Office for Health and Medical Research

Rare Diseases

Translational Research Grants Scheme
Round 9

NSW GOVERNMENT

'Building strong relationships for Aboriginal health research and innovation in NSW'

Artist: Carissa Paglino

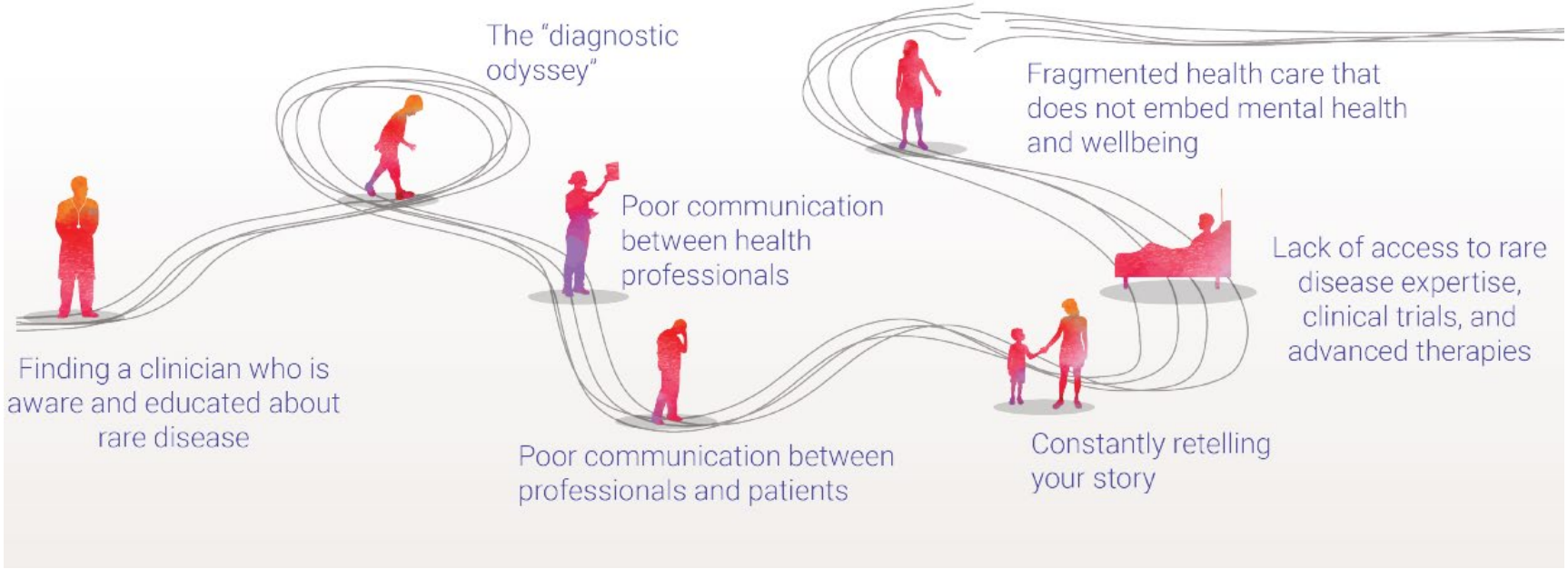
The Clinical Innovation and Research Division acknowledges the traditional custodians of the land that we work on.

We pay our respects to Elders past and present and extend that respect to other Aboriginal people present here today.

Rare Diseases

- 1 in 2,000 people
- **7,000 known rare diseases** and growing
- Individually rare, but common
- 8% of the population (around 2 million people)
- 80% have a genetic basis
- Also includes rare cancers, infectious diseases and autoimmune diseases
- Only around 5% have approved treatments
- 30% with genetic cause will die before their fifth birthday

Challenges within the health sector



National Strategic Action Plan 2020

PILLAR 1: AWARENESS AND EDUCATION

Priority 1.1: Increase every Australian's awareness of rare diseases including, where applicable, relevant prevention measures.

Priority 1.2: Ensure Australians living with a rare disease have access to information and education that enables them to be active participants in their rare disease journey.

Priority 1.3: Develop a national rare disease workforce strategy that responds to current and future demands, including the impact of genomics.

PILLAR 2: CARE AND SUPPORT

Priority 2.1: Provide rare disease care and support that is integrated and appropriate for all Australians living with a rare disease, while being both person and family-centred.

Priority 2.2: Ensure diagnosis of a rare disease is timely and accurate.

Priority 2.3: Facilitate increased reproductive confidence.

Priority 2.4: Enable all Australians to have equitable access to the best available health technology.

Priority 2.5: Integrate mental health, and social and emotional wellbeing, into rare disease care and support.

PILLAR 3: RESEARCH AND DATA

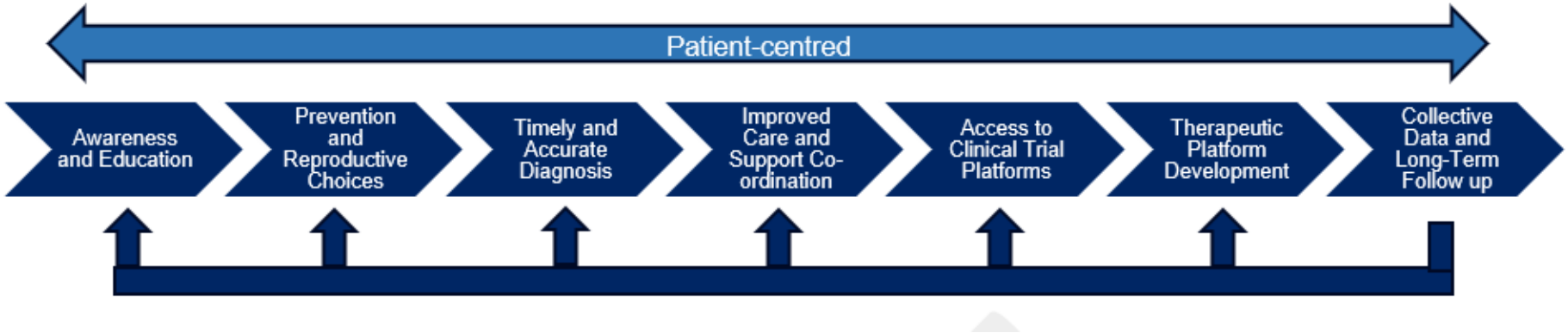
Priority 3.1: Enable coordinated and collaborative data collection to facilitate the monitoring and cumulative knowledge of rare diseases, informing care management, research and health system planning.

Priority 3.2: Develop a national research strategy for rare diseases to foster, support and drive all types of research for rare diseases, contributing to agreed priorities and systematically addressing gaps.

Priority 3.3: Ensure research into rare diseases is collaborative and person-centred.

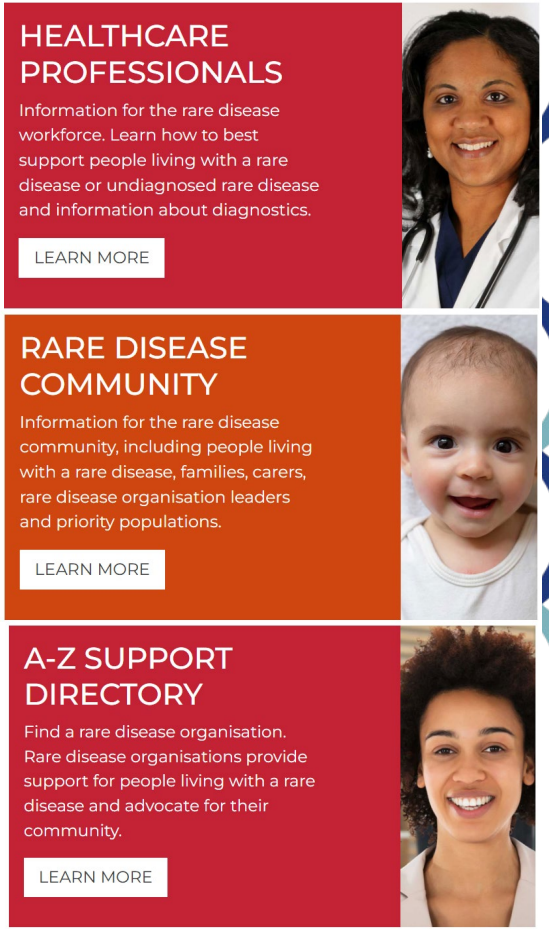
Priority 3.4: Translate research and innovation into clinical care; clinical care informs research and innovation.

The Rare Diseases Research Pipeline



Awareness and Education

- Federal investment in education resources
- Rare Awareness Rare Education (RARE) Portal
- Poor understanding by clinicians of rare diseases
- Delays diagnosis and missed opportunity for early intervention



Prevention and Reproductive Choices

- Carrier and antenatal testing and screening
- External causes for some non-genetic rare diseases, e.g. infectious diseases

Timely and Accurate Diagnosis

- Early diagnosis optimises clinical care, improves access to services and supports, can improve treatment options and access to clinical trials
- Around one third experience diagnostic delay of more than five years
- Half experience at least one misdiagnosis

Consider:

-Centres of Excellence in rare disease diagnosis and management

-Network-based care connecting clinicians with specialists, including telehealth

-Develop and implement care pathways and decision-aid tools

-Models of care supporting greater access to genetic counselling services

-Integrating advanced technologies into diagnostics

Improve Care and Support Coordination

- Can be fragmented and difficult to navigate for families
- Can impact every facet of life for those affected including education, employment, and physical and mental health
- Clear pathways to a range of services including multidisciplinary care models, peer and mental health support, disability services and financial help
- Digital tools could play a role
- Consider cultural sensitivities and vulnerable communities
- Consider support at significant life-stage transitions such as paediatric-to-adult, relocating to another area, and change in condition such as end-of-life

Access to Clinical Trial Platforms

- For around 95% a clinical trial may be their only option for accessing treatment
- Healthcare workers are gatekeepers to research and clinical trials for patients
- Standard of care should be supporting clinicians to help their patients access clinical trials where there are limited treatment options
- Initiatives could support patient recruitment through innovative trial design

Therapeutic Platform Development

- Limited commercial appeal for rare indications
- Complex and costly development

Consider platforms for:

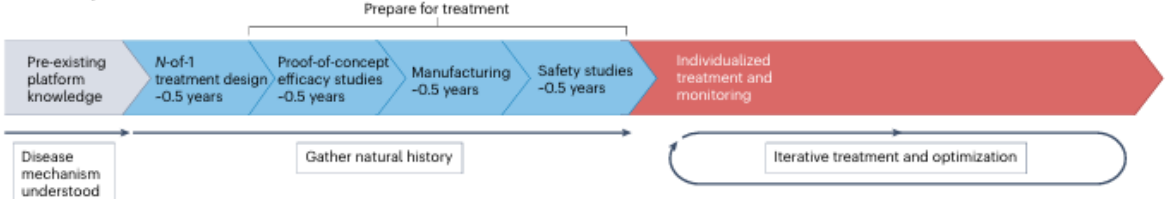
Drug repurposing

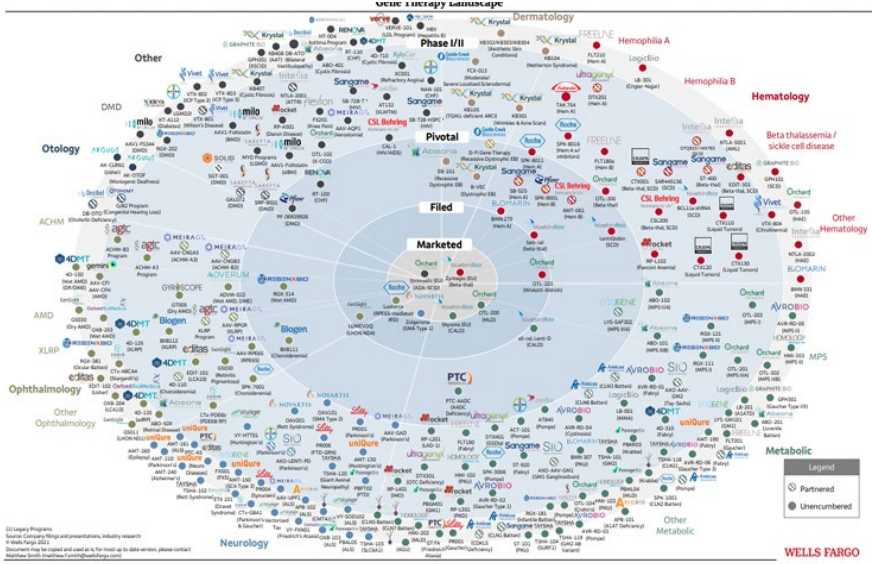
ASOs or ‘gene patches’

N=1 gene therapy

a Traditional drug approval process

b Patient-specific *N*-of-1 treatment





NEWS | 15 May 2025

World's first personalized CRISPR therapy given to baby with genetic disease

Treatment seems to have been effective, but it is not clear whether such bespoke therapies can be widely applied.

Collective Data and Long-Term Follow Up

- Supports ‘bedside back to bench’
- 2016 Australian study found 90% of people with rare diseases were interested in joining a patient registry
- Virtual registries using existing data sources and improved coding
- Biological registries can support a number of research groups to advance understanding of rare diseases
- Post-marketing surveillance

Contact us

medicalresearch.nsw.gov.au

NSW Medical Research

@MedResearchNSW

Contact us:
medicalresearch.nsw.gov.au/contact-us

Subscribe to our mailing list

Office for Health and Medical Research

NSW GOVERNMENT