

Living with Pompe disease in the UK: characterising 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 characterised by progressive skeletal and/or respiratory muscle weakness, is a devastating condition for those diagnosed and their families¹
- 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 characterise 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 of the disease
 - 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 results from interviews with people living with LOPD and qualitative results from interviews with a small sample of HCPs.

METHODS

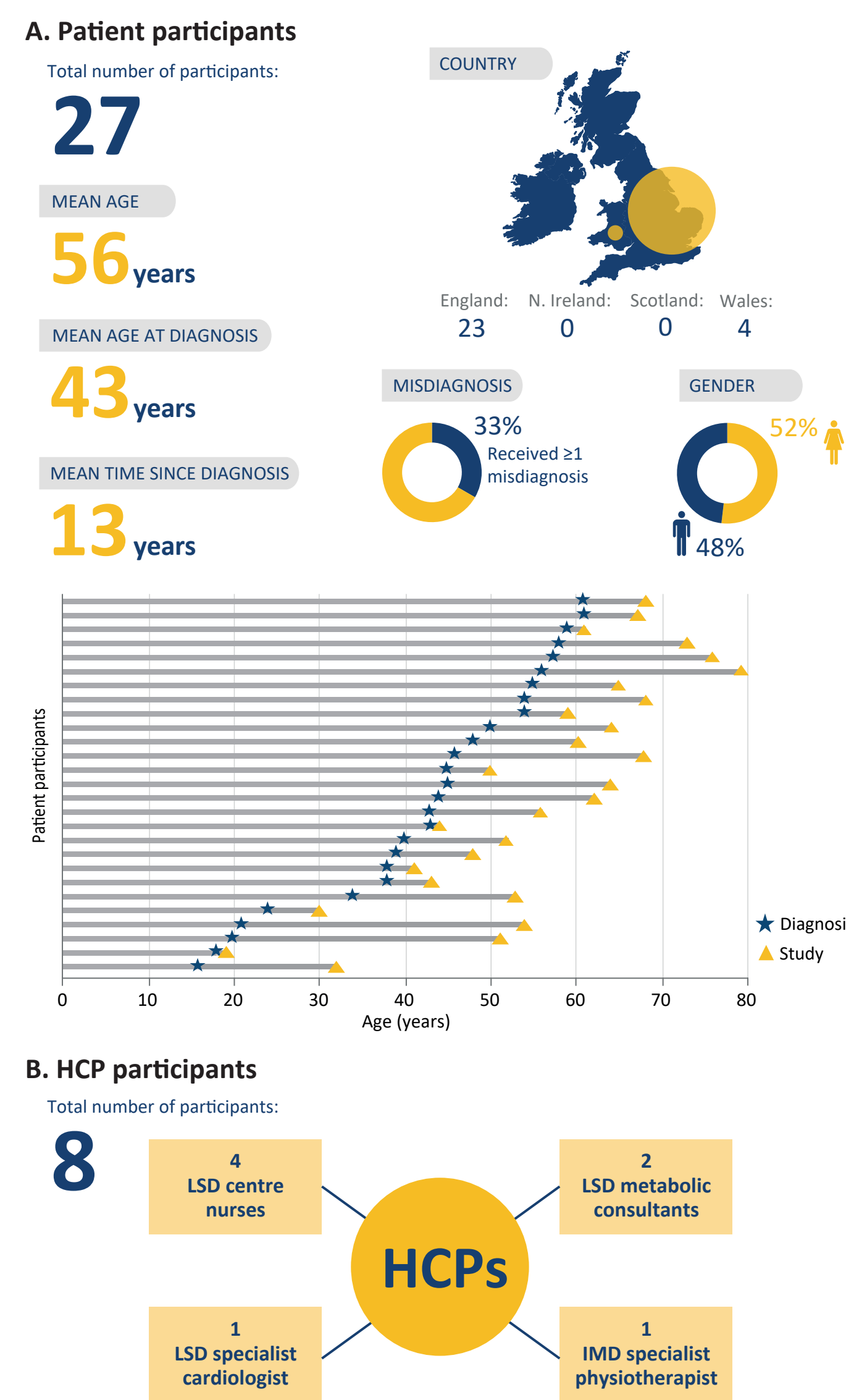
- Data were collected by conducting:
 - In-depth qualitative interviews with people living with LOPD
 - Qualitative interviews with HCPs.
- Further details on the methodology and interview discussion themes are available in the Supplement, which is accessible via quick response (QR) code.

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 voice-over by the presenting author describing in detail the archetypal emotional journey 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 2. Schematic of the archetypal emotional journey from initial symptoms to being diagnosed with and living with LOPD

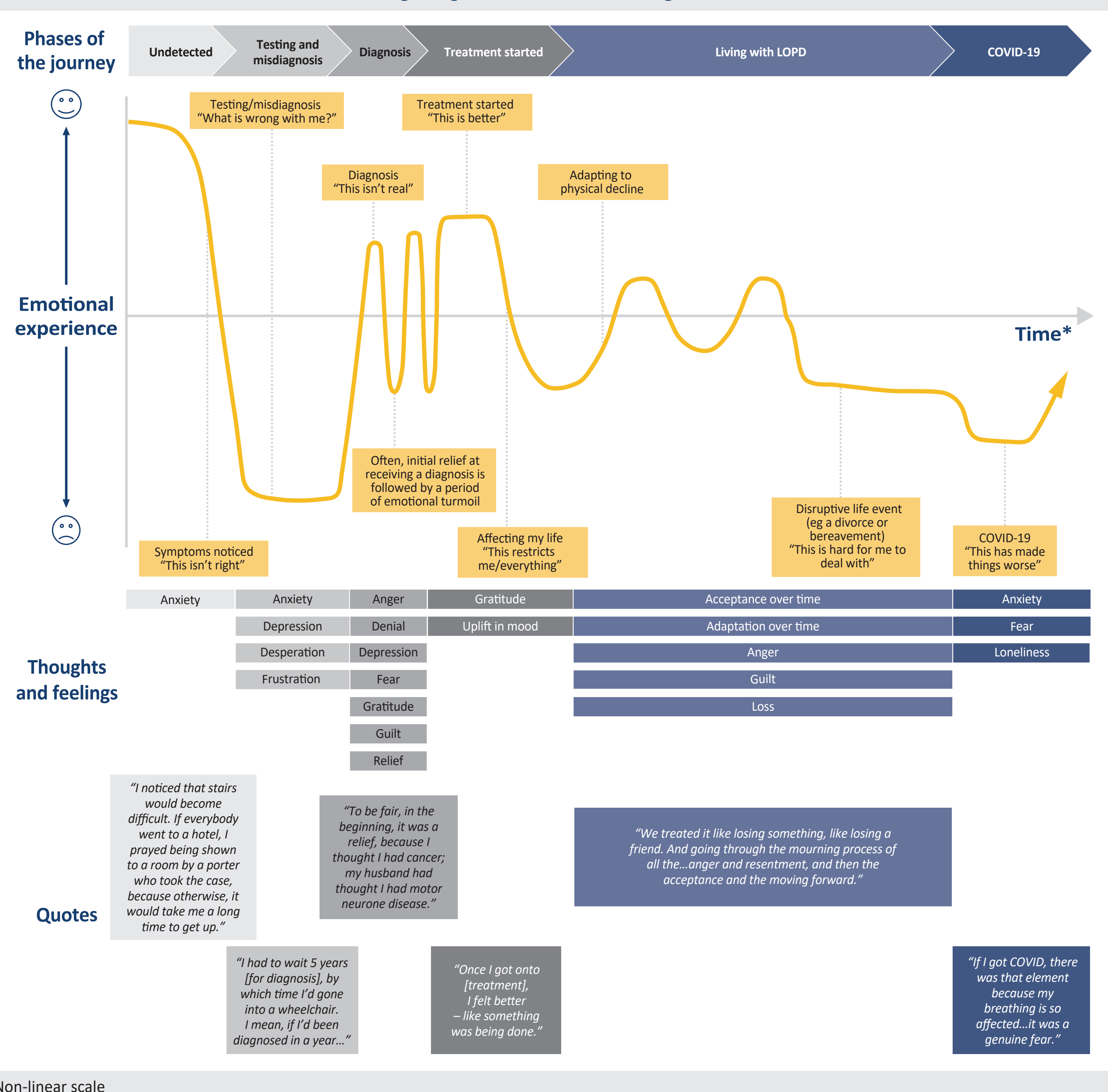
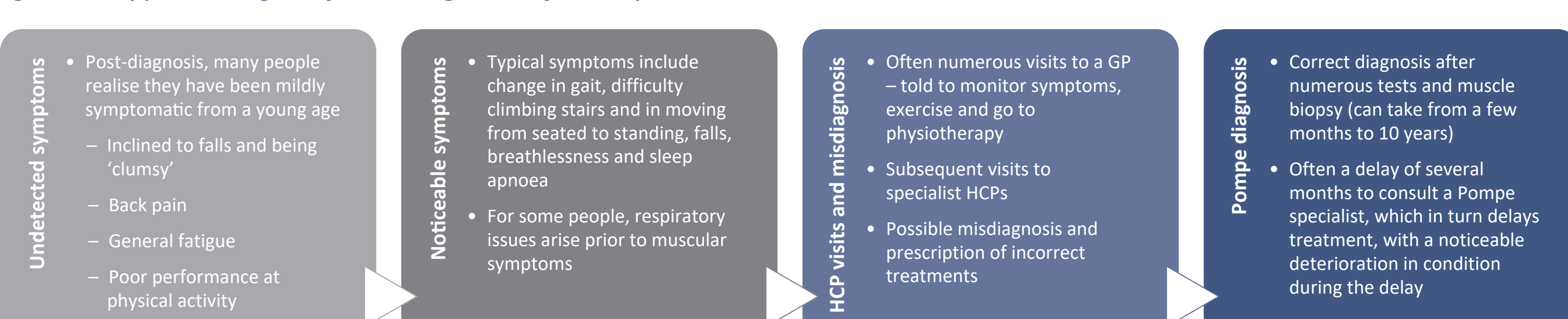


Figure 3. Typical stages of the diagnostic journey

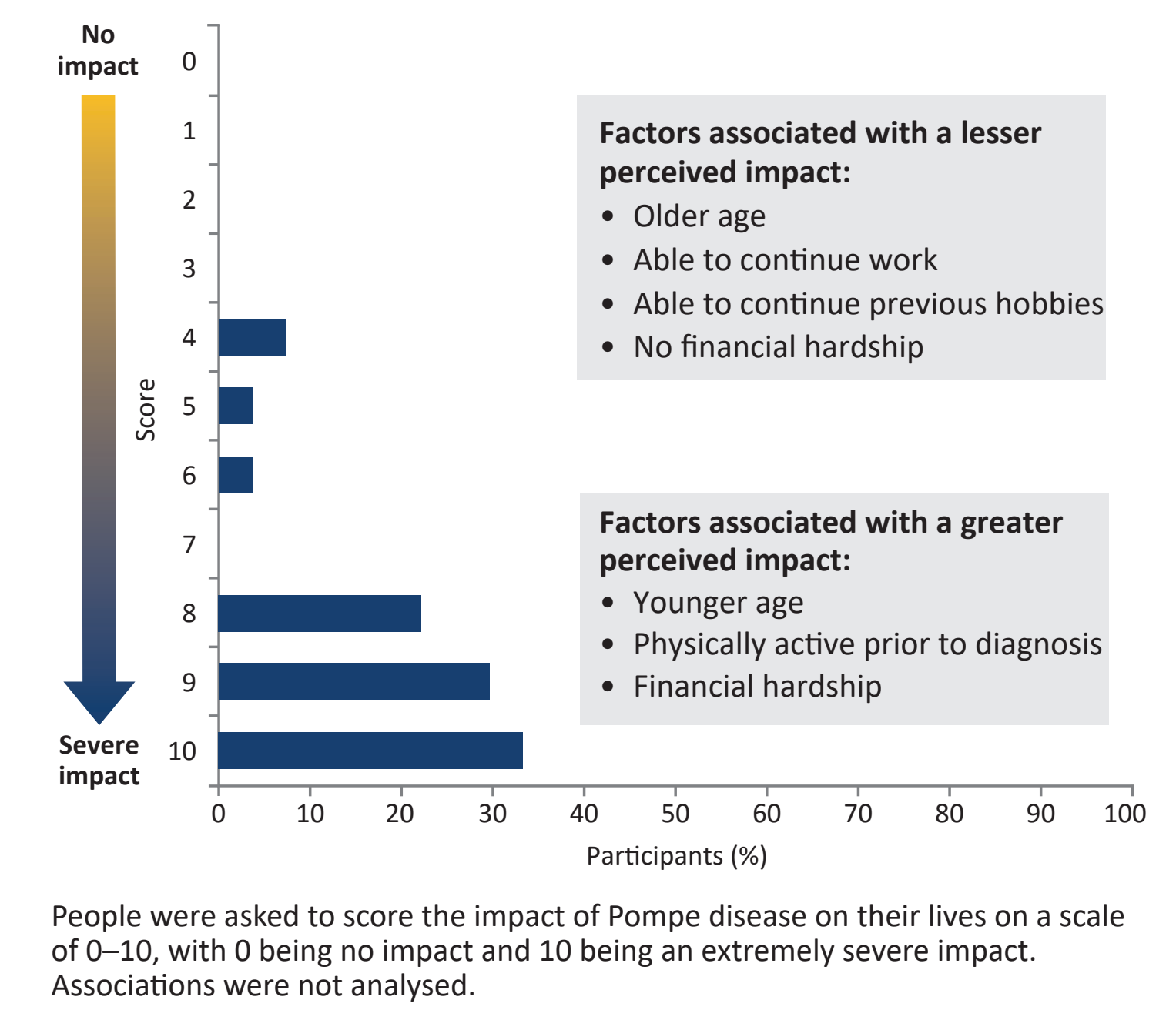


GP, general practitioner.

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 4**)
 - On a scale of 0–10 (with 0 being no impact at all and 10 being a severe impact), the mean (\pm standard deviation) score was 8.5 (\pm 1.8).

Figure 4. Perceived impact of LOPD on participants' lives

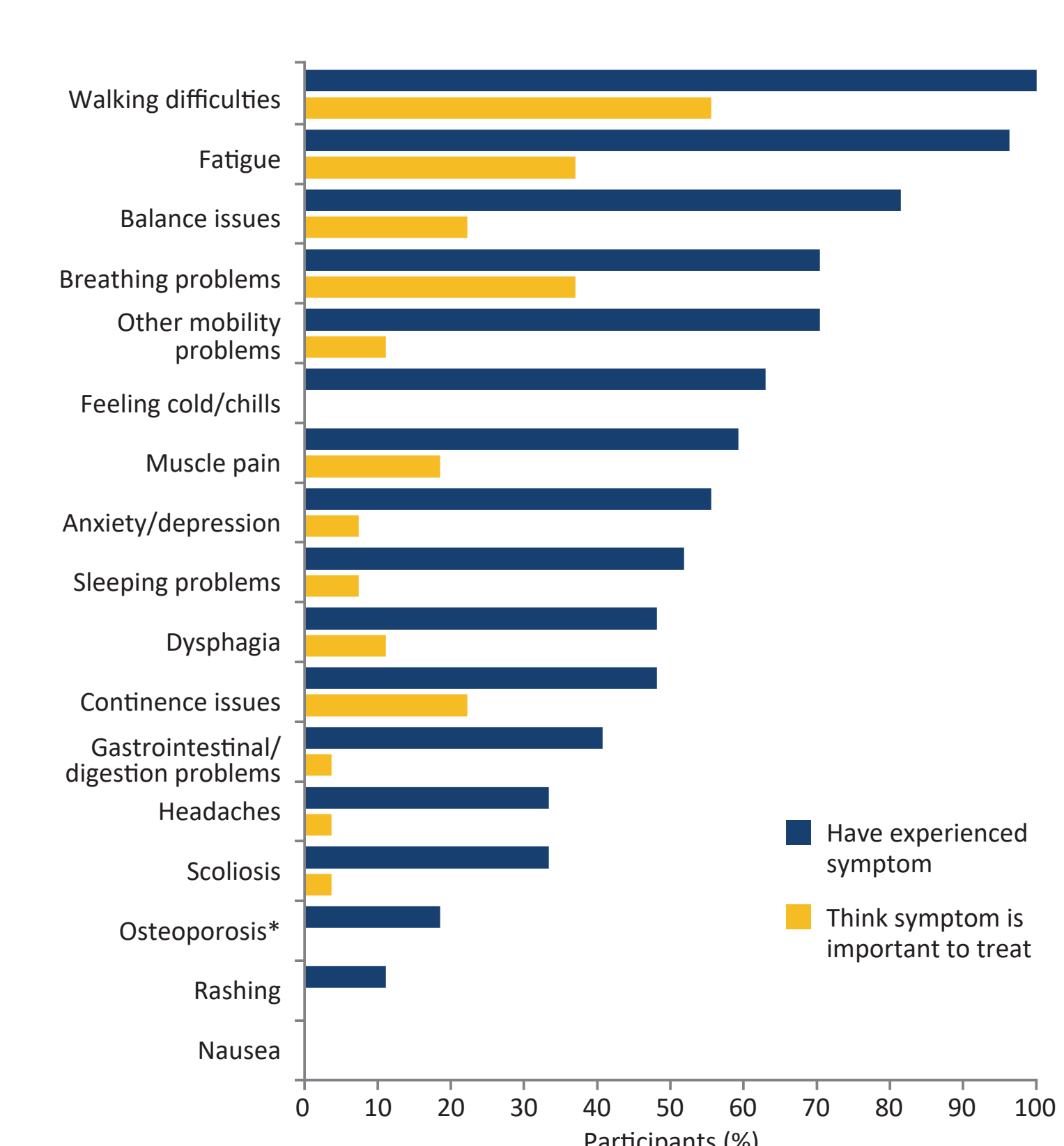


People were asked to score the impact of Pompe disease on their lives on a scale of 0–10, with 0 being no impact and 10 being an extremely severe impact. Associations were not analysed.

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 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**).

Figure 5. Frequency and importance of treating symptoms



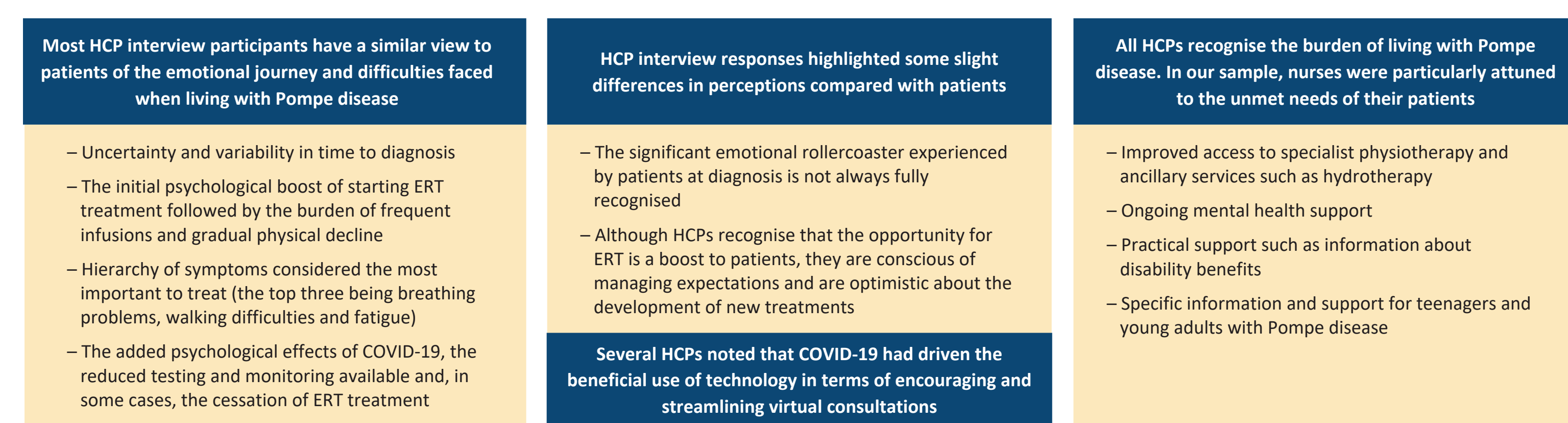
*Comorbidity (and not a symptom).

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).

Interviews with HCPs

Figure 6. Main findings from HCP interviews



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 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 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.
- Additional quantitative insights into people's experiences of living with LOPD and the impact of COVID-19 have been collated from an online survey and can be found in our poster entitled 'Quantification of the burden, unmet needs, management, and COVID-19 impact of living with Pompe disease in the UK: results of an online patient survey'.

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Narration of poster



Poster PDF + Supplement



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