Poster no. 178

Impact of mobility-aid use on late-onset Pompe disease (LOPD) patient experience: insights from patient interviews

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- **Poster PDF**
- **Supplementary material**
- **Patient quotes infographic**

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Introduction and objectives

- LOPD is a progressive disease that can lead to irreversible muscle damage.[1,2]
- The muscle damage caused by LOPD can severely impact breathing, movement, quality of life and daily functioning, placing a heavy burden on individuals, families and caregivers.[1–4]
- While clinical outcomes such as the 6-minute walk test and forced vital capacity are standard measures of disease progression, they often fail to capture patients' lived experience of LOPD, such as the emotional impact of the disease.[5]
- The Pompe Emotional Experience Research (PEER) study was a primary, qualitative interview-based study that aimed to explore the lived experience of people diagnosed with LOPD from symptom onset through diagnosis, treatment and disease progression.
- Here, we describe findings from the PEER study that explore mobility-aid use as a real-world indicator of functional decline and adaptation, emotional experiences throughout the patient journey, and the unmet needs and challenges of people living with LOPD.

Conclusions

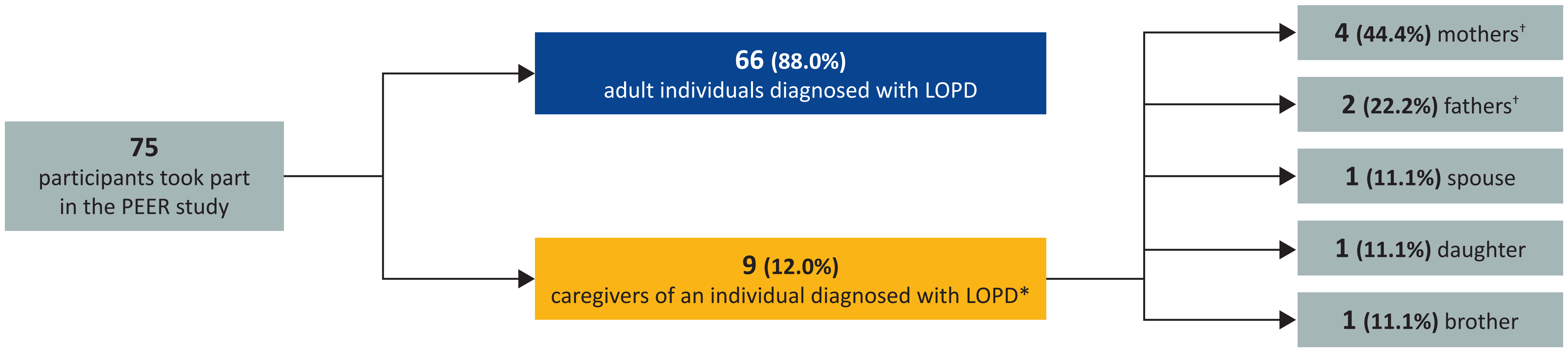
- The findings presented here suggest that mobility-aid use substantially impacts the daily lives of people living with LOPD, with mobility-aid users reporting greater functional impairment than non-users.
- Patients described their progression through everyday activities that matter most to them – things they can no longer do, fear losing, or now require extra effort, such as walking or climbing stairs.
- The qualitative insights illustrate how people living with LOPD experience disease progression in diverse ways. Small but meaningful changes in function influence daily life in ways not captured by current outcome measures.
- Overall, these findings highlight the importance of identifying the activities and experiences that matter to each individual and ensuring that comprehensive assessment and support are incorporated into routine monitoring and care, including early access to occupational therapy.

Methods

- People aged 18 or older with confirmed LOPD, as well as caregivers of individuals with confirmed LOPD of any age, in Australia, Canada, Germany, New Zealand, the UK and the USA completed the study.
- Participants completed surveys capturing demographics and mobility-aid and wearable use, which included Patient-Reported Outcomes Measurement Information System (PROMIS) questionnaires assessing self-efficacy for managing daily activities and social interactions.
- Participants also took part in patient- and clinician-informed semi-structured interviews exploring participants' diagnostic and treatment journey, and disease progression, including emotional impacts, burdensome aspects, and thoughts regarding the future.
- Additional details are reported in the **Supplementary material**, available via the QR code.

Results

Of the 75 individuals who completed the interviews, the majority were adults diagnosed with LOPD



75 participants took part in the PEER study

66 (88.0%) adult individuals diagnosed with LOPD

9 (12.0%) caregivers of an individual diagnosed with LOPD*

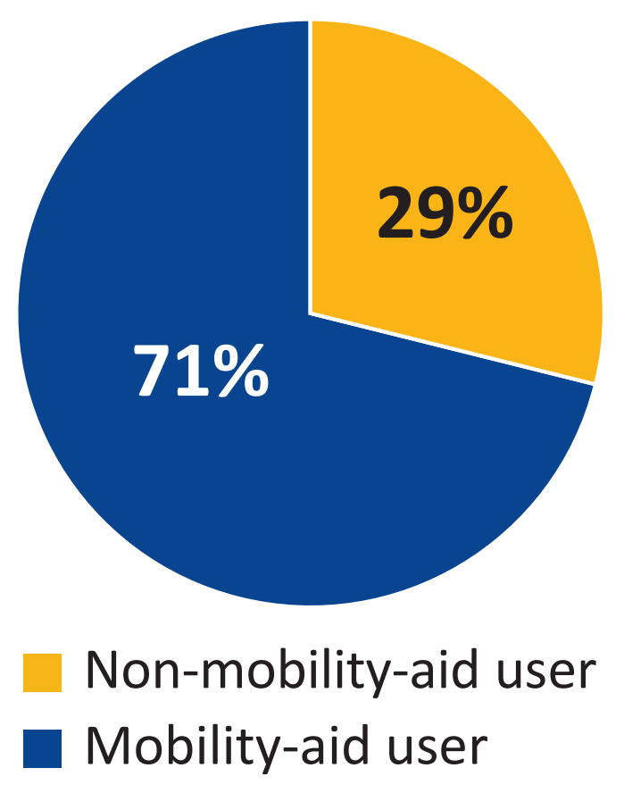
- **4** (44.4%) mothers†
- **2** (22.2%) fathers†
- **1** (11.1%) spouse
- **1** (11.1%) daughter
- **1** (11.1%) brother

*Caregivers completed surveys and interviews on behalf of individuals diagnosed with LOPD; †Mothers and fathers completed surveys and interviews on behalf of children aged <18 years diagnosed with LOPD.

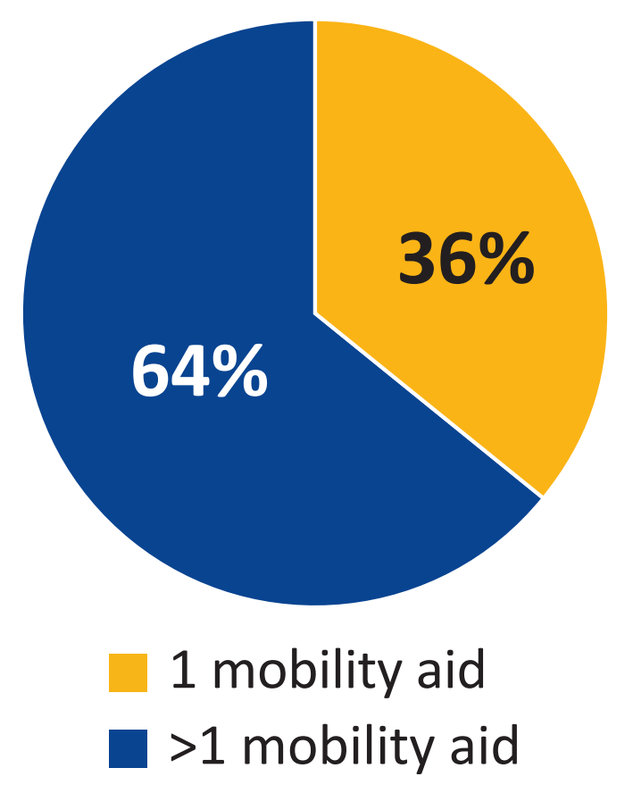
- Demographics and baseline characteristics are reported in the **Supplementary material**, available via the QR code.

Mobility-aid users reported greater functional impairment than non-users

Use of mobility aids (N=75)

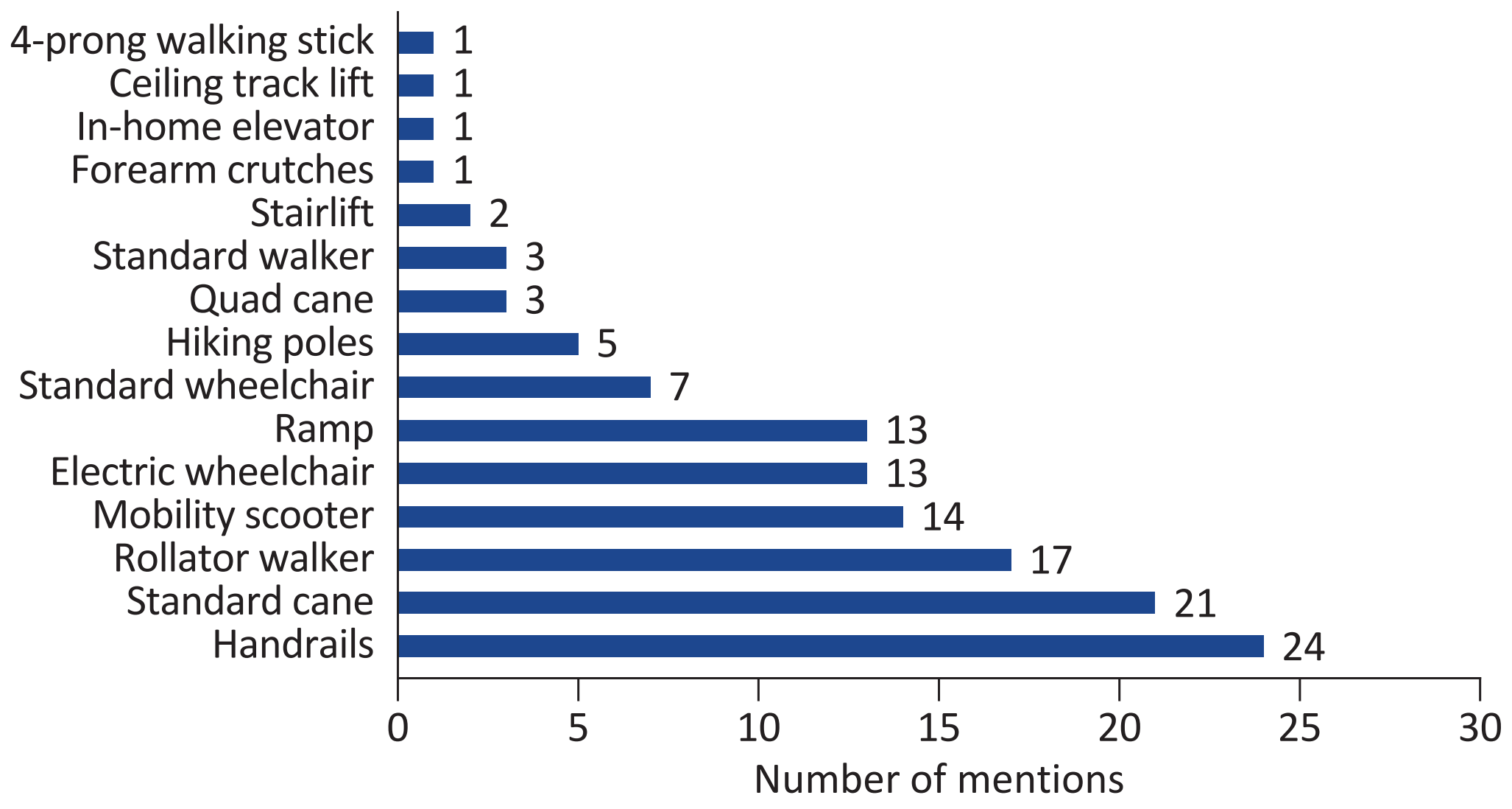


Number of mobility aids used (n=53)

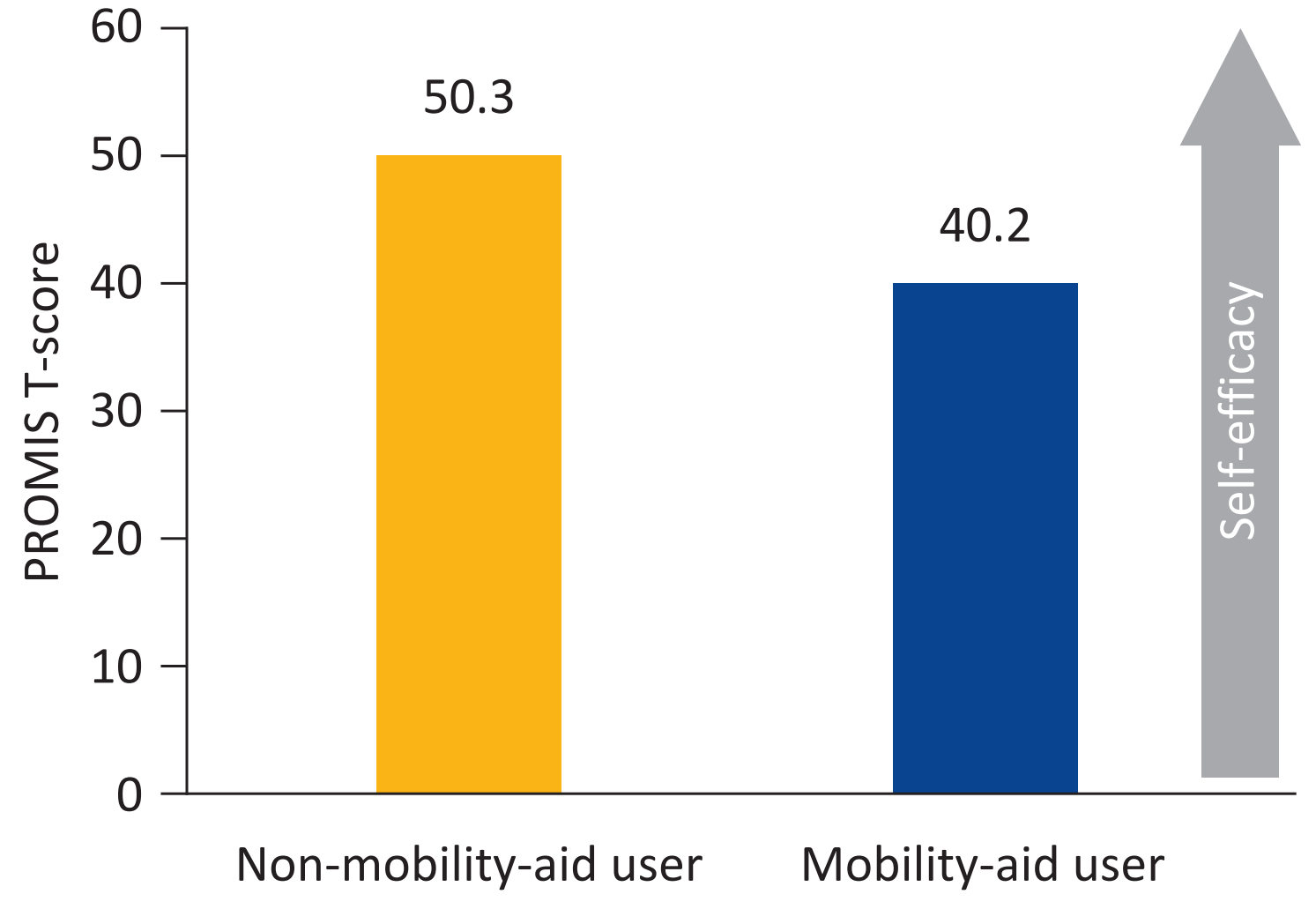


Participants reporting having accessed occupational therapy (N=75)

Types of mobility aids used



Managing daily activities



Data shown are for individuals diagnosed with LOPD, with surveys and interviews completed by the individual or by a caregiver on their behalf.

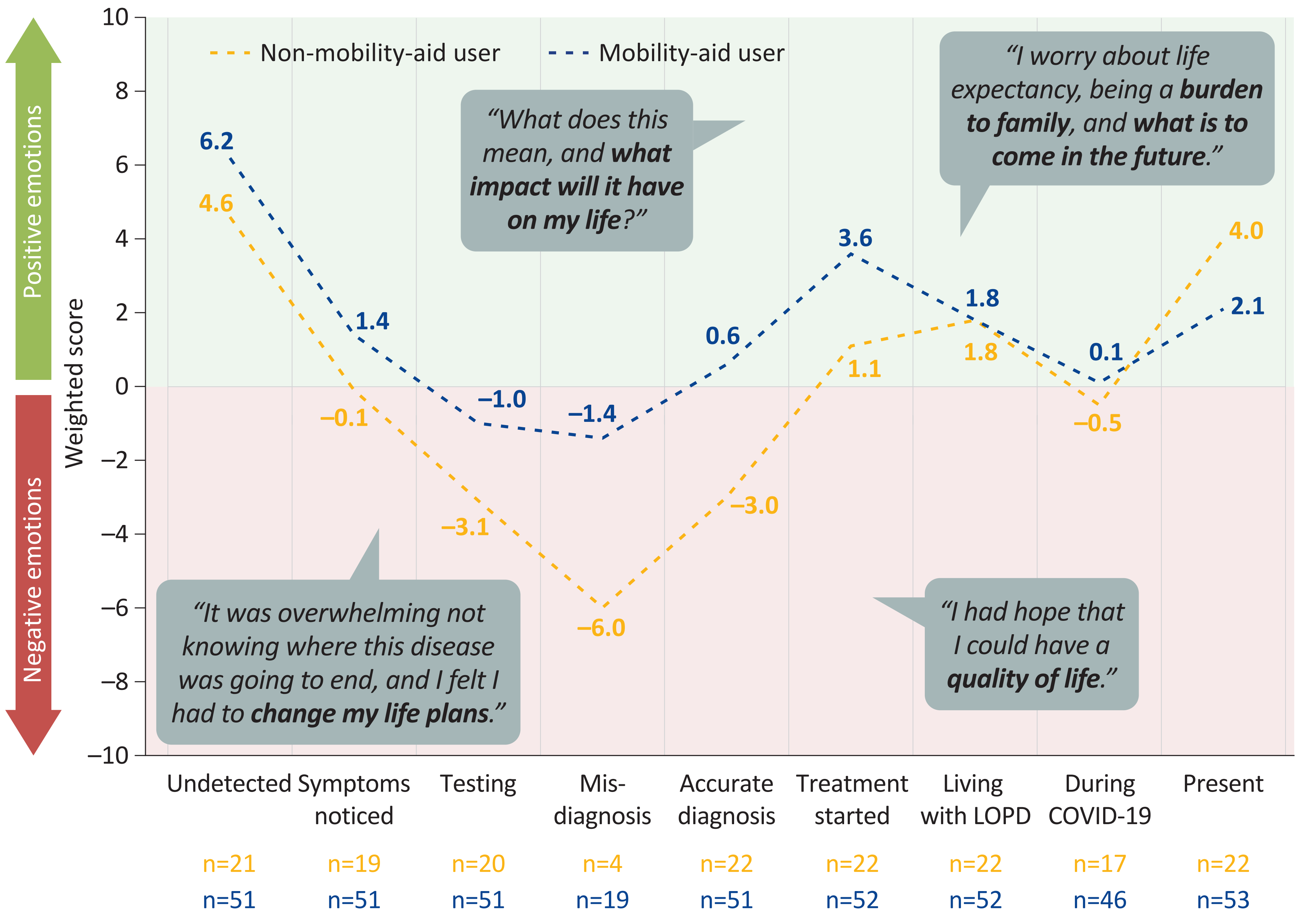
- Most participants (n=53; 71%) reported using mobility aids.
- Of the 53 mobility-aid users, there was a total of 15 different types of mobility aid used, with 19 (36%) using one aid and 34 (64%) using more than one aid.
- These data, along with patient quotes highlighting the impact of their life beyond quantitative outcomes, suggest substantial disease burden:

> *"The biggest challenge is **just functional** – walking up the stairs, getting out of a chair – it's frustrating as things are getting harder… those everyday functions."*

> *"The physical burdens, **everything I have to do has to be thought through**… like walking…"*

  - Further patient quotes are available in the **Patient quotes infographic**, available via the QR code.
- Despite the high requirement for mobility aids within this population, only six of 75 individuals (8%) reported having accessed occupational therapy, five of whom (83%) were mobility-aid users, highlighting a critical gap in supportive care.
- Mobility-aid users reported greater functional impairment, with lower mean PROMIS T-scores in managing daily activities (10.1 points lower), but minimal differences in social interactions (0.8 points lower), versus non-users.

Mobility-aid users recalled better emotional experiences at most time points throughout the LOPD journey than non-users



Zero represents neutral emotions, positive numbers represent positive emotions, and negative numbers represent negative emotions. Scores represent discrete stages; line added for visual clarity. COVID-19, coronavirus disease 2019.

- Despite greater functional impairment, mobility-aid users rated their emotional experience as better than or similar to non-users at most time points, suggesting adaptive coping or acceptance.
- Direct quotes from patients provide important insights into how they experience their functional impairment and disease progression at different points in their journey, and highlight activities of daily living as a priority for patients:

> *"The uncertainty is the worst part of the disease… **not knowing what the future holds** for symptoms, quality of life, retirement, wondering what it will look like, wondering if I am at my strongest now."*

> *"I am optimistic and **try to live day to day**. I have to consider what to do if things get worse, whether I will get home care or go into a home, having to give up my cats."*

  - For more direct quotes, see the **Patient quotes infographic**, available via the QR code.

References

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