Living with Pompe disease in the UK: characterizing the patient journey; burden on physical and emotional quality of life; and impact of COVID-19

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INTRODUCTION

- Pompe disease, a rare autosomal recessive disorder characterized by progressive skeletal and/or respiratory muscle weakness, is a devastating condition for those diagnosed and their families[1]
  - As their condition deteriorates, many people with late-onset Pompe disease (LOPD) will require the use of a wheelchair and/or respiratory support[2,3]
  - Dependency on a wheelchair and respiratory support impacts a person's ability to partake in daily activities and significantly reduces quality of life.[4,5]
- To date, few studies have investigated individuals' experiences and emotions associated with the trajectory from noticing early symptoms to receiving a diagnosis of LOPD and living with the disease.

OBJECTIVES

- To characterize the experiences of people from the UK living with LOPD
  - From first symptoms, through diagnosis and disease progression; mapping people's feelings, attitudes, emotional responses and behaviors at each stage
  - Impact of LOPD on daily activities, and the emotional and physical challenges
  - Impact of COVID-19 on daily life and LOPD care.
- To explore healthcare professionals' (HCPs) perspectives of peoples' experiences of LOPD and the impact of COVID-19.
- Here, we present interim results from interviews with people living with LOPD and qualitative results from interviews with a small sample of HCPs.

METHODS

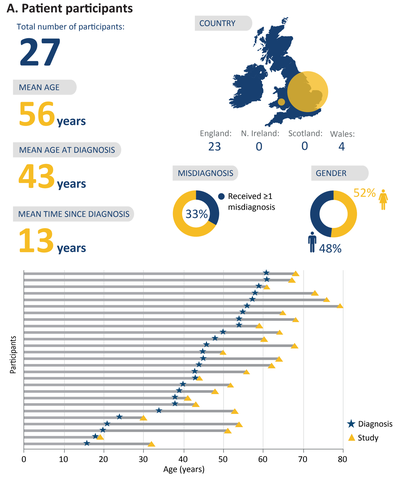
- Data were collected by:
  - In-depth qualitative interviews with people living with LOPD
  - Quantitative survey of people living with LOPD (ongoing, with results to be presented at a future meeting)
  - Qualitative interviews with HCPs.
- Further details on the methodology and interview discussion themes are available in the Supplement, which is accessible via quick response (QR) code.

RESULTS

Interview participants

- The characteristics of patient interview participants are presented in **Figure 1A**.
- The roles and workplace settings of HCP interview participants are presented in **Figure 1B**.

Figure 1. Characteristics of (A) patient, and (B) HCP interview participants

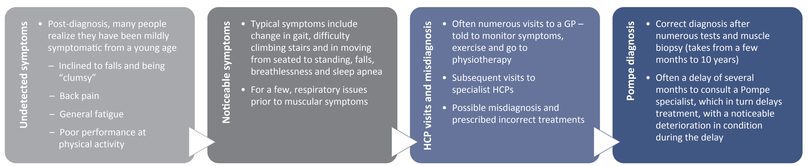


IMD, inherited metabolic disorder; LSD, lysosomal storage disorder.

Disease journey

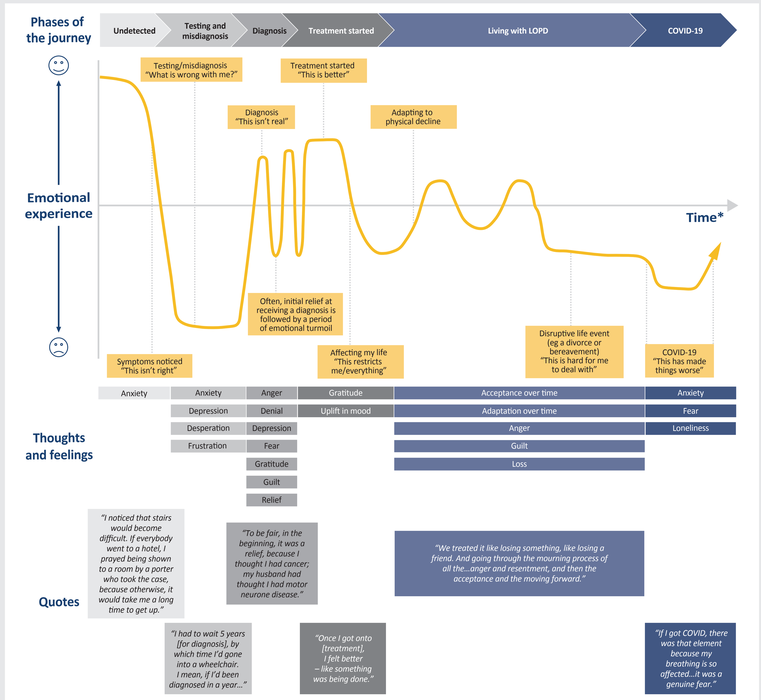
- **Figure 2** depicts some of the emotional aspects of patient participants' diagnostic journeys and of living with LOPD
  - A detailed description of the archetypal emotional journey is available in the Supplement, which is accessible via QR code.
- Typical stages of participants' diagnostic journeys are presented in **Figure 3**.
- A range of factors influenced the journey to participants being diagnosed with LOPD
  - Age at noticeable symptoms and order in which symptoms emerged
  - Time to diagnosis and early interactions with HCPs
  - Age and availability of therapy at diagnosis.

Figure 3. Typical stages of the diagnostic journey



Based on the qualitative interview study. GP, general practitioner.

Figure 2. Schematic of the archetypal emotional journey from initial symptoms to being diagnosed with and living with LOPD

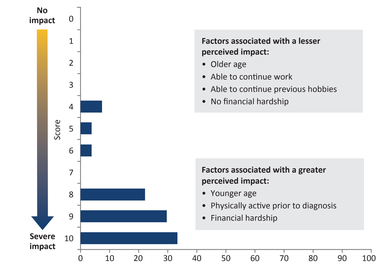


Based on the qualitative interview study. *Non-linear scale.

Challenges of living with LOPD

- Most interview participants faced challenges as their condition deteriorated, with impacts on the following:
  - Lifestyle, daily activities, social life and holidays
  - Ability to continue working
  - Dependency on others, including family members and carers
  - Family relationships.
- In general, interview participants felt that disruptive life events, such as accidents or bereavements, added to the physical and emotional burden.
- Most interview participants felt that LOPD severely affected their lives (**Figure 4**)
  - On a scale of 0–10 (with 0 being no impact at all and 10 being a severe impact), the mean (±standard deviation) score was 8.5 (±1.8).

Figure 4. Perceived impact of LOPD on participants' lives



Based on the qualitative interview study. People were asked to score the impact of Pompe disease on their lives on a scale of 0–10, with 0 being no impact and 10 being an extremely severe impact. Associations were not analyzed.

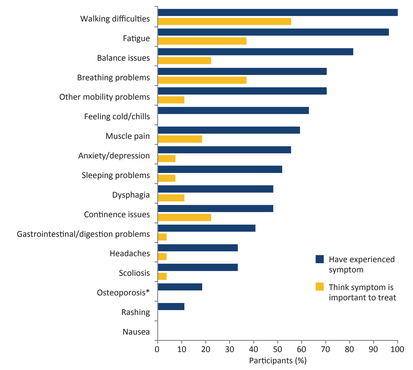
Symptoms

- The most frequently mentioned symptoms associated with LOPD were walking difficulties (reported by n=27 people; 100.0%), fatigue (n=26; 96.3%), balance issues (n=22; 81.5%) and breathing problems (n=19; 70.4%; **Figure 5**).
- Of the Pompe-specific symptoms, walking difficulties (n=15; 55.6%), fatigue (n=10; 37.0%), breathing problems (n=10; 37.0%), balance issues (n=6; 22.2%), continence issues (n=6; 22.2%) and muscle pain (n=5; 18.5%) were reported as the most important to treat (**Figure 5**).

COVID-19

- For most interview participants, the COVID-19 pandemic has been a period of increased anxiety, low mood and physical deterioration
  - Given a vulnerability to respiratory illnesses, the fear of contracting COVID-19 has been a key concern
  - Isolation and reduced contact with family and friends has been difficult
  - Valuable exercise and support facilities were unavailable
  - Reduced availability of in-home care
  - Temporary cessation of enzyme replacement therapy (ERT)
  - Reduced contact with HCPs and fewer assessments (eg lung function tests).

Figure 5. Frequency and importance of treating symptoms



Based on the qualitative interview study. *Comorbidity (and not a symptom).

Interviews with HCPs

Figure 6. Main findings from HCP interviews

Most HCP interview participants have a similar view to patients of the emotional journey and difficulties faced from living with Pompe disease

- Uncertainty and variability in time to diagnosis
- The initial psychological boost of starting ERT treatment followed by the burden of frequent infusions and gradual physical decline
- Hierarchy of symptoms considered the most important to treat (the top three being breathing problems, walking difficulties and fatigue)
- The added psychological effects of COVID-19, the reduced testing and monitoring available and, in some cases, the cessation of ERT treatment

HCP interview responses highlighted some slight differences in perceptions compared to patients

- The significant emotional rollercoaster experienced by patients at diagnosis is not always fully recognized
- Although HCPs recognize that the opportunity for ERT is a boost to patients, they are conscious of managing expectations and are optimistic about the development of new treatments

Several HCPs noted that COVID-19 had driven the beneficial use of technology in terms of encouraging and streamlining virtual consultations

All HCPs recognize the burden of living with Pompe disease. In our sample, nurses were particularly attuned to the unmet needs of their patients

- Improved access to specialist physiotherapy and ancillary services such as hydrotherapy
- Ongoing mental health support
- Practical support such as information about disability benefits
- Specific information and support for teenagers and young adults with Pompe disease

CONCLUSIONS

- Findings from the participant interviews have provided insights into the psychological and emotional impact of the diagnostic process and of living with LOPD
  - While every participant's experience was different, this study identified an archetypal emotional journey
  - The diagnostic process was long and distressing, with most participants emphasizing their desire to reduce the length of time to receiving a diagnosis, being referred to a specialist HCP, and starting treatment
  - Participants requested access to additional support at the point of diagnosis and recommended educating GPs on Pompe disease
  - For most participants, the COVID-19 pandemic has been a period of increased anxiety, low mood and physical deterioration.
- Findings from HCP interviews aligned with those from the patient participants, as well as highlighting several unmet needs and optimism for future treatments for LOPD.
- An ongoing online survey of people living with LOPD in the UK is expected to provide additional quantitative insights into peoples' experiences and the impact of COVID-19.

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References

1. Kishnani PS *et al. Genet Med* 2006;8:267–88.
2. van der Beek NA *et al. Neuromuscul Disord* 2009;19:113–7.
3. Hagemans ML *et al. Brain* 2005;128:671–7.
4. Hagemans ML *et al. Neurology* 2004;63:1688–92.
5. Hagemans ML *et al. Neuromuscul Disord* 2007;17:537–43.

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Poster PDF

Supplement

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