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Abstract

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This thesis focuses on children with severe spinal muscular atrophy (SMA) and their families. Although the disease is severe, and the families are faced with challenges in everyday life related to the progressive muscle weakness that SMA causes, knowledge of their experiences of the situation is limited. The overall purpose of this thesis was therefore to explore how families, with a child who has SMA, experience the care received and their everyday life.

The thesis encompasses two projects: a two-nationwide survey with 95 bereaved and non-bereaved parents (response rate of 84%) and an ethnographical study with two families (17 interviews and participant observations at six occasions).

The findings showed that parents were generally pleased with the care their children received. However, there were some shortcomings, especially that staff lacked knowledge about the diagnosis, leading the parents to feel that they themselves had to take initiatives for measurements and treatments (Paper II). Further, the parents reported deficiencies in coordination between care providers (Papers I–II). The parents emphasised the importance of having a good relationship with staff (Paper II), to find ways to cope with everyday life and get practical support in everyday activities, as well as social support in dealing with disease and grief (Paper III). With the new medicine for SMA, the families' narratives were rewritten, and the families were facing slow improvements; small events that made a big difference. Hope was negotiated and struggled with in different ways by different family members, but contributed to how they dealt with the disease and the outlook on the future (Paper IV).

Many of the experiences described by the families can be useful for professionals in modifying their work to support these families in accordance with their needs.

Keywords

Spinal muscular atrophy, family, advice, paediatric palliative care, health care professional, parental perception, hope, resilience

List of papers

This thesis is based on the following papers, which are referred to in the text by their Roman numerals.

I. Hjorth, E., Kreicbergs, U., Sejersen, T., Jeppesen, J., Werlauff, U., Rahbek, J., & Lövgren, M. (2019). Bereaved parents more satisfied with the care given to their child with severe spinal muscular atrophy than nonbereaved. *Journal of child neurology*, *34*(2), 104-112.

II. Hjorth, E., Kreicbergs, U., Sejersen, T., & Lövgren, M. (2018). Parents' advice to healthcare professionals working with children who have spinal muscular atrophy. *European journal of paediatric neurology*, *22*(1), 128-134.

III. Hjorth, E., Kreicbergs, U., Sejersen, T., Werlauff, U., Rahbek, J., & Lövgren, M. (2019) Parents' advice to other parents of children with spinal muscular atrophy: Two nationwide follow-ups. Submitted.

IV. Hjorth, E., Lövgren, M., Kreicbergs, U., Sejersen, T., Asaba, E. *"Suddenly we have hope that there is a future":* Two families' narratives when a child with spinal muscular atrophy receives a new effective drug. Submitted.

Contents

Abbreviations

Abbreviation	Term
HIV	Human immunodeficiency virus
IAHPC	International Association for Hospice and Palliative Care
RCFM	National Rehabilitation Centre for Neuromuscular Diseases in Denmark
SMA	Spinal muscular atrophy
SFS	Swedish Code of Statutes (*Svensk författningssamling*)
UNCRC	The United Nations Convention on the Rights of the Child
WHO	World Health Organization

Preface

I have met many families affected by severe disease in my work as a nurse at the neurological department of the Astrid Lindgren Children's Hospital in Stockholm. The patients have included children with neuromuscular diseases. Many of these children made a strong impression on me: they were often in positive spirits despite having weak bodies. One of my first encounters with the muscular disease spinal muscular atrophy (SMA) was a meeting with a 7-year-old girl who came to the hospital for her annual follow-up. She had a solid schedule for the day, planning to meet her physiotherapist, occupational therapist, physician, and dietician, get measured and weighed, and have blood samples taken. She was happy, she thought it was fun to come to the hospital and go through all the tests. She was talkative and joked with staff members, who she had met several times before. When she was in the exercise room with the physiotherapist, she was going all-in on the tests, seeing the exercises as a fun game or competition. Meanwhile, both her parents stood crying behind her. They tried to hide their tears from their daughter, but the sorrow of seeing so clearly how much muscle function she had lost since last year's tests was difficult for them to bear.

Back then, working as a clinical nurse, I had no thoughts of becoming a PhD student. But life does not always take the paths one expects, sometimes it takes turns that lead to unexpected and exciting places. When I wrote my master's thesis within the framework of the specialist education in paediatric care, I came in contact with my current supervisors. They had just initiated a survey study on parents' experiences of the care of their children with severe SMA, and I got involved in the work as a master student and research assistant. I found the research study important and saw the possibility of gaining more knowledge about a group of patients who are often disregarded in research, despite having a diagnosis that imposes great suffering and challenges on the affected child and their family. I ended up applying for a PhD education within the project.

In the middle of my doctoral education, a new drug for SMA, that could slow the progress of muscular atrophy and prolong life significantly, was introduced and

approved for treatment in Sweden. SMA, which had been associated with inevitable death, changed into a disease that, with treatment, could possibly be united with a long, good life. The introduction of the new drug treatment represented a major change of context for my thesis, something that is reflected throughout my thesis.

1. Background

1.1. Heritability, incidence, symptoms, and prognosis of SMA

SMA is a rare and severe progressive neuromuscular disease usually presenting in early childhood. With an estimated incidence of 1/11,000 live births (Sugarman et al., 2012) and a genetic carrier frequency of approximately 1 in 50, SMA was until recently the second most common fatal autosomal recessive disorder (Prior, 2008). The disease is genetic, heritable, and caused by a mutation in the survival motor neuron 1 (SMN1) gene that results in a lack of functional SMN protein. This causes loss of motor neurons, leading to gradually increasing muscle weakness. In Sweden, six to nine children are diagnosed each year (National Board of Health and Welfare, 2017), and in 2018 there were 92 children under the age of 18 years with some form of SMA (National Board of Health and Welfare, 2020b)

SMA is typically classified into one of three classes, SMA types 1–3, based on age at presentation and disease severity, although an alternative five-grade classification has also been suggested (Carré & Empey, 2016). This thesis focuses on the two more difficult types of SMA as they impact on length of life, which SMA type 3 does not.

SMA type 1 is the most severe form of SMA and presents within the first six months of life. Infants with SMA type 1 do not achieve independent sitting. Without treatment, respiratory support, and enteral nutrition, the child with SMA type 1 usually does not survive past two years of age due to breathing difficulties and difficulty in sucking and swallowing (Carré & Empey, 2016).

SMA type 2 is characterised by onset at between six and 18 months of age. Children with SMA type 2 are able to maintain a sitting position unaided, but cannot walk independently. Children with SMA type 2, like children with SMA type 1, often have difficulties clearing tracheal secretions and coughing due to weak intercostal muscles. A majority live into early adulthood and, with proper care, many live well into adulthood (Lunn & Wang, 2008; Markowitz, Singh, & Darras, 2012).

Children with SMA type 3 are often ambulatory during childhood, but may ultimately need a wheelchair. Individuals with SMA type 3 develop little or no respiratory muscle weakness, and their life expectancy is not below average.

Although children with SMA are medically fragile, they retain their cognition and verbal intelligence along with normal sensory and emotional functioning. Above-average intelligence has been noted among persons with SMA type 2 (Von Gontard et al., 2002). Severe muscular weakness can give the child difficulties in communicating.

1.1.1. SMA treatment and management – then and now

Until just a few years ago, there was no effective treatment for any form of SMA. All treatment therefore focused on preventing complications from muscle weakness and maintaining quality of life, especially through respiratory support, nutritional support, and orthopaedics/rehabilitation (Arnold, Kassar, & Kissel, 2015; Finkel et al., 2018; Mercuri et al., 2018; Tassie, Isaacs, Kilham, & Kerridge, 2013). Such interventions could extend the life span of children with SMA type 1 or 2 by several years.

In 2007, a multidisciplinary team released a Consensus Statement for Standard of Care in SMA with recommendations for managing patients with SMA, which was updated in 2017 (Finkel, Mercuri, Meyer, et al., 2017; Mercuri et al., 2018; Wang et al., 2007). The consensus suggested that treatment options should be explored together with the family and in relation to the child's potential, quality-of-life issues, and family's desires. However, different countries have different approaches and treatment strategies. For instance, the French national paediatric neuromuscular network has considered a purely palliative care approach to be the most ethical treatment for children with SMA type 1, while other countries, such as the USA, have a more proactive approach with early non-invasive ventilation and gastrostomy, leading to prolonged survival (Hully et al., 2020). Sweden has a more restrictive approach to invasive ventilation in children with severe SMA, and dialogue with the parents about treatment has been in focus.

Recently, promising treatment has become available on the market, which has changed the SMA landscape dramatically. After clinical trials that showed slowed progression of muscle atrophy, improved survival in infants and children, and in

some cases even regain of previously lost muscular functions (Al-Zaidy et al., 2019; Finkel, Mercuri, Darras, et al., 2017; Mercuri et al., 2018), nusinersen (Spinraza) was approved by the US Food and Drug Administration in December 2016, and the gene therapy drug onasemnogene abeparvovec (Zolgensma) was approved in May 2019 (Vita, Vita, Musumeci, Rodolico, & Messina, 2019).

Onasmenogene is, at the time of writing, not yet approved in Sweden, or any country in Europe, but the European Committee for Medicinal Products for Human Use in March 2020 issued a positive opinion, recommending the granting of a marketing authorisation for the drug.

Nusinersen was approved in December 2017 in Sweden. The recommendation to Swedish county councils and regions is that children with SMA type 1 and 2 should have access to this new and very expensive drug if they meet certain criteria; the child cannot be too weak, and if no effect is apparent in the evaluation that occurs every year, treatment with nusinersen should be discontinued (The New Therapies Council, 2017). Treatment has lately opened up also for patients with SMA type 3, using criteria similar to those for types 1 and 2.

Nusinersen is not curative, unless given from a pre-symptomatic stage, but can alter disease progression and reverse loss of motor function in children with SMA type 1, 2, or 3 (Darras et al., 2019; Finkel, Mercuri, Darras, et al., 2017; Mercuri et al., 2018). In the study by Darras et al. (2019), one previously non-ambulatory child with SMA type 2 achieved independent walking, something otherwise not congruent with the definition of SMA type 2 (Darras et al., 2019). Further, if nusinersen treatment is given from a pre-symptomatic stage, ambulation is accomplished in many children otherwise expected to follow the progressive and lethal course of SMA type 1 (Darryl et al., 2019).

Although nusinersen is effective in inhibiting disease progression and for many children results in a functional improvement, some children do not improve (Pane et al., 2018; Pechmann et al., 2018). For children who have already lost major motor function when nusinersen is introduced, it is uncertain when, or if, their weak bodies can regain functions that can help them move, communicate, and breathe independently.

A small number of parents in Sweden have, after discussing ethics with health care professionals, chosen not to initiate treatment with nusinersen. In other countries, some parents have terminated nusinersen therapy. Data on this have not yet been published in any study. Such choices, along with the criteria for nusinersen in Sweden and in many other countries, mean that not all children have access to nusinersen.

Even with the many benefits of the pharmacological development, there is still some uncertainty regarding the disease trajectory, which calls for more knowledge into what can be expected from the new treatment.

1.2. Care of children with SMA and their families in the Swedish welfare society

1.2.1. Health care in Sweden: a brief overview

We know that SMA has existed for many generations, but how the disease has been treated and viewed through history is hard to know. In the early 1890s, Werdnig and Hoffman described SMA as a disorder of progressive muscular weakness beginning in infancy that resulted in early death, though the age of death varied. Persons with a milder form of SMA, who survived childhood, were at this time probably socio-culturally affected by their disability, through reduced likelihood of getting work, getting married, and having children (Vikström & Haage, 2015). Until the introduction of antibiotics in the 1940s, it was unlikely that children with SMA types 1 and 2 survived childhood.

The foundation of the current welfare policies, involving social services and health care, emerged during the 20th century in the Scandinavian countries (Lindqvist & Hetzler, 2004). Protection of children came to be considered more as a task for the state than it had been before. This led to increased state influence over children's lives through legislation, preschool, school, social services, and institutionalisation of children (Berggren & Trägårdh, 2015; Sandin, 2003). This approach was also reflected in paediatric care. Children with severe diseases were, from the end of the nineteenth century, institutionalised in hospitals, separated from their parents and siblings. The reason behind this was mainly a fear of visitors bringing infections to the ward. It was also pointed out that the children

became very upset after visits, whereas they soon settled down on the ward and forgot about home when left to themselves (Hallström & Lindberg, 2015; Zetterström, 1984). However, in the middle of the 20th century, evidence emerged that children, left among strangers, experienced great suffering due to separation from their families, with long-term effects (Bowlby, 1951; Zetterström, 1984). Since then, paediatric care has been developed to involve the entire family to a greater degree (Hallström & Lindberg, 2015), with the family's and child's own perspectives being taken into account (Coyne, Hallström, & Söderbäck, 2016; Coyne, Holmström, & Söderbäck, 2018).

In Sweden, health care for children is mainly public – controlled and subsidised by the state, counties, and municipalities. All children with SMA are entitled to health care free of charge, in the form of emergency care and specialised health care, supplemented with municipal rehabilitation, where the child is entitled follow-up care from a physician, occupational therapist, physiotherapist, and dietician. In Sweden, there are seven specialised muscle teams located at different hospitals. In our neighbouring country, Denmark (where data have been collected within the framework of this thesis), the systems are organised slightly differently. In addition to emergency care, specialised health care, and municipal rehabilitation, Denmark has a national competence centre, the National Rehabilitation Centre for Neuromuscular Diseases (RCFM), which supports health care and social services by providing specialised knowledge about rehabilitation of people with rare neuromuscular diseases. All persons in Denmark with a muscular disease can be referred to the competence centre.

1.2.2. Societal support for families living with a child with severe disease or disability

1.2.2.1. Support for the child

During the 20th century, the child's position changed from being a private concern of the parents to becoming a societal matter, with the child seen more as a competent and participating citizen with their own rights (Sandin, 2003). The United Nations Convention on the Rights of the Child (UNCRC) has contributed to strengthening the child's position in society. In paediatric care, the best interest of the child should be a primary consideration, as stated in Article 3 of the UNCRC. In addition, children who are capable of forming their own views should

have the right to express those views freely in all matters affecting them. The views of the child should be given due weight in accordance with the age and maturity of the child (Article 12 UNCRC). Further, States Parties of the Convention are obliged to work actively for children's rights to health care and rehabilitation, and to prevent all forms of discrimination against children, including discrimination of disability (Articles 2, 23, and 24).

To fulfil these societal obligations, certain laws in Sweden are enacted to strengthen the rights and opportunities of children with a disease. One of the relevant acts for many children with SMA is the Act concerning Support and Service for Persons with Certain Functional Impairments (SFS 1993:387), which specifies the rights for persons with disabilities that cause significant difficulties in daily life, and guarantees the support needed in daily life. The goal is for the individual to have the opportunity to lead a life like anyone else's, e.g., with the support of a personal care assistant. In most cases, a personal care assistant has no formal nursing education. In Sweden, the costs for such personalised support are borne by the regional social insurance offices.

The Patient Act, which was enacted in Sweden in 2015 (SFS 2014:821), states that paediatric health care should work to promote what is best for each child (Ch. 1, Sec. 8) and that when the patient is a child, the child's attitude towards ongoing care or treatment should be clarified in so far as possible. The child's attitude should be given due weight, with account taken of their age and maturity (Ch. 4, Sec. 3). Further, the UNCRC became Swedish law in 2020, and the implications of that are yet to be seen in health care and society.

1.2.2.2. Support to the family

In many countries, it is established by law that it is the family that is responsible for a sick family member, a responsibility that encompasses providing financial security. However, in Sweden, as in the other Scandinavian countries, the social security system has a major responsibility for a sick person and those who care for her/him (Berggren & Trägårdh, 2015; Sand, 2010).

In Sweden, a general social insurance provides basic financial security to all families with children under 18 years. The insurance includes parental benefits, which parents are entitled to for care of a healthy child for a total of 480 days per

child. If the child becomes temporarily ill, temporary parental benefits are paid to parents who stay home from work to care for their ill child (80% of one's regular salary for a maximum of 120 days per years). Parents of a child with a disability, who needs extra care, are entitled to a child carer's allowance (omvårdnadsbidrag in Swedish) and have the right to reduced working hours until the year the child turns 19. If the child's condition is life-threatening, the parents are entitled to parental benefits for an unlimited number of days (Försäkringskassan, 2020).

Since 2009, the municipalities in Sweden are under obligation to provide social and practical support to persons who care for a family member with long-term disease or disabilities (Social Services Act SFS 2001:453). Despite this obligation, support to caregivers is given to varying degrees in different parts of the country (National Swedish Board of Health and Welfare, 2014). There are few descriptions of how caregivers in Sweden manage and experience their own situation (Sand, 2010).

Siblings' rights to support are not clearly protected. The Health and Medical Services Act (SFS 2017:30) stipulates that a child's need for information and support must be taken into account if the child's parent or any other adult with whom the child is permanently living has a severe disease or disability or unexpectedly dies. Unfortunately, siblings to children with severe disease are still not visible in the Health and Medical Services Act. However, the Act (2018:1197) on the UNCRC applies to all children, including siblings to children with severe diseases, and in particular Article 12, which states that all children have the right to make their voices heard on matters pertaining to their own situation, may be relevant for siblings.

1.2.3. Palliative care of children with SMA

1.2.3.1. Definitions of palliative care

As previously described, SMA was a life-threatening disease with no effective therapy to slow the muscular atrophy until a few years ago. Children with incurable and life-threatening disease have the right to palliative care that relieves suffering and improves quality of life. There are several different definitions of palliative care. One frequently used definition is that of the World Health Organization (WHO), which states that palliative care is an approach that

improves the quality of life of patients and their families facing the problems associated with life-threatening illness, through the prevention and relief of suffering by means of early identification and impeccable assessment and treatment of pain and other problems: physical, psychosocial and spiritual (WHO, 2002). WHO also has a specific definition of palliative care of children that has similarities with the one for adults, but emphasises that palliative care of children applies to chronic diseases, is applicable already from diagnosis, and can be given at many different care levels.

In 2019, the International Association for Hospice and Palliative Care (2019) (IAHPC) published a new and more updated definition of palliative care that includes all ages. I interpret the new definition to be more in line with the WHO definition of palliative care for children than the one for adults, in the sense that it emphasises that palliative care is applicable throughout the course of an illness and all health care settings. Unlike the WHO's definition of palliative care, the new definition by the IAHPC focuses on the relief of suffering due to *severe* illness, not *life-limiting* or *life-threatening* illnesses. An expansion of the concept of palliative care has taken place since it was first introduced as care in the terminal phase, something that is also reflected in this new definition. This expansion has led to discussions about the risk of palliative care becoming watered down, as the boundary between palliative care and other care may be perceived as unclear.

During most of my doctoral education, I used the WHO definition of palliative care for children. However, when the updated definition came from the IAHPC, I saw it as contributing to a more holistic care of children with severe diseases and emphasising the importance of basic palliative care training in health care professionals, something that I find crucial. This thesis therefore takes its starting point in the IAHPC's new definition.

1.2.3.2. Palliative care in paediatric clinics

Since palliative care is broad in its approach, aiming to relieve symptoms and improve quality of life for the child and their family, it can be assumed that a palliative care approach is practiced widely in paediatric care for children with severe diseases. However, there is a lack of knowledge about palliative care in society in general, as well as within paediatric health care (de Visser & Oliver, 2017). Common misconceptions are that palliative care is only given at the end of

life, that it represents a limitation of treatment, or that it is addressed only at persons with cancer (Boersma, Miyasaki, Kutner, & Kluger, 2014; de Visser & Oliver, 2017; Strand, Kamdar, & Carey, 2013; Weaver et al., 2015). On the contrary, paediatric palliative care is a comprehensive approach that encompasses many types of conditions, including conditions among children for which curative treatment may be feasible but can fail, conditions in which premature death is inevitable, progressive conditions without curative treatment options, and irreversible but non-progressive conditions causing severe disability leading to susceptibility to health complications and likelihood of premature death (International Children's Palliative Care Network, 2020).

Staff trained in paediatrics are often good at emphasising health and possibilities for children. The health-promoting approach is prominent, and topics like death and palliative care are often foreign to paediatric clinics. This health-promoting approach is good and important, but in some cases might discourage professionals from acknowledging issues related to death or dying, even when caring for a child with an incurable and life-threatening disease. The health-promoting approach is not, as I see it, contradictory to the palliative care approach. On the contrary, the health-promoting approach is an important part of the palliative care approach as serving to provide support to help patients live as fully as possible until death. In some cases, care could be improved by narrowing the palliative care approach and acknowledging the presence of death, when caring for a family with a child with severe disease.

Evidence shows that children with progressive neuromuscular diseases can benefit from paediatric palliative care regardless of disease trajectory (Ho & Straatman, 2013). Early integration of paediatric palliative care of children with neurological diseases can help parents as they navigate the complexities of their child's care needs (Hauer & Wolfe, 2014; Liberman, Song, Radbill, Pham, & Derrington, 2016) and to facilitate multi-professional teams discuss pain management, nutritional or respiratory support, surgical interventions, or spiritual or psychosocial needs (Hauer & O'Brian, 2011; Schwantes & O'Brien, 2014). A palliative care approach can also support parents in the many treatment decisions required regarding their child's care (Finkel, Mercuri, Darras, et al., 2017; Mercuri, Bertini, & Iannaccone, 2012).

1.2.3.3. Palliative care of children with SMA

Palliative care has been suggested in literature as a suitable care model for children with SMA, but this was before the introduction of nusinersen, which has altered the disease from acutely life-threatening to more chronic, with treatment. One might ask whether the palliative approach is suitable for children with SMA treated with nusinersen. It is likely that SMA, in future, will not be seen as a deadly disease, and children with early treatment introduction may be asymptomatic. For those children, palliative care may not be applicable. However, as the situation stands today, there are still children with SMA who have severe symptoms, meaning that the disease is still potentially life-shortening. As long as this uncertainty remains, there is a need for care that can acknowledge that death is a possible threat, with the aim of providing the conditions for a good life with the highest quality of life possible. Competence in palliative care is needed to assess these potential needs in children with SMA and their families, and to provide evidence-based holistic care, that deals with physical issues, psychological and spiritual distress, and the social needs of the child and their family (International Association for Hospice and Palliative Care, 2019).

The development of palliative care will continue, and I see it as important to further increase the knowledge of palliative care to ensure that the right care can be given to the right person at the right time.

1.3. Family perspectives of living with severe disease

Families often share a sense of belonging and a mutual and strong commitment to each other's lives. When a child has a severe disease, the entire family suffers (Wright & Bell, 2009). The perception of what a family is, how it is composed, and what its function is, varies across the world and over time. A common way to define family is as people bound by blood or by legal status. However, it has become more common to define family as a social construction, where the family members themselves define who belongs to the family (Whall, 1986). This thesis follows Wright and Leahey (2012) definition: "the family is who they say they are." In this thesis, the family as a concept is viewed from a systemic perspective and seen as a system in which each member represents one part, together

becoming more than the sum of these parts (Wright & Leahey, 2012). In a family, people are included in each other's lives and a change in the life situation of one person will affect the lives of the others (Öquist, 2008). Further, the family is seen as a system in constant interaction with its surroundings, and participates in mutual relationships with other systems, like relatives, friends, health care professionals, and colleagues (Wright & Leahey, 2012).

1.3.1. Children's perspective of living with SMA or other neuromuscular disease

Children's perspectives of living with SMA are limited in research, but children's perspectives of other neuromuscular diseases and of being dependent on medical equipment have been described. The children have described an everyday life full of daily physical training and a struggle to minimise the risk of infections (Earle, Rennick, Carnevale, & Davis, 2006). Children have emphasised the importance of social life, for instance through gaming and social media (Earle et al., 2006; Israelsson-Skogsberg, Heden, Lindahl, & Laakso, 2018), but also mention feelings of discrimination and a sense of being different (Skyrme, 2016). Being dependent on medical equipment, such as a wheelchair and mechanical ventilation, has been described from the perspective of children as something they are knowledgeable about and comfortable with. The equipment has been described as representing only a small part of the overall challenges in life (Earle et al., 2006; Israelsson-Skogsberg et al., 2018).

Studies have been conducted to measure quality of life in children with SMA. Results have shown that the mean quality of life is lower in children with SMA than in healthy children (Iannaccone et al., 2009). Interestingly, studies have shown that children with SMA rate their quality of life higher than their parents do in proxy reports (Iannaccone et al., 2009; Kocova, Dvorackova, Vondracek, & Haberlova, 2014) and that parents estimate their child's quality of life higher than the children's professional care providers do (Bach, Vega, Majors, & Friedman, 2003). Some studies have reported that the degree of physical dysfunction in children with SMA affects their quality of life (Iannaccone et al., 2009), while others report the opposite (de Oliveira & Araujo, 2011). Individuals with SMA type 2 or 3 (including both children and adults) have reported four functions that have major impact on the quality of life: the ability to use the

restroom alone, self-feeding, washing oneself, and performing transfers on one's own (Rouault et al., 2017). Low behavioural problems and psychiatric comorbidity were found in children and adolescents with SMA (Laufersweiler-Plass, Rudnik-Schoneborn, Zerres, & Backes, 2003).

1.3.2. Parental perspective of living with a child with SMA or other neuromuscular disease

Becoming a parent is a big event and in most cases associated with happiness and love. Parenthood can be experienced as unfamiliar and overwhelming and can cause stress when the parent tries to meet the demands related to parenting (Abidin, 1990; Nyström & Öhrling, 2004). Parents of a child with a chronic disease face the same stresses and challenges as other parents, with the addition of the stress caused by the disease. The disease may involve stressors that include hospitalisation, management of suffering in the child, and participation in treatment and treatment decisions. Indirect effects of the disease may affect parents' decisions about work, education, having additional children, and relying on public support (Reichman, Corman, & Noonan, 2008).

Living with a child with progressive neuromuscular disease other than SMA has been described by parents as causing anxiety, depression, and feelings of social isolation and as affecting the quality of sleep (Magliano et al., 2014; Mah, Thannhauser, McNeil, & Dewey, 2008; Yılmaz, Yıldırım, Öksüz, Atay, & Turan, 2010). Parents have described that the demands of the child's care in daily life become "normal" over time, but that they struggle with maintaining some of the old normality in their lives from before the child became ill (Mah, Thannhauser, McNeil, et al., 2008). Despite all the adversities with having a child with a neuromuscular disease, parents also emphasise the positive effects on the family, like learning to live fully in the present (Erby, Rushton, & Geller, 2006) and gaining a greater sense of compassion and understanding for others (Magliano et al., 2014; Mah, Thannhauser, McNeil, et al., 2008). Thoughts about the child's future have been described as encompassing both hope and avoidance (Erby et al., 2006), feelings of uncertainty (Mah, Thannhauser, McNeil, et al., 2008; Peay, Meiser, Kinnett, & Tibben, 2018), and fear about unclear responsibilities when the child becomes an adult (Peay et al., 2018; Yamaguchi & Suzuki, 2015). Family relationships, social support (Magliano et al., 2014; Magliano et al., 2015; Peay et

al., 2018; Thomas, Rajaram, & Nalini, 2014), and religion (Thomas et al., 2014) have been described as potentially protective factors when having a child with a neuromuscular disease.

Studies of parents of children with SMA are limited, but some have been identified. In these, parents of children with SMA have described an anxious period during the time between recognition of symptoms and receiving the diagnosis, a process which could take weeks or years (Lawton, Hickerton, Archibald, McClaren, & Metcalfe, 2015; Murrell, Lotze, Farber, Crawford, & Wiemann, 2017; Qian et al., 2015; Rallison & Raffin-Bouchal, 2013). The diagnosis has been described both as being given in a considerate manner (Lövgren, Sejersen, & Kreicbergs, 2016a) and not (Murrell et al., 2017). Parents have reported feelings of incredulity and shock at the time of diagnosis, followed by anticipatory grief from the time of diagnosis and continued grief throughout their children's lives and after their deaths (Higgs, McClaren, Sahhar, Ryan, & Forbes, 2016; Rallison & Raffin-Bouchal, 2013). Parents have also described emotional ambiguities – feelings of both joy and love and grief and sorrow – and a hope for their child to live as normally as possible, with as high quality of life as possible (Rallison & Raffin-Bouchal, 2013).

Depending on their values, some families choose palliative care with no invasive treatments, while others choose to make use of all measures available (Murrell et al., 2017; Qian et al., 2015). Parents of children with SMA type 1 have described a shift from initial sadness, frustration, and wanting to "fix" their child, to trying to shape the child's life and death in a positive way (Higgs et al., 2016). Parents of school-aged children with SMA have reported that they want their child to go to school and live like other children, even if this is sometimes hard (Yang, Mu, & Wang, 2016).

Yang et al. (2016) describe in their study, which was conducted before nusinersen was available, how parents of children with SMA were forced to change from expecting their children to grow up with a bright future, to facing the dilemma of SMA, where the experience of growing up meant an early death. The realisation that the children would never experience youth and middle age caused long-term anticipatory grief. In seemingly harmonious families, family members endured stress, strong negative emotions, and conflicts. The anticipatory grief and

knowledge of the coming death could also strengthen relationships within the family and lead to re-evaluation of meaning in life.

Unsurprisingly, research has shown that parents of children with SMA report more stress than parents of healthy children (Von Gontard, Rudnik-Schöneborn, & Zerres, 2012). However, the stress measured does not seem to be dependent on the degree of the child's physical dysfunction (Mah, Thannhauser, Kolski, & Dewey, 2008)

Compared with the context of, for instance, childhood cancer, research into experiences of the care of children with neuromuscular diseases is scarce. While practice standards for the multidisciplinary care of children with SMA have been published (Finkel et al., 2018; Mercuri et al., 2018; Wang et al., 2007), the standards of care change rapidly with new treatments. This, and the fact that SMA is rare, means that many children with SMA will receive care from providers who have little knowledge or experience of the disease. Care has been described by parents as sometimes being of poor quality, for instance when health care professionals lacked knowledge or competence about the disease (Lawton et al., 2015; Mah, Thannhauser, McNeil, et al., 2008; Murrell et al., 2017; Qian et al., 2015; Rallison & Raffin-Bouchal, 2013; Yang et al., 2016), or when care was poorly coordinated (Parker, Maddocks, & Stern, 1999). Other parents perceived problems with care, such as health care professionals behaving in an unhelpful manner (Murrell et al., 2017; Qian et al., 2015) and not giving parents enough information about the diagnosis or presenting the respiratory support options available (Beernaert et al., 2019; Lövgren et al., 2016a). However, parents have also reported positive aspects of the child's care, exemplified as experiences of receiving good support from health care professionals (Higgs et al., 2016), having their wishes concerning location of death fulfilled (Lövgren, Sejersen, & Kreicbergs, 2016b), and being involved in decisions concerning the child's treatment (Beernaert et al., 2019; Lövgren et al., 2016a).

1.3.2.1. Quality of care from a parental perspective

This thesis focuses mainly on the parents' experiences of care. To gain a deeper understanding of how care can be experienced and possibly improved, quality of care is an important concept to take into account.

Children with SMA need good, high-quality care from various highly specialised settings. Parents are vital for their children, and need to be supported so that they can in turn support their child and the rest of the family. Parents' views of their child's care are therefore of importance in offering good care to a child. There are various ways to assess quality of care and various definitions of quality of care, depending on the context and the level at which it is given, whether individual, organisational, or social. This breadth of the concept calls for different perspectives of the quality of care and thus also different ways of measuring and managing it (Blumenthal, 1996). The patient perspective of quality of care is an important aspect (Brown, 2007) and interest in this perspective may be expected to grow as education levels and access to information increase (Wilde-Larsson & Larsson, 2017).

Below, I intend to elucidate two models of quality of care and patient satisfaction (Larsson & Wilde-Larsson, 2010; Wilde, Larsson, Larsson, & Starrin, 1994; Wilde, Starrin, Larsson, & Larsson, 1993) without directly applying the models to my work, as that would be a post-construction. However, the models have inspired me and increased my understanding of the perceptions of quality of care from a parental perspective. The models and the thesis share some fundamental suppositions, such as that there is no objective truth about the degree of quality of care; the assessment is made by each individual observer. Furthermore, the observer – in this case the parents of children with SMA – brings person-related characteristics, such as sociodemographic aspects, health conditions, and personality, into the assessment of the care situation.

Based on Donabedian's model of quality of care from 1966 (Donabedian, 2005), Wilde et al. developed a theoretical model of quality of care seen from a patient perspective (Wilde et al., 1994; Wilde et al., 1993), which has also been applied to family members (Henoch, Lövgren, Wilde-Larsson, & Tishelman, 2012). The model describes the quality of care divided into four dimensions, which together make up the perception as a whole.

Larsson and Wilde-Larsson's model of patient satisfaction (Larsson & Wilde-Larsson, 2010) provides a theoretical understanding of aspects that affect patients' degree of satisfaction with received care. The model illustrates the interaction between person-related characteristics, including the person's expectations and

commitment to care, and external objective conditions, and how these together shape the cognitive interpretation of the quality of care. This cognitive interpretation, in turn, shapes the emotional response to the care situation, the result of which is the person's satisfaction with care.

1.3.3. Siblings' perspective of living with a brother or sister with SMA or other neuromuscular disease

In 2015, a literature review was conducted of existing research concerning siblings' perspectives within the familial experiences of childhood chronic disease (Knecht, Hellmers, & Metzing, 2015). The study found only nine articles, of which many applied a parental proxy perspective, a result that in itself indicates both that the research area is small and that there is a top-down view of siblings' experiences among parents, professionals, and researchers.

Excepting one study that examined behavioural problems in children with SMA and their siblings (Laufersweiler-Plass et al., 2003), no study was found that addressed the experiences of living with SMA from the perspective of a healthy sibling. The study by Laufersweiler-Plass et al. (2003) showed that the non-affected siblings had documented behavioural problems, in contrast to children with SMA. Similarly, Read, Kinali, Muntoni, and Garralda (2010) found an increased risk of emotional problems in non-affected siblings of young persons with Duchenne muscular dystrophy, another neuromuscular disorder.

Qualitative studies of siblings of young people with Duchenne muscular dystrophy (Read, Kinali, Muntoni, Weaver, & Garralda, 2011) and children with home mechanical ventilation (commonly used in neuromuscular diseases) (Israelsson-Skogsberg, Markstrom, Laakso, Heden, & Lindahl, 2019) showed that siblings are often involved in caring activities (Israelsson-Skogsberg et al., 2019; Read et al., 2011), that siblings perceived reduced parental attention (Israelsson-Skogsberg et al., 2019; Read et al., 2011), but also that the disease had positive effects on the non-affected siblings, such as increased family cohesion (Read et al., 2011) and increased personal maturity and empathy (Israelsson-Skogsberg et al., 2019; Read et al., 2011).

1.3.4. Resilience and the ability to feel hope when a child has a severe disease

Confronting severe disease and the loss of a loved child or brother/sister raises existential questions. Families who lose, or face the risk of losing, a child must deal with a deviation from what is perceived to be the natural course of life. This can lead to an anticipatory grief that arises when the child is diagnosed and continues throughout the child's entire life.

All families have resources to face adversities; some can find and use their resources easily, while others might need more support to find the ability to deal with a disease. The concept of resilience has been introduced to explain the ability to manage and cope effectively with adversity. Resilience is described as the ability to bounce back or cope successfully despite substantial adversity. In applying resilience to a family system, the concept of family resilience refers to the family's capability to regain its psychological and functional integrity after adversity (Oh & Chang, 2014; Walsh, 2016). Protective factors to family resilience can be described as a collective confidence in the family, interconnectedness within the family, a positive life view, a capability to identify and use support, open communication patterns, and collaborative problem-solving (Oh & Chang, 2014). The family's core beliefs have an impact on the optimism and hope that the individual family members experience.

There is a relationship between the concepts of hope and resilience. However, there is no clear consensus in literature on their interrelationship. It is difficult to determine which of the two concepts that contributes to the other, or if they both contribute, one to the other. Several studies show that hope is a protective factor that contributes to resilience (Earvolino- Ramirez, 2007; Gillespie, Chaboyer, & Wallis, 2007; Griggs & Walker, 2016), while others emphasise that individuals who have resilient characteristics have the ability to feel hope (Snyder, 2000). It is therefore hard to assess if a family's resilience enables family members to feel hope and have a positive outlook on the future, or if the ability to feel hope contributes to resilience.

It might be easier to understand the definition of hope by considering the consequences of absence of hope. Hopelessness is a state that causes despair, depression, and ultimately a loss of the will to live. Hope, by contrast, gives us the

courage to confront our circumstances and the capacity to surmount them. In all events, every day, hope is essential for us to keep living. When someone has a disease, hope is important in every treatment, procedure, and care plan. By feeling hope (or not), decisions can be made, and treatment programmes endured, which can have effects on the outcome of illness (Groopman, 2004).

The literature presents reviews and studies on hope related to severe diseases with varied samples of age groups, diagnosis, and different family members' perspectives. Leite, Garcia-Vivar, Neris, Alvarenga, and Nascimento (2019) describe hope within families with children living with chronic disease using a concept they call "family hope". The families used support to balance their hope, and children were described as depending on the people around them to sustain hope.

When a child is diagnosed with a severe disease, hope can arise almost immediately through the family starting to strive to survive – this can be a survival of the child with the disease, but also a survival of the entire family (Björk, Wiebe, & Hallström, 2005). Hope can remain in parents throughout the palliative phase and can take the form of hope for a cure, hope for a meaningful time with their child, and hope for a pain-free death (van der Geest et al., 2015). Hope for a cure may be one of many types of hope that support quality of life within severe diseases, and current evidence suggests it is necessary to be honest about prognosis, but that there is no need to discourage parents from hoping for a cure despite poor diagnosis (Robinson, 2012; van der Geest et al., 2015).

Parents of children with neuromuscular disease have in previous studies been described as moving between feelings of hope, avoidance, and living in the present, with the everyday aspects of life (Erby et al., 2006). Hope has also been examined by Samson et al. (2009), who, with a phenomenological approach, analysed interviews with parents of children with Duchenne muscular dystrophy. The study showed that parents' hope can be expressed in different ways with different benefits; from helping parents to absorb the initial crisis, to facilitating their adaptation or preparing them for the fatal outcome.

Within paediatric care, approaches of hope can differ between parents and professionals. While parents speak about their role as bearers of hope and protectors of their children, professionals describe a tension in maintaining hope

in the face of prognostic data. Another challenge identified for health care professionals can be parents' decisions to continue treatment even when it prolongs the child's suffering (Reder & Serwint, 2009).

Literature regarding children's experience of hope when living with a disease is scarce. However, hope has been suggested to play a unique role in the paediatric cancer context by reducing the levels of anxiety and improving youths' health-related quality of life (Lewis & Kliewer, 1996; Martins et al., 2018). A review of adolescents with chronic disease suggests that hope promotes health and is an important factor for resilience (Griggs & Walker, 2016).

Although studies involving families with severe disease emphasise the importance of hope, there is much more to explore, given the complexity of hope. SMA has recently changed as a result of new therapies; from being a progressive disease with inevitable early death to being a disease that might be united with long life. This change most likely affects families' ways to feel and express hope and makes SMA particularly interesting to focus on in relation to hope.

2. Rationale

Infantile SMA is a severe disease that has an impact on the whole family. Even though the disease is severe and families face a challenging trajectory with progressive losses, anticipatory grief, and ethical dilemmas regarding treatment, knowledge of how families experience their child's care and everyday life is limited.

The introduction of new therapies in the SMA area is rapid, and with new therapies that prolong survival, SMA has changed from being a life-threatening disease to one that may be united with long life. However, although these new therapies are effective in inhibiting disease progression and for many children result in functional improvement, some children do not improve as much as desired. Furthermore, the new therapies are not available in all countries or to all patients.

Learning from parents' experiences of care and everyday life with SMA can supply knowledge about what other parents in similar situations and health care professionals should focus on and what is important to do when caring for a child with SMA. This is particularly relevant now, when more children are expected to live longer as a result of the new therapies.

In everyday life with a disease, hope is essential for finding and maintaining the energy to live with and handle the challenges related to the disease. SMA is particularly interesting to focus on in the context of hope, since the landscape of SMA has changed in recent years. Knowledge about how families negotiate hope in everyday life during the new treatment with nusinersen is valuable in order to support families to gain resilience.

3. Aims

3.1. Overall aim

The overall aim of this thesis was to explore how families experience care and everyday life with SMA.

3.2. Specific aims

- To explore parents' reports of: 1) if health care has taken every opportunity to help the child feel as good as possible, 2) satisfaction with the various care settings, and 3) coordination of care between settings (Paper I).
- To explore parents' advice to health care professionals working with children who have SMA and their families (Paper II) and to other parents with a child with SMA (Paper III).
- To explore negotiations of hope in everyday life for families where a child with SMA received a new type of drug treatment (Paper IV).

4. Material and methods

4.1. Study design

To explore the specific aims, a variety of methods have been used. The thesis is divided into two projects: a survey study and an ethnographical study.

Table 1. Overview of the studies.

Paper	Study design	Participants	Data collection	Data analyses
I	A nationwide 11-year follow-up survey in Sweden and Denmark	Bereaved and non-bereaved parents of children with severe SMA born in Sweden between 2000 and 2010 and in Denmark between 2003 and 2013 (n = 95)	Questionnaires	Descriptive statistics, Pearson's chi-squared test, and content analysis
II	A nationwide 11-year follow-up survey in Sweden	Bereaved and non-bereaved parents of children with severe SMA born in Sweden between 2000 and 2010 (n = 51)	Questionnaires	Content analysis, descriptive statistics, and Fisher's exact test
III	A nationwide 11-year follow-up survey in Sweden and Denmark	Bereaved and non-bereaved parents of children with severe SMA born in Sweden between 2000 and 2010 and in Denmark between 2003 and 2013 (n = 81)	Questionnaires	Content analysis and Pearson's chi-squared test
IV	An ethnographical study	Two families where a child had SMA type 1 or 2, involving a total of 10 family members	Narrative interviews (n = 17) and participant observations at six occasions	Narrative analysis

4.2. The survey study (Papers I, II, and III)

The survey study is based on nationwide surveys conducted in Sweden (Lövgren et al., 2016a, 2016b) and Denmark (Beernaert et al., 2019). The surveys had an exploratory cross-sectional design and focused on parental experiences of the care of a child with severe SMA. The study was initiated in Sweden and was also conducted in Denmark, three years later. Both parents living with a child with severe SMA (here called non-bereaved) and parents who had lost a child with severe SMA (here called bereaved) were included.

4.2.1. Participants

In Sweden, all parents (legally registered as guardian) who had a child born between 2000 and 2010 and diagnosed with severe SMA were eligible for the study. Severe SMA was in this study defined as SMA type 1 or SMA type 2, if respiratory support was considered for the child during the first year of life. Children with SMA were identified by their personal identity numbers through The National Board of Health and Welfare, and their guardians' names and addresses were obtained through the Swedish Tax Agency.

Initially, 78 parents of 40 children were identified in Sweden. Parents with an identifiable phone number were invited to participate in the survey. In the Swedish sample, 61 parents of 39 children (32 deceased and seven living) participated. Of all 40 children initially identified, all but one had at least one parent represented in the Swedish survey (Figure 1). The response rate for the Swedish survey was 87%.

The Danish sample consisted of parents of children born and diagnosed with SMA type 1 or 2 between the years 2003 and 2013, for whom respiratory support was considered in the first year of life. All children were registered in National Patient Register and/or Cause of Death Register and/or at RCFM. Guardians' names and addresses were obtained through the Civil Registration System. In Denmark, 47 parents of 24 children were identified and those with an identifiable phone number were invited to participate. Thirty-four parents of 21 children (13 deceased, 8 living) chose to participate, which gave a response rate of 79%. All of the children initially identified in Denmark except three had at least one parent represented in the Danish survey (Figure 1).

In total, 95 Swedish and Danish parents (71 bereaved and 24 non-bereaved) of 60 children participated. The total response rate for the survey study was 84%.

The parents comprised almost equal numbers of mothers and fathers, with slightly more women. The mean age for the total sample was 39 years (SD 5.5). Half of the parents lived in the countryside or in a smaller town, whereas a third lived in a town with 100,000 inhabitants or more, and the rest lived in a medium-sized town. Half of the parents had a university education.

A majority of the children who were deceased had been diagnosed with SMA type 1 and had died at a mean age of 12 months (SD 19.2). The mean age of the children living with SMA was 5.7 years in Denmark and 8.7 years in Sweden.

4.2.2. The study-specific questionnaires

The questions in the questionnaires were developed specifically for this study in accordance with a method developed by Charlton (2000). The method has previously been used in construction of questionnaires for parents who have lost a child to cancer (Kreicbergs, Valdimarsdóttir, Steineck, & Henter, 2004) or suicide (Omérov, Titelman, & Nyberg, 2018), siblings who have lost a brother or sister to cancer (Lövgren, Jalmsell, Eilegard Wallin, Steineck, & Kreicbergs, 2016) and teenagers who have lost a parent to cancer (Bylund-Grenklo et al., 2014).

Two questionnaires were developed, one for bereaved parents and one for non-bereaved parents. As a first step in the development of the questionnaires, as suggested by Charlton (2000), interviews were conducted with seven non-bereaved and four bereaved parents of children with SMA, all in Sweden. The interviews focused on the experience of health care during the child's disease. Based on the interviews, themes were constructed and developed into questions, in close collaboration with staff from a rehabilitation centre and parent representatives.

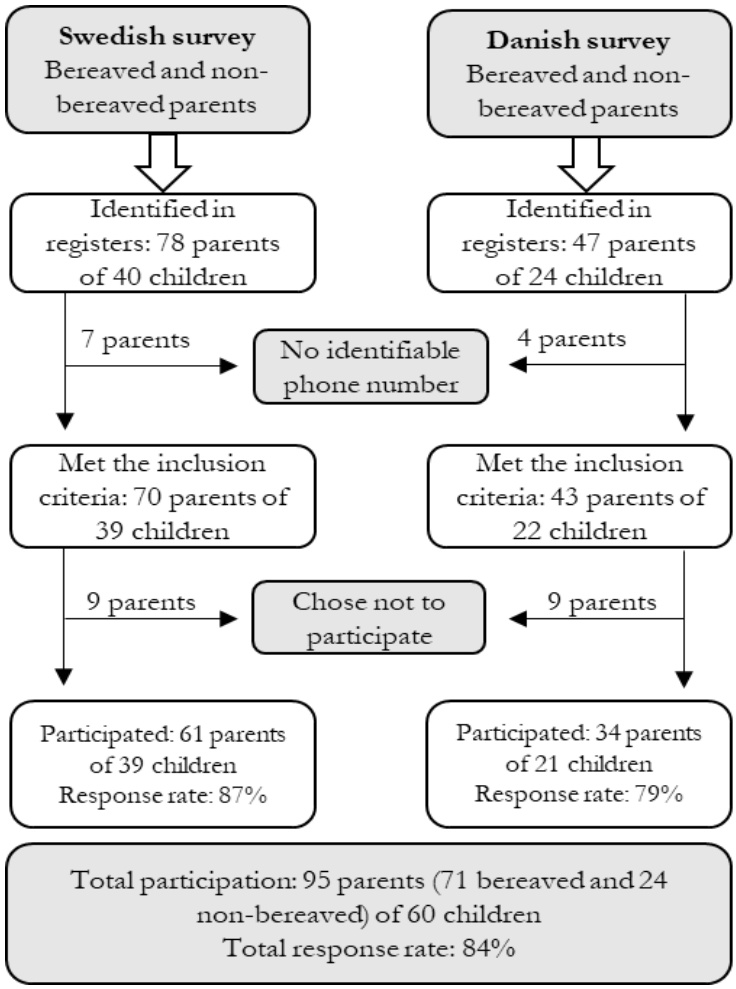

Figure 1. Selection process and participation rate.

The developed questionnaires were then face-validated with five Swedish parents of children with SMA born earlier than 2000 or later than 2010, to ensure that the questions were understood as intended. Minor changes were made after that.

The questions in the two questionnaires were largely the same, but with additional questions to bereaved parents regarding the loss of their child.

The questionnaires consisted of 59 questions for non-bereaved parents and 75 questions for bereaved parents, and included both closed- and open-ended questions.

The first part of the questionnaire focused on socio-demographics, disease-related information given to the parents, medical decision-making, and care and support received by the family. The second part involved parents' perceptions of the quality of care at the hospital and at the municipal rehabilitation centre. Questions about parents' advice to health care providers and other parents in similar situations were also included. The Swedish questionnaires were translated into Danish and cultural adaptions were made, e.g., the list of health agencies was localised.

The questions were used in the following way in the papers:

Paper 1 used questions concerning parents' experiences of coordination of care between settings and if the parents had experienced that the health care staff had taken every opportunity to help the child feel as good as possible. In connection to these two questions, the parents were given the opportunity to write free-text comments, which were included in the analysis. In addition, the study used parental reports on the care setting for their child and satisfaction with the different care settings on a scale from 1 to 5 (where 1 represented "completely dissatisfied" and 5 "completely satisfied").

Paper II was based on the parents' written responses to one open-ended question: "What advice would you give to staff working with children who have spinal muscular atrophy and their families?"

Paper III used responses to one open-ended question: "What advice would you give to other parents of children with spinal muscular atrophy?"

4.2.3. Data collection

Eligible parents were sent a letter of invitation, with mothers and fathers contacted separately. About a week later, a research assistant contacted each person by telephone and asked if they wanted to participate. If they consented, a questionnaire was sent by post to each consenting parent (February 2013 in

Sweden and April/May 2015 in Denmark). About one month after the questionnaire had been sent out, a combined thank you and reminder card was sent to the parents who had agreed to participate. If a parent did not return the questionnaire, those living in Sweden were phoned and asked if they had any particular concerns or needed assistance with completing the questionnaire.

4.3. The ethnographical study (Paper IV)

The overall purpose of this study was to explore how children with severe SMA and their family members experienced life with SMA, what they perceived as difficult in daily life, what facilitated daily life, and what kind of support they wanted from health care and society. To cover these research questions, an ethnographical approach with a focus on narrative interviews and participant observations was considered suitable. During the data collection and analysis phase, the families' narratives about hope in everyday life as a result of the new treatment of SMA stood out as significant in the data, and Paper IV therefore came to focus on that.

4.3.1. Participants

Family members from non-bereaved families in the survey study in Sweden were invited to participate in this ethnographical study. In addition, recruitment took place via the Facebook page of the Swedish patient association for SMA. To participate, a family had to include at least one child diagnosed with SMA type 1 or 2 who was between six and 19 years old.

4.3.2. Data collection

An invitation letter was sent out by post (April/May 2018) to parents who had participated in the Swedish survey, whose child was still alive and who had an identifiable home address (n = 5). The letter informed them about this ethnographical follow-up study and stated that all family members were invited to participate. Since children with SMA differ in their ability to communicate depending on how far the disease has progressed, I wanted the design of the study to be adapted to each family's wishes and each child's abilities. The text in the information letter therefore suggested interviews in the home and that I would spend time with them in everyday life, but that if the participants preferred, they could convey their experiences in other ways. Repeated interviews and participant

observations were preferred, but not mandatory. The families were also informed that they decided who they considered to be a family member and thus who would participate in the study. Approximately one week after the invitation letter was send, I called the parents to ask if their family wanted to participate in the study. This recruitment resulted in that three families consent to participate. After the recruitment of families from the survey, the recruitment of families via Facebook started, which resulted in that one family consented to participate. Of the four families that agreed to participate, one family found it difficult to meet and the mother therefore answered written questions which were sent to her via Webropol 3.0, a web-based platform. Three families where met for interviews and participant observations in everyday life.

Two of the four families were selected for Paper IV in this thesis as they consented to meet me several times: Alexander's and Isaac's families. Data from the other two families are planned to be used in future studies.

Alexander's family consisted of Alexander, who had SMA type 1, his mother, father, older sibling, and younger sibling. Isaac's family consisted of Isaac, who had SMA type 2, his mother, father, sibling, and one extended family member. To protect the families from identification, detailed characteristics of the participants are left out and pseudonyms are used. In total, 17 interviews and six occasions of participant observations collected from Alexander's and Isaac's families were included in Paper IV (Figure 2). The data for Paper IV were generated over a period of 9 months. All data in the ethnographical study were collected by me and shared with all co-authors during the entire data collection process. The co-authors provided suggestions on further ways to deepen the interviews and observations.

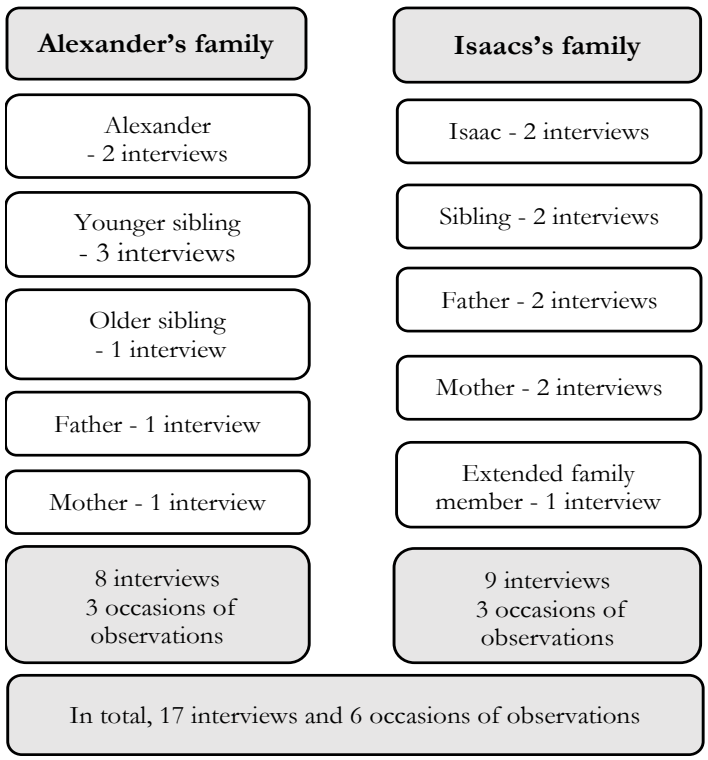

Figure 2. Number of interviews and observations included.

4.3.2.1. Participant observations

As part of an ethnographical design, repeated visits were made to the families, to gain a richer understanding of each family's experiences of living with the disease (Atkinson & Hammersley, 2007). Ethnography is a research design that can be used with benefit when exploring the nature of a particular social phenomenon, as in my case, which related to everyday life with a disease (Reeves, Kuper, & Hodges, 2008). The observations were conducted with a participant approach and data consisted of field notes. To prepare for my observations, I made pilot observations with families I didn't know, to practice the technique. I also made a home visit to a family in Denmark where a child had severe SMA. I thus had the opportunity to practice interview technique, conducting observations, and taking field notes. Participant observations can be performed in many ways. In this case, the observations were performed with the approach of a guest in the family's

homes. I participated in everyday events, e.g. sitting together in a sofa while the kids were playing or socialising during meals. During the observations, informal questions were asked about everyday events. The observations were conducted in connection with the interviews, mainly at the families' homes, though some data collection took place during leisure activities such as a football game and at one of the children's school. The encounters lasted from a couple of hours to half a day.

By taking field notes, it is possible to transform an event that takes place in the moment to an interpretation on paper that one can return to and interpret over again (Fangen, 2005). I strove to create rich field notes, including a variety of elements, e.g., the physical environment, the persons present, their activities, things that were said, and so forth (Reeves et al., 2008). My field notes switched between a) what was observed or said between the participants and directly to me in informal conversations, and b) my own reflections (Fangen, 2005). To be careful regarding my own pre-understandings, I structured the field notes in two columns, one for things that I saw or heard, and one for my own reflections. I had a notebook with me and took notes from time to time. I wanted to get as rich data as possible from my observations, but I didn't want the family to feel scrutinised or judged. I therefore explained that I took notes only to remember as much as possible of what was happening and what we were talking about, and that I didn't write anything secret or in judgment of them. Sometimes, I stepped aside (e.g., went to the toilet) to write more extensive notes. It is important to transcribe field notes as soon as possible, otherwise, it is easy to forget; quotes are especially important to write down immediately (Fangen, 2005). Therefore, in direct connection to each observation, I digitally recorded the field notes as a voice memo and transcribed them within a few days.

In order to increase my openness to the participants' stories, I prepared carefully before the meetings by making time for mental preparation. Before going to visit a family, I sat for a while to release any stress and become more present in the moment. After each interview, I followed a comparable process, reflecting on the meeting with the family, reading through the field notes, and recording more field notes on the voice recorder.

4.3.2.2. Narrative interviews

Human experiences are not easily captured, as they cannot be picked up and held or measured. One way for human beings to make sense of the world and understand lived experiences is to express them and interpret them in narratives (Greatrex-White, 2010; Polkinghorne, 1988). Narrative interviews with open questions was used in the study to allow for storied responses and promoting sharing of experiences from the perspective of the informant (Anderson & Kirkpatrick, 2016; Garro & Mattingly, 2000). In this study, interviews began with an open statement: "Tell me about yourself." Follow-up questions that were asked during the interviews focused on everyday life with the disease, experiences of support from health care and society, and expectations of treatment. No specific questions were pre-formulated, but questions were based on prepared question areas. These were inspired by reading relevant literature, by other studies' question guides, by parents' written comments in the survey study, and by conversations with parents of children with SMA, including a meeting with the Swedish patient association for SMA, during which I asked parents about things they thought were important. During the interviews, I had a mind map with the different question areas, which I looked at if I needed inspiration to come up with a new question. The ethnographical study strove to invite the children, the siblings, and the parents to tell their own stories about life and the things that mattered to them (Garro & Mattingly, 2000). The interviews were therefore mainly conducted individually, except the interviews with Alexander and Isaac as they wanted/needed an adult nearby. The siblings were interviewed individually, but in some cases they chose to sit in a room with open door. All interviews were digitally recorded and transcribed verbatim.

4.3.2.3. Interviewing children

Interviewing children is a difficult art that requires a lot of training. Children are often compliant in their communication. They often want to do what is "right" and can therefore try to understand what the adult is looking for and answer what they think the adult wants to hear. Many children are used to adults asking them questions even if the adults themselves know the answer. Through suggestive techniques and questions, children can easily be led to say things that have not occurred. Preschool children seem to be the most vulnerable to misleading information (Nicol, La Rooy, & Lamb, 2017). I was therefore especially careful

when interviewing children, making sure to ask open and inviting questions to seek the child's own perspective, without, either consciously or unconsciously, influencing the child to talk about something that may not have happened (Toeplitz-Winiewska, 2007). I also strove to make each child feel as confident as possible by being kind, calm, honest, and clear with the purpose of our conversation (National Board of Health and Welfare, 2018). Before each interview with a child, I informed the child of some basic "rules" for our interview (Nicol et al., 2017). For instance, I said that there were no rights or wrongs, that I was curious about everything the child said, and that the child should correct me if I misunderstood something. To conduct a credible interview with a child, it is also necessary to have an understanding of the child's ability to relate a story and to understand questions, based on its development (Toeplitz-Winiewska, 2007). A child's level of development is not directly associated with the child's chronological age, and can differ from child to child. I tried to adapt my language to the child I had in front of me. On some occasions, I caught myself saying an unnecessarily complicated word, and corrected myself to use a simpler word instead. To practice the technique of interviewing children, I performed some interviews about everyday life with children in my neighbourhood before I performed the interviews for the study.

4.4. Data analyses

4.4.1. Statistical analysis (Papers I–III)

Descriptive statistics were used to examine the closed-ended questions and differences between groups in Papers I-III. Pearson's chi-squared test and Fisher's exact test were used to compare expected frequencies against observed frequencies in the population (Djurfeldt, Larsson, & Stjärnhagen, 2003). Pearson's chi-squared test was used in Paper I to examine differences between Sweden/Denmark and bereaved/non-bereaved parents regarding satisfaction with care, coordination between different care settings, and whether health care staff had taken all possibilities to help the child feel as good as possible. A p-value of less than 0.05 would indicate that there was a statistically significant difference between the two groups, while a p-value above 0.05 would indicate that the differences between the groups were due to natural variation (Almquist, Ashir, & Brännström, 2014). To examine differences in smaller samples, as between the

types of advice given by bereaved versus non-bereaved parents in Paper II, the comparable Fisher's exact test was used (Djurfeldt et al., 2003).

4.4.2. Content analysis (Papers I–III)

Content analysis is a method that can be applied in various ways, from quantitative to interpretive qualitative inspired by hermeneutics, where the nature of data determines the level of interpretation (Krippendorff, 2004). As the written answers from the open-ended questions in the survey were relatively short, from single words to several sentences, content analysis that kept close to the text was considered suitable for Papers I, II, and III. Unlike in the phenomenological philosophy, Krippendorff (2004) argues that a text does not have an inherent meaning or objective qualities. Instead, Krippendorff argues that meaning in a text is always brought by someone, i.e., that the text can be read and interpreted in many different ways.

The parents' free-text comments from Danish parents was translated from Danish into Swedish by me and reviewed by one of my co-supervisors, who is bilingual. All comments were read through to get an overview and thereafter coded with a label. Comments that did not correspond to the aim were excluded. Content analysis is a method that to a large extent focuses on finding similarities in the text by coding and systematising the text (Krippendorff, 2004). Depending on the content, the data were therefore sorted into categories based on similarities. No data relating to the purpose was excluded for lack of a suitable category (Krippendorff, 2004). Since human experiences are not always single-track, it is not always easy to create mutually exclusive categories. This means that interpretation was necessary, despite efforts in Papers I, II, and III to stay as close to the text as possible. To validate the analyses, all steps in the process were critically reviewed by all authors. Adjustments and changes were made until consensus was reached. In Papers I and II, the content analysis was presented using numbers connected to the frequencies of statements. This way of quantitizing qualitative data enables visualisation of patterns in the data that otherwise might not be clear (Sandelowski, Voils, & Knafl, 2009). Further, this way to present the results increase transparency and enables for the reader to understand and judge the credibility.

4.4.3. Narrative analysis (Paper IV)

In Paper IV, narrative analysis was used on interviews and field notes, with a focus on significant events and turning points within the families' narratives (Josephsson & Alsaker, 2015; Mattingly, 1998). One core issue in narrative analysis is reconstructing otherwise isolated experiences into a coherent whole, while being cautious about the wholeness of the narrative, and not fragmenting stories and experiences into parts or categories (Creswell & Poth, 2017). The analysis was done in line with hermeneutic theory, where interpretations took shape by switching focus back and forth between the specific parts and the whole (Josephsson & Alsaker, 2015; Lindseth & Norberg, 2004; Mattingly, 1998). This way of alternating focus is relevant, as human actions cannot be understood as independent, but must be placed in the context of the whole, meaning that a specific part or quotation from an interview must be seen in the context of the wholeness of the interview. The analysis began with transcribing the first interviews, which enabled follow-up questions during subsequent visits/interviews. In total, approximately 221 Microsoft Word pages were processed, of which 177 pages consisted of interviews and 44 pages consisted of field notes. To get an overview of the data, the contents were sorted in overall topics in the software program NVivo 11. During this process, no distinction was made as to whether data came from field notes or interviews, or which family member's perspective that was being processed. Examples of topics were "about the disease", "about leisure time", "about therapy", and so on. In next phase, significant events that "stood out" in the data were observed and discussed within the research group. In this process, the narratives about hope and treatment of SMA were identified as significant. An aim was formulated for Paper IV, and data corresponding to the aim were sorted out and re-constructed into narrative vignettes (Creswell & Poth, 2017; Josephsson & Alsaker, 2015). The vignettes were processed, shortened with a focus on the purpose of the study, and translated from Swedish into English. In parallel with this process, literature on the topic was read, to facilitate understanding and interpretation of the text. The analysis then went into a more interpretive phase. Existing theories were brought into the interpretive process to apply hermeneutic interpretation to the text. The focus was on finding theories that challenged the narratives, or on finding significant events in the narratives that challenged existing theories. In line with the hermeneutic circle, the interpretation process moved between reading the text

as a whole, finding possible significant events, interpreting existing literature, and reconstructing the narrative.

The results were presented as two stories, where empirical data and theoretical perspectives were interwoven. In the vignettes, the perspectives of the various family members were carefully considered and taken in account, even if the result was presented from a family perspective. All co-authors were active during the analysis process, reading the collected data on several occasions, reflecting on possible interpretations, and critically reviewing the text that was produced.

4.5. Ethical considerations

The ethical imperative in this thesis was to avoid causing harm to those who participated. Several aspects of the research were designed to ensure that this goal was achieved. The ethical principles of *benefit* (including the principle of non-maleficence and the principle of beneficence, which refers to actions that promote the well-being of a person), *justice* (including that researchers are to respectfully listen to the person's views) and *respect* (including recognition of the person's dignity and obtaining informed consent for research) have been followed (Beauchamp & Childress, 2013; Graham, Powell, Anderson, Fitzgerald, & Taylor, 2013). Both the survey study and the ethnographical study were conducted in accordance with the Helsinki Declaration (World Medical Association, 2013). An ethical aspect that has been important to consider in this thesis is the sensitive subjects of children living with severe disease and the loss of a child.

4.5.1. Risks, burdens, and benefits for vulnerable groups and individuals

Bereaved persons are vulnerable and special considerations must be taken in research involving such persons, like carefully considering the wordings used and when is a suitable time for inviting bereaved parents. There is debate about if there is an appropriate time for inviting bereaved persons to participate in research after a loss, and – if so – when that time is. There is growing evidence that participation in research is not necessarily distressing for bereaved family members, as it can also be experienced as something positive (Eilegård, Steineck, Nyberg, & Kreicbergs, 2013; Higgs et al., 2016; Hynson, Aroni, Bauld, & Sawyer, 2006; Kreicbergs, Valdimarsdóttir, Steineck, et al., 2004; Scott, Boyle, Bain, &

Valery, 2002; R. Steele et al., 2014; Udo et al., 2019) not just for altruistic reasons, but also as a personally therapeutic experience. A common assumption is that bereaved people need time to grieve before being approached about participation in research. However, results from a study by Bentley and O'Connor (2015) indicate that participation in research is not necessarily distressing for recently bereaved family members. In the survey study in this thesis, at least six months had passed since the parents had lost their child when the invitation for study participation was sent out. The high response rate (84%) in the survey study may reflect the parents' willingness to participate in research. In their written responses in the questionnaire, many parents expressed their appreciation at being acknowledged and asked about the care of their child with SMA.

Participant observation can infringe on people's privacy (Fangen, 2005). In the ethnographical study of this thesis, Paper IV, this occurred quite literally, as I became privy to people's private lives and got intimate information about their habits and routines. This involves heavy demands on the researcher to treat the information with care.

4.5.1.1. Children in research

Research involving children is generally rather scarce, in particular research involving children with SMA. Children belong to a vulnerable group and should therefore be protected from research that could harm them (The Swedish Research Council & Uppsala University), especially considering that it is difficult for a child to foresee the consequences of participating in a study, and that children are easily influenced by others. Conducting research with children, particularly children who have a disease, requires special ethical considerations to avoid further suffering (Wolfe & Siden, 2011). However, it can also be seen as unethical to prevent children from research participation and thereby silence their voice in questions that concern them. They would then miss out on the potential benefits of knowledge that can be generated through research (Johansson & Karlsson, 2013).

4.5.2. Confidentiality

All collected data have been handled carefully to protect confidentiality. All data containing personal data have been handled in coded form and were only available

within the research group. Data containing personal information are stored on a secure server and under password protection, in accordance with Ersta Sköndal Bräcke University College's policy. As SMA is rare and the eligible families few, the risk of identification of participants in Paper IV is high. When reporting results, it was therefore necessary to slightly modify the participants' characteristics or attributes in their environment. The participants were informed of this when they got information about the study.

4.5.3. Informed consent

Information was given to the participants about the study aims, what was required of them, that participation was voluntary, that they had the right to withdraw at any time and without explanation, and how the collected data would be used. In research with children, it is important to provide this information in an age-appropriate way (Greig, Taylor, & MacKay, 2012), which was done through age-adjusted written information for children. Written informed consent was collected from all participants, and for children younger than 15 years old, written informed consent was also obtained from both parents. In the ethnographical study, verbal consent was collected repeatedly, before each visit. In the ethnographical study, one sibling declined participation. At my visit to the family, the sibling greeted me and thereafter went downstairs, keeping away from me. No field notes involving that sibling were taken.

4.5.4. Ethical approval

Both the survey study and the ethnographical study fall under the Act concerning the Ethical Review of Research (2003:460), as sensitive data were collected. Ethical approvals have been granted for both studies. The Swedish part of the survey study has been approved by the Ethics Committee in Stockholm (reference number: 2009/1702-31/2). The Danish survey has approval from the Danish Data Protection Agency (ref.nr: 2013-41-2504) and Statens Serum Institut (FSEID-00001091) (according to Danish law, ethical approval was not needed for survey studies at the relevant timepoint).

The ethnographical study has received approval from the Regional Ethical Board in Stockholm, Sweden (ref.nr 2017/985-31/2).

5. Results

The results of the four papers are presented here, merged into a coherent whole. The findings are described under four headings: Experiences of the child's care, including advice for professionals; experiences of everyday life with SMA including advice to other parents in similar situations; meaning-making in bereavement; and struggles and negotiations of hope.

5.1. Experiences of the child's care and advice to professionals

5.1.1. Parental perception of care

The parents reported that their children had been cared for in several different care settings (Paper I). The parents gave the care settings an average score of 4.1 on a scale from 1 to 5, where 1 represented "completely dissatisfied" and 5 "completely satisfied". The care settings that the Danish parents were most satisfied with were the Danish rehabilitation centre RCFM, and the neonatal care unit, followed by the paediatric intensive care unit. The Swedish care settings that parents were most satisfied with were the long-term intensive care unit followed by the paediatric intensive care unit and neonatal care unit and home care. The three settings that were most frequently reported were also the three settings that the parents were least satisfied with: the emergency care unit, the municipal rehabilitation centre, and the general paediatric unit. Throughout, bereaved parents were more satisfied with care than non-bereaved parents (Paper I). Approximately two thirds of the parents (68%) reported that the health care professionals had taken every opportunity to help the child feel as well as possible, whereas the remaining third reported the opposite (Paper I). Examples of reasons why care was not considered to be good was a perceived discrepancy between how the parents thought care should be provided and the care that the child actually received.

> I would have liked them to give him tranquilizers on some occasions, especially toward the end. They didn't agree to do what we wished because it might affect his heart activity.
>
> *Bereaved parent – Sweden (Paper I)*

5.1.2. Competence among health care professionals

The parents suggested that health care professionals needed to increase their disease-specific knowledge. The primary reason was to avoid harm to the child (Paper II). The parents experienced that the lack of knowledge could result in not getting the medical devices that the child needed, or that it took a long time before resources were put to use. Some parents also felt that health care professionals did not always listen to, or believe, the parents. They therefore gave health professionals the advice to turn to them and trust them (Paper II).

Danish parents, who in general were pleased with the RCFM, gave specific advice to other parents to make use of this existing competence (Paper III). At the same time as parents advised professionals to improve their knowledge about the disease, they emphasised that if professionals used an affected child as an opportunity to improve their competence, this must be done with respect for the child and the family (Paper II).

5.1.3. Relationship between professionals and parents

The most frequent advice to health care professionals was related to the relationship between staff and family. The parents gave advice to health care professionals to be responsive and listening to the family's concerns. They wanted professionals to be empathetic, genuine, positive, humble, considerate, and convey a sense of security (Paper II). Furthermore, parents advised health care professionals to be brave: that they should find the fortitude to be present, to remain, to talk, to ask, and to sit quietly with the family.

At the same time as the parents emphasised the importance of having a good relationship with professionals, they gave advice to other parents that they would sometimes have to fight for the child's best interests, and, in some cases, would have to make demands and critically question the care (Paper III). Always prioritising what was best for the child, e.g., prioritising life quality over a long life span, was highlighted.

In the studies for Papers I–III, the parents underlined the importance of parental involvement in the care of their child. They gave advice to other parents to take an active part in care (Paper III), and to health care professionals to include them in treatment decisions, treatment plans, and planned procedures (Paper II).

Parents also advised professionals to provide information and guidance about what parents were allowed to do in regards to nursing and treatment of their child, e.g., airway suctioning, and to demonstrate to the parents how available medical aids should be used (Paper II). The parents in the thesis encouraged other parents to make their own decisions regarding their child's care and rely on the child's ability to guide treatment decisions (Paper III).

However, mixed messages were revealed regarding parental involvement in care. The parents perceived themselves to be "experts on their own child" and wanted health care professionals to see them as such, turn to them, listen to them, and trust them (Paper II). It was also reported as somewhat problematic that the child's care required proactive parental involvement and that the parents always had to take the initiative for treatment and disease management (Paper I).

> We had to be active ourselves to get hold of the breathing aids that we needed. For example, we heard about the cough assist device from other parents whose child had trouble breathing. We can't understand why the hospital didn't offer it until we requested it ourselves.
>
> *Non-bereaved parent – Denmark (Paper I)*

5.1.4. Parents' advice on organisation of care

The parents gave suggestions for better organisation of care, for instance, better continuity of care. Since children with SMA have complex care needs, many different care providers must be involved in care. The parents experienced that there were sometimes too many different care settings and health care providers involved in the care of their child, and that it was sometimes unclear who and which hospital/setting had the overall responsibility. They therefore gave advice to health care to be clear about who had the main responsibility, and suggested that one person should be available for direct contact (Papers I–II). In Papers I–II, the parents reported shortcomings in coordination between these different health care providers, for instance in communication between different hospitals and departments, and between the muscle specialist team and the rehabilitation clinic (Paper I). Parents gave advice to professionals that they should create a special care team to follow each child throughout the entire disease trajectory and to designate a coordinator for each family (Papers I–II). When a child needs to

be hospitalised, parents highlighted the importance of letting the child go directly to the hospital ward, without having to go through the emergency unit (Paper II).

The parents gave advice to establish opportunities for care at home, from diagnosis to end-of-life. However, some emphasised the importance of having access to hospital in end-of-life, as it felt reassuring to have that option (Paper II).

One general desire was that health care should not be stingy with resources, and an issue with inequities of care and access to medical aids depending on place of residence within the country was highlighted (Papers I–II).

5.2. Experiences of everyday life with SMA and advice to other parents in similar situations

The importance of finding a sustainable way to lead everyday life was highlighted in Papers II–IV. The parents gave advice to other parents to lead as normal lives as possible despite the child's severe disease. This could include trying to see the healthy aspects in the child and not focusing only on care and treatment. Parents encouraged other parents to be at home as much as possible, as this facilitated for the family to live normally. This applied also to situations when the child was very poorly and receiving special medical care at home (Paper III).

The parents emphasised the importance of getting rest and gave advice to other parents to ask for help with practical aspects of everyday life (Paper III). When the child was hospitalised, the parents highlighted that they were often exhausted, since they had already been struggling with the care at home. They therefore gave the advice to health care professionals to assist the families with finding practical support (Paper II).

The parents emphasised the importance of not to forget to help the child play (Papers II–III). One example of advice was that toys intended for cats could be suitable for a baby with muscle weakness (pet toys are often light and easy to grip). Other piece of advice was not to forget to take care of everyone in the family, including the child's healthy siblings (Paper III). Social support was highlighted as an important factor in managing adversities in everyday life with SMA. Parents stressed the importance of getting in contact with other families in similar

situations (Paper II–III) and sharing their innermost feelings with those closest to them (Paper III).

In the study for Paper IV, the children with SMA described their disease as something unproblematic. They could both see benefits with SMA, such as sometimes getting preferential treatment. However, there were less good things about having SMA – according to Isaac, that he could not do all the things his friends could, and according to Alexander, that he sometimes got very sick. Their siblings said that having a brother with SMA felt normal, but that they could sometimes get worried when their brother was ill or sad when he was not treated fairly (Paper IV).

5.3. Struggles and negotiations of hope

In Paper IV, the two families' narratives revealed how the families used different strategies of hope, when new hope was added into their lives through treatment with nusinersen.

The narratives identified in Paper IV, whether told or enacted, demonstrated how hope allowed families to rewrite their narratives of the future. When Alexander and Isaac started treatment with nusinersen, they gained new energy. The new energy, in turn, led to new possibilities and hope (Paper IV). After several years of struggling with severe disease and striving to keep their child alive, the families had come to a turning point with a promise of a future.

The hope in Alexander's family took on a different form for each family member. The hope of a good future was something normal and natural for the younger children. Alexander's mother described the new treatment as a relief for the family as a whole. At the same time, the new situation gave her room for reflection on her own life, which caused her to feel doubt about her own life situation.

> I walk around here at home and clean and do the laundry and cook and … I don't really do anything else. I don't meet many friends; I never get up to anything. I and (Alexander's father) don't have much time together. Uhm … I don't go to any Christmas parties, like everyone else. So, it's gotten a bit … I feel like I'm almost becoming bitter, and feel that I need to make an effort so that I get more of a social life.

Alexander's mother (Paper IV)

Alexander's father said that he, due to Alexander' diagnosis, had learned not to hope for too much, but instead appreciate the good moments in everyday life, without thinking about the future. This had also become a strategy in connection with hope regarding the new treatment that Alexander received. The father said that he did not want to live by the hope of a cure for his son, but instead wanted to hold on to and appreciate the benefits already achieved.

Alexander's older sibling sometimes felt worried when thinking of Alexander's future, but tried to avoid the frightening thoughts and instead focus on the hope that nusinersen could help Alexander live a long and healthy life.

> The only thing that has been, that I think about a bit, is if he'll get to be like a middle-aged person and die when he's, like, 80 years old, or if it'll be at 15 or something. That's basically the only thing. But it's hard to say. But right now, I hope, and it looks like he'll lead a normal life, just that he has a disease and can't move in the same way.

Alexander's older sibling (Paper IV)

Isaac was confident in his hope that he, thanks to treatment, would become strong and, in the future, independent of his wheelchair and medical equipment. Isaac's mother and sibling conveyed also a conviction about a positive outcome from treatment.

> There's a lot of research on this and that they've found different ways to do it. […] So, you absolutely have to believe that. At some point … they'll find something. And then the people making the decisions have to be quick.

Isaac's mother (Paper IV)

While Isaac's father was not sure about Isaac's possibility to walk, he did not want to take this hope from Isaac. He thought it *could* be possible, but added that both he and Isaac knew there was a long way to go, given the limited muscle strength that Isaac had. Isaac's close relative was more cautious in her hopes regarding the treatment. She did not dare be filled with hope, worried that having high expectations would lead to disappointment.

Isaac's sibling did not see the disease as a threat to Isaac's life, but could feel sorry for Isaac sometimes, when he was prevented from participating in activities due to his SMA. The sibling wished that everything could be adapted for everyone, including persons with disabilities.

5.4. Meaning-making in bereavement

The findings of the papers show that the parents shared their experiences from dealing with the existential issues of losing their child. Parents who had lost their child gave advice to other parents, who were now living with an ill child, regarding how to live and create memories together, to ease any later regrets. Examples included giving as much love and closeness to the child as possible. Another piece of advice was to create memories with the child, like taking photos of the whole family together and writing a diary. Some parents gave advice to other parents about how to cope with grief. This could involve grieving together with the other parent, using culture as a source of comfort, and daring to have more children (Paper III).

As life with a severe disease can be short, the parents underlined the value of making the best of everyday life with the child, and gave advice to other parents to try to find a way to accept the situation and enjoy the good moments (Paper III).

6. Discussion of the findings

Children with SMA have been largely invisible in research, as have their families. This thesis aimed to make new knowledge available about experiences of care and living with SMA and how families negotiate with hope when a child with SMA receives a new drug treatment.

When I started working on the studies in this thesis, I did not bring any specific theory with me into the work. The work with the papers has been inductive and explorative. However, in the text below, I have described the findings in light of various existing theories, such as the quality of care model, palliative care philosophy, and theories about resilience and hope. These are discussed below, together with previous studies in the field.

6.1. Quality of care

In this thesis, the focus has primarily been on the parents' perspective of care of the child, as the parents are crucial for the child to receive the best possible care. However, one must not forget to involve the child, and should – when possible, considering age and maturity – also turn to the child.

Medical care for children with SMA has advanced, but to achieve care with high quality, the care must also be based on the unique child's needs and provide the possibility for the child and their family to narrate their own story and formulate their own needs (Svensk sjuksköterskeförening, 2019). The care of a child with severe SMA must be based on evidence, clinical experience and updated guidelines, in combination with the parents' and, when possible, the child's wishes. Health care professionals need to be active and keep themselves updated on new developments. Just a few years ago, relieving symptoms and giving life-sustaining treatment were the only options for children with SMA type 1 or severe type 2. Today, the situation has changed through medical and pharmacological developments, and professionals are required to be up-to-date with the latest guidelines.

The results of this thesis show that the parents in Sweden and Denmark were relatively pleased with the care provided, but that there was potential for improvement, which is discussed below.

Within the framework of quality of care from a patient or family member perspective (Wilde et al., 1994; Wilde et al., 1993), quality of care can be considered in four dimensions: medical-technical competence of professionals, physical-technical conditions of care organisation, an identity-orientation approach in the attitudes and actions of professionals, and socio-cultural atmosphere of the care organisation.

The care given to children with SMA and their families was rated relatively highly by the parents in Paper I. Most parents reported believing that care providers had taken every opportunity to help the child feel as good as possible. However, a third of the parents reported the opposite. Bereaved parents were generally more satisfied with the care given than non-bereaved parents. The difference between bereaved and non-bereaved parents may be related to the age of the child at death/survey follow-up, as the children of non-bereaved parents were on average much older than the children of bereaved parents. This may increase the probability of having experienced negative events at some timepoint. Non-bereaved parents have also struggled with the medical and welfare systems and daily life over a much longer time. Furthermore, the bereaved parents answered retrospectively, in terms of both time and loss, which might have influenced their perspective. Another possible explanation may be that children who had died had been given access to palliative care, which children who had grown older may not have gotten. Unfortunately, we have no detailed information on the extent to which the children received palliative care.

Though most of the parents in this study indicated that they were satisfied with the care the child had received, a majority of the free-text comments expressed a negative attitude towards the care. It is possible that some of the parents who were satisfied did not write any comments.

By applying the findings in Papers I–II to Wilde's quality of care model (Wilde et al., 1994; Wilde et al., 1993), it is clear that the dimension that most findings related to was the desire for care to have *an identity-orientation approach in the attitudes and actions of the staff***.** This encompasses parents' advice that health care professionals should be responsive, empathetic, and show warmth. The parents gave advice to health care professionals to be caring, understanding, receptive, acknowledging, and show respect, a wish that has also been expressed in other

studies to professionals dealing with families affected by severe disease (Alvariza et al., 2016; Hallström, Runesson, & Elander, 2002; Lövgren, Bylund-Grenklo, Jalmsell, Wallin, & Kreicbergs, 2016; Steele et al., 2013; van Staa, Jedeloo, van der Stege, & Group, 2011). The desire that professionals should be honest has been described before in studies of advice to staff working with children (Lövgren, Bylund-Grenklo, et al., 2016; Steele et al., 2013; van Staa et al., 2011) and is in line with previous research about hope, that underlines the importance of not conveying false information as an attempt to bring hope (Snyder, 2002). However, the balance between being honest and still instilling hope in the palliative care context is a difficult art that requires experience (Groopman, 2004).

Also in line with the dimension of an identity-orientation approach in the attitudes and actions of professionals, the results of this thesis found that parents wanted to be involved in care and treatment decisions for their child. The wish of parents of children with SMA to be involved in treatment decisions has been shown in previous studies (Higgs et al., 2016; Mah, Thannhauser, McNeil, et al., 2008). This may be explained by SMA being progressive, meaning that treatment decisions always lie ahead, and that treatment is unique for each child (Finkel et al., 2018; Mercuri et al., 2018). An inconsistency was seen in the findings; as the parents developed expertise in managing their child's condition, they wanted the staff to acknowledge this expertise by turning to them as experts. At the same time, they want the staff to be more proactive in suggesting treatments and medical equipment and always stay "one step ahead". This require a close communication between health care professionals and parents.

Several of the findings in the thesis are included in the model of family-centred care, which aims to empower family members, e.g., by professionals collaborating with parents and seeing them as experts on their child's needs (Coyne et al., 2018). In line with the findings in this thesis and with family-centred care, parents of children with SMA and other chronic diseases have previously highlighted the importance of good relationships with health professionals (Mah, Thannhauser, McNeil, et al., 2008; Smith, Cheater, & Bekker, 2013) and sufficient own knowledge about one's child's disease (Higgs et al., 2016; Hinton & Kirk, 2017; Smith et al., 2013). Several studies have shown benefits of family-centred care, e.g., reduced rates of unmet needs, fewer hospitalisations, improved physical and mental health and functional status, and fewer missed school days for children

with chronic conditions (Arango, 2011). Family-centred care can therefore be seen as suitable approach of care when treating children with complex care needs and facing important medical decisions.

In the *medical-technical competence* dimension of Wilde's quality of care model (Wilde et al., 1994; Wilde et al., 1993), the parents' desire for staff to share knowledge with them can be understood. One common topic in the findings was the problem experienced with lack of knowledge about the disease among professionals. This issue is also in line with results of previous studies of SMA and other neuromuscular diseases (Lawton et al., 2015; Mah, Thannhauser, McNeil, et al., 2008; Qian et al., 2015; Rallison & Raffin-Bouchal, 2013; Yang et al., 2016). Previous studies that have explored advice from families to health care professionals have not found this problem in paediatric oncology (Lövgren, Bylund-Grenklo, et al., 2016; Steele et al., 2013), but it has been emphasised by parents of children with long-term diseases (Hallström & Elander, 2007). One explanation may be that SMA is a rare disease and professionals who do not care for affected families frequently may feel unfamiliar with symptoms and treatments that are specific to SMA. Previous research has shown that professionals who lack knowledge about a rare disease may withdraw from situations they find unfamiliar (Grut & Kvam, 2013). In this thesis (Paper II), parents emphasised that if a child is used as a tool to increase knowledge about SMA, this must be done with great respect for the family. This ties in with the parents' reports in the study by Yang et al. (2016) stating that medical treatments of their child with SMA were based on trial and error, not evidence-based protocols.

The results of the thesis show that the parents were most satisfied with the institutions that were highly specialised (the Danish rehabilitation centre RCFM, the neonatal care unit, the paediatric intensive care unit, the long-term intensive care unit, and home care) (Paper I). This shows the importance of these children receiving specialised care with continuity.

Another topic that was raised by the parents in this thesis was the perception that care organisation could be improved, e.g., by making staff more available, and the importance of continuity in care, here interpreted in the light of the dimension of the *socio-cultural atmosphere of the care organisation* in Wilde's quality of care model. As the standard recommendations of care for children with SMA describe (Finkel et

al., 2018; Mercuri et al., 2018), children with SMA have complex care needs and are cared for by many different care providers. Implementation of a key coordinator has previously been suggested in research regarding children with other complex health needs (Carter, Cummings, & Cooper, 2007; Rallison & Raffin-Bouchal, 2013) and might lessen some of the burden on parents (Rallison & Raffin-Bouchal, 2013). It is well-known that different professions contribute to patient care and treatment with different areas of knowledge, and that teamwork can contribute with different competencies. Having good coordination between care settings has previously been described as necessary to maintain a good quality of life for children with complex care needs (Cohen et al., 2011) and is emphasised as an important part of paediatric palliative care (International Children's Palliative Care Network, 2020; WHO, 2002). The Danish rehabilitation centre serves in part as a coordinator between different care settings; a similar solution might be valued by families in Sweden (Paper II).

In February 2017, the Swedish government gave the task to the Swedish National Board of Health and Welfare to design a work process for establish a concentration of highly specialised care at the national level. Children with neuromuscular diseases are nominated to be included for specialised care at national level. The goal with this national specialised medical care is to provide good and equal care throughout the country. Collaboration is now taking place between the National Board of Health and Welfare, experts, and county councils and regions to develop these efforts (National Swedish Board of Health and Welfare, 2020a). This new organisation of care could mean improvements in some of the areas that the parents in this thesis addressed as problematic. A potential benefit would be the gathering of competence, which could contribute to rapid diagnosis, correct treatment at the right time, and adequate follow-up of treatment. However, an important task for a national care centre would be to communicate information and knowledge back to local health care providers to ensure high disease-related skills at all care levels.

The final quality of care dimension, the *physical–technical conditions of the care organisation,* can be related to the parents' desire for access to the technical aids needed by the child. One general desire was that health care would be less stingy with resources, and an issue was highlighted regarding inequities of care and access to medical aids depending on place of residence within the country (Paper

I–II). A study from Norway of parents to ventilator-dependent children shows, like this thesis, that families had to struggle to get what they wanted from community health care services (Dybwik, Tollali, Nielsen, & Brinchmann, 2011).

Beyond the quality of care model, Larsson and Wilde-Larsson's model of patient satisfaction (Larsson & Wilde-Larsson, 2010) has been useful as a framework for interpreting and understanding aspects that influenced parents' views on care and its quality. Through the use of this theoretical model, it is possible to understand that quality of care is not something constant or objective, but that the objective conditions and subjective relationships interact with the parents' personal expectations and commitment to care and that these jointly affect the parents' experience of care.

6.2. Living with severe disease

6.2.1. Finding a way to live in everyday life

Living with SMA can involve a day-to-day existence full of medical aids and care, recurring hospital visits, and life-threatening emergencies. Unlike the typical child's developmental trajectory, dependence and the need for help can increase with age in children with neuromuscular diseases. Attempting to lead as normal a life as possible, despite the child's severe disease, was a theme that appeared in both Paper III and Paper IV. The parents in Paper III gave advice to other parents to lead as normal lives as possible, and the siblings in Paper IV described it as normal to have a brother with SMA, and expressed a wish that the brother would also in the future be able to lead a normal life. Striving for normality has previously been described by families with a child with severe disease; both as a desire for sameness and normality in their lives today (Kirk, 2010; Mah, Thannhauser, McNeil, et al., 2008) and as a hope for their child to be able to lead as normal life as possible in the future (Hinton & Kirk, 2017; Rallison & Raffin-Bouchal, 2013). Penrod, Hupcey, Shipley, Loeb, and Baney (2012), in their model of caregivers' search for normality in connection to end of life, described that caregivers together with the patient go through different phases in their striving for normality as the disease progresses. What "normal" life really is, is not defined by the participants in this thesis, but maybe the striving for normality is about finding an acceptable (or normal) way of living life, that is sustainable for a longer period

of time. An essential part of the experience of health, according to Dahlberg and Segesten (2010), is related to finding a way to accept and understand oneself, in connection to one's own situation, and in interaction with other people.

Circumstances and families' challenges of coping with everyday life with SMA have drastically changed during recent years as a direct result of the new drug nusinersen. Still, similarities can be seen in the results from the survey study that was conducted before nusinersen was available and the ethnographical study of children that were treated with nusinersen. These similarities were mainly connected to strategies that increased resilience. Examples include advice given in Paper III, e.g., emphasising the importance of supporting each other and daring to feel hope for a good future, whereas in Paper IV, the families conveyed strategies that increased family resilience by seeing possibilities, and using the hope that nusinersen instilled in ways that strengthened them in coping with the disease. The results of the papers can represent the participants' own successful resilience strategies being conveyed to other families in similar situations. Strategies that increase resilience for parents of children with SMA and other chronic diseases have been highlighted before, such as the importance of access to support networks (Higgs et al., 2016; Mah, Thannhauser, McNeil, et al., 2008; Smith et al., 2013) and an ability to think optimistically (Hinton & Kirk, 2017). In line with Walsh's model of family resilience (Walsh, 2016), parents in Paper III advised other parents to actively strive to give the child the best life possible, emphasised the importance of supporting each other, and advocated daring to hope for a good future.

The children in Paper IV, both Alexander and Isaac and their siblings, seemed to have adopted their parents' strategies and positive thinking. The families thereby conveyed shared family resilience with a tendency to distinguish between matters that they could change and those they could not change, and a sense of the importance of finding and being grateful for the smaller things in life (Oh & Chang, 2014; Walsh, 2016). This family resilience might have promoted their ability to feel hope and have a bright outlook on the future. However, there is also a possibility that it is the other way around: that a family's ability to have high hopes contributes to resilience. More research would be needed to further examine the interrelationship between hope and resilience.

Despite a majority of the parents in Paper III being bereaved, most advice to other parents was not about death, but about how to live to facilitate everyday life. I interpret this mainly as a way for the parents to help other parents to ensure the best possible well-being for the child, as well as for the whole family. However, caring and involvement in care can also be seen as a way to handle one's own anxiety. The act of doing something practical for the sick child can be experienced as meaningful and ease one's own despair (Benkel & Molander, 2017). This phenomenon has also been described by Bérubé (1998), who shed light on parenting experiences from his own narratives of being a parent to a child with disability. He found that despair and worries about the future never came when he was with his son, as he was then fully occupied with taking care of present needs. Instead, existential feelings became present in calmer situations. A similar situation was shown in Paper IV, where Alexander's mother experienced more existential doubts about her own situation when Alexander's disease had become more stable.

Alexander's father described that he, thanks to Alexander's severe disease, had gained an ability to appreciate the present and not hope for too much. This way of finding meaning in adversities can be interpreted based on what Folkman (2010) called meaning-focused coping, which draws upon deep values and beliefs, where hope is an essential part. Coping here means focusing on strengths gained from life experiences (post-traumatic growth) and reordering priorities.

Hope is described to be important in managing adversities. It can exist in many different forms, capable of existing at multiple levels and express itself in many different ways (Bally et al., 2014; Leite et al., 2019; Mattingly, 2010). For many families with SMA, there has probably always been a hope, despite knowledge of the prognosis: maybe a cure will come in a miraculous way. In recent years, the hope has become more realistic, which means a shift in what to expect from the future. In Paper IV, the two families and their various family members used different strategies to cope with the new hope that was brought into their lives through treatment with nusinersen.

6.2.2. Rewriting the narrative of the future

The advances in treatment have enabled for children with previously fatal diseases like SMA to survive in the long term. This has created a population of sometimes

medically fragile children, with unknown prognosis. Uncertainty about the well-being of a child with unknown prognosis is often present in parents, even if the child is stable for an extended period of time, and such uncertainty that can cause great psychological stress (Cohen, 1995). For the two families in Paper IV, there had been uncertainty regarding the prognosis throughout the children's lives. The fact that the two boys in Paper IV could start treatment with nusinersen had affected their entire families. In Paper IV, narrative was not only a relevant research method, but can also be seen as a tool that the families used to write and rewrite their understanding of life (Alsaker, Bongaardt, & Josephsson, 2009; Mattingly, 1998). With the benefit of the new drug, the families had been able to rewrite their narrative of the future. They had gone from struggling to keep their infant children alive, to reaching a situation where there was a realistic hope for a continued good future. The hope of continued life gives strength to deal with uncertainty in a disease (De Graves & Aranda, 2008), something that was shown in Paper IV.

Paper IV showed that high expectations of a treatment were seen as a haven. While Isaac, his mother, and his sibling were convinced that Isaac would be able to walk in the future, the father and extended family member did not have the same conviction, but did not want to take the hope from Isaac. Parents of children with Duchenne muscular dystrophy have previously described the hope for future treatments, something they thought was unrealistic, but which allowed them to be supportive of their children's own dreams (Erby et al., 2006). As there are no documented long-term prognoses or statistical data on the treatment of nusinersen, it is not possible to say whether or not the family's hope of Isaac walking is realistic. However, Mattingly (2010) has described that statistics play a minor role in families' ability to feel hopeful. Clinicians and families can use contrasting language when talking about hope and make different assessments of what is realistic to hope for (Mattingly, 2010). Mattingly (2010) described how clinicians' language for statistical probabilities can differ from the spiritual discourse of hope that is common in many families. This may indicate that Isaac and his mother do not reject the medical discourse about probabilities, but that they may feel that it represents only part of the picture, or that a realistic outlook is not important (Feudtner et al., 2015).

Hope of continued good life was natural for the smaller children, while challenges and fears remained for Alexander's older sibling and parents. The fact that Alexander had become more stable gave Alexander's mother more room for reflection, which caused her to have her own personal crisis. Families are constantly in movement between change and balance, stagnation and dynamics, security and uncertainty. Families in crisis search for stability, while families that perceive stagnation seek change (Walsh, 2016; Wright & Leahey, 2012). This movement between change and balance can be interpreted as one reason why Alexander's mother's experience of own life crisis once Alexander's disease had been more stable.

In Paper IV, the siblings emphasised the importance of equality for their brothers. These narratives were noteworthy, as they were identified as representing a hope beyond that of survival – for continuing life with possibilities, dignity, and high quality. Bérubé (1998) has reflected on society's responsibility for children with disabilities. He argues that when society, thanks to early intervention programmes, surgery, and drug treatment, saves children with severe disabilities, it also has a responsibility to enable those children to flourish and lead normal lives. Children with SMA undergoing treatment with nusinersen are expected to live longer, possibly with continuing physical impairment (Pane et al., 2018; Pechmann et al., 2018), which calls for a society ready to integrate persons with disabilities. Nusinersen and onasmenogene are among the most expensive drugs in the world. By developing and providing these drugs, society has decided that these children's lives are worth investing in. Having done so, society should also give the children the continued opportunity to live a life of dignity and good quality. Disability in a society urges reconsideration and is an opportunity to rethink how we organise our lives with one another and how we want our society to be designed (Goodley, 2016).

6.2.3. Living close to death

Severe SMA can be life-threatening, and to support children with severe SMA and their whole families, acknowledging the risk of death can be positive. It is known that children living with neurological diseases can benefit from palliative care (Hauer & Wolfe, 2014; Ho & Straatman, 2013), but that there are barriers related to the misconception that palliative care means end-of-life care, which can

frighten families and professionals (Boersma et al., 2014; de Visser & Oliver, 2017; Strand et al., 2013; Weaver et al., 2015). Health care professionals in paediatric care and rehabilitation therefore sometimes distance themselves from palliative care, even if they in many cases adopt a palliative approach in their care practices.

The new treatment of SMA challenges the palliative care paradigm further, as it changes SMA from a life-shortening disease to one with uncertainty regarding progression and prognosis. The families in Paper IV were life-oriented and might not have benefited from a palliative care approach. However, needs vary between different families and an understanding of the experience of uncertainty regarding the disease may contribute to the existing palliative care paradigm. The situation with this new treatment is unique to SMA in many ways, but there are other diseases that have undergone a similar change: from being life-threatening and devastating to becoming chronic medical conditions that can be well controlled with medications. If any lessons can be learned from the introduction of a drug treatment for another disease, Human Immunodeficiency Virus (HIV), a disease that is generally known, may serve as a good example. Although effective drugs have come to curb the virus, people treated for their HIV still experience great uncertainty, fear of death, and difficulty imagining a future despite the possibility of continued life. These issues has been shown both early on in the treatment of HIV (Cochrane, 2003) and more recently (Machado, 2017).

I have no intention of comparing the effects of treatment for HIV and SMA. However, the circumstances that arise when new effective drugs are developed have similarities and open for the question of whether or not palliative care is of value. A free comparison of the treatment and palliative care paradigm shift in HIV, with those of SMA, would suggest that some families with SMA, like some people with HIV, will feel healthy thanks to treatment and perceive death as being distant, while others will feel that death is an imminent threat despite treatment. The conclusion I draw from this is that a palliative approach must be accommodating. Perhaps some children with SMA and their families will need the support that a palliative approach can provide, while others will not. This means that professionals meeting children with SMA need to have good knowledge of palliative care to be able to meet each individual family's needs. Examples could include involving professionals with competence in palliative care to discuss treatment or give psychosocial support with a focus on quality of

life. Seventeen years ago, Cochrane (2003) concluded that the paradigm of palliative care of HIV needed to be challenged and reviewed, and that the palliative care should be centred on persons and families, rather than on the disease. This is now relevant for the discussion about palliative care and SMA. Today, there is uncertainty surrounding SMA and its expected prognosis, and the care must therefore be adaptable to each child's and its family's needs.

Losing a child is probably the most painful and devastating event that a parent can ever experience, and the loss or risk of loss of a child can be associated with complicated grief symptoms (Neimeyer, Prigerson, & Davies, 2002). Questions about life and death can arise, as well as themes about meaning-making. However, Neimeyer et al. (2002) stated that people can find meaning even in the face of difficult adversities. The parents in this thesis (Paper III) gave advice to other parents regarding how to deal with, and find meaning in, grief. The results may be seen as potentially helpful for other bereaved parents in finding meaning in the death of the child. Studies have shown that parents who have lost a child with disability, despite the painful loss, have been able to find meaning and benefit in the life and death of their child and to continue to see the world as purposeful (Milo, 1997). Grieving individuals may struggle to reconstruct a personal world of meaning once it has been challenged by a loss, and some may need some support in this difficult time (Neimeyer et al., 2002).

7. Methodological considerations

7.1. Considerations on design and quality of data – survey study

Papers I–III are based on questionnaires with single items developed specifically for this study. The "perfect" survey study without errors is hard to design, and some of the strengths and limitations with this survey study are discussed below. The reason for developing questionnaires instead of using existing instruments was that there were no validated scales suitable for this specific group. However, to evaluate quality of care, one could have considered adding a previously validated questionnaire to the study-specific questions (Wilde-Larsson & Larsson, 2002).

To strengthen the credibility in the questionnaires, the questions were developed in cooperation with staff from a rehabilitation clinic and parents. The questionnaires were also validated by clinical staff, to review if the questions were relevant based on their experience, and through face-to-face validation with parents, with probing questions from the interviewer after each question or area of questions.

Since SMA is a rare disease, it is a challenge to collect big study samples. This 11-year follow-up of nationwide samples from two countries, encompassing both bereaved and non-bereaved mothers and fathers of children with SMA type 1 and severe type 2, with high response rates, might therefore be considered unique. Most studies of SMA focus on one of the classification types 1–3. However, in our study, severe SMA type 2 was included in addition to type 1. This was done as there are no clear lines between the different types of SMA, since the diagnosis is made based on maximal motor function achieved. All choices of eligibility criteria can be discussed, and this has been seen as a strength, since it expanded, rather than limiting the target population. To enlarge the study sample, one could have considered including parents of all children with SMA type 2.

As individuals can have widely different experiences, both legal guardians of each child were invited to participate in the survey. With this design, both legal guardians had the possibility to have their voices heard. One limitation with the

design of the survey is that the family members in the study are not linked to each other, and it is therefore not possible to connect family members to each other. In the tables (Papers I–III), the parents are therefore presented as individuals, not linked to their child. It is important when reading the tables to remember that the number of parents is not equal to the number of children. As mentioned before, the basic assumption in this thesis is that families can be defined by themselves. However, for practical reasons, only parents legally registered as a child's guardian were invited to participate in the survey.

In surveys, it is important as a researcher to consider who chooses to answer the survey and who does not, and whether these two groups differ, as it then could lead to errors and difficulties in generalising the findings. In this study, as in other similar surveys with study-specific questions (Bylund-Grenklo et al., 2014; Kreicbergs, Valdimarsdóttir, Onelöv, Henter, & Steineck, 2004; Lövgren, Jalmsell, et al., 2016; Omerov et al., 2013; Pohlkamp, Kreicbergs, & Sveen, 2019), the response rate was high in view of the study design. Still, 16% did not participate. We do not know why these 16% chose not to participate. Possible explanations may be that it is hard for a parent of a seriously ill child to find time for responding to questionnaires, or that the questions aroused strong feelings for some. On the other hand, a majority of invited parents chose to participate despite the sensitive topic. Previous research has also shown that participation in research for bereaved family members can be experienced as something positive (Eilegård, Steineck, Nyberg, & Kreicbergs, 2013; Higgs et al., 2016; Hynson, Aroni, Bauld, & Sawyer, 2006; Kreicbergs et al., 2004; Scott, Boyle, Bain, & Valery, 2002; Steele et al., 2014; Udo et al., 2019). Half of the parents who answered the questionnaire had studied at university, a number that is higher than the Swedish average (28%) (Statistics Sweden, 2020). This may indicate that there were more parents with a higher education level among those who answered the questionnaire than among those who did not. This, in turn, may have had an impact on the answers. However, as the total response rate of the survey was high, one can reason that the population was relatively representative.

The high response rate might also be a result of invited participants feeling acknowledged and able to relate to the questions, as they were developed for this specific group. In the free-text comments, several parents expressed appreciation at having been invited to the study. One parent wrote:

> It feels good to get this survey. My first reaction was - What?! Then we're not the only ones affected by this. That's not how it has felt. We have felt very lonely in this.

The Swedish sample of the survey had a higher response rate than the Danish sample (87% vs 79%). One modification in the data collections that could have impacted on this difference was that the Swedish parents who had consented to participate, but had not returned the questionnaire, were contacted by phone and asked if they had any particular concerns or needed assistance in completing the questionnaire.

Danish parents' survey responses were sent by post to the Danish RCFM, where the co-authors of Paper I and III worked, which may have influenced the parents' answers about this centre. Likewise, one of the researchers behind the survey in Sweden worked clinically at a hospital. Some parents may have had a relationship to him, which may have affected their answers.

7.2. Considerations on design and quality of data – ethnographical study

In Paper IV, a narrative approach was chosen to gain a deeper understanding of how families experienced and negotiated with hope when a new therapy for SMA became available. Mattingly (2010) has described narrative data collection and analysis as good candidates for understanding experiences of hope, as the method can highlight the unfinished and unpredictable qualities of life and reveal the eventualities of social life, from small moments to broad cultural histories.

The data that were collected for Paper IV consisted of digitally recorded and transcribed interviews and field notes from observations. Observations allow the researcher to collect data from the participants' perspectives, adding the extra dimension of the physical environment (Mulhall, 2003); in this case that was the families' own homes and everyday life. All interviews and observations for Paper IV were collected by me, which enabled creation of relationships.

It is important to consider which families choose to participate in studies like this one. Participation requires a good deal from the participants; they must feel that they have the time and energy to receive the researcher in their home. Being open

and sharing one's life and life situation also takes courage. Therefore, there is reason to believe that those who chose to be part of the study felt resilient, were open to communication, and were experiencing a good period in life.

The data collection for Paper IV was conducted over a long period of time (9 months), which increases credibility, as the families narrated different events in their daily lives depending on the season and how they felt at the time.

Each method carries its own weaknesses and strengths. Using a variety of methods can illuminate a research question from different perspectives and minimise the risk of errors linked to a particular method (Patton, 2015). Using multiple data collection methods, such as questionnaires, interviews, and field notes from observations, can therefore be seen as a strength.

7.3. Considerations on data analyses

Data in Paper I were analysed with both descriptive statistics and content analysis. The quantitative analyses were discussed with statisticians to determine if statistical analysis was possible. Due to small numbers, use of descriptive analysis was considered appropriate.

In Papers I–III, content analysis was conducted. Some of the written statements were quite short, e.g., "Listen to the parents". Short meaning units can result in fragmentation and therefore be difficult to interpret, something that was considered during the analysis process (Graneheim & Lundman, 2004). Both manifest and latent contents deal with interpretation, but the interpretations vary in depth and level of abstraction. Given the short statements, I have consciously strived to stay close to the data and avoid latent and more abstract interpretations. This prevented deeper interpretation of the underlying meaning. On the other hand, a latent interpretation would not have done the data justice, as the data did not have the same quality and richness as deeper interviews can have.

The identified categories were quantitized by counting the number of statements that were expressed in each category (Papers I–II). This way of quantitizing qualitative data has been discussed, and is discouraged if the number of participants is low, as there can be a risk of drawing conclusions based on misleading percentages. Numbers can have a rhetorical power, since they are

associated with scientific precision, which can lead to them being confused for truth (Patton, 2015). However, the samples in Papers I–II were relatively large for a qualitative analysis, and this kind of quantitizing was therefore considered acceptable. This way of presenting numbers can also be seen as increasing transparency with the analysis and interpretations made, and can help to deepen the understanding of patterns (Sandelowski et al., 2009).

To strengthen the credibility in Paper IV, I strived to be clear in method descriptions, interpretations, and presentation of data. This was done so that the reader can follow the research process and test the trustworthiness of the interpretations in relation to the aim of the study. I strived to be open to new interpretations, by challenging my own interpretation in dialogue with the co-authors and previous literature.

There is more than one way to interpret a text (Polkinghorne, 2007). The development of narrative interpretations are creative productions derived from the researcher's cognitive processes to recognise patterns, similarities, and differences compared with other texts. The two narratives that are presented in Paper IV, generated with two families, should be viewed as co-constructions between the participants and the researchers, where the reconstructed narratives and interpretations are created by us researchers (Mattingly, 1998; Polkinghorne, 2007). This view of knowledge is in line with the social constructivist epistemology as constructed between people, and thus not an image of reality (Burr, 2015). As I see it, this is especially relevant as regards human experiences and relationships between people, which have been the focus of my thesis. This approach means that there is no unique "right" way to interpret data, but that several different interpretations are possible. Therefore, I make no claims that the proposed interpretation in Paper IV is the only one possible. However, to enable the readers to return to the analytical steps and assess the plausibility of the offered interpretation, I have striven to be clear that ours is an interpretation based on the compiled texts (Polkinghorne, 2007).

In narrative research, there are different traditions regarding how to best "validate" the researcher's interpretations. In Paper IV, I chose to make my interpretations in triangulation with earlier theories and the other researchers in my research group. Another way could have been to let the participants confirm

my interpretation. However, I would argue that the interpretations are the researchers' own, meaning that confirmation or questioning of the participants would not necessarily improve or validate the findings.

7.4. Considerations on the credibility of the researcher

The role of the researcher must be taken into consideration and reflected upon to ensure credibility, especially in qualitative inquiry, as the researcher is an important instrument (Patton, 2015). It has been of importance for the credibility in the papers that the co-authors together covered wide experience with specific knowledge of the methods used and of SMA.

The data in the survey study was already collected when I began my PhD studies. My first interaction with the data was when I entered all the Swedish data, both open-ended and closed-ended handwritten answers, into a database. The Danish data were already entered into the database, but I went to Denmark to carefully read through all collected answers and anonymise the data, as this was required under Danish data law before it could be brought to Sweden. Both these actions were a good opportunity for me to become well-acquainted with the data.

During data collection and analysis, reflexivity is of utmost importance. As researchers, we are present in the research and must reflect on how this presence affects the creation of research knowledge. In order to strengthen the credibility of the thesis, I have striven to be aware of my own preunderstanding and possible prejudices of the context that was studied. At the start of the PhD, I had some experience of the context through my clinical experiences as a paediatric nurse. The experience of being a parent must also be taken into account. Some argue that one should try to avoid own preunderstandings as much as possible, while others emphasise the importance of taking advantage of them (Dahlberg, Dahlberg, & Nyström, 2008). My identity as a nurse and parent, and my own values and views could not be erased, but were instead seen as useful tools to understand the deeper meanings of the phenomenon that were studied. In addition, my identity as a nurse and parent, and having a similar cultural background as the participating families facilitated in establishing a relationship with the families in Paper IV (Carolan, 2003). While I tried to draw benefits from

my previous experiences, I attempted to be aware of my own preunderstandings and not make interpretations based directly thereon. This challenge has also been the subject of active discussions within our research group in relation to possible interpretations of data.

In the ethnographical study, I was dependent on my own skills in conducting interviews and observations as an instrument for data collection. To prepare for this, I took methodological courses, practiced interviewing technique with children, and performed pilot observations in families. These preparations were valuable for my ability to conduct rich observations and interviews, though my technique could have been further developed with more experience. I also had good support from my supervisors and co-authors during the process, as all the collected data were shared and we had continuous reflections and discussions on how to develop and deepen the questions.

I strove to be inviting and responsive when an informant wanted to tell me something, but at the same time careful not to intrude, showing respect for each person's integrity. I tried to appear as an interested researcher who wanted to learn from the participants. Although the situation of having a researcher in the own home was probably quite unusual for them, all three families that I visited were welcoming and the mood was relaxed.

7.5. Considerations on possibilities of transferring findings

One challenge with research is how to judge the possibility to use the findings as evidence. The survey studies were nationwide and had high response rates, which increases the possibility to generalise the findings to other different persons, settings, or times (Polit & Beck, 2016). However, the results of Papers I–III might not be directly generalisable to other countries without considering the cultural context of Sweden's and Denmark's welfare systems and care organisations. Further, after the surveys were conducted, new therapies have shown potential to change the course of SMA, with possibly prolonged survival. Therefore, the results of the survey study might not be directly generalisable to families of children with SMA who receive these new therapies. Due to differing national regulations, not all children worldwide will have access to the new therapies, and

the findings could therefore be of value for families and health care professionals who do not have access to effective therapy. Further, the findings may be useful for families and health care professionals facing similar experiences with a child who has a severe disease other than SMA.

In qualitative studies, the extent to which the findings can be transferred to other settings and groups is often referred to as transferability. In an ethnographical study, where the number of participants is small, it becomes crucial that these few participants convey a narrative that can be recognised by others, in order for the results to be interesting for others to read. The value of this kind of ethnographical study is that the method provides a deeper understanding of the experience than many other methods would generate.

To promote transferability and enable generalisation of my papers, I have endeavoured to give as detailed descriptions as possible of the samples and contexts, in order to enable for other researchers, clinicians and families to decide whether our data can be applicable for them in their settings or contexts.

8. Conclusions

The findings from the four papers in this thesis add knowledge about children's and families' experiences of care and everyday life with SMA, and of the hope that has come with the new medicine for SMA. The research showed that the parents were relatively pleased with the care given, but there was a discrepancy between how the parents wanted the care to be and how it actually was. The fact that staff often lacked knowledge about the diagnosis and that the parents felt that they themselves had to take initiatives for the best care of the child, highlights the need of highly specialised care from multi-professional teams, with highly disease-specific knowledge and knowledge about palliative care, in order to assess and meet the needs of the family. The existing information and knowledge is also important to pass on to the family and to other care settings involved in the care of the child, to achieve coherent care.

Children with SMA have complex care needs, and the thesis highlighted deficiencies in coordination between different health care providers. A desire for the establishment of a central function that could help to coordinate health care visits came to light.

The parents emphasised the importance of having a good relationship with health care staff, and care professionals were seen as having an important role in creating circumstances for a trustful and close relationship between them and the family. Parents also wanted health care professionals to see them as experts on their children, while at the same time wanting health care to take greater responsibility and be more proactive – a balancing act that underscores the importance of a close dialogue between families and health care professionals.

Parents highlighted the value of getting help with practical support with everyday activities, as well as social support for dealing with disease and grief, to promote coping with everyday life. This kind of support can be given by health care, by supporting groups in civil society, and by friends and neighbours. The parents recommended other parents to try to focus not only on illness and treatment, but also be present with their child, enjoy the child, and be active in the care of the child. Feeling that you have done everything for your child could, according to the parents, facilitate the later grief work after the child had died.

To increase resilience in families, health care professionals can take in and pass on advice from parents. Some of the advice given can be found in theories of resilience, for example the importance of utilising practical and psychosocial support, daring to feel hope for a promising future, and seeing the healthy aspects in the child. The advice given is valuable for health care professionals in their task of strengthening families living with SMA or other severe diseases.

Lastly, with the new medicine – nusinersen – the families' narratives were rewritten. The families were facing slow improvements; small events that made a big difference. These improvements gave the children more energy to play, gave the parents an opportunity to relax, and nurtured hope for the future. The hope was negotiated and struggled with in different ways by different family members, but contributed to each family member's own way of dealing with the disease and their outlook on the future. Hope does not necessarily mean a firm belief in a bright future, but can also entail a struggle. For example, the hope that the child will live can cause a parent to temporarily question their own way of life. The thesis also clarifies how hope in everyday life, whether realistic or not, has the potential to strengthen children and parents in their way of looking at the disease and future.

9. Implications and future research

The thesis provides concrete suggestions for improvement of care, from the families' own perspectives. Many of the recommendations are immediately applicable for the individual health care professional.

The parents emphasised the importance of having a good relationship with care providers, for example that health care professionals need to listen to them, show that they like the child they are caring for, and dare to stay even if it is difficult. Increased awareness of how important this is to the families might remind staff to slow down and nurture the relationships with the families they are caring for – something that does not cost money or really take any time.

A further implication of the result is that health care professionals should listen to parents' knowledge, as experts on the child. At the same time, there is a risk that parents will be given too much responsibility and health care professionals are therefore recommended to also be proactive and always try to stay one step ahead on the disease trajectory. This balancing act requires a close dialogue and a good relationship between the family and the health care professionals.

Furthermore, the result of the thesis suggests that health care professionals need increased knowledge of the specific disease. As SMA is rare, a system is required that allows specialised knowledge to reach the individual family, possibly via a highly specialised national health care team. However, as the results show, it is important that the specialised knowledge does not stay with the experts, but is passed on the family and other care settings involved in the care of the child, to achieve coherent care.

Since children with SMA have complex care needs, efforts from many different care providers are required. The parents reported deficiencies in coordination between different care providers. They suggested that health care should set up a central function to facilitate coordination of all health care visits – something that can be considered when organising health care of children with SMA and other complex care needs.

The results of the papers contribute with the participants' own successful resilience strategies, as they would be conveyed to other families in similar

situations. The results include advice on providing practical support with everyday activities, as well as social support, for instance encouraging parents to share their innermost feelings with those closest to them, as this facilitates disease management and eases later grief. The advice given can serve as important tips for both health care professionals, civil society organisations that support families with SMA, and friends and family members.

A better understanding of ways in which hope can be negotiated may be useful for health care professionals in giving support to families of children with SMA. The knowledge provided in the thesis may enable staff to nurture and normalise hope in families, as an essential aspect of managing everyday life. The thesis also clarifies how hope in everyday life – whether realistic or not – has the potential to strengthen children and parents in how they approach the disease, which staff should be aware of.

The results of the thesis can also contribute with creating a basic understanding of people's experiences in the context of families in everyday life, something that is important when developing new interventions aiming to support families. A concrete way to support families might be to implement and assess interventions for facilitating care coordination of children with SMA.

The results may also form the basis for the development of new interventions aimed at supporting families in their ability to feel hopeful, an area that is still relatively unexplored (Kaye et al., 2020). Increased knowledge about (and development of) interventions that improve resilience and ability to use hope as a strategy to cope effectively with disease may be needed.

More knowledge may also be needed about children's and siblings' experiences of living with SMA, or having a brother or sister with SMA. Long-term experiences of being treated by the nusinersen should also be gathered, perhaps in a longitudinal study. It is imperative to gain more knowledge about the paradigm shift of palliative care within a disease when new effective drugs are introduced, to provide the best support to families in times of uncertainty.

Sammanfattning

Introduktion

När ett barn får en svår sjukdom blir hela familjen drabbad. Livet tar en ny vändning och vardagen kantas kanske av sjukvårdsbesök, vårdkontakter och medicinska hjälpmedel. Spinal muskelatrofi (SMA) är en sällsynt och mycket allvarlig sjukdom som drabbar små barn. Sjukdomen är genetisk och kännetecknas av gradvis ökande muskelsvaghet. SMA delas vanligen in i tre svårighetsgrader (SMA typ 1–3) baserat bland annat på barnets ålder vid diagnos. Symtomen är liknande för alla typer, men generellt gäller att ju tidigare symtomen visar sig, desto snabbare sker muskelförsvagningen. Utan behandling avlider barn med den svåraste formen, SMA typ 1, oftast inom de två första levnadsåren, till följd av andningssvårigheter och svårigheter med att svälja. Med den andra typen av SMA, SMA typ 2, lever barn utan behandling upp till ung vuxen ålder, och med den tredje typen räknar man inte med förkortad livslängd. Detta avhandlingsprojekt fokuserar på de två svårare typerna av SMA.

Fram till för bara ett par år sedan fanns ingen effektiv behandling för någon form av SMA. All behandling inriktade sig därför mot att förebygga komplikationer av svagheten och att bibehålla livskvalitet, framför allt genom andningsstöd, nutritionsstöd, ortopedi och palliativ vård (Arnold et al., 2015; Finkel et al., 2018; Mercuri et al., 2018). Sedan december 2017 finns dock ett läkemedel registrerat i Sverige, nusinersen (Spinraza), som möjliggör inbromsning av muskelförtviningen och till och med möjlighet att få tillbaka tidigare förlorade muskelfunktioner.

Trots att sjukdomen är mycket allvarlig och familjerna ställs inför svåra utmaningar relaterade till sjukdomen, är kunskapen om hur familjer upplever vardagslivet med sjukdomen och barnets vård begränsad. Barn med neurologiska sjukdomar är tyvärr fortfarande nästan osynliga inom forskning som rör upplevelser av sjukdom och erhållen vård.

Föräldrar är en viktig del av vården av ett barn, och deras erfarenheter och råd och tips till andra föräldrar och vårdpersonal är värdefulla, både för utvecklingen av barnsjukvården och för att ge andra föräldrar stöd i sin situation.

Tidigare studier som involverar familjer med svår sjukdom betonar vikten av hopp, men det finns fortfarande mycket att utforska med tanke på komplexiteten kring begreppet hopp. SMA är särskilt intressant att fokusera på i förhållande till hopp, eftersom förhållandena kring SMA har förändrats de senaste åren; från att vara en livsbegränsande sjukdom till att bli en sjukdom som kan förenas med ett långt liv.

Kunskap om familjers upplevelser av den vård de fått, upplevelser av vardagslivet med sjukdomen, och hur familjerna på olika sätt uttrycker hopp, kan vägleda vårdpersonal i deras arbete med att ge vård av god kvalitet och öka välbefinnande hos barn med SMA och deras familjer.

Syfte

Det övergripande syftet med denna avhandling var att utforska hur familjer upplever vården och vardagen med SMA.

Metod

För att söka svar på det övergripande syftet har olika metoder använts. Avhandlingen är uppdelad i två studier: en enkätstudie (delstudie I–III) och en etnografisk studie (delstudie IV).

Enkätstudien baserades på två rikstäckande enkätundersökningar i Sverige och Danmark där föräldrar till barn med svår SMA deltog. Enkätstudien hade en tvärsnittsdesign och genomfördes först i Sverige och ett par år senare i Danmark. Både föräldrar som hade mist ett barn med SMA och föräldrar som levde med ett barn med SMA deltog (totalt 95 föräldrar med en svarsfrekvens på 84 %).

Den etnografiska studien fokuserade på hur familjer, där ett barn nyligen påbörjat behandling med nusinersen, uttryckte och hanterade hopp i vardagen. Rekrytering skedde bland dem som tidigare hade varit med i enkätstudien, samt via patientföreningen Nätverket för SMA:s Facebook-sida. Av de fyra familjer som bidrog med data till studien valdes två familjer ut till delstudie IV, då de valde att

träffa mig flera gånger. Datainsamling till delstudie IV ägde rum genom att jag besökte familjerna i deras vardag, och data bestod av fältanteckningar från deltagande observationer (sex tillfällen) i kombination med narrativa intervjuer med familjemedlemmar (17 intervjuer).

Resultat

Resultaten visar att föräldrarna som deltog i enkätundersökningarna generellt var nöjda med den vård deras barn hade fått. Föräldrar som hade mist sitt barn var mer nöjda med vården än de som levde med sitt barn. Dock framkom vissa brister, framför allt i att personal saknade kunskap om diagnosen, vilket medförde att föräldrarna upplevde att de själva måste ta initiativ till åtgärder för att deras barn skulle få den bästa vården. Då barn med SMA har komplexa vårdbehov krävs insatser från många olika vårdgivare, och föräldrarna rapporterade i studierna om brister i samordning mellan dessa olika vårdgivare. En önskan om att inrätta en samordnande funktion som kunde bistå med att koordinera vårdbesök kom fram. Föräldrarna betonade vikten av att ha en god relation med vårdgivarna. De önskade även att vårdpersonal skulle se dem som experter på sina barn, samtidigt som de önskade att hälso- och sjukvården skulle ta ett större ansvar och vara mer proaktiva.

För att klara av att hantera vardagen med ett barn med svår SMA betonade föräldrarna vikten av att få hjälp med praktiskt stöd med vardagssysslor, samt socialt stöd för att hantera sjukdom och sorg.

Föräldrarna rekommenderade andra föräldrar att inte bara fokusera på sjukdom och behandling, utan också vara närvarande med sitt barn, njuta av barnet, samt vara aktiva i vården av barnet. Att känna att man gjort allt för sitt barn kunde, enligt föräldrarna, underlätta det senare sorgearbetet efter barnet.

Med det hopp som det nya läkemedlet gav skrevs familjernas livsberättelser om. Familjernas berättelser hade skiftat från att fokusera på en kamp med dagliga utmaningar för att hålla barnet vid liv till att fokusera mer på en lovande framtid. Familjerna såg hur barnen långsamt fick mer energi av det nya läkemedlet, små händelser som gjorde stor skillnad i familjernas liv. Hoppet tog sig olika uttryck för olika familjemedlemmar, men bidrog till familjemedlemmarnas egna sätt att hantera sjukdomen och sin framtidstro.

Slutsatser och implikationer

Avhandlingen ger konkreta förslag till förbättring av vård, utifrån familjernas egna perspektiv. Många av rekommendationerna är direkt tillämpbara för enskilda medarbetare i vården.

Föräldrarna betonade vikten av att ha en god relation med sina vårdgivare, till exempel att personalen lyssnar på dem, visar att de tycker om barnet som de vårdar och att de vågar stanna kvar hos familjen även om det är svårt. En ökad medvetenhet om hur viktigt detta är för familjerna kan kanske hjälpa personalen att sänka tempot och vårda relationen till familjen – något som inte kostar pengar eller behöver ta mycket tid. En ytterligare implikation av resultatet är att vårdpersonal behöver lyssna på föräldrarnas kunskap om barnet och se dem som experter på sitt barn. Samtidigt finns det en risk att föräldrarna får för mycket ansvar och därför rekommenderas vårdpersonal att vara proaktiva, ta initiativ till nya åtgärder och alltid försöka ligga steget före i sjukdomsförloppet. Denna balansakt kräver en nära dialog och god relation mellan familj och vårdpersonal.

Resultatet av avhandlingen visar även att vårdpersonal behöver ökad kunskap om SMA. Eftersom SMA är sällsynt krävs ett vårdsystem som möjliggör för specialiserad kunskap att nå den enskilda familjen, kanske via ett högspecialiserat nationellt sjukvårdsteam. Som resultaten visar är det dock viktigt att den specialiserade kunskapen inte stannar hos experterna utan förmedlas vidare till familjen och andra lokala vårdinstanser. Föräldrarna rapporterade i studierna om brister i samordningen mellan olika vårdgivare. De föreslog att vården skulle inrätta en koordinerande funktion som kunde bistå i att samordna alla olika vårdbesök – något som kan övervägas när man organiserar vården av barn med SMA och andra komplexa vårdbehov.

Deltagarnas egna framgångsrika strategier för att leva i vardagen förmedlades till andra familjer i liknande situationer; flera av dessa råd återfinns i teorier om resiliens. De konkreta råden om praktiskt stöd med vardagssysslor, och socialt stöd för att hantera sjukdom och eventuell sorg, samt att hjälpa föräldrarna att inte bara fokusera på sjukdom och behandling, utan också vara närvarande och njuta av barnet, är viktiga att känna till för personer som möter familjer i liknande situation. Det kan gälla både vårdpersonal, civilsamhällets organisationer som stöder familjer med SMA, samt vänner och familjemedlemmar.

En bättre förståelse av hur hopp kan upplevas och användas i vardagen kan underlätta för vårdpersonal att stödja familjer med barn med SMA. Resultatet kan hjälpa personal att nära och normalisera hopp hos familjer som en viktig del i att hantera vardagen med sjukdom. Avhandlingen klargör också hur hopp i vardagslivet, vare sig det är realistiskt eller inte, har potential att stärka barn och föräldrar i hur de ser på sjukdomen och framtiden, något som är viktigt för personal att vara medvetna om.

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Experiences of care and everyday life in a time of change for families in which a child has spinal muscular atrophy

Elin Hjorth is a specialist nurse in paediatric care and has worked in paediatric neurology at Astrid Lindgren Children's Hospital for many years. This thesis focuses on children with severe spinal muscular atrophy (SMA) and their families. Although the disease can be life-threatening, and the families are faced with challenges in everyday life related to the progressive muscle weakness of the child, there is limited knowledge of how families experience their situation and the care received. The introduction of new therapies in the SMA area is rapid, and with new therapies that prolong survival, the landscape of SMA has changed dramatically for affected families.
This thesis adds knowledge about families' experiences of care and everyday life with SMA.

Ersta Sköndal Bräcke University College has third-cycle courses and a PhD programme within the field *The Individual in the Welfare Society,* with currently two third-cycle subject areas, *Palliative care* and *Social welfare and the civil society*. The area frames a field of knowledge in which both the individual in palliative care and social welfare as well as societal interests and conditions are accommodated.

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