Genomics and the brave new world of personalised medicine:

A local and global perspective.

Kathryn North
Director MCRI
Lead Investigator
Australian Genomics
The human genome is large

A human genome is 3 billion bases (A G C T)

Each copy of Tolstoy’s War & Peace has 3 million letters

So 1,000 copies has 3 billion letters

= As high as an 12 storey building
Rate Limiting factors

Cost of sequencing

Interpreting the genome
Genomic medicine in healthcare
WHAT COULD IT MEAN FOR THE PUBLIC?

- Faster diagnosis
- Improved prognosis
- Targeted therapy “Precision Medicine”
- Better manage disease risk
- Prevention
Australian Genomics
Building on local and global experience

- The Global Alliance for Genomics and Health
- Establishing Genomic Medicine in Australia
- Australian Genomics Health Alliance: NHMRC TCR
The challenge

• Data from **millions of samples** will be needed to address questions in rare disease, complex disease and cancer

• **Right now:**
  • Data is typically in **silos**: by type, by disease, by country, by institution
  • Analysis methods are **non-standardized**, few at scale
  • Approaches to regulation, consent and data sharing limit interoperability
Mission – GA4GH

• To accelerate progress in human health by helping to establish a common framework of harmonized approaches to enable effective and responsible sharing of genomic and clinical data, and by catalyzing data sharing projects that drive and demonstrate the value of data sharing.
Partner overview

500+
- Number of partner organizations
- Includes a wide variety of groups: Research Institutes, Academic Medical Centers, Universities, Disease Advocacy Organizations and Patient Groups, Funders, Life Science and Information Technology Companies, and more

46
- Number of countries in which alliance partners are based:
  - Argentina, Australia, Austria, Belgium, Brazil, Canada, China, Finland, France, Germany, Hungary, India, Ireland, Japan, Mexico, Netherlands, New Zealand, Singapore, Spain, South Africa, Sri Lanka, Sweden, Switzerland, United Kingdom, United States.

6
- Number of continents in which alliance partners are active:
  - Active throughout the globe, with a presence in 6 continents: Africa, Asia, Australia, Europe, and North and South America.
A federated platform (Exchange) to facilitate the matching of cases with similar phenotypic and genotypic profiles (Matchmaking) through application programming interfaces (APIs).

**GA4GH approach**

Don’t reinvent the wheel

Be practical – systems must work now, build a path from existing systems

Embrace federation and “data islands with analysis clouds” approach
SUMMARY:

- In 2022, genomic data on tens of millions of individuals are responsibly accessible via GA4GH standards.
  
  - Vast majority of this data has been generated due to healthcare approaches rather than research commissioned genomes.
  
  - Both research-commission genomes and secondary use of healthcare genomes for research is accessible due to the consistent application of the GA4GH APIs, SOPs and tools.
  
  - Genomics data that can be shared responsibly, are shared responsibly, meaning every qualified clinician, researcher, and corporate entity around the globe, shares and has access to, the maximal dataset that is privacy preserving within the context of the relevant and localised consent and authorization policies.
  
  - Genomic and phenotypic are integrated in clinical records and form a “healthcare learning system.”
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The Australian Health Care System

Complex and Fragmented Structure

Share of expenditure
- Hospitals
- Primary health care
- Other recurrent

Responsibility for services
- Combined private sector and public sector—all levels of government
- State and territory governments
- Private providers

Funding
- Australian Government funding share
- State/territory government funding share
- Private funding share

Health service funding and responsibilities
Australia’s Health 2014, AIHW
WHOLE OF SYSTEM CHANGE IS NEEDED
A national approach to genomics in Australia

**CHALLENGES**

- **Divide** between the **state and federal health** systems.
- Risk of duplication between states & waste of resources.
- No common **ethics, consent** or genetic test form.
- **Lack of common data storage** & difficulty in sharing data.
- **Workforce**: needs genomic literate clinicians and bioinformaticians.
- **Inequity of access** for patients.
- Inactivity, **fragmentation**, internal competition.

**OPPORTUNITIES**

- **NATA accredited sequencing facilities in all states** – need to increase sequencing capacity.
- **National healthcare system** capable of providing equity of access.
- **Quality and capability** of clinical and research networks.
- High degree of **genomic activity in Australian research**.
- Perfect time for **collaboration and linkage** nationally and internationally.
NHMRC Genomics TCR

- Demonstrate how patient benefit could be maximized through application of genomic data in one or more human diseases.
- Provide evidence to inform analysis on the cost effectiveness of implementing genomic data into the Australian health system.
- A significant increase in the understanding of practical strategies that could be used by Australian health system planners and policymakers.
- Building Australia’s research and research translation capacity in the area of genomics and healthcare.
Queensland Genomics Health Alliance commenced 2014
Sydney Genomics Collaborative commenced 2014
Melbourne Genomics Health Alliance formed 2014
Australian Genomics formed 2016
Melbourne Genomics Health Alliance gov + partner funding launched 2015
Queensland Genomics Health Alliance commenced 2016
Australian Genomics commenced 2016
Canberra Clinical Genomics launched 2016
Zero Childhood Cancer launched trials 2017
SA Genomics Health Alliance formed 2017

Mapping + piloting approaches
Sequencing, diagnostics + data infrastructure
Shared models, common leads
Building evidence + infrastructure
Global reach
Standardising practices + policy
Addressing ethical, legal & social implications
Genomics initiatives Australia
LINKED & COLLABORATIVE
Melbourne Genomics Health Alliance
Demonstration Project 2014 – 2015

- Establishing **state-wide platform** for genomic information.
- Developed **prototype** system
  - multiple organisations & different conditions.
- **Evaluate** prototype compared to standard care
  - what worked, what didn’t, potential solutions,
  - detection rate, change in management, cost effectiveness
- **Shared** approaches.

**Approach:**

- Prospective recruitment (n=315 patients).
- Five flagships – childhood syndromes, inherited neuropathy, focal epilepsy, colorectal cancer, acute myeloid leukemia
- Whole exome sequencing.
- Targeted analysis.
- In parallel with usual investigations.
Shared approaches

Common clinical consent form (germline)

Common informatics pipeline

Data standards

Curation guidelines

Common report format

Multidisciplinary review meetings
Demonstration Project key findings (2014-2015)

Patients accept genomic sequencing
More than 90% of patients consented
Following genetic counselling, 96% of patients felt sufficiently informed to consent

Patients agreed to share genomic data
98% agreed to share data for research related to their condition
93% agreed to share data for any research

Re-analysis leads to new diagnoses
12% of patients received a diagnosis from re-examination of their genomic data (this is expected to increase)

### Background
- Childhood syndromes patients (101 children).
- Seen at The Royal Children's Hospital Melbourne.
- Patients received both:
  A. Traditional diagnostic tests.
  B. WES as part of early diagnostic workup

### Numbers

<table>
<thead>
<tr>
<th>Experimental finding</th>
<th>Traditional Diagnostics (e.g. MRIs &amp; muscle biopsies, single gene test)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Rate of diagnosis</td>
<td>11%</td>
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<tr>
<td>Average cost per patient</td>
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### Experimental finding
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<th>Precision Medicine Diagnostics (Genomic Test)</th>
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When genomic testing was used:
- **Five times** more patients were diagnosed.
- The **cost** per patient was **reduced by 75 per cent**.
- Four times more patients had improved care

NB. No patients required novel expensive treatments.

**Genomic medicine produces net benefits for patients and the health system:**
- Diagnosis is faster & more accurate, with one test replacing many unsuccessful ones.
- Treatment options: no change to treatment (cost savings on unnecessary investigations), re-purposing existing medications (cost savings), or halting ineffective or harmful healthcare (cost savings) while tailored treatments are only offered to those patients who will benefit.
- Prevention: identification of disease risk permits pre-emptive treatments and disease prevention.
State-wide approach to genomic data management

1. Standardised policy and processes for data management & access (data governance)
2. Standardised policy & processes for patient consent
3. Standardised policy and processes for test ordering & reporting
4. Change control process
5. Staff to manage the data
6. Staff to manage the technology

Technology:
7. Identity & Access Management
8. Clinical Tools
   - Clinician Knowledge
   - Electronic Orders and Results
   - Clinical Decision Support Tools
9. Diagnostic Tools
   - Analysis (Pipeline) Tools
   - Curation Tools
10. Patient Tools
    - Consent
    - Results
    - Education
11. Data Access Tools
12. Master Patient Index
13. Genomic Data Repository
14. Data Integration
   - EMR (clinical data)
   - LIMS (genomic sequencing data)
   - Public variant curation data

Melbourne Genomics Health Alliance
Sydney Genomics Collaborative (SGC)
$24M GENOMIC PROGRAM 2014-2018 (NSW Health)

**Medical Genome Reference Bank.**
$10M program to build a public resource containing 4,000 whole genomes (30X) from healthy aged individuals with detailed clinical histories.

**NSW Genomics Collaborative Grants.**
$7M program to enabling NSW researchers to use WGS to identify genetic basis of disease and build capability for genomics implementation in clinical practice.

**Genomic Cancer Medicine Program.**
1. Genetic RISC in the Young Study (RiSC).
2. Molecular Screening and Therapeutics (MoST).
Australian Genomics
Building on local and global experience

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Australian Genomics has implemented a program of research that:

- **Demonstrates** how the application of genomic data impacts the care of patients across a range of disorders.

- **Identifies the practical, economic and policy implications of genomic testing** across the health system.

- **Pilots the infrastructure to support the sustainable implementation** of genomic medicine for the healthcare of Australians.

**Our clinical Flagship** projects are central to our research model: **nationally coordinated**, founded **in clinical practice**, driving data through our research programs to enable real time evaluation of the impact of genomics in patient care, coupled with united clinical and research efforts.
Australian Genomics

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
CSIRO
Rare Voices Australia
Rare Cancers Australia
Australian Mitochondrial Disease Foundation

International Partners
Broad Institute of MIT and Harvard
Baylor College of Medicine
UCL Great Ormond St Institute of Child Health
Global Alliance for Genomics and Health
Global Genomic Medicine Collaborative
Genomics England

The University of Queensland
Lady Cilento Children’s Hospital
Institute for Molecular Bioscience
QIMR Berghofer Medical Research Institute
Wesley Hospital
Royal Brisbane and Women’s Hospital
Princess Alexandra Hospital
Diamantina Institute
Pathology Queensland
Queensland University of Technology
Queensland Genomics Health Alliance
Sydney Children’s Hospitals Network
Royal North Shore Hospital
Garvan Institute of Medical Research & KCCG
Kinghorn Cancer Centre
NSW Health Pathology
Children’s Cancer Institute Australia
The University of Sydney
Children’s Medical Research Institute
University of New South Wales
Centre for Genetics Education
AIHL / Macquarie University
The Australian National University
Murdoch Children’s Research Institute
Melbourne Bioinformatics
Victorian Clinical Genetics Services
Melbourne Health / Royal Melbourne Hospital
The University of Melbourne
Walter and Eliza Hall Institute of Medical Research
Peter MacCallum Cancer Centre
Royal Children’s Hospital
Austin Health
Australian Genomics leadership

Intersection with State & International Consortia

Kathryn North, MCRI, VIC (GA4GH)
Andrew Sinclair, MCRI, VIC (MGHA, G2MC)
John Christodoulou, UoM & MCRI, VIC (MGHA)
David Thomas, Garvan, NSW (Sydney Genomics & ZCC)
Marcel Dinger, Garvan, NSW (Sydney Genomics, GA4GH)
Stephen Fox, PMCC, VIC (MGHA, ZCC)
Clara Gaff, VIC (Melbourne Genomics, GA4GH)
Nigel Laing, Harry Perkins, WA (IRDIGC)
Jozef Gecz, University of Adelaide, SA (SA Genomics)
David Hansen, CSIRO, QLD (MGHA, QGHA)
Hamish Scott, SAPath, SA (SA Genomics)
Robyn Ward, UQ, QLD (G2MC)
Deborah Schofield, USyd, MCRI (Sydney Genomics, ZCC)
Sean Grimmond, UoM, VIC (GA4GH, ICGC Med)
Natalie Thorne, VIC (Melbourne Genomics)
Deborah White, SAHMRI, SA (SA Genomics)
Our Model

Program 1: National diagnostic & research network
- Developing the best diagnostic approach for each disease area

Program 2: National approach to data federation & analysis
- Linking genome and clinical data

Program 3: Economics & health policy
- Health economics, policy development, implementation science & communication

Program 4: Genomics workforce, education & ethics
- Mapping education & training needs, addressing ethical implications of genomic medicine

Rare Disease Flagship
- Existing Activities

Cancer Flagship
- Clinically driven
- Patient focused
- 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 practice

Policy
- Practical strategies to inform Australian health system planners and policy makers
This granular representation demonstrates the many sub-projects supporting our research model, each of which focuses on specific facets of our shared goal of sustainable clinical genomic implementation for the healthcare of Australians.
Program One
A NATIONAL DIAGNOSTIC AND RESEARCH NETWORK

Purpose – the delivery of a coordinated and sustainable system for the provision of genomic testing in the clinical environment – technology agnostic.

Primary activities – supporting the delivery of the Flagship projects through a national coordination network, and developing recommendations and building evidence to inform policy change through four sub-projects -
  • Clinical Variant Re-Classification
  • National Clinical Consent
  • MSAC Application Pipeline
  • Functional Genomics
Program One
MSAC APPLICATION PIPELINE

**Purpose** – This project aims to build our capabilities and experience in successfully channeling submissions through the Medical Services Advisory Committee (MSAC) with the aim of supporting a pipeline of applications for consideration around appropriate state / federal funding of genomic testing in specific clinical circumstances.

**Primary activities** – The first application submitted by the MSAC working group relates to **genetic testing for childhood syndromes, based on MGHA Flagship data.**

- Application #1476 seeks **funding for whole exome analysis** of genomic sequencing data on children with the onset of clinical features/symptoms indicating a syndromic disorder in the first year of life.
- This application is well underway (see below), and expected to be assessed by MSAC in early 2018.
Creating a network to connect researchers who can investigate functional genomics of newly discovered genes and variants.

**Australian Functional Genomics** is being led by a group of researchers and clinicians from across Australia with the aim of integrating functional genomics into the diagnostic paradigm for managing rare diseases and cancer in Australian patients.

Register to join the network!

functionalgenomics.org.au
Program Two
A NATIONAL APPROACH TO DATA FEDERATION AND ANALYSIS

Purpose – Program 2 is focused on the development of standards and practices to exchange, share and unify genomic data from our Flagship projects: at both a national level and harmonised with international efforts. The group is developing a flexible, scalable national clinical genomics data infrastructure to unify data across our health systems to allow higher-order use of this information for research and improved healthcare outcomes.

Primary activities: five sub-projects

1/ Clinical Variant Classification & Sharing
2/ Genotype-Phenotype Data Capture & Analysis
3/ Phenotype Ontologies and eHealth
4/ Variant Pipeline Evaluation & Quality Assurance
5/ Data Governance, Aggregation & Sharing
This figure depicts the functional information flow through Program 2 tools.
Program Three
POLICY, HEALTH ECONOMICS AND IMPLEMENTATION SCIENCE

Purpose – To successfully integrate genomic medicine into the Australian health system we must understand the economic. We are analysing and modelling the health and social impact of genomic medicine to inform policy development, and – in alignment with the National Health Genomics Policy Framework, seek to build evidence to support the efficient, effective, ethical and equitable implementation of genomics into the Australian health system.

Primary activities: three domains
• Health Economics
• Policy Development
• Implementation Science
  • Health Implementation Research
  • Network Analysis: Understanding Complexity
<table>
<thead>
<tr>
<th>Australian Genomics Health Alliance</th>
<th>Addressed by Framework Priority Areas</th>
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</thead>
<tbody>
<tr>
<td><strong>Governance and Leadership Structures</strong></td>
<td></td>
<td><strong>Program One – A National Diagnostic and Research Network</strong></td>
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</tr>
<tr>
<td>AGHA National Steering Committee</td>
<td>✓</td>
<td>Mapping functional genomics capacity for variant assessment</td>
<td>✓</td>
</tr>
<tr>
<td>National Implementation Committee</td>
<td>✓</td>
<td>Establishment of clinical informatics, shared clinical informatics operational</td>
<td>✓</td>
</tr>
<tr>
<td>Community Advisory Group</td>
<td>✓</td>
<td>Consent working group: shared single clinical and research consent process/national guidelines for consent</td>
<td>✓</td>
</tr>
<tr>
<td>Independent Advisory Group</td>
<td>✓</td>
<td>Improvement cycles for standards and protocols in place</td>
<td>✓</td>
</tr>
<tr>
<td>Industry Advisory Group</td>
<td>✓</td>
<td>MSAC Working Group</td>
<td>✓</td>
</tr>
<tr>
<td>Network of National and International Partners</td>
<td>✓</td>
<td>Variant Re-Classification Working Group</td>
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</table>

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</thead>
<tbody>
<tr>
<td><strong>Program Two – A National Data Repository: Scalable, Shared and Standardised</strong></td>
<td></td>
<td><strong>Program Three – Economic Analysis &amp; Policy Implications for the Health System</strong></td>
<td></td>
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<tr>
<td>Mapping national health and genome informatic infrastructure</td>
<td>✓</td>
<td>Health Economics - Preparing data collection instruments/collecting and analysing prospective data collection/Data cleaning and quality control measures</td>
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<tr>
<td>Establish protocols approach to sharing genotype-phenotype relationships and storing and sharing clinical genomic data</td>
<td>✓</td>
<td>Health Economics - Develop cost effectiveness models</td>
<td>✓</td>
</tr>
<tr>
<td>Assess feasibility of national data standards</td>
<td>✓</td>
<td>Health Economics - Develop cost effective models of NIPT and carrier screening</td>
<td>✓</td>
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<tr>
<td>Implement pilot sharing/storage program(s)</td>
<td>✓</td>
<td>Implementation Science - supporting the retention of genomic practices once introduced into healthcare</td>
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</tr>
<tr>
<td>Develop shared analysis framework for diagnostic bioinformatics</td>
<td>✓</td>
<td>Policy - Implementation Framework for MSAC/AMIC, State and Federal Governments/ Submission of MSAC proposal for genomic testing</td>
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<tr>
<td>Develop process for continuous optimisation of diagnostic bioinformatic pipelines</td>
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<tr>
<td>Bioinformatics pipeline benchmarking</td>
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<tr>
<td>Assess effectiveness and limitations of data sharing platform</td>
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<tr>
<td>Assess requirements for national implementation</td>
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<tr>
<td>Assess then pilot international data sharing program</td>
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<tr>
<td>Digital Health Sub-Committee: scope scaled integrated genome/EHR for national implementation</td>
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<tr>
<td>Pilot interoperability of gen-phen data and EMRs</td>
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<tr>
<td><strong>Program Four – Genomic Workforce, Education and Ethics</strong></td>
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<tr>
<td>Gap and Opportunity Mapping</td>
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<tr>
<td>Needs Assessment</td>
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<tr>
<td>A framework for evaluation of genomic and genetic education activities</td>
<td>✓</td>
</tr>
<tr>
<td>Patient participation survey</td>
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Program Four
WORKFORCE, EDUCATION & ETHICS

Purpose – The successful implementation of genomics in healthcare will depend upon the availability of a workforce able to deliver genomic medicine and its acceptance on both clinical and ethical grounds by health professionals, patients and their families. Australia needs an integrated approach to workforce development in genomic medicine to minimise the risk of duplication of effort, leverage existing resources and support good clinical and ethical practice.

Primary activities: two domains
• Workforce, Education
• Ethics
MY RESEARCH PORTAL
Dynamic consent

- survey delivery linked to REDCap
- access to data and results
- link to targeted information materials & patient groups
- My genomic results
- New research
- Education and engagement

communicate study outcomes, participant's contributions to findings & new research studies

"Dynamic" consent
Our Clinical Projects

‘FLAGSHIPS’
Primary activities – Each of the Flagship projects differs slightly in design, technology application and maturity of genomic delivery in current clinical practice, but have common elements:

- Mapping referral practices, methodologies and funding for standard of care for the condition in each state
- Prospectively recruiting participants
- Applying genomic testing to demonstrate the relative diagnostic efficacy
- Capturing health costs
- Identifying impacts of genomic testing (altered management or treatment, altered diagnostic interventions or family management, improvement to patient choices)
- Undertaking participant surveys and evaluations
<table>
<thead>
<tr>
<th>Flagship</th>
<th>Duration</th>
<th>Sites</th>
<th>Number</th>
<th>Methodology</th>
<th>Lead(s)</th>
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<tbody>
<tr>
<td>RARE DISEASE</td>
<td></td>
<td></td>
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<tr>
<td>Neuromuscular Disorders</td>
<td>2016 - 2018</td>
<td>NSW, QLD, SA, VIC, WA</td>
<td>520</td>
<td>Custom Capture Panel / WES</td>
<td>Nigel Laing</td>
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<tr>
<td>Mitochondrial Disorders</td>
<td>2016 - 2018</td>
<td>NSW, QLD, SA, VIC, WA</td>
<td>210</td>
<td>Half: WES + mtDNA Half: WGS</td>
<td>John Christodoulou David Thorburn</td>
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<td>Neurodevelopmental Disorders</td>
<td>2016 - 2018</td>
<td>NSW, QLD, SA, VIC, WA</td>
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<td></td>
<td>Jozef Gecz</td>
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<tr>
<td>Epileptic Encephalopathy</td>
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<td>NSW, QLD, SA, TAS, VIC, WA</td>
<td>100</td>
<td>WES</td>
<td>Ingrid Scheffer</td>
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<td>Brain Malformations &amp; Leukodystrophies</td>
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<td>NSW, QLD, SA, VIC, WA, NT</td>
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<td>WES</td>
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<td>KidGen Renal Genetics</td>
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<td>Genetic Immunology</td>
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<td>CANCER</td>
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<td>2016 - 2020</td>
<td>NSW, NT, QLD, SA, VIC, WA, WLD</td>
<td>~300</td>
<td>RNA Seq</td>
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<td>David Thomas</td>
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<td>Germline Cancer - ICCon</td>
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<td>300</td>
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<td>Robyn Ward</td>
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Total Recruitment: ~4250 (5 years)
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<th>Flagship Lead(s)</th>
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<td>520</td>
<td>Nigel Laing</td>
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<tr>
<td>Somatic Cancer *with MGHA</td>
<td>AGHA</td>
<td>265</td>
<td>Stephen Fox</td>
<td>Mitochondrial Disorders</td>
<td>AGHA</td>
<td>210</td>
<td>John Christodoulou, David Thorburn</td>
</tr>
<tr>
<td>Germline Cancer - Paediatric / AYA</td>
<td>AGHA</td>
<td>1400</td>
<td>David Thomas</td>
<td>Epileptic Encephalopathy</td>
<td>AGHA</td>
<td>120</td>
<td>Ingrid Scheffer</td>
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<tr>
<td>Germline Cancer - ICC</td>
<td>AGHA</td>
<td>300</td>
<td>Robyn Ward</td>
<td>Brain Malformations</td>
<td>AGHA</td>
<td>120</td>
<td>Rick Leventer, Paul Lockhart</td>
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<tr>
<td>Hereditary Colorectal Cancer</td>
<td>MGHA</td>
<td>35</td>
<td>Alex Boussioutas</td>
<td>Leukodystrophies</td>
<td>AGHA</td>
<td>25</td>
<td>Rick Leventer, Paul Lockhart</td>
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<tr>
<td>Acute Myeloid Leukaemia</td>
<td>MGHA</td>
<td>45</td>
<td>Andrew Roberts</td>
<td>KidGen Renal Genetics</td>
<td>AGHA</td>
<td>560</td>
<td>Andrew Mallett</td>
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<tr>
<td>Advanced non-Hodgkin Lymphoma</td>
<td>MGHA</td>
<td>105</td>
<td>Stephen Opat Miles Prince</td>
<td>Genetic Immunology</td>
<td>AGHA</td>
<td>200</td>
<td>Matthew Cook</td>
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<td>Solid Tumour Cancers *with AGHA</td>
<td>MGHA</td>
<td>200</td>
<td>Jayesh Desai</td>
<td>Intellectual Disabilities</td>
<td>AGHA</td>
<td>50 trios (150)</td>
<td>Tony Roscioli, Mike Field</td>
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<tr>
<td>Cutaneous Malignant Melanoma</td>
<td>QGHA</td>
<td>380</td>
<td>Peter Soyer</td>
<td>Childhood Syndromes</td>
<td>MGHA</td>
<td>145</td>
<td>Sue White</td>
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<td>Lung Cancer</td>
<td>QGHA</td>
<td>~400</td>
<td>Matt Brown</td>
<td>Hereditary Neuropathies</td>
<td>MGHA</td>
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<td>Monique Ryan</td>
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<td>Metastatic Melanoma</td>
<td>SGC</td>
<td>400</td>
<td>Graham Mann</td>
<td>Focal Epilepsy</td>
<td>MGHA</td>
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<td>Patrick Kwan</td>
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<td>Complex Disorders</td>
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<td>Complex Care</td>
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<td>Perinatal Autopsy</td>
<td>MGHA</td>
<td>110</td>
<td>George McGillivray</td>
<td>Congenital Deafness</td>
<td>MGHA</td>
<td>112</td>
<td>David Amor</td>
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<td>Schizophrenia</td>
<td>SGC</td>
<td>400</td>
<td>Murray Cairns</td>
<td>Dilated Cardiomyopathy</td>
<td>MGHA</td>
<td>100</td>
<td>Paul James</td>
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<td>Mendelian Disorders</td>
<td>SGC</td>
<td>100</td>
<td>Tony Roscioli</td>
<td>Immunology</td>
<td>MGHA</td>
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<td>Jo Douglass</td>
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<td>Epilepsy</td>
<td>SGC</td>
<td>100</td>
<td>Tony Roscioli</td>
<td>Bone Marrow Failure</td>
<td>MGHA</td>
<td>150</td>
<td>David Ritchie, Piers Blombery</td>
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<td>Maturity Onset Diabetes of the Young</td>
<td>QGHA</td>
<td>490</td>
<td>John Prins</td>
<td>Kidney Genetics (KidGen)</td>
<td>MGHA</td>
<td>198</td>
<td>Catherine Quinlan</td>
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<td>Complex Neurological / Neurodegenerative disease</td>
<td>MGHA</td>
<td>90 / 20 trios (110)</td>
<td>Samuel Berkovic</td>
<td>Mitochondrial Disease</td>
<td>SGC</td>
<td>370</td>
<td>Carolyn Sue</td>
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<td>Infectious Diseases</td>
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<td>Genetic Disorders of Bone</td>
<td>SGC</td>
<td>150</td>
<td>Andreas Zankl</td>
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<td>Controlling Superbugs</td>
<td>MGHA</td>
<td>2000 - 3000 (bacterial)</td>
<td>Lindsay Grayson Ben Howden</td>
<td>Mendelian Immunodeficiencies</td>
<td>SGC</td>
<td>100</td>
<td>Tony Roscioli</td>
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<td>Nosocomial Infections</td>
<td>QGHA</td>
<td>1500 (bacterial)</td>
<td>David Paterson</td>
<td>Blinding Retinal Dystrophy</td>
<td>SGC</td>
<td>170</td>
<td>Robyn Jamieson</td>
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<td>Future Projects</td>
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<td>Additional Flagships</td>
<td>ALL</td>
<td>7000</td>
<td>TBC</td>
<td>Inherited Cardiomyopathies</td>
<td>SGC</td>
<td>160</td>
<td>Chris Sensarian</td>
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</table>
Our Growth

- **AUSTRALIAN GENOMICS LAUNCHED**
- **NHMRC TCR**
- **AUSTRALIAN GENOMICS FORMED**

- **JAN 14**, **MAY 15**, **JAN 16**, **SEP 16**, **JAN 17**, **MAY 17**, **SEP 17**, **JAN 18**

- **Melbourne Genomics Formed**
- **Sydney Genomics Formed**
- **Canberra Genomics Formed**
- **Old Genomics Formed**
- **Joint Committee on Digital Health and Genomics Formed (ADHA / Australian Genomics)**

**Staff**
- 1 in 2014, 55 in 16 months

**Investigators**
- 50 in 2014, 230 in 18 months

**Partner Institutions**
- 30 in 2014, 80 in 18 months

**Projected recruitment**
- of 2000 in two years

**Projects**
- 80% growth in 18 Months
Global Consortium
OF NATIONAL GENOMICS INITIATIVES

“No one country can do it alone”

- National Initiatives encouraged to implement tools and standards being developed by GA4GH (code of conduct, ethics, consent standards).
- Established initiatives should share best practices to assist emerging initiatives.

Participants of Meeting #2 held in May 2017 – co-hosted by Australian Genomics and Genomics England:

- Australia
- Brazil
- Canada
- Denmark
- Finland
- GenomeAsia 100K
- Global Gene Corp (India)
- Netherlands
- Qatar
- South Africa
- Switzerland
- Turkey
- United States of America
- United Kingdom
- + Global Alliance for Genomics & Health