The National Australian Citizens' Jury on using genomics in newborn bloodspot screening

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Australian Centre for Health Engagement, Evidence and Values

UNIVERSITY
OF WOLLONGONG
AUSTRALIA

The project team

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UOW research team: Di Popic, Lucy Carolan, Chris Degeling, Kathleen Prokopovich, Saniya Singh, Belinda Fabrianesi, Patti Shih, Emma Frost,

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Background

Newborn Bloodspot Screening programs

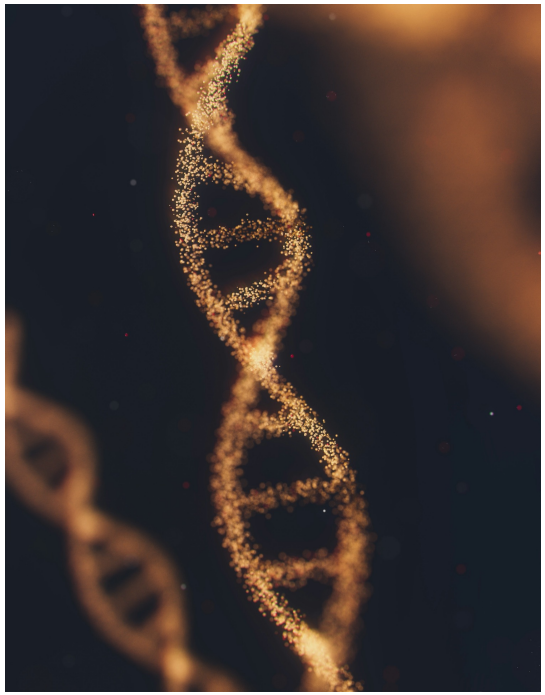
- 99% of Australian babies are screened: focus on harmful, treatable, early-onset conditions, using mostly biochemical tests
- Interest globally in incorporating genomics

https://www.vcgs.org.au/nbs-guideline/

Concerns about introducing genomics

For example...

- Quantity and type of information
- Inequitable access to follow up care
- Uncertain information, risking extended distress for families
- Costs to healthcare system
- Historical reasons for mistrust from Aboriginal and Torres Strait Islander communities



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Deliberative democracy

To support & strengthen legitimacy of representative/aggregative democracy

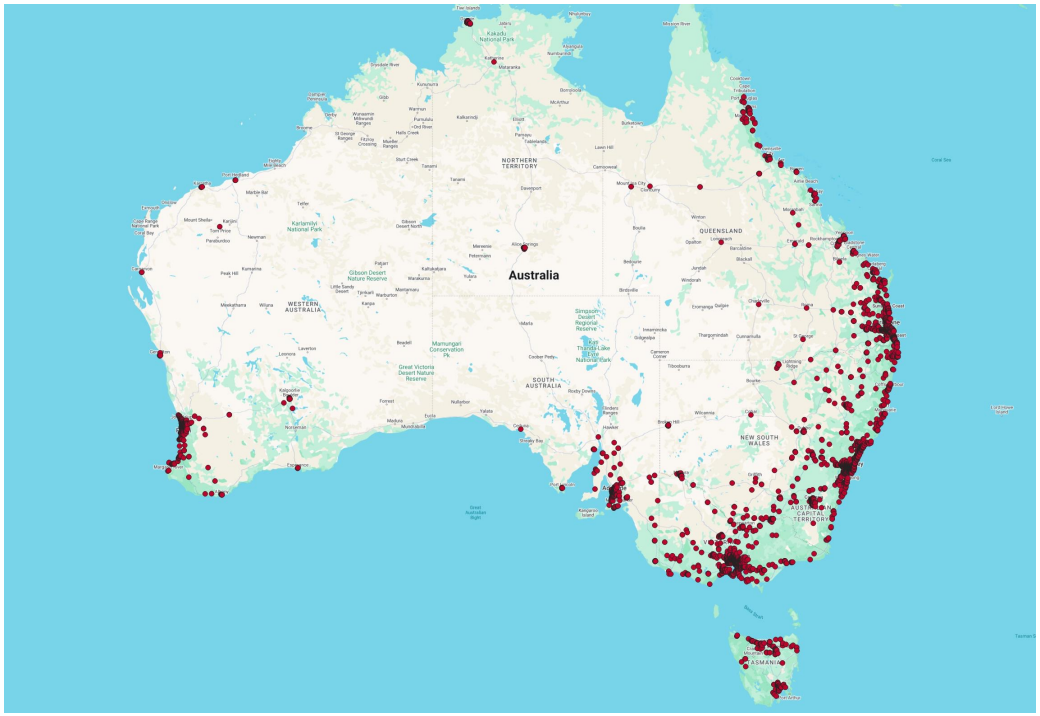
All jury photos used with consent, taken by Belinda Fabrianesi

Recruitment and selection

MANAGED INDEPENDENTLY BY THE SORTITION FOUNDATION

SORTITION FOUNDATION

- 6300 mailed invitations
- 157 unique responses (2.5% response rate)
- 30 jurors selected against eight criteria, aiming to match population proportions as closely as possible



CHARACTERISTIC	PARTICIPANTS N (%)	AUSTRALIAN POPULATION REFERENCE %
SEX		
Women	15 (50%)	45.3%
Men	14 (46.7%)	46.7%
Non-binary or other	1 (3.3%)	8%
AGE GROUP (YEARS)		
18-24	4 (13.3%)	10.8%
25-39	9 (30%)	27.5%
40-54	7 (23.3%)	24.6%
55-74	8 (26.7%)	27.5%
75+	2 (6.7%)	9.6%
ANCESTRY		
Aboriginal &/or Torres Strait Islander	2 (6.7%)	6.7%
African or Middle Eastern	1 (3.3.%)	2.9%
Asian	4 (13.3%)	14.4%
British, North American or New Zealander	17 (56.7%)	54.4%
European	5 (16.7%)	15%
Multiple ancestries (can't pick one)	1 (3.3%)	2.7%
Other	0	3.9%
DISABILITY		
Yes	4 (13.3%)	13.3%
No	26 (86.7%)	86.7%

CHARACTERISTIC	PARTICIPANTS N (%)	AUSTRALIAN POPULATION REFERENCE %
EDUCATION		
Postgraduate qualification	4 (13.3%)	9.6%
Undergraduate degree	6 (20%)	18.7%
Trade or vocational certificate or diploma	10 (33.3%)	27%
Secondary school level	9 (30.0%)	33%
Other or prefer not to say	1 (3.3%)	11.8%
GEOGRAPHY (STATES AND TERRITORIES)		
Australian Capital Territory	2 (6.7%)	1.7%
New South Wales	8 (26.7%)	31.3%
Northern Territory	1 (3.3.%)	1%
Queensland	4 (13.3%)	20.5%
South Australia	3 (10%)	6.9%
Tasmania	2 (6.7%)	2.1%
Victoria	7 (23.3%)	25.6%
Western Australia	3 (10%)	10.9%
MAJOR CITY/REGIONAL/REMOTE		
A major city	19 (63.3%)	72.1%
Regional	10 (33.3%)	26%
Remote	1 (3.3%)	1.9%
PARENT/NON-PARENT		
Parent	18 (60%)	63.5%
Non-parent	12 (40%)	36.5%

Remit and key questions

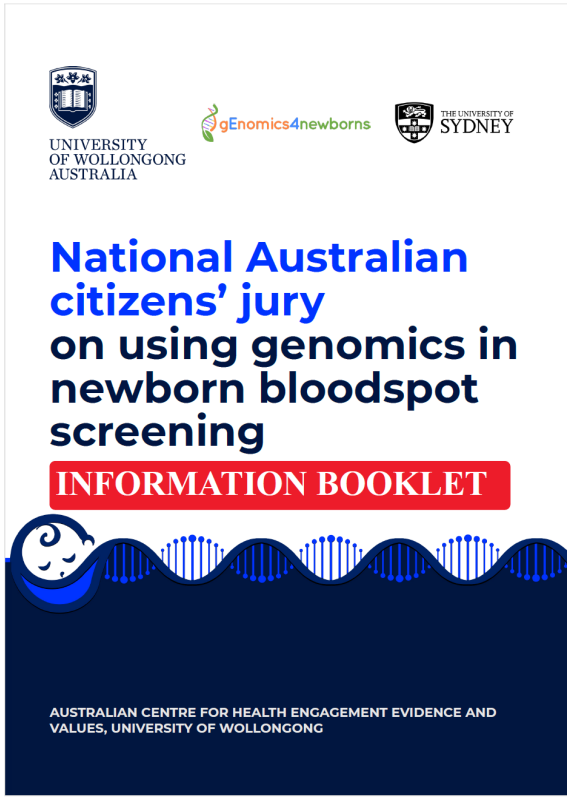
Under what circumstances, if any, should Australia use genomics in the newborn bloodspot screening program, to ensure the program remains trustworthy and effective?

- What types of genetic or genomic testing should be done in newborn bloodspot screening?
- How much of the genetic information from testing should be extracted, and how much should be reported?
- How much of the genetic information should be stored?

Online and face to face engagement

9th March to 30th March inclusive – each juror ≥24.5 hours

- All materials publicly available at: https://uow.info/genomics4newbornsjury
- Printed and online information booklet sent 2 weeks before first meeting
- Online message board system for asynchronous interaction and sharing evidence
- Presentations from 8 content experts + 3 videoed parent stories
- 3 online meetings – learning skills, deliberation, expert Q&A
- 3-day face to face meeting – structured deliberation in small groups and plenary



Stakeholders who addressed and/or observed the jury

The Minister for Health, the Hon Mark Butler MP

Human Genetics Society of Australasia

Industry Genomics Network Alliance (InGeNA)

Australasian Association of Clinical Biochemistry and Laboratory Medicine

Genomics Australia Establishment Branch, Department of Health and Aged Care*

Newborn Bloodspot Screening, Department of Health and Aged Care* - Policy and Implementation Section and Condition Assessment Section

Office of Health Technology Assessment, Technology Assessment and Access Division, Department of Health and Aged Care*

* Now the Department of Health, Disability and Ageing

Main outcome: 11 Recommendations with reasons

- Drafting in small groups and plenary
- Re-drafting based on feedback from the whole group
- Voting – 80% supermajority rule

Background issues

CORE VALUES AND PRIORITIES

- Maintaining **trust** in NBS programs
- Equal **access** to screening, support and treatment for all babies
- Ensuring **benefit** to babies and their families

(All 100% support)

Recommendations

Jurors also provided reasoning – beyond scope of this presentation

1. **Consistent** implementation across Australia (100%)
2. **Benefit to newborns** in first few years of life (100%)
3. **I**ndependent, government-mandated, Commonwealth **regulatory body** to develop frameworks and make decisions (100%)
4. **Public** funding (100%)

Recommendations

5. Consent always kept **distinct** for:

A. Immediate **clinical use**[1]

B. Retention for **future clinical use**[2]

C. Data use for **research** with appropriate oversight

(100% support)

1. *Consent for this option alone includes data destruction after a prescribed period*
2. *If ever implemented (see later recommendations)*

Recommendations

6. Reporting to parents must be in the best interests of the **health and wellbeing of the child and the family:**

- Advise that **negative** screen results will not be communicated
- **Effective** communication (simple, culturally and linguistically appropriate)
- Do **not** report **adult-onset** conditions

(87% support – 26/30)

Recommendations

7. Data governance to the same standard as any other sensitive medical data in Australia, with different data access for **different users** (100%)

8. Wraparound, culturally responsive, multi-disciplinary, adequately-funded **support services** for families experiencing true and false **positives** and for '**patients in waiting**' (100%)

9. Training in emerging technologies for multidisciplinary **health professionals** (100%)

10. High-quality **public education** program before and after birth (87%)

All jurors supported some use of genomics in newborn bloodspot screening, **provided Recommendations 1-10 were met**.

However: there was **one issue** on which they could not reach supermajority.

11. Data extraction and retention

Option 1: Whole genome sequencing where the WGS data is extracted and could be retained. [70% - 21 jurors - supported this option]

Reasons

- Potential future population health benefits from research
- Potential future clinical benefits to the child
- Potential cost effectiveness of using WGS rather than implementing a panel and making later additions
- Efficiency gains from not double handling: a whole genome sequence allows future questions to be asked of already-collected data
- Confidence in Australian data protections
- Increased flexibility of NBS, which may serve people of all ancestries more effectively.

Option 2: While the whole genome may be sequenced, only data that we already understand and can act on would be extracted or collected. [30% - 9 jurors - supported this option]

Reasons

- The significance of genomic data, which represents a whole person
- WGS not necessary in NBS – can be done when child is can consent
- Information uncertain, benefit uncertain
- Extracting and storing more information may reduce trust in NBS
- Not an appropriate mechanism to create a large WGS database, despite potential benefits, given risks of misuse
- If WGS is implemented in NBS, parents may stop participating in NBS to avoid WGS of their child

We [the minority group] are not against whole genome sequencing, however we request only targeted analysis and storage of relevant data to NBS. We [the minority group] acknowledge this will grow and evolve over time as research develops.

Implications

- Trust, equity and benefit central
- Demanding set of conditions to be met before genomics is implemented
- Jurors valued broad health and wellbeing benefits for child and family
- National consistency, national regulator, public funding
- Strong calls for parent support and public education: requires capacity building
- Consent: actionable variants vs future benefit always kept distinct
- Lack of supermajority on extent of data extraction suggests need for caution