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 lateonset 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

Phases of

the journey

Emotional

experience

••

Thoughts

and feelings

Quotes

*Non-linear scale

GP, general practitioner.

• The characteristics of patient interview participants are presented in Figure 1A.

Testing and

What is wrong with me?"

Diagnosis

Diagnosis "This isn't real"

Often, initial relief at receiving a diagnosis is

followed by a period

Fear

Gratitude

Guilt

Relief

"To be fair, in the

beginning, it was a

relief, because I

thought I had cancer;

my husband had

thought I had motor

neurone disease."

Frustration

"I had to wait 5 years

[for diagnosis], by which time I'd gone

into a wheelchair. I mean, if I'd been

diagnosed in a year...

"This restricts

me/everything"

"Once I got ont

I felt better

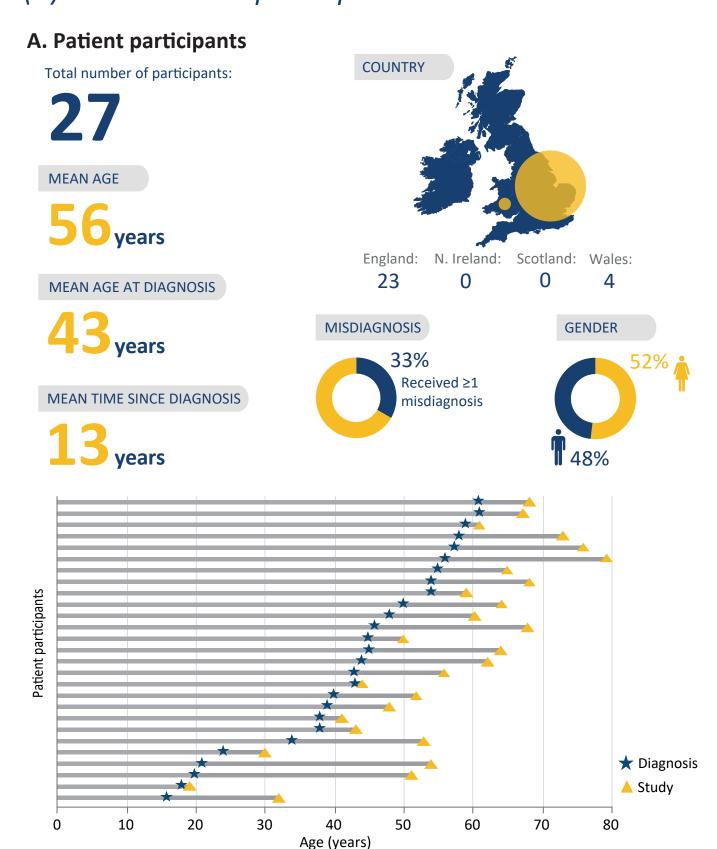
was being done.

"This is better"

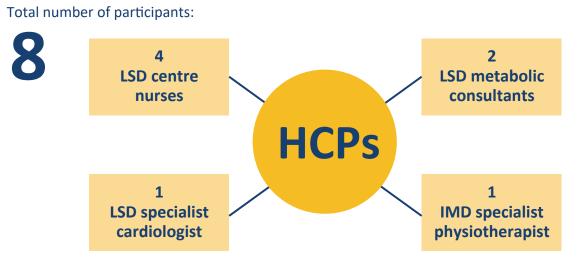
 The roles and workplace settings of HCP interview participants are presented in Figure 1B.

Undetected

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



B. HCP 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

Living with LOPD

Figure 2. Schematic of the archetypal emotional journey from initial symptoms

to being diagnosed with and living with LOPD

physical decline

Time to diagnosis and early interactions with HCPs

COVID-19

Time*

COVID-19

"This has made

things worse"

Anxiety

'If I got COVID, there

was that element

because my

affected…it was a

genuine fear."

Age and availability of therapy at diagnosis.

Disruptive life event

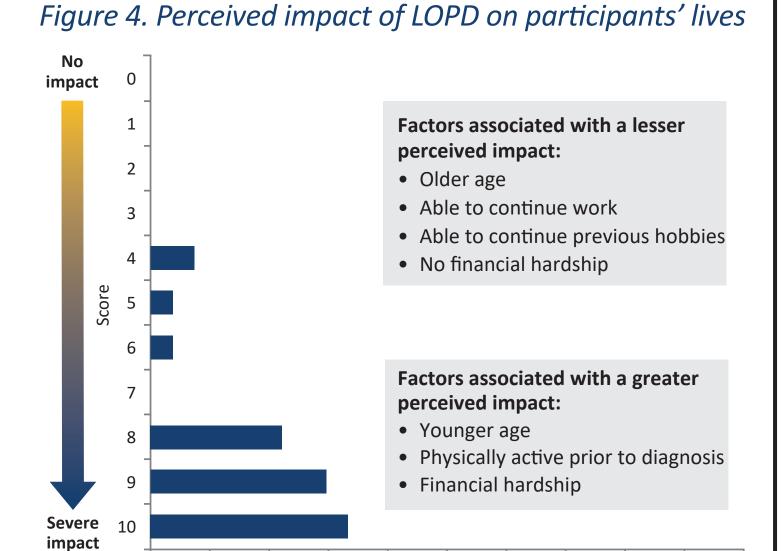
(eg a divorce or

"This is hard for me to

deal with"

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).



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.

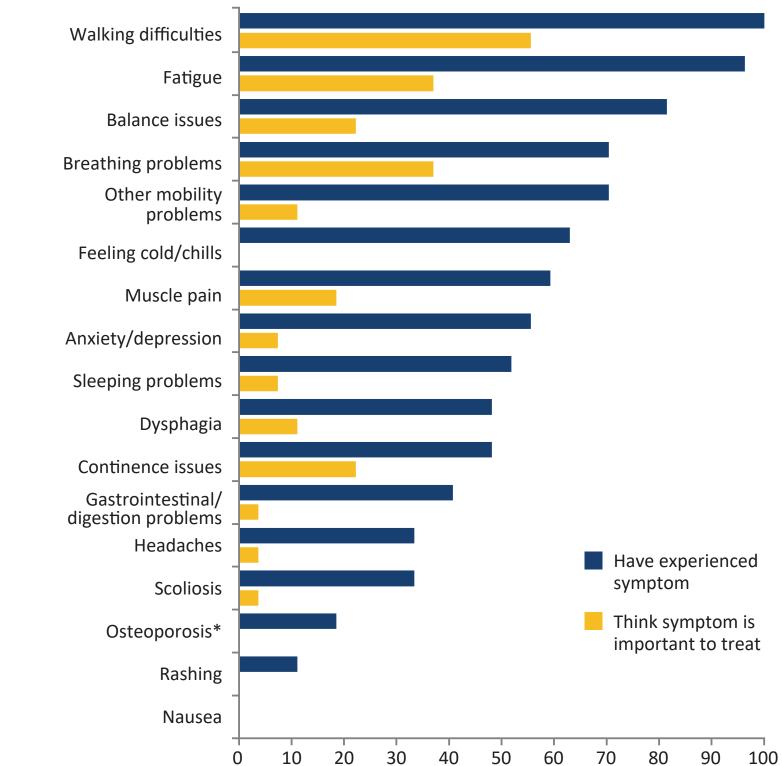
Figure 5. Frequency and importance of treating symptoms

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).



*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 when 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 with patients The significant emotional rollercoaster experienced

by patients at diagnosis is not always fully

 Although HCPs recognise 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 recognise 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

Participants (%)

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'.

Symptoms noticed

"I noticed that stairs

would become

difficult. If everybody

went to a hotel, I

prayed being shown

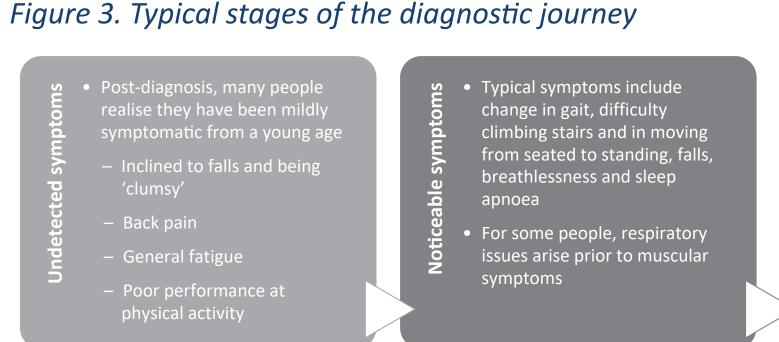
to a room by a porter

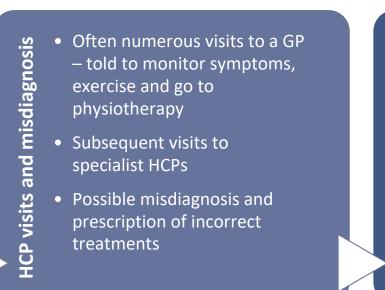
who took the case,

because otherwise, it

would take me a long

time to get up."





"We treated it like losing something, like losing a

all the...anger and resentment, and then the

acceptance and the moving forward."

numerous tests and muscle biopsy (can take from a few months to 10 years) Often a delay of several months to consult a Pompe specialist, which in turn delays treatment, with a noticeable deterioration in condition during the delay

Correct diagnosis after

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Amicus Therapeutics, Inc.

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Narration of poster







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