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Annual Report 2025

For the IPA and the Pompe community, 2025 was marked by the tragic passing of Maryze Schoneveld van der Linde and of Tiffany House.

In Tiffany's words, "It is impossible to say how much Maryze has done for the Pompe Community around the world. She was, and is, truly an inspiration to all who knew her. She worked tirelessly for patients around the world, often at the expense of her own health and needs. She will be greatly missed."

Tiffany was a driving energy connecting patients, scientists, clinicians, and industry—always pushing for better care, stronger research, and true collaboration. She believed that when we work together, we are undeniably stronger and capable of making lasting, life-saving change.

Exciting advancements continue to evolve within the Pompe community, and the IPA remains committed to monitoring these advances and cultivating relationships with all parties involved in the development of treatments or interventions for Pompe. In addition, we work closely with the Medical/Scientific and scientific communities to improve our mutual understanding of Pompe disease and the unmet needs of the Pompe Community. As you read the 2025 Annual Report, the IPA Board hopes that the scientific advances and treatment options that are coming, as well as the IPA's planned collaborations for 2025 (and beyond), will fill you with the same excitement for the future that we have.

Chairman's statement 2025

The IPA was founded 26 years ago by a small group of national organization leaders who saw the value in working together to represent the global Pompe Community. The IPA's mission is two-part: to campaign for early diagnosis and effective, affordable and safe therapies, and to strive to provide information and support to all patients, their families, and others with an interest in Pompe disease.

Specific goals/objectives that we set for ourselves were:

- Stimulate research into the causes, treatment and prevention of Pompe disease
- Stimulate the rapid application of research into the causes, treatment and prevention of Pompe disease
- Promote early and accurate diagnosis and screening programs for Pompe disease
- Support and encourage national organizations to obtain approval and reimbursement for therapies from government bodies and health providers
- Encourage other organizations or individuals to establish a mechanism for all patients to gain access to therapies, irrespective of their personal financial status
- Encourage the formation and support the continued development of national Pompe organizations
- Establish and maintain a key position with industry, researchers and individuals with interests in Pompe disease
- Provide educational and informative publications through electronic and other media

We are fortunate to not only have a treatment for Pompe disease, but to have multiple treatments and others in the pipeline. It's important to also recognize the scientists, researchers and physicians who have dedicated their lives to creating better futures for Pompe families.

With immense gratitude, we recognize the important achievements of Professor Ans van der Ploeg, MD, PhD, who has retired after many years of dedication to Pompe patients.
worldpompe.org/news/dare-to-dream/

2025 International Pompe Day—Every Move Counts!

April 15, 2025 was our 12th Annual International Pompe Day. As we look forward to new treatments like gene therapy, substrate reduction therapy, and more, it is important for patients to stay as healthy as possible while these treatments are being developed.

This year we launched the Run, Walk or Roll Event. The theme, "Every Move Counts," reminds us that whether you walk, run, or roll, every step truly makes a difference.
PompeDay.com

Special thanks to Amanda Joost and her team for organizing the 2025 Run, Walk or Roll for Pompe event!

Patient Affiliates

We currently have 67 contacts with patient organizations and individuals, representing approximately 58 countries around the world.

Community Advisory Board

The International Pompe Association (IPA) established its own Community Advisory Board (CAB) for Pompe disease in 2019 as a patient-led initiative—uniquely organized and operated by the Pompe patient community rather than being industry-driven.

Traditionally, a CAB is a neutral group formed and run by patient advocates to discuss and advise on the latest developments and challenges in medical research and procedures, facilitating dialogue with research sponsors. CAB members—typically patients or advocates—contribute expertise that helps guide sponsors in clinical program development, individual clinical trials, and other areas beyond research. Importantly, CABs ensure that clinical studies are designed to meet the real needs of patients, promoting higher-quality, patient-centered research.

The IPA CAB currently includes approximately 29 members from multiple countries across three continents, representing a broad spectrum of Pompe disease perspectives. Beginning with 19 members in 2019, the CAB has since expanded to include new members from the United States, the Netherlands, Germany, Hong Kong, Japan, Greece, France, and Australia.

The inaugural IPA CAB meeting took place on October 28, 2019, in San Antonio, Texas, as a multi-company roundtable attended by industry representatives from Spark, Sanofi, Amicus, Audentes, and AskBio.

Following the impact of the COVID-19 pandemic, CAB meetings transitioned to an online format, with one-on-one virtual meetings held with various industry partners, including:

- AskBio/Bayer (July 2020 and 2022)
- Sanofi Genzyme (September 2020; June 2021; December 2021; June 2023; and two additional meetings in 2023)
- Avrobio (two meetings in September 2021)
- Maze Therapeutics (2022)
- Shionogi (2024)
- AskBio (2024)

These virtual sessions have proven highly effective and have expanded industry engagement. In 2023, we began preparing for additional in-person CAB meetings, continuing this approach at the 2024 AMDA/IPA International Conference.

During the 2024 conference in San Antonio, two in-person, one-on-one CAB meetings were held—one with Sanofi and another with Amicus—both moderated by the IPA. CAB members also actively participated in the broader conference, strengthening engagement between the Pompe community and industry stakeholders.

In 2025, the CAB held three virtual one-on-one meetings: one with EfficientCME, an independent medical education company; one with Sanofi; and another with Denali Therapeutics.

Due to the virtual format, in which only a subset of CAB members participates in each session, the IPA is seeking additional CAB members, preferably from patient organizations, to ensure representation across the full spectrum of Pompe disease.

Prospective CAB members should:

- Be 18 years of age or older
- Possess strong English language skills
- Be able to commit to a minimum of two meetings per year
- Sign a non-disclosure/confidentiality agreement

Specific training will be provided for new members. Interested individuals should submit a letter of application outlining their relevant skills, experience, and motivation to fabiodipietro@worldpompe.org.

Through these efforts, the IPA CAB remains committed to ensuring that patient needs are prioritized and that Pompe disease research and development continue to advance with a strong patient focus.

The IPA CAB was Tiffany House's vision—bringing together patients and advocates so that our voices guide our partners' work and patient needs remain central. We now carry that vision forward, and we know she would be proud of us as we ensure this important program continues to thrive.

Current Commercially Approved Treatments

There are currently three commercially-approved treatments for Pompe disease. These treatments are not available in all countries, but efforts are underway by companies involved to continue seeking approval and reimbursement in additional countries.

Next Generation Enzyme Replacement Therapies

Amicus Therapeutics

As of November 2024, Amicus has received approval for Pombiliti + Opfolda in the European Union, the United Kingdom and the United States of America and they continue to work with other regulators and reimbursing authorities to provide commercial access in more markets.

In addition, Amicus currently has five (5) active clinical trials. These range from observational studies to pediatric trials. For more information, please visit ClinicalTrials.gov

Sanofi

Sanofi has two commercially approved enzyme replacement therapies on the market.

The first, Myozyme/Lumizyme was approved in 2006 in the European Union and United States, and continues to be used on a global scale.

The second is their next generation ERT, Nexviazyme/Nexviadyme, which was approved and commercially available in the US for late-onset Pompe in 2021. The European Union, the United Kingdom and Australia have now given their approval for the same treatment, which is known there as Nexviadyme and in Australia as Nexviazyme. Work continues with reimbursers around the world to make this treatment commercially available.

In addition, Sanofi has several clinical trials underway to evaluate its next-generation treatment in various populations, including pediatric patients. For more information, please visit ClinicalTrials.gov

Research and Drug Development

The IPA always approaches all companies or investigators active in the Pompe field to discuss their treatment development programs; many are very willing to meet with us, either by teleconference, or face-to-face when we come together for an international meeting.

Below is a list of Pompe programmes we are currently aware of; visit ClinicalTrials.gov for further details of clinical studies currently underway.

Next Generation Enzyme Replacement Therapies

Eleva Biologics

Eleva has previously announced that they are developing a moss-produced recombinant GAA; Repleva GAA/RPV-002 (glyco-improved Pompe-ERT). As of November 2024, it appears that their Pipeline is focused on Factor H and Fabry disease. However, they do still have links to publications regarding pre-clinical work in Pompe, so they will remain on our radar.

Delani Therapeutics

Denali Therapeutics is working on DNL952 (ETV:GAA), a pre-clinical, IND-enabling therapeutic candidate designed as a fusion of the enzyme acid α-glucosidase (GAA) with the company's Transport Vehicle™ (TV) platform, with the goal of delivering the enzyme broadly, including into both brain and muscle tissues, to improve upon conventional enzyme replacement therapies.

JCR Pharmaceuticals

According to JCR Pharmaceuticals website, as of October 2025, Pompe is still not included as being part of their Pipeline. We will remove it from our watchlist.

M6P Therapeutics

M6P Therapeutics is working on a next generation ERT that is "naturally produced with the highest levels of M6P as compared to other rhGAA ERTs." According to their website, M021 "normalized glycogen and significantly improved muscle strength in Pompe mice in long-term studies" and "is substantially better than standard of care ERT glycogen for substrate clearance in muscles of Pompe mice." As of October 2025, their website states that the program is in Pre-Clinical Development and it still states that they anticipate applying for Investigational New Drug status (IND) in the third quarter of 2026.

Substrate Reduction Therapies

Substrate reduction therapies (SRTs) seek to affect the disease process by reducing the accumulation of glycogen in the muscles of Pompe patients by reducing the amount of glucose that is turned into glycogen. Below we have listed the two companies who are currently working on Substrate Reduction Therapies for Pompe.

ARO Biotherapeutics

Aro Biotherapeutics is a biotechnology company pioneering the development of tissue-targeted genetic medicines with a platform based on a proprietary protein technology called Centyrins. They recently completed a Phase 1 study in healthy volunteers, which showed that "ABX1100 was well tolerated . . . and it showed durable GYS1 mRNA knockdown in muscle biopsies, with effects lasting through at least 10 weeks following a single dose."

On October 29, 2024, Aro announced the initiation of a Phase 1(b) trial of ABX1100 in late-onset Pompe patients. According to their press release: "In the planned 1b study, investigators seek to enroll adults with LOPD to evaluate the safety and bioactivity of ABX1100. More information about the trial is available at ClinicalTrials.gov using the identifier NCT06109948."

The first trial site was in Calgary, Alberta, Canada. Since then, four other trial sites have opened. Aro announced IND clearance for ABX1100 from the FDA in February, 2025.

Shionogi &Co., Ltd./Maze Therapeutics

Maze Therapeutics is developing MZE001, an investigational oral glycogen synthase (GYS1) inhibitor that aims to address Pompe disease by limiting disease-causing glycogen build-up. GYS1 is an enzyme responsible for glycogen production in human muscle cells, but not in human liver cells.

Maze completed a Phase 1 study in healthy individuals and announced positive results in February 2023.

In May 2023, Maze Therapeutics announced that it had entered into an exclusive worldwide license agreement with Sanofi for MZE001. However, this acquisition was stalled by the Federal Trade Commission (FTC), and Sanofi withdrew from the agreement in December 2023.

Following this, Shionogi & Co., Ltd. Acquired a worldwide license agreement for the rights to MZE001 on May 10, 2024. According to their press release, Shionogi is committed to advancing MZE001 and believe it has the potential to be used both as a monotherapy option and as an add-on therapy with enzyme replacement, the current standard of care, to enhance the treatment of patients with Pompe disease.

Cell and Gene Therapies (Regenerative medicine)

Regenerative medicine across rare disease has continued to gain interest. Below we have listed a number of different approaches that may each provide a solution for Pompe.

Alexion/CANbridge Care

In October 2022, AstraZeneca (through its Alexion subsidiary) acquired LogicBio Therapeutics. CANbridge holds an exclusive global license from LogicBio Therapeutics, Inc ("LogicBio") to develop, manufacture and commercialize gene therapy candidates for the treatment of Fabry and Pompe diseases, based on LogicBio's AAV sL65 technology. The company is in very early stage of development in their Pompe programme, as they continue to make progress on their Fabry disease candidate.

Amicus Therapeutics

Amicus doesn't seem to be actively working on gene therapy for Pompe disease, but they did have a poster at the WORLD Symposium in February, 2025.

Astellas

Astellas continues with its phase I/II clinical trial for LOPD adults, FORTIS. Their Gene Therapy, AT845, utilizes a muscle-directed approach with an AAV8 capsid serotype that is being investigated to determine whether it can deliver a functional GAA gene to express GAA directly in tissues affected by the disease, including skeletal and cardiac muscle.

According to ClinicalTrials.gov, the FORTIS study trial aims to recruit 11 patients at four locations (two in California, one in Utah, and one in Newcastle UK). The Primary completion date of this trial is now February 28, 2030. It is not currently recruiting.

Bayer/AskBio

AskBio, a Bayer company, has an active, but not recruiting, clinical trial listed on ClinicalTrials.gov for adults (18+) with Late-Onset Pompe Disease (LOPD) to assess their gene therapy ACT-101.

ACT-101 is infused intravenously and designed to deliver a functioning copy of the GAA gene to the liver. The goal is to restore GAA production to a level sufficient to no longer require ERT.

Only one study centre is currently active in this trial, Duke University, NC, which has enrolled 7 patients.

Crosswalk/CODEXIS/Takeda

In July 2024, Crosswalk Therapeutics acquired the Pompe disease compounds from Codexis, Inc. The compounds were part of a collaboration between Codexis and Takeda that was abandoned after Takeda announced in April 2023 that it was moving away from early-stage gene therapies.

Crosswalk's mission is to develop functional cures for rare diseases, including Pompe disease. The company's name symbolizes the idea of parents guiding their children safely through the medical care system.

According to Crosswalk's website, it is seed-stage biotech company dedicated to relentlessly pursuing functional cures for rare disease patients and their families. With an initial focus on rare diseases, we aim to expand our scope to include genetically defined common diseases in the future.

Erasmus MC – Professor Pim Pijnappel

Research continues at the Erasmus MC to study several regenerative therapies for Pompe, including stem cell regenerative therapies and RNA Oligonucleotides, as well as lentiviral gene therapy. Dr Pijnappel made a presentation at the 2024 AMDA/IPA International Pompe Patient and Scientific Conference on the status of his work. A recording of his presentation is available on the AMDA website: amda-pompe.org/conferences

GeneCradle Therapeutics

GeneCradle is working on an AAV-mediated gene therapy for Infantile-Onset and Late-onset Pompe disease. According to ClinicalTrials.gov, there are two trials based out of China that are currently recruiting. They will include 6 patients with IOPD and 33 patients with LOPD. The IOPD study was first posted on October 5, 2022, with a primary completion date of September 2024. The LOPD study was first posted on February 19, 2024 with a primary completion date of December, 2026.

Regeneron

According to the August 2024, Regeneron Corporate Presentation, Regeneron has a Pre-Ind research program for Pompe in their pipeline that is exploring CRISPR/Cas9 + AAV Transgene Insertion. This Program is in a very early stage, so we will continue to keep an eye on it as the program develops further. No new information is available on their website.

Lacerta Therapeutics

While Sarepta had a licensing agreement with Lacerta Therapeutics to develop AAV-9 gene therapy for Pompe disease, this agreement was terminated in 2023. Further, it appears that as of mid-2024 Lacerta has ceased operations. We will remove it from our watchlist.

Spark Therapeutics

Spark Therapeutics was developing SPK-3006, an investigational gene therapy for treatment of Pompe disease. However, as of July 2024 Spark announced that it has stopped its Pompe Program. According to the Community Update they provided: The decision to close our Pompe program was not related to any safety issues or concerns. Rather, the decision was part of a larger portfolio review and realignment of our strategy as an organization to ensure we're delivering impactful gene therapies to people as quickly as possible. We will remove it from our watchlist.

Campaigns

Dose Flexibility

The IPA board continues to have concerns over the inflexibility of ERT dosing for Pompe Disease. This extends beyond the current commercially-available treatment to future treatment options as well. Several treating physicians are interested in exploring higher doses, but cannot because of the prohibitive cost of additional drug. The IPA continues to raise this topic with clinicians and drug companies to explore ways forward. A major hurdle currently is the interaction between country labels for approved treatment and reimbursing authorities' ability to pay. Especially in light of the economic situation around the world, we anticipate that in the coming years it will be even more difficult to get treatments approved and reimbursed, let alone at higher doses. However, just because it is a difficult journey we face, the IPA is committed to advocating for patients around the world when it comes to equal access to optimal dosing.

Treatment Transition and Expectation Guidelines

One common question that Patient Organizations get from new patients is: "How will I respond to treatment?" Unfortunately, there is not a clear answer to this question. Patient response to treatment varies from patient to patient. In some cases, the degree of progression at time of treatment onset will affect response. In other cases, a high-sustained immune response will reduce efficacy. And in others, it is not clear why some patients respond better than others.

The only thing that *is* clear after over twenty years of experience with ERT is that patients WILL respond differently, and it is important for patients to have realistic treatment expectations. This is especially true now that there are more treatment options available to patients. Now, the questions are becoming: What criteria should patients use (in consultation with their physicians) to determine whether they should try a new treatment option? What should my expectations be if I switch treatments? How do I know if the new treatment I am trying is better for me?

These are important questions, and ones that the IPA Board is committed to helping to answer.

IPA-Erasmus Survey

The IPA/Erasmus Survey (the "Pompe Survey") was started in 2002. It is a collaboration between the International Pompe Association (IPA) and Erasmus MC.

The goal at the time it was started was to better understand, from the patient perspective, the disease burden that patients with Pompe face. The timing of the initiation of the Survey was very intentional. There was an understanding at the time that it was imperative to begin collecting this information so that we would have a patient-owned, patient-reported questionnaire and data to capture the natural history of the disease in the early days, and then to capture how that may change over time with treatment.

The IPA-Erasmus Survey has been a phenomenal success over the last twenty-two (22) years. There have been numerous peer-reviewed articles written based on the collected data—articles that each and every Pompe patient contributed to and made possible.

Today, the large number of potential future therapies makes it even more imperative that we remain strong in our commitment to the Survey, and to expanding it as appropriate. This is because each new drug that is approved will likely come with a commitment to track its clinical outcomes and patient-reported outcomes. That could mean a separate registry for each drug unless an independent registry can be developed to hold all the data. This would, necessarily, result in patient reported data being spread throughout multiple registries depending on which treatment they are on at the time, and whether they choose (upon discussion with their treating physician) to switch between therapies.

Without an effort like the Erasmus Pompe Survey in existence, it will be impossible to truly track, on a global level, how patients feel that they are doing throughout their disease and treatment experience. To that end, the IPA Board has been working very closely with Erasmus MC to update and streamline the existing Pompe Survey. The Survey is now available in the many languages.

Finally, the IPA Board continues to advocate for new modules that would allow for patients to track their own results, and are beginning work on a new Survey that will be targeted at younger patients. These meetings will continue, and we hope to also incorporate an international board of advisors in the coming years.

For more information on the IPA/Erasmus Pompe Survey, please visit the updated webpage on the Erasmus University Website: www.erasmusmc.nl/en/research/project/ipa-erasmus-mc-pompe-survey.

This website also includes a partial list of publications that have come from the data so our Members and the Patient Community as a whole can see how their contribution has led to important understanding of Pompe disease.

Independent Global Pompe Patient Data Collection Collaboration: The Octopus Project

As of November 2024, there are multiple silos of Pompe patient data around the world. These include: the Sanofi Pompe Registry, the Amicus Global Prospective Observational Registry of Patients With Pompe Disease, the IPA/Erasmus Survey Data (Patient Reported Outcomes), national data pools (ie Australia, France, Germany, the Netherlands, etc), and data collected at all of the expert centres around the world.

While data collection is imperative, it is just as important to be able to aggregate information across data silos to be able to best understand trends, and best disease management practices. This is especially vital now that patients are fortunate to have treatment options.

As a community, it is in our best interest to start collecting data in a uniform manner to allow patient data to be compared across data sets. This will allow our community to be able to collect enough data from around the world

and across the disease spectrum to better understand how treatment options, disease management approaches, and combinations of therapy affect different subsets of patients across the Pompe spectrum.

In 2023, the IPA began working with a group of expert Pompe clinicians around the world to collaborate on developing an Independent, Global Pompe Patient Registry. In addition to regular meetings with the expert clinicians, the IPA Board has been collaborating with and learning from other key parties in the rare disease registry space. These include RareX and the International Niemann-Pick Disease Registry.

All parties agree to continue working together and to meet at least biannually.

We look forward to continuing this important work and providing updates as we progress on this Project.

Communications

With the loss of Maryze Schoneveld van der Linde, our communications has been restructured. This is being managed by our Communications team, led by Annic Kobrück. Other team members are Andrea Farris and Brad Crittenden. As these changes are implemented, you'll see more regular communications through our newsletter, social media and website.

We are eager to share our member organizations' activities through our newsletter and social media channels. Let us know what you're up to!

For IPA Members only, a confidential (closed and publicly not visible) Facebook group is used for communication between the member organizations (www.facebook.com/groups/850602065054870/). If you want to join as a representative of your national patient organization, please contact the IPA Board at info@worldpompe.org.

Pompe Connections

These patient-focused publications are available on the IPA website in many languages: Dutch, French, German, Italian, Japanese, Spanish, Turkish, Arabic, Korean, Russian, Greek, Czech, Hindi, Chinese, and Portuguese. We encourage our members to share them with patients in their communities.

In addition, we would also like to ask our members, and the broader Pompe Community of patients, family members, and the medical community, to contact us at info@worldpompe.org if there are additional topics that you would like to see covered. In addition, please contact us if you would be willing to help with translations of current and future Brochures.

Meetings

WORLD Symposium 2025

Several Board Members (and members of the Pompe Community) attended the WORLD Symposium in February 2025. Attendance was a combination of virtually (following the conference) and in person. The WORLD symposium is an annual conference which brings together clinicians, scientists, industry and patients from all over the world to learn and share knowledge on all Lysosomal Diseases (LDs). Many presentations and posters highlight the current level of research activity for Pompe disease. We look forward to attending again next year.

Sanofi Rare Disease Registries Patient Council

The IPA Board has had a consistent presence on the Patient Council. The purpose of the Council is to provide the Patient Community's perspective on Sanofi's Pompe Registry.

Amicus Therapeutics Pompe Registry Steering Committee

The IPA Board has had a consistent presence on the Steering Committee. The purpose of our presence is to bring patient perspective to the Registry.

IPA AGM 2025

The IPA's Annual General Meeting in 2025 will take place virtually on November 22, 2025.

Looking ahead

The IPA welcomes ideas from the Pompe community for projects to raise global awareness, improve our support and engagement with national groups, and develop our relationships with the growing number of research and industry networks. We are a very close community and there is little doubt that Together we are Strong!

Thank you,

IPA Board

November 2025