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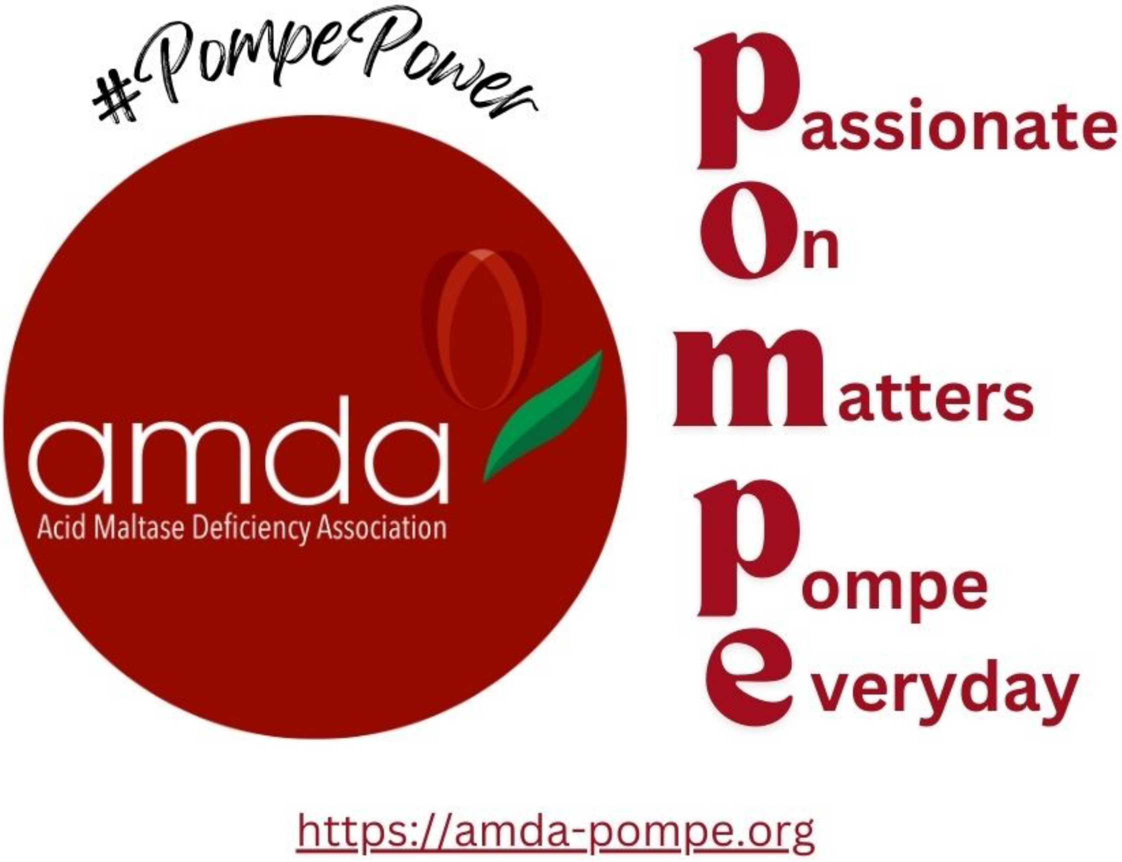
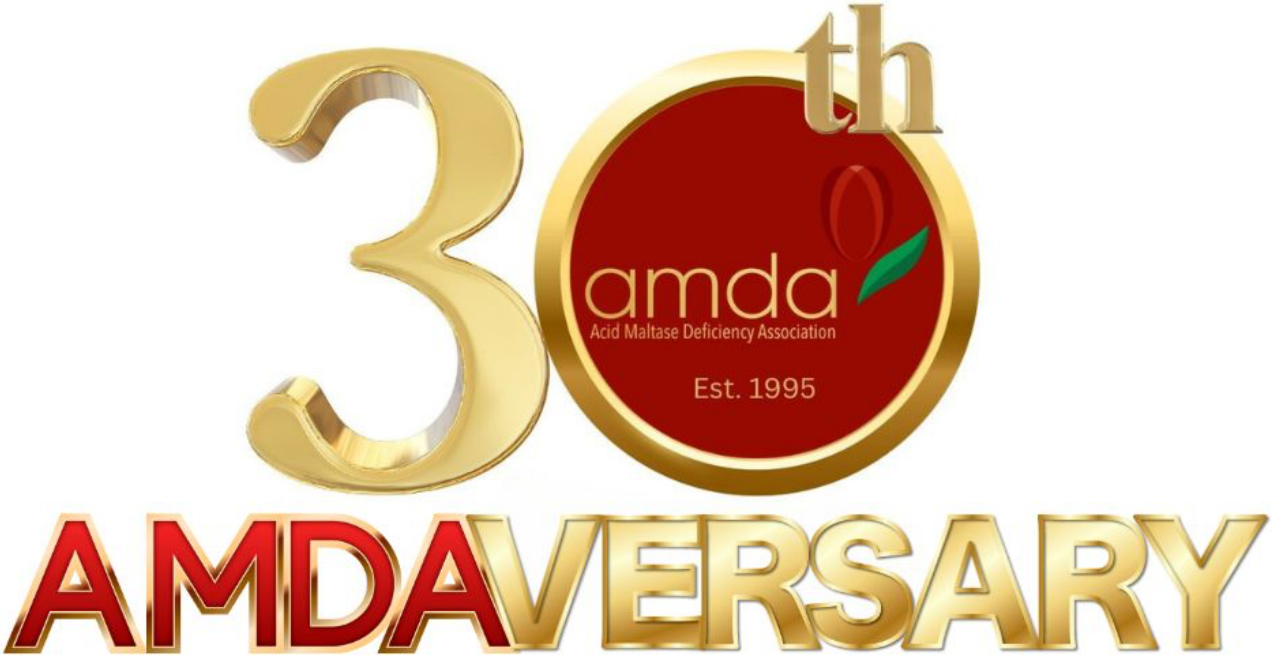
Videos Conferences News AMDA Publications
Webinars Talking With Your Pompe Peeps





May 2025

Upcoming Webinar

The Role of Genetic Counselors on the Pompe Disease Care Team

Join us **Monday, May 12, 2025** at **1 PM CT / 2 PM ET** for an informative and engaging webinar that shines a spotlight on a unique and essential healthcare provider: the **genetic counselor**. Genetic counselors are specially trained professionals who play a key role in care management, especially for individuals and families navigating complex genetic conditions like Pompe disease. In this session, you'll learn: who genetic counselors are, what they do and how they support patients, and how they can help you and your loved ones better understand and manage a Pompe disease diagnosis.

The webinar will conclude with a live Q&A session featuring our speaker, Erin Huggins—a board certified genetic counselor with extensive experience working with the Pompe disease community.

Whether you're newly diagnosed, a caregiver, or a healthcare provider, this session is a valuable opportunity to gain insights and ask questions directly from an expert in the field.

Register Here!

Webinar Details

Title: The Role of Genetic Counselors on the Pompe Disease Care Team

Date: Monday, May 12, 2025

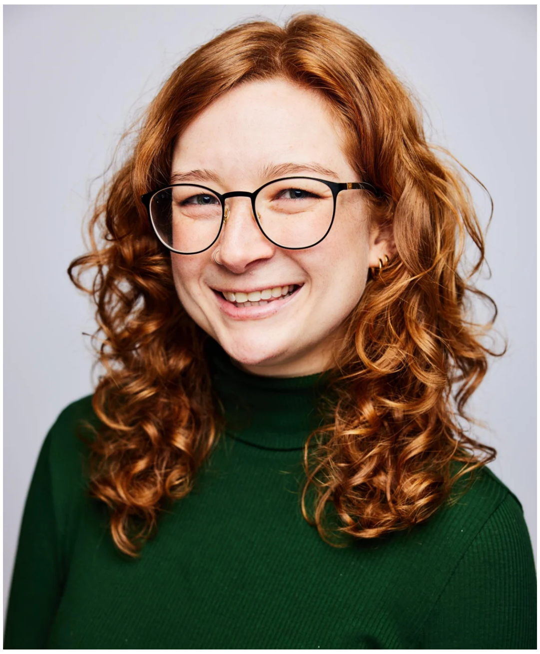
Time: 1 PM CT / 2 PM ET

Speaker:
Erin Huggins, MS, CGC

Webinar Overview:
This webinar focuses on a unique type of clinician: the genetic counselor. Genetic counselors are specially trained providers that can have an important role on your care management team.

In this webinar, we'll describe who genetic counselors are, what they do, and how they can help you and your family navigate a diagnosis of Pompe disease. At the end of the webinar, there will be a Q&A session with the speaker, a board-certified genetic counselor with several years of experience in caring for individuals with Pompe disease.

SPEAKER BIO



Erin Huggins, MS, CGC

Erin Huggins is a board-certified genetics 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.

Talking With Your Pompe Peeps!

amda
Acid Maltase Deficiency Association

Passionate
On
Matters
Pompe
Everyday

Join us for the next *Talking With Your Pompe Peeps* session!

Traveling or even just getting through the day with Pompe disease can come with unique challenges—but you're not alone. In our upcoming *Talking With Your Pompe Peeps* session, **Tuesday, May 27, 2025** at **1 PM CT / 2 PM ET**, we'll be sharing helpful strategies for managing life at home and on the go with more comfort and confidence. Moderated by **Tiffany House**, President of the AMDA and fellow community member who understands the ins and outs of living with Pompe, this session is titled **Tips & Tricks for Home & Travel**.

We invite **you** to bring your own experiences—whether it's a packing hack, a travel tip, or advice for involving caregivers—so we can all learn from and support one another. Let's connect, share, and grow—together.

**** This session will be recorded and posted on the AMDA YouTube Channel, by attending, you are giving the AMDA permission to post your image on its channel.***

Register Now!

Meeting Details

Title: Tips & Tricks for Home & Travel

Date: Tuesday, May 27, 2025

Time: 1 PM CT / 2 PM ET

Moderator:
Tiffany House

Summary:
In our upcoming discussion with the Pompe community, we’ll be sharing helpful tips and tricks for managing home and traveling with more ease and confidence. Topics will include preparing for rest breaks, packing medical essentials, and using adaptive tools to stay comfortable on the go. We’ll also talk about how to involve caregivers and plan flexibly to handle the unique challenges of traveling with Pompe disease.

2025 POMPE FAMILY MEET UP

Pediatric Pompe patients (IOPD & LOPD) and their families are invited to join us for a weekend of fun and community building

Location:
Camp Pyoca
886 E County Rd. 100 S
Brownstown, IN 47220

OCTOBER 10-12

VENUE IS WHEELCHAIR ACCESSIBLE

Registration is now officially open for Grant's Giants' 2025 Pompe Family Meet Up! Join them for a weekend of community building and fun in beautiful southern Indiana!

Who: Families affected by Pompe disease; this includes both IOPD and LOPD. **Preference will be given to Pediatric**

families, but all families are welcome to attend.

What: A weekend of community building for Pompe families.

When: October 10-12, 2025
Check In- October 10th at 3pm
Check Out- October 12th at 3pm

Where: Camp Pyoca
886 E County Rd 100 S
Brownstown, IN 47220

Please reach out to grantsgiants@gmail.com with any questions you may have.

Register Here!

May Observances

Monthly Observances

Better Sleep Month: This month is a perfect reminder to make snoozing a top priority! Sleep isn't just about catching Z's—it's a cornerstone of your overall health and happiness. From boosting mood to sharpening focus, good sleep can transform your days (and nights!). This month, explore ways to create the ultimate sleep sanctuary: dim the lights, banish screens before bedtime, and dive into the coziest pillows and blankets. Plus, don't forget the importance of a consistent sleep schedule and relaxation rituals. Sweet dreams start with smart choices! Celebrate the month by **sharing your favorite sleep tips and routines using #BetterSleepMonth.** Let’s all dream big!

Daily Observances

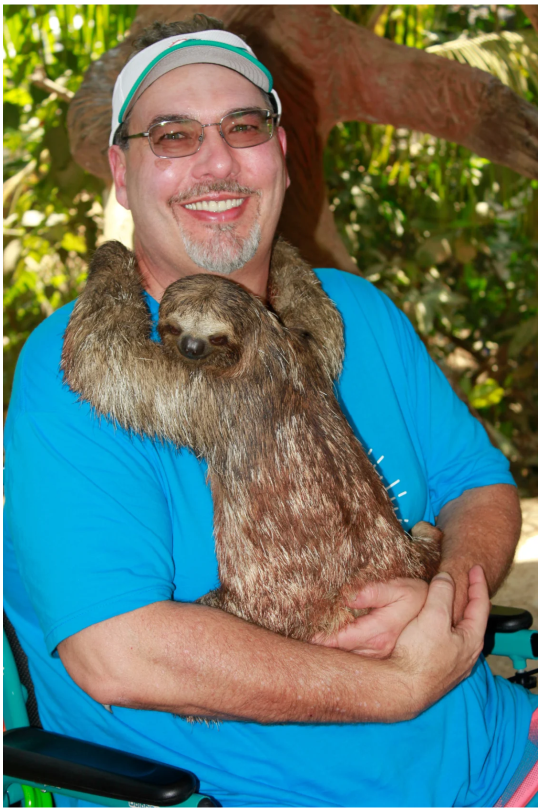
National Pet Week, May 4-10: Any time, any place, any day—our pets have a magical way of brightening our world. No matter how tough life gets, they’re always there with comfort, sympathy, and unconditional understanding when we need it most. Celebrate National Pet Week by honoring our fun-loving furry (or feathered or scaly) family members who bring so much joy to our lives. All they ask for is a snuggle… and maybe the occasional treat! **Share your pet photos on social media, #NationalPetWeek!**

National Nurses Day, May 6th/Week Observance, May 6-12: A heartfelt tribute to the incredible dedication, compassion, and expertise of nurses who tirelessly care for us in our times of need. Nurses are the backbone of healthcare, providing comfort, hope, and healing with unwavering strength and grace. This week, let’s honor the invaluable contributions of these healthcare heroes—whether it’s by expressing gratitude, sharing their inspiring stories, or simply taking a moment to say, “thank you.” **Use #NationalNursesWeek to celebrate and give a shout-out to the nurses who make a world of difference every single day.**

National Visit Your Relatives Day, May 18th: In the whirlwind of our busy lives, it's easy to lose touch with the relatives who mean so much to us. This special day serves as a reminder to pause, reconnect, and spend meaningful time with the family members we hold dear. Life is too precious not to make room for those we haven’t seen or spoken to in a while. Thanks to modern technology, staying close to loved ones—even those miles away—has never been easier. Sharing our lives across the distance can feel as though they’re right beside us, bringing families closer than ever. Let’s make the time to cherish these connections!

National Hamburger Day, May 28th: It's time to fire up those grills because burger season is officially here! Americans have a love affair with burgers that's been sizzling for generations. Even if red meat isn't your thing, there's no shortage of ways to enjoy a burger that hits the spot. Whether it's charbroiled to perfection, kissed lightly by the flames, or grilled to juicy glory, there's a flavor ready to win you over. **Share your burger masterpieces and join the party using #NationalBurgerDay on social media.** Let's make this day extra cheesy!

National Creativity Day, May 30th: Whether you're an artist, writer, musician, filmmaker, blogger, photographer, graphic designer, or one of countless other creative souls, let your imagination run wild! Dive into your creative passions and/or show support for other innovators and artists—let's make the world a more vibrant place together! **Share your creativity on our social media at #NationalCreativityDay**



Patient Spotlight

Dwayne M Wilson

Hi everyone, my name is Dwayne; I am 57 years old. I was diagnosed with Late Onset Pompe disease (LOPD) in November 2018 when I was 50 years old. I live in Southern California in the City of Irvine with my wife and mother-in-law. I am a father to four sons, still mourning the loss of our oldest son who passed away from a brain tumor at age 22, about 7 months before my diagnosis. I am a grandpa and have 2 granddaughters, ages 2 and almost 4. My passion is to connect with others to spread **Hope** and **Positivity** about living with Pompe disease and putting a smile on peoples faces.

Dwayne's Pompe story

My wife says I got Pompe disease so that I could make more friends, and she is right! Looking back, I know I was born with it.

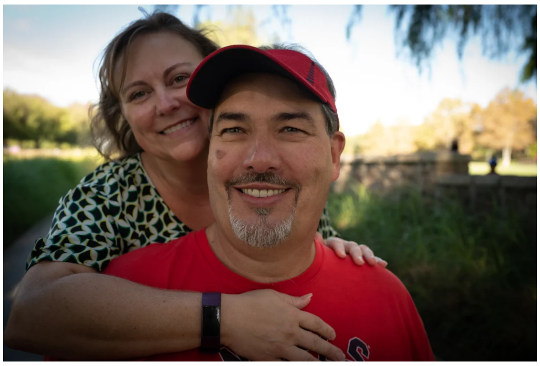
Growing up, I was always a husky kid, had difficulty with physical activities, unable to do a pull-up, push-up or do the monkey bars. I was slow and uncoordinated and had no balance. Most of the time it was just related to being overweight or out of shape as a teenager. As I entered my 20’s, I worked a fulltime job and eventually joined a gym to be healthier. I lost some weight but ended up having two knee surgeries that slowed me down even more.

In 2004, I went to my doctor to have a routine physical to apply for life insurance. While at the doctors, he noticed my liver enzymes, ALT and AST were high. Unfortunately, that denied me the life insurance that I was applying for at the time. No further tests were performed on my liver. Later that year, my father passed away from prostate cancer. After this, I went back to my doctor to have tests run to see if my prostate had any issues. My AST and ALT liver enzymes were still elevated so a liver biopsy was done. I had gallstones and my gallbladder was removed. Everything came back fine even though my liver enzymes were still elevated. The doctor just said “that’s just the way you are” at age 36.

It was not until I moved back to Southern California from Spokane, WA in 2017 that my more serious symptoms started to develop. My wife and I had moved into a second-floor apartment and after about 6 months I noticed that something was going on with my physical body. I realized that I was unable to walk up the staircase to the 2nd floor and had to grab the handrail, stepping up one leg at a time, with the same leg. I could not get up off the floor and had to push off with my arms to go from a seated position to standing like on a chair or the couch. I began to be more fatigued and exhausted by everyday activities, even noticing that when I laid down at night to sleep, my breathing was shallow.

Getting a diagnosis

In July 2018, I saw my primary care doctor who then sent me to see a Neurologist. It was then that I had an EMG and Nerve Conduction tests done to determine what was going on. The results said I had a Myopathy or a muscle weakness disease. During that meeting, I shared with my doctor that I had trouble breathing when I was in the pool with water up to my neck, it felt like someone was stepping on my chest. At that moment, the doctor said, “that sounds like Pompe disease”. He ordered a Pompe blood test and a muscle biopsy. He then referred me to a Neuromuscular specialist and on November 19th, 2018, I was diagnosed with Late-onset Pompe disease. Looking back now, since my diagnosis and my doctor discovering still high liver enzymes, I recalled they were also elevated as a teenager.



I was fortunate that it only took 6 months for my diagnosis, but once I received that diagnosis, I felt like a weight was lifted off my shoulders. So many things now made sense to me growing up as a kid. I was also relieved that there was a treatment for Pompe disease because I realized that many rare diseases do not have treatment.

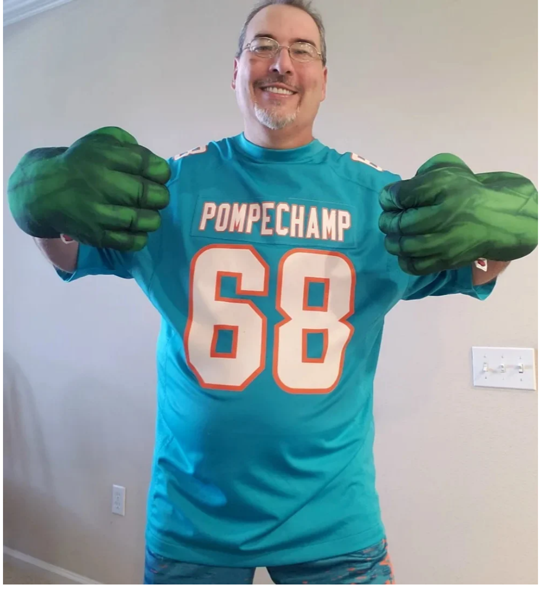
Living with Pompe disease

I like to think that everything in life happens for a reason, and I feel blessed that I was in the right place at the right time.

After diagnosis, my wife and I moved to a ground floor apartment. My doctor also prescribed a BiPap machine to assist my breathing, when sleeping. I was accepted into a clinical trial quickly, and began ERT one month from being diagnosed with Pompe Disease. I also began Physical Therapy and have done it on and off for the past 6 years. I have learned to understand how my body feels when I need to rest or to work out.

Once I had a diagnosis, I joined many Facebook Pompe support groups to become part of a community and to make friends. Having someone to talk to, listen to, and understand, really helps with my mental health. I have attended Pompe conferences to meet other patients and families going through the same journey as me. The conferences have also helped me learn more about Pompe and the on-going research for the disease.

Since being diagnosed, my perspective on life and what really is important to me, has changed. I believe that life is about having experiences and going on adventures, making memories with friends and



loved ones. Since October of 2022, I have gone to Hawaii-Oahu twice, been to Cancun and San Antonio and flew to Fort Lauderdale for a Miami Dolphins football game at Hard Rock Stadium. I traveled to Miami for a Western Caribbean cruise to hold a sloth on Roatan for my birthday last year. We even have plans for a Mexican Riveria cruise in Jan of 2026.

Being an Advocate for Pompe

With over 600 different gene mutations that can cause Pompe disease, there is not "one size that fits all "to treat a patient. Not all of us experience the same symptoms and we do not all have the same abilities or lack thereof. There is such a wide range of diagnosis for Pompe disease. We must share and advocate for each other. What works for one, may not work for another. Sharing my experience within our community is my passion. There is so much hope for future generations with newborn screening and earlier, better treatments.

With that being said, I became a patient speaker to share my story. I am an MDA Ambassador with the Muscular Dystrophy Association to spread awareness about all neuromuscular diseases. I am a writer who has written a column "On the Road to Pompe" on the Pompe Disease News website for a few years. I don't want Pompe disease to be in the driver's seat of my life. I continue to spread awareness about living with Pompe disease on social media.

You can find me on Social Media:
Instagram: @smashingpompe
X: SmashingPompe
LinkedIn: Dwayne M Wilson
Facebook: Dwayne M Wilson & Smashingpompe

AMDA

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