NOVEMBER 2023

PURA Syndrome FOUNDATION

# NEWSLETTER

## A letter from our Co-Presidents

Hello PURA Community!

It has been a whirlwind as we transitioned to our new role as co-presidents this year, all while launching a new website and planning and attending our 2023 Conference in the UK. It is challenging to navigate through running a rare disease organization but we have been building a team and network around us that will help us be as successful as possible. On the admin side, we have been implementing tools to help the foundation run more efficiently so we can put our focus where it matters in effectively supporting collaboration and community across the world. We are so grateful to the whole board, volunteers, and Global Research Network for all the ways they have been stepping up to support the PURA community this year.

We recently had some Foundation representatives attend the Global Genes Conference while we attended virtually. This was a really wonderful opportunity to hear from other Rare Disease organizations, their successes and how they learned from their failures. We are already utilizing a lot of this information in our strategic planning and are looking forward to a fruitful 2024!

One of the things we constantly say to each other is that our priority as a Foundation always has to be our PURA children and whatever is best for them. We hope you agree with that sentiment and please know that at the core of every decision we make, is each and every one of our PURA children.

Sincerely,
Liz Astridge & Eva Tucker

## PURA Awareness

We'd like to extend a heartfelt thank you to all who participated in our PURA Awareness campaign during the month of October. We had a variety of opportunities to participate and enjoyed the lead - up to our October 23rd PURA Awareness Day. A special thank you to Angelina Bueno who created all of the graphics for our social media campaign!


We are happy to announce that our 2024 PURA Syndrome Conference will take place June 21-23, 2024 in Chicago, IL, USA. Please stay tuned for more details including hotel blocks and information about the Abilities Expo happening nearby on the same weekend!

# Departures & Arrivals

## Thank you Jennifer!

We'd like to take a moment to express our gratitude to Jennifer Bierling, who is stepping down from our board. She has been a wonderful team member and was instrumental in the launch of our Global Patient Registry last year. Her hard work helped the project get off the ground and we are very grateful for that and her many other contributions throughout the years. Thank you for everything, Jen, you will be missed!

With Jen's absence opens the Secretary position on our board, so if someone you know may be willing to fill this role, please visit our website at www.purasyndrome.org to read the duties of the Secretary and how to apply.

## Introducing our new Research Liason, Jim Lagowski

We'd like to welcome our new Research Liaison and PURA Uncle, Jim Lagowski! Jim has been an instrumental part of the team so far in helping organize our Global Research Network Meeting at our annual conference, helping our board members understand some of the more scientific aspects of our Foundation, networking with other Rare Disease Organizations at the recent Global Genes conference and working with some of our PURA parents to establish a PURA Syndrome Clinicians Network. We have kept him very busy and are grateful for his contributions to the Foundation thus far. Enjoy reading his bio below and welcome, Jim!

*Jim received his M.S. in Natural Sciences at the State University of New York at Buffalo while working at Roswell Park Cancer Institute, and his B.S. in Biology, also in his hometown at SUNY Buffalo. Jim's primary training is in molecular biology, beginning his 20+ year tenure at Oregon Health & Science University (OHSU) in Portland, Oregon, USA. He worked "at the bench" in the Dermatology Research Division where he studied biochemical pathways involved in dermatological and head and neck cancers for more than 12 years. From there he moved on to the OHSU Knight BioLibrary; a university-wide human tissue biorepository, to set up and manage a small team which continues to support scientists who seek valuable specimens for their research.*

*Currently, Jim works in OHSU's Technology Transfer Department as an Agreements Manager, where he negotiates agreements between OHSU scientists and their industry and academic partners, protecting their interests in new intellectual property and facilitating the ongoing and far-reaching collaborative environment at OHSU. Gardening, pinochle, camping, and tending to his animals Blue (horse), Jack (mutt), and Boba (St. Bernard) are a few things that occupy his time when away from the office.*

## Attention PURA Siblings!

We are so excited to introduce Chrissie Brock as our new Siblings Ambassador. After meeting at our 2023 PURA Conference in the UK, Chrissie reached out to volunteer to help the Foundation to fill the role of our Siblings Ambassador. She has spent the past few months learning about the siblings programs that other rare disease organizations offer and has been putting together a plan for our PURA Syndrome siblings that will begin rolling out in the new year. Enjoy reading her bio below and welcome, Chrissie!

*My name is Chrissie. I'm the older sister to Karen, who received her PURA diagnosis the day before her 41st birthday in 2019. Karen lives in Melbourne where we were both born and brought up. I now live in England with my husband and two dogs.*

*I hope that by helping to set up a siblings network with the PURA Syndrome Foundation, we can create a community for siblings of our wonderful PURA people to meet, share stories and exchange experiences. Chrissie can be contacted at siblings@pura-syndrome.org*

# PURA SYNDROME IS TURNING 10 IN 2024!

Did you know that October 23, 2024 will mark the 10th Anniversary of the discovery of PURA Syndrome? We are putting together a year-long celebration with monthly themes so stay tuned to social media and email for more details. Interested in helping? Please contact c.thorpe@pura-syndrome.org.

ORIGINAL ARTICLE

**Whole exome sequencing in family trios reveals *de novo* mutations in *PURA* as a cause of severe neurodevelopmental delay and learning disability**

David Hunt,[1] Richard J Leventer,[2] Cas Simons,[3] Ryan Taft,[3,4,5] Kathryn J Swoboda,[6] Mary Gawne-Cain,[7] the DDD study,[8] Alex C Magee,[9] Peter D Turnpenny,[10] Diana Baralle[1,11]

## Parents & Caregivers: We need YOUR help to take our research to the next level

### Help us complete 100 Patient Registries by October 2024!

**The Foundation, in partnership with the University of Southhampton in the UK, made a full launch of the Global Patient Registry in 2022. Since then, only 35 registries have been completed with about 40 more started but incomplete. The more families we can get to complete it, the more insight it will offer researchers into PURA Syndrome. For more information and to register, please visit: purasyndrome.org/research/patient-registry/**


**Our research liaison Jim Lagowski and board member Melissa Vaught attended the Global Genes conference in September. They were able to network with other Rare Disease Organizations and brought back a lot of insights that will help the PURA Syndrome Foundation with their upcoming strategic planning session!**

***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.***