

## About the AMDA

The Acid Maltase Deficiency Association (AMDA) was founded in 1995 by Randall and Marylyn House. They became dedicated to finding answers about acid maltase deficiency, also known as Pompe Disease, when their daughter was diagnosed with the disease at the age of 12 after spending over a decade looking for a diagnosis.

The House family formed the AMDA to promote research into Pompe Disease with the goal of finding a treatment or cure. They also wanted to educate and advocate for others with Pompe Disease.

Today, Tiffany serves as the President of AMDA. She is committed to working to better the lives of Pompe patients and their families.

Today, treatment for Pompe is now a reality in the form of enzyme replacement therapy (ERT). There are also new treatments on the horizon: gene therapy, chaperone therapy, substrate reduction therapy, and many others.

Patients are living longer, healthier lives. And the AMDA is here to support and advocate for all patients and their families.

And there is also exciting on-going research for improving disease management, and improving the quality of life for all patients.



## Newborn Screening and Pompe Disease

The House family, and the majority of Pompe families like theirs, spent years trying to find out the cause of their child's many medical problems. With newborn screening and follow-up testing, parents learn early if their baby has Pompe Disease.

Pompe Disease mainly affects heart, skeletal, and breathing muscles. It gets worse over time without treatment. With newborn screening, parents do not have to search for answers. Treatment can begin early.

In the U.S., newborns are screened at birth for a number of serious medical conditions. Newborn screening for Pompe Disease is being conducted in many states. But screening is not yet available in all states. The AMDA and other organizations are working to make sure that all states screen for Pompe Disease.

Learn more about newborn screening and Pompe Disease at:

- HRSA-Pompe  
<https://newbornscreening.hrsa.gov/conditions/pompe-disease>
- Search which states test for Pompe  
<https://www.babysfirsttest.org/>



**Acid Maltase Deficiency Association**

PO Box 700248  
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**For more information on Pompe Disease, please visit the AMDA website:  
[www.amda-pompe.org](http://www.amda-pompe.org)**

# Finding out that your baby has Pompe Disease



You have waited months for the arrival of your newborn. You probably felt excited about starting your life with this new baby. Then, you learn that your baby's newborn screening test was not normal (out-of-range) for Pompe Disease. With more testing, you find out that your baby has Pompe Disease.

What do you do now? Where can you go for more information? How do you connect with other parents in the same situation as you are in now? Who can help you solve problems that may arise?

You will get through this. The Acid Maltase Deficiency Association (AMDA) is a non-profit patient support organization that can provide you and your family with the information, support, and resources you need to move forward. We are here to help you and your family as you care for baby.

**“You are not alone. We are here to help.”**

**Your baby's diagnosis of Pompe Disease may be overwhelming. You may feel sad or scared and not know where to turn. The AMDA's goal is that no family will ever be alone when questions and needs arise. There is hope for the future. We are here to help.**



## AMDA Resources

We have many resources and programs to support you and your family during the coming days, weeks, months, and years. These include:

**Patient Advocate:** The AMDA's Patient Advocate, Marsha Zimmerman, is a registered nurse. She has been working as a Pompe Patient Advocate since 2001. She has experience assisting families with everything from access to therapy to education and communication with medical professionals. She also provides emotional support.

Whatever questions or issues you may have, Marsha Zimmerman can help. Her email address is:

[marsha.zimmerman@amda-pompe.org](mailto:marsha.zimmerman@amda-pompe.org)

**Website:** Provides current research and treatment advancements, disease management, research publications, other important news.

**Newsletters:** The AMDA sends out Newsletters and News Updates through Mail Blasts to all of our Registered patients. The newsletter may include patient stories, research updates, industry updates, research study updates, recent research publications, and international Pompe news.

**Teleconference/Webinar Program:** The AMDA's teleconferences and webinars are tools to aid the exchange of information between the Pompe patient community and Pompe experts. The AMDA hosts 4 or 5 webinars a year for the Pompe community. All teleconferences and webinars are on the AMDA website.

**Patient/Scientific Conference:** The AMDA sponsors a scientific conference every 3 to 5 years. These conferences are traditionally held in San Antonio, TX. Patients, physicians, and researchers come from around the world. The conference is an opportunity to bring together the entire Pompe community to exchange information about recent developments in Pompe Disease and best practices for disease management.

**Mentor Program:** The program puts patients and their families in touch with other people who have been through similar experiences. AMDA volunteer mentors from the Pompe community are available to help patients and families in dealing with a new diagnosis. They can share experiences, feelings, and resources related to Pompe Disease. For more information, please contact [Morgan.Burroughs@amda-pompe.org](mailto:Morgan.Burroughs@amda-pompe.org)

**To receive information, register with AMDA at:  
[www.amda-pompe.org](http://www.amda-pompe.org)**