PURA FOUNDATION AUSTRALIA

Annual **Report**

24-25

PURA Foundation Australia acknowledges the traditional Aboriginal owners of the land we work on and pay our respects to them, their culture and their Elders past and present. We are committed to cultivating a safe, accessible and inclusive work environment for our Board and Volunteers that values and embraces people of all backgrounds, genders, cultures and abilities.

PO Box 90, Diamond Creek 3098
info@purafoundation.au

Welcome

The PURA Foundation Australia is a non-profit charity dedicated to providing national and local support for individuals with PURA syndrome and their families across Australia and New Zealand.

Established to raise awareness, provide education, and support research, the Foundation works tirelessly to benefit those living with PURA syndrome and their families. With guidance from both scientific and community stakeholders, we aim to accelerate knowledge, expand clinical and scientific research, develop educational resources, and ultimately support the creation of precision therapies for PURA syndrome.

We understand that caring for a child with PURA syndrome often means focusing on daily routines and responsibilities, but carer wellbeing is just as important. Connecting with other PURA families provides valuable support, shared understanding, and practical advice. By building relationships with those who truly understand the journey, families create a stronger, more resilient foundation for themselves and their loved ones.

The PURA Foundation Australia supports the PURA syndrome community both locally and globally. Our community offers a place of belonging for those who might otherwise feel isolated by rare disease, enriches ongoing medical research, and provides education about this complex condition.

When we all work together, we can achieve anything.

www.purafoundation.au

What does the PURA Foundation Australia do?

Education and Advocacy

Empowering families and the rare disease community

Awareness

Creating visibility and collaboration

Community

Creating collaborative spirit and belonging

Support

Standing beside families on their PURA journey

Research

Supporting scientific and clinical work that reflects the priorities of PURA families

Our Goals

- **To advocate for individuals with PURA syndrome and their families.**
- **To facilitate education and awareness of PURA syndrome.**
- **To advance research and precision therapies for PURA syndrome.**

Families Foundation, clinicians, and scientists as one community.

Or as we call it - Team PURA.

To assist us with our goals and our mission, please consider a donation to the PURA Foundation Australia today.

Governance

Melinda Anderson

Director - Chair

Research and Governance

It is with great pride that I present the PURA Foundation Australia's second Annual Report. Building on the strong foundations established in our first year, the past twelve months have seen significant progress in advancing our mission to support individuals affected by PURA syndrome and their families.

Our governance framework has continued to provide the structure and accountability needed to guide our growth. With effective leadership from our Board of Directors and Executive team, we have strengthened our strategic direction, ensuring every decision is mission driven and resources are directed towards meaningful outcomes for our community.

This year, our connections with researchers and clinicians have deepened. We have continued to invest in partnerships that advance knowledge of PURA syndrome and contribute to the pathway toward therapeutic clinical trials. The momentum generated by these collaborations is a source of great hope, bringing us closer to the possibility of meaningful treatments for those living with PURA syndrome.

Equally important has been our focus on families. Our community has grown stronger and more connected through outreach, events, and shared experiences. The voices of our families remain central, shaping the priorities of the Foundation and ensuring lived experience guides research and advocacy.

Looking ahead, the PURA Foundation Australia is committed to building on these achievements. We will continue to fund and support critical research, expand our advocacy efforts, and provide opportunities for families to connect, learn, and be heard. With the strong base we have created, and the growing support of our community and partners, we are well positioned to drive long term impact for those living with PURA syndrome.

Together, we are making a lasting difference in the lives of individuals and families affected by PURA syndrome.

Our Team

At the PURA Foundation Australia, our strength continues to come from our team. Every milestone we've reached this year—whether in fundraising, advancing research, or welcoming more families into our community—has been achieved together. Collaboration has been at the heart of our progress, helping us face challenges with resilience and creating deeper connections across the PURA syndrome community.
Teamwork is what turns individual efforts into collective impact. Together, we are building something greater than any one of us could achieve alone: a community defined by support, advocacy, and hope. This spirit of working side by side is what allows us to continue driving change and offering strength to families living with PURA syndrome.

Through this shared commitment, the PURA Foundation Australia is not only making progress in research and awareness but also nurturing a thriving network where families feel supported and empowered. Our teamwork is creating a legacy of care, resilience, and possibility for the future.

Melinda Anderson

Director - Chair

Natasha Keetley

Director

Neil Anderson

Director

Kirsty Truemann

New Zealand Liaison

Amy Zagato (Bowen)

Fundraising and Events

Rebecca Nunn

Parent Support Victoria Secretary

Jaime Martin

Parent Support Queensland

Te Tane Trinick

Parent Support New Zealand

Community Engagement

Natasha Keetley

Director

Community and Outreach

In its second year, the PURA Foundation Australia has continued to strengthen its presence both locally and globally within the rare disease community. Through strategic partnerships, collaborative initiatives, and ongoing outreach, the Foundation has expanded awareness, provided support to families, and further developed our PURA syndrome network.

This year, the Foundation delivered several significant presentations. We partnered with the Genetic Support Network Victoria (GSNV) to raise awareness of PURA syndrome and share the experiences of affected families on Rare Disease Day. We also participated in Rare Disease Day at Parliament House Canberra, engaging with policymakers, advocates, and other rare disease organisations to highlight the challenges faced by families and the importance of research and advocacy.

On the international stage, our Chair and Head of Research attended and presented at the Global PURA Syndrome Conference in Dallas, Texas, USA. This provided an invaluable opportunity to connect with researchers, clinicians, and families worldwide, share insights from Australia, and contribute to advancing global knowledge of PURA syndrome.

The Foundation has also strengthened its parent support network, welcoming a new representative for Queensland. This growth ensures that families across Australia have local access to guidance, peer support, and connection with the broader PURA community.

Through these efforts, the PURA Foundation Australia is building a stronger, more connected community, driving research and advocacy, and ensuring that every individual and family affected by PURA syndrome has the support, resources, and hope they need for the future.

In its second year, the Parent Liaison role at the PURA Foundation Australia has continued to be both fulfilling and deeply rewarding. Building on the foundation established in the first year, the role has strengthened support across the PURA community, with additional Support Roles in Queensland and New Zealand.

Throughout the year, we have maintained a strong focus on keeping families informed and connected. Our quarterly newsletters have continued to provide timely updates on research developments, upcoming events, and practical resources, ensuring families feel supported and empowered in their daily lives.

The PURA Parents Australia and New Zealand online support group has grown steadily, serving as a vital platform for parents to share experiences, offer advice, and build meaningful connections. The group's activity continues to reinforce a sense of solidarity and a strong support network for families.

By fostering these connections and maintaining open communication, we hope families feel more supported, informed, and connected on their journey with PURA syndrome.

Rebecca Nunn

Secretary and Parent Support Victoria

Brisbane Marathon 2025

PURA FOUNDATION AUSTRALIA

Fundraising and Events

The PURA Foundation Australia has continued to organise impactful in-person events, bringing PURA families together while raising vital funds for research. Since our formation in October 2023, the Foundation has remained committed to making a meaningful difference in the lives of individuals and families affected by PURA syndrome, guided by our mission of education, information, research, and support.

Amy Zagato (Bowen)

Fundraising and Media

In October 2024, the Melbourne Marathon Festival provided an opportunity for families to unite. Team PURA welcomed 96 participants who ran, walked, or wheeled through Melbourne, finishing at the Melbourne Cricket Ground (MCG). In the lead-up to the event, the Foundation was featured in the Herald Sun, raising broader awareness of PURA syndrome and shining a light on the strength of our community. The marathon not only strengthened connections between families but also raised $15,288 to support our clinical communication research study and advance understanding of PURA syndrome.

The Brisbane Marathon, held in June 2025, brought together 52 participants for Team PURA. Queensland families, alongside interstate participants, came together for this event, raising $10,737 specifically for epilepsy research for PURA syndrome. In addition to these events, families have provided ongoing support through individual donations, community initiatives, and other contributions, further strengthening the Foundation's ability to fund research and provide resources for the PURA community.

These events and the continued generosity of families have contributed to the growth of the Foundation, strengthened our research initiatives, and brought us closer to identifying potential treatments for PURA syndrome. They demonstrate the power of community in supporting both scientific progress and the well-being of families affected by this rare condition.

Jaime Martin

Parent Support Queensland

Queensland families have continued to demonstrate strong commitment and support for the PURA Foundation throughout the year. A major highlight was their involvement in the Brisbane Marathon Festival, which brought together families, friends, and supporters to raise awareness and vital funds. Every dollar raised from this event will be directed towards advancing research into PURA syndrome. This remarkable achievement reflects not only the generosity of our community, but also the determination of Queensland families to contribute to meaningful progress in treatments and care.

As in other states, Queensland families are geographically dispersed, often separated by significant distances. Regular engagement through Facebook groups, online meetings, and newsletter updates enables families to stay informed, share experiences, and provide mutual encouragement.

The insights and experiences of Queensland families continue to shape the direction of the Foundation. By contributing to research opportunities and awareness campaigns, they ensure their voices are heard and their unique needs represented at a national level. This collaborative approach is central to our mission, strengthening the Foundation's capacity to advocate for all families and to work towards the best possible outcomes for children and adults living with PURA syndrome.

New Zealand Engagement

Kirsty Trueman

New Zealand Liaison

The PURA Foundation Australia has continued to strengthen its connections with Rare Disorders New Zealand (RDNZ) and the wider rare community.

In 2025, the Foundation was honoured to be invited to RDNZ's 25th Anniversary Awards Ceremony at Government House, Wellington. Hosted by the Governor-General of New Zealand, Dame Cindy Kiro, the event recognised the contributions of advocates and leaders, including RDNZ founder John Forman.

This occasion provided an opportunity for the Foundation to share the story of PURA syndrome and raise awareness of the challenges faced by families living with rare conditions in both Australia and New Zealand.

New Zealand PURA families have also had the exciting opportunity to contribute to epilepsy and communication research. The findings from this work will be invaluable in addressing some of the most pressing challenges for children with PURA syndrome. We extend our thanks to the researchers, funders, and community supporters who have made this possible.

The PURA Foundation Australia remains an active member of Rare Disorders New Zealand and is committed to continuing collaboration, advocacy, and awareness-raising efforts to support people living with rare conditions. We recognise the importance of building a strong and visible PURA community in New Zealand, ensuring that families feel connected and supported.

Looking ahead, the Foundation is eager to continue engaging in joint initiatives that amplify the voices of PURA families on both sides of the Tasman.

This year, our PURA family in New Zealand grew by one, bringing our community to a total of three families. While still small in number, we see this as an exciting foundation to build upon as we look to establish a stronger collective voice for PURA families across New Zealand. Each new connection brings not only comfort in knowing we are not alone, but also strengthens our ability to advocate for awareness, support, and inclusion in research and clinical initiatives.

Although we have not yet had the opportunity to gather in person, there is a shared enthusiasm to come together. We hope that the summer months ahead will create the right opportunity to meet, check in with one another, and begin planning for the year ahead. Creating space for families to connect, exchange experiences, and consider the path forward will be an important step in shaping our future activities.

A continued highlight for our New Zealand families has been the close relationship with the PURA Foundation Australia. Being welcomed into research initiatives and collaborative projects has been a major blessing, one that ensures New Zealand families remain part of the global PURA journey. We are grateful for this ongoing connection and support.

Te Tane Trinick

Parent Support New Zealand

Research

Professor Michael Hildebrand

Head of Research

The first stages of the PURA Foundation Australia strategic research plan developed with scientific and community stakeholders to promote research in PURA syndrome have been successfully implemented over the last year.

Despite PURA syndrome being first described over 10 years ago precision therapies are not yet available for our families. The first step towards precision therapies is to describe the full spectrum of disease symptoms. PURA syndrome is a rare neuro-developmental disorder caused by pathogenic variants in the PURA gene. PURA encodes the transcription factor and RNA-transport protein PURA, which is critical for multiple cellular functions.

Most disease causing changes in the PURA gene lead to the full spectrum of disease symptoms. We have partnered with the Epilepsy Research Centre at The University of Melbourne and The Austin Hospital to analyse these disease symptoms over time by enrolling patients with PURA syndrome in the comprehensive Developmental and Epileptic Encephalopathies Research Natural History Study (DEER NHS). This first-of-its-kind international study was launched this year with funding from the National Health and Medical Research Council. Since commencing in June, 17 new families have enrolled, contributing vital data which will support the development of clinical trials for precision therapies in PURA syndrome.

To this end we have already completed studies with reported findings that support PURA gene testing for families with severe and mild disease symptoms including developmental delay, epilepsy and speech disorder. We are expanding understanding of clinical features such as speech disorder by funding our partners at the Murdoch Children's Research Institute to complete a clinical speech and language study. An incredible 102 families with PURA syndrome are in contact for this research study. This is the first systematic study of speech disorder in PURA syndrome, a feature that has often been overlooked in prior clinical research studies. These studies are helping to elucidate the full spectrum of disease symptoms in PURA syndrome.

We are also preparing for translation of these clinical and genetic findings into testing of desperately needed new precision therapies. First, we are biobanking PURA syndrome saliva and blood samples at The University of Melbourne and The Austin Hospital. Second, we have funded the development and characterisation of a pre-clinical mouse model of PURA syndrome by our partners at The Florey Institute of Neuroscience and Mental Health, Monash Genome Modification Platform and Monash University.

Together, these studies offer great promise, but we must keep up the research momentum. We look forward to reporting new findings from the Speech and Communication Clinical Study, Developmental and Epileptic Encephalopathies Research Natural History Study and pre-clinical research in 2026 and beyond.

Research

PURA related papers

Hildebrand MS, Braden RO, Lauretta ML, Kaspi A, Leventer RJ, Anderson M, Goel H, Bahlo M, Scheffer IE, Amor DJ, Janowski R, Niessing D, Morgan AT. Inherited PURA pathogenic variant associated with a mild neurodevelopmental disorder. Neurol Genet. 2024 Aug 6;10(5):e200181. PMID: 39131487.

Kaspi A, Hildebrand MS, Jackson VE, Braden R, van Reyk O, … Fisher SE, Amor DJ, Scheffer IE, Bahlo M, Morgan AT. Genetic aetiologies for childhood speech disorder: novel pathways co-expressed during brain development. Mol Psychiatry. 2023 Apr;28(4):1647-1663. PMID: 36658335.

Research Partners

- The Austin Hospital, Austin Health, Heidelberg, Australia
- Epilepsy Research Centre, University of Melbourne, Austin Health, Heidelberg, Australia
- The Florey Institute of Neuroscience and Mental Health, Parkville, Australia
- Monash Genome Modification Platform, Clayton, Australia
- Murdoch Children's Research Institute (MCRI), Parkville, Australia
- Phenomics Australia, Canberra, ACT, Australia
- The University of Melbourne, Parkville, Australia

Investigating speech and language in PURA syndrome

Prof Angela Morgan

Speech Pathology Director, Genetics of Speech MCRI.

PURA syndrome is a rare genetic condition that causes developmental delays, intellectual disability, hypotonia, and significant speech and language challenges. Many individuals remain non-verbal, while receptive language skills are often better preserved, but communication difficulties remain a core concern.

In 2024–2025, the PURA Foundation Australia, in collaboration with the Translational Centre for Speech Disorders at the Murdoch Children's Research Institute (MCRI), successfully completed recruitment for a global study examining the speech and language abilities of individuals with PURA syndrome. A total of 102 individuals participated, providing vital data to improve understanding of the condition.

The study aims to characterise speech and language profiles, guide diagnosis and prognosis, inform educational and therapy planning, and develop outcome measures needed for future clinical trials. Given the severity of expressive language challenges, the research also highlights the importance of augmentative and alternative communication (AAC) strategies.

Over the next 12 months, the team will analyse the data and prepare manuscripts to share the findings, which are expected to support better clinical care and targeted interventions for individuals with PURA syndrome.

We sincerely thank the PURA Foundation Australia and participating families for their invaluable support, which makes this research possible.

Introducing Dr Miya St John Postdoctoral Researcher Murdoch Children's Research Institute

During 2023–2024, families generously raised funds to support a 0.2 FTE postdoctoral researcher position for a study to characterise the speech and language abilities of individuals with PURA. We were delighted to welcome Dr Miya St John as the lead researcher on our communication study.

A Postdoctoral Researcher and Speech Pathologist at the Murdoch Children's Research Institute, Miya also teaches in the Department of Audiology and Speech Pathology at the University of Melbourne.

The Translational Centre for Speech Disorders research team is part of the Speech and Language Group at the Murdoch Children's Research Institute (MCRI), led by Professor Angela Morgan. MCRI is the largest child health research institute in Australia and ranks among the top three research institutes worldwide for research quality and impact. The Speech and Language Group has a strong history of collaborating with syndrome and gene support organisations. We look forward to continuing our work with this dedicated team.

World Epilepsy Day 2025

On World Epilepsy Day 2025, the PURA Foundation Australia proudly participated in the Advancing Treatments for Severe Genetic Epilepsies Public Lecture at The Florey. This internationally recognised day raises awareness about epilepsy, its impact on individuals and families, and the urgent need for research into better treatments. For the PURA community, where epilepsy affects the majority of individuals, World Epilepsy Day provides an important platform to highlight our unique challenges, amplify our voice, and foster connections that can accelerate progress towards effective therapies.

The lecture featured an insightful discussion about severe childhood epilepsy and the hopeful way forward with medical research. It also highlighted opportunities for collaboration between families, clinicians, and researchers, emphasizing the value of community engagement in shaping future treatment strategies. By linking PURA syndrome to this broader epilepsy awareness campaign, we can ensure that the specific needs of our community are considered in the development of research agendas and clinical trials.

Our Chair, Mel Anderson, participated as a panel member, providing the rare disease community perspective and sharing insights into the daily realities and challenges faced by families living with PURA syndrome. The Foundation's Epilepsy Research Lead, Professor Chris Reid, also presented, outlining his vision for advancing pre-clinical programs that will create and validate therapeutic strategies for epilepsy and other neurological disorders. These contributions ensured that the voice of our community was included in broader discussions around epilepsy.

Participation in World Epilepsy Day events strengthens partnerships with researchers and clinicians, raises awareness of the needs of families affected by PURA syndrome, and reinforces the Foundation's role as a key advocate for the PURA community within the wider rare disease and neurological research landscape.

Looking ahead, the Foundation is committed to building on this connection with The Florey and the epilepsy research community, exploring further opportunities for collaboration to drive research forward. By leveraging the awareness generated on World Epilepsy Day, we aim to accelerate progress towards better treatments and improved outcomes for individuals living with PURA syndrome. It is a privilege to work alongside a world-renowned institute such as The Florey, whose expertise and dedication are shaping the future of epilepsy research.

Together, we can ensure that the voices of families are at the heart of research, driving meaningful change for everyone affected by PURA syndrome.

Advancing Research into Epilepsy in PURA Syndrome

Prof Chris Reid

Epilepsy and Neurodevelopment Research Area Lead, The Florey

Refractory epilepsy is one of the most challenging and devastating features of PURA syndrome, yet the reasons why it occurs remain unclear. This knowledge gap continues to limit the development of effective treatments. The goal of our research program is to uncover the underlying mechanisms that drive seizures in PURA syndrome, paving the way for targeted therapies.

In 2025, three major developments marked an exciting step forward. Our research team successfully engineered a PURA syndrome mouse model based on the most commonly reported PURA variant. This model is already showing key characteristics of PURA syndrome, including epilepsy, providing an invaluable tool for future studies.

We also welcomed Jenna White as a PhD student, who secured a highly competitive PhD scholarship to lead this work. Jenna has begun characterising the PURA syndrome mouse model and will focus on studying epilepsy through detailed approaches such as EEG monitoring and single-cell brain recordings.

In addition, the PURA Foundation Australia became the first organisation globally to fund research involving pharmaceutical compounds specifically for PURA syndrome, an important milestone toward identifying potential treatments.

Exciting times for PURA Foundation Australia!

Introducing Jenna White PhD Student Melbourne University

In 2025, we were delighted to welcome Jenna White as a PhD student at Melbourne University, working on the PURA epilepsy project in collaboration with the Florey Institute and supported by the PURA Foundation Australia.

Jenna was born in Singapore and spent her childhood across Asia before moving with her family to Auckland, New Zealand. Her passion for neuroscience was sparked in high school after an optional biology course on the brain ignited her curiosity. Since then, completing a PhD in neuroscience has been her long-term goal.

During her honours year, Jenna discovered the PURA epilepsy project and was inspired by its potential to make a meaningful difference for individuals and families living with PURA syndrome. She began her PhD position at the Florey Institute in March 2025, focusing on better understanding the causes of epilepsy in PURA syndrome using a newly developed PURA mouse model.

Jenna is excited to connect with the PURA community and is committed to contributing valuable insights that will advance both treatments and care.


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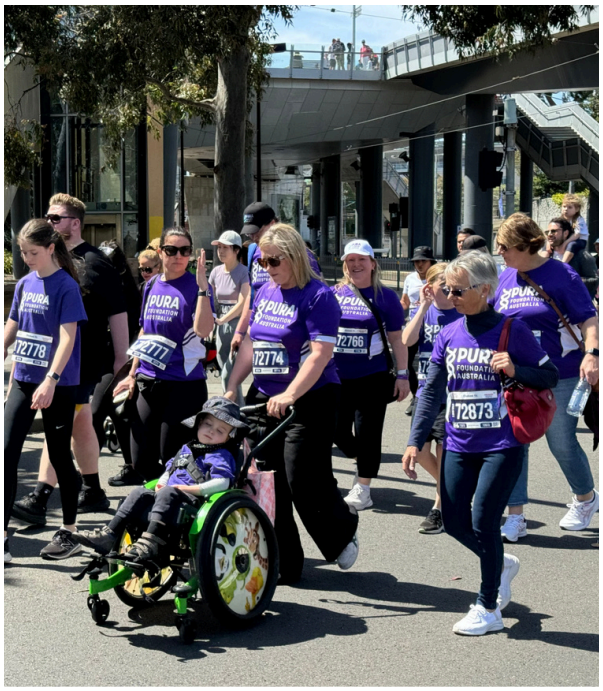
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Finance

Neil Anderson

Director
Finance and Marketing

In its second year, the PURA Foundation Australia has continued to build on the strong financial foundation established in our inaugural year. We have diversified our funding streams, securing a combination of corporate sponsorships, community-based fundraising initiatives, and individual donations. These efforts have enabled the Foundation to expand its research support, community programs, and advocacy initiatives while maintaining operational sustainability.

A highlight of this year has been the continued engagement of PURA families in fundraising activities. Families have played a central role, reaching out to workplaces, local communities, and networks to generate support. Notably, many families participated in the Brisbane and Melbourne Marathon events, helping raise both funds and awareness for PURA syndrome across Australia.

We have also seen growth in community and sponsor support, providing essential resources to fund our research grants, pilot studies, and community initiatives. These contributions have been crucial in ensuring that the Foundation can continue to support innovative research projects while expanding programs to assist families affected by PURA syndrome.

Thanks to these combined efforts, the PURA Foundation Australia has achieved both financial stability and flexibility, allowing us to respond to emerging opportunities and invest in initiatives with significant potential for the PURA community.

We extend our sincere gratitude to our sponsors, donors, volunteers, and families for their ongoing commitment. Their support ensures that the Foundation can continue to drive research, advocacy, and community engagement, making a meaningful difference for individuals and families living with PURA syndrome.

HAPPY 1ST BIRTHDAY PURA FOUNDATION AUSTRALIA

On October 1st, 2024, the PURA Foundation Australia proudly celebrated its first birthday. This milestone marked a year of progress in building awareness, supporting families, and laying the groundwork for vital research into PURA syndrome. It was an opportunity to reflect on how far we have come in such a short time and to thank the families, supporters, and partners who have walked beside us.

In just one year, the Foundation has grown into a strong and connected community. From establishing governance and fostering research collaborations to hosting family events and raising much-needed funds, every achievement has been driven by teamwork and shared purpose.

This first birthday was more than just a celebration; it was a reminder of what can be achieved when families and researchers unite.

With our community behind us, the PURA Foundation Australia is determined to continue building hope and creating change for the years ahead.

Financial Overview 24-25

Bank balance at 1st July 2024	$36,693.39
Income	
Donations (Community and Sponsorship)	$41,093.24
Expenditure	
Merchandise for Events	$3,859.30
Auditor and Accountancy	$3,734.50
Operating Expenses and Memberships	$3,868.41
Research Grant Funding	$44,075.26
Net Result	
Loss for financial year 24-25	-$14,444.23
Financial Position	
Balance as at 30th June 2025	$22,249.16

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http://linkedin.com.au/company/purafoundation

https://www.threads.com/@purafoundationau

How can you help?

Life changing knowledge and research is not possible without people like you.

Contribute towards the future of the PURA Foundation Australia and help us to assist PURA syndrome patients, and their families.

As an independent charity, we rely solely on the generosity of our supporters to help create a better future for those with PURA syndrome. Find out the ways you can help, such as a one-time gift, setting up regular donations or donating your time, by visiting our website or contacting us directly.

Our journey to help those with PURA syndrome is just beginning. Every dollar raised takes us another step closer to needed support, research, and treatments.

Please help us improve the lives of those with PURA syndrome.