



Government of Western Australia
Child and Adolescent Health Service



Integrating rare disease research, innovation, care and support:

Answering the questions families ask

Gareth Baynam

Medical Director, Rare Care Centre

Clinical Centre of Expertise for Rare and Undiagnosed Diseases

Compassion

Excellence

Collaboration

Accountability

Equity

Respect



Whole of Life Impact

People with rare diseases share the same challenges associated with chronic and complex illnesses, along with additional challenges due to the '*rarity*'

Challenges common to chronic/complex illness AND rare diseases

- Inadequate clinical management & fragmented care
- Financial burden
- Inequity of access to care depending on geography
- Lack of peer-support

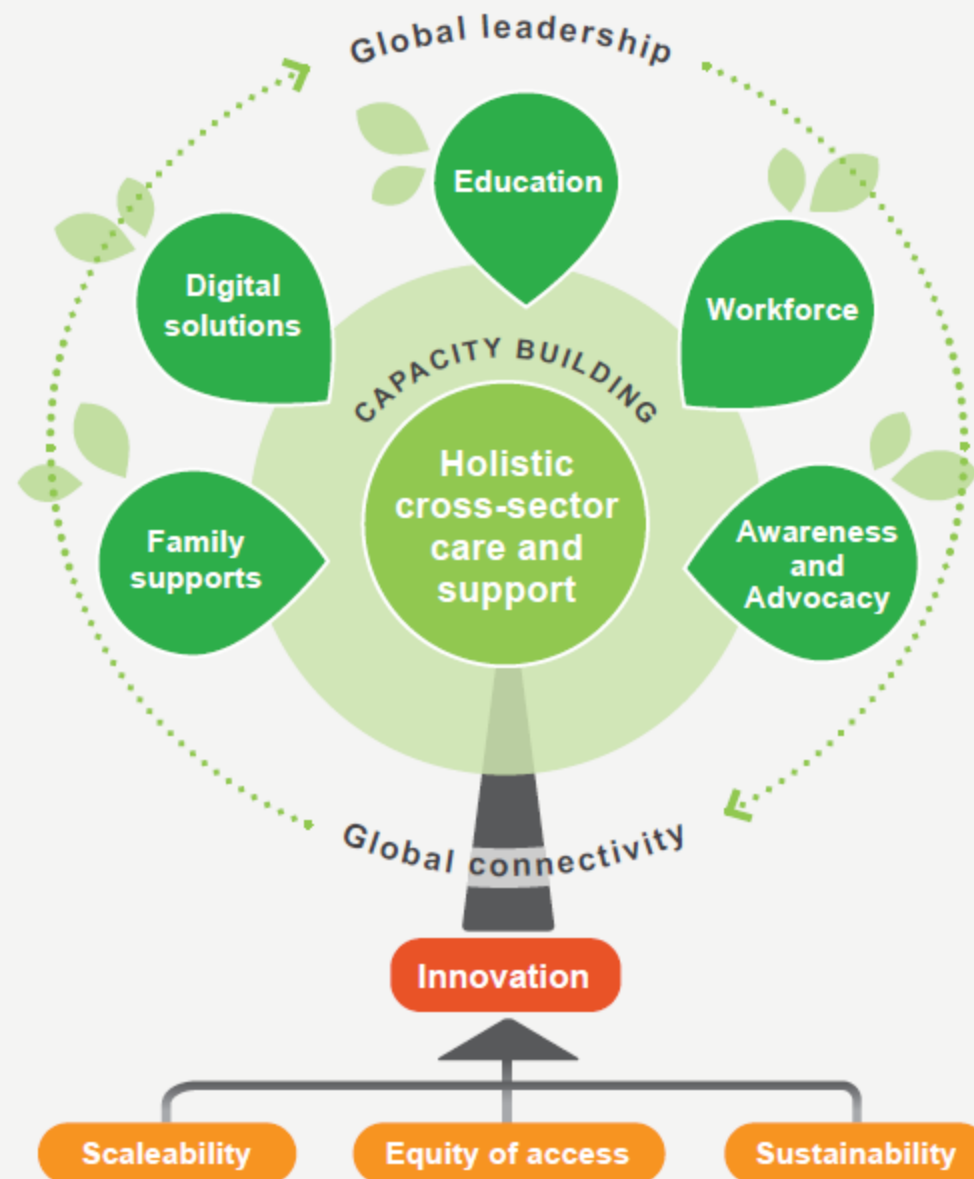


Challenges unique to rare diseases

- Lack of awareness of RD across health professionals & the broader community
- Long diagnostic journey & misdiagnosis / no diagnosis
- Lack of treatment options
- Patient or parent needs to be the 'expert' in their RD
- Higher mental health impact, isolation, stigma & discrimination

Rare Care Centre Framework

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

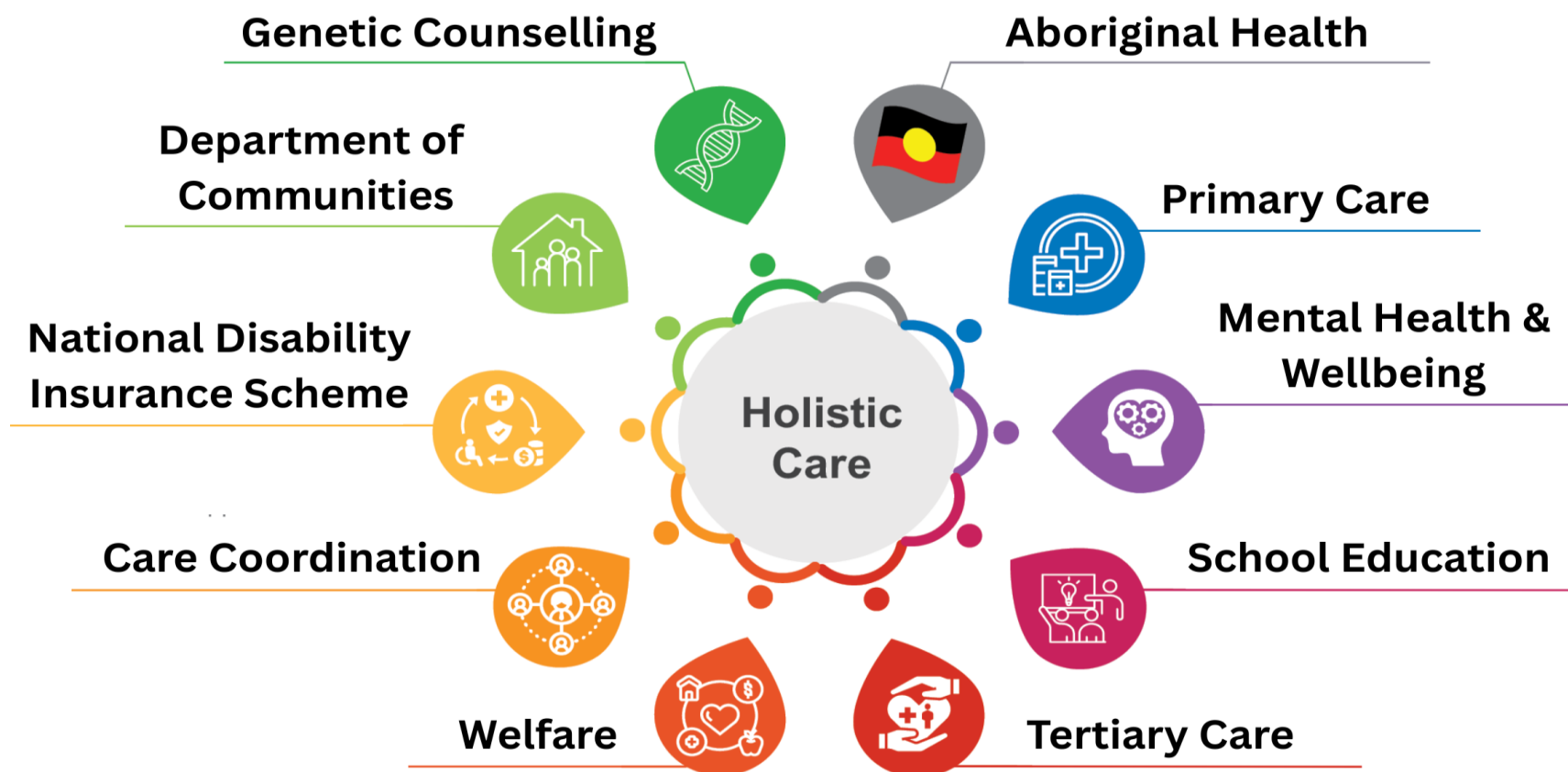


Co-design
through >200
stakeholder
meetings

Rare Care Centre

Est Feb 2022

Model of Care: Cross-Sector Care Coordination





Model of Care Features

Model of care co-designed with all sectors/agencies, clinicians and families

Referral Criteria:



Has a rare or undiagnosed disease



Identified gap/s in care across any sector/agency



Receiving ongoing paediatric care (hospital or paediatrician)



No duplication – add value



Existing sector/agency professionals remain involved in care delivery



High and low complexity conditions



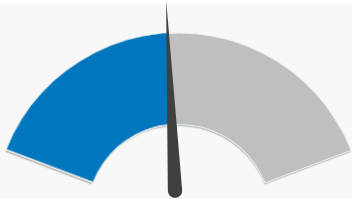
Time-limited, out-patient service (can be re-referred)



Table 1
Area of support and interventions provided.

Program Interventions										
Categories	Clinical Management		Care Coordination				Family Support & Wellbeing			
	Medical management	Genetics	Coordination of health care	Care closer to home	School	Transition to adult services	Family support	Mental health	NDIS	Welfare
Discipline	☐ Paediatrician ☐ GP ☐ Nurses	☐ Genetic counsellor	☐ Paediatrician ☐ GP ☐ Nurses	☐ Paediatrician ☐ GP ☐ Nurses ☐ Aboriginal Health Practitioner ☐ Mental Health Clinician	☐ Paediatrician ☐ GP ☐ Nurses ☐ School Teacher	☐ Paediatrician ☐ GP ☐ Nurses ☐ NDIS Coordinator	☐ Nurses ☐ Aboriginal Health Practitioner	☐ Mental Health Clinician ☐ Genetic Counsellor ☐ Aboriginal Health Practitioner	☐ NDIS Coordinator	☐ NDIS Coordinator
Interventions	☐ Referral to specialised services/resources e.g. - CAMHS - CDS - Neurosciences ☐ Facilitate investigations ☐ Facilitate admission ☐ Enable community pharmacology management ☐ Enable equipment purchase ☐ Facilitate access to clinical trials ☐ Education & sharing of information to primary teams regarding specific rare disease ☐ Facilitate access to second opinions/overseas consultations ☐ Facilitate access to clinical trials/treatments	☐ Provide genetic counselling ☐ Create visual summary ☐ Referral to specialised services/resources	☐ Provide care coordination between specialist teams ☐ MDT meetings ☐ Formal ☐ Informal ☐ Outpatient coordination ☐ Emergency Management Plan	☐ Facilitate access to GP ☐ Identifying patient eligibility for Medicare services through primary care (e.g. GP management plans, health assessments, mental health care plans) ☐ Upskill GP in medical condition ☐ Education of parents/carers about the services accessible through primary care ☐ Procedural care locally ☐ Access to specialised medical care locally ☐ Referral to specialised services/resources ☐ Access to Child Health Nurse	☐ Provide parental support & advice for school enrolment ☐ Support medical documentation to assist school documented plans (e.g. risk or medical management and individualised education plans). ☐ Liaise with school to support shared understanding of education plan between school & family ☐ Liaise to enable clinical support at school and coordinate therapy services ☐ Support the upskilling of school staff ☐ Facilitate access and transition to specialised education support personnel ☐ Liaise with school, including school psychology services to facilitate cognitive assessments.	☐ Enable access to transition resources and information ☐ Liaise with teams to assist transition ☐ Assist with adult welfare resources	☐ Provide education/upskilling to family ☐ Provide information to family ☐ Referral to specialised services/resources ☐ Enable peer group linkage ☐ Language translation	☐ Referral to specialised services/resources ☐ Enable access to MH community supports ☐ Provide psycho education and MH support ☐ Provide genetic counselling	☐ Assist families to understand NDIS eligibility application ☐ Assist families to apply for early or unscheduled NDIS plan review ☐ Facilitate access and complete appropriate referrals to NDIS Providers for Therapy and/or Community supports ☐ help build understanding of AT (Assistive Technology) re purchases and evidence required ☐ Upskill family in understanding and implementing the current NDIS plan. ☐ Upskill family to use the NDIS Portal and NDIS App ☐ Assist NDIS therapy team with reports and documentation	☐ Provide guardianship information ☐ Assist with carer payment application ☐ Provide disability support pension information ☐ Enable peer group linkage ☐ Referral to financial management services for assistance

Demographic Snapshot

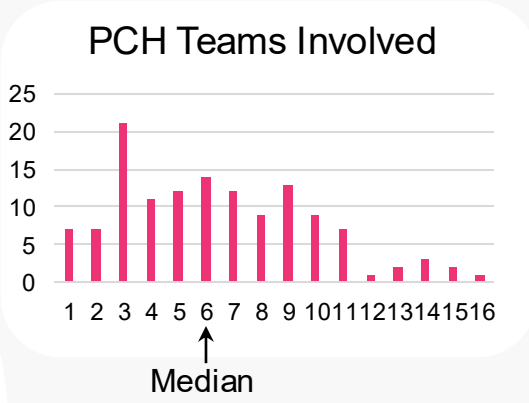
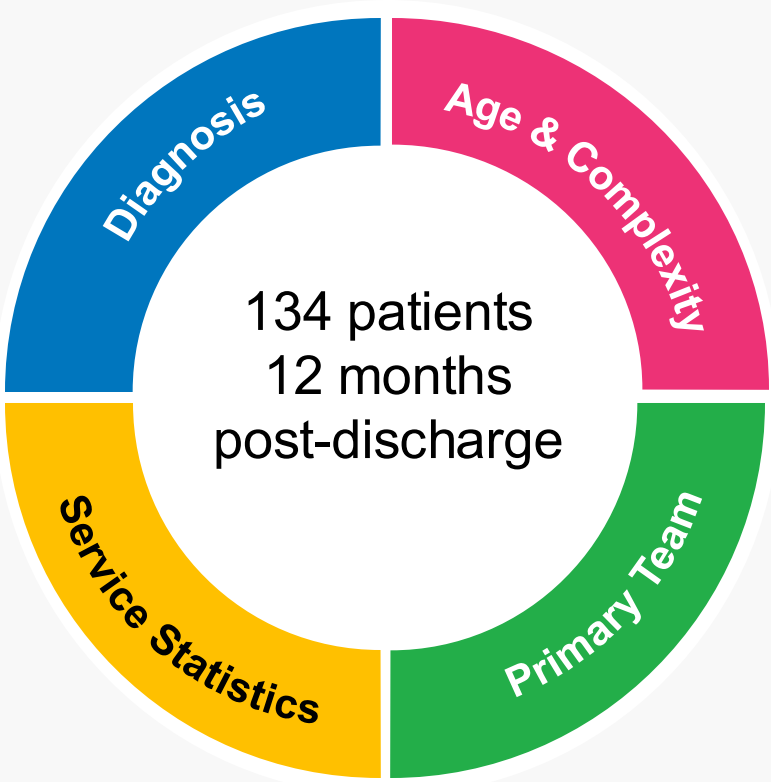


40% Undiagnosed

Most common conditions seen: 22q, CHARGE Syndrome, Tuberous Sclerosis Complex (TSC)

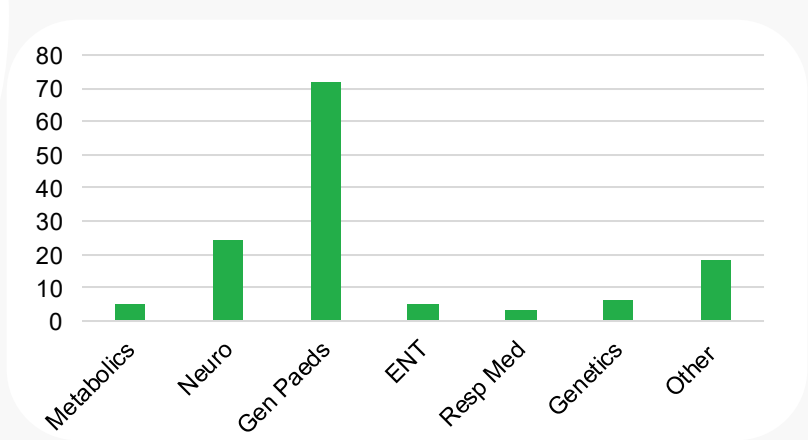
Referral Acceptance
78%

Median time on service
5.1 months



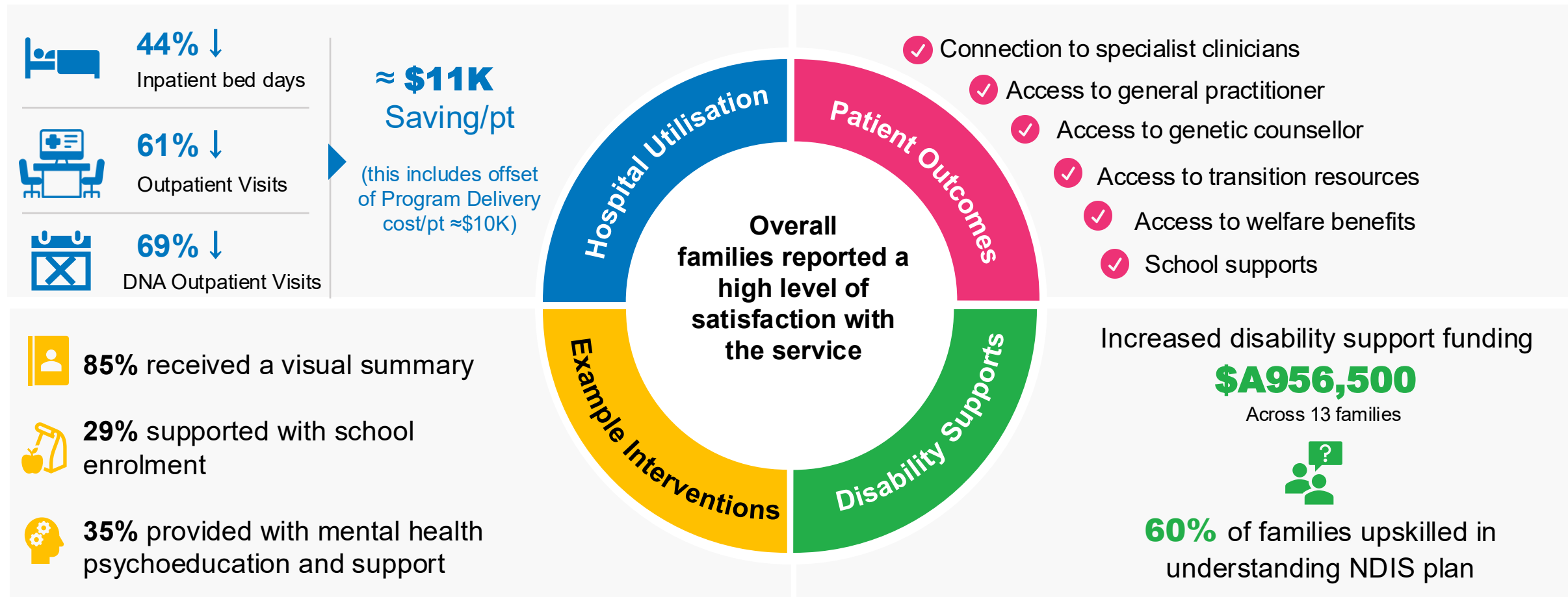
Median Age
8yrs

Age Range
0-18yrs



Impact Snapshot

(134 patients 12 months post-discharge)



Benefit Cost Ratio: \$4.70 for every \$1 spent

Costing as per CAHS Business Intelligence Unit Dec 2025

Nurse Navigator telehealth service: Feb 2024-current

The diagram illustrates the layout of the Cam RARE Patient Passport form, showing its various sections and fold lines. The form is divided into several sections:

- THIS IS ME** (Personal details): Includes fields for Name, Known as, Date of birth, UMRN, Completed by, and Updated on.
- CONTACTS** (Medical and emergency contacts): Includes fields for Medical specialist, GP Surgery, and Emergency contact, each with a corresponding Phone field.
- DIAGNOSIS / SYMPTOMS** (Primary and additional diagnoses): Includes fields for Primary diagnosis, Key clinical features / symptoms, and three additional diagnosis or symptoms sections.
- CLINICAL INFORMATION** (Medications, emergency care frequency, seizures, respiratory issues, allergies): Includes fields for Medications and dosage, Is emergency care frequently required? (Yes/No), Seizures (Yes/No), Respiratory issues, and Allergies.
- TOP 3 THINGS TO KNOW** (Key points for medical staff): Includes three numbered fields for key information.
- ADDITIONAL INFORMATION** (Additional details): Includes a large field for additional information.

The form is designed to be folded into a compact size for easy carrying. Fold lines are indicated by arrows and labels: Fold 1, Fold 2, Fold 3, and Fold 4.

Key Findings

- Access and equity
- Effectiveness
- Efficiency
- Appropriateness

Opportunities for improvement

- Integration with existing systems
- A streamlined model of care
- Embedding nurse navigators across the health care system
- National training and certification
- A mixed model of ongoing support
- Improved data and monitoring systems
- Stronger partnerships and multidisciplinary collaboration

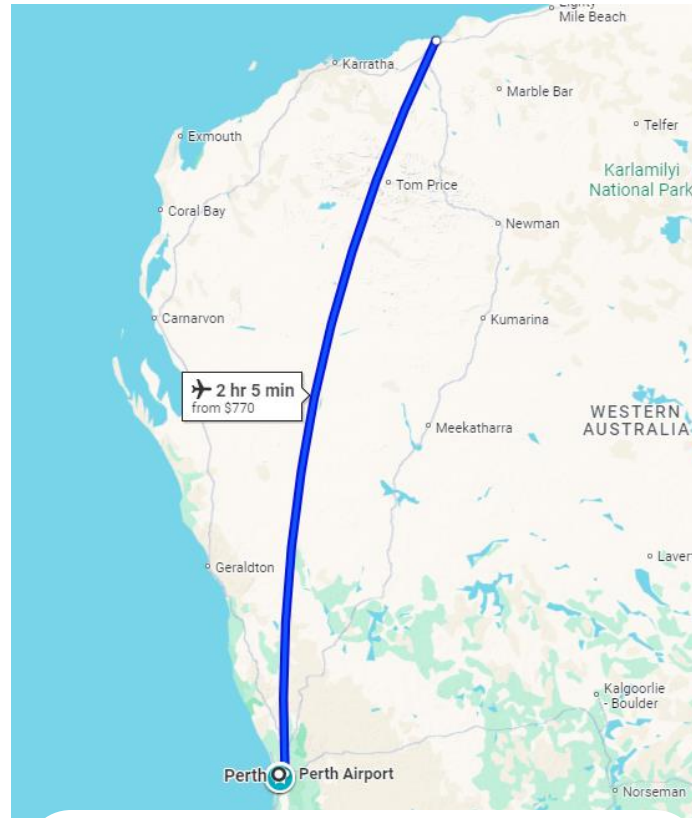
Cam RARE Patient Passport: Any rare condition, any age, anywhere

Regional Hub – Pilbara

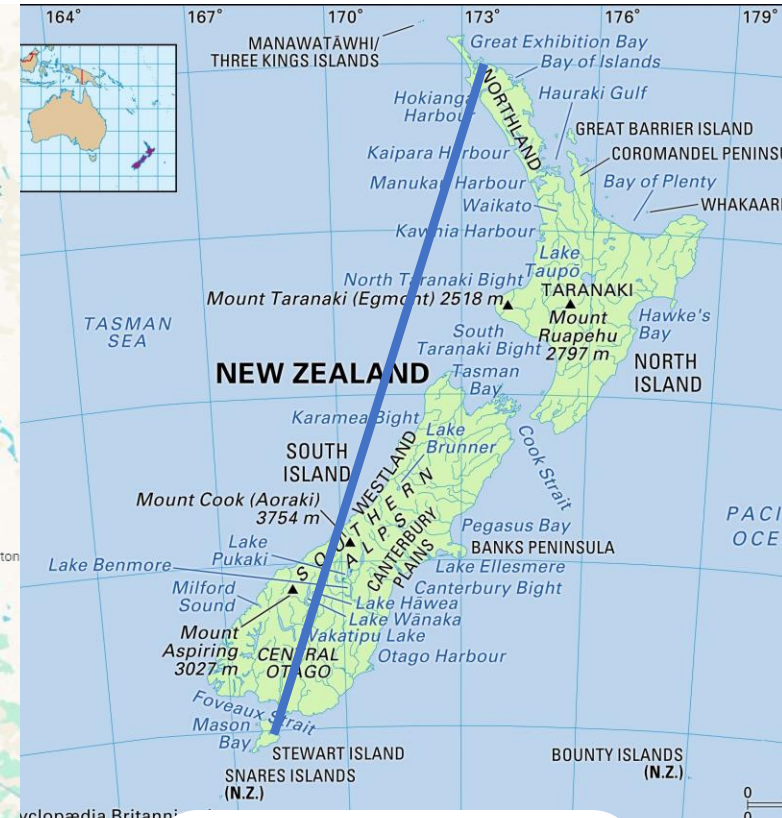
- Remote, Indigenous and Rare
- Disproportionate rates of burden of disease and health disparities, and lack of access to health care services
- Community co-designed model of cross-sector care coordination
- Site based along with outreach/telehealth
 - Equitable and direct access to care
 - Regional capacity building



The Pilbara, WA



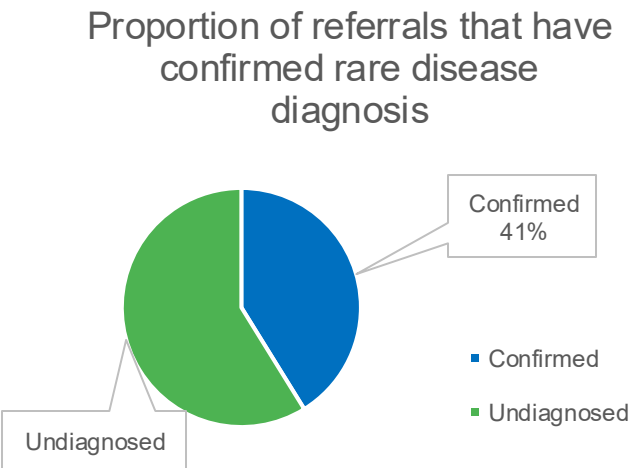
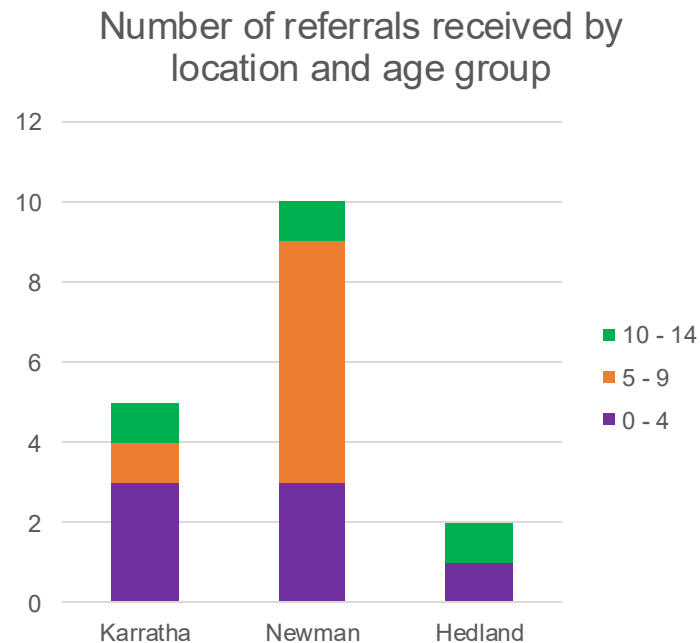
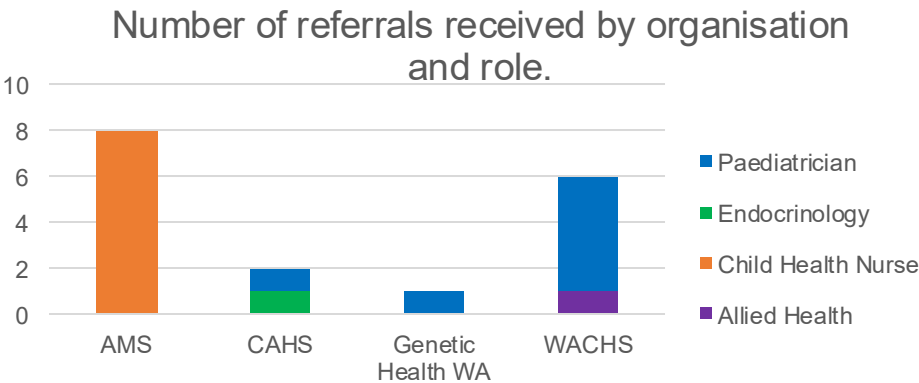
Perth, WA → Port Hedland, WA



Length of New Zealand

Rare Care Pilbara Hub

- Pilbara Hub commenced in February 2026.
- The Pilbara Hub provides clinics in Newman, Hedland and Karratha.
- The service has received 17 referrals to the end of April 2026.
- The service has seen 5 children to the end of April.
- Common gaps in care identified by referrer are family supports, medical management planning, NDIS support and education supports.



Rare Care Comprehensive Centre

RARE
REVOLUTION
MAGAZINE

RARE BITE

Western Australia's largest
philanthropic gifts is set to change the
lives of children and families living with
rare or undiagnosed diseases

RARE RARE Revolution Magazine®
18(M) fortnightly



Government of Western Australia
Child and Adolescent Health Service

Perth Children's Hospital

You are here » [Home](#) » [About Us](#) » [News & events](#) » Landmark investment transforms rare disease care for WA families

Landmark investment transforms rare disease care for WA families

October 3, 2025

A historic philanthropic donation will transform the diagnosis, treatment and support of children and families across Western Australia living with rare and undiagnosed disease (RUD).

The Stan Perron Charitable Foundation has pledged \$221.1 million over the next decade to establish the Rare Care Comprehensive Centre (RCCC) – a vital, world-leading hub at Perth Children's Hospital – to deliver holistic, lifelong care for those affected by RUD.

This transformative gift is supported by additional contributions from the [Perth Children's Hospital Foundation](#) (\$25 million) and the [University of Western Australia](#) (\$3 million).

Rare disease affects more than 300 million people globally, including over two million Australians and 63,000 children in WA.

The RCCC will address the urgent and unmet needs of these families by creating a coordinated network of care, spanning diagnosis, treatment, support, and wellbeing.

A global first in rare disease care

Building on the success of [Perth Children's Hospital's existing Rare Care Centre](#) and its internationally recognised research expertise, the RCCC is a world first initiative.

It will integrate services across health, education, disability, and social sectors, ensuring families receive comprehensive and continuous support.

The RCCC is structured around three core pillars:

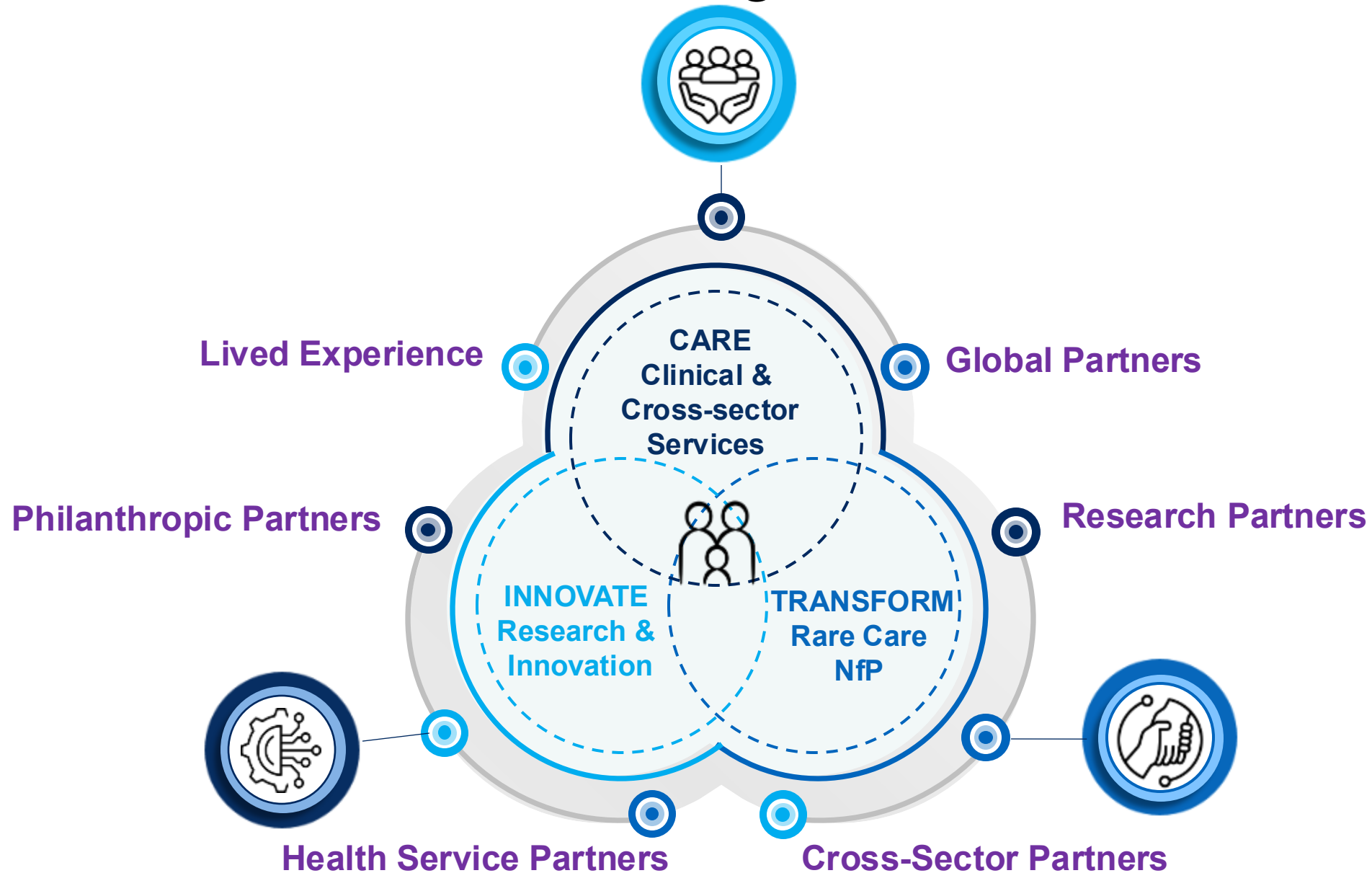
- Clinical and cross-sector services: delivering coordinated care across multiple sectors.
- Research and innovation: advancing cutting-edge science, clinical trials, and technologies.
- Rare Care not for profit: expanding impact through family support programs and initiatives such as the Global Nursing Network for Rare Diseases.

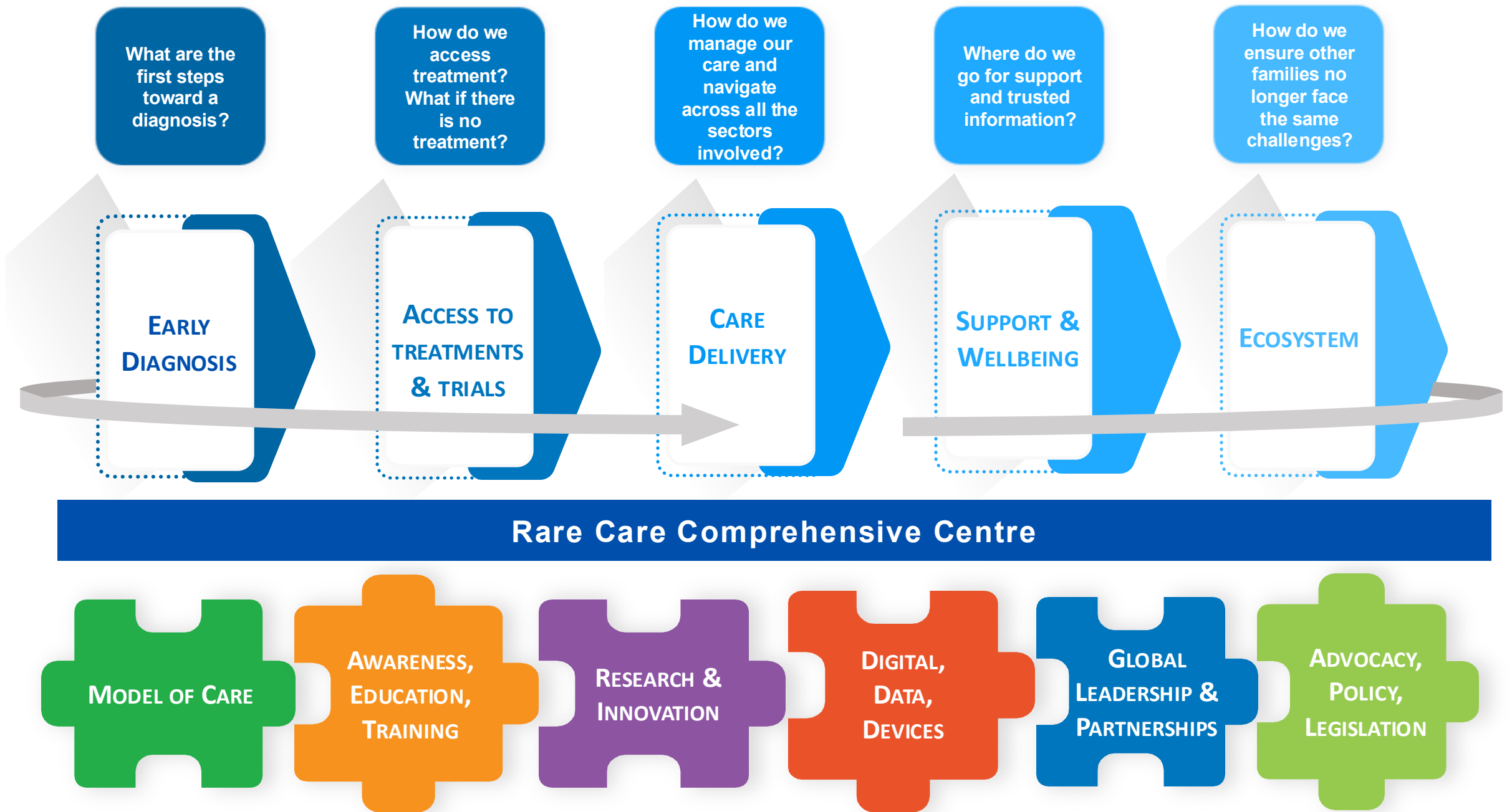


Sue Baker, Rare Care Centre Program Director, Prof Gareth Baynam, Rare Care Centre Program Director, Steve Hill, Elizabeth Perron AM, Executive Director, Stan Perron Charitable Foundation, Ron Meredith Hammett MLA, Minister for Health, Mental Health, the Gilpin family and Kaila Stevens, Rare Care Centre Program Lead (non-clinical)

[Landmark investment transforms rare disease care for WA families](#)

RCCC Structure – an integrated model





Clinical & Cross-Sector

Clinical & Cross Sector

Delivered through CAHS:

- Deliver direct care to children and families across WA through various care coordination models, with a strong focus on cross-sector (inter-agency) care coordination
- Three Programs: Cross-Sector Care Coordination, Nurse Navigator, RUD Professional Support Line
- Three Regional Hubs: Pilbara (established), Kimberley, South-West

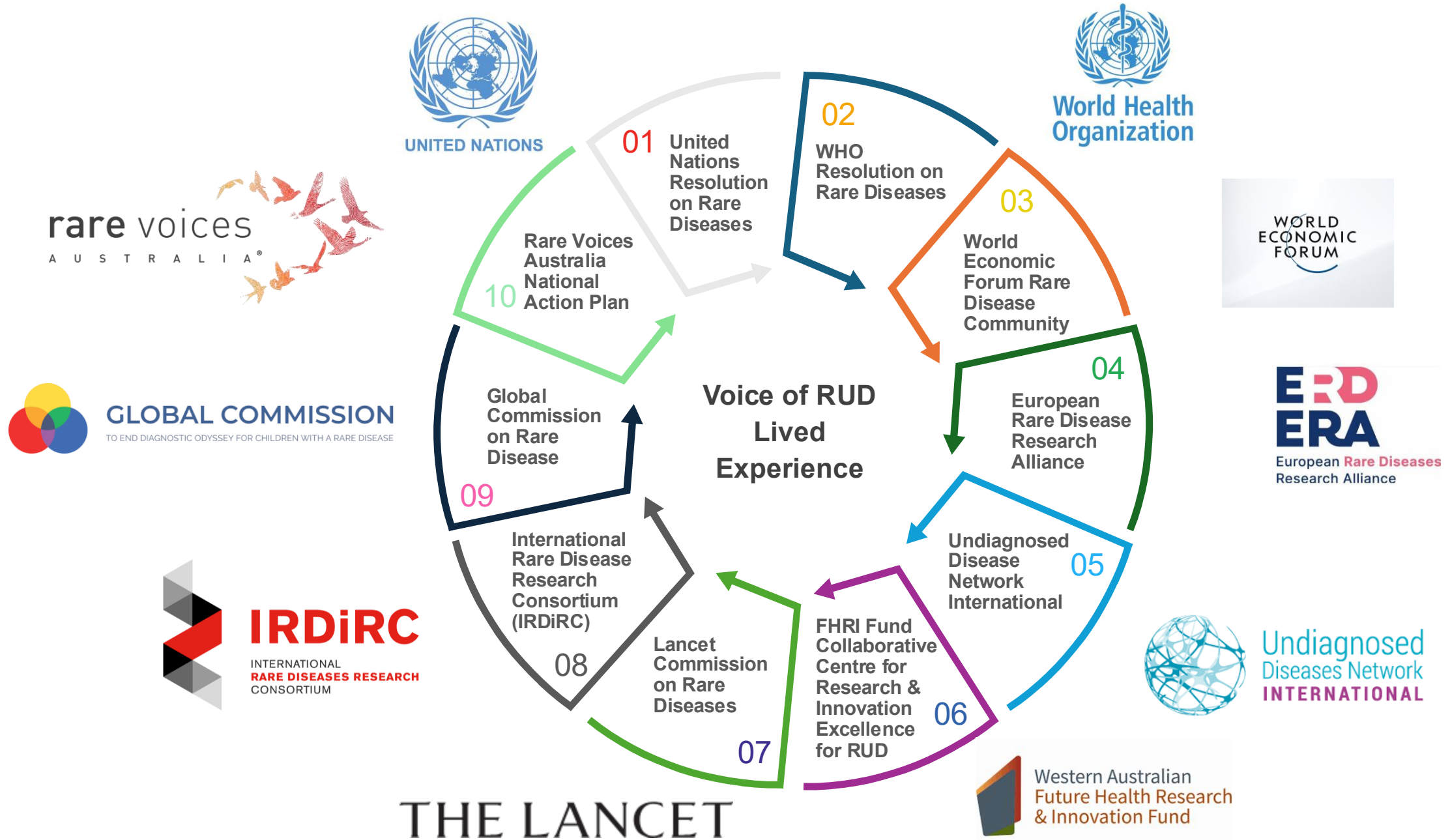
Not for Profit

Not for Profit

Delivered through new dedicated NfP:

- Integration and reporting function
- Lived experience backbone for whole RCCC
- Advocacy, awareness, education and system improvements
- Family Support Programs
- Global partnership and leadership initiatives
- Global Nursing Network for Rare Disease
- Primary Care Navigation Program – PRIMARY RARE
- Cross-sector Pathways
- Digital initiatives
- Research flagships (partial)

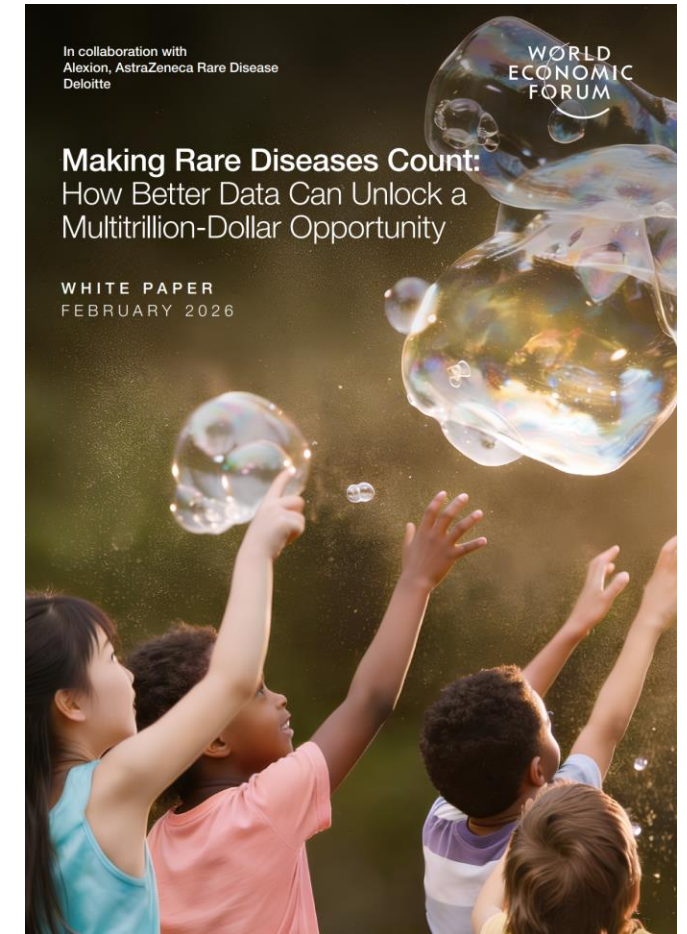
Informing a Rare Disease Research Framework



Global Priority

– enormous unmet need, staggering opportunity

- United Nations Resolution on RD
- World Health Organisation Resolution on RD
- Global Rare Disease Plan
- The Lancet Commission on Rare Disease
- World Economic Forum
 - Asia-Pacific Healthcare Sustainability, KPMG and partners
 - Rare Disease Affinity Group
 - Making Rare Disease Count



Official Launch of Australia's Top 10 Rare Disease Research Priorities

The first meeting for the new Australian Rare Disease Research Network (ARDRN) will take place on **18 September 2025 between 1pm and 2pm AEST**. The ARDRN will be facilitated and chaired by RVA and SMAC members, Clin/Prof Gareth Baynam, Dr (Elizabeth) Emma Palmer and Dr Lisa Ewans. The virtual event will also mark the official launch of Australia's Top 10 Rare Disease Research Priorities report. A discussion will be facilitated about the Top 10 priorities and the next possible best steps in terms of their implementation. All researchers are welcome to join the ARDRN. Learn more about the ARDRN and register via [this web page](#).



Australia's Top 10 Rare Disease Research Priorities

1

Development of, and access to, the best treatments and cures, including clinical trials



2

Awareness and education for health professionals and service providers



3

Diagnosis, including screening



4

Networked centres of expertise



5

Data collection and use



6

Genetic testing



7

Person-centred best practice care and support



8

Psychosocial and mental health impacts and support



9

Access to resources and information for Australians with rare disease, families and carers



10

Disease progression and, where relevant, prevention measures



Deficiencies in engagement

- Youth
- Men
- Indigenous



**James
Lind
Alliance**

Priority Setting Partnerships

[JLA Adviser Zone login](#)



[▼ About us](#) [The PSPs](#) [Top 10s](#) [JLA Guidebook](#) [▼ Making a difference](#) [▼ The JLA Lab](#) [▼ News and publications](#)

Rare Diseases Priority Setting Partnership protocol

Published: 22 April 2026 | Version: 1 - April 2026 | 10 min read

1 MOST PEOPLE LIVING WITH A RARE DISEASE AND THEIR FAMILY MEMBERS REPORT POOR MENTAL HEALTH

Mental health was estimated based on four self-reported indicators: depressive symptoms with the Patient Health Questionnaire (PHQ-8), anxiety symptoms with the Generalised Anxiety Disorder questionnaire (GAD-2), loneliness with the UCLA questionnaire and a question on suicidal thoughts over the past 6 months.

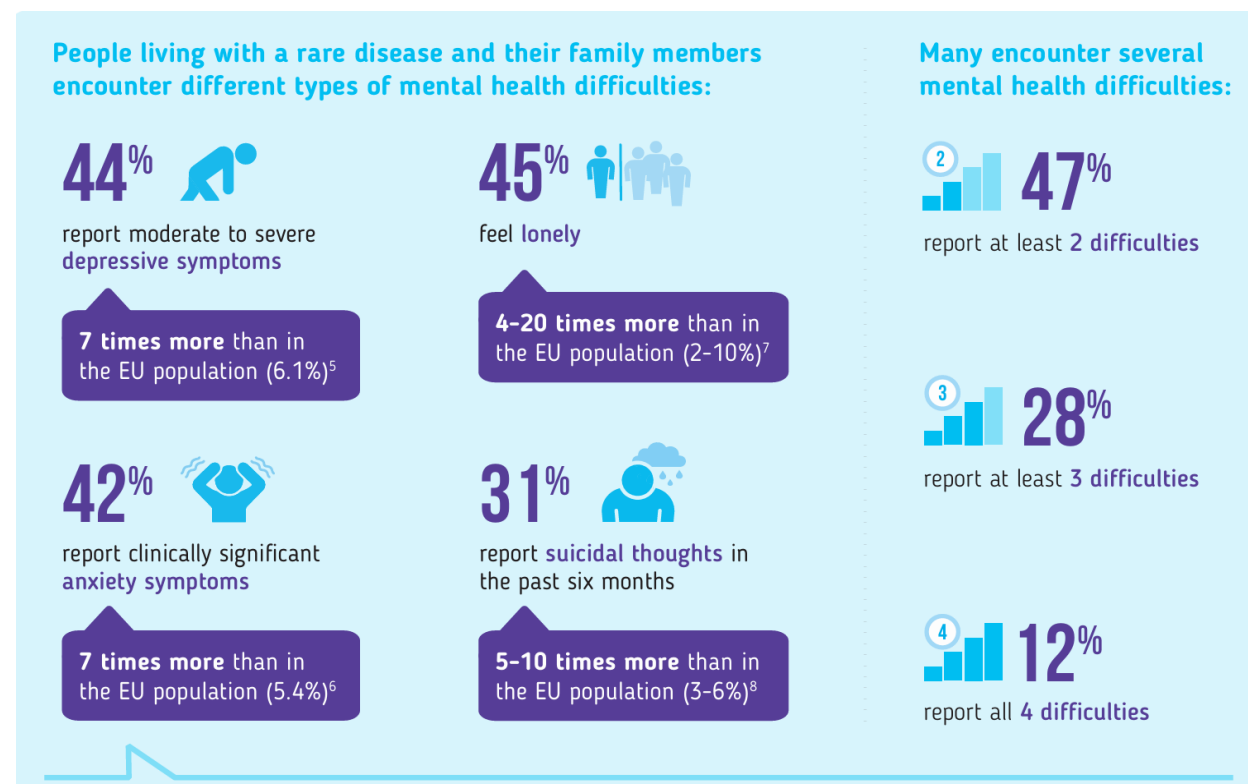


7/10

people living with rare diseases and their families report poor mental health

68% of participants reported at least one mental health difficulty among moderate to severe **depressive symptoms** (PHQ-8 score ≥ 10), clinically significant **anxiety symptoms** (GAD-2 score ≥ 3), **loneliness** (UCLA loneliness scale score ≥ 9) or **suicidal thoughts** in the past six months. All participants (n=9897).

Mental Health Burden is equivalent to extreme cohorts



Levels similar to those observed in exceptional contexts:

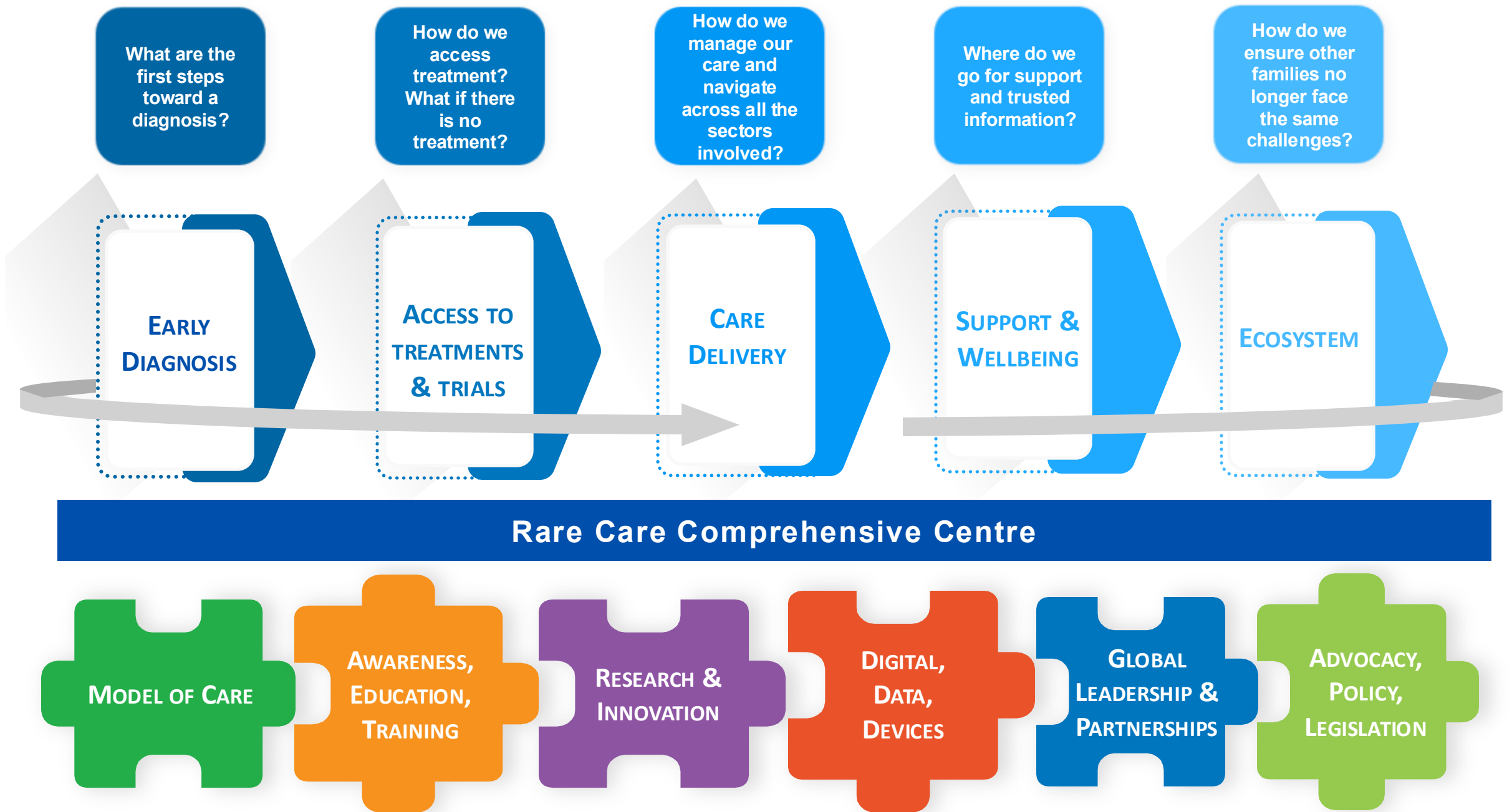
1. COVID-19 pandemic,
2. High-burden populations:
 1. Ugandans with HIV,
 2. psychiatric hospital settings,
 3. people with severe mental disorders.

[EURORDIS Rare Barometer reveals mental health crisis in rare disease community - EURORDIS-Rare Diseases Europe](#)

A starting point for a cross-sector RD Research Framework

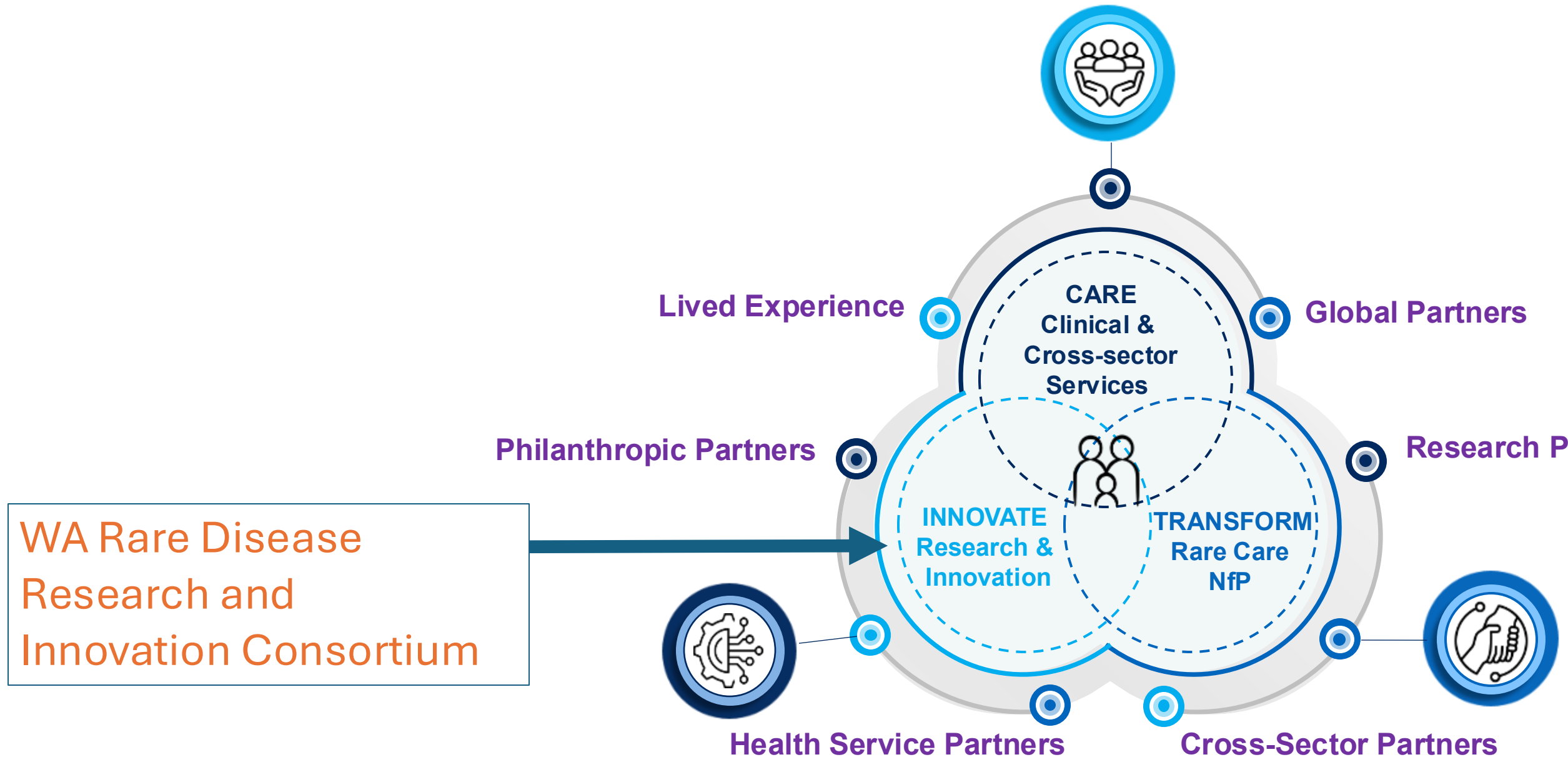
THEME AREAS	PRIORITY AREAS	United Nations CHALLENGES ADDRESSED
THEME 1: AWARENESS Advancing Awareness and Education Across Communities, Workforces, and Policy Systems	1.1 Community awareness and public education 1.2 Workforce capacity building and education 1.3 Fostering collaboration approaches across sectors 1.4 Advancing policy, regulation, legislation and advocacy 1.5 Promoting social inclusion and reducing stigma	• Diagnostic delay • Limited access to and retention of and return to employment • Psychological, social & economic vulnerability • Risk of inequality & discrimination
THEME 2: DIAGNOSIS Early Detection and Advanced Diagnosis	2.0 Rare disease coding and ontologies 2.1 Improved screening methods and programs 2.2 Advanced phenotyping and biomarkers 2.3 Access to diagnostics 2.4 Enhanced data sharing methods to improve diagnostic success 2.5 AI and ML to uncover novel diagnostic markers 2.6 Improving referral and diagnostic pathways	• Diagnostic delay • Insufficient screening • Lack of disaggregated data on people with RD
THEME 3: CARE Accelerate Development and Access to Patient Treatment, Care and Support	3.1 Culturally safe and responsive care 3.2 Innovative models of care and governance 3.3 Novel treatments and increased treatment access 3.4 Optimise access to clinical trials 3.5 AI for treatments, care and support 3.6 Enhance data sharing for improving treatment planning 3.7 Devices 3.8 Primary care 3.9 Quaternary, tertiary, second and primary prevention 3.10 Rare disease ontologies – functional annotation 3.11 Mental health and wellbeing 3.12 Metrics that matter	• Associated disability and impairment • Limited health care and barriers to access • Lack of disaggregated data on people with RD • Risk of inequality & discrimination

← Cross-sector approach →



RCCC

An integrated model



Research & Innovation Flagships

Delivered across all partner organisations through RCCC Research Consortium (CAHS, NfP, Harry Perkins, UWA, Murdoch University, Curtin University, Perron Institute, the Kids Research Institute)

Awareness – System & Community

- Increase public and policy awareness and understanding of rare diseases
- Reduce stigma, increase support and improve access to services through storytelling and public campaigns

Discovery-enabled Diagnostics

- Accelerate and improve genetic diagnosis for rare diseases
- Use advanced genomics and functional validation to find answers for families

Access to Treatments and Trials; Local Manufacturing

- Develop and deliver personalised therapies for rare diseases
- Build infrastructure for clinical trials and faster access to new treatments

Model of Care & Pathways

- Create integrated care pathways and coordination for families
- Enhance workforce training and support programs for better outcomes

AI to Unlock Rare Disease

- Harness AI to speed up diagnosis, treatment and care delivery
- Connect data and expertise for timely, equitable patient support
- Enabler that underpins the framework

What are the first steps toward a diagnosis?

How do we access treatment? What if there is no treatment?

How do we manage our care and all the agencies involved in care?

Where do we go for support and trusted information?

How do we ensure other families no longer face the same challenges?

UTOPIA · Gareth, Tudor, Tristan

genomic/phenotype synthesis, rare-pattern recognition

Treatment and trial matching, TrialR, in-silico models

cross-sector care plans

Dissemination

Multimodal AI · Tele

speech, imaging, etc... for earlier detection

personalised support for children with rare and neurodevelopmental conditions

Disabilit-EASE · Tristan, Sharon

Aurora · Timo

virtual research labs

AI-driven Drug Discovery · Tristan

Candidates, ASO
workstream – extensible to all advanced therapy types

functional-impact documentation, NDIS alignment

reduces family burden, trusted information

Cross-Sector Coding and Ontologies · Gareth, Tudor, Tristan

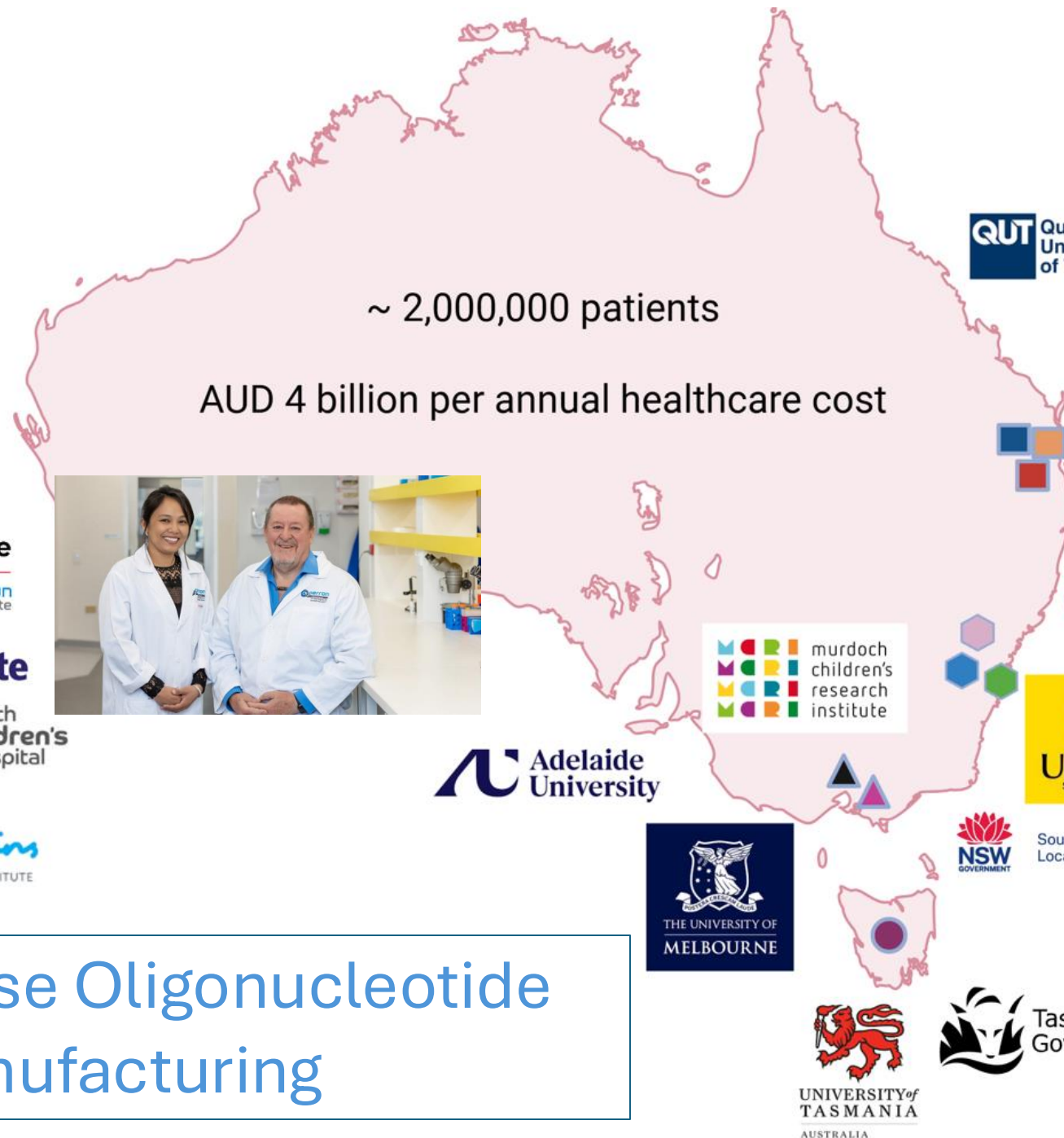
Kailume · Timo

explainable prediction

Synthetic Data Generation · Tristan, Tudor

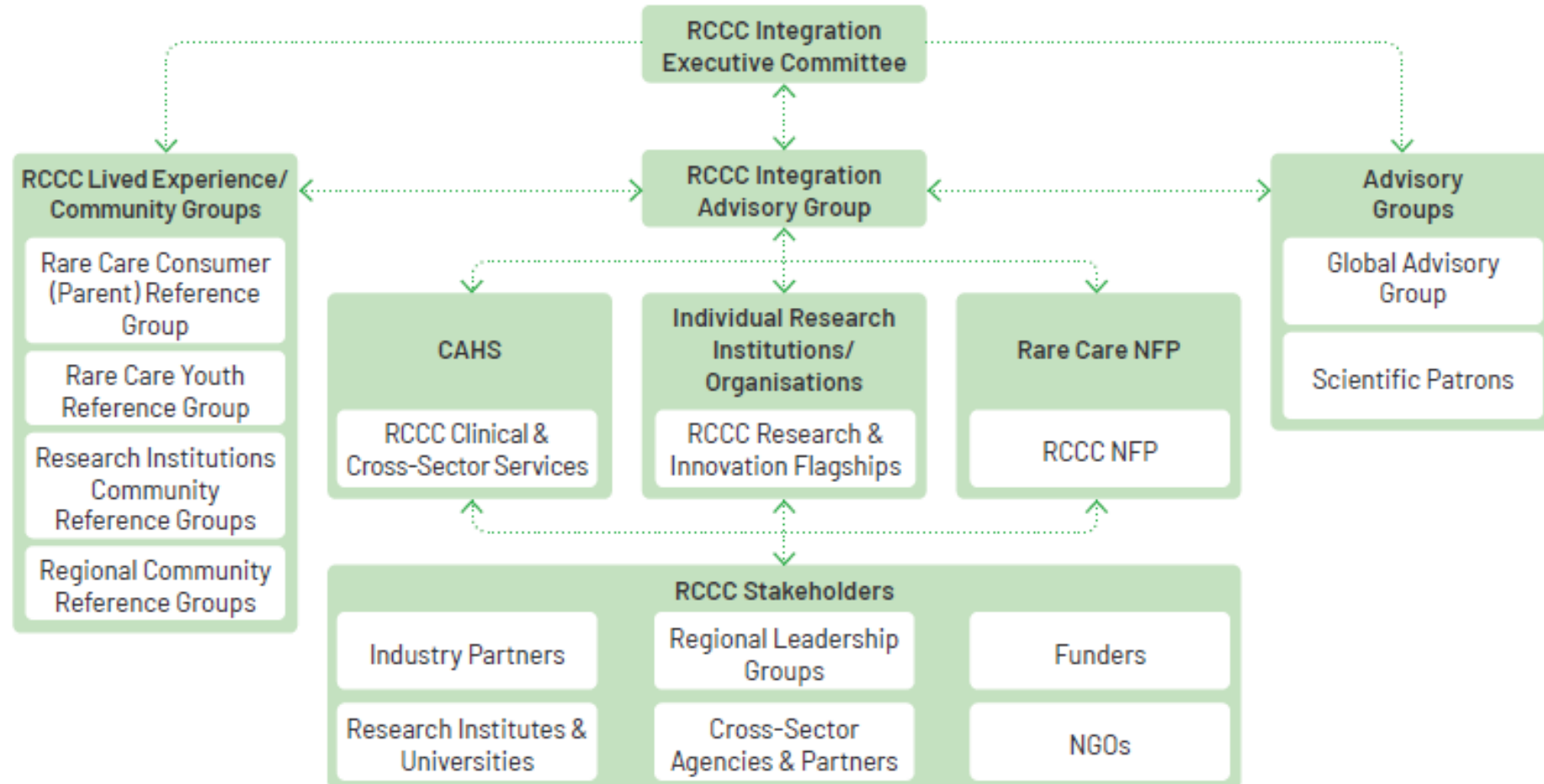
shared data environment, workforce training

coding spine, findable record



National Antisense Oligonucleotide Network and Manufacturing

The RCCC Governance Model

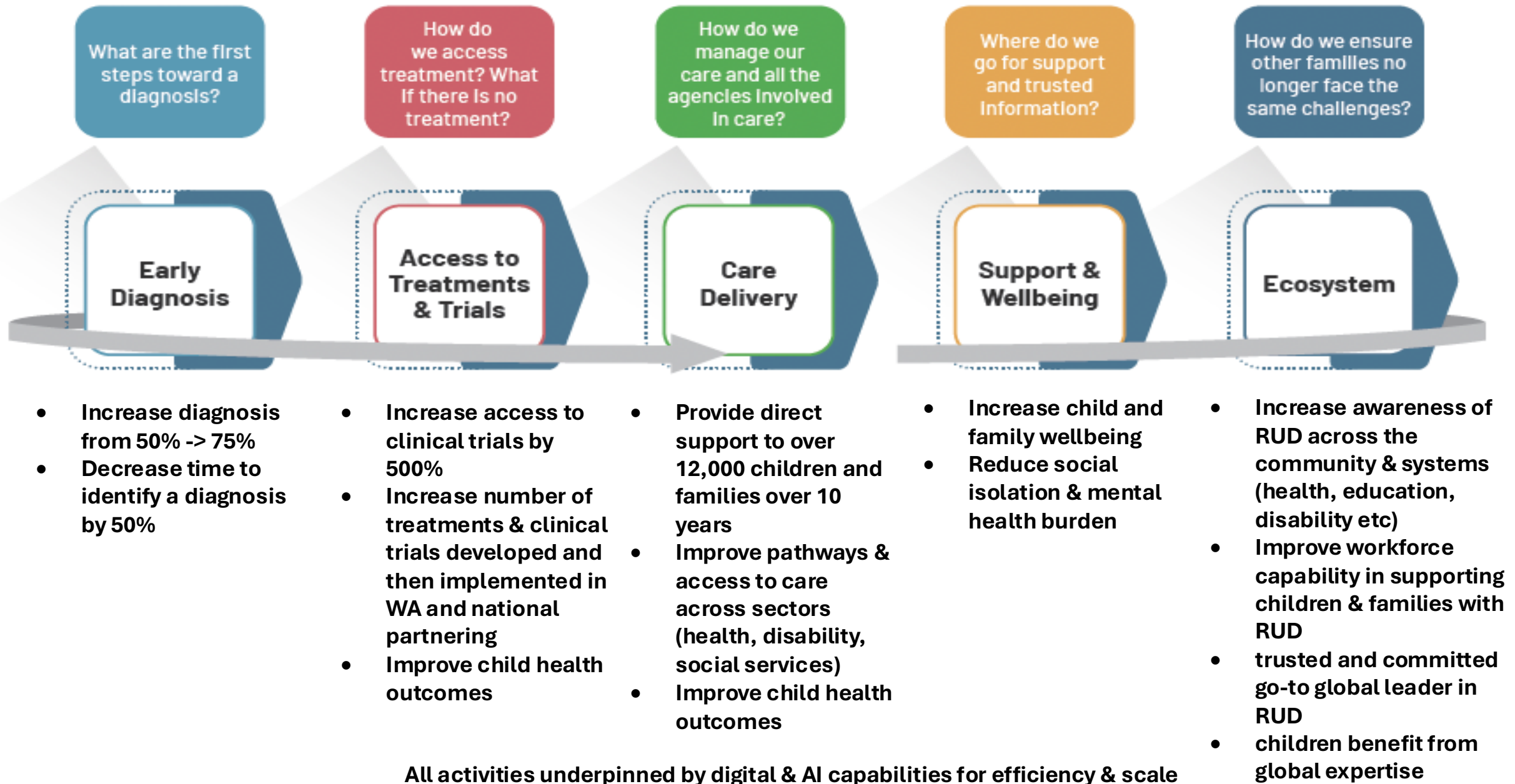


Current Status

- Network of partners: CAHS, UWA , RCCC NfP, Harry Perkins, Perron Institute, Curtin University, Murdoch University, The Kids Research Institute
- Governance structure established: interim-Steering Committee, Leadership Team, CAHS Advisory Committee, Comms & Branding Working Group
- Grant agreements in progress
- Risk analysis undertaken and planning, reporting and budget documentation significantly progressed
- RCCC branding currently in development, website development to commence soon
- Staggered commencement dates -aiming for a November start date in CAHS, Jan 2007 Universities



10-Year RCCC Impact Goals

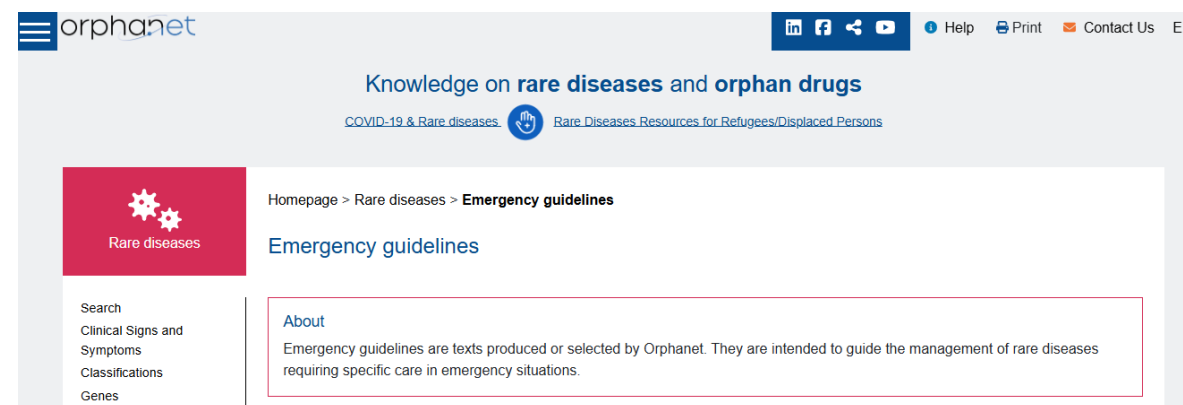


“My biggest fear is dying in
the emergency department”

Rare Care Centre, Youth Reference Group members

Emergency Department Burden of Rare Disease

- **Mortality in ED is 13 times** that of common disease
- **Bed block** due to longer and more complicated inpatient stays
- **Increased ED usage in remote regions**, WA data linkage study
- Lack of awareness of rare disease generally and e.g. of orphanet **emergency management guidelines**



[Orphanet : Diseases - Emergency guidelines](#)



Individually Rare, Collectively Common

A disease is defined as **rare** when it affects less than **1 in 2,000 people**

8,000

rare diseases
and increasing



300 million

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



80%

of rare diseases are
of genetic origin



70%

of rare diseases are
realised in childhood



5-7 years

the average time to
diagnosis



Only 5%

of rare diseases
have a prognosis
altering treatment



6 out of 10

deaths in children are
due to rare disease



The biggest cost in
health care

1.5 x

everything else
combined





Transform: the role of the NfP

Dedicated to social transformation to improve the lives of children and families affected by rare and undiagnosed diseases

NfP High level Functions

- Integration of the RCCC
- Global Networks and Partnerships
- Agility
- Safe Havens/ Chatham House Rules, Precompetitive Working, Public-Private Partnerships
- Cross-sector and System of Systems capacity and capability building