

Healthier Kids. Healthier Future.

# Genomics and Healthcare

## An Australian perspective

**Kathryn North**

Director, Murdoch Childrens Research Institute

Director, Victorian Clinical Genetics Service

David Danks Professor, University of Melbourne

Vice Chair, Global Alliance for Genomics and Health



The Children's

Excellence in  
clinical care,  
research and  
education



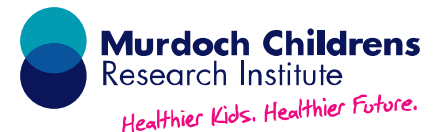
The Royal  
Children's  
Hospital  
Melbourne



Murdoch  
Childrens  
Research  
Institute



THE UNIVERSITY OF  
MELBOURNE



# Genomics and Healthcare

## An Australian perspective

- Melbourne Genomics Health Alliance
- Australian Genomics Health Alliance



# Five Year Vision

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Excel in the use of genomics to tailor health care

Clinically driven

Shared & collaborative

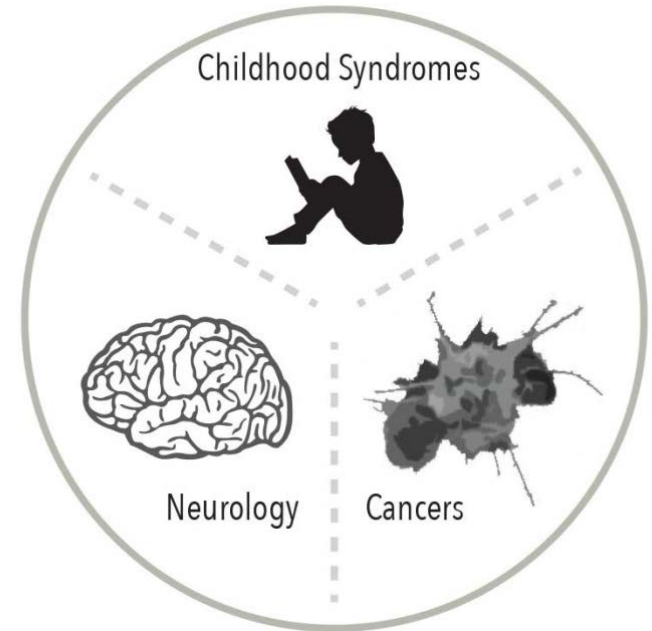
Clinical genomes available for research use

# Melbourne Genomics Health Alliance

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# Demonstration Project- 12 months



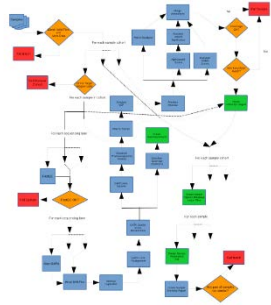
- Prospective recruitment
- Whole exome sequencing
- Targeted analysis
- In parallel with usual investigations

**n= 200 patients**

**WHOLE OF SYSTEM APPROACH**

CEO's	Gareth Goodier Kathryn North		Doug Hilton Lynne Cobiac		Stephen Smith Christine Kilpatrick	
Steering Group	Clara Gaff (program leader) James Angus (chair) Andrew Sinclair		Julian Clark Paul Waring Trevor Lockett		Mike South Ingrid Winship Sue Forrest	
NATA laboratories	Centre for Translation Pathology (Waring)		AGRF (Forrest)		VCGS (Amor/Sinclair)	
Flagship Groups	CMT (Ryan)	CS (White)	EPIL (Kwan)	CRC (Bousioutas)	AML (Roberts)	
Advisory Groups	Genomics and Bioinformatics (Taylor)	Clinical Interpretation & Reporting (James)		Community (Winship)	Information Management (Hansen)	
Technical Expertise	Pipeline platform		Variant database		Clin-Bioinf workforce	
	Computing		Database users		Sequencing	
Workshop Groups	Research Access		Ethics/Consent		Patient data	
	IM/IT		Participant survey		Evaluation	

# Systems operational



Common  
Bioinformatics  
Pipeline



Variant Curation  
Database



Multidisciplinary  
review (16)

**Bioinformatics and Genomics  
Advisory group**



Shared ethics and consent  
Agreed (draft) standards,  
guidelines, templates

**Clinical Interpretation  
and Reporting advisory  
group**

# Demonstration Project

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Enrollment complete n=220

Evaluation:

1. **Process:** barriers, issues, requirements
2. **Performance:** diagnosis, cost, time
3. **Threshold analysis:** conditions for financial viability
4. **Health economic analysis**

State Government has now  
invested \$25M for next four years  
– expand across whole of state





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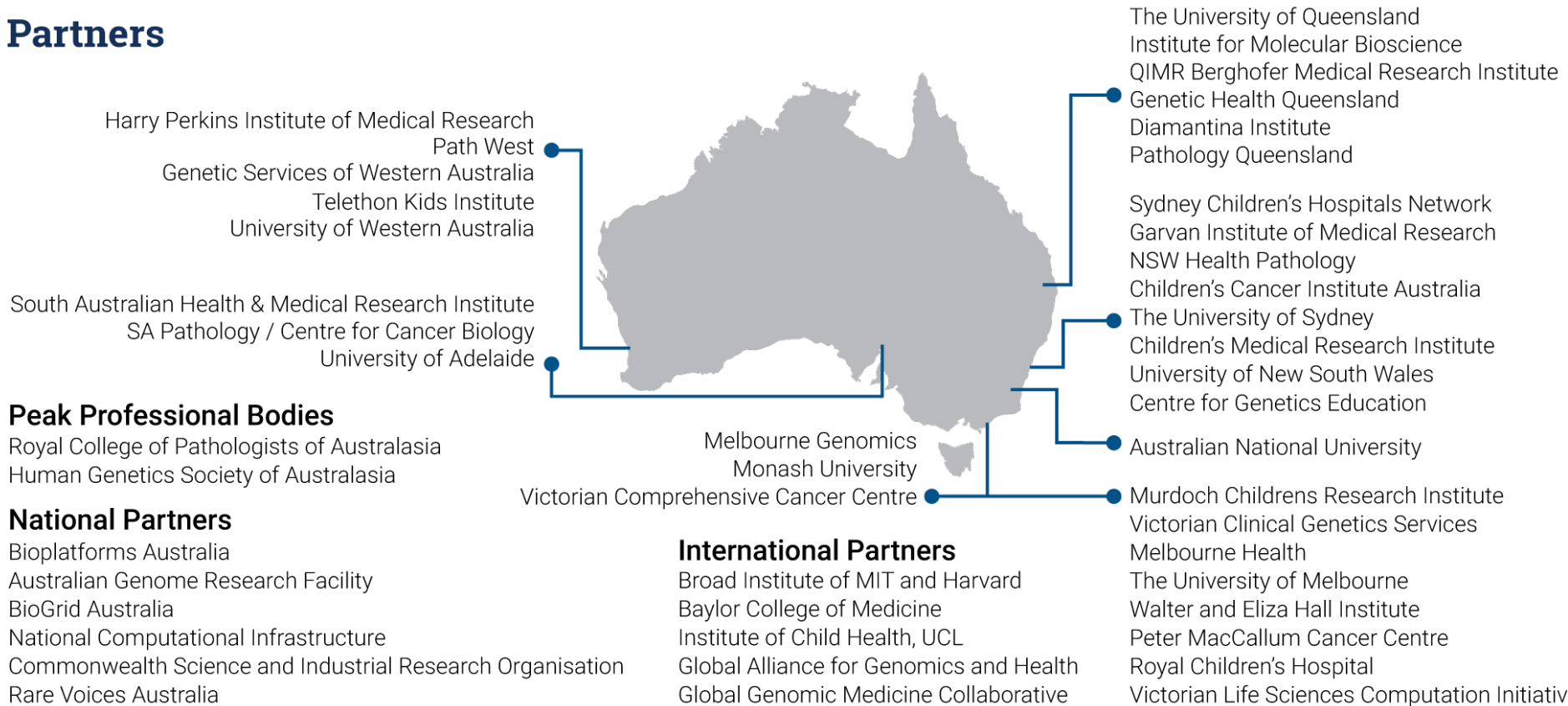


# + Australian Genomics Health Alliance

## Vision

A national collaborative diagnostic and research network

## Partners



## Peak Professional Bodies

Royal College of Pathologists of Australasia  
Human Genetics Society of Australasia

## National Partners

Bioplatforms Australia  
Australian Genome Research Facility  
BioGrid Australia  
National Computational Infrastructure  
Commonwealth Science and Industrial Research Organisation  
Rare Voices Australia

## Key mapping exercises



the workforce



genomic testing  
practices



functional genomics  
capabilities



centres of disease  
focus in genomics



technical infrastructure  
and bioinformatics  
capabilities

## Program 1

A national diagnostic and translational research network

*Developing the most appropriate diagnostic approach for each specific disease area.*

## Program 2

A national data repository: scalable, shared and standardised

*Genomic and clinical data linkage.*

## Program 3

Economic analysis and policy implications for the health system

*Health economics, policy development, communication and engagement.*

## Program 4

Genomic workforce, education and ethics

*Identify training and education requirements for health professionals and patients, theoretical ethical analysis of genomics.*

Existing Activities

Disease Area I

Rare Disease

Clinically driven

Disease Area II

Cancer

Patient focussed

Enabling research

Clinical Outcomes



Prevention



Early Diagnosis



Early Intervention



Surveillance



Targeted Intervention or Therapy

## Analysis

To provide a strong ethically informed evidence base for applying genomics to clinical

## Policy

Practical strategies to inform Australian Health system planners and policy makers

## + Where can Global Alliance Help?

An **international** data repository: scalable, shared and standardised

- **Clinical data:** Standardisation of ontologies for phenotypic clinical data
- **Genomic data:** Agreed standards and guidelines across diagnostic laboratories for variant curation and classification, and the reporting of patient results.
- Development of national **standards for patient reports**
- Development of a **data sharing** systems that allows for clinical utility throughout a patient's life
- **Registries:** National and international linkage
- Development of standards and protocols for **governance and sharing of genomic data** – national and international.
- Establishment of systems to make the **genomic data available to researchers**
- Development of robust systems and policy to ensure **data security, integrity and quality.**



*"We finished the genomic map, now we can't figure out how to fold it."*