PURA Connect

PURA Syndrome FOUNDATION

Newsletter - Spring 2026

Message from our Co-Presidents

Where Eva and I live, Spring has just arrived and we are surrounded by new growth and beauty. This description could easily be used for what's happening at the PURA Syndrome Foundation. Our board and committees have worked together to accomplish so many great things over the past year — things which have opened doors to more and more opportunities for growth, collaboration and serving our community. It is exciting, exhausting and motivating all in one!

Thanks to everyone who supported our Together for PURA 2024–2025 campaign, we're proud to announce the funding of four research grants totaling $247,000 — this marks the largest research investment in the Foundation's history. You can find more on page two of this newsletter and visit our blog for more details about the four funded projects and the researchers behind them.

Our new PURA Syndrome Registry is being tested, with a full launch planned for our PURA Conference in June. We've been working through the system carefully — testing it ourselves and consulting with our advisors, research network, and other PURA organizations to make sure it is setup to meet our community's needs. Together, the registry and biobanks will give researchers the data and samples they need to deepen our understanding of PURA Syndrome and prepare the community for future clinical trials. Please stay tuned for details!

As we shared in our Winter Newsletter, we applied for an ICD-10 code for PURA Syndrome in December 2025. Early in 2026, the CDC paused all genetic disease code requests while they develop better organizing principles for genetic conditions. Through our collaboration with COMBINEDBrain, PSF has been invited into the stakeholder conversations, including a meeting with the CDC — a chance to spread awareness about PURA Syndrome and contribute to a process that will benefit all rare disease organizations. We'll keep you posted as this progresses.

We'd like to offer a note of thanks to all who were involved in updating the PURA GeneReviews article in March — including leaders from PURA Foundation Australia and our Scientific Advisory Board members Dr. David Hunt, Dr. Lauren Sanchez, Dr. Margot Reijnders and Dr. Diana Baralle. Because GeneReviews is often the first resource clinicians consult when they encounter a rare diagnosis, this update puts current PURA Syndrome knowledge directly in the hands of the doctors caring for our families. You can view the updated GeneReviews Article here.

We are working hard to get all the final details together for our Annual Conference in Germany on June 26-28. Registration, hotel bookings and reservations for the Niessing Lab tour details can be found below. Hope to see you there!

Liz Astridge and Eva Tucker

Together, we are stronger — and together, we make a difference.

Fundraising = Research

NEW RESEARCH GRANTS FUNDED

$247,000 in Research Grants — the Largest Investment in the Foundation's History

Thanks to everyone who supported our Together for PURA 2024–2025 campaign, we are proud to announce the funding of four research grants totaling $247,000 — the largest research investment in the Foundation's history.

When we launched Together for PURA 2024–2025 to mark ten years since PURA syndrome was first named, we set out to push our research efforts to the next level. You made that happen. The four funded projects take on some of the most pressing questions in PURA science — from the brain mechanisms behind seizures, to whether shortened PURA proteins may actively disrupt cells, to measuring how PURA mutations change the way brain cells develop and communicate, to exploring the potential and the challenges of gene therapy for PURA syndrome.

Check PURA Syndrome Foundation **blog** for our big announcement with details on the exciting new research YOU helped fund through your PSF donations!

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Conference

PURA Syndrome Conference
June 26-28
Munich, Germany

Special Insider Opportunity: Tour the Niessing Lab and PURA Biobank - Sunday, June 28

Our conference is coming up and we are happy to announce an opportunity for our families on Sunday, June 28. Join us for a tour of Prof. Dierk Niessing's Research Lab and PURA Biobank with a visit to the Helmholtz Center Munich! The Munich region is home to several renowned research institutions. In addition to two leading universities — Ludwig-Maximilians University and the Technical University of Munich — and a few Max Planck Institutes, it also hosts Helmholtz Center Munich, a world-class hub for biomedical research. The research team of Prof. Niessing operates one of its two laboratories on this campus and is delighted to offer a guided tour. Participants will travel there by shuttle bus. Your visit will begin with an introduction from our PURA researcher, who will share insights into current PURA-related scientific work.

Afterward, you will join a guided tour in small groups, giving you an inside look at active laboratories where real PURA research is conducted. The tour continues to the Biobank facility, which houses several thousand patient samples — including biosamples from the PURA Biobank. You will have time to ask questions, connect with our local PURA scientists, and gain a deeper understanding of the research environment. Following the tour, the shuttle bus will return you to the conference venue. For more information and to RSVP, please click here.

Conference registration: **IN-PERSON** **VIRTUAL**

Reserve your room at the conference hotel: **HOTEL RESERVATION**

For more details about the conference, schedule, lodging, see our **Conference Website**.
If you have any questions, please contact **conference@pura-syndrome.org**.

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Conference

Celebrate the PURAPerfect person in your life with a feature in our Conference Program

This year's conference will be special in so many ways. One way you can participate is to feature your PURAPerfect Child or Relative in the conference program and online! We are now offering families and friends the ability to purchase program ads. Your support is a great way to recognize a particular family and/or child and a wonderful way to support our PURA community of families living with this rare disease. All proceeds will go directly to funding our annual conference.

Add some sunshine to our program!

Click **here** for details:

- **Full page ad $150**
- **1/2 page ad $100**
- **1/4 page ad $50**

Deadline for photo submissions is **May 15.**

Visit the **conference website** for an outline of sessions and activities, hotel booking link and additional information.

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Fundraising

TOGETHER for PURA 2026

Empowering Families, Advancing Research

Our 2026 Together for PURA Campaign launches this summer with a goal of raising $300,000 — to support critical research, empower families, and strengthen advocacy worldwide. Before we go live, we're laying the groundwork for the strongest launch yet, and we need help with two specific things:

Become a team captain. The biggest campaigns are powered by people who rally their own networks — family, friends, coworkers, neighbors. When you start a team, your community becomes part of ours, and that's how we reach $300,000. Getting started is easy: visit our 2026 Together for PURA Fundraising page, scroll to "Team Members," and click the blue "Fundraise" button. Once your team is set up, invite others to join or share your personal link or QR code.

Offer or secure a matching gift. A matching commitment — from a family foundation, a local business, or an individual donor — doubles every gift made during the campaign. Matches are one of the most effective ways to motivate giving, and we're working to line them up before the public launch. If you can offer a match or help us secure one, please reach out to Brian at fundraising@pura-syndrome.org.

Reaching $300,000 will take all of us — families and extended families, friends and neighbors, local businesses and corporate partners, restaurants, creators, and community leaders. Rare does not mean alone, and when we come together, our impact compounds. We can do this. And you can be part of it.

Every effort and every contribution makes a meaningful difference!

Join the 2026 Together for PURA Fundraising Campaign

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Expanding Our Network

Rare Disease Fair

Melissa Vaught, PSF Vice President and PURAPerfect Taylor's mom, participated in the Rare Disease Fair at the University of Washington, March 27, 2026. This event brings together patients, researchers, and advocates to foster collaboration, speed up research, and improve care access. Melissa emceed for the event and moderated a panel that included one of our PURA clinicians, Dr. Chanprasert. She also hosted a PURA Syndrome information booth at the fair to raise awareness among the attending researchers and physicians.

Photo of Melissa Vaught

American Society of Gene + Cell Therapy (ASGCT) Annual Meeting, May 11-15 in Boston, MA

This year, five PURA syndrome research presentations are being given at ASGCT 2026 — the American Society of Gene & Cell Therapy annual meeting, the largest gene therapy conference in the world.

Three of those presentations come from researchers we are funding through our Research Grant Program.

Dr. Kyle Fink (UC Davis Health) and his team — including PURA mom Celena Lozano, a neuroscience PhD candidate in his lab and a member of our board — are presenting their human stem-cell platform for studying PURA syndrome. This work is the foundation of the project we are funding.

Dr. Benjamin Cocanougher (Cincinnati Children's) is presenting his lab's broader research framework on neurodevelopmental disorders where simple gene replacement may not be enough — including PURA syndrome. Our grant supports a specific arm of that work focused on truncating PURA variants.

Dr. James Dowling (University of Pennsylvania) is presenting early findings on PURA gene therapy in a mouse model — including important safety questions that motivated and will be carried forward in the work we are funding.

Two additional PURA presentations come from other groups working in the space — including work supported by Jack's Tomorrow.

This is what investing in research looks like. Not just funding studies, but funding the science that gets presented, debated, and built on by the broader research community. Thank you to everyone who made this possible.

VIEW THE ABSTRACTS

Together, we are stronger — and together, we make a difference.

Ongoing Research

Volumetrics Study in Rare Disorders

This is open to all PURA individuals worldwide
Attention PURA Parents and Caregivers! Do you have an MRI scan of your PURA perfect individual that you are willing to share for research purposes? Dr. Jeff Waugh, a pediatric neurologist and neuroscientist at UT Southwestern in Texas, USA, is conducting a Volumetrics Study in Rare Disorders using MRI scans to understand how the brain is made differently in genetic syndromes. To learn more about this study and how to participate, see below.

Dr. Waugh is a pediatric neurologist and neuroscientist at UT Southwestern, in Dallas, Texas, USA. He uses MRI scans to understand how the brain is made differently in genetic syndromes. For many syndromes, including PURA, the brain MRI looks pretty normal. But when he combines groups of patients and compares them to groups of typically developing kids, he has found more subtle brain differences.

By finding these syndrome-specific brain differences, the hope is that we can better understand why changes in the PURA gene cause the features of this syndrome.
What would you need to do?
If you're interested in learning more, please email Dr. Waugh's research coordinator. Alyssa at (alyssa.boudreau@utsouthwestern.edu).

Developmental and Epileptic Encephalopathy Research (DEER) study for PURA Syndrome

This is open to all PURA individuals worldwide
PURA clinical epilepsy research published in 2021 highlighted the need for additional studies focussed on PURA epilepsy. Through this Developmental and Epileptic Encephalopathy (DEE) Study, we aim to understand the common PURA syndrome clinical characteristics to determine possible treatment requirements and define outcome markers necessary for future clinical trials.

Key research aims are to define the spectrum of PURA epilepsy, describing and comparing seizures and non-epileptic events seen in PURA syndrome. This includes looking at the types, frequency, patterns and severity of seizures, as well as other related issues like medications trialed, developmental delays, cognitive problems, and motor difficulties. PURA individuals with and without epilepsy can be involved.

Individuals with a confirmed diagnosis of PURA syndrome or 5q31.3 deletion syndrome (including PURA) are encouraged to join this study. To be involved in this international epilepsy research project visit the PURA Foundation Australia Epilepsy Research Page here.

GeneReviews Article on PURA in GeneReviews

Check out the March 25 update in GeneReviews on PURA-Related Neurodevelopmental Disorders.

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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.**

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.**

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