International Pompe Association

IPA Update Q1 2026

Dear reader,

We are proud to present our report on the International Pompe Survey 2024, in collaboration with Erasmus MC. “Living with late-onset Pompe disease” shows how Pompe affects patients' everyday lives and quality of life. With this report, we can also express our gratitude to the 179 patients who were willing to get involved and complete the extensive questionnaire. Here you can see how their data is incorporated into meaningful values that influence the course of the disease. We would like to thank them for their commitment.

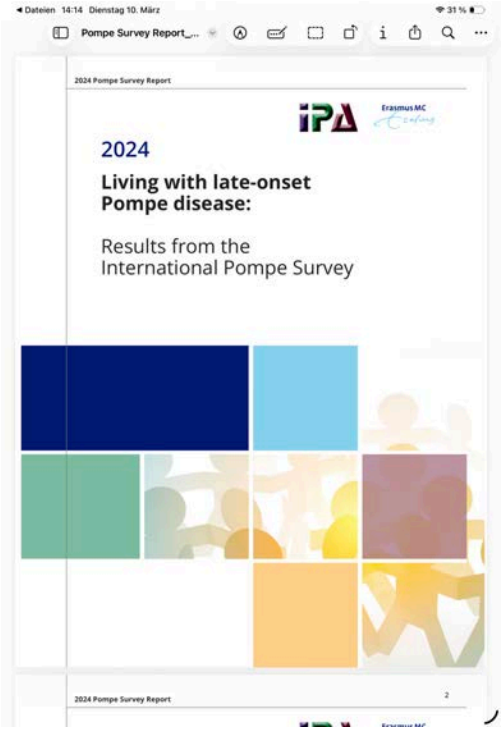
Your IPA Board

IPA Community

International

Survey: Growing global participation and next steps

The long-running international Pompe Survey by IPA and Erasmus MC, Rotterdam, continues to expand its reach and impact. Established in 2002, the survey is unique in the Pompe community: it is independent and not initiated by a pharmaceutical company, making it one of the few long-term, neutral data sources on the lived experience of people with Pompe disease.



Participation increased again. 2024 there were 179 participants, 2025, 202 participants filled in the questionnaire. The survey continues to be strongly represented internationally.

In 2025, the largest groups of respondents came from the Netherlands (74), the United States (24), the United Kingdom (17), Spain (17), Germany/Austria/Switzerland (16), and Australia (15). Participation also increased in several countries, particularly Spain and Canada.

The consent and re-consent process has recently been significantly streamlined through the introduction of digital consent using ValidSign, making participation faster and easier for patients. The survey is continuing to broaden its international reach. New participants from South Africa are expected to join the study soon, further strengthening its global perspective on living with Pompe disease.

Over the past year, several key project milestones have been reached: The organizational structure of the survey has been formalized, including a survey board and external advisors. The survey rollout in Italy and Spain has been completed. Electronic informed consent (e-consent) has been implemented and approved by the Medical Ethics Committee.

Next steps include expanding the survey into additional languages, exploring new options for securely sharing individual results, and strengthening the survey's visibility. After more than two decades of continuous data collection, the Pompe Survey remains a vital resource for understanding how Pompe disease affects patients over time. Its independent, patient-focused design continues to provide

valuable real-world insights that support research, clinical understanding, and advocacy across the global Pompe community.

Click here for the 2024 report of the survey

International

Membership fee – lowering the barrier to participation

In a move to broaden organizational participation, IPA members at the Annual General Meeting in November 2025 have unanimously voted to eliminate the annual membership fee of €100. The discussion centered on whether reducing or fully removing the fee would encourage more organizations to join and engage in IPA activities. It emerged that there also had been some inconsistency in awareness of the fee. The board confirmed that the existing statutes already allow for the fee to be set to zero.

USA

Posthumous honor for Tiffany House

The Rare Voice Awards, presented by Rare Disease Legislative Advocates (RDLA), honors advocates who help amplify the voice of the rare disease community in state and federal policy in the U SA. The 2025 RareVoice Awardees has been featured at Rare Disease Week on Capitol Hill, Washington USA on February 24-26, 2026.

The IPA is thankful for Tiffany's recognition - the 2025 Everylife RareVoice Award Recipient for State Advocacy: Patient Advocate or Organization for her role in Texas Newborn screening as Chair of the AMDA.

watch here: https://lnkd.in/e-3zUR5r

International

This April, we’re moving for Pompe. Will you join us?

The International Pompe Association (IPA) is proud to announce the Run, Walk or Roll 2026 virtual event, taking place from April 1–30 in celebration of International Pompe Day on April 15th. This year’s event brings together patients, caregivers, healthcare professionals, and advocates from around the world under one shared message: Every Move Counts.No matter where you are or how you move – running, walking, or rolling – your participation contributes to a growing global movement that keeps Pompe disease visible and the community connected.
How to participate:

→ Register

→ Log your distance between April 1–30

→ Share your story on social media and inspire your network

→ Support the cause through our fundraising page

We invite our network, patients, families, clinicians, industry partners, and allies to take part and help amplify awareness for Pompe disease on a global scale.

More information is coming soon: https://worldpompe.org/international-pompe-day/

Science and Industry

USA

Positive Phase 1b results for ABX1100 in LOPD

Aro Biotherapeutics, Philadelphia, announced encouraging topline results for ABX1100, a muscle-targeted siRNA therapy for late-onset Pompe disease. The study in 9 LOPD patients showed ~62% GYS1 mRNA knockdown, ~2.5% mean FVC improvement, and reductions in key biomarkers. ABX1100 was well tolerated with no serious adverse events. Full data will be presented at a future scientific conference.

Source: https://www.arobiotx.com/abx1100pressrelease#abx1100-phase-1...

Japan

First patients enrolled in global phase 2 trial of oral Pompe Therapy

Shionogi has enrolled the first patients in Esprit, a global Phase 2 trial evaluating S-606001, a potential first oral substrate reduction therapy (SRT) for late-onset Pompe disease (LOPD). The 52-week, placebo-controlled study will assess safety and preliminary efficacy of S-606001 add-on to ERT in adults with LOPD across the U.S., EU and UK. S-606001 works by inhibiting GYS1 to reduce glycogen buildup in muscle.

Source: https://www.shionogi.com/global/en/news/2026/03/20260319_11....

France

Presymptomatic Pompe: French data calls for clearer guidelines

A French working group is calling for binding guidelines for dealing with presymptomatic LOPD findings. This is based on data from the neuromuscular center at the Raymond-Poincaré University Hospital Raymond-Poincaré near Paris, which reports on three people with genetically confirmed LOPD who were diagnosed by chance long before the first symptoms appeared and did not require enzyme replacement therapy for years. During follow-up care, early changes were observed in some cases in tests and imaging of the muscles. However, clinical symptoms did not appear until much later. The authors point out that for people who are only genetically abnormal and do not yet have any symptoms, there is currently no clear data on the optimal time for treatment. The current recommendations of the European Pompe Consortium (EPOC) from 2024 stipulate that ERT for LOPD should generally only be started in symptomatic patients and also emphasize the importance of muscle MRI findings in detecting early muscle involvement and supporting the decision to initiate therapy. At the same time, the study shows that more and more LOPD cases are being detected very early on: through Improved enzyme tests using dried blood, genetic panels, and newborn screening in some countries. This shifts the focus toward structured follow-up examinations with clinical, functional, and radiological tests in order to find the best possible time to start therapy.

https://www.sciencedirect.com/science/article/abs/pii/S00353...

Serbia

Even a short break in treatment has lasting consequences for Pompe patients

A research team at the Neurology Clinic of the University Clinical Centre of Serbia has published findings that shed important new light on what happens when patients with a rare muscle disease called late-onset Pompe disease (LOPD) must temporarily stop their life-sustaining treatment.

The study, published in Trends in Molecular Biology (Volume 5, September 2025, pp. 143–154), followed five patients who had been receiving enzyme replacement therapy (ERT) for an average of more than four years — until an administrative supply disruption forced an involuntary treatment gap of approximately 54 days (about two months) in April and May of 2023.

The study tracked patients at multiple time points: before the interruption, immediately before restarting treatment, and again at two and six months after resuming therapy. The team measured walking ability, muscle strength, lung function, and quality of life.

The key findings were: Walking ability declined during the treatment break. On average, patients could walk about 34 metres less in a six-minute walking test after the interruption. Recovery was slow and only reached pre-interruption levels around the six-month mark after restarting treatment.

Breathing was the most vulnerable function. Lung capacity (forced vital capacity) deteriorated noticeably during the gap — and even six months after restarting ERT, it had not returned to baseline levels. This is particularly concerning, since respiratory failure is a leading cause of serious complications in Pompe disease.

Muscle strength held relatively stable during the interruption, which is consistent with findings from other research groups.

Quality of life dropped — specifically the patients' ability to carry out physical roles in daily life — and improved again within two months of restarting therapy, though again not fully recovering to prior levels.

Dr. Stojan Perić and colleagues emphasise that their results confirm what clinicians have long suspected but rarely been able to document directly: even a relatively short break in ERT — in this case, less than two months — can cause real, measurable harm that may not be fully reversible.

"These findings underscore the crucial importance of maintaining continuous ERT therapy to prevent the rapid progression of the disease that would occur without treatment," the authors write.

The study also highlights how important it is to diagnose Pompe disease early and accurately. Two of the five patients had gone undiagnosed for years because their symptoms were non-specific — they were only identified through advanced genetic testing (whole-exome sequencing).

For patients living with late-onset Pompe disease, the message from this research is clear: uninterrupted access to enzyme replacement therapy is not a luxury — it is a medical necessity. Even brief supply disruptions can have consequences that linger for months or longer, particularly for lung function. Healthcare providers, policymakers, and supply chain managers should treat ERT continuity for rare disease patients as a high priority.

Source: Impact of discontinuation and reintroduction of alglucosidase alpha in patients with late-onset Pompe disease. Trends in Molecular Biology (Trendovi u molekularnoj biologiji), Vol. 5, No. 5, September 2025

International

How much FVC improvement do Pompe patients actually notice?

In late-onset Pompe disease (LOPD), lung function is commonly measured using the FVC test. Until recently, it was unclear how much improvement patients would actually notice in daily life — the so-called clinically meaningful threshold (CMT).

Using data from the COMET study (99 adults), researchers found that an individual FVC improvement of at least 3% is needed for a patient to perceive a difference, while 2.1% is sufficient for group comparisons. These thresholds help better interpret study results — not only for Pompe disease, but for other neuromuscular diseases as well.

Source: COMET Study (comparing Avalglucosidase alfa / Nexviadyme® vs. Alglucosidase alfa / Myozyme®)
https://pmc.ncbi.nlm.nih.gov/articles/PMC12357813/

The Netherlands

ERT for lysosomal storage disorders: limitations and new directions

While enzyme replacement therapy (ERT) has significantly improved treatment for lysosomal storage disorders such as Pompe disease, its limitations have become clear after two decades — according to researchers at Erasmus MC. As patients live longer, new disease features emerge and certain tissues, such as the central nervous system, remain out of reach. Next-generation therapies are being developed to address these gaps.

In parallel, Erasmus MC researchers argue in a published letter that independently managed patient registries are essential. Manufacturer-owned registries — such as those operated by Sanofi — do not allow cross-product comparisons and lead to data fragmentation, hampering a true understanding of available treatment options. The International Pompe Survey, independently collecting patient-reported outcomes since 2002, is cited as a model.

Source: https://www.springermedizin.de/real-world-evidence-for-pompe-disease-remains-fragmented-comment/50644908

Rare Diseases Spotlight

Germany/USA

New lysosome atlas reveals cause of rare neurological disease

Together with colleagues from Stanford University, USA, researchers at the Leibniz Institute on Aging (FLI), Jena, Germany, have, for the first time, created a comprehensive cell type-specific atlas of lysosomes in the brain, the cell organelles which are responsible for degradation and recycling processes. The study shows that lysosomes in neurons differ significantly from those in other brain cells. Particularly striking is the previously little-noticed protein SLC45A1, which plays a central role in neuronal lysosomes. Mutations in this protein lead to a previously unclear neurological disease that can now be classified as a lysosomal storage disorder.

Source: https://idw-online.de/de/news864841

UK

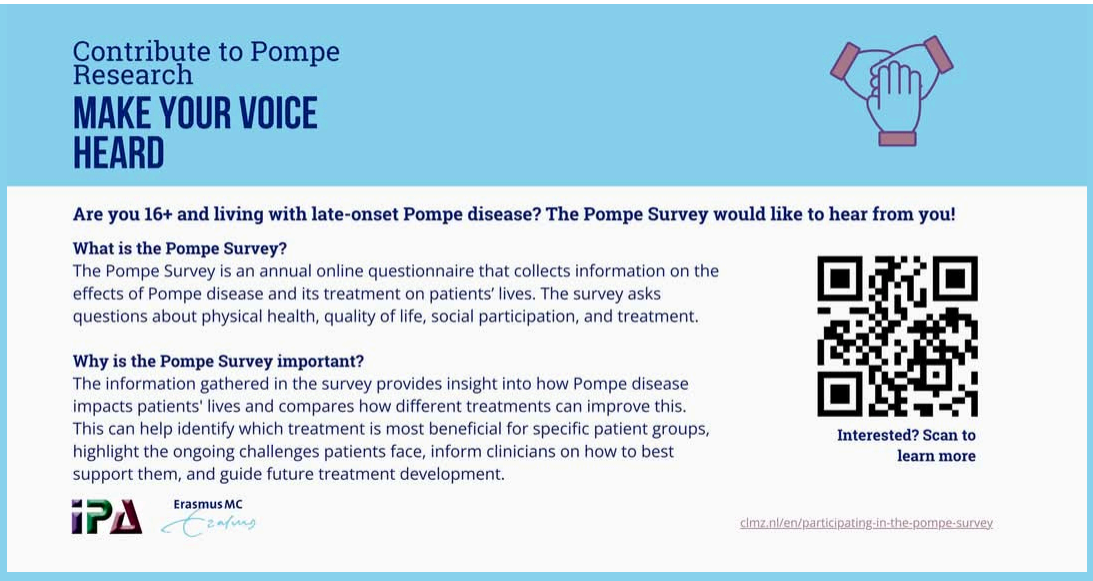
Boy with rare condition amazes doctors after world-first gene therapy for MPSII

A three-year-old boy has astounded doctors with his progress after becoming the first person in the world with his devastating disease to receive a ground-breaking gene therapy. Oliver Chu has a rare, inherited condition called Hunter syndrome - or MPSII - which causes progressive damage to the body and brain. In the most severe cases, patients with the disease usually die before the age of 20. Due to a faulty gene, before the treatment Oliver was unable to produce an

enzyme crucial for keeping cells healthy. In a world first, medical staff in Manchester have tried to halt the disease by altering Oliver's cells using gene therapy. Prof Simon Jones, who is co-leading the trial tells the BBC: "I've been waiting 20 years to see a boy like Ollie doing as well as he is, and it's just so At the centre of this remarkable story is Oliver - the first of five boys around the world to receive the treatment - and the Chu family, from California, who have put their faith in the medical team at Royal Manchester Children's Hospital.

The BBC has followed Oliver's story for more than a year - including how scientists in the UK first developed the pioneering gene therapy and how the medical trial they are conducting almost didn't get off the ground due to lack of funds.

Source: https://www.bbc.com/news/articles/c5y0y56×6ve



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