AUSTRALIAN INSTITUTE
OF HEALTH INNOVATION
Faculty of Medicine and Health Sciences

MACQUARIE
University

Implementing Genomics into Clinical Care

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The *Genomics in Australian Healthcare: Foundations to Future Conference*

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

Implementing Genomics into Clinical Care

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

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

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

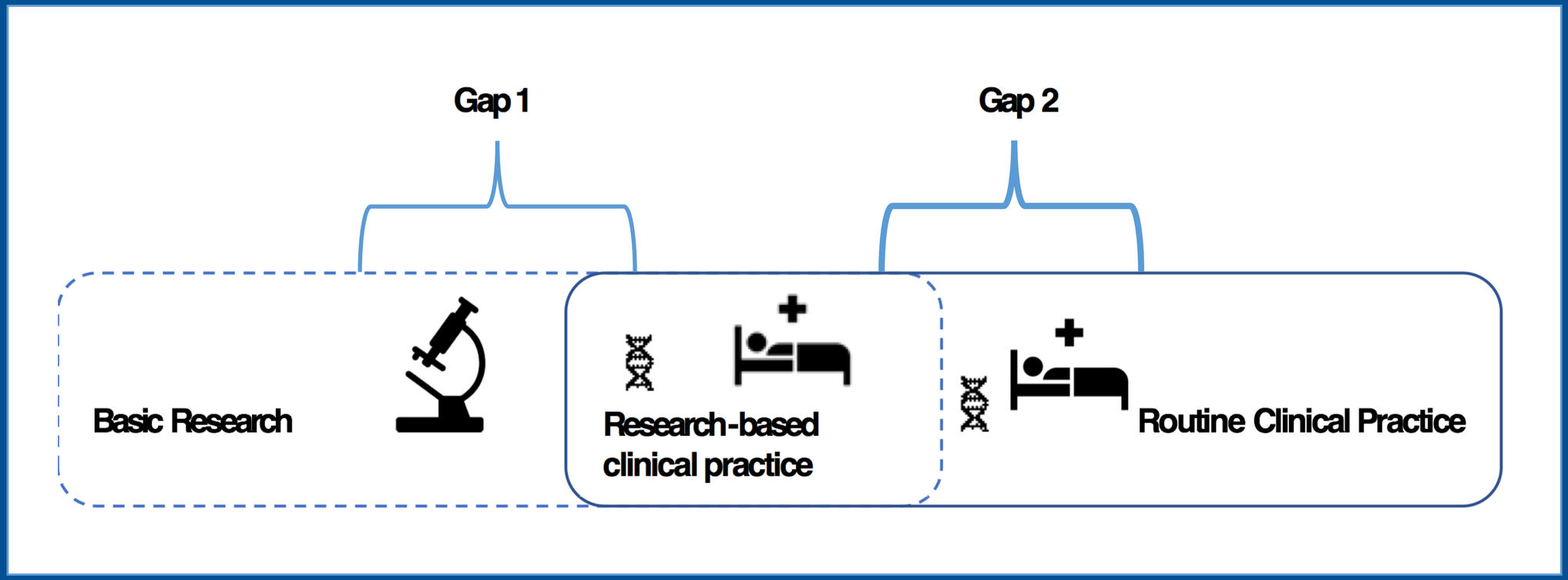
[Courtesy Nigel Laing]

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** [1-6]
- 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 [4,7]

1Khoury MJ, Gwinn M, Yoon PW, Dowling N, Moore CA, Bradley L. The continuum of translation research in genomic medicine: how can we accelerate the appropriate integration of human genome discoveries into health care and disease prevention? Genet Med. 2007;9(10):665-74.
2Shen T, Yeat NC, Lin JC-H. Clinical applications of next generation sequencing in cancer: from panels, to exomes, to genomes. Front Genet. 2015;6:215.
3Manolio TA, Chisholm RL, Ozenberger B, Roden DM, Williams MS, Wilson R, et al. Implementing genomic medicine in the clinic: the future is here. Genet Med. 2013;15(4):258.
4Roberts MC, Kennedy AE, Chambers DA, Khoury MJ. The current state of implementation science in genomic medicine: opportunities for improvement. Genet Med. 2017;19(8):858-63.
5Stark Z, Schofield D, Alam K, Wilson W, Mupfeki N, Macciocca I, et al. Prospective comparison of the cost-effectiveness of clinical whole-exome sequencing with that of usual care overwhelmingly supports early use and reimbursement. Genet Med. 2017;19(8):867–74.
6Gaff CL, Winship IM, Forrest SM, Hansen DP, Clark J, Waring PM, et al. Preparing for genomic medicine: A real world demonstration of health system change. npj Genomic Med [Internet]. 2017;2(1):1–8. Available from: http://dx.doi.org/10.1038/s41525-017-0017-4
7Powell BJ, Waltz TJ, Chinman MJ, Damschroder LJ, Smith JL, Matthieu MM, et al. A refined compilation of implementation strategies: results from the Expert Recommendations for Implementing Change (ERIC) project. Implement Sci. 2015;10(1):21.

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

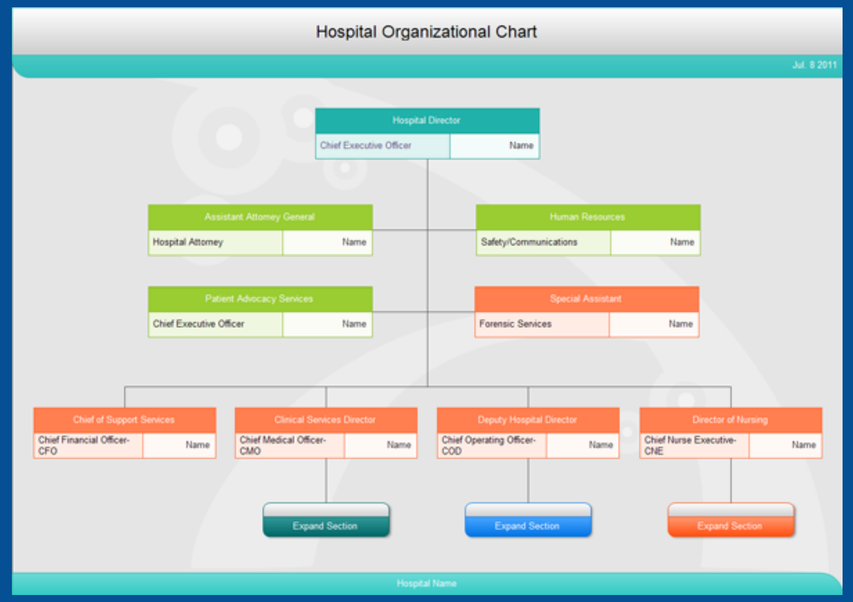
Changing behaviour is difficult and complex

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

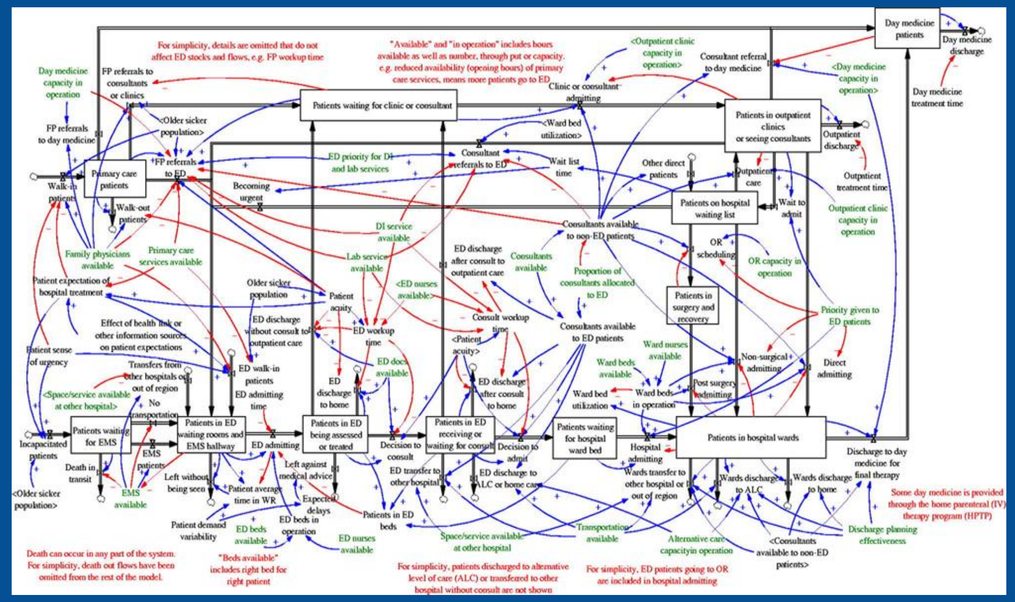
Methods used often lack the necessary components that are effective in producing behaviour change

And this challenge …

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

Implementing Genomics into Clinical Care

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

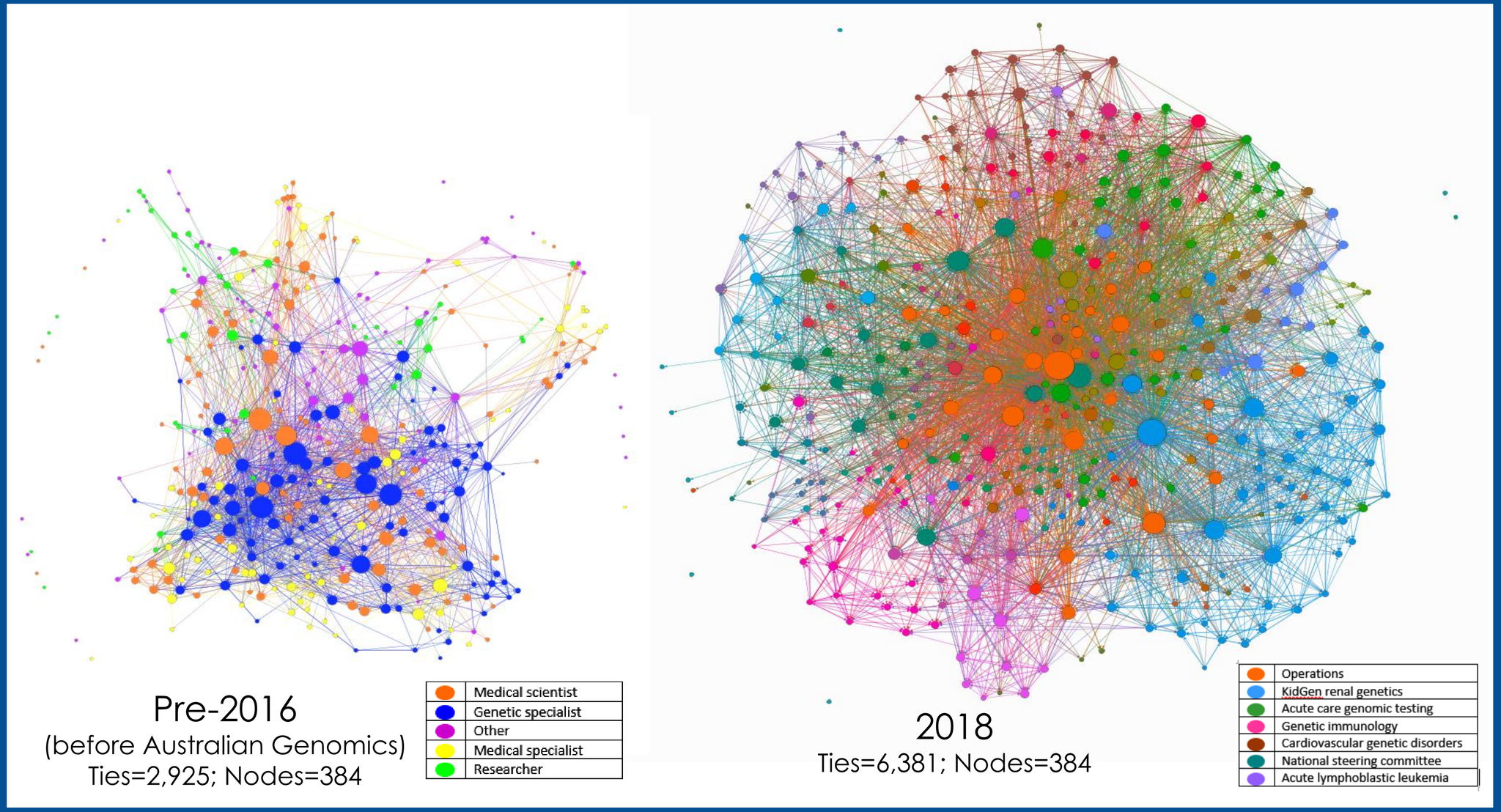
Programme one e.g. strategic landscape analysis of unmet need
Programme three e.g. Discrete Choice Experiments – what is valued
Programme four e.g. workforce development

Other

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

Implementation work covers

Australian Genomics: a learning community

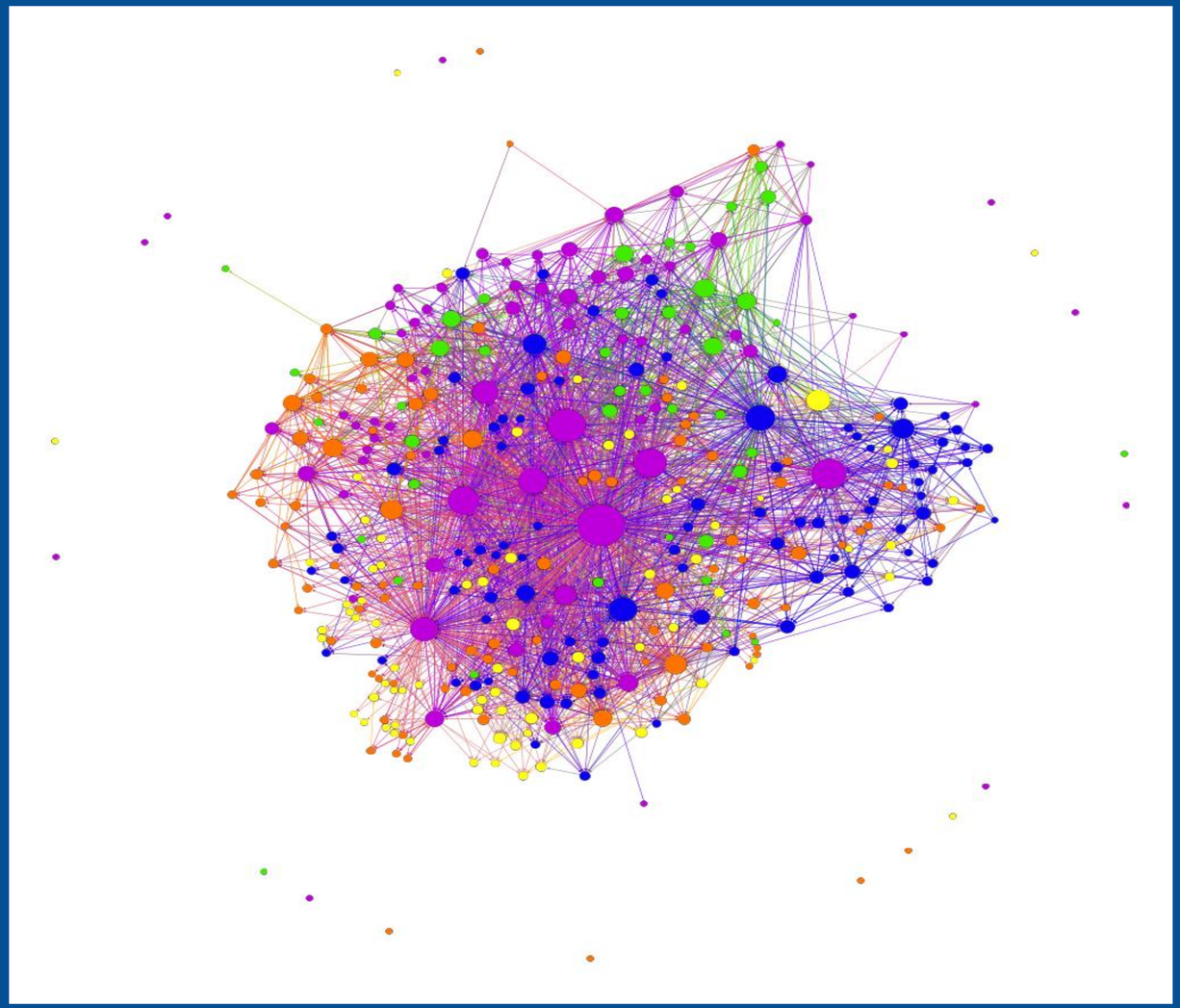


Pre-2016
(before Australian Genomics)
Ties=2,925; Nodes=384

2018
Ties=6,381; Nodes=384

[Long et al, 2019]

Australian Genomics: a learning community



[Long et al, 2019]

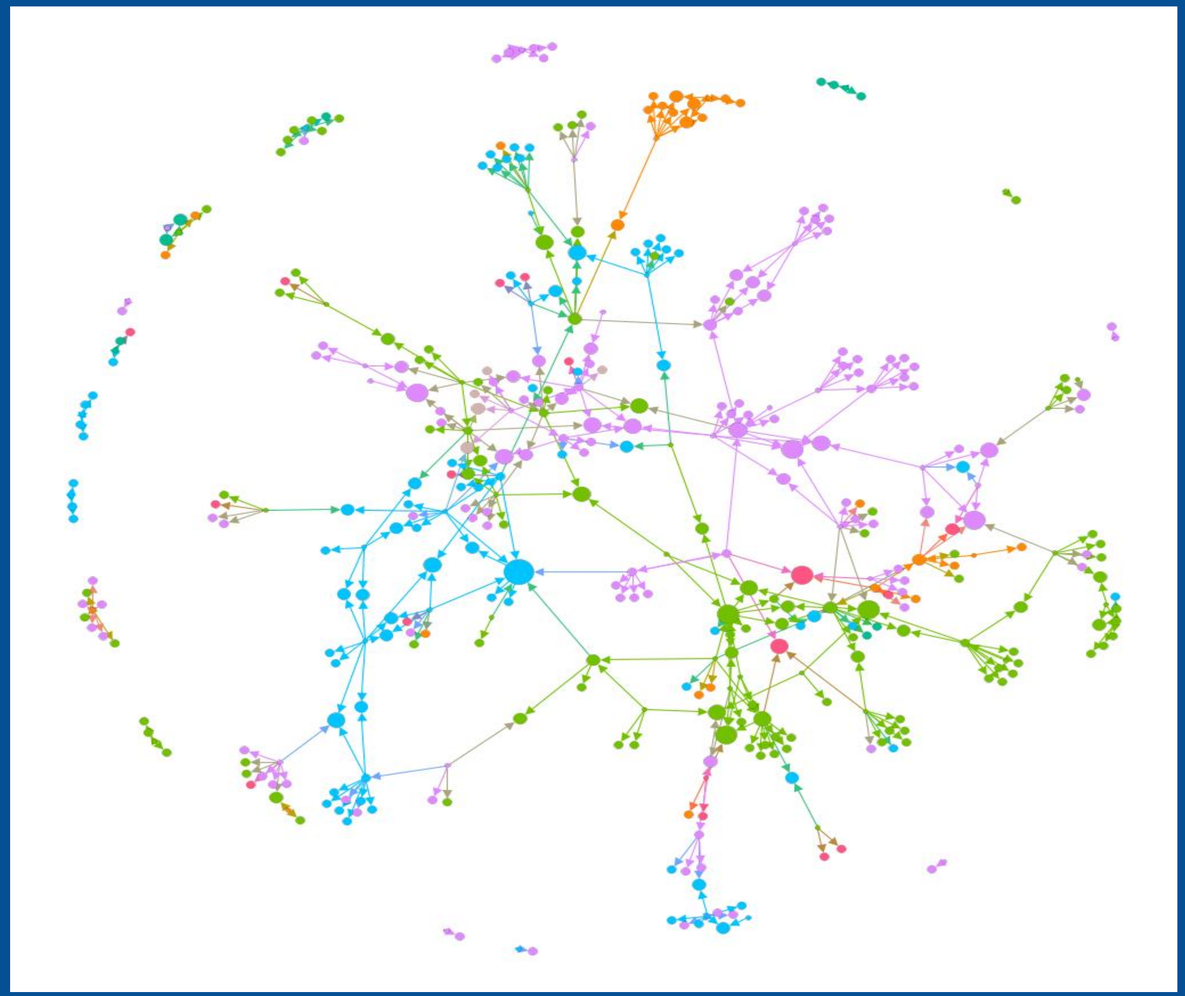
NEW TIES

People who met through Australian Genomics

Ties=3,351; Nodes=384

	Medical scientist
	Genetic specialist
	Other
	Medical specialist
	Researcher

Australian Genomics: a learning community



Collaborators from outside Australian Genomics (within Australia)

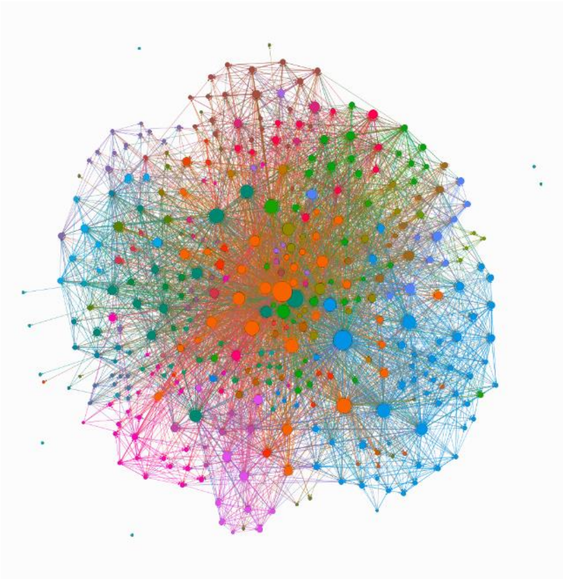
Ties=464; Nodes=412

[Long et al, 2019]

2016

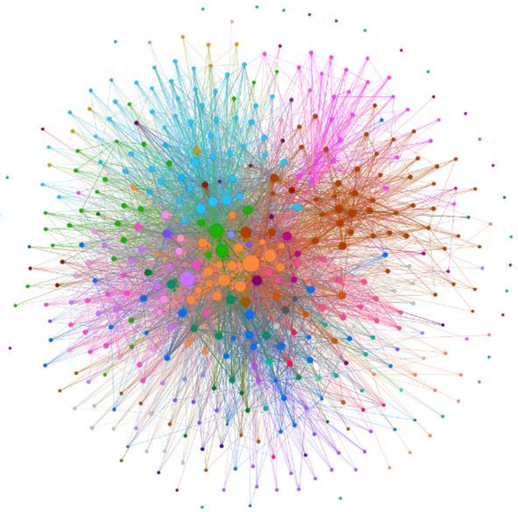
n=186
Density=2.0%
Reported ties (median) =10

2018



n=384
Density= 4.3%
Reported ties (median) = 17

2019



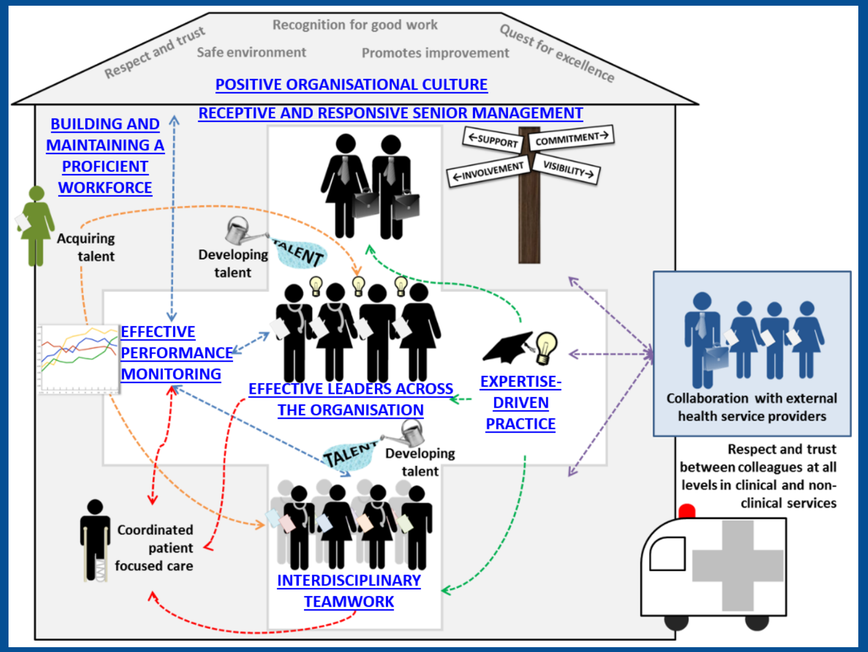
n=439
Density=3.6%
Reported ties (median) = 26

JC Long, C Pomare, Stephanie Best, et al. 2019. "Building a knowledge exchange network in Australian clinical genomics: a social network study of the Australian Genomic Health Alliance." *BMC Medicine* 17 (44)

Describing Australian Genomics as a complex adaptive system

How we are using these ideas in a complex health system

1) Understand the complex system




RESEARCH ARTICLE Open Access

High performing hospitals: a qualitative systematic review of associated factors and practical strategies for improvement

Natalie Taylor[1*], Robyn Clay-Williams[1], Emily Hogden[1], Jeffrey Braithwaite[1] and Oliver Groene[2]

Abstract

Background: High performing hospitals attain excellence across multiple measures of performance and multiple departments. Studying high performing hospitals can be valuable if factors associated with high performance can be identified and applied. Factors leading to high performance are complex and an exclusive quantitative approach may fail to identify richly descriptive or relevant contextual factors. The objective of this study was to undertake a systematic review of qualitative literature to identify methods used to identify high performing hospitals, the factors associated with high performers, and practical strategies for improvement.

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

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

- **<u>Example:</u>** *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)*
Skills
Beliefs about capabilities
Goals
Environmental context and resources
Beliefs about consequences
Emotion
Social influences
Social/professional role and identity
Knowledge
Memory, attention and decision processes
Intentions
Reinforcement
Optimism
Behavioural Regulation

Behaviour

Cane, J., O'Connor, D. and Michie, S., 2012. Validation of the theoretical domains framework for use in behaviour change and implementation research. Implementation science, 7(1), p.37.

Stage 1 (complete): Coding barriers and (mapping) enablers

Implementation	
Barriers	**Enablers**
Lack of leadership on the ground	Developing and supporting champions
CEO engagement	Support from senior leaders
DoH advocate	Advocacy to raise the genomic profile at govt level
Organisational focus on day job	Leadership from the top
Challenge for smaller genetic units to absorb workload	Developing flexible, agile dept support
Very organised, hyper accountable health systems limit potential to innovate	More control over budget

[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]

National Health Genomics Policy Framework Implementation Plan 2018-2021

Implementing Genomics into Clinical Care

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