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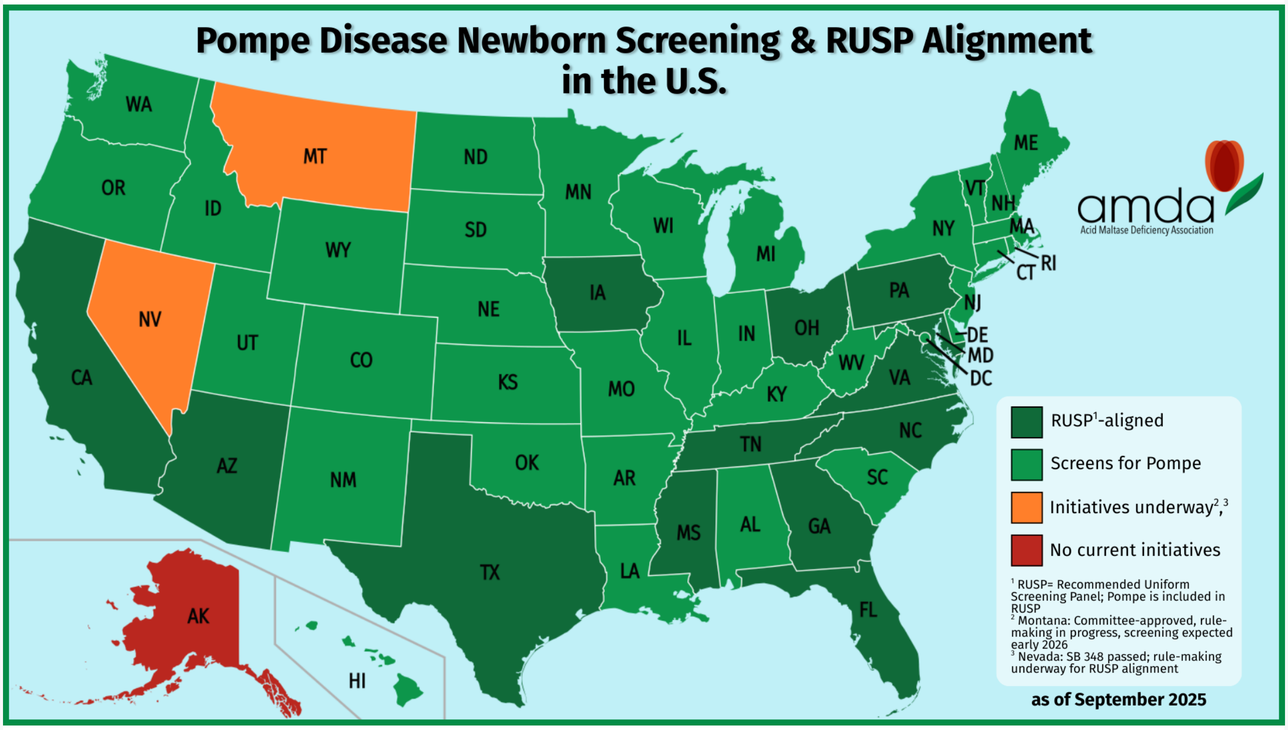
Talking With Your Pompe Peeps Conferences

September 2025

AMDA Newsletter

September is Newborn Screening Awareness Month

This month is packed with opportunities to learn, connect, and make your voice heard in the Pompe community. From research studies and surveys, to advocacy workshops and webinars, to AMDA-led programs and spotlights—you'll find plenty of ways to get involved.



Pompe Disease Newborn Screening & RUSP Alignment in the U.S.

[1] RUSP= Recommended Uniform Screening Panel; Pompe is included in RUSP
[2] Montana: Committee-approved, rule-making in progress, screening expected early 2026
[3] Nevada: SB 348 passed; rule-making underway for RUSP alignment

as of September 2025

September's Featured Article

Closing the Gaps: Pompe Disease Newborn Screening and the

Importance of RUSP Alignment

Our feature this month focuses on a critical issue: **newborn screening for Pompe disease across the United States**. We've created an updated map (as of September 2025) that shows where Pompe testing is currently included on state newborn screening panels. This resource is especially timely as we recognize Newborn Screening Awareness Month and advocate for **nationwide adoption of RUSP alignment legislation**.

Read the Full Article

In this issue: You'll also find AMDA volunteer opportunities, educational webinars, a new patient-led research initiative, and a chance to meet an artist living with Pompe who uses her creativity to inspire hope.

Disclaimer:

The content provided in this newsletter is for informational purposes only and does not constitute an endorsement or recommendation by the Acid Maltase Deficiency Association (AMDA), unless explicitly stated. Inclusion of any product, service, organization, or individual does not imply affiliation with or approval by the AMDA.

WHAT'S HAPPENING AT THE AMDA

Volunteer Opportunities: Make Your Voice Heard

The AMDA is often asked by biotech companies and other organizations for volunteers to participate in special projects, surveys, or other Pompe disease–related initiatives. If

Talking With Your Pompe Peeps — Call for Moderators & Topics

We're gearing up for upcoming *Talking With Your Pompe Peeps* sessions and want your input! If you're interested in serving as a you'd like to be considered, complete our sign-up form. If you already signed up in April, no need to do it again — you're already in our volunteer pool! Selected volunteers will be contacted directly by the project's representative.

Questions? Reach out to our Patient Advocate, **Marsha Zimmerman**, at **marzim50@gmail.com**.

Volunteer Form

moderator, or if you have topic suggestions you'd like to see covered, we'd love to hear from you.

Please reach out to **Matt Zimmerman** at **matt.zimmerman@amda-pompe.org** to get involved.

Don't Miss This Webinar

Join us for an informative webinar introducing the basics of breathing and coughing muscle weakness in neuromuscular disease. **Andrea Klein** will cover the role of muscles in breathing, the importance of blood gases, evaluation processes, appropriate interventions, and strategies for effective medical self-advocacy.

Andrea Klein, founder of **Breathe with MD, Inc.**, has nearly 25 years of experience in healthcare and is a recognized advocate for patients living with neuromuscular respiratory challenges. Living with Intermediate Collagen 6 Congenital Muscular Dystrophy herself, Andrea brings both professional expertise and personal experience to the session.

Don’t miss this opportunity to learn from Andrea and gain practical knowledge to support yourself or your loved ones. **Register now and join us!**

Event Details:

Title: Overview of Breathing Muscle Weakness in Neuromuscular Disease (NMD)

Date: Thursday, September 25, 2025

Time: 1 PM CST / 2 PM EST

Speaker:
Andrea Klein

Webinar Overview:
An introduction to the concepts of breathing and coughing muscle weakness, this presentation highlights the importance of medical self-advocacy to ensure positive outcomes. Topics include the role of muscles in breathing, the relevance of blood gases, the evaluation process, appropriate interventions, and more.

Register for this Webinar

Speaker Biography

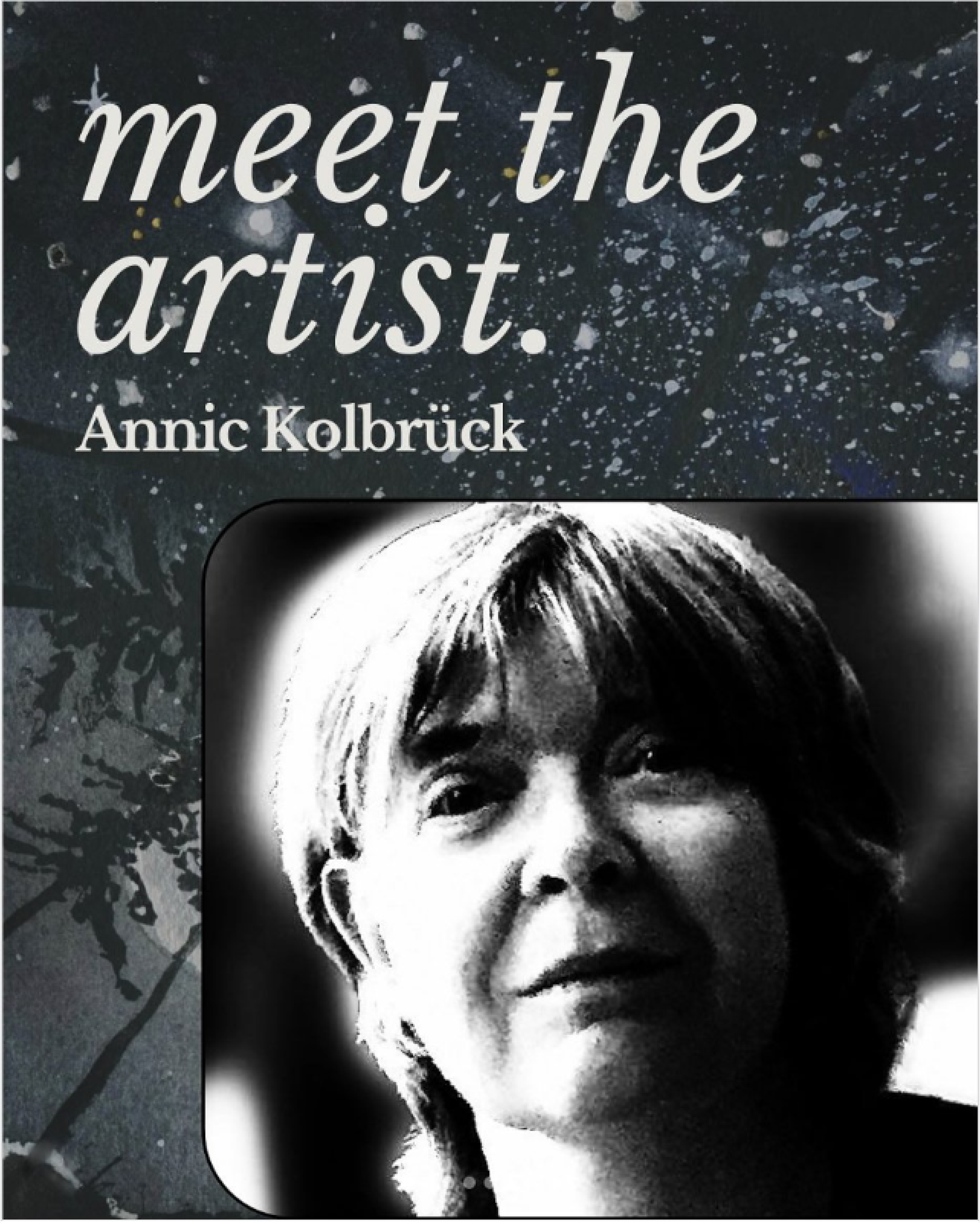
Andrea Klein

Having earned a Bachelor of Science in English-Journalism

with a concentration in Public Relations from Tennessee Technological University in 1996, Andrea has worked in the healthcare IT field for nearly 25 years. She is co-author of a chapter in the textbook “Non-Invasive Ventilation and Weaning: Principles and Practice, Second Edition,” published in 2018. She served as Ms. Wheelchair Tennessee 2017 and received the Lifetime Achievement Award at the Ms. Wheelchair America 2018 competition.

She founded Breathe with MD, Inc. in memory of her middle sister, Cheryl Lewis, who died because of inappropriate care for neuromuscular respiratory failure. Andrea has a passion for educating others about breathing muscle weakness and the importance of medical self-advocacy. She developed and implemented Breathe with MD’s “Breathing Muscle Weakness 101” online course, which has enrolled nearly 250 participants in the last two years.

Andrea lives with Intermediate Collagen 6 Congenital Muscular Dystrophy and has used non-invasive ventilation since 2013 and mechanical cough assistance since 2014.



Meet The Artist

Annic Kolbrück

Meet **Annic Kolbrück**, an artist living with Pompe disease who turns creativity into hope.

Annic has painted since her youth, and after her Pompe diagnosis in 2004, she continued her career as a journalist until her 61st birthday while also serving in the **Morbus Pompe Germany association**.

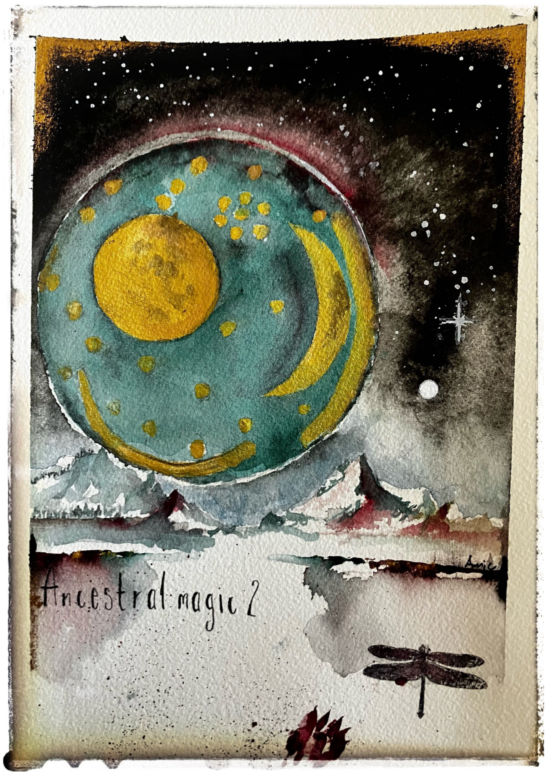
Her art is more than beautiful—it's purposeful. Each original painting **reflects optimism and resilience**, and Annic **donates 100% of the proceeds to Pompe organizations in Germany and the USA**, directly supporting research and the search for a cure.

Through her creativity, Annic shines a light for the Pompe community—proving that **art can heal, inspire, and drive change**.

Follow Annic on instagram: @annicko

Learn more about Annic on her website: https://www.annickolbrueck.de/

Explore her Artwork



Want to share your art with the Pompe community? Submit your work to us and inspire others! Email **Matt Zimmerman**, at matt.zimmerman@amda-pompe.org to be featured.

Join the Conversation!

Follow the AMDA on social media to stay updated on Pompe disease news, connect with our community, and share your story!

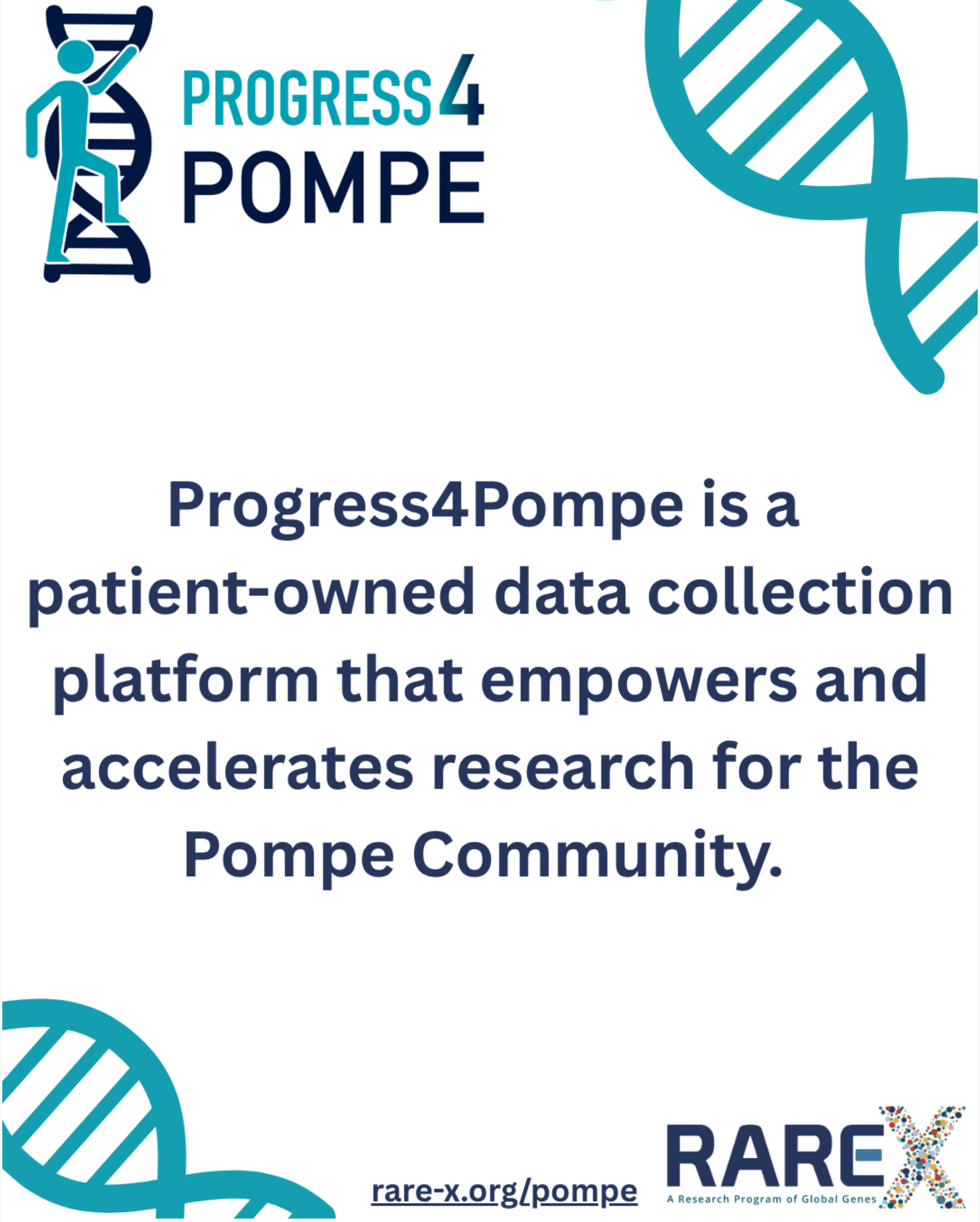
AMDA_ASSOCIATION

Instagram

Facebook

GET INVOLVED IN POMPE RESEARCH!

Contribute to Pompe Disease Research with Progress4Pompe



Progress4Pompe is a patient-owned data collection platform that empowers and accelerates research for the Pompe Community.

rare-x.org/pompe

We are pleased to share the following announcement from **Global Genes**, who are leading an important new effort to advance Pompe research. Their initiative, **Progress4Pompe**, empowers patients and families to directly contribute to scientific understanding and treatment development.

> In collaboration with Global Genes, multiple Pompe disease patient advocacy groups, including Pompe.community, Pompe Alliance, Pompe Warrior Foundation, and On a Mission for Margot, have been working together to launch a global patient-led

research initiative. With input from individuals diagnosed with Pompe disease and their families, this study now has a name: Progress4Pompe, signifying the broad impact that patient-centered research can have.

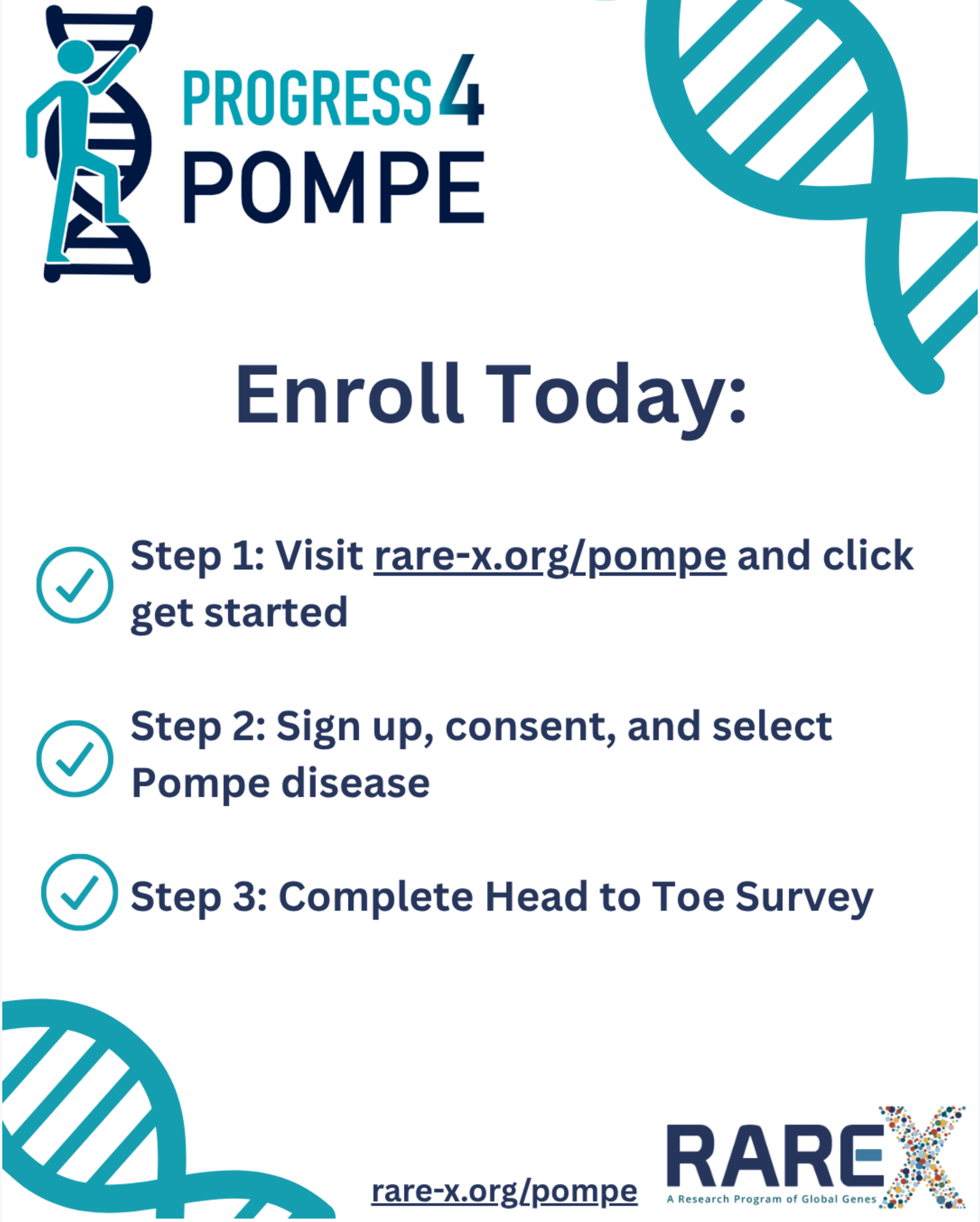
Now we need YOU to join us!

We invite you to join Progress4Pompe, a patient-owned study that can have a major impact on the Pompe Community. Together, we can achieve the following for the Pompe.community:

- Improve our understanding of the progression of Pompe disease
- Accelerate therapeutic progress for biopharma companies investing in Pompe disease
- Connect eligible patients to clinical trials

Register today by visiting rare-x.org/pompe. The sign up process will take about 30 minutes.

We will share research updates and progress. This is for everyone!



PROGRESS4 POMPE

Enroll Today:

- ✓ Step 1: Visit rare-x.org/pompe and click get started
- ✓ Step 2: Sign up, consent, and select Pompe disease
- ✓ Step 3: Complete Head to Toe Survey

rare-x.org/pompe

RARE-X
A Research Program of Global Genes

Formulation Survey

Last Chance: Make Sure U.S. Pompe Voices Are Counted

Only a handful of Pompe

UNIVERSITY of ROCHESTER MEDICAL CENTER

Developing a patient-reported outcome measure for Pompe disease

Participate in the POM-HI

patients from the United States have completed this important international survey — and the deadline is almost here. Let’s change that together.

The survey, conducted independently by **InCrowd** on behalf of a pharmaceutical sponsor, takes about **15 minutes** to complete. It is designed to capture the lived experience of people with Pompe disease and to shine a light on the **treatment needs that remain unmet today.**

Right now, U.S. representation is low. The Pompe community in Italy, Japan, Germany, the United Kingdom, and the Netherlands is already participating in larger numbers. If we want the American experience to carry equal weight, we must make our voices heard before the survey closes.

▯ Deadline: September 5

Your answers are anonymous and will never be shared individually with the sponsor.

If you tried to take this survey from our last email but got an error message, try again, the link may have been corrupted. All survey links can be found on the IPA website.

Important Disclaimer:

This survey link is intended for **Pompe patients living in**

Study: Phase 1

Principal Investigator:
Dr. Chad Heatwole, MD, MS-CI

Researchers at the **University of Rochester** are creating a patient-reported outcome measure to assess the health, symptoms, and daily impact of Pompe disease. This study aims to identify the symptoms that most affect quality of life, helping guide future research and care for the Pompe community.

What’s involved:
Participants will take part in a 30–60 minute **Zoom interview** with a study coordinator to discuss their experiences. Interviews are audio recorded, transcribed, and analyzed with strict confidentiality—no identifying information is stored with responses.

Who can participate:
Adults (18+) with Pompe disease living in the **U.S.** who speak English.

How to participate:
Reach out to the study coordinators:

Charlotte Engebrecht:
585-200-7136
charlotte.engebrecht@chet.rochester.edu

Karnavaal Al-Rubayie:
585-978-6559
karnavaal.alrubayie@chet.rochester.edu

the United States.

If you are located in **Italy, Japan, Germany, the United Kingdom, or the Netherlands**, please use the version designed for your country here: https://worldpompe.org/news/formulation-survey/

Take the Survey

MARK YOUR CALENDAR!

NORD®
National Organization for Rare Disorders

Alone we are **rare.**
Together we are **strong.**®

NORD's Newborn Screening Webinar

This **Newborn Screening Awareness Month**, the AMDA encourages you to join the National Organization for Rare Disorders (NORD®) for a virtual grassroots advocacy workshop on **Wednesday, September 10 at 11 AM CST / 12 PM EST**.

NORD staff will cover:

- Updates on the newborn screening policy landscape and NORD’s current priorities
- Tools and resources to help you advocate and share your newborn screening or rare disease story with lawmakers
- An overview of the *Living Rare Study®*, the first large-scale U.S. study following the changing experiences of people impacted by rare diseases

We hope you’ll take part in this important session to strengthen awareness and advocacy for newborn screening and the rare disease community.

Register for NORD's NBS Webinar

NORD Speakers

Leah Barber
Director of Grassroots Advocacy

Allison Herrity
Senior Policy Analyst

Darby Gavin
Senior Community Engagement Manager

2025
NEWBORN SCREENING BOOTCAMP
VIRTUAL | September 17

Newborn Screening Bootcamp: Learn from the Experts

In honor of **NBS Awareness Month**, the **EveryLife Foundation** and **Expecting Health** are hosting the **Newborn Screening Bootcamp** on **Wednesday, September 17, 2025 at 11 AM CST / 12 PM EST**. This event gives advocates and families a front-row seat to the latest in newborn screening, featuring panels with experts and patient advocates.

Learn how advocates engage with federal and state programs and discover opportunities to join NBS-related committees.

Missed last year? **Catch up on 2024 Bootcamp Highlights**

Register for 2025 Bootcamp

UNIVERSITY OF MINNESOTA
Driven to Discover®
Crookston Duluth Morris Rochester Twin Cities

WORLDFair™ 2025

September 19, 2025 I 10 AM – 2 PM CST

Pacific Standard Time: 8:00 AM until noon
Eastern Standard Time: 11:00 AM until 3:00 PM

Minnesota Landscape Arboretum

3675 Arboretum Drive, Chaska, MN 55318

WORLD Fair 2025 will be a hybrid meeting, which will invite patients, caregivers and healthcare providers from around the world to attend remotely through zoom.

A Gathering Focused on Lysosomal Diseases

WORLDFair 2025

The **11th Annual WORLDFair**, hosted by the **Advanced Therapies Department** at the University of Minnesota, will take place on:

Date: September 19, 2025

Time: 10:00 AM - 2:00 PM CT

Location:
University of Minnesota
Landscape Arboretum
3675 Arboretum Drive,
Chaska, MN 55318

This annual event brings together patients, families, healthcare providers, researchers, and industry representatives to exchange knowledge, share experiences, and explore the latest developments in lysosomal disease research and treatment.

- Remote attendance via Zoom
- Box lunch provided for in-person attendees

Intended Audience:

- Individuals diagnosed with lysosomal diseases
- Family members, caregivers, and friends
- Medical professionals and researchers
- Industry professionals
- Anyone with an interest in lysosomal diseases

Program Highlights Include:

- Educational presentations on lysosomal disorders and emerging treatments
- Patient and caregiver stories
- Networking opportunities across the lysosomal disease community
- A presentation from the **Pompe Warrior Foundation** on dietary intervention, supplementation, and ongoing research in Pompe disease

A full program with speakers and topic details can be found here.

WORLDFair 2025 Registration

CELEBRATING OUR NEUROMUSCULAR COMMUNITY

Celebrating 75 Years of the MDA

This **Muscular Dystrophy Awareness Month**, we celebrate the **MDA's 75th Anniversary** and their decades of dedication to supporting families, advancing research, and improving lives for those living with muscular dystrophy and related neuromuscular diseases—including **Pompe disease**.

Muscle health is central to both MD and Pompe, and the MDA's work in research, patient advocacy, and community support benefits the broader neuromuscular community. Their milestone is a reminder of the power of collaboration in driving treatments and resources for rare muscle disorders.

Learn more about their work and anniversary here: **mda75.org**

Monthly Observances

Newborn Screening Awareness Month: September is Newborn Screening Awareness Month, a time to highlight the importance of early detection for conditions like Pompe disease. Early screening can make a life-changing difference for babies and families. Learn more and access helpful resources through **Baby’s First Test**, **EveryLife Foundation**, and **SaveBabies.org**. See webinars mentioned earlier in this newsletter for tips on advocacy and getting involved!

National Muscular Dystrophy Awareness Month: September is Muscular Dystrophy Awareness Month, a time to shine a light on the many forms of MD and the families affected. Pompe disease is one of the neuromuscular disorders in this community, and together we share a commitment to advancing research, improving care, and raising awareness. Celebrate with us by wearing green and helping spread the word! Muscular Dystrophy Association: https://www.mda.org/

Daily Observances

Fight Procrastination Day | Sept 6th: We all put things off from time to time, but Fight Procrastination Day is a reminder to tackle tasks head-on. For those living with a rare disease, staying on top of appointments, treatments, and self-care can make a big difference. Celebrate with us by checking one thing off your list today! **#FightProcrastinationDay #TakeAction**

National Grateful Patient Day | Sept 7th: This day honors patients who show resilience and gratitude in the face of health challenges. It’s also a time to appreciate the caregivers, doctors, researchers, and advocates who provide support along the way. Celebrate with us by sharing a note of gratitude! **#GratefulPatientDay #ThankYouCaregivers #TogetherStrong**

Positive Thinking Day | Sept 13th: Positive Thinking Day encourages us to focus on optimism and resilience. A hopeful mindset can help anyone face life’s challenges, and it can be especially meaningful in the rare disease community. Celebrate with us by spreading positivity today! **#PositiveThinkingDay #ChooseHope**

National Pancake Day | Sept 26th: Fluffy, warm, and stacked high, today we celebrate pancakes in all their syrupy glory! Sometimes, enjoying the little things is just as important as the big milestones. Celebrate with us by treating yourself to a short stack and a smile. **#NationalPancakeDay #StackUpTheSmiles**

Tiffany House and Juls Williams (Liaison, AMDA) celebrating National Pancake Day, 2024.

#PompePower

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