Global Genomic Medicine Collaborative

Genomic Test Evaluation Frameworks

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Conflicts:

Chair of Medical Services Advisory Committee

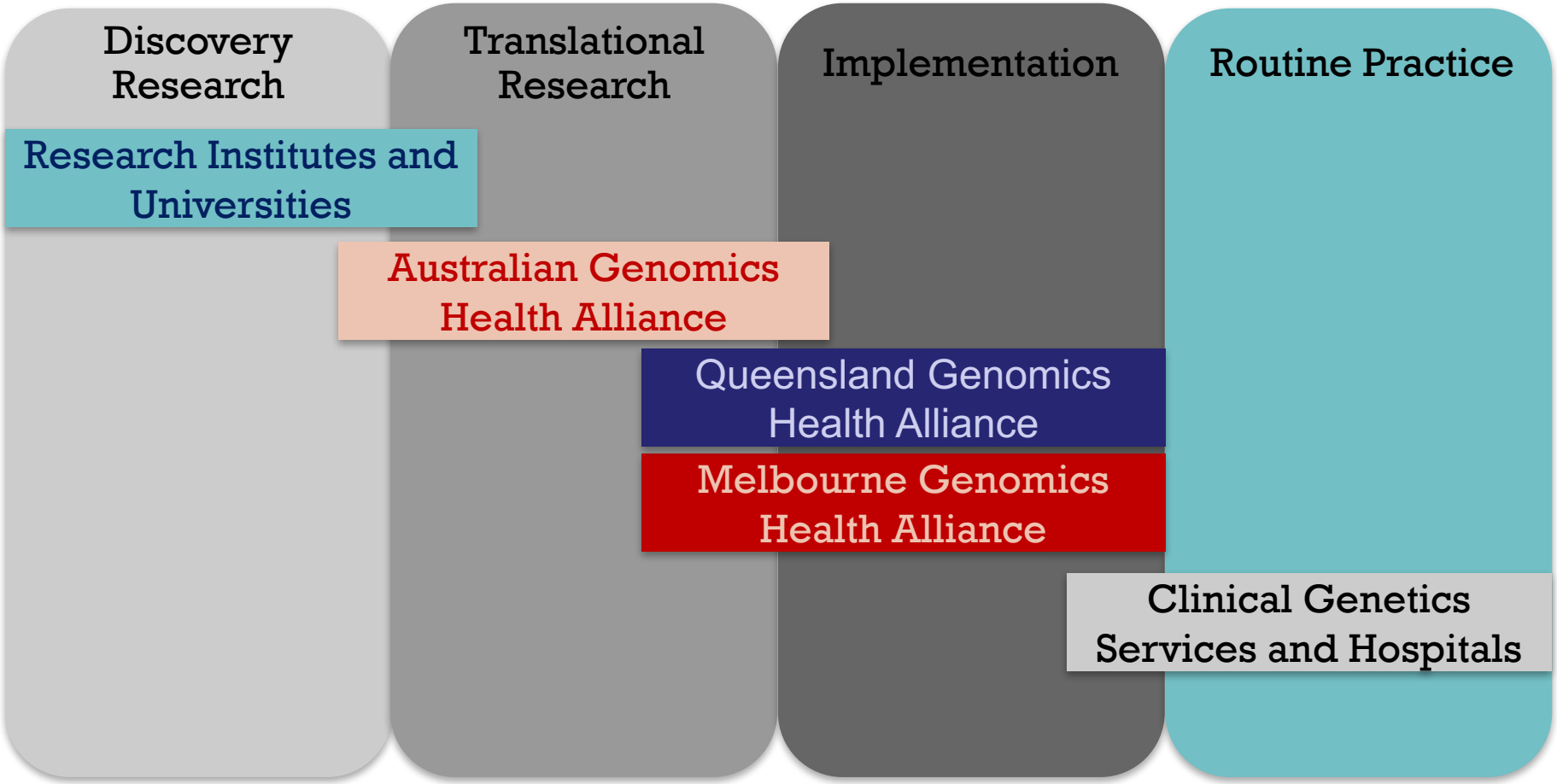
Member Pharmaceutical Benefits Advisory Committee

The University Of Queensland AUSTRALIA

Create change

Genomic health alliances in Australia

Focused on implementing genomics in practice
Whole of system change



GOOD INTENTIONS

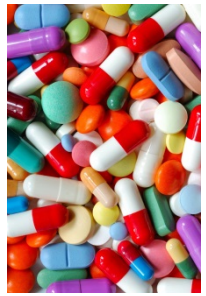
bad results

Overdiagnosis and Overtreatment

“Overdiagnosis”

Overdetection

Overdefinition



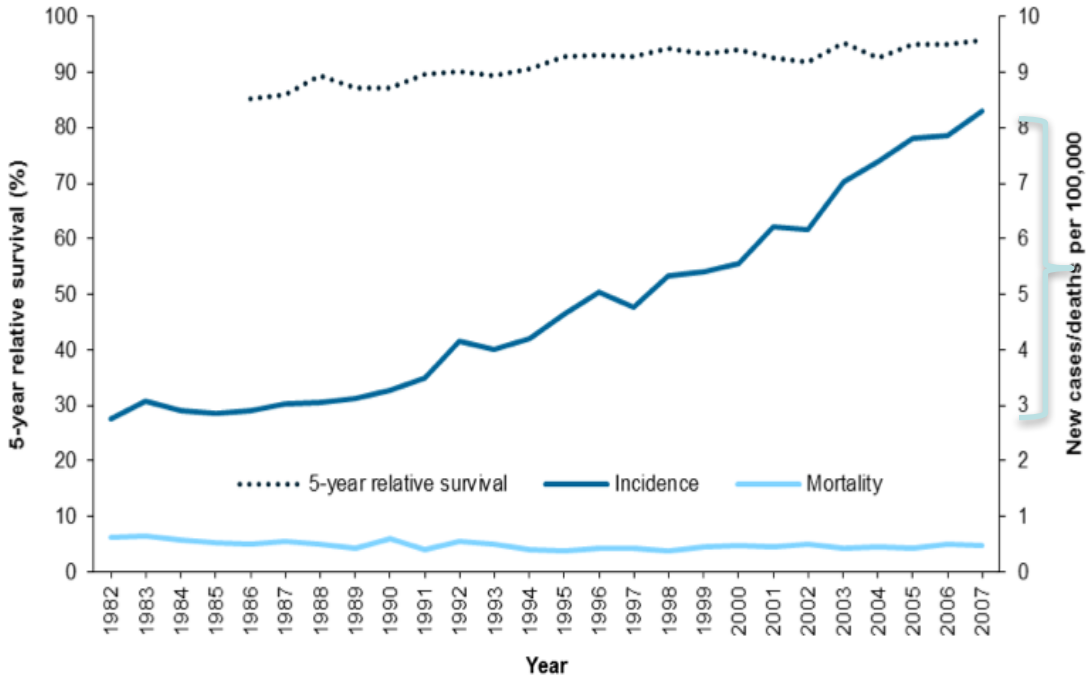
Over-testing

Over-interpretation

Over-treatment

Overdetection: thyroid “cancer"

Thyroid cancer tripled in 25 years; no more deaths

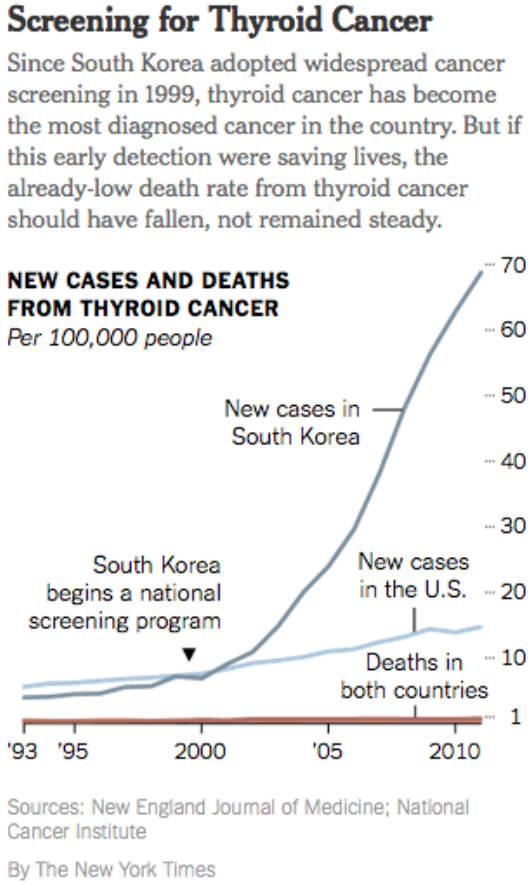


Notes

1. Incidence and mortality rates are age standardised to the Australian population as at 30 June 2001 and are expressed per 100,000 population.
2. Survival data for this figure are presented in online Table S26.3.

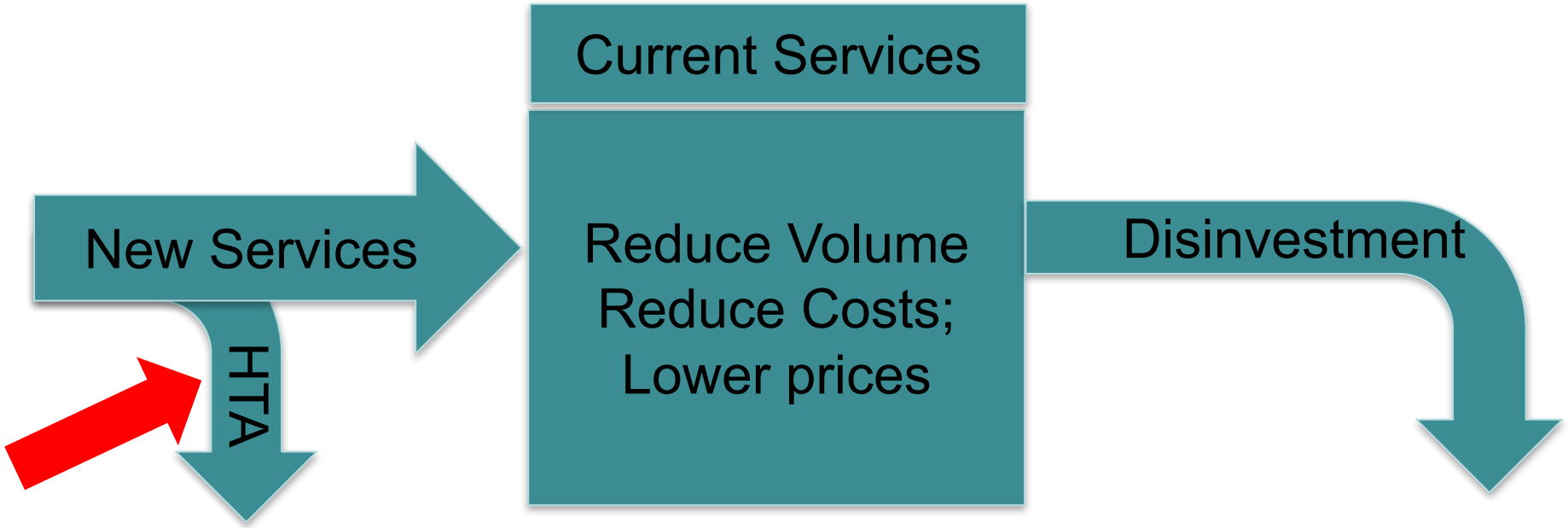
Source: AIHW Australian Cancer Database (2007); AIHW 2010b.

Figure 4.73: Yearly trends in incidence, mortality and 5-year relative survival of thyroid cancer, 1982 to 2007



Options to reduce unnecessary health care expenditure (waste)

1. Restrict uptake (**easy**)
2. Improve efficiency/costs of current services (behavior change – **hard**)
3. Disinvest in unproven technologies (**very hard**)



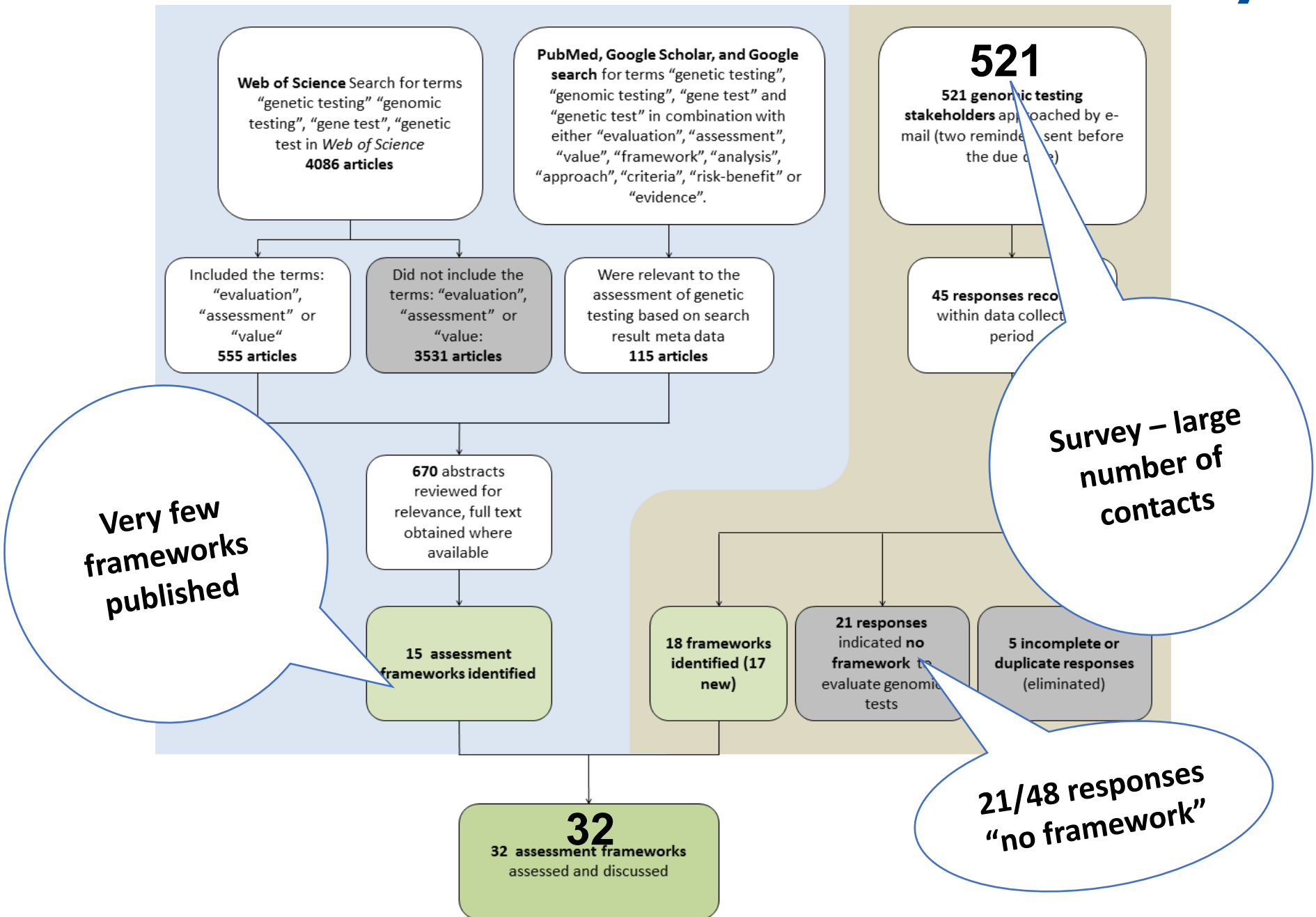
Goal: International comparative analysis of theoretical and practical genomic test evaluation frameworks

Investigators:
Andrew Mitchell (DoH) and Robyn Ward (UQ)
Sarah Nowak & Jakub Hlavka (RAND Corporation)

Funding: National Health and Medical Research Council through the
Australian Genomics Health Alliance
Supported by G2MC – contact lists

Australian Genomics Health Alliance

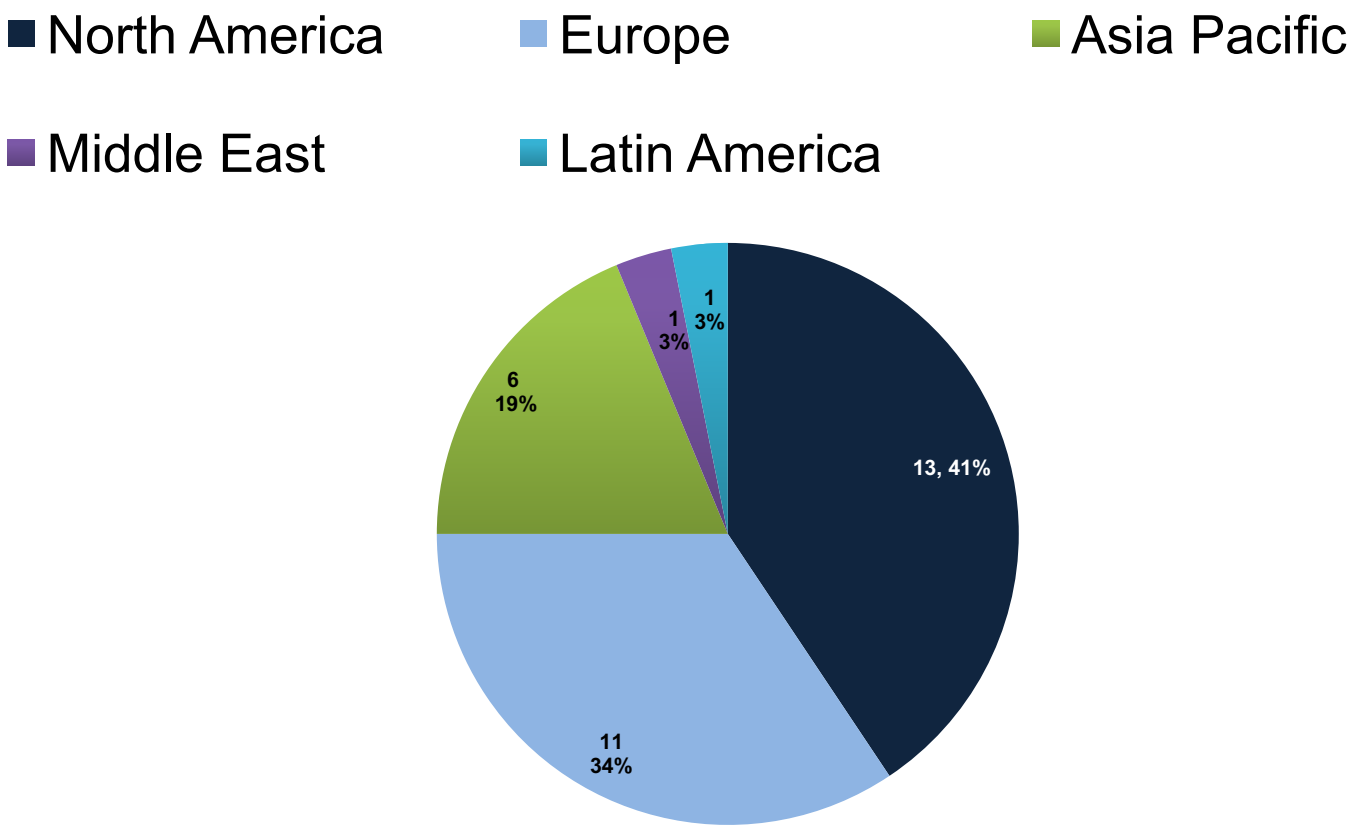
Method – literature review + survey



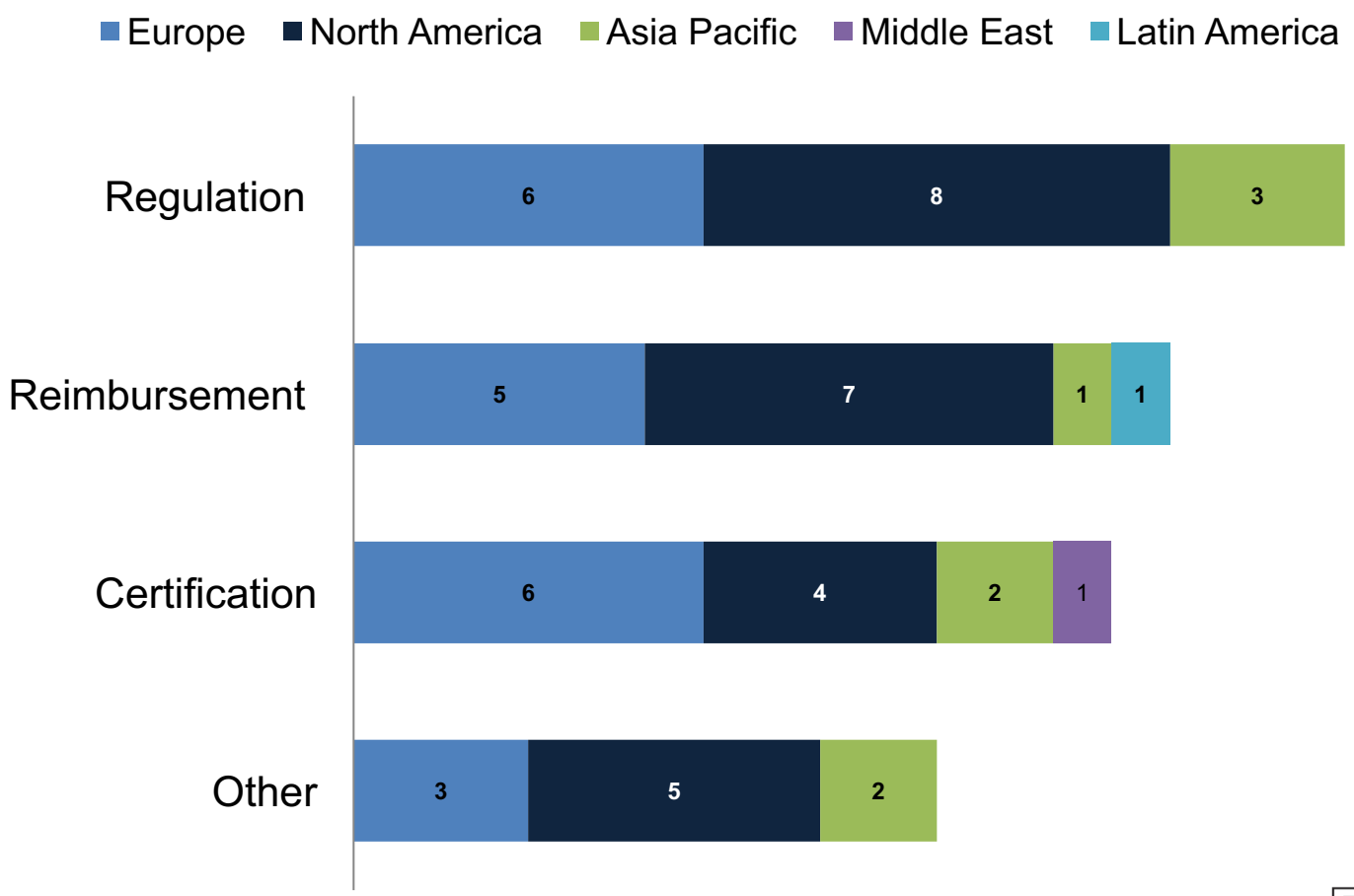
Framework categorisation

- types of decisions the framework informs
  reimbursement, certification, regulation, other
- whether the framework is specific to genetic/genomic testing or more general
- types of test covered by the framework
  single gene, multiple genes/small panel, whole genome scale, RNA expression- single gene/arrays
- test applications covered by the framework
  diagnostic, screening, predictive, prognostic, other
- framework audience
  clinicians, path labs, heath ministries, research institutes, commissioning groups, reimbursement organisations, patients
- types of criteria the framework uses to evaluate tests
  purpose, target population and intended use, population impact, seriousness, appropriateness

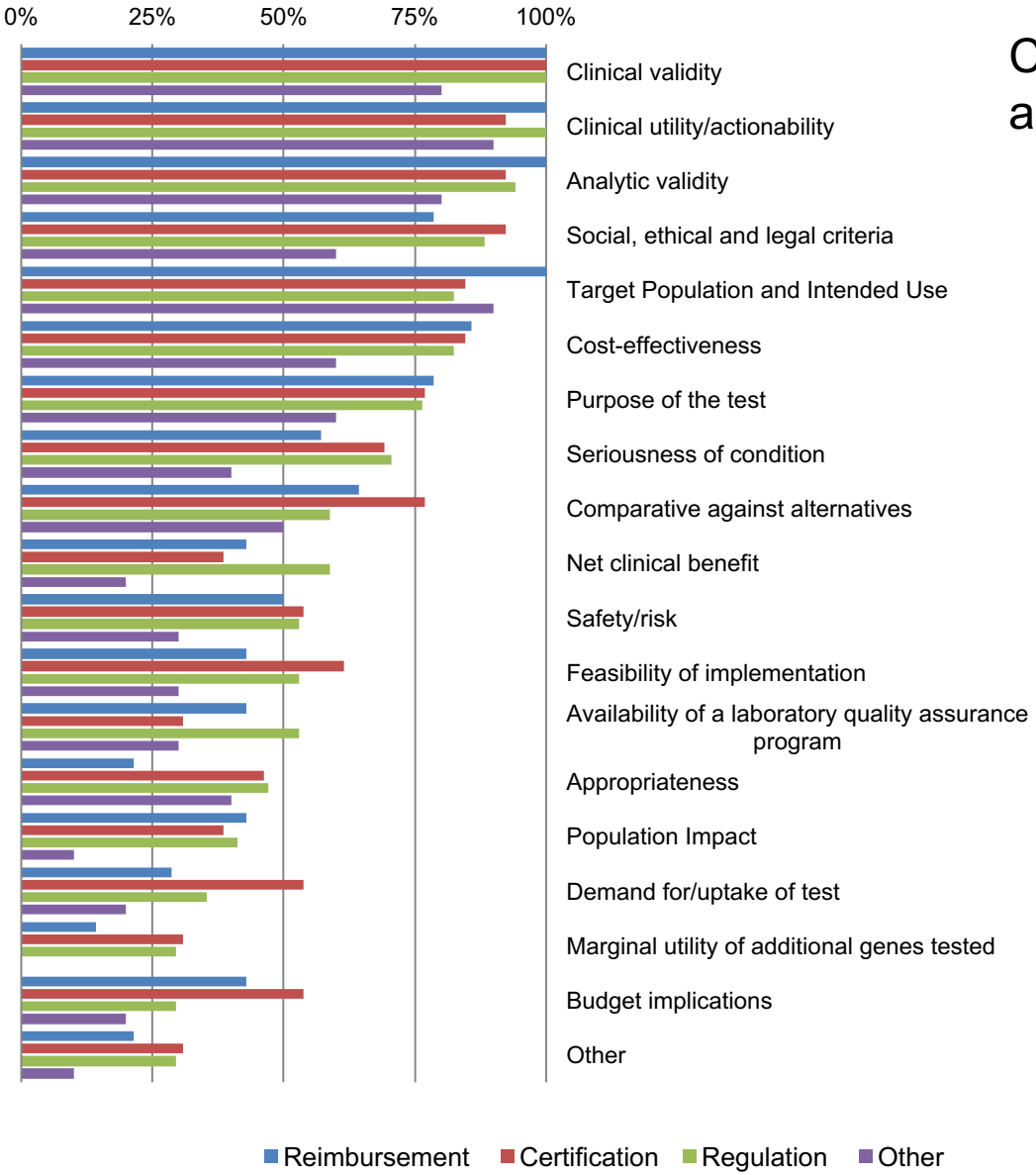
Framework sources by region



Decisions informed by framework



Assessment criteria by different test applications



Clinical validity, utility, analytical validity

Population impact budget implications

Conclusion

- Poor preparedness for implementing testing
  (50% had draft or complete frameworks)

- Frameworks evaluate genetic and genomic tests often consider only benefits and risks common to most screening, predictive, diagnostic and prognostic medical test

But

- Fail to explicitly consider risks that are specific large-scale genomic tests

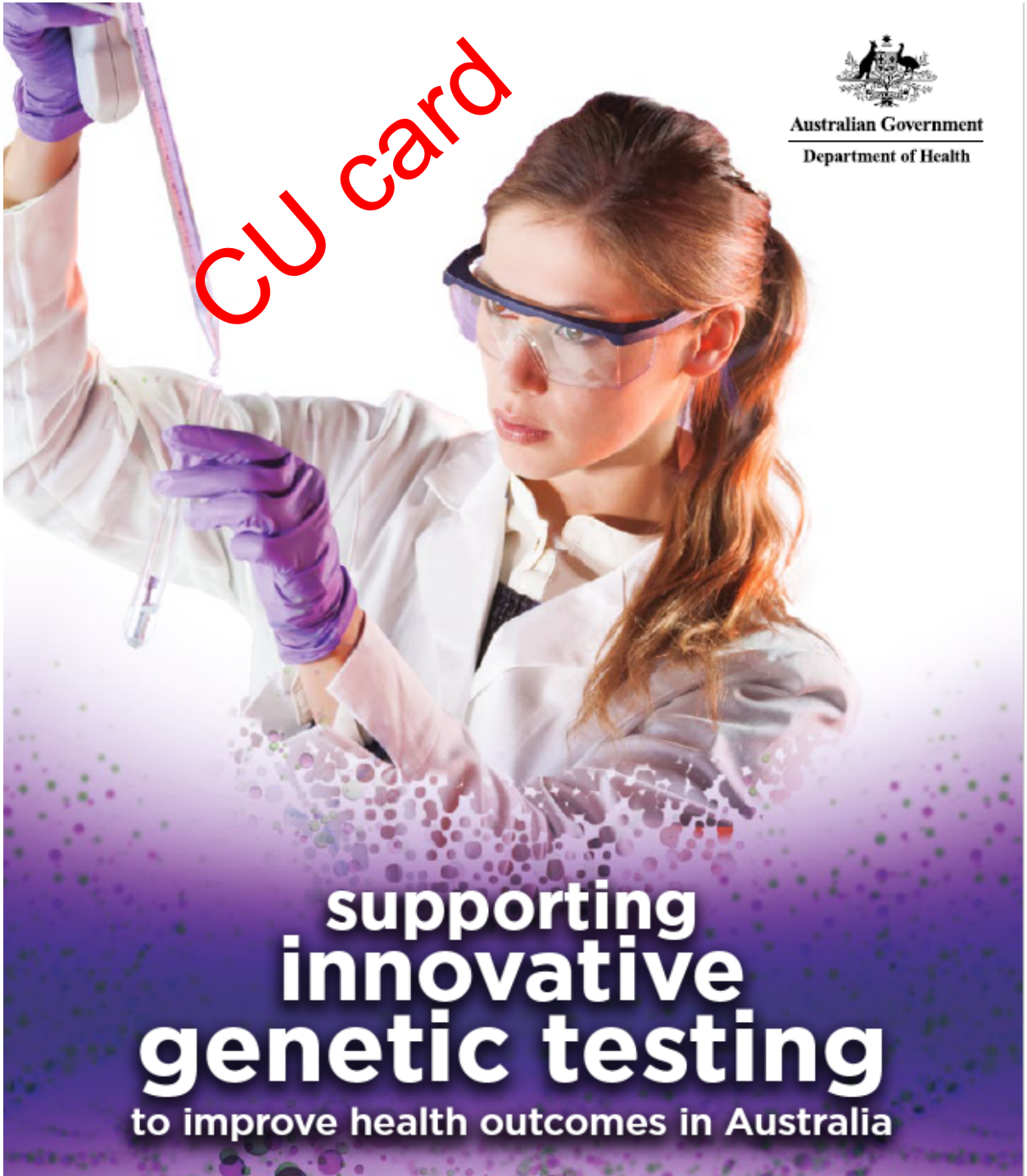
Assessing value of genomics
Australian government published expectations

- Co-dependent technology

  http://www.pbs.gov.au/info/reviews/public-consultation-pbac-guidelines-co-dependent-technology-chapter

- Clinical utility card including release of economic models

  http://www.msac.gov.au/internet/msac/publishing.nsf/Content/917C37D59F3CBF55CA257F6A0071A4A5/$File/1411-Clincial_Utility_Card-accessible.pdf


CU card
Australian Government
Department of Health
supporting
innovative
genetic testing
to improve health outcomes in Australia

Tests need to fulfil the following criteria:

- **analytical validity** can the test measure accurately and reliably the presence or absence of a particular gene or genetic change?
- **clinical validity** can the test identify a genetic predisposition or a particular clinical condition?
- **clinical utility** can the test be used to inform treatment decisions and improve health outcomes?
- **economic evaluation** can the test add value to the health of Australians at an acceptable cost?
- **government budget** can the utilisation and total cost of the test to Australians be justified by the government?

Application of the CUC proforma

- **paediatrics** genetic conditions presenting in infancy or later childhood
- **reproductive planning** families with a history of a particular genetic condition
- **cancer genetics** predisposition to certain cancers or cancer syndromes
- **rare diseases** rare and ultra-rare life-threatening or chronic conditions
- **adult genetics** conditions presenting in adulthood that may have an underlying genetic cause, such as kidney, neurological or cardiac disease

Clinical utility card + economic models

- Scope heritable mutations which predispose to disease
- Started with cancer
- Star performers – best case scenarios
- Complexity with modelling which needs to include:
  - prevention of >1 future disease
  - age specific-relative risks
  - health impact on relatives (joint production)
  - misleading test results
  - parental disutility of caring for disabled children