PURA FOUNDATION AUSTRALIA

Annual **Report**

23-24

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, present and future. 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

About Us

The PURA Foundation Australia is the only non-profit charity that has been established for individuals with PURA syndrome and their families, across Australia and New Zealand. We assist the PURA syndrome community by supporting and initiating research, raising awareness, and providing education.

This community provides a place of belonging to those who may otherwise feel isolated by rare disease, enriches the medical research being completed and educates those outside the community about the condition.

When we all work together, we can achieve anything.

www.purafoundation.au

Our Values

Respect - We welcome and value every person's unique contribution with access and opportunity for all.

Community - We work together with families, clinicians, and scientists to define and achieve shared goals.

Optimism - We collaborate to seek and find solutions, confident in our team's ability to achieve our shared goals.

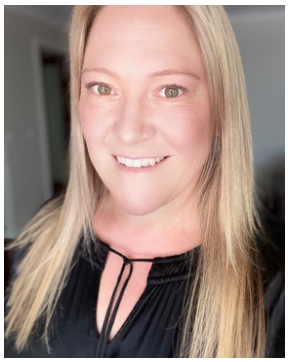
Responsibility - We understand the importance of obligation and accountability within the organisation, planning for future growth and change.

Trust - We understand the importance of transparency and communication within the organisation, and with those we serve and partner.

Our Goals

1. **To facilitate education and awareness of PURA syndrome.**
2. **To assist in the advancement of research and treatments for PURA syndrome.**
3. **To provide support for families of, and individuals with, PURA syndrome**

Governance



Melinda Anderson

Director - Chair
Research and Governance

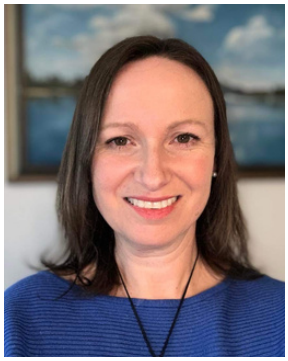
I am proud to share with you the PURA Foundation Australia's 23-24 Annual Report. The PURA Foundation Australia has had an outstanding first year, advancing our mission to support individuals affected by PURA syndrome and their families.

Effective governance is key to ensuring the Foundation's transparency, accountability, and sustainability. In our first year, we established a solid governance framework that provides clear leadership and direction, ensuring that all activities are aligned with the Foundation's mission and that resources are used responsibly.

The Board of Directors is composed of professionals with experience in management, research support, education, community outreach, and finance. Our Board works closely with our Executive team to plan for the Foundation's growth and development in the years ahead. Our outreach to families creates an environment where the voices of rare disease can be a driving force for change.

One of the Foundation's core objectives is to support and fund research into PURA syndrome. We have developed connections and started collaborations with scientific and clinical researchers who will help provide new knowledge for PURA syndrome therapeutic clinical trials and needed change for PURA patients and their families.

The first year of the PURA Foundation Australia has laid a strong foundation for future success. Our research initiatives are gaining momentum, our governance systems are in place, and we have built a supportive community of families and advocates. Moving forward, we are committed to funding our research efforts, strengthening our governance practices, and ensuring the long-term sustainability of the Foundation.



Kirsty Trueman

New Zealand Liaison

The launch of the Voice of Rare Disorders New Zealand White Papers was attended on behalf of the PURA Foundation Australia in February 2024. Two White Papers were presented in the Grand Hall Parliament: Impacts of Living with a Rare Disorder and Impact for Whānau Māori of Living with a Rare Disorder. These White Papers contained biennial survey data of people living with rare conditions, information that can be used to develop the National Rare Disorders Strategy which the government is presently drafting.

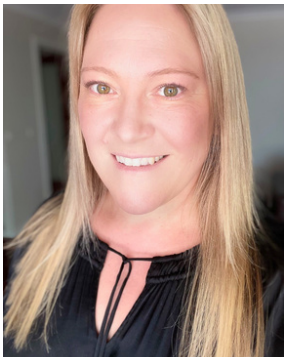
The PURA Foundation Australia was also invited to the Rare Disorders New Zealand Support Group Lead Hui in August. Main topics of discussion were the impacts to the disability sector with the change of government last year. Due to funding cuts and the reorganisation of government departments and agencies, the implementation of the Rare Disorders Strategy has in effect been put on hold. Rare Disorders New Zealand are working with the lead implementation agency Manatū Hauora on next steps to further the implementation of the Strategy. Planning is underway for Rare Disorder Month 2025 in which Rare Disorders New Zealand will celebrate their 25th anniversary.

We look forward to continuing to raise awareness and advocate for PURA syndrome in New Zealand.

Our Team

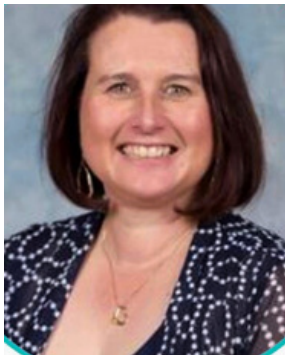
At the PURA Foundation Australia we are stronger because of our team. Every achievement, from fundraising and advancing research, to expanding our community, has been the result of collaborative effort. Teamwork allows us to support each other, stay resilient in the face of challenges, and build lasting connections within the PURA syndrome community. By working together, we not only help those with PURA syndrome but also create a supportive, thriving network that benefits everyone involved.

In the end, the power of a team is its ability to achieve more than any individual could alone. It's about building something bigger than ourselves, a community of support, advocacy, and hope for the individuals and families living with PURA syndrome. Through our teamwork, we are making a lasting difference.



Melinda Anderson

Director - Chair

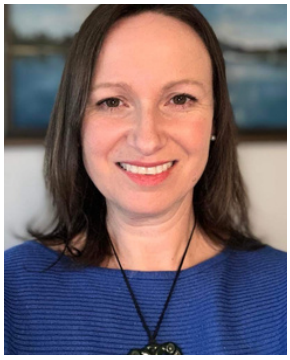


Natasha Keetley

Director

Neil Anderson

Director



Kirsty Truemann

New Zealand Liaison

Rebecca Nunn

Secretary
Parent Liaison

Amy Zagato (Bowen)

Fundraising and Events

Te Tane Trinick

Parent Support New Zealand

Research

The PURA Foundation Australia, with the input of scientific and community stakeholders, has developed a strategic research plan to promote research into PURA syndrome. It is hoped this will accelerate knowledge, create education materials, and lead to development of new treatment options for PURA.

PURA syndrome was first described in 2014 as a rare neuro-developmental disorder. The condition is 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. The specific functions of PURA protein are not well understood, nor how it is disrupted in individuals with PURA syndrome.

Through the support of new scientific research programs and development of PURA clinical studies, the PURA Foundation Australia will advance science with the aim to develop new treatment options for PURA, improving education and understanding of PURA syndrome in the community.

With the support of the PURA community, our research program will begin with funding small pilot studies. The results of these studies will guide the PURA Foundation Australia research agenda and provide a platform for researchers to obtain larger public or philanthropic grants.

Over the next 4 years, our foundation will aim to fund early-stage studies to better understand disease mechanisms and translational research programs focused on new therapies.

Foundation, families, clinicians and scientists as one community.

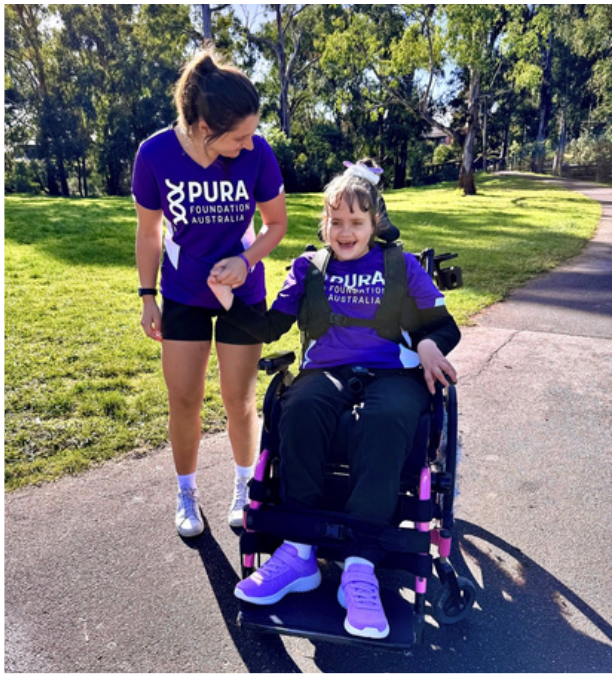
Research Affiliates:

The Florey Institute of Neuroscience and Mental Health, Melbourne Australia

Murdoch Children's Research Institute (MCRI), Melbourne Australia

The University of Melbourne, Melbourne Australia

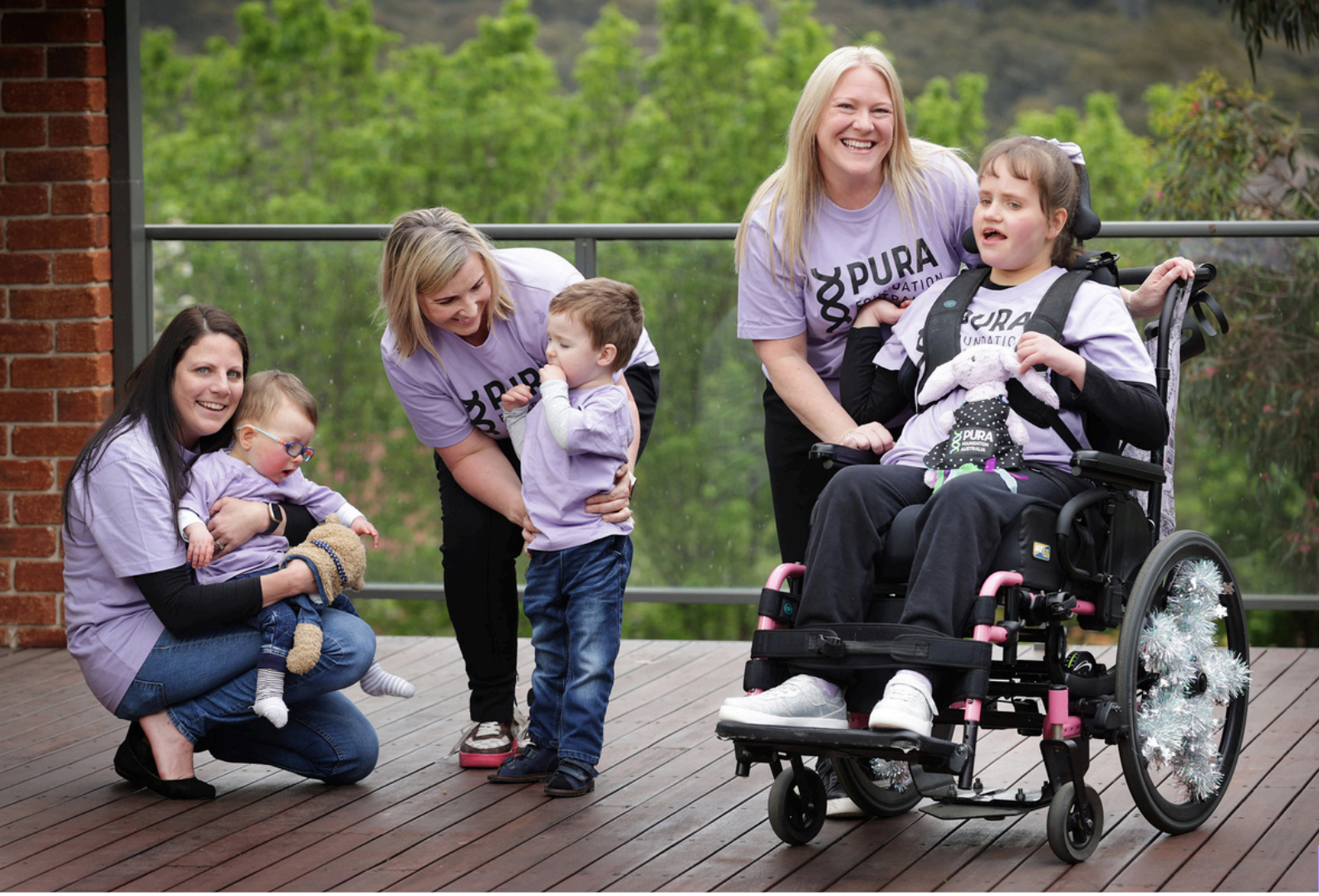
Monash Genome Modification Platform, Monash University

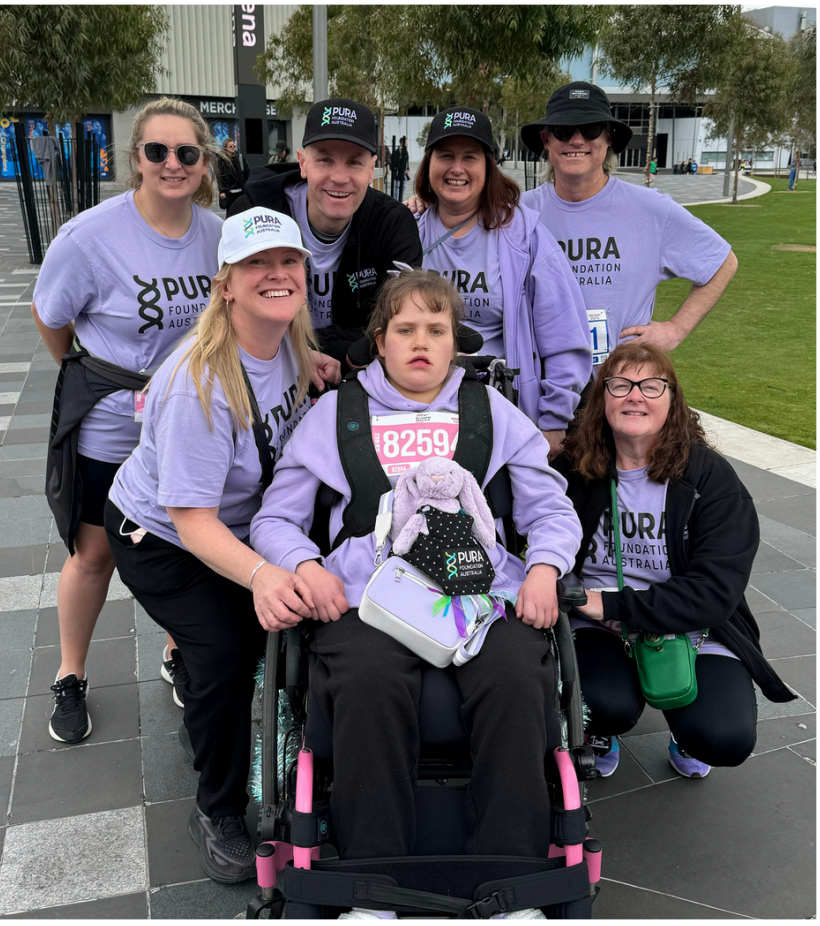


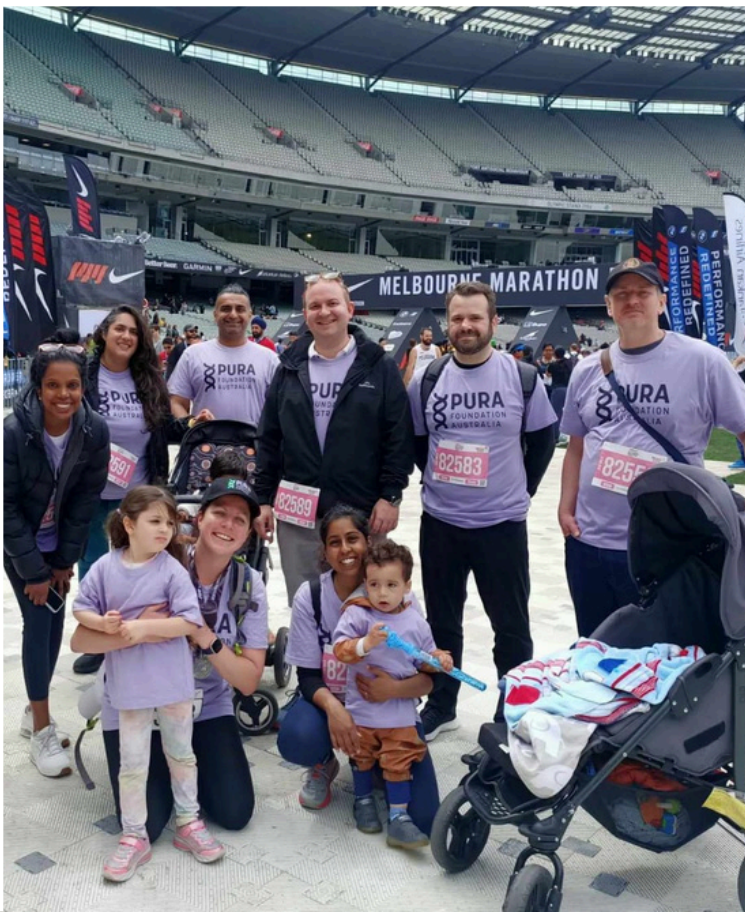
Clinical Research Publication Supported

Hildebrand et al 2024

Inherited PURA pathogenic variant associated with a mild neurodevelopmental disorder






MELBOURNE MARATHON

Research Plan

Natural History Study

With increased diagnostic testing comes additional and at times, milder cases of PURA syndrome. As such, the medical and parent community needs an updated overview of the condition. This clinical research aims to understand the full spectrum of PURA syndrome (such as hypotonia, speech difficulties, seizures, movement disorders etc.) to help guide research and provide updated education. Through our Natural History Study, we aim to understand common PURA syndrome clinical characteristics to determine possible treatment requirements, and develop education material for medical practitioners, allied health, and community members.

Lead: Professor Richard Leventer (MCRI)

Epilepsy

Translational research to identify therapies is an important aspect of research and aims to play a key role in improving the quality of life for PURA syndrome patients with epilepsy. PURA is a disorder where the epilepsy can be devastating. Once it starts, it can rarely be fully controlled. This program will support the development of pre-clinical studies to address the epileptic seizures seen in PURA syndrome. New drug development is long and costly. Repurposing of drugs can drastically reduce development time and skip many of the pre-clinical phases. We will leverage our limited resources to investigate repurposing processes by funding proof of concept epilepsy studies utilizing biological models and patient cell lines.

Lead: Professor Chris Reid (The Florey)

Communication

Speech is reportedly absent in most PURA patients, yet the linguistic phenotype has not been characterised. Speech and language abilities of PURA patients are rarely formally assessed, likely because speech is often documented as 'absent'. Unfortunately, the term 'absent speech' has not been further expanded upon to specify whether patients are truly non-verbal or minimally verbal, whether PURA patients have an ability to use alternative non-speech forms of communication such as sign language or communication devices, or whether their communication difficulties are due to severe speech motor or cognitive presentations. To better understand these phenotypes, we will increase genetic diagnostic yield by enhancing genome-wide testing methods.

Lead: Professor Angela Morgan (MCRI)

Biobank

A PURA biobank is required for better understanding of the causes, range of manifestations, and progression of this rare disease. Biological specimens are critical for genetic research, with ethics approval and protocols established to collect, analyse, and distribute biopsy specimens as appropriate. This often includes liaising with the participant's clinician and family to complete biopsy specimens at the point of surgery or sedation for other procedures. DNA will be extracted and stored from biopsied tissue to facilitate genetic testing. Cell lines will be generated from biopsied tissue to facilitate in vitro functional studies and testing of potential therapeutics.

Lead: Professor Michael Hildebrand (Univeristy of Melbourne)


PURA
FOUNDATION
AUSTRALIA


PURA
FOUNDATION
AUSTRALIA
NEIL ANDERSON
10192
HANALEI
10685
PIPER
10679
CAROLYN
10560

Fundraising and Events

PURA Foundation Australia organised impactful in person events to connect PURA families, whilst also raising funds for research in our first year.

Since our formation in October 2023, the PURA Foundation Australia has been steadfast in its commitment to making a meaningful impact on the lives of individuals and families affected by PURA syndrome. Our mission, rooted in education, information, research, and support, is the driving force behind everything we do.

In October 2023, the Melbourne Marathon Festival provided an opportunity to bring PURA families together for the first time in Australia. This inaugural fundraising event for the PURA Foundation Australia brought together a total of 66 members for Team PURA. Participants ran, walked, or wheeled through Melbourne Central Business District, finishing at the Melbourne Cricket Ground (MCG). This event aimed to raise funds to support clinical research for PURA syndrome, with fundraising efforts surpassing the initial target of $15000.

The Brisbane Marathon, held in June 2024, saw similar success with further funds raised for epilepsy research. This event provided Queensland families with the opportunity to come together and connect in person for the first time. Team PURA consisted of 70 members involved in various running and walking events. Team members were focused on reaching the fundraising target of $15000. This event also represented a milestone, as it was the first PURA event where families from interstate came together, demonstrating a united commitment to raising awareness and support for PURA syndrome.

These events have not only provided financial support for much needed research but have also helped to build community and visibility for PURA syndrome.

Amy Zagato (Bowen)

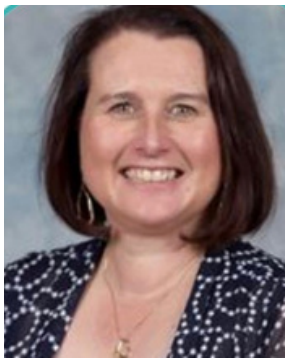
Fundraising

On February 29th the PURA Foundation Australia joined with other rare disease organisations across the globe in celebrating Rare Disease Day. It was a chance to raise awareness, share individual journeys and educate others about PURA syndrome.

For people with rare diseases, getting a correct diagnosis can take years and many visits to different clinicians. The lack of scientific knowledge and quality information on the disease often results in a delay in diagnosis.

Research needs to be international to ensure that experts, researchers, clinicians and Foundations are all connected and working as one. PURA Foundation Australia works with other global rare organisations to support research and education.

Community and Outreach



Natasha Keetley

Director
Community and Outreach

PURA Foundation Australia has made significant progress in expanding its reach and developing connections within the global and local rare disease communities. Through strategic partnerships and outreach efforts, the Foundation has been able to raise awareness, provide support, and strengthen the PURA community.

The Foundation has engaged with organisations in Australia and New Zealand that advocate for rare diseases. Within Australia we have become members of Rare Voices Australia, the national peak body for Australians living with rare disease. PURA Foundation Australia took part in the annual Rare Disease Day Parliamentary Event, hosted by Rare Voices Australia (RVA) at Parliament House in Canberra.

Insightful speeches highlighting achievements in the rare disease sector and the ongoing implementation of the Australian Government's National Strategic Action Plan for Rare Diseases were discussed. RVA's event covered important topics, such as Rare Disease Centres of Expertise and the launch of Australia's inaugural National Recommendations for Rare Disease Health Care. PURA Foundation Australia is committed to supporting RVA's advocacy, emphasising the need to improve outcomes for Australians navigating rare diseases.

Understanding the importance of finding the correct diagnosis, we have extended our outreach efforts to other rare disease organisations within Australia and New Zealand. This includes those focused on conditions with differential diagnoses for PURA as well as organisations within the Developmental and Epileptic Encephalopathy (DEE) community. Through these connections, we can educate the rare disease community about PURA syndrome and help to improve care and research opportunities within the broader community.

Rebecca Nunn

Secretary and Parent Liaison

The Parent Liaison role for the PURA Foundation Australia is both fulfilling and deeply rewarding. The main goals have involved fostering a sense of community and engagement, ensuring that families affected by PURA syndrome feel connected, informed, and supported throughout their journey.

A key focus of the work has been ensuring the timely and accurate dissemination of information to the PURA community. Through our quarterly PURA Foundation Australia newsletters, we've been able to highlight important updates, upcoming events, and offer resources for families seeking support. This helps keep families up to date and ensures that they are not navigating their challenges alone.

One of the most impactful initiatives for families has been the establishment of the secure online support group for parents of PURA patients in Australia and New Zealand. The PURA Parents Australia and New Zealand group serves as a vital space where parents can connect with one another. In this supportive and nurturing environment, families can share their experiences, insights, and advice, creating a sense of solidarity and strong foundational support network.

By fostering a sense of community and engagement, we hope to have helped families feel more connected and supported on their journey with PURA syndrome.

Finance

Neil Anderson

Director
Finance and Marketing

To ensure the financial feasibility and long-term sustainability of the Foundation, we focused on creating diverse funding avenues in our first year. Avenues included corporate sponsorships, community-based fundraising events and individual donations through family outreach efforts. These efforts have not only supported our day-to-day operations but also positioned the Foundation to provide research grants aimed at pilot studies related to PURA syndrome.

A notable success was the engagement of PURA Families in fundraising efforts. By encouraging families to reach out to their employer communities and extended networks, many took their first steps in organising donations on behalf of the Foundation. This grassroots involvement has been instrumental in building both financial support and awareness.

In our inaugural year, the Foundation successfully secured funding from six corporate sponsors for our Brisbane Marathon event. These partnerships have provided a crucial source of income, helping us to lay a solid financial foundation for future initiatives. We are deeply grateful to these companies for their support of our mission.

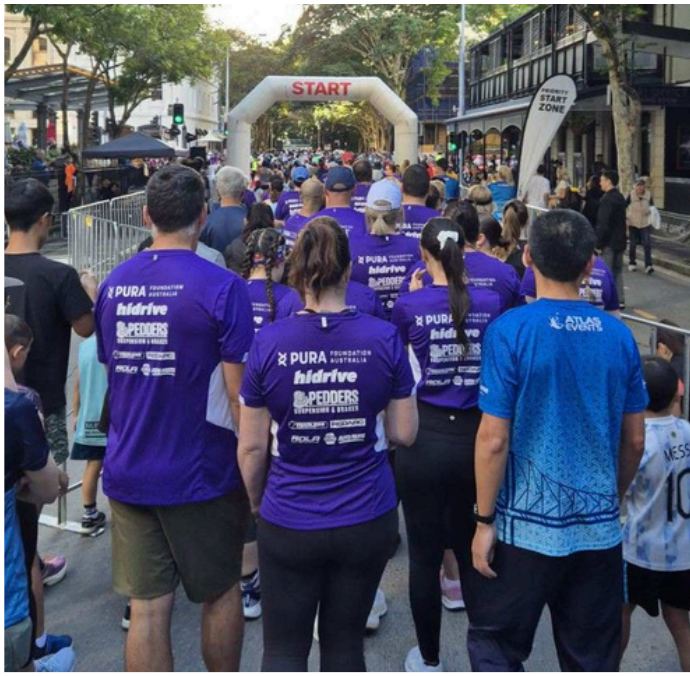
The Foundation has made remarkable progress in its first year, achieving financial stability, engaging with key stakeholders, and meeting its research goals. We are excited about the opportunities ahead and remain committed to expanding our reach, further diversifying funding sources, and supporting the families affected by PURA syndrome. As we move into our second year, we are confident that these early successes will serve as a springboard for continued growth and impact.

We would like to extend our heartfelt thanks to all our sponsors, donors, and volunteers for their unwavering support and commitment to our community.

Thank you

Sponsorship for events plays a crucial role in advancing research, raising awareness, and providing support to those affected by PURA syndrome. Rare diseases often have limited visibility, and without the backing of sponsors, holding events would be much more challenging. We would like to thank the following sponsors:

- **Hidrive**: Gold Sponsor
- **Pedders**: Gold Sponsor
- **Peerless**: Silver Sponsor
- **REDARC**: Silver Sponsor
- **Rola**: Silver Sponsor
- **NAPA Auto Parts**: Silver Sponsor
- **Jewell Projects**: Signage Sponsor



Financial Overview 23-24

Income	
Donations and Sponsorship	$43 434.13
Expenditure	
Cost of Sales (Merchandise)	$5 375.07
Operating Expenses and Membership	$1 365.67
Net Result	$36 693.39
Financial Position	
Balance as at 30th June 2024	$36 693.39

Foundation operation September 1st 2023
Launched to public October 1st 2023

Financial Overview from September 1st 2023 - June 30th 2024

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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.