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

Clinical Centre of Expertise for
Rare and Undiagnosed Diseases

Rare Care Centre

Year 2 Impact Report

February 2023-24

Contents

Cover photo with thanks to Perth Children's Hospital Foundation and Kylie Plunkett Photography

Acknowledgement of Country

The Rare Care Centre acknowledges Aboriginal people as the traditional custodians of the lands across Western Australia where our work is undertaken. We acknowledge and pay our respects 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 diseases' and 'rare and undiagnosed diseases' 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

It is with immense pride that we present you with the annual impact report of our Rare Care Centre (the Centre), now in its second year of operation. While still a new service in the landscape of rare disease care, its branches have already stretched far, reaching patients, families, and professionals across Western Australia, and globally.

From the outset, our mission has been unwavering: to support and meet the needs of children and their families living with rare diseases so that they can live the best lives possible. We do this through a commitment to identifying and bridging gaps, breaking down siloes, taking proactive approaches, striving for best practice and leveraging partnerships. In just two years, the Centre has established itself as a beacon of hope and expertise, not just within our own walls, but across the wider care community in Western Australia and internationally.

We are proud of the unique cross-sector Model of Care that has gained international praise and is being used as a blueprint to support children and their families with rare and undiagnosed diseases. We are humbled by the recognition we have received as a leader in rare disease care. This recognition has manifested in countless ways, including securing grants to expand our services, enabling us to impact even more lives with our specialised care.

We believe in the power of partnership. Our success hinges on the strength of our multi-lateral collaborations with NGOs, research institutions, education providers and care professionals across the globe. These deep partnerships include joint clinical service innovation projects, public-private partnerships, knowledge sharing, people exchange, workforce capacity building, cutting-edge technologies and international funding success.

The Global Nursing Network for Rare Diseases (GNNRD), spearheaded by the Centre, is a testament to our commitment to our global approach, an approach that is required by rarity itself. This network promotes knowledge exchange, empowering nurses worldwide to deliver the best possible care to rare disease patients. Witnessing the rapid growth of this network in the past year fills us with optimism for a future where information flows freely, empowering professionals everywhere to improve the lives of people and their families living with rare diseases.

Our impact goes beyond the boundaries of the Centre itself. We are committed to building lasting change within the wider care landscape as we extend the learnings from the extremity of rare diseases to cancer and common disease. Through dedicated training programs and knowledge-sharing initiatives, we are equipping the clinical workforce with the skills and understanding they need to provide better care for patients with rare diseases, even in settings outside of our specialty centre.

As we turn the page on another year, we do so with unfaltering dedication and hope. The stories of children and families whose lives we have affected through our clinical service, the innovation, and advances in systems, advocacy, policy, and research sparked by our partnerships and the expansion of the impact of our local and global network, fuels us forward.

This report is a testament to the collective effort of our remarkable team, our dedicated partners, and the inspiring children and families who entrust us with their care. It is a snapshot in time, capturing the early echoes of a movement, that through a combination of rare talents and kindness, is shaping the future of rare disease care, care for the common good.

We invite you to delve into the pages that follow and witness the impact in Western Australia that is also rippling outwards, leaving a trail of hope, knowledge, and a brighter tomorrow for children living with rare and undiagnosed diseases.

Take care,

Gareth Baynam
Medical Director

Welcome to the Rare Care Centre

This report marks another year in our ongoing mission to improve the lives of children and their families living with rare and undiagnosed diseases. It highlights the impact we have made thanks to your unwavering support.

63,000 children in WA with rare diseases

8,000 known rare diseases

70% of rare diseases start in childhood

50% of rare disease patients are misdiagnosed

over **50%** of rare disease patients are seeing up to 10 clinicians

there are over **300 million** people worldwide living with a rare disease

Why We Exist

Rare and undiagnosed diseases are a public health priority and are the biggest killers of children, causing 6 in 10 deaths in children, with a mortality rate 13 times higher than common diseases. In Western Australia (WA) alone, over 63,000 children are affected, often facing a long and arduous journey to diagnosis and best practice care. Rare and undiagnosed diseases are severe, chronic, often disabling and life limiting, multi-system and affect all aspects of life. Misdiagnosis and a lack of readily available treatment options add to the burden faced by families.

We envision a future where children and their families with rare and undiagnosed diseases ***live the best lives possible***. Our mission is to support them in identifying unmet needs, navigating towards the right care, and coordinating that care to ***ease the burden they carry***.

Our Approach

The Rare Care Centre (the Centre) operates within a carefully constructed Framework designed to deliver comprehensive, equitable, sustainable and scalable care to children and families living with rare and undiagnosed diseases. The Framework is underpinned by continuous innovation in all aspects of service, enabling us to bridge gaps, address unmet needs and scale our reach to support more children and families.

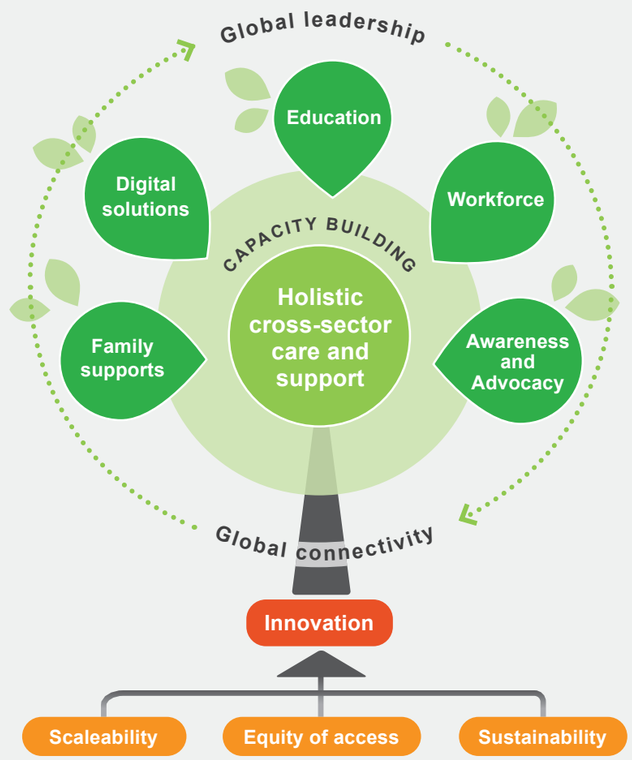
The Centre Framework is a game-changer in transforming the landscape of rare disease care, quickly becoming an international template. It delivers excellence through a unique cross-sector model to provide holistic care and support for patients and families as the critical thrust of our Centre of Expertise.

The Framework is focused on immediate impact and the future, the Model of Care is supported by data, devices and digital solutions; innovative educational and workforce capacity programs; robust family supports; awareness and advocacy; and investment in new clinical innovation and research infrastructure to accelerate advancement in rare disease care. We strive to use our learnings to influence policy and legislative change for long-term impact, while sharing these globally to maximise collective outcomes. Global partnerships are integral to our Framework as we believe in the power of “together for rare diseases”. Through our global partnerships, we share knowledge, resources and best practice, ultimately accelerating progress for children and their families living with rare diseases everywhere.

Rare Care Centre

Framework

Our vision is to ensure that children and families with rare and undiagnosed disease live the best lives possible.



Our Year in Review

135
children and families supported by the clinical service, with a subsequent reduction in hospital utilisation:

- **82%** decrease in bed days
- **16%** reduction in outpatient appointments
- **50%** decrease in emergency dept. presentations

26
Student placements accommodated

9
media campaigns about the Rare Care Centre

365
partnerships and collaborations worldwide

748
meetings held with local, national and global partners

$4+
in economic benefits for every **$1** spent

3
presentations to Parliament in Australia and New Zealand on *Rare Disease Day*

30+
grant applications submitted, contributed to and partnered on.

42
publications authored or co-authored by the Rare Care Centre

270
members in the Global Nursing Network for Rare Diseases across **37 countries**

A Year of Impact

The past year at the Centre has been one of demonstrated impact. We didn't merely set ambitious goals; we witnessed their powerful translation into real and lasting change for children and families living with rare and undiagnosed diseases.

This journey unfolded through six key strategic objectives, each serving as a pillar in our commitment to empower, innovate, and advocate. We are proud to say that at the end of our second year, we have demonstrated success against each of these pillars. In the following pages, we highlight some of our biggest achievements and impacts.

"They listened to me, broke the problem down and step by step sorted it out reaching out to the correct clinics for appointments."
Rare Care Centre Parent

Care Services

Children and families living with rare and undiagnosed diseases have multidimensional and compounding needs that impact not only their physical and mental health, but every aspect of their lives, including education, finances, social activities and employment. Our objective was to provide a state-wide holistic cross-sector care coordination model that complements existing services to address unmet needs for children with rare and undiagnosed diseases.

In our first year, we developed our novel Model of Care to reflect the unique, complex and multi-sector needs of our families, employing team members from across the sectors to provide a wrap-around service. In our second year, we have implemented and tested this model and have come away with promising feedback and demonstrated evidence of its effectiveness in providing navigation, coordination, integration and advocacy support mechanisms.

Rare Care Centre, Strategic Objectives (2021-2024)

Care Services
To provide a state-wide, holistic care model that complements existing services to address unmet needs for children with rare and undiagnosed disease.

Enduring Partnerships
To foster enduring strategic partnerships.

Research and Evaluation
To progress the understanding of rare and undiagnosed diseases by facilitating and connecting to impactful collaborative research underpinned by robust, and child/ family centred evaluation

Voice of Lived Experience
To ensure that lived experience and expertise informs the design of everything we do.

Education and Workforce Capacity
To raise awareness and fill knowledge gaps about rare disease by providing resources, support and outreach services to key stakeholder groups.

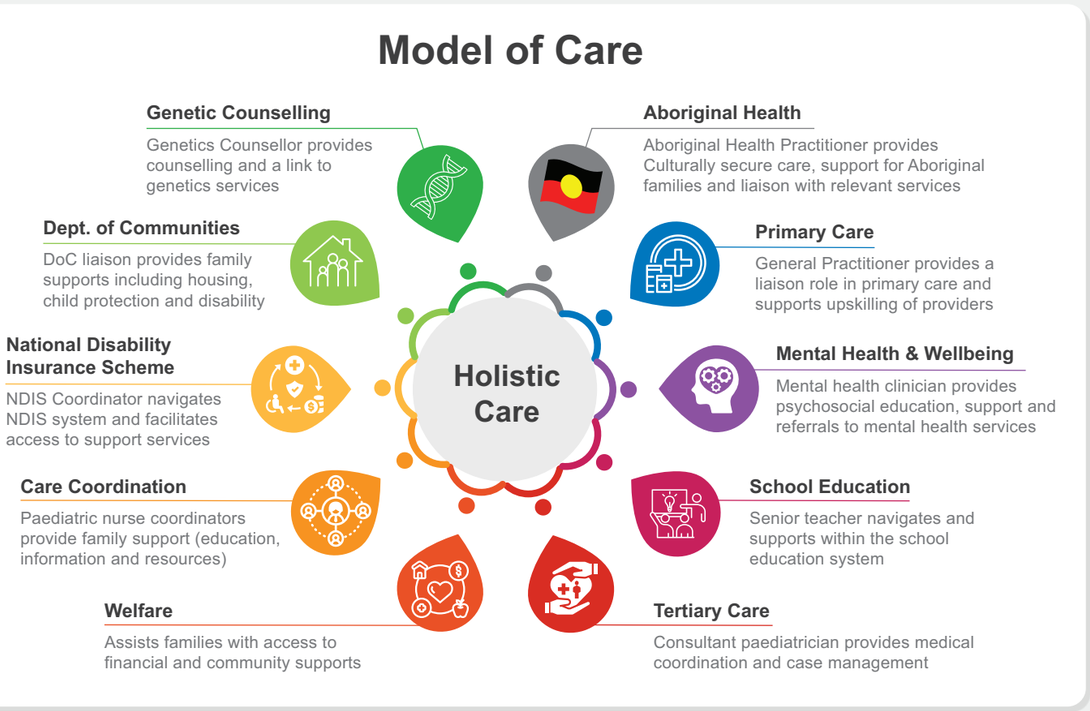
Data, Digital and Devices
To integrate existing digital technologies and facilitate the development and translation of enabling technologies.

Transforming lives: The Impact of our Unique Model of Care

Since implementing this Model of Care in the last 16 months, we have supported 135 children and families with rare or undiagnosed diseases. We continue to receive increasing numbers of referrals as awareness and benefits of our model become more widely recognised, reflecting the unmet need for comprehensive rare disease supports.

Our model is demonstrating its impact; through evaluation of the first 40 families who have now been discharged from the service we can see the following benefits.

- **Enhanced child and family experience:** family experience surveys highlight overwhelmingly positive feedback on the Centre's supportive environment, clear communication and exceptional staff which build trust and a sense of partnership with families. Families reported receiving more information, being listened to and valued, and receiving practical and emotional support improving their healthcare experience.
- **Reduced isolation and improved mental health and wellbeing:** the Centre facilitates connections to other families, support organisations and mental health and wellbeing professionals and community organisations, positively impacting on stress and wellbeing. Demonstrated through patient reported outcome measures, the Perceived Stress Scale-4 and WHO Wellbeing-5, parents report a decrease in stress and increase in overall wellbeing since receiving the Clinical Service.



"The Centre was supportive and actively listened to us... something we have been craving for the past 6 years.

Mother of a child with a rare disease

- **Increased access to the National Disability Insurance Scheme (NDIS) and welfare services:** additional NDIS funding has been secured for families ensuring that their support needs are adequately met. Families have also been upskilled to use their funding more efficiently to achieve greater value. An increased number of families have been able to access disability support pensions, carers payments and other entitlements through the navigation provided by the Centre.

Quantifiable Impact: A Model backed by Evidence

The Centre's innovative Model of Care is not just transforming lives, it is also generating economic benefits for the wider healthcare system and economy. Recent economic analysis conducted on families discharged from the Centre has demonstrated a compelling 4:1 benefit to cost ratio, highlighting the immediate and long-term financial advantages of our approach. This translates to significant cost savings while delivering superior care for children with rare and undiagnosed diseases.

The Model of Care delivers multi-pronged economic benefit, meaning tangible improvements for patients, with discharged patients experiencing an average annual reduction in hospital utilisation per child:

- Outpatient visits reduced by 3.1
- Inpatient admissions reduced by 1.15
- Number of bed days reduced by 4.9 days
- Emergency department visits reduced by 0.5

Our Model of Care provides holistic supports for families, extending its positive impact beyond healthcare costs. Fewer hospital visits mean reduced travel costs for regional patients, estimated at an average individual saving of $5250 per year. Importantly, this support allows families to miss fewer school and workdays and supports improved educational outcomes for children, leading to increased productivity and broader societal benefits.

The Centre's model has demonstrated both improved patient outcomes and significant cost savings and positions itself as a sustainable and replicable model for the future of rare disease care.

Patient & Family Story

Family B came to the Rare Care Centre after a recent diagnosis of a rare disease. Although symptoms had been present since birth, this young person had only received a diagnosis of a complex multi-system rare disease in their late teens. With over 60 appointments involving 12 medical specialties plus multiple therapists and other professionals delivering care at Perth Children's Hospital (PCH) in the previous 12 months, this young person and their mother were left with many questions and minimal answers.

The impacts of a late diagnosis include:

- Minimal access to early intervention and ongoing supports,
- Inability to access NDIS funding without a diagnosis,
- Lack of access to care close to home as therapy was only provided at PCH.

The Rare Care team were able to identify this family's most pressing needs and delivered the following outcomes:

- The Centre GP identified and upskilled a GP who had an interest in rare diseases and was local to the family, to support the young person to transition from paediatric to adult hospital and specialist services effectively.
- The Centre Genetics Counsellor, in consultation with the wider team, created a visual summary with information specific to the young person, their rare disease and its functional impact. This has been used to upskill new therapists, adult healthcare providers and the new GP so the family did not need to repeat their story or take on the role as expert in the rare disease.
- The family was referred to a rare disease specific NGO to provide community, carer and peer support; and resources, information and navigation to guide their healthcare journey.
- The Centre mental health clinician provided strategies to the young person and their family to improve their mental health and wellbeing; and created a plan to transition their mental health care to ongoing community services with links to positive mentor support groups.
- The Centre NDIS Coordinator guided the family through an NDIS plan review to access increased funding to support transition to adult services, increase therapy supports and build independence and protective behaviours. This resulted in increased social, economic and community participation for the young person.
- To support further community engagement, the Centre assisted the young person to apply for the disability support pension to promote independence and choice.
- The mother was also assisted with applications for welfare entitlements and guardianship to ensure she could provide ongoing support for her child.

"They [the team] all worked together and communicated really well. I liked the fact that everything was in one service."

Rare Care Centre Parent

Showcasing success: Recognition for our Model of Care

Our Model of Care is also gaining recognition as a leader in the field. This growing recognition underscores our commitment to building credibility and trust in rare disease care. In the short time since implementing our model, we have had many requests from organisations around the world to share our model and learnings. This spirit of collaboration is central to our approach, and we are proud to be able to share our learnings to build knowledge, generate discussion and inspire positive changes elsewhere.

Our Model of Care has been showcased at prestigious events including:

- The International Congress on Innovations and Leadership in Nursing and Midwifery
- International Rare Disease Research Consortium and RE(ACT) Congress, Berlin
- The Undiagnosed Disease Day Conference
- International Rare Diseases Research Consortium, Montreal, Canada
- The Women and Infants Research Foundation Stars Symposium
- New Zealand Parliament

We are also actively involved in knowledge dissemination through publications. The International Rare Disease Research Consortium (IRDiRC) and the European Journal of Medical Genetics (EJMG) have requested a protocol paper on the Model of Care for a special bundle issue on international best practice. This highlights the growing recognition of the potential to impact rare and undiagnosed disease care on a global scale.

Building on success: Expanding our Model to Reach More

The voices of our patients, families and partners have been instrumental in shaping and enhancing our Model of Care. In the last year we have used this feedback to direct the expansion and improvement of our Model of Care to increase accessibility and reach more patients.

With the aim of expanding our direct patient contact we are on track to increase the number of children we see through the service by 30% in our second year. To meet the demand, we have increased our clinical staffing levels, particularly of our nurses, NDIS coordinator and senior teacher.

To further increase our reach, in partnership with Rare Voices Australia, we are excited to announce the upcoming launch in April 2024 of the ***Nurse Navigator Program***, as one of two sites in a three-year pilot funded by the Australian Government. This program will be part of a suite of initiatives including a National Helpline which will be accessible to all people living with a rare or undiagnosed disease and their carers to connect them with essential resources and support.

The Nurse Navigator Program will offer:

- **Dedicated nurse support:** providing, navigation, coordination and advocacy for children and families.
- **Referral pathways:** facilitating connections and transitions to a range of service providers.
- **Tele-consults:** reducing geographic barriers and wait times to access the service.

"I love that the team understands and assists with the challenges that [a] complex rare disease brings. Nothing was ever too much trouble; the team is friendly and helpful and made a challenging move a lot easier!"

Rare Care Centre Parent

Given the success of our Model of Care and in recognition of the unique challenges and unmet needs of children with rare and undiagnosed diseases in remote regions, we are proud and excited to be developing a secondary site in the Pilbara, **Rare Care Pilbara Hub**. Children with rare diseases living in remote regions face compounded health complexities due to distance, scarcity of primary and tertiary care services, lack of community and disability supports, lack of mental health supports, and inadequate culturally safe and responsive care. Approximately 1,800 children in the Pilbara and 3,000 children in the entire Northwest of WA have a rare or undiagnosed disease and are impacted by the intersection of these challenges.

The **Rare Care Pilbara Hub** will provide supports to these children and families through:

- **Equitable access:** direct access to in-person care providers that are knowledgeable about rare disease needs and challenges, in locations that are convenient and suitable for families.
- **Improved care coordination:** facilitating seamless collaboration across health, education, disability and community sectors.
- **Hybrid model:** utilising both in-person and supported virtual appointments to allow for care close to home while leveraging the resources and expertise from the metropolitan Rare Care Centre.
- **Culturally secure care:** delivered by local staff and ensuring culturally appropriate support closer to home and on Country.
- **Regional capacity building:** opportunities for local staff to develop skills and engage in new opportunities or to return to work.

The Hub has secured $1.84 million towards its $5.8 million establishment and operational costs over five years. The project planning is already underway, and we anticipate welcoming the first family into the service by early 2025.

The achievements highlighted in this section demonstrate the growing momentum and impact of the Centre's Model of Care. We are committed to building on this success, continually expanding our reach and ensuring that every child and family affected by rare and undiagnosed diseases receives the support they deserve.

"Being part of the CRG gives me a voice which reduces my frustration at the system."

Barbara, Consumer Reference Group Member

Voice of Lived Experience

At the Rare Care Centre, we firmly believe that genuine impact cannot be achieved without authentic voices guiding the way. That's why stakeholder consultation is one of our strategic and operational foundations, to ensure lived experience and expertise informs the design of everything we do. Rare and undiagnosed diseases, by their very nature, present unique challenges and experiences that sit on top of, and compound, complexity alone. Without actively incorporating the lived expertise of patients, families, and caregivers, we risk the unacceptable outcome of creating solutions that miss the mark. The insights of children and families are invaluable in informing everything we do, from shaping our care services to establishing our priorities in research and innovation.

Amplifying Consumer Voices: The Power of Stakeholder Consultation

Our dedicated reference groups including our Consumer Reference Group (CRG) and Youth Reference Group (YRG), serve as mechanisms to amplify the voices of lived experience of families, carers, children and youth, to share their perspectives and assist with shaping services and projects of the Centre. These reference groups provide meaningful and informed feedback and input on all activities of the Centre and ensure we are acting in the interests of the families we support.

"I've had to take a massive step back from my career, but this allows me to feel like I can be part of something that's way bigger than me, a piece of something that will have value for others."

Karla, Consumer Reference Group Member

Both the CRG and YRG meet bi-monthly to:

- Provide feedback and advice on the Centre's Model of Care and other strategic initiatives.
- Contribute to discussions regarding current service and care gaps, ensuring the Centre addresses the most pressing needs of the community.
- Participate in online meetings and workshops, fostering collaboration and knowledge exchange.
- Offer feedback on documents such as policies, protocols, and resource materials, ensuring they are accessible, informative, and meet the needs of the community.
- Advocate for other children and youth affected by rare and undiagnosed diseases, amplifying their voices and needs to a wider audience.

Our reference group members are not only contributing to the Centre's success, but also finding personal meaning and purpose through their participation.

The Impact of Engagement

Our deep and meaningful engagement with our reference groups has led to several concrete improvements in the way we work to support families:

- **Post-discharge support:** Following discharge, the Centre remains a pillar of support for families through dedicated phone calls. These calls address lingering concerns, ensure families feel confident with the supports that have been put in place, and identify any emerging needs. We connect families with ongoing resources and support services, enabling a smooth transition out of the Centre service and promoting long-term wellbeing.
- **Accessibility:** We have implemented virtual appointments via video conferencing, improving access for families living in remote locations and reducing the burden of travel.

- **Empowering new voices:** Our CRG suggested applying for a McCusker Centre intern, leading to the placement of an intern who conducted a literature review and benchmarking for the development of a Family Support Program. This has accelerated program development, allowing us to plan for the program sooner. The intern has now joined the Centre as a member of staff, developing capability, retaining knowledge and bringing valuable experience and capacity.
- **Addressing unmet needs:** Based on feedback from the YRG, we are developing a new project focused on creating an Emergency Department (ED) management plan specifically tailored for children with rare and undiagnosed diseases, addressing a critical gap in care coordination.

By prioritising lived experience and amplifying the voices of consumers, we ensure that their expertise and experiences illuminate the path forward.

Beyond consultation: Empowering Change and Capacity

The impact of stakeholder consultation extends far beyond informing our internal practices. Our reference group members are not merely passive participants; they become active agents of change. For instance, Cristian, a CRG member, will soon be facilitating a Rare Care peer support group for fathers. This initiative, born from his own experiences and shared concerns within the group, will provide a much-needed space for connection and mutual support among fathers navigating the challenges of rare diseases.

Participation in the CRG has ignited a passion for advocacy within many members. Karla shared, "Participating in the CRG has inspired me to seek more opportunities for advocacy and change in the health and disability space. I now feel empowered to use my voice to make a difference for others." This exemplifies the transformative power of CRG involvement, equipping youth, parents and carers with the confidence and skills to advocate for themselves and their communities.

"Being part of the CRG for the Rare Care Centre really did change my life. It was my first step into being involved in a for-purpose organisation and it ignited my passion for being involved in this sector. It has totally changed my perspective and career path. I honestly can't thank the Centre enough, which is why I will always be happy to help in any way I can."

Cristian, Consumer Reference Group Member

Opportunities to Amplify Voices on a Global Stage

In recognition of the invaluable contributions of our CRG members who share their unique perspectives and experiences we have invested in opportunities for them to share their valuable insights on a global stage.

In 2023, the Centre co-hosted the "***Connecting Nurses Globally – A Roundtable in Rare and Undiagnosed Diseases***" event in Singapore, which established the Global Nursing Network for Rare Diseases (GNNRD). Highlighting the importance of lived experience, we invited two of our CRG parents, along with two Singaporean rare disease parents to participate in a panel discussion. The panel had an audience of over 50 attendees representing 25 countries.

Alexa's powerful story resonated deeply, emphasising the profound impact seemingly small changes in nursing care can have on a child and family's experience and outcomes, and how this could be facilitated by increased knowledge and awareness of challenges faced by children with rare and undiagnosed diseases. Her voice, along with the other panel members, helped shape the Roundtable's discussions towards tangible actions that directly benefit patients and families.

As a direct result of consumer participation, one of the guiding principles of the GNNRD established at the event is '***partnering with patients and families***'. Furthermore, a global Lived Experience Advisory Group is being formed to provide ongoing guidance to the Network, ensuring consumer voices remain at the forefront.

We are thrilled to welcome Alexa back as a consumer representative at the upcoming Middle East and North Africa Rare Disease Congress in Abu Dhabi (May 2024) where she will co-present with the Program Director on the co-design of our Rare Care Centre, further solidifying our commitment to collaborative model development and its global dissemination.

"Having a child with a rare disease has led me so many places, both good and bad, that I never imagined I would go, and being involved in the GNNRD Singapore Roundtable was another unexpected, somewhat daunting, yet exciting opportunity. To sit in a room full of medical professionals eager to learn about rare diseases so that they could provide better care for rare patients and their families was inspiring and gave me genuine hope for the future. And to realise that our perspective was not just wanted but genuinely sought after to help inform the training of nurses to meet the needs of rare patients and their families was immensely gratifying."

Alexa, Consumer Reference Group Member

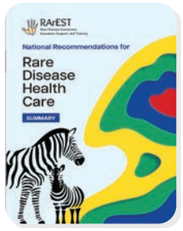
Education and Workforce Capacity

The Centre's Clinical Service directly supports up to 150 children and families each year. However, there are over 63,000 children in WA who have a rare or undiagnosed disease who also need some level of support. We understand that to have a significant impact on the outcomes of children with rare and undiagnosed diseases more broadly we need to uplift all who might provide care. This is why education and workforce capacity is a core strategic objective.

Bridging the Gap: Building Knowledge and Empowering Professionals

By equipping healthcare and other sector and agency professionals, as well as the wider community with the knowledge and resources they need, we can create a ripple effect spreading out understanding and upskilling others both within the state and beyond. We achieve this through a multi-faceted approach including building internal expertise, fostering collaboration and sharing best practice locally and globally.

In collaboration with the Royal Australian College of General Practitioners (RACGP) we developed impactful case studies on diagnosing and managing rare and undiagnosed diseases to be included in the RACGP publication “Check”. These resources highlight the importance of early recognition and holistic care, contributing directly to the knowledge base of GPs who are often the first health care professional to encounter symptoms of a rare disease.



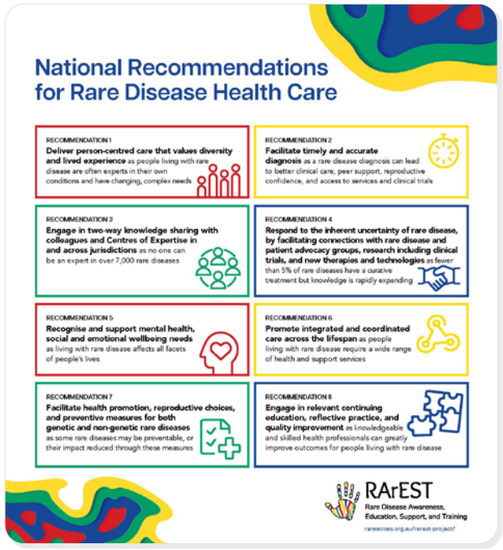
Through our partnership with RArEST (Rare Awareness, Education, Support and Training, a Commonwealth funded initiative), we also co-authored the first Australian National Recommendations for Rare Disease Health Care, which provide clear guidelines for optimal care throughout a rare disease patient's life. These valuable resources directly improve the knowledge and awareness of healthcare providers across Australia, ensuring they are better equipped to identify, support and improve the outcomes for individuals with rare diseases.

Within the WA Health system, the Centre has utilised different approaches to spread the message of rare and undiagnosed diseases such as by hosting a yarning circle at PCH, bringing together participants from education, health and research. This collaborative forum fostered open dialogue and knowledge exchange focused on improving care access and inclusivity for Aboriginal children and families in WA.

Through a roadshow, the Centre has presented on rare and undiagnosed diseases and the work of the Centre to over 25 WA Health teams, including to 32 medical consultants and registrars at the August 2023 PCH Grand Round. In this presentation the Centre Medical Director and GP focused on rare and undiagnosed disease basics, the Centre Model of Care, and the crucial role of primary care in managing rare and undiagnosed diseases. The impact across these presentations has been significant. Feedback we received highlighted the disconnect between specialists and GPs and the lack of knowledge about the resources available in primary care that could be integrated into specialist treatment plans. The audience expressed a strong desire for a similar model to be available for all children with common diseases, highlighting the broader applicability of our approach.

The Centre proactively seeks opportunities to share knowledge and expertise through a variety of national and international forums. This commitment to global collaboration cultivates awareness and inspires best practice for improved rare disease care. The Centre team have presented the Model of Care through multiple international platforms and presented three ECHO sessions as part of the Rare Disease Project ECHO® through Rare Voices Australia. This interactive format allows geographically dispersed healthcare providers to learn from each other and experts in real-time, creating a community of practice and ultimately improving care for patients with rare and undiagnosed diseases.

The Centre has also hosted the first two webinars of a series showcasing global nursing-led models of care through the Global Nursing Network for Rare Diseases (GNNRD). These webinars, available on the GNNRD YouTube channel, contribute to worldwide knowledge sharing, supporting the development of new models of care and improving care practices.



Building the Future Workforce

Investing in, and nurturing the future generation's workforce is another essential pillar of our educational strategy. We have prioritised supporting student placements across various disciplines, exposing students to the realities of rare and undiagnosed disease care, equipping them with practical experience and fostering long-term capacity building. Highlights from this year include:

- **Diverse Student Engagement:** Given the cross-sector nature of rare and undiagnosed diseases, our placements are not limited to healthcare students but we provide opportunities for undergraduate and postgraduate students from various universities, disciplines and programs, including Notre Dame Health Promotion, McCusker Centre for Citizenship Internships, New Colombo Plan scholars in partnership with SingHealth (Digital and AI), software engineering and an organisational psychology PhD student researching cross-sector work design within the Centre. These students gained valuable experience through projects and placements focused on areas like rare disease phenotype coding, clinical trials infrastructure, peer-support programs, and mental health in the context of rare diseases.
- **Empowering Medical Students:** In collaboration with the University of Western Australia (UWA), the Centre provided placements for 12 medical students undertaking their service-learning unit. Through diverse projects related to mental health, advocacy, and awareness, these students developed a deeper understanding of the needs of the rare and undiagnosed disease community, which will undoubtedly impact their future patient care.

"Working with this Centre has changed my ability to communicate with patients because my mindset has been altered by the stories and research we've heard and done for this project."

Ping, UWA Medical Student

- **Expanding Opportunities**: We recently expanded our Global Nursing Network for Rare Disease to include nursing students and midwifery students, providing them with upskilling opportunities early in their careers. Additionally, we partnered with Medics 4 Rare Diseases (M4RD) to deliver a medical student forum, providing opportunities for medical students to increase their knowledge and advocate for rare diseases early in their career.

As part of Rare Disease Day, 29 February 2024 the Centre hosted a range of activities to advocate for children and raise awareness about rare and undiagnosed diseases. Highlights from this day included:

- **Educational Scavenger Hunt:** The Centre's UWA service-learning medical students organised a successful event where 60 children participated in a scavenger hunt with educational facts about rare diseases and animals accompanied by arts and crafts with a rare animal theme, specifically the numbat, a rare animal native to WA. This idea was born through a recommendation developed as part of a literature review completed by the medical students, now published in Elsevier Rare Journal.
- **Rare Disease Day Mural:** Standing tall at the Perth Children's Hospital is a vibrant interactive mural depicting two wings created from the hand (and a foot) prints of children and families collected during Rare Disease Day 2023. This powerful symbol embodies the strength and courage of the rare disease community and serves as a daily reminder of the importance of standing in solidarity with these children and their families.
- **Hospital Exposition:** The Centre collaborated with 'Connect Groups WA' to host an expo at PCH to provide a platform for rare disease advocacy organisations to connect with healthcare workers and raise awareness of rare disease care.
- **Educational Toolkits:** In collaboration with PCH School of Special Educational Needs: Medical and Mental Health, the Centre disseminated Rare Disease Day toolkits to be incorporated into the curriculum to educate teachers and students about rare and undiagnosed diseases.

"It makes me smile and hold immense amounts of pride knowing the Rare Care Centre's initiatives are helping close the gaps in the system"

Darcy, Youth Reference Group Member [on the Rare Disease Day mural]

Data, Digital and Devices

Globally, healthcare is experiencing a paradigm shift driven by aligning technology to unmet need. We acknowledge this evolution and remain at the forefront, harnessing innovation to deliver more effective, efficient, and personalised care for children and families living with rare and undiagnosed diseases. Our strategic objective in this pillar is to integrate existing data, digital and device technologies and facilitate the development and translation of enabling technologies.
We believe technology can create significant value by:

- **Improving quality of life:** Empowering families with easy access to understandable information and streamlining communication with care professionals.
- **Enhancing clinical outcomes:** Facilitating data sharing, collaborative care, and evidence-based decision making.
- **Scaling impact:** Ensuring our solutions are sustainable and reach individuals across local, national, and global landscapes.

Over the past year, there has been a growing adoption of Generative Artificial Intelligence (Gen AI) throughout the healthcare sector. The Centre continued to be at the forefront of innovation by working to implement specific Gen AI technologies for application in WA. In a world first, the Centre is leading the application of commercially available (e.g., ChatGPT) and non-commercial large language models on whole of health system electronic health record (EHR) data. The Centre lead of data, digital and technology has delivered a rare new disease AI method (fast concept recognition). This method is faster and cheaper than using existing Gen AI technology. This method can be used with or without an EHR which means it can be deployed anywhere, including places such as low and middle-income countries that do not have an EHR or that cannot afford some Gen AI technologies.

Empowering Patients through Technology

We are committed to leveraging technology to streamline workflows, improve patient management, and empower individuals and families with rare and undiagnosed diseases. Here are some of our key initiatives:

- **Optimised Clinical Workflow:** We are undertaking the development of a Microsoft Power Apps-based solution designed to streamline clinical workflows. This solution will enhance efficiency and ensure seamless patient care coordination.
- **Rare Disease Patient Passport:** In collaboration with CamRare (Cambridge Rare Disease Network), we are implementing a patient passport for families in the Nurse Navigator Program. This passport will act as a centralised repository for critical medical and personal information, empowering patients and families to manage their care more effectively. The passport will facilitate better information flow between care providers across different sectors, both within and outside the health system. Additionally, we have formed a Working Group with several other international rare disease organisations to collaborate, and share information and learnings about patient passports. Several of these organisations will also utilise the CamRare passport as critical mass is essential in building recognition and credibility of the product. We are planning to actively recruit additional countries and organisations through the Global Nursing Network for Rare Diseases to participate in the pilot program and broaden the reach of this initiative.

- **UTOPIA Platform:** The Centre's UTOPIA platform (Unlocking Treatment Options, Personalized In-Time Access) utilises Gen AI to create semi-automated summaries of a patient's condition. These summaries leverage external knowledge bases to create a comprehensive overview, including information on clinical trials, symptom management, and disease surveillance, all presented in accessible language. UTOPIA is already impacting lives through pilot programs in Singapore: Phase 1 covers 90% of Singapore's paediatric care, and Phase 2 expands to 1.2 million EHRs across all child health conditions. UTOPIA fosters collaboration and personalised care, ultimately improving outcomes for individuals with rare diseases.

- **Patient Portal:** Providing a secure digital platform for families to share information with professionals across various sectors (e.g., medical, education, disability) is a top priority for the Centre. A mapping workshop was well attended by cross-sector partners and consumer representatives delivering surprising and valuable insights into the highest priorities and important considerations for the portal development. A partnership has now been formed with Microsoft to develop a blueprint for the implementation of an international patient portal pilot. The Patient Passport will serve as a steppingstone for this initiative, informing the design and functionality of the future patient portal.

Revolutionising Care: Phenotype trajectories in Rare Diseases

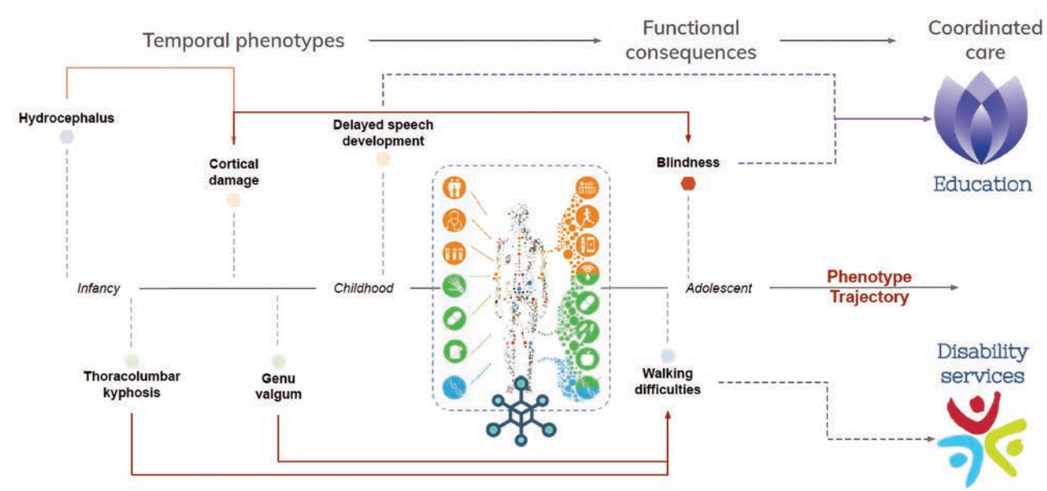
Many rare diseases are complex, with limited data available on disease progression. To address this challenge, the Centre is pioneering the development of phenotype trajectories. This approach involves breaking down a disease into its observable characteristics (phenotypes), which allows for a more comprehensive analysis and prediction of disease progression and health outcomes.

Creating computational models of phenotype trajectories enables predictions of the course of a child's health and wellbeing, including the potential functional consequences of the condition. This information can then be used to create better informed care pathways for both diagnosed and undiagnosed patients, encompassing aspects like prognosis, surveillance, and coordinated clinical and social care. The Centre is collaborating with colleagues from SingHealth Duke-NUS Institute of Precision Medicine and the Children's Hospital of Eastern Ontario to develop these models, utilising Gen AI methods to extract phenotypes from EHRs for specific disease groups.

These phenotype trajectories hold immense potential to:

- Personalise care plans for patients with rare and undiagnosed diseases.
- Improve the accuracy of prognosis for individuals with rare and undiagnosed diseases.
- Inform decisions related to disability and education services.

The Centre's research in this area not only benefits our own patients but also contributes valuable knowledge to the global efforts in rare disease.



Research and Evaluation

At the Centre, we understand that progress in rare disease care hinges on the advancement of knowledge. That's why research and evaluation are integral to our strategic objectives. We are committed to facilitating impactful collaborative research, underpinned by robust and child and family-centred evaluation.

Expanding Access: Building a Sustainable Paediatric Clinical Trials Infrastructure

In recognition of the critical role of clinical trials in advancing treatments and improving outcomes for individuals with rare and undiagnosed diseases the Centre is committed to establishing a sustainable clinical trials infrastructure to make participation more accessible for families across Western Australia. This aligns perfectly with our strategic goals of:

- **Enhancing knowledge:** Clinical trials play a crucial role in advancing our understanding of rare and undiagnosed diseases.
- **Building partnerships:** Effective clinical trials require strong collaboration between researchers, clinicians, and patient advocacy groups.
- **Improving patient care:** Ultimately, our goal is to translate research findings into improved treatment options and better outcomes for individuals with rare diseases.

Despite a global increase in rare disease trials, there remains a significant gap between the needs of WA children and available local opportunities. Only 5% of rare diseases have an existing treatment that alters the course of the disease, and for many access to a clinical trial is their only hope of a treatment.

To address the disparity of access and the unmet need, the Centre has undertaken several initiatives:

- **Hope in Clinical Trials Event:** In collaboration with the Parliamentary Friends of People with Rare and Undiagnosed Diseases, the Centre hosted a successful event for clinical trials day in May 2023. This event brought together stakeholders including parliamentarians, clinicians, senior public servants, researchers, advocates and people with lived experience, to discuss strategies for increasing access to clinical trials in WA. The event successfully generated public awareness and attracted philanthropic support to accelerate our efforts.
- **Stakeholder Engagement:** Through an extensive review process and ongoing collaboration with a diverse group of stakeholders (patients, families, researchers, healthcare providers), the Centre has identified barriers and opportunities to clinical trial access and participation in WA. This ongoing activity is crucial for developing effective strategies to streamline processes and reduce these barriers.
- **Building a Sustainable Infrastructure:** The Centre is actively working on developing a comprehensive plan to build a sustainable clinical trials infrastructure for children with rare diseases in WA. This plan will address the identified barriers and bring clinical trials closer to home for families.

The Centre is finalising a scoping paper that outlines the key findings and recommendations for building a robust paediatric rare disease clinical trials infrastructure in WA. To bring together and accelerate an integrated suite of innovation and research initiatives, we are planning to submit an application to the WA Future Heath Research and Innovation Fund to establish a Collaborative Centre for Research and Innovation Excellence for Rare and Undiagnosed Diseases.

Listening and Learning: A Patient-Centred Approach to Evaluation

At the Centre, we believe that patient-centred evaluation is fundamental to ensuring our services meet the needs of individuals and families affected by rare and undiagnosed diseases. We also know that a one-size fits all approach to evaluation will not be successful. We understand that families with rare and undiagnosed diseases are over-burdened already, and we have found that paper surveys are not always able to capture a family's experience sufficiently. Utilising the experience of our team we have been able to pivot our evaluation to utilise a variety of methods to capture a wide range of feedback and assess the impact of our programs and tailor services as needed in response to evaluation data.

- **Family Experience Surveys:** We conduct surveys to gather feedback from families who have been supported by our services. This feedback helps us understand their experiences, identify areas of success, and areas for improvement.
- **Qualitative Research:** The Centre is planning to incorporate qualitative research methods to gain a deeper understanding of family experiences and outcomes, capturing rich details not readily revealed through quantitative data.
- **Consumer Representation:** Two consumer representatives actively participate in our research evaluation working group. Their valuable insights help us craft evaluation processes, develop relevant questions, and ensure that our initiatives are truly responsive to the needs of the rare and undiagnosed disease community.
- **Health and Wellbeing Tools:** We use validated health and wellbeing tools like the Perceived Stress Scale-4 and WHO Wellbeing-5 tool to guide interventions offered to families. This helps us monitor their wellbeing and adapt our support accordingly.
- **Service Outcome Evaluations:** Planning our nurse navigator program has provided a unique opportunity to align the evaluation of our nurse-only service and cross-sector service which will enable us to demonstrate the different outcomes delivered by each service in comparison to the cost, which is crucial evidence for future investment.

IRDiRC Taskforce: Advancing Global Impact Measurement

The Centre's commitment to research and evaluation extends beyond our own endeavours. We are actively engaged in collaborative efforts to advance the field on a global scale. For example, in recognition of our expertise, the Centre Program Director was selected as a member of an International Rare Diseases Research Consortium (IRDiRC) Taskforce which is exploring the impacts of rare diseases on patients and families, identifying impact measurements and capturing potential factors that improve and/or challenge the impacts, with an intent to publish the work in 2024.

Enduring Partnerships

At the Centre, we know that we cannot achieve our mission alone. Meeting the needs of people and families living with rare and undiagnosed diseases is a global public health priority, this is reflected in the United Nations Resolution on Rare Disease, and the preparations for the World Health Organisation Resolution for Rare Disease. Collaboration is key to achieving our mission.

Our rare position as a disease-agnostic government agency grants us a unique opportunity to foster a highly collaborative environment with non-government disease-specific organisations. This has opened doors to unique partnership opportunities enabling us to leverage the expertise of a wider range of organisations. This is evidenced through numerous letters of exchange, regular meetings, and a general willingness to share information and partner with us on projects.

Fostering strong and enduring partnerships is a cornerstone of our strategic approach. Collaboration allows us to:

- **Amplify our Impact:** By working together with other organisations, we can extend our reach and ensure our efforts have a wider impact on the rare and undiagnosed disease community.
- **Share Knowledge Globally:** Partnerships facilitate the exchange of knowledge and best practices, leading to more effective care and improved outcomes for individuals with rare diseases worldwide.
- **Avoid Duplication of Work:** Collaboration helps us to identify areas where efforts can be combined, maximising efficiency, and ensuring valuable resources are used effectively.

By forging strategic and comprehensive partnerships, we extend the reach of our expertise beyond clinical care to truly deliver a Clinical Centre of Expertise. These collaborations encompass the full spectrum of our work, enabling us to deliver a more holistic approach throughout the entire patient journey. This translates to faster diagnoses, improved treatment plans, better outcomes and ongoing support for individuals and families affected by rare and undiagnosed diseases.

The Power of Partnership: A Global Approach

In the rare disease field, a global approach is essential, therefore our philosophy is "**go global or go home**" – we strive to develop resources and interventions that can be adapted and implemented across different countries and contexts. This is why we have shared our learnings and successes locally and globally and are committed to continuing to do so. The following categories describe our various types of partnerships and collaborations:

- **Knowledge Sharing –** We partner with other rare disease centres and service providers with expertise in similar areas for benchmarking best practices, sharing lessons learnt and upskilling each other. An example includes our partnership with the Syndromes Without A Name (SWAN) Clinic, Cardiff and Vale University Health Board, Wales with who we share models, business cases and evaluation and metrics frameworks, as well as collaborating on specific initiatives.
- **Collaboration on Initiatives –** We partner with a variety of organisations to collaborate on individual initiatives as rare disease care often requires us to tackle complex challenges that necessitate combined expertise and resources. An example is our collaboration with the Human Phenotype Ontology team at the Berlin Institute of Health at Charité, Germany. This collaboration helps ensure accurate and standardised representation of phenotypic data, which is crucial for advancing rare disease research.
- **Advocacy –** We believe that rare and undiagnosed diseases are “everyone’s business” so we partner with organisations to raise the profile of rare and undiagnosed diseases. An example is our collaboration with Parliamentary Friends of People with Rare and Undiagnosed Diseases, a non-partisan forum established by Members of the Parliament of Western Australia to raise awareness of and increase liaison with people with a rare and undiagnosed disease for parliamentarians.
- **Service Delivery Expansion** – We recognise that there are other specialised services that would strongly benefit our patients and families. By partnering with these organisations, we are able to expand our service, maximise our collective impact and deliver a more seamless and supportive experience for patients and families. An example from this year is our partnership with Rare Voices Australia to develop and pilot the Nurse Navigator Program.
- **Networking –** We create networking opportunities for professionals to share knowledge and resources more effectively, advocate for policy changes that benefit the rare and undiagnosed disease community and raise awareness on a broader scale. One of the biggest highlights from this year is establishing the Global Nursing Network for Rare Diseases to create a global community of practice for nurses to connect worldwide, share expertise, and access essential resources.
- **Education –** we partner with educational institutions to upskill current professionals and educate the next generation with specialised knowledge and skills in caring for individuals with rare and undiagnosed diseases. Examples include our collaboration with Curtin University who are considering rare diseases as a grand challenge that can be incorporated across the University.
- **Industry partners –** we partner with industry such as technology and pharmaceutical companies to explore new treatment options, improve access to therapy, remain at the forefront of innovation and co-fund initiatives focused on rare and undiagnosed diseases. An example is our partnership with Microsoft to develop the Rare Disease Patient Portal.

Snapshot of Rare Care Centre Partnerships and Collaborations

Global Alliance for Genomics and Health (Canada): Rare Disease Community of Interest which is co-lead by Dr Tudor Groza (Rare Care Centre Data and Technical Lead). Focused on developing standardised approaches for data sharing and leveraging phenotype knowledge to improve clinical decision-making and patient management.

Syndromes Without A Name Clinic (Wales): Sharing of models, business cases, evaluation and metrics frameworks; connection with other collaborators and creation of regular sharing forum; Collaboration on Rare Disease Patient Passport and Global Nursing Network for Rare Diseases development and implementation.

Berlin Institute of Health (Germany): Collaboration with Human Phenotype Ontology team to ensure accurate and standardised representation of phenotypic data crucial for Rare Disease research.

Rare Voices Australia (Australia): Key national partner and peak advocacy body for rare diseases in Australia co-creation of the Nurse Navigator Program; Co-authorship of the Recommendations for Rare Disease Health Care; Support and collaboration on variety of other knowledge exchange, policy and advocacy initiatives.

Parliamentary Friends of People with Rare and Undiagnosed Diseases Western Australia (Western Australia): A non-partisan forum established by Members of the Parliament of Western Australia to raise awareness of and increase liaison with people with a rare or undiagnosed diseases for parliamentarians. We have collaborated on events for clinical trials day 2023 and rare disease day 2024.

By type

10	48	306
Funders	Formal partnerships	Informal partnerships and collaborations

By location

106	63	196
WA based partners	Australia based partners *(outside of WA)*	Global partners

Explore our live partnerships and collaborations map by using the QR code.

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 via an inaugural roundtable event. This two-day event saw global leaders and nurses representing 25 low-, middle- and high-income countries across North America, Africa, Europe, Asia and Oceania come together to co-design the network and a nursing education program for rare and undiagnosed diseases.

Recognising the Critical Role of Nurses

Nurses, the world's largest healthcare workforce, stand on the frontline of patient care, serving as a crucial access point for individuals with rare diseases. They offer comprehensive and compassionate support throughout the entire care journey, encompassing screening, education, treatment delivery, monitoring, and psychosocial support. Their unique ability to navigate both clinical and non-clinical settings further strengthens their advocacy potential for the rare disease community at large.

Addressing the Need for Collaboration

Prior to the GNNRD's establishment, a significant gap existed with no mechanism to connect nurses globally and foster the exchange of knowledge and best practices in rare disease care.

Building a Network for Collective Action

The Network provides a collective global voice and strategically positions nurses to contribute to advocacy and policy platforms for rare and undiagnosed diseases. It enables nurses worldwide to connect, share expertise, and access essential resources. Through the GNNRD, nurses can:

- Engage in peer-to-peer learning through interactive forums and knowledge exchange platforms.
- Access a comprehensive library of educational resources specifically tailored to rare disease management.
- Contribute to the collective knowledge base by sharing their experiences, insights and initiatives.

A Network with Global Potential

The GNNRD represents a significant milestone in empowering nurses to deliver the highest quality care to individuals with rare and undiagnosed diseases. By fostering collaboration and knowledge dissemination, this network will:

- Improve the standard of care for individuals with rare diseases worldwide.
- Reduce diagnostic delays and improve access to specialised care.
- Strengthen advocacy efforts for increased resources and research initiatives.
- Empower nurses to become leading voices in the rare disease community.

GNNRD Achievements and Building Momentum

Although the GNNRD is in its infancy, today, it has 270 members from 37 countries with minimal active recruitment showcasing its organic growth and global demand. Momentum continues to build, and the sections below highlight some of the network's most notable achievements so far.

Strong Foundations for Global Collaboration

The GNNRD has established a robust governance structure with a Nursing Reference Group, comprising 11 members that represent the diversity of the Network's membership, providing expert input, and a Global Leaders' Implementation Group offering strategic direction and oversight.

Connecting Expertise and Empowering Nurses

The GNNRD's website and professional networking platform has been launched and serves as a central hub and virtual community of practice, facilitating connection, collaboration and knowledge sharing among nurses globally.

Delivering Education and Targeted Resources

The development of nurse-specific rare disease educational content is crucial, for the first iteration the GNNRD is partnering with Medics 4 Rare Disease while working towards a globally available accredited course.

Expanding the Network

The GNNRD is actively expanding its global footprint and working on options to build regional content and engagement. The network was successful in obtaining funding to establish an Asia Pacific regional chapter and a grant submitted to the Australian Government of Foreign Affairs and Trade has been shortlisted to facilitate a regional chapter in China. Further discussions are underway with the Middle East and North Africa, India, Brazil, and Ghana.

Looking Forward: A Strategic Vision for the Future

The GNNRD is actively developing a Strategic Framework and Operational Plan to guide its future direction and priorities. Finalising these plans and developing the structure and function of regional and national chapters will be the focus of an upcoming meeting in Abu Dhabi in May 2024 as part of the Middle East and North Africa (MENA) Rare Disease Congress. GNNRD members not attending the meeting will have the opportunity to provide feedback through virtual workshops.

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**

Our Year Ahead

The Rare Care Centre has concluded a year of remarkable achievements, a testament to the unwavering dedication of our staff, reference groups, partners, and supporters. Together, we have made significant strides towards our vision: improving the lives of individuals with rare and undiagnosed diseases. As we reflect on this successful journey, we are energized to set our sights on the horizon and embark on an exciting new chapter. We are actively developing a new three-year strategic plan to guide our future endeavours.

Next year we will focus on building on our strengths and successes. We will implement new services to expand our reach including the Pilbara Hub, Nurse Navigator Program and Family Support Program and continue to grow our initiatives such as the Global Nursing Network for Rare Diseases, Patient Portal and Clinical Trials Infrastructure.

Looking ahead, we are confident that this next year will be marked by continued growth, impactful developments, and a sustained journey towards improving the care of children and families living with rare and undiagnosed diseases.

Thank You

Dear Friends and Supporters,

With the end of another year at the Rare Care Centre, I find myself reflecting on the remarkable journey we've shared. In these pages, you have witnessed the impact of your unwavering support and how it has been woven into the very fabric of our hope, progress and dedication.

From day one, you believed in our vision, to help children with rare and undiagnosed diseases and their families live the best lives possible. Your faith, translated into action through grants and countless acts of generosity, has propelled us forward. It is truly an honour to stand with you as our partners, who share our unwavering commitment to making a difference.

We would especially like to acknowledge and thank our philanthropic funders the Angela Wright Bennett Foundation, The Stan Perron Charitable Foundation, The McCusker Charitable Foundation via the Channel 7 Telethon Trust and the Perth Children's Hospital Foundation (Principal Funder Mineral Resources Limited and Lance East Office via Telethon, Market West and other donors) without whom the Centre would not have been established. Thank you also, to other funders and supporters who have made invaluable contributions that have enabled our achievements this year.

The successes we celebrate in this report – expanded services, a thriving global network, a ripple effect of enhanced care across the healthcare landscape – are all testaments to your partnership. Every grant awarded, every donation received, every hand extended in collaboration becomes a bridge, connecting patients to vital care, researchers to new breakthroughs, and communities to a shared sense of hope.

However, numbers and statistics can only tell part of the story. The true impact of your support resonates in the smiles of families finally receiving a diagnosis, the tears of joy shed over milestones achieved, and the quiet comfort of knowing there is a network of support ready to walk with them as they navigate through challenging and complex journeys.

As Program Director, I am privileged to witness these transformations firsthand. I see the fear give way to hope, the isolation replaced by a sense of belonging, and the daunting unknown illuminated by the power of collaborative care. And in every moment, I see the reflection of your generosity, your compassion, and your unwavering belief in a brighter future for those living with rare and undiagnosed diseases.

From the bottom of my heart, and on behalf of the entire Rare Care Centre team, I thank you. Your partnership is more than just a contribution; it is the lifeblood of our mission. Together, we are breaking down walls, forging new paths, and ensuring that no one faces the challenges of rare and undiagnosed diseases alone.

As we embark on the next chapter, filled with ambitious goals and exciting possibilities, we do so with the confidence that comes from knowing we have you by our side. Thank you for being a part of this remarkable journey. Together, we will rewrite the narrative of rare and undiagnosed diseases, one life at a time.

With deepest gratitude,

Sue Baker
Program Director

Clinical Centre of Expertise for
Rare and Undiagnosed Diseases

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