Government of **Western Australia**
Child and Adolescent Health Service

Clinical Centre of Expertise for
Rare and Undiagnosed Diseases

Rare Care Centre

Year 3 Impact Report

February 2024-25

Contents

Commonly used abbreviations and acronyms

CNC	Clinical Nurse Consultant
ERDERA	European Rare Disease Research Alliance
FHRI	Future Health Research and Innovation (Fund)
GNNRD	Global Nursing Network for Rare Disease
IRDiRC	International Rare Diseases Research Consortium
PLWRUD	Persons living with rare and undiagnosed disease
CENTRE	Rare Care Centre
RD	Rare disease
RUD	Rare and undiagnosed disease
UDNI	Undiagnosed Diseases Network International

The Rare Care Centre acknowledges Aboriginal people as the First Peoples and the traditional custodians of the lands, sea, and waters across Western Australia where our work is undertaken. We acknowledge and pay our respect to the wisdom of Aboriginal Elders both past and present and pay respect to Aboriginal communities of today.

Using the term Aboriginal

Within Western Australia the term Aboriginal is used in preference to Aboriginal and Torres Strait Islander, in recognition that Aboriginal people are the original inhabitants of Western Australia. Aboriginal and Torres Strait Islander may be referred to in the national context and Indigenous may be referred to in the international context. No disrespect is intended to our Torres Strait Islander colleagues and community.

Using the term Rare and Undiagnosed Diseases

The terms ‘rare disease’ and ‘rare and undiagnosed disease’ are used interchangeably in this document for ease of reading. Unless a statement specifically refers to a characteristic of a known disease it should be considered that undiagnosed diseases are included.

Opening Letter

To our Esteemed Donors and Partners

Over three years, the Rare Care Centre (the Centre) has demonstrated significant growth, achieved remarkable milestones, and led the way globally in cross-sector care coordination. This progress is a testament to the dedication and expertise of our team, the unwavering support of our partners and funders, and most importantly, the courage and resilience of the children and families we serve.

This annual report showcases accomplishments and outlines aspirations. We are committed to tangible impact and ambitious goals. We are dedicated to supporting children and families living with rare and undiagnosed diseases, furthering our mission to meet their unmet needs so they can live the best lives possible. We are deeply grateful to the children and families who entrust us with their care, insights and vision for the future. We understand the challenges of navigating life with a rare or undiagnosed disease and remain steadfast in our commitment to developing and delivering a truly child and family-centred model of care.

Recognising the unique challenges faced by parents, we are developing a comprehensive 'Family Support Program.' Drawing inspiration from ‘gold standard’ programs internationally, we are tailoring a program to meet the needs of our WA families. Our Clinical Nurse Consultant, Anna Thetford, was awarded a prestigious Churchill Fellowship to explore and learn from these international programs. Anna will leverage these insights to implement our own impactful Family Support Program.

The Centre has been privileged to receive recognition for its work this year. A personal highlight was an invitation to attend the Rare Disease Roundtable with the Hon. Federal Health Minister Mark Butler, where I presented an overview of the challenges and complexities of rare and undiagnosed diseases and contributed to important discussions shaping future clinical and research directions. A further success indicator is the attraction of substantial funding from multiple and diverse sources. This recognition underscores the critical nature of our work and provides us with necessary resources to expand our services and reach. The Centre is deeply grateful to the WA Future Health Research and Innovation (FHRI) Fund for their generous support to develop and implement the CAHS Collaborative Centre for Research and Innovation Excellence for Rare and Undiagnosed Diseases. This Centre will serve as a pivotal hub, fostering collaboration with our partners and stakeholders across Australia and globally.

Our mission’s success is inextricably linked to strong collaborations. We have been actively working to scale our impact beyond the immediate needs of our current children and families. By sharing our knowledge, expertise, and best practices, we are benefitting children and families locally, nationally and globally. A prime example is our key partnership with the European Rare Diseases Research Alliance (ERDERA).

Looking ahead, we are excited about the future and are confident in our ability to continue to build upon our successes. A key 2025 focus is rare disease counting (coding) in the health system, and other sectors. This is so that people and families living with rare disease are not only counted, but truly count. Currently, the health system and providers are not tracking the biggest cost in our health system and the biggest killer of children – rare disease. This must change, for equity and the sustainability of healthcare, for the first 1000 days and beyond.

I want to express my deepest gratitude to our dedicated team, our invaluable partners, and the generous support of our funders, without whom this work is not possible. We hope this report provides valuable insights into our achievements and inspires continued collaboration and support in our mission to improve the lives of children and families living with rare and undiagnosed disease.

Sincerely,
Gareth Baynam
Medical Director

Welcome to the Rare Care Centre

This report reflects on another year of progress in our mission to improve the lives of children and their families living with rare and undiagnosed disease. It showcases the significant impact achieved through your valuable support.

8,000 rare diseases and increasing

300 Million people worldwide have a rare or undiagnosed disease, **2 million people in Australia**

6 out of 10 deaths in children are due to rare disease

5-7 years the average time to diagnosis

Only 5% of rare diseases have a prognosis altering treatment

70% of rare diseases are realised in childhood

80% of rare diseases are of genetic origin

The biggest cost in health care **1.5x** everything else combined

Why We Exist

Rare diseases massively impact the lives of an enormous number of children and families. In Western Australia alone, approximately 63,000 children and their families are affected.

The burden of rare and undiagnosed disease is substantial for children and families. Addressing the unmet need is a public health priority due to its impacts, including:

- **Health Burden:** Rare Diseases are often severe, chronic, and life-limiting, impacting multiple bodily systems and affecting all aspects of life.
- **Diagnostic, Treatment and Care Odysseys:** Children and their families often endure a long and arduous journey to diagnosis, facing delays, misdiagnosis, and a lack of readily available treatment, care and support options.
- **High Mortality:** Tragically, rare disease is the leading cause of death in children, with a mortality rate 13 times higher than common childhood diseases and is responsible for 6 in 10 deaths in children.
- **Significant Healthcare Costs:** Rare disease has a massive health economic burden, with healthcare costs estimated to be 1.5 times higher than the combined costs of all other diseases.

The Rare Care Centre was established to address these critical challenges. We envision a future where ***children with rare and undiagnosed disease and their families can live the best lives possible***. Our mission is to create innovative solutions for their unmet needs, navigating the complexities of the multiple government sectors and agencies, and coordinating care to ease the burden they carry.

Our Approach

At the heart of the Rare Care Centre lies a deep commitment to supporting children and families living with rare and undiagnosed disease on their unique journeys. We recognise the profound impact these conditions have on individuals and families, encompassing not only medical challenges but also significant mental, emotional, social, and economic burdens.

Across a family's journey with a rare or undiagnosed disease, they will ask many questions to try and find solutions, supports and answers. These questions and corresponding stages in the patient journey inform the pathways, programs, models of care, policies and system changes we have developed and advocate for. These questions across the patient journey are complex and multi-faceted. To address these questions effectively and meet the needs of families, our work is underpinned by six comprehensive pillars.

This report is structured around these six pillars. These pillars represent the essential workstreams that enable us to provide holistic and tangible outcomes for patients and families.

1. **Model of Care:** We are committed to further developing and delivering a world-class model of care that addresses the cross-sector and holistic needs of children and families living with rare and undiagnosed disease. This child and family-centred model focuses on:
    - Addressing unmet needs: Providing comprehensive support to address the physical, mental, emotional, financial and social challenges faced by children and families.
    - Early and accurate diagnosis: Guiding families through the diagnostic journey and connecting them with the necessary expertise.
    - Coordinated care: Facilitating seamless access to a cross-sector team of professionals, including medical, nursing, genetic counselling, mental health, disability, school education, welfare, social services and other non-governmental and disease specific supports.

2. **Awareness, Education and Training:** We strive to increase awareness of rare diseases among healthcare professionals, educators, other service providers and the broader community. Through targeted training programs and educational initiatives, we enhance the capacity of the healthcare workforce to recognise, diagnose, and manage rare diseases effectively.
3. **Research and Innovation:** We prioritise research and innovation to advance our understanding of rare diseases, develop new diagnostic tools and therapies, and improve patient outcomes for a better quality of life. We actively engage in collaborative research projects and support the translation and implementation of research into clinical practice. A 2025 priority is to deliver more rare disease clinical trials for children in WA and improve the clinical trials experience.
4. **Digital, Data and Devices:** We leverage technology to enhance patient care, improve access to information, and facilitate data-driven decision-making. This includes the development and utilisation of digital platforms, artificial intelligence, and large language models to support patient care, research, and education.
5. **Global Leadership and Partnerships:** We foster deep and meaningful collaborations with leading researchers, clinicians, patient advocacy organisations and industry worldwide. These partnerships facilitate knowledge exchange and dissemination, promote best practices, and advance the global understanding and management of rare diseases.
6. **Advocacy, Policy, and Legislation:** We actively engage in advocacy efforts to raise awareness of the challenges faced by individuals with rare diseases and to campaign for policies and regulatory and legislative change that improves access to diagnosis, treatment, and support services.

Underlying Principles

Our approach is guided by three core principles:

- **Equity of Access:** We are committed to ensuring that all individuals with rare diseases, regardless of their location, specific diagnosis, or lack of, or socioeconomic background, have equitable access to high-quality care and support services.
- **Scalability:** We strive to develop and implement sustainable solutions that can be scaled to meet the needs of the broader rare disease community, beyond the families we can see in person.
- **Sustainability:** We are committed to ensuring the long-term sustainability of our programs and services to continue providing support to the rare disease community for this and future generations.

> *"It has been heartening to watch our Rare Care Centre become a leader in the field of rare and undiagnosed diseases, playing a pivotal role in the establishment of an international nursing network and laying the foundations for a clinical centre of expertise that will have an enduring impact on helping improve the lives of children and families affected by these conditions."*
>
> **Valerie Buić**
> Chief Executive, Child and Adolescent Health Service

Year 3 Highlights

230
children and families supported by the clinical service

11
Student placements accommodated

$7.9m
successful grant applications contributed to, and partnered on

11
media campaigns about the Rare Care Centre

25
organisations interested in adopting the Centre's Model of Care

3
awards and nominations for the Centre staff and programs

$4.75
return on investment for every **$1** spent

31
presentations at conferences, seminars and academic meetings

26
children and families enrolled in the new Nurse Navigator Program

36
publications authored or co-authored by the Rare Care Centre team

608
members in the Global Nursing Network for Rare Diseases across **60 countries**

Recognition and Achievements

The Centre has gained significant recognition throughout the year, demonstrating increasing credibility and traction within the rare disease community. This is evidenced by extensive media coverage and a strong track record of successful grant applications.

Media Coverage

Media coverage plays a vital role in raising awareness about rare diseases, educating the general public, and reducing stigma associated with these conditions. Throughout the year, members of the Centre have actively engaged with the media to share our expertise and increase public understanding of rare diseases.

Professor Gareth Baynam was featured in The New Zealand Herald, ABC Science, 6PR Radio, and the Kids Research Institute Australia, raising public awareness of rare diseases, their impact on families and the importance of research. He was involved in a production of a Telethon video to showcase the impact of research on children with rare diseases, which generated vital support for the Centre's work. Rare Disease was the focus of the 2024 Channel 7 Telethon Ball. Contributions to international dissemination platforms like Orphanews and the International Rare Disease Research Consortium (IRDiRC), Medscape, Rare Revolution magazine and Podcasts have further enhanced the visibility of rare and undiagnosed disease and the Centre.

The Global Nursing Network for Rare Disease was introduced to the Council of International Neonatal Nurses via their online blog, further expanding the Network's reach and influence within the nursing community. LaunchR was promoted via newspapers and online media locally and also via the IRDiRC Media Channel internationally to increase awareness of this important initiative.

Grant Success

The Rare Care Centre has achieved significant success in securing competitive research and project funding totalling over $7.9 million across various projects. This funding success not only validates the Centre's research and program delivery priorities, but also provides crucial resources to advance its work across all pillars.

Research and Innovation: These grants enable the Centre to conduct cutting-edge research and innovation, explore novel therapeutic approaches, and contribute to a deeper understanding of rare diseases, directly informing and refining our model of care. Selected examples include:

- **CAHS Collaborative Centre for Research & Innovation Excellence for Rare & Undiagnosed Diseases:** $1.2M from the WA Health FHRI Fund
- **European Rare Disease Research Alliance (ERDERA):** $630M to European partners. Our Centre is the Australian Lead and responsible for forming the National Mirror Group (National Network into ERDERA)
- **Streamlining the development of antisense therapeutics:** Co-investigator, $5M
- **Research Project LINEAGE MRFF funded:** Co-applicant, $90K
- **Single cell CRISPR to Identify Pathogenesis with Transcriptomics (SCRIPT):** Co-investigator, $434K
- **Unravelling the Functional Significance of Novel Genetic Variants Identified in the Clinical Setting:** $139K
- **IRDiRC Stigma Taskforce (via Illumina):** Lead, $100K

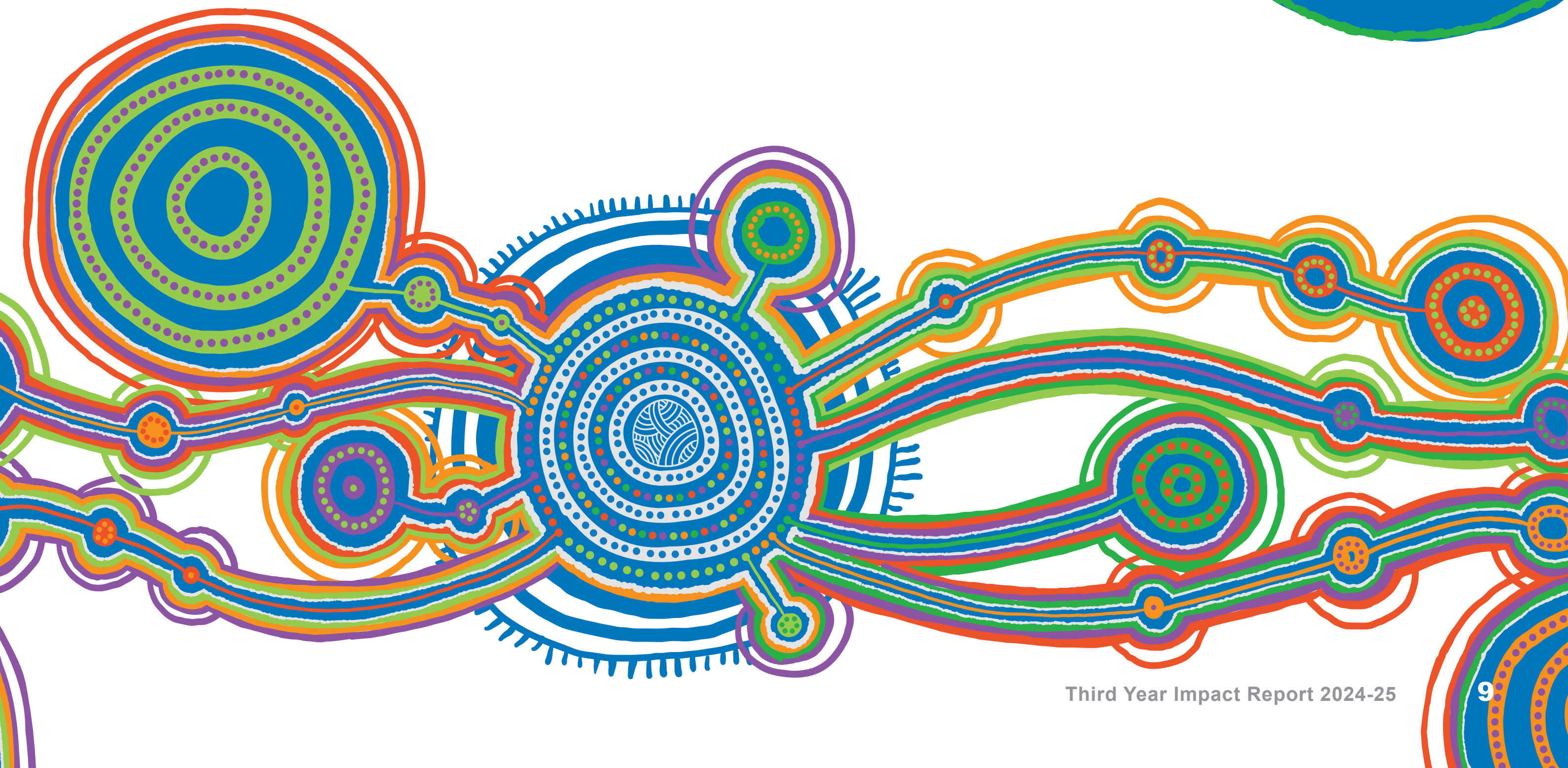
Data and Digital: These grants support the development and implementation of digital health platforms and tools to improve patient care, facilitate research, and enhance data-driven decision making.

- **Single cell CRISPR to Identify Pathogenesis with Transcriptomics (SCRIPT):** Research team member, $434K
- **Validating the feasibility in clinical workflows of the Medivox technology:** Research team member, $100K
- **Assessing the utility of AI-empowered personalised care plans for rare childhood diseases:** Lead, $100K

Global Nursing Network for Rare Disease: This recognition and successful grant acquisition underscores the acknowledgement of the significance of nursing in the care and support of people with rare disease both locally and internationally.

- **Nursing Education Program:** Stage two development of in-depth education modules: Lead, $93K
- **Asia-Pacific Group of GNNRD:** Regional needs assessment and group development workshop: Partner, $96K

Photo: Stephen Heath/PCHF



A Year of Impact

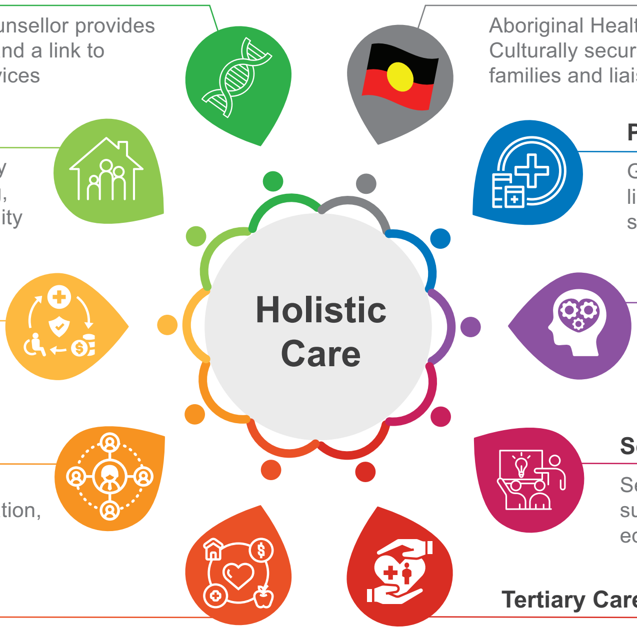
Pillar One: Model of Care

At the heart of the Centre lies a holistic, cross-sector model of care coordination designed to address the unique and multifaceted needs of children and families living with rare and undiagnosed diseases across all aspects of life – physical, mental, emotional, social and financial. This pillar is the core of what we do and why we exist. The work in all other pillars allows us to further develop and enhance our model of care so we can continuously improve care delivery for our patients and families living with rare diseases.

Over the last three years, we have developed, tested and expanded our model of care for children and families living with rare and undiagnosed diseases, demonstrating its effectiveness and significant benefits.

> *"I think it's the first time in 3 years that I have walked out of a meeting not crying due to the feeling that my requests for Skye will not be heard, I can honestly say I have never had so much help it was just such a great feeling and weight lifted off my shoulders."*
>
> Parent, Rare Care Clinic

Model of Care



Genetic Counselling
Genetics Counsellor provides counselling and a link to genetics services

Aboriginal Health
Aboriginal Health Practitioner provides Culturally secure care, support for Aboriginal families and liaison with relevant services

Dept. of Communities
DoC liaison provides family supports including housing, child protection and disability

Primary Care
General Practitioner provides a liaison role in primary care and supports upskilling of providers

National Disability Insurance Scheme
NDIS Coordinator navigates NDIS system and facilitates access to support services

Mental Health & Wellbeing
Mental health clinician provides psychosocial education, support and referrals to mental health services

Care Coordination
Paediatric nurse coordinators provide family support (education, information and resources)

School Education
Senior teacher navigates and supports within the school education system

Welfare
Assists families with access to financial and community supports

Tertiary Care
Consultant paediatrician provides medical coordination and case management

Improved Outcomes

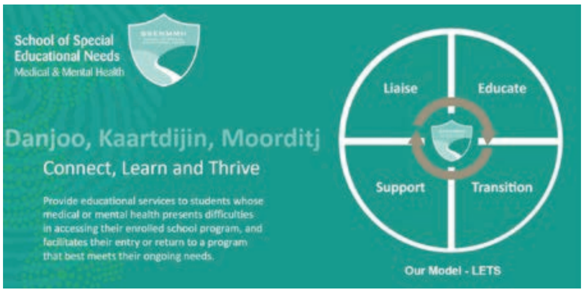
The Rare Care Centre has provided support to 256 children and families to date. A cost benefits analysis of the first 51 patients, 12 months post-discharge, revealed a significant return on investment (ROI) (social and economic) of $4.75 for every $1 spent, (at a 7% discount rate) surpassing the previous year's 4:1 ROI.

Financial savings to the health system of $13,400 per patient (over and above program cost) has been achieved through a 33% reduction in inpatient bed days and outpatient visits, and a 40% decrease in emergency department visits. This reduction in hospital visits also helps families through less travel burden, lost days from work and school, and reduced travel costs e.g. regional families travel cost is reduced by $5,200 per annum. Furthermore, we have demonstrated an average 9% reduction in parental stress, along with overall improvements in patient outcomes and reduced family burden. Examples of these include more school supports, increases in disability funding and welfare benefits, improved access to General Practitioners and care closer to home, and access to support and resources in the community. These results underscore the effectiveness of our model of care.

Expanding Our Reach and Impact

Building on this success, we have expanded our model so we can increase the Centre's reach to support more patients and families. We are pleased to provide you with an update on the significant milestones we have achieved in the past year:

- **School Supports:** The collaboration between the Department of Education's School of Special Education Needs: Medical and Mental Health (SSEN: MMH) and the Rare Care Centre exemplifies a holistic approach to supporting children with rare and undiagnosed diseases. Incorporating the SSEN: MMH vision and operational framework into the Rare Care Centre's model of care allows educational expertise to be embedded with the team. The integrated model facilitates continuous learning, minimises educational disruptions and addresses the unique challenges these children face, promoting their overall development and well-being. In 2024 this partnership ensured 111 affected children had continuity of education alongside typically developing peers with 80% attending an enrolled school within the month after this joint support.



Provide educational services to students whose medical or mental health presents difficulties in accessing their enrolled school program, and facilitates their entry or return to a program that best meets their ongoing needs.

- **Private Referrals:** We have broadened our referral pathways by accepting referrals from private paediatricians across Western Australia, increasing access to our services for children and families. To date, we have received 16 referrals from private practitioners.
- **Nurse Navigator Program:** In partnership with Rare Voices Australia (RVA), we launched a Nurse Navigator program offering nurse-led telehealth care coordination. This program provides a tailored level of support for families navigating rare disease care, acting as one of two national trial sites within RVA's Navigator Program. The program is for families whose needs are focused on nurse-led care coordination, and do not require the full cross-sector collaboration of our more comprehensive model. Our Nurse Navigator Program is one of two trial sites and works in conjunction with the RARE Helpline and other telehealth services offered by RVA. We have accepted 36 new children and families into this program.
- **Rare Care Pilbara Hub:** Work is well underway to establish the new Rare Care Pilbara Hub which will provide supports to children in remote regions living with rare diseases.

These children face compounding challenges due to distance, scarcity of health services and lack of wraparound supports. This hub will provide direct access to care providers, improved care coordination, and access to a hybrid model to allow for in-person and virtual appointments. The project is currently in co-design phase and the development of the model of care. We anticipate for the first patients to be welcomed into this new service by late 2025.

- **MindSpot Integration:** We have integrated MindSpot GP, a clinically validated online mental health support program, into our care pathways. This provides immediate access to mental health support for families, addressing the significant mental health challenges associated with living with a rare disease. We have partnered with MindSpot GP to upskill counselling staff on rare and undiagnosed disease to support more efficient and targeted resource use for enhanced child and family outcomes.
- **Patient Passport:** Building on the CamRare (Cambridge Rare Disease Network) initiative, the Centre has adapted and implemented the Patient Passport to empower families with a comprehensive summary of their child's medical and personal information. This tool, encompassing medical history, medications, appointments, and personalised care plans, facilitates seamless information sharing among care providers. Currently utilised by 22 families, the paper-based passport, chosen for its immediate impact and accessibility, has received overwhelmingly positive feedback. Parents consistently report that it simplifies communication with healthcare and support professionals, while children feel more confident in sharing their medical needs.

> "Thank you for the Arabic version of the Patient Passport for our daughter. It proved very useful when we had to call the ambulance for her. Thank you so much for your great help and support."
>
> Parent, Anonymous

A New Beginning: D's Rare Care Journey

When patient D and their mother arrived in Australia on a humanitarian visa, they were seeking safety and the opportunity to build a life closer to extended family. D's complex rare disease raised significant issues that affected their transition to Australia. D's rare disease affects their brain which impacts daily life, and along with multiple medical complexities including severe intellectual disability, delayed speech, impaired motor skills, seizures and behavioural challenges.

D and their mother were unfamiliar with the Australian healthcare system and struggling to access the support D desperately needed. At the time of referral D lacked access to crucial therapies, and documented assessments that are required to access school funding. D and their family required access to mental health and wellbeing supports with D's mother's inability to work and need to provide for D's high care needs.

Upon referral to the Rare Care Centre, the cross-sector team worked to holistically address their needs and achieve tangible outcomes to support D and their mother.

The Senior Genetic Counsellor provided crucial information about D's rare disease, using visual tools to help the extended family understand the condition, wider family planning and future pathways for D's siblings when they were ready to access more information. They addressed the mother's concerns about being a carrier, **offering compassionate support during a difficult time.**

The nursing team acted as a central point of contact, coordinating appointments with specialists, teaching D's mother how to use the 'Manage My Care' app, and advocating for D to be reprioritised on the Child Development Service waitlist. They **empowered D's mother to confidently navigate the healthcare system.**

The Centre's GP connected the family with a local doctor who had a special interest in rare diseases. This **GP was upskilled** in D's rare disease, providing them with a comprehensive visual summary of D's condition and care plan, and ongoing care in their own community closer to home.

The Senior School Teacher worked with D's school and the Education Department to secure the necessary funding and support, ensuring D **received the educational assistance they needed.**

The Mental Health Clinical Nurse Specialist connected D's mother with community services, offering much-needed **emotional support when she felt ready to access it**.

The NDIS Navigator found that D had not received any therapies since their arrival to Australia as they were ineligible for NDIS and were on long waitlists for public services. The NDIS Navigator was successful in securing Continuity of Support arrangements (CoSA) funding via the Local Area Coordinator at the Department of Communities, **enabling D to access therapies immediately**.

The impact of the Rare Care Centre's intervention was profound. D's mother expressed her heartfelt gratitude, saying, ***"My deepest gratitude for your tireless efforts in securing approval for my child's therapies. Your dedication and commitment to their well-being have been truly remarkable. I know that obtaining approval for these therapies can be a complex and challenging process, and I am incredibly grateful for the time, energy, and expertise you invested in advocating on my child's behalf. Your perseverance and unwavering support have made a world of difference in their life. Thank you again for everything. Your compassion and dedication have touched our lives in a profound way."***

Through the Rare Care Centre's holistic approach, D and their family were supported, understood, and equipped to navigate the challenges of D's rare disease. The Centre provided them with a new beginning, a foundation for a brighter future in their new home.

Pillar Two: Awareness, Education and Training

A cornerstone of the Rare Care Centre's mission is to raise awareness of rare disease, enhance professional knowledge, and build capacity within the healthcare workforce and other sectors. This is crucial for ensuring that individuals with rare disease receive timely and appropriate care, and that the Centre's expertise and learnings are effectively disseminated throughout healthcare and other systems.

While we directly support hundreds of children and families through our model of care at the Centre, we are also acutely aware that there are over 63,000 children in WA who have a rare or undiagnosed disease. Equipping the healthcare workforce and other related sectors and professionals with skills, knowledge and pathways is therefore paramount in the work we do. This scales up and scales out at a **local, national and global level**.

Locally

- **Aboriginal Health Council Western Australia (AHCWA) e-learning modules:** Developing and delivering online learning modules to enhance the knowledge and skills of Aboriginal Health Workers within Western Australia.
- **Lunch and Learn:** Sian Gannon, our Nurse Educator, presented at numerous educational sessions, including:
  - WACHM Education ‘Lunch & Learn’
  - Midwifery Complex Care Study Day
  - Curtin50 Years of Nursing conference
  - PCH Staff Development Nurse Inservice
  - WA ALIGN Symposium and Workshop, reaching over 170 healthcare professionals.
- **RACGP “Check” Case Studies:** Contributing to the development of case studies for the Royal Australian College of General Practitioners (RACGP) "Check" program to improve GP knowledge and awareness of rare diseases.
- **mEDUrare:** Through a collaboration with Syneos Health Communications, we are developing an educational animation and resource to illuminate the school experiences of children with rare diseases. This initiative aims to equip teachers, educators and peers with the tools and understanding needed to provide crucial support and foster inclusive learning environments.
- **Academic partnerships:** Our Centre builds workforce capacity and capability through fostering a vibrant learning environment. In the last year, we supported 11 students and two Aboriginal cadets (medical student, education student) to complete their service learning and internships, equipping the next generation of clinicians and educators. We also welcomed eight computer science Curtin university students to undertake learning projects with the Centre in partnership with KKH hospital in SingHealth demonstrating a strong interest in rare diseases and a commitment to improving care for individuals with these conditions, bringing our total to 37 students over the last 3 years. Our clinical team has supported 72 TAFE enrolled nursing students to gain valuable clinical experience within our weekly clinics.

> “My decision to join Rare Care stems from my aspiration to enhance diagnostic acuity using technology and improving my clinical-reasoning skills, all while contributing to a meaningful cause. Rare Care's mission aligns strongly with my values and interests, and I am eager to make a positive contribution to your work.”
>
> Joash Foo, Medical Student UWA, 2024

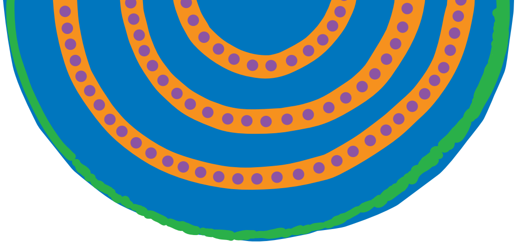
Nationally

- **Project ECHO:** Facilitating National ECHO sessions, online communities of practice that connect healthcare professionals across Australia to share knowledge and expertise in rare disease management. The Centre's Project ECHO sessions are already demonstrating tangible impact. For example, the RARE Voices Australia Advocacy Manager reported the following anecdote: ***"I have had a call to the RARE Helpline and the person heard about it from their GP in WA who had attended a Project ECHO. After this person explained some of the difficulties they were experiencing, he referred them immediately to the Helpline."*** This demonstrates how the Centre's educational initiatives are empowering healthcare professionals to better recognise and support individuals living with rare diseases.
- **National Recommendations for Rare Disease Healthcare:** Contributing to the development and dissemination of national recommendations for the diagnosis and management of rare diseases.
- **Undiagnosed Disease Clinical Framework:** Recognising the challenges faced by families of children with undiagnosed conditions, the Centre has commenced work on developing a dedicated clinical framework. This framework will provide guidance to healthcare professionals on identifying potential causes of undiagnosed disease, including recognizing parental concerns and incorporating these into the assessment process. It will also outline pathways for appropriate referral, care, and support services. This framework will serve as a valuable resource for healthcare professionals across the state. A global version is being progressed through our role in the Lancet Commission on Rare Disease.

Globally

- **Global Nursing Network for Rare Diseases – Education Program:** Leading the development of an online education program for novice and beginner nurses and midwives to upskill them in rare and undiagnosed disease knowledge and care.
- **UEMS – European Union RD Competency Program in Rare and Undiagnosed Disease:** Collaborating with the European Union of Medical Specialists (UEMS) to develop and implement specialty competency programs for healthcare professionals. Content and exam case studies have been developed, and mock exams have been held. The competency is formally recognised in the EU and USA and available to anyone globally.
- **Medics for Rare Disease (M4RD) 101 e-learning modules:** Contributing to the development and dissemination of M4RD 101 e-learning modules to enhance global awareness and understanding of rare diseases and partnering to create a 101 specifically for nurses.
- **Rare Disease Day Social Media Campaign:** The Rare Care Centre has been approached by Rare Revolution Magazine to partake in a takeover of their social media for Rare Disease Day. This opportunity will allow the Centre to further increase public awareness about rare diseases and the impact they have on people.

The Centre is committed to ongoing education and capacity building to ensure that individuals with rare diseases receive the highest quality of care and that the Centre's expertise continues to benefit the broader healthcare community.



Pillar Three: Research and Innovation

The Centre is establishing a **Collaborative Centre for Research and Innovation Excellence in Rare Diseases**. Our progress in this space allows us to continuously improve and refine our Model of Care. We are committed to driving advancements in our understanding of these complex conditions, developing new diagnostic paradigms and therapeutic approaches, and ultimately improving the lives of children and families affected by rare and undiagnosed diseases.

In 2024, Centre Medical Director, Gareth Baynam was ranked as the number 2 practicing rare disease researcher in the world, with his research mentor being number one. This commitment to research excellence is further evidenced by Gareth's recent selection for the International Research Awards on Network Science and Graph Analytics, under the category of Best Researcher Award. These prestigious recognitions highlight the Centre's dedication to cutting-edge research and its significant contributions to the field.

Collaborative Centre for Research and Innovation Excellence

The Rare Care Centre has made significant strides in advancing research and innovation in the field of rare diseases. A key achievement this year was that we successfully secured a $1.2M grant from the **WA Health Future Health Research and Innovation Fund** to establish the CAHS Collaborative Centre for Research & Innovation Excellence for Rare & Undiagnosed Diseases. This will serve as a pivotal hub for research and innovation, fostering collaboration and driving advancements in the field.

Key objectives include:

- Driving and supporting cross-sector, multi-disciplinary, culturally safe and responsive collaborative research and innovation for RUDs.
- Facilitating knowledge translation to ensure research findings are effectively implemented into improved clinical practice and inform the evolution of our model of care.
- Providing opportunities for workforce development, building the capacity of individuals and teams in research and innovation in RUDs.
- Positioning WA as a leader in the research and care for RUDs, creating an academic and practice hub for excellence and attracting world-renowned experts.
- Globally connecting rare disease innovators and innovations.

The Centre has developed a **Research, Innovation and Practice Development Framework** to guide the development of the Collaborative Centre for Research and Innovation including setting research program goals.
The significance of this work lies in its potential to:

- **Develop the science for better awareness, diagnosis and care:** Advance our understanding of rare diseases, leading to improved cross-sector diagnostic paradigms, therapeutic approaches, and ultimately, better health outcomes for children and families.
- **Reduce the burden of RUDs:** Diminish the impact of rare diseases on individuals, families, and the healthcare system by improving quality of life, reducing morbidity and mortality, mitigating the economic burden and stimulating new productivity and industry.
- **Lead and partner globally:** Position WA as a global leader in rare disease research and innovation, attracting international attention, expertise and funding.

Advancement of the Centre's Clinical Trials Program, TrialR

The clinical trial space for Rare Diseases within Western Australia has immense potential and the year has been instrumental in removing barriers for future trial success, creating key partnerships and establishing research pathways for emerging treatments.

Some of the highlights in this area have included:

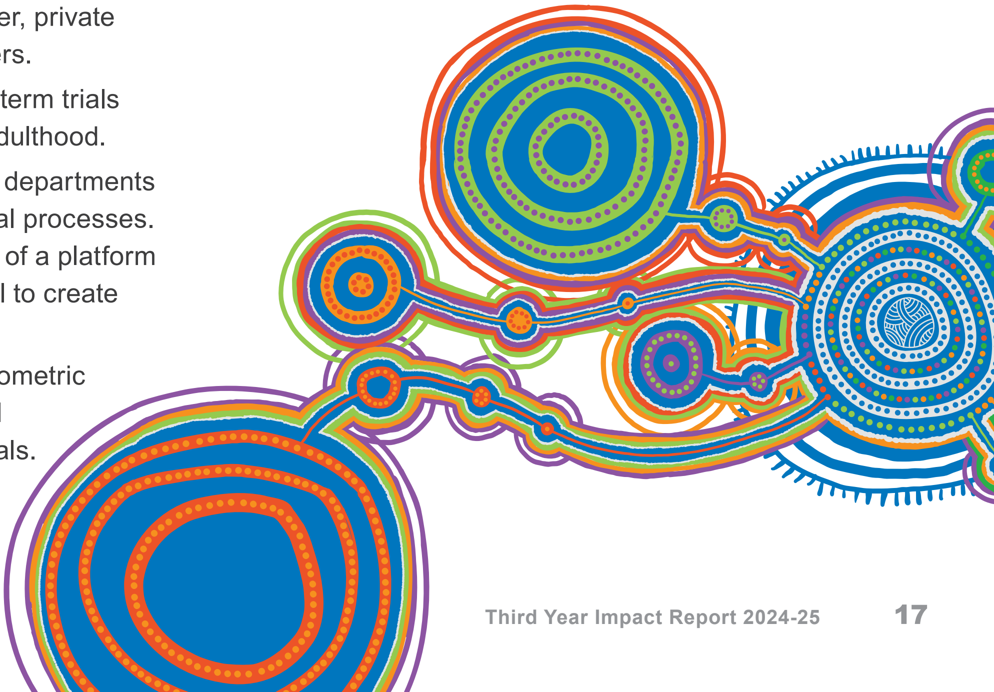
- Key collaborative partnerships with many pharmaceutical manufacturers both on a national and international level.
- Joining national and international research collaboratives to understand barriers to trials in rare diseases.
- Partnering with the Rare Disease Moonshot in the European Union and leading the Rare Disease Moonshot Australia, stimulating private-public partnerships to advance trial access.
- The commencement of infrastructure development for current and potential trials with in-hospital support, clinician engagement and access to neurocognitive assessments.
- Co-design with children and families, including families that have funded their own drug developments and clinical trials initiatives.
- The integration of current clinical trials into the Rare Care model where possible.
- Stimulated through TrialR, formation of a Rare Disease Trials Advisory Committee with multi-institutional, multi-stakeholder, private and public and multi-state members.
- Establishing connections for long term trials beyond paediatric care and into adulthood.
- Engaging the support of research departments within WA Health to assist with trial processes. For example, the commencement of a platform for clinical trials within the hospital to create clear pathways for rare diseases.
- Capacity building for better psychometric assessment capability and mental health support for rare disease trials.
- Leading a publication and international partnership on diversity, equity and inclusion for rare disease trials.

Looking ahead, the upcoming year presents a significant opportunity to initiate multiple rare disease trials. The robust infrastructure developed this year will position Western Australia as a preferred destination for families, pharmaceutical companies, and research collaborators seeking to conduct future trials.

Partnerships and Research Initiatives

We have already partnered and been engaged in research initiatives including 6 invitations to partner in multinational research and have been an author or co-author in 37 submissions and publications related to rare and undiagnosed diseases. Some research highlights include our work in:

- **Streamlining the development of antisense therapeutics for Western Australian children with rare genetic disorders:** Co-investigator, $5M
- **Research Project LINEAGE MRFF funded:** Co-applicant, $90K
- **Single cell CRISPR to Identify Pathogenesis with Transcriptomics (SCRIPT):** Co-investigator, $434K
- **Unravelling the Functional Significance of Novel Genetic Variants Identified in the Clinical Setting:** $139K



Cross-Sector Collaboration: The Deloitte Innovation Lab

The Deloitte Innovation Lab was held in July 2024, which brought together over 30 key stakeholders across public sector agencies and the broader business, philanthropic and lived experience community to identify key action areas for advancing rare disease innovation and care:

1. **Defining a dedicated working group** to plan and implement key actions and opportunities arising from the Innovation Lab.
2. **Connecting existing data** from the Department of Health and integrating data from other agencies where possible to enhance data sharing and collaboration to provide support and services for rare disease management.
3. **Progressing discussions with key senior stakeholders** to determine more formalised and tailored cross-sector (interagency/intergovernmental) governance and accountability structures for the Centre's work.
4. **Defining a plan to determine the optimal location and supporting structure** for the Centre within the wider system.
5. **Evaluating the shift from an activity-based funding model** to a recurrent or needs-based model, gathering necessary data and insights to support this transition.
6. **Determining funding and pathways** to ensure the right international experts (e.g. Medical Research Directors) can relocate to Perth to accelerate and scale the Centre's efforts.

Research and Innovation Collaboration on an International Scale

The Centre actively engages in international collaborations to advance the field of rare disease research and improve outcomes for children and families globally. Recognising the interconnected nature of research and the importance of sharing knowledge and expertise on a global scale, the Centre actively participates in several key international initiatives.

- **European Rare Diseases Research Alliance (ERDERA):** The Centre is honoured to partner as the Australian lead in the European Rare Diseases Research Alliance (ERDERA), a $630 Million 7-year project. ERDERA brings together over 170 organisations from 37 countries (in the EU and beyond), including research institutes, hospitals, universities, pharmaceutical companies, and patient organizations, with the shared goal of advancing rare disease research.

  The Centre’s role is to create a National Mirror Group, a network of rare disease researchers throughout Australia that link into ERDERA. This partnership provides us the unique opportunity to access cutting-edge research and expertise, strengthen international collaboration, create greater opportunities for our patients to participate in clinical trials and more.
- **International Rare Disease Research Consortium (IRDiRC):** IRDiRC is a global multi-stakeholder consortium to harmonise and accelerate rare disease research that represents the coordination of more than $3 Billion p.a. committed from its members into rare disease research. IRDiRC creates global taskforces to address critical unmet need. Centre Program Director, Sue Baker, is a member of the taskforce to develop a framework to assess impacts associated with diagnosis, treatment, support, and community integration that can capture changes along the rare disease patient and family journey. Centre Digital Lead, Tudor Groza is a member Functional Analysis, co-chaired by Gareth Baynam.

- **Global Alliance for Genomics and Health (GA4GH):** GA4GH supports advancing genomic data sharing for improved human health. Tudor Groza is Co-Chair of the Rare Disease Group.

We have also supported Queen's University, Belfast on their grant application for the Government of Ireland's North-South Research Programme to establish an All-Ireland Rare Disease Interdisciplinary Research and Innovation Hub to drive cross-border collaboration, innovation, research and education. We did the same for colleagues in Wales. This year the Centre Medical Director also accepted invitations to the Advisory Boards for the African Rare Disease Initiative (ARDI) and the Turkish Rare Disease Initiative (RareBoost).

By actively engaging in these international collaborations, we contribute to global efforts in rare care research. We are positioning ourselves as a global enabler and leader in rare disease research and innovation, contributing to the advancement of knowledge thus improving outcomes for children and families worldwide.

> "I wanted to extend my thanks to you and your team for the outstanding organisation of the Asia-Pacific regional leaders meeting. The event was a great success and served as an invaluable platform for sharing knowledge, experiences, and best practices in the field of rare diseases. The workshop not only provided us with a deeper understanding of the challenges and opportunities in this area but also fostered a strong sense of community among the participants. The insights and connections gained will undoubtedly have a lasting impact on our work and the broader mission of improving care for patients with rare diseases across the region."
>
> Ms Herath Mudiyanselage Chandrani Menike Herath, Faculty of Nursing, University of Colombo, Sri Lanka

Photo: Stephen Heath/PCHF

Pillar Four: Digital, Data and Devices

The Centre leverages the transformative power of digital technologies to enhance the lives of children with rare diseases and their families. This pillar focuses on developing and deploying innovative solutions that create value for families, service providers and systems.

Empowering Families and Improving Access to Information

Lyfe Languages: The Centre partners with this groundbreaking initiative to address a critical gap in healthcare communication by translating complex medical terminologies into Indigenous languages. Led by young Indigenous Language Champions, Lyfe Languages ensures culturally appropriate and respectful communication between healthcare providers and Indigenous families, fostering trust and improving health outcomes.

The Lyfe language platform currently has 8 contributing language champion users with a total of 770 translations utilising the web application many relating to Rare Disease. Lyfe languages has also created numerous animation videos to educate patients and families about their disease. Examples include *“Rare Mobs: Journey to a rare disease diagnosis”* and *“Songlines: Aboriginal and Torres Strait Islander Genetic Information”* in English audio and several aboriginal languages. Lyfe languages has created five key message posters. 2025 will see a focus on building translation data.

Advancing Precision Medicine and Accelerating Research

UTOPIA (Unlocking Treatment Options, Personalised In-Time Access): This cutting-edge digital solution utilises advanced artificial intelligence and machine learning techniques to analyse accessible patient data and identify unique "phenotype trajectories" for rare diseases. By understanding how phenotypes (diseases and disease features) evolve over time, UTOPIA aims to:

- **Enable earlier disease detection:** Identify subtle signs and symptoms that may indicate the onset of a rare disease, allowing for earlier interventions and potentially preventing disease progression.
- **Personalised care plans:** Tailor treatment strategies to the individual needs of each child based on their unique disease trajectory and response to interventions.
- **Improve disease management:** Develop more effective strategies for managing long-term health conditions and improving overall quality of life for children with rare diseases.

The Global Commission conducted a case study showing that UTOPIA has halved the time for clinicians to develop a personalised care plan by reducing the number of hours clinicians otherwise would have to spend finding and synthesising relevant information, tailoring it to the individual patient and presenting it in a format that is easy to understand. It is anticipated that UTOPIA will soon be able to create these care plans 10 times faster through replacing manual processes with technology-driven semi-automated systems.

LaunchR: This innovative partnership with multiple partners, existing and new programs at Murdoch University, the Perron Institute, Australian Kids Research Institute and international partners across 4 continents fosters a collaborative ecosystem that accelerates the translation of research discoveries into new therapies for children with rare diseases. LaunchR connects researchers, clinicians, industry partners, and patient advocates to:

- **Streamline drug discovery and development:** Facilitate access to patient data, biospecimens, and clinical trial expertise.
- **Improve access to clinical trials:** Connect eligible children with rare diseases to promising clinical trials, offering them access to potentially life-changing therapies.
- **Enhance care coordination:** Ensure seamless transitions between research, clinical care, and community support services.

Driving Innovation and Global Impact

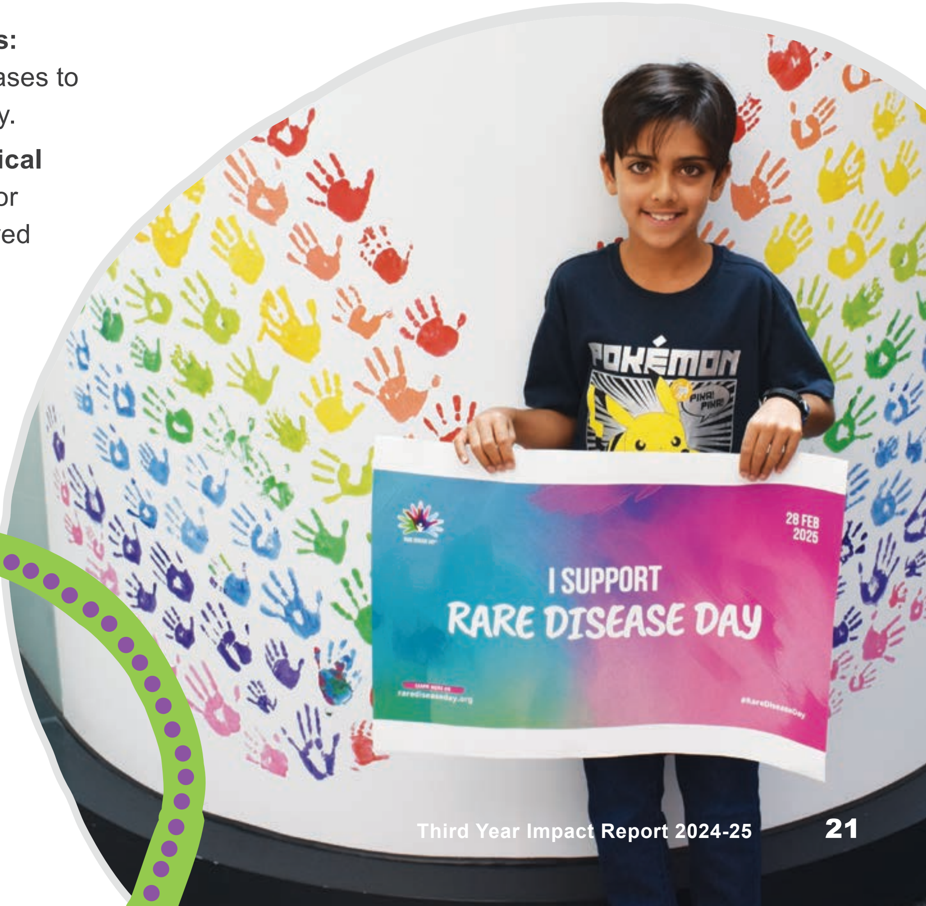
TrialR: This groundbreaking network facilitates access to clinical trials and novel therapies for children with rare disease. By leveraging digital platforms and data-driven approaches, TrialR aims to:

- **Increase clinical trial enrolment rates:** Connect eligible children with rare diseases to appropriate clinical trials more efficiently.
- **Improve geographical access to clinical trials:** Expand access to clinical trials for children living in remote and under-served communities.
- **Accelerate the development and delivery of new therapies:** Facilitate the rapid translation of research findings into real-world clinical practice.

Moonshot Australia: As the lead of this ambitious initiative, the Centre is driving a national effort to accelerate the development of innovative therapies for rare diseases by fostering strong public-private partnerships and leveraging international collaborations. Moonshot Australia aims to:

- **Advance the frontiers of rare disease research:** Support cutting-edge research in areas such as gene therapy, gene editing, and personalised medicine.
- **Improve the lives of children with rare diseases:** Deliver transformative therapies that address unmet medical needs and improve health outcomes.

By embracing the power of digital technologies and data-driven approaches, the Centre is transforming the landscape of rare disease care. Innovative initiatives like the Patient Passport, Lyfe Languages, UTOPIA, LaunchR, TrialR, and Moonshot Australia, the Centre is empowering families, accelerating research and clinical trials, and delivering precision medicine solutions that improve the lives of children living with rare disease.



Pillar Five: Global Leadership and Partnerships

At the Centre, we understand that tackling the complexities of rare diseases requires a global, collaborative approach. Our intentional and genuine partnerships allow us to share knowledge and expedite our understanding of rare diseases.

Expanding our Global Reach

We continue to expand our global partnerships with over 350 partnerships across the globe. By fostering a collaborative environment, we leverage collective expertise and resources to maximise our impact. This collaborative approach offers numerous benefits:

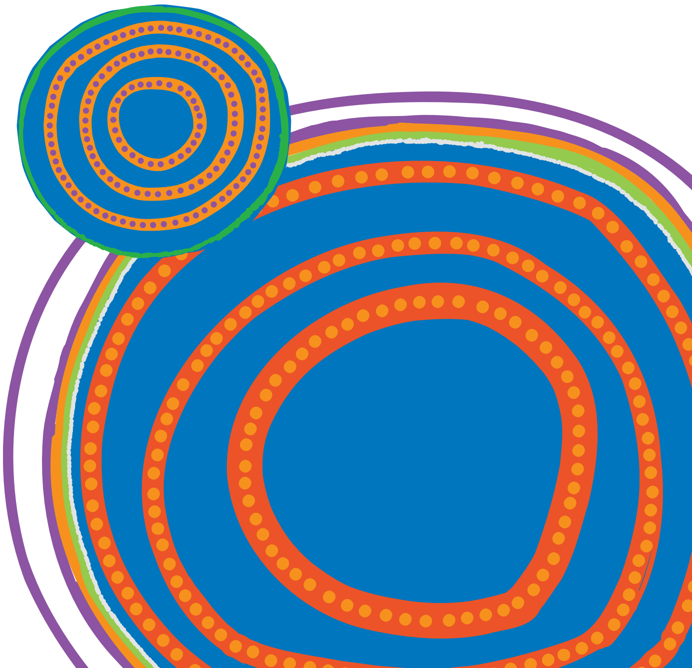
- **Amplifying Impact:** By sharing knowledge, resources, and expertise, we magnify our impact and reduce duplication of effort, ensuring that resources are used effectively and efficiently.
- **Elevating the Voice of Lived Experience:** Partnerships with patient organisations and advocacy groups empower the voices of individuals and families living with rare diseases, ensuring their perspectives are central to all our endeavours.
- **Driving Scientific Advancement:** Collaborations with research institutions, universities, and pharmaceutical and data companies accelerate scientific discovery, leading to faster development of new diagnostics, therapies, and treatments.
- **Building Capacity:** Partnerships with local, national, and international organizations enhance the capacity of healthcare professionals, researchers, and support providers to deliver high-quality care for children with rare diseases.

> *"We are partnering with you as your global leadership is evident."*
>
> Gabrielle Giolblatt, Digital Medical (DiMe) Society

Key Global Initiatives

In the last year we have also launched and expanded our global initiatives including:

- **ERDERA (European Rare Diseases Research Alliance):** As previously mentioned, the Centre is the Australian lead for this groundbreaking 7-year project that sees us connecting with over 170 leading organisations across 37 countries, providing unparalleled access to cutting-edge research, expertise, and clinical trial opportunities for Australian children with rare diseases. This strategic partnership leverages the collective knowledge and resources of a global network to accelerate rare disease research and improve outcomes for children worldwide.
- **The Global Nursing Network for Rare Disease (GNNRD):** In our last report, we shared the establishment of the GNNRD to connect and increase collaboration for nurses caring for people living with RUDs. Since its establishment, the growth in membership has increased significantly. More on the achievements of the GNNRD can be found later in this report. This global network empowers nurses worldwide to share knowledge, best practices, and support, ultimately enhancing the quality of care for children with rare diseases.



International Collaborations

The Centre continues to actively seek and foster new collaborations with leading institutions worldwide to advance research, share expertise, and improve the global standard of care for children with rare diseases.

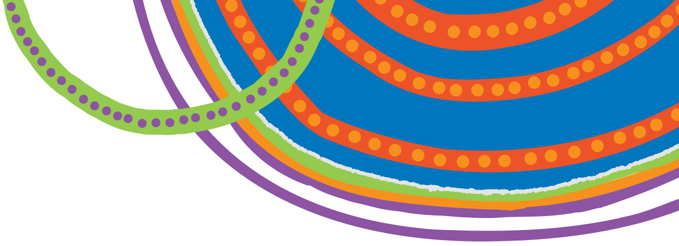
- **Children's Hospital of Fudan University, China:** We have established a partnership with this leading institution to explore joint research projects, clinical trials, and the exchange of expertise in rare disease research.
- **Aga Khan University, Pakistan:** We have extended our support to the Aga Khan University's rare disease activities and genetics and genomics nursing program, enabling them to enhance their capacity to deliver high-quality care for children with rare diseases in Pakistan. This partnership demonstrates our commitment to supporting capacity building in global health and improving access to quality care for children with rare diseases in under-resourced regions.
- **Rare Disease, New Zealand:** Professor Baynam has been appointed to the Rare Disease, NZ Clinical Advisory Panel and its Research Network Advisory fostering strong ties and knowledge exchange between our two countries. This collaboration facilitates the sharing of best practices and fosters a deeper understanding of the unique challenges and opportunities within the Australasian region.
- **African Rare Disease Initiative:** We have established a collaborative relationship with this initiative to advance rare disease research and care in Africa. This partnership reflects our commitment to global health equity and supports the development of sustainable rare disease care programs in Africa.
- **Lancet Commission on Rare Diseases:** Lancet Commission’s focus is on the most pressing issues in science, medicine, and global health, with the aim of providing recommendations that change health policy or improve practice. Professor Baynam has been invited to serve on this Lancet Commission. This prestigious appointment provides a platform for the Centre to contribute to global policy discussions and shape the future of rare disease care worldwide.
- **McKell Institute Rare Disease Research Project:** We are actively participating as a member of the reference committee for this important research project, which will inform policy recommendations for improving the diagnosis and care journey for people living with rare diseases in Australia. This collaboration will contribute to a better understanding of the policy landscape for rare diseases in Australia and inform the development of more effective and equitable systems of care.
- **New Colombo Plan Scholarships:** This Australian Government Department of Foreign Affairs and Trade funding scheme provides opportunities for Australian undergraduate students to undertake semester-based study, language training, and internships or mentorships in 40 participating Indo-Pacific locations. Together with the Sing Health-DUKE-National University of Singapore partnership, KKH Hospital and Curtin University, the Rare Care Centre has created an enduring pipeline of scholarships covering computer science and medicine and with projects ranging from digital health and AI, to virtual reality and data analytics for health system planning.

> “As a leading institution in the field of paediatric healthcare, we at the Children's Hospital of Fudan University are committed to advancing research and care for children with rare diseases. We believe that international collaboration and partnership are crucial to achieving our goals.”
>
> Yi Wang, President, Children's Hospital of Fudan University

> "Sharing an innovation journey between two health systems in our GMT+8 time zone has accelerated the impact for children and families. Leveraging on data science and clinical expertise on real world health system problems in Singapore has allowed for innovations that are improving patient care, directly and indirectly. We are excited to build on the success through our partnership in addressing the unmet needs in rare and undiagnosed disease. We look forward to increasingly delivering solutions for the first 1000 days and beyond – for equity, scale and sustainability."
>
> Saumya Jamuar, Director, SingHealth Duke-NUS Institute of Precision Medicine

- **Global Commission to End the Diagnostic Odyssey for Children Living with Rare Disease:** The global commission brings together rare disease leaders from around the world across public and industry domains to accelerate the time to diagnosis. The Commission identified UTOPIA and the Global Nursing Network for Rare Disease as global exemplars.
- **World Economic Forum (WEF) Rare Disease Initiative:** Gareth Baynam has been invited to join this initiative reflecting his domain expertise and previous WEF contributions towards healthcare sustainability and precision medicine through addressing rare disease.

By fostering global partnerships and embracing a truly collaborative approach, the Centre is driving significant progress in rare disease research, care, and advocacy so that no child is left behind.



Pillar Six: Advocacy, Policy and Legislation

Advocacy, policy, regulatory and legislative action are critical to ensuring that children with rare diseases have access to the best possible care and that their unique needs are adequately addressed within the healthcare and other systems.

Advocacy

The Centre actively advocates for the rights and needs of children with rare diseases through various channels.

- **Partnership with Industry Leaders:** The Centre actively collaborates with key industry partners to raise awareness and promote health equity for individuals with rare diseases. Recently, the Centre provided a video recording by our Medical Director, Gareth Bayman, for use at the Alexion & AstraZeneca National Conference in Sydney. This video, which highlighted the importance of improving health equity and showcased the work of the Rare Care Centre and the Global Nursing Network for Rare Disease, resonated with over 600 company employees. This collaboration demonstrates the Centre's ability to influence corporate awareness and promote a shared vision of improving the lives of people living with rare diseases.
- **Partnership with Rare Voices Australia:** Building upon the successful launch of the Nurse Navigator Program in 2023, the Centre continues to collaborate closely with Rare Voices Australia, the peak RUD national Advocacy body. This partnership provides vital support to families, including access to a national helpline for individuals living with rare and undiagnosed diseases, amplifying the voices of the rare disease community and ensuring their needs are heard and supported.
- **Parliamentary Friends of People with Rare and Undiagnosed Diseases:** In collaboration with this influential group, the Centre hosted a successful "Clinical Trials Day" event in May 2023. This event brought together key stakeholders, including parliamentarians, clinicians, researchers, and patient advocates, to discuss strategies for increasing access to clinical trials for children with rare diseases. The event successfully raised awareness and generated philanthropic support to accelerate our efforts, demonstrating the power of collaborative advocacy in driving change. Meetings with the Parliamentary leaders of the Friends Group in 2024 were focussed to building on success for scale and sustainability.
- **World Health Assembly (WHA) Resolution:** Sue Baker, Program Director and Co-Founder of GNNRD, and Kaila Stevens, Centre Program Manager, are invited members actively involved in a global coalition supporting the adoption of a WHA resolution on rare diseases. This resolution aims to address the unique challenges faced by people living with rare diseases and drive significant improvements in diagnosis, treatment, and care through the development and implementation of a global action plan. Specifically, the coalition aims to have the World Health Organization (WHO) adopt an implementation plan for this resolution. This crucial initiative aims to raise global awareness and mobilize international support for improved rare disease care on a global scale.
- **Publications and Presentations:** The Centre continues to actively engage in knowledge dissemination through the publication of 36 authored or co-authored papers and presentations at 31 conferences, seminars, and academic meetings. These efforts contribute to advancing scientific understanding, raising awareness, and influencing best practices in rare disease care, ultimately informing and shaping future advocacy efforts. Notably, a recent publication in the European Journal of Medical Genetics detailing our cross-sector Model of Care can be accessed here.

Policy

The Centre actively engages with government at all levels to influence the development and implementation of policies that prioritise the needs of children with rare diseases. Our aim in this space is to work with governments to influence the prioritisation of rare and undiagnosed diseases into all healthcare service frameworks and cross sector organisational plans. A start to this work was through the Deloitte Innovation Lab held in July 2024, attended by many members of government across different departments including health, communities, education and more. One of the actions from this included **Governance and Accountability** progress discussions with key senior stakeholders to determine where formal governance and accountability lies for the Rare Care Centre's work and services to be accelerated for impact and reach. This collaborative approach ensures that the voices of children with rare diseases are integrated into policy decisions at the highest levels.

Legislation

The development of a dedicated Rare Diseases Bill for Western Australia remains a key priority that is informed by existing rare disease legislation in our Asia-Pacific region e.g. the Philippines, Japan and India.

By actively engaging in advocacy, policy, and legislative action, the Centre is driving meaningful change to improve the lives of children with rare diseases and their families. The Centre has held meetings with various state and commonwealth government representatives covering multiple domains. The Centre's activities, impacts and leading role has opened ongoing discussions with senior Commonwealth Government officials across a variety of portfolios covering areas such as primary care, chronic disease, newborn screening and others.

Global Nursing Network
Rare Diseases

In March 2023, the Centre proudly spearheaded the launch of the Global Nursing Network for Rare Diseases (GNNRD) in partnership with Curtin Singapore and SingHealth Duke-NUS Genomic Medicine Centre. The Network was developed to connect and increase collaboration and leadership among nurses across the globe caring for people living with a rare or undiagnosed disease.

The Need for this Network

Rare diseases are individually rare, but collectively common. This means that over 300 million people across the globe live with a rare disease. Nurses are the world's largest healthcare workforce on the frontline of patient care. Nurses play a crucial role in being an access point for patients and families with RUD. The GNNRD provides a platform for nurses to connect, share knowledge, and collaborate to improve the quality of care for individuals with rare diseases worldwide. Their unique ability to navigate both clinical and non-clinical settings further strengthens their advocacy potential for the rare diseases community at large.

Building a Strong Foundation: The GNNRD Strategic Framework

The GNNRD is guided by a robust Strategic Framework (2024-2027), developed and co-designed with our Network members. This framework outlines strategic domains, goals, and priority actions to ensure the network effectively addresses the evolving needs of the rare disease nursing community.

Key Achievements

- **Exponential Growth:** The GNNRD has experienced significant growth, with membership surpassing 608 members from 60 countries. This demonstrates the increasing recognition and value of the network within the global rare disease community.
- **Brazil – Our First Country Group:** Brazil was officially recognized as the GNNRD's first country group, led by Dr. Geiza Luz. This milestone marks a significant step towards building a strong regional presence and fostering local leadership within the network.
- **Successful Regional Meetings:** The Network was invited by the Middle East and North Africa Rare Disease Congress to host its 2nd Annual Meeting in Abu Dhabi, bringing together nurse leaders from diverse backgrounds and countries with varying levels of economic development to share knowledge, discuss challenges, and develop collaborative solutions.
- **First Asia-Pacific Meeting:** A successful Asia-Pacific regional leaders workshop was held in Singapore in August 2024, fostering collaboration and developing a roadmap for the regional group.

Fostering Knowledge and Collaboration

- **Education and Training:** The GNNRD's inaugural Education and Training Sub-Committee, comprising eight dedicated nurses from diverse global locations, is diligently developing content for Module 3 of the GNNRD Education and Learning Program to enhance the knowledge and skills of nurses caring for individuals with rare diseases.
- **Inaugural GNNRD Symposium:** The inaugural virtual symposium provided a valuable platform for knowledge exchange and professional development, featuring presentations by leading experts in the field of rare disease nursing.
- **Language Translation:** Recognising the importance of inclusivity, the GNNRD has translated key education resources for nurses into various languages and implemented Wordly™, a cutting-edge real-time translation platform, to ensure effective communication and equitable access for members from diverse linguistic backgrounds.

Looking Ahead: Key Priorities

The GNNRD is poised for continued growth and impact in the coming year. Key priorities include:

1. **Strengthening Leadership:** Expanding the reach and impact of the Education and Training Sub-Committee and exploring the establishment of other key committees, such as a Research & Innovation Committee.
2. **Placing Patients and Families at the Forefront:** To empower the patient voice, the GNNRD has established a Lived Experience Advisory Group. With seven members from six countries, this group will provide critical feedback, guide network activities by ensuring activities of the network are focused on outcomes that matter to people living with RUD and facilitate the dissemination of effective solutions across diverse communities.
3. **Expanding Global Reach:** Continuing to support the development of regional and country groups, fostering strong local leadership and driving impactful initiatives within each region.
4. **Enhancing Communication and Engagement:** Optimising the GNNRD platform and developing a robust communication strategy to enhance member engagement and facilitate knowledge sharing.
5. **Defining Nursing Roles and Scope of Practice:** Collaboratively defining and promoting best practices for nursing care in rare diseases, ensuring that nurses are empowered to provide high-quality, patient-centred care.
6. **Advocating for the Rare Disease Nursing Community:** Advocating for the recognition and advancement of the unique contributions of nurses in the field of rare disease care.

By working together, the GNNRD will continue to empower nurses worldwide to provide the highest quality of care for individuals and families living with rare diseases.

For more information, to join and stay up to date with the GNNRD:

Website: **gnnrd.org**

LinkedIn: **https://www.linkedin.com/company/global-nursing-network-for-rare-diseases/**

Email: **hello@gnnrd.org**

Thank You and Looking Ahead

Dear Friends and Supporters,

Words may not fully capture the profound gratitude we feel for the unwavering support and dedication of our incredible community, and most importantly our children and families living every day with their rare or undiagnosed disease. The Rare Care Centre's journey would not be possible without the invaluable contributions of our partners, sponsors, consumers, and families. Your voices, energy, and commitment to making a difference inspire us every day.

Reflecting on the past year, we are continually amazed by the generosity, kindness, and wisdom that permeates our work. Your unwavering support provides the foundation for all that we achieve, and for that we are deeply grateful. Your continued financial contributions, the insightful conversations that spark new ideas and pathways forward, and the invaluable input and lived experience that guide the co-design of our Centre and its services empower us to focus solely on delivering meaningful, effective and tangible outcomes for our children and families.

We recognise that our work is an ongoing journey. In 2025, we are poised for another year of significant achievement. We have already commenced planning a comprehensive program of work, and we look forward to sharing our progress and achievements in the Year 4 Impact Report.

Key priorities for the year ahead include:

- **Strategic Framework Review:** Conducting a comprehensive review of the Rare Care Centre Strategic Framework to ensure it remains aligned with the evolving needs of the rare disease community and the latest advancements in research and care.
- **Pilbara Hub Development:** Continuing to build upon the successful establishment of the Pilbara Hub, expanding access to high-quality care for children with rare diseases in regional and remote areas.
- **Advancement of the Centre's Clinical Trials Program:** Progressing towards the launch of the Clinical Trials Program in 2025, providing children with access to cutting-edge therapies and accelerating the pace of medical discovery.
- **Implementation of the Family Support Program:** Establish the Family Support Program to ensure that families have access to the comprehensive support services they need, including psychosocial support, financial assistance, and access to essential resources.
- **Ongoing Evaluation and Measuring Impact:** Conducting ongoing research evaluations of the Nurse Navigator and Cross-Sector Care Coordination models to assess their effectiveness, identify areas for improvement, and ensure that these programs continue to meet the evolving needs of children and families.
- **Increased Focus on Mental Health and Well-being:** Developing a mental health wellbeing framework, tools and implementation plan, and introducing spiritual care into our programs.
- **First 1000 Days:** Further increasing impact in early life to prevent suffering and death.

We invite you to join us on this exciting journey. Your continued support is invaluable as we strive to improve the lives of children with rare diseases and their families. All conversations are valued, and every contribution brings us closer to a future where every child has access to the best possible diagnosis, treatment, care and support.

With sincere gratitude,

Sue Baker
Program Director

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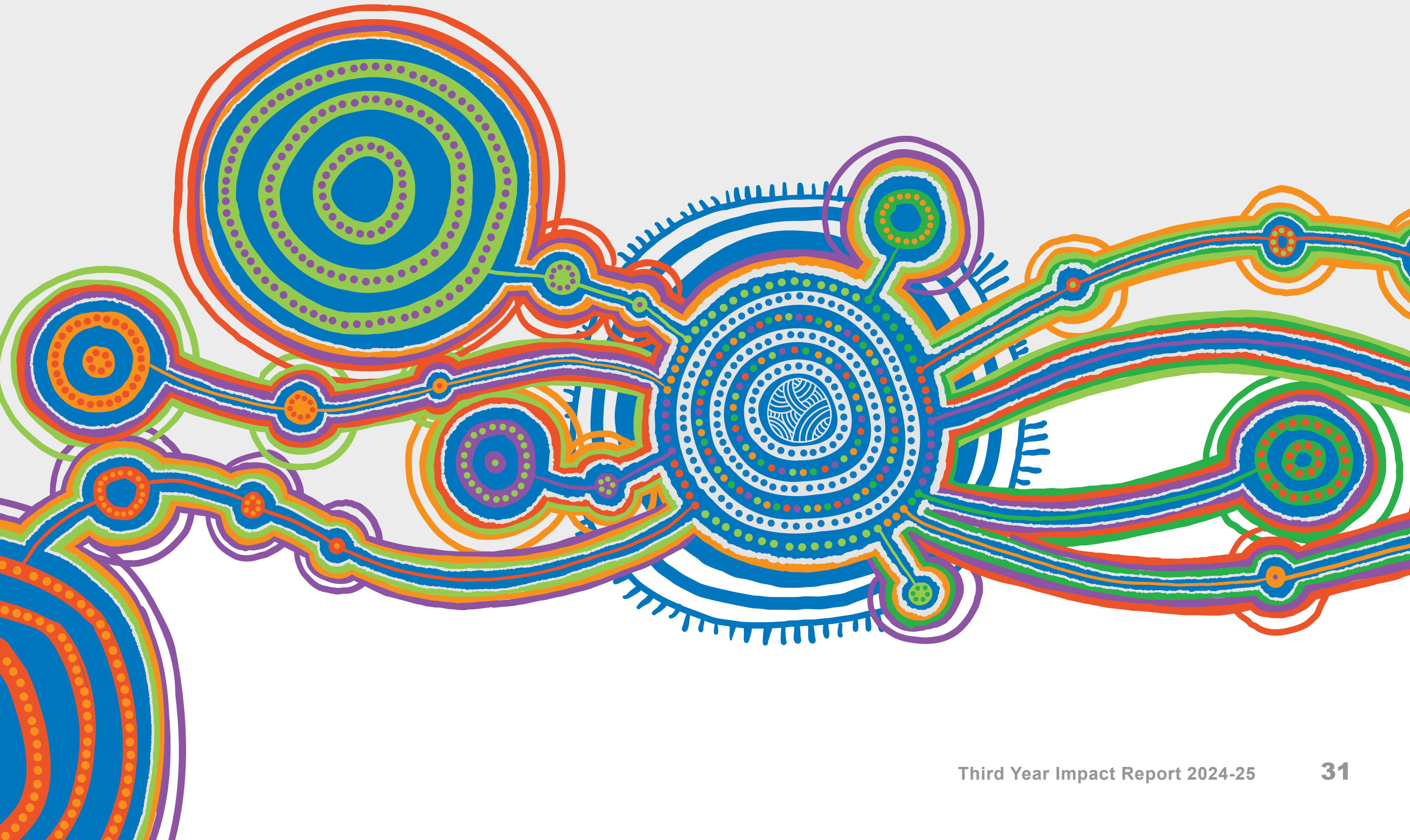

Rare Care Centre

Clinical Centre of Expertise for Rare and Undiagnosed Diseases

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