International Pompe Association

IPA Update Q2 2026

IPA Community

International

English edition of rabbit milk, hamster cells & singing quails is now available

The International Pompe Association is pleased to share that the English version of Rabbit Milk, Hamster Cells & Singing Quails, written by Erik van Uden and Maryze Schoneveld van der Linde, is now available as a PDF.

The book was first published on October 28, 2024. Maryze Schoneveld van der Linde sadly passed away on May 14, 2025. One of her great wishes was to make this important story available in English, so people around the world could learn more about the history, strength, and determination of people living with Pompe disease.

As Maryze wrote: “The book is a story of many different people, their cooperation, teamwork,perseverance, sorrow, and hope for a future.” The book tells the story of people affected by Pompe disease: individuals and families,

researchers, physicians, cooperation across disciplines, and the persistence that helped shape the development of treatment. It is a story of hardship and hope, above all a record of what shared commitment can make possible. The IPA warmly thanks Els Wiegent and Erik van Uden. Els made the English translation possible and helped fulfill Maryze's great wish. Erik supported the translation and helped make the book accessible to an international audience.

The English PDF version of Rabbit Milk, Hamster Cells & Singing Quails is now available on the IPA website, worldpompe.org.

International

Membership fee – lowering the barrier to participation

Reminder: In a move to broaden organizational participation, IPA members at the Annual General Meeting last year have unanimously voted to eliminate the annual membership fee of €100. The board confirmed that the existing statutes already allow for the fee to be set to zero.

Science and Industry

International

Home infusions can be safely implemented

Recent literature on home treatment in Pompe disease, published on PubMed showshome infusions can be safely implemented. The focus is whether enzyme replacement therapies can be administered at home in suitable individuals.The data presented suggest that home infusions are possible under clearly defined conditions and with trained staff, without increasing safety risks.

For many people living with Pompe disease, this is a significant care issue. Regular hospital-based infusions can place a burden on daily life, work, family life and mobility. The report is therefore highly relevant from a care and quality-of-life perspective.

Source: https://pubmed.ncbi.nlm.nih.gov/41631150/

France

Positive Baby-COMET Results for Nexviazyme in IOPD

Sanofi has announced positive results from the Phase 3 Baby-COMET clinical trial of Nexviazyme (avalglucosidase alfa) in infants with infantile-onset Pompe

disease (IOPD). The trial met its primary endpoint: treatment-naïve infants aged six months or younger were alive and free of invasive ventilation after 52 weeks. Secondary endpoints were also met, including ventilator-free survival at 12 and 18 months of age. Sanofi plans to submit the data to support a US regulatory application for IOPD in the second half of 2026. The results will be presented at ICNMD 2026 in Florence.

Source: https://www.sanofi.com/en/media-room/press-releases/2026/202...

USA

AskBio doses first participant in AB-1009 gene therapy trial

AskBio has treated the first participant in its Phase 1/2 PROGRESS-GT LOPD clinical trial with AB-1009. AB-1009 is an investigational AAV gene therapy for adults with late-onset Pompe disease (LOPD).

The initial aim is to assess safety and tolerability. Recruitment in the United States is ongoing. For the Pompe community, this is a relevant step because gene therapies may one day offer an approach beyond regular enzyme replacement therapy (ERT). At the same time, AskBio emphasizes that AB-1009 is not approved and that its safety and efficacy have not yet been fully evaluated.

Source: https://www.askbio.com/first-participant-dosed-in-gene-thera...

International

Shionogi launches Phase 2 trial of oral substrate reduction therapy

Shionogi has enrolled the first participants in the global Phase 2 ESPRIT clinical trial. The trial is investigating S-606001, an investigational oral substrate reduction treatment for adults with LOPD.

This approach differs from enzyme replacement therapy. Instead of replacing the missing enzyme, it aims to reduce glycogen formation. Shionogi describes S-606001 as a potential first oral substrate reduction treatment for Pompe disease.

The investigation is evaluating S-606001 as an add-on to existing ERT. If supported by evidence, it could add a new option to the Pompe treatment landscape.

Source: https://www.shionogi.com/us/en/news/2026/03/shionogi-announc...

USA

BioMarin acquires Amicus Therapeutics and adds Pombiliti + Opfolda to its portfolio

BioMarin Pharmaceutical Inc.has completed its acquisition of Amicus Therapeutics. As part of the transaction, Pombiliti plus Opfolda becomes part of BioMarin's commercial portfolio.

The combination of cipaglucosidase alfa and miglustat is approved for adults with late-onset Pompe disease who are not sufficiently benefiting from their current enzyme replacement therapy. The transaction is valued at approximately USD 4.8 billion.

For patient organizations, the acquisition is relevant because responsibilities, contacts and strategic priorities in the Pompe treatment landscape may change.

Source: https://investors.biomarin.com/news/news-details/2026/BioMar...

France/USA

Généthon and AskBio enter licensing agreement for Pompe gene therapy

The French research and biotherapy organization Généthon has entered into a licensing agreement with AskBio for the development of an investigational gene therapy for Pompe disease. According to the announcement, AskBio will take clinical development forward. The start of the clinical trial was planned for early 2026.

The news is noteworthy because it shows how academic and non-profit research can connect with industrial development in rare diseases. For Pompe disease, this strengthens the international pipeline of genetic treatment approaches.

Source: https://www.genethon.fr/genethon-conclut-un-accord-de-licenc...

International

Real-world data: switching to newer enzyme replacement therapies appears feasible

A multicenter real-world analysis published in 2026 examined the switch of people with late-onset Pompe disease to newer enzyme replacement therapies, including avalglucosidase alfa and cipaglucosidase alfa plus miglustat. The

analysis concludes that switching between different ERT products appears feasible in clinical practice. In many participants, it was associated with clinical stability. Since real-world data have so far been limited, this work provides relevant information for physicians and the Pompe community when a treatment switch is being considered.

Source: https://link.springer.com/article/10.1007/s00415-026-13689-1

Germany

New study from Bochum may support more individualized Pompe therapy

A report from North Rhine-Westphalia highlights a Bochum study that could help support more individualized treatment in Pompe disease. The background is that enzyme replacement therapy can slow disease progression, but it does not work equally well in all people with Pompe and does not fully restore muscle or organ function. The study looks at factors that may help explain differences in treatment response.
For the Pompe community, this is relevant because more personalized treatment decisions could support better monitoring, more targeted intervention and more informed decisions about treatment changes in the future.
Source: https://www.bg-kliniken.de/universitaetsklinikum-bergmannshe...

Japan

Newborn screening: Japanese analysis shows benefits and high costs

An analysis published in 2026 in the International Journal of Neonatal Screening examined the cost-effectiveness of universal newborn screening for infantile-onset Pompe disease in Japan.
The result: screening can provide substantial health benefits for children with the infantile form of the disease. It is also associated with significantly higher costs.
For patient organizations, newborn screening often remains a key issue because early treatment initiation can be critical in infantile-onset Pompe disease. At the same time, the analysis shows that health policy decisions must consider medical and economic factors.
Source: https://www.mdpi.com/2409-515X/12/2/21

International

European roadmap: Pompe gene therapy requires patient-centered centers

A roadmap published in 2026 describes the requirements for a patient-centered approach to future gene therapy centers for Pompe disease.
With the involvement of European experts, the paper identifies key elements: early and reliable diagnosis, comprehensive patient evaluation, careful selection of suitable candidates and structured monitoring after treatment.
For patient organizations, this work is particularly relevant because it shows that gene therapy is a complex care process, not only a product. Patient perspectives, follow-up care and safety structures must be integrated from the beginning.
Source: https://pmc.ncbi.nlm.nih.gov/articles/PMC12906549/

International

EU project DREAMS uses AI to support new therapies for rare muscle diseases

The European research project DREAMS aims to accelerate the discovery of new treatments for rare neuromuscular diseases. The project combines AI-supported drug discovery, stem cell data and phenotypic screening.

Pompe disease is one of the five target diseases. For Pompe research, this is relevant because the approach does not focus only on a single drug candidate. It aims to create a platform that can identify promising candidates more quickly.

Such technology-driven projects could help expand the early research pipeline in the long term.

Source: https://cordis.europa.eu/article/id/463169-accelerating-discovery-of-novel-treatments-for-rare-diseases/fr

International

Avenue Therapeutics licenses ATX-04 for Pompe disease

Avenue Therapeutics has entered into an exclusive worldwide license agreement with Duke University for ATX-04, also known as clenbuterol, for the treatment of Pompe disease. ATX-04 is an orally administered selective β2-adrenergic agonist being developed as a potential add-on treatment to enzyme

replacement therapy (ERT).
According to Avenue, clinical data generated at Duke University in people with Pompe disease receiving ERT showed improvements in six-minute walk distance, respiratory muscle strength, muscle glycogen burden, GAA activity and disease-relevant gene expression. The company plans to move ATX-04 into a late-stage clinical development program, initially focusing on Pompe disease as an adjunct to standard-of-care ERT.
Under the agreement, Avenue receives exclusive worldwide rights to develop and commercialize ATX-04 for Pompe disease and potentially other neuromuscular indications. Avenue will also take over Duke's existing clinical and regulatory assets for ATX-04, including the investigational new drug application and the U.S. FDA orphan drug designation for Pompe disease.
Source: https://ir.avenuetx.com/news-events/press-releases/detail/99...

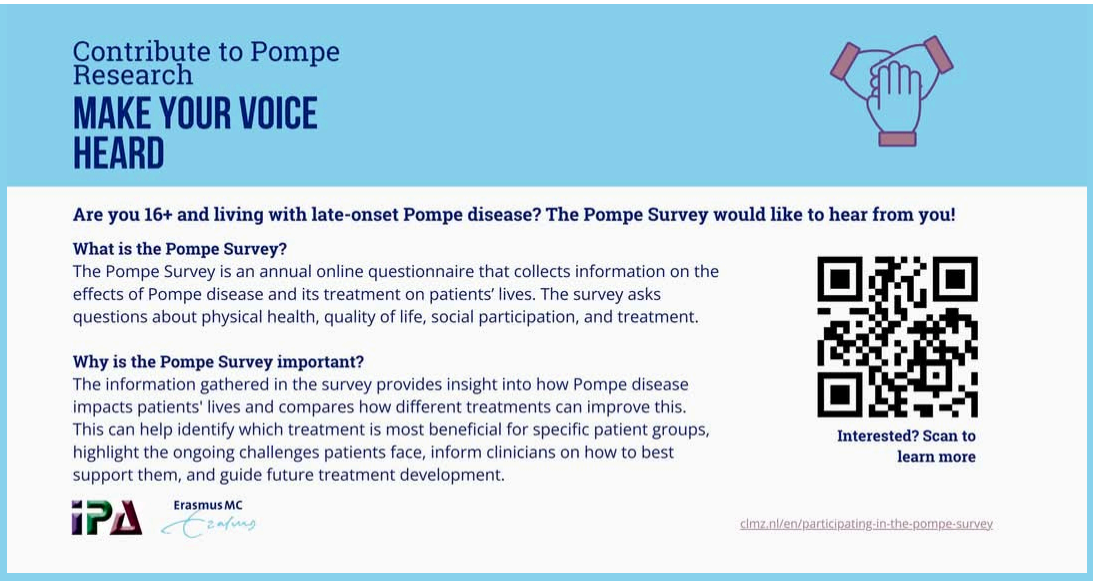
Rare Diseases Spotlight

USA

Denali receives FDA approval for first blood-brain barrier-crossing enzyme therapy

Denali Therapeutics has announced that the U.S. Food and Drug Administration (FDA) has granted accelerated approval for Avlayah™ (tividenofusp alfa-eknm) for the treatment of neurologic manifestations of Hunter syndrome, also known as MPS II.
Avlayah is the first FDA-approved biologic specifically designed to cross the blood-brain barrier and reach the brain as well as the rest of the body.
Hunter syndrome is a different lysosomal storage disorder from Pompe disease. Still, this approval is highly relevant for the broader rare disease and enzyme therapy communities. The drug uses Denali's proprietary TransportVehicle platform, which is designed to deliver large biologic medicines across the blood-brain barrier.
Denali describes the platform as a new class of biotherapeutics with potential applications in lysosomal storage disorders, neurodegenerative diseases and other serious conditions.
The approval was based on biomarker data showing a 91% reduction in cerebrospinal fluid heparan sulfate levels after 24 weeks of treatment in a Phase 1/2 study. Continued approval may depend on confirmation of clinical benefit in the ongoing Phase 2/3 COMPASS study.
For the Pompe community, this milestone matters because it shows regulatory progress for next-generation enzyme delivery technologies. These approaches may help address long-standing limitations of conventional enzyme replacement

therapy.
https://investors.denalitherapeutics.com/news-releases/news-...



International Pompe Association - IPA

Lt.gen. Van Heutszlaan 6
3743 JN Baarn
The Netherlands