Living with late-onset Pompe disease:

Results from the International Pompe Survey

2024

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Results from the International Pompe Survey 2024
2024 Pompe Survey Report

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Summary

Late-onset Pompe disease (LOPD) is a rare disorder that gradually weakens the muscles, making everyday activities harder over time. To better understand how the disease affects daily life, the IPA/Erasmus MC Pompe Survey was launched in 2002. Over time, the survey has evolved to also capture how treatments improve the real-life experiences of patients and what challenges they still face. This is the first annual report of the survey to be publicly shared. Until now, results have mainly been communicated through scientific publications and presentations. The first part of the report explains how the survey is organized and summarizes the contributions it has made to date. The second part presents the results from the 2024 survey year.

The survey is jointly run by Erasmus Medical Centre in Rotterdam, the Netherlands, and the International Pompe Association (IPA). Since 2002, more than 600 individuals with Pompe disease have participated, with an average of around 175 participants per year. By collecting data from multiple countries over a long period of time, the survey provides unique insights into the impact of Pompe disease and its treatments on patients' lives. These data are used to support reimbursement decisions, guide the development of new treatments, and support patient-informed care for Pompe disease.

The 2024 survey results show that most respondents (92%) were receiving enzyme replacement therapy (ERT). Half (50%) were being treated with alglucosidase alfa (Myozyme). In addition, 41% were using the more recently approved ERTs avalglucosidase alfa (Nexviazyme) and cipaglucosidase alfa combined with miglustat (Pombiliti and Opfolda).

The most common problems reported by patients were muscle cramps (81%), mobility limitations (76%), fatigue (74%), sleep problems (73%), and breathing difficulties (72%). Patients reported lower health-related quality of life than what is normally expected, as measured by various instruments for this (SF-36 PCS 33.2; mean EQ-5D index 0.594; mean EQ-VAS 65.5).

By following participants' experiences over time, the survey provides valuable insight into how emerging treatments, including second-generation ERTs, work in everyday life across different healthcare settings. Continued participation is essential to strengthen this evidence as new therapies become available.

In recent years, the survey has been further updated and professionalized. Customized reports based on survey data are available upon request, on a service-fee basis, to support the ongoing operation of the Pompe Survey. The survey team continues to expand its reach with the goal of including as many patients as possible. Currently, individuals aged 16 years and older with late-onset Pompe disease from 11 countries participate.

If you are interested in participating, scan the QR code for more information or visit our website.

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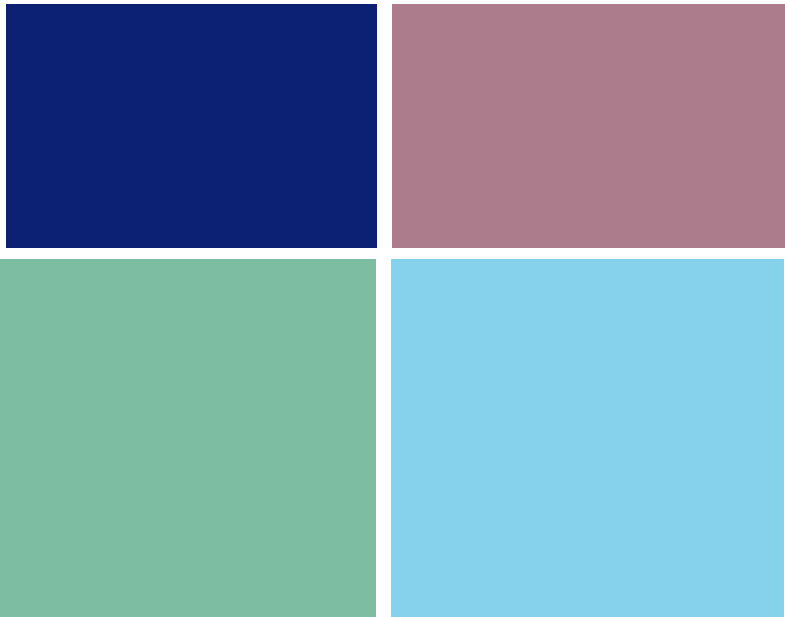


1. Introduction

Pompe disease is a rare, progressive muscle disorder. The most common form is late-onset Pompe disease (LOPD). The first symptoms of LOPD can occur at any age, from childhood to adulthood. Symptoms often start with weakness in the torso and legs, and many people also develop breathing problems. Although the way the disease presents and progresses differs from person to person, Pompe disease can make walking and breathing more difficult, and many people eventually need a wheelchair and/or breathing support. The impact this has on the lives of patients cannot be fully captured by medical tests. Only patients themselves can describe how the disease afffects their everyday lives.

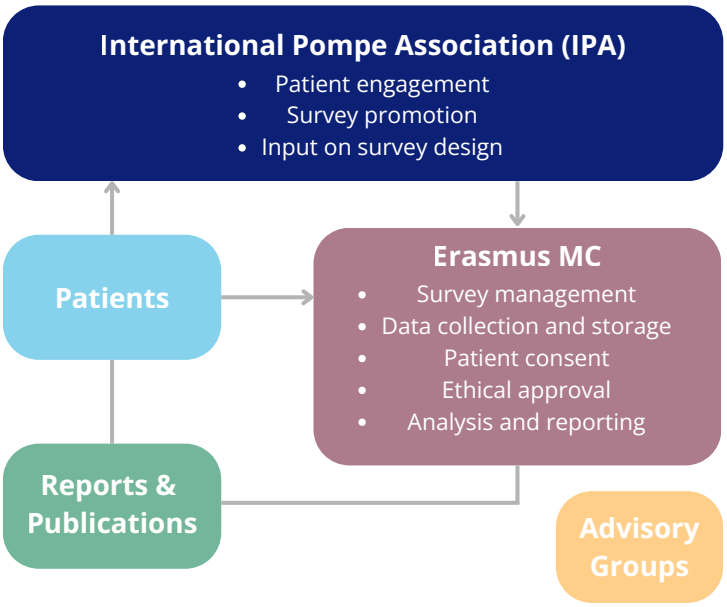
To understand the full impact of Pompe disease on patients' lives over time, the IPA/Erasmus MC Pompe Survey was established in 2002. Each year, patients from around the world are asked to complete a set of questionnaires, which measure the problems they experience and the effect of Pompe disease on their lives. These questionnaires are also called patient-reported outcome measures. With more than 600 participants since it began, the Pompe Survey is the largest study gathering patient-reported outcome data from people living with Pompe disease.

With the introduction of enzyme replacement therapy (ERT) with alglucosidase alfa for Pompe disease in 2006, the survey's focus expanded to include monitoring treatment effects. More recently, next-generation therapies such as avalglucosidase alfa and cipaglucosidase alfa plus miglustat have become available, and potential new treatments, including gene therapy, are on the horizon. As treatment options continue to evolve, it is more important than ever to collect real-world data from the patient perspective to understand how these emerging therapies affect patients' daily lives.

Since its start more than 20 years ago, the Pompe Survey has led to many scientific publications, with insights primarily shared through academic channels. This first annual report brings these insights together for survey participants, patients, stakeholders, clinicians, and other industry leaders.

The first part of the report describes the structure of the Pompe Survey and highlights how it has contributed to understanding the impact of Pompe disease over time. The second part presents key findings from the 2024 survey, including patient-reported information on treatment, physical limitations, fatigue, quality of life, participation in daily life, and the ability to perform daily activities. All results are presented for the full group of participants, regardless of treatment, country, or other characteristics.

2. Survey Organisation and Structure



The IPA/Erasmus MC Pompe Survey is an international study jointly coordinated by Erasmus MC, University Medical Center Rotterdam, the Netherlands, and the International Pompe Association (IPA). This partnership ensures that the survey is both patient-centered and scientifically robust. Erasmus MC is responsible for the day-to-day management of the survey, including patient consent procedures, ethical approval, data storage, and data analysis. The IPA supports patient recruitment and ongoing engagement, raises awareness of the survey, and works with national representatives from patient organizations around the world.

The survey is overseen by a steering board, currently composed of representatives from the IPA and Erasmus MC. The steering board provides strategic direction, ensures that ethical standards are upheld, and reviews requests for data and reports. Clinicians are consulted in an advisory role, for example via the European Pompe Consortium, to provide additional expertise. The structure of the steering board and advisory committee is currently being further formalized.

Researchers and other stakeholders may request reports or analyses from the survey, and under strict conditions also access to the survey data. All requests are reviewed by the steering board to ensure appropriate and responsible use of the data. Contributions associated with these requests help to support the continued operation and broader impact of the Pompe Survey.

Objectives

The Pompe Survey has been designed to gather data about the impact of Pompe disease directly from patients through questionnaires. This can help improve understanding of how Pompe disease affects patients' daily lives. The survey aims to learn what functional limitations and handicap patients themselves experience, how these evolve over time, and how available treatments change/improve this. This information can help doctors in the treatment of patients. Policymakers also need this information to support their decisions, and it can help industry in further developing treatments.

Study Population and Recruitment

Patients aged 16 years and older with a diagnosis of late-onset Pompe disease are eligible to participate in the survey. Patients are actively recruited by the patient organisations in Australia and New Zealand, the United Kingdom, Canada, France, Germany, the Netherlands, the United States, Italy (since 2024), and Spain (since 2025). Patient organisations in other countries, such as Belgium, support the survey by helping to raise awareness. Patients from these and other countries may participate in one of the available languages by contacting the survey team at Erasmus MC.

As part of the recruitment process, patients receive information about the survey and how their data will be used, and are asked to provide informed consent. Until recently, consent was obtained using paper forms sent by mail. From October 2025 onwards, it is also possible to provide consent for the survey electronically.

Data Collection

Participants complete the survey annually through a secure online platform or, if this is not possible, on paper. Each year, they are asked about disease-related problems and limitations (e.g., mobility, breathing, eating, sleeping, and pain), fatigue, health-related quality of life, participation in daily life, ability to perform daily activities, healthcare use, and treatment. When completing the survey for the first time, additional information is collected on demographics, age at symptom onset, diagnosis, and family history. The table below provides an overview of all included elements.

The survey is currently available in six languages: English, Dutch, French, German, Italian, and Spanish. Continuous efforts are made to further professionalize and expand the survey, including translating the survey to new languages and updating the existing surveys to reflect changing patient needs and feedback.

Ethics Approval

The Pompe Survey was approved by the Erasmus MC Medical Ethics Review Committee (NL25654.078.08) and informed consent has been obtained from all participants.

Funding

Research on Pompe disease, including the administration of the Pompe Survey at Erasmus MC is financially supported by ZonMw- the Netherlands Organization for Health Research and Development [project no. 152001005]; the Dutch TI Pharma initiative “Sustainable Orphan Drug Development through Registries and Monitoring (T6-208); “EUCLYD-a European Consortium for Lysosomal Storage Diseases” (health F2/2008 grant agreement 201678); and the Prinses Beatrix Fonds [project no. OP07-08]; MEUSIX [FP7/2007-2013, grant agreement 304999]; SSWO; Colciencias; Genzyme Corp and the International Pompe Association.

3. What we have learned from the Pompe Survey: 2002-2024

2002
Start of the Pompe Survey

The IPA/Erasmus MC Pompe Survey was created in May 2002, aiming to gather data on the natural course of LOPD and the effects of ERT.

2005
Symptoms and severity of untreated LOPD

One of the first results of the early Pompe Survey was describing the symptoms of LOPD before treatment, as well as the age symptoms occurred at[1-4]. Analyses of disease severity found that severity was related to the length of disease rather than age[5].

2006
First ERT approved

2007
Fatigue as a common symptom of LOPD

Fatigue was identified as a common symptom at all levels of disease severity[6].

2011
Survival

Following patients over time made it possible, for the first time, to study how Pompe disease affects survival and how ERT improves this[7].

2012 and further
Reimbursement

Findings from the survey, in particular survival data, have contributed significantly to reimbursement decisions in several countries[8].

2013 and further
R-PAct and Additional Survey Topics

The Pompe Survey helped form the basis for the Rasch-built Pompe-specific Activity scale (R-PAct), which is now used in several clinical trials[9]. The survey has also supported other research projects, including studies on pain and the impact of COVID-19 in people with Pompe disease[10, 11].

2013-2016
Positive effects of ERT

Findings from the survey demonstrated that ERT can improve survival, fatigue, quality of life, and participation in daily life[12-15].

2017
Cost-effectiveness

Data from the survey was instrumental to modelling studies, for example to evaluate newborn screening and the cost-effectiveness of ERT[16-18].

2018
Reduced risk of wheelchair use

Survey data was used to demonstrate that ERT reduces the risk of wheelchair use in LOPD patients[19].

2020
Minimal Clinical Difference

By combining survey responses with clinical data, the survey helped identify what level of improvement in hospital tests such as Forced Vital Capacity or the 6-minute walk test is meaningful for patients[20]. This helps to put the results of new clinical trials into perspective.

2022 & 2023
Second generation ERT released

2024
Expanding the Pompe Survey

The Pompe Survey has now included more than 600 participants over the years and continues to expand to new countries.

4. Participation in the Pompe Survey 2002-2024

Over the years, nearly 600 patients have participated in the Pompe Survey, with an average of 175 participants per year Figure 1). Participation has fluctuated over time, with the introduction of ERT leading to an increase, and the need to reconsent patients following changes to the study protocol negatively affecting participation. Other factors, including varying levels of recruitment efforts in certain years, have also played a role.

The survey was updated in 2009 and again in 2022. In 2009, it was adapted to allow online participation and to evaluate the effects of ERT. In 2022, the survey was further updated to reflect current knowledge of Pompe disease, including new treatment options, and to allow participation from additional countries. In 2022, a protocol update coincided with the introduction of a new online platform, meaning the survey was not distributed that year and the recruitment of new participants was temporarily paused. Following these updates, participation temporarily decreased, as existing participants needed to be informed and asked to provide consent again.

In 2024, the survey reopened to new participants and expansion to Italy began. Recruitment in Spain started in 2025, and efforts are underway to re-engage previous participants. As a result, we expect more than 200 participants to complete the survey in 2026.

Responding Participants by Year

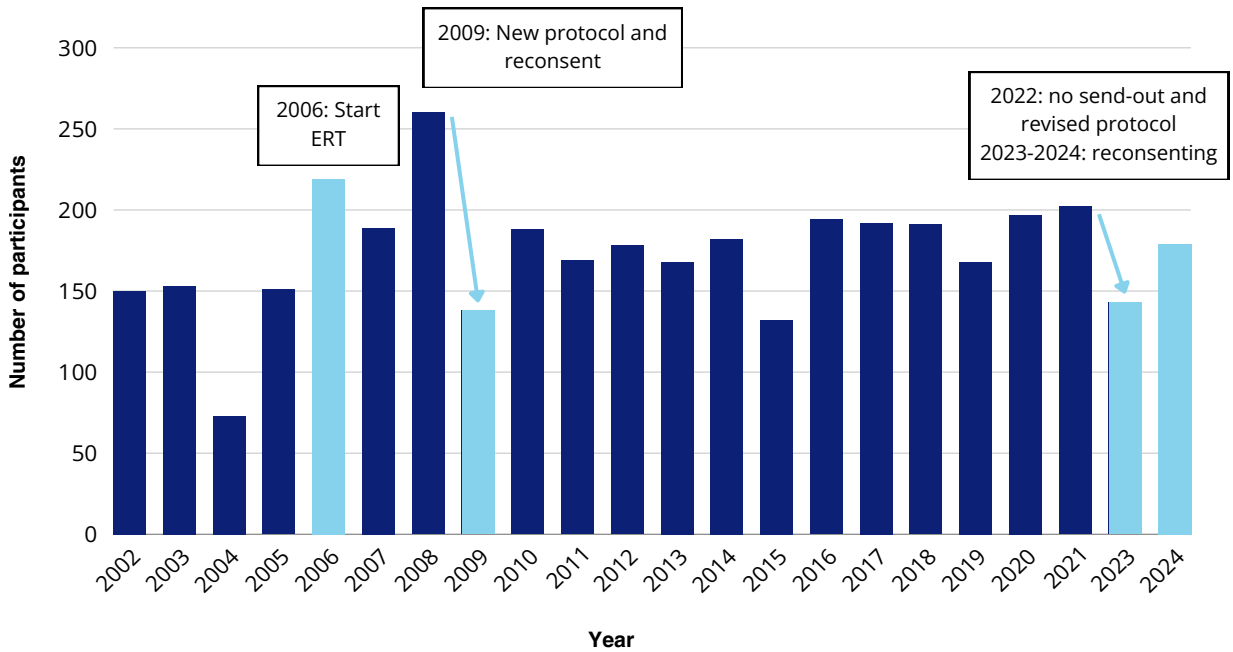


Figure 1: Number of respondents to the IPA/Erasmus MC Pompe Survey, 2002-2024

5. Characteristics of participants in the 2024 Pompe Survey

179 participants from 11 countries

40.5

Median age at diagnosis

55% female participants

Participants by Country

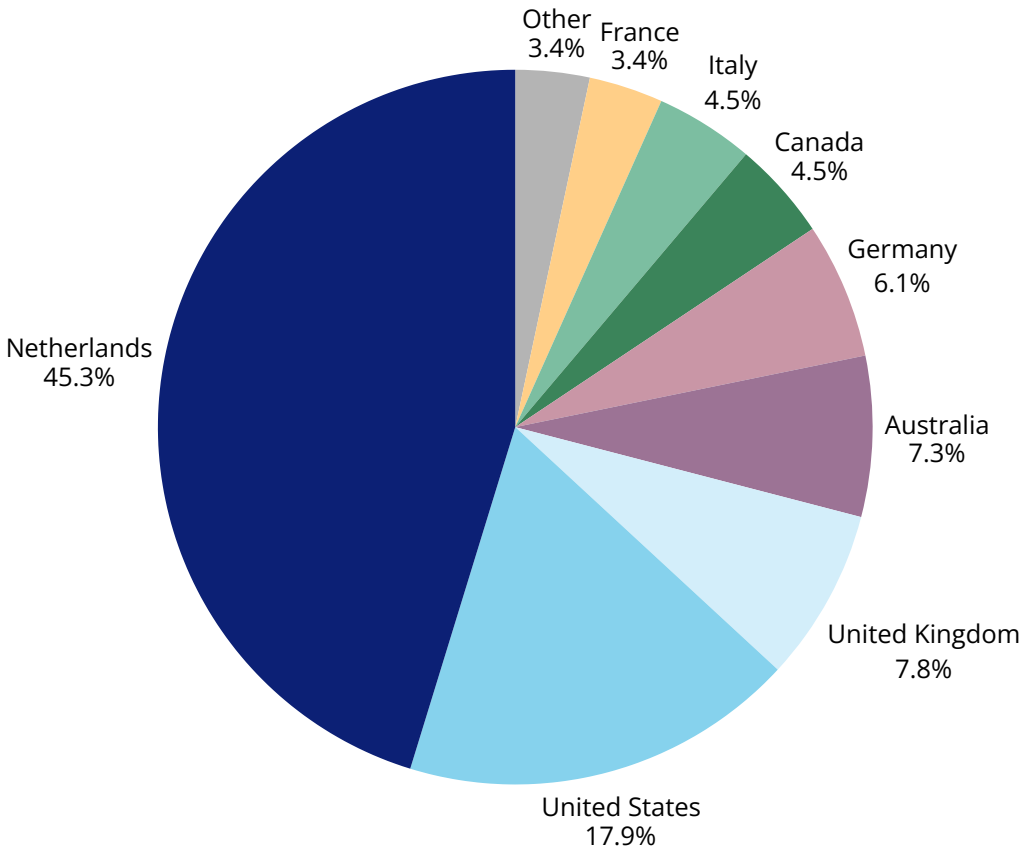


Figure 2: Distribution of Pompe Survey participants by country in 2024.

In 2024, 179 adult patients from 11 countries participated in the Pompe Survey. Most participants were from the Netherlands and United States. Participants were between 21 and 88 years old, were diagnosed with LOPD at a median age of 40.5, and had been symptomatic for a median of 13.2 years. 55% of participants were female.

6. Treatment Received

In 2024, 92.2% of participants reported being treated with ERT at the time of survey completion. An additional 5.0% had received treatment in the past, resulting in a total of 174 participants 97.2%) who had ever been treated (Table 1). Of the 165 patients receiving ERT in 2024, 40.8% were treated with one of the two newly approved ERTs. Additionally, 12.7% of participants reported receiving add-on therapies. Very few participants reported having received gene therapy.

33% of participants reported having been treated with different types of ERT in the past. This is expected to increase as more patients switch to new types of ERT.

Data was also collected on the reasons why participants stopped or paused ERT. The most common reason for pausing ERT was to switch treatments; small numbers of patients cited other reasons such as side effects, reimbursement issues, or travelling as reasons for pausing treatment and resuming at a later date.

Treatment	Number of patients (%)	Median (range)
1. Ever treated with ERT	**174 (97.2%)**	
a. Currently treated	165 (92.2%)	
Years on ERT		15.0 (0-28.0)
b. Stopped ERT	9 (5.0%)	
Years on ERT		7.7 (2.4-18.2)
2. Current ERT	**165 (92.2%)**	
Alglucosidase alfa (Myozyme)	90 (50.3%)	
Avalglucosidase alfa (Nexviazyme)	53 (29.6%)	
Cipaglucosidase alfa + miglustat (Pombiliti+ Opfolda)	20 (11.2%)	
Other	<5	
3. Never treated with ERT	5 (2.8%)	

Table 1: ERT status of participants in the 2024 Pompe Survey

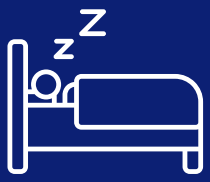
92%
of participants are on ERT

44%
of participants on treatment receive second-generation ERT

7. Problems and Limitations

81% reported muscle cramps in the last week

76% reported mobility problems



73% reported sleeping problems in the last week

72% reported breathing problems

Problems/Complaints		Number of participants (%)
	All participants	**179 (100%)**
Mobility	**Any mobility problems**	**136 (76.0%)**
	Problems walking	99 (55.3%)
	Use a wheelchair • Always use a wheelchair • Years using a wheelchair	71 (39.7%) 30 (16.8%) 9.5 (1-31)
	Cannot walk (even with aids)	37 (20.7%)
	Cannot stand up from lying down alone	25 (14.0%)
	Cannot make transfer alone	25 (14.0%)
Breathing	**Any problems breathing**	**128 (71.5%)**
	Shortness of breath: • After heavy exercise • Lying down • After light exercise • At rest	 80 (44.7%) 65 (36.3%) 63 (35.2%) 19 (10.6%)
	Use ventilation: • Years on ventilation	79 (44.1%) 14 (0-36)
	Pneumonia or other airway infection	23 (12.8%)
	Cannot lie flat on back while sleeping	48 (26.8%)

Table 2: Problems and complaints experiences by participants in the 2024 Pompe Survey

Problems/Complaints		Number of participants (%)
All participants		**179 (100%)**
Pain and cramps	Muscle cramps in the last week	145 (81.0%)
	Other pain in the last week	74 (41.3%)
Sleeping	**Any sleep problems in the last week**	**131 (73.2%)**
	Sleepiness during the day	113 (63.1%)
	Problems concentrating during the day	89 (49.7%)
	Headache when waking up	66 (36.9%)
	Nausea in the morning	22 (12.3%)
Eating	Problems swallowing	58 (32.4%)
	Problems chewing	44 (24.6%)
Hearing	Hearing problems	38 (21.2%)
Daily Tasks	Cannot dress / undress themselves	21 (11.7%)
	Cannot go to the toilet alone	19 (10.6%)
Other Problems	Scoliosis	56 (31.3%)
	Lordosis	57 (31.8%)
	Osteoporosis	52 (29.1%)

Table 2: Problems and complaints experiences by participants in the 2024 Pompe Survey

Muscle cramps were the most common problem reported by participants in the 2024 Pompe Survey (Table 2). 81% of participants reported to have experienced some form of muscle cramp in the last week. The most common locations of muscle cramps were in the shoulders, legs, neck, and back (Figure 3).

Mobility problems were the second most common problem reported by participants in the 2024 Pompe Survey. 55.3% of participants experienced problems walking, while 20.7% reported being unable to walk (Figure 4). The most common problem with walking was stepping onto curbs or walking on uneven surfaces, as well as walking with a waddling gait.

Breathing problems were reported by 72% of participants. Most experienced breathing difficulties during heavy exercise (44.7%), followed by problems when lying down (36.3%) (Figure 5). In addition, 12.8% of participants reported pneumonia or another airway infection in the past year.

Muscle Cramps in the Last Week

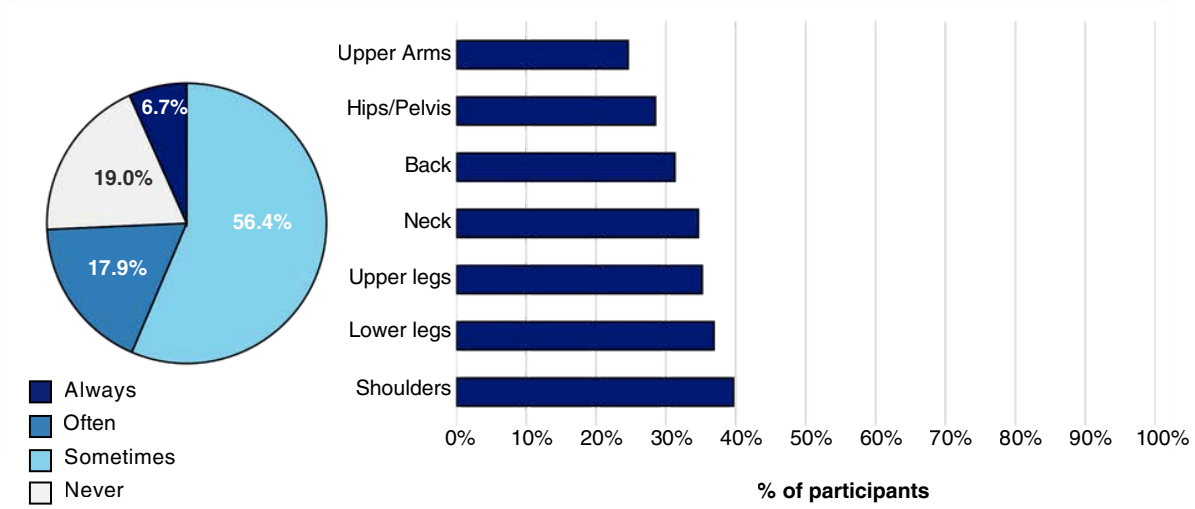


Figure 3: Muscle cramps reported by Pompe Survey participants in the past week, including overall frequency (left) and affected body regions (right).

Walking Problems

Figure 4: Walking problems reported by Pompe Survey participants in 2024, including overall walking ability (left) and the frequency of specific walking-related problems (right).

Breathing Problems

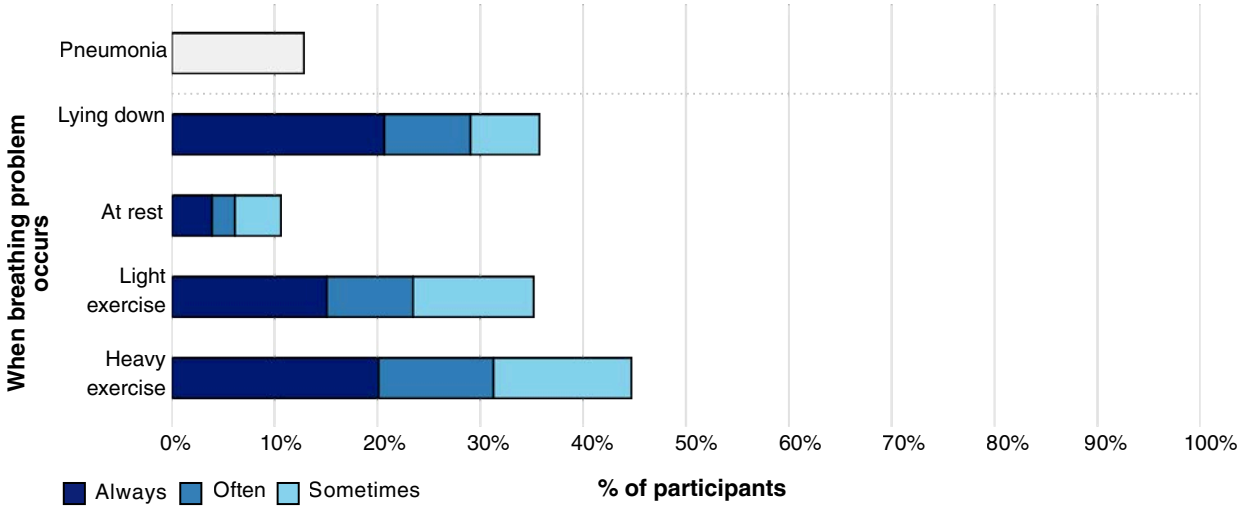


Figure 5: Breathing problems reported by Pompe Survey participants in 2024, by situation in which symptoms occur.

8. Quality of life, participation in life, activities in daily life, and fatigue

Participants completed several questionnaires that measure health-related quality of life, participation in daily life, ability to perform everyday activities, and fatigue. The following sections describe the questionnaires used and the results in more detail.

Quality of Life

SF-36

The **36-Item Short Form Health Survey (SF-36)** is a questionnaire that measures health-related quality of life in eight different areas (domains), from physical functioning to mental health[21].

The questionnaire answers can also be calculated into in two sum scores: the **physical and mental sum score**. All scores range from 0 to 100, with higher scores indicating better perceived health, and a score of 50 representing an average persons' health-related quality of life.

Complete SF-36 data were available for all survey participants in 2024 (Figure 6). Most scores were below average (45–55), indicating a reduced quality of life among participants. The lowest scores were observed for physical functioning, physical role limitations, and the physical summary score, indicating that Pompe disease has a strong impact on physical functioning. In contrast, mental health and the mental sum score were almost at the average level.

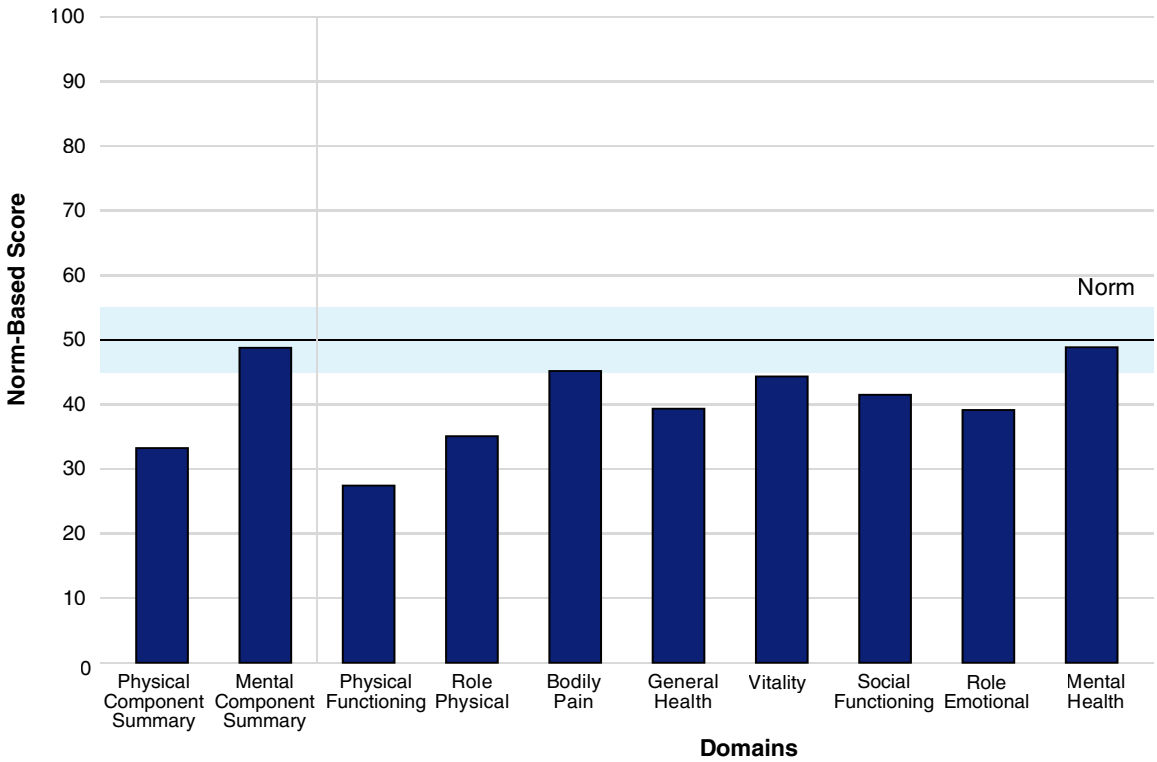


Figure 6: SF-36 domain scores among Pompe Survey participants in 2024 compared with population norms.

Quality of Life

EQ-5D

The **EuroQol 5-Dimension 5-Level (EQ-5D-5L)** is a questionnaire measuring health-related quality of life in five different areas (domains), as well as a health rating from 0 (worst) to 100 (best)[22].

Complete EQ-5D data were available for 178 survey participants in 2024. The most severe problems were reported in the areas of mobility and self-care (Figure 7) In these areas, 15.2% and 9.6% of participants, respectively, reported having "extreme problems or being unable" to perform these activities. The fewest problems were reported in the area of anxiety and depression, where more than half of participants reported having no problems.

The average health ranking score was 65.5, with a broad range (11-100), reflecting the broad range of disease severity of patients. Average scores of the general population in France, Germany, and Spain range from 73 to 75 for comparison[23].

EQ-5D-5L responses can also be combined into a single summary score which is often used in health economics research and ranges from 0 (death) -or lower- to 1 (full health). The average score of survey participants in 2024 was 0.594. In comparison, the average score in the Dutch general population is 0.892[24].

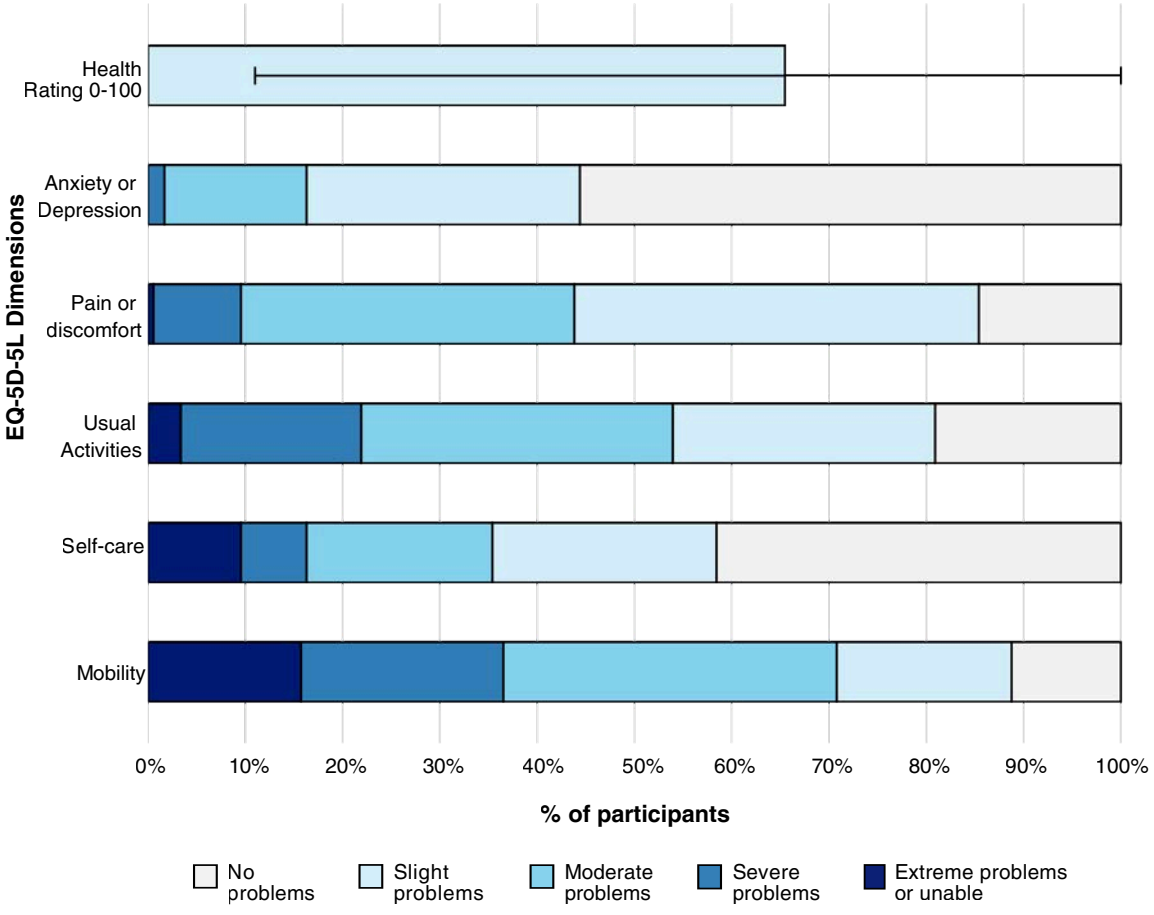


Figure 7: EQ-5D-5L health dimensions and self-rated health among Pompe Survey participants in 2024.

Participation in Life

Rotterdam Handicap Scale

The **Rotterdam Handicap Scale (RHS)** measures participation in daily life activities in people with neuromuscular disorders. Total scores range from 0 to 36, with higher scores meaning better participation in daily life[25].

In 2024, 170 participants completed the RHS, with an average score of 23.3. Most participants reported little to no difficulty with moving around indoors or taking part in indoor leisure activities (Figure 8). Greater difficulties were reported for outdoor mobility, outdoor household tasks, and work or study. Notably, 32% of participants reported that they were no longer able to perform their previous work or studies.

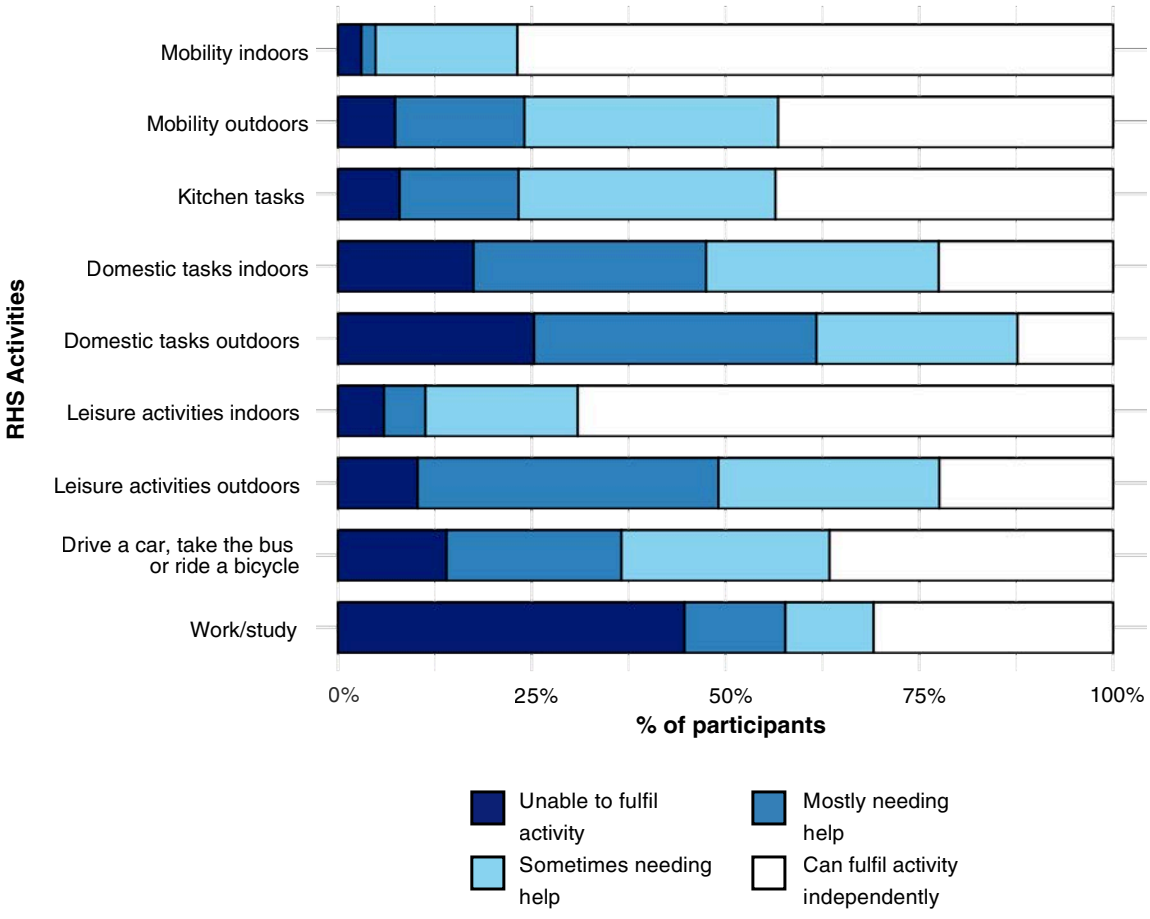


Figure 8: Ability to perform daily activities (RHS) among Pompe Survey participants in 2024.

Activities in Daily Life

R-PAct

The **Rasch-built Pompe-specific Activity scale (R-PAct)** measures how Pompe disease affects daily activities and social participation[26]. Scores range from 0 to 100, with higher scores indicating better functioning.

In the 2024 Pompe Survey, all participants completed the R-Pact, with an average score of 55.4. Most participants reported being able to comb their hair, eat (chew and swallow), and prepare a meal without difficulty (Figure 9). In contrast, most participants reported difficulty or inability with more physically demanding activities, such as running, squatting and standing up, participating in sports, gardening or yard work, and walking at a fast pace.

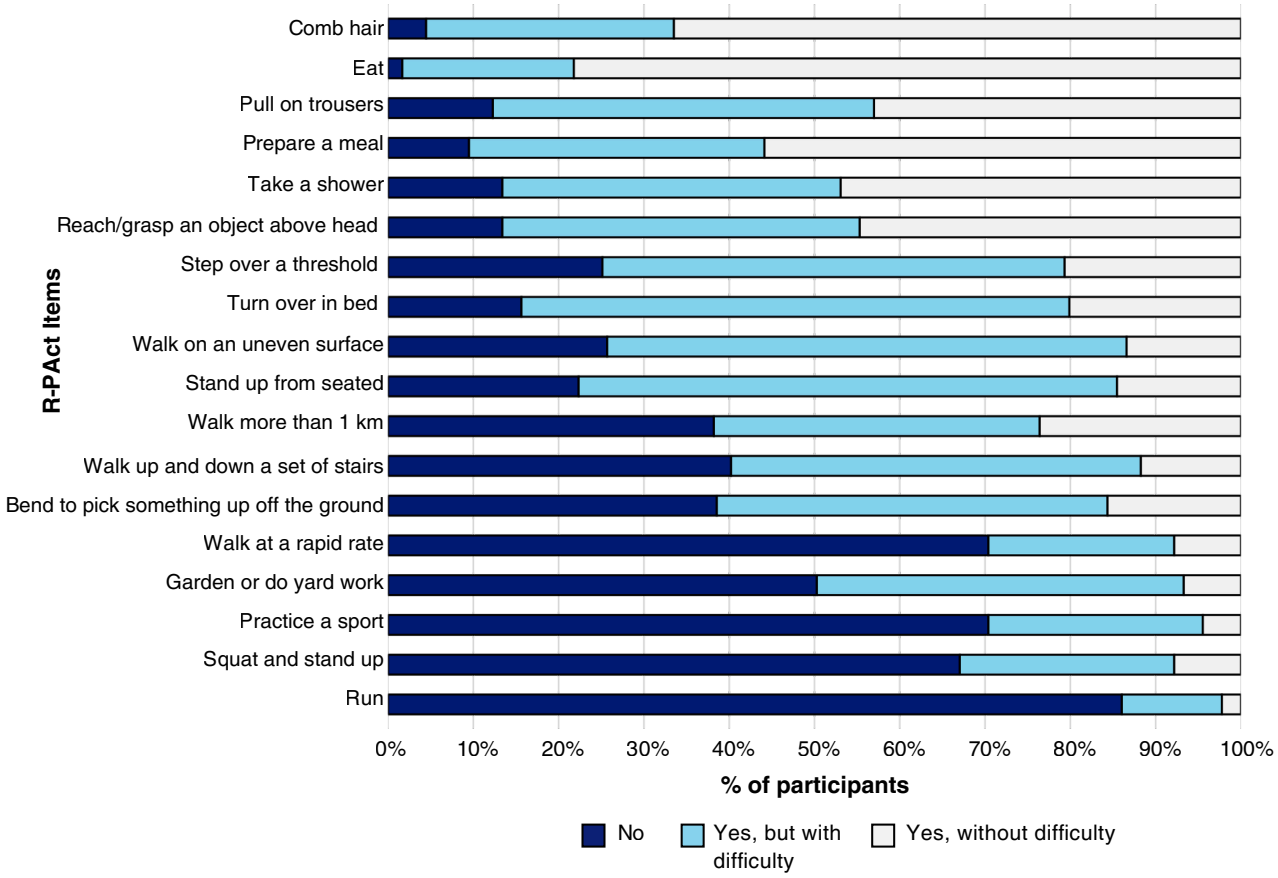


Figure 9: Ability to perform daily activities (R-PAct) among Pompe Survey participants in 2024.

Fatigue

Fatigue Severity Scale

The **Fatigue Severity Scale (FSS)** measures the severity and impact of fatigue (tiredness) on daily life[27]. Scores range from 1 to 7, with higher scores indicating more severe fatigue. Scores of 4 or higher indicate fatigue, while scores of 5 or higher indicate severe fatigue.

The average fatigue score among participants was 4.97, compared with an average score of 2.90 in healthy individuals (Figure 10). Overall, 74% of participants in the survey were classified as fatigued, and 60% as severely fatigued. Fatigue scores were similar among participants who did and did not use a wheelchair and/or ventilation.

These findings show that fatigue is a common and significant problem for people living with Pompe disease, consistent with earlier analyses from the survey[6].

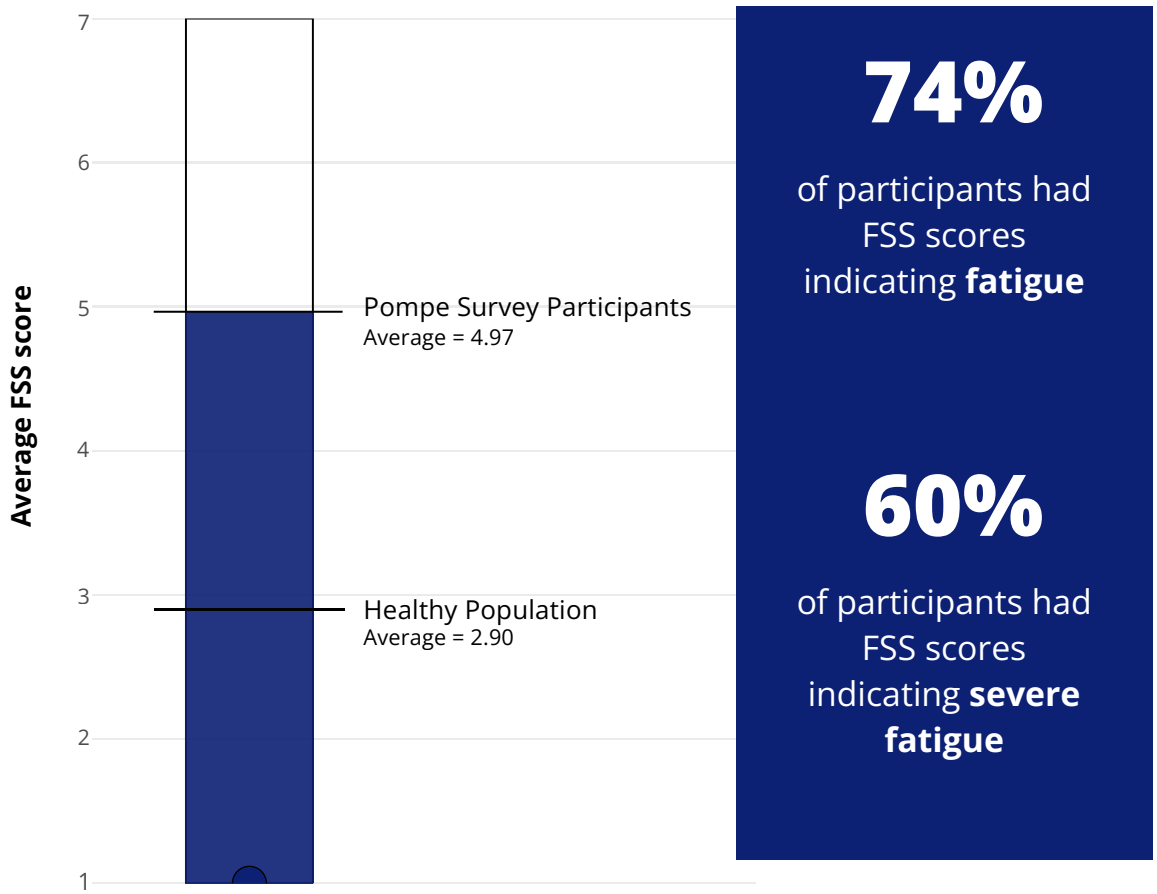


Figure 10: Fatigue Severity Scale (FSS) scores among Pompe Survey participants in 2024 compared with a healthy population.

9. Conclusion

The Pompe Survey has collected data on how the lives of adult patients with LOPD are affected since 2002. This has resulted in a wealth of information from the patient perspective, contributing to reimbursement decisions and development of new treatments. During its long duration, the survey has been updated to capture relevant information, including newly available treatments.

The results of the 2024 Pompe Survey show that, even with treatment, many people living with Pompe disease continue to face daily challenges. Problems with walking, breathing, sleep, cramps, and fatigue remain common, and these issues affect their quality of life and their ability to perform and take part in everyday activities.

By continuing to gather patients' voices, the survey contributes to a better understanding of how treatments work in real life and where more support is most needed. As new therapies are becoming available, the survey will play an important role in showing how these changes affect patients' lives, and whether patients' needs change. With more participants expected to join in the coming years, the survey will provide an even stronger platform to ensure patient experiences guide future care and research.

Over the past years, the study team has worked to revitalize and expand the Pompe Survey. Previous participants were recontacted and invited to continue their involvement, while national patient organizations supported outreach to new patients. In 2024, the survey was rolled out in Italy and in 2025 recruitment began in Spain and Belgium. E-consent has also become available in Fall 2025, making it easier and faster for new participants to join the survey moving forwards. Additional efforts to engage patients include hosting webinars, sharing information on social media, and building connections with new patient organizations and their networks.

With these developments, we expect the survey to grow further, with the aim of reaching an annual response of 200+ participants in 2026. As the survey continues to expand, the Pompe Survey team looks forward to welcoming new patients and sharing insights from the survey with the patient community as well as policy makers and industry. Customized reports from the survey are available upon request, and opportunities for research collaboration are welcomed.

10. Acknowledgements

The continued success of the IPA/Erasmus MC Pompe Survey is only possible thanks to the dedication and support of its participants. Despite changing symptoms, treatment regimens, and life circumstances, participants continue to share their perspectives and contribute to research that places patients at its center. This report would not be possible without you.

As new therapies become available, the Pompe Survey team remains committed to expanding and adapting the survey to capture patient perspectives and share these insights with the communities they come from. Your continued participation helps ensure that the experiences of people living with Pompe disease remain visible, understood, and central to future research and care.

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