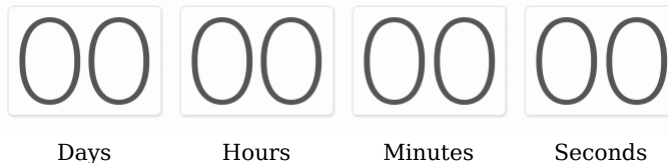




**BCH Webinar Registration**  
**Amicus Therapeutics Press Release**

**Boston Children's Hospital Webinar**

**LAST CHANCE TO  
REGISTER!!!!**



**Title:** Defining Central Nervous System Abnormalities in Infantile and Late-Onset Pompe Disease Patients

**Date:** Tuesday, October 17, 2023

**Time:** 1 PM CT, 2 PM ET

**Speakers:**

Raquel van Gool

Jaymin Upadhyay

**Webinar Overview:**

In this presentation, we will provide a short description of neurological signs and symptoms that have been reported in patients with Infantile- or Late-Onset Pompe Disease (IOPD/LOPD). We will describe our comprehensive approach toward understanding the neurobiological abnormalities in IOPD/LOPD. Finally, we will share preliminary findings from ongoing studies involving patients with IOPD/LOPD and that are taking place at Boston Children's Hospital.

**Raquel van Gool**

**Speaker Bio:**

**Raquel van Gool** is a researcher and PhD Candidate at the department of Anesthesiology, Critical Care and Pain Medicine at Boston Children's Hospital and School for Mental Health and Neuroscience at Maastricht university of the Netherlands. She completed a bachelor's program in Psychology followed by a research master's program in Cognitive and Clinical Neuroscience with a specialization in pharmaceutical development, during which she worked for several months with Boston Children's Hospital researching neuropathic conditions and lysosomal storage diseases. Raquel's current work focuses on the characterization of neurological manifestations of lysosomal storage diseases, particularly in Pompe Disease and Niemann-Pick disease Type C, as member of Dr. Jaymin Upadhyay's research team.



## Jaymin Upadhyay

### Speaker Bio:

**Jaymin Upadhyay** is a translational medicine scientist with over 15 years of experiences in the neurosciences. In 2018, Dr. Upadhyay returned to academia after spending eight years in the pharmaceutical industry. Throughout his career, he has led, or co-led multitude of investigations aimed at (i) validating new drug targets, (ii) validating novel preclinical or clinical methodologies, and (iii) translating preclinical research findings into early phase, clinical trials. Across translational biomarker studies, Dr. Upadhyay has utilized his background in neuroimaging physics and systems neuroscience and integrated this knowledge base with expertise in drug discovery and development. At Boston Children's Hospital, he serves as the principal investigator for clinical studies evaluating central manifestations in patients with lysosomal storage diseases such as Infantile- and Late-Onset Pompe Disease and Niemann-Pick disease type C.



[Register Now!!!](#)

**AMDA Webinars**

# Check Out Some of Our

# Other Wonderful Webinars

**Lentiviral Gene Therapy for Pompe Disease With Dr. Pim Pijnappel**

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**How To Manage LOPD Patients Diagnosed Through NBS With Dr. Priya Kishnani and Erin Huggins**

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**The Value of Patient Reported Outcomes & When a Hospital and a Patient Organization Work Together With Dr. Nadine van der Beek and Maudy Theunissen**

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**Insurance 101: Learning The Basics With Amy DeStefanis**

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**Aquatic Exercise: Applications for Pompe Disease**



## **Amicus Therapeutics Announces FDA Approval and Launch of New Treatment for Pompe Disease**

PHILADELPHIA, Sept. 28, 2023 (GLOBE NEWSWIRE) — Amicus Therapeutics (Nasdaq: FOLD) today announced that the U.S. Food and Drug Administration (FDA) has approved Pombiliti™ (cipaglucosidase alfa-atga) + Opfolda™ (miglustat) 65mg capsules. This two-component therapy is indicated for adults living with late-onset Pompe disease (LOPD) weighing ≥40 kg and who

are not improving on their current enzyme replacement therapy (ERT).

Pombiliti + Opfolda is a unique two-component therapy. Pombiliti is a recombinant human GAA enzyme (rhGAA) naturally expressed with high levels of bis-M6P (Mannose 6-Phosphate), designed for increased uptake into muscle cells. Once in the cell, Pombiliti can be properly processed into its most active and mature form to break down glycogen. Opfolda is an enzyme stabilizer designed to stabilize the enzyme in the blood.

“Today’s FDA approval of Pombiliti and Opfolda is a testament to the power of science, medicine, and our passionate determination to improve the lives of people living with Pompe disease. This approval embodies our Amicus spirit, passion, and resilience and is a very meaningful step for the Pompe community. I am just so immensely proud of our team, and so very grateful to everyone who has worked to bring this medicine to this approval. Most especially to all of the people living with Pompe around the world,” said John F. Crowley, Executive Chairman of Amicus Therapeutics, Inc.

[Click here for more information.](#)

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Acid Maltase  
Deficiency  
Association  
(AMDA)/International  
Pompe Association  
(IPA) Conference

**MAY 3-5  
2024**

San Antonio,  
Texas

*Stay tuned for  
more details!*



AMDA-POMPE.ORG

## 2024 AMDA/IPA Conference

### What Topics Do You Want To Hear Presentations On?

Dear Pompe Warriors and Patient Community,

We are excited to announce the upcoming 2024 Acid Maltase Deficiency Associate - International Pompe Association (AMDA-IPA) Conference, a momentous event dedicated to advancing knowledge, support, and advocacy for individuals and families affected by Pompe disease. As we gear up to make this conference an empowering experience for all, we cordially invite patients and caregivers to actively participate and shape the content of this significant gathering.

Your unique experiences, insights, and challenges provide a vital perspective that enriches the conference discussions. We sincerely value your

contribution and strongly encourage you to propose presentation topics that resonate with your interests, concerns, and aspirations related to Pompe disease.

The conference themes encompass a wide range of subjects pertinent to the Pompe community, including but not limited to:

1. Living with Pompe: Personal Stories of Triumph and Resilience
2. Advances in Pompe Research and Therapies
3. Navigating Healthcare and Access to Treatment
4. Support Systems and Building a Strong Patient Community
5. Parenting a Child with Pompe: Coping Strategies and Best Practices
6. Pompe Awareness and Advocacy: Making a Meaningful Impact
7. Overcoming Challenges in Daily Life with Pompe
8. Mental Health and Well-being for Patients and Caregivers
9. Enhancing Quality of Life through Adaptive Technologies
10. Innovations in Pompe Rehabilitation and Physiotherapy

We believe that your experiences can drive the discussions to uncover valuable insights and inspire the entire Pompe community. Whether it's a personal journey, an innovative coping mechanism, or an unexplored aspect of Pompe disease, your voice matters, and we want to hear it!

If you have a suggestion for a presentation, please reach out to the AMDA by email at [info@amda-pompe.org](mailto:info@amda-pompe.org).

Together, we can drive awareness, encourage research, and build a stronger community to tackle the challenges of Pompe disease.

We eagerly await your valuable contributions and look forward to welcoming you to the 2024 AMDA-IPA Conference.

**AMDA**

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