

## DECEMBER 2024 UPDATES

**WE WANTED TO SEND A QUICK LETTER TO FAMILIES AND CAREGIVERS TO LET YOU KNOW ABOUT A FEW THINGS BEFORE THE HOLIDAYS!**

### Global Patient Registry

We are slowly making our way to our goal of 100 completed registries but we still need as many people as possible to participate! Below are a few ways the PURA Foundation is trying to help increase our participation and why it is so important:

#### Increasing Participation

- We created a checklist for families that lists the information you need to come prepared with in order to complete the Registry. This checklist translated in Spanish, Deutsch, Polish and Italian so far. We are looking for someone to help us with translation into Portuguese. Please let us know if you need any other translations or if you are able to help us translate into any other language by emailing [families@pura-syndrome.org](mailto:families@pura-syndrome.org).
- We have people willing to help with in-person translations for Deutsch, Polish and Spanish families. Please let us know if you need any other translations or if you are willing to help meet with families to translate while they complete it by emailing [families@pura-syndrome.org](mailto:families@pura-syndrome.org).
- We have two nurses who have one-on-one appointments available to help you through the process. You can book time directly with them at our website: <https://purasyndrome.org/research/patient-registry/>
- Registry meet-ups are back! We held two in November and will be holding more in the New Year. Stay tuned for more information.



#### Why is the Registry important?

The PURA Patient Registry is vital for understanding PURA syndrome!

- It collects comprehensive data on patients, facilitating research into the disease's mechanisms and aiding in the development of targeted treatments.
- The registry accelerates the creation of new therapies by providing a resource for clinical trials,
- It helps improve patient care by developing evidence-based management guidelines and supports affected families by connecting them with valuable information and resources.

#### Get started today...

Contact the study team at [PURA@soton.ac.uk](mailto:PURA@soton.ac.uk) with the following information:

- your contact email
- the PURA study participants name
- the PURA study participants date of birth (DD/MM/YYYY)
- the PURA study participants country of residence

Please allow up to two business days for a response and please keep an eye on your junk mail in case the response does not make it to your inbox.



## Attention Caregivers!

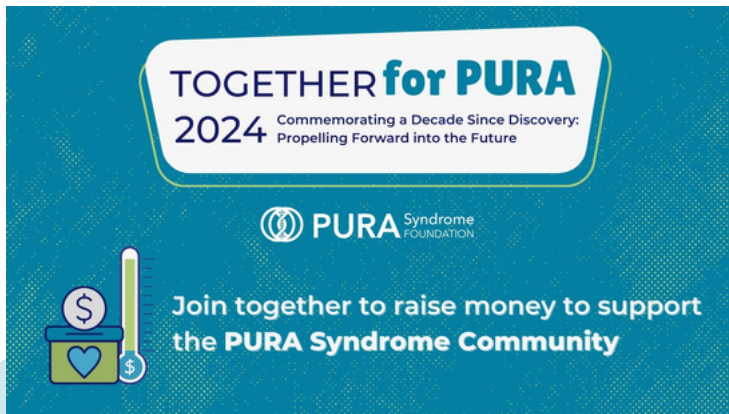
The PURA community is welcome to the first Kelleher Lab Caregiver Webinar Series on December 6, 2024 at 2:00 pm EST hosted by the Kelleher Lab at Purdue University. Attendees need to register by scanning the QR code on the left.

Save the dates for our 2025 PURA Syndrome Conference!!



## PURA Foundation Australia - Now Recruiting for Speech and Language Research for PURA Syndrome

In partnership with the Translational Centre for Speech Disorders, PURA Syndrome Australia has launched an international study looking at speech and language outcomes in individuals aged 6 months and older with pathogenic or likely pathogenic variants in the PURA gene (also known as PURA syndrome). This is a global study funded by PURA Syndrome Australia so it is open to PURA families across the globe. For more information and how to participate, please visit <https://www.purafoundation.au/>



# We are at 22% of our Goal!

We are so grateful to have raised over \$50,000 so far for our Together for PURA Campaign! We still have a long way to go but have some exciting news that will bring us closer to achieving our goal that we will be announcing in the next few weeks. Can you help us?

- Giving Tuesday is tomorrow, December 3, 2025. Encourage your family and friends to donate \$10 for 10 years of PURA Syndrome by sharing our link: <https://givebutter.com/c/pura2024/>
- We are excited to announce that the Knights of Columbus in Orleans, MA will sponsor a pancake breakfast in February 2025 with all proceeds donated to the Together for PURA campaign! Do you have a Knights of Columbus or other charitable organization in your town that might be willing to host a breakfast event for PURA? If you are interested, we can help and get the ball rolling! We can provide you with a presentation (and/or present to your organization virtually) to talk about PURA and ways they can support the Foundation. Let's make this a true Pancake Palooza for PURA in cities and towns across the world! Contact Nancy at [n.boccia@pura-syndrome.org](mailto:n.boccia@pura-syndrome.org) for details and assistance!
- Help us increase our donations by checking our corporate matching box when making a donation. If your employer does match, they will walk you through step-by-step on how to get us the donation.
- Questions? Ideas? Contact Nancy at [n.boccia@pura-syndrome.org](mailto:n.boccia@pura-syndrome.org)

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***The PURA Syndrome Foundation is a volunteer based organization. With that comes the need to recruit many hands to accomplish our mission and the goals we set forth as an organization. Please consider helping out if you are able to do so, or if there is someone in your life you think may be able to help ask them to fill out the form on our website or reach out to [volunteer@pura-syndrome.org](mailto:volunteer@pura-syndrome.org).***