

The impact of patient perception of disease progression on health-related quality of life (HRQoL) in late-onset Pompe disease



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Background and objectives

- Pompe disease is a **rare, autosomal recessive metabolic disorder**, with an estimated worldwide prevalence of approximately 1 per 4,447–37,094 cases.¹
- LOPD is progressive with **significant heterogeneity between patients**, thus it is **critical to explore patients' perspectives** to understand the true burden of disease.²
- Several studies have demonstrated the impact of LOPD on quality of life (QoL) using patient-reported outcome measures (PROMs).³
- To our knowledge, this is the **first study combining PROM data (36-item short-form [SF-36]) with insights from patients' individual self-reported perceptions** of whether their LOPD was declining, stable, or improving, in addition to data gathered through a series of **qualitative questions**.

Conclusions

- This is the first study comparing the **impact of LOPD on patient perceptions of their disease status and their SF-36 scores, and the additive impact of requiring both physical and ventilator support**.
- Considering the heterogeneous nature of LOPD, **using PROMs and individual patient perceptions of their disease provides important context for understanding its QoL impact** beyond traditional clinical measurements.
- Incorporating patients' views on their LOPD will allow for more holistic and collaborative management.

Methods

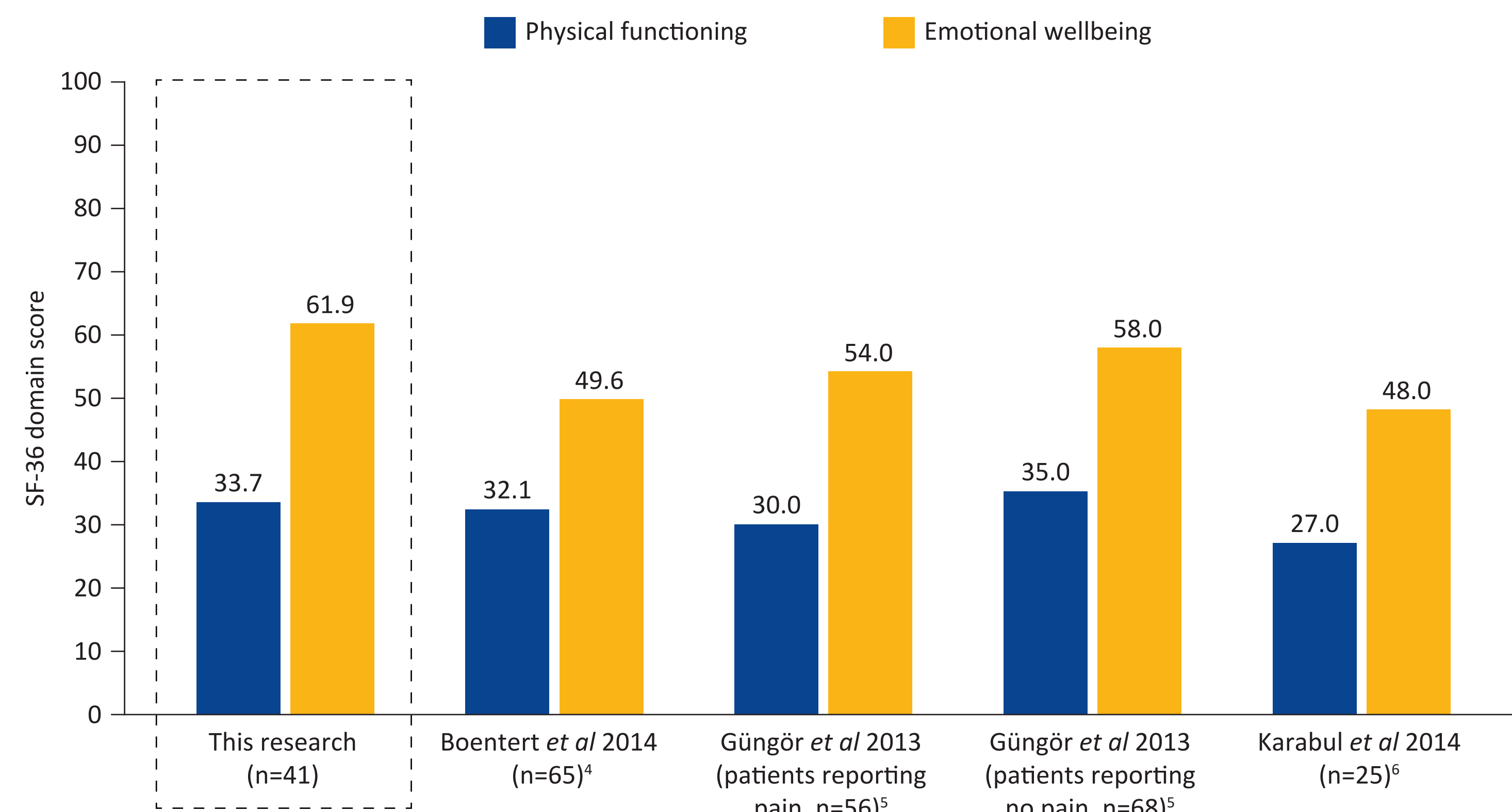
- With the assistance of patient advocacy organisations (PAOs), **41 adult participants from Australia (n=11), France (n=11), Italy (n=9), and the Netherlands (n=10)** were recruited.
- Patients with LOPD were considered eligible if they were **receiving enzyme replacement therapy and not taking part in a clinical trial**.
- The **SF-36 was used alongside a multi-modal survey** to understand how the **disease status of patients at the time of completion (ie declining, stable, or improving) related to their self-reported burden of disease and their physical functioning**, based on their 6-minute walk distance (6MWD), and forced vital capacity (FVC) results (where available).

- The SF-36 was selected as an appropriate, **validated tool evaluating patient QoL in concordance with previously published literature**.^{4–6} SF-36 component scores were calculated **for comparability with previous published LOPD literature**, and general health, physical functioning, and emotional wellbeing were used as **measurable indicators of the different aspects of patient QoL**.
- The survey collected **verbatim responses to investigate themes not often captured by PROMs alone**, including the **physical aid/ventilator requirements** and LOPD progression status of participants.
- The study was **completed online at the end of 2023**.

Results

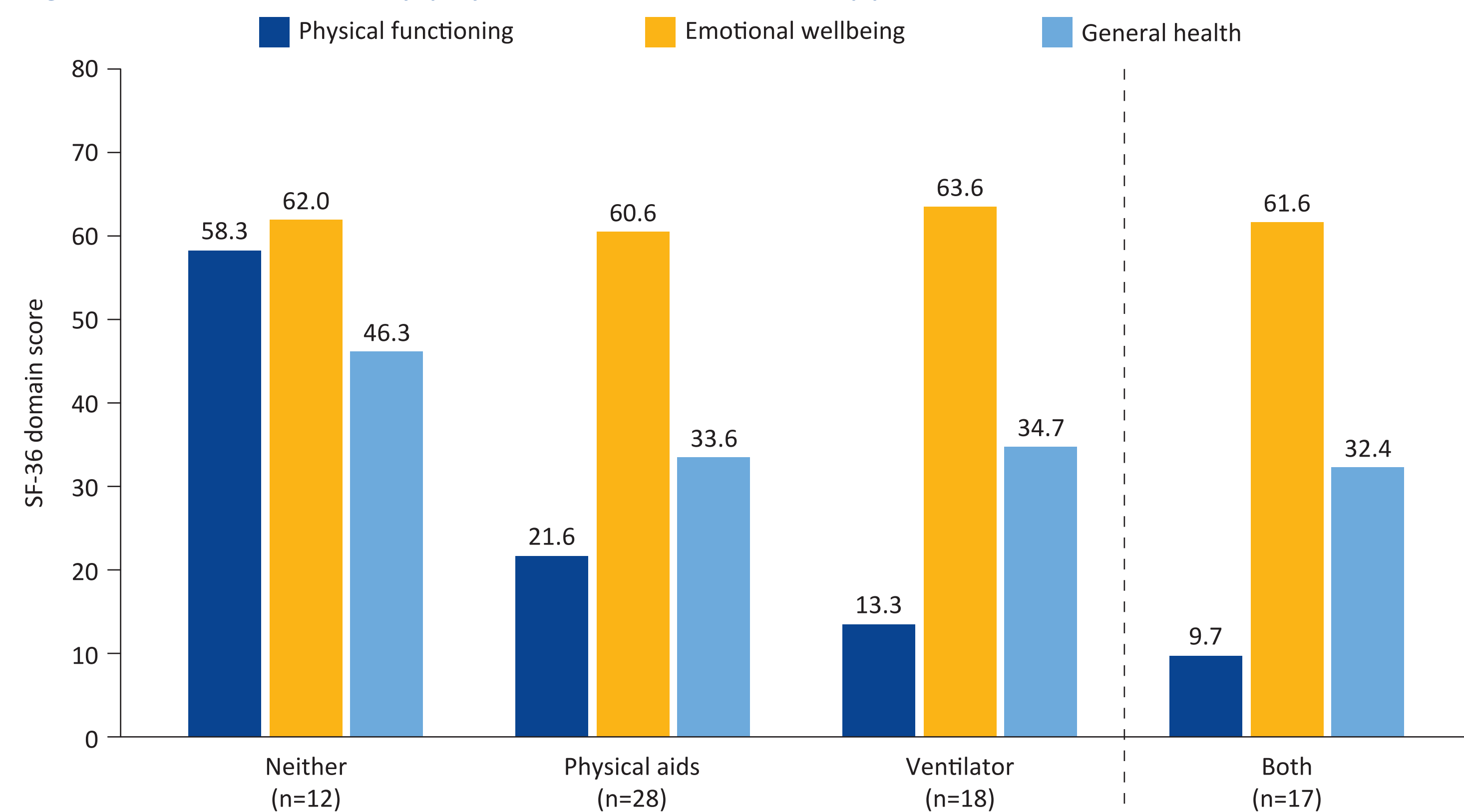
- Among 41 participants, **23 (56%) reported decline, 14 (34%) reported being stable, and only four (10%) patients felt their disease was improving**. Patients described their decline with respect to physical symptoms, such as **muscle weakness, pain, and fatigue**, as well as their ability to complete **daily activities**.
- Overall, **SF-36 scores** (each scored to a maximum of 100) **were broadly in line with previously published literature (Figure 1)**, suggesting that the survey population was representative of patients with LOPD.

Figure 1. Study SF-36 scores compared with published LOPD literature^{4–6}



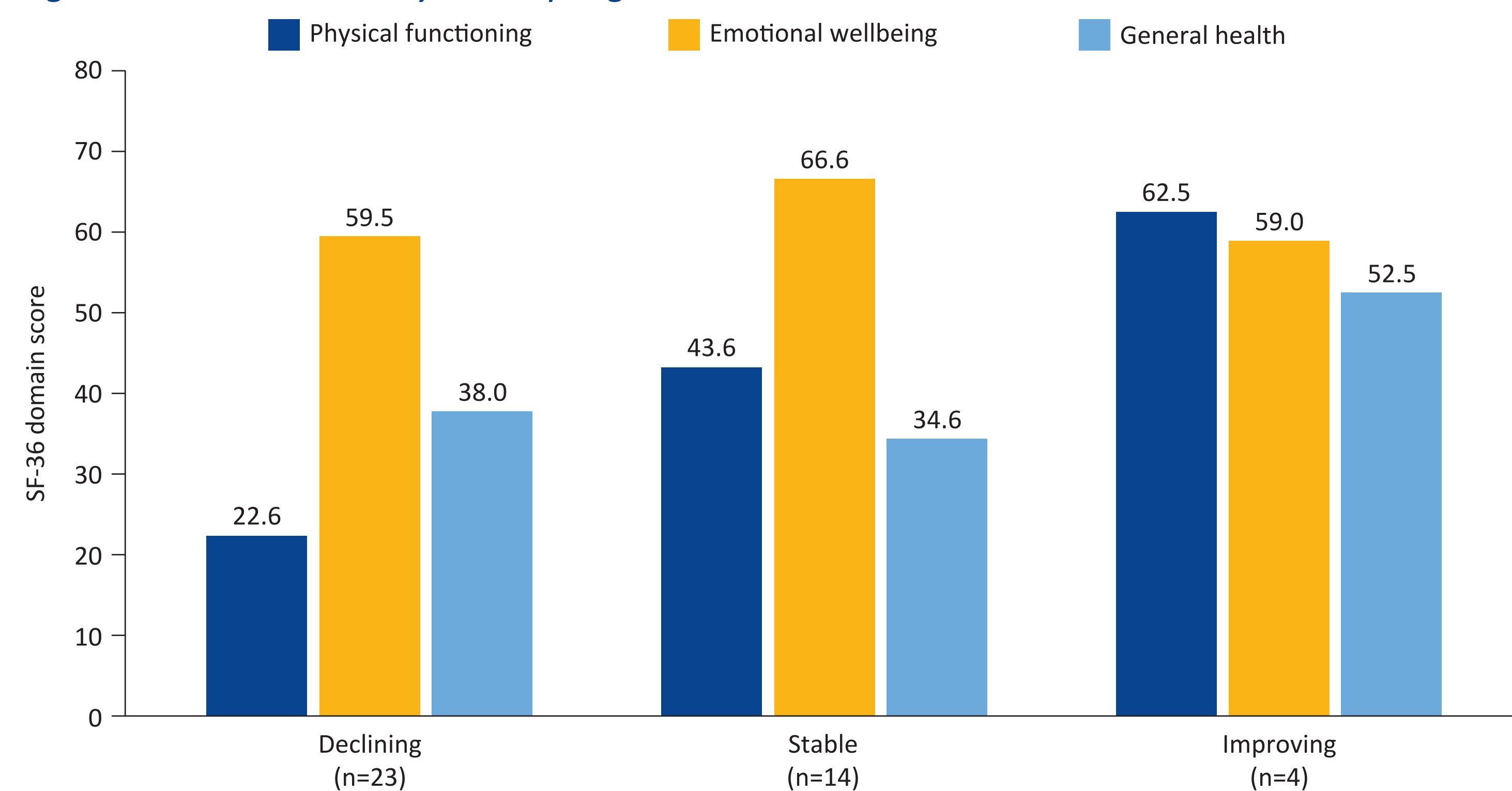
- There was an **increasing additive negative effect on SF-36 general health and physical functioning scores for individuals requiring both physical and ventilator support** compared with either being used alone, whereas emotional wellbeing remained more stable across the groups (Figure 2).

Figure 2. SF-36 scores by physical and ventilator support status



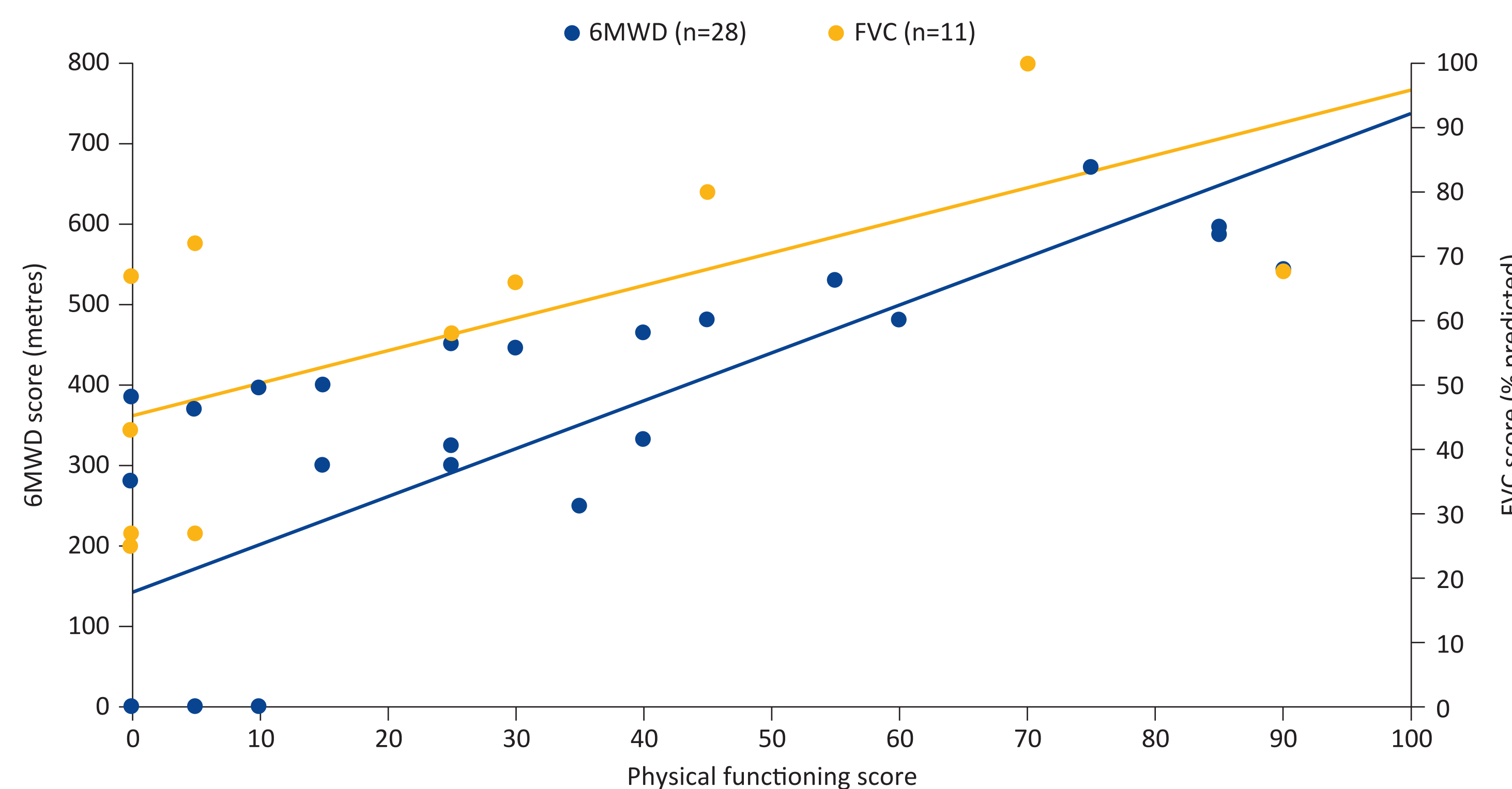
- Generally, participants **self-reporting improvement in disease status had the highest SF-36 scores**, followed by those reporting perceived stabilisation, and then those reporting decline (Figure 3).

Figure 3. SF-36 scores by LOPD progression status



- Symptomatology was found to be individualised, with participants reporting different frequencies and impacts. Most symptoms reported by patients were declining or stable. The proportion of declining versus stable symptoms was relatively similar throughout; however, less frequently reported symptoms (such as headache or speaking difficulties) were more stable than the more frequently reported symptoms (such as muscle weakness or mobility issues; Figure S1, available via QR code).
- **Physical functioning was strongly correlated with both 6MWD ($r=0.80$, $P=4.32e-7$) and FVC ($r=0.66$, $P=0.027$) (Figure 4).**

Figure 4. Association of participant SF-36 physical functioning scores and 6MWD and FVC scores



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