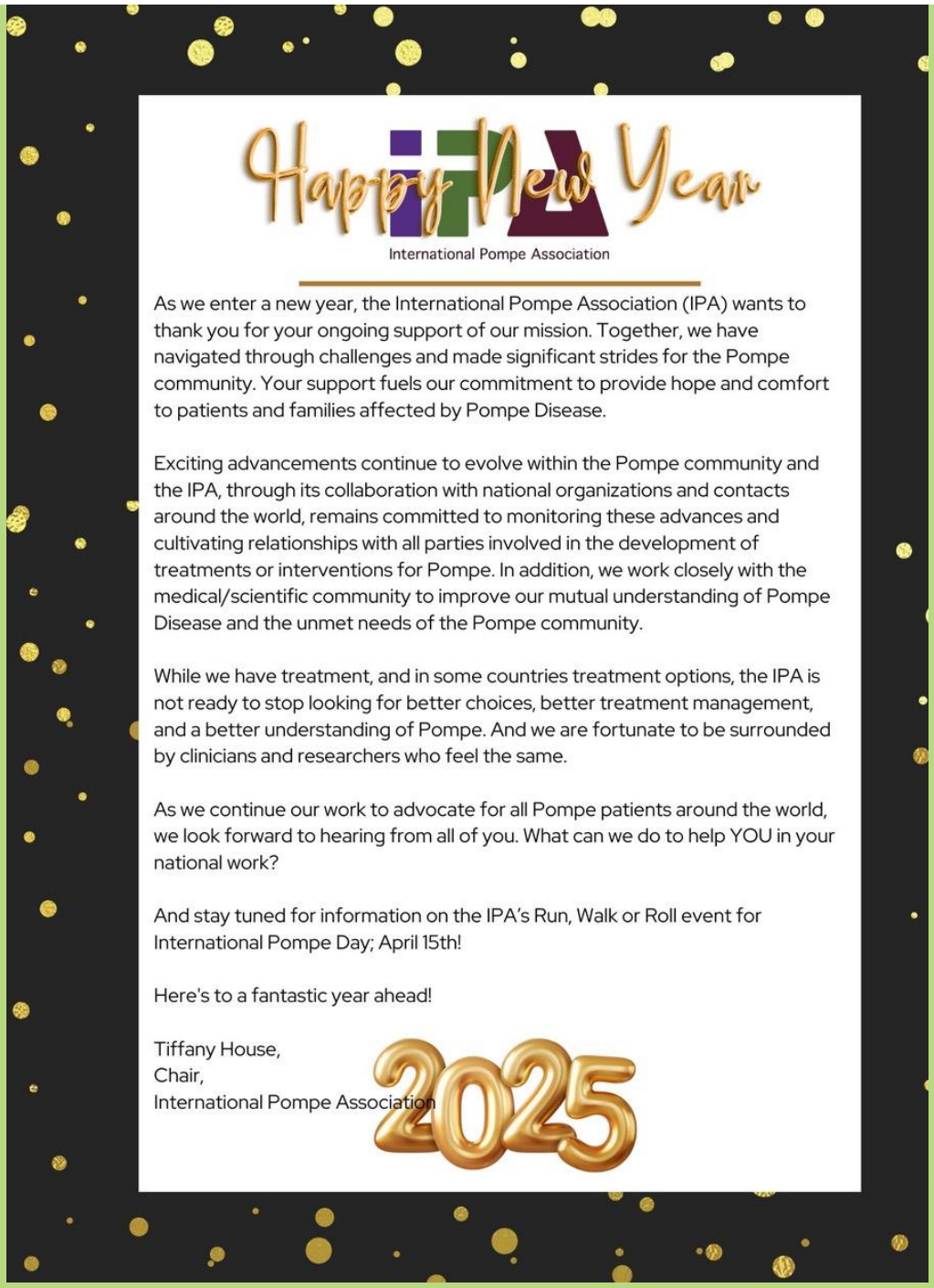
# 16.01.2025 **IPA Update Q1/2025**

## Inhalt

International Pompe Association

# IPA Update Q1 2025

International Pompe Association




The IPA annual report of 2024 can be viewed by clicking the link.

**Hopeful Times for Funding Of 2nd Generation Enzyme Replacement Therapy (ERT) Nexviazyme (avalglucosidase alfa) For Treating Pompe Disease**

The New Zealand Pompe Network (NZPN) team is ecstatic to learn that the PHARMAC Rare Disorders Advisory Committee has RECOMMENDED the funding of 2nd generation

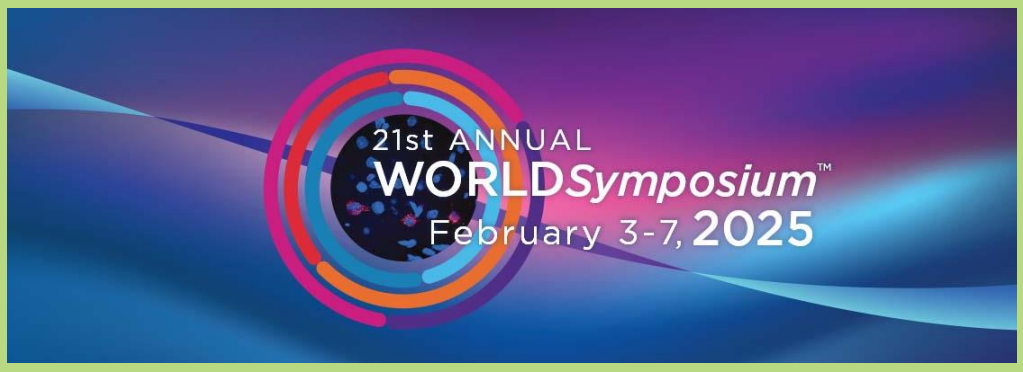
enzyme replacement therapy (ERT) Nexviazyme (avalglucosidase alfa) for treating Pompe Disease. This recommendation is for both infantile onset Pompe (IOPD) and Late onset Pompe (LOPD). Until now, only infants have been eligible for treatment in New Zealand. Currently there are 11 diagnosed patients in New Zealand. 7 on compassionate access, 4 on a clinical trial, and 1 not wanting treatment.

Allyson Lock, Executive Director of New Zealand Pompe Network said: "This recommendation has been an awfully long time coming. New Zealand is so far behind many other countries with treatments of this kind for rare disorders. A growing number of countries have treated Pompe patients since the first approved drug in 2006. There are around 80 countries treating patients. People with Pompe disease in New Zealand have had to rely on compassionate access to drugs, or participation in clinical trials. So, to have New Zealand finally acknowledging the worthiness of this drug is absolutely a game changer for those of us with Pompe, and those who will be diagnosed in the future.

I would really like to acknowledge that this has been a worldwide effort from patients, patient advocates, patient organisations and researchers. Thank you for your tireless work and dedication to people affected by Pompe disease. You are all real-life heroes!

It is our hope that PHARMAC will expedite the availability of this medicine so that when someone is diagnosed with Pompe disease their doctor can tell them that there is a treatment, and they can get it here in New Zealand. That would be an amazing outcome"!

**WORLDSymposium 2025**




WORLD*Symposium*™ is an annual research conference dedicated to lysosomal diseases. **WORLD** is an acronym that stands for ***We're Organizing Research on Lysosomal Diseases***. Since its inception as a small group of passionate researchers in 2002, WORLD*Symposium* has grown to an international research conference that attracts over 2000 participants from more than 50 countries around the globe.

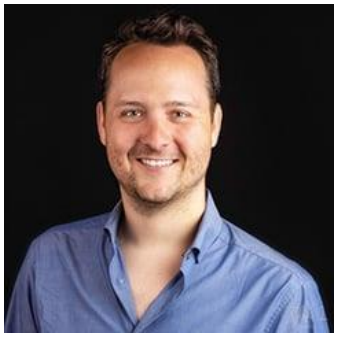
The symposium is designed for basic, translational and clinical researchers, patient advocacy groups, clinicians, and all others who are interested in learning more about the latest discoveries related to lysosomal diseases and the clinical investigation of these advances. Each year, WORLD*Symposium* presents the latest information from basic

science, translational research, and clinical trials for lysosomal diseases.

WORLDSymposium - We're Organizing Research on Lysosomal Diseases

**Ryan Colburn (USA) to Receive the 2025 Catalyst Award**

WORLD*Symposium* will present the second annual **"Catalyst" award** to Ryan Colburn, who was nominated in recognition of his efforts to instigate a shift in perspective for the prevalence of Pompe disease.



Colburn has some genetic variants, just like everyone else. In 2015 he learned that some of his variants are associated with a rare metabolic disorder, Pompe disease, and he has unapologetically altered course ever since. His professional background is in development, engineering and operations management: as applied to race cars, airplanes, rockets, satellites… and rare disease. Ryan is a driven student of process, (it is everywhere), and in this context, he is applying what he's learned along the way to contribute to the health of the rare disease ecosystem. He is passionate about empowerment and engagement, and shifting the view of patients as "subjects" to one of participants, collaborators, and partners who can help to find the most effective ways to accelerate progress on understanding and solving challenges in rare disease.

One example of Ryan's focus on shifting the perspective comes from researching Pompe disease; he grew frustrated when he realized that the widely used frequency estimate for Pompe disease of 1 in 40,000 came from two very small studies that were more than 20 years old. The results were outdated and did not factor in all the information learned in the past 20 years. He also recognized that continued use of outdated epidemiology has consequences. In 2024, after a lengthy effort to recognize updated data and methods, Ryan published "An analysis of Pompe newborn screening data: a new prevalence at birth, insight and discussion" which used improved data and framework to study prevalence at birth for Pompe disease. The new prevalence study collected and analyzed the largest relevant dataset to date, including a novel way to judge the results as a projection for prevalence at birth of Pompe. **The result is an upper bound for prevalence of 1 in 18,698 for Pompe disease, a significant difference from previous figures.** He is now working on applying lessons learned in this study to improve the state of epidemiology for an expanded set of rare genetic conditions.

Ryan will receive WORLD*Symposium*'s 2nd Annual Catalyst Award at the Be the Catalyst event at **18:00 PST on Monday, February 3, 2025.**

**Pompe kids raise funds for mega charity event for people with metabolic diseases in the Netherlands.**

It's Christmas Eve 2024. In the Netherlands we had a 6 day life radio & tv fundraising event going on. The chosen Charity this year is Metakids, the Foundation that sponsors research to metabolic diseases.

The Pompe kids (and children with other metabolic diseases) in the Netherlands have been very active to raise money: they were on television, gave interviews, organised activities etc. I am very proud of them. The Erasmus team too has been very active to raise money. There is one 'kid' especially that has been active raising money. Her name is Diede (16). She knew she would not live long as Pompe has progressed too far and caused her pain, pneumonia and total loss of hand/finger function. This young woman raised, in spite of her severe condition, over 50,000 euro 'for the children who will be born after her with Pompe Disease', as she said herself.

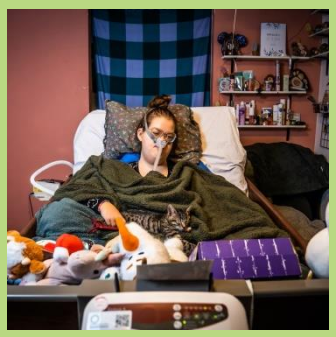
A positive and powerful vibe went through the Netherlands the week before Christmas. People can't say anymore they are unaware of metabolic diseases. Those affected and impacted feel recognised. The urgency finally seems to be clear.

The final amount of raised money to sponsor research to metabolic diseases is 11.513.485 euro.

Diede passed away at January 2nd 2025.

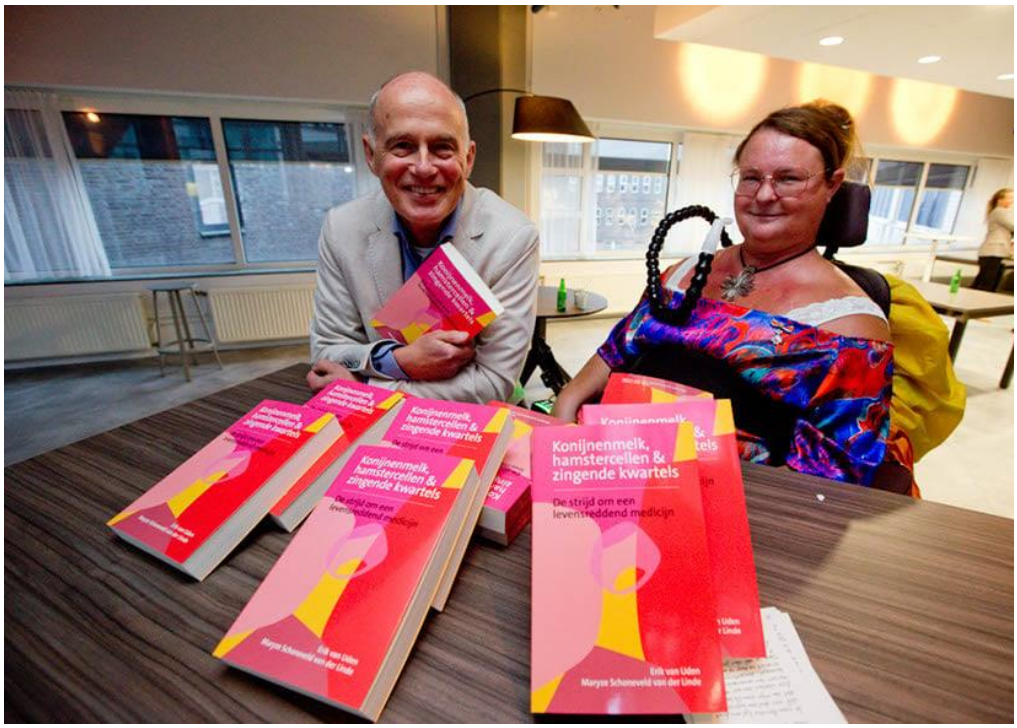
*Hanne (11) giving an interview for Dutch national radio.*

*Getting together with everyone joining the charity event.*



*Diede (16) watching the charity event while raising money herself.*

**Book written on medicine development for Pompe disease and the high impact of patients' contribution**




*Erik van Uden and Maryze Schoneveld van der Linde at the official presentation of their book at the Erasmus MC in Rotterdam in The Netherlands, where a lot of research on Pompe disease has been done.*

On Monday 28 October 2024, the book **Konijnenmilk, hamstercells & zingende kwartels – De strijd om een levensreddend medicijn** *(Rabbit Milk, Hamster Cells & Singing Quails - The Battle for a Life-Saving Medicine)* was presented.

This book, *only available in Dutch*, provides insight into the complex world of drug development. Maryze Schoneveld van der Linde and Erik van Uden worked on it for almost three years. Erik worked as Head of Communications at Spierziekten Nederland until 2021. From that position, he experienced the process of drug development against Pompe disease. Maryze, who has Pompe disease herself, actively participated in that development.

The authors used the archive of Spierziekten Nederland and the extended archive of Maryze to collect correct information. In addition, they interviewed more than 50 people who were involved in the development of the drug, namely: those with Pompe disease, scientists, doctors, people from the pharmaceutical industry and authorities.

“It was a great experience to do the writing process together. The 3-year process went smoothly: from our collaboration in writing to finding suitable interview candidates and a publisher," says Maryze.

The book is now available in bookstores. Erik: “It was encouraging to notice that everyone wanted to tell their own story. We spoke to people from the Netherlands, Belgium,

France, Italy, the UK and the US. Via Zoom of course." The book contains the historical context from Dr. Pompe to the future of genetherapy, the contributions by the many people involved in the development alternated with personal stories from Maryze.

Maryze: "With this book, we hope to inspire and motivate other people with a rare disease to take action. We all have to do that. I really believe that if you never start, there will probably be no treatment for your disease. Of course, it will not always be easy. Some already have a fairly large patient group, but sometimes it is also about individuals. Then you also have to look for other fellow patients in other countries. A reasonable command of the English language is important. It does not have to be perfect, but it does have to be understandable. In my case, my English improved with a lot of practice, because speaking, reading and writing English all day long is a good learning experience. Larger patient groups could start with a natural history study together with a medical advisor. This knowledge will help you anyway. The book shows how essential the active contribution of patients has been in Pompe disease.

Other books written by Pompe patients:

* My Fight Never Ends… My battle against Pompe disease, by Yamila Romero
* Nininha, Hugo Amaro
* One Fall, A Million Climbs – my life journey with Pompe, by Alyasghar Hebatullah
* The Cure, by Geeta Anand
* Pompe and Me, by Julie Garfield and Ben Lock
* My Heart Grew Warm, by Kasumi Yoshida
* I am a Pompe Warrior, by McKenzie Barker

*Erik and Maryze present their first book to prof. dr. Ans van der Ploeg from the Erasmus MC.*

*Another copy was presented to prof. Arnold Reuser who worked on Pompe disease his whole working life.*

*Maryze and her friend talked to former Minister of Health, Pia Dijkstra.*

**International Pompe Association - IPA**

Lt.gen. Van Heutszlaan 6
3743 JN Baarn

The Netherlands