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**July 2024**

# **AMDA Newsletter**

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## **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, 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](mailto: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.

To view the official leaflet on the survey, [click here](#).

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

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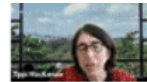
## Shionogi & Co., Ltd. and Maze Therapeutics, Inc. Announce Exclusive Worldwide License Agreement for MZE001

Shionogi & Co., LTD., and Maze Therapeutics, Inc. have agreed to develop for market, MZE001, a new substrate reduction therapy drug that could be the first ever pill take orally to treat Pompe disease.

Read the full press release at the [Canadian Association of Pompe website](#).

## PEARL Trial - PrEnAtal enzyme Replacement for Lysosomal storage diseases

### 22 First case of in-utero ERT in a fetus with infantile onset Pompe disease



**The NEW ENGLAND JOURNAL of MEDICINE**

**BRIEF REPORT**

**In Utero Enzyme-Replacement Therapy for Infantile-Onset Pompe's Disease**

Jennifer L. Cohen, M.D., Pranesh Chakraborty, M.D., Karen Fung-Kee-Fung, M.D., Maria E. Schwab, M.D., Deepika Bal, Ph.D., Sarah P. Young, Ph.D., Michael H. Gelb, Ph.D., Hamid Dhadbi, Ph.D., Alicia DiLantoni, Ph.D., Stacy Smallshaw, B.S., Felipe Moreira, M.D., Derek Wong, M.D., Catherine Lacroix, P.T., Dina El Demellawy, M.D., Ph.D., Kyle C. Strickland, M.D., Ph.D., Jane Loughhead, M.D., Anita Moon-Grady, M.D., Billie R. Lianoglou, M.S., Paul Hammett, M.D., Proya S. Kaliviani, M.D., and Tippi C. Mackenzie, M.D.

c.525\_526del;p.Asn177fs

Children with **CRIM-negative IOPD**

Pregnant female with 4<sup>th</sup> affected pregnancy

**Sibling #1:**

- Diagnosed at 8 months
- Immune reactions to enzyme
- Died (29 months)

**Sibling #2:**

- Prenatally diagnosed
- Palliative care (died at age 8 months)

**Current patient (Sib #3)- referred for fetal tx**

**Rationale for in utero ERT:**

- Known severe phenotype in this family
- Early onset cardiomyopathy
- ? Immune benefit

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University of California - San Francisco (UCSF) is excited to announce the rebranding of their phase 1 clinical trail of In Utero Enzyme Replacement Therapy as the **PEARL Trial-PrEnAtal enzyme Replacement for Llysosomal storage diseases**.

"We have rebranded our promising phase 1 clinical trial of In Utero Enzyme Replacement Therapy as the **PEARL Trial— Pr En Atal enzyme Replacement for**

Lysosomal storage diseases. We have created a website for the PEARL Trial, which is intended for a lay audience seeking information about our study and specific lysosomal storage diseases in pregnancy. Please consider visiting and sharing the new PEARL Trial website: <https://pearltrial.ucsf.edu>. The aim of the PEARL Trial is to establish the safety and efficacy of prenatal ERT for lysosomal storage diseases, including:· Mucopolysaccharidoses (MPS) 1, 2, 4a, 6, 7· Infantile-onset Pompe disease (IOPD)· Neuronopathic Gaucher disease (types 2 and 3)· Wolman disease

We have seen promising results with this approach—the first patient who was treated with in utero enzyme replacement therapy had infantile-onset Pompe disease and we saw a clear benefit of the prenatal therapy on her motor function, as we published in Cohen et al, [NEJM 2022](#). We have treated several additional patients at UCSF with no apparent safety concerns and have presented promising improvements in biomarkers at national conferences, including WORLD 2024.

Like other rare disease clinical trials, reaching eligible patients is challenging. Some participants will learn about the PEARL Trial through online searches. However, direct referral by clinicians and others in the lysosomal disease community is critical. Here are two ways you can help:1. **If you care for families who have previously experienced an affected pregnancy**, please consider sharing this information with them so that they are aware of this option for a future pregnancy.2. **Forward this message** to individuals who care for patients, diagnose, or study lysosomal diseases.

**To refer a patient to the [PEARL Trial](#)**, please contact our clinical trial manager, [Emma.Canepa@ucsf.edu](mailto:Emma.Canepa@ucsf.edu).

We are also keeping a [registry of patients](#) who are prenatally diagnosed with the above conditions so please contact us if you are aware of families who may consider participating."

-Tippi C. MacKenzie, MD and Paul Harmatz, MD



## 2024 Conference Wrap Up

We would like to thank you all for helping to make our 2024 AMDA/IPA Conference a HUGE success! With upwards of 225 in person attendees, and 80+ virtual attendees from over 13 countries, this was our largest, most successful conference yet!

We are working hard to get the session videos up on our YouTube channel, and pictures from the conference up on our website.

To view the official brochure of the 2024 AMDA/IPA Conference, [click here!](#)

If you took some photos at the conference and would like to share them, please tag us on

Facebook. <https://www.facebook.com/AcidMaltaseDeficiencyAssociation/>

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## Introducing The Talking With Your (Pompe) Peeps Series!

# Talking With Your (Pompe) Peeps



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<https://amda-pompe.org>

Based on feedback during our AMDA/IPA Conference, May 3-5, 2024, we are revamping our Mentor Program and are excited to offer the **AMDA Talking With Your (Pompe) Peeps Series**; an open forum informal peer group- we'd love to hear how the Series can be helpful to YOU.

Stay tuned for more details on our first session in August (next month), but in the meantime, we are here and available to assist you. If you are in need of help or need someone to talk to, please reach out to [Support@amda-pompe.org](mailto:Support@amda-pompe.org).

The AMDA is proud to be your premier resource center for your Pompe information; we look forward to talking with and collaborating with you.

## AMDA

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