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The symptom profile and experience of children with rare life-limiting conditions: Perspectives of their families and key health professionals



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The symptom profile and experience of children with rare life-limiting conditions: Perspectives of their families and key health professionals

Executive Summary

Background

Many non-malignant life-limiting conditions are individually extremely rare and little is known, even by professionals in the field, about the actual day-to-day symptomatology or the impact of these symptoms on the child and family. With little recorded in the literature regarding the symptoms that children with rare life-limiting conditions experience, and the associated impact of managing these symptoms on the wider family, an opportunity exists to widen the knowledge base in this area.

Methods

The study aimed to use the expert views of families and practitioners to explore and gain an understanding of the symptom experience of children with the rare life-limiting conditions of Mucopolysaccharide disease (MPS) and Batten disease and the wider family impacts of caring for and supporting these children.

The study consisted of three phases: (i) a national survey to identify which rare life-limiting conditions present symptom challenges and warrant focus during the remainder of the study, (ii) a prospective daily symptom diary completed by families over a two-month period, (iii) interviews with parents, siblings and practitioners.

The mixed method approach of symptom diaries and interviews were intended to complement each other. The information collected in the diaries informed the interview schedule, and the interviews provided an opportunity to clarify and explore issues documented in the symptom diary and explore the wider family impact.

Quantitative diary data were analysed using descriptive statistics and the qualitative interview data were generated and analysed using a grounded theory approach.

Sample

Fifty-nine people completed the survey, including 43 practitioners and 16 family members. Nurses accounted for the largest proportion of care practitioners, with hospice and community nurses accounting for 40% and 23% of respondents, respectively.

Twenty-six children, from 23 families, with Mucopolysaccharide disease or Batten disease took part in the study. Seventeen children had a Mucopolysaccharidosis, including 15 children with Sanfilippo disease, one child with Hurler disease and one child with Morquio disease. Nine children had Batten disease. Diaries were completed by twenty parents relating to 22 children (13 with Sanfilippo disease, 1 with Morquio disease and eight with Batten disease). Parents of the remaining four children chose to participate in the interview phase only.

Interviews were also conducted with eight siblings and 19 practitioners. Practitioners came from a variety of disciplines, including acute and community health, education, and the voluntary sector.

Key findings

- The study focused on the symptom profile and experience of supporting a child with MPS and Batten disease.
- A high prevalence of behavioural symptoms such as hyperactivity, repetitive behaviours and agitation were documented.
- Symptoms that parents reported as being most difficult were often those that signify a loss of a skill or ability, which, in turn, signal the progression of the life-limiting condition. Consequently, though some symptoms were particularly challenging, understanding their meaning (the threat of the child dying) was critical to understanding the experience of supporting the child.
- Parents became experts in managing the condition and its associated symptoms. This expertise often developed through necessity rather than choice.
- Siblings often manage a delicate balance between protecting their unwell sibling, their parents and themselves.
- The wider family was also clearly impacted by the child's condition.
- Practitioners often provided support which was above and beyond their usual duties to support families. Many had very narrow expertise, usually focused on controlling one or two symptoms, rather than experience of the condition in its entirety.

Findings: National survey

The national survey identified a small number of conditions which would then form the basis of the subsequent phases of the research. MPS disease and Batten disease were prioritised. These conditions were prioritised, because both parents and clinicians indicated that they present considerable symptom challenges.

Findings: Symptom profile

A total of 1150 diary entries were returned, representing an overall return rate of 93%. Of the 22 children for whom symptom diaries were kept, complete diary sets containing data for 56 days were received for 11 (50%) children. Of the remaining 11, the primary stated reason for missing diary data was because the diary was not completed when the child was staying away from home in planned respite/hospice care (54%).

Prospective daily symptom diaries, combined with interview extracts pertaining to symptom management, document the symptom profile for Sanfilippo disease, Morquio disease and Batten disease.

Sanfilippo disease: The cohort of children with Sanfilippo disease included children at different stages of the condition. A wide range of symptoms were recorded. In particular, a high prevalence of behavioural symptoms such as hyperactivity, repetitive behaviours and agitation were documented. Such symptoms were also rated high in terms of severity, alongside disturbed sleep. The most challenging symptoms were behavioural, those which prove difficult to control, and those which signify progression of the condition. Practitioners found that difficulties in communicating with the child impaired their ability to assess symptoms. The strategies employed by parents to manage symptoms were moderately effective, with no symptom or strategy standing out as more or less

difficult to manage. Overall, parents reflected on the relentless nature of behavioural symptoms, and the resulting physical and mental exhaustion in those who care for, and live with, these children.

Morquio disease: The range of symptoms experienced included pain, cold extremities (hands and feet), joint stiffness, muscle spasms, vomiting, diarrhoea, breathing difficulties and cough/cold infections. Each of these symptoms was rated as low frequency with the exception of breathing difficulties. Each symptom was rated as low severity, with the most challenging symptom being breathing difficulties. Management of breathing difficulties was regarded as working only moderately well.

Batten disease: The cohort of children with Batten disease included children at different stages of the condition (with some being very advanced and others having only received a diagnosis within the past year). Symptoms of joint stiffness, agitation, secretions, cold extremities (hands and feet) and disturbed sleep were particularly high in prevalence. Of these, joint stiffness, agitation, secretions and disturbed sleep received high severity ratings. Parents perceive their interventions to manage disturbed sleep as ineffective, and joint stiffness as only moderately effective.

Overall, symptoms which were the greatest challenge to manage in Batten disease were those which signify a loss of a skill or ability and in turn signal the progression of the life-limiting condition, and those which have the potential to lead to health-related complications and other life-threatening illnesses. Practitioners found that difficulties in communicating with the child impaired their ability to assess symptoms and also found the symptom of seizures in this population of children as difficult to manage.

Findings: Family perspectives: the key role of expertise and experiential knowledge

Parents become experts in these rare life-limiting conditions, and this expertise becomes a central component of the central part of their experiences. Parents begin to suspect that something is not quite right with their child from an early age, but rarity of these conditions precludes securing a rapid differential diagnosis. Between first noticing symptoms to the point of diagnosis parents often struggled with how friends, family and members of the public responded to them and, in particular, how their identity as a parent was challenged. These negative messages, including feeling like a bad parent, were often internalised.

However, following diagnosis the narrative arc changed. Many parents began to feel more reassured that their parenting was not to blame for any ongoing physical or behavioural difficulties. They continued to develop expertise and experience in dealing with symptoms, however, the child's ongoing symptoms continued to be difficult for other people to manage. Consequently, parents often had few people they received help from, which impacted on their daily lives and limited their opportunities to spend time together as a couple. For example, parents often took shifts in caring for the child. The majority of couples spoke about their communication with each other as 'open,' but some struggled to talk with each other about how they felt. Relational difficulties were particularly apparent when couples took different positions on making decisions about treatment choices.

The wider family system was clearly impacted upon by the child's condition. Many parents found it difficult to talk in detail with their family about the child's condition, and chose to protect them from upset. By contrast, other parents felt deserted by relatives, despite wishing for more help and support. Several families had developed expertise over generations, since some had several children affected by the condition in different parts of the family. Heritability links clearly with family members' burgeoning expertise in these rare life-limiting conditions. Talk about heritability also played a role in mediating feelings of blame, and featured in decision-making about family planning and genetic screening.

The rarity of these conditions, and the limited knowledge that many practitioners have, results in parents developing their own unique symptom management methods. Parents' expertise then becomes critical for practitioners themselves to learn from in the absence of training which adequately prepares them for supporting families affected by these rare life-limiting conditions. Becoming experts appeared to stem from necessity as much as choice, and invariably was closely related with the rarity of the condition.

At times, the development of expertise functions to isolate the family. Holding expertise within the nuclear family can preclude others from supporting the child or family. A recursive loop is therefore formed, where only those with contact with the child develop experiential knowledge, and only those with expertise can care for the child. Parents spoke of both relatives and practitioners who do not have experiential or clinical knowledge to draw upon.

Expertise and experiential knowledge is offered as a theoretical model, because it offers explanatory power in understanding a considerable proportion of the data corpus. In short, the development of expertise by parents therefore explains the tensions in accessing advice, support and help. It also explains the evolution of relationships within the couple (and the development of relational tensions or interdependence) and the wider family, where relatives do not develop such expertise. The encroachment of expertise from before the child's diagnosis anchors this learning in a period where often parents seek explanations for their child's symptoms. Expertise also explains the shift in internalising messages of parenting from others, as they develop greater understanding of the condition and have sophisticated explanations for their child's symptoms.

Findings: Sibling perspectives

Siblings spoke with clarity about their ill brother or sister's condition, and summarised complex symptomatology and their role as young carers within the family. Many spoke of emotions, combining love and compassion for their sibling with frustration at those who judge people with disabilities. Further, siblings' accounts point towards a delicate balance between protecting their unwell sibling, their parents and themselves. The emotional impact of being in a family supporting a child who has a rare life-limiting condition seemed to involve a critical combination of negative disability discourses and support through peers and family members.

Findings: Practitioner perspectives

Many practitioners expressed their expertise in relation to specific symptoms and silo expertise, rather than the whole condition. Meanwhile, other interviewees felt that the specific diagnosis was less important than the overall stance in supporting the family. A consequence of many practitioners having limited disease-specific knowledge was that they often undertook care coordination to ensure the family was well supported.

While many practitioners mentioned the presence of protocols and paperwork for documenting advance care planning discussions, none privileged this as a main facilitator in having such conversations, rather, it often came down to the relationship between family and professional. Thus, while practitioners are able to identify the value in undertaking conversations about advance care planning, many were not engaging in such discussions, even for children with clear diagnoses of life-limiting conditions.

Recommendations

- Practitioners working in specialist paediatric palliative care, generalist palliative care and generic services would benefit from developing more of an awareness of the importance of behavioural symptoms on the overall experience and challenges of having a child with MPS disease or Batten disease.
- Sharing symptom assessment and management strategies between families and practitioners would be helpful.
- It would be useful to develop opportunities for practitioners to learn from each other and augment their expertise and exposure to supporting families with these conditions.
- A sequence of case studies could be developed from the data presented in this report, which would help share the experiences of families with healthcare practitioners, such as paediatricians and hospice workers.
- A series of workshops for family members and practitioners could provide a forum for generating training materials and practice-based action plans for improving supports to families.
- Clinicians in symptom-specific specialist areas could be targeted with information about the conditions, and encouraged to consider whether the condition might be manifest in new patients.
- A standardised presentation of the study could be developed which outlines the key findings and recommendations for improving practice.
- Training should be developed which is cognisant of the relational and emotional context in which practitioners will lead advance care planning conversations.
- Training programmes should have their efficacy evaluated, with outcomes focused on communicating with parents about decline, dying and bereavement, and professional responsibilities.
- Proven interventions, such as systemic family therapy, should be offered to families with the aim of supporting the couple-relationship, wider family relationships, and enhancing communication. Outcomes should be measured, recorded and reported.
- A prospective longitudinal study is warranted looking at bereavement outcomes for families, mapping psychological morbidity.
- Further research should focus on decision-making regarding family planning, management of the condition across successive generations for those who carry the gene and communication within families regarding the heritability of conditions.
- Sibling buddying could be implemented through internet, phone, texting or face-to-face methods, each allowing for different levels of contact, and taking into account the geographical challenges presented by these rare conditions.
- Proven interventions should be offered to siblings. Siblings should also be routinely signposted to existing resources to help combat isolation.

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1 Background

1.1 Symptoms in rare life-limiting conditions

Children with rare life-limiting conditions experience a wide range of unpleasant and often undetected symptoms (Himmelstein, 2006). Many non-malignant life-limiting conditions are individually extremely rare and little is known, even by professionals in the field, about the actual day-to-day symptomatology or the impact of these symptoms on the child and family. Families consider symptom control to be a high priority (ACT, 2004; Hain, 2005; Hain et al., 2008), and symptom management is considered an area of concern in paediatric palliative care (Collins et al., 2002; Jalmesell et al., 2006; Liben et al., 2008; McCluggage and Elborn, 2006). Yet symptom management in palliative care for children with life-limiting conditions other than cancer remains largely unexplored in the scientific literature, despite its identification by families and professionals as being a key research priority for children's hospices (Malcolm et al., 2008; Malcolm et al., 2009).

UK children's hospice staff identified seizures, spasms, pain assessment/management, unidentified distress and vomiting as the symptoms which cause them the most anxiety (McCluggage and Elborn, 2006). Moreover, the task of identifying the correct symptom to treat proved more difficult and so caused more anxiety for hospice doctors and nurses than treating the actual symptom itself (McCluggage and Elborn, 2006). Managing the physical dimension of symptoms such as pain is an essential first step, but alone is rarely enough. It is important to recognise the multidimensional aspects of the symptom experience, for example, the social, psychological, cultural and spiritual elements of pain (Himmelstein et al., 2004).

Symptoms are highly subjective, and thus difficult to measure; self-report is the gold-standard for symptom assessment (Collins et al., 2002). In addition to identifying the presence or prevalence of a symptom, the frequency, intensity, or degree of distress associated with the symptom must also be determined (Collins et al., 2002; Linder, 2005). Paediatric symptom research has used health diaries as a data collection tool to identify the severity of chronic pain, medication use and parent experiences of caring for their child following surgery (Butz, 2004). Using diaries can reduce inaccurate recall of health events or behaviours and provide participants with a means of closely tracking or monitoring health or illness behaviours over time as they occur in their natural environment (Gil et al., 2000; Hunt, 2006; Maikler et al., 2007).

A wide range of symptoms are known to affect children with rare life-limiting conditions such as MPS disease (Malm and Mansson, 2010; Valstar et al., 2008) and Batten disease, yet characteristics such as the onset, timing, patterns and severity of such symptoms remain largely unknown (Siden et al., 2010). A family with experience of a life-limiting paediatric condition describe the complexity of the symptoms and their interactions, necessitating the requirement for advanced support in symptom management (Wray and Wray, 2004). Developing an accurate picture of the range of symptoms, their prevalence, severity and management issues from the perspective of families who are living with these conditions would be advantageous to ensure a broader understanding of both the symptoms experienced and their wider impact on children and families.

1.2 Families' experiences of supporting a child with a life-limiting condition

Symptoms are a family affair and impact not only on the person with the condition but also on the wider family networks (Hunt et al., 2003a). Steele and Davies (2006) explored the multidimensional impact that caring for a child with a progressive life-limiting condition has on families, including the emotional, physical, financial and spiritual impacts on their lives. Other studies have explored the experiences and impacts of supporting a child with a life-limiting condition, documenting in detail accounts of families with cystic fibrosis (Bluebond-Langner, 1996) and Duchenne Muscular Dystrophy (DMD) (Thompson et al., 1992), and spina bifida and cerebral palsy (Blum et al., 1991).

These studies fit with a wider drive of attending to the voices and opinions of family members (The Cabinet Office Social Exclusion Unit, 2008).

A recent UK-wide study (Limb et al., 2010) of the experiences of 600 families affected by rare conditions identified a number of key themes which warrant the development of interventions. Areas for practice development included delayed diagnosis, misdiagnosis and practitioners' limited knowledge about the management of such conditions. It should be noted that this survey focused on all age groups and is therefore not specific to children with rare life-limiting conditions. Nevertheless, the themes identified in this broader sample offer useful insights into stressors for families affected by rare conditions, and are reinforced by findings from condition-specific studies, for example, Batten disease (Scrambler and Williams, 2008).

Family-based interventions have been successfully trialled in palliative care settings recognising the need to intervene at the level of family rather than individual members. This recognition is part of a wider move toward systemic family therapy in palliative care (Kissane and Bloch, 2002; Mehta et al., 2009). The focus on family-wide support comes in the wake of recognising the adverse psychosocial outcomes for families caring for a child with a life-limiting condition (Hain et al., 2008) and potential to reduce morbidity in bereavement.

1.3 Sibling experiences of living with a child with a life-limiting condition

The past few years have seen an increasing interest in understanding the impact of a child's health on their siblings, notably in oncology contexts (Havermans and Eiser, 2006; Martinson et al., 1990). Research findings have led to the provision of psycho-support for siblings both for those of children with curable and incurable conditions.

Studies on the impact of non-malignant conditions are more sparse in the literature, despite the provision of the vast majority of children's hospice care being delivered to those with non-cancerous diagnoses (Sheldon and Spek, 2002). A systematic search of the literature on siblings of children with non-malignant life-limiting and degenerative conditions (such as MPS disease or Batten disease) revealed that limited work has focused on this group. The studies outlined below focused primarily on conditions such as renal failure, DMD and cystic fibrosis, and map out the limited research that has begun to explore the experiences of those with a brother or sister with non-malignant life-limiting and life-threatening conditions.

Several studies have focused on the emotional impact of having a sibling with a life-limiting condition. Research has shown that siblings report high levels of empathy and concern for their unwell sibling (Kiburz, 1994; Read et al., 2010; Taylor and Fuggle, 2001). Another consistent theme identified by (Hutson and Barlow, 2007), also involved worry about the wellbeing and prognosis of the ill child. Siblings have been reported to feel worry for their ill brother/sister (Fleitas, 2000; Hutson and Barlow, 2007), and to often feel sad (Stallard et al., 1997).

In other studies, children reported feeling anger toward their unwell sibling at times (Kiburz, 1994). This may be linked to several factors including a sense of uncertainty regarding a sibling's illness which leads to a negative impact on their emotional wellbeing and jealousy around parental attention. Fanos and Wiener (1994) suggest that there is often anger and resentment toward the unwell sibling, and provide evidence to explain this finding, reporting that they feel left out. Recent research (Knapp et al., 2010) has drawn on similes of life as a rollercoaster, with extreme dips experienced during acute episodes and the return to a higher plateau during periods of stability. Further, 64% of families agreed that they had little time left over to focus on well siblings after caring for their child with a life-limiting condition (Knapp et al., 2010).

Other studies have reported how siblings see coping as integrated into their daily routine (Brown, 2009). Several studies report higher prevalence of anxiety and depression in siblings of children with

chronic disease, life-threatening and life-limiting conditions than in control groups (Hutson and Barlow, 2007; Sharpe and Rossiter, 2002). Taylor and Fuggle (2001) also identified siblings of children with a range of chronic conditions to have increased emotional problems (though these were not considered at a clinical level of pathology). Hutson and Barlow (2007) explored the experiences of healthy siblings of children with a rare genetic form of anaemia. Siblings in the study reported efforts to contain their feelings in order to avoid adding to the families' distress. One sibling spoke of purposefully internalising their distress.

Internalising behaviour was found to have a larger negative effect on children than externalising behaviours (Hutson and Barlow, 2007; Sharpe and Rossiter, 2002). Thus, siblings with a tendency to keep their emotions to themselves are more likely to experience poorer psychological functioning than siblings that externalise their feelings (e.g. through increased aggression or agitation). Psychological functioning is linked to adjustment and coping (De Graves and Aranda, 2005).

Age has also been identified as a key factor in sibling adjustment. In a quantitative study of 252 families, data indicated that, unsurprisingly, the age of the child is associated with increased knowledge of the illness (Williams et al., 2002). Other studies have reported that younger children tend to be more positive in their outlook, including being more likely to think the illness will go away (Stallard et al., 1997). Stewart et al's (1992) study focusing on siblings of children using hospice care found that younger siblings were more likely to be accepting of their role within a family and more positive towards their unwell sibling. This may be related to the fact that many of the siblings had not discussed their sibling's illness before and therefore had perhaps protected themselves from its significance. Further, Stewart et al. (1992) also reported that older siblings demonstrated higher levels of distress than children without an ill brother/sister. Distress was defined as anger toward the sibling, feelings of being left out in the family, concerns over family breakdown and financial worries (Stewart et al., 1992). The authors conclude that distress may be greater when the sibling of a child with a life-limiting or life-threatening condition has growing awareness of the tensions that illness can bring to family life.

Other studies have highlighted communication patterns between siblings and parents. Two studies have independently identified that around 50% of their study sample of children reported communicating concerns for their siblings to their parents (Batte et al., 2006; Stewart et al., 1992). This difficulty in talking has been highlighted in terms of discrepancies between child and adult reports. For example, Sharpe and Rossiter (2002) reported that 'parent reports were more negative than child self-reports' (p699). A systematic literature review (O'Brien et al., 2009) found that parental stress was associated with poorer sibling adjustment. These studies indicate the need for research methods to seek siblings' first-hand experiences rather eliciting (what are likely to be unreliable) proxy reports from parents.

Sharpe and Rossiter (2002) recommend that intervention programmes be developed to meet the needs of siblings and families of children with a chronic illness. Two recent systematic reviews (Hartling et al., 2010; O'Brien et al., 2009) identified a total of six studies which incorporated interventions for siblings of children with non-malignant chronic illness/disability. Findings indicate positive results, for example, a family-based six-week psycho-educational-based intervention found an improvement in siblings' emotional wellbeing and coping strategies (Giallo and Gavida-Payne, 2008). Williams (2003) also reported positive effects following a five-day psycho-social intervention; sibling knowledge, mood and behaviour outcomes were all sustained at the 12-month follow-up. In contrast, Gursky's (2007) short psycho-educational intervention reported reduced anxiety levels; however, sibling anxiety remained above the normal threshold.

1.4 Advance Care Planning

Advance care planning (ACP) describes a process of discussion between families and care providers about preferences and priorities for future care. ACP is particularly important for children with a life-

limiting condition as their condition will deteriorate in the future. Discussions and decision-making about what care is wished for by the family will facilitate better care during acute and terminal episodes of care. For example, early discussions about not attempting resuscitation mean that care protocols can be drawn up and these measures will protect parents from having to make difficult decisions in the midst of an acute episode. Clinicians are known to experience difficulties in raising issues of end-of-life care when clinical management focuses on prolonging life and managing symptoms.

However, empirical studies of ACP have identified that although this is typically a difficult conversation to engage in, parents find it extremely helpful in ensuring the best quality of care for their child, and avoiding unnecessary distress (Hammes et al., 2005). Other studies report that parents appreciate open and honest communication, while recognising that pacing is important (Kirk et al., 2004). Referring to the Association of Children's Palliative care ACT pathway, Fraser et al. (2010) advise that bereavement care and ACP should be an ongoing process that commences at the time of diagnosis.

Thus, while recognised as challenging, best practice in palliative care involves undertaking ACP flexibly with families and is recommended to be initiated from diagnosis, rather than waiting for acute exacerbations to occur.

1.5 Mucopolysaccharidoses and Batten disease

Batten disease and mucopolysaccharidosis are examples of lysosomal storage diseases (LSD). They are characterised by relentless and inevitable progression from birth, leading to death in childhood, usually during the second decade of life. Both conditions belong to ACT/RCPCH group three (ACT/RCPCH, 2009).

Both conditions are caused by an inherited inability of the body to produce an enzyme. Normally, the smooth functioning of the body depends on complex metabolic pathways, which build up complex molecules from simple ones, and then modify them to act as structural elements or chemical 'messengers'. Each step in these pathways can proceed only in the presence of a specific enzyme. Each enzyme is in turn dependent on the normal functioning of a specific gene. If that gene is faulty, the enzyme itself will be absent or will not function, and consequently, the metabolic pathway cannot proceed.

In the case of lysosomal (LSD) storage diseases, the result is that quantities of the unfinished molecule accumulate. The body, lacking the capacity either to use or to destroy them, simply has to let them accumulate. It is the physical accumulation of these extra molecules that leads to the clinical manifestations of storage diseases. The clinical features of the disease are influenced by the location of the molecule deposits. Typically, signs and symptoms are mild in the first few months or years of life, before the molecule accumulates significantly. Consequently, recognition of the condition in early childhood can be preceded by the child appearing to reach developmental milestones; this is then often followed by a period of diagnostic uncertainty that can be deeply frustrating and distressing.

Sanfilippo disease and Morquio disease belong to a larger group of conditions called mucopolysaccharidoses (MPS). The molecule that accumulates is heparan sulphate (in Sanfilippo disease), or keratan sulphate (in Morquio disease), which are deposited in many different locations, including the brain and nerves. This leads, in the first instance, to behavioural disturbances, such as hyperactivity, aggressive behaviour and sleep disturbance. These become increasingly severe as the condition progresses and there is further accumulation of the molecule. Towards the end of life, as nerve damage becomes so severe so that they no longer function at all, immobility and responsiveness are more characteristic. Seizures, caused by nerve damage, are common but not universal (Malm and Mansson, 2010; Valstar et al., 2008; Valstar et al., 2010).

In Batten disease, the abnormal molecule that accumulates is lipofuscin. While it accumulates in a variety of tissues, its presence in the central nervous system is what gives the major manifestations of the condition. Seizures or visual disturbance are early signs, and cognitive changes such as slow learning, or even loss, of new skills, behaviour changes, clumsiness, and repetitive speech. Other physical manifestations include poor vascular circulation, and changes in breathing pattern.

Since the abnormal molecule is located anatomically in nerve cells, neurological symptoms are particularly severe and prominent in the course of Batten disease. Severe cognitive impairment, worsening seizures, and progressive loss of faculties are usual. Children with the disease will typically enter a palliative phase with significant symptoms such as blindness, extremely limited in mobility, with little cognitive function, and often experience severe seizures that are difficult to control (Kohlschutter and Schulz, 2009).

The hallmark of MPS and Batten disease is ongoing neuronal damage. In this respect, it is different, for example, from static injuries, such as hypoxic ischaemic encephalopathy causing cerebral palsy, or other causes of seizures. A characteristic of LSDs is that seizures become increasingly severe and difficult to control as there is relentlessly progressive damage to the neurons. This is particularly likely in Batten disease.

1.5.1 Incidence

There are limited epidemiological studies, particularly in the UK, documenting the prevalence of LSDs. A recent retrospective study analysed data on cases diagnosed in the Czech Republic between 1978 and 2008 and reported an overall birth prevalence of 12.25 per 100,000. This prevalence rate is comparable with other published European data, specifically that of the Netherlands (14 per 100,000), Italy (12.1 per 100,000) and a slightly higher rate in Portugal (25 per 100,000) (Poupetova et al., 2010).

Poupetova (2010) reported the most frequent LSDs as lipidoses (such as Gaucher, Niemann-Pick A, B and C, and Fabry disease), mucopolysaccharidoses, and neuronal ceroid lipofuscinoses with a birth prevalence of 5.0, 3.72 and 2.29 per 100,000 live births, respectively.

The MPS society (2010) report data for individual sub-diagnoses of Sanfilippo disease (1:89,000) and Morquio disease (1:220,000).

1.5.2 Summary

Thus, MPS and Batten disease are rare life-limiting conditions which are devastating and progressive in nature, with no curative treatments available. Efforts must therefore be made to achieve an advanced understanding of the assessment and management of the complex array of symptoms that children with these conditions experience. Being better positioned to support families in dealing with these symptoms will help promote optimal quality-of-life for children and their families.

1.6 Study objectives

The evidence base of paediatric palliative care is limited, however, there is a burgeoning body of research focusing on developing practice (Cooley et al., 2000; Liben et al., 2008). The rarity and complexity of conditions in this population has been cited as a difficulty in developing evidence-based practice (Emond and Eaton, 2004).

This study aimed to use the expert views of families and practitioners to explore and gain an understanding of the symptoms experience of children with rare life-limiting conditions.

Aims:

1. To use a daily symptom diary, completed by a parent or primary carer, over a two-month period to describe and document the symptom prevalence, severity and management strategies for children with MPS disease and Batten disease. The primary function of the diary was to provide prospective longitudinal understanding of the commonly experienced symptoms in children with MPS disease and Batten disease over a finite period of time.
2. To use interviews with parents, siblings and key practitioners to understand the impact and experience of supporting a child with MPS disease and Batten disease.

The mixed method approach was employed with the intent that the use of symptom diaries and interviews would complement each other. The information collected in the diaries aimed to inform the interview schedule, and the interviews would then provide an opportunity to clarify and explore issues documented in the symptom diary.

2 Methods

Three methods were used within this study, and each is described in detail below. They are presented as distinct phases in this methods section for clarity, though the symptom diary and interviews (phases 2 and 3) were conducted concurrently. The mix of methods enabled the development of a national dataset on conditions which are considered difficult to manage, alongside the in-depth exploration of symptoms as they occurred through prospective diary data, and face-to-face interviews which reflected back on experiences.

The data findings are presented thematically, that is, section 3 reports the findings from the survey, section 4 reports the symptom profile of children with rare life-limiting conditions, section 5 reports parent experiences, section 6 sibling experiences, and section 7 reports practitioner experiences.

The three phases are each described below, and illustrated in Figure 1.

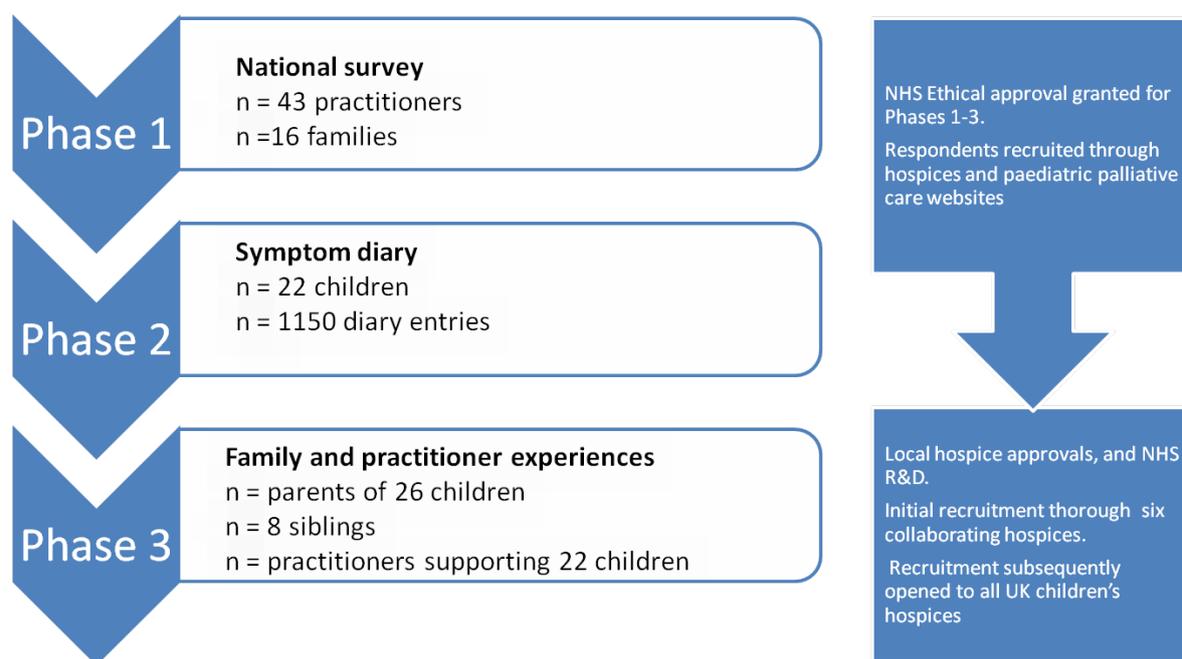


Figure 1: Methodology

2.1 Phase 1: National survey

Two surveys were designed; one for families and one for healthcare practitioners.

The survey invited respondents to nominate which rare life-limiting conditions (excluding cancer) present significant symptomatic challenges for families and professionals, and should form the focus of the subsequent research into the symptom profiles and experience of children and their families. Practitioners were invited to comment on the range of conditions they had experience of. The survey for families invited respondents to indicate which rare life-limiting conditions (excluding cancer) they have experience of. They were then asked to rate, on a 10-point Likert scale, how easy or difficult specific symptoms are to manage for the condition they mentioned. The third question asked if there were any additional symptoms related to the condition and, where possible, to describe and rate them on the same 10-point scale. The final question provided a free-text space where family respondents were invited to describe any information or support available to help manage their child's symptoms.

2.1.1 Survey procedure

Surveys were administered using online software, SurveyMonkey™, in order to gather the views of both practitioners and families. The Association for Children's Palliative Care (ACT) hosted the surveys on their website. Additionally, the national charity, Children's Hospices UK, distributed email versions of the surveys on our behalf to all Heads of Care, or equivalent, at each children's hospice across the UK and requested that the surveys be made available to professionals and families for completion either by providing paper versions within the hospice or by directing potential respondents to the online version. A reminder telephone call was made by the research team to each of the hospices to increase response rates. The time frame for completion of the survey was three weeks.

2.1.2 Survey analysis

Responses were analysed by frequency counts. Qualitative data provided in response to free-text questions were examined thematically to identify patterns within responses.

2.2 Phase 2: Symptom diary

The aim of the symptom diary was to describe and document the range of symptoms experienced by children with MPS disease and Batten disease, the severity of these symptoms and the interventions and strategies employed by families to manage them.

Gathering detail about relevant symptoms is reliant on accurate recall. Completing daily diary records facilitate the obtainment of accurate information as they allow reporting of events shortly after they occur, thus reducing errors in recall when information is gathered retrospectively. A recent study found the use of a daily symptom diary in adults resulted in increased recall of daily symptoms and increased perception of the severity of these symptoms (Ferrari and Russell, 2010).

Diaries have been used to assess symptoms in a variety of paediatric populations, including pain (Cohen et al., 2007). However, due to the rarity of these conditions, and the limited evidence base on symptoms, it was necessary to develop a prospective daily diary, since no pre-existing measure was appropriate to the study's needs.

2.2.1 Diary development

Diaries as a methodology in social research have been described extensively by Alaszewski (2006). He proposes development of diaries as a methodology based on the 'what, where and who' of the required data, proposing that they are an appropriate approach for the study of 'access[ing] those facets of social life which members of social groups take for granted and are therefore not easily articulated or accessed through research methods such as interviews ... diaries can be used to access such tacit knowledge' (p37). Further, Stone et al. summarise that diaries enable the capture of experience close to the time of occurrence thereby producing more accurate and reliable data (Stone et al., 2003). Alaszewski's guidelines for diary development were drawn upon to design the tool, in particular the structure of the record-keeping system, the guidance on using the diary and the 'training' of participants in completing the tool.

The symptom diary was developed with contributions from experts in the field. The research literature and resources available from the Society for Mucopolysaccharide Diseases (MPS Society) and the Batten Disease Family Association (BDFA) informed the initial range of symptoms to be included in the diary. The project Steering Group, which consisted of experts in policy, practice and research related to children's palliative and hospice care and parent advisors, provided additional key input to the content of the diary.

A sequence of questions was placed at the end of each week's diary to capture parents' reflections on the ways in which symptoms impacted on their child's usual activities, and the impact these symptoms have on family life. These reflections were then followed-up by the researcher when the

family interview was conducted, allowing clarification and further exploration of the wider impact of the symptoms.

2.2.2 Symptom diary pilot

A pilot study was undertaken to gauge the feasibility, utility and content of the symptom diary. Additionally, feedback was sought on the supporting documents (for example, instructions on how to complete the diary). Five families were invited to participate in the pilot study and of these, four agreed to take part. Two families had a child with Sanfilippo disease (Type A) and two families had a child with Batten disease (Infantile). Families were asked to complete the symptom diary daily for a period of one week. Each family was guided through the diary by a researcher to allow them to become more familiar with the task and to ask any questions about the process. The completed diaries were posted back to the research team and a follow-up telephone feedback session arranged.

During the feedback session cognitive testing techniques were utilised to further improve the design and development of the symptom diary. Cognitive testing is a technique which assists researchers in determining whether or not respondents understand survey questions consistently and in the way the researchers intended, and whether respondents have the information they require to answer the survey questions accurately and meaningfully. There are two main methods employed in cognitive testing: i) think-aloud techniques, where respondents are asked to verbalise their thoughts while answering the survey questions; and ii) verbal probing where respondents are asked to explain what they understand a question to mean and to describe the thought process they went through to reach the answer they provided (Alaimo et al., 1999).

A combination of the above methods was used, and was effective in identifying several areas within the diary that required improvements.

The pilot study confirmed the instructions for completing the diaries were clear and understandable and respondents felt the time taken to complete the diaries was reasonable. Refinements to the symptom diary were made based on the feedback from the cognitive testing. In particular, the addition of the symptoms: cold hands and feet, joint stiffness, disturbed sleep, hyperactivity, aggressive behaviour, repetitive behaviours (such as chewing), and choking, and a clearer demarcation of respiratory and stomach symptoms were made. Notably, the piloting phase resulted in the inclusion of behavioural symptoms which are rarely reported in the research/medical literature, yet pose challenges to families.

Seventeen core symptoms were included in the final version of the diary. The rating scale for frequency of symptoms was finalised as a five-point Likert scale, with 0 being 'not at all' and 5 being 'all of the time'. Similar changes were made to scales recording severity and effectiveness. The pilot phase played an important role in the development of the final symptom diary.

The final version of the diary consisted of a side-bound booklet printed on quality A4 paper with alternate paper colour coding of the days to allow for ease of identification. An acetate front and back cover was included to protect the diary. The front page included detailed instructions on how to complete the symptom diary and contact details for the research team should participants require any advice on diary completion. An example of a correctly completed diary for each condition was also sent to participating families.

Comments provided by families participating in the pilot study were positive and highlighted some of the benefits families experienced in completing the symptom diary:

I found the experience of filling out the diary very interesting. It would be good to look back over the weeks and see how things are changing because sometimes you get so caught up in the day-to-day management.

It would be interesting to get a profile over time and to have a bigger survey done to hopefully gain from the experience of others. (Pilot study family)

An example of the final version of the symptom diary is included in Appendix 1.

2.2.3 Symptom diary procedure

Families consented to complete symptom diaries daily for a period of eight weeks. Families were able to choose from a printed or electronic version of the diary. Printed copies were mailed to participants regularly, ensuring that new diaries arrived in time for the eight weeks to be recorded continuously. Prepaid return envelopes were provided.

The research team adopted an individualised approach to supporting families during this phase. That is, each family had a named researcher with whom they coordinated completion of the diaries and scheduling of conversations. Phone-calls and/or emails were used to facilitate adherence to diary completion (in line with Alaszewski's sense of 'training' in completing the tool), and to answer any queries arising from the diaries.

2.2.4 Inclusion criteria

- Families of children/young people who have a life-limiting condition, specified as Mucopolysaccharidosis disease (MPS) or Batten disease.
- Families of children/young people who are supported by a UK children's hospice, or condition-specific support organisation.

2.2.5 Exclusion criteria

- Children or young people who live in residential schools (to ensure the focus remained on the family impact, where parents are the primary caregivers).

2.2.6 Diary analysis

Descriptive statistics were used to describe the demographic and clinical characteristics of the participants and to summarise the frequency and severity of each symptom experienced during the 56-day symptom diary period.

The qualitative family and practitioner interview data were analysed drawing on the principles of grounded theory to explore the symptom experience.

The distinct differences in the main symptoms experienced by children with MPS disease and Batten disease necessitated analysing and reporting the symptom diary data separately for each condition. Looking at symptoms within each life-limiting condition was further complicated by differences in disease stage and rate of progression from one child to another and the resultant differences in symptoms experienced. The benefit, however, of the heterogeneous sample is that having children at different stages of MPS disease and Batten disease, and further, with sub-classifications of each condition, enables the ability to plot a broad range and breadth of symptoms within the symptom profiles.

2.3 Phase 3: Interview methodology

The interview component to the study was informed by the principles and practices of grounded theory, drawing on Charmaz's (2006) use of both Strauss and Glaser's development of this as a

methodology. Grounded theory was identified as the methodology of choice to facilitate the exploration of qualitative accounts generated by family members, relating to their experiences in supporting a child with MPS disease or Batten disease.

In practice, using grounded theory facilitates an iterative cycle of analysis of data, data collection and theory generation. Charmaz (2006) frames this as developing 'specific concepts by studying the data and examining our ideas through successive levels of analysis' (p17). The initial research idea and questions are considered 'sensitising concepts', that is, departure points for a more rigorous and thorough exploration of the topic area. Consequently, after the first round of data collection, refinements were made to the interview schedules to reflect the learning generated. Thus, as the study progressed the interview questions were shaped to enable further exploration of key concepts and issues that families raised. These included a focus on the significance of symptoms on parental and family identity and the meanings associated with symptoms (to allow the interviews to move away from a concrete recording of which symptoms occurred and an exploration of why such symptoms held significance for families).

Analysis proceeded to the development of categories, sub-categories and theoretical models that crystallise key components to the experience of supporting a child with a rare life-limiting condition.

2.4 Recruitment

Recruitment was largely conducted through children's hospices. Meetings/telephone calls with key staff in collaborating hospices were conducted to inform them of the study and gain their engagement with the study. Collaborating hospices then identified the number of children that they supported who had a diagnosis of MPS disease or Batten disease. Hospice staff then undertook to determine the age, sex and familial circumstances of the child, and determine whether they fitted the inclusion criteria.

Standardised letters of invitation and information sheets for adults (i.e. parents) and siblings were sent to key contacts within collaborating hospices. Each participating hospice forwarded these letters on to families. The information sheets clearly defined the purpose of the study and outlined the participant's involvement, including the completion of a symptom diary for eight weeks and an interview. The invitation letter requested interested families to contact the research team by telephone.

In line with ethical and legal frameworks (such as data protection) the research team did not contact families without their explicit consent. If a family made no contact within a two-week period, the hospice reminded them of the study. No further contact was made to the families after this reminder.

Researchers informed the relevant hospice if a family was willing to take part. Only the name of the family was disclosed to the hospice. This precaution was taken in order to ensure that families were not sent a reminder letter and to ensure that the hospice was aware of participation in case the family wished to discuss the study or their participation with the hospice staff.

Families who were recruited through support organisations (such as the MPS Society) were given information sheets regarding the study.

All families who made contact with the research team were given additional verbal information about the study during a telephone call. Families were offered opportunities to ask any questions and the researcher to request a formal consent form to be posted to them. Once the consent was given, two weeks of symptom diaries were posted to the family, and a follow-up conversation was arranged to help guide them through the diary process.

2.4.1 Parent interview procedure

Parents who gave consent to take part in the study were asked to participate in the interview. Interview dates were arranged directly with the parent who had taken responsibility for completing the diary. If another parent was actively involved in the care of the child (cohabiting) they were also invited to take part in the interview. Interviews were scheduled to take place following, or near the end of, diary completion so that discussion of diary entries could form part of the interview schedule.

Some families consented to take part in only an interview (n=3). These families were prompted to discuss their child's symptoms in order to ascertain an outline of the symptom profile and most challenging symptoms to manage.

All interviews were recorded and transcribed in full.

2.4.2 Interviewing siblings

To ensure a wide and encompassing understanding of how rare life-limiting conditions affect the whole family system it was important to include the views of siblings. Children were engaged with as experts on their own lives. The interview method was adapted to connect with their playfulness in a way that empowered them to communicate their views and experiences with the research team. Visual methods have been considered particularly appropriate for this task (Burke, 2008).

To facilitate this method, a card-sort technique was adopted, whereby cards with symbols representing symptoms are used to support the conversation, and thicken description of events/symptoms/views. The aim was to ensure a child-centred interaction that provided a playful component which, in turn, would enrich and stimulate the child's narrative.

The use of symbols, as a low-tech approach to ensuring that people with communication difficulties are able to have their views heard, has a lengthy history in supporting people with learning disabilities. Latterly, developments in this method have been used to augment communication in adults with communication difficulties, notably dementia and aphasia, for example, Talking Mats® and Boardmaker. Recent research has demonstrated statistically significant increases in the amount of talk by participants when using augmented communication tools (Murphy, 2009).

Card-sorts have been successfully used as a method in nursing research (Hooker, 1997) and children's services (Lewis et al., 2008). Despite this growth in card-sorts as a method, there are few markers for applying this as a research technique, with numbers of cards varying widely, from eight (Lewis et al., 2008), to 90 (Murphy, 2009). Participants use the cards to 'think physically', that is, to move the cards around to develop new and changing interactions and levels of meanings between expressed ideas. Card-sort techniques often require participants to rank responses, for example, positive/neutral/negative for Talking Mats® or in an 'x is more important than y' hierarchy (Hooker, 1997).

Analyses of responses to studies that have applied this technique are largely qualitative, informed by a thematic analysis. Analyses of card-sort activities, at an interactional level, remain under-theorised. This study adopted the card-sort from a constructionist standpoint and theorised the communication about symptoms as systemic. That is, understanding the views expressed as created in the moment, rather than reflecting a singular truth of experience. Cards were likely to help express a sense of how symptoms are likely to affect each other and have recursive relationships, illustrating how symptoms are experienced as overlapping and impacting each other.

Alongside the card-sort, a traditional interview schedule was also used to guide the conversation and to invite certain conversation about the symptoms presented on the cards. Questions were devised on the basis of a systematic review of the literature, reported above in section 1.3.

2.4.3 Sibling interview procedure

Cards were developed representing a range of physical and behavioural symptoms. Each card had a picture on one side and a descriptor on the other. The image below, for example, was used to illustrate pain.



Cards were presented at the outset of the interview, and placed on a table/carpet to enable each of the cards to be seen. The researcher then led a conversation about each symptom, eliciting the child's views of which symptoms were relevant to their sibling. Cards were left visible to the participant so that they had the opportunity to re-visit and re-narrate their relationship to each symptom as the conversation developed. Blank cards were also presented, so

that the child could draw their own representation of a symptom and use that to prompt further narratives and probing from the researcher.

In families where there was more than one sibling, the children were given the opportunity to choose whether they were interviewed together or separately. Two families had more than one sibling wishing to participate. In one such family, the mother asked that the children be interviewed together as they had a close relationship. The siblings in the second family choose to be interviewed separately, a decision that was based on the siblings' availability.

All interviews were recorded and transcribed in full.

2.4.4 Practitioner interview procedure

Each family was asked to recommend a practitioner who they felt would be best placed to contribute to a telephone interview on issues relating to their child's condition. Practitioners could come from a variety of domains including health, hospice, education, caregivers, and charities. Following the families' recommendation, the research team followed the appropriate ethical protocols to ensure that permission was obtained for each of the participating localities. Once permission was established, a written invitation was sent to the relevant practitioner including a detailed information sheet outlining the purpose of the study and their proposed involvement. The practitioner was invited to make contact with the research team informing their decision regarding participation. If a practitioner did not make contact within two weeks of the invitation, the researcher then made a telephone enquiry regarding the prospective interview. In three instances practitioners were un-contactable and subsequently the family was asked to identify a second practitioner for interview.

Where practitioners consented, a detailed interview schedule and confirmation letter was sent to them. All telephone interviews included a verbal confirmation of consent, and were subsequently recorded and transcribed in full.

2.5 Recruitment challenges

The study experienced difficulties in accessing families with a child with these rare conditions. The range of challenges to recruitment are discussed below.

For some collaborating hospices, there had been significant shifts in numbers of children due to multiple deaths. One hospice reported 'an average of 10 deaths per week' over the winter of 2009/10. Additionally, some hospices reported feeling that the diary completion may be too onerous for families, and consequently chose not to approach parents to participate. Gate-keeping by hospice staff was also evident in reported actions of staff not approaching families where the child was close to end-of-life. One hospice also expressed concerns that anonymity for families may be difficult to maintain in a study focused on rare conditions. Consequently, they did not wish to recruit

families to the study. One final factor related to hospice-project features was that some families expressed a commitment to supporting their local hospice, but no interest in contributing to wider studies. Thus, by virtue of the study being national in its scope, some parents felt that the research was not adequately connected to *their* hospice.

Some families had initially expressed a desire not to complete a diary. This feedback was integrated into the methodology offering interview-only to families keen to participate who ceased completion of the diaries during the study. Other families did not meet recruitment criteria, for example, living at home, and were consequently not eligible to participate. Adherence to this inclusion criterion was enforced to ensure that the study was able to accurately report on its aim to document the study's 'impact on families.'

Recruitment was strongest where hospice staff actively spoke with families about the study, demonstrating their own interest and enthusiasm in the study.

2.6 Final study sample size

Fifty-nine responses were received for the national survey; 43 from care practitioners and 16 from families.

Completed diaries were received for 22 children, and parents of each of these children participated in interviews. Parents of a further 4 children chose to take part in an interview only. Consequently, qualitative interviews were conducted with parents of 26 children.

Eight siblings took part in interviews.

Nineteen practitioners participated in interviews about 22 children.

2.7 Ethical considerations

This study was approved by the NHS Forth Valley ethical review committee (study number 09/S0501/60). Subsequently, local hospice approvals and NHS R&D approvals were processed and received.

Preserving anonymity is an important consideration in this study. With complex data but small sample sizes, maintaining anonymity presents a challenge in reporting the findings. Families and practitioners have been randomly assigned identifiers throughout this report.

In Sections 5, 6 and 7, identifiers from 1-23 indicate families, and 24-42 indicate practitioners. In section 4 a different approach is taken. This distinction is necessary so that participants' identities are not unwittingly identifiable through the symptom details presented. For example, only one family participated who had a child with Morquio disease; it is important that data on this family are reported, but are not explicitly tied with accounts provided at interview. Consequently, alphabetical identifiers have been randomly assigned for each family and practitioner in section 4.

3 Findings: National survey

This section reports the national survey where the conditions that form the rest of the study were defined and determined. Sections 4-7 report data on the symptom profile and family/practitioner experiences of supporting children with rare life-limiting conditions.

3.1 Clinicians

Forty-three care professionals completed the survey. Nurses formed the largest proportion of respondents, with hospice and community nurses accounting for 40% and 23% of respondents, respectively.

The life-limiting conditions most frequently identified by care professionals were mucopolysaccharidoses disease (MPS), specifically Hurler, Hunter and Sanfilippo (n=15), leukodystrophy (n=10), and Batten disease (n=9). All of these conditions fall under the broader category of progressive, inherited metabolic disorders. An additional five respondents did not clearly articulate a specific condition but instead provided responses such as 'progressive neurodegenerative' or 'progressive neuro-metabolic conditions', which could be assumed to include any of the above named life-limiting conditions. Table 1 shows the full range of responses from clinicians.

Respondents gave qualitative accounts to explain the ways in which MPS disease, leukodystrophy and Batten disease present significant symptom challenges. Thematic analysis identified three main patterns similar across the conditions: (i) the presence of difficult to manage symptoms, (ii) the limited evidence base to inform practice, and (iii) the degenerative nature of conditions and subsequent long-term impact on families. The intersections between these themes are significant, representing the combination of factors that make supporting children and families affected by MPS disease, leukodystrophy and Batten disease particularly challenging. In the sections below, direct quotations are taken from the surveys and presented in italics with some details of the respondents' professional context.

Table 1. Conditions identified by care professionals (n=43) *Clinicians could identify more than one life-limiting condition

Life-limiting condition	n
Mucopolysaccharide disease (MPS)	15
Leukodystrophy	10
Batten disease	9
Spinal Muscular Atrophy (SMA)	6
Progressive neurodegenerative/neuro-metabolic disorders	5
Edwards Syndrome (Trisomy 18)	4
Niemann-Pick disease	4
Metabolic disorders (complex)	2
Neuroacanthocytosis	2
Lissencephaly/microcephaly	2
DiGeorge syndrome	2
Huntingtons	2
Mitochondrial Chain Disease	2
Epidermolysis Bullosa junctional type	2
Inborn error of GABA/glycine metabolism	1
Sandhoff disease	1
I Cell disease	1
Non-ketotic hyperglycinaemia	1
Leigh's disease	1
Hydrocephaly	1
Anencephaly	1
West syndrome	1
Otahara syndrome	1
Undefined genetic disorders with complex epilepsy	1
Complex epileptic and epileptiform seizures	1
Lennox-Gestaut Syndrome	1
Infantile Epileptic Encephalopathy	1
Primary Pulmonary Hypertension	1
Conditions with unsuccessful bone marrow transplant (BMT)	1
Polyarteris Nodosa	1
Undiagnosed conditions with life-limiting symptoms	1
Prader-Willi syndrome	1
Wolf-Hirschorn Syndrome	1
Pallister-Killian Mosaic Syndrome	1
CHARGE Syndrome	1
Epidermolysis Bullosa mosaic type	1
Prolonged QT Syndrome resulting in hypoxic brain injury	1

3.1.1 Presence of symptoms which are difficult to manage

MPS disease, leukodystrophy and Batten disease exhibit symptoms which prove problematic for professionals and families to assess and manage. Their complexity, unpredictability and individuality pose challenges for symptom management:

All these conditions have intractable symptoms which are difficult to manage, and last for many years in some instances. The symptoms are difficult to attribute to the disease process and this makes it hard for families to receive consistent advice and support. (Profession not stated, commenting on Batten disease, leukodystrophy, Sanfilippo and Niemann-Pick Disease).

Symptoms are unpredictable, difficult to manage and there is little information around. The disease trajectory differs from child to child. (Community nurse, commenting on Sanfilippo)

The length and complexity of deteriorating symptoms, parents often met with challenges when persistent in trying to get symptoms under control. (Hospice nurse, commenting on Batten disease).

Children and young people will experience different symptoms, different degrees and different rates of progression of these conditions. This individuality makes it more difficult to control the symptoms. (Social Worker, commenting on MPS disease and Spinal Muscular Atrophy (SMA)).

3.1.2 A requirement for additional clinical evidence to support symptom management

The rarity of such conditions contributes to the limited information or clinical evidence available to support symptom identification and management for both professionals and families:

There appears to be limited evidence to support clinicians' palliative interventions in these fields. Many of us rely on our own anecdotal experience as augmented by the [VIIth edition] of the Rainbows Symptom Guide, and there is little to pull the range of treatments together. (Hospice doctor, commenting on SMA, MPS disease and progressive neurodegenerative conditions).

Symptom control can be very difficult. Sources of advice are few [and sometimes contradictory] and in my experience children can spend considerable time in hospital with unsuccessful attempts at symptom control which is dreadful for them and their families, and wasteful of resources. It can be difficult to manage dystonia, agitation, pain and fits all occurring together. (Hospice doctor, commenting on Batten disease and similar neurodegenerative conditions).

Specific challenges are hyperactivity, behavioural problems, rapid deterioration, secretion build up, feeding difficulties, seizure control and choking. As these [conditions] are very rare there is not so much information available for families. (Hospice nurse, commenting on MPS disease).

Respondents thereby identify evidence and information challenges for both families and professionals [in hospitals, though notably also hospices struggling with largely anecdotal evidence]. Evidence is identified as lacking in traditional symptom domains such as pain management, but also in wider psychosocial areas such as behavioural challenges.

3.1.3 Progressive nature of the disease and resultant impact on families

Care professionals highlighted the relentlessly progressive nature of life-limiting conditions such as Batten disease and MPS disease.

The impact on the family is huge because in these degenerative conditions the children begin life developing normally and then begin to lose skills and deteriorate slowly over a number of years. As medicine advances these children are surviving beyond their life expectancy therefore living longer with difficult symptoms which can affect their quality-of-life. (Hospice nurse, commenting on Infantile Batten disease, leukodystrophy and Sanfilippo)

I think children affected by this group of conditions suffer a lot in terms of slow degeneration and loss of skills. Perhaps the study could tell us more about their symptoms and how to manage these symptoms better. (Hospice nurse, commenting on neurologically degenerative conditions).

Respondents identified the wider systemic impact of symptoms, as impacting upon family members, which presents a further dimension when supporting a child with a rare life-limiting condition.

The progressive nature of these diseases, combined with improved medical care which means many children are living longer and the disease is progressing further into its course, has considerable consequences for symptom management and also for the impact of the condition upon the child themselves and upon the family. (Allied Health Professional (AHP) commenting on Batten disease and MPS disease)

The move from a healthy child to a child with complex emotional and physical needs, with the loss of gained developmental skills impacts on every aspect of the functioning family. (Hospice nurse, commenting on MPS disease)

3.2 Families

Sixteen families completed the survey. Thirteen respondents identified themselves as the parent of a child or young person who uses a hospice (demographic data were missing for the remaining three respondents). The life-limiting conditions cited by family respondents as being those they have experience of are listed in Table 2.

Table 2. Life-limiting conditions described by family respondents (n=16)

Life-limiting condition	n
Cerebral palsy	3
Batten disease	2
Niemann-Pick disease	2
Carbamoyl Phosphate Synthetase deficiency	1
Goldenhar syndrome	1
Deletion of part of the long arm of chromosome number 6	1
Arteriovenous malformation	1
Idiopathic Encephalopathy	1
Polyneuropathy	1
Epilepsy	1
Congenital heart condition	1
Not Stated	1

Ratings of how easy or difficult families find the management of selected symptoms were grouped into low (rating of 1-3), medium (rating of 4-6) and high (7-10) (Table 3). More than half of respondents (56%) considered seizures to be very difficult to manage. Drooling and muscle spasms were also identified as difficult symptoms to manage with 50% and 44% of respondents giving a high rating, respectively. Pain and breathing difficulties were the symptoms to receive the least number of high ratings by 25% and 19% of respondents, respectively. Nearly half of respondents (44%) described the symptom of pain as 'not applicable' and did not provide a rating.

Half of the family respondents (50%) indicated a range of other symptoms which they found hard to manage, in addition to those which were prompted on the survey. A total of 11 additional symptoms were identified by family respondents and included visual impairments/blindness, communication impairments, altered mobility, loss of motor capacity, difficulty swallowing/choking, nutrition (feeding and drinking), dementia, ataxia, hearing impairments, incontinence, and diarrhoea. All of these additional symptoms were given a high rating by family respondents indicating their difficulty in managing these symptoms. Communication difficulties were identified by four families and given a rating of 10 (very hard to manage) by each. Table 3 indicates the range of symptoms and difficulties in management.

In one family, the parent of a child with Infantile Batten disease commented that managing symptoms was not in itself difficult. Rather, they reported that the challenge came from the regular change of symptoms due to progression of the condition, for example changes in seizure activity and progressive loss of senses such as vision.

Table 3. Symptom management ratings by family respondents (n=16)

Symptom	High (rated 7-10)	Medium (rated 4-6)	Low (rated 1-3)	None (rated 0)
Pain	4	2	3	7
Nausea/Vomiting	6	2	1	7
Constipation	6	5	4	1
Drooling	8	3	1	4
Muscle Spasms	7	2	3	4
Seizures	9	2	2	3
Breathing difficulties	3	4	4	5
Secretions	6	3	2	5
Sleep Problems	5	5	5	1

Families were invited to describe sources of information or support which help them manage their child's symptoms. Families reported very little support being available, though hospice care staff, community nurses and national disease-specific organisations each provided helpful support and information. Families expressed that due to the rarity of such conditions, information and support was difficult to locate and often accessed indirectly:

I think there is information and support available but experience of the condition is limited because it is rare. Much of the support and information has been passed on second-hand for this condition. (Parent of child with Infantile Batten disease)

Families also described that while support is available to manage certain symptoms, such as medications to manage seizures, other more general symptoms, such as constipation and drooling of saliva, are often left for parents to cope with on their own:

Some medical staff are more dismissive of some symptoms, such as constipation except these can have a real effect on fun/quality-of-life when life is difficult enough. (Parent of child with deletion of part of the long arm of chromosome 6)

3.3 Summary

While a large number of rare syndromes were identified, the three most common were MPS disease, leukodystrophy and Batten disease. All three are progressive metabolic life-limiting conditions, and therefore in group three of the ACT/RCPCH categories (ACT/RCPCH, 2009).

With 59 respondents in total, this is the largest published survey of its kind identifying specific diseases and symptoms which are of most concern to families and care professionals supporting a child with a non-malignant life-limiting condition. A key finding was that families identified non-physical symptoms as particularly troublesome, for example communication impairments and impaired cognition. Symptoms that have previously been reported to be very significant such as pain were much less prominent in this survey. Indeed, nearly half of family respondents (44%) did not report pain to be a significant symptom.

Importantly, the results provide evidence of the requirement for greater understanding and awareness of the symptoms that are experienced by children with rare life-limiting conditions, and the need to centralise the experiences of parents regarding which symptoms are most challenging for them to manage. Improved assessment, management and support of the range of symptoms highlighted in this research are integral to enhance quality-of-life for these families.

Understanding which symptoms were experienced by children with rare life-limiting conditions, and which specific conditions presented challenges, is fundamental to improving healthcare. The survey identified a small number of conditions that formed the basis of the subsequent phases of the research. MPS disease and Batten disease were prioritised to reflect the desire to ensure that parents' views were privileged in the design of the following research, while taking into account the dominance of MPS disease as having the most challenging conditions in clinicians' views.

4 Findings: Prospective longitudinal symptom profile of children with rare life-limiting conditions

Data in this section are drawn from the symptom diaries completed daily by parents and from interviews with both parents and practitioners regarding the experiences of supporting a child with a rare life-limiting condition. As noted in section 2.7, the data are presented in anonymised format, with alphabetical identifiers for each family and practitioner participant. These identifiers are randomly assigned, to prevent matching of symptom descriptions with wider discussions of family impact (discussed in section 5, 6 and 7).

4.1 Participant characteristics

Twenty families kept a symptom diary. Two of these families had two children with the condition and chose to complete a separate symptom diary for each child giving a total of 22 children with symptom diary data. The sample consisted of nine girls (41%) and 13 boys (59%) with a median age of eight years, ranging from three to 21 years.

Thirteen of the 22 (59%) children had Sanfilippo disease. Of these, ten had Sanfilippo Type A, two had Sanfilippo Type B and one had Sanfilippo Type C. One of the 22 (5%) children had MPS IVA Morquio.

Eight of the 22 (36%) children had Batten disease. Of these, six had Infantile and two had Late Infantile forms of the condition.

The median age at time of diagnosis was 3.5 years (range 1.3-8.0 years) for those children with Sanfilippo disease and 2.2 years (range 0.5-6.0 years) for those children with Batten disease. The participant characteristics are shown in Appendix 2.

4.2 Symptom diary completion rates

A total of 1150 diary entries were returned out of a possible 1232 entries, representing an overall return rate of 93%. Of the 22 children for whom symptom diaries were kept, complete diary sets containing data for 56 days were received for 11 (50%) children. The primary stated reason for missing diary data was because the diary was not completed when the child was staying away from home in planned respite/hospice care (54%).

Responsibility for completing the symptom diary was undertaken by 16 mothers (73%) and six fathers (27%). The majority of families chose to complete the paper version of the symptom diary with only one family completing the electronic version. Details on symptom diary completion are outlined in Appendix 3.

In the following sections, the symptom profile for each condition is outlined in turn, with data presented to describe the frequency, severity and how the family managed the symptoms.

4.3 Symptom profile of Sanfilippo disease

Of the 13 children with Sanfilippo disease, six were girls (46%) and seven were boys (54%). Ten children had Sanfilippo Type A, two had Sanfilippo Type B and one had Sanfilippo Type C. One family had twin boys with Sanfilippo Type A. The median age at time of diagnosis was 3.5 years (range 1.3-8 years) and the median age of the children at the time of participation in the symptom diary phase of the study was 12 years (range 5-21 years).

There was significant variation in the children in terms of stage of their disease at the time of participation. Stage of disease was categorised through use of markers such as behavioural symptoms, mobility and type of feeding. Ten (77%) of the children with Sanfilippo disease were

mobile or had partial mobility with occasional use of aids such as a wheelchair. Whilst these children were mobile, some did experience limping, clumsiness, falls and a general 'slowing down', as described by their parents, indicating their mobility was on the decline. Eight of the 13 (62%) children with Sanfilippo disease continue to experience behavioural symptoms such as hyperactivity, aggression and challenging behaviour. Seven (54%) of the children with Sanfilippo disease continue to be fed orally while six (46%) children have a percutaneous endoscopic gastrostomy (PEG) in place.

4.3.1 Symptom frequency

The range of symptoms reported by families of children with Sanfilippo disease is shown in Table 4. Of the 17 symptoms assessed in the symptom diary, all were documented by families as being experienced at some point during the diary period. Pain, cold hands and/or feet, joint stiffness and disturbed sleep were the most commonly reported symptoms, being present in all 13 (100%) children with Sanfilippo disease. Also commonly reported were agitation, repetitive behaviours, secretions and diarrhoea. Coughs and colds, choking, constipation, hyperactivity, aggressive behaviour and breathing difficulties were reported less often, but still by more than half of the families.

Muscles spasms, seizures and vomiting were the least reported symptoms in the diary, and were experienced by six (46%), six (46%) and five (38%) of the children with Sanfilippo disease, respectively.

To determine those symptoms experienced most frequently over the period of diary completion, frequency data for each symptom was categorised in terms of 'low frequency' where the symptom was present for less than 50% of the diary days, or 'high frequency' where the symptom was present for 50% or more of the diary days. As outlined in Table 4, agitation was present in high frequency for 10 of the 12 (83%) children who experienced it. Hyperactivity and repetitive behaviours were also reported in high frequency, being present on 50% or more of the diary days for six out of eight (75%) children and nine out of 12 (75%) children, respectively.

Pain was experienced frequently by children with Sanfilippo disease. Parents were requested to indicate the type or source of pain on any occasion their child experienced this symptom. Responses were received for 12 of the 13 (92%) children, and reported causes of pain varied considerably. The most frequent type of pain experienced was that originating from joints or joint stiffness and occurred in six of the 12 (50%) children. Other types of pain were explained as related to constipation or bowel movements, pain from a fall or accident, pain on movement or positioning the child, ear and throat infections, headache, toothache, and post-operative pain, specifically Grommet insertion.

In addition to the 17 symptoms assessed in the symptom diary, parents were asked to document any further symptoms experienced by their child during the diary period. Eight of the 13 (62%) families listed additional symptom/s which included periods of screaming or loud shouting, limping, inappropriate laughing; dry mouth, skin rash (including nappy rash), lethargy, crying, raised temperature, shaking, and irritation at the PEG site (Table 5).

On the whole, these additional symptoms were documented as occurring infrequently, as detailed in Table 5.

Table 4: Symptom frequency in children with Sanfilippo disease (n=13)

Symptom	Symptom reported in diary n (%)	Symptom reported in high frequency in diary* n (%)
Seizures	6/13 (46)	2/6 (33)
Pain	13/13 (100)	7/13 (54)
Cold Hands/Feet	13/13 (100)	9/13 (69)
Joint Stiffness	13/13 (100)	8/13 (62)
Muscle Spasms	6/13 (46)	2/6 (33)
Disturbed Sleep	13/13 (100)	8/13 (62)
Agitation	12/13 (92)	10/12 (83)
Hyperactivity	8/13 (62)	6/8 (75)
Aggressive Behaviour	8/13 (62)	4/8 (50)
Repetitive Behaviours	12/13 (92)	9/12 (75)
Secretions	12/13 (92)	6/12 (50)
Choking	9/13 (69)	2/9 (22)
Vomiting	5/13 (38)	1/5 (20)
Constipation	9/13 (69)	1/9 (11)
Diarrhoea	12/13 (92)	1/12 (8)
Breathing Difficulties	7/13 (54)	2/7 (29)
Cough/Cold Infections	10/13 (77)	1/10 (10)

Note: *Symptom was present \geq 50% of the diary days.

Table 5: Frequency of the additional symptoms reported in the symptom diaries of children with Sanfilippo disease (n=13)

Symptom	Symptom reported in diary n (%)	Symptom reported in high frequency in diary* n (%)
Periods of screaming or loud shouting	2/13 (15)	1/13 (8)
Limping	4/13 (31)	1/13 (8)
Inappropriate laughing	2/13 (15)	0/13 (0)
Dry mouth	1/13 (8)	0/13 (0)
Rash (including nappy rash)	2/13 (15)	0/13 (0)
Lethargy	2/13 (15)	2/13 (15)
Crying	1/13 (8)	1/13 (8)
Raised Temperature	1/13 (8)	0/13 (0)
Shaking	1/13 (8)	0/13 (0)
Irritation at PEG site	1/13 (8)	0/13 (0)

Note: *Symptom was present \geq 50% of the diary days.

4.3.2 Symptom severity

Severity ratings for each symptom were categorised into two levels: low severity (where the severity of the symptom was rated as not at all, a little, or moderate severity) and high severity (where the severity of the symptom was rated as severe or very severe). Only those symptoms which were rated as severe and/or very severe on more than one occasion were categorised as high severity to allow for isolated incidents where symptoms were experienced in higher severity.

As outlined in Table 6, disturbed sleep, agitation, repetitive behaviours and diarrhoea were rated highest in terms of severity. Hyperactivity, aggressive behaviour and secretions were also rated high

in their severity by 50% of the families whose child/ren experienced them. Seizures and choking were the only symptoms to be rated low on severity throughout. It is noteworthy that the symptoms deemed most severe were largely behavioural rather than physical symptoms.

Table 6: Symptom severity ratings for children with Sanfilippo disease (n=13)

Symptom	Symptom reported in diary n (%)	Reported low* severity n (%)	Reported high** severity n (%)
Seizures	6/13 (46)	6/6 (100)	0/6 (0)
Pain	13/13 (100)	7/13 (54)	6/13 (46)
Cold Hands/Feet	13/13 (100)	7/13 (54)	6/13 (46)
Joint Stiffness	13/13 (100)	11/13 (85)	2/13 (15)
Muscle Spasms	6/13 (46)	5/6 (83)	1/6 (17)
Disturbed Sleep	13/13 (100)	5/13 (38)	8/13 (62)
Agitation	12/13 (92)	4/12 (33)	8/12 (67)
Hyperactivity	8/13 (62)	4/8 (50)	4/8 (50)
Aggressive Behaviour	8/13 (62)	4/8 (50)	4/8 (50)
Repetitive Behaviours	12/13 (92)	5/12 (42)	7/12 (58)
Secretions	12/13 (92)	6/12 (50)	6/12 (50)
Choking	9/13 (69)	9/9 (100)	0/9 (0)
Vomiting	5/13 (38)	4/5 (80)	1/5 (20)
Constipation	9/13 (69)	5/9 (56)	4/9 (44)
Diarrhoea	12/13 (92)	5/12 (42)	7/12 (58)
Breathing Difficulties	7/13 (54)	6/7 (86)	1/7 (14)
Cough/Cold Infections	10/13 (77)	7/10 (70)	3/10 (30)

Note: *severity ratings of not at all, a little or moderate **severity ratings of severe or very severe

Symptoms that were most frequently experienced or reported also tended to be those that were rated highest in terms of their severity (this was the case for disturbed sleep, agitation, repetitive behaviours and diarrhoea). Several symptoms, however, were highly reported in terms of frequency, but rated much lower in terms of severity including pain, cold hands and/or feet, joint stiffness, secretions and cough/cold infections. Figure 2 illustrates the pattern of frequency and severity, with width of each bar representing one symptom and the shading indicating the number of children experiencing low or high severity of the symptom.

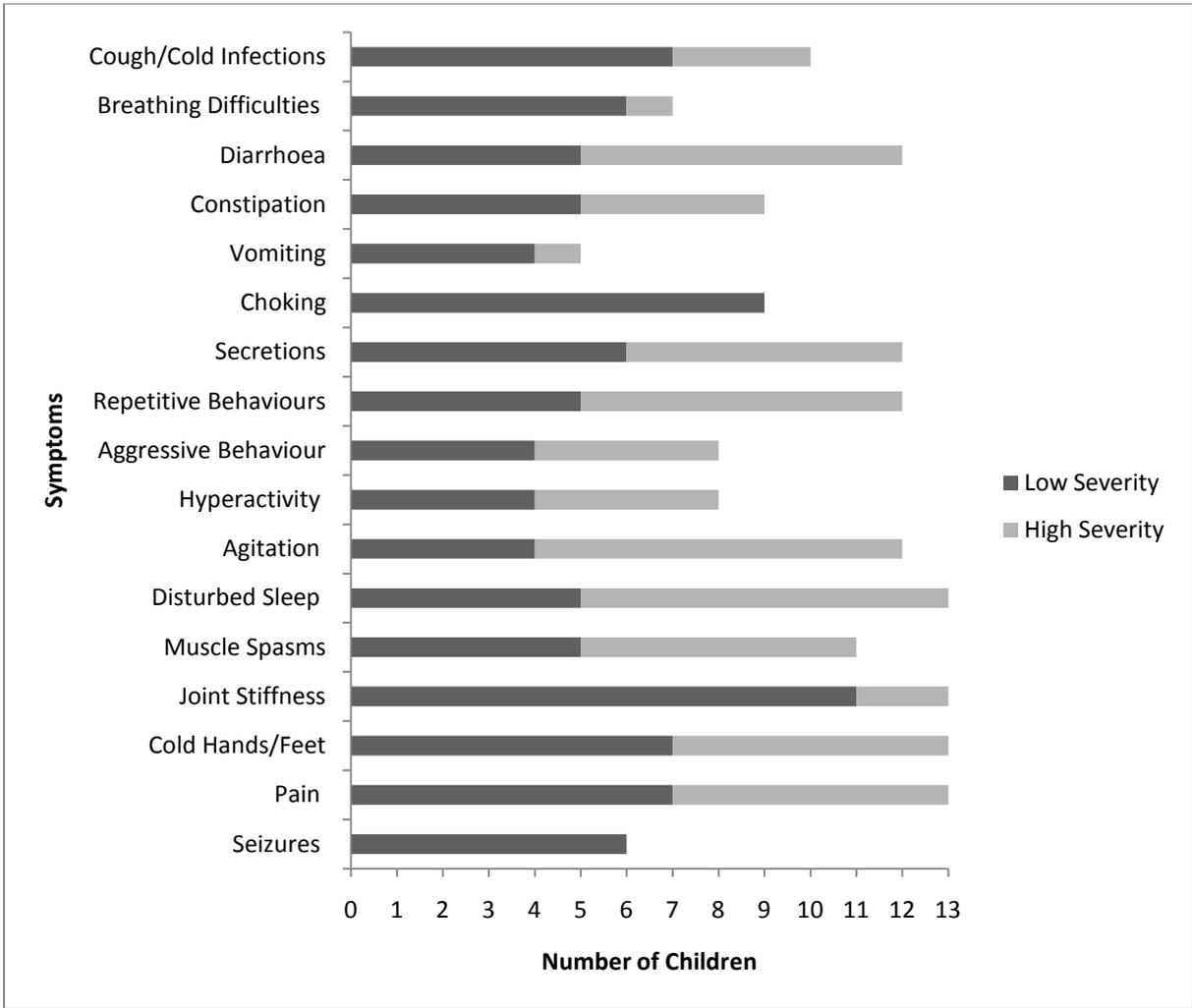


Figure 2: Symptom severity scores for children with Sanfilippo disease (n=13) Note: *Low Severity= severity ratings of not at all, a little or moderate **High Severity = severity ratings of severe or very severe.

4.3.3 Symptom management

Families of children with Sanfilippo disease used the diary to report a range of strategies and interventions to manage symptoms. These strategies were similar across families and are summarised in Table 7.

Table 7: Strategies and interventions used to manage symptoms for children with Sanfilippo disease (n=13)

Symptom	Management strategy
Seizures	Keep child safe during seizure; observe closely; give medication as prescribed; call child's name; take to hospital if needed.
Pain	Give medication as prescribed; minimise movements; change child's position as required; massage; distraction (singing, cuddles); reassurance; hydrotherapy pool.
Cold Hands/Feet	Massage and rubbing hands/feet; warm socks/footwear/gloves; keep the house warm; warm bath; use hot water bottle; put extra clothing on.
Joint Stiffness	Give medication as prescribed; heat and massage; warm bath; change positions and be gentle with movements; hydrotherapy pool; visit osteopath; give child support on movement.
Muscle Spasm	Give medication as prescribed; change child's position frequently; comfort.
Disturbed Sleep	Re-position and make child more comfortable; put child back to bed; put on music and light show; comfort; medication as prescribed (melatonin and/or chloral hydrate); reassurance; low lighting; give child safe toys; no stimulation.
Agitation	Distraction techniques; change activity; give reassurance and comfort; rest; medication as prescribed; change child's position; speak in a soft voice; give child cuddles.
Hyperactivity	Distraction techniques; offer a different activity; give reassurance and try to calm child; give melatonin before bed; outdoor play; singing.
Aggressive Behaviour	Distraction techniques; remove from situation; offer a different activity; give reassurance and try to calm child.
Repetitive Behaviours (chewing)	Give child something appropriate to chew (chewy toys; cotton apron); distraction; offer a different activity.
Secretions	Wear bibs to keep mouth and chin area clean and dry; medication; suction; neckerchief; scopoderm patch as prescribed.
Choking	Re-position; comfort child; pat back and keep child upright; feed slowly; liquidise foods; be with child and observe closely.
Vomiting	Reassurance; sips of liquids; observe and monitor closely to ensure child does not dehydrate.
Constipation	Medication as prescribed; tummy massage.
Diarrhoea	Wash, apply cream, frequent nappy changes; give fluid replacement therapy if diarrhoea is severe.
Breathing Difficulties	Change position; suction.
Cough/Cold Infections	Change position; adjust clothing and bedding; paracetamol, keep comfortable and monitor temperature; rub back, put Vicks Vaporub on chest, medication (decongestant); keep child upright.
Limping	Medication; try to walk less and at a slower pace; take rests, have a wheelchair available.
Laughing (inappropriate)	No interventions stated in the diary.
Dry Mouth	Use spongstick with water.
Skin Rash	Use steroid cream and doublebase; keep skin cool; metanium cream for nappy rash.
Lethargy/Listlessness	Comfort; re-position child; allow/encourage child to rest.
Loud Shouting	Cuddles; give soft toys to chew; distraction; talked to child; re-positioned child.

Table 7: Strategies and interventions used to manage symptoms for children with Sanfilippo disease (n=13) (continued)

Symptom	Management strategy
Crying/Whimpering	Comfort and reassurance.
Raised Temperature	Medication (paracetamol).
Shaking	Medication; change of position.

4.3.4 Effectiveness of Symptom Management Strategies

Families were asked to rate the perceived effectiveness of the symptom management strategies in the daily diary. Specifically, families were asked to indicate how well their actions worked in relieving the symptom using a five-point rating Likert scale from 'not at all' to 'very well'.

The perceived effectiveness data is presented below for those symptoms which were most frequently experienced by children with Sanfilippo disease, and those with higher severity ratings. Consequently, effectiveness is presented in relation to the following symptoms: repetitive behaviours, agitation, disturbed sleep, hyperactivity, aggressive behaviour, secretions and diarrhoea.

The interventions utilised to manage the symptom of diarrhoea appear to be the most effective, with the greatest proportion of responses (74%) being 'moderately well' and 'quite well' as illustrated in Figure 3.

As illustrated in Figure 3, there is a greater spread of responses amongst parental ratings for the management of disturbed sleep. However, the greatest proportion of responses (55%) still centres around 'a little bit' and 'moderately well'.

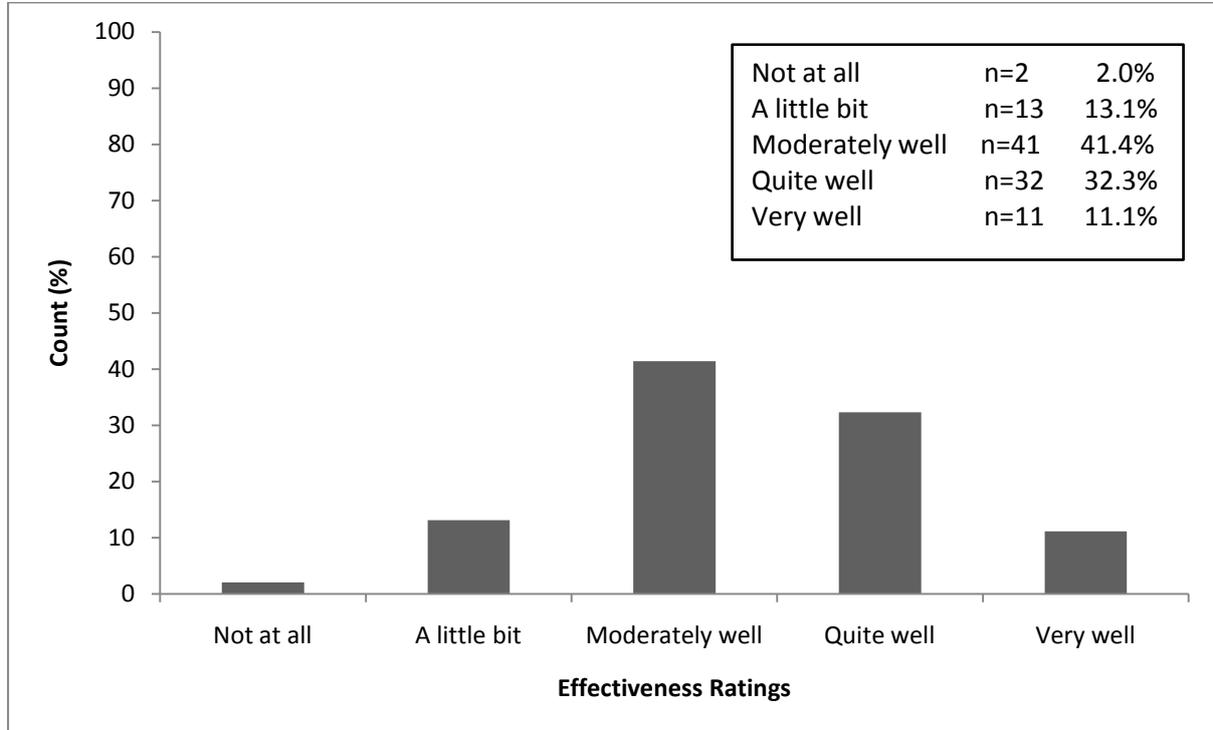


Figure 3: Perceived effectiveness of management of diarrhoea (n=10)

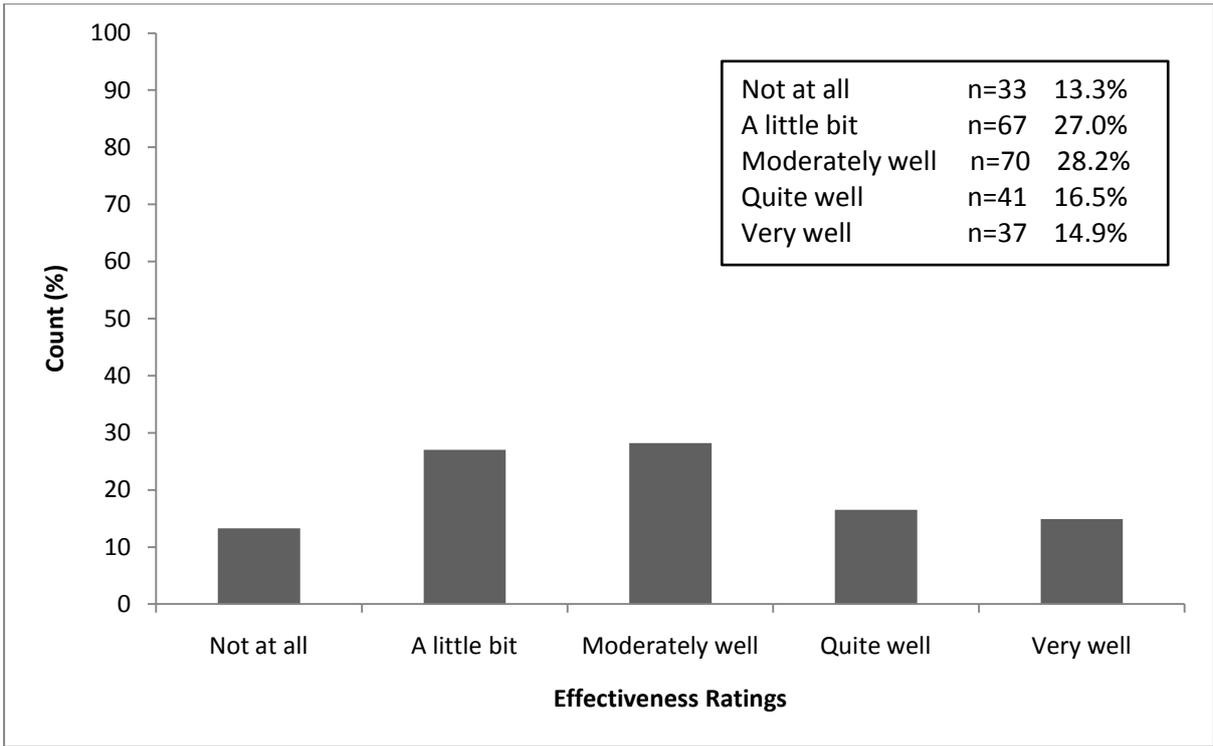


Figure 4: Perceived effectiveness of management of disturbed sleep (n=12)

The data suggest that parents perceive their interventions to manage repetitive behaviours (Figure 5), agitation (Figure 6), hyperactivity (Figure 7), aggressive behaviour (Figure 8) and secretions (Figure 9) as reasonably effective, with the majority of responses being ‘a little bit’ or ‘moderately well’ for greater than half of the responses.

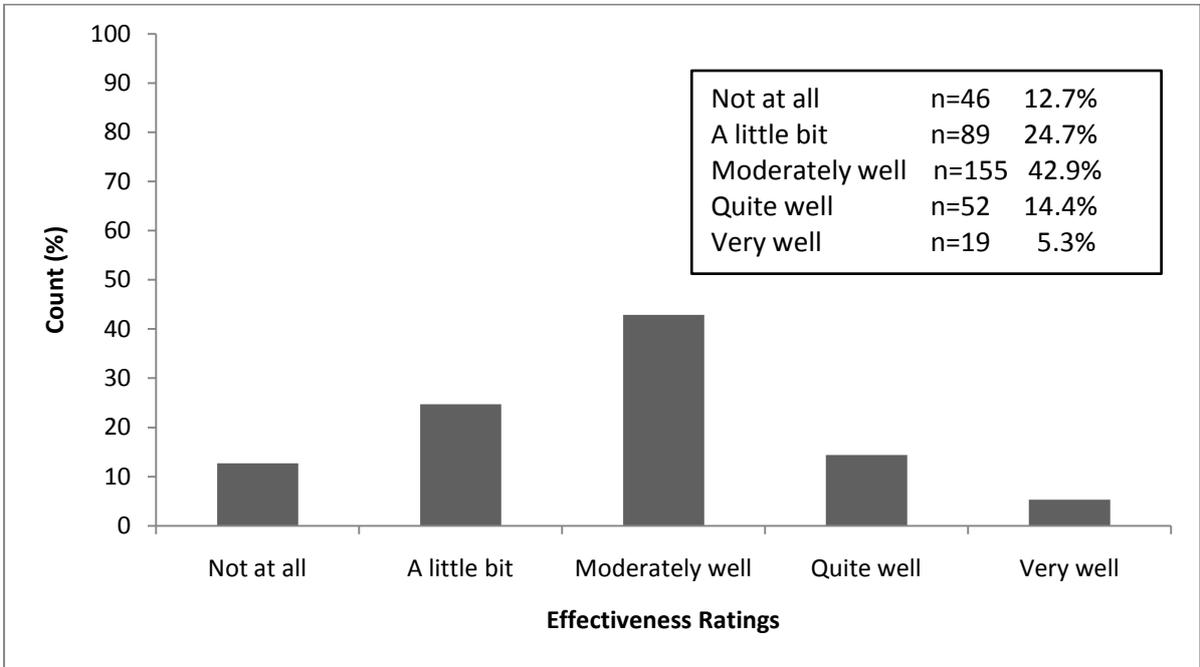


Figure 5. Perceived effectiveness of management of repetitive behaviours (n=10)

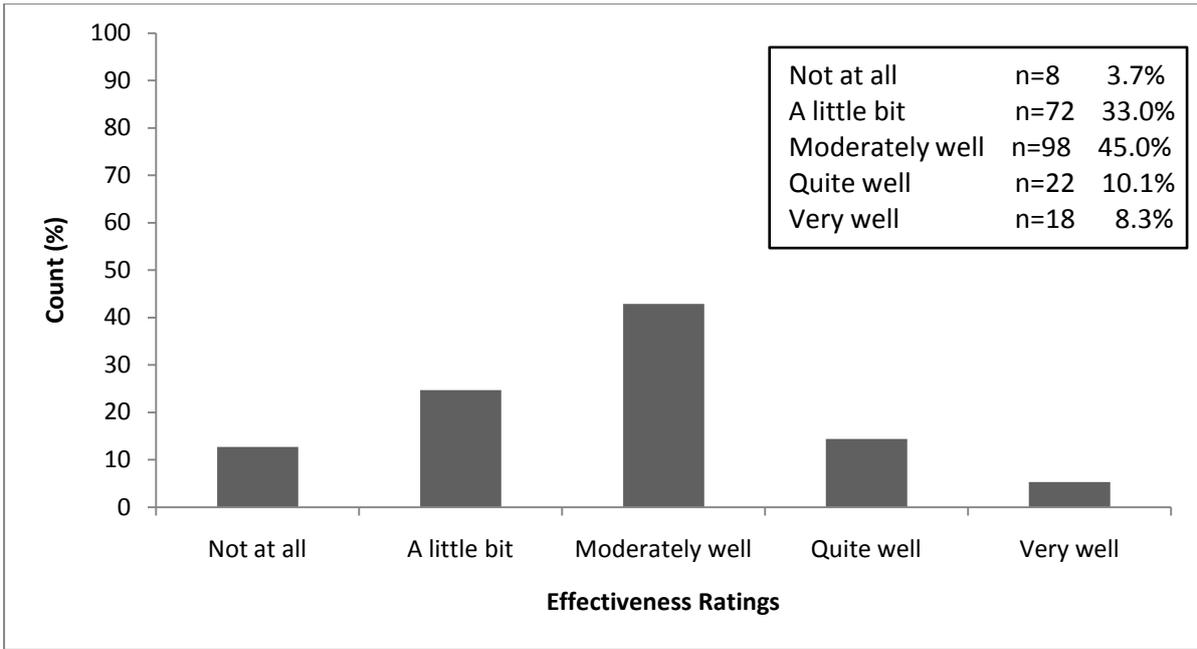


Figure 6: Perceived effectiveness of management of agitation (n=11)

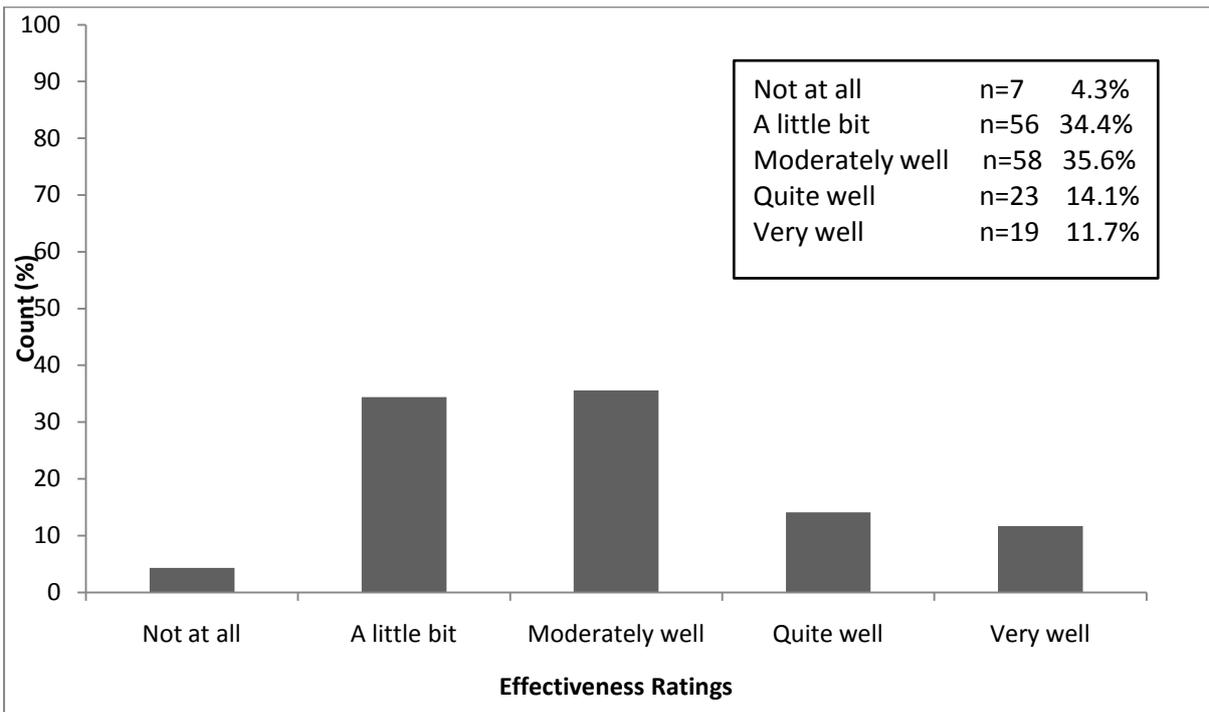


Figure 7: Perceived effectiveness of management of hyperactivity (n=7)

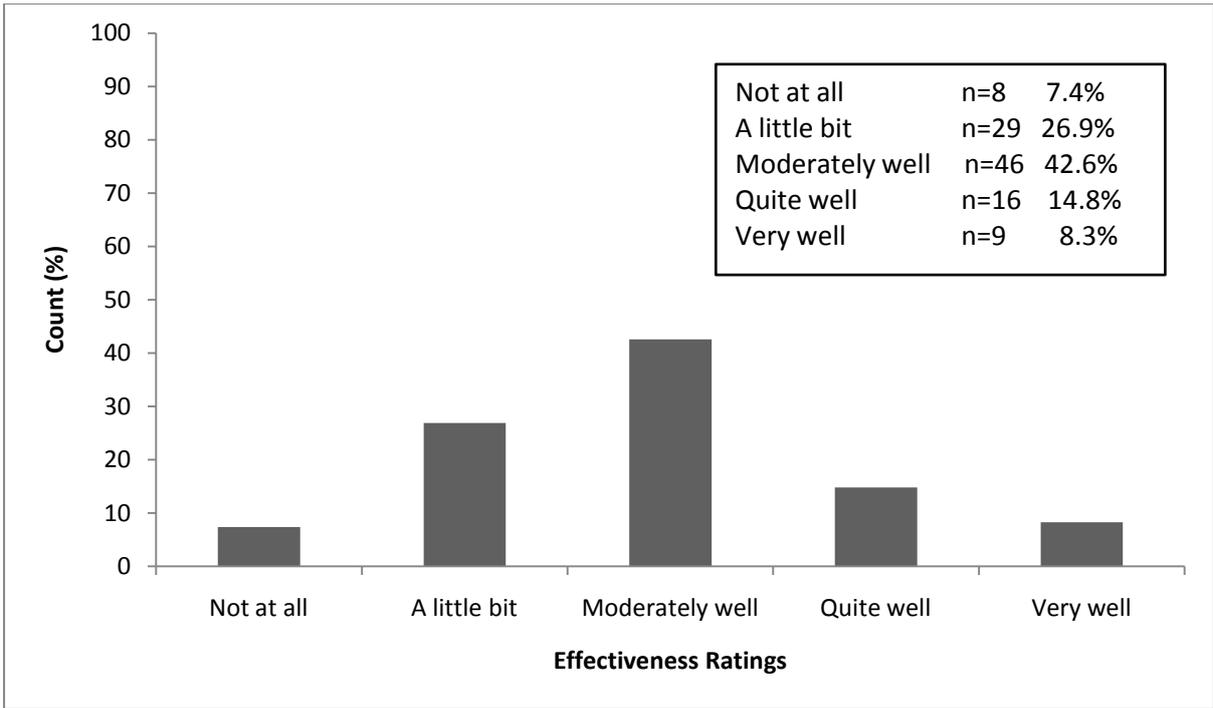


Figure 8: Perceived effectiveness of management of aggressive behaviour (n=6)

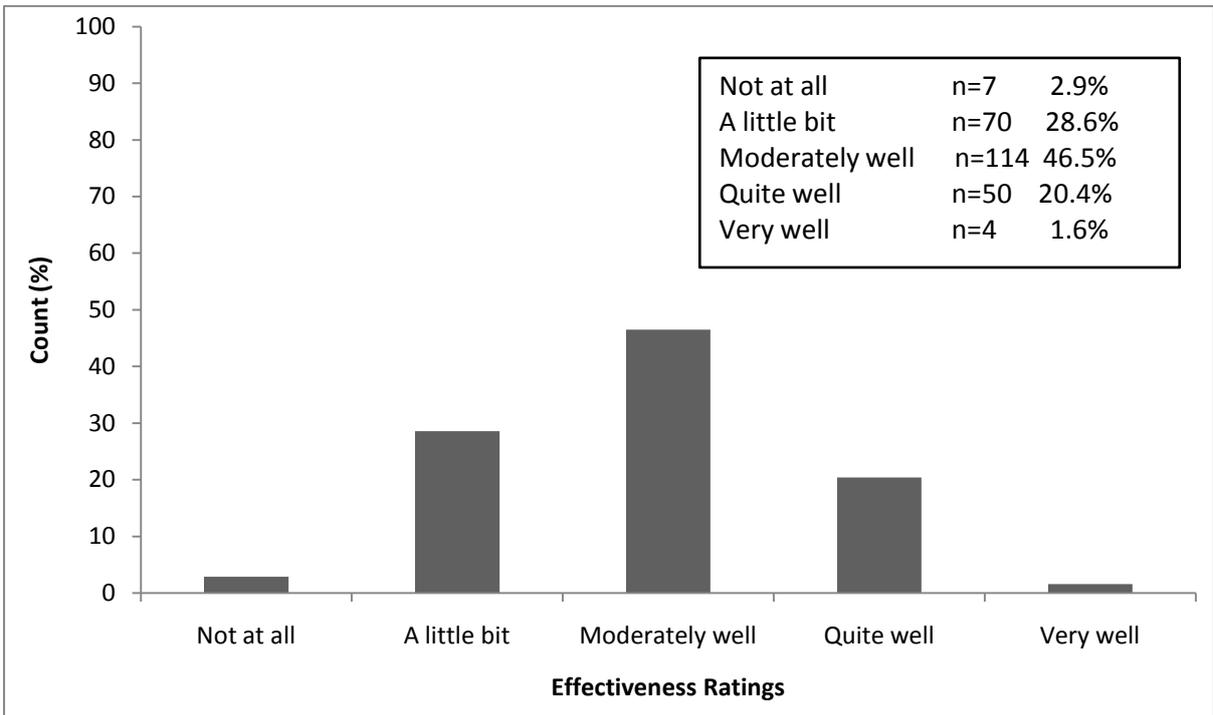


Figure 9: Perceived effectiveness of management of secretions (n=12)

4.3.5 Most Challenging Symptoms

Families were asked at interview to identify which symptoms they perceive to be the most challenging to manage and to offer their rationale as to why. Practitioners were asked the same question, regarding the most challenging symptom to manage from their point of view. Participants' views are summarised in Table 8.

4.3.5.1 Most challenging symptoms for families

Analysis of the interview data indicated three themes for families of children with Sanfilippo disease, which best summarise their perspectives on which symptoms prove most challenging to manage. These themes were: behavioural symptoms associated with condition, symptoms which prove difficult to control or manage and symptoms which signify progression of the condition.

4.3.5.1.1 Behavioural symptoms

The behavioural symptoms associated with the first and second stage of Sanfilippo disease were the symptoms most frequently described by families as being very challenging for them to manage on a day-to-day basis. Families depicted the sheer relentlessness of behavioural symptoms such as aggression and hyperactivity which are a constant feature of their daily lives.

While parents may recognise that their child does not have any control over or an understanding as to why they are behaving so aggressively, parents reported difficulties in accepting the aggression and having to continually remain calm. One father explains a desire to be more tolerant and understanding of his child's behaviour but the reality often being that his tiredness and frustration can interfere causing him to lose his temper and then feel guilty about it after the event:

Dad: I beat myself up a lot, I really do ... I should be more tolerant of them than I am, I know how they are but sometimes just whatever the situation is I'm tired or I'm frustrated because they are not doing something.

Mum: Yeah, but you are a lot better than you were.

Dad: Oh yeah, but I still beat myself up, especially after the event, you know I think why didn't you do that or why did you do that or, you know, just take those extra few seconds, take a deep breath or whatever the case is.

Mum: Count to ten.

Dad: Yeah. (Family V)

As described below, another parent also acknowledges the feelings of frustration and anger that are associated with having an aggressive child but goes on to depict what it feels like to have your own child physically beat you:

One of my doctors actually said to me 'How do you feel when she beats you up?' and do you know what, she just put it into perspective ... how would anybody feel when their child beat them up? You know, because you get frustrated, because you know she doesn't understand she's doing it, and I'm not going to lie to you, you get angry at times, because it hurts you know and it's a natural feeling, sometimes she'll just catch you unaware, or it will be ongoing, you might have a day where she's particularly aggressive towards you. I had about three weeks of it where ... and that's been quite recent, where [child] just ... it must have just been, 'Oh Mum, I'm going to be aggressive to you,' and it just seemed like I was the only one that was getting it and it was ongoing and by the end of the three weeks you were thinking ... 'How much longer can I put up with this?', you were just at your wits' end, just thinking every time I go near you, even to put socks on or anything, she'd lash out, she'd have your hair and was pulling me, trying to bite me at the same time ... that's the most challenging. (Mum, Family J)

The difficulties encountered by parents in dealing with their child's behavioural issues resonate throughout the family. Siblings are directly affected and often bear the brunt of the child's aggression. One parent describes the emotional impact felt in observing siblings being the receiver of their child's physical aggression and the need to protect other children in the family from both the physical and emotional effects of this behaviour:

From my perspective it's the aggression, I find that really hard ... to stay calm when he's aggressive and he hits out ... and particularly if he hits out at [sibling], I find that the hardest to manage emotionally ... I find it upsetting when he hits me or hits somebody else ... or other children and I do find that affects me ... the anxiety I can manage and I sort of manage it again by distracting him or ... explaining it in a very simple way, I mean he will say things over and over again and I just repeat it back to him over and over again until he sort of gets it in his head. When he is anxious and stressed he can't take stuff on so you have to give him time to process it and ... rather than having a battle and trying to force him to do something and physically making him do it, I allow him a bit of space to or I'll say to him, 'Right, five more minutes and then it's bath time.' (Mum, Family W)

Moreover, where there is more than one child with Sanfilippo disease within a family, there is the added challenge of trying to prevent the escalation of one child's aggression in order to avoid triggering aggressive behaviour in the second child:

Dad: Sleep is quite tough but then it's not all the time, but the biggest thing for me is the communication and the behavioural because when [child] does lose it, he loses it and he can get really aggressive.
Interviewer: Can you talk me through what it's like for you when he is aggressive?
Dad: It's really tough because you are trying to keep calm yourself and you are always thinking, right calm [child] down, because of [sibling] as well ... so you know, although you want to keep [child] calm for his point of view, because it can't be nice for him to be like that, and obviously you are wary of getting hit and things like that, you are also thinking of [sibling] and then [sibling] starts and they set each other off even more. And for me that's really tough and I find that difficult at times. (Family V)

An issue which permeated the interviews was the importance of being attuned to the various triggers of a child's aggression. Often symptoms such as agitation and pain would escalate and, if left unattended, would result in aggressive behaviour. In managing behavioural symptoms such as aggression and hyperactivity in children with Sanfilippo disease, parents suggest the use of distraction as a key strategy:

Mum: She'll, you know, when she's ready to ... it starts off with her scratching her head and ... being uncooperative if that makes sense. If you're asking her to do something she'll start showing signs of being stubborn ... and if she starts wanting to scrub things and, so you know that she's working up to ...

Interviewer: So do you try and dampen that down as it starts?

Mum: Yes.

Interviewer: Does that work, if you ...?

Mum: It doesn't work all the time, it doesn't work all the time, but the majority ... we've, we've got a knack now of trying to get it before it gets too far, because if it gets too far, then you've lost it, do you know?

Interviewer: And what happens when you lose it?

Mum: The aggressiveness starts with her ... she starts hitting, punching at me ... we haven't had any biting episodes lately but that used to be or ... she'll kick. (Family P)

While distraction has earned its place as a useful intervention for parents in managing aggressive behaviour and hyperactivity in children with Sanfilippo disease, the relentlessness of these symptoms is apparent as described in the extract below. The parent describes that, over time, once the use of distraction has been exhausted, parents are left to devise other strategies for avoiding or managing behavioural symptoms:

You will see that the main symptoms of [child's] illness that causes family life to be disrupted is due to behaviour, aggression and hyperactivity ... for that day [child] just didn't stop from the second he woke up and I think it's always worse when the weather is bad because we're restricted to this room ... I try changing toys around, things like that, to occupy him but it works for five minutes if you're lucky and then he is off again ... but yeah, so there are days [child] can come in from school, be all excited saying 'Hiya, hiya,' and then from nowhere there will be a big bang ... [sibling] will be lying on the floor and [child] will be hitting him round the head with the remote control that he found and it's not just once, it's repeatedly. The other day I bent down to deal with [sibling] and [child] had managed to get hold of a fork and he just started stabbing me in the head with it ... things like that and it's just, I've learnt, and [partner's] learnt, we've both learnt, that he doesn't get what he's doing, he's not doing it to be nasty ... it's just something to do so you have to try and keep your cool constantly ... and it's a case of 'Oh [child], look, what can we find?' and quite often singing gets [child's] attention a lot, if you start singing ... he will look around and that will draw his attention away and that works quite a lot ... or we say to him 'Where's the ball?' and he will start looking ... and it's constant distracting him, but there is only so many things you can distract him with and then you run out. (Mum, Family G)

4.3.5.1.2 Symptoms which prove difficult to control or manage

Some symptoms, specifically those of pain, spasm, seizures and breathing difficulties are challenging because parents find it difficult to effectively manage them. This causes anxiety in parents having to

observe their child experience such distressing symptoms and descriptions of not feeling able to effectively cope with the symptoms:

The pain ... the, the, the distress, yeah ... 'cause it's constant and I don't feel we get, are ever on top of it, I don't think we're ... 'in control'. (Dad, Family K)

The quotation below illustrates that while a parent may be able to learn to manage the symptom to a certain degree, the worry and distress for this particular parent is associated with the lack of complete control over the symptom. The worry that this symptom brings leads this parent to seek medical attention for her child when the symptom has reached a level where the parent feels that she can no longer manage:

For me it's the fits. I hate seeing it's out of my control ... that's the one thing. I can deal with everything else ... but I hate the fits. That's why, I have got a bottle of Midazolam and stuff that I can give [child], see how ... you know how it goes after a certain amount of time but ... I'm just not good with it, so I just 999 at the moment. I've only had to do it once, if it's a small fit I can deal with it ... and he has little time giggle fits and things like that. (Mum, Family Q)

Moreover, families reported that breathing difficulties for the child is difficult for the parents because of the helplessness they feel while watching their children when they experience this symptom. The parent below describes how she worries about this for herself and also for her child:

The worrying thing I think of is the, the breathing, and that really upsets me, if the child can't breathe properly, and she's got mucus on the chest already when she lays in bed, that's, that's the worry I find for [child], you know, you just can't help her 'cause there's nothing ... (Mum, Family O)

4.3.5.1.3 Symptoms which signify progression of the condition

As the condition progresses over time, children lose key abilities and skills. Each time this happens, it triggers recognition that the condition has progressed and the life-limiting nature of the condition once again becomes very apparent. As explained by the parents below, the decline of a child's mobility brings about this realisation of the condition progressing and a sense of grief:

When she goes totally off her feet that will be the real clincher for me because it's been me that has worked with her to stay on her feet as long as she has ... nobody else can walk her the way I walk and I don't like anybody saying 'Can I take the other side?', 'cause that throws me all off, her [child] and I ... we have our own means of doing it and she is comfortable with how I ... she is confident with me em, on how I walk her, it's, it's a huge struggle at the minute it really is ... but I won't give in. I know when she goes off her feet then we really are on a big countdown, and it's me that just keeps pushing and pushing, but I notice her wee legs, her knees are, they're bowing, they're coming out here and her wee feet are turning quite a bit and I would say if she's still anyway able to walk by December I think I'll be very lucky. But that'll be a real, that'll be a real hard one for me to cope with. (Mum, Family R)

I would say it's the sort of general deterioration, the sort of, the, the joint stiffness and stuff which is really hard for [child] because it's making him so immobile and making it so difficult for him to move around ... [child] wants to do things, I think he gets frightened, he's scared, yeah. He'll grab hold of you and hold you tight and he, and he trembles. Mmm, and that's very sad, really hard ... when you're used to this little thing that's quite confident and can run around and, and things, and now he can barely stand up, so it's not nice seeing him afraid. (Mum, Family E)

4.3.5.2 Most Challenging Symptoms for Practitioners

The symptoms discussed within the practitioner interviews as being those which are most challenging to manage had obvious similarity to views expressed by families. One distinctive area which emerged in the practitioner interviews, though, was that of the difficulties in assessing symptoms such as pain or distress due to communication impairments linked to the condition.

4.3.5.2.1 Behavioural symptoms

The behavioural symptoms associated with Sanfilippo disease are challenging for practitioners in a number of ways. Being one of the most significant and prevalent symptoms experienced by children with the condition, practitioners from a variety of domains will be required to support families in coping with and addressing the symptom. As explained by the practitioners below, this proves difficult as there are few interventions which successfully address hyperactivity and aggressive behaviour in this particular population of children:

Really it's been seeking information really, we've sort of involved educational psychologists in there because we didn't know whether we should be trying to modify [child's] behaviour in some way by doing things in a certain way ... so we've spoken with local educational psychologists and they really feel that they haven't got the knowledge ... you go to the MPS society again and again, they're helpful but don't have the answers. (Practitioner O)

Somebody who has extreme hyperactivity is probably the worst symptom because there's just nothing you can do to support them. (Practitioner, H)

The behavioural symptoms may have consequences on the wellbeing of the variety of practitioners and carers who are involved in the care of these children due to the physical and mental demands of managing the symptom:

The behaviour, it's really because it affects everyone who comes into contact with [child] and really because if it's out of control, it could compromise placement in school or with respite carers and then there's obviously safety issues for the child and staff and I think the affect really it has on the staff wellbeing, it's quite exhausting physically and mentally. (Practitioner O)

4.3.5.2.2 Symptoms which prove difficult to control or manage

Seizures were described as one of the chief symptoms which prove difficult for practitioners to control, due largely to their unpredictable nature within the course of this life-limiting condition. Seizure management requires careful and frequent assessment and adjustment of medications. As

described by the following practitioner, there is a requirement to be attuned to the subtle changes in the pattern of seizures:

Because it's just ... it's just ... it's, you know, it's the changes, you know, when you're dealing with you know, symptoms that are ongoing and ... you just manage them, but when the seizure pattern starts to change as it has, quite a few times, then that's when you just get a wee bit kind of you know, this isn't ... this isn't the way [child] normally is. (Practitioner N)

4.3.5.2.3 Difficulties in assessing symptoms due to communication impairments

Children with Sanfilippo disease often have problems with communication, which makes the assessment and identification of symptoms difficult. Practitioners have concerns related to assessing symptoms such as pain, noting that standardised paediatric pain assessment tools are not appropriate to use in children with Sanfilippo disease. There are often concerns that a child's aggressive behaviours may be a result of pain they are experiencing but not able to communicate to others:

We're unsure whether she's sore because she's unable to communicate that, so that's always ... we found difficult, and then sort of the assessment of that pain, can't find at all that really helps us, really to ... to know whether she is or isn't in pain. The problem is you know, you can have your smiley faces and these types of things [pain assessment tools] but even that doesn't work for her because they don't always sort of you know, physically ... we wonder whether maybe some of these aggressive outbursts or biting and pinching and these types of things are because somebody's doing something wrong and it's hurting or whether she herself is hurting, so it's difficult and it's very much trial and error. (Practitioner O)

Subsequently, the practitioner below states of the position of helplessness in terms of managing the symptom due to the lack of communication:

Yes, and I suppose that whenever [child] would take the crying spells, and often does, you feel a bit lost to help her, and [child] can't tell you what's the problem or anything like that. (Practitioner L)

The following practitioner alludes to the difficulties in assessing the pain which the child experiences due to not being able to communicate with the child:

I think the pain relief. Purely because it's difficult to know where the pain is ... [Child's] got quite a high pain threshold so when he does cry he is really in pain, and it's us getting to the bottom of what it is really and relieving it for him. (Practitioner R)

4.3.6 Summary

All 17 symptoms assessed in the symptom diary were reported by the families of children with Sanfilippo disease as being present. Of these, those present in highest frequency included agitation, hyperactivity and repetitive behaviours. Those symptoms which were rated as being most severe in

children with Sanfilippo disease included agitation, sleep disturbances, repetitive behaviours and diarrhoea.

There was significant overlap between the views of families and practitioners regarding symptoms which present the greatest challenge to manage. Behavioural issues, such as aggression and hyperactivity, which are symptomatic of Sanfilippo disease, were prioritised by families and practitioners in this study as symptoms which pose considerable challenges in their management. The relentless nature of such symptoms results in physical and mental exhaustion and the wearing down of those who care for and live with these children. Parents also spoke of the emotional challenges of managing symptoms that were indicative of the disease progressing.

Table 8. Most challenging symptoms to manage in children with Sanfilippo disease from the perspective of families and practitioners (n=13)

Child	Symptoms Occurring Most Frequently*	Symptoms with Highest Severity Ratings*	Families' views on the most challenging symptom to manage	Practitioners' views on the most challenging symptom to manage
J	Repetitive behaviours (chewing)	Repetitive behaviours (chewing)	Aggressive behaviour because it is often directed towards the parents and they are being physically attacked by their own child. The daily management of this symptom is emotionally and physically draining for the family.	Aggressive behaviour and assessing pain in a child with communication impairments are the most challenging symptoms for the practitioner.
K	Pain and secretions	Pain and agitation	Pain and distress as the parents feel that pain is never a constant in the child's life and they are never completely in control of this symptom.	Seizures because of their unpredictability.
R	Repetitive behaviours	Repetitive behaviours	Jerky movements as the parents want to make sure their child is safe but it is becoming more difficult to physically protect their child as much as they used to as the force and frequency of the movements is increasing. The loss of the child's mobility is emotionally difficult for the parents as this loss of skill signals a progression of the condition.	Loss of child's communication skills and being able to tell people what is wrong. The practitioner mentioned the child's frequent crying episodes and the challenges in not being able to say what it is that is bothering her.
D	Muscle spasms, joint stiffness and repetitive behaviours	Disturbed sleep	Challenging behaviours as the child is very energetic and the behaviours are difficult to control.	No Practitioner Interview
T	Joint stiffness	Cold hands and/or feet and diarrhoea	Loss of mobility is challenging as the child needs to be hoisted, lifted and transferred which takes a lot of time and energy for the parents.	Seizures because of the difficulties in trying to alter medication doses to control them.
E	Joint stiffness, disturbed sleep, secretions and repetitive behaviours	Joint stiffness	Sleeping difficulties as disturbed and lack of sleep impacts on the parents. Loss of mobility because the parents find it very difficult and distressing to observe the frustration and fear in their child as they experience a deterioration in their mobility.	Loss of mobility (no rationale provided in interview).

Child	Symptoms Occurring Most Frequently*	Symptoms with Highest Severity Ratings*	Families' views on the most challenging symptom to manage	Practitioners' views on the most challenging symptom to manage
O	Joint stiffness and secretions	Repetitive behaviours and secretions	Breathing difficulties because of the helplessness parents feel watching their child struggle with clearing their airway.	Periods of agitation and distress are challenging because it is difficult to determine the cause and how to deal with it. The practitioner also noted that each new symptom becomes a challenge when it initially presents.
V	Disturbed sleep, hyperactivity, repetitive behaviours and secretions	Disturbed sleep	Impaired communication because of the challenges in trying to determine what the child needs or having to guess what is wrong with the child when they are obviously upset but not able to describe what it is that is bothering them.	Assessing pain and being able to determine the source of pain is a constant challenge due to the child's communication impairments.
V	Disturbed sleep, joint stiffness and secretions	Agitation and aggressive behaviour	Aggressive behaviour because the parents are being physically attacked by their child and there is nothing they can do about it.	Extreme hyperactivity because there is very little the practitioner can do to support the child and family in managing this symptom.
P	Agitation and cold hands and/or feet	Cold hands and/or feet	Agitation because the parents are constantly trying to avoid any triggers that activate the child's agitation. This is physically and emotionally exhausting for the parents.	Extreme hyperactivity because there is little you can do to support the family in managing this symptom.
G	Aggressive behaviour, hyperactivity and agitation	Aggressive Behaviour	Aggressive behaviour because the parents are required to separate the child from the other siblings and this is emotionally challenging for the entire family.	No Practitioner Interview
W	Aggressive behaviour, hyperactivity and agitation	Cough/Cold Infection	Aggressive behaviour because the parents find it very difficult to remain calm when the child is being aggressive, in particular towards siblings.	Aggressive behaviour because of its constant nature and the fact that there are few interventions available to successfully manage such behavioural issues.
Q	Pain, cold hands and/or feet and repetitive behaviours	Repetitive Behaviours	Seizures because the parents find this a very difficult symptom to control.	Pain assessment because it is difficult to know if the child is in pain. Vomiting because it can lead to aspiration and infections.

NOTE: *Data derived from the symptom diary.

4.4 Symptom profile of Morquio disease

There was one child, a boy aged eight years, with Morquio disease, participating in the study. The child was one year old when diagnosed.

4.4.1 Symptom frequency

The range of symptoms reported by the family is shown in Table 9. Of the 17 symptoms assessed in the symptom diary, eight were documented by the family as being experienced at some point during the diary period and included pain, cold hands and/or feet, joint stiffness, muscle spasms, vomiting, diarrhoea, breathing difficulties and cough/cold infections.

To determine those symptoms experienced most frequently over the entire symptom diary period, frequency data for each symptom was categorised in terms of low frequency where the symptom was present for less than 50% of the diary days or high frequency where the symptom was present for 50% or more of the diary days. As outlined in Table 9, breathing difficulties was the only symptom present in high frequency for this child with Morquio disease and was reported on all 56 (100%) of the diary days.

The symptom of pain was experienced by this child with Morquio disease. Parents were requested to indicate the type or source of pain on any occasion their child experienced this symptom. The type of pain experienced by this child included pain related to sitting in the same position for an extended period of time; stomach pain; pain in the neck area; and pain arising during physiotherapy leg exercises.

In addition to the 17 symptoms recorded in the diary, parents were asked to document any further symptoms experienced by their child during the diary period (Table 10). Two additional symptoms were recorded for this child and included dry skin and catarrh (Table 10).

Table 9. Symptom frequency in child with Morquio disease (n=1).

Symptom	Symptom reported in diary (yes/no)	Symptom reported in high frequency in diary* (yes/no)
Seizures	No	N/A
Pain	Yes	No
Cold Hands/Feet	Yes	No
Joint Stiffness	Yes	No
Muscle Spasms	Yes	No
Disturbed Sleep	No	N/A
Agitation	No	N/A
Hyperactivity	No	N/A
Aggressive Behaviour	No	N/A
Repetitive Behaviours	No	N/A
Secretions	No	N/A
Choking	No	N/A
Vomiting	Yes	No
Constipation	No	N/A
Diarrhoea	Yes	No
Breathing Difficulties	Yes	Yes
Cough/Cold Infections	Yes	No

Note: *Symptom was present \geq 50% of the diary days.

Table 10. Frequency of the additional symptoms reported in the Symptom Diaries of child with Morquio disease (n=1)

Symptom	Symptom reported in diary (yes/no)	Symptom reported in high frequency in diary* (yes/no)
Dry skin	Yes	Yes
Catarrh	Yes	No

Note: *Symptom was present \geq 50% of the diary days.

4.4.2 Symptom severity

All of the symptoms experienced during the diary period were given low severity ratings by the family of the child with Morquio disease.

Table 11. Symptom severity ratings for child with Morquio disease (n=1)

Symptom	Severity rating*
Pain	Low
Cold Hands/Feet	Low
Joint Stiffness	Low
Muscle Spasms	Low
Vomiting	Low
Diarrhoea	Low
Breathing Difficulties	Low
Cough/Cold Infections	Low

Note: * low = severity ratings of not at all, a little or moderate; high - severity ratings of severe or very severe

4.4.3 Symptom management

Within the symptom diary, the family of the child with Morquio disease reported the range of strategies and interventions used to manage their child's symptoms. These strategies are summarised in Table 12.

Table 12. Strategies and interventions used by family to manage symptoms for child with Morquio disease as recorded in the symptom diary (n=1)

Symptom	Management strategies
Pain	Give medication as prescribed, massage, encourage child to relax, monitor and assess pain frequently.
Cold Hands/Feet	Monitor symptom.
Joint Stiffness	No interventions stated in the diary.
Muscle Spasm	Monitor symptom, massage.
Vomiting	Massage, fan to cool child.
Diarrhoea	No interventions stated in diary.
Breathing Difficulties	Change position; fan to cool child; monitor symptom.
Cough/Cold Infections	Medications (Calpol and cough medicine).
Dry skin	Use emollient cream.
Catarrh	Medication as prescribed.

4.4.4 Most challenging symptoms

During the interview, the family was asked to identify which symptoms they perceive to be the most challenging to manage and to offer their rationale as to why. The practitioner nominated by this family to participate in the project, was also asked, during their interview, to consider which symptoms related to Morquio disease are the most challenging to manage from their point of view.

Table 13. Most challenging symptoms to manage in child with Morquio disease from the perspective of the family and practitioner (n=1)

Symptoms occurring most frequently	Symptoms with highest severity score	Family's views on the most challenging symptom to manage	Practitioner's views on the most challenging symptom to manage
Breathing difficulties	Breathing difficulties	Cough/cold infections because of the worry associated with viral infections and the worry that they can lead to more serious infections and exacerbate any breathing difficulties .	Keeping the child 'healthy' to try to avoid viral (cough/cold) infections and prevent subsequent breathing difficulties .

NOTE: *Data derived from the symptom diary.

For this family, the most challenging symptom to manage was the frequent viral (cough/cold) infections their child experienced. The parents described being worried any time their child had an infection as they were aware it could lead to other serious and life-threatening conditions:

I don't like it, any, anything bad in respect to like vomit or a cold or the flu ... I, we get worried because [child] tends to get infections really, really quick and then they stay for longer and sometimes you know, you know hear, you hear some of the stories, from a, the doctors that it can just be the infection, you know, that can lead to other, other serious things which is, which is the one thing that scares me. (Dad, Family N)

The same symptom was identified by the practitioner, who described the importance of maintaining good health in this child to avoid exacerbation of his breathing difficulties.

4.4.5 Effectiveness of symptom management strategies

Families were asked to use the symptom diary to rate the perceived effectiveness of symptom management strategies. Specifically, families were asked to indicate how well their actions worked in relieving the symptom, using a five-point Likert scale from 'not at all' to 'very well'.

The perceived effectiveness data is presented for those symptoms which were most frequently experienced and those with higher severity ratings. Consequently the effectiveness is presented in relation to the symptom of breathing difficulties for the family with experience of Morquio disease.

Figure 10 suggests that parents perceive their interventions to manage their child's breathing difficulties as satisfactory. Sixty-eight percent of instances were recorded as working 'moderately well' in managing this symptom.

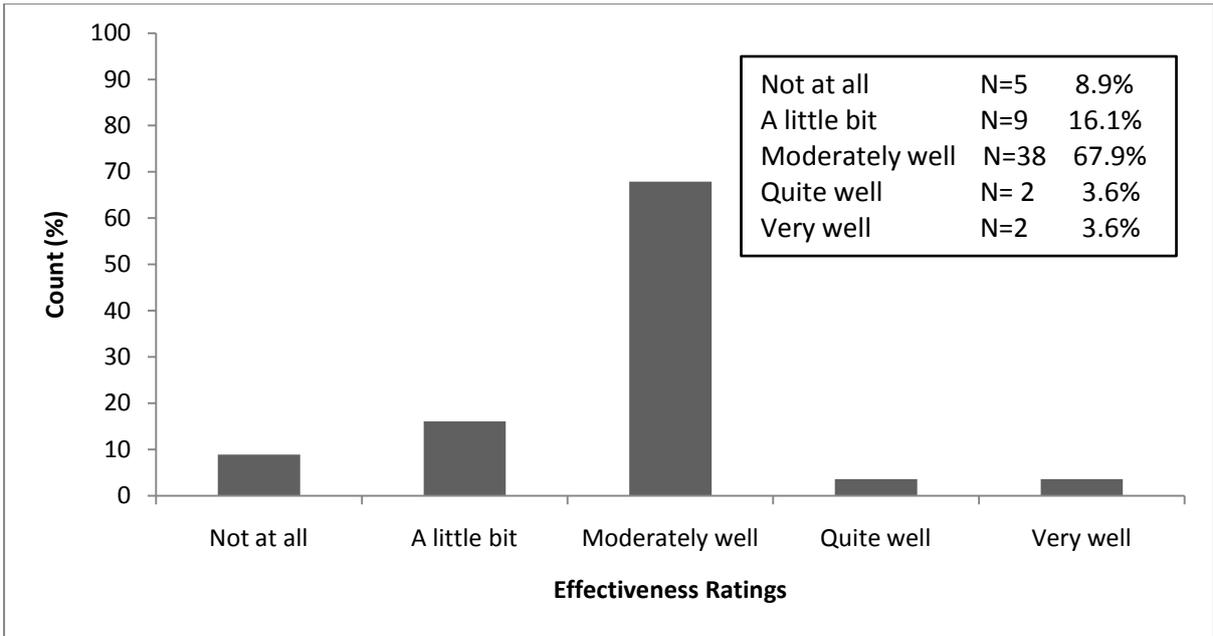


Figure 10: Perceived effectiveness of management of breathing difficulties (n=1)

4.4.6 Summary

The diary records and interview for the child with Morquio disease revealed that the symptoms of most concern to the family and practitioner revolve around the problems that viral infections can cause for this child. Both parents and practitioner spoke about their efforts to promote health and prevent infections which may lead to and exacerbate breathing difficulties.

4.5 Symptom profile of Batten disease

Of the eight children with Batten disease, four were girls (50%) and four were boys (50%). Six children had Infantile Batten disease and two children had Late Onset Infantile Batten disease. One family had twin boys both with the condition. The median age at time of diagnosis was 2.2 years (range 0.5-6 years) and the median age of the children at the time of participation in the symptom diary phase of the study was 4.7 years (range 2.5-12 years).

As Batten disease is a progressive degenerative condition, there was significant variation in the children in terms of stage of their disease at the time of participation in this study. In order to assist in categorising stage of disease, markers such as communication impairments, mobility and type of feeding were used. All of the children with Batten disease were visually impaired and/or registered blind and had verbal communication impairments. Two (25%) of the children with Batten disease were mobile or had partial mobility with occasional use of aids such as a wheelchair and the remaining six (75%) were completely immobile and relied on proper positioning and movement with aids. Four (50%) of the children with Batten disease continue to be fed orally whilst the other four (50%) children have a PEG in place. Four of the children were considered to be in the advanced stages of Batten disease by their parents and nominated practitioners or carers.

4.5.1 Symptom frequency

The range of symptoms reported in the diary is presented in Table 14. All but one of the 17 symptoms assessed in the diary were documented by families as being experienced at some point during the diary period (with the exception of aggressive behaviour). Pain and disturbed sleep were the most commonly reported, being present in the eight (100%) children with Batten disease. Also commonly reported were agitation, cold hands and/or feet, constipation and cough and cold infections. Seizures, joint stiffness, muscle spasms, secretions, diarrhoea and breathing difficulties were reported less often but still by half of the families or greater.

Choking, vomiting, repetitive behaviours and hyperactivity were the least reported symptoms in the diary, experienced by three (38%), two (25%), two (25%) and two (25%) of the children with Batten disease, respectively. It is noteworthy to highlight that the two children experiencing the symptoms of repetitive behaviours and hyperactivity had an additional diagnosis of autism and it is possible such symptoms were a manifestation of this condition rather than Batten disease.

Aggressive behaviour was not reported in the symptom diary for children with Batten disease. This symptom is common in Sanfilippo disease and was included as a core symptom in the diary for that reason, but it is not surprising it did not appear in children with Batten disease.

To determine symptoms experienced most frequently over the entire diary completion period, frequency data for each symptom was categorised in terms of low frequency where the symptom was present for less than 50% of the diary days or high frequency where the symptom was present for 50% or more of the diary days. As outlined in Table 14, disturbed sleep, in addition to being one of the most commonly reported symptoms for children with Batten disease, was also present in high frequency for seven of the eight (88%) children who experienced it. In contrast, whilst pain was reported in all eight cases, it was only reported in high frequency for two (25%) of the children. Similarly, agitation and cough and cold infections were commonly reported symptoms yet present only infrequently with one (14%) out of seven cases and one (17%) out of six cases reporting high frequency. Joint stiffness, secretions, and cold hands and/or feet were all reported as high frequency symptoms.

Pain was reported for all of the children with Batten disease, and reported in high frequency for two children (25%). Parents were requested to indicate the type or source of pain on any occasion their child experienced this symptom. Responses were received for the eight children (100%) and these varied considerably. The type or source of pain experienced included pain related to constipation or

bowel movements, pain from a fall or accident, pain related to joint stiffness, ear and throat infections, cough, toothache, and pain related to a clinical procedure, specifically the change of a PEG button and Botox injections. Two families indicated that they were not able to identify the source or cause of pain in their child on a number of occasions.

In addition to the 17 symptoms assessed in the diary, parents were requested to document any further symptoms experienced by their child during the diary period. There were no additional symptoms listed in the diary for any of the children with Batten disease.

Table 14. Symptom frequency in children with Batten disease (n=8)

Symptom	Symptom Reported in Diary n (%)	Symptom Reported in High Frequency in Diary* n (%)
Seizures	5/8 (63)	2/5 (40)
Pain	8/8 (100)	2/8 (25)
Cold Hands/Feet	6/8 (75)	4/6 (67)
Joint Stiffness	5/8 (63)	4/5 (80)
Muscle Spasms	5/8 (63)	1/5 (20)
Disturbed Sleep	8/8 (100)	7/8 (88)
Agitation	7/8 (88)	1/7 (14)
Hyperactivity	2/8 (25)	0/2 (0)
Aggressive Behaviour	0/8 (0)	N/A
Repetitive Behaviours	2/8 (25)	1/2 (50)
Secretions	5/8 (63)	4/5 (80)
Choking	3/8 (38)	0/3 (0)
Vomiting	2/8 (25)	0/2 (0)
Constipation	6/8 (75)	0/6 (0)
Diarrhoea	5/8 (63)	0/5 (0)
Breathing Difficulties	4/8 (50)	0/4 (0)
Cough/Cold Infections	6/8 (75)	1/6 (17)

Note: *Symptom was present \geq 50% of the diary days.

4.5.2 Symptom severity

To investigate parent-reported severity of symptoms over the eight-week period the diary recorded severity scores for each symptom; each were rated into the following levels: low severity, where the severity of the symptom was rated as not at all; a little or moderate; and high severity, where the severity of the symptom was rated as severe and/or very severe. Only those symptoms which were rated as severe and/or very severe on more than one occasion were included within the high severity category, to allow for the inclusion of isolated incidents where symptoms were experienced in higher severity.

As outlined in Table 15 and Figure 11, disturbed sleep, joint stiffness, secretions and agitation were rated highest in terms of severity. Seizures, muscle spasms, hyperactivity, breathing difficulties and cough or cold infections all received low severity ratings.

Similar to Sanfilippo disease, the symptoms that were most frequently experienced or reported in the symptom diary for children with Batten disease also tended to be the symptoms that were rated highest in terms of severity. This was the case for both disturbed sleep and agitation. Several symptoms reported in high frequency received lower severity ratings, including pain, cold hands and feet, constipation and cough or cold infections. Figure 11 illustrates these results with width of each

bar representing one symptom and the shading indicating the number of children experiencing low or high severity of the symptom.

Table 15. Symptom severity ratings for children with Batten disease (n=8)

Symptom	Symptom Reported in Diary n (%)	Reported Low* Severity n (%)	Reported High** Severity n (%)
Seizures	5/8 (63)	5/5 (100)	0/5 (0)
Pain	8/8 (100)	7/8 (88)	1/8 (12)
Cold Hands/Feet	6/8 (75)	5/6 (83)	1/6 (17)
Joint Stiffness	5/8 (63)	2/5 (40)	3/5 (60)
Muscle Spasms	5/8 (63)	5/5 (100)	0/5 (0)
Disturbed Sleep	8/8 (100)	3/8 (38)	5/8 (62)
Agitation	7/8 (88)	3/7 (43)	4/7 (57)
Hyperactivity	2/8 (25)	2/2 (100)	0/2 (0)
Aggressive Behaviour	0/8 (0)	N/A	N/A
Repetitive Behaviours	2/8 (25)	1/2 (50)	1/2 (50)
Secretions	5/8 (63)	2/5 (40)	3/5 (60)
Choking	3/8 (38)	2/3 (67)	1/3 (33)
Vomiting	2/8 (25)	1/2 (50)	1/2 (50)
Constipation	6/8 (75)	4/6 (67)	2/6 (33)
Diarrhoea	5/8 (63)	4/5 (80)	1/5 (20)
Breathing Difficulties	4/8 (50)	4/4 (100)	0/4 (0)
Cough/Cold Infections	6/8 (75)	6/6 (100)	0/6 (0)

Note: *severity ratings of not at all, a little or moderate **severity ratings of severe or very severe

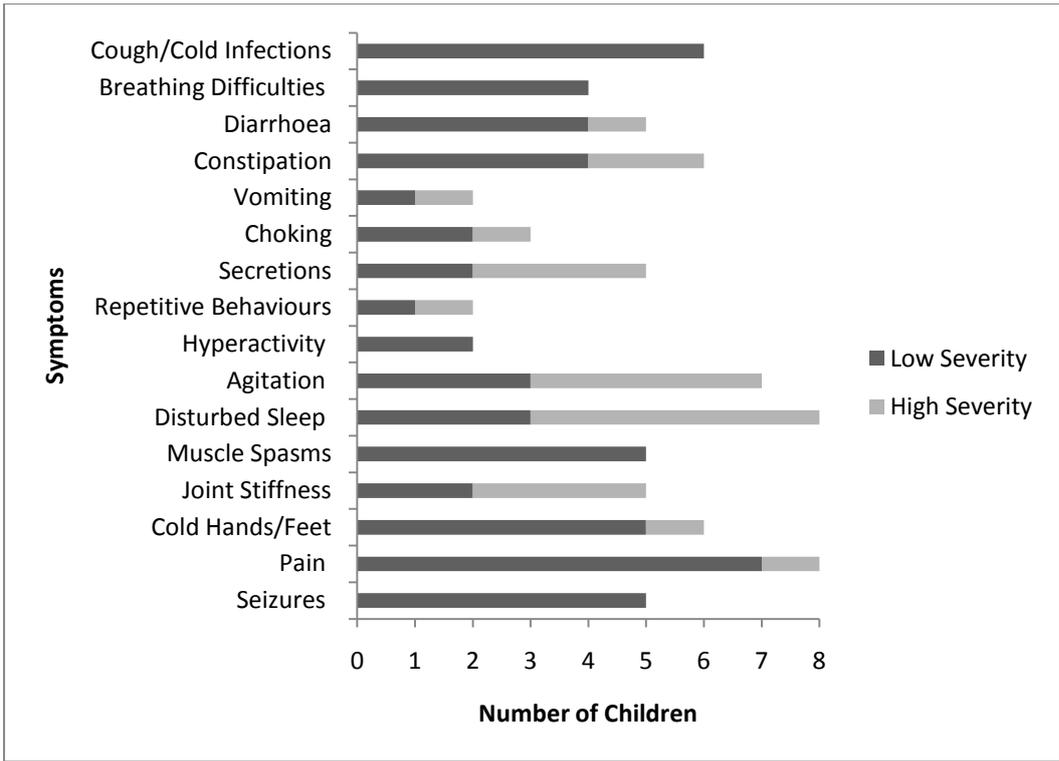


Figure 11. Symptom severity scores for children with Batten disease (n=8). Note: *Low Severity = severity ratings of not at all, a little or moderate **High Severity = severity ratings of severe or very severe.

4.5.3 Symptom management

Families of children with Batten disease used the diary to report a range of strategies and interventions to manage symptoms. These strategies were similar across families and are summarised in Table 16.

Table 16. Strategies and interventions used to manage symptoms for children with Batten disease as recorded in the symptom diary (n=8)

Symptom	Management Strategies
Seizures	Medications; suction; give child cuddles; hold child's hand and wait/observe until seizure passes; give child reassurance.
Pain	Cuddles; medications; vent PEG tube for wind (in cases where this was the cause of pain); comfort child.
Cold Hands/Feet	Extra blankets; massage and rubbing hands/feet; warm bath; extra pair of socks; warm foot/hand wear, Jacuzzi bath; heat teddy (hot water bottle).
Joint Stiffness	Stretching and passive movements; hand and feet splints; medication; massage; ensure good postural positions; hydrotherapy pool.
Muscle Spasm	Medication; stretching; massage.
Disturbed Sleep	Re-position and make child more comfortable; put quilt/blanket on child; give child dummy or favourite toy; give child reassurance; drinks; medication (melatonin); suction; soothing and comfort; soothing music; tell child a story.
Agitation	Distraction; use simple language and instructions; give child a bath; cuddles and affection; try to engage child in a favourite activity; suction; pain relief; comfort; play soothing music and/or child's favourite music.
Hyperactivity	Distraction; try to engage child in a favourite activity.
Repetitive Behaviours	Distraction; chewy toys.
Secretions	Suction; Scopoderm patch (as prescribed); Botox injection therapy (as prescribed); medications; wear bibs and keep area clean and dry.
Choking	Ensure good positioning during administration of feeds and afterwards; give oral fluids slowly; suction when required.
Vomiting	Suction; medication; slow down or stop administration of feed.
Constipation	Enema; give child extra fluids; medication.
Diarrhoea	Monitor closely; ensure fluid intake; ensure good skin care to prevent any breakdown or rashes.
Breathing Difficulties	Suction; oxygen; chest physiotherapy; antibiotics for chest infection; pep mask.
Cough/Cold Infections	Positive Expiratory Pressure (PEP) mask and chest physiotherapy; medication; suction; position changes.

4.5.4 Effectiveness of Symptom Management Strategies

Within the symptom diary, families were asked to rate their perceived effectiveness of the self-management strategies they routinely use to manage their child's symptoms. Specifically, families were asked to indicate how well their actions worked in relieving the symptom using a five-point Likert scale from 'not at all' to 'very well'.

The perceived effectiveness data are presented for those symptoms which were most frequently experienced and those with higher severity ratings. Consequently, the effectiveness is presented in relation to disturbed sleep, joint stiffness and secretions.

Figure 12 suggests that parents perceive their interventions to manage disturbed sleep as ineffective. Forty percent of instances were recorded as working only 'a little bit' in managing this symptom. However, the graph obscures individual family circumstances, in particular, one outlier, where for one of the eight families, disturbed sleep was perceived to be managed 'quite well' and

accounts for more than half of the responses (60%) in the chart. Across participants, therefore, sleep was considered on the whole difficult to manage with the strategies available to families.

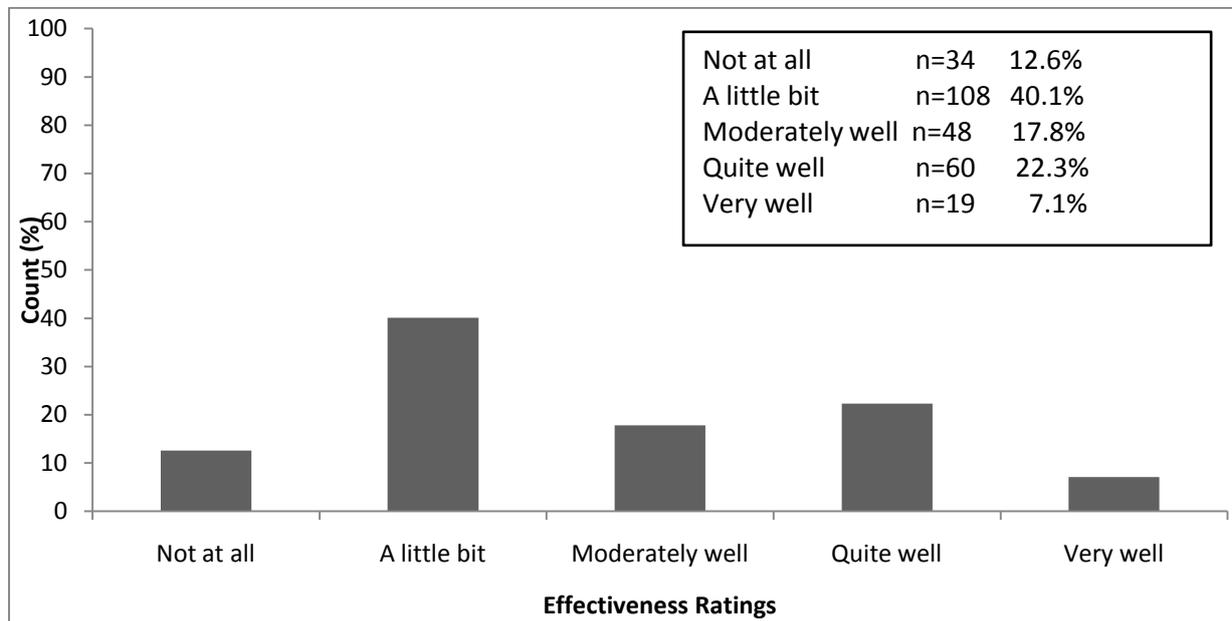


Figure 12: Perceived effectiveness of management of disturbed sleep (n=8)

A similar story is told in Figures 13 and 14 below, where for the symptoms of joint stiffness and secretions the majority of responses were centred around 'a little bit' and 'moderately well'. Once again, it appears from the chart that the responses are spread across three ratings. However, the management strategies recorded as working 'quite well' in Figures 13 (joint stiffness) and 14 (secretions) both arose from the same family which rated their management of sleep as 'quite well'. Thus, the charts illustrate that on the whole these symptoms did not respond well to management strategies employed by the families, and that much of the data rated 'quite well' stem from one outlying family.

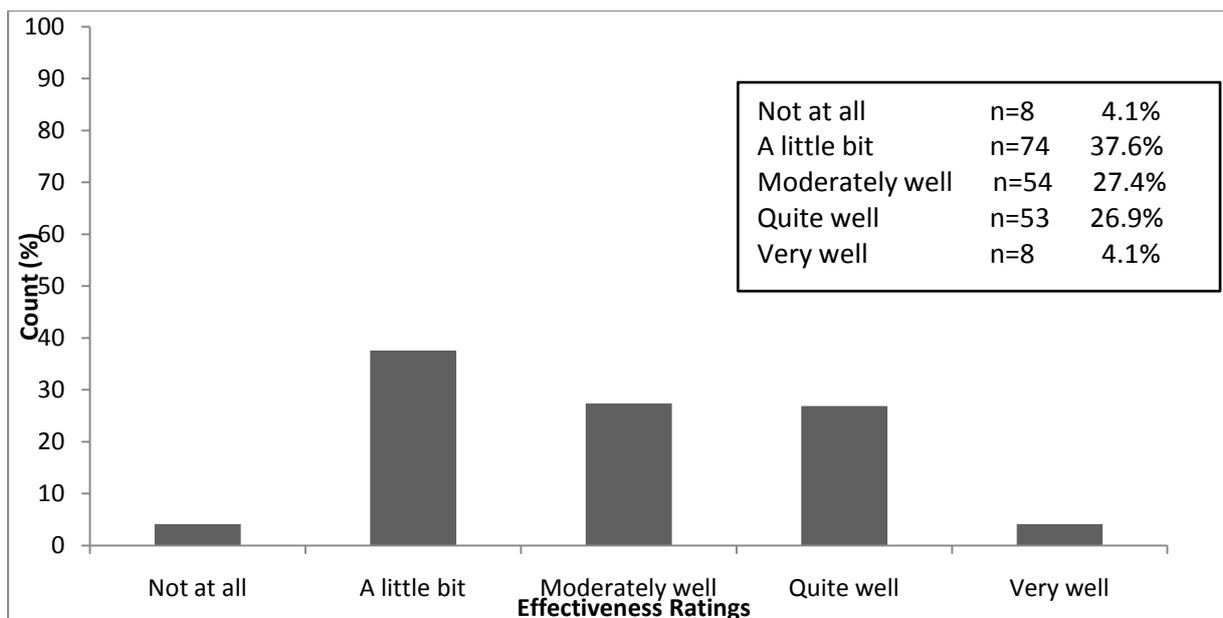


Figure 13: Perceived effectiveness of management of joint stiffness (n=5)

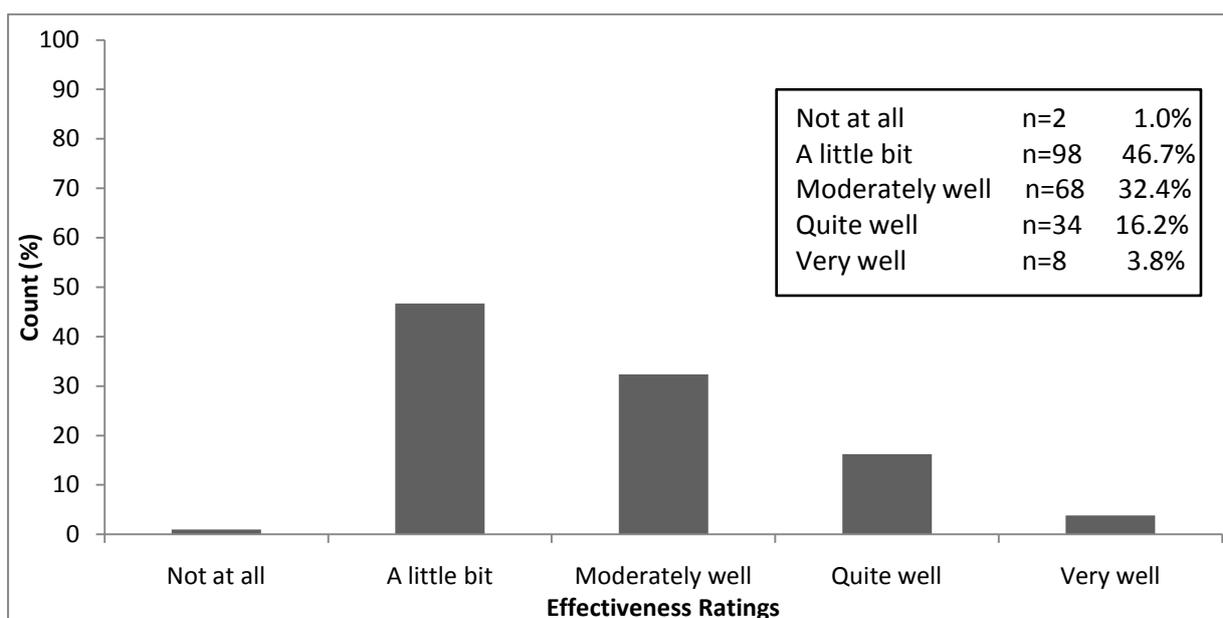


Figure 14. Perceived effectiveness of management of secretions (n=5)

4.5.5 Most challenging symptoms

During the interview, families were asked to identify which symptoms they perceive to be the most challenging to manage and to offer their rationale as to why. Practitioners were also asked, during their interview, to consider which symptoms related to Batten disease are the most challenging to manage from their point of view. Both family and practitioner views are summarised in Table 17 below.

4.5.1 Most challenging symptoms for families

The symptoms that families nominated as being the most challenging for them to manage focused around four main themes: those symptoms which signify a loss and progression of the condition;

symptoms which instil fear as parents worry they may lead to life-threatening illnesses; symptoms which lead to parental exhaustion; and symptoms which prove difficult to control.

4.5.1.1 Symptoms which signify a loss and progression of the condition

Batten disease leads to loss of sight, loss of motor skills and abilities, and mental impairment. As parents observe these losses in their child, they become very aware of the progression of the condition and this can be emotionally challenging and distressing:

Emotionally the hardest one is her, is her sight, not having her sight, 'cause obviously if she had her sight a whole new world would be opened to her so her day wouldn't be quite so bleak and eh ... you know she could be quite happily be sitting and looking about her and you know, amusing herself that way, but the fact that she's not got her sight that's quite hard, for me to deal with, emotionally probably. (Mum, Family L)

As explained by the following parent, throughout the trajectory of Batten disease children experience a series of losses in function followed by a period of time where things remain somewhat more stable. However, each time the child experiences an illness or infection, it is noticeable that they do not recover fully to the way they were prior to the illness, signalling once again to families that the condition is progressing:

So within a year everything went very rapidly and that is quite normal for late infantile Battens disease, they normally go through a rapid loss and then they would plateau for a while with ... if something, if they had say like a chest infection or some sort of infection or they get sick in any way they take a wee dip, come back up but maybe not quite where they were at and that's how it seems to go. (Mum, Family B)

4.5.1.2 Instils fear as parents know this symptom could lead to life-threatening illness

There are certain symptoms which are of particular concern in this population of children because they may lead to other infections or illnesses which can be serious and even life-threatening. Parents describe the constant worry and fear associated with these symptoms and the requirement to closely monitor their child, day and night, to try to prevent such complications from arising. In the extract below, a parent explains how the symptom of vomiting is especially worrying for their family because with the child being in the advanced stages of Batten disease the ability to cough and clear her airway is weakened and she is therefore at risk of developing complications:

I think the, the most difficult daily symptom is vomiting ... because of the fear of aspiration eh, because of the fear of aspirating and what we know can happen through that such as chest infection, collapsed lung or anything like that. I think we're especially frightened of that ... It's just a constant worry for me, especially with another part of the condition is through time their gag, gag reflex goes through time and eh, the cough reflex goes and, we think her gag's virtually away ... (Dad, Family M)

Similarly, the following parent describes how the symptom of secretions requires around the clock assessment and monitoring in order to prevent complications such as choking:

For me it would be secretions because it's a constant factor certainly ... you'll see there [in the diary] most nights, 'disturbed sleep', that's me and her [child] on the SATS monitor and the SATS crashing where her secretions build up and she chokes.' (Dad, Family B)

4.5.1.3 Symptoms that lead to physical or mental exhaustion in parents

Parents reported how the months and years of disturbed sleep has, over time, wore them down. Constant care demands meant this loss of sleep was never regained with the end result being utter exhaustion. The parent below describes how the perpetual lack of sleep took its toll and how this was visible to others:

A couple of months ago I sat with my sister at playgroup and she said 'You look awful,' ... I sat there and I said, 'I've had 12 hours' sleep in the last three nights.' (Dad, Family H)

The family quoted below explained how difficult it was for them when their child developed a new symptom, which involved months of relentless and inconsolable screaming, further impacting on their exhaustive state:

Dad: It was a different kind of scream but then it did develop into ... which is why I'm quite fed up now, it was just a ... but I'd say from February time, we did have screaming for about six months.
Mum: Yeah.
Dad: Didn't we? Day and night.
Mum: It was hard.
Dad: And I don't know if that was because his vision was failing or frustration because he couldn't do the things he wanted to do anymore. (Dad, Family H)

The parents explain further how this new type of screaming led to increasingly difficult night-times, with no-one in the family managing to get any sleep. As noted above, with a duration of six months, the cumulative impact of the symptoms was difficult for the parents to explain, and consequently difficult to manage. They spoke of trying sleep medication, but with little effect:

Dad: When he was in his bad sleep times we would have four, five nights a week where he wouldn't sleep at all. And it could be, you get the feeling of dread at about eight, nine o'clock when I think 'I've got to get some sleep in now,' and sometimes I'd just get a couple of hours' sleep and then he'd start to scream ... and we'd already given him his melatonin hadn't we?
Mum: We gave him all the medicine that we could give him. (Family H)

4.5.1.4 Symptoms that are difficult to manage or control

Some of the symptoms related to Batten disease can be difficult to manage and get under control. Seizures are one such symptom as they are unpredictable, ever-changing as the disease progresses, and very distressing for both the child and family. Whilst medication is available to control seizures, it often needs reassessed and altered as both the child grows and the condition progresses. The

parent below describes how the frequency of seizure activity in their child makes the symptom difficult to manage:

That would be the hardest thing to manage, the hardest symptom to manage, because watching the seizures is pretty horrendous because they have, although it's only momentarily, it's a cluster ... so there may be 60 to 70 in an hour. (Dad, Family M)

The distressing nature of seizure goes beyond affecting the child and family to include siblings and extended family as well. Parents have explained their difficulties in getting extended family members to agree to look after their child due to fear of what would happen should the child experience a seizure whilst the parents are away. As discussed in section 6, siblings described seizures as being the most difficult symptom for their brother or sister and described them as being 'scary' and upsetting.

4.5.2 Most Challenging Symptoms for Practitioners

The symptoms discussed within the practitioner interviews as being those which are most challenging to manage included symptoms related to Batten disease which proved difficult to control. Comparable to Sanfilippo disease, practitioners supporting children and families with Batten disease prioritised the difficulties in assessing symptoms such as pain or distress due to communication impairments linked to the condition.

4.5.2.1 Symptoms that are difficult to control

Seizures are a characteristic symptom of Batten disease, prioritised by families as being distressing and difficult to manage. Practitioners supporting children with Batten disease also nominated seizures as being difficult to control due to their variability and unpredictability. As a practitioner explains below, seizures in children with Batten disease are often distinct in character to other symptoms requiring careful assessment:

The nature of the seizures did surprise me ... 'cause you know it was slightly different from the kind of, the evolution of the seizures ... because I think, although initially, [child] had seizures which were standard with motor twitches which you could diagnose and say 'these are seizures', then they went on to have seizures in different parts of the brain I suspect, like, [child] would have bursts of laughter and almost certainly they were seizures ... and then long periods of crying ... again, you know it's very difficult to differentiate them from painful crying ... you know, but they were different and eventually they were so typical and sporadic and I saw that they could be nothing else but seizures ... so they came as a surprise because they were unusual. (Practitioner R)

4.5.2.2 Difficulties in assessing symptoms due to communication impairments

From the practitioner interviews, it was evident that there is also a challenge in terms of assessing and managing a child's pain and distress. As the practitioner has stated below, the child often cries but it is very difficult to determine the source of pain or discomfort:

Sometimes she does seem to be distressed, you know, it's very difficult for anyone involved, even her parents who are with her all the time to sometimes guess what it is that is upsetting her so, so that ... pain, or ... that can be difficult when it's just a sudden onset of distress or pain and you are not sure what's causing it. (Practitioner C)

While the practitioner above noted that assessing pain is difficult, the practitioner below also confirmed that assessing pain in a child where there are communication impairments is also difficult:

Pain ... with the child not being able to tell you that he's in pain but obviously if he's crying ... (Practitioner M)

This practitioner highlights the decline in the communication with the child and the challenges that she now faces in terms of assessing and determining the child's needs and what it is that is bothering them:

All that's gone now, there's not as good eye contact as what there would have been, you know ... you have to go in close to her to speak to her before she maybe recognises you. Yes, and I suppose that whenever she would take the crying spells, and often she does ... you feel a bit lost to help her, and she can't tell you what the problem is. (Practitioner L)

4.5.6 Summary

Sixteen of the 17 symptoms assessed in the symptom diary were reported by families of children with Batten disease as being present (with the exception of aggressive behaviour, which would not be expected and was included to capture its presence in Sanfilippo). Disturbed sleep was present in highest frequency, closely followed by joint stiffness, secretions and cold hands and/or feet. Those symptoms rated as being most severe in children with Batten disease included disturbed sleep, joint stiffness, secretions and agitation.

Symptoms which were the greatest challenge to manage in Batten disease were those which signify a loss of a skill or ability and in turn signal the progression of the life-limiting condition, or which have the potential to lead to health-related complications and other life-threatening illnesses.

Family members and practitioners for one child agreed on managing seizures as the most challenging symptom. Both groups described the distressing nature of seizures for children to experience and families and practitioners to observe. However, for the remaining children, practitioners and family members identified different symptoms as the most challenging, as indicated in Table 17.

Practitioners highlighted how profound communication impairments, characteristic of Batten disease, posed challenges in assessing symptoms in these children, notably symptoms like pain and distress.

Table 17. Most challenging symptoms to manage in children with Batten disease from the perspective of families and practitioners (n=8)

Child	Symptoms occurring most frequently*	Symptoms with highest severity ratings*	Which symptoms do families find most challenging to manage and why?	Which symptoms do practitioners find most challenging to manage and why?
A	Disturbed Sleep	Disturbed Sleep	Agitation because it is difficult to avoid triggers of the children's agitation. Parents describe using a trial and error method to control this symptom.	Loss of communication skills resulting in the children not being able to communicate their needs and feelings.
X	Repetitive Behaviours	Disturbed Sleep	Agitation because it is difficult to avoid triggers of the children's agitation. Parents describe using a trial and error method to control this symptom.	Loss of communication skills resulting in the children not being able to communicate their needs and feelings.
B	Seizures and Secretions	Secretions	Secretions because of the constant fear of the child choking. Due to the build-up of secretions during the night, parents sleep is disturbed so they can continue to monitor and assess their child.	Myoclonic jerks because the child experiences frequent jerking which leads to muscle spasm and associated pain. This then triggers the child to have seizures. The practitioner described it as a 'vicious cycle' and challenge to find medication to effectively control the symptom.
L	Joint Stiffness	Joint Stiffness	Loss of child's vision as this signifies a progression of the disease. Mum described the emotional upset she experiences in observing her child lose her sight.	Seizures because they are difficult to control and require constant reassessment.
C	Secretions	Secretions and Pain	Seizures because the parents describe not being in control of the seizures and the challenges they go through in trying to effectively manage this symptom.	Seizures because child will have a seizure that you do not immediately recognise as a seizure but it is in fact seizure activity. The child's seizure pattern changes frequently meaning you get one type of seizure under control and are then faced with a new type.

Child	Symptoms occurring most frequently*	Symptoms with highest severity ratings*	Which symptoms do families find most challenging to manage and why?	Which symptoms do practitioners find most challenging to manage and why?
M	Joint Stiffness and Secretions	Joint Stiffness	Seizures as they are a constant, unpredictable and relentless symptom to manage, despite the child being on medication to control them. Vomiting because of the fear that it could lead to aspiration and subsequent life-threatening infections.	Vomiting because the child experiences frequent vomiting which is very difficult to control. The vomiting, its severity and unpredictability, makes it even more difficult for the family to get out and about with the child.
U	Joint Stiffness and Secretions	Joint Stiffness and Secretions	Secretions because they compromise the child's airway and cause the child to have colour changes and obvious difficulties in breathing. This is scary for the parents to observe.	No Practitioner Interview
H	Joint Stiffness	Agitation	Disturbed sleep because it is a constant symptom and the parents are left completely exhausted.	Pain because it is a challenge to assess the child's pain. The child often cries but it is very difficult to determine the source of pain or discomfort.

NOTE: *Data derived from the symptom diary.

4.6 Summary

The data have highlighted the symptom profile of children and families affected by MPS disease and Batten disease. The prospective design enabled the pattern of symptoms to be revealed over an eight-week period, providing detailed descriptions of symptom frequency, severity, management and family experience. Behavioural symptoms dominated Sanfilippo disease as key concerns for parents as did the difficulties in assessing children's symptoms due to communication impairments related to the condition. The same issues were prioritised for practitioners supporting these children and families.

Sleep disturbances and the resultant exhaustion for parents and management of seizures were identified as key concerns for parents of children with Batten disease. Practitioners supporting families with Batten disease resonated with the difficulties in managing seizures as a symptom in this population. Pain was also highlighted as a concern across both MPS disease and Batten disease.

Overall, the symptom profile data are consistent with the survey phase where families prioritised symptoms such as behavioural difficulties and visual and communication impairments as being those most difficult to manage and having the most impact on quality-of-life for children and families.

5 Findings: Family perspectives of the impact of supporting a child with a rare life-limiting condition

Parents of 26 children participated in interviews focusing on their experiences of supporting a child with MPS disease or Batten disease. Families spoke at length about their experiences and touched on a wide variety of topics in describing their lives. Figure 15 illustrates some core components to the interview data, and represents a model of the inter-relatedness of some elements of their experiences. Each of the sections of the model has within it subsections that explain further the experiences of families supporting children with rare life-limiting conditions.

The model is organised in such a way that ‘family expertise and experiential knowledge’ forms the hub. Each part of the model is discussed in turn, illustrating how each component connects with the others. To help the reader navigate through the complex and rich accounts, each part of the model has a reference within it indicating which section of the report describes the data.

Parents also spoke at interview specifically about the symptoms their child experiences, and these accounts are reported in section 4.

As noted in section 2.7, the data are presented in anonymised format. To prevent matching of symptom descriptions (reported in section 3) with the data in this chapter, numerical identifiers have been randomly assigned to each family from 1-23.

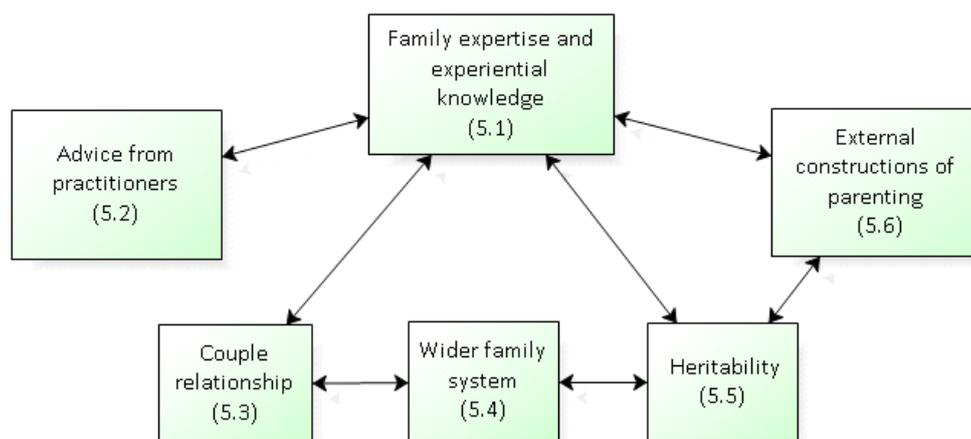


Figure 15: Family expertise and experiential knowledge

5.1 Family expertise and experiential knowledge

Family experiential knowledge occupies an important position within the model, whereby the majority of other elements of the family experience are connected to it. Its pivotal position in the model reflects the clear sense in the interview data that families had gained considerable expertise and that this is a guiding feature of their overall experience of having a child with a life-limiting condition.

As the sections below demonstrate, experiential knowledge was often visible even before a diagnosis of MPS disease or Batten disease was made. Expertise was then honed and developed through daily symptom management. This often led to parents playing a key role in educating practitioners about their child and condition. The development of expertise and experiential knowledge, though, was not always what parents consciously chose, but was a position adopted out

of necessity since the rarity of the condition meant that practitioners often had less information and experience to draw upon.

5.1.1 Parental intuition

Parents were asked at the interview to talk about their experience of their child being diagnosed with MPS disease or Batten disease. These accounts often began with parents reporting their intuition or suspicion that 'something was not right' with their child:

Up to three she was a normal child ... talking ... she wasn't making proper sentences. That's when we, I had doubts, I kept on thinking, 'Why can't she ...' She was saying things like, 'How do you do?' and, 'I want one,' but I think she thought that was a word, not a sentence, and ... and also she had this goo coming out of her nose, this green goo, all the time. (Mum, Family 22)

Similarly, the family quoted below outlines how the child's development appeared to be regressing instead of progressing:

There was just a few niggling things, her eye contact started going, she was ... as you can see in that picture she was so smiley and giggly and she stopped smiling, she stopped feeding herself and I didn't know whether it was laziness or whatever, but you, you put it down to other things obviously first ... so immediately I thought, 'She's autistic ... that's it, the MMR,' anyway so ... you obviously look on the internet, tick 9 out of 10 of the boxes as she did, poor eye contact, she lost her skills, she started, her crawling, she started banging her head a lot and was quite wobbly on her feet, you know, she started just regressing and ... children should be going forwards not backwards so, I just knew there was something wrong. (Mum, Family 18)

Parents who had older children often made reference to comparing the timing of children reaching developmental milestones. They reported that they would look to the other sibling(s) to gauge the development of their affected child, while still feeling that something was not as it should be:

She wasn't reaching the goals that [sibling] had reached. She wasn't being toilet trained, she wasn't speaking, everything was just a lot slower. And that was when we kind of realised that things possibly weren't quite the same as [sibling], but, didn't really know what was wrong with her. So, it was, it was very gradual, it wasn't something that came out and hit you overnight, sort of thing, it was, just sort of, kind of, you know, she's not quite, quite the same as [sibling]. (Mum, Family 8)

While the family above reported that it was a gradual process of identifying that something was not right, other families reported that there had been continual concern, not solely associated with how this compared to siblings' development. The following family had harboured concerns about their child since she was a toddler, yet a formal final diagnosis was only made when she was eight:

We'd been pushing and saying to people there's something not right since before the age of three ... you just were noticing things and you were putting it off thinking well, we're comparing her to [sibling] and [sibling] was very quick at everything and we think we shouldn't do that, you know not every child is the same, but you see things like play with [child] was quite sophisticated when she was little ... speech was terrible and then of course she had glue ear and they'd said that it was a real high amount that, they were surprised she even spoke because of the amount of glue in her ears so then they were saying, 'Well that's what it's been all about so she'll now be able to catch up,' and we were just going, 'No, sorry there's just ... this, this is not washing with me, there's something.' You just know. (Mum, Family 19)

Families reported that from the time that initial symptoms began presenting to the time of diagnosis of MPS disease/Batten disease, often several other diagnoses were made. Common examples were of autism and epilepsy, capturing a sense of explaining some of the symptoms but not the family's overall experience of difficulties. While these conditions may be similar to MPS disease/Batten disease, and some of the symptoms are recognisable, parents reported their scepticism that the diagnosis reflected their child's experience:

I just knew there was something, he was taking seizures and then they put it down to epilepsy and then he started kind of losing his balance, but really silly things. (Dad, Family 10)

Furthermore, some parents had worked as nurses, carers or with children with special needs. These professional experiences underpinned their parental instinct that something was not right:

Horrendous, it was awful ... I, from the very day I had him, practically knew there was something not right with [child] ... I have worked in special needs for seventeen years ... and he was hyper. He was hyper in the womb actually, he just didn't stop moving at all ... hyper from the day I had him. (Mum, Family 3)

Parental intuition and suspicions of the child's condition started the process of parents developing the expertise about the condition and the symptoms. In the subsequent section, parents describe how their experiential knowledge is developed through managing their child's symptoms.

5.1.2 Experience gained through symptom management

Families were asked to explain how they managed symptoms on a day-to-day basis. In several interviews families reported that they could recognise changes in their child's symptoms that they felt would only be recognisable to themselves as parents. They reported that this type of knowledge is learned primarily through the parent/child relationship:

It's really hard to explain because you get certain types of aggression that anybody would just see as her being aggressive, I think when you're with [child] all the time, maybe it's just instinct, being a mum or something, sometimes you'll pick up something in that and you'll think, that's not quite normal today, so then, then I might analyse 'Is she in pain, or is she just not feeling right?' and then you can pick them days out and you just think, no something's just not right with her and you may not be able to say what, but you'll know there's a slight difference and you'll know, it just ... you know mannerisms, just in her body language when she's being aggressive ... as opposed to normal days, just, she's just aggressive, it's part of the disease, you've just got to get on and deal with it, you know ... it's just one of them things. (Mum, Family 19)

Families indicated that the relationship is vital to the effective self-assessment and self-management of symptoms. In the passage below, the mother reports that prior to seeking medical advice for symptoms, she engages in self-management. Thus, before attending the hospital with a symptom, she begins the process of investigating and treating the symptom herself. The process carried out by the mother in trying to diagnose and treat her child strengthens her own knowledge base and treatment repertoire. Rather than seek external medical advice, the mother wishes to remain at home as far as is practicable and to find the solution herself:

Mum: I didn't want to be running to the hospital for something that I felt I could self-help or make right and I wanted to have that option to try to make it right because ... you know if they had said to me 'Is she having a seizure?' and I'm like, 'I don't really know!' You know, and I didn't really know and I thought well if, if, if that doesn't solve it then I'm, I'm going, we're going to have to go, you know and I would have admitted that I ... I suppose I always feel at least if I can go to hospital and say 'Look, I have tried Nurofen, I have tried that, it hasn't helped,' ... then at least ...
Interviewer: You've ruled out some of the ...
Mum: We've ruled out some of the things that they would probably go to try and then if they were giving her Nurofen then I'd be sitting there going 'Right, okay,' ... you know, I like to try and rule out the basic things first so then I have something to say rather than going ... 'Oh, well, I didn't give her Nurofen or whatever.' (Mum, Family 11)

Many parents expressed the idea that managing symptoms at home is preferable. In order to do so, parents have learned how to control symptoms, which increased their confidence in being able to assess, diagnose and treat symptoms. The family below temper this by stating that they are also aware of their limits and know when to seek medical attention:

Mum: Lots of things that the doctors have told us that we've sort of picked up on.
Dad: Yeah.
Mum: Whereas before we'd have been on the phone every five minutes to the doctor, 'What about this? What about that?' Now simple things that we can ... basically control in the house ...
Dad: You know how far you can let it go before you have to go and get seen. (Family 10)

For some families, being able to change their child's medication based on the parental observations, albeit with prior approval from their doctor, also bolstered their knowledge and confidence of symptom management:

We've got the, the, the allowance from [doctor] to change meds about the home if we feel, you know something's not quite working or we can give an extra dose and we can do all this kind of stuff. (Dad, Family 2)

Knowledge of medication and equipment surrounding symptom management was apparent in the majority of family interviews. The family below demonstrated their level of knowledge and confidence in being able to manage symptoms at home using specialist medication and equipment:

Mum: Her seizure activity and all that there, we treat it at home and if she does go into a bit of status normally the chloral ... which isn't chloral hydrate, to most children, it's chloral betaine to [child] because of her diet.

Dad: Tablet form.

Mum: It's just tablet form. We used as a sedater and we keep her sedated for maybe 24, 36, depending on how she is and, just keep her on her SATS monitor and monitor and her, her paediatricians know that we know her best, we know she'll, if she's okay and if she's not okay, if we need help, and generally it works. (Family 12)

Experience developed in managing a specific symptom leads families to understand how and when to intervene:

Mum: You have to, fine-line, you have to let her cough on her own at times ... there's times that you're dying to suction it because you know it's sitting there and you know she needs to cough and the saliva's building up but if you keep suctioning her then she'll stop coughing, she stops coughing then you're going to be in bother, you know she's going to be in bother.

Dad: It's, it's worse for her.

Mum: It's worse, like we could suction her all day long, it's handy for us, but for [child] it's not a good thing. (Family 12)

Parents' knowledge and expertise increases over time and as the condition progresses. However, families also reported that as the disease progresses and symptoms become more complex or new symptoms present, they need to learn how to manage these new symptoms. The statement from the family below outlines how their child developed a new kind of seizure activity that they were unsure how to manage:

He started having shaky ones, the first one he had, the first was when I took him to bed and his eyes slightly rolling and he started gasping for breath, I don't know what it was but we just called 999 and called an ambulance and then had had shaky ones in the taxi coming home and a couple of jerky ones at school, all different types, but we've got him on Provizan now along with his Epilim, they're doubling-up the dose and that seems to, at the moment he's on the minimal, kept him on and it seems to have worked. (Mum, Family 4)

In other cases, parents' medical knowledge and awareness helps to build and develop knowledge and symptom management for their child. Indeed, one mother diagnosed her child more quickly than medical practitioners did with a co-morbid condition.

Families were asked who they would contact for assistance if there was a particularly worrying symptom for them. In one case the mother identified that she would find her own way of dealing with specific symptoms based on knowing her child:

You manage it yourself, I mean if you've got to turn round and ask everybody how to manage each symptom you're not going to get anywhere are you ... you've just got to get up and run with it and just ... you know, you find your own ways, there's so many different things, you can find your own ways around and ... different ideas and we normally get there in the end. (Mum, Family 19)

Similarly, the statement below demonstrates clearly that parents are faced with limited sources of information. A consequence of the lack of specialists to ask is that parents develop their own internal reference guide, becoming the expert of their child's symptoms. When asked who she would approach for support and information, the following mother indicates the limited options open to her:

Probably [paediatrician] but she only works Monday, Tuesday and Wednesday, she is the paediatrician, I don't speak to the GP, the GP doesn't know anything at all. [Paediatrician] doesn't know an awful lot either [laughs]. I seem to be a bit of an expert on the MPS thing. (Mum, Family 3)

The experiential knowledge gained during the initial stages of the child's condition, including parental intuition prior to diagnosis, coupled with the experience gained through symptom management, places parents in a unique position. Not only are they learning and developing their own symptom management skills they are also educating others about the condition and how this affects their child.

5.1.3 Parents as educators

Parents felt that often practitioners looked to them to provide knowledge and insight into the condition:

You have a learning curve for a period, haven't you, where you learn what the symptoms were, what the possibilities were, was it going to be a short life ... but then having the time to explain that to everyone else, 'cause no one else had heard of it either, so we when they ask how everyone's getting on and you tell them that she's been diagnosed with [MPS disease], and then having to explain what [MPS disease] is. (Dad, Family 5)

Mum: And the nurse practitioner will say 'I've never heard of this condition'. Do you know, 'I don't know what it entails.'

Interviewer: So they're no use at all then, probably.

Mum: No they are not. Em we had to tell her what we needed them to do ... and she says 'Well, what do I need to do?' and I says 'She needs an inter-muscular injection to stop the vomiting,' and it turned out that I suspected [child] had a urine infection as well, so I tested her urine and then wrote all the results down for them, 'cause. (Mum, Family 23)

Later in the interview this family told of how the mother had attended clinical training sessions to inform and educate herself on managing co-morbid conditions. These helped her to distinguish which symptoms are related to which condition. The following quotation demonstrates with great clarity the experiential learning of parents and the magnitude of their knowledge over that of healthcare practitioners:

Mum: I've attended the training sessions to explain [child's] diabetes, how it manifests itself and I've taken time out to do that over a two-day period, just so that they understand that [child] doesn't conform to even the normal diabetes.

Dad: We have, we have nursing, we have diabetes doctors and nurses telling us what to do, and we know it's wrong, and we've got to say to them 'No, we'll do this,' 'Oh you can't do that, it's far too much', 'Look we know [child], we know what's wrong,'... 'You can't give her that,' [sounds exasperated]. (Family 23)

The rarity of MPS disease and Batten disease results in parents becoming experts in the condition which leads to them adopting, either through choice or necessity, a role of educating practitioners in their child's condition, and effective symptom management strategies.

5.1.4 Necessity in developing expertise

The impetus to develop expertise often emerged from necessity rather than by choice. The mother quoted below illustrates this necessity. Even when she is struggling, professionals rely on her expertise rather than having suggestions themselves:

You've got to know your own child to know how to manage it. I might go and say 'I'm at my wits' end and I don't know what to do about it,' but every one of them professionals, if they were having that problem, would be coming to me and going 'What do we do about ...?' (Mum, Family 19)

A number of families reported having to provide their child's care even when in hospital, because the staff are uncertain how to treat such complex rare conditions. The extract below illustrates one family's need to use their own expertise in order to care for their child while in hospital:

When it comes to a medical admission we've got to do all the work. We've got to go in with [child], stay all night, we've got to, [Mum's] got to change her and do the, administer her medication, we administer her insulin, we do her PEG feed, the nurses only come in and say 'Morning!' and away again. (Dad, Family 23)

Other parents spoke of how the rarity of the condition, and consequent lack of knowledge of the healthcare practitioners, put her in a situation where she herself was not comfortable:

And the doctors didn't know anything about it. And [exhales], that is, that is really tricky, because, I'm not particularly pushy, I'm not pushy. I don't fight particularly easy ... I don't want to be the one telling them what they should know, because, and also, you can get strange reactions, not everybody takes it very well. But they don't know about [MPS disease]. (Mum, Family 8)

Indeed, the quotation above also indicates a sense of the parent having no choice in becoming an expert in the child's condition, due to the limited exposure many doctors have to these conditions. Expertise, then, was not something which parents actively sought out to develop, but was foisted upon them through others not being able or willing to make sense of their situation.

5.1.5 Summary

The experiential knowledge gained by families often commences with their initial intuition that something is not quite right with their child. This intuition was further driven home by noticing developmental milestones which are not met or through comparisons with their other children. The rarity of these conditions, and the limited knowledge which many practitioners have, results in parents developing their own symptom assessment and management methods. Parents' expertise then becomes critical for practitioners themselves to learn from in the absence of training that adequately prepares them for supporting families affected by these rare life-limiting conditions. Becoming experts appeared to stem from necessity as much as choice, and invariably, was closely related with the rarity of the condition. The impact on families of taking on this expert role adds considerable stress and pressure onto already difficult situations, features which are discussed in more detail in the sections below.

5.2 Advice from practitioners

Families were asked about the advice they received from practitioners in the context of managing their child's symptoms. It was evident that there were a number of key components in how parents heard, and then made an active decision to either accept or reject, the advice from such practitioners. Families reported two features that were important in how advice was offered and received, namely, the practitioner's knowledge of the condition, and the practitioner's relationship with the child. Both of these features proved vital for parents to accept and implement symptom management strategies proposed by the practitioner. For some parents, their own experience of symptom management and intimate knowledge of their child meant that the advice received from practitioners conflicted with the parents' own sense of what constituted best care. Receiving differing or conflicting advice was also not uncommon, meaning that parents often had to decide themselves how to manage symptoms in the face of practitioner disagreements.

5.2.1 Knowing the condition

Families indicated that, in some cases, practitioners lacked adequate knowledge to treat the child. For some families there was a clear sense of the practitioner not knowing the child, and deferring to the parents' superior knowledge and expertise. One parent, quoted below, explains that she has

developed a relationship with a doctor who is knowledgeable about the complexity of her child's co-morbid conditions:

I'm very lucky again because I go back to [doctor], [doctor] is my main port of call and she actually is over in the [local] area, she is actually over, that Crohn's feed or that bowel problem, so she's everything combined into one, so she is, she would be my first port of call ... I just, I just stick to one person and then that makes life easier. (Mum, Family 11)

Medical and practical advice from practitioners was reported as equally important for families, as this parent explains, when speaking about someone they refer to elsewhere in the interview as their 'MPS doctor':

The advice that you get from [the MPS doctor], it would tend to be not just from her own medical background, it would also be feedback that she's had from other parents and other families, so it's not just medically based it is actually, a lot of it is practical advice as well. (Mum, Family 21)

The family below speak of their child being admitted to hospital for investigations when he developed a new symptom. The family were met with a string of doctors who lacked adequate specialist knowledge of the condition, resulting in upset for both the child and parents:

Every time we stood him up he cried his eyes out and just collapsed to the floor, literally. He was laying on the bed in the hospital in the A&E and they had about ten doctors come through, one by one, this is in the middle of the night, one by one 'Can you make him stand up so we can see?' And it just got to the point when [Dad] said 'No, I'm not making him stand up anymore until you get one doctor in here who knows anything about his condition and has a suspicion of what the hell is going on because I am fed up seeing young people come in here with not clue telling me to put my son through the pain again for no apparent reason.' It really got bad and I phoned [paediatrician] straight away and he said 'Get the doctors to call me and I will explain to them what they need to do.' And he did, so [paediatrician] is, he is the guy that seems to know what is wrong. (Mum, Family 15)

The quotation above indicates that parents are often exposed to multiple healthcare contacts before reaching someone with adequate expertise and knowledge.

Many families shared stories of a lengthy journey from noticing initial symptoms to eventually gaining a diagnosis. Gaining a definitive differential diagnosis often took months or years. The difficulty in establishing a diagnosis was attributed to the rarity of the condition. As a consequence, there was substantial delay in gaining advice and support:

*Dad: The medical professions, professionals in the community without a diagnosis is a nightmare ... 'Well, what's your child got?', 'Don't know, undiagnosed,' ... 'Ah well, I'll put you to the back list.'
Mum: 'Cause nobody knows where to slot you in, do you know what I mean, there's no slots for you, because we're undiagnosed. (Family 12)*

The mum in the quotation above indicates that the difficulties in gaining a diagnosis have had such tremendous impact that she frames this as 'we' are undiagnosed, not just the child. Other parents found that they often needed to explain the symptoms and the condition itself, to both lay people and doctors, again impacting on the availability of advice:

It did not particularly make any difference knowing what it was called ... it meant that you could say to the neighbours, 'Oh it's Battens,' and then invariably people will say, even doctors, 'Never heard of that, what's that?' 'Cause it's so rare. (Dad, Family 23)

Many families spoke of practitioners having limited knowledge and understanding of the condition, and its impact. They were quick to distinguish between practitioners who were supportive and understanding and those who struggled to understand how the condition precluded certain diagnostic tests or treatments, as the following quotation illustrates:

The GPs and the paediatricians, they're all quite good, but when you go, when you're in those kind of specialised children services, they're all quite good and they sort of know what's going on but it's when you go into the wider NHS, when you go and see about her ankle or about her ear or, you're kind of, they don't really know what's going on and they'll, they just don't understand [child's] condition or know what it is, and you find yourself telling them or giving them and booklet to make them understand what's going on. (Dad, Family 21)

The limited knowledge of practitioners of these life-limiting conditions places responsibility on the parents to navigate healthcare systems, and often to take control of the situation by demarcating what is and is not acceptable to the family. Gaining helpful advice from practitioners was often a process fraught with difficulty.

5.2.2 Knowing the child

While families reported that practitioner knowledge of the condition was important, so too were practitioner relationships with the child. Knowledge of the child appeared to be vital in how parents decided whether to accept or refute advice. The father quoted below reports how he did not take advice from a medical practitioner as he felt that that it was not specifically related to his child. Consequently, the parents' experience of the condition and their child outweighed the medical advice:

If you see somebody like orthopaedics or ENT [ear, nose and throat] or any of those people, we would always ... what they say, we would always take it with a pinch of salt, basically, 'cause a lot of the times we would know [child] better and know that the, the sort of general advice they're giving doesn't really apply to [child] so you filter out what they're saying and you ... and then as I say we know if there's a, if we still don't know then we can go to the other doctors. (Dad, Family 21)

The above point regarding the lack of advice which appeared specifically tailored to their child was described further by other families. For example, in the quotation below the mum reported that while practitioners may have experience and knowledge of these rare life-limiting conditions, they lacked the specific knowledge about how the condition and the symptoms affected the child. In this

circumstance, not knowing the child was a deterrent for the mother in fully accepting advice from the practitioner:

I'll hear what that person says and then I'll do what I think is right ... and that's the end of it ... and do you know what, at the end of the day there's nobody knows [child] like me and [partner] know her and there's nobody would know how to manage each symptom like me and [partner] would know, because they can go ...' Well, this is how we manage this symptom in Sanfilippo,' but they can't go 'This is how we've managed this symptom in Sanfilippo with [child] ... 'Cause every child's different, whether they've got Sanfilippo or not. So, you know, you've got to know your own child to know how to manage it. I might go and say 'I'm at my wits' end and I don't know what to do about it,' but every one of them professionals, if they were having that problem, would be coming to me and going 'What do we do about ...?' (Mum, Family 19)

In the following example, the mother can be heard to pity the GP in being called out to help manage a child's symptoms that would inevitably be beyond the GP's knowledge of the child:

*Dad: Like last Tuesday we ended up getting a GP out ...
Mum: Huh, poor GP.
Dad: To [child], she had a, she had a very high temperature and it was [child's] GP that came out.
Mum: But she doesn't really know her.
Dad: But she doesn't know her and she said it like, could be an ear infection or it could be just a general sort of sinus upper respiratory so they upped her antibiotics and we just got stuck in with physio and things just to make sure nothing ...
Mum: But they say 'You know her best,' and that's it. (Family 12)*

5.2.3 Knowing both the condition and the child

In some cases, families spoke of practitioners who know both the condition and their child. In these situations, parents often sought advice only from this practitioner. The family below report that the paediatrician is the first and only point of contact for their child's main symptoms:

It's just mainly her paediatrician I deal with, and I just really do what he says cause obviously he's the one that knows best but everything else I just ... I just deal with it in whatever way I think best I mean ... I think sometimes as a mother you know better sometimes [laughs]. (Mum, Family 22)

The mother is clear that the paediatrician is the main point of support and in terms of the overall care of her child; she does, however, supplement the advice given by this practitioner with her own experiential and parental knowledge:

Often, you know, [hospice], the doctors there, because they've seen [child] and they deal with [MPS disease] children all the time, you, you know they know what they're talking about, so you often ... They often sort me out, basically. (Mum, Family 22)

The mum below describes the level of contact with her child's practitioners and in the surrounding text creates a sense of how this strengthened their relationship:

Mum: You have a lot of contact with doctors. There's not a week goes by when I don't talk to, if the local paediatrician, from [specialist hospital] ring me every month, as well, so weekly contact with them.

Interviewer: To check in?

Mum: I can ring up, really, whenever I want to, to just get things changed. I mean, last week I rang up and said, 'Could we up one of them [medications], because she was coughing again', and they did it, which is great, you know? It's a good way to be, because, they know that I won't change it without their permission, but at least I know I can just ring them and say, 'Look, I feel this is, this needs to be done.' (Mum, Family 8)

For the parent above, then, there is an important intersection between practitioner expertise, accessibility and the mother's knowledge of the child's changes in symptoms.

5.2.4 Managing different advice

For some parents, their experience of symptom management, coupled with their intimate relationship with their child, often meant that practitioners' advice conflicted with what they felt was best for their child. Across the data set this created a sense of parents having to manage competing opinions on how to support their child, and draw on their own experiential knowledge and expertise.

The family below sought medical attention for their child. When the GP made a diagnosis, however, they challenged the GP's judgement and sought alternative explanations and treatments. The parents drew on their own observations and experience of the condition to diagnose the child's skin irritation as stemming from a problem with her gastrostomy:

Mum: When [child's] gastrostomy was leaking, that was a major, because the acid was burning her skin so it was leaking really, really badly and they had dermatitis and, and I said 'It's not. It's acid burning her skin,' I knew what it was ...

Dad: [voicing the GP's diagnosis] 'No it's not'...

Mum: 'Oh it's not, it's dermatitis ... give her this,' ... and I thought, 'It's not dermatitis,' what we need is to something to stop the acid from getting onto the skin in the first place. (Family 23)

Such experiences closely tie in with the data presented above regarding practitioner knowledge of the condition. GPs with limited exposure to children with gastrostomies are likely to struggle to accurately diagnose such problems. The family quoted above explained the difficulties they faced in making the decision about the best care for their child when they knew it conflicted with the advice of practitioners, as did the family quoted below:

Dad: We have diabetes doctors and nurses telling us what to do, and we know it's wrong, and we've got to say to them 'No, we'll do this,' 'Oh, you can't do that, it's far too much,' 'Look, we know [child], we know what's wrong, you can't give her that.' [sounds exasperated]

Interviewer: So you just hang up on them and do what you know you need to do.

Dad: No ... but it invariably comes down to that.

Mum: I mean if we make the wrong decision then it, we are held responsible ... but if you haven't got the help at that other end to guide you, make sure that what you're doing is right, then do you know what do we do? Do you know, we have to make the decision, I mean at the end of the day we, we're making decisions about [child] that should really be made by professionals. (Family 23)

Decision-making weighed heavily upon many families then, as they struggled to get support from non-specialist practitioners. Other families expressed similar sentiments, and the difficulties that they experienced in communicating their opinions to practitioners. The family below spoke of difficulties in managing conflicting advice. The child's consultant had told the mum to remove the nasogastric tube. Mum had warned practitioners against this, knowing that her child would need this method of feeding to maintain her weight. As a consequence the mum decided that it was best for her child to put the tube back in again. She explains her position and how she feels about taking advice from practitioners:

I says to [child's] own doctor that sees her all the time, I just said, 'Look [doctor],' I says, 'Well, Doctor So-and-so has taken her off her overnight feeds,' I says, 'She has lost weight,' I said, 'We can not afford to lose weight. Can you not get in contact with [doctor] at [hospital] and ask him for me?' And the dietician had even said, 'Oh it's fine, she'll be okay,' but then the dieticians at [hospital] had got in contact with her, she phoned me the next day and said, 'No, you need to put her back on the feeds.' I says, 'I told you this!' I feel as if half the time I don't like listening to the ones that don't deal with [child] all the time ... because, not that I don't want to listen to them but, I know they're doctors, but you know your own child. (Mum, Family 5)

For other families, managing different advice was not focused on the interplay of family knowledge and practitioner knowledge. Rather, there was, at times, a balance to be struck in mediating different practitioners' advice. For example, families would receive conflicting advice from practitioners regarding the management of their child's symptoms. The family below spoke of their experiences of mediating such different viewpoints and how they manage this. The sigh from the mother indicates her weariness in managing such tensions:

Interviewer: Do you ever get conflicting advice from professionals?

Dad: All the time.

Interviewer: What do you do?

Mum: [sighs]

Dad: Well we just ...

Mum: I would chat ... sometimes I challenge them ... I'd say to them, do you know, 'That's not what you said two weeks ago.' (Family 23)

Another parent spoke of taking charge of making treatment decisions in the face of conflicting advice:

When there was conflicting advice we would take it all on board but make up our own mind. (Mum, Family 21)

Much like the parents above, the parent quoted below also spoke of receiving differing advice and told of the effect that this had on her:

I used to back off a lot and just kind of whittle down into my shoulders and just 'Okay then, okay,' and in the back of my head I would be thinking 'But so and so told me to do this,' ... but I've learnt to stand up now and just go 'Well, actually, the doctor here, that I very much trust and value their opinion, said this.' ... completely contradicts what they said. (Mum, Family 15)

The quotation above suggests that parents sometimes appear compelled to challenge the different advice given by practitioners.

5.2.5 Summary

Parents heard, and then accepted or rejected, the advice from practitioners regarding the management of their child's symptoms. Key features which mediated the uptake or rejection of advice were the practitioner's knowledge of the condition and their relationship with the child. With parents often becoming experts there were inevitably times when they challenged the advice offered by practitioners. For some this had a feeling of liberation, for others, a sense of isolation and heady responsibility.

5.3 Couple relationship

Many parents shared accounts of their relationship during the interviews. Not all parents were in a relationship, however, and two interviewees were lone parents. Interviewees were asked to reflect on their relationship and how they managed as a couple. Those that were lone parents spoke of the impact of the child's illness on their previous relationships. Overall, the data indicate a sense of enduring tensions in parents finding time to be together. This observation is reminiscent of couples with children who are well, but warrants reporting in this dataset because this difficulty in finding time for each other was reinforced by the isolation which many felt. In turn, this difficulty is linked with the rarity of the condition and parental experiential expertise, which meant that asking others for help was difficult since few others understood how to support their child. Thus, this data set is intimately linked with the model through the rarity of the condition and the development of experiential knowledge.

5.3.1 Time together

Parents described the tension in finding time to spend with each other in the context of competing caregiving demands. For some, this was a balance that could not be struck, while others described finding ways of packing lots of activities into short periods of time. Their accounts created a sense of the pace of living being dictated by the availability of respite:

It's just so difficult, to get someone that can baby-sit in an evening to let you go to things is fraught with trying to get somebody who's capable. (Mum, Family 13)

Several parents explicitly reflected on how caring for their child resulted in changes to their relationship. The following couple (as with Family 13 quoted above) connect this with the difficulty

in finding people who are willing and able to look after their child, despite believing the child with the condition to be easy to manage compared to the siblings:

Mum: I think we've changed ... our relationship's changed.
Dad: Hm mm.
Interviewer: In what way?
Mum: Because ... it's ... I don't know ... it's because one of us is always up doing stuff.
Dad: Uh huh.
Mum: And you never get any sort of quality time ...
Dad: You never get time together.
Mum: There's very, very few people looking after [child]. That baby-sit for him and he's the easiest one to look after.
Dad: I can't understand it. Cannot understand it.
Interviewer: And is that ... that ... nobody offers or
Dad: I think they're just scared. (Family 10)

Thus, parents struggle to have time alone as a consequence of having few people around them who they can ask for support in childminding. Parents feel that few people have the appropriate skills, or feel comfortable taking on that role.

Couples talked about the ways they manage family life and their own relationship. Several couples spoke of purposefully organising their caring in shifts, so that the child is always with a parent. While this means that the child is always in the care of someone with experiential knowledge, a clear consequence is that parents rarely get time together as a couple.

Several families spoke of how they managed the division of caregiving labour. For some, this took the form of going out to work in shifts. For others this manifests in taking turns to sleep with the ill child, thereby taking responsibility for monitoring the child during the night, but also impacting on the parent's own ability to rest and have time together:

Dad: We don't really get a relationship like normal people would have, dare I say it, a cuddle in the morning and various other things, we just don't get time to do anything, it's, you know.
Mum: Well, we're, [partner] often sleeps upstairs, I sleep downstairs.
Dad: We take turns don't we? Swap beds so ...
Mum: And then, yeah, and then I'm often in with [child]. (Family 22)

Some couples share required tasks, while others reported a reliance on one parent. The excerpt below illustrates a mother reflecting on how completing symptom diaries provided an opportunity for the father to be more involved in their child's day-to-day care:

Him [partner] doing these diaries, that is something he does, because I do everything else on the boys and [partner] has been doing these religiously, haven't you? So it's been really good. (Mum, Family 6)

Thus, many couples spoke of having reduced time together and the strategies they had developed to mitigate the impact of this on their relationship.

5.3.2 Couple communication

Across the interviews there was a clear sense of parents wishing to be 'open' in communication with each other, driven by an understanding that this was a key part of coping. While couples reported open communication as a goal, some managed to do this, while others did not. The following two excerpts illustrate this drive toward being 'open':

Dad: We have to be very, very open.

Mum: We don't hold nothing back. (Family 12)

We talk to each other about anything and everything openly. (Family 15)

However, many parents struggled to talk with each other about their worries. Parents kept concerns to themselves and struggled to talk with their partner about their feelings. The following extract illustrates how the child's dad often worries about his child, yet never raises this with his wife:

Dad: [And I often think], 'Is he going to make it through the, through the rest of the day, is he going to make it through the night?' ... that kind of thing, it's in the back of my mind but I, you, I try not to show it or express it or anything like that.

Interviewer: Do you talk with your wife about that?

Dad: [Slight pause] Not really I don't think I, it's something personal I keep to myself.

Interviewer: You keep to yourself ...

Dad: Yeah ... I don't mention it ... I, 'cause I, she, I don't know ... I worry about him more than I worry about the rest of the, the other three [siblings] ... you know and it's, if, if he's not well and I'm at work he's, he's just on my mind but I'm just thinking you know, 'Should I just go home and just check up on him?' Or make an excuse or something ... and just check up on him and see how he is.

Interviewer: Do you think your wife knows that you're doing that, though?

Dad: I [hesitates] I don't know, I don't ... I'm not, I'm not sure ... but I, it's something I, I would do, but I don't mention it to anybody else. (Dad, Family 16)

Couples also spoke of adopting different stances in communicating about the condition and its impact. Some families used the language of defence mechanisms, such as denial, to articulate their ability to talk with each other. Changes in the child's health often acted as prompts for the parents' communication style to shift, as the following extract illustrates:

It was literally after I had come out of that meeting with [specialist paediatric consultant] ... that I was able to start opening up, I would start saying more to [partner] which was obviously helping her more because she felt she needed to talk about it ... and I literally was cocooned in my own little world in denial. You know, I just take it a day at a time, I still do, some days are better than others but I feel a lot more positive. I feel more positive for having [our children] than not. (Dad, Family 6)

This couple go on to discuss communication in more detail later in the interview. The following extract illustrates how discussions with clinicians had facilitated not only greater support between the couple, but how this fitted with the care of their children, referred to below as 'the boys':

Interviewer: It sounds like there's a real support between the two of you?

Mum: Yeah, yeah, I think so.

Dad: Yeah, I think it's the sort of thing that either makes you stronger or pulls you apart, thankfully it's pulled us together ... I'll cope, but again it's only since that meeting when he [consultant] dragged it out of me and I've started talking about it.

Mum: Yeah it was really good.

Dad: I probably did feel the weight lift from my shoulders, you know, I didn't feel like I was alone.

Mum: And then they sort of concentrate after they've done with us, we can work on the boys then and that's what he did, isn't it, at the next meeting, it's 'Right, you're okay, now let's do the boys.' (Family 6)

Some parents spoke of how they provided balance for each other. For the following couple, there was a sense of one allowing the other space to express frustration:

[My partner] can find it really, really, hard he'll openly say, you know, at times when it is really hard and he stresses out and snaps and that's when I've got to keep my cool and talk to him about it later and say, 'Actually, when you did that I was really upset as well, but you can't snap like that,' and it works the same in reverse as to when, you know, if I get upset about something. (Mum, Family 5)

Some couples also valued the opportunity to talk to people outside of the family. Though not everyone wished for counselling, one family reported how useful it had been for them. The following excerpt comes from a section of interview after the mother has stated that she has depression, and has sought counselling to help:

I talk to her [a counsellor] about things that I don't even want to talk to [partner] about really because it's ... I'm a very positive person and I don't like to sort of voice negative feelings so it's a sort of safe place for me to do that really. (Mum, Family 3)

One father speaks of his struggle to voice his feelings. This was initially difficult because of the emotions he was experiencing, however, as time has progressed he explains his minimal communication as characterised by a lack of time to think about his feelings:

Dad: I mean I just ... as I say I was working, I just buried my head in the sand and ... we don't really talk about it.

Interviewer: Has that changed now? Is it a conscious decision that you don't ...

Dad: You don't really have time to talk about it. (Dad, Family 10)

The above quotation is taken from a section of the interview where the parents reflect on their communication pattern. They surmise that their ability to communicate openly with each other coalesces around their ability to consider the life-limiting nature of the child's condition. Consequently, struggles in communicating with each other can result in tension because one partner is 'burying their head in the sand'. Such difficulties prompt concern about how such couples manage to undertake conversations with each other (or practitioners) regarding the future and planning for their child's end-of-life, when one parent is able to think ahead and one is not.

Indeed, some parents indicated that communication became difficult regarding treatment decision-making. One family, quoted below, disagreed on whether their child should have surgery, a decision which was focused on potentially life-and-death outcomes. Such disagreements, of course, add tremendous stress to a couple who may also be struggling to communicate about the most basic of needs/issues. The following quotation is from a dad who reflects on the struggle the couple had in making the decision, the role of other people (including family and physicians) and ultimately, his own authority within the family:

I kind of had to like stamp my authority [laughs] and it ... not in, in, in, in not that kind of term but you know, because I just felt that I wanted to do all the best that I could for him, because the doctors were saying you know 'He's got months to live,' you know, when they say that kind of thing, it really scares you kind of thing, what do you do? But then they give you the options, 'He could die on the operating table, he could become paralysed from the neck down, or he could make it through the operation,' ... and you always hope for the best really and, you know I think [my partner] was against it, 'cause a lot of people were saying 'Don't do the operation, don't do the operation,' you know, 'Do not do the operation,' and, and I just felt that I really wanted to do everything I could for him, knowing that I'd done everything and ... it was quite, it was quite tough, tough decision to make and [my partner] was against it but I just felt that, 'cause I was going by what obviously the doctors knew and did, seen other kids ... obviously if things had gone wrong I don't know how I would, how I would be reacting now, but obviously things have gone right, so I could say the decision was a good decision. (Dad, Family 16)

Thus decision-making about treatment options could prove particularly difficult for couples, and at times both highlighted and contributed to relational difficulties. Indeed, several families commented on relational breakdown, speculating that this is a consequence of communication difficulties:

I've seen couples with some of these children, I've seen the marriages break up, you come up here and suddenly that partner's not there anymore because they couldn't cope with it, is that because they don't talk about it, you know, I don't know ... we talk about every aspect of the illness. (Mum, Family 19)

5.3.3 Relational difficulties

Some of the interviewees indicated a sense of unresolved relational issues, which are brought into focus around the illness, both in managing symptoms and also in articulating notions of blame for the condition. Other couples spoke of their uncertainty about whether they would split up following the death of their child. Thus, for some couples while they try to maintain their relationship, there is a sense that all this is up for negotiation in the wake of their bereavements.

The fragility of the couple relationship is illustrated in the extract below. The dad speaks of himself as an individual after the child's death, rather than as being part of a couple. This prompts the interviewer to ask about outside support for them, and the father's response that there is little that anyone could offer to help:

Dad: And I'll probably need a few years to get away and just sort my head out, you know, no family, no friends, just, just go basically and ... who knows, we might not be together in the next couple of years.

Mum: [to the researcher] Lovely isn't it? [Laugh]

Dad: You know, it is ... No it is, no, well you take, you think, you're going to think about this ... you know, sometimes we don't see eye to eye. No, you might as well ...

Mum: Yeah ... It does go, does cause a strain.

Interviewer: Do you have any sort of support outside, you know, counselling and GPs, are they supportive?

Dad: But what, what can they do anyway? I mean they can, they can sort of guide you in what you should be doing or you shouldn't be doing, but at the end of the day you've just got to deal with it yourself. (Family 22)

In some instances, the biological father had left the family. The mother in the following quotation indicated at the start of the interview that her partner had also recently left the family, resulting in her becoming, for the second time, a lone parent with three children:

Interviewer: So has it been you on your own most of the way through?

Mum: Yeah their dad left when [youngest child] was ... two ... I think he was two, [the biological father] doesn't have anything to do with [child] and he hasn't seen them for a year I would say ... so yeah I'm used to being on my own ... our little family, 'Aren't we [youngest child]? ... got our little family, eh?' (Mum, Family 4)

Another parent had been in a troubled relationship, and had ended the marriage to her children's father:

I want the beginning of a new life where I'm head of my household and I've got my family and we have our life on our terms. (Mum, Family 9)

Parents managed their couple-relationship and their relationships with their child/ren in the wider familial context. For some there was a clear impact on the parents from the wider relational systems (a feature which is discussed in greater detail in section 5.4). One such example came as a family reflected on how they argue when they are stressed, and this is then taken out on the other sibling:

Dad: There's been certain points where it's been quite stressful and it's, it's only come out in other things, it's normally, it's discussing [child's] illness you realise that you start having a go at each other for, for something stupid and you think, 'That's not like me, it's not like [my partner], it's ... this is all sort of getting to us a wee bit.'

Mum: And then [sibling] ends up ...

Dad: Aye, and [sibling] will get the brunt of that. (Family 5)

5.3.4 Dealing with things differently

Many interviewees indicated that the couple had different ways of coping, often with one seeking more in-depth communication than the other is able to tolerate. At times this was framed as reciprocity, and at other times as a tension for the couple. The following extracts illustrate how couples view different coping styles as reciprocal and helpful, particularly when coping with feeling down, or managing the child's challenging behaviour:

Mum: Well, luckily if I'm low [my partner's] normally not low, and if he's low I'm the one that's going, 'Hey, come on, don't worry, we'll be alright, we'll get through this.' And luckily so far it's always worked out that like, hasn't it?

Dad: Yeah.

Mum: 'Cause one's not low, it's, we're not both low at the same time. (Family 22)

Dad: I might just say to him something, you know, '[Child], for God's sake will you be quiet?' or whatever, and that is when [my partner] will step in ... whereas –

Mum: Whereas we tend to do it if I am getting like that as well, [partner] will step in so and then one of us is calm. (Family 6)

My weaknesses are his strengths. (Mum, Family 12)

Other families described reciprocal coping styles in responding to physical symptoms:

Mum: In the past you've got more panicky, say if like [child] got a cold or something he'd be going, 'Get down there and get some antibiotics for her, it might go onto her chest,' and I'd go, 'Just, just let it ...'

Dad 16: No, I do though, I just ... I have worries. (Family 22)

However, not all families reported different styles as a positive feature of their relationship. Several families indicated that having different ways of coping resulted in tension and difficulty. As indicated in sections above, this would often come to the fore when parents had different ideas about what they wished to know regarding the deterioration of their child's condition, and thinking about end-of-life care. The two following excerpts illustrate speakers who adopt opposing positions in wanting to know the detail of how the illness will progress, and wishing to be protected from this knowledge:

I try to prepare [partner] 'cause I see the route we're on whereas he's really naïve, he doesn't want to look ... go there. So you have to try to find that happy balance so that then, I'm not annoying him and not getting him upset and then he thinks he can't protect me it, it ... it's difficult we just ... we just, I don't know I suppose ... we just have our own way of, of dealing with it. (Mum, Family 11)

[Partner] felt a need to really know much more in-depth exactly how the symptoms are going to alter and, you know all sorts of things, and I says you know, I am quite happy not knowing. (Mum, Family 13)

5.3.5 Summary

Many couples spoke of the struggle in finding time for each other. Many described organising caregiving as taking shifts, resulting in limited time to spend with each other. Parents often gained support only from each other, resulting in a great deal of interdependence. Their refined expertise in managing the child's symptoms was a key feature in maintaining and renewing this dependence on each other. That is, the lack of knowledge and expertise from other people in their lives actively restricts the support they are able to tap into.

Couples also spoke of the degree to which their coping styles were complementary or conflicting. Where different coping styles are reciprocal this provides strength, but where they are at odds with

each other there is potential for relational tensions to develop. Such tensions were often apparent when parents were confronted with discussions around treatment decision-making.

5.4 Wider family system

The wider family system includes grandparents, uncles/aunts and siblings of the child with the rare life-limiting condition. The data reported in this section reflects the different perspectives and roles each of these individuals takes, from the parents' viewpoint. Siblings were interviewed themselves, and their views are reported in section 6.

The actions and approaches of each member of the wider system each impacted on families' experiences of caring for their child with a rare life-limiting condition. Though each family differed in their relationship with the wider system, on the whole, parents reported working hard at supporting relatives. Consequently, many parents spoke of limiting how much they told relatives and of the impact that this had on the support they received. Families spoke of the duality of how their child's condition had changed relationships for the better, but also highlighted pre-existing relational difficulties.

5.4.1 Communication with the wider family

Though there was some variation in the data, many parents reported feeling unable to communicate the breadth and depth of their experiences with family, driven by a desire to protect others from knowing the details of what was happening. This would often take the form of actively choosing not to discuss their feelings or the health and wellbeing of their child with relatives. One parent explained this as:

We sort of just glide over for his mum and dad's sake, but they know most of what's going on, you know they just, they know. (Mum, Family 19)

Thus, there was a sense for this family that there was not a need to labour full descriptions of the child's condition, yet the paternal grandparents were intuitively able to know how things were. In other families, relatives would demonstrate an interest in hearing how the child was, but such enquiries were received as perfunctory, and did not lead to active support through offering hands-on care:

Mum: If they asked you know about [child], right, what they want to know then she does now talk to them and tell them about it.

Interviewer: Mmm, but it's mostly about them wanting to have an update on how he is?

Mum: Yes. (Mum, Family 16)

Nobody really talks, I mean you get a phone call, 'How's ... how is he today?' that's as far as it goes ... I've got a sister nearby but she never ... don't really talk much. She ... likes to think that she's the big caring aunt, you know, tell everyone else, but ... I couldn't tell you the last time she was in the house, so that's a wee sore niggly point. (Mum, Family 10)

Many families did not communicate openly about the diagnosis and subsequent implications. This resulted in parents feeling under-supported:

We felt like she [mother-in-law] could have been more supportive and ... she saw me go to hospital every week for a year to try and find out what was wrong with [child]. (Mum, Family 22)

In contrast, some parents spoke clearly about how much support they had received from relatives:

My father's been an absolute ton of support and, and I can lean on my dad an awful lot and I can say anything I like to my dad about any worries or concerns ... I've updated my dad on, on things as well, I said, 'Look Dad, you know, you'll maybe see things that, that I don't see and if you need to overstep that, that's fine ... you know you've got my permission to do so.' ... you know, that sort of way so we are a close, a close wee unit. (Mum, Family 11)

For the above family then, the maternal grandfather was an important part of the support network.

5.4.2 Openness to providing support

Interviewees reported a variety of patterns in the amount and type of support which was offered and accepted from the wider family system. For some families, there had been a conscious choice to move toward or away from other relatives:

[My husband] is an only child, my family's down in [city 500 miles away] ... so there's not much access. (Mum, Family 19)

This parent then goes on to say that the family do have a good supportive care team, constructing a sense that the two networks of support are inter-changeable or at least serve similar purposes.

Other families chose to move house, feeling unsupported by local families and therefore having less to lose. Some explained this with reference to relatives not understanding the child's lack of control over their symptoms (and not understanding the experience of living with a child with this condition):

*Dad: We moved down to [county] 'cause our family were not supportive generally, to go away, 'cause they just did not understand. When [child] went through a hyper stage they weren't being supportive, they were like going, 'Oh, shut her up,' sort of thing.
Mum: It was all, 'Bad, naughty child.' My mother was like, 'Tell her to share! She's got to learn to share.' (Family 22)*

The same parents reflected on the difficulties that the wider family have in relating to their child. The following quotation illustrates this family's hypothesis that it is a lack of understanding of their daughter and the symptoms of her condition which lead to a barrier in fostering adaptive and supportive relationships:

*Mum: Neither of them know how to connect with [child] at all ...
Dad: They won't give her a hug and all.
Mum: And my, and my mum and ... my mum's really tiny and, and [child] will throw her cup and she'll go, 'Oh! Oh!' Like that, all the time, and you'll just go, 'Oh, shh, why are you making that noise?' You know? 'She's not going, she's not trying to hurt you, so ...'
Dad: There's just no understanding, that's the thing. (Family 22)

Dad: My mum and dad don't understand it.*

Mum: [His] mum is sixty two, sixty three and she's, she can be very firm in the sense of if [child] does something it will be a snap on 'No, [child],' you know, and [child] just takes a step back as if to say 'What are you shouting at me for?' you know ... she always means well and she tries to help and she comes round once a week and spends a couple of hours here doesn't she, and helps to bath the kids things like that but she doesn't understand [child's] illness. (Family 15)

Following on from the idea that not all family members truly understand the condition, or what it is like to support children with the condition, other parents reflected on how helpful relatives sometimes were. Indeed, in one family because grandparents really understood the condition it made spending time with them as a family much easier:

We tend to go to family members because it's difficult taking [children] to friends because of their behaviour, you know, it's not everybody who understands, although they try to, but it's just getting that people who know how to help. (Mum, Family 6).

When asked about the involvement of the wider family one parent's response was to laugh at the question, following up this reaction by talking about the people who are involved in supporting the family. The parent talked about particular friends who had an understanding of children with special needs. The ease of accessing support from friends was thus related to knowing about specific symptoms and challenges, rather than the specific life-limiting condition itself:

I have friends who have special needs children because I find they understand us better than anybody does and what's really nice is instead of, as, as you would be normally with, with a special needs child in a normal situation and they start doing something very odd, whether it's chucking things about or, or that kind of thing and normally you'd be apologising all over yourself and sort of going 'Oh, my goodness!', and that's, when you're with other special needs families you never have to apologise because in two minutes it will be their one that's doing it and it'll be that one's doing it and so it's a case of, here we go and there's a kind of, it's a lovely supportive kind of accepting. (Mum, Family 13)

Knowledge of children with special needs was also indicated by other interviewees as important in providing reassurance for both the parents and the wider family that they had the skills and abilities to connect with the ill child:

My mum used to work with special needs children before we knew about [child] so she is really on that wavelength of understanding, she uses the same care in the children's nursery with the special needs children that came round ... and learning difficulty adults she works with as well, so she understands. (Mum, Family 15)

For many families, however, there was little offered by way of support. The following quotations are illustrative of the difficulties which some parents reflected on the wider family's roles:

*Dad: They're dead nice and all but they wouldn't be involved.
Mum: There's, there's no like big family feud or anything.
Dad: No, no they wouldn't be involved. (Family 21)*

My family are really close but they all find him very difficult, they are fantastic with [sibling] and they will take him off and have him overnight and then [sibling] will go and do lots of other things with my family but ... nobody really in the family can, can, cope with [child] on their own. (Mum, Family 3)

Other families struggled in the context of a range of other health concerns:

My mum's got quite a lot of her own health needs and isn't always able to ... but if my sister's ... if my sister's around if not then my brother's girlfriend if things got to crisis point, do you know, then she would, she would come round. (Mum, Family 15)

5.4.3 Managing relatives' feelings about the child's condition

Parents often took on a role of trying to manage other people's emotional response to the child's condition. As indicated in the section above, many actively chose not to discuss symptoms with parents as a way of protecting them from knowing about the progression of the disease or the day-to-day difficulties which parents were managing. As the following mother indicates, telling relatives in detail about the child's condition can be very upsetting:

I would talk to my mum like on the phone but not too much 'cause when mum gets very, very upset and she is very upset about [child] and, she finds it quite hard so it's hard talking to her when she's ... she would almost be more upset than I would be. (Family 21)

Worry about dying was clearly a concern for some relatives, and parents played a role in managing others' anxieties about this. The following extract begins with a conversation being reported, where the child's uncle was encouraged to get to know the child. Another uncle was upset at the idea of spending time with a dying child, resulting in considerable tensions:

'If you ever come over to take him out for a walk that would be really nice,' and then [child's uncle] accused [child's dad] of trying to fob his dying child off on him so we haven't seen him since either. So that's [child's uncle], [another uncle] finds it very difficult to cope with [child] just because he, he, gets, I think he just finds it distressing to think that he has got a life-limiting illness, I think he would rather not deal with it. (Mum, Family 3)

Other parents reported differences within the family. The following extract illustrates how the maternal grandmother was overwhelmed by the diagnosis, and the maternal grandfather combines stoicism taking it 'on the chin' with a sadness expressed through using the past tense to speak of the child. The reactions of these grandparents is presented with a clear sense of loss, as the mother reports hearing other people reflect on the young man the child will never become:

Dad ... has taken it very, very hard. His wife died a few years ago and he lives on his own and he still struggles. You get a lot of, 'Oh, I can't believe what a lovely young man he'd have made,' and ... and a lot of that which I find hard to listen to, that I just ... I can't think down those lines, or 'He was such a lovely boy,' and I think ... my dad just sort of takes things on the chin but he is like that, and my mum ... took it terribly. When we actually told them we thought we were going to have to call the doctor out she was that bad and she's been to the doctor's and she's on medication really, just sort of anti-depressant. (Mum, Family 1)

As reflected in the data reported above, the majority of families indicated that they did not receive a great deal of help from relatives. Some parents did speak about the people that are most supportive to them. The following parent talked about friends who take on supportive roles, and how this contrasts with her family who she feels protectiveness toward:

Mum: Funnily enough though, I wouldn't say my mum and my sister. And they'd be mortified at the thought of saying that, but I don't actually say that much to them 'cause I think the more I say things the more sometimes it upsets them, particularly my Mum although she's great with the help, I sort of hold back a bit from them really.
Interviewer: So, protecting her in a way.
Mum: Yeah. Yeah, and my sister. (Mum, Family 1)

Thus, the data indicate that much energy was required for managing communication between family members and managing others' emotional reactions to the diagnosis and ongoing progression of the condition.

5.4.4 Changes to family relationships

Many parents had spoken of the impact of the condition on their own relationship, as reported in section 5.3. Parents were also asked to reflect on whether the child's condition had impacted on relationships within the family. Several interviewees indicated that the illness had brought the family closer together, by pulling people toward them and opening lines of communication. The following family explicitly frames this as reflecting a desire to focus on the positives that the illness has through the family not the negative effects:

Dad: [It has made our family] stronger, it's pretty much healed a huge gap in terms of extended family, there was a sort of huge divide in our family and [child] has bridged that.
Mum: Not immediate family, but extended family.
Dad: I said extended family, there was a huge sort of bridge, sort of ... or there was a huge divide in extended family and [child] has bridged that, you know so she's, we always look at the positives that she's brought, rather than the negative. (Family 12)

Other families reported that the condition had not changed how the family related to each other, but that the diagnosis had firmed up hostility between individuals within the family. The child's mother is blamed for bringing illness and disability into the family:

Interviewer: Have things changed in the broader family since he was diagnosed?
Dad: Still pretty much the same really, there's not much difference. There's a, I think, I think once mother-in-law doesn't like daughter-in-law it really changes, it's really difficult.
Interviewer: I see, so that's the main factor.
Dad: Yeah I think, I think that's it, I mean that, that really changes you know ... 'Other daughter-in-laws are better.' (Family 16)

Stories of isolation were a common feature in the data. A family spoke of feeling hurt and angry because of a sense of emotional solitude from their extended family who fail to understand the magnitude of their experience:

Mum: You get angry. You go through a stage where you get very angry that no, why can't ... a while, a little while ago I couldn't ...
Dad: No one phones.
Mum: I couldn't understand why nobody loved [child] like we did, you know, 'cause ...
Dad: Or they could just phone like my brother ... I know everyone's busy and you have to think that way, but we're more heightened 'cause obviously [child's] ... But they could just phone for five minutes and say, 'How is she?' But nothing. Nothing! (Family 22)

Even in families where little had changed relationally, often there were large shifts in the pragmatics of organising everyday life. The illness was often reported to impact on the family's ability to undertake regular activities (such as shopping, cinema visits and visiting friends). In the context of family relationships, these practical difficulties led to relational tensions, as the child's needs made fundamental shifts in how the family related to the child and to each other. The following father reflects on how they coalesce around their son's needs and the parents and siblings become 'slaves' to his needs:

We are slaves to him, that's, that's the way it is, but we feel sorry for him because you know he can't do much on his, on his own. (Dad, Family 16)

Indeed, siblings often spoke of how they had been impacted by the ill child's condition. Though no siblings framed changes in relating patterns as powerfully as the father quoted above, many indicated that the ill child had taken a central position within the family, from which many if not all activities were based around. Sibling views are reported in greater detail in section 6.

The diagnosis of a rare life-limiting condition also led families to making important decisions about how many children they might have, and to prepare themselves for what the death of their child would have on them as a couple and on them as parents. This is described more fully in the heritability part of the model in section 5.5.

5.4.5 Summary

The wider family system was clearly impacted upon by the child's condition and subsequent need for care and support. The impact on the wider family and how much they were able to offer support was often, but not always, associated with the level of detail that parents felt able to give others about the child's symptoms and overall condition. Having little information or awareness of the behavioural and physical consequences of the condition meant that members of the wider family system often found it practically and emotionally difficult to support the parents and ill child.

5.5 Heritability

The heritability of the condition was raised by most families in the interviews, despite this not being something that they were formally invited to talk about. The presence of this as a strong strand of data reflects the importance of heritability in the overall experience of supporting a child with one of these rare life-limiting conditions.

5.5.1 Genes

Some families experienced difficulties in communicating the idea of genetic heritability to the wider family system. For the following family, there was a distinct sense that the grandparents could not tolerate the idea that the condition had been inherited. The child's father illustrates this in the interview by talking about how discussing the autosomal recessive genetic pattern is not accepted by the wider family:

If you try to explain to them [the family] about the genetics of one in four and all that, they really would not understand they would still turn around and say, 'No, it's ... that's got nothing to do with it,' ... I would say it's the mother-in-law, probably like my mum, you know where, where education isn't ... or because they've just like had 'normal' children and they assume or expect that, you know, their grandkids should be 'normal children.' (Dad, Family 16)

This father goes on to explain that a lack of understanding the role of genetics led to blame within the family:

[My mum] kind of like blamed it on my wife and ... 'It was your fault,' and this and that and everything else but, that's why I'm saying that they don't understand the concept of how, you know ... the, the genetics. (Dad, Family 16)

Some families reflected on blame from a different perspective, identifying that there was little they could do, since it was inherited from both parents:

[It is a] genetic one as you'll know and, fortunately it comes from both Dad and I, so then there's not any kind of, you know it's your ... any blame or any of that kind of thing. (Mum, Family 13)

Other families talked about the idea of genetics more light-heartedly:

We must have been really wicked in a past life. (Mum, Family 6)

The genetics in our family aren't exactly the, the greatest so ... [laughs]. Do you know, when bad genes were handed out we certainly got our fair share [laughs]. (Mum, Family 23)

Some families demonstrated a great deal of knowledge regarding the role of heritability, and consequently became a concern among other relatives. This was at times among the parents' own

siblings, and on occasion about future generations (that is, parents thinking about their own children's children). The following father explains how this played out in his family:

My sister's just had her third [child] actually, so they're going to be tested ... my brother wasn't particularly bothered, as brothers are, whereas my sister's been very worried about it. Whether her children are carriers, whereas until last time [my brother] came to see [our son], and he hadn't seen him for a while and I think it really hit home that, 'Oh, good Lord,' and I said, you know, 'Imagine if your ... your [children] are carriers, and they meet somebody and they are, you'd never forgive yourself, would you?' (Dad, Family 1)

The notion of guilt is clearly articulated in the above account, related to the responsibility of acting on the knowledge that one may be a carrier of the affected gene. Another family explained how they were preparing for their first grandchild, and the uncertainty about who carries the gene, and the need to prepare for the possibilities for another baby with MPS disease:

*Mum: [My husband's] son, is expecting their first son, so his grandchild ... and obviously we have explained to [my husband's son] ...
Dad: Oh, aye.
Mum: About the MPS side of it and because we don't know if [my husband's son is] a carrier or not ... and obviously for his partner as well.
Dad: We don't even know why we're carriers. (Family 23)*

Another family talked about how they had been trying to get pregnant when their first child was diagnosed, and how this impacted decision-making about having a larger family:

We were actually trying for another baby when we got [our son's] diagnosis ... so that went on hold completely. Our thoughts at the time were we are never going to have any more children, we are just going to concentrate on [child] and give him the best life we can and you know ... but then we changed our minds and realised we still want more children. (Mum, Family 15)

For some families heritability was a significant struggle to come to terms with, but nevertheless there was a sense that this led to a greater level of understanding of the condition, often over and above that of the practitioners around them. The following mum describes how her mother-in-law was aware of her child's condition long before healthcare practitioners had made a diagnosis. Yet, despite familial expertise in knowing the condition, the family communication patterns were such that such experiential learning was precluded:

[My] mum's brother had two children with [MPS disease], which I had never met. They died before I met ... but there was no support or ... I kept on asking [the relative], 'What do those girls have? What do those girls have?' And she said, 'Oh nothing, they should never have got together, that's all that was about.' And she wouldn't help and, and support me, and then when they found out [our daughter] had [MPS disease], apparently [my husband's] uncle had gone to [my husband's] mum, and said, when [our child] was two-and-a-half, 'That little girl's got [MPS disease].' So she hadn't told us. She, they could see that [our child] had [MPS disease]. (Mum, Family 22)

The sense of parents becoming the experts was articulated by other families, where they identified characteristics of MPS disease in other children in the family before professionals began to suspect the diagnosis:

*Dad: [Our niece] has to be [MPS disease], she has to be 'cause she's got ...
Mum: There was too many people that had seen it, do you know ...
Dad: She's got identical to [child].
Mum: There's too many, and if not,
Dad: The same actions, the same, the same hair.
Mum: Yeah [laughs].
Dad: The same kind of hair, that strawy hair and,
Mum: Yeah. Yeah, I mean there's too many people, it wasn't just us, it was the doctor at the hospital had ... do you know, and he didn't know what we already knew ... so you see...
Interviewer: So [the consultant] wasn't informed by that?
Dad: [The consultant] looked at the photograph and said, 'That child's got [MPS disease].' (Family 23)*

One family reported that the extended family included several other children with MPS disease; indeed dad's biological uncles had died from MPS disease as children. Most recently, dad's sister who lives in London has had a child who has been diagnosed with MPS disease. Mum told the interviewer, 'I noticed but I didn't like to say anything ... because of the shape of his backbone,' in reference to recognising the tell-tale physical signs, but not wishing to break bad news to the family. The doctors told the sister-in-law that: 'The back would go back in, but I knew from experience [that this was not the case].'

5.5.2 Not being left alone

Several families spoke about how having a child with a life-limiting condition had put them in the position of thinking about having other children. In particular, parents spoke about their worries of either having no children at all, or having an only-child, when their child with MPS disease or Batten disease died. For some, this had led to planned pregnancies to ensure that these scenarios did not occur:

We originally said 'Right, we're not going to have any more,' and just went through this whole thing of it would be selfish of us to have any more children, we've got to concentrate on [our ill child]. Once we got past that I kind of accepted the fact that one day I would end up with no children which scared the living daylights out of me because I always wanted a family. If I had more money I'd have a nice big family ... but we are both very quickly agreed that we couldn't do it again through choice ... and we both said 'We'll have the testing and if it comes back as a positive result we'll terminate,' and we just couldn't do it. We both knew that we couldn't do. We couldn't go through it twice ... and we were lucky because the next two times when I got pregnant they were both negative results so they [our other two children] are both carriers but not affected. (Mum, Family 15)

Another mother articulates a similar dilemma, wishing that her surviving child would still have a sibling to be close to in the future. She draws on her own experiences of being an only child to reflect on how lonely it might be when the ill child dies:

[Sibling] would do anything for him and that was the reason that I had [another child] was, those two were like twins, very, very close ... they still are exceptionally close ... and my fear was, when I found out about him it, to have that separation then be an only child would be too much ... when you're so close to someone ... so hence ... the third [child] coming along was for a reason so that she had someone to talk to ... discuss things with and later on when, when needs be, has that back-up other sibling, so, yeah. Whether that's right or wrong I don't know but ... I think, 'cause I'm an only child I think it's ... it would be quite lonely. (Mum, Family 4)

Another mother spoke about her desire not to have an only-child, after their child with MPS disease had died. Thus, again, the parents were keen to have another baby:

After [son] was born we had to think of, okay, some day we're going to be left with an only child, is that what we want? Do we want to just have [sibling] then some day and him going 'I've nobody to play with now,' after [our third child] appeared we, we ... again we said, 'Right, we'll be thankful now that for what we've got, we've got three beautiful children, that's it now, we're calling it quits ... and, lets just enjoy them and savouring every moment.' (Mum, Family 11)

One mother reflected on how she felt when her child was initially diagnosed. In the following quotation she indicates that on the day of her child's diagnosis she had thoughts about a desire to have other children:

I did a lot of crying and wailing, and I was saying, 'My daughter, my daughter, my little girl,' and I just talked to the doctor for about half an hour, I think. Roughly. Vaguely. And then I asked if I could have more children, because I don't suppose I can, because I thought I probably couldn't, because I didn't want my son to be alone, and I didn't want him to lose his sister and then have nobody. (Mum, Family 9).

The mother quoted above then went on to talk about how at her daughter's diagnosis she already harboured concerns that her son also had undiagnosed Sanfilippo. The desire to not be left alone herself had prompted her to seek a further pregnancy.

5.5.3 Testing other children

The discovery of an inherited condition had frequently led to discussion about genetic testing of other children in the family. The timing for testing other children varied considerably by age, but was often conducted very soon after the first diagnosis was made:

Straight on the Monday, after we got the diagnosis on the Saturday, [my wife] went down with her parents to get [our youngest daughter] tested and that took a week, so that was ... so from the Monday morning to the Friday. (Dad, Family 1)

She, when I had him diagnosed, she [another child] was six weeks old ... six months old, so we had to get her diagnosed as well in case she had it, so that was quite stressful. (Mum, Family 4)

Contrasting with the section above, for some families the diagnosis of one child also led parents to question having other children. As one couple describe, having one child diagnosed with an inherited life-limiting condition is 'terrible', and yet, an additional layer of difficulties came with processing how the discovery of a genetic condition impacted on their decision-making about having other children:

Mum: What is horrendous ...

Dad: During that [phase just after diagnosis] ...

Mum: Is it ... it just lets you get along to the second child ... and then, you know, and then you know, if you ... if we were to have known at least we'd have gone into it with our eyes open. But to have not known ...

Dad: I think ... I think if you'd have known ...

Mum: ... had it as well ...

Dad: [younger sibling] may not have been here. (Family 1)

The quotation above situates parents often in the throes of a life-stage where the diagnosis of MPS disease or Batten disease occurs when another baby has already been conceived or born, resulting in anxiety that another child may also be affected. Another family spoke of the stress of this period:

We had to wait for six months or so ... and she got tested to find out if she was ... Batten's ... as well, which was putting a strain on everybody, I think. (Mum, Family 10)

Some families tried to live with the uncertainty of not knowing a second child's health status, holding-off testing, and knowing, for as long as possible. The following quotation illustrates parents describing the time where they delayed testing their youngest child to try and enjoy her infancy without a diagnosis being made:

Dad: We just felt ... there was nothing to be gained by that test, we would just try and have her, a normal birth and, and see if we could ...

Mum: Try and enjoy her.

Dad: See if we could live with it until we seen something for ourselves, if we were gonna ... so we lived with it for about, what?

Mum: Until she was about six months and we just ...

Dad: It was too much, too much ... watching her constantly to see.

Mum: Every, everybody just kept saying, 'Oh, she's fine, look at her, she's dead alert, she's ...'

Dad: 'She's doing this, she's doing that ...'

Mum: You know, and you're like that, 'M-mmm,' but it doesn't mean anything, you know, and then it just got the better of us. (Family 2)

They go on to explain that the process of being given the diagnosis for their youngest child had been very difficult for them, but that the doctor himself had also been very upset, having been involved in the diagnosis and care of their elder son who was diagnosed with Batten disease at the age of four:

Mum: When we went in you could just see him filling up.

Dad: As I say because he had been through it with us. (Family 2)

Testing other children was, therefore, part of the ongoing challenges that parents experienced, at times alongside practitioners who they had got to know through the course of their child's illness.

5.5.4 Summary

Talk about heritability had a number of dimensions from a family perspective, including mediating feelings of blame and featuring in decision-making about family planning and genetic screening. Heritability links clearly with family members' burgeoning expertise in these rare life-limiting conditions, as the parents and other relatives learn through generations what the meaning of the diagnosis will mean. Such experiential knowledge was not heralded by all families, though, and at times learning was not passed between family members despite the clear genetic linkages.

5.6 External constructions of parenting

Families caring for a child with a rare life-limiting condition spoke about how reactions and judgements from other people (such as the general public, friends and extended family) impacted on their sense of being a good or bad parent. This was frequently linked with the rarity of the condition, in that relatives, friends and the public had such limited exposure and understanding of children with this condition that challenging behaviour was often interpreted as 'bad behaviour'. Consequently, parents often felt that they were being blamed for their child's behaviour and struggled to articulate to others why their child was different. Parents described in very powerful ways how they had been made to feel worthless, with other people giving them and their child 'filthy looks.' In short, many parents had felt as though people viewed them as bad parents.

5.6.1 Before diagnosis

The sense of being constructed as bad parents was particularly troublesome before diagnosis, and consequently, before the parent had a clear label with which to describe their child's behaviour. For a number of families the child with MPS disease or Batten disease was their first or only child, thus they had no previous experience of parenting and began to internalise negative messages. Many parents spoke of feeling judged by others:

I remember [husband's parents] thinking that we were rubbish parents ... and, and we were, sort of, because it was our first time as parents, we were thinking, well, maybe we are, and we felt belittled and all the rest of it. It was awful, it was really awful, and I think, being far away from my folks and, yes it was a really difficult time. (Mum, Family 13)

This family gave a further example of how their child's behaviour pre-diagnosis was perceived by a family member. The mother describes how she and her partner felt judged; this still felt upsetting when she reflected back on the experience:

I remember my dad being really exasperated with, with [child] once we were down on holiday and my dad was speaking to him, wanted him to do something and he, he ignored him and [laughs] dad said, 'Are you deaf? Or just ... twisted?' was the word he used, you know, meaning cantankerous, you can turn, kinda sort of that kind of thing, and there was us all upset and I think ... oh no, and we're rubbish parents and all this sort of thing and then, you know it all ... [sounds emotional]. (Mum, Family 13)

The time before diagnosis seemed critical in how other people responded to the family, and how the parents consequently felt about themselves. The family quoted below talk about their experiences of feeling judged as being bad parents prior to a diagnosis being given to their child. This mother draws on socially sanctioned ideas of good parenting (that is, breast feeding) as a way of warding off a suggestion that she is a bad parent. The accusation of poor parenting was gleaned from others' comments about her child continually having colds:

[Child] was a breast-fed baby, fully breast-fed and I just remember lots of people would say to me, 'Has [child] got the cold again?' Because she would always have a runny nose and the cold so she always looked heavy and I was like ...'Aha, yes, she's got the cold again,' I was like, you know she shouldn't, she's a breast-fed baby, breast-fed babies aren't meant to have the cold all the time and that really bugged me and I just got to the stage where I couldn't be bothered listening to people anymore I was just like ... 'Just leave me alone!' (Mum, Family 11)

Thus, despite the mother positioning herself as a good parent, the child continues to experience recurring ear, nose and throat symptoms (common to these conditions) which lead to her feeling harassed and undermined by other parents. The following parent also articulates this sense of being negatively appraised by friends:

We have fallen out with a few friends [laughs] before the diagnosis ... judging our management of him and you know, comments from friends, one friend said, 'I would understand if you put him into a Home,' so ... she's no longer a friend. (Mum, Family 15)

The above extracts clearly illustrate the impact of people's reactions to a child who exhibits a number of unusual symptoms, but has not yet been diagnosed with a rare life-limiting condition. Parents have reported being concerned about their child's unusual behaviour or inability to meet certain milestones, this sense of worry is further impacted when someone makes a negative or critical comment about the child. Parents subsequently internalise negative comments, registering a sense of blame and feeling as if they have done something wrong.

5.6.2 Practitioners

Many families experience worry about their child prior to receiving the diagnosis of MPS disease or Batten disease, due to a number of features which signify that their child may not be healthy, such as not meeting developmental milestones, ear, nose and throat infections and behaviour problems. As a result of these early symptoms, parents would often be in frequent touch with practitioners from health and social care, seeking help and advice about their child. Many parents reported, though, that far from receiving support they felt judged and isolated.

In some instances, families perceived healthcare practitioner opinions negatively. The following extract illustrates how one parent felt unheard and positioned as overly worried, when sharing her concerns with the health visitor:

We could see that he was delayed in his learning and, you know, the way that, you know, he wasn't necessarily meeting all the milestones that, you know, you could, that they should be meeting, but also health professionals were saying, 'Oh, don't worry, he'll catch up, he's just a bit behind,' which sometimes kids are, you know, these, you know, levels that they have, they do drive me insane because all kids are different. So that, that used to annoy the hell out of me. (Mum, Family 17)

Another parent described how she had been diagnosed as having post-natal depression, leading to attachment difficulties with her child, and a feeling of being unable to parent effectively. She describes how her child was taken into the care of social services, rather than being offered more

constructive support to look after her child herself. She also identifies that the child's incessant crying was interpreted as the signifier of post-natal depression rather than indicating a need for concern around the child's own wellbeing:

They were putting it down to me having post-natal depression and not managing [child]. [...] I said to them, do you know I, I had ...I did have problems 'gelling' with [child] but I had also to prove my, I felt as though I had to prove myself to everybody that I could look after this baby ... [a social services assessment showed] that I had post-natal depression and that was causing [child] to cry and I explained but I haven't had her for two weeks, so and she's still behaving the same way with my mum. (Mum, Family 23)

The sense of being judged by practitioners is articulated clearly in the excerpt above as wishing to 'prove' to people that she is a worthy parent. In the following extract, a mother talks about being negatively judged by practitioners and goes on to illustrate the frustration that went along with this sense that practitioners did not understand her daughter:

I just really couldn't work out why I couldn't get [child] to understand what you do on the potty [laugh] you know? And ... her health visitor suggested that I was being too clingy and that maybe [child] needed to be taken away from me a bit so to put her into a nursery, and so [child] was going to a nursery but they insisted she wear nappy ... not nappies, knickers, so [child] was going to school and wear knickers, and, and then they asked me did she have a an- a pet because she kept on weeing in the sandpit [...] I just think she played in the sandpit, it wasn't a connection at all, it was just ... it was just where she was obviously just relaxed playing in the sand. (Mum, Family 22)

Thus, the above parent was implicitly blamed for being overprotective, and too clingy, resulting in her child's difficulty in potty-training. The inability of a child to meet expected milestones, such as being continent in the daytime, was subsequently directed towards the parent rather than exploring the reason behind such delays.

Other parents were given reams of advice from practitioners that did not have the expected effect because the practitioner had not understood the child's condition (as discussed in section 5.2). Treating the symptoms rather than the condition itself led to much tension and upset for parents, particularly when they felt under attack for their parenting skills:

Mum: People were telling us to do and I was saying, 'But I'm doing it,' and they were like, 'But you have to be really persistent with it and you have to do it every time,' and I was like, 'But I am,' and it was, it was almost like, 'Well, you're doing it wrong,' [laughs] you know, and that, that was a real, real, hard time.

Interviewer: So that was you being told that you were doing something wrong as a mother as well?

Mum: Yeah, contradicting me as if well, you know, the way they said it was, 'Well, you have to make sure that you do it every time and you have to make sure that you do it the same and you always follow it through,' and I said, 'But I do.' I am really, really, strict with my rules [...] I know now that it was nothing I was doing wrong because these two [children] abide by it 100 percent. [Child] hasn't been on the naughty step ... for two or three weeks.

Interviewer: How do you cope with actually being told that though, that you are doing something wrong?

Mum: It's horrible, it's really horrible, especially since I am so proud with my views as to how children should behave. (Mum, Family 15)

Consequently, families reported that many practitioners reflected often negative understanding of how the child was being parented. This was particularly problematic prior to the child's diagnosis, when the parent still looked to practitioners for expert advice and guidance, and critically, before they became experts in the condition themselves.

5.6.3 After diagnosis

Following the diagnosis families often shifted in their descriptions of parenthood and their expectations of themselves and of their child. Some reported shifting feelings of initial frustrations and re-thinking their actual role as parents:

We are both really firm and set with principles on how children should be and your 'please' and your 'thank you' and all this stuff, we are quite old fashioned with it and it was always like well, 'Why doesn't [child] do it? What are we doing?' you know, and it did, that made us realise that we weren't actually doing anything wrong, it was just it was different. So in a way, that was a relief and it was a relief to have an explanation as to why he wasn't doing things that he should be doing at that age, and talking, because obviously by then he should have been talking. (Mum, Family 15)

Internalising other people's ideas of parenting continued into the post-diagnosis phase. The following extract illustrates one family's experience of being isolated by extended family members after diagnosis. The dad indicates that having a child with a disability was understood by the extended family as a consequence of being a 'bad person', creating a clearly negative construction of himself and the child's mother:

*I kind of understood the genetics part of things and how everything worked, but obviously they [extended family] don't like to really accept that kind of information if you give it to them, you know. No, unless they, they probably think unless you're a bad person you're given a ... I don't know ... [my mum] never said that to me you know, but I was just, you know, unless you're a bad person you must have a child with a disability or something like ... you must have done something **wrong** or ... you know ... there's something else, there's something else but it's not the information part, you know, that they'd like to grasp hold of. (Dad, Family 16)*

The interviews contained numerous examples of members of the public reacting to a child in a negative way and the direct emotional impact that this has on the family. In the extracts below, a negative reaction results in both anger and tears, each adding to the sense of parents themselves being vulnerable:

Dad: Because the behaviour's so bad, I think ... and because they look quite normal people think they've just got a very bad child, a naughty child, and, and it, it's ... You can't control that in the ... We, we had an instance when we, we used to live in [city], we had [child] in, still in a normal pushchair [...] minding our business, and these two, and [child] had a [dummy] in her mouth, and she looked a normal girl, nothing wrong with her by that stage, and we were pushing her along, and she said 'Get out.' This lady made a comment and I, I thought ...
Mum: No she said, 'Get out and walk!'
Dad: 'Get out and walk!' And I didn't hear, like I said, 'Sorry?' I, I went to, and I thought, 'I'm not having this,' so I said, 'What d'you say? What ...' And then she sort of back down again and I thought ... And I was so angry, I was there to sort of basically kill them which was the normal thing because they're, you know [attacking]. For about something that we hadn't done, so ...
Mum: I've, I've had it in the supermarket before 'cause [child] used to shout a lot, she doesn't do it anymore, but they make this really high pitched noise all the time, and I used to take her round the supermarket, I was in the supermarket and this woman came up, she said, 'Will you tell your child to shut up, I'm trying to do my lottery ticket.' And I was so upset, I couldn't actually speak, and I just got back into the car and just burst into tears. (Family 22)

The family above goes on to describe other members of the public as being 'nasty,' and in one instance a shop manager was called to ask the parents to quieten their daughter. Another family spoke about feeling angry and distressed when strangers react in a negative way towards them as parents:

Mum: [Partner] used to get really, really, upset when people used to give us filthy looks as if to say can't you control because ... Or why is that boy sitting in a pushchair and things like that.

Dad: Yeah, I mean you can understand it from children thinking, 'Oh, he's a big boy to be sat in a pushchair,' but you see some adults and ... they stare, if they look, it's fine because they are bound to look, but if they stare, sometimes they stare don't they and I'm like, 'For goodness' sake.'

Mum: I mean, you used to get really, really, upset and I used to say to her, 'Look, they are ignorant and we don't know these people and we are unlikely to see them again, don't let them upset you.' (Family 6)

This family go on to talk about having avoided public places in the past, but through sharing experiences with another family they have managed to reject the sense of being put into an enforced isolation:

Mum: We are a lot more calmer now.

Dad: We are a lot better now. Before we wouldn't take the boys into say, a restaurant or anything like that even just say something as rubbish as McDonalds, we wouldn't have done that before because we were being really conscious of people staring at us [...]

Mum: We don't take them out to like, public places because we used to get stared at but since we've met the [another family] who actually go to loads of places with [child] because they've got [siblings] so they've got to and we are like well, stuff other people, you know, we've got to go out as well, it's our holiday or whatever and we tend to do it.

Dad: We do so, we will sit in the restaurants now and order meals and things and ... It's not enjoyable because ... [laughs]

Mum: But, we make a point of doing it now, whereas before we wouldn't have done. So we are much more confident in public now ... I wouldn't say we are more tolerant of some people but we are more ... inclined to ignore them now. (Family 6)

The data indicate that constructions of parenting negatively impact on families caring for a child with MPS disease or Batten disease.

5.6.4 Difficulties in controlling symptoms

Families express feelings of distress and conflict as a result of not being able to fulfil their role as parent and carer of their child. This was often related to the difficulties parents experienced in managing symptoms. For example, the following mother talks about the frustration she feels when she cannot eliminate or manage a symptom, and feels that her role as a mother is tied in with being able to prevent her child from being ill:

I can't stop her crying, so I eventually went over to [city] hospital and I was like, I'm going to end up going absolutely mental here, somebody somewhere do something, am I missing something? ... I suppose as a parent I was scared, are we missing something? (Mum, Family 11)

A further family talk about not being able to stop their child from crying. The difficulty in managing this was part of the rationale social services provided for taking the child into care:

Mum: [The first symptoms were apparent] just after being born, to be honest. That didn't settle down for ... about the first four months of [child's] life was constant crying, non-stop, morning afternoon and night.

Interviewer: How were people making sense of that then, both yourselves and also the medical team?

Mum: They, they didn't, to be ... well, actually, to be ... that became quite complicated because, they were putting it down to, me having post-natal depression and not managing [child].

Interviewer: So it's not that you've got an upset baby, it's that ...

Mum: I had post-natal depression. And so much so they took [child] into care ... until I went to the doctor to seek medical attention for myself. (Mum, Family 23)

It is clear that a number of families have experienced extreme reactions to their child's unfamiliar symptoms and in many cases judgements have been made around parenting. A child's symptoms and behaviour prior to diagnosis were often associated with culturally-defined notions of good and bad parenting. The extract below illustrates a link between a child's presentation of symptoms and a cultural bias towards labelling a child as having special needs:

I know plenty of special needs children that are completely different to [child], every child is different and back then I didn't have the strength to stand up and say 'But this isn't just a special needs child, this my child, this is [child] and he is nothing like the Down Syndrome child, he is nothing like the Autistic child, he is [child] and he is like [child]' you know. (Mum, Family 15)

Thus, the mother indicates that the child's uniqueness seemed to have been lost in labelling her son as a child with special needs. This mother continues to explain how her child's behaviour was explicitly associated with what was labelled as 'bad behaviour,' and her feelings around the punishments that were trialled for his behaviour. As a growing expert in her child's condition, she indicates an awareness that being put on the 'naughty chair' for time-out would not result in changing his behaviour, but lead him to feel confused.

We've had a lot of issues with school when he started because they said with him hitting out there had to be consequence with him hitting, and the consequence was time on the chair, is effectively time out, the naughty step. You know, it's that effect, which broke my heart because I thought, I know [child] and I know that he is just going to be thinking 'Why can't I go and play?' and he won't link the fact, he'll know that someone is angry with him or upset with him and he'll sense that and he'll get sad but he won't know why. (Mum, Family 15)

The school's management strategy and labelling of the child resulted in the mother feeling frustrated and conflicted. The data illustrate how what is culturally understood about 'naughty behaviour' has a direct impact on families caring for a child with a rare life-limiting condition such as MPS disease or Batten disease.

Other parents reported instances where they internalised feelings of not being in control, leading to an overwhelming sense of guilt. In one example a father spoke of beating himself up emotionally for not being in control of his own expectations regarding his role as a parent:

I think I beat myself up more because of how they are, you know, I respond like that and then I think you idiot, you know, why did you do that, why didn't you just take those extra few seconds and be normal and calm? (Dad, Family 6)

5.6.5 Reacting to other people

Many parents spoke of how they responded to the difficulties they face in how people viewed them and their child. The following excerpt comes as the interviewer directly asks them to reflect on this:

Interviewer: So how did you guys cope with that? You know, when people were making comments out, out in public?

Mum: Very bad at first, yeah. I sometimes can't eye contact people, if I see them, sometimes they look in disgust at [child], especially when we go to London, don't they? They're really like, 'What is that?'

Dad: 'Specially children, do you know, 'specially like, you know, like younger adults or children and they look, look at them and they make nasty, sly comments. (Family 22)

Other families spoke of how they had often just accepted other people's reactions, and only rarely challenged people in their response to their child's condition.

Dad: There was one instance at the hospital toilet and I said something to this guy, and I shouldn't have done because he gave me this filthy look because the boys weren't being naughty. They were walking through holding our hands but their way of communicating as [Mum] says is noise and they were just chatting away in their own way making noise and this guy just gave this really disgusting, filthy look and we made a point of saying 'Look, that is their way of communicating, they are special needs,' and he did it again as if to say 'Yeah, right,' and I just, normally I bite my tongue, but that time I didn't and it's very rarely I lose it but, you know, when you've said something, you know, you don't want to turn round and try to think that you have to make excuses for the boys.

Mum: You shouldn't have to justify your children because they're them aren't they, it's tough, people have to deal with it [laughs]. (Family 6)

Overall, parents reported a process of coming to terms with how other people reacted to them as a family, and to their child's symptoms. The following excerpt illustrates some of this process, and the responses of other people:

Dad: If there's a leaflet she'll rip it into 15 bits and start eating and ... which is funny but, it's not when you're sat there and everybody's looking at you [laughing] ... 'Like, what's, what's up with your child?!' Control that child!' you know, and it's ...

Mum: Especially when it's one of the books to share and the doctor's waiting. [laughs]

Dad: Aye and, and we ... you're going to stop her from jumping and stuff and falling over and hurting herself or hitting somebody else or something or, you only kind of contain it because you know it's her condition and it's not something she can control. But somebody sitting on the other side of the waiting room has no idea what's going on and you're sitting there and you, you kind of get to the point where you've no shame about anything ... you don't care what people think about anything [laughs] ... you just get on with it. And she is less like that now because she is less physical. (Family 21)

The above extract illustrates how one family reached a point where they were no longer affected by public reactions to their child because they accepted that there was nothing that could be done to change her behaviour so they adjusted to their own situation.

5.6.6 Positive constructions of parenting

Throughout the interviews there were also a number of examples of families' sense of parenthood being impacted in a positive way. For example, external factors, such as sources of support and managing the realities of the condition, contributed to making families feel upbeat about their parenting. One family spoke about how relatives helped them care for their children, and provided more support over time when the care became more physically demanding. The family spoke of the way in which younger members of the family (cousins) responded positively towards the children, despite not fully understanding the illness:

Dad: [My partner's] mum is really good, she will come over and she will help [partner] as much as she can.

Mum: Yeah, my sister comes over and she's got children of her own but they are really good and I think that all the children in the family understand that [boys] are just [boys], they don't understand their illness, they just think ... [Dad interrupts]

Dad: They just accept the boys for who they are.

Mum: [Our children] are who they are really. It's great because they don't ask questions and things and like other children might do, they just accept them. (Family 6)

The family mentioned above talk about the positive impact of the support and acceptance that they received from family members. Therefore, the parents developed positive constructions of family and this seeped through to their own perceptions of parenting and coping.

Another example of positive constructs of parenthood is given below. The extract focuses on the family's response to their child being awake all night making loud noises including laughing:

Dad: What he was laughing at I don't know, but it carried on all through the night and all morning up to about lunchtime. And he got to that point again where he was so exhausted because he had not slept and he was just laughing all the time and it was really belly laughing. He was just losing it and he had worn himself out that much from lunchtime through to when he then went to bed and we just didn't know what to do that day did we?

Mum: No.

Dad: We were shouting. So from about midday through to about six thirty was just white noise and when [child] shouts, he shouts.

Mum: Poor neighbours [laughs].

Interviewer: Poor you guys, what was that like for you?

Mum: Awful.

Dad: Don't ask [Dad laughs] I couldn't even describe it, it was that bad.

Mum: We are pretty good. We do shout in the night sometimes and swear a bit but ... we are alright by the morning aren't we?

Dad: We are generally pretty good considering, you know, for the best part of eight years now we've had disturbed sleep more or less every night.

Mum: Every night, yeah.

Dad: I mean, I used to be terrible before we had the boys, if I didn't get eight hours I was a grump anyway but ... [laughs]

Mum: No, he's quite good, he's learnt.

Dad: I suppose I had to.

Mum: You have to, don't you? (Family 6)

Throughout the interviews it was recognised that a number of couples demonstrated a closeness that impacted on their ability to cope with certain situations. The couple above spoke about an extremely difficult situation where their child remained awake, making 'white noise' throughout the whole night and into the morning. However, the couple responding to each other enabled them to react positively by acknowledging their ways of coping, and reinforced their identities as being good parents. Positively connoting their own adaptation as parents was therefore part of the overall construction of parenthood.

5.6.7 Summary

Parents often struggled with how friends, family and members of the public responded to them and, in particular, how their identity as a parent was challenged. Avoiding internalising these negative messages was often most difficult before a diagnosis was made. The rarity of the condition often means that people are unable to offer explanations for why a child might have ongoing physical illnesses, behavioural quirks or use props (such as dummies or prams) which appear age-inappropriate.

Parents routinely spoke of how being viewed negatively by others was upsetting, and often resulted in them being tearful. However, following diagnosis the narrative arc changed. Many parents began to feel more reassured that their parenting was not to blame for any ongoing physical or behavioural difficulties. Moreover, as the child's condition progressed, and as the child became less active and less vocal, these challenging times were mourned, despite the less blaming reactions of others to them and their parenting style.

6 Findings: Sibling experiences

Siblings were interviewed to gain an understanding of their experiences in having a brother or sister with MPS disease or Batten disease. Of the 23 participating families, ten had siblings who met the project inclusion criteria and were therefore invited to take part in the interview. Of the ten eligible families, six agreed to participate, one declined and three were not available due to school commitments. Two families had more than one sibling wishing to participate. Siblings were invited to choose if they were interviewed separately or jointly; one dyad chose individual interviews and one sibling dyad chose a joint interview.

Consequently, eight siblings participated in an interview. Demographic characteristics of the participating siblings are summarised in Table 18. As noted in section 2.7, the data is presented in anonymised format. To prevent matching of symptom descriptions (reported in section 3) with the data in this chapter, numerical identifiers have been assigned to siblings from each family which tally with the identifiers used in section 5.

Table 18: Demographic characteristics of participating siblings (n=8)

Characteristic		n
Gender	Male	5
	Female	3
Age (years)	Median	10.5
	Range	7-12
Diagnosis of ill sibling	Sanfilippo disease	4
	Morquio disease	1
	Batten disease	1

Siblings were asked a series of questions to explore what it is like for them having a brother or sister with MPS disease or Batten disease. Siblings were asked to describe symptoms which were most difficult for their brother or sister, and to provide rationale as to why they thought these symptoms were the most difficult. Finally, siblings were asked to share the feelings and emotions they experience when their brother or sister suffer such symptoms.

The data indicated a number of categories, which are presented in Figure 16 below. To help the reader navigate through the complex and rich accounts, each part of the Figure has a reference within it indicating which section of the report describes the data.

A number of areas were highlighted that demonstrate the positive and negative impacts on siblings. These impacts include a detailed awareness of the condition and its associated symptoms, the caregiving tasks which siblings undertake, and how the condition impacts on the ability to take part in social activities. Siblings also spoke of the emotional consequences, both in terms of their own processes and in response to how the public/peers view their brother or sister, and their coping mechanisms.

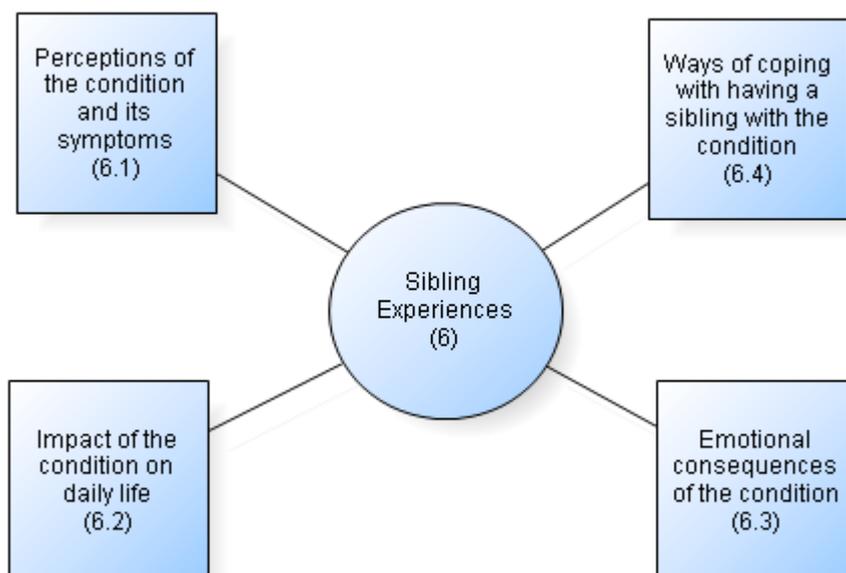


Figure 16: Sibling experiences

6.1 Perceptions of the condition and its symptoms

This section reports data on siblings’ knowledge and understanding about their brothers’ or sisters’ complex and life-limiting condition and the range of symptoms that they observe on a regular basis. This sets the scene for the subsequent section which presents evidence on how the experience of living with a sibling with a life-limiting condition impacts on their lives.

Siblings demonstrated a keen awareness of the symptoms commonly experienced by their brother or sister. Their knowledge and understanding about their ill sibling’s condition was linked with their age, with older siblings demonstrating a more lucid understanding, and articulating their feelings more clearly. Overall, siblings were able to identify relevant symptoms from the assortment of cards provided. Children explained how each symptom affects their brother or sister, as well as descriptions of some of the interventions or actions normally taken to manage the symptom.

Information regarding the life-limiting condition of a brother or sister appeared to be often acquired through parents, or the child’s primary paid carer. Practitioners supporting the child and family serve as a valuable source of knowledge for siblings, though this was not always through direct communication. As the following quotation illustrates, siblings sometimes learnt through overhearing conversations and soaking up information as adults spoke around them:

I kind of take in a lot of what people say, around, so like when there’s specialist people coming [to the home], I just take in what they say, really. (Brother, Family 8)

Other siblings sought out information proactively, particularly when a significant event had occurred, such as an acute deterioration. The following child reflects on who she speaks to when she has a question:

Sibling: Sometimes my dad or sometimes my mum, say if he's, like when he was having his operation and [Mum] went to the hospital with him and like, she went to the hospital with him for like a few hours and then she came back, like I asked her, so many, too much questions.

Interviewer: You did?

Sibling: Like 'What was he like?', 'What did they do to him?', 'How did they, how did they put the bandages round him?' and she just would answer them with me. (Sister, Family 16)

Siblings' understanding of the condition developed over time and was constantly being updated and reinforced as the condition progressed. Children spoke about their views on witnessing such changes:

Interviewer: How do you cope with all the changes that [child] goes through?

Sibling: Fine, kind of, you get used to it, and get used to change, and you recognise it, identify it, that sort of thing. You just get used to it.

Interviewer: You just get used to it. And it, is that, is that okay for you? To, you sound, it sounds like you're used to changing and things, you know, you're used to cope, coping with that?

Sibling: Yeah. Well, it kind of happens, kind, it sometimes gets better, worse, generally, more worse, but I'm kind of, that's happened for a while, so I've got used to it. (Brother, Family 8)

In one case, a sibling demonstrated with clarity an awareness of a range of symptoms that his brother experienced, identifying a range of cards depicting relevant symptoms and the care which was given in response. However, when asked about his parents' view of his knowledge, he reported that they would probably underestimate how much he knew and where he had gained his knowledge from:

Sibling: They would probably say ... [that] I don't really understand a lot about it ... that's what they would probably say.

Interviewer: And do you agree with that or do you think ...

Sibling: I kind ... I know quite a bit, but I don't know a lot.

Interviewer: You know quite a bit though ... and how, how do you know quite a bit?

Sibling: [There is a] website and it, it had a wee page on it that said, saying about the disease so I read that 'cause I ... at that point I didn't know anything about it. (Brother, Family 10)

The sibling could not then recall the detail of what he had learnt from the website, but had nevertheless signposted an important difference between what he knew about the condition and what his parents thought he knew and understood.

The data demonstrated a range of levels of knowledge among siblings, but all interviewees were able to name and talk about relevant symptoms. Several siblings stated that they felt they may know more about their sibling's condition than their parents realise. This may be a consequence of both siblings and parents adopting a protective stance toward each other, acted out through each party limiting the amount of knowledge they share with each other about the ill child's condition. Arguably then, these children demonstrated caregiving toward both their sibling and their parents.

One of the core features of siblings' talk was descriptions of the challenges they experienced in assessing their brother or sister's symptoms as a result of communication and cognitive impairments. Pain was specifically mentioned as being a troublesome symptom to recognise:

That card there says 'feeling sore' but you can't exactly tell if he's feeling sore ... 'cause he can't, he can't say anything. (Brother B, Family 13)

Some siblings described intuitively assessing whether their brother or sister is experiencing pain, which results from the close and intimate relationship family members have with the child. It is this experiential knowledge or 'knowing the child', also reflected in the parent interviews that is invoked to assess symptoms:

Well I suppose that, I think that you can just see it in her. You can just kind of tell that she is in pain. (Brother, Family 8)

Similar to the question posed to parents in the family interview, siblings were asked to describe which of the symptoms experienced is most difficult for their brother or sister to cope with. Three siblings described the symptom of seizures or 'fits'. When asked to explain why they believe seizures to be the most difficult symptom for their brother or sister to cope with, the siblings expressed factors such as their sudden onset, and alarming and scary nature. Moreover, the uncertainty of when a seizure will end, and whether or not their brother or sister is going to be okay after the event, was particularly distressing to the siblings:

It looks so scary. It's ... 'cause when he does it [has a seizure] ... he just suddenly does it. (Sister, Family 4)

Three siblings also described the symptom of coughing and difficulties in clearing their airway as the most difficult symptom for their brother or sister to cope with. The main explanation provided to support this symptom choice was the effort and struggle the siblings observed as the child attempted to clear the secretions in their throat. Thus, unlike the parent interviews, the siblings did not give voice to an underlying concern that such breathing difficulties may be a sign of the last few moments of life.

6.2 Impact of the condition on daily life

Siblings provided a range of physical, personal, social and emotional care. They spoke proudly of the caring roles they take on within the family and there was little indication of any frustration or discontent associated with such responsibilities. Siblings described the enjoyment they receive from playing with their brother or sister referring to activities such as reading a story, singing songs or being affectionate. Typical sibling rivalry behaviours and interactions where the ill-child may serve as a source of annoyance or frustration were also apparent.

The following extract illustrates a range of activities that one sibling engages in:

*Sibling: I do his milk machine, I ... do the asthma, I help Mum change his nappies, I help him, I help Mum [...] lift, lift him and I change his wheelchairs, I give him toys I ... sit on the floor with him sometimes, I hold his hand and ...
Mum: You massage him. (Sister, Family 4)*

Siblings happily talked about their experiences of caring and helping out. Caring behaviours were always described as an adjunct to the parental care the ill child was receiving. Thus, there was a clear sense that the sibling was supporting others to care rather than taking full responsibility:

Normally if [...] like Mum's out doing gardening she's 'Oh, go and check on [child] inside,' and I just run in see how his hands are, that kind of thing, if he's cold, put on a blanket. (Brother B, Family 13)

Nevertheless, many of the siblings presented as having an increased sense of responsibility for looking after their brother or sister, and as being mature for their ages:

*Sibling: 'Cause normally when Mum's reading a paper in the kitchen or Dad's watching TV and they're engrossed in something, I can just hear [child] start crying and you say to Mum...
Interviewer: Is it letting them know and things?
Sibling: Yeah, keeping an ear out. (Brother B, Family 13)*

The experience and expertise acquired through caring for her brother was described by one sibling as contributing to her career choice for the future:

Because when I'm older I, I want to like care for special needs kids or be a teacher to ... I want to do something that involves them. (Sister, Family 4)

A number of siblings reported a desire to help care for their siblings in small yet significant ways and indeed spoke about carving out a caring role in their future careers. One sibling with a keen interest in science spoke about becoming a doctor in the future. Furthermore, during the family interview the mother mentioned that her son would be a more understanding as a result of his experiences.

Siblings also spoke of the limitations in their social experiences which stemmed from the needs of the ill-child. The following extract illustrates such limitations and how it makes her feel:

*Interviewer: How does that make you feel when he's not well and he's got a cough or a cold and his mum and dad are having to stay close to him?
Sibling: Really like, upset ...
Interviewer: What's that like?
Sibling: It's like when, like, we can't like go out as a family if he's got a cough or a cold 'cause ... on Christmas we were supposed to go to my granddad but [sibling] wasn't feeling well so it was just Mum, me and my sister who went up. [...] They [parents] would probably say I was like, I felt down like, that day. It wasn't like a, a proper Christmas I would normally have. (Brother, Family 10)*

Thus, while many spoke affectionately about their caregiving roles, they also experienced limitations in their daily social activities as a result of their sibling's condition and this was described with acceptance and sadness.

6.3 Emotional consequences of the condition

Siblings expressed a range of emotions and responses during the interviews both related to their own feelings about the condition and as a consequence of other people's reaction to their brother or sister.

Siblings described feelings such as worry and sadness associated with concerns they have over their brother or sister's health. The progressive and degenerative nature of these conditions means siblings observe the changes and deterioration that happen over time. Siblings cope through both internalising and externalising behaviours. For example, during the interviews, siblings articulated a need to know that their brother or sister is okay and anxieties that arise when there is a chance that they are not okay. The emotional impact of having a brother or sister with these rare conditions is described by two of the participants below:

Well, not exactly exhausted but, once you come back from school you're exhausted and then it's just, you just feel worried and that kind of thing ... how he [ill sibling] is when you just get home ... 'cause you normally just come in the house, look around, say hello to the carers and then come and see [ill sibling]. (Brother B, Family 13)

Interviewer: Do any of these cards show how you feel when [brother] is having a bad day?

Sibling: Well if [brother]'s got like a cold or something, I'm worried and then I get a cold sore, 'cause I'm worried. (Sister, Family 4)

Sibling reports indicated powerfully the emotional repercussions, indeed one sibling's anxiety for her brother's wellbeing results in actual physical pain. During an acute episode, another sibling suggests that her emotions were stretched beyond her usual coping abilities. The extract below illustrates how this strain results in an extreme sense of worry until she is given reassurance:

We had to go like at one o'clock in the morning to the hospital 'cause he started rolling his eyes and it was either an asthma attack or a fit and, and then we needed to go to the hospital, that was quite scary [...] I just like, cried all the way there and then ... he was okay. (Sister, Family 4)

Worry about their brother or sister pervades siblings' lives. Siblings described difficulties in concentrating at school as their minds were often preoccupied with anxieties over their brother or sister:

I feel sad for [ill sibling] and I have tears going through class and stuff. (Brother A, Family 11)

*Interviewer: Is he [child] on your mind quite a bit at school?
Sibling: Yeah. Like 'What's [child] up to? What's he doing?'
Interviewer: Right, that's what you're thinking to yourself, yeah.
Sibling: But I like it when I'm at home with him, or when I'm with him 'cause I know what he's doing and I know if, if he's okay or not ...
Interviewer: Yeah, but you find when you're at school you're having a bit of a think about [child].
Sibling: Yeah.
Interviewer: Does that happen during lessons or just during breaks?
Sibling: All the time.
Interviewer: All of the time, yeah.
Sibling: But not like all **all** of the time, just as in it can happen at any time. (Sister, Family 4)*

Other siblings described how worry or concern for their sibling can impact on their social time with friends, as preoccupation and concern often dominates their thoughts as reflected in the following quote:

They [friends] would probably realise, 'cause normally I'm really cheery when I'm out playing and if I have a bad day they would know 'cause I wouldn't be as cheery ... 'cause we normally always play football and they would know if I was having a bad day 'cause I normally wouldn't play my best. (Brother, Family 10)

Such anxieties can escalate in situations where their brother or sister's health deteriorates necessitating an admission to hospital or acute care:

*I would have a little cry, 'cause I don't like it, and I don't, I don't cry at school but I just cry into myself, you know, you cry in **yourself** you know, that's why and then when I go to my bedroom I have a big cry cause [sister]'s gone away to hospital and I can't really see Mummy or [sister] cause Mummy's gone to hospital with her. (Sister B, Family 11)*

Physical separation of the family, such as when a parent has to stay in hospital with the ill sibling, was particularly distressing to the children. At times when a child's symptoms worsen or their condition deteriorates, the effects are felt throughout the family:

It's like when, like we can't go out as a family if he's got a cough or a cold cause ... on Christmas we were supposed to go to my granddad's but [brother] wasn't feeling well so it was just my little sister and I who went ... It wasn't like a, a proper Christmas I would normally have. (Brother, Family 10)

Some siblings described being observant to the concerns and worries of their parents and articulated this during the interviews. When asked to describe what it is like in their family when their brother

or sister is particularly unwell, siblings were aware of and able to articulate how the concern for their brother or sister is resonated throughout the entire family network:

Things are always a bit like 'down' ... like my Mum and Dad won't be the same and like maybe some, even sometimes my younger sister knows he's not feeling well ... she goes up to [brother], tries to get on his bed and give him a big kiss and cuddle and all that. (Brother, Family 10)

Similarly, one sibling indicated that they sometimes choose not to discuss their feelings as a way of protecting the parent. In the following excerpt, the mother interjects to ask her daughter a question about their communication. The daughter explains that her body gives her away by displaying worry through cold sores, a sentiment which was noted in a quotation presented earlier in this section:

*Mum: If it's something about [child] will you not tell me 'cause you're worried that it'll upset me?
Sibling: Yeah
Mum: But I usually get that out of you don't I? You usually tell me.
Int [to sibling]: She can winkle it out of you?
Sibling: Yeah, 'cause every time that I've got a cold sore. (Sister, Family 4)*

Thus siblings were often managing a range of complex emotions, at home and at school and were very aware of how worrying feelings could reverberate around the wider family system. The data also highlighted how siblings were affected by wider attitudes toward disabilities. Two interviewees spoke of being repeatedly teased by peers at school, because they have a 'disabled' brother or sister. In both interviews, the term 'disabled' was used by siblings to describe their brother or sister with a life-limiting condition. These children described how teachers or other authority figures had not intervened to manage the situation. This lack of timely intervention by adults represents a clear concern for children, and while it is possible that incidents were addressed later, the siblings were unaware of any such delayed action.

Siblings expressed being both confused as to why their peers behave in such a manner and feeling upset by the cruelty towards their brother or sister:

*Sibling: So, some of them just make fun of him ... he's insulting and well 'You've got a disabled brother, ha, ha, ha.'
Interviewer: And what do the teachers say about that?
Sibling: They don't do anything about it. (Brother B, Family 13)*

*Sibling B: [Name of peer]. He's in my class and he always laughs when [sister] comes into our class and he says 'I hate your sister'.
Interviewer: Why?
Sibling B: I don't know, and I ...
Sibling A: Well, first thing, how would he like it if he had a disabled sister? How would he like it if we said that to him?
Interviewer: Did you tell your teacher?
Sibling B: I did and [teacher] said 'Oh'.
Sibling A: If he had a disabled sister and someone came in and said ... 'I hear your sister's rubbish, I hate her.' How would he feel? (Family 11)*

Similar prejudice was experienced within the community. Children described experiencing or witnessing negative attitudes towards their ill sibling, and to others with a disability, and the emotions this raised. Interviewees expressed a wish for people to be more accepting and tolerant of such differences. The siblings did not describe feeling embarrassed in public when people stare or when peers tease them regarding their brother or sister with a disability. Instead they presented as being protective of their sibling and articulated a lack of understanding as to why people can be unkind:

Well, we watched this programme on the TV and it was about conjoined twins and Mum took an interest because of [sibling] and I just watched it cause there was nothing else to watch and then the mother said they were at the mall or shopping place and someone came up to them and said 'Why do you take these children here?' ... I was like 'How cruel can you be?' (Brother B, Family 13)

When [child] was wee and he used to run around he got deaf a lot, well, not completely deaf, but he lost his hearing a bit and you had to shout his name very, very loud and Mum was shouting and shouting in a cafe and this woman said 'Do you really have to shout this loud?' and Mum said 'Yes, I do,' and the woman said 'Why do you?' and Mum said 'Because he's getting deaf.' (Brother B, Family 13)

'Cause people, whenever you're walking down the street with [sibling], they look at him and then they just go like that [demonstrates turning away] ... not accepting him, it's just ... cruel. (Brother B, Family 13)

The depth of emotion expressed by this sibling is a reflection of the impact peers and the wider community have on siblings. A number of siblings demonstrated a considerable degree of empathy towards their brother or sister and indeed toward anyone with disabilities. Siblings reported a range of experiences around acceptance and the level of support received through peers, indicating an inconsistency and need for additional support.

6.4 Coping and support

Siblings reported a mixture of experiences regarding coping and support. Some siblings struggled to articulate where they gathered support from or how they coped with the long-term impact of having a sister or brother with a degenerative life-limiting condition. Siblings all spoke positively about peer relationships, but indicated that these relationships were separate from their home life. In one interview a sibling indicated that although he had a solid group of friends, he did not necessarily rely on them for emotional support:

Interviewer: What's it like for you when it sort of caring for [sibling], how it's changed your activities and things?

Sibling: Yeah, well, it limited, it limits some things. Yeah. Just limits and stuff.

Interviewer: And do you feel that you've just got used to things and you just adapt?

Sibling: Yeah.

Interviewer: Yeah, do you sort of have any sort of special, best mates? Have you got a group of best mates at school, or have you got, a particular one friend?

Sibling: I've got a best mate, and I have a group of good mates.

Interviewer: Yeah, good. Good, and do you feel that ... do you talk them about it, or do you just get on ...?

Sibling: Nah.

Interviewer: [You] do your own thing?

Sibling: Yeah. (Brother, Family 8)

In another interview a sibling indicated a strong sense of isolation. Though his communication drew heavily on non-verbal cues, the meaning was nevertheless powerful in establishing the limited time he has with friends or having a break:

Interviewer: So you don't just go away and go outside but you come in and [help]?

Sibling: M-mmm.

Interviewer: See what's happening.

Sibling: Well sort, we just go outside, come in every 10 minutes and check.

Interviewer: Check, m-mmm. Do you ever just, just go away and go to a friend's house and stuff just to get, get a break or?

Sibling: [Shakes head]. (Brother B, Family 13)

Later in the interview, this sibling goes on to say that he only has two opportunities a year to talk to somebody that really understands his situation:

Sibling: The thing ... the siblings' weekend ... they're the only ones that can understand it.

Interviewer: Yeah. That must be difficult not have, being able to talk about it whenever you want.

Sibling: Mmm ... it's normally just like two days a year. (Brother B, Family 13)

In contrast to the above data a number of siblings described their main sources of support as being family members and friends or peers. Interviewers often began the interview by asking about the siblings' friends. Responses to this opening question often led to interesting disclosures, whereby children described their best friends as being those peers who were most supportive and understanding of their ill sibling and wider family situation. It was evident that this understanding was a very important quality to them:

My best friend [name], he says that's a pity that [sister] has [MPS disease], he said that's a pity and stuff which is very, very kind. (Sister B, Family 11)

Siblings also provided descriptions of occasions where such friends take on supportive and caring roles within the child's family:

Normally my friends and all just cheer me up and stuff. Like, as I said my friend [name] he really helps ... with Mum and my wee sister and that, the same as me. When we went to [city] a couple of weeks ago he was like looking after my wee sister while my mum was changing [ill sibling] or something ... and he's really supportive when [ill sibling] is not feeling well. (Brother, Family 10)

Organised sibling support groups, provided by children's hospices or organisation and/or charities were mentioned by some siblings as being both valued and enjoyable. Formal sibling groups and networks provide a vital and valued source of support, addressing the unique psychosocial needs of this group. Siblings provided accounts of participation in sibling clubs and organised outings where they are able to meet with children who share their experiences and are able to relate to what it is like having a sibling with a life-limiting condition. The disadvantage expressed by siblings is that these meetings are infrequent, held only a couple of times a year. Moreover, children often live great distances from each other and therefore have few other opportunities to get together. Some children spoke of keeping in touch with siblings they meet through email and text messaging.

6.5 Summary

Siblings spoke with clarity about their ill brother or sister's condition, and summarised complex symptomatology and their role as young carers within the family. Many spoke of emotions, combining love and compassion for their sibling with frustration at those who judge people with disabilities. Further, siblings' accounts point towards a delicate balance between protecting their unwell sibling, their parents and themselves. The emotional impact, of being in a family supporting a child with a rare life-limiting condition, seemed to involve a critical combination of negative disability discourses and support through peers and family members.

Children's experiences indicated in this section and in Figure 16 go some way to evidence key features of sibling's lives. Rather than being a theoretical model, the figure indicates areas of connection between experiences.

7 Findings: Practitioner perspectives

Nineteen practitioners were interviewed for this study. Three practitioners spoke about more than one child. Further, two practitioners were unavailable to participate, and the mother of two children was unable to name practitioners who could speak about her children. Thus the views and experiences reported below stem from 19 interviews which relate to 22 of the 26 children recruited to the study.

The range of practitioners, their professional role and the family's rationale for inviting them to participate are presented as a table in Appendix 4. As the table indicates, several interviewees had supported a number of other children with these conditions, but that overall, this was limited to those working in paediatric palliative care settings.

The views and experiences of practitioners are presented in this report as two separate models, representing the key ways in which interviewees spoke of their views and experiences of supporting children with rare life-limiting conditions. The two models are entitled: *Practitioner Experiences* and *Advance Care Planning*, each of which is described in turn.

As noted in section 2.7, the data are presented in anonymised format. To prevent matching of symptom descriptions (reported in section 4) with the data in this chapter, numerical identifiers have been randomly assigned to each practitioner from 24-42.

7.1 Practitioner experiences of supporting families

Families were asked to nominate a practitioner that they felt would be well placed to talk about their experiences caring for a child with MPS disease or Batten disease. Therefore, the practitioners selected for interview came from a variety of professional backgrounds including nursing, special needs education, palliative care and other allied health professions. This reflects the wide range of people involved in the care of children and support of families affected by these conditions.

At the heart of the data was a sense that the rarity of these life-limiting conditions formed an important part of how the practitioner supported the child and family, and how practitioners made sense of the array of symptoms experienced by the child. Although children with MPS disease and Batten disease have an identifiable pattern of symptoms, each child's unique presentation can preclude gaining a sense of a set pattern or predicted trajectory. Such uncertainty in predicting symptoms and deterioration contributes to the overall sense of uncertainty associated with managing children with these conditions.

The data indicate that the rare and complex nature of MPS disease and Batten disease limit opportunities for developing expertise. The struggle in gaining expertise in managing the conditions is reflected in the need for regular contact with a wider group of symptom-specific experts. Indeed, many practitioners have silos of expertise, rather than disease-specific expertise. As a result, care coordination is a key role that involves knowledge sharing, advocacy and coordinating access to specialist services. By spending more time with a family, practitioners get to know a family. In the context of this study, the relationship between families and practitioners seemed very important in the overall experience of caring for the child. Moreover, the relationship between families and practitioners lends itself to emotional support and going above and beyond routine professional responsibilities. Each of these features are depicted in Figure 17, which indicates the relationship between different parts of the model.

To help the reader navigate through the complex and rich accounts, each part of the model has a reference within it indicating which section of the report describes the data.

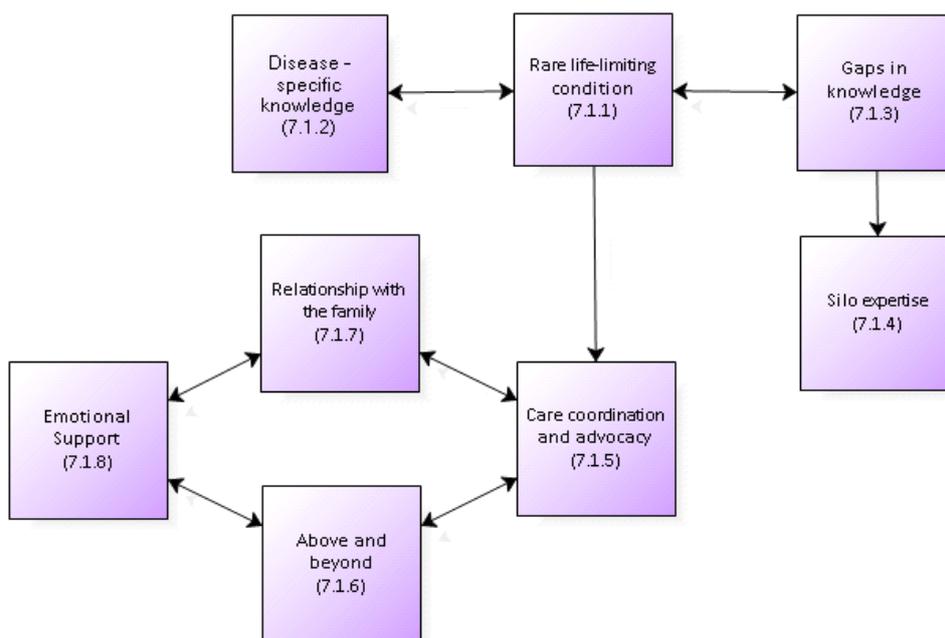


Figure 17: Practitioner experiences model

7.1.1 Rare life-limiting condition

MPS disease and Batten disease are extremely rare and complex conditions. Their rarity means that often practitioners do not have a great deal of exposure to children with the condition. In the following quotations, two practitioners from different backgrounds illustrate the rare nature of MPS disease and Batten disease:

I've had two other kids with Batten's ... for 36 years so ... That's not a lot when you think of the number of kids with cerebral palsy, autism ... and you know, Downs, you know I've worked with over the years, that's three kids with Batten's over the [years]. (Practitioner 29)

We've had quite a few over the years; well quite a few, I'd say a handful, but [child]'s our only child at the moment with Sanfilippo. (Practitioner 27)

It is not only the rarity of these conditions, but the complexity of symptoms that are key features for practitioners. The following speaker draws attention to the long-term needs of families caring for a child with MPS disease or Batten disease, extending far into the future:

It's not a terribly common disorder but these are disorders with huge implications for the child's wellbeing and constant problems and the family need to be supported for a very long time. I mean indefinitely. (Practitioner 34)

Other practitioners made the explicit link between the rarity of conditions and the challenges in symptom management:

Interviewer: What is your experience of supporting families who have a child with Sanfilippo?

Practitioner: Yeah, very limited. I have the one child with Sanfilippo that I've known for the last seven years. And [child's] disease hasn't actually followed the sort of typical progression that we'd expect. The other child that we've mentioned with MPS disease has been known to me for four years. And those are the only two children that I have ever come across in 29 years of paediatric nursing. And that's ... both been in the last seven years. My experience would be that Sanfilippo is one of the most difficult diseases to manage. (Practitioner 28)

I can think maybe one other child that I see as frequently [child] would be the one over the years I would have seen most because she's just been incredibly complex and you, you get one thing settled and it sets off something else. (Practitioner 31)

The above speaker signposts the difficulties in managing a rare life-limiting condition, where a treatment for one symptom may spark side-effects that also then require management. Rarity of the conditions means there are fewer documented management strategies to try, resulting in adding to the burden of complexity.

Thus, MPS disease and Batten disease are viewed as particularly complex due to the unique presentation of symptoms observed in children and the conditions' rarity. Practitioners frequently report that they have often seen only a handful of children with these conditions, and this impacts on their experiential and clinical knowledge of managing symptoms. However, the few practitioners who have supported children with these conditions over a number of years clearly have gained valuable experience and disease-specific knowledge.

7.1.2 Disease-specific knowledge

The data indicate that practitioners with knowledge and understanding of MPS disease and Batten disease predominantly work in specialist paediatric palliative care settings. The conditions' rarity clearly impacts on practitioners' knowledge and experience. Some practitioners had supported a number of children which had given them insight into the challenges of the condition from diagnosis through to death:

We've had obviously some with Hunter's, Hurler's and Sanfilippo ... through the hospice, and obviously I've, because they've come from respite ... come for respite for a number of years, from the hyperactive stage right to through ... to the palliative stage and end-of-life, I've got that experience of actually caring for them through the whole cycle of their conditions, really. (Practitioner 26)

Practitioners were asked to reflect on why they thought the family had chosen them to participate in the interview. This data is presented fully in Appendix 4, but the following speaker speculates that they were nominated because of their knowledge of their child's condition through a lengthy working history in a specialist paediatric palliative care setting:

Interviewer: What, in your mind, do you think that they [family] see particularly in your practice?

Practitioner: Probably caring, trusting, and a knowledge about MPS children but particularly Sanfilippo. I've got 18 years' of working at [hospice] itself. (Practitioner 42)

Other practitioners with extensive paediatric palliative care experience still felt that symptom management was an ongoing challenge:

I think it's a complex [view of the symptoms] you know. If, if you get a child in with a chest infection you could say, or a urinary tract infection, you'd say, 'Yeah, yeah we'll treat that, and that will go away,' or you know if they had a broken leg well, 'Yes,' you know, 'we can fix that for you,' and, and it's fine, there are certain things that you think, 'Yes, you can fix,' with these kids you're always waiting for them to throw the next thing at you ... You're never able to relax and say, 'That's them sussed ... we, we're okay there.' (Practitioner 31)

Other practitioners reported that while they did not have disease-specific knowledge, this was not a barrier to providing care, since their skills as clinicians in paediatric palliative care was adequate to enable them to support families. The following speaker illustrates this view:

The important thing being, I didn't make too much of Batten, but for me it was more, I have experience of supporting families who have children with the problem ... that would be life-limiting for whatever reason. Batten just was the name on the piece of paper ... in terms of, of the family's needs does not change ... so on one hand I have experience in dealing with families who have children with, with life-limiting and progressive illnesses ... but if you ask me specifically with Batten ... I didn't have experience until [this child] came along. (Practitioner 25)

Thus, not having disease-specific knowledge did not put the practitioner or family at a disadvantage. The above speaker reflects a psychosocial approach to supporting families rather than a purely medical model. That is, the approach is guided by the fact that the child has a progressive life-limiting condition, not related to the particular clinical features of the specific condition. Specialist disease-specific knowledge is therefore not privileged as important by this clinician.

Practitioners reported that even *with* direct experience of the conditions, each child is different and their own symptoms present in a unique way. A lack of continuity in the presentation of symptoms impacts further on the already uncertain nature of these rare conditions:

*Interviewer: You've worked quite closely with the other families then, as well?
Practitioner: Yes, oh yes, definitely. Yes ... symptoms that are similar, but different in each child, and I think you need to take that, you know, every child is so individual and how it affects them ... you can obviously see there is a definite pattern ... but obviously it is so different in each child. (Practitioner 38)*

Just because a child's diagnosed with a MPS condition that it's not going to run a course that you think ... I did ... every child is different and every family's different and you might expect that all the symptoms that you've described [child] has got but that doesn't mean to say another child with [MPS] condition will show or you know present as he's done ... and I think what you mustn't do is assume that he's going to go down this road, because he might not and for the families it's very important to listen I think, to listen to them and actually support them where they're at [...] I think the thing I have learned from [child] is that you take each family as it is and support them where they're at now. (Practitioner 26)

The above speakers therefore indicate that while they are familiar with these rare life-limiting conditions, such knowledge does not always provide adequate preparation for supporting all families with a similar diagnosis. The final quotation above cautions against such approaches, and serves as a reminder that a tailored approach is required for each family according to their needs. Disease-specific knowledge serves an important role in preparing clinicians to support children, and enables them to view the complexity and variability of presentation.

Connected integrally with the rarity of the condition and disease-specific knowledge is the articulation of practitioners' gaps in knowledge and experience in supporting children with MPS disease and Batten disease.

7.1.3 Gaps in knowledge about MPS disease and Batten disease

Talk about gaps in knowledge was prevalent across interviews, since many practitioners were not specialists in the specific conditions. Such gaps were indicated, for example, when interviewees spoke about which symptoms they found to be most challenging to manage. Data presented in section 4 indicated the symptoms which practitioners and families found to be most challenging for each child in this study. Practitioners reported that often the challenge of symptoms was not the symptom itself, but rather the difficulty it presented as they grappled with the limits of their knowledge. The majority of responses were framed as connected to the individual's lack of experience with these conditions and consequently, about gaps in their knowledge, limitations to their clinical skills and challenges in finding supportive work environments.

Gaps in clinical skills were often related to the management of specific symptoms. One practitioner, for example, spoke of finding visual impairment difficult to manage due to having no prior experience dealing with it as a symptom. The following quotation thus illustrates the position of the majority of speakers who have a narrow field of expertise, rather than specialist paediatric palliative care:

Interviewer: Could you tell me a little bit about the symptoms that concern you the most with [child] and why?

Practitioner: I think it's the visual impairment, because I'm not used to it. (Practitioner 24)

Other interviewees spoke of gaps in their skills regarding how they might prepare families for the transition to end-of-life care. Discussions of end-of-life planning are explored further in section 7.2, focusing on advance care planning. The speaker below, however, indicates that they struggle with limited skills and knowledge about how to lead such conversations:

[Talking about death and dying] is something that never comes or rarely comes up that ... paediatricians need to and, I don't know, the training for it, it doesn't come up in true palliative care setting, but I think it's, although we all know how to set up the end-of-life care along with all the policies and forms, I think that the, the distressing part of it and how to bridge that distress with professionals ... is something we haven't, we don't address, and maybe partly because we don't admit to it. (Practitioner 25)

Many practitioners reflected on the training which would support them in their role and this was often in relation to knowledge about the condition, its symptoms and how they might manifest in

different children. The following two excerpts illustrate the training needs of practitioners in non-palliative care settings:

More information on what to expect. Access to others who work in similar settings. (Practitioner 24)

What he has, what type it is, what, how it progresses to that and what happens as it, the stages it goes through, because I realise that [child] is, he's slowly deteriorating, that his organs are going to shut down, but we don't know exactly what happens after that, I know he is going to pass away, but how it, how it is going to happen [...] when he does pass away, he might end up, someone says he might end up taking a massive seizure, and that would be the end of it, but it's not always a seizure. (Practitioner 36)

Training which centres on the family and ways in which to manage their transition through the condition was recommended by a number of practitioners:

Practitioner: There could be better training.

Interviewer: Okay, specifically around ... a certain area or just generally?

Practitioner: I think just ... it ... sort of even just the talking to families that ... that you know, have just had that awful diagnosis and how to ... how to ... how to help them or ... how to talk through things like that, it's not something that as medical professionals we really get taught, it's just something that you ... you work your way through ... you know, on the medical side of things we're ... we're quite well trained, but it's ... helping with the emotional and psychological side of it that the families deal with. (Practitioner 40)

One practitioner talked about their experiences with a family struggling to come to terms with their child's life-limiting condition:

I've seen with the family how they've gone from real denial, into sort of wanting the best and making the most out of having [child]. We've had a bit of training on what it feels like ... so it's been really interesting for me, obviously not, you know, difficult for them, but it's been interesting to see that change actually. Especially in [Mum], so I've learnt a lot from that. And obviously working with the other professionals and carers involved in the family ... that's been good. And also just getting the experience of, you know, how things work in that area, you know, with the doctors and you know, how you get your support. It's always good for experience for other families which you can take with you. (Practitioner 38)

The desire to share experiences with other practitioners is mentioned a number of times as an important learning opportunity. Thus, the absence of a work environment where learning from others was possible also presented a difficulty for practitioners. Several interviewees spoke of being encouraged to participate in discussions with other practitioners to gain support in their work:

We've always been encouraged to take part [...] and allowed, I think would be the word that I would probably to use, to get involved with, you know, sort of areas outside of [the hospice]. (Practitioner 42)

Such meetings and networks enable practitioners to gain valuable support from each other:

As physios don't get a lot of formal support through our regular training, it's really, you know ... [laughs] ... It's an area that's lacking ... and I think through experience, 'cause I've been working for 30 years as a physio now, so a lot of it is just things that you've picked up a long the way. (Practitioner 40)

Thus practitioners spoke about gaps in their symptom knowledge, a shortfall of opportunities to discuss their experience and practice with other professionals, and struggles with their knowledge of specific symptoms and clinical skills. Presenting these issues as gaps was linked with the rarity of the conditions, and often with quite narrow expertise.

7.1.4 Silo expertise

The interview data indicate that many practitioners have symptom-specific experience and expertise rather than experiential knowledge of the conditions themselves. Thus, interviewees would talk about knowing a great deal about managing one symptom (such as visual difficulties or seizures), but not how that fits with the condition as a whole. This is termed silo expertise, to reflect the idea that each of the practitioners interviewed demonstrated professional expertise, but that this was often related to a narrow tranche of knowledge, rather than about the condition as a whole.

The following clinician describes her expertise in autism and how this enables her to support children with Batten disease who struggle to communicate and relate to others. The practitioner indicates that expertise relating to autism has been helpful in supporting the family:

We would pass on ideas to [the parents] or within our practice that we've picked up from working with children generally with different issues. Like the sensory issues ... that we use with children with autism that we've passed on and you know, they use with ... [the children]. I suppose really our experience of working with children with autism has given us a huge repertoire of things to try. (Practitioner 24)

The above speaker contextualises her knowledge about autism by stating early on in the interview that she has no other experience of supporting children with Batten disease, or indeed with visual impairments:

Interviewer: What is your experience of supporting families who have a child with Batten disease?
Practitioner: This is the first experience. Yeah, this is the only one ... so none really ... [laughs]
Interviewer: It's so rare, this is the thing. How do you feel that the care needs of the children with this condition differ from the others that you have at the school, from your experience?
Practitioner: I think really the visual impairment is the huge, huge difference ... for us, because we've never had a child with a visual impairment before. (Practitioner 24)

Thus, while an experienced practitioner with specific expertise, this knowledge is focused on specific symptoms rather than the wider range of effects which the conditions are known to have. The following interviewee also indicates the limits to their expertise, and how this triggers referrals on to other services. Despite speaking as a physiotherapist, when a child experiences an acute exacerbation of secretion and breathing problems she refers the child on:

I'm aware of, as the child progresses with their disorder breathing is a difficulty and then if they're ill, they potentially could have more secretions, but more of an acute physio role rather than my role in the family ... but it'd usually be, if they're that bad they would be hospital admitted and then get treatment that way. Yeah, 'cause with being in our position we're not really confident with the respiratory side of things ... we don't do it enough that we would, call on our acute physios. (Practitioner 37)

Interviewees reflected on the limits of their knowledge of these rare conditions. Several speakers indicated how they are able to provide information on the most common symptoms, but without specialist medical training are constrained to signposting families to other services and sources of information. The following clinicians speak about the limits of their expertise:

If I wasn't happy with ... how things were with her fitting because I wouldn't really be experienced enough to know, you know, about changing those sort of medications. I would be more experienced with the pain side of things. (Practitioner 38)

Our knowledge is sometimes limited ... on that, because obviously we're not ... we're not medically, trained, some of the things we are able to give information on because it's well known, but usually it's sort of signposting them saying you know, you're probably better off speaking to your specialist about that ... or speak to the physio under you know, the specialist centre, 'cause they'd be best advise you on that. (Practitioner 35)

Silo expertise also meant that practitioners had limited knowledge related to symptom interpretation. The following speaker reflected on their experience of observing a seizure for the first time and the need to check their interpretation with a colleague. Thus, the impact of having narrow ranges of knowledge had considerable far-reaching consequences when practitioners were unclear as to how to make sense of new symptoms:

*Interviewer: You noticed [child] having a seizure?
Practitioner: Yes, well, as far as I'm aware it was a seizure.
Interviewer: As far as you're aware.
Practitioner: Yes.
Interviewer: Okay.
Practitioner: It was just a real difference in his behaviour and I got somebody else to watch and she agreed with me that that's what was happening. (Practitioner 24)*

A few exceptions to the sense of silo working were visible in the data, however. For example, some practitioners had specialist experience of supporting children with these rare life-limiting conditions. Some speakers quoted in sections above reported having decades of experience in paediatric palliative care, and knowing many children with MPS disease or Batten disease. One interviewee

reflected on how her experience in supporting many families with an MPS condition had led her to state that she was confident in supporting children with this condition:

At one time I think I had six children on my case-load ... and I know several others within my team who have children with the same condition so ... I would think I am quite aware of it now just because there are so many here ... I'm pretty confident in supporting them. (Practitioner 37)

Examples of clinicians who have in-depth knowledge of the conditions were rare, however. Consequently, much of the data illustrated the narrow focus of practitioners' expertise. One of the products of having limited knowledge of the conditions, but knowledge of *part* of the symptomatology, was that practitioners often adopted a key role in care coordinating. Indeed, as the model in Figure 17 indicates, it is the silo expertise and rarity of the condition which act as prompts to taking on a role of care coordination, which then leads to a range of other key dimensions of care that the practitioner undertakes.

7.1.5 Care coordination and advocacy

The dominance of silo expertise suggests a link between particular knowledge around rare life-limiting conditions and being positioned as mediators, advocates and coordinators. Thus, throughout the interviews practitioners reported that both they and families took an active role in linking other agencies together, for example, through sharing information and coordinating treatment plans. Many practitioners found the need to source more specialist knowledge and this invariably led them to becoming more involved in care coordination. Indeed, practitioners indicate that care coordinating is an important component of how they support families:

I was a kind of central point from where they kept going to the different specialities. (Practitioner 25)

Sharing information is also driven home by the following speaker, who illustrates that this is one of the central features of their role:

I carry out lots of different roles, but really communication is the key. So I would say I liaise between home and school, medical staff, the local respite carers, the hospice, psychology services and other sort of professionals and with that's included [the hospital], the MPS society for the specialist advice. So having done that it's really sort of reporting and sharing information regarding the symptoms and making suggestions of what we could do, and asking, obviously, for advice from people. (Practitioner 28)

Care coordination involves supporting families with both medical and social issues. The following two excerpts illustrate the role that practitioners have in supporting the family beyond clinical symptom management:

I do home visits and I just spend time with them listening to their concerns, taking their concerns on board and doing really, sort of advocacy, a little bit of advocacy for them with professionals. (Practitioner 31)

I've only had the one child [with Batten disease]. I think because, I've always made myself very available to them, so that, you know, if situations arise they know that they can get in touch quite quickly. I don't give many patients' families my work mobile number ... you know, unless I feel that there's a need. So they, from the start, they've had, my work mobile and been able to contact me, even if it's just with a question, or you know, something, and I think because I'm quite happy to come to other appointments with them to sort of advocate a certain, about when we're looking for equipment or wheelchairs ... or things that, I make time to come to all these appointments with them to ... to make sure that their ... their child is ... is getting what they need. (Practitioner 40)

Advocacy and connecting different parts of the support team with the family were common features among this interview cohort. Additionally, the speaker above was not unique in offering their mobile phone number to families, in order to ensure that the family could access them or their expertise at any time. At the heart of this part of the model is a sense that practitioners view the family in its widest context, and offer systemic support by drawing together key services required for the care of the child and family. Care coordination therefore shifts the focus away from individual physical symptoms (such as that made possible by silo expertise) into a domain where wider psycho-social and medical needs are met.

7.1.6 Above and beyond

The data indicate that in many cases practitioners went above and beyond their formal duties in offering support to families. That is, they took on roles which are greater than what might be expected in their employment contract. Examples of going above and beyond often related to the lengths they would go to in undertaking care coordination and, by virtue of their close relationship with families, offer support outside regular working hours. Some interviewees suggested that going above and beyond is a reflection of two contrasting approaches to supporting families at the end of life. The following clinician explains the difference between palliative care and palliative medicine:

I'm of the controversial opinion that palliative care is different from palliative medicine ... you cannot administer care without actually becoming involved to a certain extent ... whereas palliative medicine just straightforward symptom control [...] you can do that without becoming involved with the family. So do you, do you see ... a big difference there [...] but actually to administer effective care the family have to trust you ... and it's very difficult to have a relationship of trust without having some involvement and, and I'm not suggesting you know you become part of every family but, you, you have to have some level of involvement which is why I can give them my mobile number and know that they won't ring me unless there's a problem. (Practitioner 31)

Practitioner 31 offers her mobile phone number to families. The speaker positions this as part of their 'controversial' approach to care, and is a signifier of going 'above and beyond' that which is traditionally offered by practitioners. The following quotation illustrates this further, as another speaker reflects on giving out their personal contact details in order to enable families to contact them in an emergency or in order to offer the family respite:

*Interviewer: Do you have any contact with [the family] at the weekends or anything?
Practitioner: Generally not, I have to say I did give them my phone number for if they needed to talk through the summer holidays I told them when I was going away because they were really quite low as we broke up and I was quite concerned ... They've not rung me. They were very concerned that they wouldn't, they said that you know, 'It's your break', I said, 'No, look if you need to talk about anything, if you want me to come for an hour and give you a break ...' you know, 'ring me ...' but they've not. No, they're not the sort of parents that would take advantage, I think. (Practitioner 24)*

Going above and beyond appeared to be intimately bound to the positive relationship that practitioners had developed with family members. As the following excerpt illustrates, professional boundaries were often re-drawn to accommodate the relationship:

And there are no boundaries or barriers between ... the family and us and you know we do what we need to do and yeah, I know that we sometimes bend the rules a bit ... But that's ... you know, that's just ... we just do what needs to be done, and as I say, that's the tone in this [organisation]. (Practitioner 29)

Maintaining contact with families out-of-hours and developing a more personalised relationship with the family were thus key exemplars of going above and beyond. As the model in Figure 17 indicates, going above and beyond has a recursive interaction with the provision of emotional support and the clinician's relationships with the family. It is also closely tied-in to care coordination, as practitioners take on tasks in advocating for families.

7.1.7 Relationship with the family

The practitioner model in Figure 17 illustrates a cyclical flow between roles in care coordination, providing support above and beyond their role and the relationship with the family. Practitioners described their position as being a 'central point' and that they 'work closely' with families. Many of the practitioners interviewed had known their families for many years, and had consequently built a closer relationship over time, or had played a key role in the child's diagnosis, which had also strengthened the bond between practitioner and parents:

I think I do know them well, let me put it this way I've probably got closer to them than, compared to a lot of my other patients. (Practitioner 25)

I was the lead ... in her diagnosis. And holding the reins for her considerably complex treatments, so I'm probably the person that she's ... and the family have come to know best I would guess. (Practitioner 34)

Interviewees directly cited their relationship with the family as informing their practice:

I just text her maybe on a weekly basis over the holidays to say 'How is he? How's the boy?' [...] So it's that kind of relationship. (Practitioner 29)

Practitioners also spoke of how going into the family's home had contributed to enabling them to develop a close relationship. This alongside making themselves available to the family (going 'above and beyond') strengthened the bond:

I'm sort of like the most consistent person who goes into the house, probably [...] And just being involved in her care and everything, and experience of the condition you know, with other families, and just probably 'cause we're available 24 hours, seven days a week [...] There's, you know, there's a good relationship, you know, sort of, the trust has built up. And obviously, you know, I sort of ... got the best interest of the family and the child. (Practitioner 38)

For some interviewees, the process of getting to know the family and learning about the condition had resulted in them reflecting more widely on their identity as clinicians. The following speaker muses over the impact of supporting a family with a child with a rare life-limiting condition, and that maintaining a 'professional distance' does not always feel appropriate:

Interviewer: What have you learned through your experiences from working with this family?
Practitioner: Of course I learned about the disease a lot ... I think very much my experience and how much I should be trying to help them ... I think it's been instrumental in them being able to go through ... I think reluctantly so I probably allowed to open up a bit more to the family ... normally you know you keep all the professional distance and such like to be objective but I felt with a family like the [family] had two children ... I've probably learned a lot that bringing some of the guards down is not such a bad thing after all. (Practitioner 25)

Thus, the relationship with the family not only affects practitioners' roles in care coordination, but also, critically, can impact on how they view themselves and the appropriateness of traditional professional boundaries. Associated with the notion of the relationship feeding into different ways of practicing, the following speaker illustrates the idea that the relationship is closely tied in with a sense of honesty and openness. Indeed, honesty was a common trope in how practitioners described their relationship with parents and families:

We share everything with the parents, whether it's good or bad, we don't just tell them we've have a good day, if things have been not so good. And right at the beginning, you know, I would say to [family] that I'm going to be really honest with you and ... tell you if ... if [child's] been upset or if there's been something wrong, I said ... and you know I'd really appreciate it if you do too, because that's the ... the best way of working together for the benefit of the child. We also have the welfare of the parents very much embedded into the way that we work. (Practitioner 24)

Practitioners' accounts also indicated the recursive connection between their relationship with the family and the role they play in care coordination. The following speakers identify that through developing a positive relationship with the parent/s, they come to know a great deal about the family's circumstances, social needs and emotional state:

I [work closely] with family, you kind of get an overall picture, everything that's going on with them really, try and help them in all areas whether it be [the child's physical ability] but also the social needs as well. (Practitioner 37)

I think I would say I know them well. Well, Mum especially. Because we tend to actually ... she tends to be the one that contacts and she ... she's sort of tended to be the one at home when you ring and it's a relationship that's developed over seven years and I think she feels free to say whatever it is and would always ring if there's anything, and she's always keen to share what's been happening. And I think over the years I have become aware of the impact that [child's] condition has on the whole family. As much as you can, you know, I see her in school, I've seen her at home, I hear what's happening, I hear from the respite carers so I think I have a pretty good awareness ... It's never the same as, obviously, living in that situation ... but as much as I can do I think. I think I do understand their experience because I've seen, you know, because I've witnessed so much of it first hand ... in school, home, wherever, I ... you can't put yourself in somebody else's shoes but you can imagine. It's exhausting, very wearing and it's ... no matter what they do it's not going to make a ... a difference to the end outcome, so ... I think I'm aware. (Practitioner 28)

The above lengthy quotation depicts the connection between developing a strong relationship with the family and the provision of emotional support.

7.1.8 Emotional support

It is perhaps not surprising that practitioners reported emotional support as part of their unique relationship with families in this study. Examples of emotional support include making themselves available, listening, and sharing their sense of empathy with a family. As with the section above, the provision of emotional support indicates a shift in models of working, from a pure medical model focused on symptom management to one which is cognisant of psychosocial needs:

Interviewer: And how well would you say you know the family?

Practitioner: I think very well I would say. Yes ... I think fairly well, I think 'cause I've always been available to them, as I said, you know, that Mum's quite often phoned up, even just to sort of sound things off or just to, you know, if she's a little bit concerned about something and, so you know, I think we've had a very good open rapport and ... and very ... you know, I think she's ... she's quite happy to just pick up the phone if there's anything concerning her. (Practitioner 40)

Some practitioners went as far as to say that they had a full understanding of the family's experience, indicating an acute awareness of the emotional component of supporting a child with a rare life-limiting condition:

Interviewer: How well would you say that you can understand their experience in caring for the child?

Practitioner: Fully understand.

Interviewer: Would you say that's through the experience of working with the boys in particular?

Practitioner: Yes, yeah and talking and listening. Mainly listening. (Practitioner 42)

Emotional support provides families with an outlet for some of the build-up of pressure associated with caring for a child with a rare life-limiting condition. A number of practitioners reported examples of giving emotional support through active listening and expressions of empathy. Some practitioners had organised additional levels of care to support families in anticipatory grief and to bolster emotional resources in advance of bereavement (referred to below as 'the path that is expected'):

We have now got somebody going out who is family support worker who is going to take on a role of the pre-bereavement role ... with [the family] although he hasn't labelled it as that. But he's started to go working with [Mum] now and he will sort of be talking to her and obviously as these feelings are coming up, he's getting to know her because obviously [child] is going down the path that is expected. (Practitioner 26)

The following speaker illustrates their understanding of the impact of the life-limiting condition on the emotional needs of the family. The quotation comes from a section in the interview when the practitioner is asked about the most difficult part of the condition for the parents to manage:

*Practitioner: His whole deterioration is, for [Mum], she's losing her little boy ... all the symptoms are expected but they're still hard to cope with when your little boy doesn't recognise you and he's now starting to have seizures and ... you know, he's got pain, she doesn't know where, you know, if he's crying and you can't relieve it ... you know, so probably [...] I think probably all of it really for her ...
Interviewer: It's the emotional impact rather than [...] the sort of physical symptoms?
Practitioner: I think it's equal, I think it's all of it [...] we mustn't underestimate the emotional side of her little boy deteriorating. (Practitioner 26)*

The above speaker highlights the impact that the chronic deterioration of a child over a long period of time has on the emotional needs of the family. The above speaker goes on to explain how important it is for them to emotionally support the family:

It's very important to listen I think, to listen to them and actually support them where they're at. And for [Mum], as I say, she wants somebody who is very open and she wants to know things as they're going to be [...] I mean I couldn't lie to [Mum] because ... any trust we had [...] I think the thing I have learned from [Mum] is that you take each family as it is and support them where they're at now. (Practitioner 26)

The example above highlights the significance the relationship with the family has on practitioners' outlook in supporting a family through emotional support.

7.1.9 Summary

Practitioners had a range of knowledge and experience in MPS disease and Batten disease, though many expressed their expertise in relation to specific symptoms and silo expertise, rather than the whole condition. Meanwhile, other interviewees felt that the specific diagnosis was less important than the overall stance in supporting the family.

A consequence of many practitioners having limited disease-specific knowledge was that they often undertook care coordination to ensure the family was well supported. This care coordination took

the form of advocating for the family, and often included elements of care which were above and beyond what practitioners typically provide. Thus, the practitioner model in Figure 17 has a recursive link between care coordination, the relationship with the family, going above and beyond and providing emotional support.

7.2 Advance Care Planning

Practitioners were asked at what point they would be involved with the family in talking about advance care planning (ACP). Their responses were revealing, both in terms of their ideas of what this constituted and how it should or could be undertaken. Practitioners' views also have wider significance for their overall role in supporting families with a child who has a rare life-limiting condition, as indicated in the practitioner experiences model (section 7.1).

ACP is typically used to describe clinical pathways and protocols, which guide and document family decision-making about treatment and care wishes as their child's condition deteriorates. The term is used here to incorporate this meaning, but also to describe informal discussions with families about preparing for their child's deterioration and, ultimately, their death. ACP is thus used in an inclusive fashion, reflecting the ways in which respondents talked about their roles with families, and includes conversations from the outset of diagnosis, not just at the end stage.

The model described in this section of the report (Figure 18) is derived from the interview data, and suggests that there are a range of features which impact on practitioners' abilities to undertake conversations around ACP. The model in Figure 18 below illustrates the different components, framing them as barriers and facilitators. The duality of barriers and facilitators, however, should not be read as absolutes, but as representing a range of features that frame practitioners' abilities in talking about ACP. Thus, 'accepting that decline cannot be prevented' is presented as a barrier, but conversely 'accepting the inevitability of decline' is a facilitator. The model, however, is derived from the ways in which practitioners spoke, and their implicit framing of features as affordances or constraints on ACP.

Much of the experience gained by practitioners in relation to ACP was drawn from supporting children and families with other conditions. This is due to the rare nature of the conditions explored throughout this study, resulting in many practitioners having limited exposure to other children with the same condition. Very few practitioners (n=4) spoke of supporting other children with this condition in relation to their experiences of ACP.

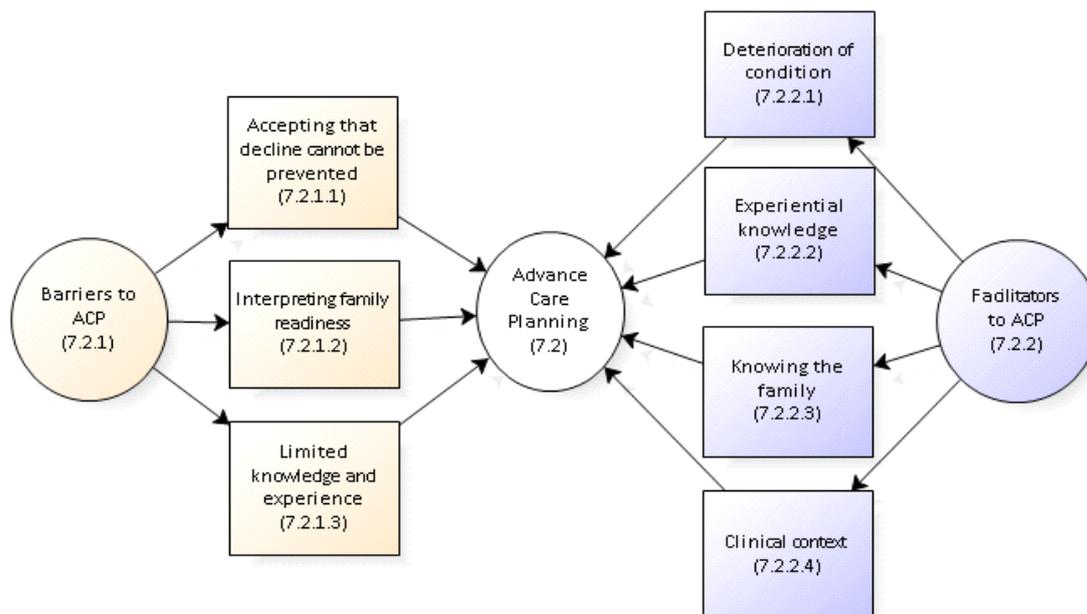


Figure 18: Advance Care Planning

To help the reader navigate through the complex and rich accounts, each part of the model has a reference within it indicating which section of the report describes the data.

7.2.1 Barriers to Advance Care Planning

7.2.1.1 *Accepting that decline cannot be prevented*

One core feature indicated by practitioners in relation to ACP was a reluctance to accept that the child had a degenerative condition, and the practitioner felt powerless to prevent decline. Some interviewees explained the difficulty in accepting inevitable decline and death with reference to how difficult this is for junior members of staff. The following excerpt indicates this difficulty and identifies the dearth of appropriate training to address such challenges:

The main training needs ... within the hospital, juniors in the hospital it is a changed mind-set, from treatment to palliative and that's a big step for a junior doctor ... even for some of the senior doctors ... oh yeah, that's, that's a really big step. (Practitioner 31)

The above interviewee provided additional detail on the challenges of accepting that decline is inevitable by indicating the tensions between families who have expressed their wish to have no further treatments offered, and staff who find it challenging to stop offering interventions. The following quotation draws from the practitioner's wider experience in supporting families at the end of life, not solely the family involved in this study:

I was actually a barrier between the, the child and the junior staff, so when I went to the consultant she said, 'Yeah, the parents said they didn't want anything,' we shouldn't be doing any of that, but the, the registrar who's not made the jump between, you know I, I do everything in my power to make children better and to, when the parents have decided to withdrawn treatment, let's just wait for this child to die. (Practitioner 31)

Practitioners find it personally and professionally taxing to change care-plans in recognition that there is no further treatment available for the child. One interviewee explained this with reference to the 'hurt' that families and practitioners feel, which creates a barrier to undertaking timely and sensitive ACP conversations:

It's an admission that you can no longer help the family to cure the disease or whatever it is ... and I think it hurts the family the most, but it hurts the caring physician, too and that aspect of it I, is probably poorly addressed ... but I think we have to facilitate why we don't do it well and I think if you explored the 'why' it's because there is a lot of hurt involved there. (Practitioner 25)

Thus accepting that little could be done to offer curative treatment presents a difficulty in engaging in ACP conversations at both personal and professional levels.

7.2.1.2 Interpreting family readiness

Many practitioners were reluctant to talk about the future with families, finding it easier to locate conversations in the here and now, rather than open discussions about likely decline and subsequent decision-making. For some interviewees, not engaging in ACP conversations was explained with reference to having determined that the family was not ready to engage in such conversations:

It depends, every family's different ... for some families we never complete it properly because they're not ready to. (Practitioner 26)

Other interviewees expanded the point indicated in the above extract, creating a distinct sense that the onus was on families to dictate when the time was right to engage in such conversations. Ultimately, this meant that practitioners did not position themselves as responsible for ensuring that ACP discussions occurred, and placed responsibility for this on the family themselves. In response to a question about when they might raise ACP with the family, the following practitioner responded:

At any time that the family raised it. (Practitioner 28)

Further, other practitioners suggested that the timing of entering conversation about ACP was when family and care providers were in agreement about the palliative nature of the care required. The following professional reflected on this issue, in relation to a child in the third phase of Sanfilippo disease:

Well it's difficult, actually, to pinpoint it, but really depends on the family, and when the doctors and the family agree that the child is ... is for palliative ... only for palliative care. (Practitioner 38)

While waiting for agreement that the child requires palliative care has meaning in conditions which may have been open to curative treatment (such as organ failure or cancers), it is hard to understand how this applies to children with conditions which are, by their very nature, life-limiting and degenerative. Thus, there is a sense that practitioners find it hard to enter discussions about ACP, and place responsibility for the timing on other people, rather than themselves. This implication that other people should take the lead in commencing conversations is particularly pertinent given the broad definition of advance care planning, and supporting families to prepare themselves for their child's decline.

7.2.1.3 Limited knowledge of Advance Care Planning

Linked with the idea discussed above that practitioners struggle to identify themselves as responsible for introducing the idea of ACP, some had limited knowledge of what ACP is. Consequently, some practitioners did not position themselves as having a role in supporting families to make plans for the end of life. Indeed, some interviewees expressed clearly that ACP was not undertaken.

One interviewee asked for clarification about what ACP was, and then indicated her view that this dimension of care fell under someone else's remit. In the following excerpt, the speaker indicates a view that ACP is something which occurs at a particular stage of the child's deterioration, rather than being integrated from the initial diagnosis of a life-limiting condition:

Can you just explain what you mean a bit more? We do have a good palliative care team, so I guess if the family were, if we knew it was getting to that stage that's where we would refer to ... It is, yeah. But again, it's up to the family whether they accept that or not. (Practitioner 37)

This interviewee captures a number of key points drawn out in the model in Figure 18. The speaker indicates limited knowledge of ACP; ACP is undertaken by *someone else*, rather than being held as a joint responsibility by everyone involved in the child's care, and positioning the family as responsible in guiding discussions. The speaker goes on to reflect further about whose responsibility it is to undertake ACP conversations, indicating that it is the professional who is most involved:

You know, I think it's whoever you, you know ... whoever's in the main ... looking after [child] at that stage, I guess. (Practitioner 37)

Other interviewees felt that ACP should happen only toward the end of the child's life, rather than as part of generalist palliative care activity, as an ongoing conversation which is begun when it is clear that the child has a life-limiting condition. The following healthcare professional illustrates the need for it to be undertaken by an experienced practitioner and refers to the recent diagnosis of a child with a rare life-limiting condition:

*Practitioner: I think there needs to be, you know, it needs to be somebody with more specific experience.
Interviewer: At what point would you be involved with the family in talking about advance care planning?
Practitioner: I'm not sure about that, I have to say. With everything being so new, I don't know. (Practitioner 24)*

7.2.2 Facilitators of Advance Care Planning

A range of factors acted as facilitators to families and practitioners engaging in conversations about ACP.

7.2.2.1 Deterioration of condition

Often, acute episodes led to the discussion of ACP, as crises prompted urgent decision-making. The following quotation takes an extreme, though not unusual, case to illustrate sudden deterioration as a trigger to discussing end-of-life decision-making:

I think the trigger with [child] was because [child] had been intensive care with pneumonia which had then ... I think the family were very aware that, this could happen again and ... we'd said, 'Right, we needed to sit down and talk about what would happen. You know, or how far would you want to go the next time? Do you want to go as far as intubation? And maybe we should document that...' In that case, it, yes, it's the crisis that, that gives you permission. In other cases it's just, you know things are, are going downhill and, 'Do you think it'll be useful in the school if I had a little plan of, of what to do?' ... again, I have said to, to parents that if, 'If your child is taken to hospital they will resuscitate them, they will do a full resuscitation, unless you have made it clear that you don't want that done.' (Practitioner 31)

Thus, the above speaker marks out ACP discussions not just as the territory of the medical team, but something which warrants discussion in a range of contexts in which the child is likely to be, such as school. Other practitioners explained that noticing deterioration in a child was likely to lead to ACP conversations:

I think when it became, or becomes apparent that [child] started to become dependent on technology, so gastrostomy feeding, needing suctioning, that there's obvious signs of marked deterioration ... I would see as an appropriate time to introduce that subject. (Practitioner 28)

Markers of deterioration are often integrated into ACP paperwork (such as that promoted by the ACT (ACT, 2004)). In the quotation below, although deterioration acts as a facilitator, the speaker nevertheless continues to view this as someone else's job, as noted in the barriers section above:

We've noticed a marked difference in [child's] mobility, swallowing and [child's] just slow deterioration that, you know, it was inevitable that they [family] were going to have to have that, that conversation with the consultant. (Practitioner 27)

7.2.2.2 Experiential knowledge

For many interviewees the ability to engage in ACP conversations was explained with reference to their experiential knowledge, built up over often lengthy careers in palliative care. Some interviewees reflected on how their skill-set had increased over years of experience, making them much more able to confidently undertake difficult conversations with families in the present day. Indeed, the following practitioner explicitly references their own (and their team's) growing knowledge of these rare life-limiting conditions:

[The] professionals in my capacity do agonise about which, over time and having to deal with more patients like [this child] ... You're talking about 'end-of-life' and that whole planning for the end of life, it's always extremely difficult and I'd [sighs] ... I, I know myself ten years ago, it is something you don't want to do and you keep ... you know you keep pushing it for another day for a later day, for a later day, and actually, that day never comes until they come in with an acute illness and then it's someone who they don't know who ends up having to do it. (Practitioner 25)

Thus the above speaker highlights the tensions in not feeling able to undertake ACP conversations, that is, the risk that if someone known to the family does not lead the discussion, then it is likely to be done during an acute crisis by someone who does not know the parents or child.

For some practitioners, their experiential knowledge enabled them to see ACP not as an ultimate act of decision-making, but as a process which could be reviewed and repeated at any time. The following interviewee begins their account by reporting her own speech in a conversation she has had with parents:

'You're free to change you're mind and to call the ambulance if you want to, as long as you know what will happen ... when they arrive, or would you like us to let the ambulance people know that if they're called, that they are to make sure she's comfortable and maybe give her a bag and mask but you don't want her taken to the hospital or you don't want her intubated when she gets there,' and allow those conversations just take ... you always say to the parents, 'Look,' you know, 'this is written down, it's not written in stone ... you know, and at any stage you decide that you want more done or less done then you're her mum and dad, and you have the right to do that.' (Practitioner 31)

For some interviewees, talking about ACP was clearly framed as requiring attention from very early on in the disease's progression. The following speaker indicates, clearly and compassionately, that their knowledge of the specific life-limiting condition enabled them to be able to undertake conversations and ACP. Their experiential knowledge of the condition meant that they were able to hold an appreciation of the tensions in talking about decline with parents who witness their child appearing very active:

From the beginning ... just by, just talking with them and, you know seeing what they want, just going at their pace really. Answering questions and concerns when it needs to be addressed ... It's very difficult to look forward in the child's condition when they're actually running around and you're actually saying, 'Well, they will be, you know, less mobile, they will be this, they will be that.' You know, when actually, you know, they've got an awful lot of living to do first ... We do support any, you know, any changes that do happen ... as they go through their condition. (Practitioner 42)

Experiential knowledge also meant that some interviewees reflected on the difficulties in talking with parents about a child's deterioration and death. The following quotation is noteworthy since the speaker indicates the role that ACP documents and protocols should have in facilitating such conversations, and yet it is often practitioner distress and denial of decline which precludes discussions:

Although we all know how to set up the end-of-life care along with all the policies and forms, I think that the, the distressing part of it and how to bridge that distress with professionals ... is something we haven't, we don't address, and maybe partly because we don't admit to it but, but there is a denial. (Practitioner 25)

Thus, for some practitioners, while they 'know' about what to do, and have an awareness of the policies and procedures, a barrier to undertaking best practice care is the emotional toll this takes on practitioners. A further interviewee spoke candidly about her own past difficulties in discussing ACP with families. She reflected on how this was not a strongpoint for herself, and something that others in the care team are better at:

I find it difficult to bring things up ... Yeah. But there are ... there are some that are much better. And very often there will be people that bring out different things that you know ... we all work with the children and we all work with the families and we don't always just work with our own contact families ... so there will be people on the team who work with the families and can probably, you know, offer different things ... working as a team. (Practitioner 38)

For this practitioner, team-working acted as a facilitator to undertaking ACP conversations, since colleagues could be called upon to have difficult conversations. Reflexivity of practitioners, in recognising their own barriers through years of experience, thus has the potential to facilitate colleagues to step in to support families in ACP.

7.2.2.3 Knowing the family

In line with best practice, 'advance decisions evolve over time through the development of a trusting relationship and an ethos of shared decision making' (ACT, 2010), many interviewees reflected on how their relationship with the family was instrumental in enabling conversations about ACP. Thus, in addition to experiential knowledge of children's palliative care, knowledge of the family was also a key component:

It's difficult to say when the times comes 'cause, if, if you get to, to know the families, you, you get to, get the vibes of, it's time for you to say something, or in the family they'd actually quite like to say something but they're not quite sure how to ... how, who to do it, so that's just a bit of people-watching really. (Practitioner 31)

Other interviewees framed their relationship with the family in terms of the ability to be 'open and honest', and without such characteristics, ACP discussions would be impossible:

It's much easier done in the cold light of day by people they know. (Practitioner 25)

Some practitioners illustrated how strong the relationship was between family and professional, for example the following speaker who talks about co-signing the 'do not resuscitate' (DNR) order:

I personally would say I know them better than any professional most probably involved with them ... to be honest, because ... they tend to, when [child], when [the parents] had the DNR they wanted me to sign it because they have known me so long and, you know, so I do feel that I do know them better than most. (Practitioner 27)

The following speaker also illustrates the power of knowing the family, and consequently having a good awareness of their comfort with different levels of discussion and decision-making. The speaker illustrates that for some families going into fine detail is appropriate. Knowing the family enables the practitioner to judge at what level parents are able to plan:

So it's very much gentle talking to them and really knowing the family and knowing where they're at because obviously, some families have started talking about that pre- sort of, bereavement. Sort of care, they couldn't cope with it, other families really want, almost have it down to a fine art so when the child dies they know which funeral director and we can, you know, if they want that arranged we can, you know, go along those lines, sort out what funeral director they want, what songs they want at the funeral, almost, really down to everything ... what the child ... they want to wear, you know, after they ... some families want that in huge, huge detail. (Practitioner 26)

One further professional expressed how she frames the support for the child as end-of-life care. At a time when the child was critically unwell, she had cuddled up to the family and initiated a conversation about the family's wishes for their child's funeral. This proximity, of 'cosying up in bed' appears closely related to the practitioners' relationship with the family:

What we do now is providing is end-of-life care [...] We were thinking this could be the end of [child]. We're talk ... this is about two years ago as well, you know. But I mean, [the parent] had spent the night and was sitting in a wee bed in the room and ... [parent] was in jammies [pyjamas] and this is about half eight in the morning and you know ... I just, we just kind of cosied up in the bed ... And we talked about [child's] funeral. (Practitioner 29)

7.2.2.4 Clinical context

In some instances, the clinical context of care provision was a core factor in how ACP was undertaken. In particular, distinctions were drawn between the palliative care sector and acute care sector. Some interviewees were employed by specialist children's hospices, and explained that their organisations have taken a lead in developing operational strategies. Some interviewees' organisations were referred families from tertiary care specifically in order to undertake ACP with them. Thus, while the data indicate that not all professional groups (and clinical contexts) see themselves as responsible for undertaking ACP, other organisations are viewed as specialists:

*Practitioner: Within our care plans we do address things such as resuscitation policies and things and we work with the specialist centres so they ... the specialist centres usually tell the families to contact us ... with regard to setting up a care plan ... to include sort of end-of-life.
Interviewer: So that's them, the clinicians, ducking it and asking you to do the [ACP]?
Practitioner: Yes! [laughs]. (Practitioner 35)*

The following speaker positions their context as being influential. Her role as a hospice nurse, who is involved in outreach care in people's homes, means that she is able to use this element of her professional care and knowledge of the family to integrate into informal and ongoing ACP discussions:

We can be proactive about things which I ... sort of being proactive as in you know if they're losing their swallow then talking about gastrostomies, not leaving it too late ... And those things, and hoisting ... you know talking about hoisting. Before we actually, before you actually need it ... Talking about it, getting the child used to it and getting the family used to because it ... it ... all these things that are really huge for the family and if you can sort of be proactive and not actually leave it until when you actually need it, then I think that's really important. Yeah, and I think that just comes with the experience of working with other families and seeing them go through it, and knowing how things are going to progress, whereas the families who are going through it aren't always aware of why you're doing things and the importance of it all. (Practitioner 38)

7.2.3 Summary

Often, practitioners that have regular contact with the family, and describe having an 'open and honest' relationship, will talk about wider support issues and planning for the future. Those practitioners who are familiar with a palliative care setting (hospice nurses, specialist consultants) may talk more openly about ACP, but the starting points are variable and often family-led. Although being family-led fits with the palliative care ethos, it does not necessarily prepare families adequately and often places responsibility on parents for initiating discussions. While many practitioners mentioned the presence of protocols and paperwork for documenting ACP discussions, none privileged this as a main facilitator in having such conversations, rather, it often came down to the relationship between family and professional. Thus, while practitioners are able to identify the value in undertaking conversations about ACP, many were not engaging in such discussions, even for children with clear diagnoses of life-limiting conditions.

8 Reflections on participating in the study

Several participants reflected on the process and experience of taking part in the study. With daily diaries to complete for a period of eight weeks, feedback on this part of the methodology was particularly interesting to note. Many families provided reassuring feedback that the daily completion of symptom diaries was not onerous:

It only takes 5-10 minutes at the end of the day, it's no bother at all. The more you do it, the better you get. The diary is quite structured and it's clear what you are meant to fill in.

(The diary is) simple and straightforward, it only takes a few minutes each day.

Not complicated and not time consuming, it doesn't even take five minutes. I keep it in view so as not to forget about it. It's so easy!

Other parents reflected on the research process as a whole:

It's quite nice to be able to give something back and to be able to help other families who are going through what we are.

I have found it to be a really interesting experience and I would be happy to speak to other families who are thinking about taking part.

One family spoke very powerfully about the sense that no one around them truly understands what it is like to support their daughter. In the following quotation, the mother reflects on the process of having completed the daily symptom diary and taken part in the interview about her daughter and the impact on the family:

Mum: No one has actually, I mean apart from you, no one has actually come in and said, do you know, 'Give us a breakdown,' like the diaries, give us a breakdown of what [child's] routine actually is. And ... 'cause I don't recall anybody ever doing that have they? There's nobody said, 'Tell us exactly what [child's] routine is day-to-day.'

Interviewer: Do you think that would be helpful for people that are involved in her care then, to really understand?

Mum: I think it probably would be but do you know, the one thing we've found here is that they're not interested.

Dad: That's just what I was going to say.

Mum: In knowing ... and I think probably [our support worker] would even say the same, in that respect, do you know, they don't want to learn because therefore, if they learn then ...

Interviewer: Then they've got to do something.

Mum: They've got to do something about it. (Family 15)

For the family quoted above there is a sense that not understanding what daily life is like for the family is a marker of services' lack of interest and of the minimal support they receive. It is noteworthy that the parents believe that in telling others about their experience, the account would be so compelling that they would be offered additional supports.

9 Discussion

The study has contributed to the evidence base of the symptom profile and family experiences of supporting a child with a rare life-limiting condition. The study's primary contribution was in shifting the focus away from that reported in the extant literature (McCluggage and Elborn, 2006), notably, the study looked broadly at the experience of symptoms, to include behavioural as well as medical concerns. Consequently, this is the first research which reports the lived experience and consequent impact of the full range of symptoms of children with MPS disease and Batten disease.

By following families prospectively over two months and documenting the symptoms children experience together with the associated impact on their families, the study has made contributions to a significant gap in knowledge related to the symptom profile of these rare life-limiting conditions from the family's lived experience. The qualitative component of the study contributes to the development of a better understanding of the *meaning* of the symptoms recorded in the daily diary. Interviews enabled the in-depth exploration of the overall illness experience and impact of caring for a child with one of these rare life-limiting conditions. The interviews produced new insights in their own right, focusing on the wider ramifications and meanings of symptoms and the family experience.

In the cohort of children with Sanfilippo disease, which included children at all three stages of the condition, the presence of a wide range of symptoms was confirmed. In particular, a high prevalence of behavioural symptoms, such as hyperactivity, repetitive behaviours and agitation, were recorded. Such symptoms were also found to be rated high in terms of severity, alongside disturbed sleep.

The cohort of children with Batten disease also included children at different stages of the condition (with some being very advanced and others having only received a diagnosis with the past year). Symptoms of joint stiffness, agitation, secretions, cold extremities (hands and feet) and disturbed sleep were particularly high in prevalence. Of these, joint stiffness, agitation, secretions and disturbed sleep received high severity ratings.

Parents and practitioners were asked to identify those symptoms which pose particular challenges in their management. Both groups identified similar challenges in managing some symptoms. For those supporting a child with Sanfilippo disease, there was acknowledgement from both parties of the significant difficulties in managing behavioural symptoms, such as hyperactivity and aggression. For both Sanfilippo disease and Batten disease, specific symptoms were identified as being troublesome for parents and practitioners to control and effectively manage, namely seizures, pain and muscle spasms.

The data also demonstrate distinct differences between parent and practitioner viewpoints in which symptoms prove most challenging to manage. Families struggled with symptoms that they felt signified the progression of the disease or which posed an acute threat to their child's health. Families also struggled with the physical and mental exhaustion arising from the relentless and constant nature of the symptoms whilst practitioners struggled with assessing symptoms in these children due to communication impairments associated with the disease process. This signifies some disjuncture between the differing perspectives of the symptom profile and impact of supporting a child with a rare life-limiting condition. This finding was signalled in the Phase 1 survey, and driven home by the symptom diary and qualitative data in Phases 2 and 3.

A major finding of a recent study (Parkes et al., 2009) identified that psychological symptoms in children with cerebral palsy, specifically hyperactivity and emotional disorders, were key predictors of high stress in parents, and may be more important than the severity of the child's physical impairments. This finding provides further evidence of the importance of considering both physical and non-physical symptoms. On the whole, families did not perceive the various management

strategies they employed to manage these symptoms as being particularly effective, suggesting the need to further explore services and support to help families cope with managing such symptoms.

Effective management of challenging behaviours presents an ongoing difficulty. Practitioners from a variety of disciplines are committed to helping children and families but feel there is little they can offer in terms of specific interventions. Challenging behaviours are likely to be caused by numerous factors which could be a direct result of the disease process where abnormal metabolites are deposited within the central nervous system and lead to cognitive impairment (Bax and Colville, 1995).

Parents described that their child's frustration can often escalate into aggression. Similarly, aggressive behaviour can often signal that the child is trying to communicate something to the parent or caregiver. For example, pain or distress may manifest in aggressive behaviour. The complexity of symptom interaction therefore necessitates all involved in the care of children with conditions such as Sanfilippo disease to collaborate in documenting strategies which work and those which do not, to develop a greater understanding of this symptom and successful interventions to support the child and family. A study conducted nearly two decades earlier, surveyed families of children with MPS disorders to explore the nature and prevalence of behavioural problems in this population and found similar results to the current study with behavioural issues and sleep problems being highly prevalent and parents reporting that they received little or no support in the management of these difficult behaviours (Bax and Colville, 1995). Such findings, in conjunction with the current study, add further weight to the necessity in directing efforts to help families manage what is a devastating and exhausting symptom.

Practitioners supporting children with these rare life-limiting conditions are met with further challenges in assessing symptoms, arising partly from difficulties in communicating with children who are cognitively and/or sensory impaired. This finding resonates with the work of McCluggage and Elborn (2006) who described how anxiety arose in hospice nursing staff when trying to assess the needs of children with life-limiting conditions who could not communicate verbally. Struggles in effectively communicating with the child may be a reflection of practitioners lacking the experiential knowledge and the intimate relationship that families have with the child, thereby stressing the importance of practitioners listening to and learning from families. Studies in children with neurological impairment have demonstrated the need for practitioners to have knowledge of the science of pain relief, familiarity with the condition, and knowledge of the individual child for effective management of pain (Hunt et al., 2003b)

As children lost abilities, such as walking or being able to see, this brought about realisation of the life-limiting nature of these conditions and intensified the emotional distress parents felt. This acknowledges the importance of ongoing support for these families as they experience grief and losses throughout the trajectory of their child's illness. Menezes (2010) identified several 'moments of realisation' or times when the life-limiting nature of the child's condition was further illuminated to families, heightening their uncertainty and the need to talk. The 'acute loss of abilities' was one such moment which is echoed in this study's findings. Practitioners supporting families should be cognisant of such times along the illness trajectory which are key opportunities to facilitate communication within the family system (Menezes, 2010).

The rarity of these conditions means that there is a lack of information on symptoms. Parents are faced with challenges in terms of getting the proper support, equipment, medication and a comprehensive understanding of their child's needs. Many practitioners had specific, silo expertise, which meant that they had generalist knowledge rather than disease-specific knowledge. This meant that there was greater emphasis on parents becoming experts in their own right, as clinicians referred to their experiential knowledge. Access to help and support are important elements of families' experiences, and access is mediated by the family's expertise in managing the condition and

with the role of members of wider family system. Thus, the uptake and accessibility of support is mediated by the availability of people with the right skill mix to support the family, which itself is impacted by the rarity of the condition/s. There is a considerable risk then of both families and practitioners feeling isolated and unconnected with others with similar experiences.

Families developed a great deal of expertise through their experiences of supporting their child. The findings from this study, however, go beyond that conceptualised as the expert patient as 'autonomous', 'independent' (Shaw, 2004), or engaging in self-care/self-management (Badcott, 2005). Rather, unlike these other depictions of experiential knowledge, this study does not imply an ideological imperative that the family take on a position of expert. Expertise in the context of this research is derived through necessity, which itself stems from the rarity of the condition. Expertise is then associated with both management of symptoms and technical understandings of the condition (Tyreman, 2005).

The life-limiting condition has impacts which reverberate around the wider family system (Steele and Davies, 2006). Many families experience complicated relationships with extended family members compounded by the nature of their child's condition. Such complex relationships hinder access to familial support and impact parental wellbeing. Stress is a known correlate of decreased family functioning (Streisand et al., 2011), and was evidenced in the data regarding strain on the parents' relationship. Offering family therapy to those most at risk of complicated bereavement outcomes is recommended in paediatric palliative care prior to the child's death (Bartell and Kissane, 2005), and has proven outcomes in significantly reducing distress, depression and pathological grief in palliative care populations (Kissane et al., 2005).

Recent trends in research have plotted the sequelae of inherited conditions across generations and impacting on relationships. The majority of these studies have focused on adult populations, for example, women affected by the BRCA breast cancer gene (Boenink, 2011), and document the need for support for families affected by rare conditions (Atkin et al., 1998; Senior et al., 1999) such as genetic counselling (Sarangi and Clarke, 2002). The population described in this study, however, differ by virtue of the different patterns of heritability, notably the autosomal recessive inheritance of the conditions. Thus, there is limited literature which is directly relevant for families or clinicians to draw upon in navigating the difficult terrain of family planning in current and successive generations.

Siblings appeared to have a reasonable amount of knowledge about the ill child's condition, appropriate to their age (Williams et al., 2002). Siblings demonstrated that they are keenly aware of their brother/sister's symptoms. Many spoke about the caregiving tasks which they undertake routinely, supporting their parents in a range of tasks which impact on their own engagement in activities, whether or not the term young carer would be appropriate (Thomas et al., 2003). However, some reported isolation, and that they would learn about the condition/symptoms from listening in on adult conversations. Interventions in paediatric oncology settings have suggested that the development of health information resources (such as interactive DVDs) can be useful in increasing feelings of control (Dragone et al., 2002).

Many reported worry, sadness and anxiety, clearly demonstrated in the literature (Hutson and Barlow, 2007; Sharpe and Rossiter, 2002) regarding typical internalised behaviours for siblings of children with life-limiting conditions. While siblings often spoke of a friend who was a key source of support to them, many also indicated a sense of isolation (Fleitas, 2000; Stallard et al., 1997) indicating ongoing support needs for the brothers and sisters of those with life-limiting conditions.

Isolation also emerged from the sibling data as a component of their experience. Organisations such as ACT, Batten Disease Family Association and the MPS society provide valuable resources for parents to support the siblings of children with life-limiting conditions. Some specialist palliative care

centres and charitable organisations, such as SIBS, offer a range of support services specifically for siblings. The fundamental challenge in supporting siblings of children with a rare life-limiting condition is the availability, accessibility and continuity of services available to them. Siblings' isolation may also be intensified as they keep worries to themselves, to prevent additional burden on their parents (Batte et al., 2006; Stewart et al., 1992).

Limitations

The national survey in Phase 1 had a short turn-around time which may have impacted on the size and heterogeneity of the sample.

Due to the constraints of numbers of children affected by these rare life-limiting conditions, theoretical sampling, as described by Glaser and Strauss, was not possible. That is, specific recruitment of children at a particular stage of the condition was not feasible. Linked to this, staff at collaborating hospices often chose not to approach families where the child was at the end of life, resulting in symptom data and family experiences which have less focus on this period.

Similarly, the recruitment strategy for practitioners resulted in a partial sample. Asking families to nominate someone who could reflect on the experience of supporting them and their child will have influenced the accounts from practitioners. For example, the importance of the relationship in how practitioners spoke of families and their role may be an artefact of the sampling strategy. That is, asking families to nominate a practitioner for the interview led to interviews with those who are particularly personable and relate well to the families.

It is important to note that parents may have reported symptoms differently in the diary. That is, one parent's understanding of 'severity' may be different to another's. One such example is that for one parent severity may indicate level of impairment, while for another it may be a proxy for distress. Future studies may wish to focus on distress as a specific variable.

Moreover, some of the additional symptoms listed by families could be interpreted by practitioners or clinicians as being related to the disease process or to other documented symptoms. However, the language and descriptions noted in this report are those used by families. For example, two families listed 'inappropriate laughing' as additional symptoms. This 'inappropriate laughing' could be related to seizure behaviours, but it is not possible to be certain.

Summary

When a child's life-limiting condition is rare and its trajectory is unknown, families tread unfamiliar and unpredictable paths. This study has documented for the first time the symptom profile and family experiences of supporting a child with MPS disease and Batten disease. The study has foregrounded the symptoms and experiences of concern to parents, and in doing so has focused on behavioural symptoms and the impact on the wider family system. The experiences reported here are likely to be of help to families who are newly immersing themselves in supporting a child with one of these conditions, and provide guidance on suitable interventions to support the family and professional networks. There is clear benefit to researching paediatric palliative care populations, and the following recommendations suggest a number of fruitful paths for future studies and interventions.

10 Recommendations

Symptoms

The study's findings indicate a need for paediatric palliative care services to adopt a broader paradigm than the biomedical model. Previous research has failed to recognise the challenges families face in addressing behavioural symptoms, and consequently, this has led to a vacuum of enhanced supports for families. The data suggest a range of recommendations that would be fruitful in developing practice.

Recommendation 1: Practitioners working in specialist paediatric palliative care, generalist palliative care and generic services would benefit from developing more of an awareness of the importance of behavioural symptoms on the overall experience and challenges of having a child with MPS disease or Batten disease. Specific training should be rolled out to these clinicians, with emphasis on the severity, frequency, management and family impact.

Recommendation 2: Sharing symptom assessment and management strategies between families and practitioners would be helpful. This could sit alongside and complement biomedical approaches described in the Rainbow Symptom Management Guide. Critically, assessment and management strategies would reach beyond physical symptoms, such as pain and seizures, to include behavioural concerns, such as agitation, hyperactivity and sleep disturbances. Outcome measures should be developed and utilised to determine effectiveness of specific symptom management approaches.

Learning from each other

Practitioners often feel isolated, and due to the rarity of the conditions there were infrequent opportunities to meet with others who are also supporting families affected by MPS disease or Batten disease. Isolation was a factor for a broad range of practitioners, such as care workers, educationalists, hospice staff and allied health professionals. While some networks already exist, for example, email discussion lists, these are not routinely used by the broad range of practitioners caring for these children and families.

Recommendation 3: Develop opportunities for practitioners to learn from each other and augment their expertise and exposure to supporting families with these conditions. Technology should be used to implement this recommendation, for example, webinars (web-based seminars), to enable practitioners across the UK to communicate with each other. Such seminars could contribute to continuing professional development for staff. Practitioner networks could be coordinated by a children's hospice, a children's palliative care charity or a condition-specific charity, and would complement the written information available through publications such as Linchpin.

Some practitioners and many families had developed expertise which could be harnessed in supporting other clinicians and newly diagnosed families.

Recommendation 4: A sequence of case studies could be developed from the data presented in this report, which would help share the experiences of families with healthcare practitioners, such as paediatricians and hospice workers. The aim of these case studies would be to support practitioners to develop a rounded understanding of the experience of supporting a child with a rare life-limiting condition. Consequently, the case studies would include focus on family impact and behavioural (as well as physical) symptoms.

Opportunities for training, led by family members, would be valuable for practitioners in a range of settings, for example special schools, hospices and community allied-health professional teams.

Recommendation 5: A series of workshops, convened regionally across the UK, of family members and practitioners could provide a forum for generating training materials and practice-based action plans for improving supports to families. Action-plans and training materials would address both symptom management and family experience. This work would ideally be led by one coordinating organisation.

Families find the time before diagnosis very difficult and wish for a more rapid accurate differential diagnosis. Many specialities are likely to be involved in the child's care in diagnosing and treating specific symptoms (such as ENT clinicians, and autism specialists).

Recommendation 6: Practitioners involved in routine developmental monitoring (such as health visitors and GPs) and clinicians in symptom-specific specialist areas could be targeted with information about the conditions, and encouraged to consider whether the condition might be manifest in new patients. This approach to targeted information distribution takes cognisance of the rarity of conditions and the multiple healthcare practitioners with whom families are in touch before securing the final diagnosis.

The findings from this study add significantly to the evidence base regarding the symptom profile and family experience of supporting a child with MPS disease and Batten disease.

Recommendation 7: A standardised presentation of the study could be developed which outlines the key findings and recommendations for improving practice. This could then be presented by members of this project team and steering group to a range of UK-wide networks including, but not limited to, families, paediatric palliative care clinicians, paediatricians, educationalists in special schools and condition-specific charities.

End-of-life care planning

There are opportunities for improving practice around planning for the child's deterioration, and in particular, advance care planning. The study identified difficulties in some couples in treatment decision-making which exacerbated pre-existing relational difficulties. Combining this with practitioners' struggles to raise the topic means that advance care planning is likely to be sub-optimal, and undertaking conversations about death, dying and bereavement continues to prove problematic.

Understandings of advance care planning, in its widest context (moving beyond viewing this as protocolised care), is necessary.

Recommendation 8: Training should be developed which is cognisant of the relational and emotional context in which practitioners will lead ACP conversations. This will complement, rather than replicate, the protocol-based approaches to supporting practitioners to carry out advance care planning with families.

Training would helpfully focus on the following areas, all of which should be evaluated for outcomes and impact on adjustment in the lead up to and following bereavement:

- Talking with families about advance care planning (focusing on practitioner responsibility for leading conversations).
- Managing emotional dilemmas (for example, the tension between active care in the face of palliative needs).

- Education for practitioners who are not palliative care specialists on the role they have in supporting families in talking about advance care plans.

Recommendation 9: Training programmes should have their efficacy evaluated, with outcomes focused on communicating with parents about decline, dying and bereavement, and professional responsibilities.

Recommendation 10: Research should look at bereavement outcomes for families comparing those who have been supported by staff with the training described above and those who have not. Outcomes could also be assessed related to staff-reported self-efficacy. For example, hospices operating across more than one site could trial training in one location and location outcomes between sites.

Supporting families

The impact on families and wider relationships was stark. The data suggest a number of recommendations to shore up support for families supporting children with these rare life-limiting conditions. Supportive interventions should be available throughout the child's illness, ensuring that families are offered input at times of crisis, for example, during the multiple losses that families experience as the child's condition progresses, and during bereavement.

Recommendation 11: Proven interventions, such as systemic family therapy, should be offered to families with the aim of supporting the couple-relationship, wider family relationships, and enhancing communication. Families should be screened (using tools such as the family relations index) to identify those most likely to benefit from such interventions.

Recommendation 12: Outcome measures should be used to determine the effectiveness of interventions, for example, establishing the impact on parenting self-efficacy (Guimond et al., 2008), or other markers of psychological wellbeing, such as anxiety, depression and relationship satisfaction.

Recommendation 13: Prospective data of bereavement outcomes for families should be routinely collected. These measures would map psychological morbidity. The data would increase the evidence base regarding the longer term sequelae of supporting a child with a rare life-limiting condition. It would also guide the provision of interventions.

The genetic basis of the condition and implications for passing on the gene was a concern for many families, both in the present day and also in thinking forward to future generations within the family.

Recommendation 14: Further research should focus on decision-making regarding family planning, management of the condition across successive generations for those who carry the gene and communication within families regarding the heritability of conditions.

Siblings would benefit from regular and ongoing opportunities to share feelings, concerns and reduce anxiety. Connecting siblings with each other will enable them to share their condition-specific understandings, recognising the knowledge which many had built up. Thus, buddying has a stronger potential than other approaches (e.g. mentoring) since it recognises the level of knowledge that siblings develop over the course of years.

Recommendation 15: Sibling buddying could be implemented through internet, phone, texting or face-to-face methods, each allowing for different levels of contact, and taking into account the geographical challenges presented by these rare conditions. These virtual/distant methods will augment (rather than replace) the annual get-togethers and more formal supports that hospices or condition-specific charities organise. Buddying can

add to other e-supports, such as www.sibs.org.uk, by supporting the development of new relationships between people who have experience of similar conditions.

Psycho-education and psycho-social approaches have proven positive outcomes for siblings.

Recommendation 16: Proven interventions should be offered to siblings. Further work should be conducted to establish those families who are most likely to reap the greatest benefits from such interventions, for example, families where children internalise their emotions, or are isolated geographically, living in remote/rural areas.

Recommendation 17: Siblings should also be routinely signposted to existing resources to help combat isolation. This may include, but not be restricted to, information sheets by ACT (www.act.org.uk). Further, new resources could be developed using media familiar to children and young people, such as the development of DVDs drawing on a psycho-educational format to address concerns such as internalised worry, isolation and bullying.

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12 Appendices

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Appendix 1: Symptom diary example



Children's Hospice Association Scotland
Sharing the Caring

THE SYMPTOM DIARY



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How to fill in your diary

- Part A focuses on some of the symptoms associated with MPS disease and Batten disease. Part B is a weekly reflection on how the symptoms impact on your child's usual activities and the impact these symptoms have on your family life. We want to learn about all your child's symptoms and health behaviours.
- It would be helpful if you try to complete the diary around the same time each day. If you are not able, or forget, to complete the diary one day, don't worry – just try to fill it in as soon as you can.
- Please don't worry about spelling, grammar or 'best' handwriting but try to write as clearly as you can, using a pen.
- Please remember to fill in the date, time and your name on each diary entry in the space provided.
- There is a space for comments at the end of each diary entry so please use this space if there is anything you would like to explain in greater detail. This space will also be useful to write any questions you may have for the researcher.
- Remember to return the completed diaries in the prepaid addressed envelope every two weeks.
- Please describe your child's symptoms in your own words or as you usually describe them. You do not need to use medical terms or jargon.
- If you have any questions or would like further help completing the diary, please telephone 📞 Cari, Gillian, Sally or Liz on 01786 849260. If they are not available, please leave a message and they will return your call as soon as possible.

Please tell us as much as you are able – no matter how minor it may seem. We would rather have too much information than too little!

DAY 1

Date _____ Time _____

Name of person completing the diary _____

PART A: Please tell us about the symptoms your child experienced today. There is space at the end of this section to add any additional symptoms your child experienced which are not described below.

Symptom	Frequency	Severity	Management	Effectiveness
	How often did the symptom occur? 0=Not at all 1= A little of the time 2=Some of the time 3=Most of the time 4=All of the time	How severe was the symptom? 0=Not at all 1=A little 2=Moderate 3=Severe 4=Very severe	What did you do to try to relieve the symptom?	How well did your actions work in relieving the symptom? 0=Not at all 1=A little bit 2=Moderately well 3=Quite well 4=Very well
Seizures	0 1 2 3 4	0 1 2 3 4		0 1 2 3 4
Pain <i>What do you think the pain was related to?</i>	0 1 2 3 4	0 1 2 3 4		0 1 2 3 4
Cold hands and feet	0 1 2 3 4	0 1 2 3 4		0 1 2 3 4
Joint stiffness	0 1 2 3 4	0 1 2 3 4		0 1 2 3 4
Muscle spasms	0 1 2 3 4	0 1 2 3 4		0 1 2 3 4
Disturbed sleep	0 1 2 3 4	0 1 2 3 4		0 1 2 3 4
Agitation/Distress	0 1 2 3 4	0 1 2 3 4		0 1 2 3 4
Hyperactivity	0 1 2 3 4	0 1 2 3 4		0 1 2 3 4
Aggressive behaviour	0 1 2 3 4	0 1 2 3 4		0 1 2 3 4
Repetitive behaviours(chewing/rocking)	0 1 2 3 4	0 1 2 3 4		0 1 2 3 4
Secretions/Drooling	0 1 2 3 4	0 1 2 3 4		0 1 2 3 4

Symptom	Frequency	Severity	Management	Effectiveness
	<i>How often did the symptom occur?</i> 0=Not at all 1= A little of the time 2=Some of the time 3=Most of the time 4=All of the time	<i>How severe was the symptom?</i> 0=Not at all 1=A little 2=Moderate 3=Severe 4=Very severe	<i>What did you do to try to relieve the symptom?</i>	<i>How well did your actions work in relieving the symptom?</i> 0=Not at all 1=A little bit 2=Moderately well 3=Quite well 4=Very well
Other Symptom: -----	0 1 2 3 4	0 1 2 3 4		0 1 2 3 4
Other Symptom: -----	0 1 2 3 4	0 1 2 3 4		0 1 2 3 4
Other Symptom: -----	0 1 2 3 4	0 1 2 3 4		0 1 2 3 4
Other Symptom: -----	0 1 2 3 4	0 1 2 3 4		0 1 2 3 4
Comments:				

Part B: Complete this section **once** at the end of each week.

B1. Please tell us about your child’s general activities this **past week** by circling one of the numbers in each row that fits best. For each activity, we are interested in finding out how this past week has been in comparison to what you would describe as your **child’s average or usual pattern**. If you are unable to assess any of the activities please circle N/A at the end of each row and tell us why.

	A lot worse	A little worse	Much the same/Usual	A little better	A lot better	*Not able to assess
Sleep Pattern	0	1	2	3	4	N/A
Nutrition (eating and drinking or being fed)	0	1	2	3	4	N/A
Behaviour	0	1	2	3	4	N/A
Mobility (moving about; changing positions or being transferred to a different position)	0	1	2	3	4	N/A
Communication						
i) Your child was able to understand you and others	0	1	2	3	4	N/A
ii) You and others were able to understand your child	0	1	2	3	4	N/A
Taking part in play and other social activities	0	1	2	3	4	N/A
Other? (Please add)	0	1	2	3	4	N/A
Other? (Please add)	0	1	2	3	4	N/A

Note: If you are unable to assess any of the above activities please tell us why?

B2. Did you contact anyone for advice or assistance regarding any of the symptoms your child experienced in the past week? Yes No

If yes, can you tell us who you contacted and why?

B3. Please tell us if any of the symptoms your child experienced this past week stopped your child or any member of your family from doing something you would usually do? (example - go to school, go to work, go shopping, visit family/friends etc)

B4. Please tell us in what ways your child's health and symptoms over the past week has affected you and other members of your family. The researcher might ask you about this when you see them. If you need more space please continue on a spare sheet.

B5. If there is anything else you would like to tell us about your child this week, please do so. If you need more space please continue on a spare sheet

Appendix 2: Symptom diary participant characteristics (n=22)

Child	Gender	Age (years)	Age at diagnosis (years)	Life-limiting condition
1	Female	15.0	8.0	Sanfilippo disease Type A
2	Male	14.0	4.0	Sanfilippo disease Type A
3	Female	13.0	1.3	Sanfilippo disease Type A
4	Female	8.0	3.0	Sanfilippo disease Type A
5	Female	13.0	3.5	Sanfilippo disease Type A
6	Male	16.0	2.0	Sanfilippo disease Type B
7	Female	9.0	4.0	Sanfilippo disease Type A
8*	Male	7.0	3.5	Sanfilippo disease Type A
9*	Male	7.0	3.5	Sanfilippo disease Type A
10	Female	21.0	6.0	Sanfilippo disease Type A
11	Male	5.0	1.5	Sanfilippo disease Type B
12	Male	7.0	7.0	Sanfilippo disease Type C
13	Male	12.0	2.5	Sanfilippo disease Type A
14	Male	8.0	0.5	Morquio disease
15*	Male	4.5	3.0	Batten disease (Infantile)
16*	Male	4.5	3.0	Batten disease (Infantile)
17	Female	9.0	6.0	Batten disease (Late Infantile)
18	Female	3.5	1.5	Batten disease (Infantile)
19	Male	8.0	5.0	Batten disease (Late Infantile)
20	Female	5.0	0.5	Batten disease (Infantile)
21	Female	12.0	1.0	Batten disease (Infantile)
22	Male	2.5	1.5	Batten disease (Infantile)

Notes:

- The numbers in the column entitled 'child' represent individual cases. Those highlighted with an * indicate a family where there are two siblings affected with the condition.
- Numerical identifiers used in this table do not correspond with identifiers with quotations, to preserve the anonymity of respondents.

Appendix 3: Symptom diary completion details for study participants (n=22)

Child	Life-limiting condition	Symptom diary days N (%)	Reason for missing data	Diary completer
1	Sanfilippo disease	56/56 (100)	N/A	Mother
2	Sanfilippo disease	53/56 (95)	Reason not given	Mother
3*	Batten disease	56/56 (100)	N/A	Mother
4*	Batten disease	56/56 (100)	N/A	Mother
5	Sanfilippo disease	55/56 (98)	Family social outing	Mother
6	Batten disease	56/56 (100)	N/A	Father
7	Batten disease	56/56 (100)	N/A	Mother
8	Batten disease	56/56 (100)	N/A	Mother
9	Batten disease	56/56 (100)	N/A	Father
10	Sanfilippo disease	49/56 (88)	Child in planned respite/hospice care	Father
11	Sanfilippo disease	46/56 (82)	Child in planned respite/hospice care	Mother
12	Morquio disease	56/56 (100)	N/A	Father
13	Sanfilippo disease	49/56 (88)	Reason not given	Mother
14	Batten disease	52/56 (93)	Child in planned respite/hospice care	Mother
15	Sanfilippo disease	56/56 (100)	N/A	Mother
16*	Sanfilippo disease	50/56 (89)	Child in planned respite/hospice care	Father
17*	Sanfilippo disease	50/56 (89)	Child in planned respite/hospice care	Father
18	Sanfilippo disease	56/56 (100)	N/A	Mother
19	Sanfilippo disease	32/56 (57)	Diaries not received	Mother
20	Sanfilippo disease	53/56 (95)	Child in planned respite/hospice care	Mother
21	Sanfilippo disease	35/56 (63)	Diaries not returned due to demands on mother	Mother
22	Batten disease	56/56 (100)	N/A	Mother

Notes:

- The numbers in the column entitled 'Child' represent individual cases. Those highlighted with an * indicate a family where there are two siblings affected with the condition.
- Numerical identifiers used in this table do not correspond with identifiers with quotations, to preserve the anonymity of respondents.

Appendix 4: Practitioner roles and parents' rationale for inviting them to participate

Practitioner identifier	Profession/ work setting	Reason for being nominated	Practitioner's previous experiences of supporting families with this condition
1	Educationalist	<i>We work very closely with them, I have done for a year...and the school that I work in have a policy of working very, very closely with parents anyway. And it's just the way that...it's...it's the practice of the organisation that I work for ... generally we take children with autism ... and we're not a state school so we are very autism specific ... we share everything with the parents, whether it's good or bad, we don't just tell them we've have a good day, if things have been not so good. And right at the beginning, you know, I would say to [parents] that I'm going to be really honest with you.</i>	<i>This is the first experience. Yeah, this is the only one. So none really.</i>
2	Paediatric palliative care consultant	<i>I've stayed in touch with them, I'm the one who they have contacted for all the inter-current illnesses so probably I've had the longest contact with the family.</i>	<i>This is my first experience of Battens [disease][...] I was working in a metabolic unit where there were Battens who came through but I was not responsible for them. You know I dealt with them, got in, got to know them when they were on the ward but not over a long period of... this is the first family.</i>
3	Carer	<i>I've been [child's] key worker for quite a number of years; also I've been at the hospice since it opened which is 21 years. So we've had, I've had a number of children come through here with MPS conditions. So I think [family], appreciated the fact that I'd seen the condition before and that I could talk from experience, although every child's different [...] I think she thinks I've got sort of that empathy that I've been there and she's got confidence in what I say [...] I hope I do best practice in the respect that I treat [child] as an individual. And the family as an individual, but I have got a lot of hospice experience.</i>	<i>I've had a number of children come through here with MPS conditions. Well we've had obviously through some with Hunter's, Hurler's and Sanfilippo</i>

Practitioner identifier	Profession/ work setting	Reason for being nominated	Practitioner's previous experiences of supporting families with this condition
4	Palliative Care Nurse	<i>I've actually known the family for quite a long time. [Child]'s been coming to us since 1999 so he was five and a half then. I think I've got a really good relationship with the family; I used to be a key-worker for them until [child] went on to a teenage team and they trust me, they know that I'm open, honest with them, and, you know, that we all care for [child] in his best interest and with the needs that they've got for him as well.</i>	<i>A handful [...] we have had quite a few children with Sanfilippo [...] so quite a lot of experience.</i>
5	Community children's nurse	<i>I think it's probably I'm that child's care coordinator. And I think that the mum feels quite comfortable talking to me and she probably knows that I'm fully aware of the problems that they face day to day...caring for their child, so...felt that I'd be able to sort of perhaps express their feelings as well as my own.</i>	<i>Very limited. I have the one child with Sanfilippo that I've known for the last seven years.</i>
6	Educationalist	<i>I think [family] would have nominated us because we have such a close relationship. [laughs]. You know, we care, you know for [child] and we know [child] in exactly the same way that she does. Now obviously, she's his mother so her emotional attachment to [child] quite different from ours. But I do think that we know [child] very, very well and every little movement and noise, that you know, we know what that means...as much as [Mum] does. And also, I think I mean I do think that she understands that we kind of share her pain as well, you know because we are so emotionally involved at a different level from her[...]I think she just feels comfortable with us, I think that's probably it in a nutshell, she just feels comfortable with us.</i>	<i>Two other children with Batten disease [...] I've worked with kids who've had Battens, different kinds of Battens</i>

Practitioner identifier	Profession/ work setting	Reason for being nominated	Practitioner's previous experiences of supporting families with this condition
7	Educationalist	<i>We care for the child, when he's in, for about six hours a day. It's quite a long time, I think they trust...must trust us and we have, or I have, quite you know, a lot of experience with children [...] we have experience looking after children who have got life-limiting conditions.</i>	<i>Not Batten disease, no. But we have experience looking after children who have got life-limiting conditions.</i>
8	Paediatric palliative care consultant	<i>I think I sort of support them. I do home visits and I just spend time with them listening to their concerns, taking their concerns on-board and doing really [...] a little bit of advocacy for them with professionals I think.</i>	<i>I can think maybe one other child that I see as frequently [child] would be the one over the years [...] My other kids with MPS, we have one at the minute 'cause we've had several who've died in the last two or three years.</i>
9	Paediatric palliative care nurse	<i>I was the first person that sort of introduced them to [hospice] [...] and I think that has a you know, a big bearing on how parents sort of cope really [...] then, kind of listen to what the family want and do things the way the family want me to do them, you know I'd like to think basically that they do trust me, they leave [child] now...I mean [Mum] found it really hard obviously at first, couldn't leave [child] at all but, you know, they'll leave [child] in our care now [...] but I think, I mean they're a family that bond and get on well with everybody. But it's just that you know, I'm sort of contact for them. I do...I think they trust us now really.</i>	<i>I worked as a school nurse prior to working at the hospice and, you know, I've worked with children with obviously Battens, I've got on my contact list I've got children with...a child with juvenile Batten's.[...] I've worked with children with Hunter's, I've got a young lady, a 16 year old, with Hurler's at the school I worked with, and Batten's as well. So yes I've had you know, a fair bit of experience.</i>
10	Community children's nurse	<i>I've worked with [family] for quite some years and they [we] have a lovely relationship, a really, a real lovely relationship and we see each other a lot.</i>	<i>I have [supported] others, another children who is in a different stage of it [MPS]</i>
11	Consultant Paediatrician	<i>I'm not sure it's a matter of what I do well, it's just that I was the lead is [...] in her diagnosis. And holding the reins for her considerably complex treatments, so I'm probably the person that she's...and the family have come to know best I would guess.</i>	<i>It's a reasonably big part of my job. It's not a terribly common disorder [I have supported] about 10.</i>

Practitioner identifier	Profession/ work setting	Reason for being nominated	Practitioner's previous experiences of supporting families with this condition
12	Paediatric palliative care	<i>My understanding is because of the support that I've been giving the family, they felt after speaking with you, that I would be a good person to speak to in terms of supporting individuals, advocating, on behalf of them and their...some of the complexities of the issues.</i>	<i>Well we...we sort of support them from all areas, you know, they usually come to us, from diagnosis so it's taking that initial call you know supporting them, making sure that they're in touch with their specialist centres.</i>
13	Carer	<i>Longest carer who has been working with the family and knows them very well.</i>	<i>None</i>
14	Physio-therapist	<i>I think because it work closely with family, you kind of get an overall picture, everything that's going on with them really, try and help them in all areas whether it be the child's physical ability but also the social needs as well [...] Making sure that they've got the right support, you know for education at home.</i>	<i>We seem to have quite a few ... on our case-loads compared to I think the general, well the UK population, there seems to be quite a lot in, in this area. I've got several children, some younger, some older, or in mainstream school, there's two actually in the special school that I'm in at the moment [...] and I offer support to at least three others in mainstream schools.</i>
15	Children's palliative care nurse	<i>[Practitioner has been involved with the family] about seven years and our organisation's been involved for longer than that [...] so I would think probably because that, I'm sort of like the most consistent person who goes into the house, probably [...] just because, well, just the professional support isn't it? And just being involved in her care and everything, and experience of the condition, you know, with other families, and just probably 'cause we're available 24 hours, seven days a week. Not me personally but you know, my team. So, maybe because of that, I don't know.</i>	<i>At the moment we've only actually got two children on the books with the condition, but I've been involved with the end-of-life care and sort of quite a long period of care before end-of-life care with two other families. So yeah, so quite a lot of experience I suppose which is not, you know, it's quite a rare disease still, isn't it?</i>
16	Carer	<i>Over the years we have formed a very close bond, like the child is just like part of our family now, so we are very close. Well I would say that, you know, I treat their child the same as I treat me own, you know, and care for her to the best of my ability.</i>	<i>Practitioner: I have only, I only have dealt with [child], and then I would be in a classroom with them in a different setting you know, at school. Interviewer: So is it just [child] that you have come across that has Sanfilippo then? Practitioner: No the other two boys in school have it.</i>

Practitioner identifier	Profession/ work setting	Reason for being nominated	Practitioner's previous experiences of supporting families with this condition
17	Physio-therapist	<i>I think because ...I've always made myself very available to them, so that, you know, if situations arise they know that they can get in touch quite quickly. I don't give many patients' families my work mobile number... you know, unless I feel that there's a need, so they...from the start they've had, my work mobile and been able to contact me, even if it's just with a question, or you know, something, and I think because I'm quite happy to come to other appointments with them to sort of advocate a certain about when we're looking for equipment or wheelchairs or things that I make time to come to all these appointments with them to make sure that their child is getting what they need.</i>	<i>Interviewer: You've mentioned that this is the only family you [work with]? Practitioner: At the moment that I have, yes. I had one quite a number of years ago in the past but because it's not a very common condition. It's not, but so I can really only speak of...the present family that I've been working with.</i>
18	Carer	<i>I have known the family for years and have good communication skills with [child].</i>	<i>None</i>
19	Children's palliative care nurse	<i>I've worked at the house for, [hospice] now for 18 years [...] So, whether it's just that, experience-wise there's probably MPS and other conditions but when we first have a family to the house, we sort of put them aside, two contacts, so that the family finds it easier to relate to, two people in general, 'cause otherwise they're looking at a team of perhaps 50 people [...] probably caring, trusting, and a knowledge about MPS children but particularly Sanfilippo.</i>	<i>Practitioner: A knowledge about MPS children, but particularly Sanfilippo. Interviewer: Particularly Sanfilippo. Practitioner: Yeah. Interviewer: So you have experience of other MPS conditions? Practitioner: Yes we do.</i>

Appendix 5: Members of the project steering group

Name	Affiliation
Dr Bryony Beresford	Senior researcher, University of York
Linda Buchanan	CNS pain management, Royal Hospital for Sick Children, Edinburgh
Francis Edwards	Palliative care nurse consultant for Scotland
Libby Gold	Children's Hospice Association Scotland
Anne Harris	Rainbow Trust
Dr Jeremy Keen	Consultant Physician in Palliative Care, Highland Hospice
Katrina McNamara-Goodger	ACT, Head of Policy and Practice
Zoe Picton-Howell	Parent
Karen Sinclair	Palliative care nurse consultant for Scotland
Carrie Upton	Chaplain, Royal Hospital for Sick Children, Edinburgh

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