Poster no. 130

Living with Pompe disease in the UK: characterizing the patient journey and burden on physical, emotional and social quality of life

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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]
  - Ultimately, 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 considerably 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 and behaviors at each stage
  - Impact of LOPD on daily activities, and the emotional and physical challenges.
- Here, we present interim results from interviews with people living with LOPD; interviews with healthcare professionals (HCPs) and a quantitative survey of people living with LOPD are ongoing, with results to be reported at a future meeting.

Figure 1. Interview study flow

People were invited to participate in the study by the two Pompe disease patient advocacy organizations in the UK*

Respondents completed a screening questionnaire to assess their eligibility†
n=27

Each eligible participant kept a preinterview diary of their experiences of living with LOPD
n=27

Each eligible participant took part in a 1-hour, in-depth, qualitative video or phone interview between June and July 2021
n=27

Eligibility criteria
- ≥18 years of age
- Diagnosis of LOPD
- UK resident
- Provided written informed consent

*Pompe Support Network and Association for Glycogen Storage Disease (UK); †More than 27 respondents were screened and eligible for the study; however, it was not possible to arrange interviews with all the respondents within the alloted time.

METHODS

In-depth interviews of people living with LOPD

- In line with British Healthcare Business Intelligence Association (BHBIA) guidance, all participants provided informed consent before any study-related activities.
- Screening questionnaires and interviews were scheduled with study participants (**Figure 1**).
- The 1-hour qualitative interviews were audio-recorded and transcribed
  - Interview discussion themes are available in the Supplement, which is accessible via QR code.
- Repeating themes were organized into theoretical constructs for analysis.

Quantitative survey of people living with LOPD

- In line with BHBIA guidance, all participants provided informed consent before any study-related activities.
- Adults with diagnosed LOPD who live in the UK were invited to participate in an online survey by the two patient advocacy organizations in the UK: Pompe Support Network and Association for Glycogen Storage Disease (UK).
- Survey topics are available in the Supplement, which is accessible via QR code.

RESULTS

Interview participants

- The characteristics of interview participants (n=27) are presented in **Figure 2**.

Disease journey

- **Figure 3** depicts some of the emotional aspects of 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 4**.
- 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 2. Characteristics of interview participants

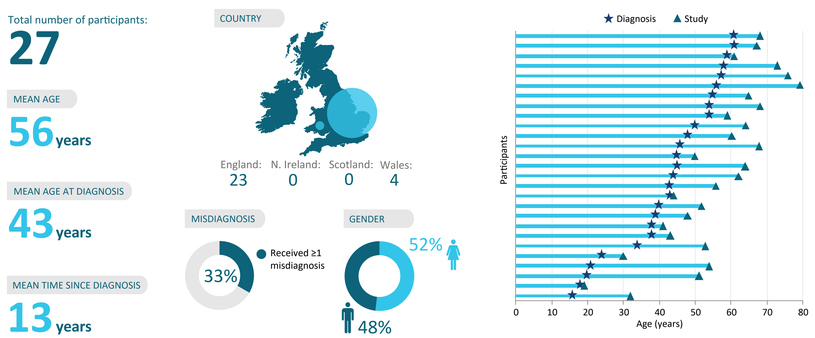
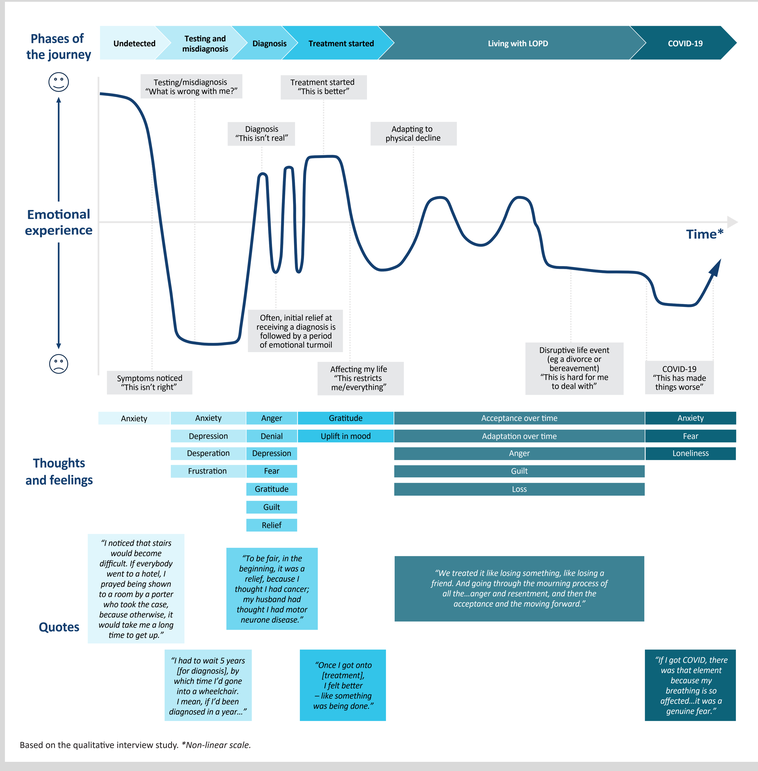
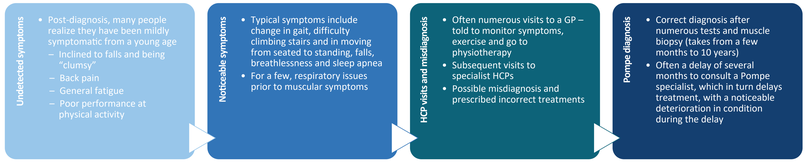


Figure 3. Schematic of the archetypal emotional journey from initial symptoms to being diagnosed with and living with LOPD



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

Figure 4. Typical stages of the diagnostic journey

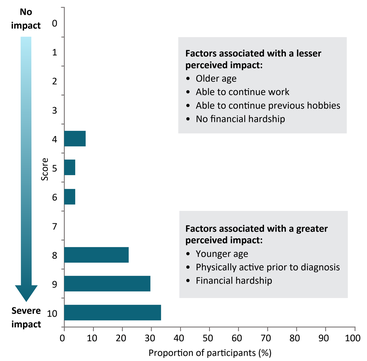


Based on the qualitative interview study. GP, general practitioner.

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 5**)
  - 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 5. 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.

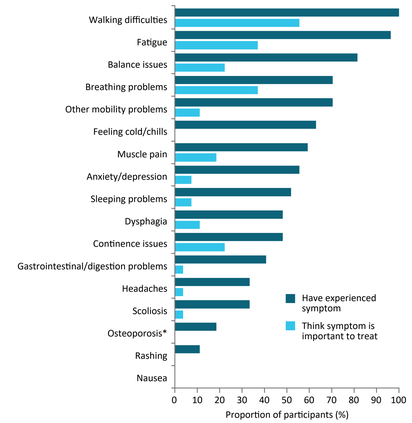
Symptoms

- 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 6**).
- 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 6**).

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
  - Reduced contact with HCPs and fewer assessments (eg lung function tests).

Figure 6. Frequency and importance of treating symptoms



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

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 generally 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 general practitioners on Pompe disease.
- The ongoing survey is expected to provide additional quantitative insights into the experiences of people living with LOPD.
- There remain multiple unmet needs throughout the disease journey for people living with LOPD.

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