

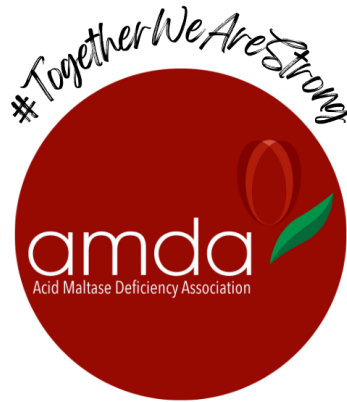


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September 2024

AMDA Newsletter

Boston Children's Hospital Brain Imaging Research Study Looking For More IOPD Patients

Jaymin Upadhyay and his team at The Boston Children's Hospital are looking for more IOPD patients to enroll in their Pompe Disease Brain Imaging Research Study. If you have a family member or know of someone diagnosed with IOPD please consider enrolling/informing them about enrolling in this study. See flyer below for more information.

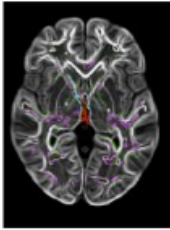
"All research procedures (non-contrast musculoskeletal and brain MRI) will be paid for by the study. They or their clinician will have access to their MRIs. There is a up to \$1500 (travel) + \$250 study stipend. If participants do not or cannot undergo MRI, they can still enroll in the study and undergo other non-invasive study procedures.

Our age range is 4-55 years of age for the entire study, but we are especially looking for IOPD patients to enroll."

-Jaymin Upadhyay, PhD

Assistant Professor, Harvard Medical School

Department of Anesthesiology, Critical Care and Pain Medicine



Thank you for your interest in our research study! Inside you will find the answers to many frequently asked questions about the study.

For more information, please contact our research group at

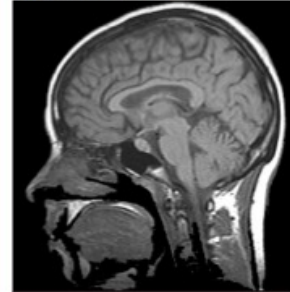
Email:
Jaymin.Upadhyay@childrens.harvard.edu
Wells.A.Herlan@childrens.harvard.edu

Phone:
917-736-1541



Pompe Disease Brain Imaging Research Study

 Boston Children's Hospital
Until every child is well™



Division of Genetics & Genomics,
Department of Medicine
&
Department of Anesthesiology,
Critical Care
and Pain Medicine

Q: Who is eligible?

A: Patients diagnosed with Infantile-Onset or Late-Onset Pompe Disease (IOPD/LOPD) or healthy individuals.

Q: What does this study involve?

A: During this study, we will ask you to complete questionnaires, cognitive task, motor task, give a blood and urine sample, and undergo MRI and fNIRS.

The study procedures will occur over the course of a single day. The amount of time will vary depending on whether you have a IOPD or LOPD diagnosis.

Participation in this research will not alter or interfere with any of the standard clinical procedures or treatment that one would normally receive if not partaking in this research. All research studies will take place at Boston Children's Hospital, McLean Hospital or New York Medical College.

Q: What if I live out of town, can I still participate?

A: Yes! If a patient lives outside of the Boston metropolitan area, our lab can help coordinate your visit to Boston. We will also provide reimbursement for travel-related expenses.



Q: Will I get paid to be part of this study?

A: Yes! Patient or healthy subject who participates in this study visit will be paid up to \$200.



Q: What is an MRI scan?

A: MRI (Magnetic Resonance Imaging) is a medical imaging technique that can take pictures of your body.

Q: What are the risks associated with MRI?

A: MRI does not involve any radiation and so there is no risk to most people. It does however involve a strong magnetic field, which although not harmful by itself, may cause implanted medical devices that contain metal to malfunction in patients who have them. Patients with any such devices are not eligible for the study. We will not inject or use any tracer or contrast agent in this study.

Q: What is an fNIRS scan?

A: fNIRS (functional near infrared spectroscopy) is an imaging technique that can also take pictures of your brain. fNIRS involves wearing a cap.

Q: What are the risks associated with fNIRS?

A: The risks are minimal, but does involve a laser. fNIRS is a very safe brain imaging technique.

AMDA/IPA International Pompe Patient & Scientific Conference 2024



Exercise Recommendation
for L



With:
Keyuna (Coach K) Milam

2024 AMDA/IPA International Conference Recordings Now Available

The recordings of the 2024 AMDA/IPA International Patient and Scientific Conference are now available to view on the [AMDA YouTube channel](#).

Explore the workshop on **Exercise Recommendations for LOPD** with Keyuna "Coach K" Milam presented at the conference. Coach K has exercise recommendations for all levels of impairment. Check the video out above!

Additional videos of the in-person workshops will be added at a later date.

2024 AMDA/IPA Conference Playlist



Join us **Tuesday, September 24, 2024** at **12 PM CT** for the second session of our *Talking With Your Pompe Peeps Series*. Moderated by **Tiffany House**, President of the AMDA, this session is titled, **Balancing Pompe (School, Work & Life): How Do You Balance Yours?** This Open Forum Discussion will focus on exploring the various ways members of the Pompe community juggle all of the things life throws their way. Come join the conversation!

****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 it's channel.***

[Register Here!](#)

Meeting Details

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

Date: Tuesday, September 24, 2024

Time: 12 PM CT

Moderator: Tiffany House

Get To Know The Moderator

Tiffany House

Tiffany House is **President, Acid Maltase Deficiency Association (AMDA)**, Inc., San Antonio, Texas; a non-profit advocacy organization for patients with Pompe Disease.

Hosts/spearheads biannual conference, communicates with industry, current on latest advancements in treatment and patient care, attends various scientific/patient conferences, and is Chair or member of various Working Groups or Committees.



Chairman/Board Member, International Pompe Association (IPA), Baarn, the Netherlands; an international Pompe patient support organization. The mission of the IPA

is to “Campaign for early diagnosis and effective, affordable and safe therapies. Strive to provide information and support to all patients, their families and others with interests in Pompe Disease.” Board Member; participates in regular conference calls with Board and industry, annual meetings, and face-to-face meetings with industry and scientific/medical experts.

Active/Voting Member, Lysosomal Disease Network (LDN) External Advisory Committee (EAC), National Institutes of Health (NIH) Rare Diseases Clinical Research Network (RDCRN), University of Minnesota, Minneapolis, Minnesota; participates in annual meetings and communications, evaluates Pilot Project proposals and Fellowship applications, reviews clinical research and opportunities with a focus on MPS, Pompe, and Fabry diseases and two cores: administrative and education.

Member, Lysosomal Storage Disorders (LSD) Advocacy Coalition; participates in monthly meetings to work together on state and federal issues such as Medicare Home Infusion, medical research and Newborn Screening as well as state and federal issues.

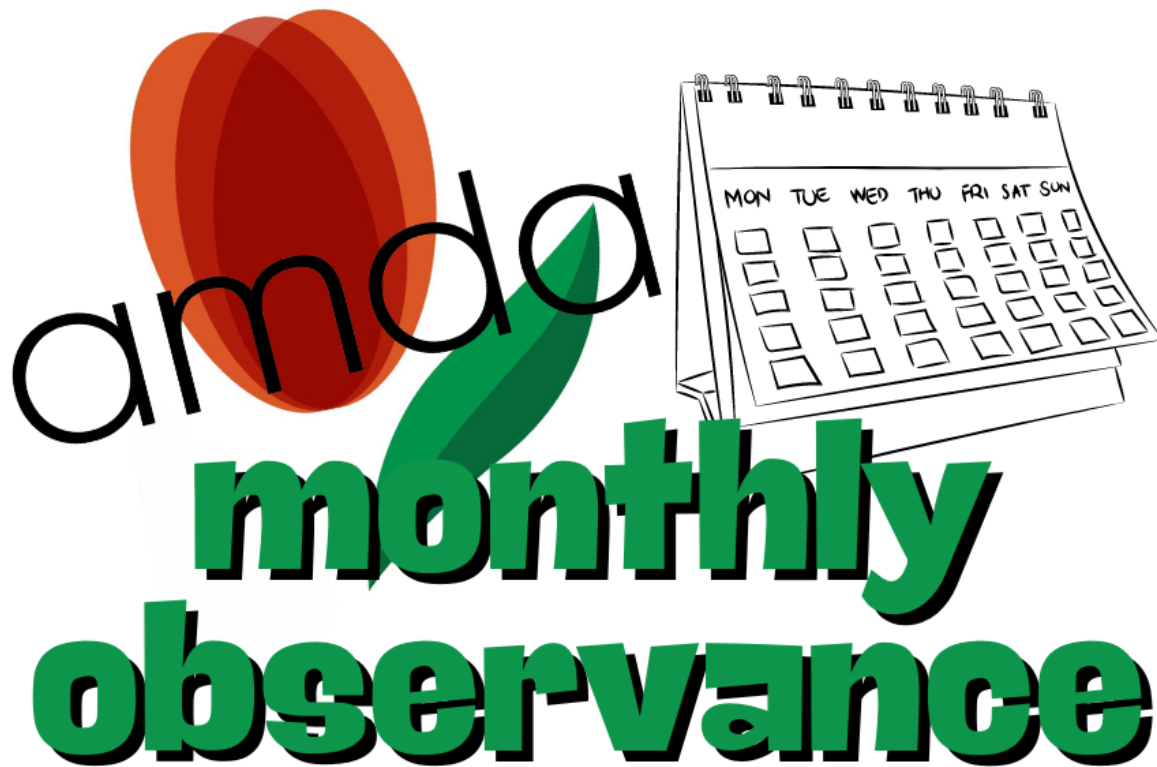
Panelist, Bayer Pompe Lived Experience Expert (LEE) Standing Expert Panel (SEP); collaborates with a broad geographical spread of experts to obtain ongoing deep insights into the disease characteristics, patients unmet needs and preferences, the current standard treatments, and the potential impact of novel therapies.

Newborn Screening Advisory Committee member.

Patient Representative/Voting Member of Four Advisory Committees (AC), Food and Drug Administration (FDA), Washington, D.C.; serves as the “voice of the patient.” Active team member reviewing Lumizyme (Pompe treatment). Committed to email correspondence, teleconferences, and in-person meetings in D.C. Lumizyme AC Voting Member, evaluated application with recommendations, October 2008. Patient Representative Role guest speaker, FDA Patient Representative workshops, 2009 and 2010.

Patient Representative, Niemann-Pick Type C and NAGS Deficiency AC, January 2010; Patient Representative, Direct-to-Consumer Genetic Testing AC, March 2011.

Tiffany received her Juris Doctorate (Suma Cum Laude) in 2011 at St. Mary’s University School of Law, San Antonio, Texas and an M.A. in English, 2007



National Disease Literacy Month

National Disease Literacy Month was created to improve one's overall understanding of their diagnosis and the services needed to make care decisions, focusing on children and their knowledge of the conditions they have and the care they need every day. Over 90% of children do not understand their disease or chronic illness. **Visit our [webpage](#) for resources:**

- [Talking With Your Pompe Peeps Session with Colleen Sackos: The Elementary School Experience: Building Success for IOPD/LOPD Children Through Collaboration and Partnership!](#)
- [AMDA Publications - AMDA Pompe \(\[amda-pompe.org\]\(http://amda-pompe.org\)\)](#)
- [Links - AMDA Pompe \(\[amda-pompe.org\]\(http://amda-pompe.org\)\)](#)
- [Youtube Channel](#)

If you have further questions or need additional assistance, please contact us at: info@amda-pompe.org

Newborn Screening (NBS) Awareness Month

On June 3, 2013, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) made the recommendation to add Pompe Disease to the federal Recommended Uniform Screening Panel (RUSP). In 2015, Pompe Disease was added to the RUSP. Despite this addition, screening for Pompe has not been implemented by every U.S. State and territory.

The AMDA is a strong advocate and supporter for the EveryLife Foundation's NBS campaign to include Pompe Disease to each State's NBS. "Early diagnosis and intervention through newborn screening saves lives." Explained Tiffany House, a Texas advocate and president of the Acid Maltase Deficiency Association. "Even a few days difference in the initiation of treatment can have a profound impact on the path that these children's lives will take."

Please refer to the links for more information:

[Newborn-Screening-and-Pompe-Disease-Final-post-04.05.24.pdf \(amda-pompe.org\)](#)

Newborn Screening - Pompe Disease - EveryLife Foundation for Rare Diseases: <https://everylifefoundation.org/newborn-screening-take-action/pompe-disease/>

TREAT YOURSELF!



National Ice Cream Cone Day
September 22nd



National Pancake Day
September 26th



National Coffee Day
September 29th



Celebrating National Ice Cream Cone Day, National Pancake Day, and National Coffee Day is a perfect way to treat yourself and embrace simple joys. These fun food holidays offer a chance to slow down, indulge in your favorite comfort foods, and create small moments of happiness. Whether it's enjoying the cool sweetness of an ice cream cone, savoring a stack of fluffy pancakes, or relishing a warm cup of coffee, each celebration is a reminder to take a break from the daily grind and appreciate life's little pleasures.

Treating yourself to these fun traditions is not just delicious—it's a way to recharge and celebrate everyday delights!

Invitation to Enroll Into the IPA/Erasmus MC Pompe Survey

If you have been diagnosed with Pompe disease and are at least 16 years old or older and live in the US, the AMDA would like to invite you to participate in the IPA/Erasmus MC Pompe Survey (the Pompe Survey).

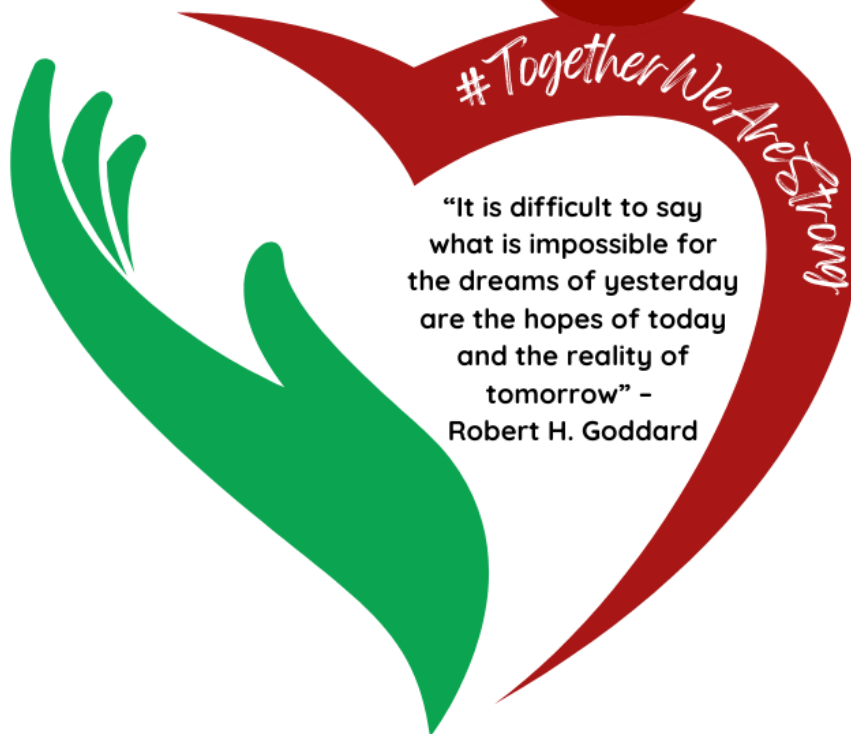
The Pompe Survey is a collaboration between the Pompe Center of the Erasmus MC and the International Pompe Association (IPA) that started in 2002. Over the past two decades many patients from all over the world have participated in this annual survey. The data collected by this survey is very useful for both clinicians and researchers, so we encourage participation.

If you would like to participate in the Pompe Survey, please email Marsha Zimmerman (marsha.zimmerman@amda-pompe.org) with your full name and home address.

The AMDA will provide this information to the Erasmus/IPA Pompe Survey team, and they will send you the Pompe Survey consent form to sign.

If you have any questions, please email Marsha. She will be able to help you.

If you are struggling or just interested in talking to someone, please contact us at support@amda-pompe.org. We have patients or family members ready and willing to talk to you. You don't have to feel alone!



#TogetherWeAreStrong

“It is difficult to say what is impossible for the dreams of yesterday are the hopes of today and the reality of tomorrow” - Robert H. Goddard

AMDA

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