



2023 ANNUAL REPORT

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Table of Contents

Letter from the Co-Presidents	3
PURA Syndrome Clinician Network	4
New Sibling Ambassador	5
Self-Care for Caregivers	6
Strategic Planning	7
Research Update	8
Registry and Biobank Update	9
Conference Update	10
Financial Report	11
Board Update	12
Special Thanks	13

Letter from our Co-Presidents

Dear Friends of the PURA Syndrome Foundation,

It is with deep gratitude and appreciation that we are again writing this letter to all of our families, friends, supporters, and donors. With any transition, our entrance into the roles co-presidents was filled with a lot of conversations, establishing relationships and learning about what we needed to do to build on the work of those that came before us. All these things together will help us to steer the PURA Syndrome Foundation in a positive direction to support the needs of our community while ensuring long term growth and sustainability.

One of the large projects we embarked on late in 2023 alongside the Board of Directors was strategic planning, which took place over a four month period. We began by determining the pillars of the Foundation we wanted to focus on: Family Support, Research, Global Patient Registry, Biobank, Financial Sustainability, Infrastructure and Process, Data Management and Marketing/Communications. From there we did a SWOT analysis (strengths, weaknesses, opportunities, and threats) on each of these pillars which then we converted to key action steps with concrete timelines, milestones and leadership roles. We are hopeful that by putting this strategic plan in place we will have better transparency in how our Foundation is working towards achieving our mission!

Another key project we have been focusing on is expanding resource offerings to our community. In 2023 we began reaching out to clinicians who work with PURA patients and are officially launching our PURA clinician directory. We are hopeful that this will not only help families when seeking out physicians, but it will increase communications between clinicians who are working with PURA patients in order to better serve our population. We also announced our new sibling ambassador, Chrissie Brock, who you will read more about later in this report. Siblings play a big role in our PURA community and we are excited to be offering opportunities for them in the near future.

We are grateful for each of you who continue to support us in furthering our mission to drive research, raise awareness in the general community, as well as connect, educate, serve and empower families impacted by PURA Syndrome.

Sincerely,

Liz Astridge & Eva Tucker

Co-Presidents, PURA Syndrome Foundation

PURA Syndrome Clinician Network

After discussing with our Global Research Network and talking to parents, our Foundation decided to launch the PURA Syndrome Clinician Network. We began reaching out to physicians and building the network in the Fall of 2023 and plan on launching shortly.

This network will consist of doctors across the globe who have more than one patient with PURA Syndrome and have been highly recommended to us by another clinician or caregiver. The purpose of this is twofold: 1) To provide a resource for families that they can check if they are looking for primary care or a particular specialty for their child and 2) To provide a resource for clinicians that will allow for open communication between them so that they can better care for our PURA family members.

While our initial launch will primarily have clinicians from the United States and Europe, we are looking to expand this to aid our global community. If you have a clinician you would like to recommend for our PURA Syndrome Clinician Network, please pass along their name, email address and country to: president@pura-syndrome.org.



New Sibling Ambassador

Late in 2023 we introduced our new Sibling Ambassador, Chrissie Brock. Her younger sister Karen received her PURA diagnosis the day before her 41st birthday in 2019. Karen lives in Melbourne where they were both born and brought up. Chrissie now lives in England with her husband and two dogs. She hopes that by setting 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.

We are so excited to be offering this outreach to the many PURA siblings in our community. Our goal is to offer a variety of different ways to support siblings of all ages, including meet ups, podcasts, books, websites, and other resources to help along your journey.

Chrissie has been hard at work putting together these initial offerings and has many ideas on how to expand them going forward. We will begin offering meet ups for adult siblings first, and will add on different age groups. Please continue to check back to our siblings page at www.purasydrome.org/siblings as we will be periodically adding more resources and regular meet ups to the calendar.



Self-Care for Caregivers

The PURA Foundation partnered with Behavioral Change Specialist and author of The Kindness Method Shahroo Izadi to create self-care resources for our PURA caregiver community that one can do in the comfort of their own home!

Shahroo began by surveying our parents and from those responses, created resources for our PURA caregiver community. We wanted to provide different types of formats for those in our community who might like to sit down and read/write (workbook pdfs) or for those of who may only have time to listen (audio files) while doing a myriad of other things.

We hope you are enjoying these resources, as this is just the beginning of providing our community with materials to help along their caregiving journey. If you have yet to utilize these amazing resources, they can be found on our website at: www.purasyndrome.org/oxygenmaskproject



on-the-spot tools
BITESIZED MOTIVATIONAL RECORDINGS

Strategic Planning

In the Fall of 2023, our PURA Syndrome Board began meeting once a month (in addition to our once a month board meetings) to embark on strategic planning for the Foundation. We began by choosing the pillars that we wanted to focus on, which were Financial Sustainability, Family Support, Infrastructure & Process, Research, Registry, Biobank, Data, and Marketing/Communications. From there, we set a goal and then did a SWOT analysis (strengths, weaknesses, opportunities, and threats) for each of these pillars. From there we created key action steps with a timeline, leadership, resources and ongoing actions. All of this will be uploaded into a special program that will help track our progress and keep our group on task. We look forward to sharing more of this with you at our conference in Chicago at the end of June!

Pillars							
Financial Sustainability	Family Support	Infrastructure & Process	Research	Registry	Biobank	Data	Marketing/Communications
Goal:	Goal:	Goal:	Goal:	Goal:	Goal:	Goal:	Goal:
Key Action Steps:	Key Action Steps:	Key Action Steps:	Key Action Steps:	Key Action Steps:	Key Action Steps:	Key Action Steps:	Key Action Steps:

Marketing and Communications

<p>STRENGTHS</p> <p>What are my strengths? What is going well?</p>	<p>WEAKNESSES</p> <p>What are we missing? What is not going well?</p>	<p>OPPORTUNITIES</p> <p>How can we improve or make things easier?</p>	<p>THREATS</p> <p>What challenges are we facing? What could go wrong?</p>
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Research Update

**MESSAGE FROM CHAIR AND
COORDINATOR OF GRN
MATT GUILLE**



**Professor of Developmental Genetics
at University of Portsmouth**

In late 2023, Matt Guille and our GRN compiled the following research summary:

Our overall aim is to understand the mechanisms that link variants of the Pura gene with the pathogenicity experienced by patients and the natural history of the disorder. We will then use this knowledge to support patients by investigating the best management practices and establishing treatment strategies for the disease. To achieve this aim, we have several objectives on different but interconnecting levels that together make our current strategy:

1. **At the Patient Level**, the GRN is gathering patient data and tissue samples to enable future experiments such as testing whether findings in cellular and animal models of PURA syndrome recapitulate those in patients. This is being achieved by the Patient Registry, Natural History of the Disorder Study and Biobank.
2. **At the Research Animal Model Level**, the GRN is establishing models of PURA syndrome. Human stem cell iPSC, mouse, zebrafish and Xenopus models of the disease are now available within the GRN.
3. **At the Phenotypic Level**, the GRN is analysing the changes in established models that may cause the pathogenicity of PURA variants. We have specialised imaging, behavioural (including seizure analysis), biochemical, cell biology, transcriptomic, proteomic and metabolomic techniques to analyse these changes available within the GRN.
4. **At the Molecular Protein Level**, the GRN is examining the pathogenicity of new and well-known PURA variants by understanding the relationship between changes in the sequence of PURA protein, cellular function and animal model phenotype. We are using structural biology, predictive algorithms, classical molecular genetics approaches, gene and base editing to make and test the function of new and existing PURA variants.
5. **At the PURA Protein Family Level**, the GRN is investigating whether other members of the PURA protein family could compensate for PURA variants in patients. We have lines of Xenopus in which these PURA protein family members are knocked out to determine whether their functions overlap with PURA, and cell lines in which their ability to compensate for PURA loss can be tested.
6. **Sharing Resources:** The GRN supports fostering research by collaboration and exchange of resources, subject to normal academic (not for profit organisations) or commercial agreements with the originating laboratory.

Research Update

Registry Update – *Need more participation!*

The PURA syndrome global patient registry was formally launched in January 2022 in partnership with University of Southampton and the Clinical Informatics Research Unit (CIRU). Working in collaboration with families, clinicians and researchers, this project collects clinical data on PURA syndrome from affected patients globally throughout the course of their lives. As of the end of 2023, we only have 78 patients enrolled with only 47 of those enrolled having a majority of their data entered. The more data we have, the more likely our researchers will be able to define the full spectrum of the disorder, and also characterize the natural history (what happens across age and life) for PURA syndrome. **We need as many families as possible to participate.** For more information and how to sign up, please visit our website at: www.purasyndrome.org/registry

Biobank Update from Dr. Dierk Niessing – *Launching Soon*

After the ethics approval of the PURA-Biobank in 2022, the Biobank team was busy establishing procedures and protocols as well as a registration website. The current development team consists of the PURA Fellow Simone Riebe, the lab head Dierk Niessing and the coordinator of the Biobank facility at Helmholtz Zentrum Munich, Dr. Bettina Lorenz-Depiereux. In addition, the team is grateful for the ongoing support they receive from PURA mother, Melinda Anderson (Australia). In 2023, the team had to overcome unexpected hurdles and delays due to a complete reorganization of the Biobank facility at Helmholtz Zentrum Munich. Once the IT integration of Biobank and Patient Registry has been finalized and contractual details are settled, the Biobank is ready for launch. The Biobank will be physically located in Germany but will serve PURA research worldwide. The Biobank team expects the PURA Biobank to start soon and will announce it accordingly.

Conference Update

Our annual conferences are an important time to gather together families and researchers to connect, share, and learn from one another. The theme of our annual conference in June 2023 was Forward Together Again. The conference, which took place in person for the first time since 2019, was in Hinxton, UK from June 23–25, 2023. It was a wonderful opportunity to reconnect with our fellow PURA families and friends. We offered a virtual option for those who were unable to attend in person and made any recorded sessions available to those who couldn't join live. Visit puraconference.org to view past recordings.

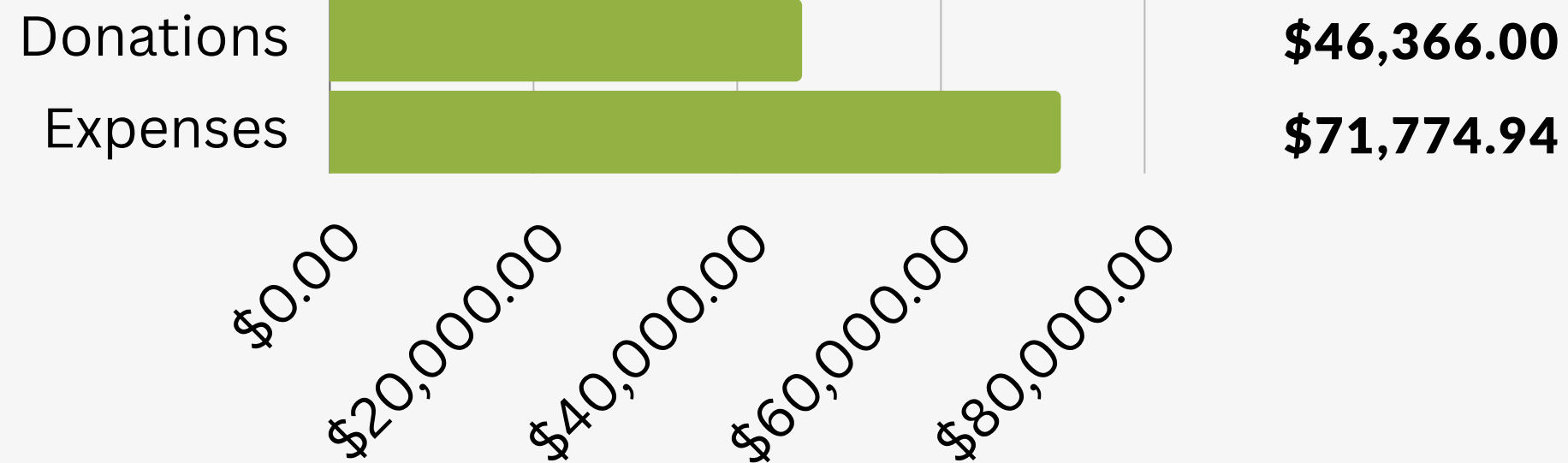
We are looking forward to our 2024 conference which will take place June 21-23, 2024 in Chicago, Illinois, USA. We have a wonderful array of speakers mainly from the Chicago area and a few of our GRN members will be joining us from Europe to give updates on our PURA research. This year's conference is also the same weekend as the Abilities Expo and is within walking distance to our conference hotel. The Chicago Abilities Expo is free to attend and will have products and technology to view and demo, as well as workshops to attend, or simply connect with others in the disability community.

The next conference will be back in the UK in 2025.

puraconference.org



2023 Financial Report



2023 was a year of high spending by the foundation. This was due to a few factors, including:

- Two of our research grants approved in 2022 were paid out in 2023.
- Our conference is back to being held in person so conference expenses are increasing. While most of our labor is volunteer, the venue and other costs will remain higher now that we are in person.



In order to ensure the sustainability and growth of the PURA Syndrome Foundation, we will be ramping up fundraising activities in 2024, working towards the goal of funding more research grants and being able to hire a full-time executive director to help manage the day-to-day activities of the organization and better serve our community. Please contact us at info@pura-syndrome.org if you are interested in hosting a fundraiser and visit our website purasyndrome.org for ideas.

Board Update

In 2023, we were delighted to add Nancy Boccia to our Board of Directors. Nancy has been instrumental in the planning of our conferences since 2021 and has been a wonderful addition to our PURA Executive Board.

We remain grateful to all of the board members past and present who have contributed their time and energy in the service of our beautiful PURA community. Thank you to our board members who have stepped down over the past year for your service and commitment to the PURA Community: Jennifer Bierling, Caroline Thorpe and Mel Wardhaugh.

2024 Board of Directors:

- Liz Astridge, Co-President
- Nancy Boccia
- Damon Fisher, Treasurer
- Eva Kittay
- Eva Tucker, Co-President
- Melissa Vaught



LIZ ASTRIDGE, CO-PRESIDENT



NANCY BOCCIA



DAMON FISHER, TREASURER



EVA KITTAY



EVA TUCKER, CO-PRESIDENT



MELISSA VAUGHT

Special Thanks

Thank
you!

- Global Research Network
 - Prof. Matthew Guille
 - Dr. David Hunt
 - Prof. Diana Baralle
 - Sophie Powell
 - Dr. Margot Reijnders
 - A/Prof. Rob Rouhl
 - A/Prof. Rick Leventer
 - Prof. Guido Rubboli
 - Katrine Johannesen
 - Annie Godwin (PhD)
 - Prof. Dierk Niessing
 - Sabrina Bacher
 - Sandra Burczyk
 - Estera Jeruzalska
 - Carolin Ketteler
 - Dr. Kathi Zarnack
 - Melina Klostermann
 - Dr. Robert Janowski
 - Dr. Gabriele Anton
 - Simone Riebe-Züfle (fellow of PURA Syndrome Foundation)
- Nancy Boccia for her continued support organizing our annual conferences
- Jim Lagowski for his support to the Global Research Network
- Mel Anderson for her ongoing support to the Registry and Biobank
- Ceciel van Hoeckel and Mel Anderson for their diligence and hard work tracking and maintaining statistical data on PURA

2023 Donors

\$5,000-\$10,000

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- Eric Knapp & Debbie Hussey

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Special Thanks

thank you

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<\$200

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- Zelde Krulewitz
- Verena and Christian Kummerfeldt
- Aaron Lagowski
- Alison Lagowski

Special Thanks

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Thank You