PURA Connect

PURA Syndrome FOUNDATION

Newsletter - Winter 2026

Message from our Co-Presidents

We are hitting the ground running in 2026! It has been a very busy few months at the PURA Syndrome Foundation with a lot of items being checked off on our strategic planning to-do list from **approving new research grants** to **beginning to onboard our new registry platform** after an extensive selection process.

Working with Odylia Therapeutics, the PSF completed a research landscape analysis of PURA Syndrome in February 2025 to **identify research gaps and strategically move forward with our research** efforts. Our Request for Proposal (RFP) process brought forth exciting research opportunities that aligned with some of the gaps presented in the landscape analysis. Due to to success of the Together for PURA fundraiser and the quality of the submissions we received the board unanimously voted to **fund over $200,000 in grants to further our knowledge of PURA and develop more treatment options**. We look forward to announcing more about these projects once the contracts are finalized. *A special thanks to Kathryn and Ashley from Odylia for their support over the year to develop our research strategy and put the plan into action. While our scope of work with them is complete, we look forward to keeping this momentum going in 2026 and working with them again on future projects!*

We have been looking for ways to provide more family support and will be partnering with **UniteUs** this year, which helps connect families with **social service supports**. While this platform is specifically US-based, we are looking for options to support families around the world, so if you have any suggestions, please contact us at president@pura-syndrome.org.

Our conference planning for Germany is well underway and we hope to see many of our worldwide families in **Munich in June**. **Registration is now open** – more information about that and more is in this newsletter, or can be found at puraconference.org.

At the end of 2025, we were absolutely humbled at the outpouring of support for our **PURA Syndrome Awareness Day** and **Giving Tuesday/End of Year fundraisers**. We had two donors generously step up and provide $5,500 matches to help us reach our $11,000 fundraising goal for each campaign. We are so excited to say that we met and **exceeded our goals!** We are so grateful for all of those who are helping raise awareness and contributing to our mission and would not be able to accomplish any of these priorities without your support. We will continue to ensure that we, the PSF board, and our advisors keep what's best for all individuals with PURA at the forefront of minds when making decisions.

And finally, we are happy to announce that we will be **launching a PURA Syndrome podcast** this year to help our families get more detail and understanding about the projects we are working on this year. Stay tuned for more details!

Sincerely,
Liz Astridge & Eva Tucker
Co-Presidents, PURA Syndrome Foundation

Our mission is to support and drive research, raise public awareness and connect, educate, serve, and empower families impacted by PURA Syndrome.

AWARENESS

World Rare Disease Day - February 28

Rare Disease Day is fast approaching!

In less than four weeks, the PURA Syndrome Foundation will join together with other rare disease organizations across the globe in celebrating World Rare Disease Day. Rare disease day is dedicated to raising awareness among the general public and key decision-makers about rare diseases and disorders and their impact on patient's lives. World Rare Disease Day gives a public and social platform for rare stories of hope, struggle, and resilience.

When an individual is impacted by a rare disease, awareness is critical in increasing support, raising funds, increasing research opportunities, breaking down social barriers, and educating the general public. We invite our families and supporters to join the Foundation in raising awareness for PURA Syndrome on Saturday, February 28, 2026. Specifically, we ask you to participate in at least one of the following ways:

1. Follow the PURA Syndrome Foundation on social media. Like, comment, and share some of our Rare Disease Day posts. You can find us on Facebook and Instagram.

2. Tell your story. As February 28 approaches, consider sharing your #PURAPerfect story on social media, via video, through a letter or email, or get creative! We have seen families create educational materials to send to their child's school and even lobbying for better healthcare with local and state legislatures. We would love to hear your stories! Tag us on social media or email communications@pura-syndrome.org if you are interested in sharing your story on the Foundation's social media pages.

3. Spread awareness on social media. The PURA Syndrome Foundation has created a social media template to assist you in creating awareness. Click here to download the Zebra graphic OR create a personalized image with our PURA frame using the free photo editing software Canva:

- Visit the following link to add a photo of your family or the #PURAPerfect individuals in your life to this template OR if you are unable to do it yourself, please email a photo to info@pura-syndrome.org.
- Upload the personal photo of your choice
- Add your personal photo to the social media template and size appropriately.
- Download your image to your phone or laptop

Now you're ready to share your image(s) to social media. Don't forget to tag the Foundation!

4. On February 28, wear your rare disease or PURA Syndrome t-shirts, bracelets, and buttons to raise awareness in your local community. Need gear? Purchase items at our Zazzle store and a portion of proceeds will be donated to the PURA Syndrome Foundation.

5. Consider a donation or start a fundraiser to benefit the PURA Syndrome Foundation by visiting https://givebutter.com/2026RDD.

Visit our website for more ideas to spread awareness

2026 FUNDRAISING

Help us raise $5,000 by Rare Disease Day
February 28, 2026

In celebration of this day, help us raise $5,000 to advance research and improve care for individuals and families affected by PURA Syndrome.

While PURA Syndrome is rare, our community is strong, joyful, and determined. Funds raised directly support research, increased understanding, and better therapies, resources, and care at every stage of life.

Rare Disease Day reminds us that progress happens when we come together. Every contribution brings us closer to greater awareness, better outcomes, and a brighter future for those living with PURA Syndrome.

Please share our link and encourage family & friends to contribute: **https://givebutter.com/2026RDD**.
Thank you for standing with our community and helping ensure every individual with PURA Syndrome is seen, supported, and empowered to thrive.

Help us reach $5,000 by Rare Disease Day!

PURA SYNDROME • RARE DISEASE DAY
I SUPPORT RARE
PURA SYNDROME
RAREDISEASEDAY.ORG

I ♥ someone rare

PURA Syndrome FOUNDATION

RARE DISEASE DAY®

Alone we are rare, together we are strong.

RESEACH & REGISTRY UPDATES

NEW GLOBAL REGISTRY and U.S. BIOBANK PARTNERS

After carefully evaluating our options, the PSF board decided to partner with **COMBINEDBrain** and **Matrix** for our updated **Global Registry platform** as well as a **US-based Biorepository**. Matrix is a **sophisticated, customizable and user-friendly technology platform for the registry**. COMBINEDBrain is a non-profit organization devoted to s**peeding the path to clinical treatments for people with rare genetic neurological disorders** by pooling efforts, studies, and data.

By contracting with Matrix under COMBINEDBrain umbrella for the registry we get access to their platform at a much reduced rate, and have additional benefits including access to standardized surveys to help us be more **clinical trial ready** and access to **multiple language translations** — while also being able to customize our registry to our needs now and in the future. The platform has additional features to **manage health data** and the option to share with your own doctors, as well as, to see how your data compares to other participants' anonymized data in the registry.

For those who are not familiar, the **Global PURA Registry** collects clinical data on PURA syndrome from affected patients around the world throughout the course of their lives. This will enable researchers to define the full spectrum of the disorder, and also **characterize** the natural history **(what happens across age and life**) for PURA syndrome. This is important to many aspects of PURA research from **improving guidelines for care** to establishing outcome measures for future clinical trials. Our hope is that this new registry will position us to not only **enable more PURA specific research**, but to also be able participate in **more cross disease research** with our rare diseases that share similar characteristics like epilepsy, hypotonia and communication issues.

The **PURA Biorepository** involves collecting a combination of prospective and retrospective samples from globally consented PURA syndrome patients. The samples will be used for research with the long-term goal to understand and develop treatment strategies for PURA syndrome or related disorders. Along with this new U.S.-based biobank, there are also PURA biobanks in Germany and Australia to serve our global community of families and researchers.

We'll talk more at the upcoming conference about all of these — addressing data privacy, how data and bio samples are used, and answer your questions so that you can make informed decisions on how you want to participate.

Watch out for more details over coming months and feel free to reach out to president@pura-syndrome.org if you are interested in helping test the new registry.

Matrix

COMBINED

PURA Syndrome FOUNDATION

RESEACH & REGISTRY UPDATES

Expanding Our Network

PSF leadership has been actively engaging with the broader research and translational ecosystem through attendance at the American Epilepsy Society Annual Meeting, the Ultragenyx Rare Bootcamp, the Epilepsy/DEE Data Commons Planning Meeting, and the COMBINEDBrain Annual Summit. These meetings allow us to stay closely aligned with advances in neuroscience, drug development, regulatory expectations, and patient-centered research. Our goal is to put the right pieces in place now so that when therapeutics emerge, our patient community is coordinated, prepared and ready to move from scientific discovery to patient care. Let us know if there is a conference or meeting you highly recommend by emailing science@pura-syndrome.org.

COMBINED

AES AMERICAN EPILEPSY SOCIETY

ultragenyx

DATA FOR THE COMMON GOOD

PURA Disease Concept Model

Over the past year, the PSF has taken deliberate steps to ensure our community is research-ready, not just hopeful. A key effort underway is the development of our Disease Concept Model (DCM) through a collaboration with COMBINEDBrain. The DCM is a structured framework that allows us to align PURA biology with the symptoms that matter most to PURA perfect individuals and their families. Through this research study, we will learn how our community's lived experiences determine clinically meaningful endpoints, helping researchers, industry, and regulators speak the same language. This work is foundational for preparing us for clinical trial readiness as future therapies arise.

We will soon be recruiting PURA perfect families interested in contributing to the DCM. Stay tuned for more details in the near future if you're interested in being part of this important next step.

UPCOMING EVENTS

National Caregivers Day - February 20

"There are only four kinds of people in the world: those who have been caregivers, those who are currently caregivers, those who will be caregivers, and those who will need caregivers."

Rosalynn Carter, former US First Lady and passionate advocate for caregiving

PURA Syndrome Foundation has partnered with Shahroo Izadi to create self-care resources for our PURA caregiver community. Click here for free resources.

Rare Disease Day - February 28

Raising awareness and generating change for the 300 million people worldwide living with a rare disease, their families and caregivers. Check out our website for information and celebration ideas along with our Rare Disease Day Fundraising Campaign. Also, see the Rare Disease Day website

GrandPOPS Virtual Meetup - February 28

Mark your calendars -- February 28 at 1 p.m. CST, we will host a virtual GrandPOPS Meetup. Join us on Zoom to share our stories, photos, joys, and concerns. If your special grandchild is available, we would love to them, but we welcome photos too. If you (or your parents) haven't signed up for our email list, go to our website or email me at grandpops@pura-syndrome.org

Virtual Meetup for Families from India - March 1

Several families from India have recently contacted us with interest in connecting with one another! We will be hosting a virtual meet-up for any of our families from India on Sunday, March 1, at 9:00 am IST. If you would like to join this meet-up please be sure to register here.

UPCOMING EVENTS

Register now for the **PURA Syndrome Conference, June 26-28 in Munich, Germany**. Join researchers, experts and our PURA community for in-depth sessions that will focus on:

- Research and studies about PURA
- Parent Discussions
- PURA Community Social Time
- And so much more!

All sessions will be in English with German captions on screens. If you additional language support beyond google translate, please contact conference@pura-syndrome.org. The conference and sessions are open to family, caretakers, researchers, clinicians, and friends. For those unable to attend in person we will also offer a virtual option, and many of the sessions will be recorded to view at a later date.

IN-PERSON CONFERENCE REGISTRATION

VIRTUAL CONFERENCE REGISTRATION

Book your room at the Conference Hotel!

The conference takes place from June 26-28, however, we have a reduced rate from Thursday through Sunday night for anyone who would like to stay longer. Please click this link to book our Conference hotel.

For ADA-compliant rooms or rooms with connecting doors, kindly contact the Hotel Reservations Team at: reservation@himunich.com

If participants would like to reserve a twin room, they can also contact the Hotel Reservations team at: reservation@himunich.com

If you have any questions, please contact conference@pura-syndrome.org.

PURA IN THE NEWS

Spotlight on Celena Lozano

We are thrilled to celebrate our very own **Celena Lozano** – **PURA mom, board member, and neuroscience PhD student** – for winning a Summer Writing Award through the National Association of Science Writers (NASW) 2025 Perlman Virtual Mentoring Program! In her **award-winning article, Celena offers a powerful and insightful look at the future of gene therapies for rare neurodevelopmental disorders like PURA Syndrome.** She highlights the gap between media hype and the current scientific reality, calling for stronger collaboration and clearer communication among researchers, clinicians, journalists, policymakers, and rare disease families. Celena's NASW piece, *"When Will Our Kids Be Next? What Baby KJ's Story Means for the Future of Gene Therapy,"* can be read here: NASW Article

Celena later expanded her perspective into a compelling opinion piece published by STAT, titled "*After CRISPR Cured Baby KJ, What Comes Next for Rare Diseases and Personalized Gene Editing Drugs?"*, available here: STAT Article

Photo of Celena Lozano PURA mom, board member, and neuroscience PhD student

Local News Feature

PURA Family shares their story on a local news station for PURA Awareness Day. PURA perfect Brandon and his family share their story on a local news station for PURA Awareness day. Watch the video here. Read the story here.

Celebrating a new, faster path to gene-editing medicines on demand

The US FDA recently released a new regulatory roadmap for personalized therapies. In the linked piece, Fyodor Urnov raises important points about how this pathway may differ for metabolic versus neurological disorders and highlights voices from the rare disease community, including Celena Lozano's. Innovation is moving quickly, but families, equity, and clear communication must stay at the center of these discussions. The article can be read here.

Every story matters.

REMINDERS

PURA Syndrome Clinician Directory

A common question we hear from new families is: "*How can we find doctors who are familiar with PURA Syndrome?"* To help answer that, we've been building a **Clinician Directory** — a growing list of healthcare providers who have experience treating multiple individuals with PURA Syndrome and are experts in their respective specialties.

This directory serves a dual purpose:

✓ Helping families connect with knowledgeable providers

✓ Enabling clinicians to connect with one another to improve care and advance research

📍 You can find the **Clinician Directory** in the *For Families* section of our website. If you know of a clinician who should be added to the directory, please let us know by contacting us at **president@pura-syndrome.org**.

Get Involved in PURA Research

The PURA Syndrome Foundation encourages all PURA caregivers and individuals to consider participating in **research studies** as they become available.

We currently have two open research opportunities. While the Foundation is not always affiliated with or sponsoring these studies, we share them with the hope that participation will contribute to better understanding, care, and outcomes for our children and community.

🔍 To **explore current opportunities**, click here. Your participation can help drive progress and make a lasting impact.

PURA Syndrome Foundation Needs YOU!

The PURA Syndrome Foundation is a **volunteer-based organization**, which means we rely on the time, talents, and generosity of people like you to help us achieve our mission and reach our goals. As our community continues to grow, so does the need for many hands and open hearts to support our efforts — from event planning and fundraising to outreach and advocacy.

If you're able to help, or if you know someone who might be interested in getting involved, we'd love to hear from you!

👉 Please fill out the volunteer form on our website or email us directly at volunteer@pura-syndrome.org.

Every step forward—every data point, donation, and shared story—brings us closer to better care and brighter futures for individuals with PURA Syndrome.
Thank you for being part of this community.