

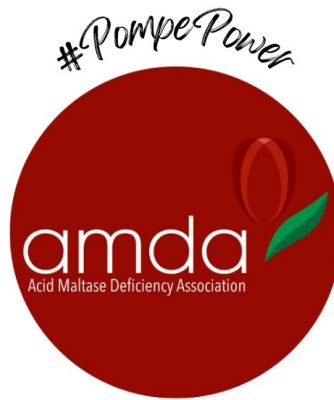


**Videos**

**Conferences**  
**Webinars**

**News**  
**Talking With Your Pompe Peeps**

**AMDA Publications**



**p**assionate  
**O**n  
**m**atters  
**p**ompe  
**e**veryday

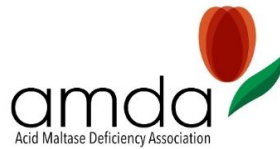
<https://amda-pompe.org>

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**January 2025**

# **AMDA Newsletter**

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## HAPPY NEW YEAR

As we enter a new year, the Acid Maltase Deficiency Association (AMDA) wants to thank you for your unwavering support and dedication. Together, we have navigated through challenges and made significant strides for the Pompe community. Your support fuels our commitment to provide hope and comfort to patients and families affected by Pompe Disease.

Exciting advancements continue to evolve within the Pompe community and the AMDA, through its involvement with the International Pompe Association (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 community to improve our mutual understanding of Pompe Disease and the unmet needs of the Pompe community.

The AMDA was established in 1995 to assist in funding research and to promote public awareness of Pompe Disease. This year marks the 30th Anniversary of the founding of the AMDA. We are proud of our journey so far, and eager to continue working for Pompe patients and their families; watch our video, "The History of the AMDA: 1995 - 2024": <https://www.youtube.com/watch?v=xzbvrVabfA8>

It is incredibly rare for a rare disease to have a treatment, let alone treatment options. Not only are there currently three commercially approved treatments in the U.S., but there are multiple new approaches in clinical trials and pre-clinical research.

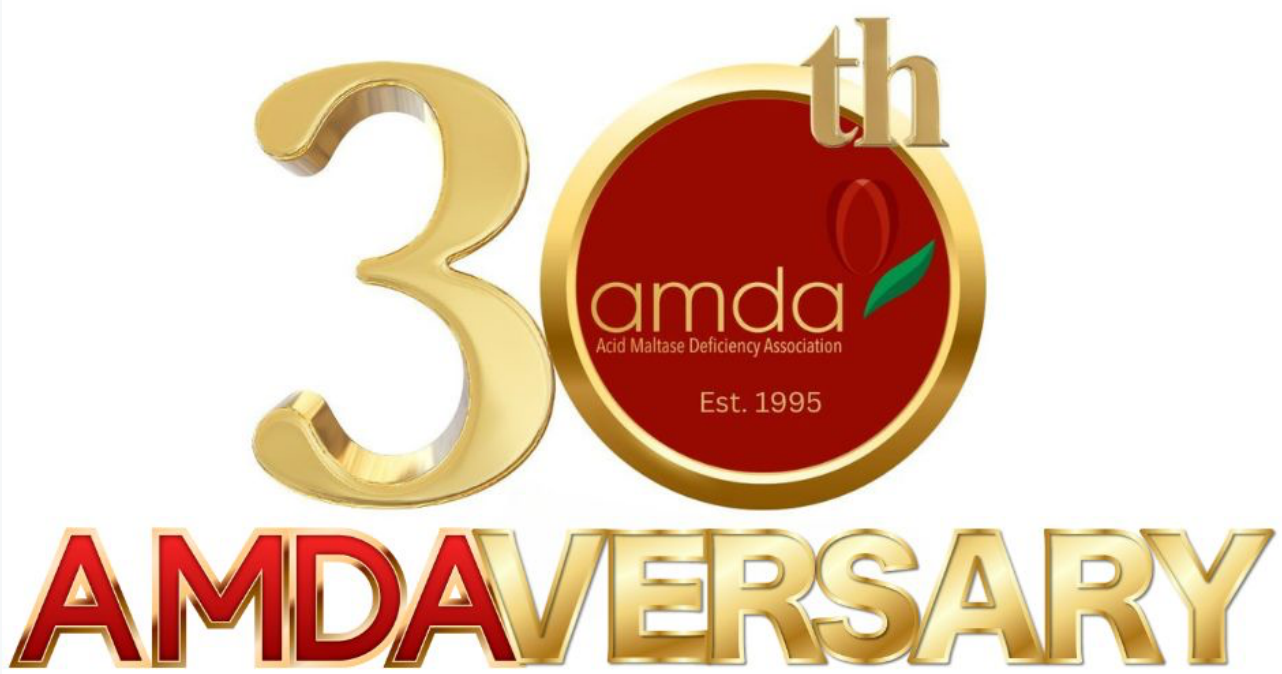
But we are not ready to stop looking for better choices, better treatment management, and a better understanding of Pompe. And we are fortunate to be surrounded by clinicians and researchers who feel the same.

The AMDA remains committed to providing the most accurate and current news on Pompe Disease and to listen to our community on what they need and want to know. You are not alone; we are here to help. The AMDA is POMPE: Passionate On Matters Pompe Everyday!

Here's to a fantastic year ahead!

Tiffany House,  
AMDA President





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### Upcoming Webinar

## The ABC's of Pompe Disease

Join us Monday, January 13, 2025 at 1 PM CT / 2 PM ET for a comprehensive introduction to Pompe disease with Erin Huggins, a PhD student in Molecular Genetics and Microbiology at Duke University. Topics of discussion include the underlying cause of Pompe disease, genetics/inheritance, newborn screening, treatment/management, and more! There will be a Q&A session after the presentation.

[Register Here!](#)

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## Webinar Details

**Title:** The ABCs of Pompe Disease

**Date:** Monday, January 13, 2025

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

**Speaker:** Erin Huggins, MS, CGC

### Webinar Overview:

Are you looking for information on Pompe disease or trying to navigate a new diagnosis? Overwhelmed by the amount of information online and the sea of jargon and terminology? Whether you are dealing with a diagnosis, are a family member or a caregiver, or even a medical provider who just wants a refresher on the basics, this webinar is for you! We will cover the "ABCs" of



Pompe disease including its underlying cause, genetics/inheritance, newborn screening, treatment/management, and more. Information will be presented in a thorough, easy-to-follow manner and there will be time for questions at the end. We hope you'll join us!

## **Erin Huggins, MS, CGC**

Erin Huggins is a board-certified genetic counselor in Durham, NC, USA. She received her B.S. in Biology from Coastal Carolina University in 2016 and her M.S. in Genetic Counseling from the University of South Carolina in 2018. From 2019-



2024, Erin was a member of Dr. Priya Kishnani's Pompe Disease Clinical and Research Team at Duke University, where she participated in numerous research activities related to lysosomal and glycogen storage diseases, with her primary interests in newborn screening for Pompe disease and clinical variant interpretation. In addition to her research role, Erin provided clinical genetic counseling for patients across the lifespan with a variety of inherited metabolic disorders. In 2024, Erin stepped down from her role on Dr. Kishnani's team to pursue her PhD in Molecular Genetics and Microbiology at Duke University, where she continues to contribute to research related to Pompe disease.

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## **2025 is the year of #PompePower!**

Living with Pompe undeniably comes with challenges, but it also cultivates incredible strength and resilience. It teaches the power of perseverance, the value of adaptability, and the importance of celebrating every victory, no matter how small. Each day you face obstacles and push through, you redefine what strength truly means. Your journey inspires others, proving that even in adversity, there's power, purpose, and the ability to rise beyond limits.

For 2025, when posting on social media about Pompe disease or the AMDA, include #PompePower to share your strength with us!

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At the cusp of a new year, we find ourselves filled with aspirations and the desire to embrace change. **Resolutions** are more than mere promises; they are commitments to personal growth and well-being. While we set goals for our careers, relationships, and health, it is crucial to anchor these resolutions in self-care. **Self-care** isn't a luxury; it's a necessity. It's about recognizing and honoring our needs, whether it's through mindful relaxation, healthy habits, or nurturing our passions. By investing in self-care, we cultivate resilience and enhance our capacity to meet life's challenges with grace. As we navigate our resolutions, let's remember to celebrate small victories and cherish moments of contentment. This year, let's commit to **being kinder to ourselves, embracing each step of our journey, and prioritizing our overall well-being:**

**New Year Resolutions:** Symbolizing fresh starts and affirmations for the year ahead. New year's resolutions help kick-off healthy habits placing us squarely on a path of health and renewal. New beginnings don't always have to do with our physical self, sometimes we challenge ourselves to new adventures or lifestyles or all the things we want to do

differently this time. Be on the lookout for our February *Talking With Your Pompe Peeps* series with tips to help guide you through yours!

**Hobby Month:** The perfect time of year to pick up a new hobby or get back into an old one that you haven't thought about in a while! The great thing about having a hobby is that it can take your mind off the pressure of work or other daily stresses. Finding a good hobby and giving yourself time to take part in it on a daily or weekly basis is sure to make you happier, and may even be healthier.

**Self-Care Month:** It's much too easy to get carried away by life's troubles and forget to appreciate yourself. Self-Care Month is a time to appreciate yourself and invest time in conscious physical, spiritual, and psychological growth. It involves prioritizing your happiness and well-being. Your needs won't take care of themselves, pay attention to yourself on this amazing holiday.

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## Check out these Wellness and Fitness videos on our YouTube Channel!

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### AMDA/IPA International Pompe Patient & Scientific Conference 2024



Exercise Recommendation  
for LOPD

With:  
Keyuna (Coach K) Milam

Milam

### Exercise Recommendation for LOPD with Keyuna "Coach K" Milam

**Title:** Exercise Training for  
Patients with Pompe disease  
**Speaker:** Keyuna (Coach K)

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### Talking With Your Pompe Peeps Session 2 with Tiffany House

**Title:** Balancing Pompe (School,  
Work & Life): How Do You  
Balance Yours?

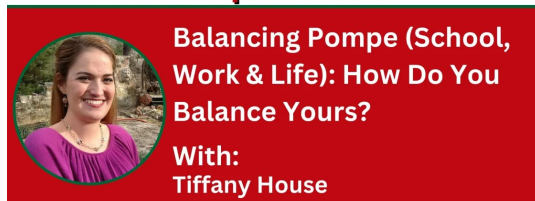
**Date:** Tuesday, September 24,  
2024

**Moderator:** Tiffany House

#### **Summary:**

This Open Forum Discussion will focus on exploring the various

### Talking With Your Pompe Peeps Series



Balancing Pompe (School,  
Work & Life): How Do You  
Balance Yours?

With:  
Tiffany House

ways members of the Pompe community juggle all of the things life throws their way. Come join the conversation!

**AMDA/IPA International Pompe Patient  
& Scientific Conference  
2024**



**Diet and Exercise  
Interactions for the  
Treatment of Pompe Disease**

**With:  
Dr. Mark Tarnopolsky**

**Diet and Exercise  
Interactions for the  
Treatment of Pompe  
Disease with Dr. Mark  
Tarnopolsky**

**Title:** Diet and Exercise  
Interactions for the Treatment

of Pompe Disease

**Speaker:** Mark Tarnopolsky, MD, PhD

**Summary:**

Pompe disease causes progressive muscle weakness due to issues like impaired autophagy, mitochondrial dysfunction, inflammation, and reduced protein synthesis, which exercise training can help improve. Studies show that endurance and resistance training (ENDUREX) enhance fitness, function, and strength in late-onset Pompe disease (LOPD) patients. A higher protein intake, greater than 1.2 g/kg/day with high biological value proteins, is recommended, especially post-exercise, along with monitoring and addressing vitamin D and B12 deficiencies. Although the ketogenic diet showed no benefit in mice, a ketone precursor, 1,3 butandiol, improved autophagy and function.

**Aquatic Exercise Webinar**

**Title:** Aquatic Exercise:  
Applications for Pompe Disease

**Speaker:** Dr. Kendra Lucas, PT,  
DPT, Aquatic Physical Therapist  
at Kettering Health, Ohio

**Webinar Overview:**

The unique properties of water provide surprising benefits for individuals struggling with gait difficulty, muscle weakness, cardiorespiratory impairments, balance problems and other symptoms associated with Pompe disease. Aquatic physical therapy offers a safe alternative to land-based programs. The water's buoyancy off-loads joint pressure, allowing you more freedom to move with less energy exertion. Join Dr. Lucas to learn much more about how aquatic exercise can help you stay active and healthy so you can live your best life.

**AMDA Webinars**



**Aquatic Exercise:  
Applications for Pompe  
Disease**

**With:  
Dr. Kendra Lucas**

**Respiratory Muscle  
Strength Training in Pompe  
Disease Webinar**



## AMDA Webinars



Respiratory Muscle  
Strength Training in  
Pompe Disease  
With:  
Dr. Harrison Jones

**Title:** Respiratory Muscle  
Strength Training in Pompe  
Disease

**Speaker:** Harrison N. Jones,  
PhD, BRS-S, CCC-SLP

We also have mini fitness videos and tips on our:

Instagram:

[@amda\\_association](https://www.instagram.com/amda_association)

Facebook:

[Acid Maltase Deficiency Association - AMDA](https://www.facebook.com/AcidMaltaseDeficiencyAssociation)



## Daily Observances

**National Peanut Butter Day, January 24th:** What is your favorite high-protein peanut butter snack? **Let's swap and share recipes!** Do you prefer creamy or chunky? With or without jelly, and what flavor jelly?



**National Croissant Day, January 30th:** Croissants are flaky buttery, crescent-shaped pastry that are crispy on the outside and soft on the inside. How do you like to enjoy yours?

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## 2024 At A Glance

2024 was a busy year, here is what the AMDA was up to:

- AMDA Hosted Webinars: 1 (due to planning/hosting 2024 AMDA/IPA International Pompe Patient & Scientific Conference, May 2024)
  - Attended Other Webinars: 5
  - Hosted TWYPP Series: 3
  - Non-Industry Meetings: 14
  - Meetings with Industry: 32
  - AMDA Meetings: 52
  - Panels/Working Groups Collaboration: 35
  - Hosted 2024 AMDA/IPA International Pompe Patient & Scientific Conference: 1
  - Attended Other Conferences, Symposiums, Coalitions & Consortiums: 19
- 

## 2024 AMDA/IPA Conference and Pull for Pompe Photo Galleries Now LIVE!

Photo galleries for the 2024 AMDA/IPA International Pompe Patient & Scientific Conference and 2024 Pull for Pompe fundraiser event are now up on the AMDA website! To view these photo galleries go to the links below.

[2024 AMDA/IPA International Pompe Patient & Scientific Conference Photo Gallery](#)

[2024 Pull for Pompe Photo Gallery](#)

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## Why should I get genetic testing if

# I already have a diagnosis?

NORD (National Organization for Rare Disorders) has created a new animated video, with both English and Spanish versions, to explain why it may be important for people with rare diseases to have genetic testing, *even if they already have a diagnosis*. The videos are free and available to all on the [NORD YouTube Channel](#).

## WHAT DOES the GENETIC TESTING PROCESS LOOK LIKE, and WHY is GENETIC COUNSELING IMPORTANT?



### AMDA

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