Implementing Genomics into Clinical Care

Jeffrey Braithwaite
Janet C Long
Stephanie Best
Natalie Taylor

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Thank you

Professor Kathryn North
Dr Tiffany Boughtwood
Associate Professor Zornitza Stark
Hossai Gul
Professor Robyn Ward
Professor Nigel Laing
The National Steering Committee in Genomics
The Australian Genomics Health Alliance and all involved …
Associate Professor Clara Gaff
The MacKenzie’s Mission team (Professor Edwin Kirk, Professor Martin Delatycki)
Lots of other people …
Implementing Genomics into Clinical Care

A presentation and discussion in three parts:

Part 1: A primer
Part 2: Progress with implementation
Part 3: Discussion: Q and A
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Part 1: A primer
Aims of Australian Genomics

• “Australian Genomics uses its national footprint and diverse network of partners to drive research through the clinical flagship projects, and the program areas that model the infrastructure supporting genomic health service delivery.”

• “The flagships and programs intersect across multiple projects.”
National approach to data federation & analysis
Establishing standards & processes to capture and use genomic & clinical data
Evaluation, policy & ethics
Building evidence for scalable, sustainable and equitable genomic healthcare
Genomic Workforce & Education
Mapping workforce education & training needs for effective delivery of genomic healthcare

National diagnostic & research network
Driving a coordinated & sustainable system for genomic healthcare

RARE DISEASE FLAGSHIPS

CANCER FLAGSHIPS

REPRODUCTIVE CARRIER SCREENING (MACKENZIE'S MISSION)

CLINICAL OUTCOMES

PREVENTION
EARLY DIAGNOSIS
EARLY INTERVENTION
MONITORING
PRECISION THERAPY
REPRODUCTIVE CONFIDENCE

ANALYSIS to provide a strong, ethically informed evidence base for applying genomics to clinical practice
A big issue

The big issue is changing the funding for these networks from research funding from the Australian Genomics NHMRC grant, or the MRFF Australian Genomics Health Futures Mission grants, such as Mackenzie’s Mission, into **sustainable funding of standard of care genomic diagnostics** through State and Federal Health Departments.

[Courtesy Nigel Laing]
MSAC applications

• Australian Genomics has submitted an MSAC application for Childhood Syndromes which has been approved

• Other Flagships are also heading towards MSAC applications
The problem in a picture: making it look easy

[Best et al, Deep inside the genomics revolution, in press]
The problem in words with references: it’s harder than that

- Advances in genetic and genomic research have promised to transform future approaches to disease prevention, detection and treatment \(^1\text{-}^6\).

- However, the process of routinely integrating these changes into existing healthcare systems has been slow and challenging, with health systems struggling to keep up with the exponential speed at which the genomics evidence-base is evolving \(^4\text{-}^7\).

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It doesn’t just happen …

Hope is not a plan
Why is it difficult for healthcare professionals?

They are already very busy before genomics arrives.

Changing behaviour is difficult and complex.

People often rely on intuition rather than gaining a clear understanding of key barriers to change.

Methods used often lack the necessary components that are effective in producing behaviour change.
We think healthcare works like this:

But it actually works like this:

So we can’t change it just by asking, telling, demanding or insisting it change
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Part 2: Progress with implementation
Implementation Science (programme three)-Australian Genomics

**Complexity:** Understand Australian Genomics as a complex adaptive system with a view to leveraging emergent features

**Implementation of genomics:** Understanding service pathways and clinical processes with the aim of co-developing an implementation plan to facilitate adoption of genomics in clinical practice across Australia that is cost-effective, clinically useful and feasible

### Flagships

- **Ultra rapid acute care** barriers and enablers to implementation
- **KidGen:** e.g. role of the MDT, process mapping
- **Mackenzie’s Mission** e.g. Referrers’ attitudes and perspectives

### Programmes

- **Programme one** e.g. strategic landscape analysis of unmet need
- **Programme three** e.g. Discrete Choice Experiments – what is valued
- **Programme four** e.g. workforce development

### Other

- **Melbourne Genomics** e.g. knowledge translation
- **Laboratories** e.g. Communication for variant prioritisation
- **New study:** Active implementation of Mitochondrial guidelines

### Implementation work covers

- Policy
- Organisation
- Clinician/lab
- Individual / community
Australian Genomics: a learning community

Pre-2016
(before Australian Genomics)
Ties=2,925; Nodes=384

2018
Ties=6,381; Nodes=384

[Long et al, 2019]
Australian Genomics: a learning community

NEW TIES

People who met through Australian Genomics

Ties=3,351; Nodes=384

[Long et al, 2019]
Australian Genomics: a learning community

Collaborators from outside Australian Genomics (within Australia)

Ties=464; Nodes=412

[Long et al, 2019]
Describing Australian Genomics as a complex adaptive system
How we are using these ideas in a complex health system

1) Understand the complex system

2) Develop principles of implementation

- Management approval and ongoing support
- Commitment amongst members of the target group
- Use of boundary spanners
- Mapping of guidelines onto local problems
- Process mapping with supporting audit data
- Adopting the perspective of the target group
- Acknowledging the complexity of implementing evidence (i.e., changing behaviour) in practice
- A monitoring plan
- A flexible approach that is driven by local context
- Co-production and design to combine theoretical and contextual expertise
- Incorporation into established structures

[Taylor et al, 2014, 2019]
3) Use determinant frameworks

- **What do determinant frameworks do?**
  - Specify determinants that act as barriers and enablers that influence implementation outcomes. These can be manipulated to increase the likelihood of change.

- **Example:** Theoretical Domains Framework (TDF)
  - 12/14 determinants of change
  - Separately mapped to behaviour change techniques (BCTs)

### TDF version 2 (Cane, O’Connor et al. 2012)

<table>
<thead>
<tr>
<th>Determinants</th>
</tr>
</thead>
<tbody>
<tr>
<td>Skills</td>
</tr>
<tr>
<td>Beliefs about capabilities</td>
</tr>
<tr>
<td>Goals</td>
</tr>
<tr>
<td>Environmental context and resources</td>
</tr>
<tr>
<td>Beliefs about consequences</td>
</tr>
<tr>
<td>Emotion</td>
</tr>
<tr>
<td>Social influences</td>
</tr>
<tr>
<td>Social/professional role and identity</td>
</tr>
<tr>
<td>Knowledge</td>
</tr>
<tr>
<td>Memory, attention and decision processes</td>
</tr>
<tr>
<td>Intentions</td>
</tr>
<tr>
<td>Reinforcement</td>
</tr>
<tr>
<td>Optimism</td>
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<tr>
<td>Behavioural Regulation</td>
</tr>
</tbody>
</table>
## Stage 1 (complete): Coding barriers and (mapping) enablers

<table>
<thead>
<tr>
<th>Implementation</th>
<th>Barriers</th>
<th>Enablers</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Lack of leadership on the ground</td>
<td>Developing and supporting champions</td>
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<tr>
<td></td>
<td>CEO engagement</td>
<td>Support from senior leaders</td>
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<tr>
<td></td>
<td>DoH advocate</td>
<td>Advocacy to raise the genomic profile at govt level</td>
</tr>
<tr>
<td></td>
<td>Organisational focus on day job</td>
<td>Leadership from the top</td>
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<tr>
<td></td>
<td>Challenge for smaller genetic units to absorb workload</td>
<td>Developing flexible, agile dept support</td>
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<tr>
<td></td>
<td>Very organised, hyper accountable health systems limit potential to innovate</td>
<td>More control over budget</td>
</tr>
</tbody>
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[Taylor, Long, Best et al, 2019]
Stage 2 (proposed): Align relevant service level barriers to individual level TDF-coded barriers

Service Level barrier
- Organisations don't understand how transformative genomics is
- The day job – awareness clinicians are already overloaded
- Lack of funding/ facilities/ overstretched resources/funding model/time poor

Clinician barrier(s)
- Physicians lack knowledge of value of genomics
- Perception of an organisational pre-occupation with fundamental health care/high thru put specialities
- Words more than action

TDF coding
- Beliefs about consequences
- Goals/goal priority
- Intentions

At the service level there is awareness of clinicians workloads.
From the clinical perspective there is thought that some organisations are focused on ‘basic' healthcare (not innovation) and care that is easy to sign off through Medicare

[Taylor, Long, Best et al, 2019]
Stage 3 (proposed): Aligning relevant service level barriers to suggested enablers

**SL barrier**
- Organisations don’t understand how transformative genomics is
- The day job
- Lack of funding/ facilities/ time poor/ overstretched resources/funding model

**Clinician barrier(s)**
- Physicians lack knowledge of value of genomics
- Pre-occupation with fundamental health care/high thru put specialities
- Words more than action

**TDF coding**
- Beliefs about consequences
- Goals/goal priority
- Intentions

**Enablers**
- No enabler
- Leadership from top (CL)
- Funding++/health econ data/ decreasing cost of sequencing (SL)

CL = clinician level ideas
SL = service level ideas

[Taylor, Long, Best et al, 2019]
Stage 4/5 (proposed): Mapping enablers to BCTs and developing theory informed strategies

**SL barrier**
- Organisations don’t understand how transformative genomics is
- The day job
- Lack of funding/facilities/time poor/overstretched resources/funding model

**Clinician barrier**
- Physicians lack knowledge of value of genomics
- Pre-occupation with fundamental health care/high thru put
- Words more than action

**TDF coding**
- Beliefs about consequences
- Goals/goal priority
- Intentions

**Enablers**
- No enabler
- Leadership from top (CL)
- Funding+/health econ data/decreasing cost of sequencing (SL)

**BCTs**
- N/A
- Social support (practical)

[Taylor, Long, Best et al, 2019]
Stage 6 (proposed): coding barriers and enablers to key issues

**SL barrier**
- Organisations don’t understand how transformative genomics is
- The day job
- Lack of funding/facilities/time poor/overstretched resources/funding model

**Clinician barrier**
- Physicians lack knowledge of value of genomics
- Pre-occupation with fundamental health care/high thru put
- Words more than action

**TDF coding**
- Beliefs about consequences
- Goals/goal priority
- Intentions

**Enablers**
- No enabler
- Leadership from top (CL)
- Funding++/health econ data/decreasing cost of sequencing (SL)

**BCTs**
- N/A
- Social support (practical) or look at Kok et al

**TSci Key issues**
- Market analysis (need and values of target audiences)

[Taylor, Long, Best et al, 2019]
The way forward?

• Many of us think that we will need a mix of Federal and State funding of genomic diagnostics in Australia

• And funding to support workforce education and capacity-building

• At this point, it is not clear how this will work

[Courtesy Nigel Laing]
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Part 3: Discussion: Q and A
Discussion points

• Implementation may end up being a big challenge

• Turning one of the most important research projects for Australia and internationally into routine care will not be easy

• There are many natural experiments in the world and amongst our own Australian flagships

• But we’re not yet clear, despite having a policy plan, what the journey will be like, and where we will end up, by the end of the 2020s