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Living with late-onset Pompe disease in the UK: interim results characterising 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 characterised 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

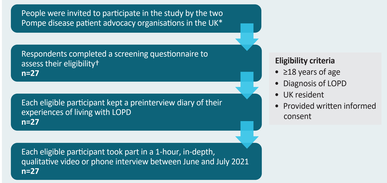
- To characterise the experiences of people from the United Kingdom (UK) living with LOPD
  - From first symptoms, through diagnosis and disease progression; mapping people's feelings, attitudes and behaviours at each stage
  - Impact of LOPD on daily activities, and the emotional and physical challenges.
- Here, we present interim results from interviews with people living with LOPD; interviews with healthcare professionals (HCPs) are ongoing, with results to be reported at a future meeting.

METHODS

In-depth interviews of people living with LOPD

- The qualitative interview study received the appropriate ethical approvals.
- 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 presented in **Table 1**.
- Repeating themes were organised into theoretical constructs for analysis.

Figure 1. Study flow

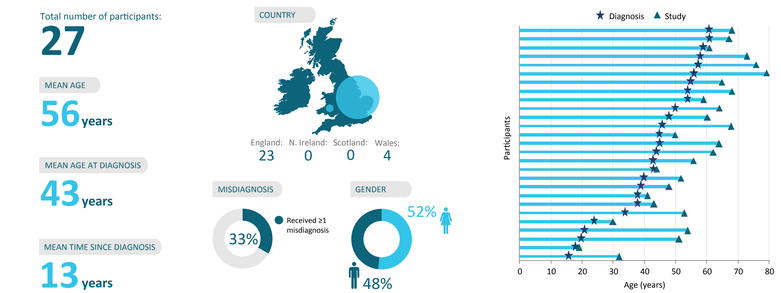


*Pompe Support Network and Association for Glycogen Storage Disease (UK); †More than 27 respondents were screened and eligible for the study; however, it was not possible to arrange interviews with all the respondents within the alloted time.

Table 1. Interview discussion themes

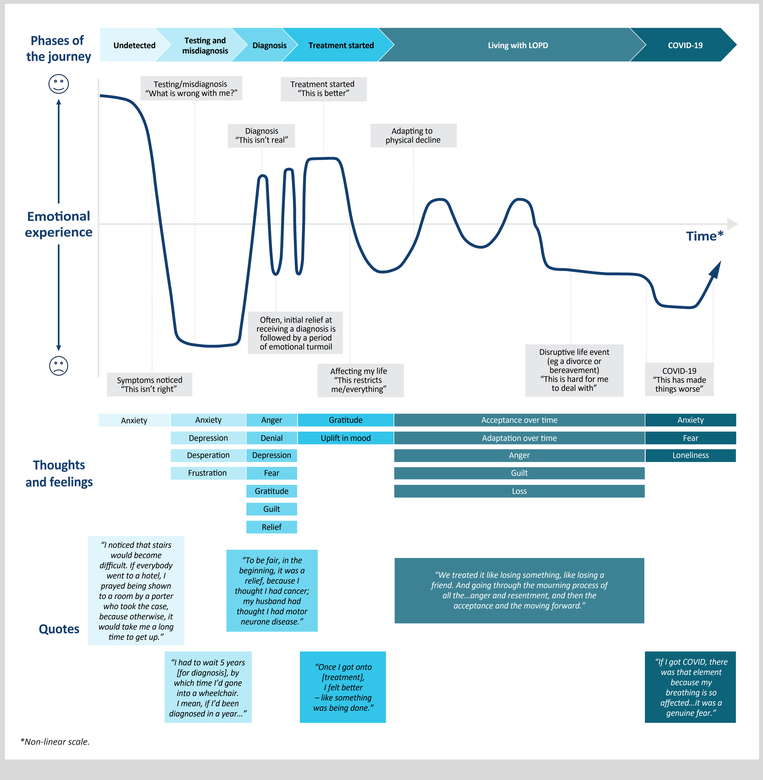
Discussion theme	Discussion topic(s)	Example question(s)
Personal context	People were asked to (1) introduce themselves, (2) provide some background on their experiences with LOPD, and (3) discuss the perceived impact of COVID-19 on their care and emotional wellbeing	"Tell me a bit about Pompe and you"
Prediagnosis	People were asked to (1) draw a timeline from first symptoms to now, and (2) describe their symptoms and experiences before receiving a diagnosis of LOPD	"Describe the thoughts and feelings you experienced alongside the symptoms"
Diagnosis	People were asked to describe their experience of the diagnostic process, from first consultation with an HCP to receipt of the correct diagnosis	"Tell me how your diagnosis was arrived at" "Describe your feelings on first hearing the diagnosis"
Post-diagnosis	People were asked to describe (1) how the disease has progressed since diagnosis, and (2) the accompanying emotions	"How would you describe living with Pompe to someone who does not know the condition?"
Living with LOPD	People were asked to describe their experience of living with LOPD, including (1) the symptoms and severity, (2) their emotional state, and (3) any challenges	"What symptoms do you live with now?" "What aids, if any, do you use?"
LOPD disease management landscape	People were asked to (1) describe their interactions with HCPs, and (2) discuss the advantages and disadvantages of their current disease management, including any unmet needs	"What, if any, additional support/help would you like in relation to your treatment?" "What could improve the experience for you?"
Comorbidities	People were asked to describe their other conditions	"Do you have and receive treatment for any other conditions?"
Future hopes and expectations	People were asked about their hopes for the future management of their condition	"How would you like to see treatments for Pompe disease developing in the future?"

Figure 2. Characteristics of study participants



RESULTS

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



*Non-linear scale.

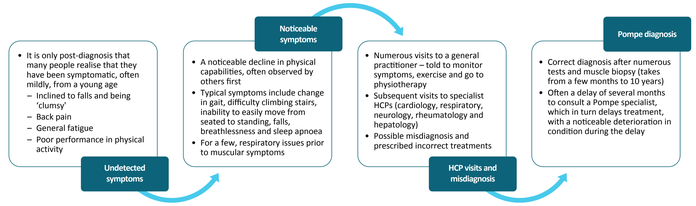
Study participants

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

Disease journey

- **Figure 3** depicts some of the emotional aspects to participants' diagnostic journeys and of living with LOPD.
- 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.

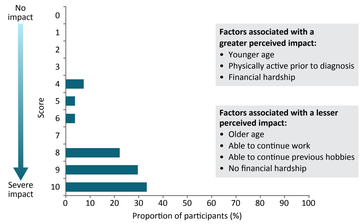
Figure 4. Typical stages of the diagnostic journey



Challenges of living with LOPD

- The majority of 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, participants felt that disruptive life events, such as accidents or bereavements, added to the physical and emotional burden.
- Most 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



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 analysed.

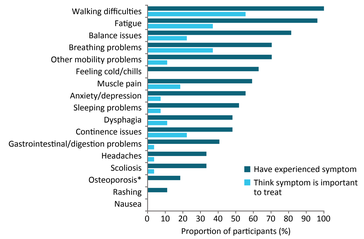
Symptoms

- 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 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



*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 emphasising 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.
- There remain multiple unmet needs throughout the disease journey for people living with LOPD.

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