Understanding challenges for ultra-rare lysosomal storage disorders: Patient and caregiver experience of care and support through the disease journey

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Background

Lysosomal storage disorders (LSD) are autosomal recessive lysosomal storage conditions, caused by lysosomal enzyme deficiencies, that are life-limiting and characterised by progressive neurological and cognitive deterioration, including other body systems.[1,2]

The exact prevalence of ultra-rare LSDs is difficult to determine but is estimated at <1 in a million.[3]

Ultra-rare disorders have a profound impact on patients and their caregivers but information about the experience of care and support received by patients and their families during the disease journey is limited.

Objectives

- To gain an **understanding of patient and caregiver experiences** of living with an ultra-rare lysosomal storage disorder
- To gather insights into **how best to support these patients and their families**

Methods

Members of MPS Society UK — Parents/caregivers of members with eligible ultra-rare disorders (Table 1) were invited to take part.

10 — Telephone **semi-structured interviews** were conducted between June–September 2020.

Qualitative data collected included questions about their experience with the **care and support** received since diagnosis.

Thematic analysis — Responses were analysed by applying an inductive thematic-content approach.

Table 1. Eligible ultra-rare disorders

AGU (Aspartylglycosaminuria)
Fucosidosis
Geleophysic dysplasia
GM I Gangliosidosis
LAL-D (Lysosomal acid lipase deficiency)
ML I (Mucolipidosis I; Sialidosis)
ML II (Mucolipidosis II; I-Cell disease)
ML IV (Mucolipidosis IV)
MLD (Metachromatic leukodystrophy)
MPS IX (Natowicz)
MPS VII (Sly)
MPS IV B
MSD (Multiple sulfatase deficiency)
Winchester

Results

Patient demographics

11 Patients

55% female

Born 1992—2017

5 Disorders (Table 2)

7 live patients
Mean (±SD) current age: 14.6 (±9.3) years, range 4.8—28.2)

4 deceased patients
Mean (±SD) age at death: 8.6 (±8.0) years, range 1.8—19.0)

2.7 years Mean (±SD) **age at diagnosis**: 2.7 (±3.7) years, range 0.1—12.5)

Table 2. Ultra-rare disorders of patients included in this study (n=11)

Disorder	Patients
Fucosidosis	4
GM I Gangliosidosis	1
LAL-D (Lysosomal acid lipase deficiency)	2
ML II (Mucolipidosis II; I-Cell disease)	3
MSD (Multiple sulfatase deficiency)	1

Diagnosis experience

Lack of support during diagnosis delivery:

Medical language was not clear
"We did not understand the explanation of the condition"

Lack of empathy and sympathy
"Heartless and cold"

Perceived lack of interest
"Parents hear their child has a life-limiting condition once, doctors say it every day"

Insufficient or conflicting information
"We had to google it"
"I felt lied to"

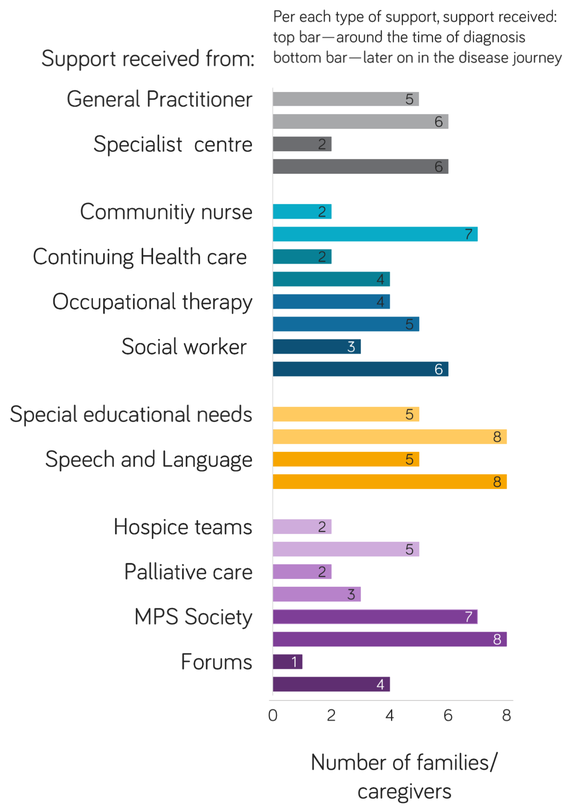
Support families and caregivers would have liked around the time of diagnosis:

- ✓ Diagnosis explained in **lay terms**
- ✓ **Emotional support** during and after receiving the diagnosis
- ✓ Specialist **training** for clinicians to deliver it
- ✓ **Tailored accurate information** on the disease, its progression and research/ trials
- ✓ Information on **sources of support and services**

Disease journey

The type of care and support that was needed changed as the disease progressed (Figure 1).

Figure 1. Number of families/caregivers that reported receiving support by type of care & support service (n=10).



- Caregivers reported that the most support received around the **time of diagnosis** was provided by **General Practitioners** (GP) and the **MPS Society** (Figure 1).

  "Our GP was there for life, he was the focal point"

- The role of **specialist centres** and **community services**, such as social workers and hospice teams, became more prominent as the disease progressed (Figure 1).

  "The specialist was key to keep us sane"

- Caregivers reported receiving **emotional support** from the MPS Society and international forums, later on in the disease journey

  "Without the MPS Society I don't know where we would be today"

Support families and caregivers would have liked throughout their disease journey:

- ✓ **Access to counselling services** throughout the disease journey and from the point of diagnosis
- ✓ **Support with paperwork** to access services, available help and schooling
- ✓ Provision for patients **transitioning into adulthood**

References: 1. Poswar FO, et al. Lysosomal diseases: Overview on current diagnosis and treatment. Genet Mol Biol. 2019; 42:165-177. 2. Giugliani R, et al. Neurological manifestations of lysosomal disorders and emerging therapies targeting the CNS. Lancet Child Adolesc Health. 2018; 2(1):56-68. 3. www.orpha.net, accessed 07.01.21.

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Key finding

- The care and support needed by families and carers of patients with ultra-rare disorders changes during the disease journey

Conclusions

Caregivers of patients with ultra-rare diseases face constant challenges over time.

Providing **care and support tailored to different stages of the disease journey** is needed to improve the quality of life of patients and their families.