien zu identifizieren, mit denen sich betroffene Familien erfolgreich an veränderte Situationen anpassen; Faktoren zu identifizieren, die die Anpassung an veränderte Situationen beeinflussen und Erfahrungen der betroffenen Familien mit dem Schweizerischen Gesundheitssystem zu verstehen.
Diese Erkenntnisse sind die Voraussetzung für eine mögliche Verbesserung der pflegerischen Versorgung der Betroffenen sowohl in Spitälern und Institutionen als auch in deren häuslichem Umfeld. Sie fliessen in die zukünftige Entwicklung von familienzentrierten pflegerischen Angeboten für junge Menschen mit neuromuskulären Erkrankungen und für deren Familien mit ein.

3. Allgemeine Informationen zur Studie

Hintergrund

Diese pflegewissenschaftliche Studie untersucht Erfahrungen von Jugendlichen und jungen Erwachsenen, die an einer neuromuskulären Erkrankung leiden. Das „Erwachsenwerden" ist eine Zeit des Übergangs vom Kindes- zum Erwachsenenalter und gleichzeitig eine Zeit der Entwicklung und Entfaltung der eigenen Persönlichkeit. Junge Menschen, die an einer chronischen Erkrankung leiden, die sich bereits im Kindesalter manifestiert, erleben diese Zeit anders als ihre gesunden Mitmenschen. Für Personen, die an Muskeldystrophie leiden bedeutet es, dass in einer Zeit, in der von ihnen erwartet wird, dass sie zunehmend unabhängiger werden, ihre körperliche Einschränkung und Abhängigkeit von Unterstützung zunimmt. Genetische Erkrankungen, die sich im Kindesalter manifestieren, haben zudem grosse Auswirkungen auf die Familie oder auf

nahestehende Personen. Aus diesem Grund werden Erfahrungen von Familienmitgliedern und nahestehenden Personen in die Untersuchung mit eingeschlossen.

Methodisches Vorgehen
Diese pflegewissenschaftliche Studie hat einen qualitativen Ansatz. Das bedeutet, dass Daten in erster Linie durch Einzelgespräche mit den betroffenen jungen Personen und deren Angehörigen erhoben werden. Die Anzahl der Teilnehmende wird auf ungefähr 40 Personen (8 bis 15 Familien) geschätzt. Teilnehmenden machen während des Gesprächs Angaben zu ihren Erfahrungen und zu ihrer Person. Das Gespräch dauert ungefähr eine Stunde. Einzelgespräche mit Kindern werden nur nach informierter Zustimmung des Kindes und des jeweiligen Erziehungsberechtigten durchgeführt. Die Interviews werden an einem von der teilnehmenden Person gewünschtem Ort durchgeführt, vorzugsweise in deren familiären Umfeld.

Ablauf der Studie
Die Gesamtdauer der Studie ist auf drei Jahre festgelegt. Die Planungs- und Vorbereitungsphase begann im September 2013. Der Beginn der Datensammlung ist auf den September 2014 festgelegt und die Datensammlung dauert ungefähr ein Jahr. Der Durchführungsort der Datensammlung ist die Deutschschweiz. Gleichzeitig mit der Datensammlung werden die Daten analysiert, sodass erste Ergebnisse im Frühjahr 2016 vorliegen. Eine Vorstellung der vorläufigen Resultate ist für das Frühjahr 2016 geplant, zu der alle Studienteilnehmenden eingeladen sind. Das planmäßige Studienende wurde auf September 2016 festgelegt.
Wir machen diese Studie so, wie es die Gesetze in der Schweiz vorschreiben. Ausserdem beachten wir alle international anerkannten Richtlinien. Die zuständige Kantonale Ethikkommission hat die Studie geprüft und bewilligt.

4. Ablauf für die Teilnehmenden

Sie sind an einer Teilnahme an dieser Studie interessiert und haben bereits Kontakt mit der Studienleiterin aufgenommen. Diese hat Sie dann kontaktiert und Ihnen mit Ihrer Zustimmung diese schriftliche Studieninformation und Einwilligungserklärung per Post zukommen lassen. Sie werden in den nächsten Wochen erneut kontaktiert und bei bestehendem Interesse können Sie mit der Studienleiterin einen Termin für ein Gespräch vereinbaren.

Mit Ihrer Einwilligung zur Teilnahme an dieser Studie erklären Sie Sich zu einem ungefähr stündigen Einzelgespräch bereit, dass eine wissenschaftliche Mitarbeiterin mit Ihnen führt und das auf Tonband aufgenommen wird. Mit der Durchführung dieses Interviews endet Ihre Studienteilnahme.

5. Rechte der Teilnehmenden

Sie nehmen nur dann an dieser Studie teil, wenn Sie es wollen. Niemand darf Sie dazu in irgendeiner Weise drängen oder dazu überreden. Sie müssen nicht begründen, warum Sie nicht mitmachen wollen. Wenn Sie Sich entscheiden mitzumachen, können Sie diesen Entscheid jederzeit zurücknehmen. Sie müssen ebenfalls nicht begründen, wenn Sie aus der Studie aussteigen wollen.
Sie dürfen jederzeit alle Fragen zur Studie stellen. Wenden Sie Sich dazu bitte an eine der Personen, die am Ende dieser Studieninformation genannt sind.

6. Pflichten der Teilnehmenden

Wenn Sie bei der Studie mitmachen, müssen Sie bestimmte Regeln beachten. Dies ist notwendig für Ihre Sicherheit und Gesundheit. Wir werden Sie dabei so gut wir können unterstützen. Als Studienteilnehmende/r sind Sie verpflichtet:

- Sich für ein stündiges Gespräch zur Verfügung zu stellen und Angaben zu Ihren Erfahrungen und Ihrer Person zu machen;
- Informationen über die Studie mittels Studienflyer an Personen weiterzuleiten, die ebenfalls wichtige Gesprächspartner für die Studie sein könnten;
- Den Anweisungen der Studienleiterin zu folgen und sie über Änderungen im Befinden zu informieren, die für die Studie relevant sind.

Wenn Sie diese Pflichten nicht beachten, können Sie Haftungsansprüche verlieren.

7. Nutzen für die Teilnehmenden

Die Teilnahme an dieser Studie nützt Ihnen und Ihrer Familie nicht direkt. Sie tragen aber dazu bei, dass in Zukunft andere Familien mit Muskelerkrankungen unterstützt werden können.
Die Studie liefert wichtige Erkenntnisse über die Erfahrungen von jungen Menschen mit neuromuskulären Erkrankungen und deren Familien über den Übergang vom Kind zum Erwachsenen. Diese Erkenntnisse sind die Voraussetzung für eine mögliche Verbesserung der pflegerischen Versorgung der Betroffenen in dieser Lebensphase. Sie fliessen in die zukünftige Entwicklung von familienzentrierten pflegerischen Angeboten für junge Menschen mit neuromuskulären Erkrankungen und für deren Familien mit ein.

8. Risiken und Belastungen für die Teilnehmenden

Die Teilnahme an dieser Studie sollte für Sie kein Risiko darstellen. Sie erklären sich zu einem Gespräch bereit, das ungefähr eine Stunde dauert. Dieses Gespräch kann jedoch verschiedene Emotionen auslösen. Insgesamt gehen wir aber davon aus, dass es sich positiv auswirkt, wenn Sie Sich in einem persönlichen Gespräch über Ihre Erfahrungen ausdrücken können. Im Rahmen von Fragen oder beim Auftreten von negativen Emotionen während oder nach dem Gespräch verweisen wir Sie an eine entsprechende Beratungsstelle weiter.

9. Ergebnisse aus der Studie

Die Studienleiterin wird Sie während der Studie über alle neuen Erkenntnisse informieren, die den Nutzen der Studie oder Ihre Sicherheit und somit Ihre Einwilligung zur Teilnahme an der Studie beeinflussen können. Sie werden diese Information mündlich und schriftlich erhalten.

10. Vertraulichkeit der Daten

Wir werden für diese Studie Daten zu Ihren Erfahrungen und zu Ihrer Person erfassen. Diese Daten werden wir verschlüsseln, d.h. wir werden anstelle Ihres vollen Namens nur eine dreistellige Zahl verwenden, um Sie zu kennzeichnen. Zudem werden in den Interviews Personen-, Orts- und Institutionsnamen, sowie Datumsangaben und andere Sachverhalte, die eine Identifizierung der Person ermöglichen würden, verschlüsselt. Einzig die Studienleiterin weiss, wer oder was sich hinter dieser Verschlüsselung verbirgt. Andere Wissenschaftler, die an dieser Studie beteiligt sind, werden nur mit den verschlüsselten Daten arbeiten.
Alle Personen, die mit der Studie in irgendeiner Weise zu tun haben, verpflichten sich absolute Vertraulichkeit zu wahren. Wir werden Namen von Teilnehmenden nirgends, in keinem Bericht, keiner Publikation, nicht gedruckt und nicht im Internet, veröffentlichen.
Es kann sein, dass die Studie während des Ablaufs überprüft wird. Dies können die Behörden tun, die sie vorab kontrolliert und bewilligt haben. Auch diejenige Institution, die die Studie bezahlt, kann den Ablauf überprüfen lassen. Sie alle sorgen dafür, dass die Regeln eingehalten werden und Ihre Sicherheit nicht gefährdet wird. Dazu muss die Leiterin der Studie eventuell Ihre persönlichen Daten für solche Kontrollen offenlegen. Ebenso kann es sein, dass im Fall eines Schadens ein Vertreter der Versicherung Ihre Daten ansehen muss. Das darf dann aber nur die Daten betreffen, die unbedingt gebraucht werden, um den Schadensfall zu erledigen.
Die erhobenen Daten müssen aus Gründen der Nachvollziehbarkeit zehn Jahre im abgeschlossenen Archiv der ZHAW aufbewahrt werden.

11. Weitere Verwendung von Daten

Sie können jederzeit aus der Studie aussteigen, wenn Sie dies wünschen. Sie müssen Ihre Entscheidungen nicht begründen. Wenn Sie später aussteigen, werden wir Daten, die wir bis dahin erhoben haben, zu Ende auswerten. Diese Daten stellen einen wichtigen Bestandteil der Ergebnisse dar und können nicht mehr ausgeschlossen werden, weil sonst die ganze Studie ihren Wert verlieren würde. Wir werden Ihre Daten dann jedoch vollständig anonymisieren, d.h. wir werden endgültig Ihren Namen und die Verschlüsselungsnummer darauf löschen. Niemand wird danach mehr erfahren können, dass die Daten von Ihnen stammten. Prüfen Sie bitte, ob Sie damit einverstanden sein können, bevor Sie bei der Studie mitmachen.

12. Entschädigung für Teilnehmende

Wenn Sie bei dieser Studie mitmachen, bekommen Sie dafür keine Entschädigung. Allfällige Reisespesen werden nicht übernommen.

13. Deckung von Schäden

Falls Sie durch die Studie einen gesundheitlichen Schaden erleiden, haftet die Institution, die für die Durchführung der Studie verantwortlich ist. Diese Haftung gilt aber nur dann, wenn Sie nachweisen können, dass der Schaden auf die Studie und damit verbundene Handlungen zurückzuführen ist. Die Zürcher Hochschule für Angewandte Wissenschaften (Technikumstr. 71, CH-8401 Winterthur) hat eine Versicherung bei der Zürich Versicherungs-Gesellschaft AG (Mythenquai 2, 8002 Zürich) abgeschlossen, um im Schadensfall für die Haftung aufkommen zu können. Wenn Sie einen Schaden erlitten haben, so wenden Sie Sich bitte an die Leiterin oder Prüferin der Studie.

14. Finanzierung der Studie

Die Studie wird mehrheitlich von der Studienleiterin, Frau Waldboth, finanziert, die im Rahmen ihrer Doktorarbeit von der Zürcher Hochschule für Angewandte Wissenschaften teilfinanziert ist. Ein Ansuchen für eine weitere Finanzierung durch einen zusätzlichen Sponsor ist zukünftig angestrebt.

15. Kontaktpersonen

Bei Fragen oder Unklarheiten, die während der Studie oder danach auftreten, können Sie Sich jederzeit an eine dieser Kontaktpersonen wenden:

Studienleiterin
Veronika Waldboth
Wissenschaftliche Mitarbeiterin
Zürcher Hochschule für Angewandte Wissenschaften
Institut für Pflege
Technikumstr. 71
CH- 8401 Winterthur

Tel. +41 58 934 6499
Email: veronika.waldboth@zhaw.ch

Prüferin der Studie
Prof Dr. Romy Mahrer-Imhof
Leiterin Master of Science in Pflege
Zürcher Hochschule für Angewandte Wissenschaften
Institut für Pflege
Technikumstr. 71
CH-8401 Winterthur

Tel. +41 58 934 6344
Email: romy.mahrer@zhaw.ch

Einwilligungserklärung für Eltern von betroffenen Personen

Erwachsen werden mit einer neuromuskulären Erkrankung

Studie: „Erwachsen werden: Erfahrungen von Menschen mit neuromuskulären Erkrankungen und deren Familien“

- Bitte lesen Sie dieses Formular sorgfältig durch.
- Bitte fragen Sie, wenn Sie etwas nicht verstehen oder wissen möchten.

Titel der Studie: Erwachsen werden mit einer neuromuskulären Erkrankung Erwachsen werden: Erfahrungen von Menschen mit neuromuskulären Erkrankungen und deren Familien	
Verantwortliche Institution (Sponsor)	Zürcher Hochschule für Angewandte Wissenschaften Departement Gesundheit Institut für Pflege Technikumstrasse 71 8401 Winterthur
Ort der Durchführung:	Schweiz
Leiterin der Studie	Veronika Waldboth
Teilnehmerin / Teilnehmer	Name und Vorname (bitte in Druckbuchstaben): ***X*** _____ Geburtsdatum (Tag /Monat/ Jahr): ***X*** _____ Geschlecht (bitte ankreuzen): ☐ weiblich ☐ männlich

- Ich wurde von der unterzeichnenden Studienleiterin mündlich und schriftlich über den Zweck, den Ablauf der Studie, über mögliche Vor- und Nachteile sowie über eventuelle Risiken informiert.

- Meine Fragen im Zusammenhang mit der Teilnahme an dieser Studie sind mir zufriedenstellend beantwortet worden. Ich kann die schriftliche Studieninformation vom 25.03.15, Version 3 behalten und erhalte eine Kopie meiner schriftlichen Einwilligungserklärung. Ich akzeptiere den Inhalt der zur oben genannten Studie abgegebenen schriftlichen Studieninformation.

- Ich nehme an dieser Studie freiwillig teil. Ich kann jederzeit und ohne Angabe von Gründen meine Zustimmung zur Teilnahme widerrufen, ohne dass ich deswegen Nachteile bei der weiteren medizinischen Betreuung erleide.

- Ich hatte genügend Zeit, meine Entscheidung zu treffen.

- Ich bin darüber informiert, dass eine Versicherung Schäden deckt, falls ich nachweisen kann, dass die Schäden auf die Studie zurückzuführen sind.

- Ich weiss, dass meine persönlichen Daten nur in verschlüsselter Form zu Forschungszwecken weitergegeben werden können. Ich bin einverstanden, dass die zuständigen Sponsoren der Studie, der Behörden und der Kantonalen Ethikkommission zu Prüf- und Kontrollzwecken in meine Originaldaten Einsicht nehmen dürfen, jedoch unter strikter Einhaltung der Vertraulichkeit.

- Ich bin mir bewusst, dass die in der Teilnehmerinformation genannten Pflichten während der Studie einzuhalten sind. Im Interesse meiner Gesundheit kann die Leiterin der Studie mich jederzeit von der Studie ausschliessen.

Ort, Datum	Unterschrift Studienteilnehmerin/Studienteilnehmer
X _____	***X*** _____

Bestätigung der Studienleiterin: Hiermit bestätige ich, dass ich dieser Teilnehmerin/diesem Teilnehmer Wesen, Bedeutung und Tragweite der Studie erläutert habe. Ich versichere, alle im Zusammenhang mit dieser Studie stehenden Verpflichtungen gemäss dem geltenden Recht zu erfüllen. Sollte ich zu irgendeinem Zeitpunkt während der Durchführung der Studie von Aspekten erfahren, welche die Bereitschaft der Teilnehmerin/des Teilnehmers zur Teilnahme an der Studie beeinflussen könnte, werde ich sie/ihn umgehend darüber informieren.

Ort, Datum	Unterschrift der Studienleiterin

Zürcher Hochschule für Angewandte Wissenschaften
Institut für Pflege

Florence Nightingale School of Nursing and Midwifery
King's College London

Studieninformation für Sorgeberechtigte von Kindern von 8 bis 13 Jahren

Erwachsen werden mit einer neuromuskulären Erkrankung

Studie: „Erwachsen werden: Erfahrungen von Menschen mit neuromuskulären Erkrankungen und deren Familien"

Sponsoren:
Institut für Pflege an der Zürcher Hochschule für Angewandte Wissenschaften (ZHAW) in Winterthur (CH) und Florence Nightingale School of Nursing and Midwifery am King's College in London (UK)

Inhaltsverzeichnis

Sehr geehrte Damen und Herren

Wir sind vom Institut für Pflege der Zürcher Hochschule für Angewandte Wissenschaften in Winterthur. Diese pflegewissenschaftliche Studie ist die Doktorarbeit in Pflege von Frau Veronika Waldboth und wird in Zusammenarbeit mit dem Institut für Pflege der Zürcher Hochschule für Angewandte Wissenschaften (ZHAW) in Winterthur und der Florence Nightingale School of Nursing and Midwifery am King's College in London durchgeführt. Frau Waldboth arbeitet als wissenschaftliche Mitarbeiterin an der ZHAW und wird das Projekt in der Schweiz umsetzen.

1. Auswahl der Personen, die an der Studie teilnehmen können

Es können alle in der Schweiz ansässigen Personen an dieser Studie teilnehmen, die an einer neuromuskulären Erkrankung leiden oder Angehörige (Eltern, Geschwister und andere nahestehende Angehörige) einer Person sind, die an einer neuromuskulären Erkrankung leidet. Die von der neuromuskulären Erkrankung betroffene Person muss zwischen 14 bis 30 Jahre alt sein und die ersten Symptome der Erkrankung müssen bereits im Kindesalter aufgetreten sein (zwischen 0 bis zehn Jahren). Derzeit soll eine mässige bis starke körperliche Einschränkung vorliegen.
Nicht teilnehmen dürfen Personen, die kein deutsch sprechen und die an einer ausgeprägten geistigen Einschränkung leiden, die es der Person nicht ermöglicht zur Teilnahme an der Studie einzuwilligen.

2. Ziel der Studie

Ziel dieser Studie ist es, die Erfahrungen von Menschen mit genetisch bedingten neuromuskulären Erkrankungen zu erfassen, die spezifisch für den Übergang vom Kindes- zum Erwachsenenalter sind. Feinziele dieser Studie sind es, die Herausforderungen zu verstehen, mit denen betroffene Familien während dieser Entwicklungsphase konfrontiert sind; Strategien zu identifizieren, mit denen sich betroffene Familien erfolgreich an veränderte Situationen anpassen; Faktoren zu identifizieren, die die Anpassung an veränderte Situationen beeinflussen und Erfahrungen der betroffenen Familien mit dem Schweizerischen Gesundheitssystem zu verstehen.
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3. Allgemeine Informationen zur Studie

Hintergrund

Diese pflegewissenschaftliche Studie untersucht Erfahrungen von Jugendlichen und jungen Erwachsenen, die an einer neuromuskulären Erkrankung leiden. Das „Erwachsenwerden" ist eine Zeit des Übergangs vom Kindes- zum Erwachsenenalter und gleichzeitig eine Zeit der Entwicklung und Entfaltung der eigenen Persönlichkeit. Junge Menschen, die an einer chronischen Erkrankung leiden, die sich bereits im Kindesalter manifestiert, erleben diese Zeit anders als ihre gesunden Mitmenschen. Für Personen, die an Muskeldystrophie leiden bedeutet es, dass in einer Zeit, in der von ihnen erwartet wird, dass sie zunehmend unabhängiger werden, ihre körperliche Einschränkung und Abhängigkeit von Unterstützung zunimmt. Genetische Erkrankungen, die sich

im Kindesalter manifestieren, haben zudem grosse Auswirkungen auf die Familie oder auf nahestehende Personen. Aus diesem Grund werden Erfahrungen von Familienmitgliedern und nahestehenden Personen in die Untersuchung mit eingeschlossen.

Methodisches Vorgehen
Diese pflegewissenschaftliche Studie hat einen qualitativen Ansatz. Das bedeutet, dass Daten in erster Linie durch Einzelgespräche mit den betroffenen jungen Personen und deren Angehörigen erhoben werden. Die Anzahl der Teilnehmenden wird auf ungefähr 40 Personen (8 bis 15 Familien) geschätzt. Teilnehmende machen während des Gesprächs Angaben zu ihren Erfahrungen und zu ihrer Person. Das Gespräch dauert ungefähr eine Stunde. Einzelgespräche mit Kindern werden nur nach informierter Zustimmung des Kindes und des jeweiligen Erziehungsberechtigten durchgeführt. Die Interviews werden an einem von der teilnehmenden Person gewünschtem Ort durchgeführt, vorzugsweise in deren familiären Umfeld.

Ablauf der Studie
Die Gesamtdauer der Studie ist auf drei Jahre festgelegt. Die Planungs- und Vorbereitungsphase begann im September 2013. Der Beginn der Datensammlung ist auf den September 2014 festgelegt und die Datensammlung dauert ungefähr ein Jahr. Der Durchführungsort der Datensammlung ist die Deutschschweiz. Gleichzeitig mit der Datensammlung werden die Daten analysiert, sodass erste Ergebnisse im Frühjahr 2016 vorliegen. Eine Vorstellung der vorläufigen Resultate ist für das Frühjahr 2016 geplant, zu der alle Studienteilnehmenden eingeladen sind. Das planmäßige Studienende wurde auf September 2016 festgelegt.
Wir machen diese Studie so, wie es die Gesetze in der Schweiz vorschreiben. Ausserdem beachten wir alle international anerkannten Richtlinien. Die zuständige Kantonale Ethikkommission hat die Studie geprüft und bewilligt.

4. Ablauf für die Teilnehmenden

Ihr Kind ist an einer Teilnahme an dieser Studie interessiert und Sie oder Ihr Kind haben bereits Kontakt mit der Studienleiterin aufgenommen. Diese hat Sie dann kontaktiert und Ihnen und Ihrem Kind diese schriftliche Studieninformation für Sorgeberechtigte, eine Einwilligungserklärung, sowie falls zutreffend eine Studieninformation für Kinder von 11-13 Jahren per Post zukommen lassen. Da Ihr Kind jünger als 14 Jahre nicht volljährig ist, müssen Sie als Elternteil oder Sorgeberechtigte Person mit seiner Teilnahme an dieser Studie einverstanden sein und schriftlich dazu einwilligen. Die Studienleiterin wird Sie und Ihr Kind in den nächsten Wochen erneut kontaktieren und bei bestehendem Interesse und mit Ihrer Einwilligung und der Einwilligung Ihres Kindes mit Ihrem Kind einen Termin für ein Gespräch vereinbaren.
Mit der Einwilligung Ihres Kindes zur Teilnahme an dieser Studie erklärt es sich zu einem ungefähr stündigen Einzelgespräch bereit, dass eine wissenschaftliche Mitarbeiterin mit ihm führt und das auf Tonband aufgenommen wird. Mit der Durchführung dieses Interviews endet die Studienteilnahme.

5. Rechte der Teilnehmenden

Ihr Kind nimmt nur dann an dieser Studie teil, wenn es teilnehmen will. Niemand darf es dazu in irgendeiner Weise drängen oder dazu überreden. Sie oder Ihr Kind müssen nicht begründen, warum es nicht mitmachen will. Wenn Ihr Kind sich entscheidet mitzumachen und Sie dieser Teilnahme zustimmen, können Sie und Ihr Kind diesen Entscheid jederzeit zurücknehmen. Sie und Ihr Kind müssen ebenfalls nicht begründen, wenn Ihr Kind aus der Studie aussteigen will.

Sie und Ihr Kind dürfen jederzeit alle Fragen zur Studie stellen. Wenden Sie Sich dazu bitte an eine der Personen, die am Ende dieser Studieninformation genannt sind.

6. Pflichten der Teilnehmenden

Wenn Ihr Kind bei der Studie mitmacht, muss es bestimmte Regeln beachten. Dies ist notwendig für seine Sicherheit und Gesundheit. Wir werden Ihr Kind dabei so gut wir können unterstützen. Studienteilnehmende sind verpflichtet:

- Sich für ein stündiges Gespräch zur Verfügung zu stellen und Angaben zu ihren Erfahrungen und ihrer Person zu machen;
- Informationen über die Studie mittels Studienflyer an Personen weiterzuleiten, die ebenfalls wichtige Gesprächspartner für die Studie sein könnten;
- Den Anweisungen der Studienleiterin zu folgen und sie über Änderungen im Befinden zu informieren, die für die Studie relevant sind.

Wenn die teilnehmende Person diese Pflichten nicht beachtet, kann sie Haftungsansprüche verlieren.

7. Nutzen für die Teilnehmenden

Die Teilnahme an dieser Studie nützt Ihrem Kind und Ihrer Familie nicht direkt. Ihr Kind trägt aber dazu bei, dass in Zukunft andere Familien mit Muskelerkrankungen unterstützt werden können. Die Studie liefert wichtige Erkenntnisse über die Erfahrungen von jungen Menschen mit neuromuskulären Erkrankungen und deren Familien über den Übergang vom Kind zum Erwachsenen. Diese Erkenntnisse sind die Voraussetzung für eine mögliche Verbesserung der pflegerischen Versorgung der Betroffenen in dieser Lebensphase. Sie fliessen in die zukünftige Entwicklung von familienzentrierten pflegerischen Angeboten für junge Menschen mit neuromuskulären Erkrankungen und für deren Familien mit ein.

8. Risiken und Belastungen für die Teilnehmenden

Die Teilnahme an dieser Studie sollte für die Teilnehmenden kein Risiko darstellen. Ihr Kind erklärt sich zu einem Gespräch bereit, das ungefähr eine Stunde dauert. Dieses Gespräch kann jedoch verschiedene Emotionen auslösen. Insgesamt gehen wir aber davon aus, dass es sich positiv auswirkt, wenn sich Ihr Kind in einem persönlichen Gespräch über seine Erfahrungen ausdrücken kann. Im Rahmen von Fragen oder beim Auftreten von negativen Emotionen während oder nach dem Gespräch verweisen wir Ihr Kind an eine entsprechende Beratungsstelle weiter.

9. Ergebnisse aus der Studie

Die Studienleiterin wird Sie und Ihr Kind während der Studie über alle neuen Erkenntnisse informieren, die den Nutzen der Studie oder die Sicherheit Ihres Kindes und somit seine Einwilligung zur Teilnahme an der Studie beeinflussen können. Sie werden diese Information mündlich und schriftlich erhalten.

10. Vertraulichkeit der Daten

Wir werden für diese Studie Daten zu den Erfahrungen und zur Person Ihres Kindes erfassen. Diese Daten werden wir verschlüsseln, d.h. wir werden anstelle des vollen Namens nur eine dreistellige Zahl verwenden, um Teilnehmende zu kennzeichnen. Zudem werden in den Interviews

Personen-, Orts- und Institutionsnamen, sowie Datumsangaben und andere Sachverhalte, die eine Identifizierung der Person ermöglichen würden, verschlüsselt. Einzig die Studienleiterin weiss, wer oder was sich hinter dieser Verschlüsselung verbirgt. Andere Wissenschaftler, die an dieser Studie beteiligt sind, werden nur mit den verschlüsselten Daten arbeiten.
Alle Personen, die mit der Studie in irgendeiner Weise zu tun haben, verpflichten sich absolute Vertraulichkeit zu wahren. Wir werden Namen von Teilnehmenden nirgends, in keinem Bericht, keiner Publikation, nicht gedruckt und nicht im Internet, veröffentlichen.
Es kann sein, dass die Studie während des Ablaufs überprüft wird. Dies können die Behörden tun, die sie vorab kontrolliert und bewilligt haben. Auch diejenige Institution, die die Studie bezahlt, kann den Ablauf überprüfen lassen. Sie alle sorgen dafür, dass die Regeln eingehalten werden und die Sicherheit der Teilnehmenden nicht gefährdet wird. Dazu muss die Leiterin der Studie eventuell persönlichen Daten Ihres Kindes für solche Kontrollen offenlegen. Ebenso kann es sein, dass im Fall eines Schadens ein Vertreter der Versicherung Daten Ihres Kindes ansehen muss. Das darf dann aber nur die Daten betreffen, die unbedingt gebraucht werden, um den Schadensfall zu erledigen.
Die erhobenen Daten müssen aus Gründen der Nachvollziehbarkeit zehn Jahre im abgeschlossenen Archiv der ZHAW aufbewahrt werden.

11. Weitere Verwendung von Daten

Ihr Kind kann jederzeit aus der Studie aussteigen, wenn Sie oder Ihr Kind dies wünschen. Sie oder Ihr Kind müssen diese Entscheidungen nicht begründen. Wenn Ihr Kind später aussteigt, werden wir Daten, die wir bis dahin erhoben haben, zu Ende auswerten. Diese Daten stellen einen wichtigen Bestandteil der Ergebnisse dar und können nicht mehr ausgeschlossen werden, weil sonst die ganze Studie ihren Wert verlieren würde. Wir werden die Daten Ihres Kindes dann jedoch vollständig anonymisieren, d.h. wir werden endgültig die Namen und die Verschlüsselungsnummer darauf löschen. Niemand wird danach mehr erfahren können, dass die Daten von Ihrem Kind stammten. Prüfen Sie und Ihr Kind bitte, ob Sie damit einverstanden sein können, bevor Ihr Kind bei der Studie mitmacht.

12. Entschädigung für Teilnehmende

Wenn Ihr Kind bei dieser Studie mitmacht, bekommt es dafür keine Entschädigung. Allfällige Reisespesen werden nicht übernommen.

13. Deckung von Schäden

Falls Ihr Kind durch die Studie einen gesundheitlichen Schaden erleidet, haftet die Institution, die für die Durchführung der Studie verantwortlich ist. Diese Haftung gilt aber nur dann, wenn Sie nachweisen können, dass der Schaden auf die Studie und damit verbundene Handlungen zurückzuführen ist.
Die Zürcher Hochschule für Angewandte Wissenschaften (Technikumstr. 71, CH-8401 Winterthur) hat eine Versicherung bei der Zürich Versicherungs-Gesellschaft AG (Mythenquai 2, 8002 Zürich) abgeschlossen, um im Schadensfall für die Haftung aufkommen zu können. Wenn Ihr Kind einen Schaden erlitten hat, so wenden Sie sich bitte an die Leiterin oder Prüferin der Studie.

14. Finanzierung der Studie

Die Studie wird mehrheitlich von der Studienleiterin, Frau Waldboth, finanziert, die im Rahmen ihrer Doktorarbeit von der Zürcher Hochschule für Angewandte Wissenschaften teilfinanziert ist. Ein Ansuchen für eine weitere Finanzierung durch einen zusätzlichen Sponsor ist zukünftig angestrebt.

15. Kontaktpersonen

Bei Fragen oder Unklarheiten, die während der Studie oder danach auftreten, können Sie Sich jederzeit an eine dieser Kontaktpersonen wenden:

Studienleiterin
Veronika Waldboth
Wissenschaftliche Mitarbeiterin
Zürcher Hochschule für Angewandte Wissenschaften
Institut für Pflege
Technikumstr. 71
CH- 8401 Winterthur

Tel. +41 58 934 6499
Email: veronika.waldboth@zhaw.ch

Prüferin der Studie
Prof Dr. Romy Mahrer-Imhof
Leiterin Master of Science in Pflege
Zürcher Hochschule für Angewandte Wissenschaften
Institut für Pflege
Technikumstr. 71
CH-8401 Winterthur

Tel. +41 58 934 63 44
Email: romy.mahrer@zhaw.ch

Einwilligungserklärung für Sorgeberechtigte von Kindern von 8 bis 13 Jahren

Erwachsen werden mit einer neuromuskulären Erkrankung

Studie: „Erwachsen werden: Erfahrungen von Menschen mit neuromuskulären Erkrankungen und deren Familien“

- Bitte lesen Sie dieses Formular sorgfältig durch.
- Bitte fragen Sie, wenn Sie etwas nicht verstehen oder wissen möchten.

Titel der Studie: Erwachsen werden mit einer neuromuskulären Erkrankung Erwachsen werden: Erfahrungen von Menschen mit neuromuskulären Erkrankungen und deren Familien	
Verantwortliche Institution (Sponsor)	Zürcher Hochschule für Angewandte Wissenschaften Departement Gesundheit Institut für Pflege Technikumstrasse 71 8401 Winterthur
Ort der Durchführung:	Schweiz
Leiterin der Studie	Veronika Waldboth
Teilnehmerin / Teilnehmer	Name und Vorname der/des Studienteilnehmerin/Studienteilnehmers /(bitte in Druckbuchstaben): ***X*** _____ Geburtsdatum (Tag /Monat/ Jahr): ***X*** _____ Geschlecht (bitte ankreuzen): ☐ weiblich ☐ männlich

- Ich und mein Kind wurden von der unterzeichnenden Studienleiterin mündlich und schriftlich über den Zweck, den Ablauf der Studie, über mögliche Vor- und Nachteile sowie über eventuelle Risiken informiert.

- Meine Fragen im Zusammenhang mit der Teilnahme meines Kindes an dieser Studie sind mir zufriedenstellend beantwortet worden. Ich kann die schriftliche Studieninformation vom 25.03.2015, Version 3 behalten und erhalte eine Kopie meiner schriftlichen Einwilligungserklärung. Ich akzeptiere den Inhalt der zur oben genannten Studie abgegebenen schriftlichen Studieninformation.

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Ort, Datum ***X***_____	Name und Vorname der sorgeberechtigten Person der/des Studienteilnehmerin/Studienteilnehmers (bitte in Druckbuchstaben): ***X***_____ Unterschrift der sorgeberechtigten Person der/des Studienteilnehmerin/Studienteilnehmers: ***X***_____

Bestätigung der Studienleiterin: Hiermit bestätige ich, dass ich dieser Teilnehmerin/diesem Teilnehmer und deren sorgeberechtigter Person Wesen, Bedeutung und Tragweite der Studie erläutert habe. Ich versichere, alle im Zusammenhang mit dieser Studie stehenden Verpflichtungen gemäss dem geltenden Recht zu erfüllen. Sollte ich zu irgendeinem Zeitpunkt während der Durchführung der Studie von Aspekten erfahren, welche die Bereitschaft der Teilnehmerin/des Teilnehmers zur Teilnahme an der Studie beeinflussen könnte, werde ich sie/ihn und seine/ihre sorgeberechtigte Person umgehend darüber informieren.

Ort, Datum	Unterschrift der Studienleiterin

Zürcher Hochschule für Angewandte Wissenschaften
Institut für Pflege

Florence Nightingale School of Nursing and Midwifery
King's College London

Studieninformation für Teilnehmende von 14-17 Jahren

Erwachsen werden mit einer neuromuskulären Erkrankung

Studie: „Erwachsen werden: Erfahrungen von Menschen mit neuromuskulären Erkrankungen und deren Familien"

Sponsoren:
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Inhaltsverzeichnis

Lieber/e Jugendliche

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Einschränkung und Abhängigkeit von Unterstützung zunimmt. Genetische Erkrankungen, die sich im Kindesalter manifestieren, haben zudem grosse Auswirkungen auf die Familie oder auf nahestehende Personen. Aus diesem Grund werden Erfahrungen von Familienmitgliedern und nahestehenden Personen in die Untersuchung mit eingeschlossen.

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Diese pflegewissenschaftliche Studie hat einen qualitativen Ansatz. Das bedeutet, dass Daten in erster Linie durch Einzelgespräche mit den betroffenen jungen Personen und deren Angehörigen erhoben werden. Die Anzahl der Teilnehmenden wird auf ungefähr 40 Personen (8 bis 15 Familien) geschätzt. Teilnehmende machen während des Gesprächs Angaben zu ihren Erfahrungen und zu ihrer Person. Das Gespräch dauert ungefähr eine Stunde. Einzelgespräche mit Kindern werden nur nach informierter Zustimmung des Kindes und des jeweiligen Erziehungsberechtigten durchgeführt. Die Interviews werden an einem von der teilnehmenden Person gewünschtem Ort durchgeführt, vorzugsweise in deren familiären Umfeld.

Ablauf der Studie
Die Gesamtdauer der Studie ist auf drei Jahre festgelegt. Die Planungs- und Vorbereitungsphase begann im September 2013. Der Beginn der Datensammlung ist auf den September 2014 festgelegt und die Datensammlung dauert ungefähr ein Jahr. Der Durchführungsort der Datensammlung ist die Deutschschweiz. Gleichzeitig mit der Datensammlung werden die Daten analysiert, sodass erste Ergebnisse im Frühjahr 2016 vorliegen. Eine Vorstellung der vorläufigen Resultate ist für das Frühjahr 2016 geplant, zu der alle Studienteilnehmenden eingeladen sind. Das planmäßige Studienende wurde auf September 2016 festgelegt.
Wir machen diese Studie so, wie es die Gesetze in der Schweiz vorschreiben. Ausserdem beachten wir alle international anerkannten Richtlinien. Die zuständige Kantonale Ethikkommission hat die Studie geprüft und bewilligt.

4. Ablauf für die Teilnehmenden

Du bist an einer Teilnahme an dieser Studie interessiert und hast bereits Kontakt mit der Studienleiterin aufgenommen. Diese hat Dich dann kontaktiert und Dir mit Deiner Zustimmung diese schriftliche Studieninformation und Einwilligungserklärung per Post zukommen lassen.
Die Studienleiterin wird Dich in den nächsten Wochen erneut kontaktieren und bei deinem bestehenden Interesse kannst Du mit ihr einen Termin für ein Gespräch vereinbaren.
Mit Deiner Einwilligung zur Teilnahme an dieser Studie erklärst Du Dich zu einem ungefähr stündigen Einzelgespräch bereit, dass eine wissenschaftliche Mitarbeiterin mit Dir führt und das auf Tonband aufgenommen wird. Mit der Durchführung dieses Interviews endet Deine Studienteilnahme.

Es kann sein, dass wir Dich von der Studie vorzeitig ausschliessen müssen. Das kann deshalb geschehen, weil sich Dein gesundheitlicher Zustand verschlechtert hat und wir Dich keiner zusätzlichen Belastung aussetzen möchten.

5. Rechte der Teilnehmenden

Du nimmst nur dann an dieser Studie teil, wenn Du es willst. Niemand darf Dich dazu in irgendeiner Weise drängen oder dazu überreden. Deine laufende medizinische Behandlung geht genau gleich weiter, wenn Du nicht mitmachst. Du musst nicht begründen, warum Du nicht mitmachen willst.
Wenn Du Dich entscheidest mitzumachen, kannst Du diesen Entscheid jederzeit zurücknehmen. Du musst ebenfalls nicht begründen, wenn Du aus der Studie aussteigen willst.
Du darfst jederzeit alle Fragen zur Studie stellen. Wende Dich dazu bitte an eine der Personen, die am Ende dieser Studieninformation genannt sind.

6. Pflichten der Teilnehmenden

Wenn Du bei der Studie mitmachst, musst Du bestimmte Regeln beachten. Dies ist notwendig für Deine Sicherheit und Gesundheit. Wir werden Dich dabei so gut wir können unterstützen. Als Studienteilnehmende/r bist Du verpflichtet:

- Dich für ein stündiges Gespräch zur Verfügung zu stellen und Angaben zu Deinen Erfahrungen und Deiner Person zu machen;
- Informationen über die Studie mittels Studienflyer an Personen weiterzuleiten, die ebenfalls wichtige Gesprächspartner für die Studie sein könnten;
- Den Anweisungen der Studienleiterin zu folgen und sie über Änderungen im Befinden zu informieren, die für die Studie relevant sind.
- Den Anweisungen der Studienleiterin zu folgen und sie über den Verlauf Deiner Erkrankung zu informieren und neue Symptome, neue Beschwerden und Änderungen im Befinden zu melden.

Wenn Du diese Pflichten nicht beachtest, kannst Du Haftungsansprüche verlieren.

7. Nutzen für die Teilnehmenden

Die Teilnahme an dieser Studie nützt Dir und Deiner Familie nicht direkt. Du trägst aber dazu bei, dass in Zukunft andere Familien mit Muskelerkrankungen unterstützt werden können.
Die Studie liefert wichtige Erkenntnisse über die Erfahrungen von jungen Menschen mit neuromuskulären Erkrankungen und deren Familien über den Übergang vom Kind zum Erwachsenen. Diese Erkenntnisse sind die Voraussetzung für eine mögliche Verbesserung der pflegerischen Versorgung der Betroffenen in dieser Lebensphase. Sie fliessen in die zukünftige Entwicklung von familienzentrierten pflegerischen Angeboten für junge Menschen mit neuromuskulären Erkrankungen und für deren Familien mit ein.

8. Risiken und Belastungen für die Teilnehmenden

Die Teilnahme an dieser Studie sollte für Dich kein Risiko darstellen. Du erklärst Dich zu einem Gespräch bereit, das ungefähr eine Stunde dauert. Dieses Gespräch kann jedoch verschiedene Emotionen auslösen. Insgesamt gehen wir aber davon aus, dass es sich positiv auswirkt, wenn Du Dich in einem persönlichen Gespräch über Deine Erfahrungen ausdrücken kannst. Im Rahmen von Fragen oder beim Auftreten von negativen Emotionen während oder nach dem Gespräch verweisen wir Dich an eine entsprechende Beratungsstelle weiter.

9. Ergebnisse aus der Studie

Die Studienleiterin wird Dich während der Studie über alle neuen Erkenntnisse informieren, die den Nutzen der Studie oder Deine Sicherheit und somit Deine Einwilligung zur Teilnahme an der Studie beeinflussen können. Du wirst diese Information mündlich und schriftlich erhalten.

10. Vertraulichkeit der Daten

Wir werden für diese Studie Daten zu Deinen Erfahrungen und zu Deiner Person erfassen. Diese Daten werden wir verschlüsseln, d.h. wir werden anstelle Deines vollen Namens nur eine dreistellige Zahl verwenden, um Dich zu kennzeichnen. Zudem werden in den Interviews Personen, Orts- und Institutionsnamen, sowie Datumsangaben und andere Sachverhalte, die eine Identifizierung der Person ermöglichen würden, verschlüsselt. Einzig die Studienleiterin weiss, wer oder was sich hinter dieser Verschlüsselung verbirgt. Andere Wissenschaftler, die an dieser Studie beteiligt sind, werden nur mit den verschlüsselten Daten arbeiten.
Alle Personen, die mit der Studie in irgendeiner Weise zu tun haben, verpflichten sich absolute Vertraulichkeit zu wahren. Wir werden Namen von Teilnehmenden nirgends, in keinem Bericht, keiner Publikation, nicht gedruckt und nicht im Internet, veröffentlichen.
Es kann sein, dass die Studie während des Ablaufs überprüft wird. Dies können die Behörden tun, die sie vorab kontrolliert und bewilligt haben. Auch diejenige Institution, die die Studie bezahlt, kann den Ablauf überprüfen lassen. Sie alle sorgen dafür, dass die Regeln eingehalten werden und Deine Sicherheit nicht gefährdet wird. Dazu muss die Leiterin der Studie eventuell Deine persönlichen Daten für solche Kontrollen offenlegen. Ebenso kann es sein, dass im Fall eines Schadens ein Vertreter der Versicherung Deine Daten ansehen muss. Das darf dann aber nur die Daten betreffen, die unbedingt gebraucht werden, um den Schadensfall zu erledigen.
Die erhobenen Daten müssen aus Gründen der Nachvollziehbarkeit zehn Jahre im abgeschlossenen Archiv der ZHAW aufbewahrt werden.

11. Weitere Verwendung von Daten

Du kannst jederzeit aus der Studie aussteigen, wenn Du dies wünschst. Du musst Deine Entscheidungen nicht begründen. Wenn Du später aussteigst, werden wir Daten, die wir bis dahin erhoben haben, zu Ende auswerten. Diese Daten stellen einen wichtigen Bestandteil der Ergebnisse dar und können nicht mehr ausgeschlossen werden, weil sonst die ganze Studie ihren Wert verlieren würde. Wir werden Deine Daten dann jedoch vollständig anonymisieren, d.h. wir werden endgültig Deinen Namen und die Verschlüsselungsnummer darauf löschen. Niemand wird danach mehr erfahren können, dass die Daten von Dir stammten. Prüfe bitte, ob Du damit einverstanden sein kannst, bevor Du bei der Studie mitmachst.

12. Entschädigung für Teilnehmende

Wenn Du bei dieser Studie mitmachst, bekommst Du dafür keine Entschädigung. Allfällige Reisespesen werden nicht übernommen.

13. Deckung von Schäden

Falls Du durch die Studie einen gesundheitlichen Schaden erleidest, haftet die Institution, die für die Durchführung der Studie verantwortlich ist. Diese Haftung gilt aber nur dann, wenn Du nachweisen kannst, dass der Schaden auf die Studie und damit verbundene Handlungen zurückzuführen ist.
Die Zürcher Hochschule für Angewandte Wissenschaften (Technikumstr. 71, CH-8401 Winterthur) hat eine Versicherung bei der Zürich Versicherungs-Gesellschaft AG (Mythenquai 2, 8002 Zürich)

abgeschlossen, um im Schadensfall für die Haftung aufkommen zu können. Wenn Du einen Schaden erlitten hast, so wende Dich bitte an die Leiterin oder Prüferin der Studie.

14. Finanzierung der Studie

Die Studie wird mehrheitlich von der Studienleiterin, Frau Waldboth, finanziert, die im Rahmen ihrer Doktorarbeit von der Zürcher Hochschule für Angewandte Wissenschaften teilfinanziert ist. Ein Ansuchen für eine weitere Finanzierung durch einen zusätzlichen Sponsor ist zukünftig angestrebt.

15. Kontaktpersonen

Bei Fragen oder Unklarheiten, die während der Studie oder danach auftreten, kannst Du dich jederzeit an eine dieser Kontaktpersonen wenden:

Studienleiterin
Veronika Waldboth
Wissenschaftliche Mitarbeiterin
Zürcher Hochschule für Angewandte Wissenschaften
Institut für Pflege
Technikumstr. 71
CH- 8401 Winterthur

Tel. +41 58 934 6499
Email: veronika.waldboth@zhaw.ch

Prüferin der Studie
Prof Dr. Romy Mahrer-Imhof
Leiterin Master of Science in Pflege
Zürcher Hochschule für Angewandte Wissenschaften
Institut für Pflege
Technikumstr. 71
CH-8401 Winterthur

Tel. +41 58 934 63 44
Email: romy.mahrer@zhaw.ch

Einwilligungserklärung für Teilnehmende von 14-17 Jahren

Erwachsen werden mit einer neuromuskulären Erkrankung

Studie: „Erwachsen werden: Erfahrungen von Menschen mit neuromuskulären Erkrankungen und deren Familien“

- Bitte liess dieses Formular sorgfältig durch.
- Bitte frag, wenn Du etwas nicht verstehst oder wissen möchtest.

Titel der Studie: Erwachsen werden mit einer neuromuskulären Erkrankung Erwachsen werden: Erfahrungen von Menschen mit neuromuskulären Erkrankungen und deren Familien	
Verantwortliche Institution (Sponsor)	Zürcher Hochschule für Angewandte Wissenschaften Departement Gesundheit Institut für Pflege Technikumstrasse 71 8401 Winterthur
Ort der Durchführung:	Schweiz
Leiterin der Studie	Veronika Waldboth
Teilnehmerin / Teilnehmer	Name und Vorname (bitte in Druckbuchstaben): ***X*** _____ Geburtsdatum (Tag /Monat/ Jahr): ***X*** _____ Geschlecht (bitte ankreuzen): ☐ weiblich ☐ männlich

- Ich wurde von der unterzeichnenden Studienleiterin mündlich und schriftlich über den Zweck, den Ablauf der Studie, über mögliche Vor- und Nachteile sowie über eventuelle Risiken informiert.

- Meine Fragen im Zusammenhang mit der Teilnahme an dieser Studie sind mir zufriedenstellend beantwortet worden. Ich kann die schriftliche Studieninformation vom 25.03.2015, Version 3 behalten und erhalte eine Kopie unserer schriftlichen Einwilligungserklärung. Ich akzeptiere den Inhalt der zur oben genannten Studie abgegebenen schriftlichen Studieninformation.

- Ich nehme an dieser Studie freiwillig teil. Ich kann jederzeit und ohne Angabe von Gründen meine Zustimmung zur Teilnahme widerrufen, ohne dass ich deswegen Nachteile bei der weiteren medizinischen Betreuung erleide.

- Ich hatten genügend Zeit, meine Entscheidung zu treffen.

- Ich bin darüber informiert, dass eine Versicherung Schäden deckt, falls ich nachweisen kann, dass die Schäden auf die Studie zurückzuführen sind.

- Ich weiss, dass meine persönlichen Daten nur in verschlüsselter Form zu Forschungszwecken weitergegeben werden können. Ich bin einverstanden, dass die zuständigen Sponsoren der Studie, der Behörden und der Kantonalen Ethikkommission zu Prüf- und Kontrollzwecken in die Originaldaten Einsicht nehmen dürfen, jedoch unter strikter Einhaltung der Vertraulichkeit.

- Ich bin mir bewusst, dass die in der Teilnehmerinformation genannten Pflichten während der Studie einzuhalten sind. Im Interesse meiner Gesundheit kann die Leiterin der Studie mich jederzeit von der Studie ausschliessen.

Ort, Datum ***X*** _____	Schriftliches Einverständnis: Unterschrift Studienteilnehmerin/Studienteilnehmer ***X*** _____
	☐ Mündliches Einverständnis: Herr/Frau ***X*** _____ hat sein/ihr Einverständnis aufgrund körperlicher Gründe mündlich gegeben. Siehe Audiofile Nr. ***X*** _____

Bestätigung der Studienleiterin: Hiermit bestätige ich, dass ich dieser Teilnehmerin / diesem Teilnehmer Wesen, Bedeutung und Tragweite der Studie erläutert habe. Ich versichere, alle im Zusammenhang mit dieser Studie stehenden Verpflichtungen gemäss dem geltenden Recht zu erfüllen. Sollte ich zu irgendeinem Zeitpunkt während der Durchführung der Studie von Aspekten erfahren, welche die Bereitschaft der Teilnehmerin / des Teilnehmers zur Teilnahme an der Studie beeinflussen könnte, werde ich sie/ihn umgehend darüber informieren.

Ort, Datum	Unterschrift der Studienleiterin

Zürcher Hochschule für Angewandte Wissenschaften
Institut für Pflege

Florence Nightingale School of Nursing and Midwifery
King's College London

Studieninformation für Teilnehmende über 18 Jahren

Erwachsen werden mit einer neuromuskulären Erkrankung

Studie: „Erwachsen werden: Erfahrungen von Menschen mit neuromuskulären Erkrankungen und deren Familien"

Sponsoren:
Institut für Pflege an der Zürcher Hochschule für Angewandte Wissenschaften (ZHAW) in Winterthur (CH) und Florence Nightingale School of Nursing and Midwifery am King's College in London (UK)

Inhaltsverzeichnis

Sehr geehrte Damen und Herren

Wir sind vom Institut für Pflege der Zürcher Hochschule für Angewandte Wissenschaften in Winterthur. Diese pflegewissenschaftliche Studie ist die Doktorarbeit in Pflege von Frau Veronika Waldboth und wird in Zusammenarbeit mit dem Institut für Pflege der Zürcher Hochschule für Angewandte Wissenschaften (ZHAW) in Winterthur und der Florence Nightingale School of Nursing and Midwifery am King's College in London durchgeführt. Frau Waldboth arbeitet als wissenschaftliche Mitarbeiterin an der ZHAW und wird das Projekt in der Schweiz umsetzen.

1. Auswahl der Personen, die an der Studie teilnehmen können

Es können alle in der Schweiz ansässigen Personen an dieser Studie teilnehmen, die an einer neuromuskulären Erkrankung leiden oder Angehörige (Eltern, Geschwister und andere nahestehende Angehörige) einer Person sind, die an einer neuromuskulären Erkrankung leidet. Die von der neuromuskulären Erkrankung betroffene Person muss zwischen 14 bis 30 Jahre alt sein und die ersten Symptome der Erkrankung müssen bereits im Kindesalter aufgetreten sein (zwischen 0 bis zehn Jahren). Derzeit soll eine mässige bis starke körperliche Einschränkung vorliegen.
Nicht teilnehmen dürfen Personen, die kein deutsch sprechen und die an einer ausgeprägten geistigen Einschränkung leiden, die es der Person nicht ermöglicht zur Teilnahme an der Studie einzuwilligen.

2. Ziel der Studie

Ziel dieser Studie ist es, die Erfahrungen von Menschen mit genetisch bedingten neuromuskulären Erkrankungen zu erfassen, die spezifisch für den Übergang vom Kindes- zum Erwachsenenalter sind. Feinziele dieser Studie sind es, die Herausforderungen zu verstehen, mit denen betroffene Familien während dieser Entwicklungsphase konfrontiert sind; Strategien zu identifizieren, mit denen sich betroffene Familien erfolgreich an veränderte Situationen anpassen; Faktoren zu identifizieren, die die Anpassung an veränderte Situationen beeinflussen und Erfahrungen der betroffenen Familien mit dem Schweizerischen Gesundheitssystem zu verstehen.
Diese Erkenntnisse sind die Voraussetzung für eine mögliche Verbesserung der pflegerischen Versorgung der Betroffenen sowohl in Spitälern und Institutionen als auch in deren häuslichem Umfeld. Sie fliessen in die zukünftige Entwicklung von familienzentrierten pflegerischen Angeboten für junge Menschen mit neuromuskulären Erkrankungen und für deren Familien mit ein.

3. Allgemeine Informationen zur Studie

Hintergrund

Diese pflegewissenschaftliche Studie untersucht Erfahrungen von Jugendlichen und jungen Erwachsenen, die an einer neuromuskulären Erkrankung leiden. Das „Erwachsenwerden" ist eine Zeit des Übergangs vom Kindes- zum Erwachsenenalter und gleichzeitig eine Zeit der Entwicklung und Entfaltung der eigenen Persönlichkeit. Junge Menschen, die an einer chronischen Erkrankung leiden, die sich bereits im Kindesalter manifestiert, erleben diese Zeit anders als ihre gesunden Mitmenschen. Für Personen, die an Muskeldystrophie leiden bedeutet es, dass in einer Zeit, in der von ihnen erwartet wird, dass sie zunehmend unabhängiger werden, ihre körperliche Einschränkung und Abhängigkeit von Unterstützung zunimmt. Genetische Erkrankungen, die sich im Kindesalter manifestieren, haben zudem grosse Auswirkungen auf die Familie oder auf

nahestehende Personen. Aus diesem Grund werden Erfahrungen von Familienmitgliedern und nahestehenden Personen in die Untersuchung mit eingeschlossen.

Methodisches Vorgehen
Diese pflegewissenschaftliche Studie hat einen qualitativen Ansatz. Das bedeutet, dass Daten in erster Linie durch Einzelgespräche mit den betroffenen jungen Personen und deren Angehörigen erhoben werden. Die Anzahl der Teilnehmende wird auf ungefähr 40 Personen (8 bis 15 Familien) geschätzt. Teilnehmenden machen während des Gesprächs Angaben zu ihren Erfahrungen und zu ihrer Person. Das Gespräch dauert ungefähr eine Stunde. Einzelgespräche mit Kindern werden nur nach informierter Zustimmung des Kindes und des jeweiligen Erziehungsberechtigten durchgeführt. Die Interviews werden an einem von der teilnehmenden Person gewünschtem Ort durchgeführt, vorzugsweise in deren familiären Umfeld.

Ablauf der Studie
Die Gesamtdauer der Studie ist auf drei Jahre festgelegt. Die Planungs- und Vorbereitungsphase begann im September 2013. Der Beginn der Datensammlung ist auf den September 2014 festgelegt und die Datensammlung dauert ungefähr ein Jahr. Der Durchführungsort der Datensammlung ist die Deutschschweiz. Gleichzeitig mit der Datensammlung werden die Daten analysiert, sodass erste Ergebnisse im Frühjahr 2016 vorliegen. Eine Vorstellung der vorläufigen Resultate ist für das Frühjahr 2016 geplant, zu der alle Studienteilnehmenden eingeladen sind. Das planmäßige Studienende wurde auf September 2016 festgelegt.
Wir machen diese Studie so, wie es die Gesetze in der Schweiz vorschreiben. Ausserdem beachten wir alle international anerkannten Richtlinien. Die zuständige Kantonale Ethikkommission hat die Studie geprüft und bewilligt.

4. Ablauf für die Teilnehmenden

Sie sind an einer Teilnahme an dieser Studie interessiert und haben bereits Kontakt mit der Studienleiterin aufgenommen. Diese hat Sie dann kontaktiert und Ihnen mit Ihrer Zustimmung diese schriftliche Studieninformation und Einwilligungserklärung per Post zukommen lassen. Sie werden in den nächsten Wochen erneut kontaktiert und bei bestehendem Interesse können Sie mit der Studienleiterin einen Termin für ein Gespräch vereinbaren.

Mit Ihrer Einwilligung zur Teilnahme an dieser Studie erklären Sie Sich zu einem ungefähr stündigen Einzelgespräch bereit, dass eine wissenschaftliche Mitarbeiterin mit Ihnen führt und das auf Tonband aufgenommen wird. Mit der Durchführung dieses Interviews endet Ihre Studienteilnahme.

Es kann sein, dass wir Sie von der Studie vorzeitig ausschliessen müssen. Das kann deshalb geschehen, weil sich Ihr gesundheitlicher Zustand verschlechtert hat und wir Sie keiner zusätzlichen Belastung aussetzen möchten.

5. Rechte der Teilnehmenden

Sie nehmen nur dann an dieser Studie teil, wenn Sie es wollen. Niemand darf Sie dazu in irgendeiner Weise drängen oder dazu überreden. Ihre laufende medizinische Behandlung geht genau gleich weiter, wenn Sie nicht mitmachen. Sie müssen nicht begründen, warum Sie nicht mitmachen wollen. Wenn Sie Sich entscheiden mitzumachen, können Sie diesen Entscheid

jederzeit zurücknehmen. Sie müssen ebenfalls nicht begründen, wenn Sie aus der Studie aussteigen wollen.
Sie dürfen jederzeit alle Fragen zur Studie stellen. Wenden Sie Sich dazu bitte an eine der Personen, die am Ende dieser Studieninformation genannt sind.

6. Pflichten der Teilnehmenden

Wenn Sie bei der Studie mitmachen, müssen Sie bestimmte Regeln beachten. Dies ist notwendig für Ihre Sicherheit und Gesundheit. Wir werden Sie dabei so gut wir können unterstützen. Als Studienteilnehmende/r sind Sie verpflichtet:

- Sich für ein stündiges Gespräch zur Verfügung zu stellen und Angaben zu Ihren Erfahrungen und Ihrer Person zu machen;
- Informationen über die Studie mittels Studienflyer an Personen weiterzuleiten, die ebenfalls wichtige Gesprächspartner für die Studie sein könnten;
- Den Anweisungen der Studienleiterin zu folgen und sie über Änderungen im Befinden zu informieren, die für die Studie relevant sind.
- Den Anweisungen Ihrer Studienleiterin zu folgen und sie über den Verlauf der Erkrankung zu informieren und neue Symptome, neue Beschwerden und Änderungen im Befinden zu melden.

Wenn Sie diese Pflichten nicht beachten, können Sie Haftungsansprüche verlieren.

7. Nutzen für die Teilnehmenden

Die Teilnahme an dieser Studie nützt Ihnen und Ihrer Familie nicht direkt. Sie tragen aber dazu bei, dass in Zukunft andere Familien mit Muskelerkrankungen unterstützt werden können.
Die Studie liefert wichtige Erkenntnisse über die Erfahrungen von jungen Menschen mit neuromuskulären Erkrankungen und deren Familien über den Übergang vom Kind zum Erwachsenen. Diese Erkenntnisse sind die Voraussetzung für eine mögliche Verbesserung der pflegerischen Versorgung der Betroffenen in dieser Lebensphase. Sie fliessen in die zukünftige Entwicklung von familienzentrierten pflegerischen Angeboten für junge Menschen mit neuromuskulären Erkrankungen und für deren Familien mit ein.

8. Risiken und Belastungen für die Teilnehmenden

Die Teilnahme an dieser Studie sollte für Sie kein Risiko darstellen. Sie erklären sich zu einem Gespräch bereit, das ungefähr eine Stunde dauert. Dieses Gespräch kann jedoch verschiedene Emotionen auslösen. Insgesamt gehen wir aber davon aus, dass es sich positiv auswirkt, wenn Sie Sich in einem persönlichen Gespräch über Ihre Erfahrungen ausdrücken können. Im Rahmen von Fragen oder beim Auftreten von negativen Emotionen während oder nach dem Gespräch verweisen wir Sie an eine entsprechende Beratungsstelle weiter.

9. Ergebnisse aus der Studie

Die Studienleiterin wird Sie während der Studie über alle neuen Erkenntnisse informieren, die den Nutzen der Studie oder Ihre Sicherheit und somit Ihre Einwilligung zur Teilnahme an der Studie beeinflussen können. Sie werden diese Information mündlich und schriftlich erhalten.

10. Vertraulichkeit der Daten

Wir werden für diese Studie Daten zu Ihren Erfahrungen und zu Ihrer Person erfassen. Diese Daten werden wir verschlüsseln, d.h. wir werden anstelle Ihres vollen Namens nur eine dreistellige Zahl verwenden, um Sie zu kennzeichnen. Zudem werden in den Interviews Personen-, Orts- und Institutionsnamen, sowie Datumsangaben und andere Sachverhalte, die eine Identifizierung der Person ermöglichen würden, verschlüsselt. Einzig die Studienleiterin weiss, wer oder was sich hinter dieser Verschlüsselung verbirgt. Andere Wissenschaftler, die an dieser Studie beteiligt sind, werden nur mit den verschlüsselten Daten arbeiten.
Alle Personen, die mit der Studie in irgendeiner Weise zu tun haben, verpflichten sich absolute Vertraulichkeit zu wahren. Wir werden Namen von Teilnehmenden nirgends, in keinem Bericht, keiner Publikation, nicht gedruckt und nicht im Internet, veröffentlichen.
Es kann sein, dass die Studie während des Ablaufs überprüft wird. Dies können die Behörden tun, die sie vorab kontrolliert und bewilligt haben. Auch diejenige Institution, die die Studie bezahlt, kann den Ablauf überprüfen lassen. Sie alle sorgen dafür, dass die Regeln eingehalten werden und Ihre Sicherheit nicht gefährdet wird. Dazu muss die Leiterin der Studie eventuell Ihre persönlichen Daten für solche Kontrollen offenlegen. Ebenso kann es sein, dass im Fall eines Schadens ein Vertreter der Versicherung Ihre Daten ansehen muss. Das darf dann aber nur die Daten betreffen, die unbedingt gebraucht werden, um den Schadensfall zu erledigen.
Die erhobenen Daten müssen aus Gründen der Nachvollziehbarkeit zehn Jahre im abgeschlossenen Archiv der ZHAW aufbewahrt werden.

11. Weitere Verwendung von Daten

Sie können jederzeit aus der Studie aussteigen, wenn Sie dies wünschen. Sie müssen Ihre Entscheidungen nicht begründen. Wenn Sie später aussteigen, werden wir Daten, die wir bis dahin erhoben haben, zu Ende auswerten. Diese Daten stellen einen wichtigen Bestandteil der Ergebnisse dar und können nicht mehr ausgeschlossen werden, weil sonst die ganze Studie ihren Wert verlieren würde. Wir werden Ihre Daten dann jedoch vollständig anonymisieren, d.h. wir werden endgültig Ihren Namen und die Verschlüsselungsnummer darauf löschen. Niemand wird danach mehr erfahren können, dass die Daten von Ihnen stammten. Prüfen Sie bitte, ob Sie damit einverstanden sein können, bevor Sie bei der Studie mitmachen.

12. Entschädigung für Teilnehmende

Wenn Sie bei dieser Studie mitmachen, bekommen Sie dafür keine Entschädigung. Allfällige Reisespesen werden nicht übernommen.

13. Deckung von Schäden

Falls Sie durch die Studie einen gesundheitlichen Schaden erleiden, haftet die Institution, die für die Durchführung der Studie verantwortlich ist. Diese Haftung gilt aber nur dann, wenn Sie nachweisen können, dass der Schaden auf die Studie und damit verbundene Handlungen zurückzuführen ist. Die Zürcher Hochschule für Angewandte Wissenschaften (Technikumstr. 71, CH-8401 Winterthur) hat eine Versicherung bei der Zürich Versicherungs-Gesellschaft AG (Mythenquai 2, 8002 Zürich) abgeschlossen, um im Schadensfall für die Haftung aufkommen zu können. Wenn Sie einen Schaden erlitten haben, so wenden Sie Sich bitte an die Leiterin oder Prüferin der Studie.

14. Finanzierung der Studie

Die Studie wird mehrheitlich von der Studienleiterin, Frau Waldboth, finanziert, die im Rahmen ihrer Doktorarbeit von der Zürcher Hochschule für Angewandte Wissenschaften teilfinanziert ist. Ein Ansuchen für eine weitere Finanzierung durch einen zusätzlichen Sponsor ist zukünftig angestrebt.

15. Kontaktpersonen

Bei Fragen oder Unklarheiten, die während der Studie oder danach auftreten, können Sie Sich jederzeit an eine dieser Kontaktpersonen wenden:

Studienleiterin
Veronika Waldboth
Wissenschaftliche Mitarbeiterin
Zürcher Hochschule für Angewandte Wissenschaften
Institut für Pflege
Technikumstr. 71
CH- 8401 Winterthur

Tel. +41 58 934 6499
Email: veronika.waldboth@zhaw.ch

Prüferin der Studie
Prof Dr. Romy Mahrer-Imhof
Leiterin Master of Science in Pflege
Zürcher Hochschule für Angewandte Wissenschaften
Institut für Pflege
Technikumstr. 71
CH-8401 Winterthur

Tel. +41 58 934 6344
Email: romy.mahrer@zhaw.ch

Einwilligungserklärung für Teilnehmende über 18 Jahren

Erwachsen werden mit einer neuromuskulären Erkrankung

Studie: „Erwachsen werden: Erfahrungen von Menschen mit neuromuskulären Erkrankungen und deren Familien“

- Bitte lesen Sie dieses Formular sorgfältig durch.
- Bitte fragen Sie, wenn Sie etwas nicht verstehen oder wissen möchten.

Titel der Studie: Erwachsen werden mit einer neuromuskulären Erkrankung Erwachsen werden: Erfahrungen von Menschen mit neuromuskulären Erkrankungen und deren Familien	
Verantwortliche Institution (Sponsor)	Zürcher Hochschule für Angewandte Wissenschaften Departement Gesundheit Institut für Pflege Technikumstrasse 71 8401 Winterthur
Ort der Durchführung:	Schweiz
Leiterin der Studie	Veronika Waldboth
Teilnehmerin / Teilnehmer	Name und Vorname (bitte in Druckbuchstaben): ***X*** _____ Geburtsdatum (Tag /Monat/ Jahr): ***X*** _____ Geschlecht (bitte ankreuzen): ☐ weiblich ☐ männlich

- Ich wurde von der unterzeichnenden Studienleiterin mündlich und schriftlich über den Zweck, den Ablauf der Studie, über mögliche Vor- und Nachteile sowie über eventuelle Risiken informiert.

- Meine Fragen im Zusammenhang mit der Teilnahme an dieser Studie sind mir zufriedenstellend beantwortet worden. Ich kann die schriftliche Studieninformation vom 25.03.2015, Version 3 behalten und erhalte eine Kopie meiner schriftlichen Einwilligungserklärung. Ich akzeptiere den Inhalt der zur oben genannten Studie abgegebenen schriftlichen Studieninformation.

- Ich nehme an dieser Studie freiwillig teil. Ich kann jederzeit und ohne Angabe von Gründen meine Zustimmung zur Teilnahme widerrufen, ohne dass ich deswegen Nachteile bei der weiteren medizinischen Betreuung erleide.

- Ich hatte genügend Zeit, meine Entscheidung zu treffen.

- Ich bin darüber informiert, dass eine Versicherung Schäden deckt, falls ich nachweisen kann, dass die Schäden auf die Studie zurückzuführen sind.

- Ich weiss, dass meine persönlichen Daten nur in verschlüsselter Form zu Forschungszwecken weitergegeben werden können. Ich bin einverstanden, dass die zuständigen Sponsoren der Studie, der Behörden und der Kantonalen Ethikkommission zu Prüf- und Kontrollzwecken in meine Originaldaten Einsicht nehmen dürfen, jedoch unter strikter Einhaltung der Vertraulichkeit.

- Ich bin mir bewusst, dass die in der Teilnehmerinformation genannten Pflichten während der Studie einzuhalten sind. Im Interesse meiner Gesundheit kann die Leiterin der Studie mich jederzeit von der Studie ausschliessen.

Ort, Datum ***X***_____	Schriftliches Einverständnis: Unterschrift Studienteilnehmerin/Studienteilnehmer ***X***_____
	☐ Mündliches Einverständnis: Herr/Frau ***X***_____hat sein/ihr Einverständnis aufgrund körperlicher Gründe mündlich gegeben. Siehe Audiofile Nr. ***X***_____

Bestätigung der Studienleiterin: Hiermit bestätige ich, dass ich dieser Teilnehmerin/diesem Teilnehmer Wesen, Bedeutung und Tragweite der Studie erläutert habe. Ich versichere, alle im Zusammenhang mit dieser Studie stehenden Verpflichtungen gemäss dem geltenden Recht zu erfüllen. Sollte ich zu irgendeinem Zeitpunkt während der Durchführung der Studie von Aspekten erfahren, welche die Bereitschaft der Teilnehmerin/des Teilnehmers zur Teilnahme an der Studie beeinflussen könnte, werde ich sie/ihn umgehend darüber informieren.

Ort, Datum	Unterschrift der Studienleiterin

Zürcher Hochschule für Angewandte Wissenschaften
Institut für Pflege

Florence Nightingale School of Nursing and Midwifery
King's College London

Erwachsen werden mit einer neuromuskulären Erkrankung
Interviewleitfaden

1. Begrüssung der teilnehmenden Person

   „Guten Tag (Anrede, Name teilnehmende Person). Herzlichen Dank, dass Sie Sich bereit erklärt haben / du dich bereits erklärt hast an dieser Studie teilzunehmen."

2. Information über:

   - Inhalt und Ablauf des Interviews
   - Möglichkeit der Unterbrechung des Interviews falls gewünscht
   - Freiwilligkeit der Teilnahme
   - Tonbandaufnahme des Interviews
   - Anonymität der Transkription
   - Vertraulichkeit der Daten

4. Nachfragen, ob Fragen offen sind

5. Kontrolle Einwilligung

6. Start der Tonbandaufnahme

7. Start des Interviews:

I. Für Kinder < 12 Jahren

Aufwärmen mit Kartenspiel

Zeichnung von Familie erstellen und erklären lassen

II. Für Jugendliche ab 12 Jahren und Erwachsene

Einstiegsfrage: „Wie sieht ein normaler Tag in Ihrem / Deinem Leben aus?"

Vertiefung zu Themenbereich 1: Erleben des Alltags
- Familie, Sozialleben und Freunde
- Wohn- und Arbeitssituation
- Gesundheit
- Herausforderungen im Alltag
- Umgang mit Herausforderungen

Vertiefung zu Themenbereich 2: Erwachsen werden
- Unterschied zwischen Leben als Kind / Jugendlicher zum Leben als Erwachsener
- Veränderungen
- Herausforderungen
- Umgang mit Herausforderungen
- Erwartungen und Ziele

Abschluss: „Möchten Sie / möchtest Du noch etwas ergänzen?"

7. Dank und Abschied

Florence Nightingale School of Nursing and Midwifery
King's College London

Zurich University of Applied Sciences
Institut of Nursing

Becoming an adult affected by neuromuscular disease
Topic guide

1. Welcome the participant

   Good day (title, name of participant). Thank you that you are willing to participate in this study.

2. Give information about:
   - Content and structure of the interview
   - Possibility of a break during the interview (if needed)
   - Voluntary participation
   - Recording of the interview
   - Anonymity of the transcription
   - Confidentiality of the data

3. Ask if the participant has questions

4. Check informed consent

5. Start the recorder

6. Start the interview

   I. <u>For children < 12 years</u>
   - Warm up activity with card game
   - Child draws the family and talks about the drawing

   II. <u>For adolescents >12 years and adults</u>

   Opening question: How does a normal day look like in your life?

   Detailed questions for theme 1: everyday life
   - Family, social life, friends
   - Living and working situation
   - Health
   - Challenges in everyday life
   - Coping with challenges

   Detailed questions for theme 2: Becoming an adult
   - Differences between life as a adolescent and life as an adult
   - Change
   - Challenges
   - Coping with challenges
   - Expectations and objectives

   Conclusion: "Would you like to add any additional information that has not been discussed before?"

7. Thank you and good bye

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Florence Nightingale School of Nursing and Midwifery
King's College London

Erwachsen werden mit einer neuromuskulären Erkrankung
Interviewleitfaden

1. Begrüssung der teilnehmenden Person

   „Guten Tag (Anrede, Name teilnehmende Person). Herzlichen Dank, dass Sie Sich bereit erklärt haben / du dich bereits erklärt hast an dieser Studie teilzunehmen."

2. Information über:

   - Inhalt und Ablauf des Interviews
   - Möglichkeit der Unterbrechung des Interviews falls gewünscht
   - Freiwilligkeit der Teilnahme
   - Tonbandaufnahme des Interviews
   - Anonymität der Transkription
   - Vertraulichkeit der Daten

4. Nachfragen, ob Fragen offen sind

5. Kontrolle Einwilligung

6. Start der Tonbandaufnahme

7. Start des Interviews:

I. Für Kinder < 12 Jahren

Aufwärmen mit Kartenspiel

Zeichnung von Familie erstellen und erklären lassen

II. Für Jugendliche ab 12 Jahren und Erwachsene

Einstiegsfrage: „Wie sieht ein normaler Tag in Ihrem / Deinem Leben aus?"

Vertiefung zu Themenbereich 1: Erleben des Alltags
- Herausforderungen im Alltag
- Umgang mit Herausforderungen
- Familie (Perspektive der Eltern, Geschwister, Unabhängigkeit)
- Sozialleben (Freundschaften, romantische Beziehungen)
- Wohnsituation (Selbstständigkeit)
- Schule oder Arbeitssituation
- Gesundheitssituation (Pflege, Spital, Hausarzt)

Vertiefung zu Themenbereich 2: Erwachsenwerden
- Veränderungen in der Phase des Erwachsenwerdens
- Herausforderungen (körperlich, emotional, sozial, kognitiv)
- Umgang mit Herausforderungen
- Herausforderungen für Familie

Abschluss:

- Erwartungen und Ziele („wenn Sie/ Du die Wahl haben/ hast, wie wäre es?")
- „Was ist für Sie/ Dich wichtig im Leben?"
- „Möchten Sie / möchtest Du noch etwas ergänzen?"

Sensibles Thema: Möchten Sie/ möchtest Du mehr darüber erzählen?

7. Dank und Abschied

Zürcher Hochschule für Angewandte Wissenschaften
Institut für Pflege

Florence Nightingale School of Nursing and Midwifery
King's College London

Kartenspiel zum Aufwärmen für Kinder (<12 Jahren)

Studie: „Erwachsenwerden mit einer neuromuskulären Erkrankung"

Welches ist Dein Lieblingsspiel?

Was machst Du gerne in Deiner Freizeit?

Hast Du ein Lieblingstier?

Was ist Dein Traumberuf?

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Institut für Pflege

Florence Nightingale School of Nursing and Midwifery
King's College London

Erwachsen werden mit einer neuromuskulären Erkrankung
Demografische Angaben

A. Junger Mensch mit einer neuromuskulären Erkrankung

	ID	
	Datum der Erhebung (TT/MM/JJ)	
1	Geschlecht	□1 Weiblich □2 Männlich
2	Alter in Jahren	_____ Jahre
3a	Typ der neuromuskulären Erkrankung	□1 Muskeldystrophie Typ Duchenne □2 Spinale Muskuläre Atrophie □3 Unbekannter Typ □4 Andere:
3b		_____
4	Zivilstatus	□1 Ledig □2 In Partnerschaft □3 Verheiratet / eingetragene Partnerschaft □4 Geschieden □5 Verwitwet
5a	Wohnsituation	□1 Lebe mit Familie □2 Leben in Wohngemeinschaft / mit Freunden □3 Lebe in Institution / betreutes Wohnen □4 Lebe alleine □5 Andere:
5b		_____
6	Wie viele Personen leben in Ihrem Haushalt?	_____ Personen
7a	Beschäftigung / Beruf	□1 Schüler/in □2 Student/in □3 Rentner/in □4 Arbeitslos □5 Dauerhaft erwerbsunfähig □6 Hausfrau/Hausmann □7 Erwerbstätig □8 Anderes:
7b		_____

7c	Falls erwerbstätig: Angabe des Berufs	_____
7d	Stellenprozente	□1 Vollzeit (80-100%) □2 Teilzeit (>80%)
8	Schulabschluss	□1 keine abgeschlossene Grundschule □2 Grundschule □3 Lehre □4 Gymnasium □5 Hochschulabschluss
9	Wie bewerten Sie Ihren derzeitigen körperlichen Gesundheitszustand?*	□1 Schlecht □2 Angemessen □3 Gut □4 Sehr gut □5 Ausgezeichnet
10	Wie bewerten Sie Ihren derzeitigen geistigen und emotionalen Gesundheitszustand?*	□1 Schlecht □2 Angemessen □3 Gut □4 Sehr gut □5 Ausgezeichnet

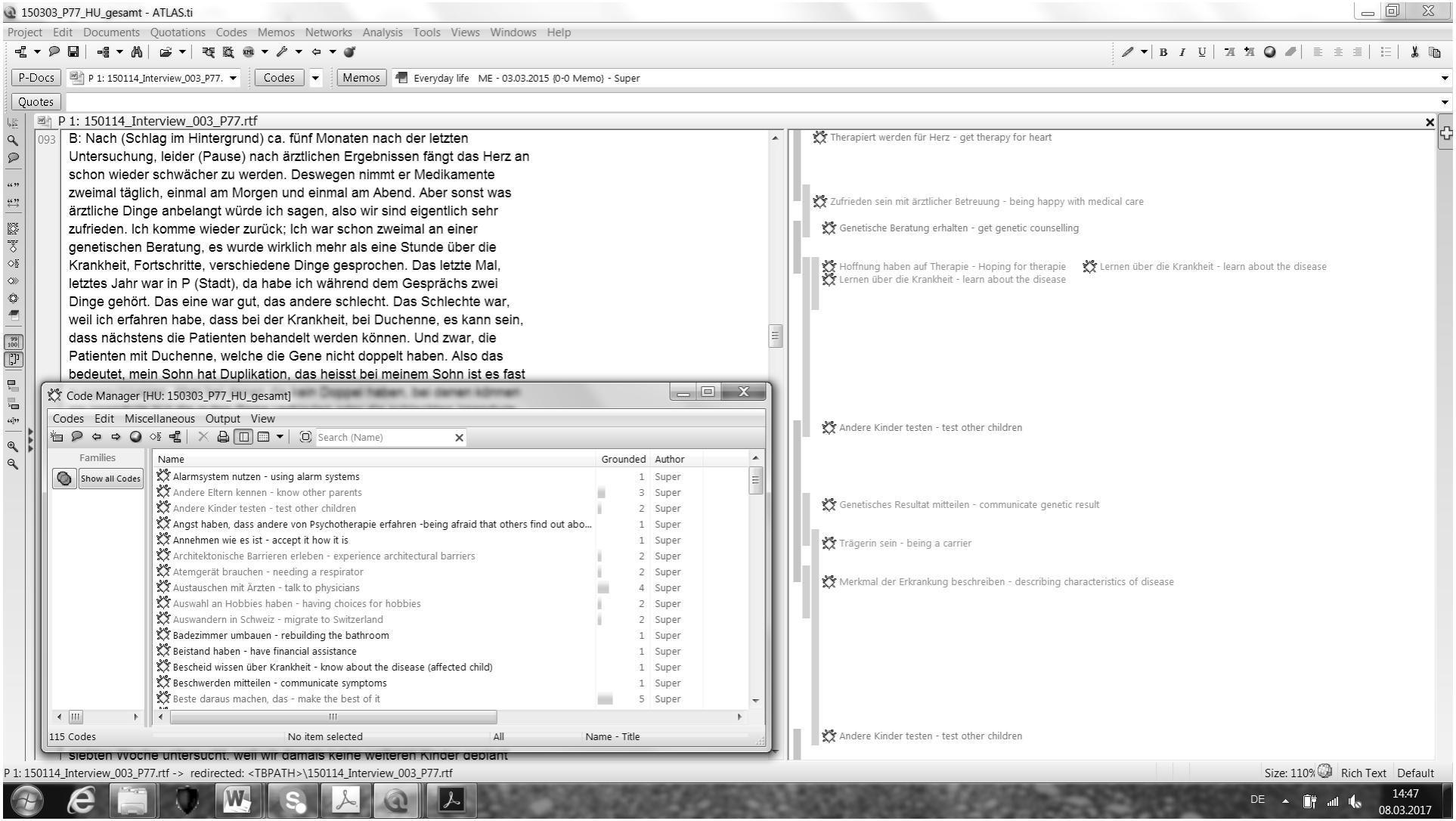
B. Eltern, Geschwister und nahestehende Angehörige

	ID	
	Datum der Erhebung (TT/MM/JJ)	
1	Geschlecht	□1 Weiblich □2 Männlich
2	Alter in Jahren	_____ Jahre
3a	Beziehung zur Person mit der neuromuskulären Erkrankung	□1 Mutter □2 Vater □3 Schwester □4 Bruder □5 Andere:
3b		_____
4a	Typ der neuromuskulären Erkrankung	□1 Muskeldystrophie Typ Duchenne □2 Spinale Muskuläre Atrophie □3 Unbekannter Typ □4 Andere:
4b		_____
5	Zivilstatus	□1 Ledig □2 In Partnerschaft □3 Verheiratet / eingetragene Partnerschaft □4 Geschieden □5 Verwitwet
6a	Wohnsituation	□1 Lebe mit Familie □2 Leben in Wohngemeinschaft / mit Freunden □3 Lebe in Institution / betreutes Wohnen □4 Lebe alleine □5 Andere:
6b		_____
7	Wie viele Personen leben in Ihrem Haushalt?	_____ Personen
8a	Beschäftigung / Beruf	□1 Schüler/in □2 Student/in □3 Rentner/in □4 Arbeitslos □5 Dauerhaft erwerbsunfähig □6 Hausfrau/Hausmann

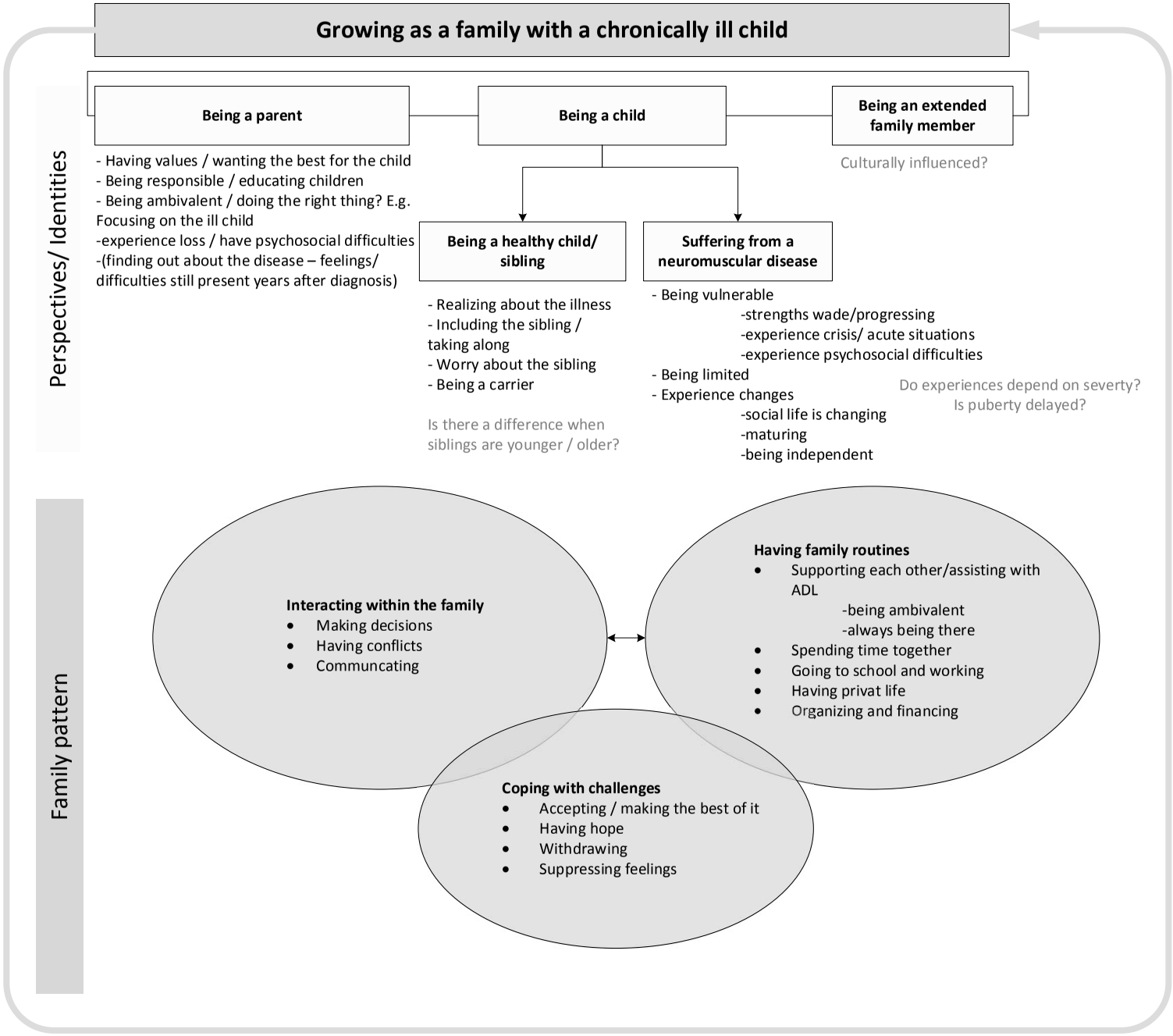
		□7 Erwerbstätig □8 Anderes:
8b		
8c	Falls erwerbstätig: Angabe des Berufs	
8d	Stellenprozente	□1 Vollzeit (80-100%) □2 Teilzeit (>80%)
9	Schulabschluss	□1 keine abgeschlossene Grundschule □2 Grundschule □3 Lehre □4 Gymnasium □5 Hochschulabschluss
10	Wie bewerten Sie Ihren derzeitigen körperlichen Gesundheitszustand?*	□1 Schlecht □2 Angemessen □3 Gut □4 Sehr gut □5 Ausgezeichnet
11	Wie bewerten Sie Ihren derzeitigen geistigen und emotionalen Gesundheitszustand?*	□1 Schlecht □2 Angemessen □3 Gut □4 Sehr gut □5 Ausgezeichnet

* Adapted from: The Data Resource Center for Child and Adolescent Health. (2012). National Survey of Children's Health. Retrieved 27.02.14, from http://www.nschdata.org/Content/LearnAboutTheSurvey.aspx

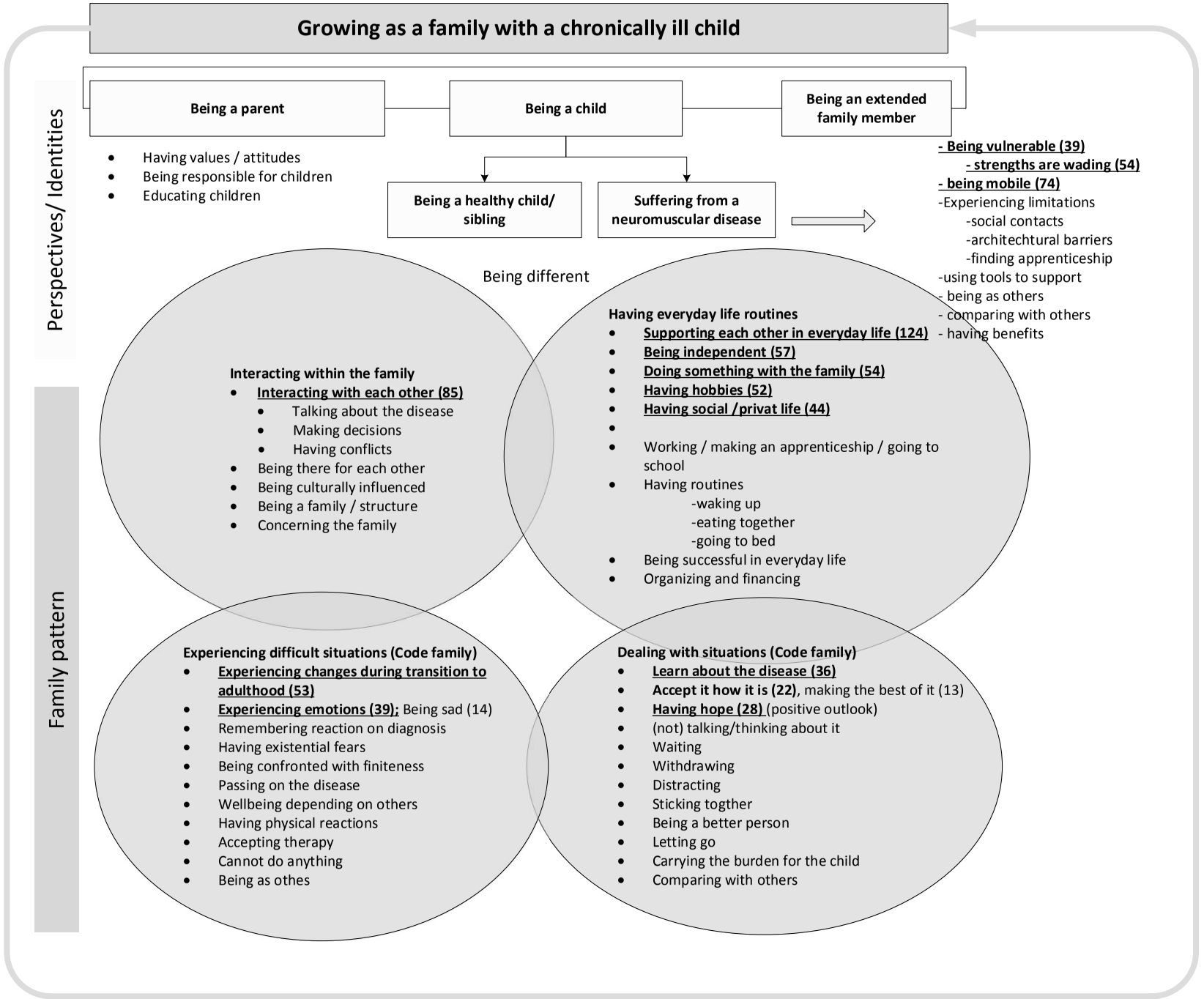
Appendix 2.8: Process of analysis



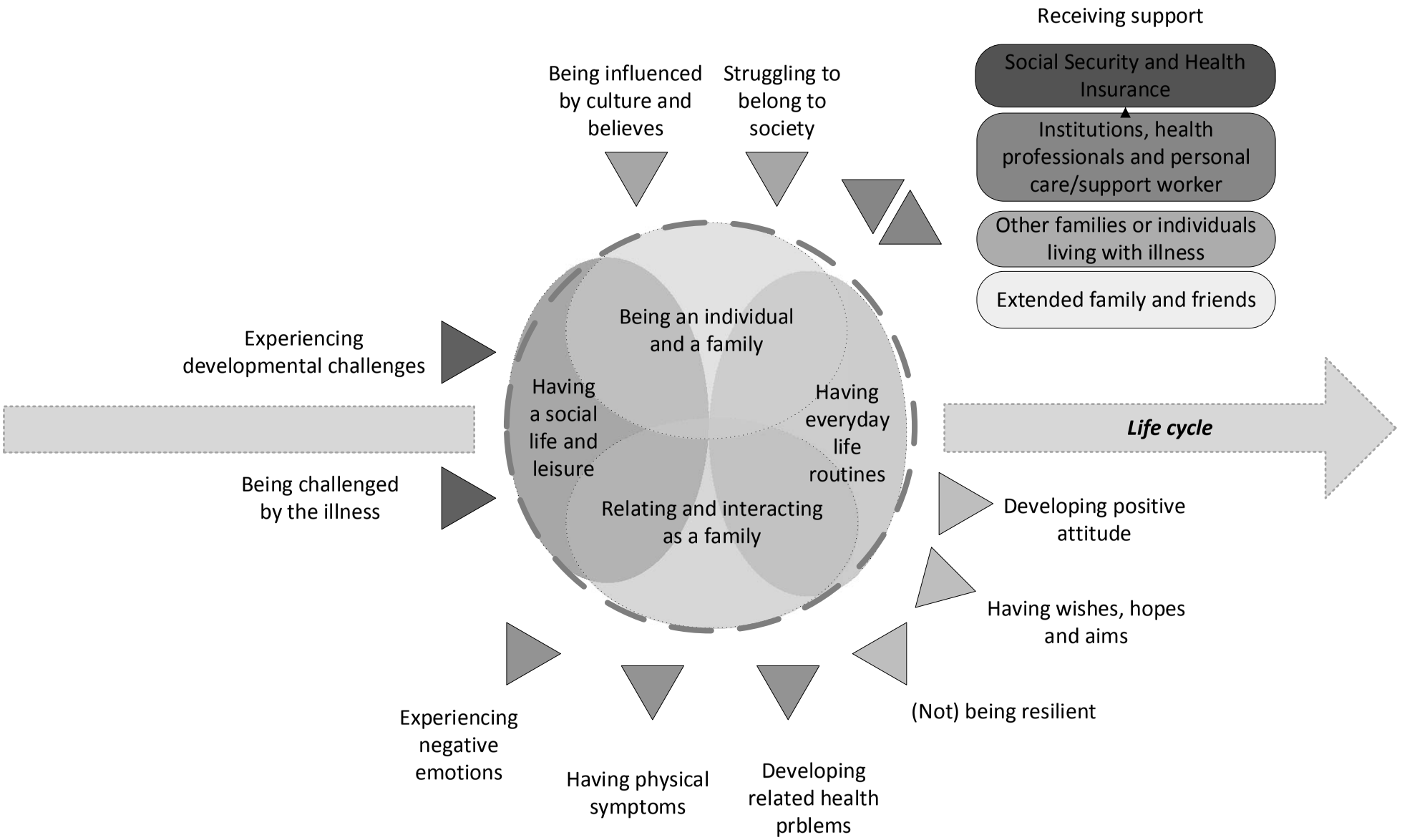
03.03.15_Coding 1rst Interview with Atlas.ti



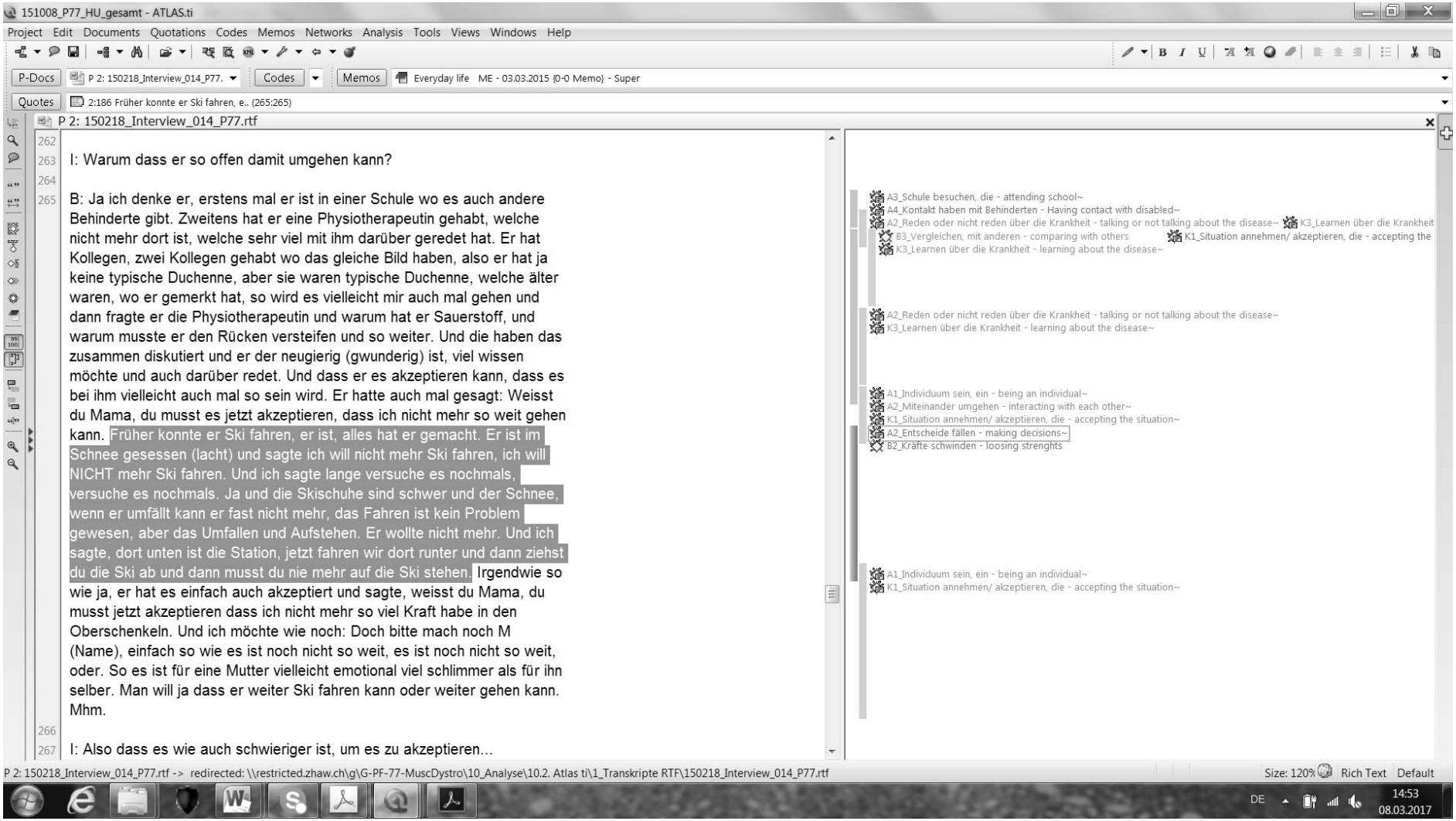
20.4.15_Diagram of analysis of two interviews with parents



7.7.15_Diagram of analysis of 10 interviews (parents (3), affected individuals (4), siblings (3)



A family with an family member living with neuromuscular disease moving through the life cycle



08.10.15_Coding of interview with Atlas.ti

Deutsch	English
Herausforderungen in der Entwicklung erleben	**Experiencing developmental challenges**
Die Kategorie „Herausforderungen in der Entwicklung erleben" beschreibt Herausforderungen oder Schwierigkeiten der Familienmitglieder in der Entwicklung während des Erwachsenwerdens einer jungen Person, die mit einer neuromuskulären Erkrankung lebt. Die Folgenden Codes sind Bestandteil dieser Kategorie (figure 1). Die Codes „sich von der Familie loslösen" und „selbständig sein und werden" sind die stärksten Codes und sie beeinflussen sich gegenseitig. „Mit Gleichaltrigen umgehen" ist der drittstärkste Code, und der ist verbunden mit „in einer Partnerschaft leben", „Sexualität erfahren", und „planen Kinder zu haben" und „ die Krankheit weitervererben". „Reifer werden" versus ein „Kleinkind bleiben" beeinflussen „selbständig sein und werden" und sind beeinflusst durch „erfahren, dass der Körper sich verändert".	The category „experiencing developmental challenges" describes challenges or difficulties of family members regarding their development during the transition into adulthood of a young individual living with neuromuscular disease. The following codes are properties of this category (figure 1) The codes „ Detaching from the family" and "being and becoming independent" are strong codes and they are mutually influencing each other. "Interacting with peers" is the third strongest code, and it is associated with "living in a partnership", "experiencing sexuality" and "planning to have children" and "passing on the disease". "Becoming mature" versus "remaining a toddler" are influencing "being and becoming independent" and are influenced by "experiencing that the body is changing"

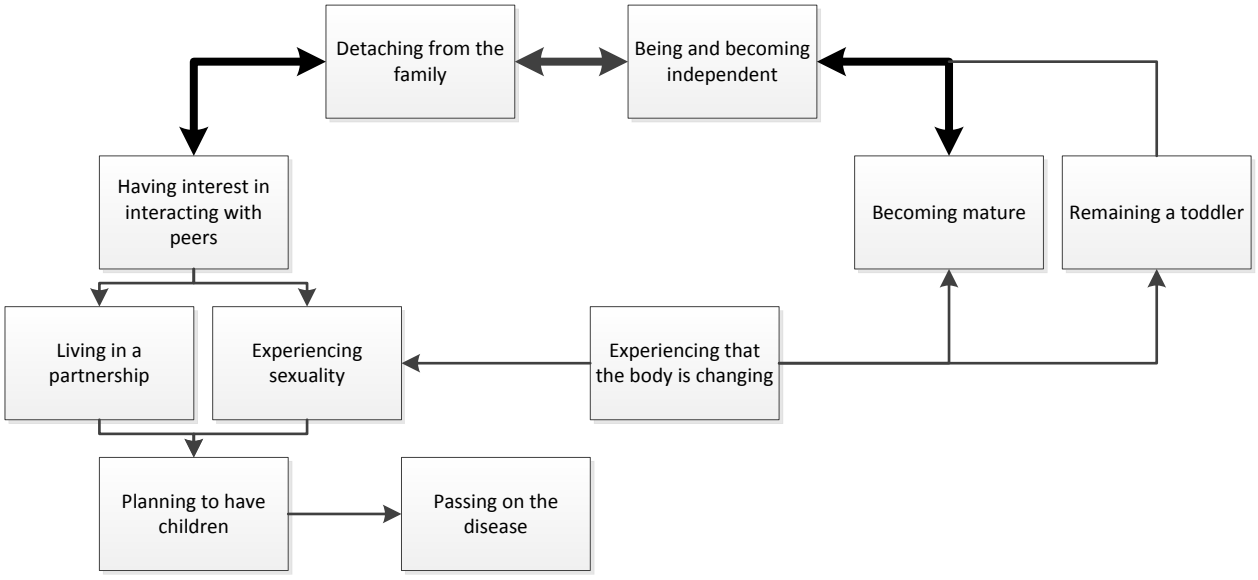
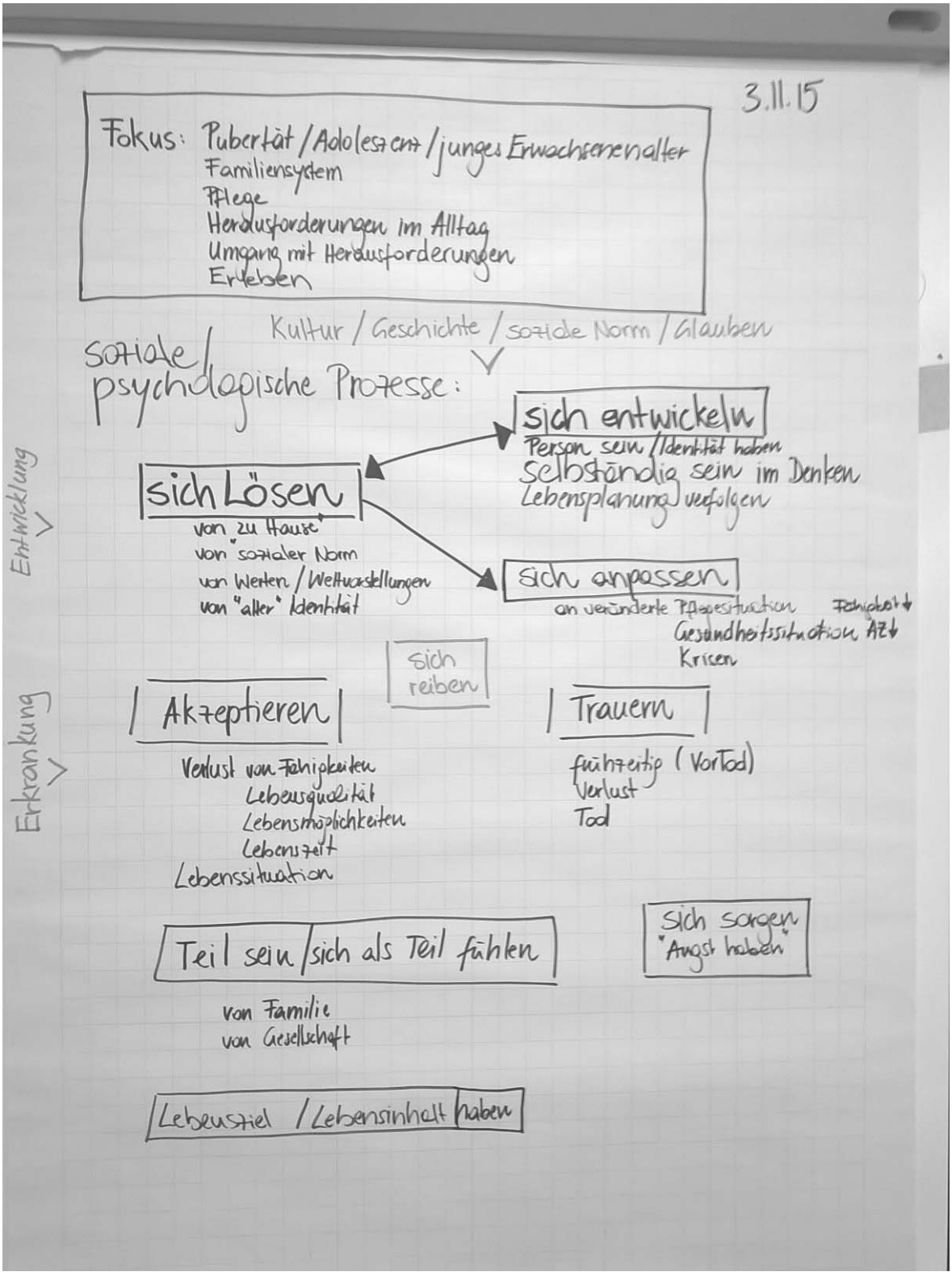


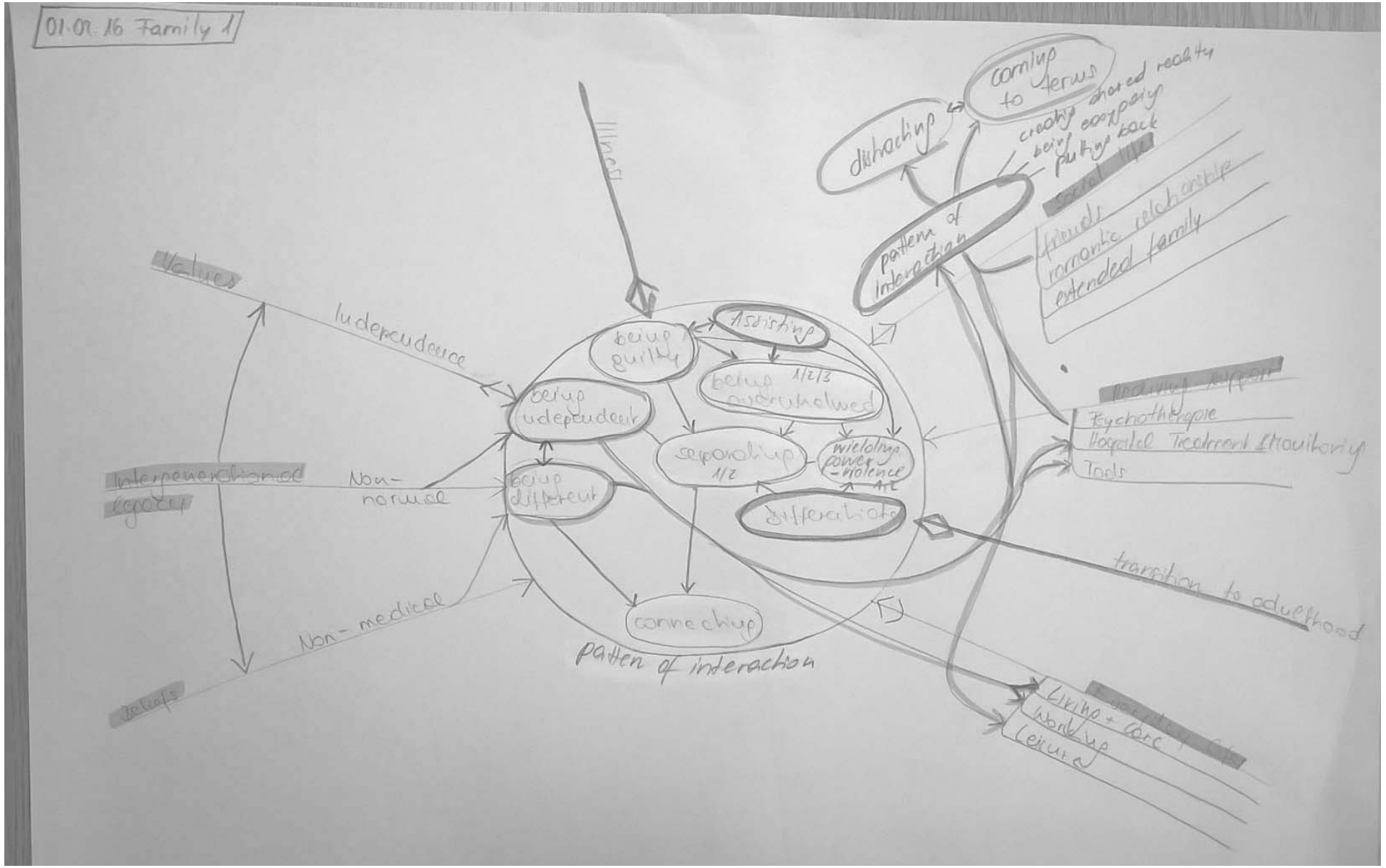
Figure 1: Experiencing developmental challenges

Deutsch	English
Sich von der Familie loslösen	**Detaching from the family**
„Sich von der Familie lösen“ beschreibt den Prozess des Übergangs in eine neue Phase des Familienlebens; Es ist der Übergang von Familie mit Kindern, zur Familie mit Jugendlichen bis hin zur Familie mit jungen Erwachsenen.	“Detaching from the family” describes the process of the transition into a new stage of the family life; It is the transition from being a family with children, to being a family with adolescents up to being a family with young adults.
Eine Eigenschaft dieses Prozesses ist, dass er eng verbunden ist mit der Entwicklung der Selbstständigkeit der Kinder. Die Konsequenz dieses Prozesses ist, dass Kinder selbstständiger werden und sich von den Eltern distanzieren. Eltern, im Gegensatz, haben mehr Zeit für sich.	A property of this process is that it is closely connected with the development of the independence of the children. As a consequence of this process children become more independent and they distance themselves from their parents. Parents, in contrast, have more time for themselves.
Eine Eigenschaft dieses Übergangs ist, das es als Schwierigkeit erlebt wird sich von der Familie zu lösen. Für Eltern ist es schwierig, sich von den betroffenen Kindern oder Jugendlichen zu lösen. Sie sehen es als Ihre Pflicht für Ihre Kinder zu sorgen.	A property of this process is that family members experience it as difficulty to detach from the family. They see it as their duty to care for their children.
Da die körperliche Beeinträchtigung und gleichzeitig die Angewiesenheit auf Unterstützung zunehmen, sind einige Eltern, die die Pflege ihrer Kinder übernehmen nicht entlastet. Im Gegensatz dazu nehmen Pflegetätigkeiten mehr Zeit in Anspruch.	As the physical limitation and the need for assistance is increasing, some parents that care for their children not relieved. In contrast, caring activities take more time.
Des Weiteren erleben Eltern das Loslösen ihrer gesunden Kinder als Dilemma. Sie möchten einerseits, dass ihre gesunden Kinder ein eigenes Leben führen und ausziehen, Arbeit finden und eine eigene Familie haben. Andererseits, werden die Geschwister auch gebraucht, um die Unterstützung für das kranke Familienmitglied zu leisten und die Eltern zu entlasten.	Furthermore, they experience the detachment of their healthy children as dilemma. On the one hand they want their healthy children to have their own lives, including moving out, have a job and an own family. On the other hand, they need their ill child’s siblings to assist him or her and to relieve the parents.
Physische Entfernung, dadurch hervorgerufen, dass die betroffenen oder gesunden Kinder	Physical distance, created by the ill or healthy child not living in the same physical

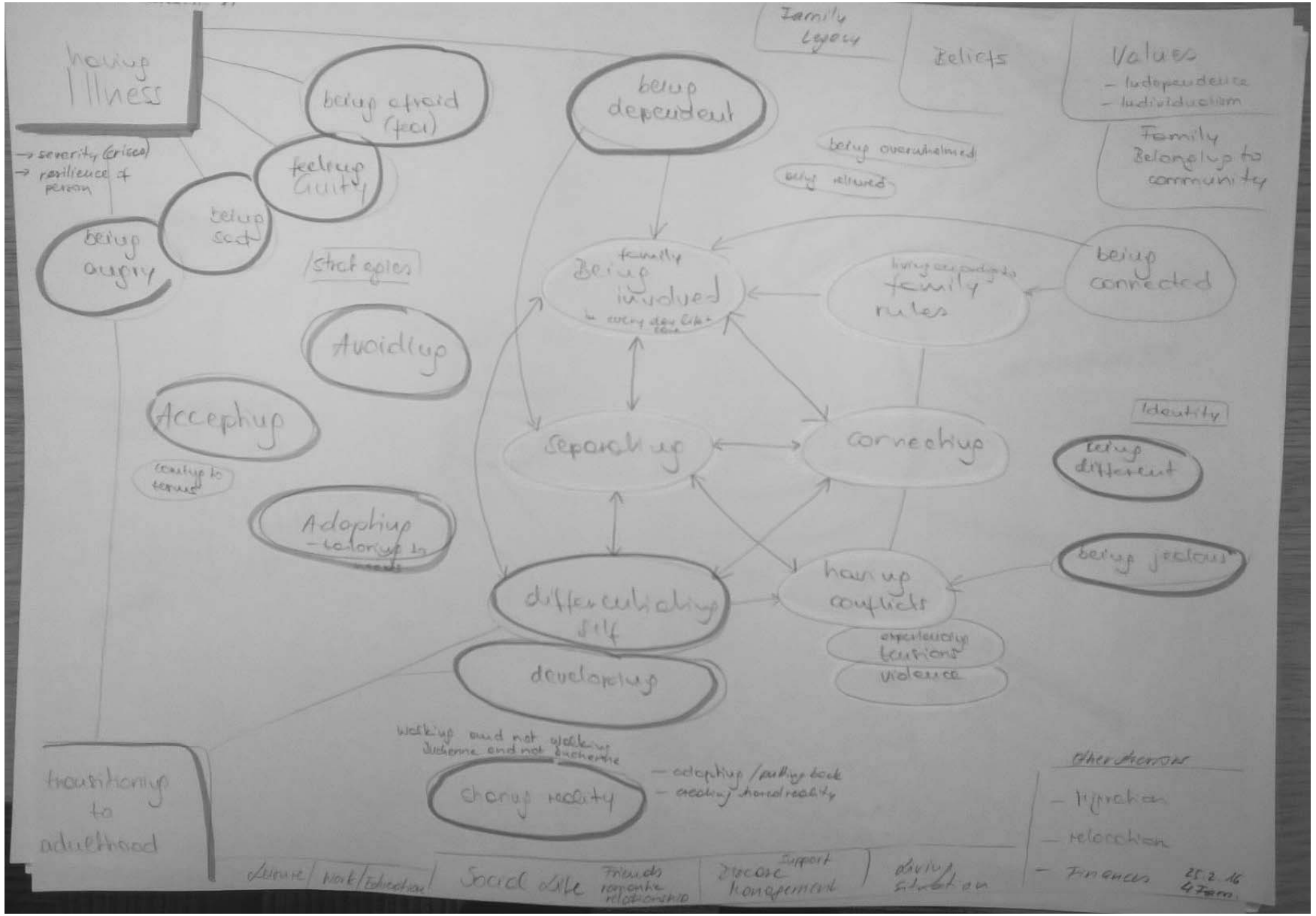
nicht mehr im selben Umfeld / zu Hause wohnen können oder wollen, wie der Rest der Familie, beeinflusst den Loslösungsprozess. Einige Betroffene wollen mit ihren Eltern wohnen bleiben, andere, die bereits unter der Woche in einer Institution leben, wollen am Wochenende bei ihren Eltern bleiben. Andere möchten ausziehen und selbständig wohnen mit 24 Stunden Betreuung. Immer zu Hause sitzen ist nicht cool. Sind mehrere Kinder von der Erkrankung betroffen sind, sind sie meist an den gleichen Orten untergebracht. Einige sind in verschiedenen Institutionen untergebracht.	environment / home as the rest of the family, because they had to or wanted to, influences the detachment process. Some affected individuals want to stay with their families, others, who live in institutions during the week, want to stay with their families on the weekend. Some also want to move out from home, and live independently with 24 hour assistance. Always being at home is “not cool”. Is more than one family member affected by the disease, they are normally living at the same place. Some live in different institutions.
Selbständig sein und werden	**Being and becoming independent**
Der Code selbständig sein beschreibt die Bedeutung der Selbständigkeit für Familienmitglieder während der Phase des Aufwachsens. Der Wunsch nach Selbständigkeit wird von Seiten der Familienmitglieder als stark beschrieben. Es ist bedeutend, selbständig zu sein. Selbständigkeit bedeutet Freiheit. Eine Mutter ist stolz, da sie als alleinerziehende Mutter die Situation im Griff hat und weiss wo sie Hilfe holen kann falls nötig. Gesunde und betroffenen Kinder haben den Wunsch selbständiger zu werden, unabhängiger von den Eltern zu sein. Dazu gehört einen Job zu haben, selbständig zu wohnen, einen eigenen Haushalt zu haben und eine Familie zu gründen. Kranke Kinder wollen sich selbständig Assistenzen organisieren, selbständig mobil sein (e.g. vorzeitige Autoprüfung), selbständig Entscheidungen treffen und so leben wie sie das möchten, Finanziell unabhängig von den Eltern zu sein. Einige betroffenen Kinder sind noch	The code being and becoming independent describes the meaning of independence for family members during the transition into adulthood. The desire for independence is described as to be strong by all family members. It is important to be independent. Independence means freedom. A mother is proud to be independent, as she as a single mother is handling the situation well and knows where to ask for help if necessary Healthy and ill children have the wish to become more independent from their parents. This includes having a job, living independently and having an own household and family. Ill children want to organise assistance themselves, being mobile independently (e.g. early driver licence), making own decisions, live how they want, being financially independent from parents. Some individuals are still independent, can eat



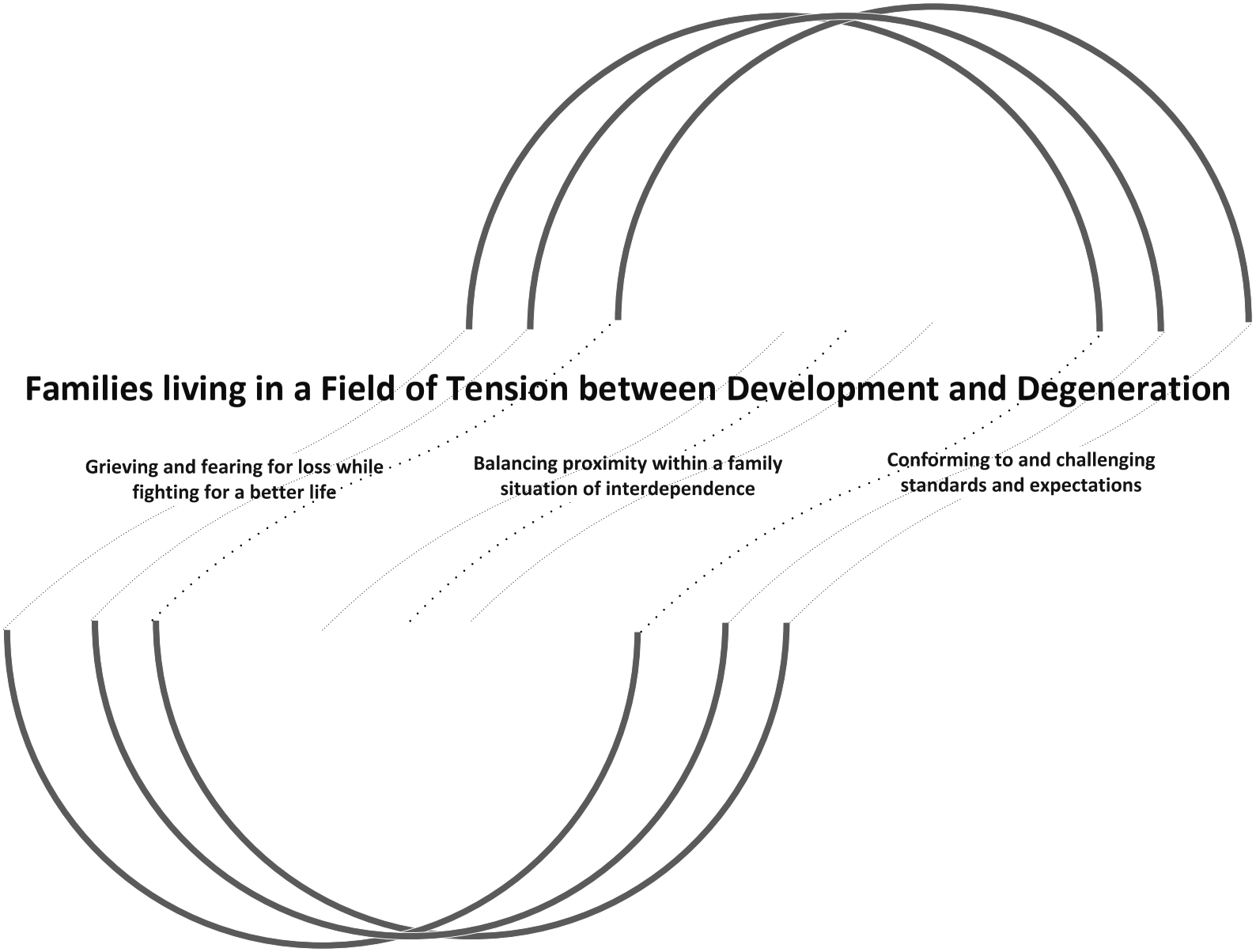
3.11.15_Notes from discussion on flip chart



01.02.16: Analysis of family system

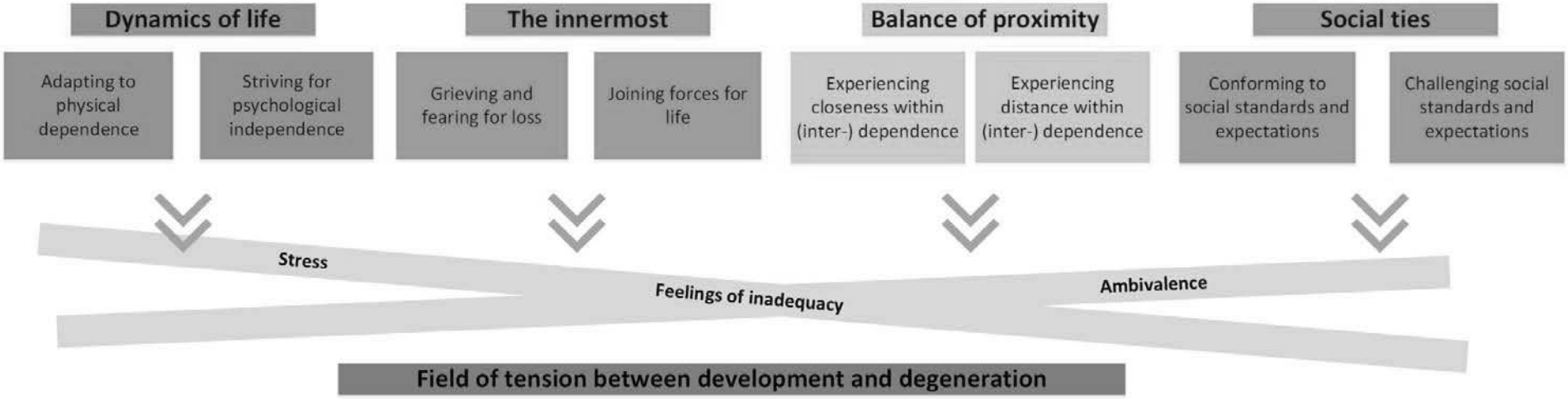


25.02.16_Analysis of families systems



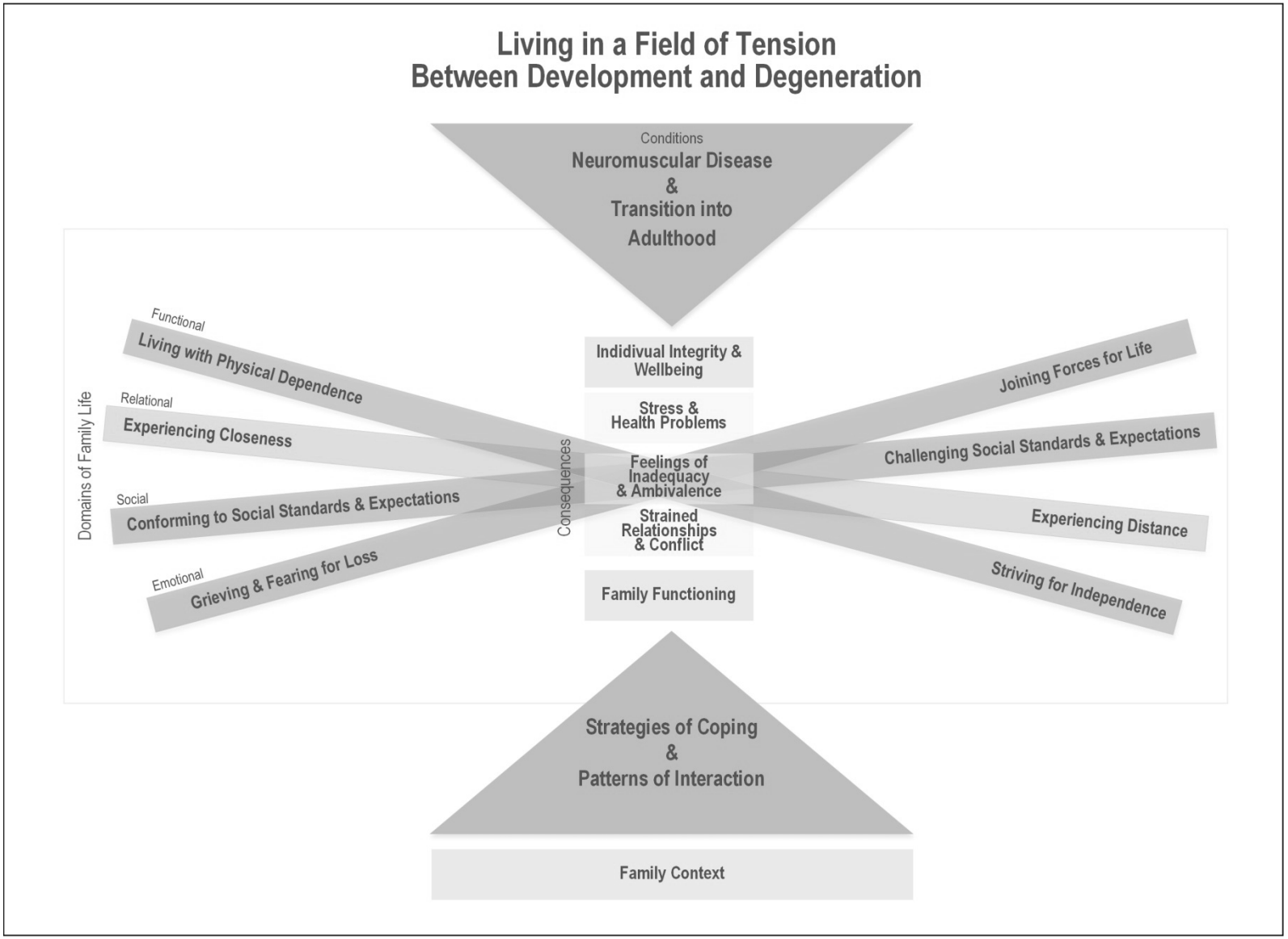
20.07.16_Model version 1

20.07.16_walo



28.09.16_walo

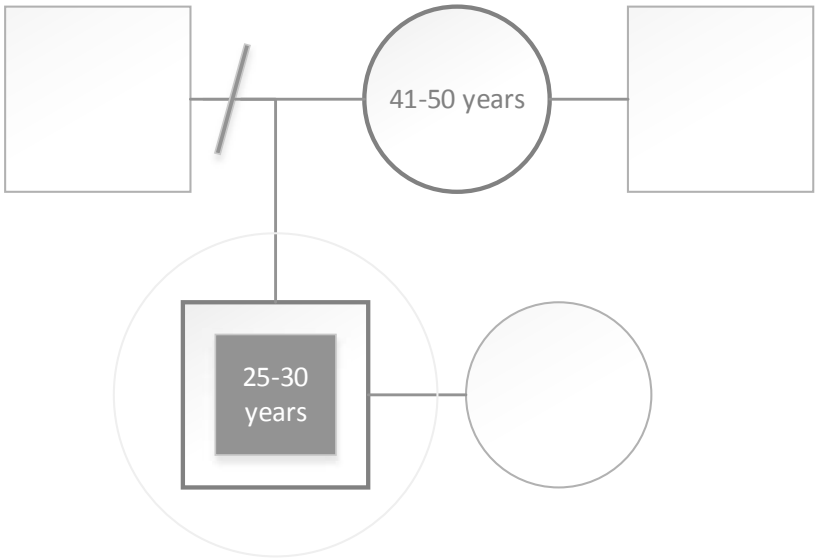
28.09.16_Model version 2


Living in a Field of Tension
Between Development and Degeneration
Conditions
Neuromuscular Disease
&
Transition into
Adulthood
Domains of Family Life
Functional
Living with Physical Dependence
Relational
Experiencing Closeness
Social
Conforming to Social Standards & Expectations
Emotional
Grieving & Fearing for Loss
Consequences
Indidivual Integrity & Wellbeing
Stress & Health Problems
Feelings of Inadequacy & Ambivalence
Strained Relationships & Conflict
Family Functioning
Joining Forces for Life
Challenging Social Standards & Expectations
Experiencing Distance
Striving for Independence
Strategies of Coping
&
Patterns of Interaction
Family Context

20.2.17_walo

Appendix 3: Family genograms

Family 1:



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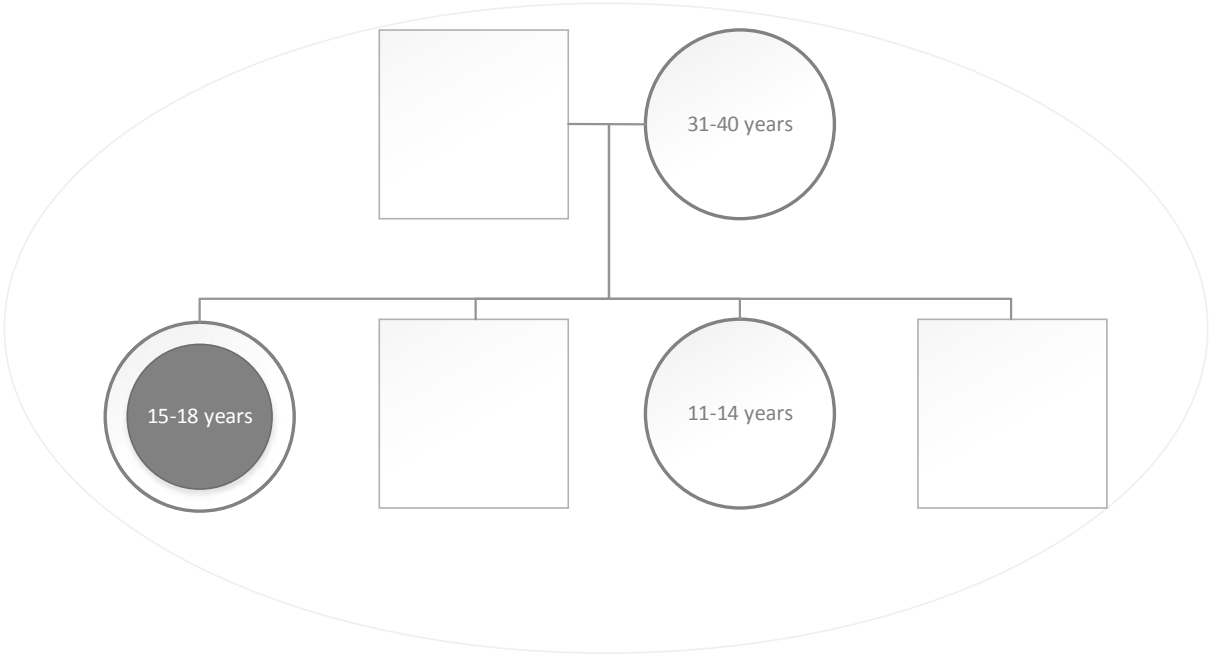

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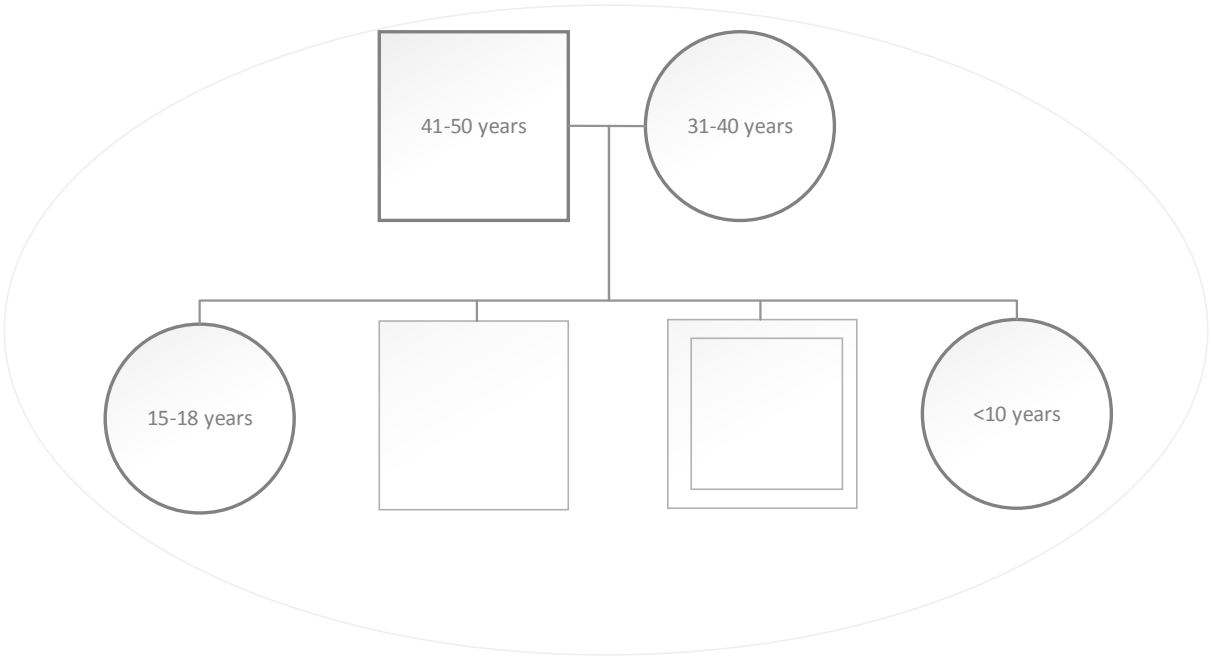

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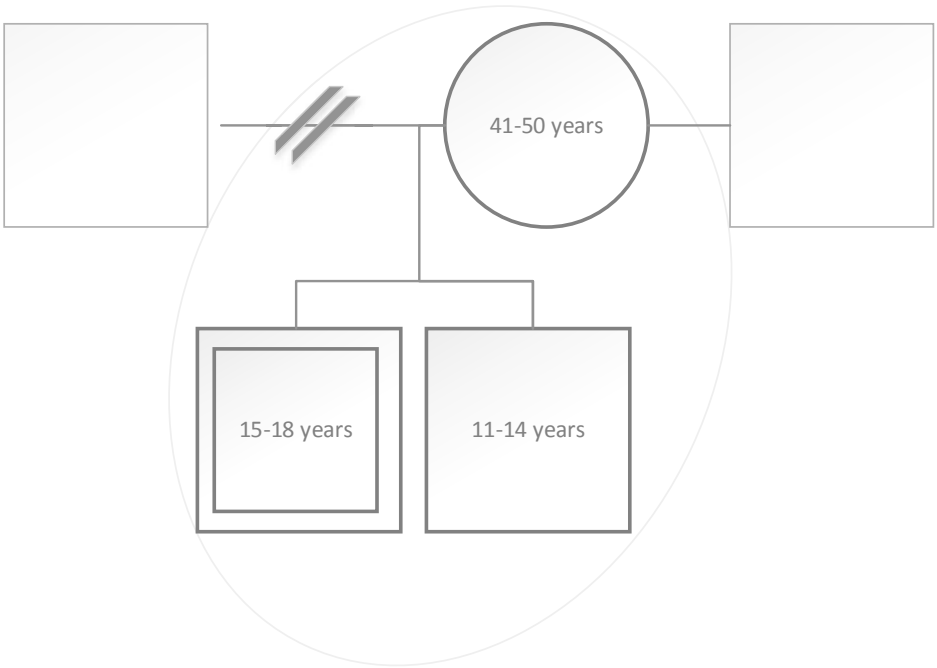

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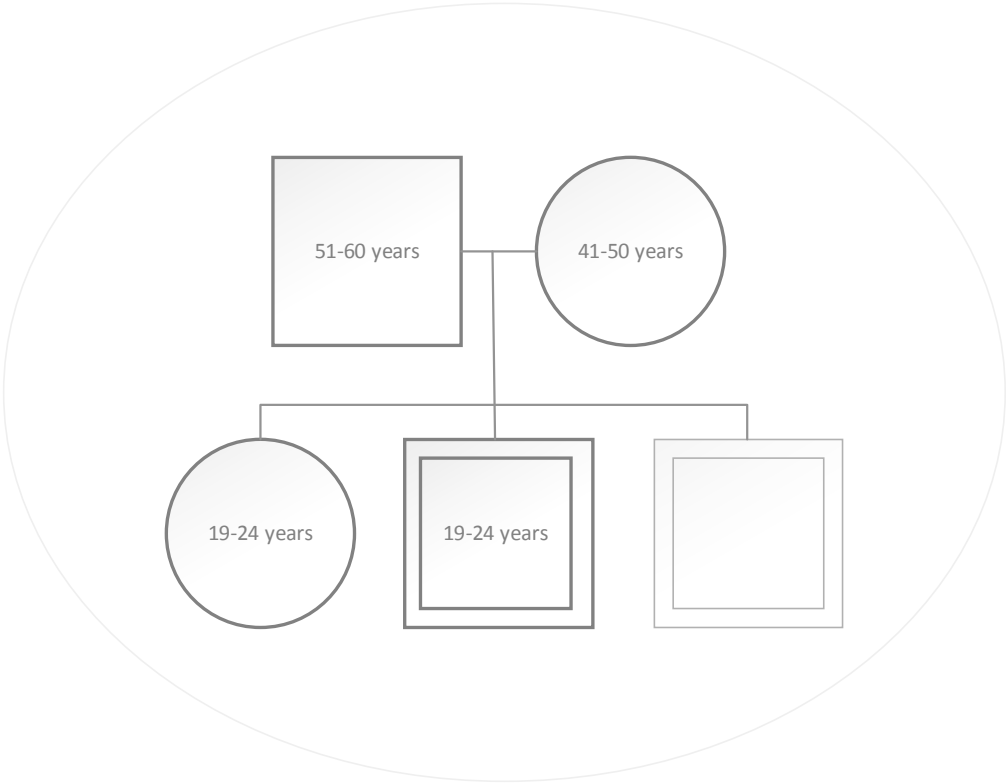

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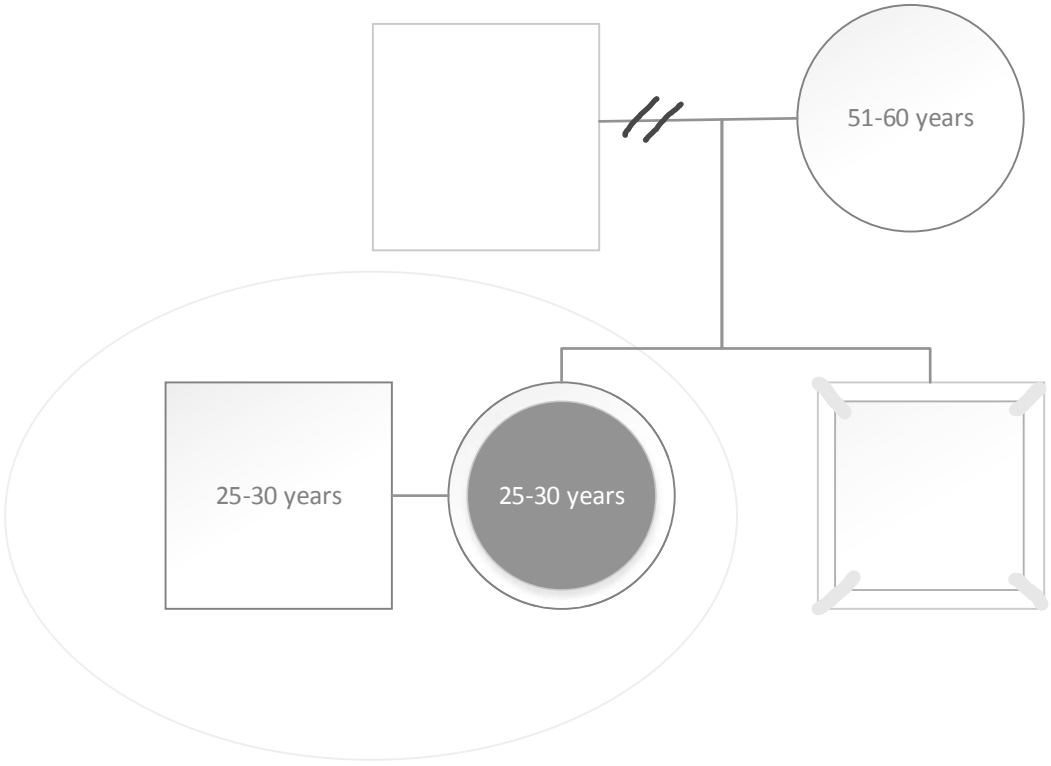

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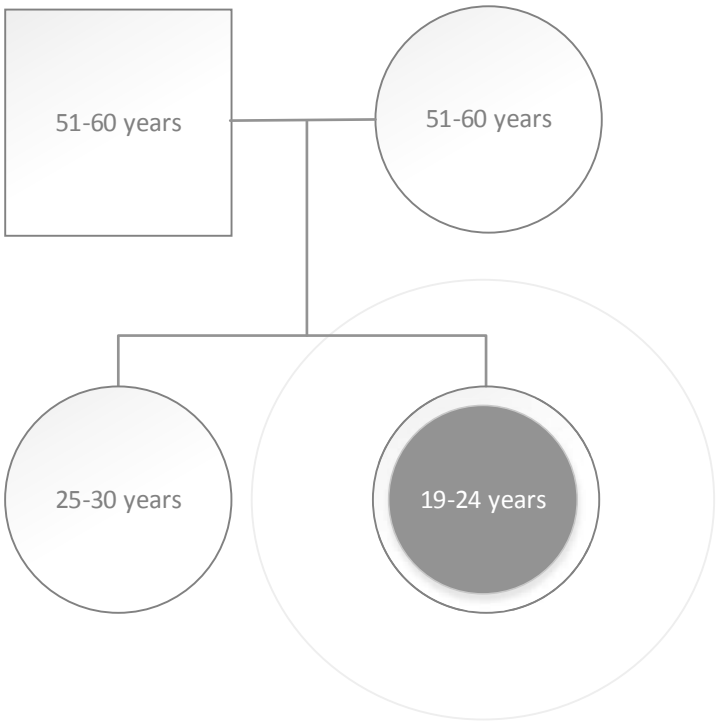

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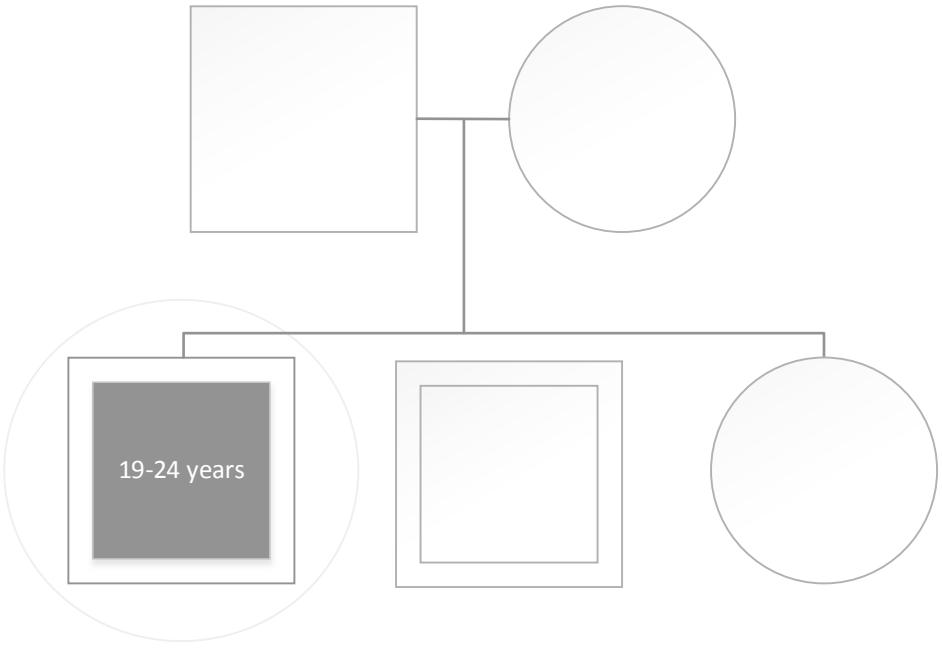

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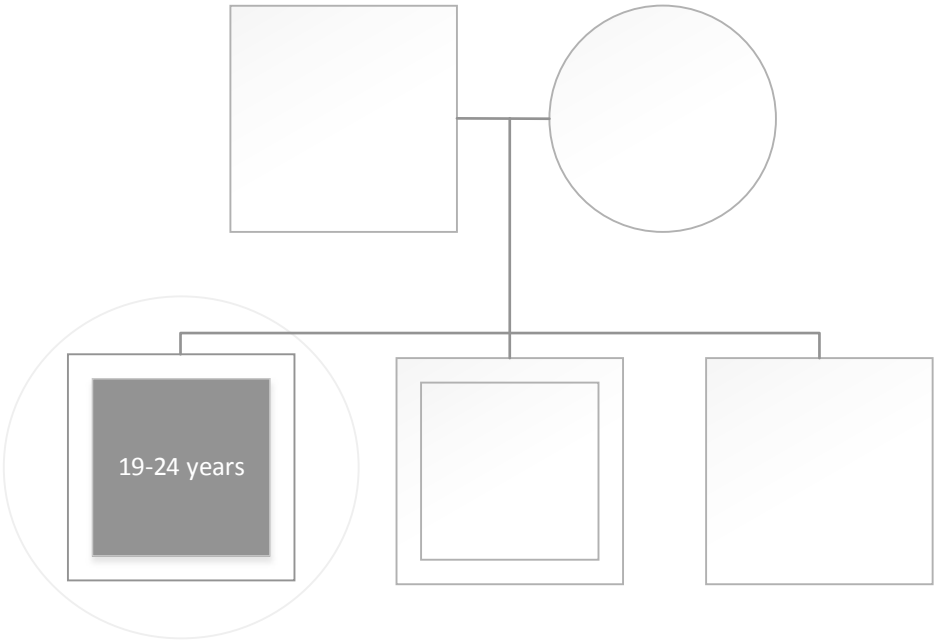
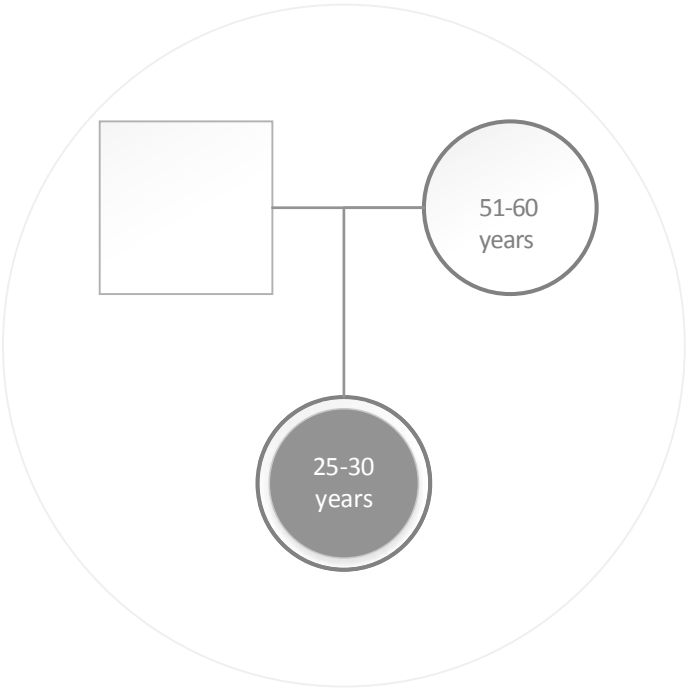


Familie 10:



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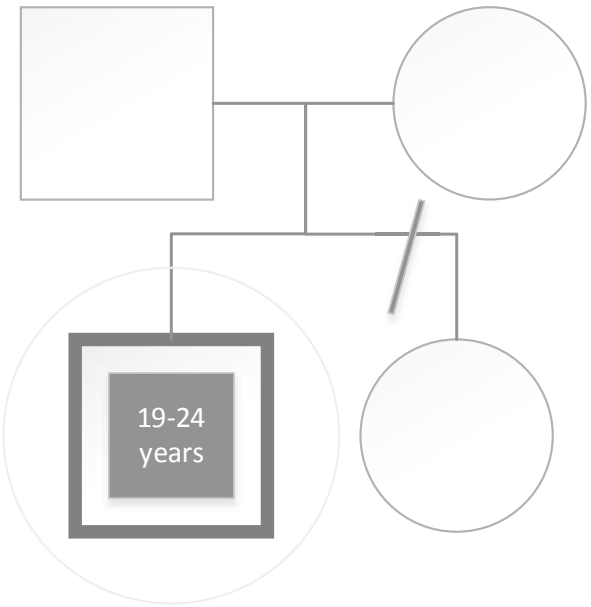

Familie 12: