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

Many non-cancerous life-limiting conditions are individually extremely rare. Little is known, even by professionals in the field, about the actual day-to-day presentation of symptoms or the impact of these symptoms on the child and family. This study aimed to use the expert views of families and practitioners to gain an understanding of the symptom experience of children with mucopolysaccharidosis disease (MPS) and Batten disease.

Key Findings

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

Background

Symptoms in rare life-limiting conditions: A wide range of symptoms are known to affect children with rare life-limiting conditions, yet characteristics such as the onset, timing, patterns and severity of such symptoms remain largely unknown (Siden et al., 2010). 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).

Family experiences of supporting a child with a life-limiting condition: Symptoms have an effect on both the individual with the condition and the wider family networks (Hunt et al., 2003). 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. Family-based interventions have been successfully used in palliative care settings recognising the need to intervene at the level of family rather than individual members (Kissane and Bloch, 2002; Mehta et al., 2009).

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. Despite these being difficult conversations to engage in, parents appreciate open and honest communication (Fraser et al., 2010).

Mucopolysaccharidosis and Batten disease: MPS and Batten disease are rare life-limiting conditions which are devastating and progressive in nature, with no curative treatments available. The conditions lead to death in childhood, usually during the second decade of life. They are considered 'storage diseases', because certain molecules made by the body begin to accumulate or store themselves in the brain and nerves. The consequence is that children begin to show behavioural disturbances such as hyperactivity, aggressive behaviour and sleep disturbance. Children with MPS will become immobile and unresponsive towards the end of life, as nerve damage becomes more severe the child. Seizures, caused by nerve damage, are common but not universal. Children with Batten disease will experience seizures or visual disturbance early on, and show cognitive changes such as slow learning, or even loss, of new skills, behaviour changes, clumsiness and repetitive speech.

By understanding how to assess and manage these complex symptoms, and the experience of supporting the child, services will be in a better position to support these families.

Methods

The study used three methods for data collection:

(i) A national survey. This was used to identify which rare life-limiting conditions the study would focus on. Forty-three practitioners and 16 family members participated in the national survey.

(ii) A daily symptom diary was kept over a two-month period. Completed symptom diaries were received for 22 children, totalling 1150 diary entries. This represents a completion rate of 93%.

(iii) Interviews with parents, siblings and practitioners. Parent/s of 26 children were interviewed. Eight siblings and 19 practitioners also participated in interviews.

Ethical approval was granted by NHS Forth Valley ethics committee, NHS R&D offices and individual hospices.

Findings

The findings are presented in relation to reporting the symptom profile of MPS disease and Batten disease. Second, the accounts of families (parents and siblings) are presented, before the final section which outlines the experiences of practitioners.

Symptom profile

Sanfilippo disease: The children who took part in this study were at different stages of the condition. A wide range of symptoms were recorded by their parents in the symptom diaries. In particular, behavioural symptoms such as hyperactivity, repetitive behaviours and agitation were documented as occurring most often. These symptoms (and disturbed sleep) were also recorded as being severe. 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 because of communication impairments (such as loss of language or understanding) made it more difficult to assess symptoms. The strategies used 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 occurring infrequently 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 occurred particularly often. 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, those which have the potential to lead to health-related complications and other life-threatening illnesses (such as a severe chest infection). Practitioners found that communication impairments (such as the child's loss of language, understanding or vision) made it more difficult assess symptoms. Practitioners also found it difficult to manage seizures in this population of children.

Family perspectives: The key role of expertise and experiential knowledge

Families spoke at length about their experiences and touched on a wide variety of topics in describing their lives. Figure 1 shows some of the key areas families spoke about, each of which is connected with the development of expertise and experiential knowledge by them.

Parents become experts in these rare life-limiting conditions, and use this expertise to manage their role as a caregiver. Between first noticing symptoms to the point at which a diagnosis is made, 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. However, following diagnosis many parents began to feel more reassured that their parenting was not to blame for any ongoing physical or behavioural difficulties.

Without a diagnosis it is a nightmare. (Dad, Family 12)

The development of expertise functions, at times, to isolate the family. Holding expertise within the nuclear family can prevent others from supporting the child or family. A clear cycle is therefore formed, where only those with contact with the child develop the necessary knowledge, and only those with expertise can care for the child. Since these conditions are inherited, families often had experience of children across generations. However, this did not always result in receiving help.

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)

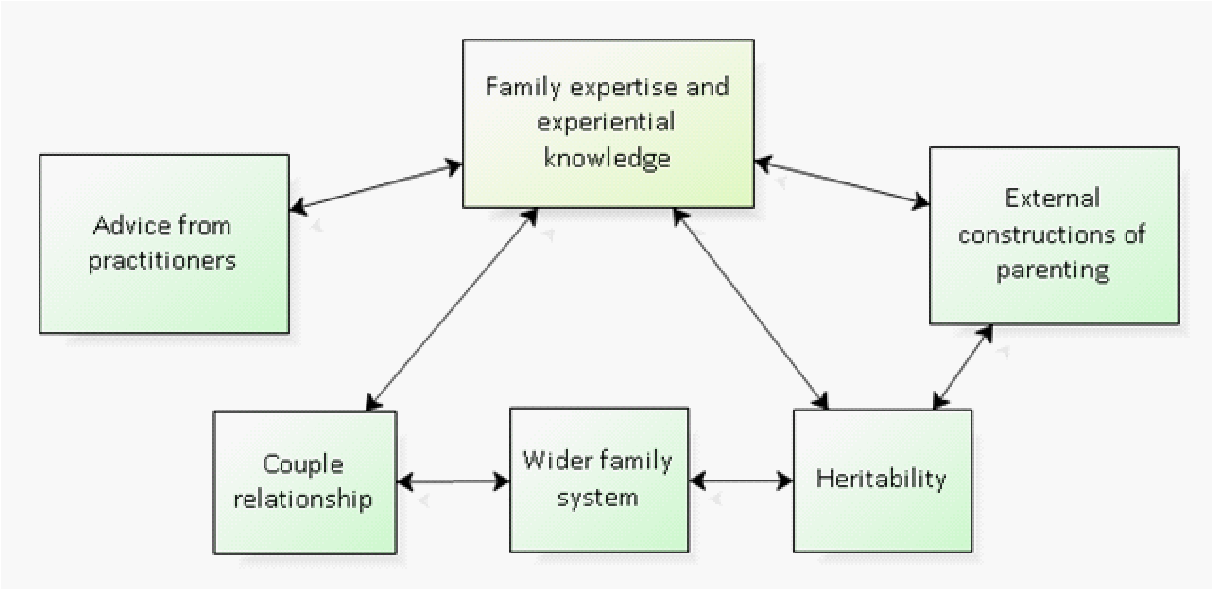

Figure 1. Family expertise and experiential knowledge

Many parents spoke with each other openly, but others did not.

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.
Int: 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. (Family 16)

Parents often felt as though they were on their own in managing symptoms, finding that practitioners turned to them for answers.

There's nobody would know how to manage each symptom like me and [partner] would know [. . .] I might go and say 'I'm at my wit's 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)

Sibling experiences

Siblings spoke with clarity about their ill brother or sister's condition, and summarised complex symptoms 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. Siblings' accounts also 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 combination of people speaking negatively about disability and accessing support through friends and family members.

Each of these issues is shown in figure 2 below.

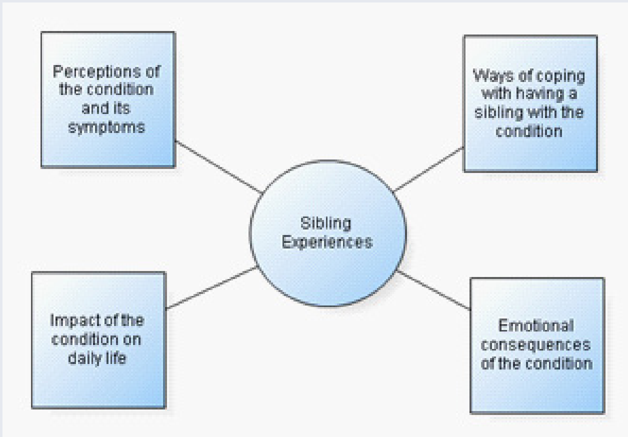

Figure 2. Sibling experiences

Siblings had a great deal of insight into symptoms that their brother or sister experienced. Some spoke of what it was like seeing them unwell.

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

Several siblings stated that they felt they may know more about their sibling's condition than their parents realise. This may be because siblings and parents try and protect each other by limiting how much they talk to each other about the ill child's condition.

Siblings also spoke of feeling isolated, and the limitations in their social experiences which stemmed from the needs of the ill-child.

Siblings were often managing a range of complex emotions, at home and at school and were very aware of how worrying feelings could echo around the wider family system. Two interviewees spoke of being repeatedly teased by peers at school.

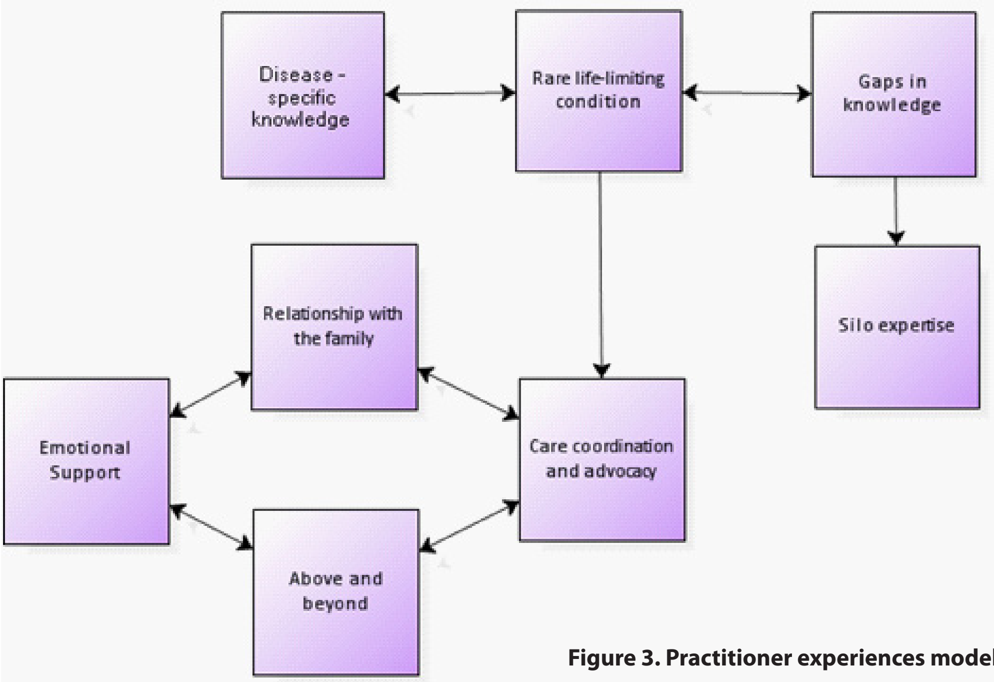

Figure 3. Practitioner experiences model

Practitioner experiences

Families each nominated a practitioner to be interviewed. These practitioners had a wide variety of backgrounds, such as children's palliative care, community children's nurses and education. The interviews showed two core strands, one of which relates to practitioner's overall roles (Figure 3) and the second focuses on advance care planning (Figure 4).

Many practitioners spoke of having expertise in only one or two symptoms, rather than the whole condition. This is named in the model as 'silo' expertise, indicating a narrow range of knowledge.

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)

A consequence of many practitioners having limited condition-specific knowledge was that they often undertook care coordination to ensure the family was well supported. In many cases, practitioners went above and beyond what would be expected of them in their role.

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 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. (Practitioner 24)

Advance Care Planning:

Practitioners were also asked about advance care planning. Figure 4 represents the issues which help and hinder talking about decline and dying.

Many practitioners had limited experience of talking with families about preparing for a child's death. While practitioners knew the importance of talking about advance care planning, many found it hard to admit to themselves that the child was dying, and avoided these conversations.

It's very much gentle talking to them and really knowing the family and knowing where they're at. (Practitioner 26)

The most important feature in helping have such conversations was the relationship between family and professional.

[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)

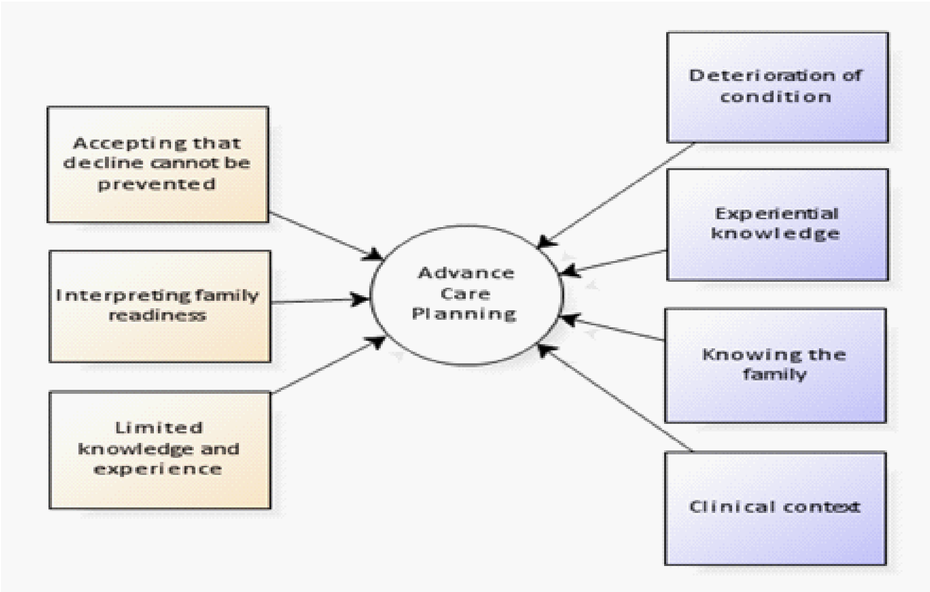

Figure 4. Advance care planning

Training needs were identified by some participants:

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)

Practitioners found it easier to have conversations about the child's deterioration if they knew the family well.

It's very much gentle talking to them and really knowing the family and knowing where they're at. (Practitioner 26)

Some practitioners waited until families spoke about planning for the child's condition to decline before talking with them about end-of-life planning, or waited for a clear sign of decline:

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)

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

The data suggest a range of recommendations that would be fruitful in developing practice:

- 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.
- Sharing symptom assessment and management strategies between families and practitioners would be helpful.
- Further training should be offered to practitioners to support them to have conversations about advance care planning. This training should be evaluated to determine its impact on practitioner self-efficacy and on family members following the death of their child.
- A sequence of case-studies could be developed from the data, which would help share the experiences of families with healthcare practitioners, such as paediatricians and hospice workers.
- Further interventions are required, for example family therapy (to support the couple-relationship, wider family relationships, and enhance communication), and bereavement support.
- Siblings should be offered support, for example, buddying. Siblings should also be routinely signposting to existing resources to help combat isolation.

Project Team

This research was led by Dr Liz Forbat (Senior Research Fellow and Co-Director of the Cancer Care Research Centre). The research team was Dr Cari Malcolm, Gillian Anderson, Sally Adams and grant-holders Professor Faith Gibson, Dr Richard Hain and Anthea Morley.

The project was funded by the Children's Hospice Association Scotland.

This report is distributed by the Cancer Care Research Centre at the University of Stirling.

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