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

**Clinical Centre of Expertise for**
**Rare and Undiagnosed Diseases**

# Rare Care Centre

## Year 4 Impact Report

February 2025-26

# Contents



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

## Using the term Aboriginal

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

## Using the term Rare and Undiagnosed Diseases

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

# Opening Letter

**To Our Esteemed Donors and Partners,**

Every day, we meet children and families living with rare and undiagnosed disease who are navigating uncertainty, complexity and fatigue, often across multiple services and systems. Year 4 has reinforced a simple truth: when care is coordinated, when expertise is connected, and when families are holistically supported, outcomes improve.

This has been a pivotal year for the Rare Care Centre.

The Centre has continued to deliver cross-sector coordinated care while demonstrating measurable system value and growing national and international influence. Families are experiencing clearer pathways and reduced fragmentation. Clinical trial capability has advanced. Research and innovation translation has strengthened. Workforce education has expanded. Our work is increasingly recognised not only for compassion, but for rigour, standards and impact.

Importantly, this impact is being demonstrated in both child and family experience and system performance. Reductions in hospital utilisation, improved coordination across agencies and strengthened disability and education engagement are contributing to better outcomes for children, while also delivering measurable value to the broader health and care systems. Broader community impact is being driven through increased family wellbeing.

Building on the outcomes for children and families delivered through our foundational philanthropic partnerships, a defining milestone of Year 4 was the Stan Perron Charitable Foundation's landmark $221.1 million donation to fund the establishment of the Rare Care Comprehensive Centre, alongside $25 million from Perth Children's Hospital Foundation and $3 million from The University of Western Australia.

This unprecedented commitment is a profound endorsement of the outcomes of the Centre demonstrated to date. It reflects trust in the team, in our partners and in the measurable difference being delivered for children and families across Western Australia, and beyond.

This year also saw strong performance in competitive research funding, continued growth of the Nurse Navigator Program, launch of the Pilbara Hub and sustained contribution to global rare disease leadership. Recognition through major awards and nominations acknowledged individual and collective effort, but more importantly signalled credibility within the broader health and research and innovation ecosystem.

Rare Care's influence continues to extend beyond Western Australia. Through global partnerships, policy contributions and the Global Nursing Network for Rare Diseases, we are contributing to international efforts to improve diagnosis, care and equity for people living with rare disease. This year also saw the launch of the Wales Digital Rare Care Centre. Critically, our global reach is grounded in local delivery and shaped by the lived experience of Western Australian children and families.

To every donor and partner, thank you. Your generosity, collaboration and kindness have enabled this progress. Year 4 demonstrates what is possible when compassion and sustained commitment is matched with evidence, coordination and shared purpose.

The pages that follow set out the impact delivered this year across each stage of the rare and undiagnosed disease journey.

Sincerely and with gratitude,

**Dr Gareth Baynam**
Medical Director, Rare Care Centre

# Welcome to the Rare Care Centre

**This report marks another year of progress in our mission to improve the lives of children and families living with rare and undiagnosed disease across Western Australia (WA).**

**8,000** rare diseases and increasing

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

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

**5-7 years** the average time to diagnosis

**Only 5%** of rare diseases have a prognosis altering treatment

**70%** of rare diseases are realised in childhood

**80%** of rare diseases are of genetic origin

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

## Why We Exist

Rare diseases, while individually rare, are collectively very common. They affect thousands of children in WA and millions worldwide. The vast majority are chronic, severe and multi-systemic, often presenting in early childhood. Yet only a very small proportion have an effective treatment, and the average time to diagnosis remains more than five years.

Rare and undiagnosed diseases are the leading cause of childhood mortality, responsible for a staggering 6 in 10 childhood deaths. Hospital mortality rates for children with rare diseases are significantly higher than for children with common conditions. The economic burden is immense, with rare diseases accounting for a disproportionate share of paediatric hospital expenditure.

Despite this scale, most rare diseases remain largely invisible within health system data. They are not adequately captured by local and international classification and coding systems, meaning their true prevalence, cost and impact are not tracked and have historically been greatly underestimated. Advances in diagnostic testing, data linkage, treatment, care and support are progressively surfacing the unmet need and have made it clear that there is so much to be done. Approaching rare disease as a collective and discrete domain requiring comprehensive and targeted action is increasingly being prioritised as a global health inequity challenge. The scale of the unmet need and parallel opportunity for tractable outcomes that avert suffering and death is becoming clear.

Behind the statistics are children and families navigating complex health, disability, education and community systems, often without a clear pathway. Care is frequently fragmented. Eligibility criteria do not reflect complexity. Services are designed for common conditions and linear pathways, not uncertainty or rarity. Families are left to coordinate across silos, advocate repeatedly, bear the brunt of re-telling their stories, and absorb the emotional and financial strain.

Rare and undiagnosed diseases affect more than health alone. They influence schooling, social participation, employment, mental wellbeing and family stability. Without coordinated action, unmet need compounds over time, increasing both personal and system burden.

## Our Approach

The Rare Care Centre (the Centre) exists to respond to this structural challenge.

We operate as a cross-sector Clinical Centre of Expertise, working across health, disability, mental health, education and community systems to deliver coordinated, equitable and sustainable care. Our role is to reduce fragmentation, address unmet need, and strengthen the system around each child and family.

Our work is guided by the Patient Journey Framework, developed to reflect the real questions families ask and the stages they move through:

- What are the first steps toward a diagnosis?
- How do we access treatment and what if there is no treatment?
- How do we manage our care and navigate across all the sectors involved?
- Where do we go for support and trusted information?
- How do we ensure other families no longer face the same challenges?

These questions define the stages of the journey:

1. Early Diagnosis
2. Access to Treatment & Trials
3. Care Delivery
4. Support & Wellbeing
5. Ecosystem

The Patient Journey Framework recognises that this is not a linear path. Needs evolve, sometimes quickly and without warning. A new symptom, a missed referral, a transition to school or a change in service access can shift the entire trajectory of care.
Our model is designed to respond to that reality.

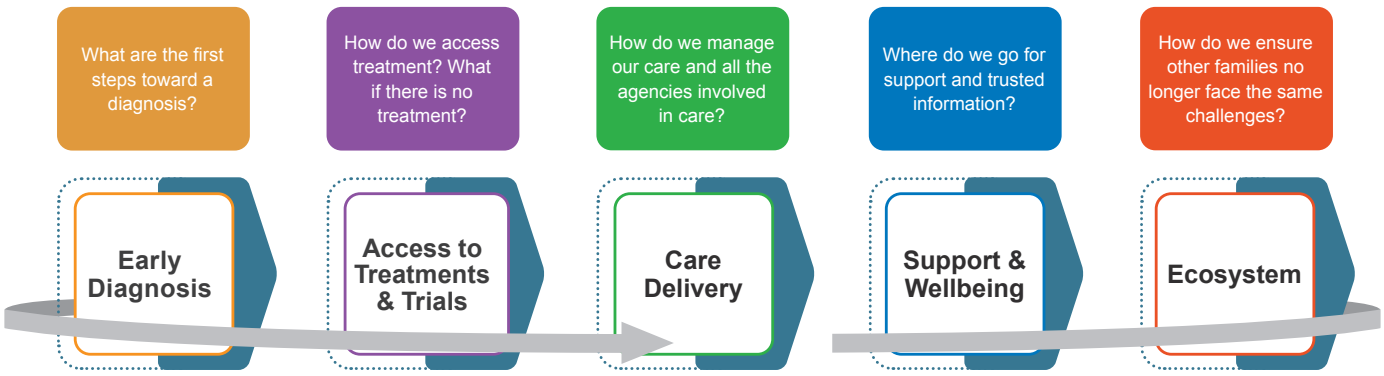
Underpinning this work are critical enablers that strengthen impact across every stage: Digital & AI and Global Leadership & Partnerships.

In Year 4, the Centre continues to mature as both a clinical service and a system reform initiative. We deliver cross-sector care coordination directly to children and families, while strengthening care pathways, clinical trial readiness, workforce capability and international collaboration.
Our approach combines evidence, collaboration and lived experience to improve outcomes locally, while contributing to advancement in rare disease care globally.

The pages that follow outline the measurable impact delivered this year across each stage of the Patient Journey Framework and the critical enablers that support it.

*"I would like to thank the many dedicated CAHS clinicians, researchers and staff who have worked tirelessly for many years to improve outcomes for families living with RUD. This extraordinary financial contribution from the Stan Perron Charitable Foundation, and the support and collaboration of all funding and research partners gives us a unique opportunity to deliver life-changing benefits for families."*

Michael Hutchings, CAHS Acting Chief Executive



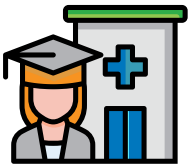
# Year 4 Highlights

**From local coordination to global leadership, Year 4 demonstrated sustained growth, measurable value and expanding influence.**

**411**
children and families supported through cross-sector care coordination

**11**
university student placements

**$15.3m**
successful grant applications contributed to, and partnered on



**36**
enrolled nursing student placements

**34**
conference presentations (local, national and international)

**18**
international meetings

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

**43**
presentations delivered about our Model of Care and Nurse Navigator Program

**6**
major awards and nominations for Centre staff and programs

**30**
publications authored or co-authored by the Rare Care Centre team

**824**
members across 66 countries in the Global Nursing Network for Rare Disease (GNNRD)

**TOTAL $249.1M** funding received for the WA Rare Care Comprehensive Centre

**$221.1M**
funding from Stan Perron Charitable Foundation

**$25M**
funding from Perth Children's Hospital Foundation

**$3M**
funding from the University of Western Australia

# Recognition and Achievements

**The Centre continued to gain national and international recognition throughout the year, reflecting growing credibility, trust and influence across clinical care, research and innovation and system reform. It signals confidence in the model, the strength of partnerships and the measurable impact being delivered for children and families living with rare and undiagnosed disease.**

## Media Coverage

In October, the Stan Perron Charitable Foundation announced a landmark $221.1 million donation to fund the establishment of the Rare Care Comprehensive Centre (RCCC). This transformative gift, together with $25 million from Perth Children's Hospital Foundation and $3 million from The University of Western Australia, brings total committed support to nearly a quarter of a billion dollars over 10 years.

This funding will enable the establishment of the RCCC, strengthening diagnosis, treatment access and coordinated care for children and families living with rare and undiagnosed disease in WA, while positioning the state as a global leader in integrated rare disease care.

Elizabeth Perron AM, Executive Chair of the Stan Perron Charitable Foundation, said the donation honours her father's legacy:

*"Western Australia is so fortunate to have world-class researchers and practitioners who work tirelessly to address some of the most complex and rare diseases that affect children around the world. The Stan Perron Charitable Foundation is very pleased to have the opportunity to provide direct support for their important work, ensuring that Western Australia remains at the forefront of this vital field of medical research."*

The announcement received extensive coverage across major television, print and digital media, including Seven News, Nine News, 10 News and The West Australian. Coverage focused on the practical impact the RCCC will deliver for families who currently face prolonged diagnostic journeys and fragmented care pathways.

Beyond this milestone, the Centre's leadership and expertise continued to attract national and international attention. Dr Gareth Baynam and Dr Andy Poh featured in the "In Scientific Dialogue" podcast, discussing how rare diseases challenge health systems to rethink care delivery. Rare Revolution Magazine profiled WA's approach to equitable innovation in rare disease care, reinforcing the Centre's growing global relevance.

This visibility strengthens public understanding, builds confidence among families and partners and reflects increasing recognition of the Centre's expertise in delivering coordinated, family-centred care.

## Notable Awards

Recognition during the year reflected both individual leadership and collective impact.

Dr Gareth Baynam was a finalist for the 2026 Australian of the Year (WA) acknowledging his sustained leadership in ensuring that children with rare and undiagnosed disease are not left without answers or coordinated care. Through his work founding the Undiagnosed Diseases Program WA, co-founding the Undiagnosed Disease Network International, contributing to Australia's first National Strategic Action Plan for Rare Diseases and leadership in the International Rare Disease Research Consortium (IRDiRC), Gareth has helped shape diagnosis pathways, health delivery reform and research priorities nationally and globally.

Internationally, the Centre played a central role in Medscape's Rare Disease Education Platform receiving a 2025 Silver Anthem Award in the *Health: Education or Literacy* category. Medscape has a reach of more the 13 million active healthcare professionals. The Anthem Awards recognise global purpose-driven initiatives delivering measurable social impact. The Centre contributed through development of rare disease nursing education modules, steering committee leadership and lived experience contributions from our Nurse Educator. This collaboration strengthens the global nursing workforce and directly supports earlier recognition and coordinated care for rare disease patients.

These acknowledgements are not about profile alone. They reflect the growing maturity of the Centre and its contribution to shaping rare disease care locally and globally.

## Grant Success

The Centre continued to demonstrate strong performance in competitive funding environments, contributing to and partnering on research and innovation projects totalling $15.3 million for the year.

Successful projects include:

- Streamlining the development of antisense therapeutics for Western Australian children with rare genetic disorders, $5 million
- National Rare Trials Infrastructure, establishing Western Australia as a coordination hub for adaptive platform trial capability, $1 million
- Pharmacogenomics for youth mental health, Stan Perron Charitable Foundation, $4.8 million
- Aboriginal and Torres Strait Islander partnerships for paediatric lung health excellence study, NHMRC, $2.5 million
- Single cell CRISPR to Identify Pathogenesis with Transcriptomics (SCRIPT), $434,000
- Developing manufacturing of GD2 CAR T-cells for neuroblastoma, $497,320
- FHRI PromethION24, $703,000
- Tsinghua collaboration adapting large language models for rare disease diagnosis in Asian populations, $100,000
- Developing an Adaptive Rare Diseases Platform Trial (ARDaPT), Women and Infants Research Foundation, $45,000
- Functional significance of novel genetic variants identified in clinical settings, $139,000

These grants span therapeutics, digital health, genomics, Indigenous partnerships and clinical trial infrastructure. Collectively, they accelerate access to diagnosis, enable local manufacturing capability, strengthen research translation and position WA as a coordination hub for rare disease innovation.

## Publications

The Rare Care team contributed to 30 peer-reviewed journal articles during the year, advancing knowledge in rare disease diagnosis, workforce capability, digital innovation and research prioritisation.

Selected publications include:

- Rare and Undiagnosed Disease: A Learning Program for Nurses and Midwives. Authored by Sue Baker, Kaila Stevens and Dale Pugh and published in Nursing Reports.
- A systematic assessment of large language models' knowledge of rare diseases: How much do large language models know about rare disease?. Primary author Tudor Groza with contributions from Tristan Carlisle and Gareth Baynam and published in Human Genetics and Genomics Advances.
- Australia's top 10 rare disease research priorities: a priority setting partnership. Authored by Gareth Baynam et al and published in Journal of Rare Diseases.

These publications demonstrate the Centre's commitment to rigorous evidence generation while maintaining a clear focus on clinical translation and system improvement.

# Jake's Story

**From diagnosis to whole family support**

Jake and his family were referred to the Centre when he was five years old following a diagnosis of White-Sutton Syndrome, a neurodevelopmental disorder characterised by global developmental delay, intellectual disability, autism spectrum disorder and visual, hearing and feeding difficulties. Like many families adjusting to a rare disease diagnosis, Jake's parents were concurrently managing medical uncertainty, schooling decisions and disability supports.

Before referral, Jake had frequent contact with Perth Children's Hospital (PCH), including multiple outpatient appointments and an emergency department presentation. Even with a confirmed diagnosis, the complexity of Jake's condition meant his family were experiencing gaps in information, coordination and practical guidance. In particular, they wanted more information about the future implications of Jake's diagnosis and to understand options around medical surveillance, education and disability supports.

Over the year that Jake was supported by the Centre, the team worked in partnership with his family to provide holistic family-centred support and structured care coordination.

Early discussions focused on ensuring the family had clear, accessible information about White-Sutton Syndrome and its implications. Rare Care communicated recommended surveillance to Jake's treating team, facilitated a referral to Neurology and arranged key investigations including a renal ultrasound and echocardiogram to support proactive monitoring of his condition.

The Genetic Counsellor worked with Jake's parents to understand the genetic basis of his condition, including considerations for family planning for themselves and their other children to enable informed decision-making and appropriate referrals for Jake's siblings.

The Mental Health Clinical Nurse Specialist provided resources and support for mental health and wellbeing for the whole family, including connecting Jake's dad to the Rare Care Dad's Connect Group. The Senior Teacher discussed available education options with Jake's parents, and in conjunction with the broader team provided written advocacy to support school funding applications aligned with their decisions. Once an autism spectrum disorder (ASD) assessment was finalised Rare Care submitted documentation to both the National Disability Insurance Scheme (NDIS) and Jake's school, strengthening his access to developmental and educational supports and minimising administrative burden for his family.

Medical care coordination remained central throughout. The Centre nurses and paediatrician liaised with specialist clinics, ophthalmology, radiology and Jake's GP to streamline communication and appointments. The Centre GP, in consultation with the broader team developed a visual summary which outlines the effects of White-Sutton Syndrome on Jake to help tell his story. The visual summary was provided to his GP to upskill them in his condition and support continuity of care. This document can be provided to other members of Jake's care team to minimise the number of times his family have to repeat their story.

When initial NDIS requests for equipment and meal supports were declined, the NDIS Navigator assisted Jake's family to review their plan, liaised with the Local Area Coordinator regarding reassessment of funding, and worked with the nursing team to provide practical guidance on feeding safety, continence supports and after-school care to Jake's family.

Jake was discharged with clearer documentation, established surveillance pathways and strengthened education and disability supports. In the five months following discharge, he required three outpatient appointments, reflecting a reduction in hospital use.

Jake's journey illustrates the role of the Centre in providing families with clearer pathways, improving access to services and increasing their confidence in navigating systems.

# A Year of Impact

**The Rare Care Centre's work is grounded in the Patient Journey Framework, which recognises that children and families navigating rare and undiagnosed disease move through distinct stages of a complex journey. While no two experiences are the same, common needs emerge across Early Diagnosis, Access to Treatment & Trials, Care Delivery, Support & Wellbeing and the broader Ecosystem that they navigate.**

In previous Impact Reports, our work was presented through the Centre's foundational pillars. That structure reflected how we organise the Centre and deliver our services. This year, we have made a considered and intentional shift in how we tell our story. We are now reporting against the stages of the Patient Journey Framework to better reflect our impact on children and families.

While we have always worked across the full breadth of the journey, it has become increasingly clear that this is the most meaningful way to demonstrate impact.

The focus is on what has changed for families, clinicians and the system as a result of the Centre's work.

In addition, this section outlines the impact of work across critical enablers, Digital & AI and Global Leadership & Partnerships, which strengthen delivery and expand reach across the entire journey.

# Early Diagnosis

Children and families living with rare and undiagnosed diseases often face a prolonged and complex path to diagnosis, commonly referred to as the diagnostic odyssey. This can involve years of uncertainty, misdiagnoses, repeated investigations and procedures and fragmented clinical management. The impact extends beyond the child's health. It affects mental wellbeing, family stability, financial security and trust in the health system.

Uncertainty generates ongoing anxiety and distress. When answers remain elusive, families can feel isolated and unsupported. Earlier diagnosis changes this trajectory. It enables timely and tailored management of symptoms, supports informed life and family planning, reduces mental health burden and connects families to relevant communities. Diagnosis also creates the foundation for targeted therapies, personalised care and access to clinical trials.

## Strengthening Early Suspicion and Referral

Earlier diagnosis begins with earlier suspicion. Children cannot enter a diagnostic pathway unless rare disease is considered in the first place. Limited awareness and training across the health workforce remain significant barriers.

In Year 4, the Centre continued to build capability to recognise signs of rare disease across disciplines and care settings.

Our GP, Dr Claire Bowden, delivered the session "When Common Symptoms Point to Uncommon Diseases: A GP's Detective Toolkit" to clinicians nationally through the 2025 Rare Disease Project ECHO series, co-hosted throughout the year by our Nurse Educator, Sian.

Across the year, the Rare Care team delivered 43 presentations to more than 927 health professionals and partner organisations locally and nationally. These sessions extended beyond hospital settings, including presentations at the WA Health Improvement Summit and other broader system forums, ensuring rare disease awareness reaches generalist and cross-sector audiences.

Internationally, the team presented across nursing, medical, biotechnology, scientific and digital health forums and participated in 20 conferences and symposia. This sustained engagement strengthens rare disease awareness, recognition capability and referral pathways both locally and globally.

## Undiagnosed Hackathons: Accelerating Answers

Globally, an estimated 350 million people live with an undiagnosed condition, approximately 75 percent of whom are children. While genomic sequencing can provide answers for around 40 percent of individuals, nearly 60 percent remain without a diagnosis.

The Undiagnosed Hackathon, led by the Wilhelm Foundation, is a global collaborative initiative designed to address this gap. The Centre Medical Director, Professor Gareth Baynam, serves as Advisory Board Member and Scientific Co-Lead.

The Hackathon model brings together concentrated expertise, data and technology in an intensive 48-hour environment to find a diagnosis that has remained elusive despite persistent investigation. Insights generated are translated beyond individual cases, strengthening diagnostic practice globally.

In 2025, the second Undiagnosed Hackathon was hosted at the Mayo Clinic in Minnesota, USA. The event convened 130 collaborators from 15 countries, including clinicians, geneticists, bioinformaticians, molecular biologists, scientists, developers and AI specialists. Twenty-nine individuals with undiagnosed diseases participated.

During the event, the bell rang six times, marking confirmed diagnoses for those individuals. Nine additional participants progressed with defined follow-up pathways. Within three months, a further diagnosis was achieved through continued collaboration.

Dr Tudor Groza (Rare Care Centre, Digital and AI advisor) tested and refined advanced AI approaches during the Hackathon, including


early application of Alpha Genome technology. These tools are now being deployed in WA to support additional case investigations. Outcomes and insights contributed to international scientific dissemination, including coverage in the prestigious medical journal, Nature.

A scientific and ethical blueprint has since been developed with international partners to support scaling of the Hackathon model globally. The third Hackathon was held in February 2026 in Hyderabad, India. The Centre is a keen collaborator with the Wilhelm Foundation on this important initiative.

As Professor Baynam reflected:

*"The Undiagnosed Hackathons are unique. Nowhere else on the planet is there such a concentration of data, new technology and medical and scientific expertise brought together, and with the children and families present, to condense what otherwise takes years or decades into a 48-hour period. This not only changes the lives of the children and families at the hackathon but also amplifies globally through the hackathon's international network."*

## Year 4 Impact

**In Year 4, the Centre strengthened early diagnosis through:**

- **Workforce education and awareness building**
- **National and international capability development**
- **Development and deployment of advanced genomic and AI tools**
- **Direct participation in global collaborative diagnostic acceleration models**

**Earlier diagnosis remains the entry point to better care. The impact delivered this year reflects a deliberate focus on reducing delay, strengthening referral pathways and accelerating answers for children and families.**

# Access to Treatments & Trials

For many families, diagnosis does not bring resolution. After years seeking answers, they may learn that no approved treatment exists, or that emerging therapies and clinical trials are inaccessible due to geography, cost or timing.

Approximately 95 percent of rare diseases currently have no prognosis-altering treatment. For many children, a clinical trial represents the only pathway to a potential treatment.

The gap between diagnosis and access to treatment remains one of the most urgent challenges in rare disease care.

## Structural Barriers to Treatment

Traditional drug development models were not designed for rare conditions. Large, randomised trials, extensive toxicology programs and commercial investment exceeding $1 billion per drug are rarely feasible for very small, dispersed populations limiting advancements in treatment options for many diseases.

Even where scientific advances have resulted in a clinical trial, accessibility can remain constrained by infrastructure, workforce capacity and regulatory complexity. In WA, limited local trial availability has historically, frequently required families to consider travelling or relocating interstate or overseas, adding financial and social strain to already complex circumstances.

Improving access requires deliberate infrastructure and coordinated system design.

## Advancing Rare Disease Trial Capability in WA

In Year 4, the Centre focused on building sustainable rare disease clinical trial capability in WA through a multi-institutional network called TrialR. TrialR brings together people living with rare disease, multiple metropolitan and regional hospitals, trials centres, medical research institutes and universities, clinicians and scientists.

TrialR is strengthening trial readiness, access and workforce capacity. Establishing these foundations positions WA to host and initiate more rare disease trials locally.

A key milestone was collaborating with the PCH Neurology, Oncology and Research Departments to deliver the first clinical trial for Angelman Syndrome in WA. Targeted resourcing has created protected time for clinicians, built governance capability and embedded trial delivery within existing services.

## ARDaPT: Adaptive Trial Infrastructure

In 2025, Dr Brad MacDonald received the Women and Infants Research Foundation (WIRF) Research Acceleration Award to progress the Adaptive Rare Disease Platform Trial (ARDaPT).

ARDaPT establishes a reusable master protocol capable of evaluating multiple therapeutic interventions across rare diseases. Adaptive platform designs improve efficiency while maintaining scientific and ethical rigour.

During Year 4, Brad:

- Commenced the development of Governance processes
- Mapped regulatory and implementation pathways
- Progressed trial infrastructure design
- Presented the project at the WIRF Symposium

This infrastructure strengthens trial readiness and is expected to reduce approval timelines and increase WA's capacity to attract industry and investigator-initiated trials.


## Building a Trial Ecosystem

Effective trial delivery depends on coordinated system support.

In Year 4, partnerships were strengthened across the Child and Adolescent Health Service (CAHS) clinical trials infrastructure, oncology, nursing, anaesthetics, theatre services, psychology support and the CAHS Research Centre. The Centre maintained membership of the Australian National Paediatric Trial Network and the WA Country Health Service (WACHS) Clinical Trials Group.

International engagement continued through collaboration with the Rare Disease Moonshot initiatives, the European Rare Disease Research Alliance, Uncommon Cures and leading hospitals in China.

The team engaged with more than 20 national and international industry partners and undertook feasibility assessments for future studies at PCH.

## Equity in Trial Access

Rare diseases are characterised by inequity. This inherent inequity can be compounded by intersecting factors such as Indigeneity and remoteness, which also impact access to clinical trials. Addressing inequities in access to rare disease clinical trials is critical, as they are often the only potential treatment pathway, and their success depends on enrolling sufficient numbers from already small, highly specific patient populations.

The Centre led authorship of an international publication on diversity, equity and inclusion in rare disease trials, accepted by the Orphanet Journal of Rare Diseases in February 2026.

Work also commenced with Lyfe Languages to support improved access to culturally secure information about trials for Aboriginal and Torres Strait Islander families.

## The Human Imperative

Behind infrastructure are families seeking options.

As Steve Hille, father of Mina and CEO of FOXG1 Research Foundation Australia, reflected:

*"Trials are the only option and the only hope we have... The outcome of a trial is a treatment, but the trial must happen to get there. We aren't asking for anything special for Mina or kids like her, we want her to have the same opportunities as a child without a rare disease."*

### Year 4 Impact

**In Year 4, the Centre:**

- **Continued to build foundational rare disease trial infrastructure in WA**
- **Supported access to a first in WA disease trial at PCH**
- **Advanced adaptive platform design through ARDaPT**
- **Strengthened national and international partnerships**
- **Engaged industry partners and conducted feasibility assessments**
- **Embedded equity considerations into trial development**

**Year 4 focused on building practical readiness and strengthening WA's capacity to deliver more timely and equitable access to rare disease trials and treatments.**

# Care Delivery

**Children and families living with rare and undiagnosed disease often navigate multiple teams and systems at once. Care Delivery focuses on the practical delivery of care, improving coordination, integration and navigation, strengthening continuity and reducing avoidable burden on children, families and the health system**

## Clinical Services – Impact at a Glance

### Reach and Access

**411** Families supported through the Cross-Sector Care Coordination model

**78** Families supported through the Nurse Navigator Program

**247** Children maintained continuity of education through collaboration with School of Special Education Needs: Medical & Mental Health

**36** Nursing student placements strengthening future workforce capability

### Complexity of Care

Children supported by the Centre are managing multi-team, cross-sector care.

**6**
Median number of CAHS teams involved

**16**
Maximum CAHS teams involved for one child

This level of system involvement underscores the coordination burden placed on families.

### Financial impact

(based on utilisation metrics):

**$11,030** approximate savings per patient, including offsetting the program cost

### System Impact

Measured reductions in hospital utilisation:

**44%** ↓ **decrease** in inpatient bed days

**69%** ↓ **decrease** in non-attendance at outpatient appointments

**61%** ↓ **decrease** in outpatient visits

### Who We Support


Average time engaged with the Centre: **153 days**

Metro: **89%** | Regional: **11%**

**40%** undiagnosed at time of engagement


These results reflect reduced hospital dependence, improved appointment engagement, strengthened GP access, better use of disability funding and better alignment across education and health systems.

Care delivery in Year 4 demonstrates measurable system efficiency and improved family experience for children with rare and undiagnosed disease.

## Cross-Sector Care Coordination Program

To date, 411 children and families have been accepted into the Cross-Sector Care Coordination Program, with 156 families currently active and 255 families discharged.

Demand for our services has increased substantially as awareness of the Centre grows and improved outcomes for children and families are demonstrated. To respond, clinical capacity has expanded through increased staffing across key roles including Paediatrician, Clinical Nurse, Mental Health Clinical Nurse Specialist, GP, and administrative support. Service delivery has also been extended through additional clinics, including a fortnightly clinic at REACH Mt Lawley and a new monthly clinic at the Cockburn Health and Community Hub which commenced in December 2025.

A qualitative evaluation of the Cross-Sector Care Coordination Program was undertaken to seek feedback from parents and carers, with the report finalised in December 2025. Feedback was consistently positive, particularly regarding feeling supported, reducing stress and improving navigation of complex systems.

*"To have the rare care team and education support... that's been the lifechanging factor... I think it's important to stress the fact that just having access to the team alleviated some stress and felt like I was supported. It just felt like there's help, even if they weren't able to resolve our issues, they were trying really hard to do so and were on our side. I think that reduces stress levels and feels like you're not alone."*

*"From the get-go, I was blown away by how much support there was. I felt like I've been banging my head against the wall for a few years trying to get any help or support at all, and immediately when I got the very first phone call from the Rare Care team, I was blown away at how many professions are part of the team. It was a big relief and felt like somebody was finally on our side."*

## NDIS Support

Families of children living with rare or undiagnosed disease often face significant challenges navigating the NDIS, particularly when translating complex medical information into disability-focused language. To address these barriers, the Centre has implemented supports designed to bridge the gap between Health and NDIS, build family capability and inform system-level improvement.

Within the Cross-Sector Care Coordination Program, the team:

- Works directly with families to prepare NDIS applications, ensuring documentation reflects functional need
- Assesses whether plans meet requirements and supports Change of Circumstance requests where needed
- Coordinates referrals for additional appropriate assessments to strengthen evidence for reviews
- Identifies suitable local services and resources that can be accessed via NDIS funding
- Builds parent and carer capability to understand and use plans effectively

Within the Nurse Navigator Program:

- NDIS support was incorporated after the program identified that many families faced challenges navigating the scheme
- The team identifies families who would benefit from additional coaching and capacity building

Across the wider system, Centre staff also:

- Participate in local and national NDIS platforms to represent the needs of children with rare and undiagnosed disease
- Provide expert clinical insights to other health and community systems, including WACHS and CAHS pathways
- Collaborate with not-for-profit organisations including Rocky Bay, Kiind, RippleAbility, Childhood Dementia Initiative and Alike to advocate for the needs of children with rare and undiagnosed disease in the disability space
- Contribute expert advice to the Rare Voices Australia (RVA) Rare Disease Disability Network
- Engage directly with decision-makers, including meetings with Hon Dr Anne Aly MP and WA representatives for Thriving Kids

## Nurse Navigator Program

The Nurse Navigator Program which commenced in 2023, approaches the end of its initial Australian Government grant funded period in June 2026. A comprehensive external final evaluation report on The Rare and Complex Disease Telehealth Nurse Program (Navigator Project) has recently been completed, confirming that telehealth nurse navigation addresses critical service gaps, improves patient experiences and offers potential for cost-effective system benefits. As a result, this service will be embedded ongoingly as part of the clinical services delivered by the Centre.

Demand for the Nurse Navigator Program increased during Year 4. The program recruited a second part-time clinical nurse and added NDIS Navigator support.

Program reach to date:

- 78 children referred
- 30 currently in the program
- 48 discharged

Families can be referred to the program by PCH clinicians, external paediatricians and via the RVA helpline. Once accepted, families receive an initial telehealth assessment which informs tailored interventions delivered to address gaps in their care. Navigation and coordination of services form a significant part of the interventions delivered.

Children supported through the program were under the care of an average of six PCH teams. The Nurse Navigator delivered an average of 14 episodes of contact per child (to the family or other service providers) and 13 interventions per child. Interventions include rare disease information and resources, mental health referrals, welfare information, liaison with school, GP access support, care coordination between teams and appointment coordination.

*"It was extremely helpful having access to an experienced nurse who understood both the medical and disability aspects to help obtain, summarise and present the information in the passport. I think this is why the passport is so successful. I was also extremely grateful on a parent level for the understanding, sensitivity, advice and input. It's a mentally challenging task listing your child's difficulties and weaknesses whilst also trying to show their strengths.*

*We are only 10 years into our medical journey, and it can be so overwhelming trying to remember everything for every situation. I know it's going to make our medical journey going forward much easier and less stressful and gives me added peace of mind should we need to suddenly present for Emergency Care. I'm very grateful to have come across this service and appreciate having the opportunity to be part of it."*

## Patient Passport

All Nurse Navigator families are provided with a Patient Passport, the most frequent intervention delivered through the program.

The Patient Passport, developed by CamRARE and adapted in partnership for the Centre, provides a concise strengths-based summary of complex medical, developmental and personal information that might be difficult to communicate, particularly in high-stress or time-critical settings.

To date, 62 Patient Passports have been completed. Feedback has been consistently positive, with families describing improved emergency department experiences and reduced burden from repeatedly recounting complex histories. The Passport has also been used to support communication and safety across school, daycare, community and allied health settings.

The effectiveness of the Passport is in part due to the information being completed by an experienced Nurse Navigator. Clinicians and families have highlighted the value of clinical expertise, advocacy and sensitivity in producing a Passport that is accurate, usable and tailored.

Following the positive feedback from families, the passport will now be rolled out more widely via the Cross-Sector Coordination Program in 2026.


RARE Patient Passport

THIS IS ME

Name

Known as

Date of birth

UMRN

Completed by

Updated on

CONTACTS

Medical specialist

Phone

GP Surgery

Phone

Emergency contact

Phone

DIAGNOSIS / SYMPTOMS

Primary diagnosis

Key clinical features / symptoms

Weblink to condition information

Additional diagnosis or symptoms

Additional diagnosis or symptoms

Additional diagnosis or symptoms

CLINICAL INFORMATION

Medications and dosage

Is emergency care frequently required?

No

Yes (see emergency care record on next page)

Seizures

No

Yes (see neurology section on next page)

Respiratory issues

Allergies

TOP 3 THINGS TO KNOW

1

2

3

PRESENT THIS PASSPORT TO MEDICAL OR CARE STAFF

ADDITIONAL INFORMATION

MORE ON NEXT PAGE >>

EMERGENCY CARE RECORD

HISTORICAL INSTANCES OF ED VISITS

Average frequency of emergency care

weekly monthly annually less often

Date

Presentation / symptoms

Treatment

Outcome

MY NORMAL

Height

Weight

Pain

Neurology & neurodivergence

Major surgery history

Implants / lines / tubes

Mobility

Sensory impairments

Equipment & devices used

Toileting

Additional information about me or about caring for me

CARING FOR ME

Key things to know about caring for me

Communication

Eating & drinking

Likes

Dislikes

This passport template has been designed by

CAMRA

www.camraredisease.org

DISCLAIMER

This document reflects the patient or caregiver's understanding of their condition and is to be used as a tool to aid communication of medical and care needs.
CamRARE is not liable for any losses or damages from the use of this passport or its information.

Piloted by the Rare Care Centre, Perth Children's Hospital, Western Australia

## Pilbara Hub

The Rare Care Pilbara Hub is a collaborative initiative between CAHS and WACHS, integrating the specialist expertise of the Centre at PCH with local Pilbara expertise and networks. In 2025, the team focused on development, planning and service design to support effective and sustainable delivery.

Key establishment activities included:

- Community engagement through the On- Country Yule River Bush Meeting (23 and 24 July 2025) and IBN Group Members Consultation Meeting.
- Stakeholder engagement across approximately 30 services in the Pilbara
- Two co-design workshops in Karratha and Port Hedland
- Governance established through a Cross-Sector Governance Group and Model of Care Advisory Committee, with the first two meetings of each group held
- Capacity building through a Client Referral Workshop in Newman (December), attended by 15 participants

A locally based workforce and subsequent capacity and capability building are a cornerstone of the Hub's model. Employment of a Clinical Nurse Specialist, Senior Project Officer and Aboriginal Health Practitioner along with securing a locally based Paediatrician to deliver clinics was a crucial step towards service commencement.

Following detailed planning and stakeholder engagement, we were pleased to deliver our first patient clinic in Karratha in February 2026. Monthly clinics have been scheduled in Karratha and Port Hedland, with bi-monthly clinics to be delivered in Newman. The Hub is delivered as an outpatient model under the Activity Based Funding Framework. Formal in-kind support commitments at clinics have been secured from the Department of Education's School of Special Education Needs: Medical and Mental Health (SSEN:MMH), the Department of Communities, and the National Disability Insurance Agency (NDIA) to ensure Pilbara families have access to a truly cross-sector place-based approach.

Currently, two thirds of referrals received for the Pilbara Hub are for Aboriginal families reflecting a strong and effective referral pathway from the Aboriginal Community Controlled Health Organisations in the region. The delivery of patient clinics in the Pilbara supports culturally secure care on Country and closer to home, enabling equitable, timely and direct access to services for children and families.

# Sawyer's Story

**Preparing for a rare transition**

Sawyer is a 15-year-old living in regional Western Australia. He and his mum, his sole parent and primary advocate, have navigated years of uncertainty related to his rare chromosomal duplication and complex developmental needs.

Before being referred to the Centre, Sawyer's care was spread across multiple health services in the metropolitan and regional area. With the lack of communication between services, his mum managed appointments, assessments and funding applications largely on her own, often without a clear pathway or consistent support.

In late 2024, a Clinical Geneticist referred Sawyer to the Centre. From the time the referral was accepted and the team heard the family's priorities at the triage call, the team reviewed previous reports, identified gaps in assessment and prepared for his first appointment. When Sawyer attended clinic in March 2025, there was already a structured plan in place aligned with his family's priorities.

Several gaps became clear. Sawyer had not yet received an ASD assessment, which was required to support appropriate school placement and strengthen his NDIS eligibility. His mum had been seeking funding without the documentation needed to demonstrate the functional impact of his condition. Sawyer's support at school was limited with no education assistant in place, and care in his regional area was fragmented with no consistent GP overseeing management.

Given Sawyer's age and his upcoming transition to adult services, it was crucial to set Sawyer and his mum up for success in the next phase of his life.

The Rare Care team coordinated a formal ASD assessment. This provided the clinical evidence required to secure appropriate education and NDIS supports. The Centre Paediatrician linked Sawyer with ongoing developmental monitoring and eye health checks, and the team developed a visual summary to assist clinicians and care providers in quickly understanding his needs.

The Centre GP identified and upskilled a local GP with relevant information about Sawyer's condition and explained mechanisms such as chronic care management plans to enable his care to be anchored closer to home.

Following his assessment and Centre advocacy, funding for his education support school was secured. Sawyer's NDIS plan was appropriately strengthened reflecting the clearer documentation provided, and after working with the NDIS Navigator his family was better prepared for future reviews. The Centre team worked with Sawyer's mum, providing mental health and wellbeing support and regular follow-ups and guidance regarding guardianship planning, welfare entitlements and community services.

During a hospital admission, the Centre coordinated social work input, Keeping Kids in No Distress (KKIND) services and therapy dog visits to support both Sawyer and his mum while they were away from their regional community.

Sawyer was discharged from the Centre with established local supports, stronger documentation and greater preparation to transition to adult services. Sawyer's GP has become an essential part of his team and helps to bridge the divide between rural and metropolitan, and paediatric and adult services.

# Support & Wellbeing

**Care does not end with a diagnosis or coordination. Rare and undiagnosed disease affects the whole family in a myriad of ways. Support & Wellbeing focuses on the family as a whole, reducing isolation and building sustainable support networks around children, parents and siblings.**

## Churchill Fellowship: Strengthening Family Support Models

In 2024, the Centre Clinical Program Lead Anna Thetford was awarded a Churchill Fellowship to undertake an international exploration of family support programs for rare disease.

This work was completed in 2025 and included visits to Centres of Excellence in Romania, Sweden, Norway, Wales and Ireland. Anna's findings were formally published by the Churchill Trust and are publicly available. In November 2025, Anna was awarded the Winston Churchill Medallion in recognition of the quality and impact of this work.

The Fellowship identified several consistent principles across high-performing family support programs:

- Families must be placed at the centre of program design and delivery
- Lived experience should shape education, activities and workforce skill mix
- Clear program structure and intentional design improve engagement
- Addressing practical barriers, including logistics and access to care supports, directly improves participation and outcomes
- Sustainability requires measurable outcomes, embedded research and strong cross-organisational partnerships

In 2026 Anna will build on these findings to develop a WA-specific family support program as a core next step to expand the Centre's service offering.


## Sibling Support

Siblings of children and adolescents living with rare disease often experience overlooked emotional and social impacts.

A structured sibling support program will be delivered as the first part of a comprehensive family support program. In February 2026 Centre team members undertook SIBS training, a preventive group intervention designed for siblings of children with physical or cognitive disability run by a team from Norway. The program includes structured sessions for both siblings and parents supporting communication, coping strategies and peer connection.

This work builds on the Churchill Fellowship findings and reflects the Centre's focus on whole-of-family wellbeing.

## Dad's Connect Group

Fathers of children with rare and undiagnosed disease often report social isolation and limited access to peer support.

Since its implementation in 2024, the Dad's Connect Group has grown to include 30 fathers and is supported by a Mental Health Clinical Nurse Specialist and two fathers who co-facilitate the group. The group provides peer connection, mental health support, shared problem-solving and a safe space for open discussion.

This initiative recognises that targeted supports for specific family members can reduce isolation and strengthen overall family resilience.

As Cristian, one of the dads who co-facilitates the group, reflected:

*"Although we all have busy lives with many challenges, we are still getting dads showing up to the events. I think this speaks to the value that people are getting out of it. We have no agenda or structure around the group but there are always great conversations had, with a shared understanding of the challenges we all face. Funny and not so funny stories are shared, great ideas and pointers about where to find useful information or help and indeed examples of the frustrations we have to deal with in the world of caring and advocating for your child. I think the most important part of the group is the unspoken understanding that there is zero judgment amongst us, a natural empathy for what other dads are going through and the ability to be able to talk freely without having to explain every aspect."*

### Year 4 Impact

**In Year 4, Support & Wellbeing efforts:**

- **Explored internationally recognised family support models**
- **Strengthened evidence-informed design of future support programs**
- **Prepared for structured sibling support programs**
- **Sustained a dedicated fathers' peer support network**

**This year reflects a shift toward structured, research-informed and sustainable wellbeing programs that consider the needs of the whole family embedded within rare disease care.**

# Ecosystem

**Ecosystem reflects the Centre's role in addressing the challenges families face at a system level. Changing the ecosystem means raising awareness, strengthening workforce capability, influencing policy and contributing globally so that future children and families no longer experience the same challenges.**

## Raising Awareness

### Rare Disease Education for Schools

In collaboration with SSEN:MMH and Syneos Health Communications, the Centre developed and launched a rare disease animation and accompanying e-learning resource to raise awareness amongst teachers and students in educational settings.

The animation was inspired by a real letter written by a child with a rare disease. The child was not asking for sympathy, but simply to be seen and understood. With more than 70% of rare diseases presenting in childhood, many children move through school unnoticed, not because they are invisible, but because their experiences are not yet recognised or understood.

To bring this reality to life, the animation features a character named Dia. Dia represents children living with rare disease. Her diamond-like form reflects quiet strength, shaped under pressure. She intentionally has a gender-neutral and culturally ambiguous appearance, allowing children to see themselves, or someone they know, in her story. Her rare disease is symbolised by the backpack she carries, a simple metaphor for the additional weight many children manage each day.

Rather than relying on narration, music carries Dia's emotional journey from isolation toward connection. The animation was developed in consultation with local and international teachers, students and people with lived experience to ensure authenticity and relevance.

The resource:

- Encourages more inclusive and informed classroom environments
- Supports teachers to better understand the day-to-day impact of rare disease and discuss this with their students
- Combines visual storytelling and music to convey both emotional and practical realities
- Is freely available and captioned in 20 languages

Since its release on 1 September 2025, the animation "The Weight of Rare Disease" has received 1,036 views. The Centre has distributed the resource to schools, educators, health professionals and advocacy organisations to support more inclusive learning environments.

The Centre's collaborative efforts and the creative vision of Syneos Health Communications were recognised when *The Weight of Rare Disease* was awarded Gold in the category of *Diversity and Inclusion in Creative Communication* in the 2026 PM Awards – an annual event recognising excellence across pharmaceutical marketing and healthcare communications in the United Kingdom.

## Rare Disease Day 2025

On 28 February 2025, the Centre participated in the global "Light Up for Rare" campaign, organising a total of 32 Perth buildings and landmarks to be illuminated in Rare Disease Day colours, including:

- Matagarup Bridge
- Optus Stadium
- Council House
- Perth Train Station
- Great Northern Highway Port Hedland Interchange Bridge
- PCH "Fizz"

This initiative aligned WA with the global Rare Disease Day movement, increasing public visibility and solidarity.


The Centre also:

- Hosted a Rare Recipe Cook Off at PCH, raising awareness of rare disease amongst staff
- Participated in the Rare Revolution social media takeover campaign, sharing Rare Care Centre content across the Rare Revolution social media channels
- Coordinated a Global Rare Disease Day montage video through the GNNRD
- Co-hosted a craft corner within “Fun on Four” at PCH

These activities strengthened local engagement while connecting WA to international awareness efforts.

## Education and Workforce Development

Rare diseases are frequently under-recognised in clinical practice because many health professionals have limited exposure to them in training and rarity may go unrecognised in day-to-day care. Strengthening awareness and education helps build clinicians’ index of suspicion, supporting earlier recognition, more appropriate referral and a more responsive system.

Throughout 2025, the Rare Care team continued to invest in workforce awareness and capability building. This included hosting information stalls at three events: the PCH Nursing Expo, the PCH International Disability Day event, and the SMARTcare7 Notre Dame Nursing Society event. Across these settings, the team engaged nurses, students and allied health professionals in practical conversations about rare disease recognition and the realities faced by children and families.

The Centre’s mascot, Zebedee, played a simple but effective role in this work. By explaining why the zebra is the symbol for rare disease the team created an accessible entry point for discussion about rarity, diagnostic delay and the importance of recognising when a presentation falls outside typical patterns.

This awareness-building work helps strengthen the foundations for earlier diagnosis and more informed care. It also contributes to a growing workforce engaged in rare disease practice, which is seen reflected in increasing nursing interest in the GNNRD.

Internationally, the Centre has partnered with the European Board of Rare and Undiagnosed Diseases (UEMS) to develop a European Certificate Examination in Rare and Undiagnosed Diseases (EC-RUD). The examination, a postgraduate initiative, has been developed to support harmonised knowledge, shared standards and consistent recognition of advanced competencies in rare and undiagnosed disease medicine across Europe, and is accessible by clinicians globally.

Sometimes when you hear hoof beats, it will be a zebra (rare disease), not a horse

Aligned with UEMS educational objectives and Council of European Specialist Medical Assessments (CESMA) quality standards, EC-RUD provides a structured mechanism to assess core knowledge and clinical reasoning in this highly specialised field, while fostering an international community of practice.

In 2025, our Nurse Educator, Sian, and Medical Director, Gareth, contributed to this work through membership of the Examination and Education Steering Committee. The written examination was undertaken for the first time by 13 international candidates in October 2025.

The EC-RUD represents an important step in strengthening shared training frameworks, benchmarking expertise and advancing clinical capability in rare and undiagnosed disease care which can be translated globally.

## Advocacy, Policy and System Reform

### World Economic Forum

In 2025, the World Economic Forum (WEF) reaffirmed its focus on rare diseases through the formation of the WEF Rare Disease Affinity Group under its Health for All initiative focused on global health inequity.

Medical Director Gareth Baynam is a member of the Affinity Group and co-author of the WEF white paper on Global and Multi-stakeholder Investment in Rare Diseases: “Making Rare Diseases Count: How Better Data Can Unlock a Multitrillion-Dollar Opportunity”.

This white paper, addressing data inequalities impacting the rare disease community and the opportunities to improve health and wellbeing broadly if these are addressed will be published on Rare Disease Day 2026, and contributes to global strategy, cross-sector investment frameworks and implementation pathways.

## World Health Assembly Resolution on Rare Diseases

In May 2025, the World Health Assembly (WHA) unanimously adopted the WHA Resolution on Rare Diseases, formally recognising rare diseases as a global health priority. The next phase involves development of a Global Action Plan for Rare Diseases.

As an early member of the Coalition for Advocacy for Rare Disease Equity (CARE), Centre Co-Director Sue Baker was invited to speak at the international webinar: *Moving the WHA Resolution on Rare Diseases Forward: From Promise to Action* (January 2026).

Her contributions focused on collaboration, implementation and the role of healthcare practitioners in translating policy into action. This engagement positions WA within the global policy dialogue on rare disease system reform.

*“Your perspectives on collaboration, implementation, the Global Action Plan and the role of healthcare practitioners and the clinical community in these initiatives were incredibly insightful and helped shape a rich and meaningful dialogue. Your expertise and commitment to improving the lives of people living with a rare disease are greatly appreciated. We look forward to working together to ensure that the WHA Resolution translates into tangible progress at all levels.”*

Alanna Miller, Global Policy Lead for Rare Diseases International

## Rare Diseases International – Lancet Commission

Medical Director Gareth Baynam was selected as a Commissioner and Steering Committee member for the *Rare Diseases International – Lancet Commission on Rare Diseases*.

As one of 27 Commissioners representing six continents, Gareth leads the Diagnosis theme, developing evidence-based and equity-focused international recommendations that can be adapted globally.

In November 2025, the Lancet Commission, Rare Diseases International and the Hong Kong Genome Institute co-hosted the International Genomic Medicine Symposium. Gareth presented on leveraging emerging technologies, including AI, to transform diagnostic pathways.

Following this event, Gareth visited Peking Union Medical College Hospital, Beijing and Children's Hospital of Fudan University, Shanghai to consolidate longstanding partnerships and support the launch of the *Undiagnosed Disease Network for Children* in China, spanning leading children's hospitals across the country.

## Year 4 Impact

**In Year 4, the Centre:**

- **Launched globally accessible rare disease education resources for schools**
- **Increased public visibility through Rare Disease Day initiatives**
- **Strengthened clinical workforce awareness and engagement**
- **Contributed to global economic and policy frameworks through the WEF**
- **Engaged in implementation dialogue following adoption of the WHA Resolution**
- **Led international diagnostic reform through the Lancet Commission**
- **Expanded long-standing partnerships across Europe, Africa, the Americas and the Asia-Pacific**

**To change the outcomes for future families our impact must extend to the ecosystem in which we operate including education systems, global policy platforms, workforce development and international research collaboration.**

# Critical Enablers

**The Centre's impact is amplified by cross-cutting themes of work that underpin the Patient Journey Framework and catalyse outcomes across each stage. These critical enablers strengthen how the Centre delivers care, builds capability, translates innovation and influences the broader system for children and families living with rare and undiagnosed disease.**

In Year 4, these enablers continued to mature, helping the Centre deliver better care today while building foundations for the next stage of growth.

## Digital & AI

### Turning complex information into practical support

Rare and undiagnosed disease generates complex information. Families are often asked to repeat their story across services and hold the burden of being experts in their own rare disease. Clinicians must interpret specialised evidence that is spread across different systems and sources. The Centre's digital work is focused on reducing this burden, improving clarity and helping the right information reach the right person at the right time.

The Centre's approach to digital tools and AI is grounded in practical benefit. The goal is straightforward: make rare disease information easier to understand and easier to use, so families can access support faster and clinicians can make decisions with greater confidence.

### Functional Consequences Reporting

One of the most practical examples is the Centre's AI-enabled pipeline that produces Functional Consequences reports. These reports take complex clinical information and translate it into clear, structured descriptions of how a child's condition affects everyday life. This is particularly important for disability-related systems such as the NDIS, where eligibility and supports often depend on describing functional impact in plain language, not just providing a medical diagnosis.

Each report describes impact across six day-to-day areas: communication, social interaction, learning, mobility, self-care and self-management. The content is based on clinical evidence and aligns with recognised international frameworks, including the World Health Organisation's International Classification of Functioning, Disability and Health (WHO ICF) and World Health Organisation Disability Assessment Schedule (WHODAS 2.0), to support consistency and reliability.

Most importantly, the reports are designed to reduce load on families. By automating and standardising a process that is usually time-consuming and emotionally difficult, this approach supports more timely access to services, and helps families, planners and service providers build a shared understanding of a child's needs.

### RACE: Rare disease Assistant for Clinical Evidence

The Centre is also developing digital tools to support clinicians with the evidence burden that sits behind rare and undiagnosed disease care. Clinicians often need to quickly locate and interpret information from multiple sources, including clinical guidelines, genetic data and emerging scientific literature.

Rare disease Assistant for Clinical Evidence (RACE) is designed to support this work. In simple terms, it uses an AI-supported approach to review and summarise evidence from trusted sources through three lenses: clinical, genetic and scientific. It then combines those findings into structured recommendations, with transparent sourcing so clinicians can see where the information comes from and make their own judgement.

This supports faster evidence appraisal, more consistent decision-making and stronger confidence in clinical reasoning, while maintaining human oversight.

RACE is currently undergoing clinician testing to validate its accuracy.

### Variant Effect and Phenotype Discovery

A further area of work is the development of a pipeline that helps researchers better interpret uncertain genetic variants by linking them to clinical presentation. This is highly technical work, but the intent is practical: improve our ability to understand what a genetic finding might mean for a child's symptoms and care.

The pipeline brings together different evidence sources, including evolutionary signals, laboratory data and clinical phenotype mapping using the Human Phenotype Ontology. This supports more integrated interpretation and, over time, contributes to improved knowledge and diagnostic capability.

### Digital Health Measurement Collaboration

This year the Centre partnered with the Digital Medicine Society's Digital Health Measurement Collaborative Community (DATAcc) to develop a core set of digital clinical measures for paediatric rare diseases. The purpose of this work is to strengthen consistency in how outcomes are measured and reported, which supports therapy development and better evaluation of what works.

The Centre contributed at strategic and operational levels and was used as a real-world example of an innovative approach to patient care journeys.

### Digital collaborations and partnerships

Year 4 also included ongoing collaboration across research, technology and implementation partners, including the Wilhelm Foundation; Rare Compute; Rare Molecule Foundry; Fujitsu; A*STAR and KKH Women's and Children's Hospital, Singapore; LeadPath Healthcare Innovations, Dubai; University of Texas; UCLA; Dell Medical School; Peking Union Medical College Hospital; Tsinghua University; Children's Hospital of Fudan University, Shanghai; the University of Tasmania; and youth innovation partners through Bloom.

These partnerships are building practical capability around diagnosis, data systems and future workforce skills in AI-health, while supporting research translation.

## Global Leadership & Partnerships

### Strengthening local care through international connection

Rare disease is global, and progress is accelerated when systems share knowledge rather than working in isolation. In Year 4, the Centre continued to play a significant international leadership role, contributing to initiatives that strengthen care, pathway development, research collaboration and system reform. This work does not sit separate from local delivery. It strengthens the knowledge, partnerships and momentum that improve outcomes for children and families in WA.

### Global collaboration to advance rare and undiagnosed disease

A major milestone this year was the formal partnership announced on Global Undiagnosed Day, 29 April 2025, between Rare Diseases International – the global peak body for rare disease, the Wilhelm Foundation – the global peak body for undiagnosed disease and the Centre. This collaboration brings together global leadership in rare and undiagnosed disease to improve diagnosis, care and support for people and families worldwide.

For the Centre, this partnership is significant because it moves beyond connection into shared delivery. It positions the Centre as a key partner in strategic initiatives spanning clinical service development, care pathway design and proof of concept work for the Global Network for Rare Diseases. It also reflects growing international confidence in the Centre's model, expertise and capacity to contribute practical leadership in this field.




## Rare and Undiagnosed Disease Care Pathway

Through this partnership work, the Centre is co-leading development of a high-level, disease-agnostic care pathway for rare disease. The aim is to create a pathway that can be adapted across different health systems and geographies while still reflecting the realities people living with a rare disease face.

The Centre's contribution operates at three levels.

1. a Senior Project Nurse is leading the research, evidence gathering and pathway drafting.
2. Medical Director Gareth Baynam chairs the international Working Group, which brings together global experts to shape the pathway's core content and direction.
3. Centre Co-Director Sue Baker chairs the Review Group, which provides structured review and challenge to ensure the pathway is practical, relevant and applicable across different contexts.

Together, these groups involve 52 global stakeholders, bringing perspectives from multiple countries, systems and lived experience contexts. This layered approach helps ensure the pathway is not developed in isolation but is tested against the realities of care delivery across diverse settings.

The value of this work is greater consistency in how families are supported, clearer shared language across the journey and a stronger foundation for improving rare and undiagnosed disease care internationally and locally.

## International taskforces and commissions

The Centre's leaders also contributed to a range of international taskforces, commissions and advisory groups focused on persistent challenges in rare disease. These roles are important because they shape the frameworks, recommendations and partnerships that influence how health systems respond to rare and undiagnosed disease over time.

This work includes leadership and participation in International Rare Diseases Research Consortium taskforces and other global advisory groups, with a focus on translating evidence into practical recommendations. It includes executive membership, co-chairing and membership of taskforces progressing work on stigma, care coordination, diagnosis-to-therapy pathways and preparedness for genetic N-of-1 treatments.

The Centre's international leadership also extended through direct collaboration with major institutions and networks. In 2025, Gareth was invited to work with leading children's hospitals across China to establish the Undiagnosed Disease Network for Children, with the Centre's cross-sector care coordination model adopted as a foundational framework. Anna Thetford presented the Centre's model at the 2nd International Conference on Clinical Research Networks for Rare Diseases in Germany, sharing practical experience from WA with an international audience.

This leadership was further reflected in appointments to a range of international groups and initiatives, including the Lancet Commission on Rare Disease, the Wilhelm Foundation Advisory Council, the WHA Rare Disease Resolution Coalition, the European Rare Disease Research Alliance, RareBoost, the African Rare Disease Initiative and the RareCompute Foundation.

These partnerships matter because they do more than raise the Centre's profile. They connect WA into global networks of expertise, strengthen local capability and ensure that children and families here benefit from international knowledge, collaboration and momentum. In Year 4, the Centre continued to demonstrate that global leadership is one of the ways local impact is strengthened.

# Global Nursing Network
## Rare Diseases

### Mobilising the world's largest healthcare workforce

Nurses are the world's largest healthcare workforce and are on the frontline of patient care. For many people living with rare and undiagnosed disease, a nurse is the most accessible health professional and sometimes the only one available. That makes nursing capability, connection and leadership critical to improving outcomes for children, adults and families living with rare and undiagnosed disease.

Since launching in March 2023, the Global Nursing Network for Rare Diseases (GNNRD) has continued to grow in both reach and influence. By February 2026, the network has reached 824 members across 66 countries. This growth reflects more than membership numbers. It signals increasing recognition of GNNRD as a trusted global platform for nursing education, collaboration, advocacy and leadership in rare disease care.

In Year 4, that growth translated into practical outputs. The GNNRD launched its online Resource Hub, providing free rare and undiagnosed disease materials for nurses around the world, many translated into multiple languages. The network was also identified in Rare Diseases International's public survey of rare disease expertise, centres and networks as one of the most frequently cited international networks respondents were members of, a notable achievement for a network that has existed for less than three years.

GNNRD also contributed to global policy activity through Rare Diseases International's coalition supporting the WHA rare disease resolution adopted in May 2025. This is important as it positions nurses within the broader international movement to improve recognition, care and support for people living with rare disease.

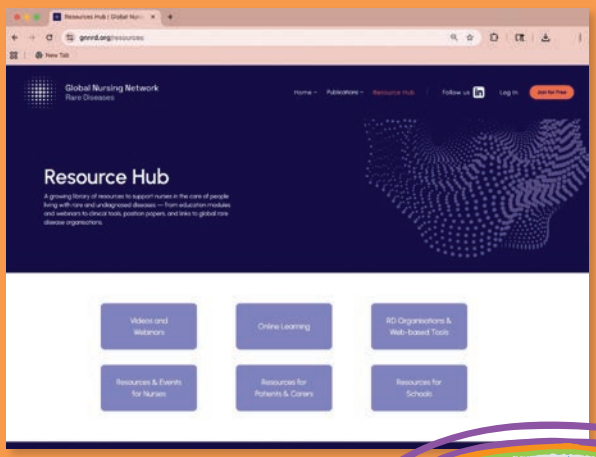
Education remained a major focus throughout the year. In partnership with Medscape, the GNNRD developed three free educational video resources for nurses, helping strengthen awareness and practical understanding of rare disease care. The network also made strong progress on its Comprehensive Learning Program; a three-tier certificate pathway designed to build nursing capability over time. Content for the first level has now been written, reviewed and is being built into a modern e-learning platform. In parallel, GNNRD partnered with Medics for Rare Disease to adapt the well-regarded Rare Disease 101 modules for nurses, with launch planned for Rare Disease Day 2026.

The network also supported broader awareness work by hosting and promoting *The Weight of Rare Disease* animation and maintaining regular communication with members through quarterly newsletters. GNNRD members also contributed to international knowledge sharing through presentations and participation at conferences including the SingHealth Nursing Conference and the World Nursing & Healthcare Summit.

## Launch of
## RD 101 (Nursing)

A freely-available foundational eLearning course developed in collaboration with **M4RD**


## New online resources hub




## Global Engagment and Representation


Geisa Luz & the Wilhelm Foundation, Brazil


Jasmine Goh, SingHealth Nursing Conference

## Global connections for patient outcomes

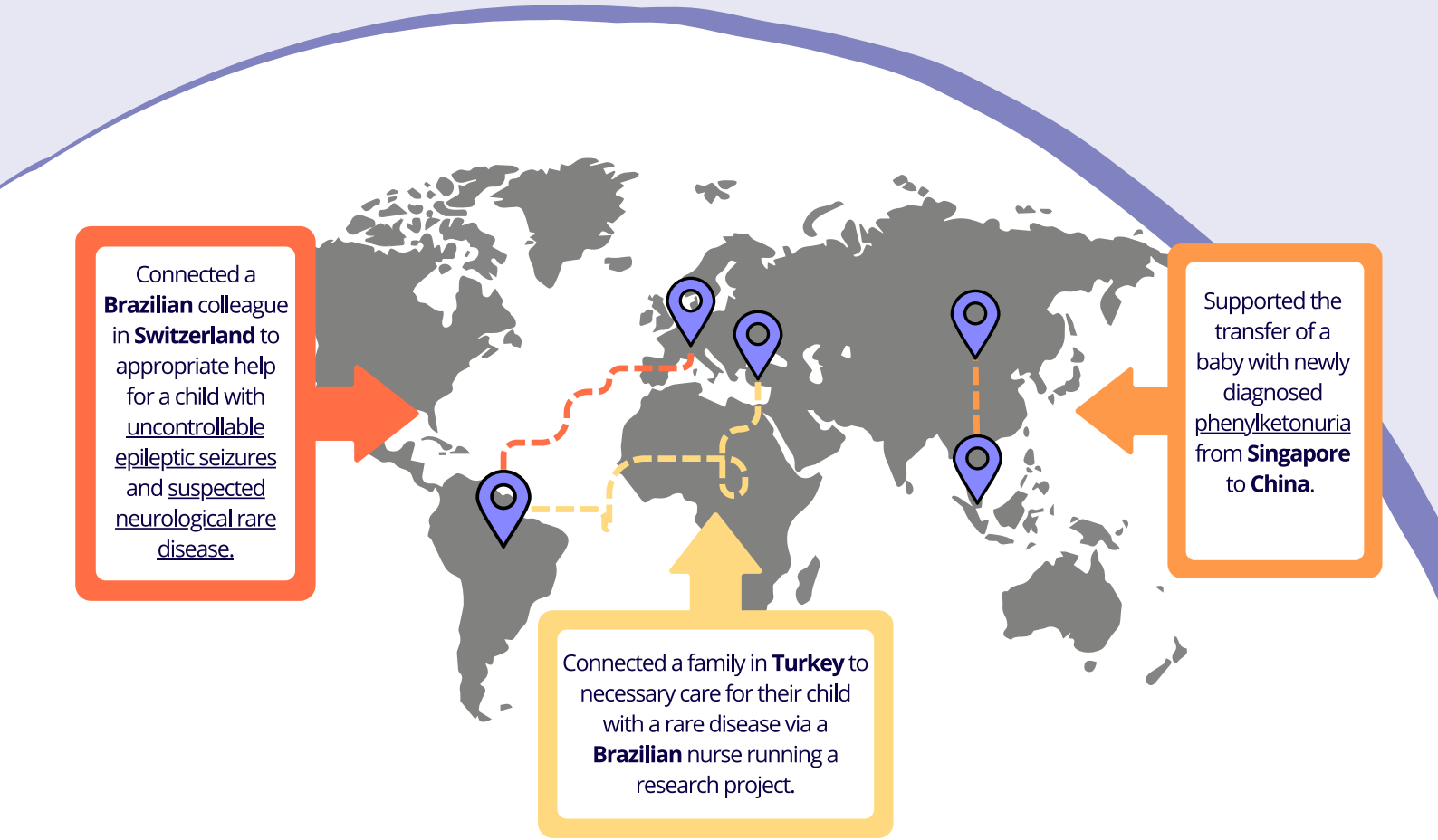
One of the most important strengths of GNNRD is its ability to connect nurses across countries in practical ways that directly support patient care. Through its Clinical Questions space and secure networking platform, nurses have been able to seek advice, share expertise and help connect families to appropriate care beyond their own health system or geography.

In 2025, this included examples such as connecting a Brazilian colleague in Switzerland to appropriate support for a child with uncontrollable epileptic seizures and suspected neurological rare disease, supporting the transfer of a baby with a newly diagnosed rare disease from Singapore to China, and linking a family in Turkey to necessary local care through a Brazilian nursing research project. These examples demonstrate that global nursing connection is not abstract. It can directly influence access, continuity and coordination for children and families.

## Removing language barriers with Wordly

In 2025, GNNRD introduced Wordly, an AI-enabled real-time translation and captioning platform to support multilingual collaboration. Wordly provides live captions, subtitles and transcripts in more than 50 languages, enabling nurses and lived experience contributors to participate more fully in meetings, webinars and project work.

This has strengthened equity within the network by making it easier for nurses to contribute in their preferred language and to engage in complex discussions without the requirement of English fluency. It has also improved the quality of collaboration across committees and projects by widening participation and reducing reliance on interpreters.



Feedback from members has been strongly positive. As Geisa Luz from Brazil reflected:

*"Using Wordly truly made me feel part of a global network of nurses who share the same values and are driven by the same purpose. It was more than just a translation tool, it was a bridge that enabled meaningful dialogue across different languages and cultures, strengthening our collective voice in rare diseases. For me, it was also an opportunity to feel less alone in a universe where so many patients live isolated due to the lack of knowledge and awareness about their conditions. Thank you Wordly, for helping us connect beyond borders."*

## Committee leadership and delivery

As the network has matured, so too has its committee structure. In Year 4, the work of the committees was central to turning GNNRD from a growing network into an active delivery platform.

The Nursing Reference (Leadership) Group continued to provide leadership, governance and quality oversight across the network's work. Members supported review processes, contributed to content development and acted as champions for GNNRD within their own countries, helping extend the network's credibility and reach.

The Education and Learning Committee had a particularly productive year. Its work included strong input into the curriculum and content for the Comprehensive Learning Program, webinar planning and development priorities for future resources. The appointment of a student nurse representative also broadened perspective and helped ensure the program remains relevant to the future nursing workforce.

The Research and Innovation Committee was established during the year, strengthening the network's research foundations. Its first major project focuses on defining rare disease nursing roles, scope and competencies. This work will support future position papers and recommendations, helping clarify the role of nurses across rare disease care internationally.

The Lived Experience Advisory Group continued to play an important role in ensuring that the voices of people living with rare and undiagnosed disease, and their families, remain embedded across the network's work. During the year, members contributed to setting research priorities, reviewing care pathway work and informing the design of educational resources, including the rare disease education animation and the Comprehensive Learning Program.

## Regional growth

Regional network development also accelerated during 2025.

The Brazil Regional Group maintained active engagement through scientific events, education activities and patient and family initiatives, supported in part through partnership with the Association of Families, Friends and People with Disabilities, Serious and Rare Diseases (AFAG). This regional activity has strengthened local education, advocacy and workforce development while remaining connected to the broader mission of GNNRD.

The Asia-Pacific Regional Group formally commenced in April 2025, following planning in Singapore in 2024. Throughout the year, the group contributed to conferences, newsletters, education and advocacy activities. Its growing role reflects the importance of regionally led collaboration within a global network.

## Year 4 Impact

**In Year 4, GNNRD continued to strengthen its role as a practical global platform for rare disease nursing by:**

- **growing to 824 members across 66 countries**
- **expanding free multilingual resources for nurses**
- **progressing a structured global learning program**
- **contributing to global policy activity through the WHA rare disease resolution**
- **reducing language barriers through Wordly**
- **strengthening governance, research and lived experience input through its committees**
- **enabling practical cross-border connections that directly supported patient care**

**As Alicia Truelove (member of Education and Learning, and Research and Innovation Committee) from the United States reflected:**

***"What a privilege it's been to serve the global rare and undiagnosed disease community and to collaborate with brilliant nurses united in their passion for rare."***

**GNNRD has moved beyond being an emerging network. In Year 4, it continued to demonstrate that when nurses are connected, equipped and supported, they can play a powerful role in improving rare and undiagnosed disease care around the world.**

Zohra Hasan Ali, supported by the GNNRD to become Pakistan's first Advanced Clinical Genomics Nurse.

# A ‘Comprehensive’ Look Ahead

**Year 4 marks an important transition point for the Centre.**

Over the past four years, the Centre has shown that cross-sector coordinated care can improve outcomes for children and families living with rare and undiagnosed disease. It has also shown that current capacity does not match the scale of need. Too many children still face delayed diagnosis, fragmented care, and uneven access to therapies, trials and support.

The landmark commitment of $221.1 million from the Stan Perron Charitable Foundation, together with $25 million from the Perth Children’s Hospital Foundation and $3 million from The University of Western Australia, creates the opportunity to respond at a different scale. It enables the Centre to move from a proven model to a comprehensive, integrated ecosystem with the reach, structure and long-term stability needed to deliver greater impact across WA.

## From Rare Care Centre to Rare Care Comprehensive Centre

The shift to the Rare Care Comprehensive Centre (RCCC) is not a change in purpose. It is a change in scale, integration and permanence.

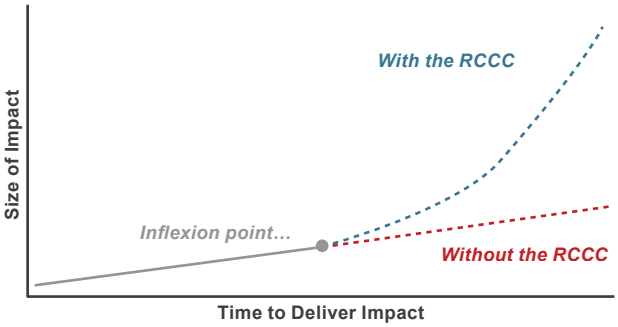
The Centre has already demonstrated the value of cross-sector coordinated care. The RCCC builds on that foundation so more children and families can access earlier diagnosis, more consistent care, stronger support and better connection to innovation and research. At its core is a simple principle: families should not have to hold the systems together themselves.

Children with rare and undiagnosed disease often fall outside eligibility criteria designed for more common conditions. As a result, parents are left navigating multiple systems, repeating information and advocating for support at every step. The RCCC is designed to reduce that burden by bringing care, support, research and system change into one more connected structure.

## A Rare Inflection Point

Western Australia is uniquely placed for this next step. The clinical model is established in practice, partnerships across health, education, disability and research are in place, world leading research is already being undertaken and the case for expansion is supported by evidence.

The investment now committed allows the Centre to move from serving hundreds of families to building a system capable of reaching thousands, without losing the integrity of the model.




Without this scale, progress remains gradual and uneven. With it, earlier diagnosis, coordinated care and access to innovation can become more consistent across the state rather than dependent on geography, timing or circumstance.

## A Clear and Practical Vision

Our vision remains unchanged: to ensure that the 63,000 children and their families living with rare and undiagnosed disease in WA live the best lives possible. What the RCCC changes is our capacity to realise that vision in a more meaningful, equitable and scalable way.

In practical terms, this means families being met earlier by professionals who understand rare and undiagnosed disease, reducing years of uncertainty and repeated referral. It means better use of digital tools and coordinated pathways within a structured statewide system. It means schools, disability systems and health services working with a clearer shared understanding of a child’s needs.

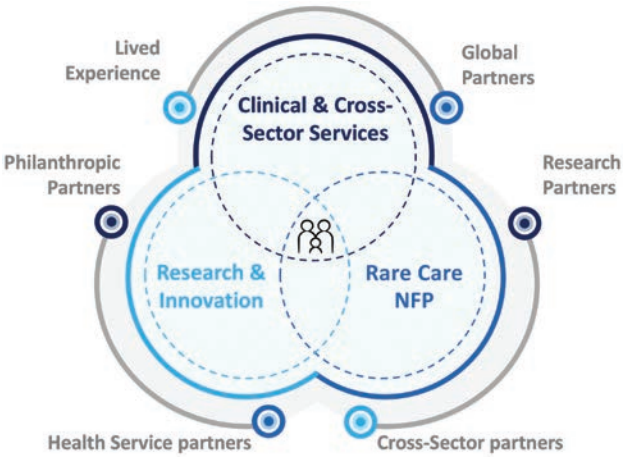
It also means stronger access to clinical trials and emerging therapies, so families are less likely to be forced to relocate or fundraise to access care. Regional families will be better supported closer to home through regional hubs, workforce development and formalised cross-sector partnerships. Nurses and allied health professionals will be better equipped to recognise rare disease earlier and support families across the full journey. Lived experience will continue to shape service design so programs reflect what families actually need.

This next stage is not about adding new layers. It is about reducing fragmentation and building a system that works together around the child and family.

## Delivering the Vision Through an Integrated Structure

Improving outcomes for children with rare and undiagnosed disease requires more than strong individual programs. It requires those programs to work together as one system.

The RCCC brings together three essential components: Clinical and Cross-Sector Services, Research and Innovation, and an enabling Not-for-Profit platform. These are designed to operate as an integrated structure rather than separate streams of work.



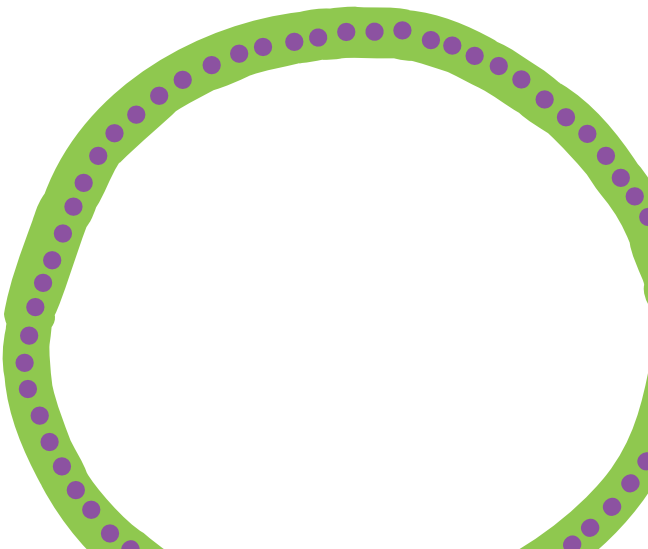

Each component strengthens the others. Clinical and Cross-Sector Services provide the frontline of direct support, bringing together health, disability, education, mental health and community systems into a coordinated model aligned to the Patient Journey Framework. Research and Innovation is embedded within service delivery so clinical challenges inform research priorities and discoveries are translated back into care pathways and practical tools. The Not-for-Profit platform helps sustain and scale impact through family support programs, education, navigation, resources, communications, partnerships and philanthropy.

The strength of the RCCC lies in this integration. Care identifies need. Research and innovation develops the solutions. The enabling platform helps ensure those solutions are sustained, scaled and translated into broader system change.

## An Integrated Commitment

The RCCC represents the next stage of the Centre's development. It builds on strong foundations, measurable outcomes and established partnerships, while recognising the responsibility that comes with significant philanthropic investment.

Its purpose remains the same: to deliver an equitable, agile and enduring system that recognises and responds to the unique needs associated with rare and undiagnosed disease and takes meaningful action to support children and families across the full journey.



# Thank You

**Dear Friends and Supporters,**

As I reflect on our fourth year, I am struck by the strength of the community that stands behind the Centre and the children and families we serve.

This year has demonstrated what can be achieved when compassion is matched with determination, and when commitment is supported by evidence. Across clinics, communities and global partnerships, we have seen families experience clearer pathways, reduced isolation and greater confidence navigating complex systems. The impact outlined in this report represents countless hours of collaboration, persistence and shared purpose.

The landmark support announced this year, including the Stan Perron Charitable Foundation's donation alongside contributions from Perth Children's Hospital Foundation and The University of Western Australia, is a profound endorsement of the work delivered to date. It reflects trust, and it places responsibility on all of us to steward that trust carefully and with integrity.

I am deeply grateful to every donor and partner whose generosity has made this possible.

I am equally grateful to the families who place their trust in us, often during some of the most uncertain moments of their lives. Your insight, resilience and willingness to share your lived experience continue to shape our programs and strengthen our model of care. You remind us daily why this work matters.

To our clinical, research and administrative teams, thank you for your dedication and professionalism. The progress achieved this year has been built on quiet persistence, thoughtful innovation and a shared commitment to doing the work well.

Year 4 has laid stronger foundations. The systems, partnerships and infrastructure now in place provide real confidence that the impact being delivered today can be sustained and strengthened over time. It is a privilege to be part of this journey alongside you.

With sincere gratitude,

**Sue Baker**
Co-Director, Rare Care Centre

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Clinical Centre of Expertise for
Rare and Undiagnosed Diseases

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