The experiences of mothers and young people with neuromuscular disorders: Pushing back and pulling away through transition.

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Contents

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Abstract

The number of young people with neuromuscular impairments reaching adulthood has increased due to advances in medical science. As young people with progressive degenerative neuromuscular conditions continue to live longer than previously anticipated, their needs for services, and opportunities to live the extended period of their lives in ways that are meaningful for them need to be met, with primary, secondary and respite services in place to meet the needs of a growing and emerging group of young adults. Qualitative research about the experiences of these young people is limited. The experiences of the mothers of children and young people neuromuscular impairments is currently underrepresented in the literature.

A study using case study methodology was undertaken to explore the experiences of five mothers of children: and five young adults with neuromuscular conditions. Eight cases were developed using a range of data sources including: one-to-one interviews with five mothers and one young adult, online open-ended questionnaire responses from young people, artefacts such as photos and video clips held by mothers, and email correspondence with participants. Each of the cases are presented as a journey, for the mothers starting with the antenatal and birth stories, followed by caring for their children in early and later childhood years and for the young adults', their journeys through adolescence and early adulthood. Data were also triangulated deductively using three theories: identity theory; Transition theory; and the ABC-X Theory of Family Stress and Coping.

The findings showed that being the mother of a child with a neurological disorder had a significant impact on their identity as mothers. Within-case themes for the mothers were the journey to parenthood; parental hopes, dreams, and aspirations; the importance of family and social support; and facing the future and continuity of care. Identity, as a disabled person, coloured the lives of the young adults, although they were each determined to live as normal a life as possible. For young people, the immediacy of short-and medium-term goals were important. The young people were aspirational for their futures with each identifying clear educational and career goals. The mothers were determined that their child's potential would be maximised, their goals reached, and that their children would not be held back by budgetary constraints or limitations in service provision. The key theme to emerge in the cross-case analysis was the pushing away and pulling back through transition; to and from independence for the young people approaching and entering early adulthood, and for the mothers pushing their child to independence whilst also pulling them back due to dependency; becoming a mother of a disabled child and an expert parent; and holding onto the child and mutual dependency.

The original contribution this research makes to the field, is in expanding understanding of the maternal perspectives of raising a child with a neuromuscular impairment and how identity theory can be used to conceptualise these experiences; and of the voices of young adults growing up with a neuromuscular impairment of their hopes and aspirations for their academic, career and personal hopes of the future.

Key words:

Adolescents, young adults, neuromuscular disorders, identity, transition, mothers, case study methodology.

Chapter 1: Introduction

1.1 Introduction

In this chapter I will explain the background and context for this research study and identify the key policies that have been influential in raising the profile of children and young people with continuing health needs, including progressive and non-progressive neuromuscular disorders. I will discuss the impact of local and national policies which relate to the health and wellbeing of young people with chronic and complex medical conditions, and how these policies have influenced the services that young people are able to receive, as they grow up with neuromuscular impairments. In the chapter I will also address the impact of a diagnosis of a neuromuscular impairment for the young person, their parents and siblings and will discuss the expected development of young people as they move through their adolescent years to adulthood. This introduction will conclude with an outline of each of the chapters.

1.2 The reasons for undertaking the study.

As a health professional, I have had many years of professional practice and experience of working in different roles as a qualified adult nurse, children's nurse, and a health visitor. These professional roles have provided me with opportunities to meet and provide care for children from birth to late teenage years who have continuing healthcare needs, both in acute healthcare settings and in the community. Some of these children and young people had been given a diagnosis of a progressive or non-progressive neuromuscular disorder, which is a diagnosis given to an individual who has either been born with or who has an acquired a neuromuscular impairment. Whilst some infants may have sustained a brain injury in utero, others may have sustained an injury to their brain which causes ongoing damage, due to a premature birth or because of an injury during a traumatic delivery at birth. Other infants and young children may have been born following a normal pregnancy and have had an uneventful and expected antenatal and postnatal period, with their subsequent developmental achievement being within the normal expected time frame. However, as their early childhood continues, a small number of children develop symptoms which go on to lead to a diagnosis of a genetic disorder of a progressive, degenerative, and life-limiting neuromuscular disorder.

At the start of the study, my intention was to explore the experiences of ten young people aged between 16 and 21 years in relation to their preparation for, and their experiences of, their transition from child to adult services. I was interested in this subject area due to my

experiences of working as a Community Children's Sister for nine years, which led to many opportunities for me to observe the impact that leaving one service, where young people have been well known and supported for the duration of their childhood years by health professionals and moving into adult services has had for young people and their parents. Currently in most NHS services, during their teenage years, young people and their parents must leave the services that they have had confidence in over many years and move to new services where adjustments and new therapeutic relationships need to be built. During my professional life, I was also aware of the anxieties and stress that parents can experience as their children move from child to adult services, and that the need for support and guidance is not just important for young people but is also vital for parents and primary caregivers. The voices of children and young people as service users of health and social care provision are important, as only they can give an account of their needs, wishes and experiences of care from their own perspectives.

One weekend I was working at a local children's hospice with a 17-year-old young woman with severe cerebral palsy, who had no verbal communication abilities but who communicated by eye gazing to an alphabet chart on her wheelchair. She was approaching her transition to adult services and she spelt out for me *"let me tell my story – don't ask my mum".* This comment crystallised for me the importance of providing opportunities for young people with neuromuscular impairments to have the opportunities to be heard for themselves, and for health professionals to develop greater awareness of young people's needs and the needs their parents and wider family prior to their move to adult services.

Whilst it is true that some young people have their journey to adult services well timed and, coordinated and with appropriate transition services being available for young people to move on to, the accounts from the literature, national policies, and personal accounts from young people, tell a different story, where young people have recounted poor transition experiences and, in some cases, a lack of appropriate services for young people to move on to once they have reached adulthood. The literature on the importance of transitions planning has demonstrated that there are long term health implications when young people's experiences of transition to adult services are poor. The evidence that these negative experiences can affect compliance and the future well-being of young people with chronic health issues is compelling (Watson 2000, Bell 2007, Baines 2009, Bell et al 2010, Berg-

Kelly 2010, Crosby-McKenna 2010, Perry et al 2010, McKenzie, et al 2011, Moola et al 2011, Björquist et al 2014, Baldanzi, et al 2016, Hagell et al 2019).

The needs of children and young people cannot be considered in isolation, as they each have primary care givers throughout their childhood years, who coordinate and provide the continuous daily care required until and often beyond the time that adolescents reach adulthood and the parents' needs also need to be considered. However, the recruitment of young people who met the inclusion criteria for the study was more challenging than I had anticipated (these challenges are discussed in detail in the methods chapter, see 3.6.2).

The aim of the study was to explore the experiences of young people with a neuromuscular impairment growing up into adulthood, and to understand the experiences of their parents or primary caregivers. The study objectives were to:

- Explore and understand the perspectives of young people with neuromuscular conditions.
- Explore and understand the experiences of parenting for parents or primary caregivers of young people with neuromuscular conditions.
- Gain an understanding of the issues and challenges encountered by young people with neuromuscular conditions and their parents or primary caregivers.
- Explore the transition experiences of young people with neuromuscular conditions moving to adulthood.

1.3 Key policies on transitions between child and adult services

Since the millennium, the health and wellbeing of children and young people has continued to be identified as being of increasing importance for the future of the UK. In the past 16 years, prominent clinicians have sought to continue to raise the profile of young people's health to policy makers at both a national and local level; with the aim of providing the consistency and the quality of services required to meet the specific needs of young people with complex and continuing health needs (Aynsley-Green et al 2000, McDonagh et al 2006, McDonagh 2007, McKenzie et al 2011). Their aims have been supported by multiple policies and guidelines from a range of Government Departments, Charities and local guidelines which have been developed to address the needs of children and young people within complex and continuing healthcare needs (see for example: National Service Framework

2004, DH 2007, DH 2008, DH 2008, Building on the Foundations: The Need for a Neuromuscular Service Serving Patients in the NHS South Central Region 2009; RCN 2013; Department of Education 2014; Quality Commission 2014; NHS South East Coast Strategic Clinical Networks 2015; Chambers et al 2015; NICE 2016; The Children and Families Act 2014, Together for Short Lives 2018).

The National Service Framework for Children, Young People and Maternity Services (DH 2004) was a comprehensive and far-reaching document which highlighted the long-term impact of inequalities in health for children and young people. This policy framework introduced the concept that health and social services were to become child focused and child centred, with health and social care provision working closely together to meet the needs of children and their families. This 10-year strategy was innovative in that it recognised the importance of the antenatal period and the long-term effects which can occur when infants are born prematurely. The framework had 11 standards, with standard 8 specifically focussed on the needs of children and young people with complex health needs. This standard was designed to be transformative for children and young people and emphasised the needs for flexible and appropriately funded care packages designed individually and holistically to meet the needs of each child with complex healthcare conditions. It acknowledged that some children and young people will go on to need palliative and end of life services, which were to be planned and delivered at a time that would meet the needs of each child and their family. This document was far reaching and an important seminal policy which paved the way for subsequent polices to be developed; however, although it was due to be revised after 2014, this has not yet taken place.

The 2007 Aiming High for Disabled Children Department of Health policy document (DH 2007) aimed to transform services for disabled children and their families. It provided a commitment of three years of essential additional funding of 370 million pounds to invest in services for disabled children. This included increased funding for the provision of short break respite care. The policy acknowledged the needs of parents to be able to access work and announced the additional financial commitment of 35 million pounds to contribute towards the funding of suitable childcare facilities to meet the needs of disabled children and enable parents to work. The gap in transition services for young people moving from child to adult services was also recognised and the Department of Health announced a new ‘Transition Support Programme’ funded by an additional 19 million pounds to provide better

support for young people and their parents as they approached their transition to adult services. This policy has raised the profile of the needs of disabled children and their families.

The DH (2008a) published guidance for commissioners and providers of health services on the needs for young people with complex health needs or a disability as they approach their move to adult services. This policy document sought to address the issues that had been highlighted by young people and their parents concerning their transition from child to adult services. This was the first major document that acknowledged the feelings of loss that young people can experience as they move to an unfamiliar adult service and specified the need for transition to be a process which is planned and prepared for over a long period of time. "Growing up matters: better transition planning for young people with complex needs" (Commission for Social Care Inspection 2007) provided guidance for social care providers of the transitions needs for young people with continuing healthcare needs. This guidance identified that the period of transition between services can pose risks for young people and their families which need to be mitigated against. The policy advocated that transition should be planned for each young person and their parents, with the child and their needs placed at the centre of the plans and arrangements made. This endorsed the guidance from the DH in standard 8 of the NSF for Children, Young People and Maternity Services (2004). This standard within the NSF recommended that a health transitions plan should be developed in collaboration with each young person. This was important and necessary guidance for professionals, however more than a decade after these publications, transition is still not working well for all young people, although examples of good practice are evident in different parts of the country.

A second document produced by the DH (2008b) built on the previous documents and focused on the needs of children and young people with complex and life-threatening conditions. It aimed to provide these children with packages of care that were adequately funded and resourced and were developed in conjunction with health and voluntary organisations. The focus in this document concerned the integration of services to meet the needs of children and recognised the importance of meeting the spiritual and psychological needs for the child and family. In this policy, the identification of one key professional to coordinate the individualised package of care for the child was acknowledged, which was to include advance care planning to prepare for the child's death as part of palliative care. This

document identified the increased numbers of children and young people living to adulthood due to advances in medical care and acknowledged the resource and funding concerns that needed to be in place to meet the care needs of each child and young person.

The Walton Report commissioned by the Muscular Dystrophy Campaign (2009) was undertaken by an influential and wide reaching all-party parliamentary group (APPG) inquiry which examined the needs of individuals with Muscular Dystrophy. This report sought the views of a wide range of individuals affected by neuromuscular conditions, their relatives and specialist health staff working within this specialist field of practice. The report made seventeen recommendations which sought to raise the profile and highlight the needs of patients with all forms of neuromuscular conditions, and established guidance for improving the care of patients with neuromuscular impairments. The inquiry report highlighted the wide variations in services available for patients across the country with families reporting that they were either having to fly to another part of the country or to face having to drive for very long distances to be able to access the specialist support and treatment from one of the few neuromuscular specialist services available in the country. This report identified both the positive and negative experiences of children and adults in respect of the quality of care that they had received. This was an important and influential report which focussed on the needs of one population group, however as children and adults with Muscular Dystrophy are continuing to live much longer into their adult lives than previously, the needs for specialist services will continue to grow rather than diminish.

The Royal College of Nursing (2013) produced a policy document aimed at nursing staff which identified the specific needs of adolescent patients as they approach the age of leaving children's services and moving to adult services. It built on the previous documents in identifying the importance of the transition arrangements being planned and prepared for, with the young person and their family being at the centre of the planning arrangements. This document identified the importance of collaboration with the appropriate adult services, with an identified member of staff being the co-ordinator for the planning and communication between services, to ensure that each young person has a good experience of their transition to adult services and has the opportunity to meet healthcare professionals in the new service prior to their health care being handed over. Case studies of good and poor practice are cited to enable nursing staff to understand the risks and the benefits to each young person of a well-planned and well managed transition. This document was crucial for driving forwards

initiatives to create nursing posts for specialist transition nurses which are not yet available for all young people.

The Children and Families Act (2014) was an important piece of English legislation which focused on the needs for children who have a special educational need or a disability, and children who were going through the process of being adopted. This legislation mandated that transition could extend up to the age of 25 years if needed and ensured that children and their families would have a voice in the decisions that were made concerning their care. This legislation paved the way for a single assessment, the Education, Health and Care plan (EHC), to be completed, which would cover the social care, health and education needs for each child and would ensure that parents were given more help and support when caring for a disabled child. This has transformed the experiences of parents and young people and has reduced the repetition of assessments which families had previously experienced. However, currently this only applies in England and it would be beneficial if all children and young people in the UK were covered by this legislation.

The Care Quality Commission (2014) was commissioned by the government to look at the progress that had been made in the transition arrangements for 40,000 children and young people with continuing and complex health conditions. They reported that despite the earlier guidance, only 50% of the 180 participants interviewed had received support from a health care professional prior to their transition. It echoed the recommendations from each of the previous guidance for the development of transition clinics and ensuring that each young person and their families are allocated a healthcare professional to take the lead in coordinating and supporting young people in their journey to adult services. The findings indicated that improvements were still needed to ensure that parents and young people received the support that they need in planning and preparing for the future.

A project by NHS South East Coast Strategic Clinical Networks (2015) looked at the transition needs of young people living in the South East of England. It endorsed the recommendations from previous documents for transition for each young person to be planned and purposeful, with specific transition pathways identified for young people with epilepsy (two separate pathways for those who do or do not have complex needs), asthma or diabetes. These pathways identify the need for family support for the young person as they move towards adulthood and identified the need in each pathway for transitions clinics to be

established as one step to smooth the transition path from child to adult services. This has financial and resource implications for the providers of healthcare services, but once in place, improves the experiences for young people and their parents.

Together for Short Lives is a UK charity who provide information and guidance for parents who have children with life-limiting or life-threatening conditions. This important charity provides information and support for families and guidelines and information for professionals who are providing care for children and young people receiving palliative care. In 2015, they provided guidelines on jointly commissioning palliative care for children and young people from birth to 25 years including short breaks (Together for Short Lives 2015). In this guidance they identified the need for health and wellbeing boards, local authorities, and clinical commissioning groups to jointly commission palliative care services for children and young people. This also included recommendations about transition to adult services and has developed a *"pentagon of support"* (page 37), as a visual representation of the support needed by young people and their families which encompasses the domains of housing, work/life balance/social care, healthcare, and education but at all times keeping the young person at the centre of the planning and decision making. This enables a holistic assessment to be completed on the needs of each young person.

The NICE (2016) guidelines again highlighted the needs of parents and their children, for support and planning to be done during the months and years leading up to their transition to adult services. The guidelines advised that this process of preparation should begin for each young person at the age of 13 or 14 years which would allow for the necessary preparation to be performed in a timely and age-appropriate way. For the first time, the individualised timing of the transition preparation process considered the emotional and psychological readiness of the individual young person to make their transition to adult services. This guideline moves away from the traditional model of provision for children's services, whereby transition taking place at a set age, and instead advocated a move towards a more flexible, individualised, and holistic approach, appropriate for each individual young person. However, the traditional provision of services in health and social care is usually predicated on set inclusion and exclusion criteria of the ages at which an individual would meet the designated threshold for a service. Allowing young people to remain in a service beyond the age of 18 years requires the resources to be in place to enable ongoing support for some young people and their parents and families to be continued into early adulthood (Aldiss et al

2016). The reality for many families remains that their experiences of transition have not benefited from these guidelines, and whilst improvements have been made in some services with specialist transition staff being appointed to concentrate on supporting parents and young people with their transition journey, this does not reflect the experiences for all young people. Some hospital trusts have developed adolescent inpatient units which cater specifically for adolescents and young adults until their early to mid-twenties and staffed by both adult and children's trained staff. This allows a bridge between children's and adult services and allows young people and their parents to become prepared for new services for the future (Viner 2007, Viner 2018).

Each of the guidelines and policies outlined above, have been published over the past decade and each have brought the health and wellbeing of children and young people to prominence. The main objective of each of the policies has been to raise discussions concerning funding and service reorganisation to bring about benefits for children and young people. The guidelines and polices have identified the needs of parents and primary care givers and have recognised the stressors that families face as they care for a child or young person with a continuing healthcare need. However, the experiences of some young people and their parents' remains below what has been outlined in the guidelines and legislation and continues to be an unmet need for many families.

The issues concerning the transition of adolescents to adult services remains a key topic in health policies and continues to be an important area of research. Fergan et al (2014) reviewed 18 qualitative studies concerning the experiences of young people moving to adult services. Young people felt that they had not been adequately prepared for the transition to a new service; and were concerned about having to adjust to developing new relationships with staff and achieving responsibility for their own health. The findings identified the importance of health professionals acknowledging the vulnerable stage of physical and emotional development that teenagers are going through in their adolescent years, the importance of partnership working between young people and health professionals, and health staff considering the vulnerability and feelings of loss that young people may experience as they move between services. However, provision for adolescent health care is different in the USA and Canada which will be considered in the next section.

1.4 Specialist healthcare provision for adolescent patients with neuromuscular disorders

In the United States of America (USA) and Canada, adolescent health is a specific medical speciality and provides clinicians who have the clinical skills and expertise to work with and care specifically for teenagers. In the UK there are some clinicians who also specialise in the health and wellbeing of young people, and these staff have been at the forefront of developing inpatient adolescent units which have been designed to meet the physical, emotional and psychological wellbeing of young people when inpatient care is required (Aynsley- Green et al 2000, Viner 2008, Yassaee et al 2018, Viner 2018). However, these specialist adolescent services are few and far between and most commonly are in place for specific conditions such as teenage cancer units within specialist tertiary units. The emphasis within specialist adolescent provision is the acknowledgment that young people need a service that is different from the toys and noise of a children's ward and yet has the increased support not readily available in adult wards. Adolescent units act as a bridge between the familiarity of children's services and the increased expectations for the young person to become more autonomous in adult services, whilst at the same time providing a more relaxed and flexible environment than is available on adult wards (Viner 2007). Where adolescent units are available, they have received positive appraisals from the young people who have used them (Viner 2007). The experienced staff provide support for young people and their parents to prepare them for their transition to adult services by encouraging young people to be more actively involved in the decisions about their care and to develop their confidence in communicating their needs and wishes with medical staff (Viner 2008). From the literature concerning young people's transition to adult services, one of the main issues identified concerned the levels of long-term support needed for both young people and their parents. Parents' anxieties were centered on the appropriate level and provision of care packages that would be needed to meet the specific requirement for their child, when continuing care and supervision into adulthood was going to be required to ensure that the complexities of their child's needs were met throughout their lifespan (Thackeray et al 2015).

1.5 Disability legislation

Disability rights and legislation has significantly changed for the better within the UK over the past decades, with legislation (The Mental Capacity Act 2005, Convention on the rights of persons with disabilities 2009, The Equality Act 2010, The Children and Family Act 2014,

and the Welfare Reform and Work Act 2016), which are promoting and protecting the rights of individuals with disabilities to live, and work within their communities. Amongst the many issues that have been identified by research are the experiences of stigma that have been identified by individuals with neuromuscular conditions and members of their families.

The issues concerning stigma worldwide are complex and vary from between different countries, ethnicities, and religions. Within society, stigma resulting from the attitudes, knowledge, and behaviour of others towards an individual or group who are different from the accepted norm can cause isolation, hurt, and fear to those to whom the stigmatisation is directed focussed at. Stigma may be because of a lack of knowledge concerning a condition or social group which may need to be addressed by providing the information required to address the knowledge gap or legislation to protect those at risk from being targeted (Thornicroft et al 2007) and to provide the support needed for young people with neuromuscular conditions to enable them to access Further and Higher Education and work opportunities. Experiencing stigma can have long term emotional and psychological consequences for the individuals affected and their close family members or their carers [1] (Bombard et al 2009). Employers have a responsibility to ensure that their practices are inclusive and non-discriminatory and that their recruitment and employment practices comply with the law. Employers also must ensure that their employees receive training on stigma in the workplace and that they make reasonable adjustments to allow employees with disabilities to be able to undertake their work responsibilities with the support that they need in place.

The complexities of living with a neurological disorder where the symptoms change and deteriorate over time and new symptoms emerge, may require repeated re-adjustment by the young person and their parents or carers which can be challenging emotionally and physically as they come to terms with learning to use new equipment and to develop their own expertise with managing their changing circumstances (Kinali et al 2006, Wagner et al 2008, Mah et al 2008). Young people may need time away from work to attend health related appointments and may need periods of time away from the workplace when their health deteriorates. Each of the young people who participated in this study had clear aspirations for their careers but were realistic as to how their disabilities might potentially impact on their employment goals:

[1] The religious perspectives are explored in greater detail in chapter 3.

"*I doubt it will be easy to find a good employer who will understand"* (Elizabeth aged 21 years).

Young people with neurological impairments can experience a myriad of other conditions. Each condition manifests itself with specific symptoms and treatment regime necessary to manage them and can require input from specialist clinicians and therapists which involves different clinics appointments and investigations. The different conditions that the young people in the study were living with included: Duchenne muscular dystrophy, autism, epilepsy, cerebral palsy, visual impairment, significant hearing loss, urinary incontinence, obsessive compulsive disorder, enteral feeding via a gastrostomy tube[2], and respiratory impairments.

1.6 Defining neuromuscular disorders.

In the following section I introduce neuromuscular disorders, how they are diagnosed and briefly discuss the types of disorders that were relevant to this study (rather than using a glossary of terms). Neuromuscular disorders are a large and complex group of conditions that affect any part of nerves or muscles and can be either genetic or non-genetic in origin and are either progressive or non-progressive in their trajectory (Muscular Dystrophy Canada 2016). There are 162 identified conditions that are classified as neuromuscular disorders, and these have been divided into the subcategories of skeletal muscle disorders: neuromuscular joint disorders, peripheral nerve disorders, motor neurone disorders and genetically determined ataxias (abnormal movements of the body) (Muscular Dystrophy Canada 2016). Most currently available epidemiological data from the Muscular Dystrophy charity has demonstrated that overall, within the UK, the number of children born with neuromuscular conditions is continuing to increase. It has been estimated that there are over 70,000 adults and children in the UK with a diagnosis of one of 60 muscle wasting conditions with approximately 100 boys born in the UK each year with Duchenne Muscular Dystrophy (Muscular Dystrophy 2015) and worldwide it is estimated that inherited neuromuscular conditions affect one in 3,500 children (Strehle 2009). The challenge for healthcare commissioners and providers in the UK, however, concerns the lack of a national database to confirm the numbers of children and adults living in the UK with a neuromuscular condition

[2] A gastrostomy tube is an artificial feeding tube which is inserted into the stomach via a surgically made stoma through the abdominal wall. This enables nutrition to be delivered into the stomach when the patient is unable to meet their own nutritional needs orally.

(Neuromuscular Campaign 2010). It is thought that some of the known increase in incidence attributable to sanguineous marriages[3], with new diagnoses in young children increasing in some areas of the country where there are more marriages between parents who are first cousins than in others. Marriage between blood relatives increases the risks of a faulty gene being present; for example, if the condition is an X linked recessive disorder such as is the case with Duchenne Muscular Dystrophy, both parents need to be carriers of the faulty gene and if this gene is present within a family, related adult members have an increased chance of passing this condition to their unborn child, compared to unrelated parents (Finkler et al 2003, Fraser et al 2011).

There continues to be evidence that parents of some specific religious and ethnic backgrounds are unaware of their risks in relation to the transmission of autosomal recessive conditions (James et al 2006, Chapple et al 1995, Finkler et al 2003). Pre-conceptual care provides potential parents with the opportunity to know about and to be able to access genetic advice and possible screening if required. The provision of this health screening is important for their individual decision making in relation to family planning; in order that their own potential risks in passing on a genetic condition to an unborn child can be known and informed decisions made. Knowledge concerning genetic inheritance and the associated risks of transmission may not only have long term consequences for the parents and for the future of their children but also has significant implications for future health service provision. (Fraser et al 2011) This includes the need for increased children's hospice provision and increased availability of geneticists to diagnose and support families where children have been born with genetic conditions (Botkin et al 2015).

Progressive and degenerative neuromuscular disorders (some of which form the group of diseases that come under the wider umbrella term of Muscular Dystrophies) are genetic and over time cause a progressive loss of muscular functions which result in a gradual deterioration in gross motor skills such as sitting unsupported and walking unaided, and fine motor skills such as pincer finger movements which enable a child to tie their shoe laces, do up buttons on clothing and hold a pencil (Strehle 2009, Camfield et al 2011, Fraser et al 2011, Cerebra 2012). These conditions increase in severity over a prolonged number of years and ultimately lead to a shortened life span and death, usually from respiratory failure (Strehle

[3] Sanguineous marriages are where the marriage partners are first cousins.

2009, Camfield et al 2011, Fraser et al 2011, Cerebra 2012). Non-progressive neuromuscular disorders, such as Cerebral Palsy where the initial injury to the brain before birth, during delivery or after birth, varies widely in the severity and the resulting impact that is had on each child depending on which areas of the brain have been damaged and which limbs are affected. Some children and adults who are less severely affected with Cerebral Palsy have a normal life expectancy and can achieve the individual educational, sports career or independent living goals that they have set for themselves, although they may need some additional support particularly with their mobility. However, children with more severe impairments may have a shortened lifespan due to repeated chest infections, and significantly reduced mobility. These deteriorations in their physical health often results in young people needing to move from walking independently, to using a wheelchair for some or all of the time. Some children and young people may go on to develop another added complication of the development of epilepsy resulting in seizures due to the areas of neurological damage that have occurred in their brains before, during or after their birth (Hutton et al 2006).

1.6.1 The diagnosis of neuromuscular disorders

The timing of a diagnosis of a neuromuscular condition varies from child to child and depends on the disorder and the presenting symptoms. Some neuromuscular conditions are diagnosed soon after birth when the infant presents with poor muscle tone and difficulties with nutritive sucking and swallowing (Menezes et al 2012, Guglieri et al 2015). Other children may be asymptomatic initially but gradually start to demonstrate delayed or impaired neurological development (Menezes et al 2010, Papa et al 2016). Children, usually boys but in rare cases girls, who are carriers of one affected X chromosome and may exhibit some signs of muscle weakness (Andrews and Wahl 2018), who are affected by Duchenne Muscular Dystrophy (DMD), exhibit normal development in the early months of life with the associated acquisition of the expected milestone development such as sitting, pulling to stand, crawling and learning to walk. In the weeks and months of early childhood, children affected with DMD start to lose the large and fine motor skills that they had previously acquired (Glasper 2008, Papa et al 2016,). For some children, the development of symptoms that lead to the establishment and confirmation of a diagnosis may be lengthy, protracted and extremely stressful and may require travelling long distances to specialist centres to enable a diagnosis to be confirmed (Poysky et al 2009, Menezes 2010).

The assessment and investigations required to establish a diagnosis will vary depending on the disease being screened for. The first stage for all assessments begins with a thorough antenatal and postnatal history being taken, together with a detailed physical examination performed by an experienced clinician. Blood samples to enable gene testing are often required to confirm or to rule out a genetic cause of the presenting symptoms in a child. In addition, electromyography (EMG) and nerve conduction studies can be used by doctors to confirm the diagnosis. A muscle biopsy may or may not be required to confirm the suspected diagnosis by examining the muscle fibres in minute detail and in addition a nerve biopsy may be useful in confirming a diagnosis for those children whose presenting clinical picture remains undiagnosed (Laing 2012, Menezes et al 2012,). These investigations are invasive and take time to be completed and it can be very stressful for the parents and family as they wait for test results to become available.

The diagnosis of a genetically inherited condition can have significant psychological impacts with widespread ramifications for the child, the parents, and the wider family. For some families there may be tensions and the apportioning of blame when the condition is the result of an autosomal dominant genetic disorder, which may add significantly to any feelings of guilt and responsibility for either or both of the biological parents. For families it is usually a relief to have the confirmation of a diagnosis, which in turn may enable the family to access specific help and support from charities or organisations which specialise in their specific condition. James et al (2006) explored the experiences of 151 families with X linked and autosomal recessive disorders in the USA. The findings from this study concurred with the findings from other research studies. Families may require the expertise of genetic specialists and counsellors to help them to come to terms both with the diagnosis and the feelings of guilt and blame that parents may experience because of the diagnosis; together with the need to support parents to develop their knowledge and understanding in relation to reproductive risk (Chapple et al 1995, James et al 2006, Bombard et al 2008, Bushby et al 2010). The psychological impact of having a diagnosis needs to be understood by health professionals, as the parents come to terms with the caring responsibilities that disruption to the future that they had envisaged for themselves as parents and for their child.

1.6.2 Duchenne Muscular Dystrophy

Duchenne Muscular Dystrophy (DMD) is an X-linked recessive genetic condition carried on the X chromosome. In two thirds of the diagnosed cases, the different gene is inherited from

the mother, whilst in one third of cases it is a new gene mutation. It is a severe form of muscular dystrophy. The disease which most commonly occurs in boys, causes severe progressive muscle weaknesses and muscle wasting due to the lack of an essential protein called dystrophin which is essential for maintaining muscle fibres. The muscle weakness and muscle wasting are most commonly diagnosed in young boys between the ages of 2 and 4 years of age, when children who have attained their early gross motor skills or sitting unsupported, pulling to stand and walking independently, start to become unsteady and have difficulty in pulling up to stand and frequently lose their balance. The disease progresses rapidly with most boys becoming wheelchair dependent before their teenage years. Approximately 100 boys are diagnosed with DMD in the UK each year (Guglieri et al 2015, Muscular Dystrophy UK 2019). Due to advances in treatment options and current clinical trials, the life expectancy of men with DMD is continuing to increase, with advances in gene therapy giving future hope for the boys and men living with DMD (Guglieri et al 2015, Dowling et al Cohn 2017, Muscular Dystrophy UK 2019).

1.6.3 Cerebral Palsy

Cerebral Palsy is a non-genetic and non-progressive brain disorder which occurs due an injury to the brain taking place in the antenatal or postnatal period of a child's life. The extent of the brain injury which has been sustained will determine the extent of the disability that the infant or child is diagnosed with but can range from mild weakness in one limb to four limb quadriplegias, whereby the child's movement is severely affected, and they are wheelchair dependent. There are approximately 1,700 new cases of cerebral palsy in children diagnosed each year (Cerebral Palsy UK 2019). Children with cerebral palsy may also suffer from epilepsy, visual impairments, speech difficulties and urinary incontinence. The damage sustained is lifelong but symptoms of muscle weakness or muscle stiffness or muscle spasms can be improved by physiotherapy, hydrotherapy, and the use of splints to support the affected limb and prevent increased stiffness or deformities from developing. Speech and language impairment and difficulties with feeding orally can be improved with support and advice from Speech and Language Therapists (NICE 2019).

1.6.4 Autism

Autism spectrum disorder (ASD) is a wide-ranging developmental disorder where children display difficulties with speech and language, socialisation and play and communication with others. Young children may display difficulties with maintaining eye contact with their

parents or siblings and have significantly delayed speech and language development. The severity of the difficulties that the child is experiencing varies widely from one child to another. Some children attend mainstream schools with additional support, whilst other children who are more severely affected will be supported in their education through special needs schools. Children diagnosed as being on the autistic spectrum may have difficulties in regulating their emotions and expressing their needs and feelings verbally (National Autistic Society 2018).

Although there is no known cause of autism, some families have more than one child who is diagnosed as being on the autistic spectrum and each child may have different needs which can be complex for the parents to manage. Parenting courses run by children's centres can be helpful to give parents techniques and strategies to support their child and to establish a daily routine for the child and the family.

Children diagnosed as being on the autistic spectrum often have disrupted sleep patterns and either have difficulties in settling to sleep or wake very early in the mornings. Medication such as Melatonin can be helpful in enabling the child to settle to sleep in the evening, and a good bedtime routine and good sleep hygiene can also be helpful and supportive for this routine (Goldman et al 2014).

1.6.5 Epilepsy

Epilepsy is a neurological condition whereby individuals experience epileptic seizures. Over 500,000 people in the UK have a diagnosis of epilepsy (Epilepsy Society 2016). There are many different types of seizures with some children experiencing more than one seizure type. Treatments for epilepsy include anti-epileptic medication, which can cause drowsiness and weight gain. Some children with epilepsy are sensitive to flashing lights or strobe lights and find that these types of lighting can induce seizures. If seizures are occurring from one specific area of the brain, for some children, neurosurgery to remove the affected area of the brain can be an option. For some children, a vagus stimulation device (VNS) can be inserted into their chest, which sends electrical signals to the child's brain via the vagus nerve in their neck which can reduce the number and the intensity of the seizures. If a child or an adult who has a vagus stimulation device in situ has a seizure, a special magnet can be passed over the VNS which can reduce the duration of the seizure.

It is important that the assessment of each young person is carried out holistically and for parents and professionals to understand both the developmental and cognitive stage for each young person with a neurological condition. Adjustments to the advice and provision of services and levels of support needed will need to be tailored to meet individual needs.

In the next section I discuss adolescent development and the challenges for young people with neuromuscular impairments in achieving physical independence from their parents and immediate family.

1.7 Child Development

The purpose and importance of developmental assessments is explained by Bellman et al (2013). All children develop at slightly different rates, however there are accepted normal ranges for the attainment of speech and language acquisition and the development of fine and gross motor and play skills (Sheridan 2016). Speech and language development are dependent on the child's ability to hear, and therefore the new-born hearing screening programme, assesses new-born infants for their abilities to respond to sound appropriately (Sheridan 2016). If hearing loss is present further investigations will be performed to identify the cause of the hearing loss and ascertain whether interventions such as a cochlea implant might be appropriate or whether the parents want any intervention to be done to restore hearing for their child to enable speech to develop. One of the young people in the study who had a significant hearing impairment, identified her need for support with attending university lectures, and was aware that without the support, her learning experiences would have been significantly reduced.

Health Visitors and General Practitioners are the key health professionals in the child's early life, who monitor and assess each child's development during the first 5 years of their lives. If any deviation from the normal expected development is identified, the child is referred to a Community Paediatrician for further investigation and assessment and referral to specialist services as required.

Having a definition of adolescence is useful for providing a clear definition of what is understood by the term "adolescence". Having a clear definition provides the parameters by which clinicians can make sense of the term and this can then be used consistently in all

documentation. The WHO defined adolescence *"as the period in human growth and development that occurs after childhood and before adulthood, from ages 10 to19 years"* (WHO 2016). Whilst it is well known that during this long period in human development huge biological and psychological changes occur both in the brain and the body, the emotional, psychological, religious and cultural aspects of a young person's life are also crucial components of a young person's development and can strongly influence their psychological development, including their individual development of relationships and friendships, together with their development of their own identity which includes their sexuality (Kaplan 2004, Viner et al 2005, Coleman 2011). Each child and young person need to be assessed holistically by health and social care professionals to ensure that each aspect of their physical and mental health have been assessed and taken into consideration, and that young people are active participants in the decisions that are being made for them and about them (National Service Framework for Children, Young People and Maternity Services 2004, CQC 2014, NICE 2016).

These policies identified the importance of the different needs of young people with continuing healthcare needs being met across the UK, to give children the best chances to maximise their potential with the support that they need from specialist therapists and services. However, inequalities in service provision continues to exist, with long waiting lists for assessments and treatments to begin (Hillis et al 2016, Rosen-Reynoso et al 2016).

1.8 Adolescent development

The expected course of development for teenagers in western cultures is that young people will achieve the expected stages of adulthood, which includes the completion of their education, find employment, and eventually move away from the parental home to live independently with autonomy over their choices and decisions and establishing intimate relationships with another person (Christie et al 2005). However, when a young person has a chronic illness that requires ongoing care and intervention; these choices and ambitions may be hampered by the need to have ongoing support for activities of daily living, which due to their illness they are unable to perform for themselves and need the ongoing support of paid carers or family members (Akre et al 2015). For young people with progressive degenerative conditions, their dependency on others will continue to increase rather than decrease as their muscle function deteriorates, however young people can be supported to move to adapted

living accommodation and have the support of carers, to help them to develop independence, although for this to happen, the funding to employ support staff needs to be in place with a sustained level of funding to enable the required level of support to be continued for the duration of the young person's life.

For parents or carer givers the realisation that the ongoing complex needs of their child is going to pose limitations on their child's desire and ambitions for independence can be difficult to cope with. As parents grow older, the practicalities of providing long term care for their child may be challenging as they themselves become less physically able. This can cause some parents to develop increasing levels of stress and depression and to feel increasingly isolated (Akre et al 2015). Knowing when and how to "let go" as a parent and allow young people to make their own decisions and accept the consequences of those decisions are dilemmas faced by most parents. Consequently, to support both young people and their parents or carers in this developmental progression, the transition to adult services and the age and the stage at which this occurs needs careful planning, preparation and organisation to ensure that the process is anticipated and goes as smoothly as possible, with a specialist transitions coordinator available for each young person to guide them and their parents or primary caregiver through the move from one service to another (McDonagh 2007, Bushby et al 2010, Camfield et al 2011, Akre et al 2015).

1.9 Thesis chapters overview

In chapter 2 the literature review will be presented. At the start of the chapter the review strategies and processes which were used for this research study are shown, with literature concerning the impact for a child and their parents of having a neuromuscular condition, and the physical, emotional, and psychological impacts than can affect children and their parents explored. In the second section of the chapter, the literature related to the experiences of a young person having a diagnosis of epilepsy and the impact for parents of delivering enteral feeding for their children is presented and discussed. This is followed by studies concerned with the experiences of men and boys of growing up with Duchenne muscular dystrophy; the experiences of young people of transition to adult services; and role of parents becoming "expert parents", when they are caring for a child with complex needs. In the final section of this chapter, the literature concerning the sexual health of young people with physical

disabilities is presented and this chapter is concluded with a review of current policies related to the needs of young people with long term conditions.

In chapter 3 I present the methodology and methods that have guided the planning and delivery of this research. The chapter includes a discussion on the epistemological and ontological position that I have adopted in this research study. I present the history of case study methodology and explain why this methodological approach was adopted for this study, what recruitment strategies which I used to advertise for and recruit participants who met the inclusion and exclusion criteria for the study and the challenges which arose during the recruitment phase of the study and explain how these were overcome. The considerable ethical challenges that I encounter when undertaking the study are discussed at length. Detailed data collection strategies are presented followed by data analysis processes which I used to analyse the data from the interviews and the questionnaires. The steps that have been undertaken to demonstrate reflexivity and rigour in this research are presented, with discussion as to the influence of my previous professional roles and experiences of working as a nurse and how this may have influenced my understanding of the data. In the final section of this chapter, I present the three frameworks used during data analysis.

Chapter 4 presents the findings chapter in which the eight cases with the ten participants are be presented individually, with a sequential pattern of the earliest recollections of the parents or the young person presented first, followed by their experiences of the early childhood years and then for the young people, their experiences of going to university and how their neuromuscular conditions has affected their experiences of being an undergraduate student. In this chapter a table giving the biographical details of each of the participants is presented. The two pivotal cases where both a mother and her young adult child were participants, which gives a unique perspective from growing up with and supporting a child living with a neuromuscular condition.

In the second half of this chapter, a cross-case analysis is presented. The sequence of becoming the mother of a disabled child, becoming a caregiver and an expert parent, holding onto the child and mutual dependency and lastly the child becoming a young adult has been followed to present and discuss the data gained from each of the participants.

In chapter 5, I draw the thesis to a close by considering my contribution to knowledge and to the current conversation of the experiences of the mothers and their children of growing up with a neuromuscular impairment. The chapter includes recommendations for practice and current policies; recommendations for future research; study limitations; plan for the dissemination of the research findings; and reflections of my experiences of undertaking this research.

1.10 Chapter summary

In this chapter I have introduced this research by outlining the reasons why I chose to undertake this study. The key current legislation and guidance concerning the health and wellbeing of children and young people with complex and continuing healthcare needs have been identified and discussed along with disability legislation. The chapter concludes with an overview of neuromuscular conditions including diagnosis processes that are currently used and an overview of the neuromuscular conditions relevant to this study followed by an introduction to child and adolescence development.

Chapter 2 Literature review

2.1 Introduction

In this chapter I first explain my review strategies in relation to the literature that is presented here. I critically examine the literature concerning adolescent development and the experiences of young people with complex and chronic illness and the impact that this has on their families. Literature on the transition experiences from child to adult services for this population is examined. The review also addresses some additional health issues that young people with neuromuscular disorders deal with daily including epilepsy, enteral feeding, intermittent urinary catheterisation and becoming wheelchair users. Literature on experiences of parents of children with neuromuscular disorders is also discussed.

2.2 Review strategies and history

The review of the literature was wide ranging and took place over the entire journey of the PhD study. Review strategies and processes were modified over time from the research proposal development, over the period of undertaking the research and considering the findings. The initial searches were used to develop my research proposal and were focused, at that time, primarily on transition experiences of young people with neuromuscular disorders. The review became data driven when analysis started (see methods chapter for detailed analysis stages). As the study progressed and particularly during data analysis, searches were carried out on very specific subjects for example, different types of neurological conditions and the implications of these on parents their families and the young people themselves, birth trauma, achieving urinary continence through intermittent self-catheterisation, living with epilepsy, parental experiences of having a child diagnosed with autism, children's experiences of using a wheelchair, the experiences of parents and young people of enteral feeding, sexual health for young people with neuromuscular conditions, the role of children's hospices and expert parents. During this time literature related to the theories I used during analysis was also retrieved and scrutinised.

Searches were carried out using CINAHL, Medline, PubMed, and Ebsco databases at different stages between February 2012 and August 2019. Multiple search terms were used to search for literature and changed according to which specific area was being considered, at the different stages of the research, particularly during data analysis. During the analysis stage – when examining the data through theoretical lenses – I searched for literature where

identity theory, Transitions Theory (Schlossberg 1981), the ABC-X theory of family stress and coping (Hill 1949), had been used in qualitative research concerning the experiences of children and young people who had chronic illnesses.

There is limited research currently available that explores the needs and wishes of young people with neuromuscular impairments from their own perspectives and most of the research literature comprises quantitative studies of large cohorts. This lack of qualitative studies may in part be due to the challenges faced by both young people and researchers of wanting to include young people in research (UNICEF 2013, National Children's Bureau 2016). However, the body of research where researchers are developing expertise in conducting research with children under the age of 18 years is continuing to increase, and therefore through the publication of more research studies involving children and young people, their voices are reaching a wider audience in health and social care. Having the findings from research conducted with children and teenagers is imperative for influencing the design of services for young people in primary and secondary care (Kirk 2007, Shaw et al 2011, Twycross 2012, and Cheetham et al 2013). There are experienced researchers who are working collaboratively with young people as co-researchers on some projects (Moules et al 2012). This type of participatory research develops the knowledge and skills of young people and enables them to be active participants in the design of future research projects.

The guidelines from the National Children's Bureau and research from Kirk (2007), Shaw et al (2011), Cheetham et al (2013) and Twycross (2012), alert researchers to the specific needs of children and young people, when undertaking research, which include the importance of informed consent, confidentiality and power relations between the researcher and the young person or child as the participants. There is consensus from this research that children and young people are entitled to have their voices heard and should be facilitated in their inclusion in research. A UK study by Fraser et al (2011) confirmed the increase in referral rates to a children's hospice in Yorkshire of children and young people with progressive degenerative neuromuscular conditions. For example, the findings from the study identified that survival rates of young men with Duchenne Muscular Dystrophy have significantly increased over recent years, which highlighted the need for services to be available for young people living into their adult years with their life limiting condition. These studies confirm that the needs for young people with neuromuscular conditions will continue to increase, and

that their needs in childhood and adulthood will need to be met in the immediate and longer term.

2.3 Impact of having a child with a diagnosis of a neuromuscular condition.

Following the assessment and tests required during the diagnosis process; the confirmation of a genetically inherited disease may mean that parents must come to terms with the news that they have more than one child affected with the same condition and have to watch the trajectory of the disease process unfold with each of their children. This process can be devastating for the child and the family as they come to terms with the diagnosis and prognosis of an identified life-limiting condition and for some families can cause immense strain on the parent's relationship, their religious faith, and relationships within the wider family (Fraser et al 2011, Moss et al 2012). Accurate diagnosis through genetic testing is not only important for the affected child to confirm the diagnosis but also for the parents to be able to make informed decisions with regards to any future planned conceptions (Bombard et al 2009, Menezes et 2012, Papa et al 2016).

The significance and impact of a diagnosis of a neuromuscular condition is ongoing throughout the lifespan of the person affected and often requires extensive adjustment and support both emotionally and psychologically (Poysky et al 2009). Parents and extended family members need time and support to come to terms with the established diagnosis together with the implications for the child's immediate and longer-term future, as the child will require the provision of specialist education, health, and respite service to be available. Parents will commonly go through a process of bereavement as they come to terms with a different expectation of their child's life and future from the one that they had anticipated and planned for during the pregnancy (Chapple et al 1995, Finkler et al 2003, Poysky et al 2009, Moss et al 2012). As the different diseases develop, the gradual deterioration of muscle function leads to the loss of previously attained muscular functions and skills such as sitting unsupported, pulling to stand, and walking unaided and have a life-changing effect for the child and their family (Mah 2008, Hodges et al 2010,). Although many affected young people, their families and clinicians take a keen interest in the ongoing advances in medical science and in particular gene therapy which may offer enhanced symptom management or halt the disease progression, some young people may be in denial and not want to think ahead to the future or may not want to take the risks of proposed high risk interventions such as

bone marrow transplantation which carry an uncertain success rate and may lead to death (Guglieri et al 2015). Currently there are different scientific research projects that are underway worldwide investigating the replacement of the faulty gene or the development of a replacement for dystrophin in the case of Duchenne Muscular Dystrophy (Kazuki et al 2010, Guglieri et al 2015, Muscular Dystrophy UK 2016). Stem cell research is also offering hope for the future, however although some pre- clinical trials using animals showed promising results the future, the longer-term results and efficacy in humans remains uncertain (Guglieri et al 2015).

Although some families may choose to accept the diagnosis and may not want to have any further investigations; other families do seek answers for symptom management or treatment options for their child. They may want explanations together with a prognosis for their child's diagnosis (Finkler et al 2003, James et al 2006). For some children, the process of securing and confirming a diagnosis can be protracted with the investigations required taking long periods of time for the results to be available which can heighten fear and anxiety for parents and lead to significant stress and apprehension during the period of waiting for the diagnosis to be confirmed (Poysky et al 2009, Laing 2012,). Once the test results are available, some results confirm the news that the infant or young child has a rapidly progressive neurological condition such as the more severe form of Spinal Muscular Atrophy or Juvenile Battens Disease; which will result in a significantly shortened life expectancy and early death (Qianet al 2015). Other children and teenagers may have years of life on a palliative care journey with treatments, interventions, and inevitable deterioration still ahead of them. Parents may have to come to terms with and have to cope with more than one child in the same family being affected by a specific genetic condition with all the resulting adjustment and acceptance that will be required, with many parents describing their adjustment of expectations and future hopes and plans for their child. If the child becomes a wheelchair user, adaptations to their home may need to be made or the family may need to be rehoused or purchase a suitable house that can be adapted to accommodate a wheelchair and additional equipment such as a hoist for safely moving their child between the bed and their chair or helping them with meeting their personal hygiene needs (Finkler al 2003, James et al 2006).

The rise in incidences of confirmed diagnoses of some neuromuscular conditions, together with the known increase in survival and life expectancy rates due to advances in technology and pharmacology, have resulted in the recognition and acknowledgement of the need for

funding and providing appropriate and adequate health and social care services to meet the specific needs of this client group. The call for further research to provide treatment options for individuals with neuromuscular impairments continues to be advocated (Strehle 2009, Muscular Dystrophy Campaign 2010).

The above studies offer a global perspective of the complex multi-factorial challenges that parents face when searching for an explanation for their child's neuromuscular impairment. When a genetic cause for the child's neuromuscular difficulties is being considered, there can be a protracted delay in investigations being performed and results becoming available. The importance of the roles played by geneticists and clinical staff in supporting parents to help each child in the family to adjust to having a sibling with a neuromuscular impairment are presented by Chapple et al (1995) and Poysky et al (2009). These qualitative studies by Chapple et al, Finkler et al and Poysky et al, identify the importance of the parents being supported in helping each child in the family to be included and for the whole family to adjust to the unexpected reality of having a child with a neuromuscular condition. The findings from these studies included raising awareness for professionals of the complexities for families adjusting to the changes needed in moving forwards as a family unit. In addition, Finkler et al and James et al acknowledged the feelings of guilt, blame and stigma which parents of children with a genetic cause for their neuromuscular impairment had reported, and who needed support to address the impact that the diagnosis had had for the parents and the families. These studies have shown that parents need ongoing support from the time of diagnosis to accept and adjust to caring for a child with a neuromuscular condition and to enable the child and any siblings to move forward as a family unit.

2.3.1 Treatments for neuromuscular conditions

There are currently advances in the pharmacological science that provides medication regimes to treat neurological and neuromuscular conditions. These include the use of glucocorticoids which are known to improve muscle function and reduce muscle degeneration, but long-term use of any steroids can have severe side effects which include excessive weight gain, immunosuppression which reduces the ability of the individual to fight infection and loss of bone density. Intrathecal Baclofen where the drug is injected directly into the cerebral spinal fluid, or deep brain stimulation using electrodes that are implanted deep within the brain, have the potential to extend and improve the quality of life

for those diagnosed with the condition (Guglieri et al 2015); however, the costs and availability and moral and ethical implications of genetic testing, including the issue of presumptive information concerning the genetic risks to other blood relatives or the potential legal challenges of being able to receive future treatments still need to be addressed (Botkin et al 2015). These new interventions which are used to manage the symptoms that occur for children with some neuromuscular conditions have been instrumental in the management of the debilitating symptoms such as tremors that can occur. These studies showed a reduction in the decline of muscle strength, which had the additional benefits of reducing the risk of the development of scoliosis. There was also an improvement in respiratory function in the young people receiving these treatments, however the side-effects of the long-term use of this group of drugs are still being evaluated (Bushby el 2010).

Each of the reviews by Guglieri and Bushby present an update of the advances that have been made in the treatment of Duchenne Muscular Dystrophy. The use of steroids can have a significant impact on the child's body image and emotional wellbeing but are an essential part of the treatment regime and are necessary to slow muscle deterioration. Parents and clinicians can face difficult decisions in seeking to balance the risks and the benefits of using steroids to treat children and young people with some forms of neuromuscular impairments. The importance of ongoing research for symptom management and improving the quality of life for children and young people with progressive neurodegenerative conditions from the international research community in this specialist field is evidenced, with research continuing to improve the outcomes for young people living with progressive degenerative neuromuscular conditions.

2.4 Experiences of caring for children with a neurological disorder.

There is an increasing evidence base of research which has looked at parental experiences of caring for children with a continuing health need. This research has captured some of the stress, issues and concerns that parents have to cope with day by day, which includes the anxiety of learning new skills and knowledge related to the child's condition and the uncertainties of future treatments, which cause significant levels of parental anxiety and fears for the future. (Fereday et al 2009, Benson 2010, Nelson et al 2015, Cuzzocrea et al 2016, Findler et al 2016, Hsiao 2018, Pedrón et al 2019). Some of the main issues related to the

psychological impact for parents of having a child with a neuromuscular disorder and the mental health issues for young people and their parents will now be considered.

2.4.1 Psychological impact of having a child or of being a young person with a neuromuscular condition.

The complex wider social and psychological issues that may arise for young people and immediate family members associated with the diagnosis of a neuromuscular condition have been reported in previous research (Bombard et al 2009, Botkin et al 2015). These studies have identified some of the complex and often hidden issues that arise for families and include the challenges and long-term implications of potential and actual experiences of genetic discrimination, for example the implications in finding a future partner or needing to declare their genetic inheritance in health screening or occupational health screening (Botkin, et al 2015). For professionals and for families, the ethical dilemmas of whether and when to screen for genetic differences and the consequences that can arise from advances in genetic testing need careful consideration and discussion prior to testing. Some of the advantages for pre-conceptual decision making include the knowledge that predictive testing for some genetic disorders such as Huntington disease can be carried out prior to a conception, or embryos can be screened for any genetic difference prior to implantation. However, the ethical implications of these interventions are strongly debated and contested by some religious or faith groups which believe that the unborn child has a right to life (Botkin et al 2015). These advances in genetic screening allow individuals who are known to have a family history of a disease to discover whether they are carriers of the gene that causes the condition and then to make informed decisions with regards to family planning and risks in terms of a planned pregnancy (Heller et al 2016). A positive result obtained from genetic screening may have implications for individuals in terms of higher premiums being charged for health insurance or increased difficulties in obtaining health insurance which may have unforeseen consequences for travel plans or employment opportunities and may have both a socio-economic and a psychological impact on the individual. Where children are being screened for a genetic condition, the legal and ethical implications of a child having the capacity to consent to the screening and the potential future implications of a positive result being obtained needs to be carefully considered (Botkin et al 2015). The rise in self-screening kits for a range of medical conditions that can be purchased over the internet need to be used with caution. A positive result may result in distress and anxiety which needs careful follow-

up by trained healthcare professionals, with further investigations often needed to confirm the diagnosis. During the process of counselling families, the ethical issues of who is responsible for informing or withholding information about genetic risk for relatives should not be overlooked and may be difficult when individuals do not realise that the diagnosis of a genetic disorder in a sibling or close family member may have direct implications for their own current and future health (Forman et al 2013, Howard et al 2013, Abul-Husn et al 2014, Botkin et al 2015).

These studies raise awareness of the significant needs that parents and carers can have for their own mental and physical wellbeing. The studies by Botkin et al (2015) and Bombard et al (2009,) identify the immediate and longer-term emotional and psychological implications for parents of having genetic testing performed on their child and themselves, when seeking to confirm the diagnosis for their child's neurological impairment. The findings included reports of discrimination in employment, which had a significantly detrimental impact on young people striving to achieve their career ambitions. Existing research identify both the need of genetic screening and the importance of long term follow up being made available in all areas of the country to support the child and the parents, as they adjust to the diagnosis, and as parents make pre-conceptual decisions about any future pregnancies. Howard et al (2013), raise concerns about home testing kits for genetic diagnosis and identify the needs for an increase in specialist genetic specialists to be able to support individuals who receive a positive result of a genetic condition from a home testing kit.

The research by Botkin (2015) reminds health professionals of the legal, ethical, and psychosocial implications of genetic testing for causes for neuromuscular impairments in children and adolescents and opens the discussion for the wider implications for genetic testing to be considered more widely. The world is continuing to change as advances in genetic medicine are developed, however, the implications of genetic testing on the emotional and psychological wellbeing of the individual family members requires skilled discussions to be undertaken by highly experienced health professionals, prior to genetic testing taking place.

2.4.2 Mental health in children and young people with chronic conditions

Most children and young people with neuromuscular conditions will become increasingly dependent on technology as their disease progresses and their health deteriorates. Due to the increase in muscle weakness, many children and young people with neuromuscular impairments will eventually need to use power assisted wheelchairs for their mobility and independence, and in addition may need invasive or non-invasive ventilation to support their respiratory function; both of which have a significant impact on their abilities to socialise, travel and obtain employment or continue into further or higher education. The inevitable and increasing dependency on parents and carers for their individual activities of daily living to be met for young people with progressive and degenerative neuromuscular disorders can have a negative effect on their mental health and overall wellbeing (Bushby et al 2010, Read et al 2010, Vuillerot et al 2010). There is known to be an increased incidence of depression and risk of suicide in young people with chronic and complex illnesses (Greydanus et al 2010), which directly impacts the quality of life that young people with neuromuscular conditions are experiencing, which is reflected in the psychosocial management needs of this client group. The mental health of children and young people are currently receiving increased media and political attention. The numbers of young people with mental distress is continuing to increase and the lack of suitable inpatient and outpatient Child and Mental Health Services (House of Commons 2019) are increasingly stretched in some areas of the country with long waiting times for support and treatments which has a negative effect on children and young people.

As their dependency on others increases and their individual level of independence decreases, it can be very difficult for some young people and their parents to adjust to the changing balance of caring needs and responsibilities with parents describing themselves as *"being the lifeline"* (Mah et al 2013 p 983) for their child or children, with parents feeling the need to take on the responsibility for facilitating effective communication between different health professionals to ensure that their children receive the high levels of care and intervention that they need (Mah 2008, Read et al et al 2010, Muscular Dystrophy 2010, Vuillerot et al 2010, Bushby et al 2010).

Quality of life indicators which includes the whole person in the context of their physical and mental health, their spiritual being, and feelings of physical, social and, community belonging. Each of these becoming are recognised and acknowledged as being one means by

which important measures of psychological wellbeing can be measured (Huismann, et al 2012). Research has identified that for parents and for young people with a neuromuscular condition, stress levels are very often high with ongoing adjustment by the individuals and their families being required as the trajectory of their illness progresses (Mah et al 2008, Poysky et al 2009). As the disease processes continue, for many children and young people with neuromuscular conditions, increasing interventions are required to sustain their life with parents and carers needing to develop medical skills and knowledge which are far beyond the expected remit of parents. The skills needed will vary but may include the ability to administer intravenous or rectal medication to control seizures, control pain or manage distressing symptoms such as excessive secretions and muscle spasms. As muscular function weakens, young people with neuromuscular impairment may develop an unstable swallow mechanism which significantly increases their risk of aspiration of food or fluids into their lungs leading to repeated chest infections and respiratory compromise. Following full assessment by a Speech and Language Therapist and a Dietician it may be deemed to be safer to move from an oral intake of food and fluids to the administration of enteral feeds via either a nasogastric, gastrostomy or a jejunostomy tube to meet the full nutritional requirements of their child (Mah et al 2008, Read et al 2010, however the acceptance of the change in the child's condition and the move to a different type of nutritional intake can be difficult for parents to adjust to as it is an additional daily reminder of the gradual deterioration in their child's health (Strehle 2009, Yang et al 2010).

There are strong links between mental health and wellbeing of children and young people growing up with a neuromuscular impairment, as well as the burden of responsibility for the parents to meet the complex daily needs of their children continues throughout the child's life. The literature review by Patel et al (2010) identified the increased suicide risk for adolescents with chronic illnesses and highlights the issues of depression and anxiety which can be present. The authors advocate for screening for depression to be undertaken on a regular basis to support the mental health of young people living with chronic illnesses. The clinical priority can be for health professionals to focus on the physical challenges for young people, whereas the emotional and mental wellbeing of these young people is also crucial. Services need to be available for children and young people to access when needed, which does not have the long waiting times which is currently the situation for Child and Adolescent Mental Health CAMHS services in many areas of the country. The quantitative study by Mah et al (2009) looked at the responses from 109 families where the parents had

completed either the Parenting Stress Index or the Index for Parents of Adolescents questionnaires and explored the stress of parents and their quality of life when caring for a child with a neuromuscular disease. The studies by Mah et al (2008) and Poysky et al 2009) focussed on the needs of parents and identified the levels of stress experienced by parents as they cared for a child with a progressive degenerative neuromuscular condition who required mechanical ventilation at home and raise awareness for services that parents also need specific, tailored, and on-going support to meet their own emotional and psychological needs.

2.4.3 Enteral feeding via a gastrostomy tube

Enteral feeding[4] via a surgically inserted gastrostomy tube[5] enables the delivery of medication, fluids, and balanced nutrition when a child or adult is unable to swallow or unable to meet their total nutritional needs orally. This method of artificial feeding may be either temporary or permanent. As the numbers of children and adults receiving enteral nutrition via a gastrostomy tube have continued to rise, the benefits and the challenges for the individual and their carers has become an area for research. The benefits of enteral nutrition are the assurance that the nutritional needs of a child or adult can be wholly met via an artificial means; however, research and clinical experience has demonstrated that some of the challenges of gastrostomy feeding include abdominal pain, diarrhoea, soreness and infections of the gastrostomy stoma site, leakage of gastric fluid or feed from the gastrostomy tube or from the stoma site which can stain and damage clothing (Fereday et al 2009, Craig 2013, Wu Wu et al 2013). The spontaneity of social events may be tempered by the need for parents or carers to find appropriate places for the enteral feed or medication to be delivered, which may cause reluctance to use a gastrostomy tube in public places or may be a barrier to the child being accepted into a pre-school nursery (Grzybowska-Chlebowczyk et al 2015). Prior to a child or young person being discharged from hospital with a gastrostomy tube in situ, parents or carers need to be trained and assessed as competent in its use and to know how to trouble shoot and deal appropriately with issues such as a blocked gastrostomy tube or leakage from the tube if it should occur (Pedrón-Giner et al 2013, Guidelines and Audit Implementation Network 2015, Nelson et al 2015).

[4] Enteral feeding is artificial feeding when the individual is unable to meet their nutritional needs orally.
[5] A gastrostomy tube is an artificial feeding tube which is surgically inserted into the stomach by a surgeon.

The experiences of 24 parents of providing gastrostomy feeding for their children was researched in a UK study (Brotherton et al 2007). Whilst parents valued oral feeding and understood the necessity and benefits of it, this was edged with sadness as the need to commence enteral feeding constituted a deterioration in their child's overall condition. The difficulties of sourcing childcare options when others may not feel confident to use a gastrostomy tube, together with the issues of disturbed sleep for themselves were also identified. The research evidence on sleep demonstrates the importance of good quality uninterrupted sleep each night for the mental and physical wellbeing for both adults and children (Bonnet et al 2014). When parents have interrupted sleep for months and years, they can become exhausted mentally and physically as they struggle to cope with the relentless effects of poor-quality sleep. The training and ongoing support for parents being available in the evenings and weekends, provided essential levels of reassurance for parents that they were not alone and could access expert advice out of hours if needed without having to resort to taking their child to the nearest hospital (Nelson et al 2015).

Pedrón-Giner et al (2013) researched the distress experienced by 92 primary caregivers of children receiving enteral nutrition and reported that the mothers reported high levels of anxiety from the stress of administering enteral feeds for their children, which in turn had negative impacts on their mental health. The mothers felt burdened by the level of responsibility and demonstrated symptoms of anxiety and depression. The dilemma for parents and clinicians, is ensuring that primary care givers have the knowledge, training, confidence, and equipment to be able to deliver the enteral feeds needed for the child, with expert support available when needed.

The psychosocial aspects of feeding children with a neurological disability for the parents, can identify high levels of both maternal stress and feelings of guilt that their child had reached the point of needing enteral nutrition (Craig 2013). The importance of good preparation before the commencement of enteral feeding, together with ongoing support, considering the emotional and psychological wellbeing of the parents are essential, to encourage a positive outcome for the child and the wider family to be achieved. The use of toys with enteral feeding tubes inserted and age-appropriate books with stories and pictures of enteral feeding, can be used to help both the parents and the child to gain confidence with enteral feeding before it is commenced.

The quality-of-life experiences for Polish families following the insertion of a percutaneous endoscopic gastrostomy tube (PEG) in 302 children aged between 7 months and 20 years of age, was explored by Grzybowska-Chebowczyk et al (2015). The study identified the challenges experienced by some of the parents of finding a pre-school setting which would accept a child with a gastrostomy tube in situ. The majority of families reported an increase in their quality of life following the insertion of the gastrostomy tube and were satisfied with the outcomes, however the need for ongoing support from clinical staff remained.

Whilst the nutritional indicators for enteral feeding to be commenced may be straightforward to establish, the lived experiences of parents delivering artificial nutritional support for their child are important to understand, in order that any potential issues can be addressed and to enhance compliance of the enteral feeding regime required for the child (Boland et al 2017). This Irish study also identified that the key issues for 88 participants had been their experiences of poor discharge planning and an inconsistency in the advice given prior to discharge. This raises the importance of ongoing support from a single point of contact being available for patients and their carer givers, as one means of combating high stress levels and improving satisfaction with home enteral nutrition. This was relevant as in the study two of the mothers who were delivering enteral feeds to their children at home, and one of the young people had carers at university, who had been trained to take responsibility for their enteral feed requirements and to safely manage the care of the gastrostomy tube.

These studies have raised awareness for clinicians, of the issues around the emotional and psychological wellbeing of parents when providing complex care at home. The qualitative study Fereday et al (2009), explored the experiences of 34 parents who were providing enteral feeding for their children at home, and reported the conflicting emotions that parents reported of juggling their role of being a parent whilst also learning the medical skills necessary to give their child the enteral nutrition which was essential for their child's survival but was not a role they had envisaged as being part of their parenting journey. A qualitative study by Boland et al (2017) explored the experiences of 88 adult and paediatric patients following their discharge from hospital following commencing enteral feeding and identified the need for greater information and follow up support to be made available. The quantitative, longitudinal, multi-centre prospective study by Pedrón et al (2019) followed 93 children following the insertion of a gastrostomy tube for the delivery of enteral nutrition and reported the benefits to each of the children in terms of their improved nutritional status and growth, at

six months and one year after the gastrostomy feeding had been started. These studies have identified both the benefits and the challenges associated with enteral feeding. They have explored the levels of stress and anxiety that parents can experience in relation to providing enteral feeding for their children at home and highlighted the importance of on-going emotional and psychological support being made available for parents and for the child or adult receiving the enteral nutrition when needed.

2.4.4 The experiences of parents and their children of having epilepsy.

The large numbers of young people growing up with epilepsy has increased the need for further research to be done to understand this young adult group. Adolescence is known to be a time of biological, emotional, and psychological development (Christie et al 2006, Coleman 2011), where young people are keen to fit in with their peers and be able to take part in the social activities that their friends are enjoying. However, a diagnosis of epilepsy can lead to bullying or teasing, social isolation and reluctance to be open with others about the safety needs of the young person in the event of seizures occurring (Geerings et al 2015, Benson et al 2016, Lambert et al 2016,). A review of the risk factors which have been linked to sudden unexpected deaths in individuals with epilepsy (SUDEP) (Brotherton et al 2006), identified that compliance with medication and seizure control were significant risk factors in sudden unexpected deaths. However, abnormalities with cardiac rhythms can also be a contributory factor.

The importance of providing the level of information and knowledge needed to improve outcomes for young people and their parents was reported in a systematic review of mixed methods studies of the needs of young people with epilepsy (Lewis et al 2010). Young people reported health professionals being more concerned with medication compliance in seizure management and prevention, than taking a holistic view of the young person which took account of their individual lifestyles and concerns. The findings from this systematic review identified that young people were seeking greater information concerning how to live their lives as a young person with epilepsy and had felt that the information that had been provided by health professionals had not met their specific needs. The complexities of providing information about epilepsy and seizure management should not be underestimated when there are known to be around 38 different seizure types and 30 different known epilepsy syndromes and the information for each young person and their parents will vary according to the cause

of their seizures (Epilepsy UK 2018). The importance of psychosocial adjustments for young people was found to be crucial for improving the outcomes for young people in the longer term. Here the importance of providing a holistic assessment of the lives of each young person is evident, with active listening important to enable each young person to feel that they have been heard, and that health professionals are interested in them as individuals without the sole emphasis being on their epilepsy.

Crosby-McKenna (2010) explored the effective and timely transition of young people with epilepsy to adult epilepsy services, through a survey of 266 young people aged between 12-20 years with epilepsy in the UK. The epidemiology of epilepsy is important as it is estimated that there are 40,000 young people living with epilepsy in the UK, all of whom will need to be supported as they approach adulthood and move to adult services. The findings from this large survey concur with the findings from other studies in identifying the importance of adequate and age-appropriate transitions services being available for young people, which can provide the information that young people have requested, by practitioners who are knowledgeable and confident in working with teenagers and are available to provide information concerning contraception and the consumption of alcohol. Young women with epilepsy require specialist advice on oral contraception options as some are contraindicated with some epilepsy medication (Reimers 2016). The role of transitions coordinators and the development of transitions teams were also identified as important for supporting young people and their parents to develop effective professional relationships with clinicians within adult services. As with the provision of any health services, the need for such a service needs to be verified through the development of a business case, and the evidence of the numbers of patients with an identified disease to warrant the financial investment in tailor made adolescent services. This may lead to families having to travel longer distances to access a specialist service for adolescent epilepsy transition services; however, telephone support between young people and specialist services may be one approach which could help to bridge the gap and improve the experiences of transition to adult services for young people and their parents (NICE 2016).

A systematic review of 19 research studies explored the views of young people with a diagnosis of epilepsy, with the aim of understanding the ways young people managed their epilepsy (Lewis et al 2014). The issues of compliance with medication, and the correlation with the occurrences of seizure activity was identified by health professionals and parents.

The recommendations were that further research should be conducted to improve long term outcomes, particularly as each year there are deaths of adults and children during epileptic seizures which may be preventable. Compliance with prescribed treatments is a complex area which will vary for each young person. Good communication skills of the health professional and the confidence of the young person to be able to articulate the challenges that they might be facing, are essential if good seizure management is to be achieved.

The issues of transition for any young person with a continuing healthcare need are vital and are not reducing. A review of 49 studies on the transition experiences of young people with epilepsy confirmed the importance of looking at the holistic experiences of young people, which must include their emotional and psychological wellbeing and not just focus on seizure management, and compliance with their treatment plans (Geerings et al 2015). The impact for young people and their parents of leaving services where they are known and have formed levels of trust with health professionals and moving to new services can be very significant with poor transitions experiences having a negative impact on the health of some young people (NICE 2016, Rajendran et al 2016, Andrade et al 2017, Camfield et al 2019).

A large study investigated factors that affected the self-esteem of 15 young people with epilepsy in Singapore (Chew et al 2019). There was a link confirmed between the severity and frequency of seizures and a lower level of self-esteem and higher symptoms of stress than for young people with less severe disease. The recommendations were that greater emphasis should be placed on providing individualised support for parents and young people who have a diagnosis of epilepsy. As with other studies and policies, the importance of individualised care for young people with epilepsy, provided by health professionals with expertise in the field of neurology was identified.

The impact on the whole family when a child is diagnosed with epilepsy was examined in a UK study by Harden et al (2016). This overview of the literature of 21 studies explored the views of young people with a diagnosis of epilepsy and the views of their parents. Their findings concurred with the experience of the young woman in this study who had developed epilepsy in her mid-teenage years, in identifying the disruption to her child and adolescent years from having seizures. Young people who may have wanted to learn to drive like their peers, are unable to do if they are having seizures, and a diagnosis of epilepsy will prevent young people from pursuing certain career paths which includes becoming a pilot or joining

the armed services. These barriers cause limitations for young people at a crucial time in their emotional and psychological development when they are seeking increased independence from their parents (Christie et al 2005). The risks to young people with epilepsy of living in student accommodation at a university or college or moving to share accommodation with friends when they are having seizures, can cause great anxiety to the young person and to their parents, who worry about nocturnal seizures which may cause injuries or death. The stress for parents and children when their child had a diagnosis of epilepsy and their experiences of stigma and negative social experiences was also explored in a study by Benson et al (2016).

The issues concerning epilepsy transition services was the focus of a small study with 10 adolescent participants in the UK (Dhanjal et al 2017). The findings from the study highlighted the areas of improvement that were needed in relation to ensuring that lifestyle factors were covered by staff in their conversations with young people during the transitions period. These findings enabled the further development of the existing service in that location. A checklist was developed and implemented to ensure that each of the areas of information and guidance were documented as being covered with each young person with epilepsy who was going through that transitions service. Although this was a small telephone survey, the implications are important for all young people with epilepsy and concur with the findings from other studies where young people had identified their needs for further specific information to be provided; to enable them to have the information to guide their decisions for their futures.

The findings from a recent study in the UK of the transition's experiences of young people with epilepsy (Teh et al 2018), concurred with previous studies where participants wanted more information on contraception, career choices and the genetic transmission of epilepsy. Whilst the Teh et al (2018) study revealed a high satisfaction rate with the service, the needs of young people for further information concerning contraception, the inheritance risks of epilepsy and careers advice were also identified as unmet needs for some. The complexities of the transitions process for young adults can be improved by the support of specialist nurses and meeting multi-disciplinary teams within a transition's specialist clinic setting. Health professionals need to be able to signpost young people to reliable online information, as an adjunct to giving information in a hard copy. The aim of these two approaches would be to empower young people to access information at a time and in a format that is appropriate for

their specific needs; and to advise young people against using information on the internet which might not be reliable, or evidence based. However, eventually, each young person with epilepsy will need to make the transition to adult services and become more autonomous with their own care.

When young people with epilepsy fail to make the transition to adult care, the effects on their emotional and psychological wellbeing can result in a negative impact on their independence, socialisation, and quality of life. A review of a range of transitions programmes from child to adult services for young people with epilepsy, from eight different countries (Camfield et al 2019), identified that the key factors to influence the success of transition from child to adult services included the individual readiness of the young person to make the transition, the level of involvement of the parents as they continue to provide support but expect their child to take on more responsibility for their own care, and the provision of age appropriate information for each young person about their disease, and management.

The links between loneliness and young people with epilepsy becoming independent from their parents (Geerlings et al 2019) is significant for young adults on their journeys to adulthood. Those who were still living with their parents reported higher levels of loneliness, which was attributed in part to the challenges for young people with epilepsy to use public transport and their fears of having a seizure in public, which contributed to their willingness to socialise outside of their homes. Having a supportive but not overprotective family was identified as being a protective factor for young people preparing for their transition to adulthood. However, seizure control is also an important factor in parents having the confidence to allow their children to have more freedom to go on school trips or move away from home to attend university.

Each of these studies have identified some of the emotional and psychological challenges for parents and their children once a diagnosis of epilepsy has been made. Parents need support and guidance from health professionals to have the skills and confidence to know when to transfer more autonomy to their teenage children with epilepsy, whilst in turn, young people need to demonstrate their ability to comply with their treatment plans and have the knowledge and understanding to manage their lifestyle choices. Young people need to have access to reliable evidence-based information to enable them to make informed decisions about their lifestyle choices.

This wide range of studies from a number of countries identify the multiple challenges that young people living with epilepsy face, as they learn to live with the condition and to prepare for their transition to adulthood and adult services. A quantitative study by Geerings et al (2019) recruited 59 young adults with epilepsy and explored their transition to adulthood. They found that a failed transition to independent living away from their families increased social loneliness. They recommended that health care professionals working with young people with epilepsy should be specifically asking young people about their social relationships and working with young people of their plans for transition to adulthood. A quantitative study by O'Toole et al (2019) explored the communication between 47 children and 72 parents where the child was living with epilepsy. The study identified the importance of open communication about the condition for the levels of acceptance and emotional and psychological wellbeing for the children and young people concerned. The health promotion issues of contraception and pre-conceptual care for young women with epilepsy were considered in a review of 19 studies undertaken by Lewis et al (2010) and identified the importance of communication and preparation for young people with epilepsy moving from child to adult epilepsy services. These studies have identified the physical, emotional, and psychological complexities for young people living with epilepsy and the concerns of their parents is seeking to support their children in moving towards increased independence. Each young person's experience and risks from this condition will vary, but the needs of health professionals to provide the education and support needed for young people to become independent from their parents remains paramount.

2.5 Achieving urinary continence through intermittent self-catheterisation.

Whilst urinary continence is not in itself a life limiting condition, it adds to the challenges of daily life for young people who are already managing other complexities of their health care. When a young person is intermittently self-catheterising in order to empty their bladder several times each day, they are at an increased risk of developing urinary tract infections which can increase the risk of sepsis which can be life threatening (NICE 2018). The management of urinary incontinence in children and adults has changed significantly over the past decades, with new ways for young people to achieve urinary continence through

intermittent self-catheterisation or through the surgical development of a Mitrofanoff [6]stoma, where paralysis, bladder abnormalities or abnormal nerve functioning prevents a child or adult from being able to void their bladder and achieve urinary continence for themselves. Two of the young people in this research were intermittently self-catheterising each day to achieve urinary continence.

In a study of 20 Canadian children with Spina Bifida, and 31 parents of children with Spina Bifida, experiences of intermittent self-catheterisation of using either a hydrophilic (self-lubricating) urinary catheter or a washed and reused PVC urinary catheter to achieve urinary continence were explored (Chick et al 2012). It was noted that from previous studies, children and young people with Spina Bifida had rated being incontinent at school as extremely stressful. The study established that once young people had learnt to use self-lubricating urinary catheters; this increased their independence and opened other opportunities such as camping to them, as they would no longer have to find suitable facilitates to wash and air-dry urinary catheters. However, funding arrangements need to be in place for self-lubricating catheters to be made available through the local continence services, and provision made for the collection and disposal of used urinary catheters. Schools need to have the toilet facilities available for children and young people to have the space and handwashing facilities available to enable them to use urinary catheters safely at school.

A study of 11 children with Spina Bifida in India who had undergone the surgical procedure for the formation of a Mitrofanoff catheterisable stoma (Reddy et al 2015), demonstrated that following the procedure, each of the children were able to perform intermittent self-catheterisation independently, thus increasing their self-confidence and improved their own body image of being able to be continent at school and in social situations. However, young people need to be taught how to handle urinary catheters and to be rigorous with their hand hygiene techniques to minimise the chances of introducing infection into their bladder via the Mitrofanoff stoma, which in turn could cause renal damage. The ability to handle a urinary catheter is a fine motor skill, and for young people with impaired fine motor skills, or a progressive degenerative disorder, they may not have the dexterity to become independent with using the Mitrofanoff stoma. However, the benefits are that the urinary continence will

[6] A Mitrofanoff stoma is a surgically created passage often using the appendix to create a urinary diversion for patients with abnormal bladder function. The stoma has a one-way valve which enables intermittent self-catheterisation to be take place between 4-6 times per 24 hours to achieve urinary continence.

prevent skin soreness from developing from urine when the child wears nappies or continence pads.

There is limited research available concerning the experiences of young people living with the need to perform intermittent urinary self-catheterisation. However, the studies that are available have considered the emotional wellbeing and increase in self-esteem for children and young people who have a functional cause for their urinary incontinence. The surgical advances which have enabled children and young people to benefit from the construction of a Mitroffanoff stoma have been explored by Reddy (2015), however, the benefits of being able to achieve urinary continence through intermittent self-catheterisation are balanced with the practicalities of the child or young person having the dexterity with their fine motor skills to be able to perform the necessary task and the potential risks of urinary tract infections of sepsis developing if hygiene practices are poor. These surgical options for urinary incontinence offer hope for young people in their journey towards independence and adulthood and remove the need for the removal of continence pads throughout the day, and the anxieties concerning the risk of odour from urinary incontinence.

These studies have identified that the numbers of young people growing up using intermittent self-catheterising will continue to increase, and further research into the requirements for these young people as they move to adult services will be needed.

2.6 Parents' experiences of developmental delay and autism.

The experiences of 26 parents whose children were disabled but who did not have a confirmed diagnosis and are known as SWAN (Syndrome without a Name) demonstrated the stresses that families live with when there is no diagnosis or prognosis for their child (Coates-Dutton 2015). With the advances nationally and internationally in the field of genomics, more families are choosing for their child's DNA to be sequenced in the hope of finding causes and answers to their child's developmental differences (100,000 Genomes project 2019). The scientific breakthroughs which are gathering pace in the field of genetics and gene therapies give hope to some families that treatment options for their child might become available, and for children with epilepsy genomics provides information for clinicians regarding the efficacy of different treatment options for seizure management, which includes the use of cannabis oil for children with fragile epilepsy where they have multiple seizures each week.

Online support groups provide essential support for parents whose children where a diagnosis is not available for their child or where there are other parents whose children have the same clinical presentation as their own child (www.undiagnosed.org.uk). This parent-to-parent support is important as parents have the empathy and understanding with one another, that health professionals are unable to give.

A Canadian study explored the levels of stress and distress that parents can experience when their child is diagnosed with developmental delay and disability; particularly at key moments in their child's life such as when they start school. The findings from this large study of 155 mothers highlighted the importance of the coping strategies that the mothers had developed and the importance of parents feeling and becoming empowered in their individual situations (Minnes et al 2015).

The ways in which parents who have children with additional needs cope, has been identified as being crucial for the emotional and psychological wellbeing for the parental relationship and for each of the children in the family. Cultural perspectives on the ways in which 220 parents of children with disabilities in south India coped day to day with caring for a child with additional needs, identified the importance of relationships between the parents, support from the wider family and the local community as protective factors in influencing how parents coped and adjusted to having a child with special needs (Gothwal et al 2015).

A UK study explored the experiences of 38 parents from 36 families of young people with Autism aged between 15 and 21years. The findings identified the importance of parents being supported in their parenting roles as they prepared and supported their children through the process of transition to adult services. Parents identified their experiences of a lack of support during the planning processes for their children and reported feeling overwhelmed in seeking to communicate with different services and to coordinate the future care for their child. The findings indicate the need for better support for parents and young people with Autistic Spectrum Conditions is needed during the transition process to adult services. However, the need for adequate training to ensure that the staff who are in post have the confidence and currency of information to be able to provide the specialist support needed, remains unmet for some families (Spiers 2015). The numbers of adults and children who are living with a diagnosis of Autism in the UK is estimated to be more than 700,000 (Crane et al 2016),

which makes this a significant health need, with high levels of health and social care resources required to support children and families once a diagnosis of Autistic Spectrum Disorder (ASD) is made. The severity with which individual adults and children are affected varies significantly, with children who are most severely affected being educated within specialist education provision and requiring high levels of support from parents or carers for the duration of their lifespan. The beginning of this journey starts with health professionals or parents first noticing that young children have poor eye contact with their primary care givers, and in addition may have delayed speech and language, poor socialisation and play skills with siblings or other children, or other development of behavioural difficulties. However, the path to establishing a diagnosis can be lengthy with long waiting lists for children to be seen by specialist Paediatricians (Minnes et al 2015).

A large UK study of 1047 parents of children with Autism, examined their experiences of obtaining a diagnosis for their children (Crane et al 2016). The findings revealed that the average length of time between parents first seeking help from a health professional and finally having a confirmed diagnosis for their child was 3.5 years. The challenges for parents concerned the interventions which could be offered once a diagnosis of autism had been confirmed; with many parents reporting high levels of dissatisfaction with the levels of support offered to them following their child's diagnosis. The levels of ongoing support vary widely geographically in the UK, with some specialist nurses available to provide ongoing support for parents, whilst in other areas, parents reported feeling lost and frightened as they continued with their unexpected parenting journey.

The abilities and challenges for children and young people living with developmental delay or autism vary widely, however the needs for ongoing support is undeniable. A quantitative study by Mines et al (2015) recruited 155 mothers of 113 boys and 42 girls to explore the predicators of distress in parents of children with developmental delay and identified the importance of providing ongoing support for mothers, as the wellbeing of the mothers correlated with the improved outcomes for their children.

A qualitative study undertaken by Spiers (2015) when interviews were conducted with 38 parents to explore their experiences of parenting a child on the autistic spectrum. Parents identified their needs for ongoing support in preparing their children for their move to becoming young adults. The complexities of establishing a diagnosis for the child and the

high levels of stress that parents can experience in parenting a child with a diagnosis of Autism, can be considerable and parents identified the needs for ongoing support and advice to be made available to them, to aid them in their parenting journey of their child with continuing care needs. The ongoing and changing needs for young people growing up with developmental delay for continued and high-level support throughout their lifespan, and support for parents to develop and underpin their parenting are reported in these research studies.

2.7 Adolescents and neuromuscular conditions

For any child or young person who is living with a chronic health condition, the importance of the role played by their parents or primary care giver in providing for their ongoing health needs should not be underestimated and needs to be recognised by health and social care professionals (NICE 2016). Kingsnorth et al (2011) explored the notion of parents being considered *"transition experts"* when supporting their child with additional healthcare needs as they moved between children's and adult services within a rehabilitation hospital. The data was obtained from 10 parent – led peer support group sessions. Although this research related to one specific setting, the findings that parents need ongoing support in preparing for their child to move between services concurs with the findings from UK research and policies (NICE 2016). The evidence shows that the numbers of inpatient stays for children and young people aged between 0-14 years of age with neurological conditions is continuing to rise in England, with the highest numbers of children having a diagnosis of epilepsy or cerebral palsy (Jarvis et al 2018), which has implications for current and future service planning. As these young people reach adulthood, their needs for hospital admission will not diminish and will need to be met in adult services.

2.7.1 Experiences of young people with neuromuscular conditions

Although there are many published research studies which have explored the experiences of young people with a range of chronic illnesses, there is limited published research concerning the experiences of young people with neuromuscular disorders. Research undertaken on behalf of the Muscular Dystrophy Campaign with 40 young men aged between 15 and 33 years of age of their experiences of growing up with Muscular Dystrophy, identified the challenges that the young men had experienced socially and emotionally and their desires for increased independence but highlighted their support needs from parents or carers to enable

this hope to become a reality for them (Abbott et al 2010). The findings revealed wide variations in provision between geographical regions in the packages of care and levels of support that were available for young adults with Muscular Dystrophy and their families. This variation in provision is in sharp contrast to the National Service Framework for Long Term Conditions (2005) which aimed to remove the disparity in service provision in different parts of the country. The NICE guidelines on the transition of young people from children's to adult services (2016) identified the importance of good multi-agency working between services and the fundamental principle of involving young people in all aspects of their care, including the planning and preparation of transition arrangements to adult services and advised that transition takes place to adult services should take place at a time when a young person is developmentally ready rather than at a predetermined age (McDonagh 2006). If this new approach were to be implemented across the country, it would necessitate changes in local service provision, which would have significant financial, training and resource implications and would require an expansion of appropriately qualified staff to resource the new services.

The three identified threads of neuromuscular disability, gender, and their experiences of becoming adults, for 11 young men aged 16-27 years with Duchenne Muscular Dystrophy (DMD) in Canada was explored by Gibson et al (2014). The study identified that as more young men with DMD are living into their adult years, these young men were able to demonstrate through audio diaries, photographs and individual interviews, their abilities to intersect the three dimensions of their lives. They demonstrated the similarities of their experiences of being at university, having social lives, interests and hobbies which aligned with their non-disabled peers, whilst also acknowledging their differences of needing support with personal care from carers, which they had needed to adjust to, after having many years of their parents being their main source of support. As with other research, the essential hygiene, nutrition, and activities of daily living needs of young men with deteriorating muscle function will continue to be needed to be funded and resourced, to enable young people with DMD and other forms of Muscular Dystrophy to be active participants in their lives and to reduce social isolation and loneliness. One of the challenges for young men with DMD once they leave school, is that they may not readily have access to the physiotherapy and hydrotherapy that they have benefited from during their childhood years, which can have a negative impact on their comfort and range of movement in their limbs.

The psychological and emotional wellbeing of young men growing up with the progressive, degenerative and life limiting neurodegenerative condition of Duchenne Muscular Dystrophy (DMD), is of crucial importance as both they and their families come to terms with the shortened life expectancy of this disease, which does not have a cure at present.

The impact on siblings of having a brother with Duchenne Muscular Dystrophy (DMD) was examined in a UK study by Read et al (2010). The primary carers within the family provided information about the demographics of the family, and the siblings, the parents and teachers completed the Strengths and Difficulties Questionnaire (SDQ) to assess the level of any emotional difficulties experienced by the siblings in the previous 6-month period. The findings indicated that the siblings from that study were well adjusted psychologically.

Neurodevelopmental, emotional, and behavioural issues in DMD relating to underlying dystrophin gene mutations was researched in a large UK study by Ricotti et al (2015) with 130 male participants aged between 5 and 17 years were recruited from four European Countries. The findings provided the information that the participants who had a mutation at the end of the DMD gene had an increased risk of a neuropsychiatric disturbance. Over one third of the participants demonstrated at least two measures of emotional, behavioural, or neurodevelopmental issues which included symptoms of both internalising and externalising their problems. The authors concluded that boys with DMD are at an increased risk of mental health issues which increased for those with the specific gene mutation on the DMD gene, warranting possible intervention and treatment as appropriate.

An Iranian study exploring the quality of life in young males with DMD Gholamreza et al (2016) with 85 male participants aged between 8-18 years was the first Iranian study looking at the specific needs of this client group. The participant group were matched with 136 neurotypical peers. The findings showed that the self-assessment of their quality of life for the boys with DMD was comparable with the results for their non-affected peers in most of the assessed areas. A lower level of satisfaction was shown in the two sections “physical health” and “friends”. The findings from the parents demonstrated that parents’ assessment of their child’s quality of life was lower than was demonstrated from the findings from young people. The importance of psychological support for young people and their parents was identified as an area which would benefit from being developed.

An Italian study explored the mental health of a large sample of 47 boys with DMD aged between 2 and 18 years using the Wechsler Intelligence Scale or Griffiths scale to assess the cognitive ability of each boy and the Child Behaviour Check List (CBCL), the Youth Self Report (YSR) and the Strength and Difficulties Questionnaire (SDQ) to assess behavioural and emotional difficulties (Colombo et al 2017). In common with other studies which have examined the links between chronic illness and mental health concerns of anxiety, depression, and internalizing problems; the findings from this cohort indicated higher levels of emotional stress and distress for the participants than amongst their peers.

As more men with DMD live longer lives, they could become role models for boys and adolescents. Abbott et al (2014) in a UK study, interviewed 37 young men with DMD aged 15 years and over, and 58 family members looking at the experiences of young men moving from childhood to adulthood. The participants identified that they needed more physical, emotional, and psychological support than they had received to help them to negotiate their transition to adulthood. Employment and career opportunities were identified as areas of life that had been missing for many young and older men. Education and health professionals need to be able to talk to young men with DMD about all current and future issues in their lives, which includes opportunities for young men to be able to meet together and socialise with one another.

Hamdani et al (2015) interviewed 11 young men with DMD aged 16 to 27 years in Canada to explore their individual experiences of moving from childhood to adulthood. The authors identified that the aims of many current policies concerning the transition of young people with chronic illness to adulthood, have been written with the expectation for young people to become more independent over time. However, they argue that this trajectory does not meet the needs of young people living with a progressive degenerative condition, where their dependence on others will increase rather than decrease over their lifespan. As with previous studies, these findings highlight to health and social care providers that the needs of young people with progressive degenerative diseases are different from other young people with chronic illness, who are anticipated to have many years in adulthood to look forward to. However young men with DMD need both social opportunities and practical support to enable them to live their lives in ways that are meaningful to them, and to enable their physical, emotional, and psychological needs to be met.

In a Japanese study seven men with DMD aged between 20 and -48 years were interviewed about their experiences of understanding their disease and living with their condition (Fujino et al 2016). Key stages of the disease progression such as starting to use a wheelchair or a ventilator, heralded times of fear and anxiety for these men, who had not spoken to their parents of their concerns because they did not want to distress them. Each identified that they would have benefited from having professional support at these key periods of transition.

Skyrme (2017) interviewed nine boys and young men aged between 10 and 21 years and one young woman with Muscular Dystrophy in a UK study about their experiences of living with a progressive degenerative condition. Young people were reticent to discuss their difficulties with friends and did not want to be noticeably different from their peers and wanted to "fit in" but had experienced social isolation from being unable to participate in social activities or socialise at friends' houses due to physical barriers such as stairs which they could not navigate as wheelchair users. The research sought the young people's views on being participants in research projects to treat the symptoms of their muscle wasting disease, with most young participants wanting movement in their hands and arms to be restored to increase their independence with feeding but also to enable them to hug their parents. Providing opportunities for young people to speak freely about their experiences of living with their disease, allowed greater insight into how young people viewed their lives and their hopes for the future.

These different studies have given an international perspective on the lives and experiences of young men with DMD growing up into adulthood, and of moving forwards to an unexpected period of lifespan of their lives, which for previous generations had not been available to them. Young people with progressive degenerative neuromuscular conditions can now make plans for their early to mid-adult life, which in their early childhood would not have been presented as a viable option to their parents when the diagnosis for their child had been confirmed. This increase in life span has become a reality for many young people due to major advances in medical science which includes non-invasive ventilation, and new drug therapies. These interventions have increased the life span for young people with many different types of Muscular Dystrophy, and therefore the social and emotional needs of young people with progressive degenerative disorders will continue to rise rather than diminish. However, when young people with progressive degenerative conditions leave school, they

can become socially isolated when no longer with their peers (Abbott et al 2014), and they need services which are tailored to meet their needs as they reach adulthood.

The quality-of-life indicators are an important reminder to clinicians to closely monitor the emotional and psychological wellbeing of young men growing up with DMD and to provide services to meet the specific needs of this client group. The risks for the development of poor mental health for boys growing up with DMD are identified, with the needs for ongoing emotional and psychological support throughout their lives being advocated. The life expectancy of young men with DMD is continuing to increase and therefore the cohort of adult men with DMD is growing across the world. Whilst the treatment options for the management of the physical impairments are extremely important, the social and emotional needs of young men for relationships and being able to live a life that is meaningful for them is also crucial for their psychological wellbeing.

2.8 Young people's experiences of transition to adult services

Within the UK and other Western countries, there are currently two opposing professional perspectives concerning the timing at which young people move to adult services when they have continuing healthcare needs. One perspective from specialists in adolescent medicine advocates for the recognition that the maturation of the prefrontal cortex in the frontal lobe is a biological process which varies in its timing from one individual to another and therefore the timing of the individual transition should be an individualised stage of development, as opposed to the current threshold for many services whereby young people are required to make the transition to adult services at a predetermined age (McDonagh 2007, Campbell et al 2016, Willis et al 2018). Other paediatricians argue that making the transition to adult services at the legal age of 18 years, avoids ambiguity and confusion for young people, their families and for professionals, and times well with young people leaving school and preparing for the next stages of their adult lives (Berg-Kelly 2010). A predetermined age for transition should allow for the planning and preparation for each young person, with the aim that each young person develops their own knowledge of their health issues and treatment interventions and allows for young people to gradually assume increasing levels of responsibility, confidence and competence in the management of their own health; whereas during their childhood that level of knowledge and responsibility had usually been held by the parents or primary caregivers (McDonagh 2006, McDonagh 2007). Arguably however,

this approach does not allow for the variations in abilities of individual young people, who due to additional considerations which may include a learning disability, social or emotional challenges or an overall deterioration in their health; may need the ongoing support from health professionals who have known them for many months or years and can offer guidance beyond the biological age of 18 years (Kerr et al 2018).

As more young people with chronic and complex health conditions grow up to become adults, their experiences of transition to adult services influences their engagement with the health services that can continue to support them into adulthood. Bryant et al (2009) reviewed 17 studies concerning the transition experiences of young people with chronic illnesses and identified lack of transitional support as being a key theme to emerge from 5 of the 17 studies that they reviewed. Some young people felt that staff did not have enough time to spend with them to plan and prepare for their move to adult services and identified concerns that some staff had not received training in adolescent physical and emotional development. The implications for this affect both staff and patients, as staff being adequately trained can enhance their confidence when working with young people, which in turn may improve understanding and communication between young people and clinical staff and may have a positive outcome for compliance and attendance at future appointments (Viner 2019).

When considering the needs of young people, a joint UK and Australian study explored the experiences of 21 young people in the UK and 39 young people in Australia with HIV of their transition to adult services (Bundock et al 2011). The study noted that having the experience of a well-planned transition can have a positive effect for young people with complex and chronic health needs. These finding concur with the findings from other studies and current polices which advocate for the needs of adolescents with continuing healthcare needs to be recognised, and for health professionals to work with young people and their families throughout the years of their adolescence to prepare them for transition to adult services.

In addition, Allen et al (2011) used face-to-face interviews with 46 young people and 39 mothers to explore the changing needs of parents when supporting their children with diabetes to make the transition from child to adult services. During this transitional period parents and their children need to adjust to the young person developing increasing autonomy over their own health. This change in role requires the parents to adjust to a new parental role

of stepping back and allowing their child to take the lead with their health and to take responsibility for their actions and inactions and the consequences that may result. The research identified that parents also need support in the adjustment period of transition between services that identified that future services need to acknowledge and accommodate the roles that parents continue to play in supporting their children. The role of parents in supporting their children with any chronic condition needs to be acknowledged, with young people being given the opportunities to gain experiences and have opportunities to have more control over their health compliance, knowledge of their disease, and develop their confidence and competence whilst still in children's services, to prepare for the expectations of independence in adult services.

Irrespective of the nature of the chronic condition that young people are living with, the same themes around the needs for support for both young people and their parents / carers continued to emerge. In a UK study, Price et al (2011) undertook a qualitative study using semi-structured interviews which explored the transition experiences of 11 young people with diabetes. The two dominant themes that emerged, concerned the need for transition services to be developmentally appropriate and the importance of the communication skills used by health professionals when working with young people. The cognitive development of a young person may not match their chronological age, with some adolescents being less mature and others more mature than their biological age. Therefore, the information that is provided for young people about their medical conditions needs to be titrated and adjusted to meet their individual needs and adjusted accordingly.

The recurring theme of young people having access to staff who have the knowledge and communication skills to work effectively with teenagers, are repeated in different studies and policy documents (DH 2007, DH 2008a, DH 2008b, DfE 2014, NSF 2016,). Evidence continues to suggest that this aspect of care is crucial for enhancing the transition experiences for young people and for enabling young people to develop their own knowledge and self-care skills to manage their health needs for the future. However, the costs of introducing new services at a time when the NHS is in a large financial deficit can be challenging. Robust business cases justifying the needs of one patient group over another need to be developed and presented to local Commissioners and the funding agreed before new services can be introduced. The training needs of staff in adult services, who may be inexperienced in caring for young adults with some specific conditions need to be met, to ensure that young people

receive the optimal levels of care and support in adult services. Many young people are articulate and well informed about their medical condition and treatment regimes which can be extremely important for developing the skills and knowledge of new staff providing care and support for them.

One of the most significant challenges for service users, commissioners and service providers concerning the transition to adult services for any young person, is the lack of consistency between health, social care and education providers regarding refining and defining the age at which young people are expected to move between services (NICE 2016). Within healthcare providers both locally and nationally there are wide variations between the expected ages at which young people move between services, ranging from eighteen years in some services, to mid-twenties for others and this differs between NHS service provision and charities such as children's hospices, with come clinicians allowing young people to stay in children's services longer than others (Aldiss et al 2016). These wide variations and lack of consistency cause uncertainty and confusion for young people and for their parents and carers. Some of the studies that explored the experiences and longer-term outcomes for young people with different chronic illnesses; highlights the growing evidence that for some young people moving from a service which has a philosophy of family centred care to an adult service which focuses on the individual has significant implications for poorer compliance and longer-term health outcomes. Young people disengaging with health services and for some young people ceasing to comply with their treatment regime which in the most severe circumstances can result in the loss of a transplanted organ (Watson 2000).

The process of transition has been defined *as "the purposeful, planned movement of adolescents and young adults with chronic physical and medical conditions from child centred to adult oriented healthcare systems"* (The American Society of Adolescent Medicine 2003 p1). This definition is endorsed by current UK policies on transition for teenagers with ongoing health care needs (RCN 2004, DH 2008, DfE 2014, NICE 2016). For all teenagers, the process of transition from childhood to adulthood not only includes the transition between medical services for those with ongoing medical needs, but also includes the development of their own identity; the search for a "significant other" in terms of a partner; the ability to move out of the family home and to live more independently and to find employment (Christie et al 2005).

A debate exists around the definitions and ages for transition to adult services. Local examples of the current discussions and variations that exist, include whether provision for young people should extend to nineteen, twenty-one or twenty-four years of age. One local children's hospice extended inpatient provision to nineteen years of age and outpatient support and access to day events up to the age of twenty-four years. This decision was made to fill some of the current gaps in respite provision in the local area for young people with life-limiting illnesses. For each of the local health services, the commissioned service and the funding attached to that service determines the inclusion and exclusion criteria that are applied to the admission of teenagers to a service, with variations existing between specialities and between primary, secondary, tertiary and charity provision (*Together for Short Lives* 2015). This can vary across the country, so if families move from one geographical location to another, they may find that their choices of provision are different to what they have been used to, which creates inequality for young people with high levels of need.

For some chronic but more common diseases, such as diabetes or Cystic Fibrosis, the numbers of young people moving into adult services are larger. In many areas this has resulted in transition clinics and services being commissioned and resourced at a local level (Dugueperoux et al 2008, Allen et al 2011). However, for young people with rare or more complex health needs, which includes progressive neuromuscular disorders, the number of young people requiring the transition to appropriate acute and respite care services is smaller which may make the demands for specialist services for some groups of young people less evident. The recent changes in the NHS and the introduction of the centralisation of services; may require young people and their parents to make much longer journeys to access suitable provision. Where this is the case, the expense of travelling much longer distances, and the additional physical impact and resulting fatigue from having to undertake more arduous journeys in another part of the country to access the services that they need can take its toll on parents and young people.

The Muscular Dystrophy campaign published advice for young men with Duchenne Muscular Dystrophy (DMD) in relation to transition (2010) and identified the aspects of life that are most important for young men with DMD namely wanting and seeking to be in a relationship with another young person, have a job, being able to socialise and have a friendship group. A report from the same charity undertook a survey of 850 people living with a disease of their muscles and showed that 60% of the participants to the survey rated

their transition experiences as being *"poor" or "very poor"* (Muscular Dystrophy Campaign 2008); an experience which has been echoed in other research studies which have investigated the experiences of young people with ongoing health needs (Wedgwood et al 2008, Tuchman et al 2008, Taylor 2009). This concurs with the view that much more could be done to develop transition services and the ways in which young people and their families are prepared for their move from one service to another. This includes the advice that each young person should have an allocated health professional who coordinates the transition arrangements and works with each young person over a prolonged period of time to help them to prepare emotionally and psychologically for leaving a familiar service and moving onto a new service within adult provision. McDonagh (2006) argued that some professionals feel under prepared and find working with teenagers difficult, and proposed that better education and understanding of the neurophysiological, biological, emotional and psychological changes that take place in the adolescent brain, particularly the maturation of the pre-frontal cortex and the impact that this can have on behaviour and compliance for some young people may significantly help health, education and social care professionals to understand some of the decisions that young people make (Blackmore 2009, Blakemore et al 2010). Extensive research by Blakemore in cognitive neuroscience of the physiological changes that take place in the teenage brain during the adolescent years, have been fundamental in developing professional knowledge concerning the neurophysiological brain development of young people. This scientific evidence has helped professionals to develop new ways to communicate with young people and to address their needs in different and more appropriate ways, such as recognising that teenagers have difficulty in focussing on the longer term but are more focussed on the here and now and their immediate or shorter-term goals and aspirations (Christie et al 2005, Burnett et al 2009, Blakemore 2018).

However, as with the commissioning of any new service, evidence of the need for a new service through epidemiological data and the development of a strong business case, need to be provided to secure the required funding to resource new adolescent units. Ongoing audit and evaluation of the effectiveness of any new service, including evidence of patient satisfaction, are required to ensure ongoing funding in a financially constrained and competitive healthcare environment. The definition from the NSF for Children, Young People and Maternity services (2004) defines adolescence as being from *"the beginning of the tenth year to the end of the nineteenth year"* (p18). Epidemiological data for the UK continues to show growth in the overall population numbers of young people with those age

parameters and therefore the need for services that meet the specific needs for young people appears to be robust (Office for National Statistics 2016).

There is evidence of the longer-term impact on compliance and health outcomes for some young people once they have moved from one health service to another (Watson 2000, Bell 2007, Price et al 2011). These findings have identified the growing numbers of young people needing to make the transition to adult services and has demonstrated that a poor experience of transition can lead to a significant deterioration in the health and longer-term outcomes for some young people. The themes that repeatedly emerge from these documents continue to discuss the importance of adequate and planned preparation for transition for each young person and their primary care givers. The planning and preparation for transition includes having appropriate and adequately resourced services for young people to transition to; the need for comprehensive documentation to demonstrate what has been discussed and which may include the provision of a health passport which the young person can keep with them. Different policy documents have highlighted the importance of having one healthcare professional who can act as the coordinator for the duration of the move between services (RCN 2004, DH 2008, RCN 2013).

One important variation between child and adult services concerns the differences between the philosophies of care between children's and adult services. Within children's services, inpatient provision has twenty-four open visiting for parents and the children grow up into teenagers with trust and confidence in healthcare professionals that they have known for a long time (Bell 2007, Dugueperoux et al 2008, Berg-Kelly 2010, Fair et al 2010, Bundock et al 2011). When young people then move to adult services; the philosophy of care focusses on the individual patient rather than the wider family, and within adult services it is the norm for inpatient provision to have restricted visiting hours with limited opportunities for young people to have a relative or close friend to stay with them. This change in philosophy in adult services, which focusses on the individual rather than the whole family may be welcomed by some young people who are ready and willing to take on responsibility for their day-to-day healthcare and are able to discuss and negotiate a treatment regime that fits with their life style and make compliance daily more achievable. Examples of this include the timings of medication, the frequency of certain types of medication required to manage their condition or the types of physiotherapy required to improve or maintain their lung function. However, research has demonstrated that some young people struggle to cope with the competing

demands being made of them from their healthcare needs, their educational demands and their need to develop their own identity and to fit in with their peer group. This on occasions results in young people not feeling able to or being equipped with the knowledge and skills to comply with their recommended and prescribed treatment plans, which can lead to a deterioration in their overall health, and which may be preventable (Bell 2007, Berg-Kelly 2010, Bundock et al 2011).

The research literature on the experiences of young peoples in transition to adult services has highlighted the importance of ensuring that young people, with a range of chronic health conditions, get the support that they and their parents or carers need in planning and being prepared for their transition to adult services. Compelling evidence from the studies discussed, has shown that when young people have a poor transition experience, this can have negative and long-term consequences for young people continuing to engage with health and social care services and consequently this can have significant adverse effects on their physical and emotional health. The age of transition is debated, with acknowledgement that the young person should be adequately prepared and emotionally ready for their transition to adult services, and there should be flexibility as to the age at which young people move to new services, rather than this important stage being expected at a pre-determined age. However, this change will have resource implications for inpatient and outpatient children's services, which are currently commissioned from birth to 18 years of age.

2.8.1 The role of the transition's coordinator

The current guidance concerning the transitions journey for young people with continuing healthcare needs in the UK, is that a transitions coordinator should be allocated to each young person to guide and support them through the transitions process (RCN 2004, RCN 2014, NICE 2016). The multiple layers of complexities that arise for young people with complex health needs may include the acquisition of knowledge about their disease, medications, and the interventions necessary to monitor and control their health; the development of clinical skills to administer injections or nebulised medications as required, whilst also juggling the demands of obtaining or further their education and meeting deadlines for the submission of academic work. However, although being allocated a transitions coordinator provides ongoing support for young people, they are not available for every speciality or in every NHS service. Therefore, some young people are not enabled and supported in their transitions

journey in the ways in which young people with access to specialist transitions staff are. For each service, there needs to be enough young people with a specific health condition to be able to justify employing a transitions coordinator.

A qualitative study from the USA (Annunziato et al 2013) compared the experiences of two groups of young people who had received liver transplants. The first group had not had the support of a Transitions Coordinator, whilst the second group, a year later, had been supported by a transitions' coordinator using a social media platform, text messaging, telephone calls and emails. In the first group four patients had died post-transplant, but in the second group there had been no transplant failures, no deaths and no young people becoming lost to follow up in the adult post-transplant clinics. Although this study had a small sample size, the findings demonstrated that the availability of a transitions' coordinator had a positive impact on the lives, experiences, and outcomes of the young people in the study. Further guidance of the role of transitions coordinators would be useful for staff who might be interested in moving into that role as part of their future career development.

This study together with the national guidelines has identified the importance of the role of a transitions co-ordinator who can hold the whole picture of the health needs for each young person and can act as a central point of contact for the young person, parents and health and social care staff to plan for and communicate the future care plans for the individual. Although some of the services in the UK, have transitions co-ordinators in post, this is not the case for all services, so not all young people have equity of service provision. The transition from child to adult services comes at a time of immense biological changes that occur for all young people during puberty. The hormonal changes co-inside with feelings of uncertainty as young people seek to establish their own identity and develop confidence in their abilities to make their own decisions and become more independent from their parents. The philosophies of care change from family centred care in children's services, to autonomy and individualised care in adult services. Transitions Co-ordinators provide a useful bridge between the two different services and by working with both the young person and their parents, they can support the young person to become more confident in their decision making and parents can start to gradually relinquish some of the responsibility that they have carried for their child.

2.8.2 The availability of services for young people

Revised understanding of the neurophysiological changes that occur in the pre-frontal cortex of the brain and the implications that this has for decision making for each young person as they move through the period of adolescence has been identified from more recent research (Christie et al 2006, Blakemore 2009, Blakemore 2010). Recognition that the complete maturation of the prefrontal cortex occurs later than previously known, has raised questions, and has encouraged ongoing debate as to whether young people should be allowed to stay in the services where they have built helpful therapeutic relationships with the staff and are well known, until they have reached their early to mid-twenties and are ready emotionally and psychologically to take on more of their own care (Viner 2008).

The increasing body of scientific evidence now points to transition being a stage of development rather than as previously thought; at a pre-determined age (Sawyer et al 2012). This is currently causing debate in services across the UK to determine where the inclusion and exclusion criteria for health services for young people should be revised and should continue to be set or have some flexibility to accommodate the specific needs of young people who need more time in a service that is familiar to them and their families (NICE 2016).

However, as identified from recent advances in cognitive neuroscience, the ages at which young people are ready and can take responsibility for their actions and decisions about their healthcare will vary according to their individual biological development. For young people with progressive degenerative diseases, the loss of fine motor skills over time reduces their independence of performing key aspects of their care for themselves; however, their cognition is often unimpaired, and they are pivotal for training carers and informing health staff about the ways in which they wish the specific aspects of their personal care to be undertaken. For this increasing dependence on others to be supported, young people need adults who they trust and can build a rapport to be identified, and trained appropriately, in order that young people can become physically independent from their parents and move into adapted accommodation where their ongoing health care needs can be met by people other than their parents. This releases the parents from the daily responsibility of caring and the physical strain of moving and lifting their adult children at a time in their lives when they may be becoming frailer themselves. Young people's expectations of having their own homes

and being independent from their parents is also a powerful driver for the need for social care and housing services to meet the needs of young people with neuro muscular impairments to become adults and live away from their parental homes in suitable and adapted accommodation and with the trained carers that they need to support them (McGrath et al 2009, Stokes et al 2013).

Changes in local demographic data and patterns of referral of young people with progressive neuromuscular disorders were examined for one specific children's hospice in the north of England (Fraser et al 2011). This retrospective cohort study looked at the referrals that had been made for these specific diseases between 1987 and 2010. They concluded with two important findings. Firstly, they noted from their research that there has been an increase in the numbers of children from south Asian backgrounds being diagnosed with progressive neuromuscular disorders and secondly that the median age for survival for boys and young men with Duchenne Muscular Dystrophy (DMD) has increased to 24.7 years for those born between 1980 and 1989 and that this is continuing to rise. Therefore, as more young men with DMD are living into adulthood and more children are being born with a diagnosis of a neuromuscular disorder who will require palliative care provision, a review of current palliative care services will be needed to ensure that adequate services are available to meet the future needs of this group of children. These findings add to the current discussion within the world of paediatric palliative care, for the inclusion criteria for children and young people to meet the threshold for palliative care services to be revisited, as the needs for hospice services continues to increase in some areas of the county (*Together for Short Lives 2017).*

A Canadian study explored the needs of teenagers and young adults with cerebral palsy aged between 13 and 32 years of age in relation to their need for acute healthcare services (Young, et al 2007). This large study of 587 young people and 477 young adults identified that young people and young adults with cerebral palsy have complex and continuing healthcare needs that need to be provided for with services that are comprehensive as they grow up into adulthood. They identified that at the time of the study, there were gaps in the health provision available in adult services to meet the needs of young people needing to make the transition between services. These findings concur with the findings from other studies exploring the lived experiences of children and young adults of living with cerebral palsy which advocate for increased services for those young adults who are most severely disabled by their condition. Young people with cerebral palsy vary widely in the level of disability that

they are living with, with some young people having additional complexities of vision or hearing impairments or epilepsy, which also need to be taken into consideration when their preparation for transition to adult services is planned. The complexities for young people with cerebral palsy is that the different services that they use may have different thresholds for transition to adult services, which can mean that they make the transition to adult services for one speciality whilst remaining in children's services for a different aspect of their care.

Research has identified the complexity and the importance of neurodevelopment during adolescence. Cognitive neuroscience is an important and growing area of research into the impact that the physiological changes in brain function has on the developing brain during puberty and the adolescent years. The research findings add weight to the current professional position nationally and internationally, that transition to adult services should be a stage of change which is flexible and titrated to meet the individual needs of a young person. This is quite different from the current situation in many clinical services in the UK, which require young people to move to adult services at a pre-determined age.

2.8.3 Transitional resistance to adult care

Many of the studies reviewed also highlighted the needs for young people and parents to be more thoroughly prepared for their journeys of transition between health and social services, just as young people are prepared throughout their childhood for their transitions within educational provision. The Royal College of Nursing issued guidance in 2004 and 2013 highlighting the importance of young people being involved with and supported in their transition between services and that the transition experience should be planned for well in advance of the transition taking place. The guidance advises that initial conversations concerning the future plans for transition to adult services should start in the early teenage years and during mid and late adolescence should continue to build the knowledge, skills, and confidence of the individual young person, until by late adolescence, they are prepared and ready to move over to adult services. However, the reality for many young people is that the process is rushed, with a lack of information or conflicting information made available to them, which in turn reduces their confidence and readiness for the transfer to adult services when the time is deemed suitable. Some young people and parents have reported good experiences with excellent planning and preparation made available to them, which increased their self-esteem, knowledge base and skills and helped parents to prepare for relinquishing

some of the elements of control for their child's care that they had previously held responsibility for.

The Nuffield Trust (2016) issued guidance concerning new models of care for child health services which acknowledged the improvements that have been made in child health provision within the UK over the past decade but highlighted the needs for improvements to be continued and for the health needs of young people to be prioritised and inequalities in funding and resourcing of services for young people to be addressed. They proposed that a national children and young people's strategy should be introduced to challenge and change the ways in which current provision for young people's health and social care needs are met and resourced. One example of the inequalities of funding to support children with continuing healthcare needs, is the funding and resourcing of Community Children's Nursing teams. This inequity of provision results in parents seeking help by telephone from their local children's ward if the need arises; or taking their child back to hospital when the issue that is causing concern could have been dealt with at the child's home if staff had been available to attend to them out of hours.

The Care Quality Commission (2014) considered the current situation for many young people concerning their experiences of transition to adult services. They identified a continued lack of support and preparation for young people with continuing healthcare needs and identified the importance of family members or carers also needing to be supported during the transition process. They identified that the current structure of commissioning arrangements does not include the financial and staff resources required to prepare and support young people with their transition and that this needs to be addressed by local Joint Health and Wellbeing Strategies (JHWS) and in doing so that the health needs for children and young people should be prioritised.

Viner (2018) discussed the key needs for young people in relation to transition and identified what young people have identified that they want in relation to their transition needs. These identified requests identified the importance of a holistic individualised approach being provided for each young person. He endorsed the need for professionals to be trained in all aspects of adolescent health in both children's and adult services and proposed that the development of a shared philosophy between them would be beneficial for both young people and the staff caring for them.

Throughout the literature reviewed, several themes emerged highlighting that there is evidence from the data concerning the compliance of some young people with their necessary treatment regime which have resulted in detrimental effects to their health of moving from a service where the relationships with healthcare professionals were established and trusted; to a service where more autonomy was given to each young person for their health and wellbeing than the family centred care which forms the basis of the philosophy of children's health services (Watson 2000, Bell 2007, Berg-Kelly 2011). However, at some time, all young people who survive to adulthood will be required to move to adult services, and the more preparation that can be provided in advance of this taking place the better. The date of transfer between services should be known in advance and therefore should not come as a shock to the young person or their parents. This transition can be celebrated as a positive step for the young person moving into adulthood; but can cause a mixture of emotions for their parents as they relinquish their control over the day-to-day management of their child's health. The needs for any young person with a chronic illness are different from their neurotypical and unaffected peers; as they navigate the normal upheavals of the teenage years, with balancing the lifestyle choices of smoking, drinking alcohol, sexual health and the use of recreational drugs against the specific risks of their disease.

For young adults with progressive neuromuscular impairments, their levels of dependency for help with meeting their personal care needs and the management of the technology required to sustain their lives such as non-invasive ventilation will increase as they age due to the disease trajectory, as this can be difficult and distressing for the young person and their family to adjust to. Young people in these circumstances need to have the assessment, funding and appropriate resourcing for their individual healthcare needs to be met throughout their lifetime, by competent and caring support staff.

2.8.4 The experiences of using a wheelchair.

The experiences of children and young people of using an electric powered indoor /outdoor wheelchair was explored in a UK study with 9 children aged between 7 and 16 years (Gudgeon et al 2015). The study sought to gain an understanding of the participants of moving from a manual wheelchair to a powered wheelchair and the support and training needs to enable young people to become confident and proficient in using a powered

wheelchair. Certain aspects of daily life which included the negotiation of roadside kerbs, created stress for parents and young people initially. The findings demonstrated that young people described a range of experiences which had been both negative and positive as they negotiated a new life with increased freedom associated with learning to use an electric powered wheelchair.

A UK study interviewed 11 children and 24 parents concerning their experiences and quality of life assessments of using a wheelchair using the HQRoL (Health Related Quality of Life) framework (Bray et al 2017). The three themes of participation, self-worth and feeling fulfilled and health and functioning emerged from the data. These findings are a reminder of the specific needs of children and young people who have progressive diseases who become wheelchair dependent after previously being able to walk unaided or with support. Becoming wheelchair dependent can affect the self-esteem and confidence of young people as they become more dependent on others for assistance in their daily lives and as they seek to remain socially active with their friends. New opportunities at local sports centres may offer wheelchair basketball which children and young people can be encouraged to try.
The benefits and the challenges of becoming wheelchair users and the enhancement that results for the young person's independence are identified in these studies from the UK.

The above studies and guidance documents identify the importance of the transition experiences for young people and their parents. The timing of starting the preparation is of particularly importance; with the recommendations from the NICE guidelines (2016) that this process should start at the beginning of the adolescent years and be built on gradually to build the confidence of young people and their parents ahead of their move to new and unfamiliar services.

2.9 Parents experiences of their child being diagnosed with cerebral palsy.

The experiences of parents whose children had been diagnosed with cerebral palsy have been explored in different studies across the globe which gives a global perspective. An Australian study focussed on the coping, resilience and coping abilities and strategies of 102 parents who had children with Cerebral Palsy (Whittingham et al 2013). The grieving process for parents of their journey of emotional and psychological adjustment to the loss of the child that they had anticipated during their pregnancies was explored. The findings were consistent

with other studies and identified that parents continued to grieve in different ways and to different degrees for the losses in physical and cognitive development of their children and were able to discuss their own feelings of anxiety, stress and elements of guilt following their child's diagnosis. The need for sensitive communication between parents and professionals was identified, with the acknowledgement that specific events during their child's development could act as triggers for exacerbations of sorrow and readjustments to parental expectations and acceptance of the realities for their child's abilities. These findings concur with the findings from other studies which have also considered the emotional and psychological effects on parents and primary carers of a diagnosis of cerebral palsy, with some mothers feeling that the cause of the impairment for their child could somehow have been prevented by something that they did or did not do during the pregnancy.

Lima et al (2016) explored the issues of parental stress and support for caregivers of children with Cerebral Palsy in Brazil. This large study recruited 100 participants of whom the majority were the mothers of the children, although some grandmothers were also participants in the study. The study aimed to ascertain the role that parental educational level, family income, parental occupation and material status played in the ways in which families adapted and coped with having a child with Cerebral Palsy. The severity of the child's impairment did not seem to have a significant bearing on the family's ability to cope, but the level of social support was much more important.

The needs for ongoing support following the diagnosis of Cerebral Palsy for the whole family concur with the findings from other studies in this area of research; and are important for service providers to take account of when health and social care services are developed and resourced. Many families struggle to find respite care for their children who have a diagnosis of Cerebral Palsy and current funding cuts have affected the provision of holiday clubs and after school activities for children with additional needs across the UK (Special Educational Needs 2018, NICE 2019). The emotional and psychological impact on parents of having a child diagnosed with Cerebral Palsy can be enormous. With high levels of stress and anxiety and experiences of grief and guilt evidenced from the findings in the research.

2. 9.1 Authoritative parenting when a child has a disability.

The stress that can arise within families when a child has a disability was explored in a UK study by Woolfson et al (2006). In this study, 53 parents whose children had been diagnosed with a developmental impairment were recruited to a study with 60 parents whose children had no obvious developmental issues. The findings from the study showed that the parents of children with a developmental disability demonstrated higher levels of stress than parents of children with no disability. Some parents with children in the younger age group found using an authoritative parenting style helpful, but for parents of children in the older age group the reverse was true. For parents of the non-disabled children, the parenting styles did not appear to make a difference.

This study is helpful in reinforcing for health and educational professionals the importance of offering parenting courses and advice to support parents in providing a nurturing but appropriate parenting style with consistency and boundaries when their child is identified as having a disability. However, the experiences from parents indicate that although such parenting courses might be available in their local area, there are often long waiting lists for spaces on these courses to become available. Health Visitors have expertise in parenting skills and techniques for young children and might be helpful in providing information and advice in the interim period until the parents receive a place on a specialised parenting course.

2.10 Expert parents

For the parents of many children with complex medical histories which require constant assessment and management by default, these parents become the experts in their child's wellbeing as they navigate the complexities of learning medical terminology and skills to keep their child alive and negotiate and communicate with different health and social care professionals across primary, secondary, and tertiary health services, education provision and social care.

A large study from the Netherlands explored the position of parents as experts for their children with complex needs (De Geeter et al 2002). Data for this study was collected from 723 questionnaires that were sent to parents who had been purposefully identified as having a child with multiple disabilities. Two research groups were created with one group of parents

within special schools being offered the opportunities to meet regularly with school staff and to be equally involved with setting the educational goals for their child; whilst the second group of parents no changes were implemented. The findings reported that the parents who were more involved with the goal setting for their child increased their own knowledge concerning the options and possibilities. The findings identified the importance of parents being recognised by professionals as being the experts for their children, with their knowledge, views and experiences being taken seriously and factored into the individual decisions being made about each child.

The ways in which the parents of children with complex health needs are supported to be fully involved in the decisions that are made concerning their child in health and social care in the UK, was investigated by McNeilly et al (2017). This large study included 77 parents of children and young people with a range of impairments in Northern Ireland. The data was gathered using a mixed method approach of both surveys and semi-structured interviews which aimed to explore the lived experiences of parents of being involved with the decisions that had been made about their child. Some of the factors that caused levels of stress for parents will be discussed below.

One area in which parents become 'expert' is that of managing enteral feeding. For parents, the realisation that their child is unable to take sufficient calories orally to meet their own nutritional needs through eating orally and the decision for enteral feeding to be commenced can cause distress with many parents associating the commencement of enteral feeding with further evidence of deterioration in their child's condition. A Spanish study exploring the levels of distress exhibited by 58 mothers of children with neurological impairments who were being enterally fed, found that 53% of the participants demonstrated high levels of stress and anxiety from the levels of responsibility they had taken on in delivering the enteral feeding that their children required (Pedron-Giner et al 2013, 2019). The recommendations included providing greater support for parents when their child is being enterally fed. One of the challenges, however, is understanding the ways that parents want additional support to be available for them, and whether if additional support services were developed, parents would use them and find them to be beneficial in providing the advice and reassurance needed to improve their confidence. In the UK there are home enteral nutrition specialist nurses who parents can contact for extra support, although this may not be available in all countries.

There is little research on the concept of the 'expert parent', however, as many more children are growing up dependent on technology for their survival, parents are needing to develop expert knowledge which often goes beyond the expertise of their GP's and local hospitals. Smith et al 2013 undertook a review of 34 studies of parents' experiences of caring of their children with chronic conditions. Parents identified not always being able to gain the information about their child's condition and not feeling valued in the vital role that they played in caring for their child. Caring for a child with invasive or non-invasive ventilation, is a highly specialist area of medical care, and for young people with progressive degenerative neuromuscular conditions, mechanical ventilation becomes their lifeline as their respiratory muscles weaken, and they become unable to effectively breath unaided. Parents take on roles and responsibilities that are alien to them, but which are essential to enable their child to live at home rather than remain in hospital or move to residential care. However, the levels of stress that this can cause can be very significant, and parents need ongoing support to support their mental health and wellbeing.

2.11 The role of children's hospices

The numbers of very sick children who are technology dependent in the UK are increasing year on year, as technology advances and children who have a shortened lifespan are living longer (Fraser et al 2011, Fraser et al 2014). These children need high levels of technological expertise. In more recent years, and the development of community children's nursing teams and children's hospices have increasingly provided opportunities for children to be cared for at home with a comprehensive package of care. Individualised care packages involve training parents, care assistants and nurses to have the knowledge and confidence to look after children who are ventilator dependent or have tracheostomies at home and at school where appropriate. Part of the planning of a package of care for technology dependent children and young people involves local children's hospices, where the training of the family and care staff can be continued prior to their transfer home and the family and siblings can visit more easily, stay overnight if needed and relax in an environment that is less clinical and acute than a hospital ward.

For many years, successive policy documents have advocated that sick children should be cared for at home wherever possible, but this is not without its challenges, and a care package for a child who is technology dependent can take many months to plan and prepare for. This

may require the family to be rehoused if the existing housing is not suitable, or for the existing home to be adapted to enable the necessary equipment to care for the child to be accommodated. Either of these options can take many months or years to accomplish, during which time, a child who is medically stable may need to remain in hospital or in a children's hospice. This becomes extremely costly in financial terms but also in the emotional and psychological costs of the family being separated for long periods of time and the stress and distress that this can cause to the parents and the siblings in the family of them being separated.

The first children's hospice in the UK opened in the 1980's with a steady increase in numbers over the past decades, however they are fewer in number than adult hospices and are spread out geographically and families may have to travel long distances to access the specialist services and facilities that are available there. Forty – four children's hospices now exist in the UK, and they offer high quality palliative and end of life care for children with life limiting conditions and support from art therapists, play therapists and Psychologists for the parents and siblings in the family. Hospice provision enables parents to meet one another, to share ideas and provide valuable support for one another. However, each children's hospice sets inclusion and exclusion criteria for acceptance of a child for their services, and whilst many children and young people have very significant physical and learning disabilities, they may not be living with a life limiting condition and therefore would not meet the threshold for acceptance.

A study by Champagne et al (2012) in Canada explored the experiences of 33 parents from 25 families whose children had received respite care from a children's hospice. The findings demonstrated that the parents found great benefits from allowing their children to receive respite care at the hospice; particularly the high-quality care that their children received from the staff at the hospice and the opportunities for the parents to rest. The importance of rest for primary care givers is crucial for the ways in which parents' cope with the relentless responsibility of caring for a child with complex needs. However, many families struggle to find family members or friends who are willing to care for their child on a temporary short-term basis when they are technology dependent or have an unstable medical condition.

One UK study explored the role that children's hospices can play in moving a child whose life is dependent on technology, from hospital to a children's hospice and preparing the child

and the whole family to move home (Price et al 2017). Interviews conducted with 5 parents and focus groups with 26 professionals explored their experiences of providing and receiving this "stepped down" approach to the provision of care. The parents who participated reported that their children had stayed in hospital from between 3.5 months to 2 years whilst the planning and preparation for their child to be transferred to the local children's hospice had taken place. These lengths of time disrupt family life dramatically and may cause financial and relationship difficulties for the parents. All the parents reported the benefits of being in the more relaxed and calmer environment of the children's hospice.

Children's hospices have the additional benefits of play specialists, music therapists, soft play areas, specialist play equipment which can accommodate a wheelchair and a relaxation room with fibre optic lights and choices of relaxing music to enhance the quality of the children's lives. However, depending on the complexity of the child's condition, there may be a delay whilst the hospice prepares the staff and the equipment required to prepare for the child's transfer from the acute hospital setting. The results of this study demonstrated the perceived benefits of the parents of moving from an acute hospital unit to a children's hospice in the transition phase of the child and their family moving to their home.

When children are diagnosed with life limiting illnesses, the views of their parents in the care that is offered to them and is available for them is paramount. The children's hospice movement aims to provide high quality care to the child, siblings, parents, and extended family for the duration of the child's childhood, until either they die, or they are transferred to be under the care of an adult hospice as a young adult. Children's hospices have strict criteria for the thresholds of acceptance of a child to receive the high-quality provision available within a children's hospice. This criterion includes that a child has a life limiting condition and has a shortened life span. Each application is reviewed by the hospice team and if assessed as being suitable, a decision is made as to the number of days of respite care which can be offered to support the child and family. Some children's hospices also have community nursing staff who can offer a designated number of respite support in the child's own home (Ling 2012, Brombley 2015).

The role of children's hospices in providing high quality expert care and support for children with life limiting conditions and their families is crucial. Children's hospices provide high quality respite, palliative, and end of life care for children across the Western world,

however, the need for more hospices to meet the growing needs of children whose lives will be shortened by their disease has been identified by researchers in the field. Parents and siblings benefit from the psychological and emotional support, which is provided from hospice staff, however the strict thresholds for referral acceptance, mean that some families will not meet the inclusion criteria for their child to be able to benefit from these specialist services.

2.12 Sexual health and intimacy experiences for young people with physical disabilities

The importance of sexual health and the sexual experiences of young people with complex health needs is an important area of research, as health professionals consider the physical, emotional, psychological, and sexual health of young people. The research findings and discussion have highlighted a less well identified issue which is central to the lives of all young adults.

A Canadian study focused on the sexual experiences of 11 young people with Spina Bifida aged 16 to 25 years (Heller et al 2016). The findings identified concerns from young people that their physical disabilities might influence potential sexual partners before being recognised for who they were as individuals. However, following sexual experiences, the participants were able to recognise a rise in their confidence and that they felt more empowered to discuss their disability and sexual needs than previously. In common with other research studies, the participants expressed their wish to increase their knowledge about the mechanics of having sex and ensuring their sexual health within a nurturing and supportive relationship. This study reminds health professionals of their responsibilities to provide sex and relationship education to young people with physical and intellectual disabilities, to enable them to have the knowledge and confidence to have an intimate physical relationship when the time is right for them.

A large American study explored the range of sexual experiences that young people with physical disabilities had the opportunity to experience (Khan et al 2018). The findings demonstrated that those young people who had self-identified as having a severe physical disability had a significantly lower level of sexual experiences than their neurotypical peers. The recommendations were that this knowledge should be used in the development of future

information and polices which are developed to support the psychological and emotional wellbeing of adolescents.

An American study by Secor-Turner et al (2017) used data submitted as part of a wider study called *"My Path"* which researched the experiences of transition by young people with impaired physical mobility. The findings concurred with previous studies on the same topic which stated that young people felt under prepared and lacked the information that they needed about sex and sexual health, to act as a basis for a healthy lifestyle. The challenges for health, education and social care providers are to know how and when this level of information should be provided and to ensure that this matches the cognitive abilities of the individual young person. This American study has implications world-wide, as parents and health and social care providers acknowledge the needs of young people with physical disabilities to be as well informed as their non-disabled peer group. Additionally, some parents may need support and information from health professionals to prepare them to answer questions from their children, which must include information about contraception and sexual health protection and any contraindications with contraceptive options and the medication that their child has been prescribed (Dickson et al 2017).

Many young people with physical disabilities are sexually active and will become parents themselves as adults. Young people with genetic conditions need to have the opportunities to have genetic counselling offered to them and be able to make informed decisions about their plans for future pregnancies. In the past decade there have been several governments and local polices seeking to highlight and address the needs of adolescent health (Care and Quality Commission 2014).

Some services offer transitions clinics which are jointly staffed between adult and paediatric health staff and are valuable in smoothing the transition from one service to another. Transition clinics offer the opportunity for young people and parents to meet and begin to develop confidence and trust in staff who are new to them, and for young people to develop their confidence in seeing health professionals on their own and becoming more involved in the decision-making processes concerning their health and the care options that they are able to receive (Kennedy et al 2008, Baines 2009, Allen et al 2011, Berg-Kelly 2010, Bundock et al 2011). However, the costs of setting up and resourcing transition clinics together with the rarity of some medical conditions, results in transitions clinics not being available in all areas

of the country or necessitates young people and their parents travelling for long distances to enable them to access specialist provision to meet their specific needs (Muscular Dystrophy Association 2009). The commissioning of services for young people is a complex issue and where a particular disease is rarer, expertise for the treatment and management of that specific condition is centralised which causes challenges for patients needing to travel in order to access specialised expertise and care to meet their specific health needs (Healthcare Commission 2007).

Parents and carers are crucial for the survival of children with ongoing healthcare needs during the childhood of each young person and the knowledge of the condition and the technical skills needed to care for the child are complex and arduous including the use of enteral feeding equipment, suction machines, care of tracheostomies and for some young people with neuromuscular conditions the necessity for non-invasive or invasive ventilator support.

The demands on the NHS and cuts to budgets in national and local services, have continued to have a negative impact on the daily experiences of young people with continuing care needs. Current reports from NHS England of the plans to make drastic cutbacks to current services to make major financial savings and to improve efficiencies in services, (The Guardian 2016) will require ongoing adjustment for health staff and their patients and families. The commissioning of services is crucial within health and social care provision and yet the current service provision in many areas is that young people are expected to move to adult services after the age of sixteen years for inpatient provision or up to eighteen years in some services and nineteen years in others.

Some have suggested that a paradigm shift is required when the planning and commissioning of services to meet the needs of young people with disabilities (McDonagh et al 2006, Hamdani, et al 2011) and yet with budget constraints and many Trusts currently overspent and in financial difficulties; the complexities of addressing health needs and commissioning health services to meet the needs of the local population cannot be underestimated.

The NHS Five Year Forward review (NHS England 2014) identifies the importance of patient safety, clinical effectiveness, and patient experiences, as being key objectives to be achieved by the NHS in England over the next five years. However, the complexity for young

people with neuromuscular disorders is that the numbers in this client group may preclude the development of specific services in each area which results in young people having less clinical contact and input from specialist health professionals in their local area and necessitates significant financial expenditure to gain the reviews and treatment that they require.

One key transition for young people who have been able to access respite services through their local children's hospice, is the transition from child to adult hospice inpatient and outpatient services. However, the referral and acceptance criteria by hospices varies widely and differs across the country, which for some young people results in them being left without the ongoing psychological and physical support that they have come to know and benefit from in the services provided by their local children's hospice. The emphasis within the children's hospice philosophy and provision is the longer-term palliative care journey from diagnosis to end of life care which may be many years, whereas the focus in adult hospices is symptom management and end of life care.

Some children's hospices have extended their service provision to include young adults with life limiting illnesses up to the age of twenty-five years but by no means all have been able to provide and resource this. However due to the charity status of and their reliance on charitable donations, children's hospices need to be able to clearly identify which services and employment of professionals that they can afford to provide, and this service provision includes the employment of clinical psychologists, music, and art therapists in addition to nursing and healthcare support staff (*Together for Short Lives 2015).*

The emphasis from successive governments from national policies from the Department of Health and from local policies during the past decade has been and continues to be the importance of seeking and capturing the views and experiences of service users and the strong message that service users should have access to local health and social care services that are tailored to meet their individual needs (, DH 2008, Health and Social Care Act 2012). However, capturing the views of service users is only one part of the process; funding and resourcing what is requested may not be financially achievable.

Viner (2007) undertook a large UK study of 8855 young people aged twelve to seventeen years who had been inpatients in either child, adolescent, or adult wards. The objective of the

study was to see whether adolescent inpatient wards make a difference in relation to the quality of the care received and the overall patient satisfaction of their inpatient experience. The conclusions from both the younger and the older adolescents in the study were that dedicated adolescent wards improved the involvement of the young people in their overall care including their involvement in their decision making and that their patient satisfaction was higher than their experiences of being on either a children's ward for the younger teenagers or an adult ward for the older ones.

Research from studies which have considered the needs of young people in relation to their experiences of transition to adult services have consistently agreed that preparation should begin early and should focus on developing each individual young person's knowledge about their medical condition, medication and required therapies such as physiotherapy and build on their existing knowledge and coping abilities with the focus of building their independence (RCN 2004, van Staa et al 2011).

It is to be hoped that the new guidance from NICE (2016) concerning the transition process for young people moving from children's to adult services which highlights in 11.2 that *"transition support is developmentally appropriate taking into account the young person's maturity, cognitive abilities, psychological status, needs in terms of long-term conditions, social and personal circumstances, caring responsibilities and communication needs"* (p4) will help to move this complex debate forwards and will be influential in raising awareness of this important issue and overall that the standards of provision and the transition experiences of young people with continuing healthcare needs will be improved.

Sexual intimacy is a normal part of the human experience, and adolescents commonly crave the experience of being in an intimate relationship with another young person. The research highlights the specific needs for disabled young people to have access to appropriate sexual health information and services, to enable them to make informed decisions concerning their sexual health and sexual activity. Young people reported needing access to information and had felt less informed than their non-disabled peers. As part of sexual health and decision making, young people with a genetic cause for their disability also need access to genetic counselling services.

2.13 Chapter Summary

This chapter has discussed the strategies that were used to undertake a review of the research literature and current polices concerned with the experiences of children and young people with neuromuscular conditions. I have considered the literature concerning the experiences of young people living with different health conditions. The literature presented has explored the experiences of young people and their parents of managing complex and fluctuating health issues and the physical, psychological, and emotional impact that this can have for the whole family. The literature has identified the current positions of commissioners and government in relation to the provision and support that should be available for young people with complex and chronic illnesses and has examined the impact that these policies have on the daily lives of individual young people. The consistency of the themes that have emerged concerning the need for robust transition planning and preparation for both young people and their parents is compelling. The importance of health staff receiving adequate training about the complex biological and physiological changes that take place during adolescence and the impact that the neurophysiological changes have on the decision making and coping abilities of young people supports the need for appropriate services for young people to move on to. Several of the appraised studies have identified the need for further research to be carried out and for service provision to be reviewed and improved where necessary. These findings have been endorsed in recent policy documents concerning the health and wellbeing of young people in the UK (DfE 2014, NSF 2016), however current financial constraints within the UK and the increasing competition for funds to resource services across the life span; continues to pose challenges for the commissioners and providers of health services to meet the specific needs of young people.

Chapter 3: Methodology and methods

3.1. Introduction

In this chapter the study design, methodological underpinning of the research and the methods and that were chosen to undertake the study are presented. I begin the chapter by identifying the aims and objectives of this research study. The chapter then progresses with a discussion of the epistemological and ontological position that I adopted, followed by discussion on the study design and decisions that were made as the study progressed and how I came to use case study methodology. This is followed by a discussion on methods of sampling and recruitment and the challenges that arose in relation to this study. The data collection methods and the stages of the data analysis are then presented.

The aim of the study was to explore the experiences of young people with a neuromuscular impairment growing up into adulthood, and to understand the experiences of their parents or primary caregivers. The study objectives were to:

- Explore and understand the perspectives of young people with neuromuscular conditions.
- Explore and understand the experiences of parenting for parents or primary caregivers of young people with neuromuscular conditions.
- Gain an understanding of the issues and challenges encountered by young people with neuromuscular conditions and their parents or primary caregivers.
- Explore the transition experiences of young people with neuromuscular conditions moving to adulthood.

3.2 Epistemological perspective and research approach

Epistemology is defined as *"a branch of philosophy concerned with the theory of knowledge. It attempts to provide answers to the question, "how and what, can we know?"* (Willig 2008 p2). With regards to neuromuscular conditions, health professionals generally understand the physiological processes concerned with genetic and non-genetic neuromuscular conditions. This is the basis of the *biological evidence* for the symptoms and disease progression that young people are living with; however, another important aspect of concerns the lived experiences of young people with neuromuscular conditions experience. A further important aspect of the findings concerns the lived experiences of young people experiencing their disease in their own bodies. This can only be understood through qualitative research which

gives participants the opportunities to express their life journeys in their own words. The recounting of their experiences is unique to each individual, and through qualitative research, quality of life and hopes and dreams can be explored, and greater understanding gained of the priorities of each young person in living their lives to the best of their abilities.

My position of being an experienced nurse with extensive knowledge in this field, and twenty-six years' clinical experience of working in a range of health care settings in both primary and secondary care, gave me one professional perspective. In undertaking this study, I have attempted to gain an understanding of the experiences of the participants, through the paradigm of qualitative research and the lens of previous clinical experiences, and in doing so, I have recognised that there are different realities for the participants of their experiences, which are presented in the two findings chapters. As a nurse, on the one hand, I lean towards positivism in the sense of recognising the evidence of medical and scientific research and the evidence that underpins the establishment of medical a diagnosis of individual children and young people in this field. However, as a qualitative researcher, I lean towards the paradigm of constructivism (Flick 2009), whereby I recognise and acknowledge that there can be multiple realities for different individuals as they live their lives with the same diagnosis, and that each individual constructs their own reality of their life and circumstance and that this changes over time (Willig 2010, Petty et al 2012).

Constructivism is an epistemological position which acknowledges that knowledge is generated by individuals (Willig 2010). This is a philosophical and scientific position that knowledge arises through a process of active construction (Mascolol et al 2005). Stake (1995) identifies that: *"A constructivist view encourages providing readers with good raw material for their own generalizing. Constructivism helps a case study researcher justify lots of narrative description in the final report" (p102)*.

The principles of constructivism are that knowledge is constructed and not transmitted and that the initial understanding that is gained by the researcher is local rather than global (Wilson 1996: 23). For this study, I was constructing new knowledge and understanding of the experiences and challenges of the participants that they faced in their day-to-day lives of living with neurological condition or being the mother of a child with a neurological condition. Using a constructivist approach allowed me to interpret the narratives and

observations from the data sources and the richness of the data allowed the voices of the participants to speak to the readers themselves.

3.3 Ontological position

Ontology is defined as *"the science or study of being or existence and its relation to non-existence……Ontology deals with what is real (versus fiction or appearance), what is the nature of reality or matter"* (LoBiondo-Wood et al 2006 p 134). Ontological positions can be either "realist" whereby it is accepted and acknowledged that the world is orderly or that different objects and aspects of life have a causal relationship one with another; or "relativist" where there is recognition that a range of different interpretations can be applied to different situations (Willig 2008). Ontologically I have adopted a relativist position, and in doing so acknowledge and accept that whilst different participants in the study may have the same diagnosis; there are likely to be wide variations in the trajectory of their individual disease progression and family, social and cultural influences which will result in different interpretations and range of experiences being applicable to each individual family.

Therefore the relativist constructivist position that I have taken in this research, has meant that the richness of the data that I have gathered from the interviews and the questionnaires, together with my observations during each of the interviews and the additional data of photos and videos which two of the mothers chose to show me; have enabled me to combine the ontological relativist position of acknowledging the differences and uniqueness of the experiences of each of the participants together; with the epistemological constructivist position of constructing knowledge about the lives of families living with a progressive or non-progressive neurological condition.

3.4 Qualitative paradigm and study design

Qualitative research enables researchers to explore the experiences, feelings and emotions concerning a specific aspect of the lives of different groups of people (Silverman 2016). Although the numbers of participants in qualitative research are generally small, the richness and depths of the data obtained through qualitative research approaches, enables researchers to compare the findings with other qualitative research study participants and gain a deeper understanding of the lived experiences of individuals who share the same disease, social

circumstances or life experiences (Gerrish et al 2006, LoBiondo-Wood et al 2006, Benner et al 2008, Flick 2009, Silverman 2016).

In qualitative research the researcher seeks to understand the natural setting where participants are living their lives, have the lived experiences of being patients, relatives or participants choosing to live in a certain location and adopting a particular lifestyle, which is applicable to the research (LoBiondo-Wood et al 2006). The wide variety of qualitative research methods allows researchers to select a research methodology that provides the best platform for the investigation, exploration and understanding of the area to be studied and to provide illumination and explanation for the often complex and multi-factorial issues that are taking place and contributing to the lives and experiences of the research participants (Gerrish et al 2006, LoBiondo-Wood et al 2006, Joubish et al 2011). This research study aimed to add to the body of knowledge and to address the current gap of qualitative research concerning the experiences of young people with neuromuscular impairments. Choosing a qualitative methodology for this study was also influenced by my desire to collect the narrated 'stories' of my participants. Using a research diary in the form of notebooks to document my thoughts and ideas from the beginning of my PhD journey was useful for designing the propositions for the study and for making notes before and after each of the interviews. It also helped me to document my thoughts before and after discussions during supervision which enabled me to confirm my epistemological and ontological positions as the research study continued (Clark 2009, Engin 2011).

The overarching consideration that governs research projects concerns the planning and the design of each study. In the design and planning of any research project, the attention to detail and the choices that are made for the data collection and analysis of the data obtained are crucial for the abilities of the researchers to be able to answer the questions posed for the study with the greatest clarity possible (Green et al 2004, Hennink et al 2011, Silverman 2014). Therefore, the research questions being posed, and the information being sought should determine the methods and methodology chosen to structure and guide the research processes for the researcher and be best able to answer the research aims and objectives (Green et al 2004, Hennink et al 2011, Silverman 2016). The initial stages of the consideration and planning of the design of a research study, which culminates in the final choices of methodology should allow for the data collection and the results yielded and

analysed to provide answers to the questions posed in the study with the greatest clarity possible (Pope et al 2006, Bowling 2009, Polit et al 2012).

In the design of this study, both the methods of data collection and the time and location of the data collection were carefully considered. I was aware that many young people with neuromuscular disorders have limited mobility and some are wheelchair users. In addition, many young people have busy lives with school, college, university, work, or social commitments in addition to attending outpatient appointments with therapists and medical clinicians. Therefore, the intention was to design a data collection method that provided flexibility for the young people that would enable them to answer the open-ended questions being posed over a specific time frame of four weeks; at a time and pace that would suit each young person's specific needs considering potential fatigue and needs for rest and would enable them to carefully consider their responses to each of the questions being posed. As part of the decisions made regarding the design of the data collection methods chosen, I did not want to cause additional stress to young people or their parents or carers, by requiring them to meet me as the researcher on a specific day or to travel to a specific location to be able to participate in this study if they chose to do so. Therefore, the flexibility of using computers for data collection provided privacy and was a medium that the young people were familiar with from school and, with the advances in technology such as the use of *"eye gaze"* communication programmes (Borfestig et al 2016), enabled potential participants to have the freedom to be honest about their thoughts and feelings about their individual lives and ambitions which were then sent confidentially to the researcher.

The design of the study was finalised following careful consideration and consultation with professionals working in this area of practice, and from personal previous clinical experiences of working with young people with neuromuscular disorders. Consideration was given to which methods of data collection would yield the best response rate and the richness of data that is being sought to attempt to give meaning to the lived experiences of this specific group of young people and their families.

There were several different approaches that could have been taken within the paradigm of qualitative research, which include grounded theory, ethnographic research, case study, and phenomenology (LoBiondo-Wood et al 2006). Although grounded theory and phenomenology, as potential methodologies for this study, were considered in the early

stages of the study, the decision to use case study methodology was to enable a detailed examination of each case individually and then of the eight case studies collectively, seeking to understand the similarities and differences between the different participants and how these related to existing research and theories.

3.5 Case Study methodology

Case study methodology *"investigates a contemporary phenomenon within its real-life context, when the boundaries between phenomenon and context are not clearly evident"* (Yin 1994 p135), and thus can be used to provide an in-depth study of one or more individuals, organisations, or situations to gain understanding and clarity of the similarities and differences that are apparent. Yin's (2014) definition of case study methodology fitted well with the study aims and objectives. Case study research looks specifically at the context of the experience or the situation under scrutiny and considers what bearings the context has on the overall data that is collected and allows a holistic yet flexible approach to be used (Clarke et al 2006). This methodology focuses on the minor and major details of each case and seeks to gain understanding of how an organisation or individual participants live their lives and to ascribe meaning to the findings gathered from the data (Willig 2008).

The complexities together with the similarities and the differences, in the lives of young people with neuromuscular impairments and their parents was worthy of scrutiny and needed to include an exploration of the small details of their lives, in addition, for parents, to the large events which they had faced and adjusted to over the years of their children's lives. Being able to meet two mothers with younger children in their own homes, enabled observation of *"the real-life context"* identified by Yin (1994 p135), and gave a different perspective and an added dimension of understanding, which I was unable to gain from the interviews with the three other mothers which took place outside of their homes. I was mindful that each young person and parent was an individual, and each of their family units differed widely, therefore their experiences were unique even when their diagnosis was the same. Case study provided me with the opportunity to explore the details of the complexities for young people and their parents of living with the complexities of a neuromuscular impairment.

3.5.1 Case study boundaries

A clear definition of what the case or cases are in case study research is needed at the start of each research study, in order that the researcher is able to complete the research within the allocated time frame for the research (Stake1995, Baxter et al 2008, Simons 2009). In addition, having clear boundaries, also enables researchers to be able to balance their time effectively and ensure that the stages of data collection, analysis and writing up the research is able to be completed in the time frame set (Merriam 2009, Stake 2013, 2014). At the outset of research using case study, the definitions of what the *"case"* or *"cases"* in the study are were clearly identified and defined, with clear boundaries for what are being identified as each *"case"* being stated (Hancock et al 2011, Simons 2009, Merriam 2009, Stake 1995, Yin 2009).

For this research study, central to the cases were a child or young adult who had a neurological impairment. I initially planned to recruit only young adults and any of their parents or primary caregivers (e.g. foster parents or Grandparents), as participants in the research. Due to the difficulties in recruiting young people who met the inclusion criteria for the study, ethical approval was sought to include mothers of children younger than sixteen years of age. This approval was granted, and three mothers of younger children were subsequently recruited, each of whom had sons aged seven years, seven years, and fifteen years of age respectably, who each had neurological impairments. Fathers were included in my recruitment approaches, and would have been welcomed as participants, however, unfortunately no fathers came forward to take part in the study.

Additional data that was used as it was made available for each of the cases, which included an interview with the Transitions Coordinator at a local children's hospice who had known Lauren for several years, email correspondence from both Elizabeth and her mother, emails and text messages from Lauren, and photos and short video clips of Lucas and Andrew which were shown to me by their mothers (see methods and finding for more detail of case material).

3.5.2 History of case study methodology

Case study, as a research approach, has been used in medicine, psychology and sociology for many years, and provides an approach by which situations or the lives of individuals or

organisations can be used as individual or collective case studies (Stake, 1995, Hancock et al 2006, Yin 2014, Silverman 2016). This methodological approach enables a depth of knowledge and understanding to be gained, and comparisons made to develop new meaning and understanding of a specific situation (Stake 1995, Hancock et al 2006, LoBiondo-Wood et al 2006, Simons 2009, Yin 2014, Silverman 2016, Thomas 2016). The history of case study methodology extends back over many years and originally was used in qualitative psychology and nursing research (Anthony et al 2009). Trustworthiness of this methodology is a concern and can be answered by the transparency and robustness of the study design and the meticulous execution of each stage of the data collection and data analysis of the study which in turn demonstrate the rigour and reliability of the research processes (Yin 2014).

Case study as a methodological approach is recognised to have gained in popularity in recent years, with more research using case study methodology being published (Anthony et al 2009, Hyett et al 2014, Yin 2014). The flexibility of case study methodology, in being able to be adapted to suit the research question, is acknowledged as being an advantage and can be viewed as one of the positive aspects of choosing this research approach (Thomas 2016). However, one of the main criticisms of case study has been related to whether case study should be recognised as being a research method or a methodology (Hyett et al 2014). This has been considered by different authors with the conclusion being that case study as a methodology must be supported by the robust framework of the choice of the case or cases being researched and a clear application of the principles of case study methodology evidenced (Hyett et al 2014, Yin 2014, Thomas 2016).

Further criticisms of case study, as a research methodology, has concerned the generalisability of the findings from small cases, although it has been argued that each case used in case study research should be viewed holistically for its uniqueness (Thomas 2016). An alternative perspective concerning the generalisability of the findings from case study research proposes that it is not always necessary for researchers to be able to generalise from the findings of research rather that the richness of case study methodology can provide insight and a new perspective of the research issue in its own right (Simons 2009, Thomas 2016). These new perspectives can add to the worldwide conversation of the research area and can act as a starting point for new research to be planned and completed. This perspective was useful in understanding and appreciating the value of the findings of the research study,

as in one piece of work, it enabled the voices of mothers and young people to be heard, understood and comparisons between their experiences to be made.

3.5 3 Types of case study design

Case study methodology is known and understood for its own design, data collection methods, and analytic processes and is still evolving as a research approach. The emergence of *"pragmatic case study"* (Fishman 2013), was initially designed to be used in research in psychotherapy, whereby rich data from case study methodology from each individual case is presented together with detailed information about what the psychotherapist did in terms of identifying and carrying out work with an individual client. This led to the development of a specific format which was used for reporting on pragmatic case studies in psychotherapy, whereby the treatment plan and course of the therapy delivered to the client was presented. This approach added to the rigour of case study methodology and allowing for greater comparison to be made between cases and for deeper analysis across cases to be carried out (Widdowson 2011).

Generally, there are three recognised major types of case study research designs. These are case studies that are exploratory, explanatory, or descriptive (Hancock et al 2011, Yin 2014). In exploratory case study research, the researcher is planning future research questions and identifying whether a future research study on a topic would be feasible. In the second type, that of explanatory case study research, the aim of the research is to discover the links between events and the outcomes for individuals. Descriptive case study aims to describe the depth of a phenomenon which is taking place in one specific setting (Hancock et al 2011, Thomas 2016). In addition to these three types of case study Stake (1995) identified *intrinsic case study*, whereby the researcher becomes interested in another aspect of a situation which they need to understand, aside from the issue that they had first intended to study. This issue then becomes the *intrinsic case study*. A fourth category of case study identified by Stake is *instrumental case study* which enables researchers to explore an issue which is linked to the original focus of their research.

For this study I used an explanatory case study approach identified by Yin (2014). Yin identified that there can be two main uses for explanatory case study. The first is to provide detailed descriptions and explanations of the lives of the participants and the second is that

explanatory case study can also be used to identify research questions which could be the focus of future research. For this research study, I have used explanatory case study to provide detailed descriptions and explanations of the lives of the five mothers and five young people who are growing up with a neuromuscular condition.

3.6 Methods

3.6.1 Recruitment

Initially I intended to recruit up to 10 young people with neurological disorders. The original inclusion criteria were that young people should be between the ages of 16 to 21 years of age and be living with a progressive or non-progressive neuromuscular condition. Before embarking on this study, my understanding from the literature was that young people may be viewed as a hard-to-reach group in terms of recruitment and data collection (Sydor 2013, Bonevski et al 2014). Given the nature and topic of the research study, the recruitment of the participants needed to be purposive, whereby participants who met the inclusion criteria for the study could be approached (Gerrish et al 2006, Boswell et al 2007, Suri 2011). However, finding young people with neuromuscular impairments who met the age criteria for inclusion in the study became more challenging than anticipated. See Figure 1 for depiction the recruitment and data collection processes and decisions that were made along the way (also see Boxes 1 and 2 below).

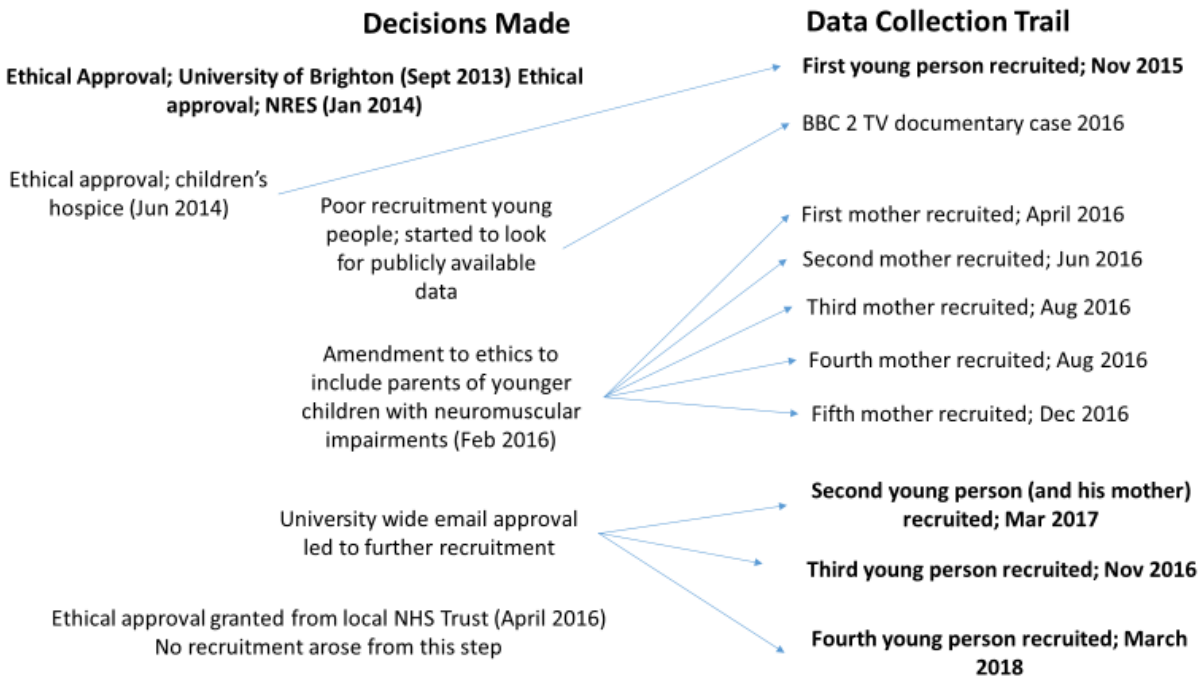

Figure 1: Flow chart of recruitment and data collection process

Recruitment of 5 young people:

- Lauren was recruited to the study from the local children's hospice in 2015. Her questionnaire was returned in November 2015.
- Nathaniel experience was from a BBC 2 TV documentary "Children of Helen House" and the documentary was transcribed in 2016.
- Elizabeth was recruited from the flyer advertising the study. She returned her questionnaire in November 2016.
- Edward and his mother were recruited following a university wide email in 2016. He returned the questionnaire in March 2017.
- Suzie and her mother were recruited to the study by word of mouth from a university colleague and Suzy attended for interview in March 2018.

Box 1: Recruitment processes for the young people

Recruitment of 5 mothers:

- Amanda was recruited by word of mouth through a colleague from the university wide email and did her interview in April 2016.
- Kim – was recruited by word of mouth from a colleague from the university wide email and did her interview in June 2016.
- Connie was recruited by word of mouth through a colleague and did her interview in August 2016.
- Caroline was recruited from the flyer advertising the study and did her interview in August 2016.
- Melanie was recruited from the university wide email and did her interview in December 2016.

Box 2: Recruitment process for mothers

Having worked as Community Children's Sister and as a children's nurse caring for many children with neuromuscular conditions in their homes, in inpatient wards and at a children's hospice, the starting point for the recruitment processes was to approach consultant Pediatricians and a local children's hospice for an initial conversation prior to applying for site specific research and development approval. I prepared and delivered a presentation of my research plan to a team of clinicians at a local children's hospice and to pediatricians at a regional specialist centre for children with severe neurological impairments. I then met with one of the consultant Pediatricians at a specialist regional children's hospital and discussed my research plan and proposal with him. At each of these events I received an enthusiastic

response, however detailed discussions then took place as to how the recruitment to the study could be carried out once site-specific research and development approval had been granted. The main concerns that were raised were related to potential the risks of coercion, if I as the researcher had presented the participant information sheets (See Appendix A and Appendix B) and consent forms (Appendix C and Appendix D) to the parents and young people myself. At each of the meetings, the professional opinions voiced were that the clinical staff who had an established relationship with the potential participants, should act as the *"gate keepers*" for the recruitment process. The ethics of whether clinicians should be placed as or should choose to be the gatekeepers for the recruitment of participants in research has been considered by different authors. Sharkey et al (2010) argue that for clinicians to act as gatekeepers to participants being offered the opportunity to take part in research is ethically indefensible, as it prevents suitable participants from having the autonomy to choose to take part in research projects if they wish to do so. However, as this study concerned children or young people, the strengths of this approach were that the clinical staff were known to the parents and young people in the relevant services and could hand out the flyers advertising the study (Appendix E) and the consent forms (Appendix C and D) and the information literature during outpatient clinics or inpatient admissions.

However, the limitations to this approach were that I was reliant on the good will of busy clinical staff, who were working under time pressures in their relevant services, to have the time to give out the recruitment packs to potential participants. In addition, the clinical staff may have been unable to answer any questions that parents, or young people may have had, which may have acted as a potential barrier to recruitment. If I had been permitted to attend clinical areas with relevant clinical staff, I could have worked with the staff to answer questions and allow potential participants to meet me and be able to put a face to the name that they were seeing on the participant information literature. Although I had been given an honorary contract with the local NHS trust for the duration of the data collection year, I was not permitted to attend in patient or outpatient areas to meet potential participants.

Prior to starting the study, through the local NHS commissioning unit, special needs schools in the local area were approached by a lead consultant Pediatrician, and one of these schools used their Twitter account and school newsletter to advertise the study. Through this approach, one young person contacted me via email and expressed her willingness to participate in the study, however in the following weeks, she withdrew from the study citing

academic pressure in the run up to her important exams as preventing her from having the time to participate in the study. A second young person who had previously attended the same school and who saw the announcement on Twitter, contacted me by email and became one of my participants. The children's hospice identified 26 potential participants who met the inclusion criteria for the study, and gave a participant information pack to each family, but this only yielded one young person coming forward as a participant. The one-page recruitment flyer, which provided details of the research study and my contact details was sent to the hospice (Appendix E).

Although the involvement of an advisory group is recommended by the National Institute of Health Research when a study idea is being developed (Involve 2011); in this study the development of an advisory group with young people, parental and staff representation at the children's hospice was not permitted prior to the proposal being submitted for ethical approval. This was denied because the Senior Management Team felt that to have an advisory group ahead of ethical approval being granted could have raised expectations of the young people, which could potentially have been damaging if ethical approval had not been granted. After discussion with my supervisory team, I gave the first draft of the flyer to the three non-disabled teenage children of a colleague and requested that they should give constructive feedback on the design of the leaflet. The young people advised a change in font style to make the advertising leaflet more visually appealing to teenagers, and changes in colour in the font for the different questions to make it more interesting to complete for young people. This feedback was useful and highlighted the importance of being able to undertake a pilot study of proposed research questions and approaches, in order that any potential barriers can be identified and addressed prior to the data collection being commenced. In response to the feedback given, I then amended the first draft and printed copies of the second version and included it in the information packs which were given to clinicians to give out to potential participants.

Due to the difficulties that I was experiencing with the recruitment of participants, I sought advice from my Supervisors and from the Doctoral College. Following advice from the Doctoral College, in 2016 a university wide email was sent out advertising the study (Appendix F), which yielded one mother of a child with cerebral palsy responding to the email and volunteering to become a participant in the study. Personal contacts from colleagues resulted in the recruitment of the remaining four parents and two of the young

people. As can be seen in Figure 1, recruitment of mothers with children with neurodegenerative diseases was much more successful from the university wide email that I was given permission to send. This may have been because the colleagues who passed on the information about my study to their friends knew me and could provide some level of reassurance about the aims of the study, the inclusion criteria for the study and could vouch for me as a health professional and member of the academic staff at the university.

3.6.2 Challenges in recruitment to the study

Aa well as the challenges outlined above, significant challenges in accessing potential participants were the requirement to use *"gatekeepers"* to different services to be able to access parents or young people to give them the information about the study (Heath et al 2007, Walker et al 2011), despite having been given an honorary contract by one of the local NHS Trusts. Finding ethically acceptable means by which potential participants can be approached and accessed, is a key element in the research process which must be addressed and overcome by any researcher if data collection is to be achieved, and the outcomes for a research study to be reached. Undertaking research with children and young people poses a separate set of challenges due to the need for obtaining parental consent for those under the age of 18 years and needing to ensure that young people have the mental capacity to consent for themselves to participate in the research being proposed (Fraser et al 2003, Graham et al 2015).

The issues of power and the potential issues of coercion are important issues to be considered in any researcher/participant relationship and need to be acknowledged and addressed. When the researcher is known to the potential participants there is the potential risk of participants feeling obliged to participate and may have concerns that if they choose not to do so that there may be adverse consequences to their relationship to the health professional concerned or to the care that they receive; even though the participation information sheets should make it clear that this would not be the case (Heath et al 2007).

The specific challenges with this study, were that despite going through the research and development procedures in different local services (both NHS and non-NHS) and being given the necessary approvals, access to meeting potential participants to the study was denied. The restrictions prevented opportunities for me to meet potential participants to be able to explain

the study. Therefore, I had to explain the study and leave the advertising information with busy clinical staff and ask them to make the initial approaches to potential participants on my behalf, which resulted in a prolonged delay in the recruitment stage of the study. I felt powerless to effect any changes and felt that I needed to comply with the restrictions being placed on my access to potential participants, in order that I would have some opportunities to recruit to the study, although the proposed route had been unplanned.

A local consultant Paediatrician identified six young people from his case load who met the inclusion criteria for the study and information packs and consent forms were sent out by post to them, but none were returned. The barriers to accessing potential participants did cause significant delays with recruitment which might have been avoided, if direct contact to potential participants had been permitted. One of the barriers to recruiting potential participants may have been the busyness and complexities of trying to juggle the demands of their daily lives which are likely to have been significant contributing factors. This was confirmed by one parent and her daughter during the data collection phase of the study, whereby changes in her daughter's medical condition, admissions into hospital and frequent hospital appointment culminated in considerable delays in their availability to participate in the study. However, later, the young person made email contact to confirm that she still wanted to participate and subsequently was able to return her completed questionnaire.

Another local consultant Paediatrician who specialised in neuro-disability and ran four clinics each week; was enthusiastic about the study and offered to speak to young people and parents/caregivers who met the inclusion criteria for the study and was hopeful that more young people would want to be participants in the study. Unfortunately, this did not result in any participants being recruited. One reason for this is likely to be that families are busy with their lives and may not have had the time or emotional capacity to engage with the study at the time at which they were initially approached, although at a different time in their lives, there may have been a different response. Avoiding the summer months when young people are particularly busy with exams was also a consideration in the timing of aiming to recruit participants to the study.

The decision to use the data from one young man from the television series *"Children of Helen House"* (a of BBC programme series featuring children and young adults receiving hospice care), came about because at that time I was struggling to recruit young people to the

study. Ahmed (2010) argues that using documentary evidence in research is acceptable and valuable in research and can be used in a wide range of research studies. In addition, as a young 21-year-old, Nathaniel gave an interesting perspective of the importance of sexuality from a male point of view, which as a researcher enabled me to gather important data with which to construct further understanding of the lives of both young men and young women living with progressive degenerative neuromuscular impairments. Choosing to include Nathaniel's experiences, also enabled the study to have more of a gender balance with two young men and three young women being participants in the study.

3.6.3 Snowball techniques in recruitment

Three of my participants were recruited using the snowball technique. Snowballing is a recognised and approved method within both qualitative and quantitative research, which is used effectively for recruitment with *"hard to reach groups"* (Gerrish et al 2006). Using this recruitment technique for research, one recruited participant gives information about the study to their friends or colleagues who meet the inclusion criteria for the research study in the hope and expectation that others will also choose to become research participants in the project (Sadler et al 2010). It is recognised that there are a wide range of contributing factors such as culture, belief systems, stigmatisation or low literacy levels which may make the recruitment of participants to some research projects more challenging than for others and that recruitment from some specific age groups which include children and young people may pose additional challenges (Sadler et al 2010). One of the advantages of using snowballing to access participants for research studies is that this approach can be seen as an ascending strategy, whereby individuals who work in the specific area of research, or have the specific condition being researched, know of others who fit the inclusion criteria for the study and can pass on information about the study on behalf of the research team (Atkinson et al 2001).

The advantages with utilising snowballing techniques from specific population groups are that a recruited participant is more likely to know how and where additional potential participants may be found and invited to participate in the research study (Sadler et al 2010). However, one potential disadvantage or limitation may be that the recruited participant may not want to be involved in recruiting others due to the time factors involved or not wanting to lose friendships or create any negative feelings within the identified group. Another known

limitation of using the snowballing technique is that it would not necessarily lead to an incidental sample of participants being recruited to a study (Atkinson et al 2001, Streeton et al 2004, Perz et al 2013).

For this study, the nature of the topic being explored meant that it was always going to require purposive sampling to be utilised. Purposive snowballing recruitment strategies are a non-random approach to seeking participants to meet the inclusion for a research study. (Valerio et al 2016). For this study I approached schools and a children's hospice and I met with three different Paediatricians who provided clinical expertise for children and young people with neuromuscular conditions which was the purposive approach of seeking services where young people with neuromuscular conditions received care. In addition, I met one of the mothers through a contact with a senior colleague at the university, and she in turn offered to speak to other mothers who she knew to verbally advertise the study more widely. One of the special need's schools advertised the study through their school Twitter feed and a previous pupil who had attended that school but who was in her first year at university, saw that Twitter announcement and contacted me via email and became one of the participants in the study. The advantage in taking this approach in recruitment was that I knew that I was advertising the study to potential participants who had an interest in the research area, however the disadvantages were that I had limited the potential participants who might have been interested in being participants in the study.

The limitations of using snowball techniques for recruitment include selection bias, whereby social networks will influence the participants who are contacted and given the opportunity to participate in the research, and some contacts may act as "gate keepers" and not pass on the information to as many potential participants as might be available, due to wanting to being protective of their contacts, which can limit the potential for recruitment to the study (Atkinson et al 2001, Streeton et al 2004).

3.6.4 Challenges for participants in being able to participate in the research.

Once recruited as participants to the study, the correspondence from young people from their emails, which were sent in addition to their responses in the questionnaire, gave an insight into the complexities of their lives. The information that they provided demonstrated how hard it was for them to maintain a social life, continue with their education and manage the

changing demands of their disease which for some involved the need to learn how to use new equipment to manage their symptoms.

These experiences caused unforeseen and significant delays in starting the data collection phase of the project and took time to resolve. However, as the health of two of the young women became more stable, they felt able to resume their correspondence with me and eventually returned their completed questionnaires. Our email correspondence during this period was also used as data in this study.

3.7 Data collection

The study was initially designed to use an on-line data recruitment and data collection method for young people, to enable them to be involved as research participants at a time and at a location that was suitable for each person. This method of data collection was designed to be inclusive and to allow young people with a neuromuscular condition and a wide range of verbal communication abilities to participate in the research study using their own computer, rather than having to rely on others to interpret their communication through using Makaton, alphabet charts or other communication methods such as PECS (Picture Exchange Systems). By using their own computers, it enabled young people greater security, confidence, and confidentiality to communicate for themselves, as opposed to using another person to interpret for them (Nind 2008).

Although focus groups were considered for gathering data from the young people, for many families, public transport could have been challenging due to requirements to use a wheelchair to facilitate their mobility and would have taken precious time, which the parent may have been unable to give, and therefore this data collection method was discounted. In addition, the use of focus groups could have compromised parents' confidence to speak freely and openly about their experiences. From the first design plans, the study was specifically developed to minimise the expectation being placed on families or young people to have to incur the extra inconvenience to travel to meet with the researcher and participate in the research. The difficulties that some young people have as part of their neuromuscular condition identified that face-to-face interviews or participating in focus groups would be difficult for some young people; therefore, it was concluded that email correspondence together with completing a questionnaire electronically and returning it to the researcher via

email would be the easiest and most cost-effective mechanism for young people to be able to respond for themselves. In addition, for some potential participants in the study, their impaired verbal communication skills would have made interviewing or focus groups inappropriate data collection methods to adopt for this study. Consequently, being mindful of enabling the study to be as inclusive as possible for any young person wishing to contribute to the research, on-line data collection methods with the use of computer software packages such as the relatively new "eye gaze" technology were chosen to enable young people to contribute at a time and location most suited to their varying needs (Borgestig et al 2016).

Being able to meet two of the mothers in their own homes, provided additional valuable information for me with regards to the complexities of their lives, as the mothers showed me the challenges that they faced in storing the equipment that they needed to care for their children in the garden shed, under the stairs and in their kitchens and living rooms. These visible reminders of boxes of single use disposable supplies were a necessity, but were also a constant reminder to the parents, that their parenting journeys were different to their wider family and friends. By their own admissions, each of these mothers' lives were regulated by the times of each day when their children needed medication or a medical intervention such as an enteral feed, which were time critical. The mothers mourned the lack of spontaneity that their friends and family members were able to enjoy, as each outing from the family home had to factor in the time, location and resources needed to ensure that their children's medical needs were met. The third and fourth interviews with mothers took place at the university on a date and at a time of their choosing, and the final interview took place in a coffee shop which was chosen by the mother.

3.7.1 Interviews

Data collection from each of the mothers was conducted through face-to-face semi-structured interviews to enable a comprehensive exploration of their parental experiences of providing ongoing care for a young person in their family with a neuromuscular condition. One young person, whose mother was also a participant in the study, chose to have a face-to-face interview and to meet me as the researcher. Face-to-face interviews provide opportunities to observe the non-verbal communication of the participants, which otherwise is hidden from the researcher with, for example, telephone interviews (Novik 2008). In these individual interviews with the mothers, the stress and distress that they had since the births of their children and continued to experience in their parenting journeys were obvious, and it was

clear to see that, despite their devotion to their children, they continued to experience high levels of anxiety and stress on a daily basis (Dickson-Swift, James et al 2009, Holtan et al 2014).

An interview schedule (Appendix G) was used to guide the interviews, although during each of the interviews the participants volunteered more information and two mothers were keen to show me around their homes, to help me to gain a more complete picture of their lives and the impact that having a child who required specialist standing and seating equipment had on their living arrangements. Both mothers were keen for me to meet their sons and showed me pictures and videos of their children on their mobile phones, for example: *"If you've got a minute, I must just show you one little clip of footage on my phone", "I've got lots, but I want to show you this particular one - just to get a feel for who Lucas is"*.

Being invited to conduct the interviews in their homes enabled both mothers to have more control over the interview process and we were able to start and stop the recordings as required, when they needed to answer their mobile phones or attend to their children. Being a guest in their homes enabled the two mothers to feel safe in their own space. One mother had a qualified nurse in attendance with her son Lucas on the day of the interview and she introduced the nurse to me. Both mothers became distressed at different points in the interviews when they were showing me pictures of their children; however, being in their own homes enabled them to go the kitchen or bathroom and regain their composure before choosing to continue with the interviews, despite being offered the opportunity to terminate the interview if they wished to do so. Using qualitative research methodology and inviting participants to share their stories with someone that they had not met before, enabled the interview process to be positive experience for the mothers where they were listened to without judgement or criticism and could tell their stories to someone that they would not meet again. This appeared to be therapeutic for them both (Birch et al 2000).

This approach to interviewing these two mothers in their own homes was akin to phenomenological interviewing, whereby the researcher is keen to understand the *"life world"* (p140) experiences of participants, and to understand their lived experiences and their *"worlds"* (p141) from their own perspectives (Bevan 2014). Being invited to see their homes gave a new lens by which to understand the complexities of the lives that they were describing through their narratives. In both interviews, as we talked, they pointed to

photographs in frames on their walls in their sitting rooms of their children, which had been taken at different stages in their lives and they used these to show me how much their children had changed over the years of their lives and how proud each of the mothers were of their sons.

The benefits of narration and how this enables understanding to be developed by and from the individuals themselves has been well documented (Bowling 2010, Silverman 2016, Polit et al 2017,). However, the skills of the researcher need to be able to take account of the context in which the narrative takes place, the substance of the information that is being imparted by the participant and the audience to which the information is being given (Gubrium et al 1998). These skills also include the ability to use field notes and observations of the body language and nonverbal communication of the person recounting their experiences, together with a comprehensive analysis of the data obtained, to be able to build up a more comprehensive understanding of the impact that the re-telling of the experiences has had for the participants (Gubrium et al 1998).

Due to my awareness that some young people with neuromuscular impairments have difficulty with oral communication due to their muscle weakness, I wanted to be as inclusive as possible and to give young people a choice of either completing an open-ended questionnaire or being interviewed by me. Three young people completed the questionnaire, I interviewed one young person and the data from the fifth young person came from the TV documentary as explained above.

3.7.2 Use of the internet to collect data.

There is a growing knowledge base concerning the use of the internet as a tool for gathering data for research (Rabiee et al 2005, Nind 2008, Ison 2009). Different authors have identified that qualitative research using the internet falls broadly into three main categories. The first requires the passive analysis of information gathered from forums already established such as discussion groups for specific diseases. In the second category, active analysis can be achieved whereby the researchers contribute to the online discussions seeking clarification of the views expressed online. In the third identified category, the researcher(s) identifies themselves to the online community using the specific forum and used the internet for a more traditional data collection approach such as the completion of semi structured interviews or surveys (Eysebbach et al 2001, Markham et al 2008, Hine 2010).

However, potential issues that can arise from undertaking research using the internet have been considered by an increasing number of authors. Consideration has been given to the complex issues of privacy, security, ethical considerations, research relationships and the importance of ensuring that work produced because of using the internet for data collection in qualitative research can be judged as being meaningful (Eysenbach et al 2001, Markham et al 2008, Hine 2010). Initially using the internet was considered as a means of advertising the study and recruiting potential participants, as many young people are regular users of the internet and gain information and support from accessing different specialist websites and forums concerning their specific medical condition. However, my initial plan to use this route for recruiting potential participants via chat rooms or forums for young people with neuromuscular impairments was discounted early in the planning process, due to the complexities of using this approach, and the potential ethical difficulties of ensuring privacy and informed consent for potential participants.

It was important to consider that some potential participants to this research study may not have had the necessary IT skills to be able to participate in the study, (as it was designed to collect data from young people on-line via the internet). Consideration was given as to whether English was their first language or that they may have low literacy levels or a learning difficulty, which would have made participating in a study requiring on-line data collection methods difficult. In addition, some potential participants may not have had access to the internet at home and may have had to rely on the IT facilities at their school, college, university, or respite facility in order to participate in completing the questionnaire if they chose to do so. These situations provide some explanation for the complexity that was a part of the difficulties in recruiting young people as participants for the study. Potential participants were also given the options of emailing their completed questionnaires to me or sending their questionnaires back to me in the post in the stamped addressed envelopes provided. Three of the young people chose to email their questionnaire back to me and one young person chose to come and meet me in person at the university.

3.8 Triangulation and case study methodology

The purpose of triangulation in case study methodology is to provide an evidence base for the results presented in the final research report. It is a trusted and acknowledged mechanism for

cross-checking the data and the analysis presented in the research and enables different perspectives of the data to be presented (Stake 1995, Simons 2009, Yin 2014). There are four types of triangulation used in case study methodology which are recognised in the research literature (Stake 1995, Simons 2009, Yin 2014, Thomas 2016). The first of these four types is data triangulation, whereby different types of data which have been obtained for the study e.g. interview transcriptions, field notes, memos, data from medical records or questionnaires are appraised and compared within each of the cases. This process adds rigour to the analysis of the data overall (Stake 1995, Simons 2009, Yin 2014, Thomas 2016). Data triangulation was one of the main triangulation approaches used in this research study and is consistent with using case study methodology (Yin 2014). Data triangulation was achieved by using data sources from transcribed interviews; online questionnaires; field notes taken after face-to-face interview; emails; notes from conversations and memos that were written at different points in the research process; the television documentary of the young person with Duchenne muscular dystrophy, and photos and video clips which two of the mothers had shown me from their mobile phones. A second type of triangulation is that of data source triangulation, which was also used, whereby I interviewed different participants (primarily mothers and young adults) to gain a range of perspectives of the experiences of watching a child become a young adult with a neuromuscular condition or being a young person striving to achieve their own personal goals, with the physical and emotional challenges placed on them from living with a neuromuscular condition.

A third triangulation approach is that of theory triangulation, where theory is used deductively to make sense of the data (Stake 1995, Simons 2009, Yin 2014, Thomas 2016). In this study I used the three theoretical frameworks of identity theory, the ABC-X model of Family Stress and Coping (Hill 1958) and Transitions Theory (Schlossberg 1981) to offer different perspectives on the data. When more than one researcher is involved in a research project, this is referred to as multiple researcher triangulation, in this situation more than one researcher scrutinises and interprets the data seeking to agree on the meaning of their combined interpretation of the data (Stake 1995, Flick 2018). Some degree of multiple researcher triangulation can take place in a PhD study. However, although this is limited, for this research study this was achieved through intensive discussions with my three supervisors throughout the research journey and during data analysis. Discussions with other PhD students and in research forums which are run by the Doctoral College, also provided

opportunities for me discuss my data and my interpretation of it and to gain different perspectives from others experienced in conducting qualitative research.

3.9 Propositions and case study research

Case study methodology benefits from propositions or objective criteria and theoretical concepts being developed which are then used to guide data collection and analysis, which all contribute to trustworthiness, reliability, and congruence (Amerson 2011; Yin, 2014). Propositions or frameworks provide direction to the research (Yin 2014) and are developed from the literature and the researchers own perspectives of the concept and experience within the field, which allow the researcher to link the data and relate it to the theory, thus initial pattern matching can commence (Yin 1994, 2003, 2014). Propositions can be used in a similar way to a hypothesis in quantitative research and offer statements of the relationship between the data and the theoretical frameworks. They direct the researchers' attention to something that should be examined and provide the rationale and direction for the data collection; they are the basis for initial data analysis and are supported or rejected during data analysis (Baxter et al 2008, Yin 1994, 2003, 2014). Propositions increase the ways in which researchers can manage the complexities of the work necessary in conducting research and the likelihood of bringing the project to completion (Baxter et al, 2008).

Yin (2014) identified the importance of propositions in case study research as directing the attention of the researcher and the reader to the aspects of the literature which should be addressed within the scope of the study. However, Yin also provided the caveat that propositions may not be appropriate or needed when the case study is exploratory. For this study, from my clinical experience and the literature I identified the following issues as the propositions for this study:

- The voice of young people with neurological conditions is not well articulated in the literature.
- Young people need the opportunities to talk about their lives and tell their stories from their perspectives.
- Research literature on the experiences of young people with chronic and complex health needs indicated that young people and their parents are still experiencing a lack of support with the planning and preparation for their move to adult services.

- For young people who would previously have expected to have died during their adolescent years, the extension to their life span needs to be supported with facilities and services, to enable them to benefit from the additional months and years of life.

3.10 Ethical considerations

When undertaking research with human participants, ethical approval from a regional NHS ethics committee and local approval for site specific research and development committees is required. Any research study involving potentially vulnerable participants requires additional scrutiny to ensure the safety and protection of the participants. There is a tension between the requirement of local and national guidelines to seek the involvement and the views of service users and the protracted systems that need to be successfully navigated in order to secure the necessary approvals for research projects. Navigating and meeting the necessary requirements for ethical approval to be secured resulted in longer periods of time being required which resulted in unavoidable delays to the recruitment of participants. The literature and local and national guidance concerning gaining consent from young people both in clinical practice and for the purposes of undertaking research are comprehensive and clear with regards to the processes and safeguards for ensuring that informed consent is gained from parents or carers and from young people themselves (United Nations Convention of the Rights of the Child 1989, GMC 2007. BMA 2008, GMC 2008, Lambert et al 2011, RCN 2012).

Over the past two and a half decades, polices concerning the rights of children and the importance of children being given the opportunity to participate in research has been debated, considered, and published. One of the seminal pieces on this subject is the United Nations Convention on the Rights of the Child (1989) which states in article12 that: *"All children and young people who are capable of forming their own views have a right to be heard in decisions that affect them. Article 13: All children and young people have a right to have their views heard" (*p5).

The juxtoposition between seeking to understand in more detail the needs of families who have children with life-limiting conditions, balanced with the methodological challenges that research of this nature creates has been identified by other authors (van Staa et al 2011, Kirk

et al 2014,). The sensitive nature of asking questions of those with life-limiting illnesses or questioning those who love and care for them is complex and yet without seeking to consult and explore these issues services may have difficulty in growing and changing to meet the needs of this group of children and young people. The epidemiologcal evidence currently available demonstrates the growing need for services to meet the rise in prevelance of children being born with and living with life-limiting conditions in England (Fraser et al 2011).

Although research projects involving children and young people are often more complex than research with adults, there is wide spread recognition of the importance of the voices of childen and young people being sought and being heard. Therefore the planning and design of research studies involving children should take into account their cognitive abilities and use a range of data collection methods cognicent with their abilities with the aim of yielding a rich source of data to explore issues that affect this client group.

A review of the current local and national guidance on conducting research with children and young people and gaining their consent (GMC 2007, GMC 2008, BMA 2008, RCN 2012, National Children's Bureau 2016, Brady et al 2017) identified the recommendation of the development and inclusion of a distress protocol with clear processes for signposting participants onto appropriate support if they become distressed after answering any of the questions posed. Therefore, this was developed with the aid of the lead supervisor (Appendix J).

Informed consent and the capacity to consent were important considerations in the design of the study. The Mental Capacity Act (2005) came into law to provide protection for the most vulnerable members of society over the age of sixteen years within the UK and was designed to protect and restore power to those who are assessed as lacking the capacity to consent for themselves. The Mental Capacity Act has five key principles whereby those caring for individuals who are vulnerable must provide a range of opportunities and tools to enable people to make decisions for themselves with support wherever possible. For young people with progressive and degenerative neuromuscular conditions, their verbal capacity will gradually become more impaired as their disease worsens, however their cognition and intellectual abilities are often unimpaired, and they need to be offered the opportunity to participate in the research for themselves using the available computer technology if they

choose to do so (Ison 2009). I was mindful that at the stage of young people being presented with the information about the study, that it was vital that the individual mental capacity of each young person to consent to participation in the study, had been assessed by a parent or professional who knew them well, and that the responsible adult could ensure that they were able to make a coherent decision for themselves. Parents or caregivers also signed the completed consent forms for their child to participate in the study (see Appendix C and D), in addition to completing consent forms for their own participation in the research. By taking this course of action, I was ensuring that the adults who knew the young people best, had been able to make the decision as to whether each young person had the mental capacity to be able to consent to being a participant in the research.

3.10.1 Ethical approval from the NHS regional ethics committee

I applied for ethics approval from the NHS regional ethics committee for the south coast of England and I attended the committee hearing in November 2013. I was advised that the committee had concerns about the wording in one of the questions for the young people in which I had intended to ask what their *"hopes and dreams"* were for their futures. The committee felt that the wording of the question could have caused emotional distress to the young people, and therefore required that the wording of that question should be amended to *"ambition"*. The committee also requested some revisions to the information sheets for participants which I amended and resubmitted in December 2013. This caused a further unforeseen delay in ethical approval being granted and thus delayed the recruitment phase of the study. I applied for and received ethical approval and indemnity insurance cover from the University of Brighton in 2013.

I resubmitted the revised documentation to the regional NHS ethics committee and received final ethical approval for the research study from the NHS Health Research Authority (IRAS Project ID: 116662). I then applied for research and development approval at the local children's hospice and the local NHS children's hospital, and these were subsequently granted.

3.10.2 The benefits of taking part in the research.

The opportunities and the therapeutic benefits for individuals to provide an account of their own lives and lived experiences, and how these experiences enable understanding to be developed by and from the individuals themselves, has been well documented (Bowling

2010, Polit et al 2012). Research exploring the therapeutic benefits and challenges of undertaking qualitative research interviews, indicates that a therapeutic benefit for participants can be demonstrated (Birch et al 2000). Therefore, researchers need to acknowledge the positive effects which can result for participants by being actively listened to and being able to share their experiences with a researcher who in some cases is also unknown to them (Rossetto 2014). However, the aspect of qualitative interviews which may be unrecognised by novice researchers is the issue of emotional stress which the researchers themselves can experience, and need to be able to recognize in themselves and deal with appropriately to protect their own mental health and wellbeing. In this study, providing parents with the opportunities to narrate their birth histories and relate their child's health journey evoked painful memories for three of the mothers and at different points in their recall, they became emotional but although given the opportunity to stop their interviews, they each chose to continue with contributing to the research. Researchers are human beings with their own personal histories and need to be emotionally robust to deal with some of the complex and emotional information provided by participants, and to be able to process the information through the provision and process of supervision (Ross 2017).

3.11 Data analysis processes

Data analysis is one of the most challenging aspects of undertaking case study research (Yin 2014). The advantages of having more than one case within a case study research project is that the evidence obtained is more compelling and adds to the robustness of the study overall (Thomas 2006). However, the disadvantages that need to be considered are that the more cases that are included as part of case study project means there is a need for increased time to collect, transcribe and analyse the data; this may require extensive resources depending on the amount of data obtained (Eisenhardt 1989, Thomas 2006, Houghton et al 2012).

Different approaches to the analysis of the data in case study research have been identified by Stake (1995) and Yin (2014). Stake identified that there is no set time in the case study research process when the analysis of the data should be commenced. Stake recommended that initially the researcher should note the overall first impressions of the data, and then move onto the detailed analysis of the data as the research process is continued, when each line is considered and the whole data set is disassembled and then reassembled to make sense of what the data is saying. In case study research the analysis is a continuous process, when

new meanings can be attributed to the data as the analysis process is continued, with the emphasis being on creating understanding, looking for patterns and ultimately to make sense and create meaning from each case (Stake 1995).

Yin (2014) advocates that an analytic strategy should be adopted by the researcher but acknowledges that the analysis stage of case study methodology is often the least well defined and articulated. He suggests that a starting point for researchers should be where the researcher seeks for patterns in the data and develops a scheme of documenting the data in tables or flow charts to aid with the objective examination of what the data is saying (Yin 2014). Yin acknowledges that computer software programmes such as NVivo and HyperRESEARCH can have their place in the analysis process. However, these cannot replace the work of the researcher in developing a strategy for detailed and rigorous analysis of the data (Crowley et al 2002). I chose not to use qualitative data management software, such as NVivo. Instead, I made a conscious decision to undertake the analysis of the data manually myself to ensure that I had not missed any crucial aspects of the data which I may have missed by being confined to primarily working at a computer screen and not going back and forth (physically and spatially) repeatedly and thoroughly through data from the cases (Crowley et al 2002).

Yin (2014) presents a number of strategies that can be used in the analysis stage of the research; the first being for the researcher to use the propositions that led originally to the study as the guide for the analysis. For this study, the propositions that were developed was a recognition of the importance of young people with neurological impairments, with or without impaired verbal communication skills, and their mothers, to have the opportunity to speak their own stories and for health staff to learn from their experiences. The second approach to the analysis involves looking at the data *"from the ground up"* (p136) looking for the emergence of concepts from the data and seeking explanations for the relationships between the different concepts that have been identified. The third approach involves developing *"a case description"* (p139) to organise the data.

One of the difficulties with the analysis stage of case study research, is that there is not one single definitive approach which is applicable for all case study research, however the aspect, which is consistent with each author, is the agreement that the analysis should be a dynamic process throughout the research processes. In this study, the analytic plan which I developed

and followed, started with notes that I made following each interview and then I made further notes as I re-listened to the recordings of each interview and considered whether my first impressions matched my later review of the data. As codes emerged, I created tables and then cut and pasted sections of the data into the tables to show how sections of data from the different participants matched what I believed the data was saying (Appendix K). This was then further developed (Appendix L) and I was able to see the similarities in the journeys that each participant had taken from their narrative and I looked for meaning and connotations to help understand and construct a 'story' from what each participant had said. As each concept emerged, I returned to the literature to search for similarities in the experiences of other parents and young people living with neuromuscular conditions. Using a white board and different coloured pens enabled a visual representation of the timeline of events as reported by the participants, starting with the pre-conceptual period and birth stories from the mothers and recollections from early childhood from the young people (see Appendix J). This approach is supported by Simons (2009) and Thomas (2010) who identify the usefulness of this method for enabling new insights to be revealed as analysis progressed. I combined these stages with the steps of thematic analysis based on Braun et al (2006, 2013) and Fereday et al (2006) (expanded below). The steps of analysis for the three open-ended qualitative questionnaires also followed the steps of thematic analysis from Braun and Clarke (2006). For each of the questionnaires I carried out line-by-line coding, and then collapsed these codes into themes.

Generally, case study methodology is used to understand what the issues are that match the experiences of other cases, but also aims to identify which aspects are unique to each case. The researcher must search for each of the aspects that are relevant to the case and think broadly about other aspects which might have a bearing on the findings, for example politics or economic factors which may have an influence on the case being studied (Stake 1994). The researcher is responsible for deciding which aspects of the case will need to be explored and will need to provide a sound justification for the areas that are included (Simons 2009, Silverman 2013).

There are multiple ways in which a case can be reported. Case(s) may be presented as a journey, a narrative or story following a timeline, themed or categorised, or pattern mapped. It is important that the researcher displays enough evidence from data, providing as much detail of the case(s) as possible so that the reader may draw their own conclusions from the

evidence presented. For example, Stake (1994) recommends presenting cases as vignettes or a brief descriptive story using episodes to illustrate aspects of the case to provide the reader with a 'story' they can recognise. In keeping with case study methodology, as much detail as possible is provided about each of the cases, in order that the reader can also make a judgment as to the authenticity of the data and the findings (Stake 1995, Yin 2014). It is also understood that in case study research a massive amount of data is generated (Hancock and Algozzine 2006, Thomas 2016) much or which cannot be used in one piece of work. That is the situation in this study, where there is a huge richness in the quality and volume of the data obtained from the participants, which I plan to use in writing future academic papers.

The steps that were performed in the analysis of the data were as follows. Each face-to-face interview with the five mothers and one young person was recorded and I then transcribed each recoding verbatim. Doing the transcription alone, gave me the opportunity to concentrate on the content and listen to the inflexions in the voices as they recounted their individual accounts of their lives to me. The television documentary was also transcribed verbatim which yielded twelve pages of data which were then analysed following the same steps as the interview data.

Each transcript was line-numbered to ensure easy location and retrieval of data. These were printed off and laid out side by side to enable me to move easily from one to another (which was more difficult to do on the computer screen). I used different coloured highlighter pens to go through each line of data one by one and used different colours to represent different codes which I had developed e.g. *"impairment" "loss" "new beginnings".* I repeated the same process with each of the questionnaires from young people, which enabled me to compare the responses from each question in sequence and to note the similarities and the differences in the responses from each participant; for example, each young person highlighted the importance of their parents and siblings for their support but also for the enjoyment and enrichment to their lives that they received from being with their family members.

Initial 'naïve' (Eisenhardt 1989) reading of the transcripts and open-ended free text questionnaires led to conceptualising the experiences as a 'journey'. As the narratives from the mothers started with their pregnancies, it became logical to view the data as a life journey for each participant starting with the antenatal period of the pregnancy (where this

information was available) and then continue through the early weeks and months of the child's life, including the development of symptoms and the confirmation of a diagnosis and then continuing through early and late childhood. During this stage of the data analysis, some early initial broad themes were created (Appendix K).

More detailed analysis of the data was then performed using both the six phases of thematic analysis following Braun and Clarke (2006, 2013) and the six stages of thematic analysis of Fereday and Muir (2006); both influenced my decision making during the data analysis process (see Table 1 below).

Braun and Clarke 2006	**Fereday and Muir- Cochrane 2006**
Phase1: Familiarising yourself with your data	Stage 1: Developing the code manual
	Stage 2: Testing the reliability of the code
Phase2: Generating initial codes	Stage 3: Summarising data and identifying initial themes
Phase3: Searching for themes	Stage 4: Applying template of codes and additional coding
Phase4: Reviewing themes	Stage 5: Connecting the codes and identifying themes
Phase 5: Defining and naming themes	Stage 6: Corroborating and legitimating coded themes to identify second-order theme
Phase 6: Producing the report	

Table 1: Braun and Clarke (2006) and Fereday and Muir- Cochrane (2006)

As discussed above, in the early stages of the analysis process, I read and re-read the transcripts of the interviews and questionnaires. Throughout the process I repeatedly listened to the recordings of each of the interviews to immerse myself in the data and to think carefully about what I was hearing from the mothers, and the one young person who also chose to be interviewed. In this first stage of the process, I went backwards and forwards through each of the transcripts and the three questionnaires and developed codes for each section of the data. In this stage I looked for words and phrases which represented what the 'stories' of the participants. My clinical background enabled me to understand the medical terminology such as *"gastrostomy tube" "total parentral nutrition" "bladder augmentation*" and *"Mitrofanoff"* without becoming distracted or needing to look for definitions of those words. As I read and re-listened to the data, I was able, at this initial stage, to develop some early codes and assign these to the data.

In stage two of the analysis process, I worked through the transcripts and the questionnaires one by one. I worked through each of the sets of data and highlighted different sections with highlighter pens and applied codes throughout each set of data. The analysis of the four questionnaires (Appendix H) from the remaining young people was undertaken after each of the questionnaires were each printed off to allow me to write notes and prepare them for the second phase of the data analysis process. The questionnaire consisted of a mixture of closed and open questions to try and gain as much of an insight into the lives and experiences of the young people who were growing up with a neuromuscular impairment as they negotiated their own individual paths through school and higher education and made plans for their future.

At this second stage I tested the reliability of the codes that I had developed by discussing with my supervisors the codes that I had developed. Fereday et al (2006) identify the importance of confirming the reliability of the codes being assigned to the data, which when there is more than one researcher on the research team can be carried out by another member of the team. However, undertaking PhD study is a solitary process, but the importance of supportive supervision is crucial, by coding the transcripts myself and then discussing these at length with my supervisors, I was able to test the reliability of the codes that I was developing during stage two of the analysis processes.

In the third stage of the data analysis, I looked again at the data and searched for potential themes which matched the codes which I had assigned in stage two of the analysis process and began to apply quotes from the data to the separate codes. I moved backwards and forwards between the different transcripts which I had spread out over a large table. I cut the transcripts into the different coded sections and grouped them together, to ensure that each of the sections then matched the codes ascribed (Appendix L). At this stage I was able to see whether other codes would have also been applicable and make any modifications needed. At this stage I started to identify the key themes which then formed the structure from which I subsequently write the cross-case analysis chapter. This third stage of the process involved rechecking the data and identifying initial themes which were appropriate for the data. At this stage, I was able to look again at the themes which I had allocated to the data and again revisit the transcripts which I had highlighted with different colours, and make sure that the colours in the different transcripts matched the themes allocated to the different sections of

the transcripts. This visual display of different highlighted colours was very helpful for making the different themes stand out, and by spreading them out across a long table I could more easily assign each segmented to the allocated coloured section.

In the fourth stage of the data analysis, I revised both the codes and the initial themes that I had assigned to the data. At this stage I again checked the extracts of the data which I had applied codes to make sure that they were the most appropriate to have chosen from the data that I had gathered from the participants. By doing this, I was able to visibly demonstrate that the codes had been applied appropriately to the themes (see Appendix M). At this stage, I cut the coloured segments of the transcripts and laid them on large sheets of flip chart paper and secured them with staples and glue and was able to stand back from them and move backwards and forwards between them, to ensure that different sections of the narratives had been matched to the codes that I had developed.

In the fifth stage of the analysis process, I refined each of the themes that I had identified and revised the names that I had given for each of the themes (Appendix M). I looked for and identified patterns which had evolved from the data, which led to the decision to present the findings in the form of a timeline starting with the antenatal and birth stories from the mothers and moving on to the early and later childhood experiences from both the mothers and the young people's perspectives. At this stage in the analysis, it became even more obvious that a chronological timeline was present in each of the interviews and questionnaires from young person, with the mothers starting their recollections with their pregnancies and the births of their children, and the young people recalling their levels of mobility and independence in their early childhoods, and how these had changed as they had grown up and what the impact of this had been on their lives.

In the sixth and final stage of the analysis process, I moved between the different themes that I had identified from the data and grouped the different themes into clusters to check once again that the codes and the themes that I had assigned to the data confirmed the findings from the data. This also involved the deductive application of identity theory, family stress and coping theory, transitions theory and as a way of explaining and understanding the experiences of the participants. At this stage, having intentionally had some time away from the analysis, I had gained new perspective and was able to look afresh at the data and analysis that I had completed.

Finally, the findings were interpreted using analytical techniques to include pattern matching across the cases (cross-case analysis) looking to provide an explanation about the cases and identify a set of causal links (Yin 1994 p 102). It was at this stage, late in the analysis, that I identified the significance of the two pivotal cases and conceptualised about the complexity of transition for these families (see Findings Chapter on Cross-case analysis).

By the conclusion of the analysis process, it is important that all prior knowledge is brought to the study, that all the relevant evidence has been included and that all of the major rival interpretations for explanations for the findings have been dealt with (Yin 1994, Yin 2014, Thomas 2015). This process ensured that the data analysis was detailed and thorough and enabled the essence of the realities for parents and young people to be understood and related to policies and guidelines in addition to the findings from other published research studies. I have used the examples of the photos and video clips of the children which two of the mothers showed to me, and the excerpts from the emails that I received from the participants, to reveal more of the lives and experiences for each of the families of living with or supporting a child or young person with a neuromuscular impairment.

3.12 Reflexivity in research

Reflexivity in research concerns the recognition and acknowledgement of the importance of the position of the researcher and the effect that their knowledge and presence during the data collection processes and at every step of the research process has been. The significance of the researcher or researchers needs to be acknowledged, understood, and presented as part of the wider discussion and the lens through which the study has been undertaken and the data findings presented and interpreted needs to be identified and acknowledged (May 2002, Flick 2009, Silverman 2016). The researcher cannot change who they are or the experiences that they have previously had, but the ability to honestly reflect on themselves as individuals and professionals and to be able to analysis the relationships that have emerged between the researcher and the participant is essential. Reflexivity also changes with each research project and as it is the relationship which occurs between the researcher and the participant and this will change from one research project to another, but within the same project, this interaction can change with each interview or interaction with participants (Dodgson 2019).

I came to this project with twenty-six years of clinical experiences of providing care for children and adults with neuromuscular conditions both in acute and community settings and I had a parent who died of a neuromuscular condition in old age. The knowledge that I have as an experienced adult nurse, children's nurse and Health Visitor was extremely useful for understanding the terminology that the parents used. Undertaking one of the interviews with the mother of a severely disabled seven-year-old boy in her home at her request, reminded me of the stress that parents face with trying to store the large quantities of medical equipment and disposables that are a necessity for delivering enteral feeds, intravenous medication and fluids and suctioning secretions to maintain the patency of the child's airway. Having worked as a Community Children's Sister for nine years, I had a wealth of experience of visiting families caring for their children with neuromuscular impairments at home and supporting parents as they watched the gradual decline of their children's health as the weeks and months of their children's lives progressed. As a nurse, when the mothers were explaining aspects of their children's health and medical interventions, I was familiar with the medical terminology and they did not have to spend time explaining the different aspects to me, which I hope added to them feeling able to be comfortable in their presence.

At times, I found it difficult to separate my previous clinical experience as a nurse, with my new role as a researcher and had to remind myself to be objective, and not to respond to the narratives of the mothers in the ways that I might previously have done as a nurse. I was mindful of the advantages and the potential disadvantages of being a researcher with some medical knowledge, although the parents seemed reassured to know that I was a nurse and had had experience of caring for children with neuromuscular impairments both as inpatients and in the community.

As an experienced community children's nurse, I was comfortable with having difficult conversations with parents in their own homes, however on these occasions my role was that of a researcher and not of a nurse and therefore this changed the dynamic between the mothers and me, and I had to adjust to this new professional position. I gave each of the mothers the choice of where they wanted their interviews to take place. Two mothers chose to meet me at the university and their interviews were conducted in a seminar room where their conversations with me could be confidential. Two of the mothers requested that their interviews should be conducted at their homes and the fifth mother asked for her interview to

be conducted in a coffee shop that she knew well, and she felt comfortable to meet me in that location. When the two mothers became distressed during their interviews with me in their own homes, they were comfortable in their own domains and we could stop and start the interviews at their pace. The ways in which I responded to their distress was by being mindful of the distress protocol, turning off the recording and offering each mother the opportunity to terminate the interview at that point. This approach varied significantly from the ways in which I would have reacted if I had been in the role of a community nurse with a continuing professional relationship with the child and parent, where we could have continued the conversations over subsequent visits if the mothers had requested this.

The interview with the mother in the coffee shop took place in December on a weekday, but the coffee shop was busy with customers and the noise levels was loud and disruptive. Although this mother chose a table at the back of the shop, the noise levels were high, which on the one hand ensured some level of confidentiality but on the other hand, at times made it difficult to hear what she was saying, and I had to reflect back what I thought that she had said. When I listened back to the recording in order to start the transcribing of the interview, the music playing and the sounds of china rattling, and background conversation was very apparent. However, this mother chose this venue as a setting that she was familiar with and she felt relaxed and able to speak with me there.

Conversations with my supervisors were useful during my supervision meetings, to enable me to regain the researcher perspective over the clinical perspective, and to refocus my understanding from a more objective position and to discuss the themes that had emerged from the data and be able to link these with the literature that I had sourced and reviewed. These supervision sessions also enabled me to speak about my own feelings from hearing the mother's stories and meeting some of their children (Ross 2017).

Parahoo (2006) defines reflexivity as a continuous process whereby the researcher uses reflection to recognise the role that they have played in the research study and their interaction with participants. This was important as I conducted the interviews in four different settings and I was mindful that in the homes of two of the mothers, that I had to not become distracted by the medical equipment as a nurse medical charts, but rather use my observations and knowledge of these items, to understand the high levels of stress that both of these mothers were displaying, as they made decisions about the necessary interventions for

their children, which in the past would only have been made in hospital by medically trained health staff.

3.13 Rigour and trustworthiness

The credibility of qualitative research is demonstrated through the criteria used by the researcher for ensuring rigour in the research process and the analysis of the data. However, there is not a clear consensus between authors as to the different criteria which need to be met. Rigour and trustworthiness are important aspects of undertaking qualitative research whereby the research can clearly evidence each of the steps that were taken to undertake the research and analyse the data and can demonstrate the deep interaction that the researcher has had with the data (Maher et al 2018, Noble et al 2015).

Noble et al (2015) propose that four criteria are necessary for ensuring that the rigour and trustworthiness of the processes have been followed in qualitative research. These are: *Truth or value*: whereby the researchers accurately present the views, experiences, and perspectives of the participants, whilst at the same time also honestly presenting any issues which may potentially have introduced bias into the analysis. At this stage, my previous extensive experiences as a nurse have been acknowledged and the benefits of the medical knowledge that I hold in understanding the terminology used by the participants presented. I needed to ensure that I did not allow this previous clinical knowledge and experiences to cloud what was emerging from the data or to pre-judge what findings emerged as the analysis processes continued. The second is *Consistency:* whereby the decision trail of the researcher is presented with clarity and detail, in order that a different researcher unconnected with the research could follow the steps of the research process and should be able to reach similar conclusions to those of the original researcher. Details of this are also evidenced in my extensive research diary entries (Clark 2009, Engin 2011). Thirdly is that of *Nutrality:* at this stage, there must be clear evidence of the philosophical position adopted by the researcher. This should be distinct from the experiences or the views of the participants. I have adopted a constructivist epistemological position whereby I have acknowledged and recognised that the knowledge presented has been constructed by myself based on the participant narrated construction of their lives. As a nurse, it was challenging to hear the concerns from the participants of their experiences of health care which they had received from the NHS. *Applicability*: is the fourth and final stage of the process to demonstrate rigour and

trustworthiness and at this stage the researcher considers whether the findings could be applied to other client groups. I believe that there is applicability of the findings from this research to other young people with a range of long-term conditions and their families.

Ensuring credibility involves checking that the research findings resonate with others who understand the field of practice or have expertise in the research methodology. I presented early findings from this study at the University of Brighton annual Doctoral College Conference, and one of the audience members was a senior Physiotherapist who had had extensive experiences of working with working with children and adults with neuromuscular conditions. She confirmed that the findings from my data matched with her clinical experiences and echoed her professional concerns for young people reaching adulthood with neuromuscular impairments where the services may be very different from the level of services that young people have been able to access during their childhood years.

3.14 Theoretical frameworks

In this study I have used three theoretical frameworks as part of the theory triangulation process. The data was examined deductively through the theoretical lenses of Identity Theory (Burke et al 2009), the ABC-X model of family stress and coping (Hill 1949, 1958) and Transition's theory (Schlossberg 1981).

3.14.1 Identity theory

The understanding of what identity means is complicated and has been extensively explored by different eminent authors. Identity theory refers to the complexities of considering what makes an individual able to ascribe a position or a role to themselves and the links between the mind and their consciousness (Burke et al 2009). These ideas, of who each person is, changes over time from their birth to their death and alters during their lifespan depending on whether they are describing themselves as the role that they play in their personal or professional lives. The premise behind identity theory is the understanding of the complex interaction between the brain and the mind (Place 1956, Feigel 1958 and Smart 1959 cited by Burke et al 2009). Mead (1934) identified that as the environment continuously changes for each person, so their behaviour to their circumstances evolves to meet the new circumstances.

In identity theory, Mead (1934) identified the ways in which the human mind is linked to the brain and to the "self" of each person and how this is changed and shaped by the society that the person lives in and their interaction with others. The ways in which parents view themselves, altered as their individual situations changed throughout the years of their children's lives. Mead (1934) identified the relationships that exist between the mind of an individual and how this relates to their environment, he proposed that this complex relationship between a person and their environment and how this gives meaning to the individual will change and develop dependent on the stage of life that the person is in. This relationship was later identified as "symbolic interaction" by Blumer (1962) who used this as his interpretation of the work of Mead.

Stryker and Burke (2000) continued the development of the theory of identity and linked behaviours with the ways in which individuals behave in the society that they are living in and further strengthened the earlier links of Mead of "symbolic interaction" by linking the ways in which people react to both physical and social objects and to ways that individuals react and respond to the identities and roles held by others in society, for example whether the role of the police for upholding the law is valued and upheld or whether medical advice given by a doctor or a nurse is adhered to by a patient. Stryker and Burke (2000) as one of the first authors of identity theory, to recognise that people may hold multiple identities in their lifespan, which are linked to their professional and personal roles and their positions in their local communities.

3.14.2 Theories of transition

Transition is a process that each human being goes through multiple times during their lifespan. For each of the participants, both the parents and their children were making transitions through the different phases of their lives, and through this process, their individual identities were being shaped and changed as the young people moved from childhood to adulthood.

Schlossberg (1981) developed Transition theory as a mechanism by which the changes that adults face in their lifetimes, which move from the mundane and routine phases of life through to extremes of both positive and negative experiences that can affect the lives of individuals. Schlossberg categorised the transitions that take place during adulthood into the

three separate areas of "*individual", "relationships" and "work"* (Schlossberg 1981) and acknowledged that each adult human is constantly moving into and out of experiences of transition, but at the beginning of each new phase of transition, the individual needs time to acclimatise themselves to the new situation and to feel comfortable in it. The ways in which adults adapt and cope with each new episode of transition is determined by the four "S"'s of *"situation" "self" "support" and "strategies"* (Anderson et al 2012).

Schlossberg (2011) identified that the impacts of transition experiences in adulthood can vary dependent on whether the transition was "*anticipated" "unanticipated" or a "non-event"* (e.g. if an anticipated promotion does not come to fruition), and acknowledged that the ways in each individual copes with transitions in their lives will vary significantly, but the recognition of the importance of reaching out to others for support to cope with transition, can be an important mechanism by which adults cope with new phases in their lives.

Wilkins et al (2006) used Schlossberg's Transitions Theory (1981), as the theoretical framework with which to explore the experiences of the siblings of children who had been diagnosed with cancer, and to understand how siblings can make a healthy or unhealthy transition through the uncertainty of being in a family where one child was receiving treatment for cancer.

3.14.3 ABC-X model of family stress and coping

The ABC-X model of family stress and coping (Hill 1949, 1958) was developed to understand the experiences of families following the second-world war, whereby Hill sought to understand why stressful events in their lives might result in a crisis developing in one family but not in another. Hill looked at the ways in which the wider family and the local society, built resilience for some families to enable them to cope with the adversity that they faced. This was highly relevant to the families in these case studies as each family had faced significant levels of stress in relation to the health and wellbeing of their children, and therefore this theoretical perspective was used in the data analysis from the mothers, to develop understanding of the events that took place in each of the families.

This framework defines the ABC-X as the following:

A= the specific stressor event

B= the available resources that each family has which enables them to cope with the event.

C = the individual perceptions of the event

X= the endogenous variable for each individual and the family unit.

This framework was particularly useful to analyse the parental experiences of the five parent participants.

This theory has been used in several studies, for example, Bigalke (2015) used the ABC-X model of family stress and coping in a large study of 115 participants to explore the parents' experiences and levels of stress following their child being diagnosed with cancer in the USA. The finding demonstrated the importance of the relationship that parents had with the medical staff who were treating their children and that this had a direct impact on the levels of stress that the parents experienced and the ways in which each of the participants coped with this stressful event in their lives.

In a study in the USA Riper (2007) used the same theoretical model to explore the experiences of 76 families of their children being diagnosed with Down's syndrome. Riper identified that families move through the adjustment and adaptation phases when becoming the parents of a child with Down's syndrome and found that contrary to some previously held positions, many families found that having a child with Down's syndrome was a positive experience for them and brought them closer together, despite the anxieties of caring for a child with a continuing health care need.

Guyard et al (2017) also used the ABC-X model of family stress and coping (Hill 1949) to explore the adaptation of families who had teenagers (aged between 13 and 17 years) with cerebral palsy, in a large European multi-centre study of 286 families from Ireland, France and Denmark. Their findings demonstrated that the ways in which the family functioned was an important predictor of the levels of parental levels of distress. The authors used this theoretical model to explain well-being for children growing up with an ongoing disability.

3.15 Chapter summary

In this chapter I have presented the research methodology and methods that I used in undertaking this research study. I have addressed the ethical considerations in relation to this

study and how these were overcome. I have explained my rationale for choosing case study as the methodology used for undertaking this research study, with its benefits of being to view each case of each family separately and to conduct a cross case analysis looking at the similarities and the differences in the experiences of the participants. I have considered the importance of reflexivity in the role of the researcher and the advantages and possible limitations of having previous relevant clinical experience to use as part of the lens that I used to make sense of the data. Three theoretical frameworks were identified that were used deductively during the data analysis. I have presented the four separate steps which I followed to ensure that rigour and trustworthiness have been presented accurately and honestly and the standards expected from qualitative research have been met.

In keeping with case study methodology, the findings and literature are presented together in the next chapter.

Chapter 4: Findings and discussion

4.1 Introduction

In this chapter firstly I present the eight case studies individually, as discussed in chapter 3, in keeping with the principles of presenting case study findings, as much detail of the narratives as possible are presented to enable the reader to also make a conclusion as to the validity of the data and the findings (Stake 1995, Yin 2014) (see Table 2). In the second section of this chapter, I present a cross-case analysis and synthesis of the eight cases.

In cases one, two and three the mothers were the only participants; cases four and five had both the mother and their child as participants, and the participants in cases six, seven and eight were young adults, all aged 21 at the time of the data collection. The combination of these cases provided a unique opportunity to relate the views of mothers with younger children at an earlier point in their journeys to adulthood; with the views of mothers whose children were on the cusp of adulthood; and to consider those with the experiences of teenagers or young adults who were living with a progressive or a non-progressive neuromuscular impairment.

The first two cases are two mothers who both had seven-year-old boys with complex medical needs. The child in the first case study (Lucas) was on a palliative care pathway and required qualified nurses to care for him at home twenty-four hours a day due to the severity and unstable nature of his condition. He had required numerous admissions to hospital and his mother (Caroline) had had to adjust to having nurses and carers in her home caring for Lucas day and night.

In the second case, the parents had two boys, both with Autism; but the older child (Andrew) had additional complex medial needs which were unstable, and he had required numerous inpatient admissions to the local district general hospital and to a tertiary specialist children's hospital. The mother (Connie) was a qualified nurse who had anticipated being able to return to work part-time following the birth of her second child; however due to the severity of her older son's condition this had not been possible. Case three presents the experiences of a mother (Amanda) of having her16 year-old son (Orlando) who had a severe learning disability and a neuromuscular impairment, low bone density, epilepsy and limited verbal communication abilities.

The fourth case presents (Edward) a 16-year-old young man with Cerebral Palsy. Edward was on the cusp of transition to adult services and his mother (Melanie) expressed her feelings looking back at her pregnancy and the unexpected premature delivery of her son; and the journey that they, as a family, had gone through in the years of his childhood and adolescence. This case study enabled a comparison of the concerns of both the mother and of her son as they each looked ahead to his adult life and considered some of the potential barriers that could arise for him in seeking to achieve his goals and ambitions for his future. The fifth case presents Kim and her 17-year-old daughter (Suzie). Suzie also had a diagnosis of cerebral palsy, but in addition she had significant visual impairment, short term memory difficulties and epilepsy. As in the case of Edward above, this case study enabled the perspectives of both the mother and the young person to be expressed and the similarities in their concerns to be appraised.

Cases six, seven and eight present the perspectives of three young people in their early adult years each of whom were living with progressive, degenerative neuromuscular impairments which were life limiting. Each of these young people were ambitious, aspirational for their futures and each focused on achieving their education and career goals. The first two young women (Elizabeth and Lauren) were at different universities as undergraduates, studying for degrees in their chosen subjects; whist the final case (Nathaniel) and his mother (Madeline) presents a young man who had graduated from university and had returned to live at his parents' home. Throughout the findings, pseudonyms have been used to protect the confidentiality and anonymity of the participants.

Table 2: Overview of Cases

Cases	Overview of each case	Data Collected
Case 1 Single mother with no other children. Father had regular contact with his son.	Caroline – mother of seven-year-old Lucas with severe neurological impairment following a virus at the age of five months. Lucas had severe epilepsy, required total Parenteral Nutrition (TPN), had severe visual impairment, had no verbal communication, and required 24-hour care by qualified nurses at home.	Interview with the mother. Field notes related to photographs and video clips on the mother's phone.
Case 2	Connie – mother of Andrew aged seven years who had been born prematurely at 36 weeks gestation. Andrew was	Interview with the mother.

Two parent family with a younger son aged 5 years	diagnosed with SWAN (Syndrome without a name), autism, required gastrostomy feeding, had asthma, severe developmental delay. Connie had a younger son Bobby aged 4 years with high functioning Autism.	Field notes related to family photographs presented by the mother.
Case 3 Single mother with younger daughter aged 7 years. Father visits his son each week.	Amanda – mother of 15-year-old Orlando who was born at term but had an undiagnosed genetic disorder. This resulted in global delay, epilepsy and speech and language delay. Amanda had a younger daughter aged 7 years who had no health issues.	Interview with the mother. Younger child aged 7 years was present during the interview.
Case 4 Two parent family with older daughter.	Melanie – mother of 16-year-old Edward who was born prematurely at 27 weeks gestation. Edward – 16 years of age with Cerebral Palsy, bladder augmentation requiring intermittent self-catheterisation four to six times a day to achieve urinary continence. Melanie had an older daughter who had also been born prematurely but had no health issues.	Interview with the mother. Completed questionnaire from Edward.
Case 5 Two parent family with older daughter.	Kim – mother of 17-year-old Suzie who was born prematurely at 30 weeks gestation. Suzie – 17 years of age with Cerebral Palsy, epilepsy, and visual impairment. Kim had an older daughter who was also born prematurely but had no health issues.	Interview with the mother. Interview with the daughter.
Case 6 Two parent family with younger daughter.	Elizabeth aged 20 years with Triple X Syndrome, hypermobility, Asperger's Syndrome, deafness in one ear, a progressive neurological disease (with no confirmed diagnosis), mild Cerebral Palsy, urinary incontinence requiring a suprapubic urinary catheter, sleep apnoea, spondylosis of the lower spine, Obsessive Compulsive Disorder and chronic pain. Elizabeth had a younger sister who had no health issues.	Phone call with the mother. Emails from the mother. Completed questionnaire from the daughter. Emails from the daughter.
Case 7 Two parent family. 5 younger siblings.	Lauren aged 21 years with Congenital Myotonic Muscular Dystrophy, severe Scoliosis and Restrictive Lung Disease. Lauren had four younger siblings with no health issues.	Completed questionnaire from the young person.
Case 8 Mother also appeared in the TV documentary.	Nathaniel – 21-year-old with Duchenne Muscular Dystrophy and his mother Madeline. Nathaniel had been born at term and achieved normal development for the first fourteen months of his life. From then on, his balance and previously acquired gross motor skills declined and he was referred by the GP for further investigations. A diagnosis of Duchenne Muscular Dystrophy was made when he was 17 months of age.	TV documentary.

Common within-case themes for the mothers were: the journey to parenthood; parental hopes, dreams, and aspirations; the importance of family and social support; and facing the future and continuity of care. Individual sub-themes included: adjusting to expectations, deterioration in health and palliative care needs, dealing with stigma, effect on siblings; and children are a blessing. For the young adults, the common within-case themes were: hopes, dreams and determination; the importance of social relationships and having 'normal' experiences; importance of family and friends; making career choices; and transition to adult services. relationships, family, transition to adult services, and making career choices. Individual sub-themes included: increasing independence; parental sacrifices and effect on siblings; living with progressive deterioration; and dealing with stigma (see Table 3 below).

Table 3: Within-case themes

Mothers	**Common within-case themes**	**Individual Sub-themes**
	Journey to parenthood	Adjusting to expectations
	Parental hopes, dreams and aspirations	Deterioration in health of child and palliative care needs
	The importance of family and social support	Dealing with stigma
	Facing the future and continuity of care	My children are a blessing
		Effect on siblings
Young Adults	**Common within-case themes**	**Individual Sub-themes**
	Hopes, dreams, and determination	Increasing independence
	The importance of social relationships and having 'normal' experiences	Parental sacrifices and effect on siblings
	Importance of family and friends	Living with progressive deterioration
	Making career choices	Dealing with stigma
	Transition to adult services	

4.2 Caroline

4.2.1 Introduction

Caroline was the mother of seven-year-old Lucas who had a severe neurological impairment following a virus at the age of five months. He also had severe epilepsy and was virtually blind. In addition, Lucas had no verbal communication abilities. Due to the severity and unstable nature of his health he required 24-hour care by qualified nurses at his home. He was unable to feed orally and received his nutrition from total parenteral nutrition (TPN) via a central line. Although he had previously attended a local special needs school; the

deterioration in his health and the unstable nature of his medical condition had meant that being part of the school community had become almost impossible. Lucas lived with his mother in the south of England. One of the downstairs reception rooms had been turned into his bedroom, to allow ease of access and to accommodate the large volumes of equipment and supplies of disposable equipment that was required to meet his extensive care needs. Although his parents had separated, his father visited each week. Lucas had two older half-brothers who both lived with their mother in a separate town.

4.2.2 Journey to parenthood

Caroline was 41 when Lucas was born, and he was her first child:

> *I didn't ever want to have children. I had such a busy happy life. We were both totally shocked to find out at the age of 41 that I found myself pregnant, but over the moon.*

This unexpected pregnancy had been welcomed and Caroline and her partner had looked forward expectantly to the arrival of their baby. After a normal pregnancy, Lucas had been born at term following an uneventful antenatal and postnatal history. However, within five months he contracted a virus which marked the onset of his life with a severe disability. *"Lucas was left like a floppy rag doll. It also affected his visual field, there is nothing structurally wrong, and it's the messages being sent from the brain to the eye".*

Following Lucas's unexpected illness and the realisation that their son had been left with a permanent neurological deficit which they had been unprepared. Caroline and her partner had to make the unexpected and difficult psychological and emotional adjustment from being the parents of a normal infant, to becoming the parents of an infant with profound and complex health needs. For mothers of children with physical disabilities, levels of emotional stress are known to be higher than for parents of non-disabled children, particularly when the future for their child is uncertain Yamaoka et al 2016):

> *It took it's toll on us.* (Lucas's dad) *wanted life to just carry on and I* (Lucas's mum) *needed to totally focus on Lucas; so, we went our separate ways but it's all fine.* (Lucas's dad) *sees Lucas once a week. Last year when Lucas was in hospital, he came quite frequently, we thought that we were going to lose him, so primarily it is me and Lucas here.*

Relationship breakdowns in families who are caring for a severely disabled child are not uncommon (Mencap 2006, Contact 2017). However, despite the challenges and the ongoing

stress of the situation; Caroline and Lucas's father had reached an amicable solution to enable them both to continue to be involved with being parents to their son. The experiences that Caroline reported concur with the stresses that other parents with children with complex needs have also identified (Miller et al 2009, Menezes 2010). These experiences were endorsed in the findings of a report produced by Contact (2017) in which a profile of families in the UK caring for disabled children was presented. The findings from the report identified that children with disabilities were more likely to grow up in a household with one parent than their non-disabled peers. Although the day-to-day decisions and care for Lucas were taken by Caroline; his biological father remained part of the decision-making processes.

4.2.3 Parental hopes, dreams and aspirations

> *I had my dreams and aspirations – me and Lucas being beach bums – we lived on the beach as a family - rock pooling of an evening all year round and I love the sea. The beach to me now is inaccessible and it's upsetting.*

The dreams that Caroline had had for Lucas from his birth were strongly linked to her own childhood experiences of living by the sea and the loss continued to be painful. As the family lived in a town by the sea, the views of the beach were a regular occurrence and continued to be a reminder of what Caroline could no longer take for granted as an activity she could share with Lucas:

> *I remember the year that Lucas should have started walking, we lived opposite a park and I saw these little tots taking their first steps in walking and that was SO painful, to know that that wasn't going to happen for me with Lucas. I'll always be glad that Lucas doesn't know.*

The ways in which each parent copes when their child is identified as being on a palliative care pathway is unique. The importance of maternal mental health for mothers who have children with additional needs has been explored in work by Findler et al 2016a, 2016b; and Yamaoka et al 2016. Both studies identified the psychological stress that parents of children can experience when their child has a disability. Despite the challenges, Lucas's mother was able to focus on what were for her the positive aspects of her son's illness and disability. She had been able to recognise the strengths within her that had enabled her to cope and come to a point of being able to accept that Lucas would not recover from the catastrophic brain damage that had occurred from the virus. However, whilst speaking about Lucas, Caroline was able to take the position of being grateful that Lucas was unaware of his difference, and

therefore that he could not become distressed by comparing himself to other children of his age:

> *I'll always be glad that Lucas doesn't know - because I think that my worst nightmare would be to think that Lucas knew what was wrong compared to how he used to be – and it being locked in here* (mother points to her head), *I think that that would be quite evil for him to endure. I'm happy that it happened when it did and not after a longer time – for him and me selfishly.*

Symptoms of stress are commonly reported by parents when their child has complex needs and these levels of stress increase in relation to the degree to which the child is dependent on technology to keep them alive; and the amount of time that it takes for parents to meet the technological needs for their child (Brenner et al 2015, Lazzarin et al 2017). Lucas was dependent on technology which sustained his life, which included a central line for the administration of his nutrition and the complex combinations of drugs which were necessary to manage his dystonia. Caroline, who had no previous medical training or experience, had unexpectedly become the "expert parent" for Lucas. She felt that being a mature parent had given her extra abilities to cope with the stress of being Lucas's mother and to manage the technical complexities of his healthcare needs. Studies which have explored the experiences of parents whose children are discharged home dependent of technology, have agreed that parents face many hours each day using equipment to sustain their children's lives which correlates with the reality that Caroline faced once she had succeeded in being allowed to take Lucas home (Brenner et al 2014, Lazzarin et al 2017).

Using transitions theory (Schlossberg 1981), provided a mechanism to understand how Caroline's experience fitted with her *anticipated* stage of transitions theory and how her welcomed transition to becoming a parent, had moved swiftly into her *unanticipated* phase of the Transitions theory, as she adjusted to the reality that unexpectedly she had become the mother of a child with complex continuing healthcare needs which were subsequently diagnosed as being life limiting.

4.2.4 Social support: *"I wasn't alone".*

For Caroline, the opportunities to meet other parents with young children with special needs who were in similar parenting circumstances to her own, provided an immediate psychological lifeline for her as she had spent the previous year feeling isolated and afraid for the future, unsure whether any recovery would be possible for her son:

> *When Lucas was 14 or 15 months old - I took him along to a baby signing course. I had never interacted with another family. I was just over awed, almost in tears of joy. It was just a revelation to realise that **I wasn't alone** and there was support. It was just lovely to realise it wasn't going to be as lonely a journey as I thought it was.*

Using Transitions theory, it became apparent that the *"support"* aspect of the theory had significantly affected the way in which Caroline adapted and developed her coping strategies to deal with her new situation. This new group welcomed her, and she felt able to be part of their world and they became part of her world, which provided validation for her and for her young son. Her identity changed from being the mother of a "normal" baby, to be the mother of a child with deteriorating and fluctuating health needs and thus identity theory, made her experiences clearer as her role changed both to herself and within her local community. In this new role, she adapted to learn new medical terminologies and lost the freedom and spontaneity that she had enjoyed before Lucas had become ill.

For parents learning to adjust to a world of disability takes time; and being in contact with other families in similar situations can provide important social and emotional support; where parents can empathise and support other parents of children with complex needs (Miller et al 2009, Yamaoka et al 2016). This resonated with Caroline's experience as she adjusted and reshaped her world of being the mother of Lucas as a young child with a severe neurological impairment. Whilst health professionals can offer professional advice from the wealth of knowledge and experience that they have of working with children with neurological impairments, parents benefit greatly from the understanding and sense of connectedness that comes from being with other parents who have experiences that are similar to their own, and they can empathise and support each other practically, emotionally, and psychologically.

With her change of identity, Caroline's world had become open to new social support networks, and to socialising with other parents who also had children with special needs, from whom she gained support, advice and guidance. Caroline had to learn a new medical language to understand the diagnosis, symptom management and interventions necessary to support her child's health needs and to become the expert parent for her child.

4.2.5 Deterioration health of the child

> *We went into hospital for the umpteenth time with him vomiting. He was seen by an amazing surgeon and she put a PIC[7] line into Lucas and she put on TPN actually. They quickly decided to change the gastrostomy for a Jej[8].*

The deterioration in Lucas's ability to maintain his own nutritional needs orally or via a gastrostomy tube inserted into his stomach; resulted in the insertion of a jejunostomy tube which reduced Lucas's ability to vomit and thereby to retain the calories from the enteral feed. This new development required Caroline to learn new technical skills to enable her to care for the jejunostomy tube and to continue to care for Lucas at home. However, research exploring the experiences of parents who have children who are technology dependent and receiving enteral nutrition at home; have demonstrated that parents in this situation have increased levels of stress and anxiety due to the weight of responsibility they take on as they learn new skills to be able to administer enteral feeds and to deal with any problems that may arise (Pedrón-Giner et al 2013, Brenner et al 2015, Lazzarin et al 2017).

Here, transitions theory identifies the significance of adults moving into a stage of *"unanticipated transition",* and at this stage in her patenting journey, Caroline needed to make a further major transition to accepting that in order for her son to be able to go home from the hospital, the severity and unpredictability of his illness meant that she needed to accept that qualified nurses would be providing 24-hour care in her home:

> *So last February – Lucas had got to the point where we'd got carers, and we had a small carer package giving us three nights a week because Lucas was so bad.*

Caroline had to come to terms with accepting that Lucas's increased care needs were beyond what she could manage alone at their home. However, being able to have Lucas at home rather than in hospital was dependent on carers being trained and able to care for Lucas in his own bedroom for alternate nights to enable Caroline to sleep.

As the mother of a very sick and medically unstable child, Caroline lived with high levels of stress which were juxtaposed with her gratitude for the provision of the high levels of

[7] PIC line is a peripherally inserted intravenous cannula which enables medication, nutrition and fluids to be inserted directly into the blood stream.

[8] Jej is a jejunostomy which is a feeding tube which is passed via a surgically made incision in the abdominal wall. The feeding tube is passed through the stomach and into the jejunum section of the small intestine and which increases the chances of the enteral feed, fluids and medication to be absorbed.

ongoing support that enabled Lucas to be at home rather than in hospital (Brenner et al 2015). However, this also had an impact on her personal life and left her feeling exposed to the scrutiny of others in her own home:

> *I'm surrounded by nurses – which we couldn't be at home without them – sometimes it just drives me to distraction because there is NO privacy.*

4.2.6 Further deterioration in health and palliative care

Lucas's health had continued to deteriorate to the point of hospitalisation and the need to be ventilated and at one stage he spent three months in a high dependency unit. Caroline struggled with the desire to have her son free from the complexities of medical interventions. These feelings were contrasted with Caroline's conflicting emotions of the precariousness of technology and medications that were keeping him alive. Caroline loved her son deeply but struggled with all the technology that had become a necessity to keep him alive and whilst she understood the necessity of the central line for total parenteral nutrition (TPN) and for intravenous drugs to be administered, it was difficult for her as a mother to reconcile seeing her child attached to this equipment (Brenner et al 2015). She wanted to have the spontaneity of being able to cuddle her child without having to ensure that the central line administrating his nutrition did not become dislodged:

> *It's a funny old life but it's the life that we've got but I sometimes just look at all those tubes and I wish that I could just rip them all out and have him au natural but they're his life support – they keep him going you know.*

The family stress and coping theory (Hill 1949) provided a theory to make sense of the narrative provided by Caroline; whereby the stressful event (variable A), combined with the resources that were available to Caroline (variable B) which were her friendship group of other mothers who also had children with special needs, and Caroline's own perceptions of her changed circumstances (variable C) of being the mother of a child with a life limiting illness, were added to variable X which represents the degree of the crisis that the family has faced, which for Caroline was the end of her relationship with Lucas's biological father. This sequence of the ABC-X model of family stress resonated with each of the stages of Lucas's life that Caroline had wanted to share with me through the interview.

4.2.7 Summary

This case has presented the journey of Caroline's life from becoming a mother and the following seven years of her son Lucas's life. It demonstrates the complexities for both

parents of adjusting to the realisation that their child who had been born perfectly healthy in every way after a normal pregnancy had become severely disabled following a viral illness, with no hope of recovery. During the subsequent years, both parents had come to terms with the knowledge that their son was on a palliative care journey and would have a shortened life. For Caroline, the importance of having social contact and developing friendships with other parents in similar situations; provided a valued source of emotional and psychological support; whereby the parents in this group could offer support to each other and provide information and exchange knowledge as needed. The palliative care journey and the funding of a care package to enable Lucas to receive high quality specialised support in his own home, enabled Caroline, Lucas, and their extended family to be able to spend quality time together in the community as opposed to living out the remainder of Lucas's life in hospital.

4.3 Connie

4.3.1 Introduction

Connie was the mother of Andrew aged seven years. Andrew had been born at 35 weeks gestation by emergency caesarean section due to intrauterine growth restriction. He had been diagnosed with SWAN (Syndrome without a Name), autism, asthma, severe developmental delay, abnormalities of his feet which required the use of specialist boots to support the muscles in his feet, and impaired vision which was corrected using glasses. Andrew was doubly incontinent. He had no verbal communication abilities but was able to communicate to a limited extent using a computer speech device which was called "his voice" by the family. He was eternally fed via a Gastrostomy – Jejunostomy (GJ) tube. He had significant gastrointestinal abnormalities and had had prolonged inpatient admissions to a tertiary specialist children's hospital.

Andrew attended a local special needs school and was in the class for children classified as having profound multiple learning disabilities (PMLD). Due to the extent of his disabilities Andrew received one-to-one support from a learning support assistant during the school day. Connie and her husband were also parents to a younger boy aged four years who had been diagnosed with high functioning autism but was able to attend a mainstream school.

4.3.2 Journey to parenthood: A *"very bumpy"* journey

Connie was 39 years old when Andrew was born and 41 years old when her younger son Bobby was born. Prior to becoming pregnant, Connie had worked as a specialist nurse in adult nursing at a large NHS hospital. Her husband worked full time for a local university. Although Andrew had been born five weeks prematurely, initially his difficulties were unknown, and his parents believed that he was a healthy infant. However, as early concerns around his developmental delay and muscle weakness arose, Connie and her husband sought more specialist opinions. They found the protracted processes of seeking support from their GP and waiting for appointments with specialist paediatricians and subsequent investigations to establish a diagnosis and prognosis for their son extremely stressful:

> *I think initially it was around being believed. It's been **a very bumpy journey**. The anxiety related to having a three month wait for it to be reviewed in London – and then the uncertainty of who you believe.*

The experiences of Andrew's parents concur with those of other parents who had waited for long periods of time for specialist investigations and test results in the hope of establishing a diagnosis for their child (Kishore et al 2011, Neece 2014). However, for Andrew's family, the genetic screening, which had taken many months before the results became available, had not led to a diagnosis being confirmed. This had led to great disappointment and concern for both Connie and her husband, as the lack of a formal diagnosis increased the uncertainty of Andrew's prognosis. For Connie and her husband, Andrew's development of additional symptoms posed more questions and greater uncertainty for his long-term future.

As Andrew had developed additional symptoms and had failed to meet the expected fine and gross motor development for his chronological age; an increased number of health professionals at a local and a tertiary level became involved in Andrew's assessment and ongoing care. This caused extra challenges and stresses for Connie in terms of effective and timely communication between professionals; due to the lack of a coherent working team; and an increased burden of maternal responsibility to ensure that each professional had access to the most current information about Andrew.

4.3.3 Dealing with stigma.

> *It is really interesting how people's views are different and the impact that it has. People will see a disabled child when you're in a wheelchair but when you're in*

a pushchair his level of disability hasn't changed but as he's in a pushchair therefore he must be ok and he's just misbehaving or whatever.

Andrew's parents had made the emotional and psychological adjustments between previously having felt comfortable with the normal appearances of being parents pushing their child in a pushchair, to the unwanted attention from passers-by to Andrew being in a wheelchair. They had struggled to get funding for a wheelchair for him once he had outgrown being able to be seated safely in a double pushchair with his brother; because Andrew was able to walk short distances unaided, he did not initially meet the criteria for the local wheelchair service.

Andrew can be in his wheelchair and Bobby can walk; but I can't manage them for an hour or two at a playground on my own, but obviously the membership for the zoo costs money whereas going to a playground doesn't. It is the more structured things that tend to cost money – so yes, the cost implications of that.

The ABC-X Model of Family Stress and Coping (Hill 1949), identified ways in which families can cope with stressful events in their lives when they are given the appropriate information and support. The X in the model represents issues which can cause individual families to experience levels of crisis that the family unit are unable to manage and cause the family structure to break down. For example, Andrew's parents used their benefit payments to pay for additional help at home during the school holidays to enable Connie to take both of her sons on trips out of the home and to give her support, without which she had not felt able to manage the needs of both her children.

Starting to use the wheelchair for Andrew was an important turning point for Connie and her husband in their acceptance that their son was disabled. Despite intensive physiotherapy from birth, the challenges that Andrew had faced with his physical mobility and low muscle tone could not be overcome sufficiently to enable him to walk unaided for long periods. In addition, one benefit of moving to using a wheelchair provided the visual cues to the public that Andrew had special needs, which might not have been obvious from his physical appearance. This provided some validation for Connie and her husband of the electronic support that Andrew needed to have with him:

Quite a few people comment that he tends to have his "voice" (electronic speech device) with him – it's "ooh – he's even got a computer with him out shopping……" and it's actually said like that so it's not a compliment – it's kind of – "oh you've got a child addicted to a computer……"

Connie and her husband still felt that they were viewed with curiosity by members of the public; this added to their levels of stress of being parents to both Andrew with severe autism and additional complex needs and his younger brother Bobby. Research exploring the experiences of parents of children who are diagnosed with autism, confirm that parents can suffer from high levels of stress which can affect the whole family (Postorino et al 2015, Roblyer 2017).

4.3.4 Parental hopes, dreams, and aspirations

> *I'm probably going to have the rest of my career out now because I can't see how I could juggle working and the complexities of Andrew's needs.*

Connie had prioritised the needs of her children over and above her aspirations for her career. She had accepted a different future for herself than the one that she had previously envisaged and planned for. She had initially anticipated returning to work as a school nurse, which would have provided the flexibility to enable her to care for her children in the school holidays. For many families, who have a child with continuing healthcare needs, working parents require employment flexibility to be in place to enable parents to attend the numerous healthcare appointments needed to monitor their child's condition or to take them to therapy appointments (DeRigne et al 2010, Stabile et al 2012). For Andrew's family, having one parent not working also had significant financial implications as shown in other studies (Burton et al 2017). Connie had given up her job as a nurse and devoted her time and energy to caring for her two sons, who both had been diagnosed with continuing health care needs, however this had financial implications for the whole family:

> *"financially obviously that does have an impact because obviously we were hoping to have 2 salaries but I guess in a way our life style has changed anyway having disabled children – for instance holidays abroad – holidays away aren't possible at the moment – the only thing that we do is go down to K's parents for 2 – 3 days and that's as far as we can venture with the boys anyway – so life circumstance has dictated that as well as finance – but we certainly wouldn't have the money for foreign holidays and things like that really"*

In common with other parents who have children with continuing health care needs, Connie had identified the financial pressures for her family (Beresford et al 2007, DeRigne et al 2010, Contact 2017). The costs of having a child who, for example, is incontinent, is considerable, as the local continence service for children only allowed for Andrew to be provided with four nappies per 24 hours. This was insufficient for his needs; and his parents

were funding the additional nappies needed each day. Their financial costs also included the costs of travelling to medical appointments both locally and at tertiary centres, the cost of car parking and the wear and tear on their car of needing to travel for long distances (Stabile et al 2012).

4.3.5 Facing the future and continuity of care.

> *I remember a conversation with our Neurologist. He said we will certainly know more about his future by age 15. He is slipping further and further behind his age range. I suspect that he will need lifelong learning and obviously me and my husband have had to get our heads round that.*

As the years passed, and the severity of Andrew's long-term needs became more apparent, Connie and her husband became increasingly anxious about Andrew's future:

> *I do feel quite fearful for the future as to how his needs will be met with everything being cost cutting and everything being reduced.*

The anxiety of Andrew's parents is shared by many other parents of children who have complex needs, with regards to the long-term funding for the support and care that their child will need for the duration of their lifetime. Research on the resources needed to support children and young people with disabilities through to adulthood, demonstrated that at the current time there are insufficient resources to meet the needs of disabled children and their families in the UK (Contact 2017). For this family, the changes to local services had meant that Connie and her husband had had to form new levels of confidence and trust in different services than those that they have known previously which was challenging for them and their son, as Andrew displayed high levels of stress and anxiety when the familiarity of attending appointments with health professionals changed to unfamiliar settings:

> *Continuity is really important. Change in venues upsets him so much because he is so anxious about hospital appointments. Different people have got different objectives for him and working towards things in different ways doesn't really help.*

As an experienced mother of a child with complex needs, Connie was able to clearly understand how Andrew reacted to new unfamiliar clinical surroundings and new medical staff, as she sought to work in partnership with medical staff, which is a factor upheld in other research related to the needs of children with complex needs (De Geeter et al 2002). Connie expressed her frustration at the lack of communication between professionals and the need for therapy and medical staff to have clear and agreed objectives for Andrew to work

towards both at school and at home. For both Andrew and his parents, the importance of being able to build trust with the same staff was important for enabling progress to be made with clarity of objectives being made available.

4.3.6 *"My children are a blessing".*

Despite the challenges of bringing up two children with additional needs, Connie was able to recognise and appreciate the benefits that being the mother of these two young boys had been for her and for their friends, family, and local community:

> *I think that **my children are a blessing**. I'm amazed by how they cope with life and the way they are with other people. There is so much that the world can learn from them and people do learn from them.*

Connie was able to reposition herself as a mother of two disabled children who was able to see their worth and be appreciative of her privileged role as being their mother (Green 2007, Ryan et al 2008). As a family, they were regular attenders at their local church and received acceptance and support to enable Andrew to attend a small group in the Sunday school which was specifically for children with additional needs.

The theories of identity theory, family stress and coping and the transition theory each enabled me to gain greater understanding, and to make sense of the experiences that Connie and her husband had had, as they adjusted to Andrew's changing health needs. Connie's previous identity as a specialist nurse, with autonomy in her practice and previously being used to having a certain level of professional respect from her colleagues and patients; came to an unexpected end, as she made the decision to stay at home full time due to the demands of Andrew's health. This decision was made due to the multiple hospital appointments that Andrew needed to be taken to, which for Connie made returning to her previous professional life untenable.

Transition's theory provided a means for understanding how Connie and her husband had moved from the *"anticipated"* transition of becoming parents, to the *"unanticipated"* transition of a premature delivery and their gradual transition to becoming the parents of a child with special needs. Connie's major source of "*support"* came from the local network of other mothers of children with special needs, who became her trusted friends. They supported

each other and did not judge each other's parenting styles or their children's behaviours which was an important source of support for her.

Each of the mothers and the young people identified the importance of their family and friends for providing essential support to them. Connie had had limited contact from her own mother after her son's diagnosis, which had been a source of great sadness for her and her husband. The importance of family and social support for parents caring for a child with a disability has been identified in different studies from both a UK and a global perspective (de Araújo et al 2016, Bray et al 2017, An et al 2018). The needs of parents for support, acceptance, feeling included in their community and social circle, were each of enormous importance in helping parents to build resilience in themselves and for their families and to continue to cope during the complexities and stresses of being the parents of children with continuing care needs.

The ABC-X model identified the stressful event of Andrew's birth and subsequent diagnosis (variable A), with the resources of their personal Christian faith and the support from their local church and friendship group (variable B), the parents' perceptions of their family's circumstances (variable C) and (variable X) the amount of crisis which resulted, which unfortunately for this family contributed to the estrangement of the maternal grandmother, who resisted Connie's attempts for her mother to develop a relationship with her grandson, and at the time of the interview, this situation remained unresolved. They did have a great deal of support from the paternal grandparents, who visited regularly even though they lived in another part of the country.

4.3.7 Summary

This case study has outlined the journey for one family of having two children with additional needs, one of whom had complex and lifelong physical health needs and a severe learning disability. The journey to parenthood as mature parents had taken an unexpected turn as each of their two children had been diagnosed with additional needs. The journey following Andrew's premature birth had been frustrating, confusing, and concerning for Andrew's parents as they sought answers for the range of symptoms and increasing levels of need for Andrew; and as they established professional relationships with a range of health and education professionals.

Initially they had believed that Andrew's developmental progress would be normal for his age, however as he failed to achieve the expected gross and fine motor skills in the first year of his life; Connie and her husband had to readjust their hopes, dreams, and expectations for their first child. As the gaps between Andrew's abilities and his peer group grew, Connie and her husband began to accept the realisation that Andrew would need lifelong care and would be unable to live independently. However, Connie was able to express her feelings that both her children were "a gift" and that family and friends had all benefited from getting to know them. They had all learnt essential skills of how to communicate with Andrew as a non-verbal child by observing his body language and the few Makaton[9] signs that he had learnt at school which enabled him to convey some of his needs.

4.4 Amanda and Orlando

4.4.1 Introduction

Amanda was the mother of a fifteen-year-old son Orlando and a seven-year-old daughter Lucy. The children had different biological fathers. Amanda had had a difficult relationship with her daughter's father and was no longer in contact with him. She was separated from Orlando's father but maintained close contact with him. Orlando had an undiagnosed genetic disorder which resulted in global delay, epilepsy, speech and language delay, and low bone density which required regular intravenous treatment. He was identified as being part of a group of children called SWAN (Syndrome without a Name). Amanda had had a normal pregnancy, and Orlando had been born at term with no identified issues at the new-born checks or at the routine six-week development check. Lucy had no health issues and attended a mainstream primary school.

4.4.2 The journey to parenthood

Although Amanda's relationship with Orlando's father had ended, he had moved to an address in the local area and continued to see his son each week. Consequently, both parents had maintained a relationship which enabled Orlando to benefit from the love and care from both of his parents:

> *Basically, by the time he was two I was on my own – and so I was doing all the appointments and everything on my own. I'm really lucky in that his Dad has him*

[9] Makaton is a language system of communication which uses signs and pictures to enable children or adults who are nonverbal or who have limited verbal communication skills to communicate their wishes to others.

for a week in the summer….I'm a single parent – it's the three of us. My parents help us quite a bit because obviously I'm on my own. Orlando's father sees him once a week.

A UK profile of families who have children with a disability, confirmed that parents are more likely to be single parents if they have a child with a disability than those families without children with additional needs (Contact 2017). However, although living on her own with both children, Amanda was able to receive and benefit from the practical support offered by her parents and from Orlando's biological father who all lived locally.

In the early weeks following Orlando's birth, Amanda was unaware that he had any developmental impairments:

He was my first child. The Health Visitor picked up - at three months old he wasn't hitting his milestones. I work in a special needs school now and I have a lot of knowledge but at the time I didn't know - I had had no other experience.

At an early stage, the Health Visitor was able to identify that Orlando had low muscle tone and was developmentally delayed for his chronological age; therefore, appropriate referrals were made for Orlando to see a community Paediatrician for further investigation. Amanda was aware that as a first-time mother, she did not have any previous experiences to draw upon which would have alerted her to any concerns related to Orlando's development.

4.4.3 Parental hopes, dreams, and determination

It would be lovely if he could do something that would help him to earn some money, but I don't know whether he would be able to. It would be really nice if he could be involved in something as an adult whereby, he could contribute in some way.

Amanda understood that Orlando would need lifelong care and support. Her concerns regarding Orlando's future concurred with the views of other parents who needed long term care and whose futures were uncertain (Kingsnorth et al 2011). Whilst Amanda was aspirational for Orlando within his abilities; as a teacher working at a special school, she was also realistic about some of the challenges ahead:

Until I am one hundred percent happy, he's not going anywhere. I'm hoping that there will be support and there will be money available to support that. The first stage for him is making the transition to the college – which is already starting.

In common with many other teenagers, the gap between school and leaving education was being met for Orlando by the provision of a place at a specialist college locally which could meet his specific needs. Amanda was determined to ensure that Orlando was provided with the best possible opportunities:

> *He will go to College and will have another three years. They're hoping to get some places up to twenty-five but there's a big "don't know" –the transition is something that you have to really really fight for. Sometimes if you don't kick up a fuss you don't get the things you need and that is just a real shame.*

For Amanda, a future dilemma was the choices that she would need to make if the options for support for Orlando were not suitable to meet his needs. She recognised the importance for herself and her own career and being able to go to work: *"I have never wanted to not work";* whilst acknowledging that she might need to face a future as a long-term caregiver.

For many parents who have children with complex continuing health needs, having employment which allows the flexibility to be able to attend medical appointments with their child, whilst continuing with their career can be difficult to achieve (DeRinge et al 2010, Stabile et al 2012, Burton et al 2017). This is particularly true when the health needs for the young person are met by a range of health professionals in both local and tertiary centres, which requires the attendance at numerous review appointments both close to home and further afield, such as in Orlando's case.

Amanda's experiences of the complexities for her as a single parent of attending multiple appointments for Orlando on her own is not uncommon; and correlates with the findings from other research studies in this field (Giovagnol et al 2015). The strain for parents coping with having one or more children with additional health needs can be immense and the complexities of their children's health can cause physical, emotional and psychological symptoms to become evident for the parents, which can manifest itself in the symptoms of headaches, anxiety, tearfulness and poor sleep (Barfoot et al 2017).

4.4.4 The importance of family and social support *"I'm very lucky"*

> *I think that I'm very lucky because I have really amazingly supportive parents. When you have moments – when it does all become overwhelming; I've really learnt through the help of my parents how to manage is to look at the practicalities. I'm not capable of ignoring the emotional factors because that is not in my makeup.*

The uncertainty of being a single parent for two children, when one of the children has complex continuing health care needs, can bring immense levels of stress for the parent concerned (Hsiao 2018). Amanda's immediate family and close circle of friends were an important source of strength and support for her:

> *I just met a group of four or five other parents - mums who live in the area where I live, so that's really good. We get together socially now, and we've all got children of different ages but we share - we pool our information – so that's quite useful again just for practical and emotional things.*

The ABC-X model of family stress and coping (Hill 1949) provided a lens by which I would make sense of Amanda and Orlando's experiences. Amanda had thought that her young baby was "normal", but the assessment of the Health Visitor started the processes of assessment which led to the unexpected diagnosis of low muscle tone and severe developmental delay (variable A). The resources available to the family (variable B) were the maternal grandparents who lived locally and who consistently gave physical, emotional, and financial support to the family. This was augmented by the provision of respite care when Orlando became a teenager, which enabled Amanda to spend time with her younger child (variable C). Amanda was aware that her son was on the cusp on transition to adult services and that the future funding and provision of services for her son were uncertain (variable X). The outcome of the early months following Orlando's diagnosis was the ending of the relationship between Amanda and Orlando's biological father, but he had remained in close contact with the family and visited each week.

International studies have provided insight into the life changing impact that having a child with a disability can have on the parents and wider family, with the emotional impact lasting for months and years for the parents (Whiting 2012, Whiting 2014, Packingham 2016, Yamaoka et al 2016, Contact 2017, House of Commons 2019). The impact on the mental and physical health of the study participants included their experiencing depression, anxiety, low mood, back pain, feelings of isolation and despair and fear for the future, which correlates with the findings from different studies by authors across the globe (Pedrón-Giner et al 2013, Whiting 2014, Yamaoka et al 2016, Packingham 2016, Findler, Jacoby et al 2016, Contact 2017). With her extensive experience of working as a teacher in a special needs school, Amanda had both professional and personal experiences of understanding the long-term benefits and impacts for parents and siblings of supporting a child with a disability in their

family and was acutely aware that adjustments and her decisions as a parent to ensure the best outcomes for her son would need to continue for the long term into the future.

4.4.5 Summary

This case study has demonstrated the complexities for Amanda, as a single parent, in caring for her teenage son with a significant learning disability, impaired mobility, speech and language delay and epilepsy, whilst also caring for her young daughter and holding down a career as a teacher in a special school. Amanda identified the importance of social and family support and her overarching anxieties concerning the longer-term future for Orlando's future which included the funding of appropriate services to support Orlando educationally, medically, and socially as he made the transition from childhood to adulthood.

Amanda, Orlando and Lucy had been supported through the roles that the maternal grandparents and the extended family members had contributed in providing practical, emotional and physical support which had enabled Amanda to continue with her roles both as a mother to two children as a teacher working within the field of specialist education. The social impact of being able to form a friendship group locally with other mothers in a similar situation to her own; had provided solidarity and sense of inclusion and acceptance through shared knowledge and expertise were essential for Amanda's psychological and emotional wellbeing.

This case study has raised the important issue for Amanda; of having a son with long term health needs who was on the cusp of adulthood. Her concerns were related to how her teenage child's specific health, education, employment, and social care needs would be provided for in the long term and whether the provision offered will be sufficient and tailored to meet the young person's individual needs. When considering the human costs; Amanda was anxious as to the personal costs for her as Orlando's mother, of potentially needing to give up her career to care for Orlando; if the future funding or provision available were to be insufficient to meet Orlando's long-term needs.

4.5 Edward and Melanie

4.5.1 Introduction

Edward was a 16-year-old boy who had been born prematurely by caesarean section following the obstetric emergency of a placental abruption[10]. Early brain scans following Edward's birth showed areas of ischemia in the brain which indicated that significant neurological damage had taken place and a diagnosis of Cerebral Palsy was made. Edward had an older sister Claire, who had also been born prematurely. She was six years older than Edward; but had grown up with no lasting impairments from her premature birth. Edward's disabilities included: urinary incontinence due to impaired bladder function; and a weakness on the left side of his body which had resulted in reduced mobility and function of his left hand and left leg.

The anticipation of giving birth and assumptions of having a normal child are part of achieving the identity of becoming a mother. However, for the mothers in this study, their reality of this anticipated journey and the development of their identities as parents was disrupted. The research literature on 'birth stories' is extensive and demonstrates how childbirth is a highly emotive event that can involve complications. Three of the mothers including Melanie, had experienced premature labours and deliveries and this had impacted on their anticipated identity as mothers (Arnold et al 2013, Steyn et al 2017, Ionio et al 2016). Melanie, Connie, and Kim had each had an emergency caesarean section due to foetal distress, which for each set of parents had resulted in their babies being born prematurely.

4.5.2 Parental aspirations

Work by Huang et al (2010) explored the experiences of mothers following the confirmation that their child had been given a diagnosis of Cerebral Palsy. The researchers identified that the confirmation of the diagnosis is a time of great distress for families when the future of the deficit is unclear and the prognosis for the child uncertain. However, Melanie's focus and determination in supporting Edward was all-consuming and unrelenting and contributed to Edward's abilities to successfully manage his disabilities:

> *We just had the attitude from the beginning – Edward can do it unless he proves that he can't. He didn't walk until 3 and a half and then one-day Edward just got up and walked across the room!*

[10] Placental abruption is an obstetric emergency where the placenta comes away from the uterine wall without warning leading to haemorrhage and the risk of death to the infant and the mother.

Both of Edward's parents had the optimism and expectation that their son would defy the predictions that had been made by medical staff in the hours following his birth. Many parents who have children with a disability report high levels of stress but also a sense of achievement when their child reaches milestones that were previously predicted to be unobtainable for them (Hastings et al 2002, Findler et al 2016 a,b). This concurred with the experiences of Melanie and her husband when, although developmentally delayed with fine and gross motor skills, Edward had gradually attained the skills which had been predicted by health professionals as being unachievable for him.

4.5.3 Edward's hopes, dreams, and determination

Edward identified that his personal aspirations were centred on his natural talent and passion for sport. Edward's ambitions were to play cricket professionally for the British disabled cricket team, but that he was also keen on travelling:

> *I just love sport. For cricket I play for ……and for football I play for ……disability side. I want to play cricket for England or reach the Paralympics. Also, to get a good job. I also am interested in History and Politics.*

Edward demonstrated the strength of his commitment to sport and his ambitions to achieve playing one of his chosen sports at a very high level; whilst at the same time thinking about his longer-term career. As with many other young people at the age of sixteen, Edward was exploring career choices and ambitions for his future and had identified some possible options to pursue which included his talent for sport, although he was less clear about the type of paid employment that might have been of interest to him. Being able to decide on a career choice has been identified as being one of the key tasks expected of young people during their adolescent years (Christie et al 2005).

Edward had not allowed his physical weakness to deter him from his own personal goals. By playing for teams that accommodated and encouraged his participation in both football and cricket. Edward's passion for sport had been rewarded by being part of a team and gaining the social, emotional, and physical benefits from being part of local teams. Research which explored the determinants of social participation for young people with Cerebral Palsy identified the importance of sports activities to promote a sense of wellbeing and inclusion (Kang et al 2010). These findings resonated with the positive experiences that Edward had

gained from being active in both local cricket and football teams. However, due to the complex bladder surgery that he had undergone, contact sports carried potential risks of damage to his augmented bladder, which Edward and his parents had had to weigh up and balance and to find a compromise which was an acceptable risk. However, they remained very proud of what Edward aspired to do:

> *Since the surgery, he can't do rugby or hockey which for Mr Sporty wasn't ideal – but he got used to it. He plays football and cricket – he's in goal!*

Although Edward had expressed a desire to play sport professionally, his mother had concerns as to whether his continuing health needs could limit her son's opportunities to travel to some countries. The links between young people with a disability and their interests in playing sports and how this helps to develop their identity of themselves is relevant here for this young man and his family (Fitzgerald et al 2009). The role that the family can play in supporting the young person's participation is crucial with Edward's mother making the commitment to drive long distances to enable her son to attend practice sessions and matches as he worked towards his ambition to play for England in the disabled cricket team. This support from his mother being willing to take the time and spend the money necessary for his sports equipment and clothing had strengthened the bond between Edward and his mother and developed his self-esteem as he continued to develop in his natural talent for sport.

Parents and children's experiences of intermittent self-catheterisation identified the importance of the child or young person being self-reliant and competent to undertake their own care; to increase their own self-esteem and independence from parents or carers (Chick et al 2012). However:

> *several times when we go to the GP – they're lovely but they - but you know it's much too complex – the complex bladder issues are too much for them so they just send him to the …….*[Hospital]
> *but I'm tied and the older Edward has got – it's awkward for me doing the willy stuff - he's very good and he accepts it*

In travelling abroad for any prolonged period, Edward would need access to regular supplies of disposable urinary catheters, hand sanitation gel or handwashing facilities and a means by which the used catheters could be disposed of safely:

> *We go to India a lot and he likes that, but I know the Australian system and the medicals you have to go through and everything and I don't think that he'd get in – I don't know – so I do hope that that doesn't cause problems.*

The balance between aspiration and reality for Edward's future ambitions was dependent on whether his continued health issues would prove to be a barrier to him playing sport in Australia. From the time of Edward's birth his parents were determined to support his aspirations in whatever way they could. However, for Edward, the complexities of coming to terms with his lifelong physical weaknesses had been challenging:

> *When they did his profile for the disabled cricket, they picked up on his left hand and he was really upset about it. He said, "I spend my life to make it so that nobody notices anything; and they knew exactly what to ask me to expose all of my weaknesses". For some reason it really upset him, so I said, "But it worked because you got your grading to enable you to play cricket" and he said "yeah".*

Melanie was able to comfort Edward and provide the emotional and psychological support that he had needed following the crucial selection process, which determined whether or not Edward would gain a place on the cricket team for disabled players that he yearned to play for. This conversation had enabled Edward to gain a more positive and enabling perspective of the selection processes he had been through with the cricket selectors (Hanisch 2013).

4.5.4 Increasing independence

At 16 years old, Edward should have become more independent from his parents. However, although he was able to insert and remove the intermittent urinary catheters for himself; he had not been trained to insert a semi-permanent indwelling urinary catheter alone when he went into urinary retention. This was an important consideration for Edward becoming more independent (Björquist et al 2014). On these occasions, difficulties with intermittent self-catheterisation resulted in the necessity for an indwelling urinary catheter to be inserted;[11] and had resulted in an ongoing dependency between Edward and his parents that was unanticipated. Melanie and her husband were working towards changing this, however.

A systematic review which was completed by Lindsay (2016), explored the experiences of children and young people of growing up with Cerebral Palsy. This work identified the risks for young people becoming socially isolated and needing the support of their friends and family to cope with pain, and physical skills needed to manage their condition and developing independence. Both Melanie and her husband had had to learn the skill of

[11] Intermittent self-catheterisation involves the young person inserting a urinary catheter from the penis to the bladder between four and six times per 24 hours to drain urine from the bladder to avoid stagnation of the urine and the risk of urine infections from developing. The catheter is removed once the bladder is empty.

catheterising their son when needed (Chick et al 2012). They had taken on the role of "expert parents", developing the skills and knowledge to intervene when Edward developed urinary retention. This was more complex than it might be for others, due to the complex bladder surgery that Edward had had earlier in his childhood, and the risks of damage to his enlarged bladder.

Edward had also needed to incorporate the long-term effects of the Cerebral Palsy into his daily life and adjust as necessary, *"I struggle to walk long distances, have to self-catheterise four times a day. Walking on hills, or uneven ground is harder.*

The weakness on the left side of his body limited Edward's abilities to undertake some of the physical activities and required consideration when taking part in some outdoor pursuits. However, Edward was undeterred by his physical limitations and continued to work hard to play football and cricket and to enjoy recreational activities with his friends (Lindsay 2016). Both Edward and his mother had been impressed with the planning and organisation of the transition process and had felt fully included in the discussions and decisions that were being made about his care.

4.5.5 Transition to adult services

As a young person at the age of 16 years old, Edward had begun but not completed the process of making the transition to specialist urology services within adult services. He and his mother had had a positive experience of meeting the specialist transitions nurse who was co-coordinating his move from child to adult services. Although Edward's first clinic appointment in adult services had yet to take place, the planning and preparation had resulted in a positive experience for both Edward and his mother, where they had been involved and collaborated with by an experienced specialist nurse and their experiences concurred with the guidance from the Royal College of Nursing for preparing young people to move to adult services (RCN 2013).

4.5.6 The importance of family

Edward identified the importance of his friends and family and identified some of the ways in which he enjoyed his free time:

> *Meeting friends at the park or beach is fun! Enjoying a chat over coffee or a drink down the local pub is always a welcome suggestion too!! I love family*

holidays and spending time together generally as a family. We watch films together, have music sessions and play games together.

The close bond that Edward enjoyed with his friends and family provided him with essential emotional support. He was able to identify a range of social activities which enhanced his life and enabled him to relax and enjoy the company of his friends and family. Having a friendship group prevents social isolation and the risks of poor mental health and provided an additional network of support for Edward beyond his immediate family; which were important for the mental and psychological wellbeing of young people, as demonstrated by the findings from a European longitudinal study exploring the quality of life for adolescents with Cerebral Palsy (Colver et al 2015). Loneliness for children or adults is known to have a detrimental effect on mental health and psychological wellbeing. For young people with disabilities it can become more challenging for them to develop friendships and relationships and to feel accepted by their peers (Kelly et al 2016), however Edward attended a main stream school and appeared to have a small but very supportive group of friends, who supported him by offering to carry his school bag at school and accepted that his lower limb weakness reduced his speed of walking but encouraged him to enjoy recreational activities with them and he enjoyed socialising with these friends outside of school hours.

4.5.7 Parental sacrifices

The complexities of caring for children with continuing health care needs often results in parents making sacrifices of their own career progression to allow for the flexibility needed to take their children to a wide range of review appointments:

I did have a career till Edward was born – so now I run my own business, but I have a very flexible attitude to it because I've had to fit everything in medically.

The changes to the economic position for families where one or more child has a disability can be significant and detrimental to the economic prosperity of the family (De Rigne et al 2010, Stabile et al 2012, Burton et al 2017). The positivity with which Melanie was able to adapt to the change in her circumstances as a parent, enabled her to maintain an economic presence within the family; whilst remaining in control of the medical appointments that Edward needed to attend. This flexibility enabled Melanie to provide the transport and ongoing emotional support for Edward of being able to attend inpatient and outpatient appointments with Edward as needed.

4.5.8 Effect on siblings

Edward's mother had been very aware of the challenges that she had faced as the mother of two children, both of whom had been born prematurely and who could both have had long term health needs. As a young child Edward had taken the priority for Melanie's attention during hospital admissions and her older daughter had remained at home with her father. However, it was not until her daughter Claire was at university, that Melanie became aware of the impact that having a younger brother with additional health needs had had on her daughter:

> *As a family it's had a massive impact on my daughter – she's five years older so she understood everything. It was only within the past 3 years or before she went to uni that she admitted that she did resent him. I wanted her to get involved with different organisations for siblings, but she wouldn't.*

The experiences that Edward's sister had had is common and many parents feel torn when attempting to meet the needs of each of their children (Gursky 2007). However, Melanie showed sensitivity to the needs of both of her children and had worked hard to try to ensure that their individual needs had been met.

There is a large body of research that has been conducted looking at the experiences for children of having a sibling with a continuing health care need. Quality of life indicators were important measurements of the emotional and psychological implications for children. Positive findings concerning children with disabled siblings becoming more considerate and caring were noted in a number of the studies (Meltzer et al 2016, Shivers et al 2017, Perenc et al 2018, Takataya et al 2019). Two of the parents commented on the kindness and thoughtfulness of the younger children in their family towards their older disabled brother or sister, even though they did have times of being jealous of the sibling that received the most parental attention in the family. However, some studies have reported that in families that had disabled children, the unaffected siblings had increased levels of behavioural difficulties themselves as compared to other families (McCullough et al 2011, Giallo et al 2014, Hastings et al 2014, Fullerton et al 2016, Siminghalam et al 2018, Joosten et al (2019).

Identity theory provided a lens for understanding Melanie's change of identity as she gave up her previous identity of being an employee to becoming self-employed which gave her the flexibility to attend appointments with Edward. Transition's theory (Schlossberg 1981) also enabled and understanding of Edward's journey through adolescence, as he aimed to achieve his goals of achievement in sport, whilst also juggling his continence needs.

4.5.9 Summary

This case has shown a young man on the cusp of adulthood who was still negotiating the skills needed to provide his own care needs and to become independent from his parents. Edward remained determined to achieve his own personal goals and aspirations of playing sport at a high level and aiming to succeed in his career as he navigated his own path to adulthood. His friendship groups and his family provided ongoing and essential support for him as he moved from childhood into becoming a young adult; with all of the emotional, biological, and psychological changes that take place for all young people during the years of adolescence.

The resolve of both parents and their son to overcome the challenges that had arisen from the diagnosis of Cerebral Palsy; which had had a lasting effect on Edward's mobility and urinary continence, was evident as they sought to enable Edward to live as independently as possible; and to work towards the ambitions that he had identified for himself. The challenges that Melanie and her husband had experienced in the early years when they had needed to provide care and attention for both of their children during the times when Edward had required frequent hospital admissions; highlighted the difficulties for the whole family of meeting the needs of both Edward and his sister.

4.6 Suzie and Kim

4.6.1 Introduction

Suzie was a 17-year-old young woman who had been born prematurely following an emergency caesarean section due to pre-eclampsia. Early brain scans following her birth revealed that Suzie had had two cerebral bleeds which had caused irreversible neurological impairment and led to the diagnosis of Cerebral Palsy being made. Suzie's disabilities included: a weakness down the right side of her body with a significantly weakened right hand, and a weakness of her right leg and foot. These muscle weaknesses reduced her physical mobility and caused her to walk with a limp. She had a permanent visual impairment which also impeded her physical mobility. At the age of 15- years she developed epilepsy. The brain damage that was identified at birth caused Suzie to have some short-term memory impairment and learning disabilities. Suzie had an older sister who had also been born prematurely but who had no continuing health issues.

In keeping with other research on traumatic birth experiences (Coates et al 2014, Woodward et al 2014, Hawes et al 2016, Hall et al 2015), three of the mothers were able to recollect clearly and in great detail, the sequence of events that had led up to their children being born prematurely. Post traumatic trauma experienced by the mothers of premature infants is well known in the literature (Beck 2004, Shaban et al 2013). The feelings that these women had experienced many years before had shaped their identities leaving them with feelings of loss and guilt because their children had been born prematurely (Hall et al 2017, Hawes et al 2016, Coates et al 2014, Woodward et al 2014). Kim was able to recount with clarity the trauma and stress of her daughter's premature birth and the first few days of her life, when her daughter was critically ill and there were doubts as to whether her life could be saved by the specialist staff in the neonatal intensive care unit.

4.6.2 Hopes, dreams, and determination

Suzie was clear about her own ambitions for the future: *"For the past couple of years, I have been wanting to be a radio presenter".* However, in pursuing this goal, Suzie acknowledged some of the difficulties that she had experienced with her education:

> *I had to go through reception year twice; because I had forgotten how to read and how to write – I had to learn all of that over again so that stopped me from being with my own age. I'm ok with learning but it's just the issue of memory because I can't remember things very well but that's due to the brain bleed - I sometimes use those flash cards with a picture or a phrase to help me remember.*

As a young adult, Suzie demonstrated both insight and acceptance into her own learning needs, through some of the adaptations and coping strategies that she had employed successfully to address these issues. For Suzie, the learning disabilities that she lived with each day, required a longer-term approach to enable her to accept a slower pace for her to achieve the learning that needed to take place in each subject area. Suzie was aware of the impact that her visual impairment and the day-to-day fluctuations in her vision had on her ability to read, focus and to walk safely independently. The individualised transition journey from childhood to adulthood for young people with Cerebral Palsy, may result in young people continuing to require support and for the young person to accept the additional support needed, beyond the age of their able - bodied peers, as identified in the findings from a study by Björquist et al (2014).

4.6.3 Parental aspirations

Suzie's parents had a strong vision and ambitions for their daughter; however, they were practical and philosophical in their approach to supporting Suzie:

> *We've got more and more **aspirational** for her. We would like her to be as fully independent as possible and taking care of herself and having her life and not her life with her parents, but we don't know yet about the feasibility of that.*
> *She wants to go to university. She does struggle academically because of her brain injury and she may have the entry requirements to do what she wants but it might take her longer.*

Kim and her husband had recognised and accepted that the timeline for Suzie's academic attainment would be different from her older sister or her peer group and that she would follow her own trajectory which they fully supported. With their acceptance they were able to take away the pressure for Suzie to achieve academically in the same time frame as her older sister. The experiences of *"chronic sorrow"* were identified in work by Whittingham et al (2013) which considered the experiences of parents of children with Cerebral Palsy, as parents experienced and expressed sorrow when their child was unable to achieve their goals and independence at the same stages as siblings or peers.

4.6.4 Adjusting to expectations.

Despite her initial very poor prognosis, Suzie had continued to make progress in each area of her life throughout her childhood and adolescence. As a family they adjusted their expectations and time frames and came to the acceptance that Suzie's journey to adulthood would take place at a different pace to some of her peers. Over time it had become apparent to the family that Suzie's visual impairment was going to have an ongoing impact on her mobility and independence:

> *She needs support in a crowded space but it's mainly because of her eyesight – although her gait is affected and her balance. If she fell, she could really badly hurt herself. I'd say it's the vision more than anything else and coping with crowded spaces more than walking*.

The anxiety and the unpredictability of the diagnosis of epilepsy heralded another unexpected and unwanted development in their daughter's health. It is not unusual for parents to be frightened about seizures as epilepsy carries a mortality risk. People with epilepsy need to have family, friends or carers who know how and when to intervene in the event of a seizure, as identified in the findings from a study exploring self-esteem in young people with epilepsy (Lewis et al 2010, Chew et al 2017). Therefore, the development of epilepsy during her

adolescent years had impeded the steps towards independence that Suzie was aiming for. She needed to have others around her who could look out for her and care for her appropriately if a seizure should occur, as identified in a review by Surges, Thijs, Tan and Sander (2009) into unexpected deaths for individuals with epilepsy. Suzie's mother recounted the fear and anxiety that the diagnosis of epilepsy had had for her and her family: *"we are very very frightened about epilepsy"*.

For parents or carers, watching a young person have [12]a "tonic-clonic" seizure can be deeply upsetting:

> *The seizures were very distressing for her and for us because she was fully conscious through them. It was horrible because she lost her gag reflex, she had involuntary vocalisations plus the things that go with muscle effects of having a seizure.*

The interrelationship between the medication, seizures and deterioration in her physical health resulted in another obstacle for Suzie to overcome. The loss of sensation in her hand and arm had had a detrimental effect on her fine motor skills which consequently adversely affected her ability to meet her own self care needs such as fastening buttons on clothing which increased her dependence on others. This caused an unexpected and unwanted barrier to her previously attained independence as demonstrated in work from Kang et al (2010), and Lindsay (2016), which explored the experiences of young people growing up with Cerebral Palsy:

> *We've got more and more aspirational for her. As she's got older, she did so much better than we ever anticipated. We would like her to be as fully independent as possible and if that's 100% independent – fantastic – and maybe that's achievable with some support, but if she was even 50% independent – meaning maybe having two nights away from home by herself and taking care of herself and making her own decisions in life – and having her life and not her life with her parents – but we don't know yet about the feasibility of that.*

Suzie's mother was facing the challenges of supporting Suzie to become more independent of her parents as her older sister had done. Both Kim and her husband were moving towards the stages of both *"expected transition"* and *"unexpected transition",* as they were uncertain as

[12] A type of seizure where the individual falls to the ground and loses consciousness. There is violent shaking of all four limbs and the individual may suffer urinary incontinence. The individual may bite their tongue or sustain other injuries. Once the seizure has ceased, the person will be drowsy and disorientated initially.

to what steps would be necessary or achievable in order for Suzie to achieve the independence that both she and her parents had wanted for her.

4.6.5 Developing independence.

Suzie was aspirational with her plans to move away from her parental home and to live independently:

> *Well apparently, I will be getting a Social Worker next year to obviously help me with the transition from living at home to living on my own – so yeah, I have to wait for that.*

As a young person with long term health needs, the allocation of a Social Worker would enable Suzie to have impartial advice on the benefits system and support Suzie to achieve her goal of having her own independence. However, the effects on her memory and the ability to meet her own needs without prompts and reminders from her parents were a concern for her mother:

> *Sometimes I ask "have you had your medication today" but she won't remember if she's had it, so she does rely on us to help her with that* (Suzie's mother).

As part of her journey to independence, Suzie had experienced challenges which her parents and healthcare professionals worked to help her to overcome:

> *We had lots of conflict when she was younger. She didn't want to do any physio and we had to have a gap in physio for a while.*

Conflict between teenagers and their parents is a normal dynamic, however then the conflict concerns children or young people choosing not to comply with recommended treatment regimes, this can be very difficult for the parents to know how to respond. Kim and her husband faced challenges in wanting the best outcome for their daughter whilst bearing in mind her views and being prepared to make compromises. Suzie's experience correlate with the findings from a study exploring the experiences of health and well-being of young people growing up with Cerebral Palsy (Björquist et al 2014).

The adjustments made by Suzie's family were evident in the ways in which they adapted their parenting style to meet her needs:

> *I think the isolation is the biggest challenge for us – we are the sort of family that just gets on with it as if there is nothing wrong.*

For parents with children with continuing healthcare needs, needing the support and advice from other families as part of a social network is common. Suzie's family demonstrated high levels of cohesion and resilience in striving to continue with their plans and to adapt as necessary to accommodate the special needs of their daughter. This was shown in the challenges for Suzie of being intolerant to using the splints that had been provided to support her weakened right hand. Her parents had to balance her discomfort and refusal to wear these as against the recommendations by Therapists. As a young adult she was demonstrating her ability to be active and autonomous in her decision-making and make her own choices about compliance with treatments.

Two of the mothers were juggling their identities of being the mother of a child with a continuing health care need, together with their roles as professionals holding down full-time jobs in stressful and demanding roles. Kim, who had previously worked as a qualified nurse, revealed her need to separate herself from looking at research on her child's condition and to consciously be a *"parent rather than a nurse".* For each of the mothers, their anticipated transition to parenthood became unanticipated as their child's diagnosis was revealed and the reality of their worlds changed to include hospital appointments, medication and learning technical skills to support their child's medical needs. Kim and her husband worked closely together to support their daughter and continuously looked for solutions which would enable their daughter to reach her future goals of greater independence.

4.6.6 The importance of family

As with many young people, Suzie had a strong attachment with her family and enjoyed having time with them relaxing and socialising:

> *With my family I like to go shopping or to the park or the beach because we live an hour away from London. We go for days out in central London quite regularly because my sister lives there now – we visit her quite regularly.*

Maintaining and sustaining the bond with her older sister was an important part of her life as a young person. Despite the limitations of her physical health and stamina, Suzie was able to take pleasure in a wide range of activities outside of her life at school, which contributed to her emotional and psychological wellbeing, a factor identified as one of the key determinants of health for young people with Cerebral Palsy (Kang et al 2010). Her relationship with her

parents was strong and she demonstrated a growing insight and maturity in how she appreciated the support that they were giving to her:

> *As a teenager I find my parents incredibly annoying – so I don't tend to listen to them too much but when I do it actually does help. It's my dad that has actually helped me to go the gym a lot more. That's to help me to build up my muscles a bit more – get a bit stronger.*

The ABC-X model of stress and coping provide a lens to gain understanding of the journey of this family. The stressful event (variable A) of a premature birth and a very turbulent neonatal journey when the parents were unsure whether Suzie would survive, together with the resources of strong family cohesion (variable B), the parents' perceptions of their daughter's health and continuing healthcare needs (variable C) and variable X (the outcome), which resulted in the parents making the decision that Suzie's father would become the parent who stayed at home and supported Suzie by taking her to numerous hospital appointments and overseeing the co-ordination of the different services who were providing healthcare for Suzie.

The focal theory of adolescence identified the benefits for young people of being able to concentrate on one aspect of life at a time, but the unexpected diagnosis of epilepsy when Suzie was 15 years old, made this theory unsuitable for this young person as she adjusted to the medication and implications of having seizures as a teenager.

4.6.7 Summary

This case study has shown the complexities that this family had faced when supporting their 17-year-old daughter with Cerebral Palsy, visual impairments, and a relatively recent diagnosis of epilepsy. As a young adult, Suzie was testing out ideas for her future and had clear ideas as to the ways in which she wanted her career to go. She acknowledged the physical challenges that she faced and the issues that her learning disability and visual impairment had posed for her education; but was able to demonstrate her resilience in seeking strategies to enable her to adapt and cope with the deficits of her short-term memory impairment.

Kim and her husband continued to support Suzie with her ambitions to become more independent; whilst balancing this with their overarching parental concerns of needing to

prioritise Suzie's safety particularly in relation to her seizures and memory impairment. Both the parents and their daughter were optimistic and aspirational for the future and were taking small steps to enable her to gain the increasing independence that she craved.

4.7 Elizabeth

4.7.1 Introduction

Elizabeth was a young 20-year-old woman with a complex medical history. She had been diagnosed with Triple X syndrome (see glossary for details), hypermobility, Raynaud's disease, Asperger's Syndrome, and deafness in one ear. In addition, as a teenager she had also been diagnosed with a progressive neurological condition which she described as being similar to Multiple Sclerosis, but which was still being investigated and had not been formally diagnosed. At birth Elizabeth had been diagnosed with mild Cerebral Palsy which in late adolescence had resulted in her bladder becoming affected. This had been treated by the insertion of an indwelling supra pubic catheter. Her physical mobility was also adversely affected by Spondylosis of the lower spine and chronic pain. Elizabeth's sleep pattern had been affected by sleep apnoea and she was waiting for a non-invasive ventilator mask (Continuous Positive Airway Pressure CPAP) to support her breathing during sleep. Elizabeth identified that she had been diagnosed Obsessive-Compulsive Disorder and anxiety which affected her mental health and psychological wellbeing and resulted in further aspects of her health that she needed support with.

4.7.2 Hopes, dreams, and determination

Despite the complexities of her medical history and co-morbidities, Elizabeth presented as a determined and focused young woman who had clear goals and aspirations for her future. Elizabeth had experienced a recent deterioration in her health which had led her to undergo emergency surgery[13] which had required her to have a prolonged absence from her attendance at university. This unexpected surgery had taken longer than anticipated to recover from; but despite this, she had continued as an undergraduate student.

Elizabeth demonstrated her determination to succeed with her academic goals by returning to university and being willing to re-do the second year of her degree which had been

[13] The emergency surgery had been for acute urinary retention and for the insertion of a supra pubic urinary catheter.

interrupted by the unexpected serious and life-threatening illness of sepsis. By taking this course of action, Elizabeth demonstrated a high level of tenacity and determination to overcome the adversity of her ill health and to continue with her degree studies at university.

Elizabeth's recollection of her school years showed a young person who was engaged in activities which were normal for a young person of her age. She had attended a mainstream school and studying a range of subjects in the sixth form between the ages of 16-18 years of age:

> *Until three years ago I was fairly average really. Although born with the majority of these conditions, apart from my deafness none really affected me or at least I didn't know of them.*

However, as a teenager, Elizabeth noticed deterioration in her memory function which had a detrimental effect on her ability to study and subsequently led her to make changes to her academic subject choices which were better aligned to her strengths and abilities. In taking this course of action, Elizabeth demonstrated a high level of maturity in recognising the impact that her impaired memory function had had on her ability to continue with her original choice of subjects. She adapted her subject choices accordingly to ensure that she was able to gain the grades needed to secure a university place for her chosen course.

4.7.3 Progressive changes in physical health

Over the period between the ages of 18 and 21 years; Elizabeth had noticed a gradual but significant deterioration in her physical health. These changes had led to an adverse effect on her daily life and had made her more disabled than she had been accustomed. The changes in her muscle strength had changed Elizabeth's body and had forced her to adapt to new ways of being mobile and to accept a new level of mobility and independence:

> *Now I find it hard doing simple things and have ended up going from running and athletics to a walking stick which now is only for indoor use as I need a wheelchair full time.*

Despite this deterioration in her health, Elizabeth had adapted to the changes in her health by learning the new skills needed to become a proficient wheelchair user. Elizabeth acknowledged the challenges of undertaking tasks in her daily life that she had previously taken for granted and had been able to perform with ease:

> *Small things like washing my hair I find tiring and when it's not washed for a while you just feel manky. I have a catheter to take care of, have to make sure*

that I don't fall asleep in certain positions, daily medication which I have to take three times a day or more depending on the issue.

I have support workers from the social services who come twice a week currently and then I usually think I'm doing fine but I think everyone knows that I'm not always.

Whilst it is acknowledged that young people with chronic illnesses can experience a lower quality of life than their non-disabled peers (Garnefski et al 2009); for Elizabeth, the provision of additional support to develop her life skills and to cope with the mundane daily tasks of life, enabled her to continue to play an active role in her own life and to develop her newfound independence of living away from home for the first time. In accepting the help from support workers, their intervention enabled an objective assessment of Elizabeth's abilities to meet her own hygiene and nutritional needs to be performed and for additional support to be put into place as needed.

As Elizabeth's muscular strength had continued to weaken, the need to pace herself and adapt to a reduced level of strength whilst adapting to her changing medical needs had become a necessity. This adaptation to a continued health decline and the increased requirements to attend medical appointments also impacted on her wider family: "*It is becoming the new normal for us"* (Elizabeth's mother); as they also adjusted to seeing Elizabeth in a new perspective with an increasing level of dependence that they had previously not known, such as "*Frequent hospital trips and admissions. I have to remember to plug my chair in to charge otherwise I won't be able to go out".*

The complexities for Elizabeth of having changed physical health needs impacted significantly on her life in general. Despite these changes, Elizabeth showed determination to overcome her increasing physical challenges and to continue to be as independently mobile as possible:

If my muscles are tired or weak that day, it's harder to get to my chair but I dislike using my chair inside as I want to walk as much as I can for as long as I can.

Elizabeth illustrated the complexities of her daily life as her health had continued to deteriorate in early adulthood:

I have a catheter to take care of, have to make sure I don't fall asleep in certain positions, daily medication which I have to take three times a day or more

depending on the issue. Frequent hospital trips and admissions. You get used to it. I have to remember to remember to plug my chair in to charge otherwise I won't be able to get out etc.

These excerpts show how different her life was from her peers. She had multiple facets to juggle and fit in with her busy life as a university student.

However, in spite of these challenges, Elizabeth demonstrated a positive acceptance of the benefits that having a wheelchair available to her when needed; whilst at the same time striving to maintain her ability to walk when able to do so. She was able to appreciate the benefits and the sense of freedom that the use of the wheelchair had provided for her:

I love trying new things: wheelchair basketball etc. With wheelchair basketball I love the flow of the wheels and how easy it is to move. I'm not a great team player due to my deafness but it's nice to have that social outing and also for others to try things like that.

4.7.4 Socialising, having friendships and family.

Work presented by Rossetti et al (2018) exploring the importance of friendship for disabled and non-disabled students, confirmed from their work that friendship promotes the identity of being accepted as part of a group and provides a benefit to the emotional and psychological wellbeing for the young person. In adopting this attitude to her changed circumstances, Elizabeth demonstrated her determination to seek out the positive aspects of her life and was seizing new opportunities to become part of a different social group and to meet new people as new opportunities became available to her:

New things haven't got around to too much apart from wheelchair basketball which is really nice. Although I don't want to be normal, these give me an opportunity to be just like others and do stuff people of my age would do – my chance to have a life.

Elizabeth had grown up in a rural setting and she had struggled with the move to the city:

Living in a city it is hard to be outdoors in the greenery but it's better than nothing. I prefer being outside.

Whilst acknowledging the challenges of her transition to living in anew urban environment, here Elizabeth was able to focus on the positive aspects of seeking outside spaces which were more comfortable for her; and she was able to recognise the importance of being able to see plants and trees which enhanced her emotional and mental health:

> *I like just being with my friends no matter what we do, I just like seeing them. My family is the same, I just like seeing them and being with them and knowing they are there.*

Elizabeth's friends and family provided a constancy and stability for her at a time when she was changing emotionally and psychologically as she became an adult and as she juggled her undergraduate experiences with the uncertainty of her changing health status. Quality of life measurements are key to understanding emotional and psychological wellbeing. A systematic review by Travlos et al (2017) explored this issue in relation to young people with neuromuscular impairments who were wheelchair users. The researchers acknowledged that advances in medical science are improving symptom management, such as the non-invasive night ventilation support that Elizabeth was waiting for; and improving the quality of life for young people. The data presented identified the importance for young people of *"living well in the present"* (Travlos et al 2017) as identified by Elizabeth.

4.7.5 Making career choices.

Elizabeth was aspirational for her future and had had some volunteering experiences *"working with children with special needs and disabilities"* which had helped to shape her ideas and plans for the years ahead; and which had enabled her to formulate a clear ambition for her career.

However, she was also trying out other ideas and alternative career possibilities; although her decision-making processes were coloured by the realisation that her health was fragile and labile which made planning for the future more challenging than for her peers:

> *But then I want animals too – so I'm not really sure. I would like to work in healthcare stuff too so that I can change things for others or at least improve others 'experiences. But since becoming iller; after university I'll have to see where and how I am before anything else. I doubt it will be easy to find a good employer who will understand.*

The development and the barriers to career aspirations for young people with complex levels of disability was explored in work by Rojewski et al (2012). Their work identified the importance of gender in career aspirations within this client group and identified that female students are more likely to have high career aspirations, in comparison to their male colleagues. Elizabeth, despite the deteriorating nature of her neuromuscular impairment, remained determined, focused, and aspirational for her future.

4.7.6 Transition to adult services

As identified in the literature, eventually each young person will need to make the transition to adult services. Unfortunately, Elizabeth's experience of her transition to adult services had not been positive:

> *Although I have already moved basically over to adult services, I feel like everyone kept going on about transition services and transition help, but it all seems to be words on paper and not a real thing. Both with audiology, CAMHS, etc. I feel there is a difference a very big difference. Adult services I'm not seen as a person I am an object or just another "special needs case", right care and choice don't apply and generally I don't feel you matter. Children's services were more relaxed in terms of environment and didn't treat you like you were thick if you didn't understand something, felt more like a person and as though since you were classed as a kid people actually wanted to help you.*

Here Elizabeth had moved through the stage of *"anticipated transition"* but had found herself in an unanticipated situation, where she was still navigating her individual position and identify in each of the separate services that she had moved to. As a young adult receiving care in adult services, she needed to become familiar with the expectations of new services for her to be more autonomous in her decisions about her care. Unfortunately, she had not received the planning, preparation and support which could have smoothed the path of transition for her, as she developed confidence in new health staff and new services.

The expectation from clinicians are that young people will continue to develop their abilities to take increasingly responsibility for their health care needs. However, young people may struggle to comply with a complex treatment regime and to adjust their behaviours and choices accordingly to accommodate the needs of their medical condition, such as their consumption of alcohol and the associated interaction with pharmacological interventions which may adversely affect the efficacy of their drugs and have a significantly detrimental effect on their health (Watson 2000). Elizabeth had experienced a continued deterioration in her physical health and had become increasingly reliant on technology and the support of carers to maintain her health and daily routines. She needed ongoing support to adjust to the transition to receiving care in different adult services (Soanes et al 2019, Soanes et al 2019).

4.7.7 Summary

Elizabeth presented as a determined young woman, who despite living with multiple comorbidities and fragile health; was resolute in aiming to achieve her own personal goals despite the challenges of her deteriorating health. Her story highlighted her resilience with in facing multiple challenges with her health but how, with support, was enabled to move towards meeting her career aspirations. Identity theory enabled an understanding of the different stages that Elizabeth had gone through as she moved from identifying herself as an active young person who enjoyed sports and walked unaided, to developing progressive muscle weakness which caused her initially to use a stick to support her with walking, to using a wheelchair.

4.8 Lauren

4.8.1 Introduction

Lauren was a 21-year-old woman, the eldest of five children. She had been born with Congenital Myotonic Muscular Dystrophy,[14] severe scoliosis, and restrictive lung disease. These conditions resulted in her being wheelchair dependent using a powered wheelchair. Lauren was fed enterally via a gastrostomy tube (PEG).[15] Despite the challenges of her limited mobility and deteriorating progressive degenerative neuromuscular condition, Lauren demonstrated high levels of determination and perseverance to achieve the goals that she had set for herself. She lived at home with the support of her parents and carers and she attended a local university as an undergraduate student. She was in her first year studying for a BA Hons in Counselling, Coaching and Mentoring.

Although able to communicate verbally; Lauren did not live locally and therefore she chose to participate in the research study by completing the questionnaire and returning this electronically via email. We also communicated via text messages. Lauren presented as a bright and articulate young woman, who had been identified as an Ambassador for the local

[14] An estimated 9,500 people in the UK have a form of myotonic dystrophy (Muscular Dystrophy UK 2017). The congenital form of Myotonic Muscular Dystrophy is the most severe form of this condition and is diagnosed in the early weeks or months after a child's birth; less severe forms of the condition may not be diagnosed until later in life.

[15] A PEG is a feeding tube which is placed through the abdominal wall and situated in the stomach, to enable nutrition and fluids to be delivered into the stomach, when an individual is unable to meet their own nutritional needs orally due to an unstable suck or swallow resulting from impaired muscle function.

children's hospice, where she was well known as she had been a service user of the hospice for respite care for many years since her early childhood:

> *Lauren was one of our group of young people who are between 16-25 years who we take things up with and talk about the services that we provide, and how they can be improved and whether we are providing a service that is useful for them.* (Sophie – a Transitions Coordinator at a children's hospice).

4.8.2 Living with progressive deterioration

As a younger child, before the progression of the Congenital Myotonic Muscular Dystrophy, Lauren had been more mobile and had had greater muscle strength which enabled her to participate in a wider range of childhood activities: "*When I was younger, I found it a lot easier to sit up for longer periods of time".* However, the trajectory of increased muscle weakness had adversely impacted on her life: "*My disability affects my daily life in every way you can think of".* The progression of her muscle weakness had led to Lauren being totally dependent on her family or on carers to meet all her needs, as she was no longer able to feed or dress herself, or to meet her own hygiene needs. Her progressive muscular deterioration had resulted in Lauren's inability to be able to sit up for long periods and she had become wheelchair dependent. Of most concern was the impact of Lauren's weakened muscles on her respiratory function; with the increased risk of her developing of respiratory infections and ultimately respiratory failure (Arens et al 2010). The impact that the progressive degenerative nature of her disease and corresponding weakness of her muscular strength had been life-changing:

> *My CMMD and scoliosis means that I struggle to sit up for longer than a couple of hours so finding places to lay down when away from the house is sometimes difficult.*

The complexities of living as a wheelchair dependent, undergraduate student who needed to attend lectures, seminars and student activities was apparent in her narrative. Lauren identified an ongoing concern and major barrier in terms of her daily activities as being: "*accessibility of places/transport for my wheelchair".* The importance of access and mobility; was only part of the daily issues for Lauren; she also had to deal with: "*breathing impairment gives me headaches in the best case and hospitalisation in the worst".*

For the treatment of her restrictive lung disease, Lauren was dependent on nebulised medication every four hours which had to be planned into the itinerary of each day; which she described as: "*problematic".* Lauren also needed to have privacy for her nebuliser to be

set up and for the administration of the drugs needed to treat her respiratory disease. Although nebulisers can run off a battery; more usually the machine is connected to a mains electricity supply: "*I have recently had to buy my own battery-operated nebuliser to have my medication on the go"*.

Reflecting on the changes in her physical deterioration and the realisation of her increasing dependence on others was difficult: *"I was also "younger" so people were more able to/allowed to lift me etc".* Consequently, Lauren was totally dependent on others to assist her to change position or transfer herself from her wheelchair to her bed or to the floor to enable her limbs to stretch out and to ease discomfort:

> *It is especially hard as there is no means of hoisting in public areas so, in order to lay down, I must rely on my parents to do so as carers are not allowed to lift me. I have to plan outings in advance to ensure I have people available to support me which means I occasionally miss out.*

4.8.3 The importance of family and friends

Lauren's family worked together with her and health professionals to develop solutions to some of the challenges and developed solutions that enabled Lauren to participate in life and in the opportunities presented to her:

> *Most of the time I use my adapted vehicle (bus) to get from A-B as I can easily get my wheelchair in and out. My dad and Uncle have built me a bed which is bolted to the floor to accommodate my laying down meaning we can take longer journeys. I also have a collapsible bed which I take to certain places like the cinema*.

The challenges of being able to enjoy socialising and outings with friends and family are identified. Lauren's physical weakness made sitting up for long periods uncomfortable for her and her father and her uncle had worked with her to find a practical and comfortable solution to meet her needs. These adaptations enabled Lauren to go out and to enjoy the activities that others of her age would take for granted:

> *Meeting friends at the park is beach is fun! Enjoying a chat over coffee or a drink down the local pub is always a welcome suggestion too! I love family holidays and spending time together generally as a family. We watch films together, have music sessions and play games together.*

The limitations of Lauren's physical abilities to sit up for long periods caused her to adapt to her deteriorating physical condition. She was aware that she needed to adapt her socialising in person in relation to her physical limitations. With the wide range of internet opportunities

to connect with other people across the world, to reduce loneliness and to maintain social and emotional contact with others, Lauren had become an avid and proficient user of social media which she was able to do whilst lying down:

> *Sometimes my condition means that it is sometimes difficult to see people in person and social media is a happy medium so that I can still keep in contact with the people I care about.*

The importance of social media for young people with additional health needs for maintaining friendships and providing opportunities for the development of new interactions with others is well recognised (Näslund et al 2013, Hemsley et al 2015, Hynan et al 2015). Social media provides opportunities for young people to join social groups and forums and to interact with other young people in similar circumstances to themselves. This provides opportunities for information exchanges, support, and friendships to become established, however this relies on the technology and infrastructure being available and updated to meet the needs of the fast-changing world of information technology.

Lauren and her family had carried out modifications to augment her ability to engage in hobbies that had become impossible for her to do alone, and thus enabled her to gain a sense of achievement and fulfilment and which enhanced her mental health and wellbeing:

> *I love photography, though it's not easy for me; I cannot take photos myself. My family and friends and I now have a system where they take video footage as opposed to photos, then I capture stills on my laptop from the film - it's not quite the same but it does feel more as though I am taking the photos myself!*

Being able to join in with the important normal social activities of eating and meeting with family and friends in a social situation were important; however, for a young person wanting to fit in with their peer group, having an enteral feeding tube was another visible sign of her being different to her peers and yet was an essential aspect of her daily life that her friends did not have to consider for themselves as they were able to meet their own care needs:

> *My feeding tube[16] sometimes leaks which ruins clothing and can be embarrassing in public. I cannot do any personal care for myself so relying on carers to be available or on time can be frustrating (the same applies for attending Uni).*

[16] A feeding tube might be a nasogastric tube which is a fine tube inserted into the nose and passed down into the stomach which need to be changed frequently; or a gastrostomy or jejunotomy which are placed into the stomach or small bowel following a surgical procedure.

Lauren's identity had become that of a young person who was reliant and dependent on others to meet her needs. She was aware that receiving enteral feeds in public was different from her peers, but she needed to have confidence in her carers to be able to manage her feeding tube and support her, if her feeding tube leaked in public which she had previously found to be embarrassing.

Body image for adolescents is an important and often underestimated issue which is closely linked with self-esteem and self-acceptance for young people. Any obvious variations from what are perceived to the "norm" can lead to bullying, rejection or becoming socially isolated from friendship groups and peers (Pinquart 2012). For young people with progressive degenerative conditions, their changing body shape, and physical abilities due to the progressive weakening of their muscles, gradually affects all the muscles in their bodies resulting in increasing difficulties in speaking and swallowing their own saliva, resulting in increasing difficulties in communication. Lauren was reliant on others to intervene and support her with her body image issues which were related to her enteral feeding.

4.8.4 Hopes and dreams for the future

Lauren has clearly defined goals and career ambitions for her life:

> *I would like to become a counsellor for other young individuals with similar disabilities. I would also like to live independently, by which I mean my own house with live-in carers. I hope to be able to become a qualified counsellor and practice from my own office at home for ease.*

In common with many other young people, Lauren also highlighted her desire to have a family of her own in the future: "*Like any other girl my age, I want a family someday too as well as a disability dog"*. For many young people with a diagnosis of a progressive life limiting disease, planning for an uncertain future can be difficult. However, advances in technology have led to an increase in the predicted life span for some young people, which raises the importance of appropriate and adequate funding and support to enable young people to enter the work force or pursue further or higher education opportunities to enable them to achieve their individual goals (Gibson et al 2007):

> *My main goal in life is something a lot of people take for granted – to have my own house. Not shared accommodation or residential with communal areas and on call staff but my own place with 24/7 live-in carers. On call staff would not meet my needs. I want somewhere I can decorate myself, furnish myself, and call*

mine, somewhere I can call home. The dream would be to have my own build to fully accommodate my needs!!

4.8.5 Experiences of transition to adult services

Lauren's feelings about her transitions experiences were "*Not the best. The closest "appropriate" respite service is 3 hours away!! I hardly see any of my health professionals as much as I used to, funding cuts left, right and centre."* Through the lens of identity theory, it became clear that Lauren had always identified herself as being disabled, but as her muscle strength had deteriorated over the years, she was able to recount how her ability to sit up in her wheelchair for long periods had changed and her identity became one of a young person who was totally dependent on others to meet all of her needs.

4.8.6 Summary

This case study has demonstrated Lauren's perseverance and determination as a young person living with a life limiting progressive degenerative neuromuscular condition. Lauren had worked hard to achieve her ambitions in pursuing her education, career, and personal life goals despite the complexities of her medical condition and the progressive nature of her neuromuscular impairment. Her close-knit family and friends provided essential daily support for Lauren to enable her to attend university and live her life to the full, meeting friends, enjoying hobbies and activities, and relaxing with friends and family.

4.9 Nathanial

4.9.1 Introduction

Nathanial, aged 21 years, was living at his parents' home. He had been born with Duchenne Muscular Dystrophy (DMD), was wheelchair dependent and used a powered wheelchair. He was reliant on his parents and carers to meet his physical, hygiene and nutritional needs.

As a young child, his development had initially been normal, and he had achieved his fine and gross motor skills at the expected times. However, Nathanial recalled the changes that he experienced as he went through his early childhood years:

Up until about six I was fairly normal running about. I would fall over a bit and I'd bang my head quite a few times.

Cognitively he was unimpaired and had successfully completed a degree at university:

> *Nathaniel needs everything doing for him and I think people forget that because he can have an intelligent conversation and when the weather is fine he will go out in the village and people think that he is reasonably independent.*

Nathanial's mother was acutely aware of being the mother of a child with a life limiting illness and of needing to value the times that they were able to spend together:

> *I sort of look at things and think if I don't have length of time with Nathanial, I will have depth of experience with him. He has a sense that he hasn't got a long life.*

In common with all men with Duchenne Muscular Dystrophy, although his body had become physically frailer with significantly reduced muscle tone, Nathanial's mind was unaffected, and he presented as a bright, articulate young man who had graduated from university; and was aspirational for his future. However, the gradual erosion of his muscle strength had rendered Nathanial dependent on others to meet all of the fine and gross motor tasks that he had previously been able to do for himself: "*Mum do you think you could turn the page for me?"*

4.9.2 Dealing with progressive physical disability.

As a young man who had been a talented artist, the progression of his disease with the associated decline in muscle function had caused Nathanial considerable distress as he readjusted to the reduction in fine motor skills in his hands:

> *My arm just wants to go off and do a painting. Just being able to go and create something with your hands is something that most people should aspire to. I get down and have a cry and get it out of my system it does actually help quite a bit I think to cry.*

One of the many challenges for Nathanial had had in adjusting to the changes in his body, were the adaptations that he had made by learning to hold a paint brush with his mouth and thereby to continue to express his artist talent. However, he needed the support of others to be able to access the paints he needed and was reliant on others to be available to help him when he wanted to paint; which reduced the spontaneity of being able to paint when he wanted to, which he had previously enjoyed.

The impact on the self-identity of the young adults manifested in several ways and included: dealing with pain, loss of physical strength and independence, and dealing with stigma. The body of literature concerning young men living with Duchenne Muscular Dystrophy has

explored the physical, emotional, and psychological implications of living with this condition and acknowledges the difficulties for young men of living with pain and increasing disability (Abbott et al 2015, Pangalila et al 2015). Young people living with Muscular Dystrophy identified the emotional and psychological effects of living with stigma as a young adult, which had as much of an impact as coping with the deterioration of their physical strength (Setchell et al 2017) and correlates with the experiences that Nathanial described of standing out as being different at university and encountering difficulties with making friends. Young men living with Duchenne Muscular Dystrophy have grown up with the identity of being unlikely to live into adulthood, but more recently due to significant advances in medical science, they can now look forward to a period of lifespan which had previously been unavailable to them. With this comes the need for young men to develop a new sense of their own identity and their place in the world which includes forming relationships and becoming sexually active (Gibson et al 2014, Zamani et al 2016, Abbott et al 2019). Nathanial had graduated from university and was living at home with his parents. He was adjusting to the changes in his physical health and was making plans for his future.

4.9.3 The importance of social relationships and having 'normal' experiences.

The emotional and psychological aspects of life for a young person of growing up experiencing increasing dependency on others, rather than gaining independence can be difficult for the young person and for their parents; as they face the future with a life limiting progressive disorder (Hodges et al 2010, Landeldt et al 2018). A major issue for Nathanial was his desire to develop his friendship group and increase his social circle and to gain confidence with talking to women:

> *I've not had much luck talking to people - especially girls. Even if you are just talking to them saying hello and passing the time of day, they are a bit off hand with you - it's very disheartening. I think maybe I'll try to find another route to try and solve the problem for me.*

The development of relationships with others can be difficult at a time in life when all young people in their late teens and early 20's are establishing their identity and sexuality (Christie et al 2005, Gibson et al 2014, Kelly et al 2016). As the life span for young men with Duchenne Muscular Dystrophy continues to lengthen due to advances in medical science, a new generation of adolescents are becoming men and need to go through the processes of transition to adulthood in all areas of their lives and can look forward to an extended

longevity to their life span than was previously expected for them (Viana et al 2007, Gibson et al 2014).

The importance of social interaction and intimacy with others and peer support are crucial for all young people for improving their self-esteem, confidence, and sense of belonging within a social group or community. However, for young men with DMD, their reliance on others for transport and for meeting physical and emotional needs may be a barrier for them being able to participate in the social activities of their friendship group, which may lead to a negative effect on their emotional and psychological health and wellbeing (Gibson et al 2007). When reflecting on the physical aspects of being able to have sexual relationships, Nathanial was honest and realistic; *"I want things to happen now whilst I am able to feel like normal and can enjoy it".*

In seeking a female partner, Nathanial was candid as he considered what attributes he was seeking in a potential sexual partner; "*I'm not expecting a Miss World or anything – but it's about being normal really. I want to have the experiences that other people do".* The drive for Nathanial to have the same experiences as his friends in having an intimate relationship with a girl, is a normal part of the expected development of young people of his age and is one of the "tasks of adolescence" identified by Viner et al (2005).

The desire to have a first sexual encounter had become the predominant issue for Nathanial and he was willing to push the boundaries and explore the possibilities available to him to make this desire a reality, which included considering using a sex worker who had experience of working with disabled clients to enable Nathaniel to have his first sexual experience. Here Nathanial presented a clear example of his determination and ability to overcome the obstacles that had arisen to achieve the goal that he had set for himself:

> *It's that word isn't it – prostitute - which seems very derogatory, but yeah, I know it's not what most 21-year-olds would be doing. I want my first experience not be something I regret or feel I can't talk about or feel guilty you know…*

Whilst Nathanial acknowledged that his proposed solution to meet his sexual need might be different from other young men at his age, it had not been a rushed decision and Nathanial had taken time to research the options available to him before reaching the decision to proceed with contacting a sex worker who had had experience with working with clients with

physical disabilities. Nathanial had consulted his mother concerning his plans of which she was totally supportive:

> *Like all children he has to grow up and do his own thing, but it is quite difficult. I hope that he won't be upset or disappointed. I think with all fantasies that the reality is often very different from what one imagines. Sometimes it is better to hold onto the fantasy than live with the reality.*

However, in seeking to support Nathanial, his mother agreed that she would be willing for a sex worker to visit their home, but she chose to be away from the house at that time.

4.9.4 Dealing with stigma.

> *But the main thing that people need to understand is that disabled people aren't different or a different type of person. It's not that I didn't try at University and stuff. I think that there should be more than this option because it's not just that you shouldn't or that you're not allowed to do this, or you can sit in silence and feel like a freak.*

Nathanial's comment of *"feel like a freak"* demonstrated that for some young people, their feelings and experiences of stigma can be self-stigma where the young person views themselves as being different because of their experiences from others (Corrigan et al 2002). Nathanial had the confidence to articulate the issues of sexuality that were of increasing concern to him at that stage in his life, as he was aware of his peer group being sexually active and he felt left out and different from his friends.

The emotional aspects of growing up with a progressive degenerative condition are as important as addressing the physical manifestations of the muscle decline that young people experience as they grow up through adolescence; but may be overlooked or minimised by health professionals (Setchell et al 2018). The side effects of the treatments that young people may be prescribed such as the use of steroids can delay the onset of puberty and cause mood swings. Young people with muscular dystrophy are often smaller than their peers, which can adversely affect their self-esteem and make them feel even more different from their friends. Stigma from others can add to the emotional burden of growing up being different, whereas as Nathanial identified, young people with physical disabilities want to be accepted and treated the same as their peers.

Through identity theory, it became clear that in his early childhood years, Nathanial had identified as a child who was able to walk independently and play with his friends, to becoming a child who fell over frequently and became progressively weaker, until he became reliant on a powered wheelchair for his mobility and his independence. However, he was a talented artist, and used his mouth to hold each paintbrush once he was no longer able to use his hands to control the fine motor skills needed to paint. His own views of his own identity were enhanced by his artistic talent, and some of his paintings were sold at a charity event to raise money for the children's hospice that he had been attending for many years for respite care.

4.9.5 Summary

This case study has raised the important issue for Nathanial of his sexuality and sexual needs as a young person with a progressive degenerative neuromuscular condition and other physical disabilities. The life span for young people with different types of Muscular Dystrophy are increasing and young people can now make plans for the increase in their life expectancy, which for Nathanial included being able to be sexually active and being able to enjoy the intimacy of a sexual relationship. Nathanial's mother was aware of the reduced life span that her son was living with and was keen to support him in living his life to the fullest, which included him exploring different options for sexual intimacy.

In supporting this group of young men, health professionals need to be sensitive to the holistic needs of all young people including the psychological wellbeing for young people with physical disabilities, which includes information regarding their sexual health.

4.10 Cross-case analysis

In the following section of this chapter, I present a cross-case analysis of the eight cases. As discussed in the methods chapter, there are few guidelines for conducting cross-case analysis when using case study methodology. The cases have been presented in a specific order, to enable the reader to gain an understanding of each participants' story, starting with the antenatal and birth stories, and moving onto accounts from three mothers of their children's early childhood and then their primary school years. This was followed by two pivotal cases, those of Melanie and Edward and Kim and Suzie (see Figure 1), as they represented both the experiences of the mother and the young adult within case and enabled the reader to gain both

perspectives. The final three cases were the stories from three 21-year-olds of growing up and negotiating adulthood with a progressive degenerative neuromuscular condition. Three significant themes arose from the cross-case analysis. These were pushing back and pulling away during transitioning; becoming a mother of a disabled child and an expert parent; and holding onto the child and mutual dependency.

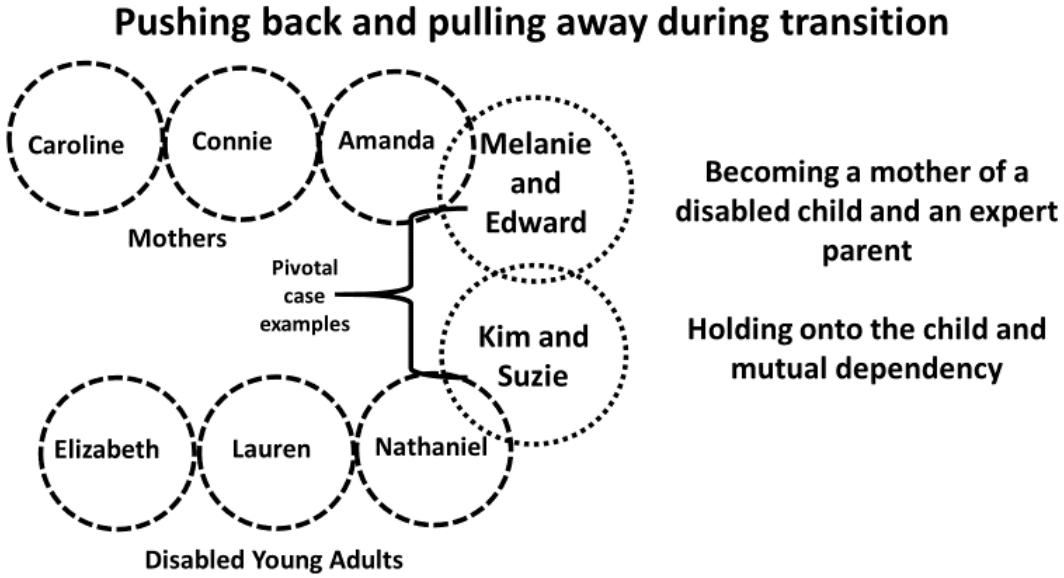

Figure 2: Pushing back and pulling away during transition.

4.10.1 Pushing back and pulling away during transitioning.

For two of the three parents (Connie, and Amanda) of the younger children with degenerative neuromuscular conditions, the early indications of being torn between wanting their child to become independent in the future and at the same time holding on to them because of anxieties about safety were always present. For example:

> *So, it would be really nice if he could be involved in something as an adult that involves routine whereby, he could contribute in some way. In an ideal world that would be amazing and somewhere to live where he could be safe and ok. I mean that until I know that there would be anywhere that I am 100% happy with – that he's not going anywhere (Amanda-Orlando's mother).*

> *Andrew is slipping further and further behind his age range really although he is still making progress which of course I am thankful for – but I do suspect that he will need lifelong learning and obviously me and my husband have had to get our heads round that. (Connie - Andrew's mother).*

Both mothers had begun to look ahead to the future and had identified the specific needs that both their sons would have and were apprehensive as to how their children's specialist needs would be met.

Two of the young adults (Elizabeth and Lauren) identified their wishes for independence whilst also acknowledging their on-going needs for support to meet their specific physical requirements:

> *My main goal in life is actually probably something a lot of people take for granted - to have my own house. Not shared accommodation or residential with communal areas and on call staff but my own place with 24/7 live-in carers. On call staff would not meet my needs and, truthfully, I want somewhere I can decorate myself, furnish myself. I want somewhere I can call mine, somewhere I can call home (Lauren).*

> *Thankfully only recently I have been lucky and managed to get social care support as before it was my mum looking after me. They help me make food and learn recipes, help me take my bins and laundry and also just getting out and about. I guess it's reassuring for my mum to know someone is checking up on me now and again and it's nice for me to know someone is coming even if I feel bad for sitting whilst they do stuff (Elizabeth).*

These plans are part of the normal tasks of adolescence identified by Viner and Christie (2006), where neurotypical young people should achieve independence from their parents as an essential part of their journey from childhood to adulthood. However, these young people has the same desires as their peers, but needed additional and continued support to achieve their aspirations.

The two pivotal cases demonstrated a pattern of pushing back and pulling away that was occurring during the stage of transition for both young adults (Edward and Suzie) in these cases. The young people strove to gain independence from their parents, *"I'm looking forward to getting away from my parents – yeah"*. Suzie felt that her parents *did* encourage her to be more independent and that when she was prepared to actually listen to this it helped to give her a sense of independence, and she also recognised herself as a difficult teenager – confirming that she was a normal teenager who was seeking to achieve the tasks of adolescence (Viner and Christie 2006, Kaplan 2003):

> *My parents just try to give me a bit of encouragement now and again but sometimes I just don't take it on board because as a teenager I find my parents incredibly annoying – so I don't tend to listen to them too much, but when I do, it actually does help and it's my dad has actually helped me to go the gym a lot*

more – that's to help me to build up my muscles a bit more and to get a bit stronger (Suzie).

Suzie's mother confirmed and recognised this rebellious aspect as part of normal teenager behaviour, her desire to move away, become independent and develop a career path for herself:

She wants to go to university, and she wants to get all of her GCSEs, but she gets frustrated with adults and adults giving her feedback that she doesn't want to hear (Kim – Suzie's mother).

Kim also recognised that the physical difficulties that Suzie had would impact on her future independence and achieving her personal goals; even though Suzie had identified the steps that she wanted to take to achieve the independence that her older sister had achieved, "…*she wants to live up to that and saw that her older sister left home to go to university at 18 and so of course Suzie wants to do that".* However, she acknowledged that this would not be easy for Suzie:

She does struggle academically because of her brain injury. She may have the entry requirements to do what she wants but it might take her longer, but she wants this to happen when she's 18 like every other young person, – and she might not be there yet - we are trying to – you know - she's not very happy that she's doing GCSEs split over 2 years.

In a similar way, Melanie, Edward's mother, was seeking to support her son's career aspirations whilst at the same time recognising that his aspiration for sporting success would not be easy. However, her determination to support him involved sourcing a new school where he would be supported to develop his skills for playing cricket and enable him to work towards his ambition of playing cricket professionally for the England disabled cricket squad.

"He's going to go to a beautiful new school and because of his cricket and their senior cricket person – and they are totally accepting of his disabilities and he's going to flexi board during the week which I think will be good for him".

Whilst for the mothers there was a need to remain close to their children even though they were transitioning to becoming young adults. The mothers were pulling their child back (from independence) due to fears of mistakes being made in care which could result in a deterioration of their child's health:

Because of Suzie's memory problems she does need help sometimes. She can arrive at an appointment and have forgotten what she was going to say even though she might have – she's written it down or she might have a conversation which is a perfectly legitimate conversation which might be way ahead in the future – epilepsy medication appointments – she's talked about how she's going

to manage it when she goes to university – but she's not there yet so she doesn't need to worry about how she's going to manage it yet. She gets stressed and she gets anxious about things that are quite far into the future – so we are letting go but we're still there – if that makes sense?" (Kim – Suzie's mother)

This concept of pushing back and pulling away attempts to show the complexity of the transitioning from adolescent to young adult for parents and children with neurodegenerative disorders.

Pushing back and pulling away occurred at several levels. For the young people with progressive degenerative disorders, as their peers were developing increasing levels of independence during their teenage years, they reported their increasing dependence on their parents, siblings, and caregivers due to the trajectory of their disease processes. Their experiences correlate with studies which have explored the lived experiences of young people with chronic illnesses and the ways in which young people adapt to a new level of normal in their lives where they become accustomed to living with an increased level of dependence on others (Taylor et al 2008, de Ridder et al 2008, Gudgeson et al 2015, Travlos, et al 2016).

The young adult pushing back and pulling away in the context of their parental dependency is a normal stage of adolescence, with young people seeking greater independence from their parents (Christie et al 2005). In the two pivotal cases, both mothers wanted to push away the young adult to become more independent but also pulled back, in addressing their own 'caring' needs. The mothers were aspirational for the future of their children, although uncertain as to how their hopes and dreams for them could be realised (Turnbull et al 2014). They were determined to maximise the potential for their child and showed great perseverance and tenacity in seeking the best clinicians and became the champion for their children. The mothers talked about "*fighting"* for their children and feeling the need to be *"brave"* to strive for the best interventions for their child.

4.10.2 Becoming a mother of a disabled child and an expert parent.

Identity theory provided a mechanism by which the mothers' perceptions of themselves, and the ways in which they adopted and adapted to their new roles altered while they were adjusting to their new positions within society, of being mothers of children with additional needs (Burke et al 2009, Polger 2009). Each of the mothers had experienced stigma, social isolation, and emotional distress (Green 2003) as a result of becoming identified at a 'mother

of a disabled child'. The need for mothers to devote time to a disabled child leading to resentment of the disabled child by siblings has been identified in the literature (Emerson and Giallo 2014, Begum et al 2011, Hartling et al 2010). For parents trying to divide their time between the children in their family; the complexities of caring for a child with additional health care needs and the time that is required to provide complex and time-consuming care for their child; often results in the unaffected child having less time with their parents than their affected sibling (Hartling et al 2010, Begum et al 2011).

The mothers' identities were also reshaped to that of, 'becoming both a caregiver and expert parent' in the specific condition of their child. Collectively the expertise required ranged over: cerebral palsy; autism, visual impairment, Duchenne Muscular Dystrophy, Congenital Myotonic Muscular Dystrophy, epilepsy, urinary incontinence, obsessive compulsive disorder, scoliosis, and restrictive lung disease. One of the 'new' identities that emerged for all the mothers in this study was that of becoming an "expert parent" (De Geeter et al 2002). As such they had developed a level of expertise and clinical competence that they had not envisaged. Their experiences resonate with the experiences identified in other research which has explored the parental experiences of their child's diagnosis being given to them (Rahi et al 2004, Wright 2008). The following excerpt is an example of the level of expertise the parents needed to achieve:

> *We have 5 Foleys at home and we can put Foleys in ourselves – we haven't let Edward do it because really the continence nurse said the timeframe - by the time you've been to the GP – been sent to the local hospital and then they decide that he has to go to A&E at the children's hospital, then they've got to find a surgeon to put the catheter in under Entonox or something – he will have gone further into retention. (Melanie Edward's mother).*

The mothers were also highly expert in their knowledge of the multiple, complex, and ever-changing medication regimes that their child required, demonstrated in the following extract:

> *He used to wear three Clonodine patches – he now wears seven – they're 200 micrograms each – so we went from 600 – we're now on 1400, one of those changes since we went back – so there have been significant changes (Caroline – mother of Lucas).*

Caroline did not have a medical background, but it was extremely important to her that she could make sense of the treatments that were being prescribed to manage her son's seizures, but also to enable her to be part of the decision-making processes. Connie, on the other hand, was a qualified nurse and had a wealth of experience of working in the NHS, but as a parent

she recognised the limitations of the local health care services and the potential impacts that different systems had had for her son's treatment. "*I've got a file of all Andrew's letters that I take to all appointments so if anybody says something or if something crops up that's involved another practitioner that they haven't got in their notes I've got a copy".*

4.10.3 Holding onto their child and mutual dependency.

The mothers were pulling their children back from reaching increased independence because of their need to still be in control of their child's daily medical care. Through necessity, the mothers had become the main caregiver and an expert parent for their child and had developed technical skills and knowledge to meet their child's complex needs. They expressed hope that their children would achieve some level of independence from them, whilst recognising that their children would continue to require support and care in the longer term:

> *He really likes routine – so it would be really nice if he could be involved in something as an adult that involves routine whereby, he could contribute in some way. In an ideal world that would be amazing and somewhere to live where he could be safe and ok. I mean that until I know that there would be anywhere that I am 100% happy with – that he's not going anywhere" (Amanda – mother of Orlando).*

> *I'm quite aware that because of the level of disability that Andrew has that the chances are that he's going to need lifelong input – I think that it is unlikely that he will ever live independently – somebody can prove me wrong, but I think that with his level of disability that would be the case. So, I think that it is probably unlikely that he will hold a job. I do suspect that he will need lifelong learning and obviously me and my husband have had to get our heads round that" (Connie – mother of Andrew).*

Both Amanda and Connie were looking ahead to their children's futures and anticipating the levels of support that would be needed for their sons in the longer term. They had identified the emotional adjustment that they as parents were in the process of working through, as they identified their roles in being involved with securing appropriate services to meet their children's needs as young adults.

In the two pivotal cases, Melanie was supporting Edward to become more independent from her, aiming at a gradual loosening of their interdependent relationship. However, due to complex bladder augmentation surgery Edward remain physically dependent on his mother for catheterisation. For Kim and Suzie, although Suzie craved greater independence from her

parents, her mother was concerned that Suzie's impaired short-term memory would prevent her from remembering to take her ant-epileptic medication, without which she was at significant risk of having further seizures. In accordance with expected development during adolescence, the young people were straining forwards and striving for further independence as they had seen their older siblings and friends successfully achieving (Christie et al 2005), whist the mothers were pulling their children backwards, because of fears that their children's medical needs could be met by them once they had left home.

Strong emotional bonds between children and their mothers are an expected and desired manifestation of secure attachment (Sheridan 2016). These strong emotional bonds provide nurturing and protection for the child during the childhood years and enable the child to explore the world around them and gradually develop independence as they move through their adolescent years (Kaplan 2003). However, when a child has a continuing or deteriorating health issue, the parents' knowledge, skills, and care for their child are heightened, and create a level of mutual dependency whereby the child is reliant on their parents for meeting their medical needs and for transportation to and from medical appointments and social activities in specially adapted cars which can accommodate their wheelchairs. These needs reinforce the parents' identities of being the parents of children with continuing health care issues, for whom being needed would continue throughout their child's life.

Each of the young people identified their parents as being an important source of practical and emotional support for them in their daily lives and in their recreational and social activities, whereas for non-disabled adolescents, their social interaction with their friends becomes gradually more independent from their parents as they approach adulthood:

> *Most of the time I use my adapted vehicle (bus) to get from A to B as I can easily get my wheelchair in and out; my Dad and Uncle have built me a bed which is bolted to the floor to accommodate my laying down needs meaning we can take longer journeys (Lauren).*

> *Thankfully only recently I have been lucky and managed to get social care support as before it was my mum looking after me. They help me make food and learn recipes, help me take my bins and laundry and also just getting out and about. I guess it's reassuring for my mum to know someone is checking up on me now and again and it's nice for me to know someone is coming even if I feel bad for sitting whilst they do stuff (Elizabeth).*

The mothers were acutely aware of the level of dependency that their child had on them for their health needs to be met, but this also served to reinforce the mother's identities and the vital roles that they played in providing for or training others to meet their children's continuing health needs. One of the mothers (Amanda) stated her future resolve to sacrifice her own career in order that her son's future needs could be assured, if suitable services which were acceptable to her could be provided. She highlighted the importance of the management of her son's life and how much she would be prepared to sacrifice to enable him to achieve his aspirations:

> *The worst possible case would be that he would be somewhere where he is just at home or just in a home or in a room and didn't go out and do stuff. I will do everything in my power to make sure that that doesn't happen. To the point that you even think that maybe if at some point you had to that maybe I would not work but I have never wanted to not work because I am too motivated – but you just think – well - you never lose that sense of responsibility I suppose – I would never – I would never walk away (Amanda – mother of Orlando).*

The adolescent years are known to be a period where biological, emotional, and psychological changes take place in the body and minds of all children as they grow up from childhood to adulthood (Christie et al 2005). During the adolescent years, young people are trying out different identities for themselves through their associations with different friendship groups, participation in different social and sporting activities and acceptance or rejection of religious or political beliefs (Christie et al 2005). Research into the lived experiences of young people with cerebral palsy (Kang et al 2010, Björquist et al 2014, Gilson et al 2014, Lindsay 2016, Rapp et al 2017) identify the physical factors of pain, reduced mobility, psychological problems, and parental anxiety for their child were all significant factors for teenagers growing up and seeking to establish their independence as young adults. All young people strive for independence and to changing their identity from being a child to becoming an adult. For example, Nathanial wanted to form an intimate relationship with a female partner but had had difficulties in achieving this personal goal. Identity theory provides a mechanism by which Nathanial could change his self-identity and those around him can form a new identity for him as a young man actively seeking to form a relationship with another person and to be able to be on a par with his friends. Furthermore, young men with Duchenne Muscular Dystrophy are not impotent and can be sexually active with support and can become fathers (Abbott et al 2019). However, for a young person with a chronic or life limiting illness, rather than developing greater independence from their

parents, they remain closely connected through the necessity of needing practical support to manage their health needs.

4.11 Chapter summary

In this chapter the eight cases have demonstrated the importance of the relationship between young people and their parents for emotional, psychological, and physical support in managing their daily lives. The young people demonstrated their motivation and aspirations for their futures but were realistic about the support that they would need to enable them to achieve their academic and career goals. The mothers had developed high level skills to manage the complexities of their children's medical care and had developed the expertise necessary to care for their children at home. Each of the mothers had gained boundless support from other parents also caring for children with continuing health care needs and valued the support that they also received from their families and close friends.

In the cross-case analysis conceptualisation of the pushing forwards and pulling backwards between parents and their disabled children as they move through their adolescent years and approach adulthood is presented. The levels of interdependency between mothers and their children have been identified, with recognition as to the difficulties for young people of following their yearned for paths to greater freedom from their parents. All parents carry with them hopes for their children's futures, but when a child has a continuing or deteriorating health trajectory, parents' emotional ties to their children remain strong as they too found it difficult to have the increased freedom from parenting duties which they may also have expected as they aged and approached retirement. The young people in this study remained reliant on their parents for physical and emotional support, whilst dreaming of having homes of their own which they could decorate to their own tastes and build their own nuclear families. The mothers wanted more independence for their children and were looking for solutions to help their children to move forwards but were themselves being held back by the level of responsibility that they held to ensure that their child's needs were being met to their maternal satisfaction and demonstrating a level of mutual dependency which both held them and repelled them.

In the next chapter I will consider how the findings of the study add to the existing knowledge surrounding young people and their families living with neuromuscular

impairment and make recommendations for policy and practice. I will discuss the strengths and the limitations of this research and will consider further areas of research in this subject area.

Chapter 5 Conclusion

5.1 Introduction

In this final chapter I address the contribution that this research makes to the field of mothers of children with neuromuscular disorders and young people living with neuromuscular impairments. I will provide recommendations for practice and policy and future research. In the chapter I will also make some personal reflections on my experiences of undertaking this research and present a dissemination plan. Limitations of the study will be addressed.

5.2. Contribution knowledge

The experiences of children and young people growing up with a neurological impairment are important, and in order for young people to be enabled to maximise their potential, their stories need to be told and the information disseminated to a wider audience of service commissioners and providers. This study is unusual in that the voices of both young people and mothers are represented in the same piece of work, with both participant groups identifying the needs for further support and services to be available to them for their adult years.

This work offers a considered and detailed account of the experiences of young people with neuromuscular disorders and their mothers, within the context of their wider systems of family, social contacts, educational experiences, and health care challenges. The cases were diverse in terms of their stage of transition and their personal experiences. Being the mother of a child with a neurological disorder impacted significantly on the identity of the mothers, once a diagnosis was established. Identity, as a disabled person, coloured the lives of the young adults, although they were all determined to live as normal a life as possible. The immediacy for young people concerned their short-and medium-term goals. Each young person was aspirational and ambitious for their futures with clear goals that they were striving to achieve. For each young person, their immediate family and close friends provided essential support for them, during a crucial time in their lives when they were striving towards their personal goals but were being hampered but undeterred by the physical constraints of their neuromuscular impairments. The mothers were each immensely supportive and protective of their children, but their focus was more concerned with the longer-term needs of their children and how these would continue to be met as adults. Each mother was determined that their child's potential would be maximised, their goals reached,

and that their children would not be held back by budgetary constraints or limitations in service provision, to the point of a willingness to forgo their own careers in order to ensure that their children's needs were met in the longer term.

The study has demonstrated the significant challenges experienced by the five mothers of becoming an expert parent in terms medical knowledge and practice skills, which they had not expected to need to develop, and a new medical language that they needed to learn in their journey to being the parent of a child with a neuromuscular condition. There were tensions identified between health professionals and parents in terms of their roles, in that being the '*expert parent'* the parents needed to work collaboratively with professionals, whilst at the same time having to demonstrate and prove their competency to undertake quite complex medical interventions to enable their children to remain at home.

The conceptualisation of young people pushing towards and yearning for greater independence whilst being constrained or pulled back by their parents, is a unique contribution to this area of research, and is worthy of future study. This study adds to the existing body of knowledge on this subject area of the experiences of young people growing up with a neuromuscular impairment, and the journeys that their mothers had taken in providing care for their children. This study offers both parents and young people's perspectives and has considered the similarities and the differences in the concerns faced by each and the levels of interdependency.

5.3 Methodological contribution

This research is unusual in that the eight case studies presented using case study methodology, vary between the first three cases where the mothers of younger sons with neuromuscular impairments tell their stories and were at an earlier stage in their parenting journeys, followed by the two pivotal cases where the mothers and their teenage children were participants and told their own stories individually and therefore were not influenced by each other's contributions and recollections, and lastly, three cases where young people over the age of eighteen years of age were the participants. In case study, cases are normally similar in terms of content of material and participants, for example teachers working in a secondary school or nurses working in a specific speciality which gives one dimension, however in this case study, having both young people and mothers, together with the professional endorsement of the experiences recounted from a transitions coordinator at a

children's hospice has provided a three dimensional view of the living with or supporting a young person with a neuromuscular impairment. It has been this three-dimensional view which has enabled the pushing forwards and pulling back during transition and towards gaining independence for these young adults to be revealed and presented and has highlighted the complexities and interdependencies between young people and their mothers.

5.4 Recommendations for practice and policy

The numbers of children and young people growing up with a neuromuscular impairment is continuing to rise and therefore the needs of this important group of young adults needs to be listened to and addressed by health and social care professionals. The following recommendations are made as a result of this study:

- Birth support for mothers needs to be available. All mothers need the opportunities to recount their birth stories to Midwives, Health Visitors and GPs, in addition to their own friends and family members, and in doing so, to be heard and to be given the opportunities to make sense of the outcomes for their children, especially when the pregnancy ends with an unexpected premature birth. It was evident that the experiences that the mothers had had during their pregnancies and the delivery and early weeks of their children's lives had had a very significant and long-lasting impact on them. Having the opportunities to revisit their pregnancy and birth experiences when needed for their own emotional and psychological wellbeing, would be beneficial for the mothers to know that this option was open for them.
- Mothers need support to prepare for their child moving to adult services and enable them to achieve their own and their child's hopes and dreams. This could be supported by each young person being allocated a transitions specialist health professional who could facilitate and co-ordinate the planning and preparation of their transition to adult services and could work with each young person and their parents to ensure that adequate preparation has been provided. Although there are Transitions Coordinators available in some services – this is not available to all young people. Respite care services for children and young adults need to be adequately funded and resourced, so that young people and their families can access services close to their homes and not have to travel long distances to be able to access the specialist care that they need. As this cohort of young people are now living into adulthood, the need for respite provision to meet the specialist needs of young people

with neuromuscular conditions will continue to increase and their specialist needs will need to be provided for and adequately funded.

- Whilst each of the young people focused on their medium-term goals for their lives, their mothers were extremely concerned about the funding to meet their children's long-term needs. Funding support is needed for the duration of the children's lives. This will have financial implications for the NHS, social care and for charities who provide high quality care for children with neuromuscular impairments. Funding for children's hospices need to robust and continued, in order that young adults can continue to be supported beyond the age of 18 years.
- Staff in adult services may have education and training needs to enable them to care for young adults with diseases which they may not have encountered previously. Adult services may have limited experiences of caring for young adults with progressive neuromuscular conditions and may need training, equipment and facilities to provide the essential care that young people will continue to need, which includes access to physiotherapy, hydrotherapy and occupational therapy services.

Both the parents and young people identified their needs for adequate planning and preparation to have taken place, before the transition to adult services in both health and social care took place. Their *"anticipated"* transition to adult care was often replaced by *"unanticipated"* events, with parents reporting that the care for their children was fragmented and poorly co-ordinated and they had lacked a single professional who could act as the co-ordinator for the communication between different health and social care teams.

The findings from this research could be beneficial to Commissioners during the planning of future of services to meet the requirements of young people with continuing health care needs. If current and future services are to have the flexibility to enable young people to make the transition to adult services at a developmental stage that is appropriate for them, as opposed to the current situation of transition taking place at a service driven age, this will be a major shift for some services, which will need to be adequately and appropriately resourced.

5.5 Recommendation for future research

The original aim of this study was to enable young people with verbal or non-verbal communication abilities, to be able to give their own perspectives and life stories via the use of their computers or electronic communication devices. The initiative for this study came from speaking with a 17-year-old young woman at a children's hospice who was unable to speak verbally but spelt out from an alphabet chart on her wheelchair *"let me tell my story – don't ask my mum".* However, as discussed, access to this groups were fraught with difficulties due to various approaches to 'gatekeeping' which could have been reduced if I had sought permission from specialist charities which have access to families across the UK. This could have enabled a larger number of participants to have been included in this study.

Future research involving fathers and siblings would enrich this field of study by exploring the paternal experiences of caring for children with progressive or non-progressive neuromuscular conditions. This project has identified the experiences and the needs of a small number of families. Although the findings from this study concur with previous studies, future research would enable a clearer picture to emerge of the complexities of the lives and experiences of young people and their parents, siblings, and other caregivers of growing up with a neurological impairment and could add to the current body of knowledge concerning this subject area; with the findings used to inform the planning, design, and implementation of new services for young people in this client group.

The need for the commissioning and provision of specialist services for children and adults with Muscular Dystrophy is recognised nationally; however, the provision of specialist clinicians, nurses, and therapists to meet the unique needs of this client group has meant that services have been centralised which results in long journeys for many families to access the specialist care needed (Muscular Dystrophy.UK 2016. A report from the National Confidential Enquiry into Patient Outcome and Death (NCEPOD) (2018) examined the quality of care received by children and young adults up to the age of 25 years with a chronic neurodisability. This report identified as one of the principal recommendations that the transition planning and coordination of young people should be a multi-agency approach which is led by a clinician with a clear plan for each young person provided and must include primary and secondary care providers.

Evidence suggests that across the country, the need for hospice provision for children and young people with life limiting illnesses is continuing to grow (Norman et al 2014, Fraser et al 2014, Jarvis et al 2018). The psychological and emotional needs for young people beyond the age of 18 years have been acknowledged, and it has been identified that many young people are not ready for the transition to adult services to take place at that age. Although legally adults; young people aged between 18-25 years do not fit comfortably within children's services, and therefore a redesign of services to meet their specific needs has been required. This re-evaluation of the needs of young people for continuity of care and support have led to some services being expanded to provide ongoing care for this client group when needed; and thus, to allow more time for the young person to acquire the skills and confidence needed before transferring to a new service (Care Quality Commission 2014). One current point of view is that transition to adult services for young people with continuing health care needs should be when the young person is ready for the move to adult services to take place, rather than being at a pre-defined age of 18 years for each young person (NICE 2016), which would allow for a developmentally appropriate approach to be implemented, however current service provision has pre-set thresholds for the ages that young people leave health and social care services and changes to this will need to be adequately funded and resourced.

The epidemiological evidence concerning the needs of young people with life limiting conditions in the UK has demonstrated that the numbers of children and young people requiring specialist services has continued to grow (Fraser et al 2014). Therefore, the funding streams to support children and young people with shortened life spans must continue to be a priority for health and social care providers. Health and social care services also need to train staff to have the specialist skills and knowledge needed to provide the high-quality care needed to support young people and their parents in inpatient and community settings both in children's services and adult health and social care.

Young people with neuromuscular conditions are important and deserve to have parity of opportunities to enable them to access further and higher education. Each of the young people aspired for their futures but were realistic that for these to be realised, health, social care and housing provision would need to be funded and available to meet their future needs. Parents wanted the reassurance that their children's future needs would be assessed, funded, and resourced, so that when they were no longer able to meet their children's physical needs, that

skilled and compassionate carers would continue to care for their children for the duration of their life span.

5.6 Dissemination plan

Several publications are planned to enable the voices and experiences of these participants to be heard and to add to international conversations within the academic and professional community of the needs of young people with neuromuscular conditions and the experiences of their parents and siblings. Potential journals include: Journal of Adolescent Health; Journal of Children and Young People's Nursing; and Child: health, care, and development. The possibility of writing a book for health care students is also being considered, to raise awareness of the needs of young people with neuromuscular impairments. Another potential avenue for raising awareness of the experiences of children growing up with a neuromuscular deficit, might be to write one or more than one children's book, to help younger children to understand the needs of their peers of needing the help of equipment such as a wheelchair, to live their lives to the full.

5.7 Limitations of the research

This study was limited due to the small number of participants within each case, although, using case study methodology enabled me to explore in depth the complex and multifactorial issues of a small number of participants living with a neuromuscular disease. Cases with a larger data source – such as sibling or father interviews would have enhanced the richness of the data. It would have been particularly useful to have had fathers as participants in the research and to gain the paternal perspective of the parenting journey of being the parent of a child with a neuromuscular impairment. The challenges encountered with recruiting participants to the study limited the diversity of potential participants to the study. With hindsight, I could have approached some of the national charities including Muscular Dystrophy UK and Scope, to advertise the study more widely nationally, which potentially could have resulted in more participants coming forward. This approach would have negated the necessity to use gatekeepers, as families could have made the decisions to participate in the research for themselves. This also offers the opportunities for further research, where collaboration with national charities could widen recruitment possibilities, which would enable more young people and their families to be included and to have their voices heard.

The data collection method of completing open-end qualitative questionnaires by young people could be achieved from any part of the country and would not have limited young people from across the UK from taking part. The interviews with parents could have been completed by telephone and audio recorded, which would have removed the necessity for parents to travel to meet with me. Using social media groups to advertise the study might have been one approach which would have negated the use of "gatekeepers" to access potential participants, however due to inexperience and concerns about the management of this recruitment approach, this was not used for this study.

5.8 Personal reflections on undertaking the research.

As an inexperienced researcher, I encountered many challenges including: recruitment of participants; delay in gathering data; grappling with consideration of different qualitative approaches to use in the early stages of the study; and becoming familiar with case study methodology and the complexity of this. Considering different qualitative research methodologies was interesting and informative and doing extensive reading around case study methodology helped me to make the decision that using case study was the most appropriate methodology for answering the questions posed for the study.

Returning to my propositions which were developed early in the study and focused on the transition of young people to adult services, I conclude that these statements have been upheld based on my study findings:

- *The voice of young people with neurological conditions is not well articulated in the literature.* This study can contribute to the worldwide conversation on the experiences of young people living with progressive and non-progressive neuromuscular conditions. As this emergent group of young adults continues to grow, health professionals need to know how services can meet the needs of this group of young people. My theoretical position was a recognition of the importance of young people with neurological impairments, with or without impaired verbal communication skills, and their mothers, to have the opportunity to speak their own stories and for health staff to learn from their experiences.
- *Young people need the opportunities to talk about their lives and tell their stories from their perspectives.* Only young people themselves can explain to health

professionals what their experiences and needs are, and how health and social care services can be adapted or developed to meet their needs.

- *Research literature on the experiences of young people with chronic and complex health needs indicated that young people and their parents are still experiencing a lack of support with the planning and preparation for their move to adult services.* This study has demonstrated that the complexity of transition for this group of people needs to be better considered in practice, policy and future research.
- *For young people who would previously have expected to have died during their adolescent years, the extension to their life span needs to be supported with facilities and services, to enable them to benefit from the additional months and years of life.* The numbers of young people who are growing up into adulthood with progressive neurodegenerative impairments are continuing to grow, and services including children's hospices need to adapt to meet this increase in expected numbers of adolescents and young adults for the future.

5.9 Conclusion

This study has made an original contribution in developing understanding of the maternal perspectives of raising a child with a neuromuscular impairment and how identity theory can be used to conceptualise these experiences. The study has contributed a wider understanding of the needs of parents in adjusting to the loss of their *"anticipated"* and *"unanticipated"* experiences during their pregnancies of having a normal child, and both the challenges and the positive experiences that the mothers reflected on, of being part of a community of mothers who provide ongoing intimate care for their children, in order to ensure that their children's health and emotional and psychological needs have been met. The voices of young adults growing up with a neuromuscular impairment have been sought and heard, with greater understanding gained of their hopes and aspirations for their academic, career and personal hopes of the future gained.

The conceptualisation of the level of pushing forwards and pulling back through transition, from independence and into adulthood has been helpful for understanding the depth and complexities of the relationships between young people and their parents but in particular with their mothers, each of whom had made sacrifices and adjustments to their own careers and aspirations to ensure that the complexities of their children's health and emotional needs

could be met. Further research making this aspect the focus of the child-parent relationship is warranted to understand how these needs could be met better in the future.

This study has identified some of the requirements of the parents and young people whose needs will have to be met throughout their lifespan. It has shown the levels of love and unwavering support that the parents have shown to their children and the parents' concerns for the funding and provision of services to meet their children's needs for the future. This research has enabled children and parents to tell their own stories in their own words and that in the future this information will be disseminated to a wider audience and can influence the planning of future services.

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Appendix A: Participant Information Sheet for parents / carers

Patient information sheet for parents / carers (version 3)

Study title: A study to explore the experiences of young people aged 16 – 21 years with neuromuscular disorders of their journeys of transition from childhood to adulthood.

Centre number: 13/LO/1553

IRAS project ID: 116662

Principal researcher: Laura Gilbert

Invitation and brief summary:

My name is Laura Gilbert and I am a part time Doctoral student from the School of Nursing and Midwifery at the University of Brighton. I am interested in understanding the experiences of young people with neuromuscular disorders as they move from childhood to adulthood.

What's involved?

I will be asking those young people who choose to be involved in the study questions concerning their hobbies, interests, their experiences at school / college or in the work-place and what their ambitions for their future are.

What would taking part involve?

I have devised an online set of questions that young people can complete and return to me either by email or by post in a stamped addressed envelope which will be provided by the researcher.

I will be asking young people questions on two occasions – one year a part – as I want to try and gain a better understanding of what changes for them in their lives over a period of twelve months.

I have chosen to use this method of obtaining information so that young people can complete the questions at a time that suits them, rather than having to travel or be available to meet with me at a time which might not be convenient for them; and that using computers will enable those young people with impaired verbal communication skills to use a medium that enables to communicate fluently for themselves.

For parents / carers who choose to participate, I would like to meet them either at their homes or at a hospital or children's hospice, to ask them some questions about their experiences of living with a young person with a neuromuscular disorder.

As part of the final document, anonymised quotations will be used to ensure that the views of young people and their parents / carers can be heard but they will not be able to be identified.

If your son / daughter do not wish to participate in the study but as the parent or carer of a young person you wish to participate, you would be most welcome to do so.

Your name or the name of the young person will have been provided by a Doctor, senior Nurse or Specialist Nurse only after they have spoken to you to explain this research study and then only if you or the young person has indicated that you wish to participate.

If you or the young person chooses to take part, participation in this study **is entirely voluntary and both the young people and the adults are free to withdraw from the research at any time without giving a reason.** Any decision to withdraw from the research **will not in any way affect the care that the young person will receive.**

What are the possible benefits to taking part?

The possible benefits of taking part is that it will give young people and their parents / carers the opportunity to "have a voice" and the use of computer technology provides the opportunity for young people with impaired verbal communication skills to have an equal opportunity to express their views and feelings of living with a neuromuscular disorder.

What are the possible disadvantages and risks of taking part?

Young people may not wish to be asked about their ambitions for their future however they have the options of either choosing not to answer any of the questions posed or to withdraw from the study without any consequences if they found any of the questions difficult or potentially distressing.

If you have any concerns about this study, you may contact Professor Julie Scholes by telephone on 01273 644078 or by email at j.scholes@brighton.ac.uk

Thank you for taking the time to read this information sheet.

Mrs Laura Gilbert

Appendix B: Patient information sheet for young people

University of Brighton

Patient information sheet for young people (version 3)

Study title: A study to explore the experiences of young people aged 16 – 21 years with neuromuscular disorders of their journeys of transition from childhood to adulthood.

Centre number: 13/LO/1553

IRAS project ID: 116662

Principal researcher: Laura Gilbert

Invitation and brief summary:

My name is Laura Gilbert and I am a part time Doctoral student from the School of Nursing and Midwifery at the University of Brighton. I am interested in understanding the experiences of young people with neuromuscular disorders as they move from childhood to adulthood.

What's involved?

I will be asking those of you who choose to be involved in the study questions concerning your hobbies, interests, your experiences at school / college or in the work place and what your ambitions for your future are.

What would taking part involve?

I have devised an online set of questions that you can complete and return to me either by email or by post in a stamped addressed envelope which will be provided by the researcher.

I will be asking each young person that chooses to be part of the study questions on two occasions – one year a part – as I want to try and gain a better understanding of what changes for you in your life over a period of twelve months.

I have chosen to use this method of obtaining information so that each young person who chooses to contribute to this study can complete the questions at a time that suits them, rather than having to travel or be available to meet with me at a time which might not be convenient for them; and using computers will enable those young people with impaired verbal communication skills to use a medium that enables to communicate fluently for themselves.

For parents / carers who choose to participate, I would like to meet them either at their homes or at a hospital or children's hospice, to ask them some questions about their experiences of living with a young person with a neuromuscular disorder.

As part of the final document, anonymised quotations will be used to ensure that the views of young people and their parents / carers can be heard but they will not be able to be identified.

If you do not wish to participate in the study but your parent or carer does wish to participate, they would be most welcome to do so.

Your name or the name of your parent / carer will have been provided by a Doctor, senior Nurse or Specialist Nurse only after they have spoken to you to explain this research study and then only if you or your parent / carer has indicated that you wish to participate.

If you or parent / carer choose to take part, participation in this study **is entirely voluntary and you and your parents / carers are free to withdraw from the research at any time without giving a reason.** Any decision to withdraw from the research **will not in any way affect the care that you are currently receiving or will be receiving in the future.**

What are the possible benefits to taking part?

The possible benefits of taking part are that it will give you and your parents / carers the opportunity to "have a voice" and the use of computer technology will enable you to express your views and feelings about iving with a neuromuscular disorder at a time that is suitable for you.

What are the possible disadvantages and risks of taking part?

You may not wish to be asked about your ambitions for their future however you have the options of either choosing not to answer any of the questions posed or to withdraw from the study without any consequences if you do not wish to answer any of the questions posed.

If you have any concerns about this study, you may contact Professor Julie Scholes by telephone on 01273 644078 or by email at j.scholes@brighton.ac.uk

Thank you for taking the time to read this information sheet.

Mrs Laura Gilbert

Appendix C: Consent form for parents

University of Brighton

Centre Number: 13 /LO/1553

Study Number: 116662

Researcher: Laura Gilbert

(Version 3)

CONSENT FORM FOR PARENTS OR CARERS TO PARTICIPATE IN THE STUDY

Title of Project: A study to explore the experiences of young people aged 16 – 21 years with neuromuscular disorders of their journeys from childhood to adulthood.

Name of Researcher: Laura Gilbert

Please initial all boxes

1. I confirm that I have read and understand the information sheet dated 4.12.13 (version 3) for the above study. I have had the opportunity to consider the information, ask questions and have had these answered satisfactorily. ☐

2. I understand that my participation is voluntary and that I am free to withdraw at any time without giving any reason, without the medical care or legal rights of the child / young person that I have parental responsibility for being affected. ☐

3. I agree to anonymised quotations from participants being used in the write up of the study ☐

4. I agree that the information provided can be shared with the three PhD supervisors who are overseeing this project: Dr Kay De Vries, Dr Chris Cocking & Dr Emily McWhirter ☐

5. Following completion of the study for this PhD, the thesis will be made available via the repository at the University of Brighton via the internet, as well as through the British Library. ☐

Name of Participant	Date	Signature

Name of Person	Date	Signature

Appendix D: Consent form for young people

University of Brighton

Centre Number: 13 /LO/1553

Study Number: 116662

CONSENT FORM FOR YOUNG PEOPLE TO PARTICIPATE IN THE STUDY

Title of Project: A study to explore the experiences of young people aged 16 – 21 years with neuromuscular disorders of their journeys from childhood to adulthood.

Name of Researcher: Laura Gilbert

Please initial all boxes

6. I confirm that I have read and understand the information sheet dated 4.12.13 (version 3) for the above study. I have had the opportunity to consider the information, ask questions and have had these answered satisfactorily. ☐

7. I understand that my participation is voluntary and that I am free to withdraw at any time without giving any reason, without my medical care or legal rights being affected. ☐

8. I agree to anonymise quotations from participants being used in the write up of the study. ☐

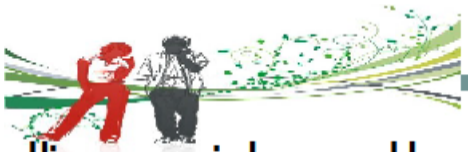
9. I agree that the information provided can be shared with the three PhD supervisors who are overseeing this project: Dr Kay De Vries, Dr Chris Cocking & Dr Emily McWhirter ☐

10. Following completion of the study for this PhD, the thesis will be made available via the repository at the University of Brighton via the internet, as well as through the British Library. ☐

Name of Participant	Date	Signature

Name of Person taking consent.	Date	Signature

Appendix E: Recruitment flyer as part of the recruitment process



Hi my name is Laura and I am a nurse / lecturer at the University of Brighton.

I'm doing a research project for my PhD and would really like your help.

Are you between 16—21 years old?

Do you have any condition that affects your neurological system ?

Would you be able to take part in some research to help me to understand a bit more about what it feels like to be a teenager and have a neurological condition?

Would you be able to answer some questions via email and then email them back to me and tell me about yourself? This would normally be on just one occasion and you can email back your answers within 4 weeks.

I'd really like to talk to parents and carers too, so if you think you might be able to help me; I would love to hear from you.

If you would like to be part of this study then please contact me by email .

Here is my email and address:

Mrs Laura Gilbert
University of Brighton
Westlain House
Village Way
Falmer
Brighton BN1 9PH
01273—644186
Email:
l.gilbert@brighton.ac.uk

May 2015 (Version 5)

Appendix F: email sent to the whole university as part of the recruitment process

Mon 15/02/2016 10:32

Laura Gilbert

Research study conerning the expereinces of young people with neuromuscular conditions.

To uni info

Message Leaflet for potential participants version 6.pdf (707 KB)

Do you know a young person aged 16 – 21 years with a neuromuscular condition?

I am undertaking a research project for my M.Phil / PhD for which I have ethical approval from the University of Brighton and the NHS Regional Ethics Committee.

This research study aims to explore the experiences of young people with neuromuscular conditions growing into adulthood and as part of that life journey, their experiences of transition from children's to adult services.

Participation in this study would involve completing an on line questionnaire and then emailing this back to me.

Please share the attached leaflet advertising the study with any young person with a neuromuscular condition or any personal contacts that you might have where this is appropriate.

If you know anyone who is interested in taking part in this study, or who would like any further information please can you ask them to contact Laura Gilbert at l.gilbert@brighton.ac.uk.

With many thanks
Laura Gilbert

Appendix G: Interview Schedule for parents/carers

Questions for the parents / carers in either face to face or telephone interviews: (These questions may change as the study continues)

1. Please can you tell me about yourself, your child and your family?
2. What medical condition / illness does your child have?
3. At what age was your child diagnosed?
4. What have been the challenges for your child and for your family since the diagnosis?
5. What hopes and dreams do you have for your child?
6. What hopes and dreams for their future does your child have?
7. What will you and your child need for these to be achievable?
8. What are your feelings about your child moving to adult services in the future?
9. If they have already moved to adult services – what has their and your experience of this been?
10. Is there anything else that you would like to tell me about your child and your family?
11. Is there anything you would like to ask me?

Appendix H: Questionnaire for young people:

Please answer these questions as fully as you can but you DO NOT have to answer any questions that you do not want to.

Please take your time and answer these questions when you feel well enough and able to do so.

Please email your answers back to me within one month.

Please give me as much information as you can for each of the questions.

If I need further clarification, I might need to contact you again by email which I hope will be acceptable for you.

1. 1. **Please can you tell me about yourself –** how old are you? Are you male or female?

2. What hobbies and interests do you have?

a) Where do you go to do these hobbies?
b) What do you most enjoy about these hobbies?
3. What do you enjoy doing with your friends / family?
a) What do you most enjoy about those things?

3.Do you attend school / college / university and / or do you have a job?

a) If you are at school, college or university what subjects are you studying at the moment? Are you studying full or part time?
b) If you have a job – where are you working at the moment and how many hours are you working each week?
c) What would you like to do in the future?

4. What medical condition / illness do you have?

5. Does your illness affect your daily life – if so how?

a) What helps with this?
b) Is there anything that makes things more difficult for you?

6. How has this changed whilst you have been growing up?

7. Who or what helps you in your daily life?

8. What are your ambitions for your future?

9. What will you need to help you to achieve these ambitions?

10. How do you feel about moving to adult services in the future?

a) What are you looking forward to about moving to another service?
b) If you have already moved to an adult service – what has your experience been?

11. **Is there anything else that you would like to tell me about yourself or to ask me?**

Thank you VERY much for taking part in this study.

Best wishes: Laura Gilbert: l.gilbert@brighton.ac.uk

Appendix I: Interview schedule for young people

Please can you tell me about yourself? How old are you?
What hobbies and interests do you have? Where do you go to do these hobbies? What do you enjoy doing with your friends and family? What do you most enjoy about these things?
Do you attend school / college/ university? Do you have a job? What subjects are you studying?
What medical conditions do you have?
How does your medical health affect your daily life? What helps with this? Is there anything that makes things more difficult for you?
How has this changed whilst you have been growing up?
Who or what helps you in your daily life?
What are your ambitions for the future?
What will you need to help you to achieve your ambitions?
How do you feel about moving to adult services in the future? What are you looking forward to about moving to another service? If you have already moved to an adult service – what has your experience been?
Is there anything else you would like me to tell me about yourself? Is there anything you would like to ask me?

Appendix J: Distress protocol

Distress protocol to be followed if participants become distressed during participation in the research study.

(This has been adopted and revised from the original version and is being used with the permission of Dr Chris Cocking – Lead Supervisor for this research study)

***(*© Chris Cocking, School of Nursing, University of Brighton).**

Protocol to follow if participants become distressed during participation:

This protocol has been devised to deal with the possibility that some participants may become distressed and/or agitated during their involvement in my research.

There follows below a three-step protocol detailing signs of distress that the researchers will look out for, as well as action to take at each stage when conducting the face to face or telephone interviews with parents or guardians.

The PI (Laura Gilbert) is a part Doctoral student from the School of Nursing and Midwifery at the University of Brighton and a Senior Lecturer in Health Visiting and Child Health. She is a qualified Adult Nurse, Children's Nurse and a Health Visitor and is registered with the NMC and has experience in monitoring and managing situations where distress occurs.

Laura Gilbert has completed qualitative research with children and teenagers on one previous occasion for her MSc in Paediatrics.

Mild distress:

Signs to look out for:

1) Tearfulness
2) Voice becomes choked with emotion/ difficulty speaking
3) Participant becomes distracted/ restless

Action to take:

1) Ask participant if they are happy to continue
2) Offer them time to pause and compose themselves
3) Remind them they can stop at any time they wish if they become too distressed

Severe distress:

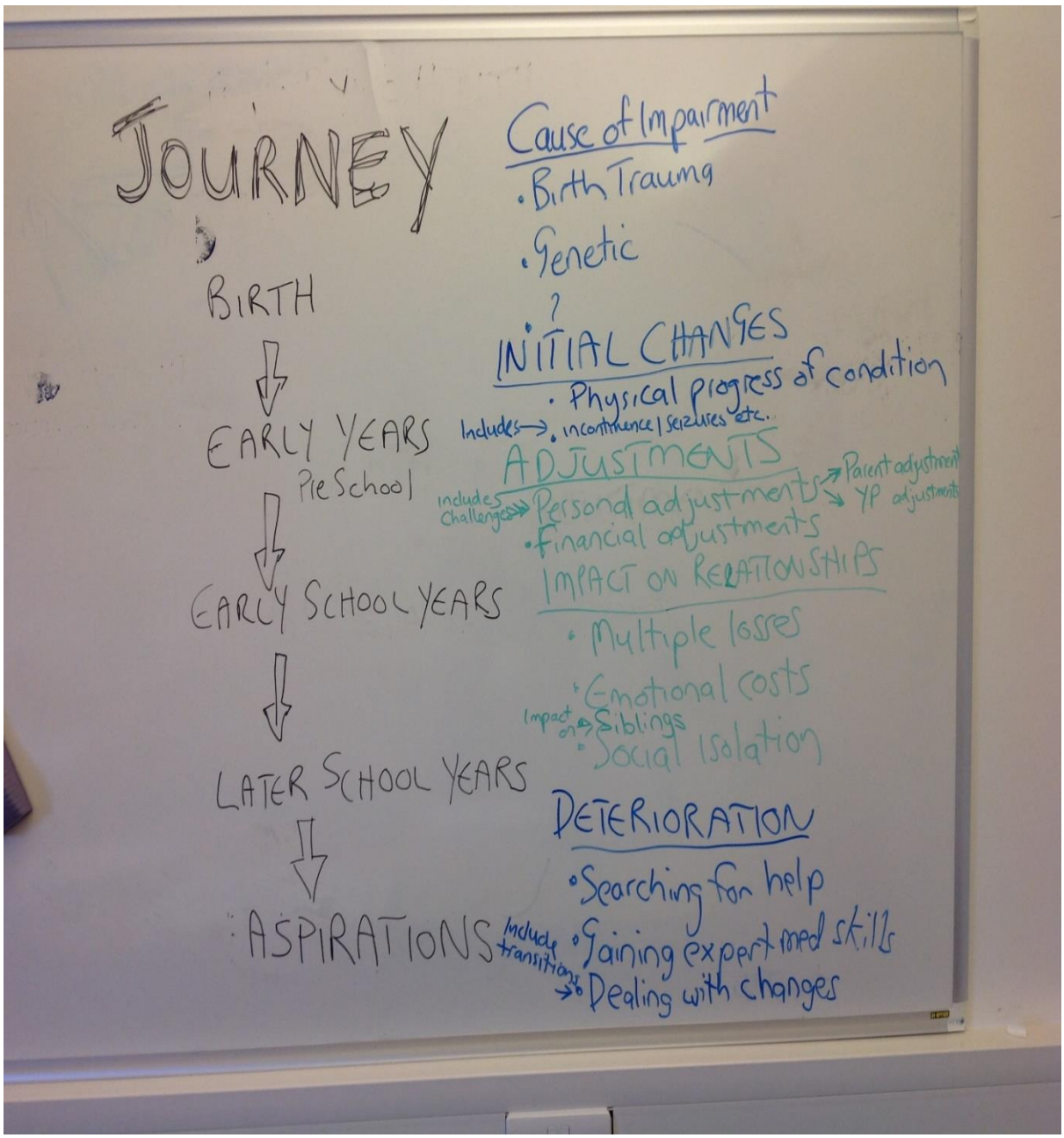
Signs to look out for:

1) Uncontrolled crying/ wailing, inability to talk coherently
2) Panic attack- e.g. hyperventilation, shaking, fear of impending heart attack
3) Intrusive thoughts of the traumatic event- e.g. flashbacks

Action to take:

1) The researcher will intervene to terminate the interview.
2) The debrief will begin immediately
3) Relaxation techniques will be suggested to regulate breathing/ reduce agitation
4) The researcher will recognize participants' distress, and reassure that their experiences are normal reactions.
5) Young people who are attending …… Hospice Care for Children to be referred to Head or Deputy Head of Care or Transition Coordinator for further support.

Appendix K: Example of early analysis



Appendix L: Example of the development of themes as part of the data analysis

PARENT INTERVIEW WITH CB Summer 2016

CATEGORY	THEME 2	THEME 1	DATA
	Impairment	virus (16) initial changes (21-24) getting to know (73) physical effects (313-315	*"He contracted a virus when he was 5 1/2/ old"* *"he was left like a floppy rag doll…very limp with no tone….it also affected his visual fields – it's the messages being sent from his brain to the eye – so we don't believe that L sees any further than sort of a hand in front of his face….."* *"and we'd all take a little packed lunch – most of the kids sadly weren't eating"* *"but immediately after the virus his hands were like this* (mother clenched his hands to show me) *we tried with physio – I did it every day with him – just trying to open them & he's now got nice supple hands – he doesn't like anything in them – trying to get him to hold a rattle – he's not – so I'm really excited to get the sand & water table back)*
	Loss	Relationship breakdown (14) Emotional cost (27) New relationship (34-36) Loss of a child (33) Social isolation (48)	*"dad and I sadly parted…."* *"and it took its toll on mum and dad…"* *"I do have a partner of 3 years now – we live separately but that suits us both – we are very set in our ways & very mature you know…"* *"we thought that we were going to lose him….."* *"until then I had never interacted with another family…."*
	New beginnings	Importance of being part of a group (54-55) Emotional & psychological support (63-65) Wanting life to be normal (28) Outcome (30)	*"I rang my mum & L's dad – and was almost in tears of joy at the fact of having met other families with other children in the similar situation as myself……."* *"there was a need for a group – a regular time, place & venue that we could go & meet & support each other…."* *"Dad kind of wanted life to just carry on & mum needed to totally focus on L…"* *"so we went our separate ways but it's all fine – Dad sees L once a week – um – 'cos he works - & more if he has time off……"*

Appendix M: Examples of data coding and theme development

4.3.4 Figure	**PARTICIPANT 1 – NATHANIAL 21 YEARS WITH DUCHENNE MUSCULAR DYSTROPHY**	**PARTICIPANT 2 – AMY (PARENT OF OLIVER AGED 15 YRS)**	**PARTICIPANT 3 – LYNDSEY AGED 21 YEARS WITH CONGENITAL MYOTONIC MUSCULAR DYSTROPHY**
Independence	*"Mum do you think you can turn the page for me?"*	*He's very long limbed and he's hypermobile – in that it's sort of genetic…..basically he's very tall and he's walking with a wide gait – he started off needing sort of Afos for his ankles and now we've got them up to here"* (mother points to half way up her own leg), *so by the time he was about 7 he needed a wheelchair when we went out and about because he can walk – we've got stairs – he can go up*	*I cannot do any personal care for myself so relying on others to be available or on time can be frustrating.*
Implications of the disease	*It's just very debilitating. My arm just wants to go off and do painting. It doesn't affect anything else – your brain and mind are just as efficient.*	*About four days after his fourth birthday he was diagnosed with epilepsy. The other major thing with him is to do with sort of receptive and expressive language.* *We were sent to …..street and they were sort of developing a group for children who had delayed speech and language – so I remember that we turned up and I was on my own with him and there were all these other parents and their children could kind of do stuff and Oliver couldn't even be in the room and it was like too much and I remember being quite – actually getting very emotional about it..* *he's got very low bone density …..so he has calcium infusions at the hospital twice a year.*	*My disability affects my life in every way that you can think of. Accessibility of places /transport for my wheelchair. Breathing impairment gives me headaches in the best case and hospitalisation in the worst. I struggle to sit up for more than a couple of hours, so finding places to lay down when away from the house is sometimes difficult. My feeding tube sometimes leaks which ruins clothing and can be embarrassing in public.*
Setting of own goals	*I've not had much luck talking to people really – especially girls – even if you are just talking to them saying hello and you know passing the time of day – they are a bit off hand with you – it's a bit – very disheartening actually but I think maybe I'll try to find another route to try and solve the problem for me.* *(Narrator: The other route is a radical one. Nick has been talking to Chris about using his savings to pay for a prostitute).*	*It would be lovely if he could do something that would help him to earn some money, but I don't know whether he would be able to. "it would be really nice if he could be involved in something as an adult that involves routine whereby he could contribute in some way. In an ideal world that would be amazing and somewhere to live where he could be safe and OK*	*My main goal is actually something a lot of people take for granted – to have my own house. Not shared accommodation or residential with communal areas and on-call staff but my own place with 24/7 live-in carers. On call staff would not meet my needs and truthfully, I want somewhere I can decorate myself furnish myself… I want somewhere I can call mine, somewhere I can call home.*
Hopes and desires	*I'm not expecting Miss World or anything – it's about being normal really. I want to have the experiences that other people do…*	*Sister aged 8 years "be a fireman.." Mum:" Be a fireman? He loves fire engines"* *The head of O's school said "can you envisage him living somewhere else?" and it's – it's really hard and yes of course one day that will have to happen but that is a big thing and um – um I'm hoping that there will be support – and there will be – you know money available for some sort of support because I can do what I can but you know if somebody else has a child that gets to 18 – and you know is able to sort of fend for themselves – they support them but it's very different both financially and emotionally and um you know – psychologically".*	*I would like to become a counsellor for other young individuals with similar disabilities.* *Like any other girl my age I want a family someday as well as a disability dog.* *I hope to become a qualified counsellor and practice from my own office at home for ease.*
Stigma		*The Health Visitor was quite old fashioned and was kind of basically I remember even at the time when he was three months old saying you know – I got the notes a few years later to*	

		see- and she was writing how I was in denial around his special needs – and I wasn't in denial – I just didn't know" I think that for me a lot of time it was other people's responses – I think when you are a parent it is very difficult and I kind of – to cut a long story short – my way of approaching it now is particularly children – other children and when they are with their parents I say "hello" and then they kind of join in or they get shy and hide behind their mums and everything but that kind of helps me because it is a positive way of dealing with that.	
Communication		*He started off with PECS* (picture exchange system) *when he was very little and that didn't really – we tried it at home but I think in the home setting it doesn't always work…..but at school it worked very well and he understands Makaton and he has now – he can do sort of three word sentences – that's over fifteen years – but we've got to that and he will talk about things he can see, he will talk about things he likes, um – he really really likes music and he can sing and he knows the words to a lot of songs we sort of do….*	