



2022 ANNUAL REPORT

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WWW.PURASYNDROME.ORG

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The Year in Numbers

620

KNOWN CASES OF PURA
SYNDROME WORLDWIDE

306

PATHOGENIC VARIANTS
IDENTIFIED

53

COUNTRIES WITH KNOWN
CASES OF PURA

20

RESEARCHERS IN GLOBAL
RESEARCH NETWORK

9

STUDIES PUBLISHED IN
2022 REFERENCING PURA*

8

WORLDWIDE PURA
SYNDROME AMBASSADORS

4

RESEARCH GRANTS
IN 2022 BY PURA
SYNDROME FOUNDATION

\$69,849

DOLLARS RAISED IN 2022
BY PURA SYNDROME
FOUNDATION

*<https://pubmed.ncbi.nlm.nih.gov/>

Letter from Liz & Eva

Dear Friends of the PURA Syndrome Foundation,

The PURA Syndrome Foundation hopes this message finds you well. It is with deep gratitude and appreciation that we are writing this letter to all of our families, friends, supporters, and donors. 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.

As we have embarked on this new role as co-presidents over the past few months, we move forward humbly building on the work of those that came before us to get the PURA Syndrome Foundation to where it is today. We move forward from the pandemic and look forward to being back together again at the upcoming conference in the UK. Then back together in the U.S. in 2024.

You may have noticed we recently launched a new logo and website. While our original logo will remain a treasured symbol of PURA Syndrome, our new logo symbolizes the core elements of the foundation's mission — community and research — and the new format works for our growing needs. The new website is designed to help us better organize our information and serve the needs of our community — at every stage of diagnosis. Continuing to build out the resources on the site is going to be a key focus for us and we appreciate your feedback and support.

In this report we will share a lot of exciting progress, including the launch of the global registry, biobank, affiliate and ambassador programs. None of which would be possible without the PURA parent warriors and caring researchers around the world that have contributed their time and energy to making it happen.

We continue to be amazed at your continued support and are poised to make exciting investments in research in 2023. We remain deeply committed to our mission and are looking forward to the many opportunities to connect again in-person.

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

Ambassador Program

During 2022 we launched the Ambassador Program. As our community grows around the world we, at the PURA Syndrome Foundation, want to be a part of this amazing experience and assist in every way we can. We have developed this program to help practical matters in the most direct and effective way possible. This consists of having a representative, who is the person of reference, for different regions and for specific topics. They are known as our PURA Ambassadors.

They cover many different functions including:

- welcoming new families
- helping families at any point in their PURA journey
- helping to answer questions directed to the region or topic
- hosting online meet-ups where families can gather together and talk about different topics, help each other, share their experiences, meet other families, among many others
- assisting with translations
- they are working on providing resources to assist our families
- and guiding in any way possible among many others

These are the regions we are currently working with: Austria, France, Germany, Italy, Spain, UK, US and a topic based one which is called Caring Together. This last one is to promote the power of us joining together, as families, to support each other, and to be there for each other. This could be when you are newly diagnosed, when you are going through a new stage, if you have lost your PURA child or you are simply at a point in your life when you feel like having a community surrounding you could help or to share your experience.



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SPAIN



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SOUTH AMERICA



JENNIFER HEROLD
GERMANY



ALBERTO CASALICCHIO
ITALY



MEL WARDHAUGH
UNITED KINGDOM



MELISSA VAUGHT
UNITED STATES

Thank You
Ambassadors
and special thanks
to Caroline Thorpe
for her dedication
to this program.

Affiliate Program

The PURA Syndrome Foundation is a global non-profit organisation. Our mission is to support and drive research, raise awareness in the general community, as well as connect, educate, serve, and empower families impacted by PURA Syndrome. As we have grown, the need has come up for members of our community to incorporate their own charitable organization in their home country, affiliated with the PURA Syndrome Foundation, but independent in operations.

One of the key objectives for the Foundation and the Affiliate is to work together towards common goals and to direct individual accomplishments toward fundamental objectives, cooperating with each other to succeed in these principles.

The primary purposes of the Affiliate, which must be reflected in its organisational documents, must be to further the mission of the Foundation to support research, educate and raise awareness about The PURA Syndrome Foundation.

We are currently finalizing our initial agreements and will be announcing the first PURA Syndrome Foundation affiliates soon. Contact info@pura-syndrome.org if you are interested in learning more about this program.



Research Update

**MESSAGE FROM CHAIR AND
COORDINATOR OF GRN**

MATT GUILLE



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

With the advent of gene editing, which works extremely well in the frog, Matt sought collaborations with clinical geneticists to test whether the frog could be a useful tool to help them diagnose and understand rare genetic diseases.

It is wonderful to be able to report some significant findings from the Global Research Network (GRN) as the labs involved finally completed their recovery from the research delays caused by the pandemic. In particular, the molecular activity of PURA in cells was reported by a team led by Dierk Niessing in “Nucleic Acids Research” at the beginning of 2023. In this work, Dierk and his team used a number of tools, both new and established, to take a genetic “snapshot” inside cells to get new insight about how PURA behaves in the cell, as well as to pull out 236 potential protein targets from more than 55,000 PURA binding sites they identified in genes. This work gives us some deep insights into the mechanism by which PURA acts within cells and what it is doing, as well as some new leads to pursue as we seek these answers, which will ultimately tell us why PURA’s reduced expression creates such a disruption in cellular function and human development.

These are some big, albeit first steps in the long road to intervention, where the last sentence of the paper states it well by saying: “While this study opened the door for an unbiased and systematic understanding of molecular PURA-dependent pathways, it will require more research on both ends, clinical and non-clinical, to convincingly connect our findings with the symptoms in PURA Syndrome patients.” One of our current challenges in bridging that gap between non-clinical basic science and clinical intervention is the need for access to patient data and samples, and we are taking steps with both the Patient Registry and the Biobank, set up by GRN members, to continue to seek more input from patients, their families, and their doctors. It’s exciting to be fully “back to the bench,” and with our continued efforts to build these patient-based resources, and with new data arising from cell-based studies, our knowledge of PURA will grow and further inform progressive studies to be undertaken by the GRN (and other) labs going forward.

The GRN members are all looking forward to being able to meet in person for the first time in years (for the first time ever for some newer members!) later this year and we are excited to hear of the advances made in understanding this disease that are not yet ready for publishing, but are ready to share.

Thank you all for your inspiration and support, we look forward to meeting many of you in June.

Research Update

Registry Update – *Launched in 2022*

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 proposes to collect clinical data on PURA syndrome, from affected patients globally throughout the course of their lives. This should enable researchers to define the full spectrum of the disorder, and also characterise the natural history (what happens across age and life) for PURA syndrome.

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

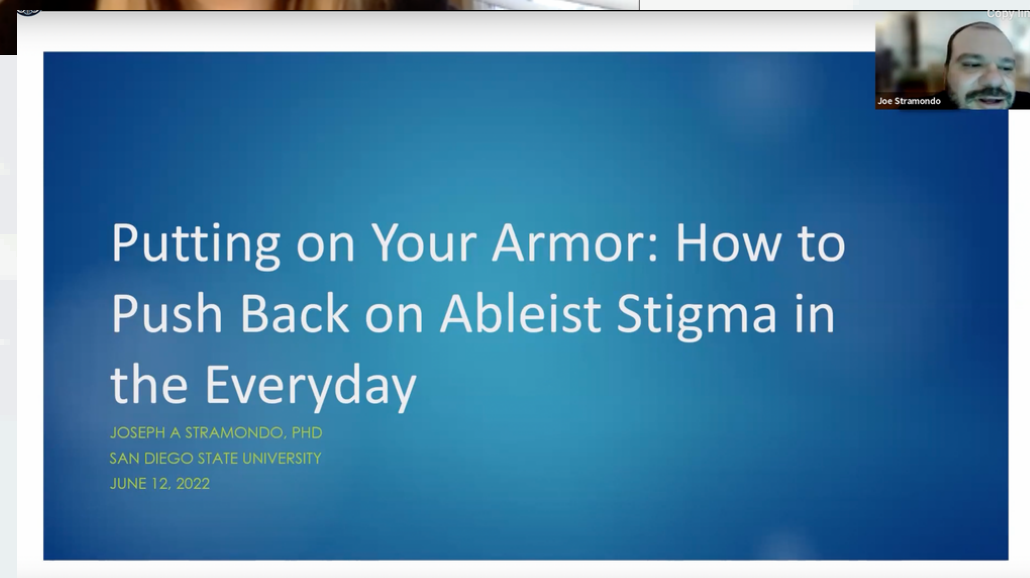
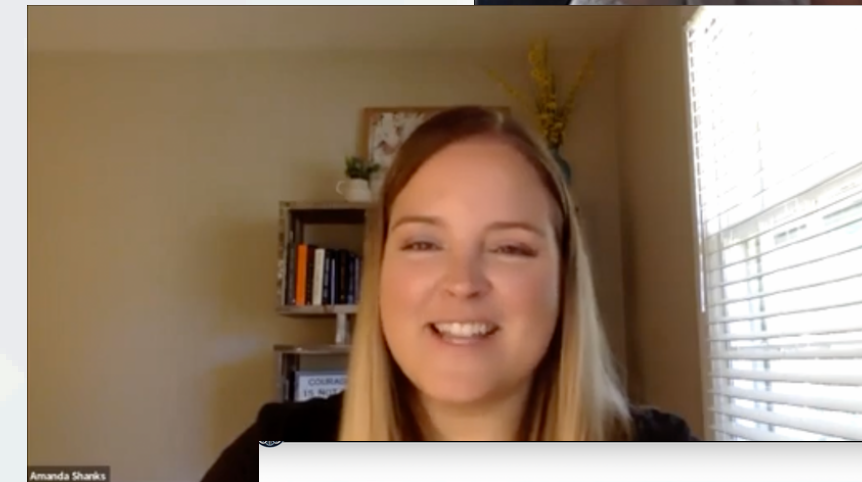
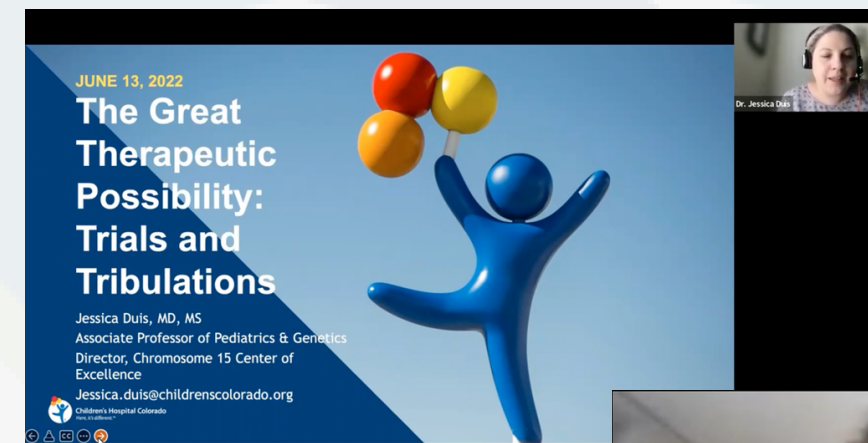
After the ethics approval of the PURA-Biobank in 2022, the Biobank team was busy establishing procedures and protocols as well as a registration website. The current development team consists of the PURA Fellow Simone Riebe, the lab head Dierk Niessing and the head of the Biobank facility at Helmholtz Zentrum Munich, Dr. Gabriele Anton. In addition, the team is grateful for the ongoing support they receive from PURA mother, Melinda Anderson (Australia). 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

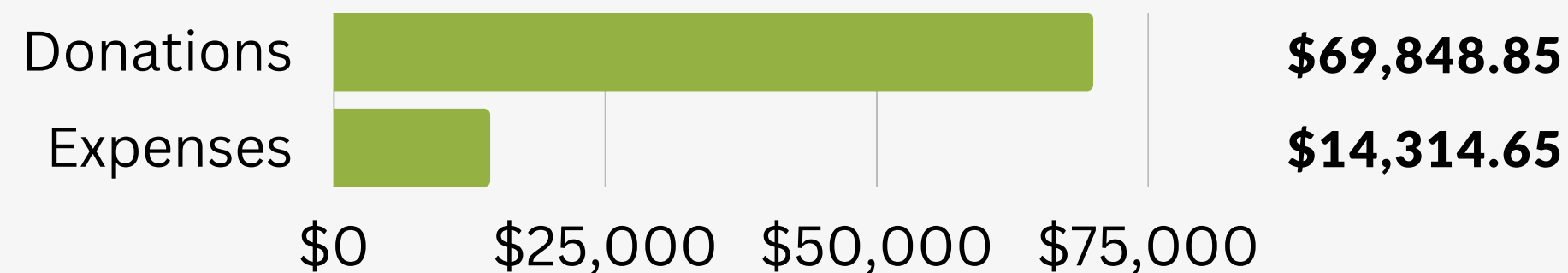
Our annual conferences are an important time to gather together families and researchers to connect, share, and learn from one another. In 2021 and 2022, we held virtual conferences that brought together the PURA Community from around the world over the course of a week in June. The virtual format allowed many to attend that wouldn't have been able to travel to be together in person. We recorded most of the sessions to make the presentations available to those who couldn't join live. Visit puraconference.org to view past replays.

The theme of our upcoming annual conference in June 2023 is Forward Together Again. This is a very fitting theme for this year in general, as we are moving forward from the shut downs and cancellations that the pandemic thrust upon us. Our in-person gatherings that provide meaningful connection and education for families are coming back and have taken place in different cities around the world. Our conference, which will take place in person for the first time since 2019, is in Hinxton, UK from June 23–25, 2023. We are excited to welcome many wonderful speakers and are offering a full schedule of research sessions to get everyone up to speed on what is happening. It will be a wonderful opportunity to reconnect with our fellow PURA families and friends. One good thing that came from the pandemic is that our conference committee is comfortable with online technology, so there will be a virtual option available for those who are unable to attend in person. The next conference is anticipated to be in the U.S. in 2024.

puraconference.org



2022 Financial Report



2022 was a year of unusually low spending by the foundation. This was due to several factors, including:

- Because of the generous support of our volunteers, we incurred minimal expenses to host our 2022 virtual conference.
- One of the research grants approved in 2022, wasn't paid out until 2023.
- In addition to the annual conference, we were focused on launching the new website and ambassador program in 2022, which was done with all volunteer labor.
- Foundation support for research nurse for the registry and a fellow for the biobank paid out prior to 2022 had not been completely spent down.



In order to ensure the sustainability and growth of the PURA Syndrome Foundation, we will be ramping up fundraising activities in 2023, 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

It takes a lot of heart and dedication to build and grow a global patient organization. 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: Amanda Shanks, Jon Erickson, Todd Vaught

2023 Board of Directors:

- Liz Astridge, Co-President
- Jennifer Bierling, Secretary
- Damon Fisher, Treasurer
- Eva Kittay
- Caroline Thorpe
- Eva Tucker, Co-President
- Melissa Vaught
- Mel Wardhaugh



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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
 - Katrin 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

2022 Donors

\$5,000-\$10,000

- Anonymous Donor
- Eric Knapp & Debbie Hussey

\$1,000-\$4,999

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- Eva Kittay
- Emily & Robin Klem, Renee Laiuvara
- Amy Lagowski

Special Thanks

thank you

2022 Donors

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- Peter Bowes
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- Alessandra Cancado
- Philippa Catt
- Shea Chambers
- Franck Chauvel
- Clarec Cherry
- Paula Cherry
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- Shirley Cook

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- Michelle Cox
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Special Thanks

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- Shannon Welch
- Caroline White
- Alex Willis
- Richard Worf
- Stephen Wyness
- Marvin & Pamela Yeargain
- Farrah Youngerman
- Eva Zhung

Thank You