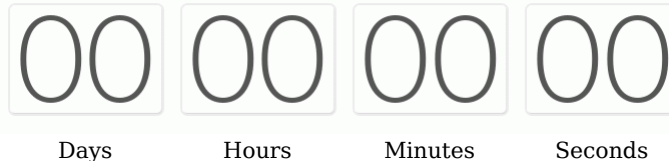




[Duke Webinar Registration](#)
[Spark Therapeutics Webinar Registration](#)

**Webinar: How to Manage LOPD Patients Diagnised
through NBS**

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



Title: New insights into Pompe disease since the advent of NBS

Date: Tuesday August 10, 2023, at 1 pm CT/ 2 pm ET

Speakers:

Dr. Priya Kishnani, MD

Erin Huggins, MS, CGC

Webinar Overview:

An overview of the lessons learned from newborn screening for Pompe disease and its impact on disease management.

**Dr. Priya Kishnani,
MD**

Speaker Bio:

Dr. Priya Kishnani, MD, is Chief of the Division of Medical Genetics in the Department of Pediatrics, and Professor of Molecular Genetics and Microbiology at Duke University Medical Center. She holds certifications from the American Board of Medical Genetics and the American Board

of Biochemical Genetics. She holds numerous appointments at Duke University Medical Center, serving as Director of the Glycogen Storage Disease Clinical and Research Program, Director of Clinical Trials, and Director of the YT and Alice Chen Center for Genomic Research and of the Metabolic Clinic.

Her passion is to establish the highest quality of care and treatment by understanding the emerging natural history of individuals through investigator-initiated studies and clinical research trials with a focus on glycogen and lysosomal storage disorders. She has published extensively on these conditions, through research publications, textbooks, and scientific reviews. Dr. Kishnani was involved in the clinical development of alglucosidase alfa for the treatment Pompe disease (PD), resulting in its US Food and Drug Administration (FDA) approval in 2006, and in the development of avalglucosidase alfa-ngpt as a second-generation enzyme treatment, which was approved by the FDA in 2021. She also focuses on next-generation therapies such as gene therapy and other modalities to treat PD.

As well as having an interest in long-term complications of PD, Dr. Kishnani and her team at Duke University played an integral role in the nomination and approval for the addition of PD to the Recommended Uniform Screening Panel for newborn screening in the United States in 2015.



Erin Huggins, MS, CGC

Speaker Bio:

Erin Huggins is a board-certified genetic counselor at Duke University in the Division of Medical Genetics, Department of Pediatrics. 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. She provides clinical genetic counseling for patients across their lifespan with a variety of inherited diseases including glycogen storage diseases, lysosomal storage diseases, and other inborn error of metabolism. In addition to her clinical role, Erin is a member of Dr. Priya Kishnani's research team and is involved in a number of research activities related to metabolic disease. Her primary areas of interest are newborn screening for Pompe disease and clinical variant interpretation in rare diseases. Erin is originally from Charleston, SC, and currently resides in Durham, NC.



[**Register Now!**](#)

September Webinar

Spark Therapeutics Webinar: Trial Expectations

Title: Spark Therapeutics Webinar: Trial Expectations - What to Expect

Date: September 27, 2023, at 1 pm CT/ 2 pm ET

Speakers:

Jacose Bell

Webinar Overview:

This presentation, developed by Spark Therapeutics, takes audience members through the introductory science of AAV gene therapy. We will examine how AAV theoretically works in a liver-directed approach, how that is different from other investigational forms of cell and gene therapy, and what the challenges and potential risks are with AAV gene therapy. The presentation will then focus the remainder of its time on the role of clinical research and things people should consider if they are interested in participating in clinical research for investigational gene and cell therapies.

Jacose Bell

Speaker Bio:

Jacose Bell is Sr. Patient Advocacy Lead at Spark Therapeutics. Jacose joined Spark in April of 2021, and has had the privilege of supporting relationships and partnership with the greater Pompe community ever since. Jacose is passionate about raising awareness and providing education about AAV gene therapy as part of that role.



Register Now for the September Webinar!!!



IPA Calls to Action

CALL to participate in IPA Project

Every Move Counts

Happy International Pompe Day!
#TOGETHERWEARESTRONG



At International Pompe Day the International Pompe Association kicked off the Every Move Counts project. This project focuses on raising awareness of the importance of movement. We all have different abilities and levels of disability, but the important thing to remember and focus on is that “Every Move Counts.” We should celebrate what we can do, not focus on what we can’t.

We want to show all the ways that individuals in the Community show that Every Move Counts. Several of you already submitted valuable contributions that will be used in this project. More participants are still needed to reach our goal. · Do you have modified physical therapy that you can share? · Do you keep moving by playing with your dog? · Do you focus on music or other hobbies to keep yourself moving? The ways that we can show that Every Move Counts will be as diverse as our community! By sharing how YOU believe Every Move Counts, you can help others in our community! If you are interested in being part of this project, please reach out to us at info@worldpompe.org.

IPA is collecting these stories, videos, pictures, etc. and will then work with experts in the field to compile a set of resources that will be available on the IPA website. We will need representatives from all around the world, and across the disease spectrum so please consider joining our Project!

Every Move Counts also relates to developments in Pompe treatment and management, and our daily lives. Therefore, our SECOND project is that we are requesting submissions for a 2024 Calendar that we will create. We would like to ask our community to submit artwork for a calendar that we will distribute to our members and to others in the community to remind us throughout next year to not give up—Every Move Counts! In research, in drug development, and in our lives. So please submit artwork to

info@worldpompe.org that shows what Every Move Counts means to you!

Every Move Counts Calendar Submissions

Every Move Counts also relates to developments in Pompe treatment and management, and our daily lives. Therefore, our SECOND project for International Pompe Day is that we are requesting submissions for a 2024 Calendar that we will create.

We would like to ask our community to submit artwork for a calendar that we will distribute to our members and to others in the community to remind us throughout next year to not give up—Every Move Counts! In research, in drug development, and in our lives. So please submit artwork to info@worldpompe.org that shows what Every Move Counts means to you!

Together
WE ARE
STRONG



Patient Stories

Share Your Pompe Disease Story!

We want to hear your story! Share your experiences living with Pompe Disease to inspire and support others in the community. Whether you're a patient, caregiver, or family member, your perspective matters. Your privacy will be respected, and your story can make a difference.

Why Share Your Story?

Living with Pompe Disease can present unique obstacles and triumphs that only those who have experienced it firsthand truly understand. By sharing your journey, you can offer valuable insights and inspiration to individuals and families who may be newly diagnosed or seeking support in their own path. Your story has the power to make a meaningful impact on the lives of others and contribute to a greater understanding of Pompe Disease within the broader community.

How to Participate

We invite you to share your journey with us in the form of a patient story. Whether you are a patient yourself, a family member, or a caregiver, your perspective is valuable. Your story could encompass various aspects, such as:

1. **Diagnosis and early experiences:** How were you or your loved one diagnosed with Pompe Disease? What were your initial thoughts and emotions?
2. **Navigating treatment and management:** Share your experiences with various treatments, therapies, or lifestyle adjustments you have incorporated into your daily routine.
3. **Overcoming challenges:** Tell us about the challenges you faced and how you found strength and resilience to overcome them.
4. **Moments of triumph:** Share your moments of victory, big or small, that have brought joy and fulfillment into your life.
5. **Support and community:** Highlight the impact of support groups, family, friends, or healthcare professionals on your Pompe journey.

Your Privacy Matters

We understand that sharing personal stories requires trust and vulnerability. Rest assured, we are committed to maintaining your privacy and will anonymize any information shared if you wish to do so. Your story will only be used for the purpose of raising awareness about Pompe Disease and inspiring others within our community.

How to Submit Your Story

Submitting your story is simple. You can either write your story in an email or send us a recorded video. Please feel free to include any pictures or visuals that you believe would complement your narrative.

Email your patient story to info@amda-pompe.org. If you are sending a video, please upload your video to youtube and send a link with your submission.

Your story can make a difference, and we can't wait to hear from you!

Thank you for being an integral part of our Pompe Disease community.

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

**MAY 3-5
2024**

San Antonio,
Texas

*Stay tuned for
more details!*

**SAVE
THE
DATE**



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