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

Clinical Centre of Expertise for
Rare and Undiagnosed Diseases

Rare Care Centre

First Year Impact Report

Update for Philanthropic Funders

Feb 2022-2023

Contents

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.

Welcome to the Rare Care Centre

We aspire to improve the lives of children and their families living with rare and undiagnosed diseases in compassionate, sustainable and equitable ways.

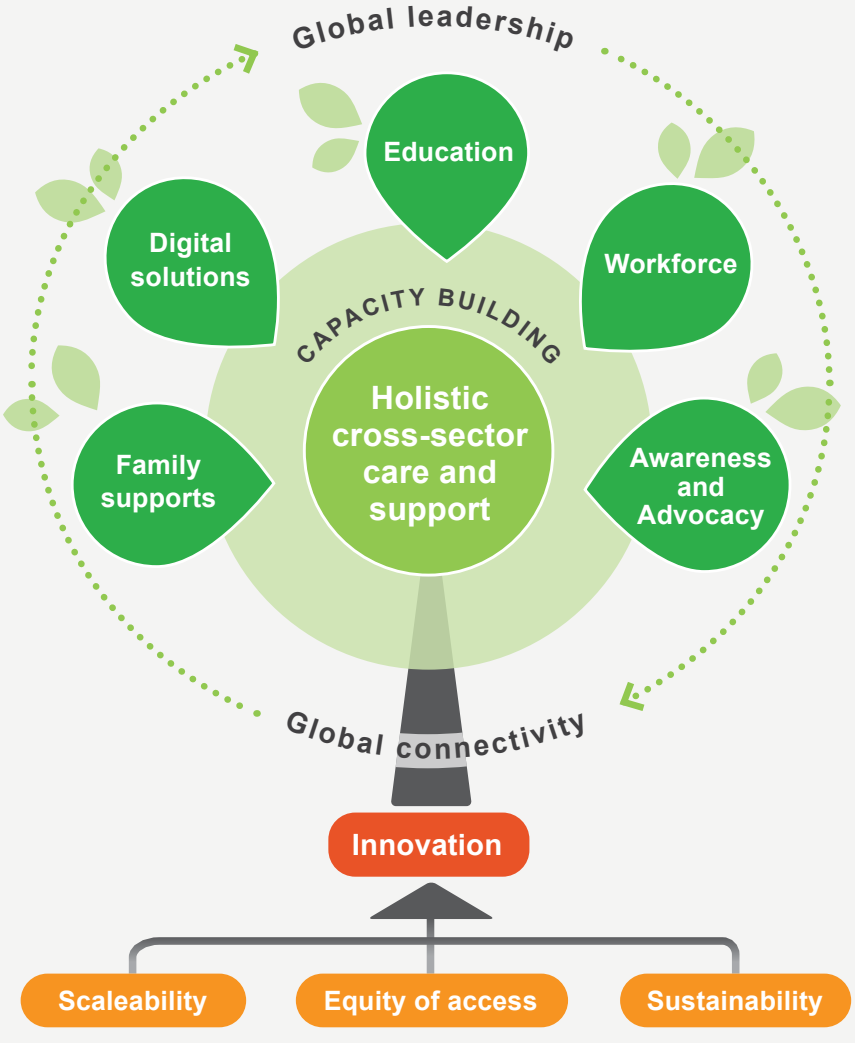
We know that rare and undiagnosed disease care is more effective if managed in centralised centres of expertise. Centralised centres offer a unique opportunity from which knowledge, expertise, capacity, and enhanced capability for identifying, managing and treating rare and undiagnosed diseases can be coordinated. In our first year the Clinical Centre of Expertise for Rare and Undiagnosed Diseases, known as the Rare Care Centre has developed a world first transformative, co-designed cross-sector Model of Care, which helps children and families dealing with rare and undiagnosed diseases to live their best lives possible. By providing holistic care and support in an integrated and responsive service, we put choice and control back into the hands of families.

We act locally and think globally. Supported by innovative digital solutions, new educational paradigms, family support programs, global professional networks, new clinical research infrastructure and policy and legislative change, our model of care is a game changer, and is fast becoming an international template.

Our operational framework looks to the future by prioritising the need to build capacity through education, workforce development, awareness raising and advocacy.

Rare Care Centre
Framework

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



"For the families of children living with a rare or undiagnosed disease, knowing there is a dedicated centre that aims to provide a comprehensive and coordinated treatment plan and deliver access to a global network of specialists and families living with the same diagnosis, is a game changer."

Valerie Jovanovic, Chief Executive, Child and Adolescent Health Service

We are proud to receive validation from industry and the highest calibre scientific leaders across the world. However, nothing is more motivating, and more grounding, than the voices of our families.

"I have hope now" - These words spoken by the mother of the first child seen through the Rare Care Centre, continue to drive us forward to improve the clinical outcomes and quality of life for children and families living with rare and undiagnosed diseases.

The rare disease community has played a critical role in our first year, co-designing the service and providing support. We are so grateful for the gracious and generous input from all the inspiring health professionals across the breadth of specialities at the Child and Adolescent Health Service, as well as the valuable advice and support from leaders across education, community services, disability, mental health, Aboriginal health and refugee health sectors. We are equally grateful to our public, private and philanthropic partners. The establishment of the Rare Care Centre would not have been possible without the catalytic philanthropic support of 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.

Our Vision

Children with rare and undiagnosed diseases and their families live the best lives possible.

Our Mission

We exist to support children living with rare and undiagnosed diseases and their families to identify their unmet needs, navigate to the right care, and coordinate that care to reduce the burden on children, parents and carers.

Our Guiding Principles

Established by the Rare Care team, these guiding principles underpin our decision making, shape the way we deliver all services and functions and provide the benchmark for how we interact with all families, partners, colleagues and stakeholders.

1. Adding value to the lives of children and families is at the forefront of all that we do.
2. Care will be delivered with compassion, imagination and a focus on equity, to leave no one behind.
3. Care will be delivered in a culturally safe and responsive way.
4. Our services will be enabled through trusted partnerships with aligned organisations and service providers.
5. We will respect ancient and old ways and embrace new and innovative ways to create knowledge.
6. We will be kind to each other and to all with whom we share a journey.

Our Strategic Objectives

1 **Care Services**

To provide a state-wide holistic care model that complements existing services to address unmet needs for children with rare and undiagnosed diseases and their families.

2 **Stakeholder Consultation**

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

3 **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.

4 **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.

5 **Integrated Digital Technology and Innovation**

To integrate existing digital technologies and facilitate the development and translation of enabling technologies.

6 **Enduring Partnership**

To foster enduring strategic partnerships.

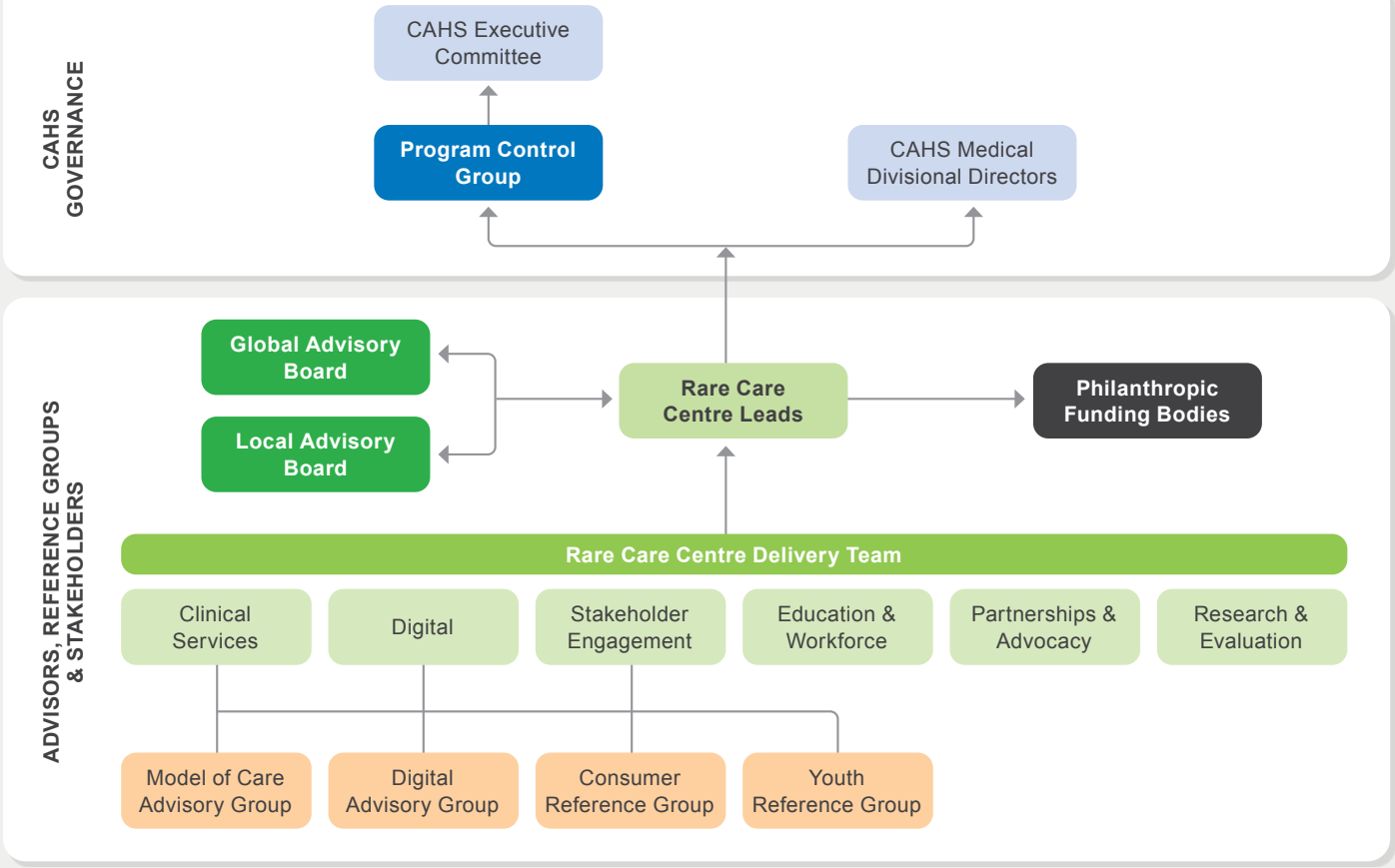
Governance and Management

The Rare Care Centre is governed by the Rare Care Program Control Group, a cross-sector strategic group that provides oversight of the Centre's development and implementation and ensures effective cross-sector integration. Meeting monthly, the Group functions under the authority of the Child and Adolescent Health Service Executive Committee with oversight from the Medicine Division Co-Directors and Executive Director of Perth Children's Hospital and Neonatology.

Several groups of expert stakeholders who connect through formal and informal mechanisms are also available to the Rare Care Centre. These include advisory boards, reference groups and technical advisory groups including:

- **Model of Care Advisory Group:** A cross-sector operational group that provides advice and input into the development of the model of care and the implementation of the clinical service. Sectors involved include Perth Children's Hospital clinical specialties, Aboriginal health, refugee health, primary care, NDIS, Department of Communities and Department of Education.
- **Digital Advisory Group:** A cross-sector operational group with clinical and technical expertise that provides advice on the development of the digital solutions and devices work undertaken by the Rare Care Centre.
- **Consumer Reference Group:** A group of parents/carers with lived experience of caring for a child with a rare or undiagnosed disease in WA and who have informal links with other families. The group provides feedback and input on all activities of the Centre and ensures we are acting in the interests of the families we support.
- **Youth Reference Group:** A group of young people aged between 12-17 who have a rare or undiagnosed disease and have informal links with other young people in WA. The group provides feedback and guidance on all activities of the Centre and ensures we keep patients' voices at the forefront.
- **Local Advisory Board:** A group of WA based local leaders from various sectors who provide strategic advice specific to the WA context on a semi-regular basis.
- **Global Advisory Board:** A high-level group of world leaders in rare and undiagnosed disease who provide strategic advice, early insights and assistance with international partnerships on a semi-regular basis.

Rare Care Governance and Stakeholder Overview



The Centre is co-led by Dr Gareth Baynam and Sue Baker.

Dr Gareth Baynam is the Medical Director for the Rare Care Centre. A clinical geneticist, intrapreneur, digital health expert and professor working in Western Australia, Gareth has a long-standing commitment to health, education, social system transformation and improved Indigenous health care.

Gareth works to develop and deliver innovative, high-value and sustainable rare and undiagnosed disease healthcare through public-private-philanthropic and cross-sector partnerships with all stakeholders including Aboriginal health leaders and the community. He was awarded the WA Health Minister's Award for his contribution to the health of the WA community. Among current activities, Gareth is a practising Clinical Geneticist, a Clinical Genomics Policy Advisor at WA Health, the Director of the Undiagnosed Diseases Program, and a Chair of the Interdisciplinary Scientific Committee of the International Rare Diseases Research Consortium.

Sue Baker is the Program Director for the Rare Care Centre. As a senior nurse leader with 40 years' experience in children's health care services across both the UK and Western Australia, Sue has been at the forefront of innovative service design, implementation and evaluation, particularly for vulnerable cohorts of children and families. Having held a variety of Director and Executive level positions in WA, Sue has extensive experience in leading complex projects and successfully securing research and innovation grants and funding. Sue has received multiple health awards including the prestigious honour of the WA Nurse/Midwife of the Year and holds key positions on global rare disease organisation advisory groups.

Our Organisation

The Rare Care Centre is part of the WA Government Department of Health's Child and Adolescent Health Service and is based at the Perth Children's Hospital. The Child and Adolescent Health Service is the leading service provider for paediatric health care in WA, and is made up of four service areas: Neonatology, Community Health, Child and Adolescence Mental Health Services and Perth Children's Hospital.

As WA's dedicated paediatric hospital, Perth Children's Hospital delivers emergency, inpatient, ambulatory and outpatient services, state-wide outreach and mental health services, neonatology intensive care, and the state's only paediatric trauma centre. A centre of excellence for teaching and research they partner in major paediatric research and education initiatives with Telethon Kids Institute and the State's universities.

"The more we connect with the Rare Care Centre, the more we realise that it is a Centre of Innovation."

Chief Technical Officer of Microsoft Australia and New Zealand

Living with Rare and Undiagnosed Diseases

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

Rare and undiagnosed diseases are one of the biggest killers of children. The mortality rate is 13 times higher than for children with all common diseases combined.

In WA there are 63,000 children with rare and undiagnosed diseases. On average it takes between five and seven years for a child with a rare disease to get an accurate diagnosis with many being misdiagnosed along the way, sometimes multiple times. This is one of the reasons that rare and undiagnosed diseases represent the highest cost to our health system.

A rare disease is defined as a medical condition with a specific pattern of clinical signs, symptoms and findings that affect less than 1 in 2,000 people. There are more than 8,000 different rare diseases with some currently being undiagnosable. Some people are suspected of having a rare disease due to their symptoms, however remain undiagnosed.

While there are clinical differences between rare diseases, when we look at them together there are many common challenges and experiences for children and families living with rare and undiagnosed diseases. In addition to the challenges experienced by families of children with medical complexity with or without disability, families tell their story many times, to many different teams on multiple occasions. They also navigate multiple complex sectors and systems such as health, education, community services, NDIS, and social services in their search for appropriate care and management. Children and families living with rare and undiagnosed diseases have additional challenges that are specific to the rarity, which can include:

- Parents are often the expert in their child's disease
- There is a long journey to receive a diagnosis
- They experience multiple misdiagnoses and inappropriate clinical management
- There is a lack of treatment options
- They experience isolation, stigma and discrimination

Prior to the Rare Care Centre being established, many WA families coped alone with the immense burden created by these challenges.

Our Work

Gareth Baynam

Medical Director

“The globally connected Rare Care Centre provides a unique opportunity to ensure children across WA receive the very best care and support that they truly deserve. Together, guided by the local and global voices and strength of children and families living with rare and undiagnosed diseases, the Rare Care Centre team have started a partnership journey, a transformation. Listening to those voices and empowered by our philanthropic partners, our colleagues, and the cumulative experience and wisdom of all those we have consulted within Health, across other systems and in the NGO sector, we have succeeded in achieving a key step of the delivery of our clinical service.”

Sue Baker

Program Director

“We have seen the rippling effect of the United Nations (UN) Resolution on Rare Diseases and the formation of the Global Network for Rare Diseases in December 2021; a partnership between the World Health Organisation (WHO) and Rare Disease International (RDI). We are building trust through delivering tangible outcomes at speed to compassionately address the unmet needs of children and families living with rare and undiagnosed diseases. We feel such gratitude to be part of the Rare Care Team, and to have the opportunity to learn, contribute and serve the needs of children and families living with rare diseases.”

Care and Support

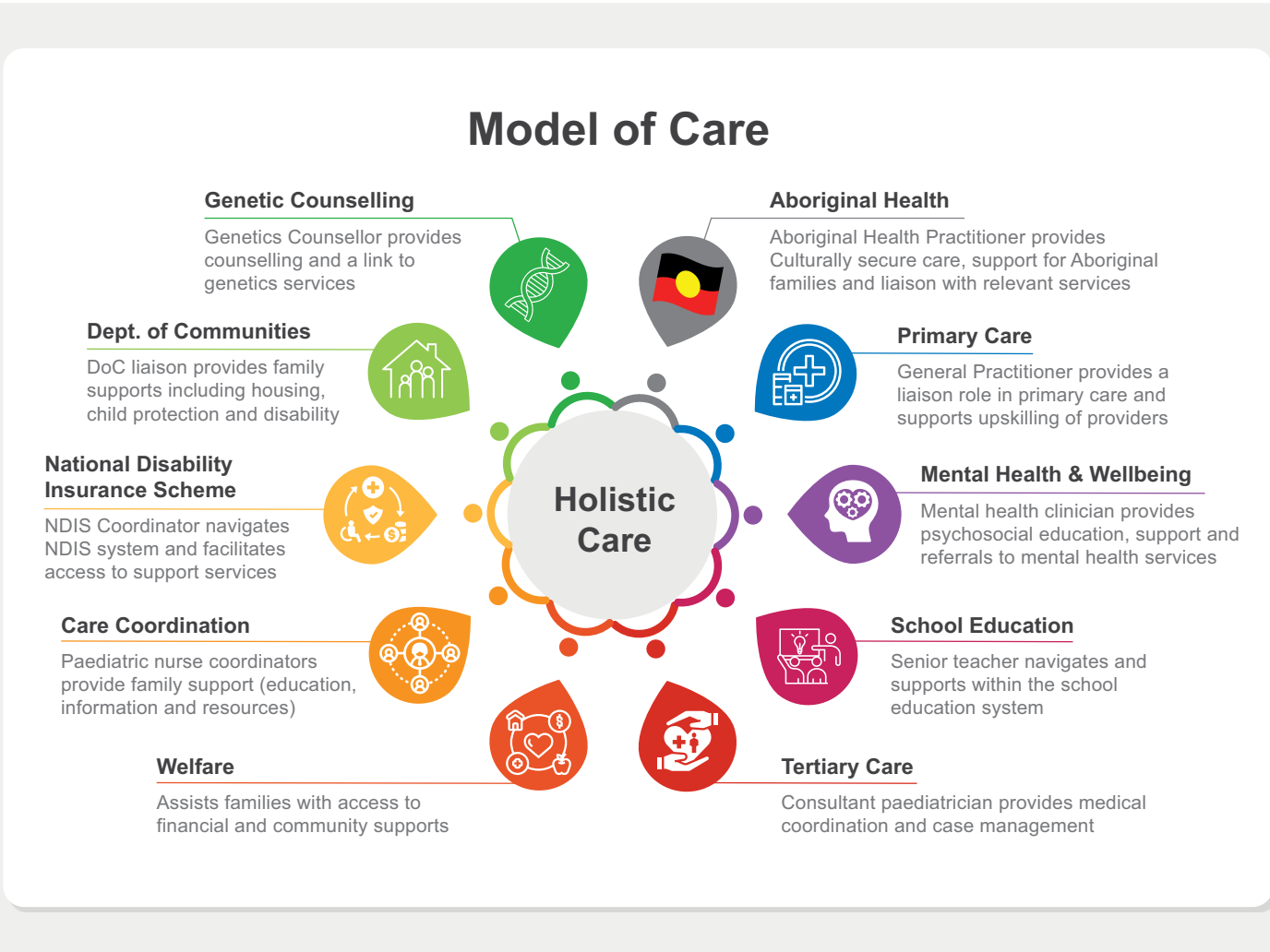
We provide care and support for children and families living with rare and undiagnosed diseases, aiming to empower them to live their best lives possible. Our work involves improving support and care coordination, enabling community resources, and accessing clinical trials and research.

Our focus is to deliver care that is compassionate, equitable, holistic and culturally secure. In the first year we have collaborated with existing Aboriginal and Non-Aboriginal community and public-private partnerships, initiatives and services, to tailor our Model of Care to Aboriginal health, mental health and regional and remote health care.

By partnering with existing services, we have concentrated on collectively addressing the unmet needs of children and families through aligning pathways, simplifying systems, informing policies and growing and integrating technologies.

Model of Care

Children and families living with rare and undiagnosed diseases have multidimensional needs that impact not only their physical and mental health, but every aspect of their lives, including education, finances, social activities and employment. In the first year we prioritised developing a novel Model of Care that reflects their unique, complex and multi-sector needs.



Our process to design the Model of Care included identifying key areas of concern, gaining a deeper understanding of children and families' experience and needs, leveraging internal and external expertise at both operational and strategic levels and undertaking extensive consultation locally, nationally and globally. To ensure the voice of children and families is always at the heart of everything we do, we partnered with our Consumer Reference Group and Youth Reference Group, both of which contributed valuable insights, advising us what would and would not work for them.

Our consultation told us that families who are engaged with various services across multiple sectors have difficulties navigating and accessing the right resource or service at the right time. Services that families often access include education, health including mental health, primary care and genetics services, NDIS and community services. We also found that for some families, particularly those from culturally and linguistically diverse communities, knowledge of available resources and services is lacking.

The Rare Care Centre approach is to bring key sectors together to form one team that families can access. We facilitate the sectors interacting directly with each other, through the right contact point, using the right language, at the right time, bridging gaps that previously existed.

The Model of Care is a state-wide, cross-sector care coordination service that has been designed to reduce the amount of time families spend searching for the right assistance to meet their needs, as well as increase their awareness of, and access to services available to them.

The Model adds value without duplicating existing services. It provides navigation, coordination, integration and advocacy support mechanisms, and accommodates children with both high and low complexity. To deliver the cross-sector service, professionals from each sector were employed, including:

- Consultant Paediatrician
- Clinical Nurse Consultant
- Paediatric Nurses
- General Practitioner
- Genetics Counsellor
- Mental Health Clinical Nurse Specialist
- Aboriginal Health Worker
- Senior School Teacher
- NDIS Support Coordinator
- Welfare Officer
- Department of Communities Coordinator

The Model of Care has been developed within the Department of Health's Activity Based Funding Framework which focuses on the patient, recognises the complexity of their needs and provides appropriate funding, enabling sustainability and scalability.

In recognition that families accessing the Rare Care Centre spend significant time in hospital, we partnered with North Metropolitan TAFE – Roaming Education and Community Health (REACH) which provides an offsite space to run outpatient clinics. While providing a setting that is easier to access and more comfortable for families, it also delivers opportunities to increase the knowledge and experience of the next generation of healthcare providers as our nursing team provides supervision and mentorship for enrolled nursing students.

Digital Solutions and Devices

Globally the emergence of a new wave of healthcare innovations based on digital technology has raised expectations that systems will be able to offer more effective, efficient, and personalised services. The Rare Care Centre has leveraged this shift to drive innovation through digital solutions and devices that are scalable and sustainable. These will create value by improving the quality of life and clinical outcomes for children and families living with rare and undiagnosed diseases in the local, national and global arena.

"Your initiatives in Western Australia serve as a model for the world, especially in working with rare and Indigenous peoples."

Dr William (Bill) Gahl, Director of the National Human Genome Research Institute, USA National Institutes of Health

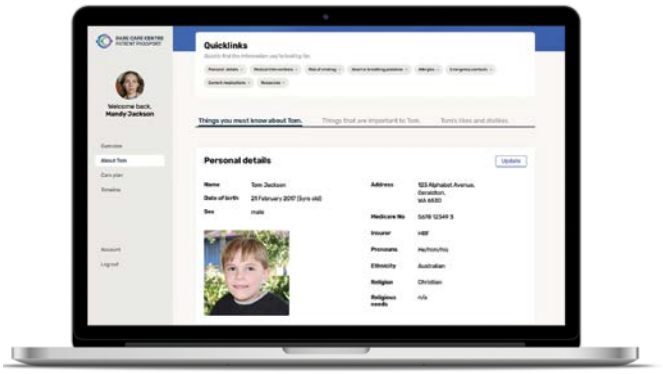
In the first year we prioritised integrating new and existing digital technologies into our care workflow. We have also established a Digital Advisory Group comprising clinical and technical experts from both public and private service providers across multiple care sectors. Our Medical Director is leading the Data, Digital, and Technology Taskforce for the World Health Organisation Global Network for Rare and Undiagnosed Diseases. This position further provides us with a global connection for collaboration, sharing of solutions, and generating new knowledge.

Three key innovative digital projects have commenced this year and are currently in development.

UTOPIA

UTOPIA (Unlocking Treatment Options, Personalised In-Time Access) is a crucial component of our digital solutions. Aiming to deliver new insights and improve clinical outcomes, UTOPIA connects a variety of existing health data sources to provide shared understanding and integrated access. The first product of UTOPIA will be a semi-automated comprehensive patient summary. Summaries are unique to each patient and are pre-populated from recognised data sources to create an efficient care summary for children and families, making it easier for them to share their care story.

In our first year we have completed the scoping for the UTOPIA platform, developed governance, and sought approvals for its development and implementation. Receiving approval of the UTOPIA Concept Approval Request from Health Support Services was a key milestone and UTOPIA was selected as one of the top five digital priorities for WA Department of Health.



Rare Diseases Patient Portal

Following community co-design, a proof of concept has been developed for a digital Patient Portal. The Portal aims to provide a central source of truth accessible by both families and care professionals, which enables exchange of clinical and support information across sectors (such as health, education, communities etc) in real-time. Importantly the portal ensures the voices of the child and family are highlighted. We have also commenced the process of seeking potential funders.

Virtual Digital Twin Project

A partnership with the Hub for Immersive Visualisation and eResearch (HIVE), Curtin University and SingHealth will create 'Digital Twins' from multiple types of imaging data, including ultrasound, CT, and MRI from people with rare and undiagnosed diseases. These will be presented in an interactive digital learning environment (virtual reality or augmented reality) for improved education of healthcare professionals as well as increased engagement with children and families. Clinicians will be able to present and discuss results in a tangible and personalised way. The first stage of visualisation has been completed.

Education and Workforce Capacity

The Rare Care Centre has developed and partnered in an array of education initiatives to build capacity and increase knowledge and awareness of rare and undiagnosed diseases across the healthcare workforce. These programs and resources also provide information on the challenges of living with rare and undiagnosed diseases and ways to improve care for children and families. All programs are designed to be adapted and scaled up to suit national and global organisations for future sustainability across rare and undiagnosed disease education.

The Rare Care Centre has a focus on upskilling GPs as they are often the first medical contact point for a family. The first initiative targeting GPs was updating the HealthPathways resource provided by the Western Australian Primary Health Alliance, which assists GPs in early diagnosis and appropriate referral pathways for the family, as well as providing links to resources that both the GP and family can utilise in disease management. The updated pathway was launched with an education webinar covering the importance of rare and undiagnosed disease awareness and providing appropriate support in how to effectively navigate the pathway. This resource is being adopted nationally and is under consideration for implementation in New Zealand. The second initiative has been formal accreditation of the Rare Care Centre as a GP training provider. Working with WA General Practice Education and Training, we are already educating GP trainees in WA providing clinical supervision specific to rare and undiagnosed diseases in an extended skills placement.

As recognised experts in rare and undiagnosed diseases, the Rare Care Centre has received multiple invitations to provide education to other professional bodies such as presenting an educational webinar for Palliative Care Australia which was attended by 286 healthcare professionals globally, and viewed over 250 times on YouTube.

"The RArEST project has benefitted immensely from the Rare Care Centre's specialist knowledge and networks. Beyond their medical expertise, we have learned from their genuine appreciation for the experiences of people living with rare disease. Their generous involvement means that health professionals across Australia will be better equipped to deliver the best possible care."

Dr Elizabeth (Emma) Palmer, Clinical Geneticist and Co-Lead of RArEST

Education Partnerships

The Centre is now a key part of the growing national and global network of organisations working to build capacity and educate to better the lives of children and families living with rare and undiagnosed diseases.

We are proud to play a pivotal role in a Commonwealth funded project called the *Rare Disease Awareness, Education, Support and Training* Project or RArEST, a collaboration between Australia's Peak Advocacy Group for rare diseases – Rare Voices Australia, University of New South Wales, University of Western Australia, Macquarie University and the Rare Care Centre. It has recently launched a Rare Disease Project ECHO which is an internationally validated framework and methodology that develops skilled communities of practice to connect and empower healthcare workers. To date five sessions have been provided on a range of topics relevant to rare diseases. These sessions were very successful with over 101 people registered, 44 of whom were from WA. Initially aiming to be national, the Rare Disease Project ECHO is rapidly expanding into the international arena. This style of education and connection is a catalyst for knowledge sharing and supporting better recognition and outcomes for children and families living with rare and undiagnosed diseases.

Working with RArEST we are also co-authoring a national recommendation document specifically for children and their families, which represents the first step in nationally standardising the approach for rare and undiagnosed disease management. We were able to contribute learnings from our cross-sector Model of Care, which represents best-practice care coordination for children and families living with rare and undiagnosed diseases in a novel, co-located, cross-sector team approach.

RArEST has enabled other educational collaborations such as working with Medics for Rare Disease (M4RD), a leading education organisation in the UK, providing expertise and advice in reviewing and adapting learning e-modules for the Australian context. These modules for healthcare professionals are free, interactive and designed with adult learning principles. We are also focused on working with M4RD to build the knowledge and skills of the future medical workforce in rare and undiagnosed disease.

Future Workforce

Recognising that future healthcare workers are a vital part of building awareness and capacity within the healthcare system we have undertaken the following initiatives.

- Partnered with the Department of Health Graduate Development Program and the University of Western Australia by providing placements for a Master of Public Health student and Graduate Development Program Officer in 2022.
- Hosted a booth at the UWA Medical Student Scholarly Activities expo, resulting in 12 students registering and being accepted to complete their service-learning unit in the Rare Care Centre in 2023.
- Hosted five Colombo scholarship recipients in collaboration with Curtin University, Curtin Singapore and SingHealth. Each student was designated a project that was designed to utilise an innovative technology to assist healthcare professionals to process and handle rare disease patient data more effectively to improve patient outcomes. These projects are now being expanded together with leading colleagues from Singapore's national precision medicine project (PRECISE).

- A range of new partnerships with Edith Cowan University are also in progress, including through their Centre for Precision Health that has identified rare diseases as a key expansion focus.
- Development of an online international medical student forum in partnership with M4RD to promote learning, resource development and exchange between medical students and rare and undiagnosed disease experts is underway. Members of this group are drafting a white paper on the importance of rare and undiagnosed education and how they sought this education outside their curriculums. Currently there are members from Australia, the United Kingdom, Ghana, and China.
- The Rare Care Centre is also one of four groups that have partnered to create a novel training accreditation for specialisation in rare and undiagnosed diseases, under the European Society of Medical Specialties. The five-year course (Competency) is recognised formally throughout Europe and the USA and is available for doctors globally. The syllabus, organisational accreditation package, governance and examination structures are complete, with final exam content as the last stage near completion.

"The Rare Care Centre demonstrates a commitment to providing inclusive, excellent and joined up care for all those impacted by rare conditions. Around the world families are crying out for the same fundamental things from their healthcare systems: joined up and compassionate care. And the team behind the Rare Care Centre are listening."

Dr Lucy Mckay, CEO Medics for Rare Disease

Advocacy and Awareness

Our advocacy for children and families living with rare and undiagnosed diseases works on several levels. Integral to addressing their unmet needs is the ability of the Rare Care Centre team to individually advocate on the behalf of families with a range of services such as NDIS, education, welfare agencies and other community services.

By building partnerships, raising awareness, creating new or modifying existing policies and creating or enhancing referral pathways we also advocate for long term systemic changes within and between services and sectors (e.g. health, education, disability) to enable recognition and improved outcomes for children and their families via improved support, care, clinical trials and research.

The Rare Care Centre has forged links with many rare and undiagnosed diseases groups worldwide. These relationships ensure a broader advocacy so the collective voices of children and families living with rare and undiagnosed diseases in Western Australia are heard nationally and globally.

A key advocacy event was the *"Parliamentary Friends of Rare and Undiagnosed Disease"* where we informed WA State politicians on the impact that rare and undiagnosed diseases have on children and families, and the common challenges they face. Over 70 Members of Parliament attended including the State Premier Mark McGowan.

Medical Director Gareth Baynam was a keynote speaker at the 2022 national Rare Voices Australia Summit where he presented on the impact of the Centre and our Model of Care. Through this advocacy a new partnership was established with legal experts working pro bono to scope existing national policies and legislation pertinent to meeting the needs of people and families living with rare and undiagnosed diseases.

Alongside our advocacy and awareness building, a strong focus for the Rare Care Centre is to strengthen collaborations that share knowledge and create improvements in health and community services, locally, nationally and internationally. In our first year, the Rare Care Centre's Directors have forged forward with this priority by building new and nurturing existing partnerships. More than 183 meetings with multiple stakeholders including 16 key Non-Government Organisations were held. Of these organisations and key leaders, 106 were local, 34 were national, and 43 represented global connections.

Leadership Roles

The Rare Care Centre is affiliated with over 50 rare and undiagnosed diseases committees, advisory boards, working groups and taskforces in various capacities at Founder, Chair, Director, Advisor, and Board Member levels. Medical Director Dr Gareth Baynam has extensive leadership roles across local, national and global organisations including:

- Chair of the International Rare Disease Research Consortium (IRDiRC) Interdisciplinary Scientific Committee
- Chair, Functional Analysis Taskforce, IRDiRC
- Director, Undiagnosed Disease Network International (UDNI)
- Member of the WA Department of Health High Cost Highly Specialised Cell and Gene Therapies Clinical Program Steering Committee
- Country Coordinator for the Orphanet Advisory Board
- Member of the Advisory Board for Edith Cowan University Centre for Precision Health
- Mentor for the Australian Clinical Entrepreneur Program
- Invitation to contribute on Indigenous aspects of health equity planning for the New Zealand Government

Program Director, Sue Baker has been offered key positions on:

- Global Panel of Experts, WHO Global Collaborative Network for Rare Disease
- Framework to assess impacts associated with diagnosis, treatment, support, and community integration Task Force, IRDiRC

Knowledge Sharing

Presentations and Publications

We also have an ongoing commitment to increase understanding of rare and undiagnosed diseases within diverse audiences via roundtables, research, presenting at seminars, conferences or symposiums and co-authoring publications. In our first year, over 25 presentations were delivered and over 15 publications are in print, with more in progress.

- MC Palliative Care Australia Event – *The Diagnostic Odyssey of Genetic Diseases, Family perspectives on genetic testing.*
- Publication in Brief (WA's legal magazine): *Is it time for a Rare Diseases Act?*
- Baker S and Baynam G. (2022): *Towards a Global Nursing Network for Rare and Undiagnosed Diseases.* Poster presentation 11th International Conference on Undiagnosed Diseases.
- Final stage draft of a framework for Aboriginal genetic health care for the Australian Department of Health.
- Economist Impact: *Connecting the Dots: Embedding Progress on Rare Disease into Healthcare.*
- Invited submission for WHO Global Scientific Committee on equitable genomic healthcare.
- Digital Twin – Science on the Swan abstract accepted.
- World Health Assembly, Geneva.
- *Stigma and Rare Diseases – A call to Global Action and Towards a Global Nursing Network for Rare and Undiagnosed Diseases* abstracts and posters - 11th International Conference on Undiagnosed Diseases, Vienna
- Dr Gareth Baynam was a keynote speaker at the 2022 Rare Voices Australia Summit presenting the Model of Care and impact of the Centre

Journal Publications

- PubMed (nih.gov) Position statement: *Rare disease education in Europe and beyond: time to act.* 2022
- PubMed (nih.gov): *mEDUrare: Supporting Integrated Care for Rare Diseases by Better Connecting Health and Education Through Policy.* 2021
- PubMed (nih.gov): *Childhood rare diseases and the UN convention on the rights of the child.* 2021
- PubMed (nih.gov) Manuscript *3D Facial Analysis for Rare Disease Diagnosis and Treatment Monitoring: Proof-Of-Concept plan for Hereditary Angioedema.* 2023
- *A parent first: Exploring the support needs of parents caring for a child with medical complexity in Australia.* Among authors: **Theresa Abbott**, **Sue Baker**, (2022). Journal of Pediatric Nursing 2022 YJPDN-0290.
- *Further evidence for distinct traits associated with RBM10 missense variants*. Poulton C, et al. Among authors: **Baynam G.** Clin Genet. 2022. PMID: 35645043
- *Medical Comorbidities in MECP2 Duplication Syndrome: Results from the International MECP2 Duplication Database*. Ta D, et al. Among authors: **Baynam G.** Children (Basel). 2022. PMID: 35626810
- *Unlocking sociocultural and community factors for the global adoption of genomic medicine*. Chediak L, et al. Among authors: **Baynam G.** Orphanet J Rare Dis. 2022. PMID: 35549752
- *Common data elements to standardize genomics studies in cerebral palsy*. Wilson YA, et al. Among authors: **Baynam G.** Dev Med Child Neurol. 2022. PMID: 35441707

- *Association between craniofacial anomalies, intellectual disability and autism spectrum disorder: Western Australian population-based study*. Junaid M, et al. Among authors: **Baynam G.** Pediatr Res. 2022. PMID: 35352007
- *A brief history of MECP2 duplication syndrome: 20-years of clinical understanding*. Ta D, et al. Among authors: **Baynam G.** Orphanet J Rare Dis. 2022. PMID: 35313898
- *Functional validation of variants of unknown significance using CRISPR gene editing and transcriptomics: A Kleefstra syndrome case study*. Fear VS, et al. Among authors: **Baynam G.** Gene. 2022. PMID: 35176430
- *CRISPR single base editing, neuronal disease modelling and functional genomics for genetic variant analysis: pipeline validation using Kleefstra syndrome EHMT1 haploinsufficiency*. Fear VS, et al. Among authors: **Baynam G.** Stem Cell Res Ther. 2022. PMID: 35139903
- *Epidemiology of Hospital Admissions for Craniosynostosis in Australia: A Population-Based Study*. Junaid M, et al. Among authors: **Baynam G.** Cleft Palate Craniofac J. 2022. PMID: 3513007
- *Research on rare diseases: ten years of progress and challenges at IRDiRC*. Monaco L, et al. Among authors: **Baynam G.** Nat Rev Drug Discov. 2022. PMID: 3507916

Our impact is being felt further afield as an increasing number of local, national and international organisations have requested letters of support from the Rare Care Centre on applications for funded research. In our first year these have included:

- Centre for Population Genomics for Australian Genetic Diversity Database "OurDNA" project
- The Healthy Strides Foundation – To support the healthy strides foundation for Channel 7 Telethon trust Research Grant for the project “The Power of One”, personalised allied health interventions for rare and neurological disorders
- Kalparrin – Telethon Grant Application
- Murdoch University – Channel 7 Telethon Foundation Project – Treatments for Those Who Have None – extended platform
- Jenny Downs – Application for Fellowship ‘A knowledge to practice roadmap for improving quality of life in children with disability’
- Rocky Bay – Early start prevention program
- National Computational Infrastructure and the Australian National University
- Genome Canada for EpiSign International: Health System Impact Assessment and Expanding Clinical Utilization of Epi/Genomic Testing in Rare Disease and Beyond
- Rare Disease Wales – Rare Disease Digital Health and Wellness Community (Dialect) Node: Speaking the language of rare disease
- Rare Voices for Primary Health Care Quality and Coordination Rare and Complex Disease Telehealth Nurse Program Grant Opportunity

The Centre is being recognised as a global exemplar with many enquiries and requests for information. In our first year the Model and related suite of Centre corporate documents have been requested and shared 11 times locally and nationally, and 19 times internationally over five continents. The Rare Care Centre was also the subject of the keynote presentation of the International Rare Diseases Meeting, REACT in Berlin in 2023.

Our Impact

"I feel different now that I have come here, because this is the first time that anyone has looked at my experience holistically. I felt valued and listened to by Rare Care".

Measuring Our Impact

Continuous evaluation and quality improvement are key to ensuring the Rare Care Centre is delivering high value healthcare, positively impacting children and families, and having a broader global impact. The Centre Evaluation Framework outlines two distinct components: service evaluation and impact evaluation. Service evaluation includes assessment of the interventions delivered through the cross-sector Model of Care, as well as measuring the outcomes of other capacity building areas including education and workforce capacity, digital solutions, research, advocacy, and partnerships. Impact evaluation includes assessing benefits for children and families living with rare and undiagnosed diseases, economic impact and workforce design. By establishing evaluation frameworks within the Model of Care, we utilise continuous quality improvement to achieve best practice alongside positive outcomes for children and families.

To evaluate our impact on children and families we utilise two validated Patient Reported Outcomes Measures and have designed a bespoke Patient Reported Experience Measure in the form of a family survey. Data from our clinical service is captured in a custom-made REDCap (electronic data capture system) form which has also been approved as a clinical registry.

We have partnered with Western Australian academic institutions and non-Government rare diseases oganisations to ensure the evaluation is meaningful. Working with the Future of Work Institute at Curtin University, the Rare Care Centre has secured a PhD student to evaluate the workforce design of the Centre given the innovative cross-sector team model. A cost benefit analysis of the Centre will also be conducted by an external economics expert.

Impact on Children and Families

Over our first year, the Rare Care team has worked holistically alongside children and their families to enable a range of positive outcomes. For our families who have carried such a large burden for a long time, the impact of these outcomes is huge. Examples of these outcomes include:

- Enrolment and attendance at school with full-time special needs education support
- Connected families to local GPs to support coordinated care closer to home
- Facilitated transition to adult services and employment opportunities
- Secured increased NDIS core funding to ease the financial burden
- Enabled families to fully access appropriate supports with their NDIS package

- Advocated for in-home monitoring devices allowing parents to sleep and improve their wellbeing
- Provided wellbeing education and support for families and improved access to appropriate mental health services
- Improved access to genetic counselling services
- Coordinated outpatient appointments to reduce hospital attendances
- Facilitated access to education support services outside of school
- Provided visual summaries of information on the child's rare or undiagnosed disease to upskill school staff, medical staff, carers and community therapy providers
- Facilitated access to specialist medical care and therapy support

Global Impact

Meeting the needs of people and families living with rare and undiagnosed diseases is a global public health priority, and critical in ensuring medical, social, and human rights are met. The World Health Organisation, European Commission, World Economic Forum, United Nations and Australian and international peak rare diseases advocacy groups recognise globally networked Centres of Expertise for Rare Diseases as an urgent high-need priority.

The need to better support children and families living with rare and undiagnosed diseases has been recognised in WA, following the recommendation from the Ministerial Council for Precision Health. The Council was established to advise the State Government on opportunities to further develop and support precision health advances in the state. From all proposals within the entire WA health system, the Clinical Centre of Expertise for Rare Diseases (the Rare Care Centre) was ranked as the number one initiative.

Family Story

One of the earliest families to access the Rare Care Centre described several years of repeating their story in multiple consultations and communications with 36 different professionals from a variety of health, education, primary care and NDIS services. The family has 14 teams involved in their care and support at Perth Children's Hospital alone, with no designated care coordinator. The number of appointments with various health teams was extremely high, and with many on sequential days this required multiple trips to hospital, time off work for the parents and interruptions to schooling. The disease is so rare that it is not included for automatic eligibility under the NDIS.

The Rare Care team enabled and empowered many changes which resulted in the following positive outcomes for the family:

- The Nursing team significantly reduced the number of both scheduled and subsequently missed appointments through rescheduling coordination.
- Led by the nursing team, collaboration between health teams helped to coordinate appointments and admissions to appropriate times that suited the family and avoided missing school and therapy appointments.
- The cross-sector team, led by the Genetics Counsellor, developed a visual summary of the child's rare disease and story, the medical complexities and educational implications of the disease, which could be used by medical teams, education staff, community therapists and in-home support providers. The visual summary conveyed key information in an easy-to-understand format and reduced the need for the family to continually repeat their story.
- The Paediatrician facilitated access to a developmental assessment resulting in a successful enrolment in a school with appropriate funding.
- The Senior Teacher developed individual school plans that supported school transition, maintained engagement and minimised disruption during planned surgical admissions.
- The Senior Teacher also facilitated a case conference to school staff as required and provided family tours of school options in their local area to help them make a decision on where to enrol.
- NDIS providers coordinated therapy goals and strategies with the child's educational plans.
- An increase in NDIS funding resulted in 10 additional hours of in-home support/week.
- The Paediatrician facilitated a priority referral to Child Developmental Services in the local community to support ongoing management and additional assessments.
- A mental health and wellbeing assessment for the family resulted in linking family members to appropriate community supports and services.

Our Partners

In our first year of operation, priority was given to establishing a range of key partnerships including the following:

- Partnership with peak international organisations: WHO, WEF and UNICEF.
- Official and founding partner in the UEMS Competency for Rare and Undiagnosed Diseases (globally available training for doctors formally acknowledged in the EU and USA).
- Chair of the International Rare Diseases Research Consortium Interdisciplinary Scientific Committee via Medical Director. This consortium represents $3 billion p.a. committed to rare diseases research.
- Improving care for remote region and Aboriginal children living with rare and undiagnosed diseases. Partnering with Roy Hill Community Foundation and their commercial partners in the Pilbara, such as Pilbara Faces (3D facial analysis), Pilbara Voices (Aboriginal translations of medical terms) and Pilbara DNA (access to clinical whole genome sequencing) to build on our world leading initiatives.
- Partnering with the Indian Organization for Rare Diseases and the State Government of Telangana to identify the rare diseases patient population in all cities and villages.
- Partnering with the Undiagnosed Diseases Network International on 'Champions Initiative', partnering rare and undiagnosed disease experts in high income countries with those in low/middle income countries to support capacity building.

Letters of Exchange that partner the Rare Care Centre with leading organisations around the world have the ability to collectively amplify advocacy, support, knowledge sharing and collaboration.

Established Letters of Exchange

Prader-Willi Research Foundation, Australia

Rare Disease Awareness Education Support and Training (RArEST), Australia

Rare Disorders New Zealand, New Zealand

Rare Voices Australia, Australia

Good Sammy Enterprises, Australia

Agrenska Centre, Sweden

Casa dos Raros, Brazil

Rare Disease Nurse Network, UK

In Progress Letters of Exchange

Children's National Hospital, Washington USA

Fudan Children's Hospital, China

KK Women and Children's Hospital, Singapore

Brazilian Paediatric Society, Brazil

Centre for Rare Disease, Newcastle University, UK

Mayo Clinic, USA

Critical Path Institute, USA

Syndromes Without a Name Clinic, Cardiff

University of Cambridge – CONCORD study, UK

Queen Sylvia Hospital, Sweden

Rare Diseases Malta

Genetic Alliance, USA

ISONG, USA

Boston Children's Hospital, Harvard Medical School, USA

Children's National Hospital, Washington USA

Uncommon Cures, Washington USA

Child Development Program, Queensland Health, Australia

Our Future

While we are proud of all we have achieved to date, we are really just getting started. Plans are already in place for many further developments and initiatives.

A significant milestone will be achieved in March 2023 when we partner with Curtin Singapore and SingHealth Duke-NUS Genomic Medicine Centre to host the first global roundtable to connect nurses working in rare and undiagnosed diseases in Singapore. 'Connecting Nurses Globally – A Roundtable in Rare and Undiagnosed Diseases' invites senior dynamic nurses from across the globe to collaborate in the design and implementation of a global nursing network. The group will also lead development of an international curriculum for rare and undiagnosed diseases nursing education – something that does not currently exist. Nurses hold a crucial role at the forefront of care supporting children and families living with rare and undiagnosed diseases and undertaking many roles beyond healthcare provider such as educator and counsellor. In many low to middle-income countries and remote regions, nurses are the only readily accessible health professionals. The Roundtable and network development has been supported by multiple private and philanthropic entities which has enabled us to provide sponsorship funding to nurses who otherwise would not have been able to attend.

Other key initiatives and projects we will be progressing in the upcoming year are:

- Leading the planning of an inaugural Asia-Pacific Consortium for Medical Devices in Rare Diseases with the aim to co-design a framework for the progression of medical devices strategies.
- Expanding our clinical service for children and families.
- Developing pathways to support children and families engaged with the Rare Care Centre to better access services delivered by partner organisations such as carer mental health and supported employment.
- Developing an Aboriginal Health Worker Rare Disease e-module learning package in partnership with the Aboriginal Health Council of Western Australia to upskill the Aboriginal Health Workforce in a culturally secure and responsive way.
- Developing a business case for proposed funding from philanthropic stakeholders for the Pilbara Hub, a satellite site of the Rare Care Centre in the Pilbara to enable children and families with rare and undiagnosed diseases to access locally focused healthcare and support services close to home.

"I have hope now."

Mother of a child with a rare disease

- Progressing the mEDUrare initiative which aims to connect health and education through policy, resources and other practical and sustainable measures. This will include the creation of an educational video for teachers and students showing a school day in the life of a student with a rare disease. This video aims to educate, increase awareness and reduce stigma.
- Developing peer support programs starting with a peer group for fathers.
- Commencing evaluation on the work design of the Rare Care Centre by a PhD student in conjunction with the Future of Work Institute at Curtin University.
- Developing clinical trials infrastructure to support families to be able to access clinical trials and treatments locally in WA.
- Publishing a Model of Care protocol paper to share the development and implementation of the Rare Care Centre Model of Care internationally.

There are many opportunities still to discover and so much more to do. We need the continuing support of our partners, our trusted advisors and specialists and our local communities. Together we can create new knowledge, collaborate, and share solutions globally to enhance the lives of all children and families living with rare and undiagnosed diseases.

Clinical Centre of Expertise for
Rare and Undiagnosed Diseases

Key2creative_43046_05/23