Medical Journal of Australia

MJA The Medical Journal of Australia

RESEARCH OPEN ACCESS

Genomic Newborn Screening: Verdict From an Australian Citizens' Jury

Yves Saint James Aquino[1] | Joanne Scarfe[2] | Diana Popic[1] | Lucy Carolan[1] | Chris Degeling[1] | Kathleen Prokopovich[1] | Margaret F. A. Otlowski[3] | Saniya Singh[1] | Belinda Fabrianesi[1] | Kaustuv Bhattacharya[4,5] | Kristi Jones[6,7] | Ainsley J. Newson[7] | Patti Shih[1] | Bruce Bennetts[6,7] | Emma Frost[8] | Zornitza L. Stark[9,10] | Kristen Nowak[11,12] | Louise Healy[13] | Sarah Norris[2] | Stacy M. Carter[1]

[1]Australian Centre for Health Engagement, Evidence and Values, University of Wollongong, Wollongong, New South Wales, Australia | [2]Leeder Centre for Health Policy, Economics and Data, University of Sydney, Sydney, New South Wales, Australia | [3]Centre for Law and Genetics, University of Tasmania, Hobart, Tasmania, Australia | [4]Genetic Metabolic Disorders Service, Sydney Children's Hospitals Network, Westmead, New South Wales, Australia | [5]UNSW Sydney, Sydney, New South Wales, Australia | [6]Sydney Children's Hospitals Network Randwick and Westmead, Westmead, New South Wales, Australia | [7]University of Sydney, Camperdown, New South Wales, Australia | [8]University of Wollongong, Wollongong, New South Wales, Australia | [9]Victorian Clinical Genetics Services, Melbourne, Victoria, Australia | [10]University of Melbourne, Melbourne, Victoria, Australia | [11]Department of Health, Government of Western Australia, Perth, Western Australia, Australia | [12]University of Western Australia, Perth, Western Australia, Australia | [13]Rare Voices Australia, Melbourne, Victoria, Australia

Correspondence: Stacy M. Carter (stacyc@uow.edu.au)

Received: 23 May 2025 | **Revised:** 6 February 2026 | **Accepted:** 17 February 2026

Keywords: genetic testing | health policy | mass screening | public health | qualitative research

ABSTRACT

Objective: To support a nationally representative group of Australians to make informed, reasoned recommendations on the use of genomics in newborn screening programmes.

Design: Hybrid Citizens' Jury method.

Setting, Participants: Thirty Australian adults recruited by random ballot invitation and stratified selection against population-based demographic targets of age, sex, ancestry, highest level of education, location of residence (state/territory, urban/non-urban), experience of disability and parent/non-parent.

Main Outcome Measures: Jury recommendations with reasons.

Results: The jury made 11 recommendations. The jury agreed whole genome sequencing could be used in the programme, but only if conditions were met regarding national consistency, benefit, Australian Government oversight, consent, reporting to parents, data protection, supporting parents and the healthcare system, and parent and public education. All of these conditions were agreed by consensus, except reporting to parents and parent and public education, where there was a supermajority (24/30) in agreement and minority dissent. The jury were split on Recommendation 11: how much genomic data should be extracted and retained. Nine jurors supported whole genome sequencing only if data extraction and retention were limited to interpretable, actionable genetic information; 21 jurors supported a more expansive approach.

Conclusions: To maintain public trust in Australian newborn screening, programmes should take a more conservative approach to data extraction and storage until concerns are addressed and safeguarding conditions implemented. Jurors' key concerns include identifiability of genomic data, risk of data misuse and potential to undermine trust and participation in newborn screening.

JEL Classification: Environment and public health, Medical genetics, Statistics, epidemiology and research design, Ethics and law, Health services administration

Plain Language Summary

The known: Some evidence exists regarding public attitudes on genomics in newborn screening. We lack evidence about informed, reasoned public views.

The new: Over 3.5 weeks, a national citizens' jury, comprising a representative sample of 30 Australians, engaged with information and experts and then developed recommendations. Jurors supported whole genome sequencing in newborn screening if there is national consistency, public funding, regulation, data protections, equitable support services, well-trained professionals, high-quality education and nuanced consent processes; they disagreed only on the acceptable extent of data extraction.

The implications: This study provides concrete public recommendations to inform national health policy on using genomics in newborn bloodspot screening programmes.

1 | Introduction

Rapid advances in genomic sequencing capabilities and precision treatments are driving stakeholder interest in introducing genomic sequencing to newborn screening [1]. Potential benefits include (i) increasing the number of conditions (and genetic variants that indicate the risk of a condition) screened for by newborn screening; (ii) potentially improving the disease course of childhood-onset conditions through earlier diagnosis and treatment; and (iii) potentially ameliorating burdens on affected individuals, families and health systems [1–3].

However, these potential benefits should be balanced against numerous ethical, legal and social implications [4–7]. These include increased quantity and type of information, inequitable access to accurate diagnosis and treatment, risk of distress due to long-term uncertainty for families and increased overall healthcare system costs. In Australia, genomic research remains a culturally sensitive topic for Aboriginal and Torres Strait Islander peoples. Disrespect of cultural and spiritual beliefs and failure to prioritise the interests of some Indigenous communities have resulted in Indigenous people's mistrust of genetic research. Indigenous-led projects are working to rebuild this trust [8, 9].

Historically, the Australian newborn bloodspot screening programme has received high levels of public support, with about 99% participation rate annually [10]. It is important to engage with the public and understand social values when considering the possible integration or use of genomics in the programme.

We convened a national citizens' jury to discuss the potential use of genomics in newborn screening as part of a larger, multimethod project (Section S1). Deliberative democratic methods originate in political science and government—they are systematic approaches to support representative, diverse members of the public to become informed, deliberate together and influence policymaking [11, 12].

2 | Methods

We used the Hybrid Citizens' Jury method [13], which conforms to the Organisation for Economic Co-operation and Development (OECD) good practice principles for deliberative democracy [14]. The jury discussed the question: 'Under what circumstances, if any, should Australia use genomics in the newborn bloodspot screening programme to ensure the programme remains trustworthy and effective?' (Section S2). Deliberative recruitment and sampling methods involve random ballot invitation followed by stratified selection according to demographic criteria. This aims to provide all community members with an equal opportunity to participate and to recruit a diverse small group that matches population demographics.

An Expert Reference Group was formed to guide the study design, including the development of background information for jurors. The group consisted of experts in Indigenous genomics, disability bioethics, patient advocacy, screening programme management, health policymaking, clinical genetics and the genomics industry (Section S3).

2.1 | Juror Recruitment

The independent, not-for-profit, deliberative democracy recruitment agency Sortition Foundation [15] recruited 30 Australian residents. To ensure each resident had an equal invitation chance, invitations were mailed between 1 January and 14 February 2025 to 6300 households randomly selected from the Australia Post database. Each state and territory received the number of invitations proportional to its population size. One adult (aged ≥ 18 years) from each invited household was eligible to participate.

To increase the likelihood of recruiting at least two jurors identifying as having Aboriginal or Torres Strait Islander ancestry, Sortition Foundation sent 630 invitations to select postcodes that have both a high proportion of residents with Aboriginal or Torres Strait Islander ancestry and ready transport routes to Canberra. Separate yarning circles for Aboriginal and Torres Strait Islander peoples were also planned (Section S1).

Participants were excluded if they were elected representatives from any level of government, paid employees of any political party, unable to speak English, experts in genomics or health policymaking, people with relevant advocacy roles (e.g., rare disease advocates) and persons with experience of a rare, genetic condition either personally or among their close friends or family. Lived-experience perspectives were included in the evidence provided to jurors.

Sortition Foundation used an algorithmic system [16] for the stratified random selection of 30 participants by age, sex, ancestry, highest level of education, location of residence (state/territory, urban/non-urban), disability experience and parent/non-parent (Section S4).

Each juror received $1025 as compensation for their participation, plus centrally arranged travel and subsistence for the

face-to-face meeting. Extensive efforts were made to enable participation, including lending digital devices and providing data packages for internet access, Zoom training, assisting with logistics and funding special travel needs.

2.2 | Jury Planning and Procedure

The entire jury process took 22 days (9–30 March 2025): 19 days online and 3 days face-to-face in Canberra (Section S5). We shared resources via the secure VisionsLive platform, with juror interaction via message boards [17]. Synchronous online discussions were undertaken via Zoom. Facilitation was led by SMC (experienced deliberation facilitator), CD (experienced deliberation facilitator), and LC, YSJA, EF, BF, KP and PS (qualitative researchers with deliberation experience). SS, a clinical psychologist, was available in all sessions for participant well-being support.

The process involved six core steps for deliberation: understanding purpose; relationship building; skill development; information inputs, group dialogue and deliberation; group decision-making; and closing [18]. Online resources and activities were aimed at building skills and knowledge on both the process (e.g., ground rules) and the topic in question (e.g., genomics and newborn screening).

Jurors watched video presentations by seven content experts (KB, BB, KN, ZS, KJ, MO and AJN). Peer reviewed by all content experts and members of the reference group, the video presentations covered topics including background information about Australian newborn screening programmes, genetics and genomics, and ethics and law (Section S5). Experts discussed three testing options, namely single gene tests, gene panels (sequencing DNA regions or chromosomes) or whole genome sequencing. One video recounted three stories from parents with differing experience of screening and/or parenting a child with a rare genetic condition. After each video presentation, jurors asked the experts questions. Questions not addressed due to time constraints were sent to the relevant experts and their responses uploaded as text to VisionsLive.

During the 3-day face-to-face meeting, jurors met for about 18.5 h in total. Observers from organisations interested in genomics or newborn screening (Section S3) attended and agreed to a formal observer protocol (Section S6). The opening ceremony included presentations (https://uow.info/nsw-data-jury) by Mark Butler (Minister for Health, Disability and Ageing), Nicole Millis (Rare Voices Australia) and Natasha Heather (Human Genetics Society of Australasia). After activities to establish working relationships and revise key hopes, concerns and questions, jurors had a final opportunity to engage with the experts. On Day 2, jurors discussed and documented their views and values through a combination of plenary and world café-style [19] sessions addressing three key questions:

- What types of 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?

Jurors discussed reasons to use, or not use, single gene tests, gene panels or whole genome sequencing. Jurors then identified other concerns they felt required recommendations, grouped these into six topics and self-selected to working groups (4–8 jurors each) to draft recommendations for each topic.

On Day 3, the jury discussed how much data should be extracted from whole genome sequencing in newborn screening, as there was no consensus. Working groups then revised their recommendations based on written juror feedback from Day 2. A plenary session finalised recommendations through cycles of feedback, re-drafting and voting. Facilitators implemented minor edits (SMC, CD, YSJA) for ease of reading, with edits approved by the whole jury. A recommendation was included if it was supported by a supermajority (≥ 24 jurors). A subgroup of jurors presented the final recommendations to the observers in a closing ceremony.

2.3 | Analysis

Data for analysis included the final jury recommendations, the reasons for the recommendations (recorded in templates) and the reasons recorded during the world café-style and plenary sessions. YSJA and SMC worked with the raw data to summarise and describe jurors' main concerns and reasons and transcribed these into Excel. There were no conflicts to resolve, as the analysis was descriptive.

This article complies with the CJCheck guidelines [20] and the OECD Good Practice Principles [14] (Sections S7 and S8, respectively). Additional data were responses to a paper-based, anonymised, evaluation survey at the end of the face-to-face process (Section S9).

2.4 | Ethics Statement

This project was approved by the University of Wollongong Social Sciences Human Research Ethics Committee (2024/241).

3 | Results

Of the 157 unique eligible respondents (2.5% response rate, within the range expected in sortition processes [16]), 30 were selected against demographic characteristics of the Australian population (Table 1; Section S4), with a good match to targets on all criteria.

All jurors participated online and attended the final 3-day face-to-face deliberations in Canberra. The jury made 11 recommendations (Table 2). The recommendations with goals and reasons are presented in Section S10.

The jury reached consensus (30/30) on eight recommendations, supermajority (≥ 24/30) on Recommendations 6 and 10 and non-supermajority (< 24/30) on Recommendation 11. For Recommendation 6, a supermajority recommended that

TABLE 1 | Demographic criteria and participant characteristics.

Characteristic	Participants	Australian population reference [21–24], %
Total number of participants	30	
Sex [21, 24]		
Women	15 (50%)	45.3
Men	14 (46.7%)	46.7
Non-binary or other	1 (3.3%)	8
Age group (years) [21]		
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 [21]		
Aboriginal and/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 (cannot pick one)	1 (3.3%)	2.7
Other	0	3.9
Disability [21, 22]		
Yes	4 (13.3%)	13.3
No	26 (86.7%)	86.7
Education [21]		
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) [21]		
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 [21]		
Major city	19 (63.3%)	72.1
Regional	10 (33.3%)	26
Remote	1 (3.3%)	1.9
Parent/non-parent [21, 23]		
Parent	18 (60%)	63.5
Non-parent	12 (40%)	36.5

parents must not be informed about adult-onset conditions, and that informing must be 'in the best interests of the health and well-being of the child and their family'. Four dissenting jurors argued that parents should be informed about adult-onset and non-treatable conditions, uncertain information and/or screen-negative results. In Recommendation 10 (education), only four jurors voted 'uncertain' because they wanted genomics in newborn screening to be included in the high-school curriculum. The jury, thus, had a shared commitment to informing and educating parents and the public, but had different views on how to achieve such education.

The jury supported some use of genomics in newborn screening, provided Recommendations 1–10 were met. The only area where the jurors could not reach supermajority was in relation to the amount of data extracted and retained from whole genome sequencing as part of the screening workflow (Recommendation 11). Twenty-one jurors (non-supermajority) supported extraction and retention of all data; 9 jurors supported whole genome sequencing only if data extraction was limited to information that we already understand and can act on. This minority acknowledged that an entire genome may be sequenced initially, but argued that only a select set of genes should be analysed, with uninterpreted or raw data discarded. The jurors who supported more expansive data extraction emphasised the inclusion of more clinical conditions, potential research benefits that could include saving more lives and the utility of the information for a child's future

TABLE 2 | Final jury recommendations.[a]

We, the jury, deliberated at length about the ways genomics could be used in the newborn bloodspot screening programme. We carefully compared options ranging from single-gene tests to sequencing the whole genome and from comprehensive data extraction to targeted data extraction. We contrasted information of immediate benefit to the child, information that may help the child later in life and information use for research.

We agree that the following are important:

- Maintaining trust in the newborn bloodspot screening programme (Support 30/30).
- Equal access to screening, support and treatment in the newborn bloodspot screening programme for all babies in Australia (Support 30/30).
- Ensuring that screening provides benefit to babies and their families (Support 30/30).

We make the following recommendations:

Overarching conditions

Recommendation 1 (Support 30/30): We must ensure that the use of genomics in newborn bloodspot screening is provided in the same way across Australia, so that all Australian babies and families who can benefit have access to the programme.
Recommendation 2 (Support 30/30): Use of genomics in newborn bloodspot screening must benefit the newborn at the time of screening or in the first few years of life.

Government oversight

Recommendation 3 (Support 30/30): We recommend an independent national regulatory body mandated by the Australian Government to develop frameworks and make decisions on an ongoing basis, regarding but not limited to the following:

- To ensure the newborn bloodspot screening programme is uniform across states and territories.
- Access to and storage of data, including guidelines for how data cannot be used.
- Length of time for storage and destruction of data.
- Oversight over the conditions screened, including addition of new conditions where applicable.
- Periodic review of the programme's operation and objectives to ensure it remains trustworthy, effective and fit for purpose.
- Ensuring that access to the programme and supporting healthcare services remains equitable across diverse cultural, linguistic, geographical and socio-economic groups.

Recommendation 4 (Support 30/30): We recommend the newborn bloodspot screening programmes remain publicly funded.

Consent

Recommendation 5 (Support 30/30): Parents must be given the following options to support informed consent. These options should be given before the birth.
A. Consent for participation in newborn bloodspot screening for immediate clinical benefit to the child (this means the data will be destroyed after the prescribed period unless consent is given for points B and C below).
B. Consent for participation in newborn bloodspot screening for future clinical benefit for the child.
C. Consent for use of data for research as governed by ethics and government policy.
Participants must have the right to withdraw consent at any time for B or C and must be made aware of this right. This would include destruction of data from programme databases or research databases as requested by the participant.
Once the child reaches an appropriate age, they should be able to give or withdraw consent.

Reporting to parents

Recommendation 6 (Support 26/30, supermajority): We recommend that the information reported to parents must be in the best interest of the health and well-being of the child and their family. During the consent process, parents must be told that if the child needs no further testing or follow-up, the parents will not hear from the programme again.
Information should be provided in simple terms, in multiple languages and with available cultural services and support.
It should not include information about adult-onset conditions.

Data protection

Recommendation 7 (Support 30/30): We recommend all data collected or generated in newborn bloodspot screening must be protected to the same standard as other sensitive medical data in Australia. The jury recognises existing frameworks to control access to data. The jury emphasises the importance of ensuring that parties seeking access to data are governed clearly under these frameworks.

Supporting parents, healthcare system

(Continues)

TABLE 2 | (Continued)

Recommendation 8 (Support 30/30):

- Parents and children must have access to equitable and appropriate support services, taking into consideration the diverse cultural needs of the whole population. It is essential to ensure these services are comprehensive and adequately funded.
- Health services must provide holistic support, treatments, communication and coordination for continuity of care. This includes addressing positive results, false positives and the needs of patients-in-waiting. To support genomic newborn bloodspot screening, allied health, counselling support and peer-support services for newborns and their families are essential.
- Access to services must be provided in a culturally sensitive manner, such as appropriate language, translation and sensitivity to diverse religious, cultural and gender needs.

Recommendation 9 (Support 30/30): Training for healthcare professionals from relevant disciplines must be kept abreast with emerging technologies in newborn bloodspot screening.

Parent and public education

Recommendation 10 (Support 26/30, supermajority): We recommend that we must have an ongoing antenatal and post-partum education that provides clear and concise information about risks, benefits and outcomes of the genomic newborn bloodspot screening programme. This information should be available in, but not limited to, pamphlets, advertisements and government websites. Government websites must be equitably accessible by all people. Information should be updated regularly.

Data extraction and retention

Recommendation 11: Recommendation about different ways to use genomics in the newborn bloodspot screening programme. We, the jury, contrasted two broad testing and data extraction options, understanding that the whole genome may be sequenced in either workflow in the laboratory.

- **Option 1:** Whole genome sequencing where the whole-genome sequencing data are extracted and could be retained (21/30 jurors, non-supermajority, supported this option).

Jurors raised reasons in favour of sequencing the whole genome and extracting all available data, including:

- The benefits for the whole population if whole genome sequence data are available for future research, including potentially saving more lives and including more conditions. The group took these potential benefits seriously and did not want to limit Australians' access to these benefits.
- The benefits for the child in the long-term, providing more information for the child's future clinical care, more quickly.
- Future-proofing the system and increasing cost-effectiveness by implementing whole genome sequencing sooner, 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.
- A more expansive approach increases the flexibility of the programme and may serve people of all ancestries more effectively.

- **Option 2:** Although the whole genome may be sequenced, only data that we already understand and can act on would be extracted or collected (9/30 jurors supported this option).

Jurors raised concerns about sequencing the whole genome and extracting all available data, including:

- The significance of genomic data, which represents a whole person.
- Concern that whole genome sequencing is not necessary in the newborn bloodspot screening programme and that this may not be the best time to sequence their genome—this could be done when they are old enough to consent.
- Concern about certainty of information and benefit from testing and follow-up.
- Concern that extracting and storing more information may reduce trust in the screening programme.
- Concern that this is not an appropriate mechanism to create a large database of whole genome sequences despite the potential benefits, in light of the risks of misuse.
- Concern that if whole genome sequencing is implemented in newborn bloodspot screening, parents may stop participating in newborn bloodspot screening to avoid whole genome sequencing 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 newborn bloodspot screening. We [the minority group] acknowledge this will grow and evolve over time as research develops.

[a]This table presents the verbatim recommendations on which the jury voted. The goals and reasons for these recommendations are in Section S10. We qualified support for the recommendations as consensus (30/30), supermajority (≥ 24/30) or non-supermajority (< 24/30).

clinical care. The jurors who supported more restrictive data extraction were concerned about uncertainty in interpretation of genomic data, the risk of data misuse, unnecessary data extraction, timing (more expansive sequencing could be done in adulthood) and the risk of revealing too much information about a neonate without their consent. Fundamentally, this group was concerned about compromising trust and participation in newborn screening.

Jurors agreed that genomics in newborn screening must benefit the newborn at the time of screening or in the first few years of life (Recommendation 2) and that information reported to parents must be 'in the best interests of the health and well-being of the child and their family' (Recommendation 6). However, they were divided on collective, long-term or less certain benefits (e.g., from research), which helps to explain the jury's split position on data extraction and retention (Recommendation 11). In particular, the jury disagreed on whether the newborn screening programme was the correct setting to collect genomic data for potential future benefit. Overall, the jury was unified in a child-centred approach to benefit that also recognised the value of other benefits to the child's family, including benefits beyond medical treatment (e.g., community and peer support for parents and caregivers). Jurors acknowledged that these benefits do come with risks and agreed that holistic support must be provided to patients-in-waiting and those who receive false-positive screening results (Recommendation 8).

4 | Discussion

To our knowledge, this study is the first nationally representative deliberative democratic process designed to develop recommendations on the potential use of genomics in newborn screening. Our study suggests that informed Australians support the use of genomics in newborn screening, but only if the following requirements are met: national consistency, public funding, a national regulatory body, strong data protections, equitable and appropriate support services, well-trained professionals, high-quality education for parents before and after a birth, and nuanced consent processes.

Previous studies have investigated public perspectives on the use of genomics for newborn screening through a range of methods, including online surveys [25], interviews [26] and focus group discussions [27–29]. These studies suggest varying public sentiment on this topic. A Canadian online survey suggested that integrating genomics into newborn screening might promote distrust and reduce participation [25], similar to the concerns expressed by the minority position on Recommendation 11, and consistent with this jury's focus on maintaining trust.

In common with our study, Genomics England's public dialogue explored the question 'What are the implications for the NHS (National Health Service) and society of using whole genome sequencing for newborn screening?' [30]. This public dialogue between 133 participants included information inputs and exchange with multiple experts. The main differences between our study and the Genomics England dialogue [30] are that (i) their dialogue was described as a qualitative study, not a deliberative democratic process; (ii) their findings were generated by researchers from recorded discussions (participants did not reach consensus or make recommendations themselves); and (iii) participants in their process spent less time learning and deliberating ($\geq$12 h) compared with jurors in the Australian process ($\geq$24.5 h). The conditional support reported in the Genomics England dialogue [30] is similar to this Australian jury's minority position in Recommendation 11, which is discussed further below. We emphasise that the more permissive majority position on Recommendation 11 was conditional on Recommendations 1–10 being met, which is no small task for Australia given the limits of our current approach to seeking consent for newborn screening, and the absence of a national approach for genomic data governance. As participants in the English dialogue did not have an opportunity to create and vote on recommendations regarding safeguarding conditions, they may have taken a more cautious view.

Our study contributes to the policy environment for the potential use of genomics in newborn screening in Australia and offers a model that could be implemented in other jurisdictions. Since 2018, Australian newborn screening has operated under a National Policy Framework [31], which articulates a set of criteria for adding or removing health conditions from newborn screening. However, implementation is the responsibility of each state and territory, which led to some inconsistency in conditions screened and methods used. Reflecting recent policy initiatives for newborn screening more generally (e.g., a nationally agreed decision-making pathway [32] and moves to improve national consistency and expand conditions screened [33]), the jury unanimously supported a national approach to the use of genomics in newborn screening to ensure consistency across states and territories (Recommendation 1). They also recommended an independent national regulatory body to oversee the use of genomics in newborn screening (Recommendation 3). A number of existing bodies could potentially fulfil this role, including the Newborn Screening Program Management Committee [32], Genomics Australia [34] and the Cancer and Population Screening Committee [35].

In alignment with the National Policy Framework, jurors accepted that the benefits of newborn screening can be described in terms of health and well-being benefits for the child and the child's family (Recommendations 2 and 6). Acknowledging the need to manage the impacts of false-positive screening results and patients-in-waiting, the jury's recommendation for holistic support (Recommendation 8) requires capacity building in state- and territory-based health infrastructure.

Consent provisions were central (Recommendation 5). Despite the disagreement on the proper extent of data extraction and retention (Recommendation 11), all jurors agreed that consent to the use of whole genome sequencing for currently actionable genetic variants should always be kept distinct from consent to extract and/or retain sequencing data for potential future benefit (Recommendation 5).

The lack of consensus on Recommendation 11 reflects a central tension that animated the process: the potential that introducing genomics could erode existing high public trust in the programme. Although all jurors agreed about the benefits of whole genome sequencing, enough were opposed to extracting and retaining all genomic data that the jury could not reach a consensus. This finding suggests the need for caution in pursuing whole genome sequencing in the programme given informed citizens' concerns about the significance of genomic data, the risks of uncertain information in a population screening programme, the risks of misuse of a large genomic dataset and the non-necessity of generating and retaining comprehensive genomic data in the neonatal period.

4.1 | Limitations

Although best practice methods were applied in our recruitment process, selection bias is inevitable. Respondents may have been particularly civic-minded and interested in genomics and/or newborn screening. This concern is reduced by the final group being demographically diverse and reflective of the Australian population by age, sex, ancestry, highest level of education, location of residence (state/territory; urban, regional, rural), experience of disability and parent/non-parent status.

The jury formulated recommendations for the Australian healthcare system, in which newborn screening is free for all newborns and downstream healthcare is provided to citizens with substantial government subsidy. The jury recommendations and reasons, as well as the authors' insights from the study, may not be generalisable to all healthcare contexts.

The study required jurors to engage with highly complex topics. Willingness and ability to contribute to the deliberative process may vary among participants. We mitigated this concern by providing the jury with diverse evidence in multiple modalities, sufficient time to engage with evidence through the online phase and opportunities to ask experts for clarification or additional information. In addition, a distress protocol (Section S11) and a dedicated research team member ensured that jurors who felt overwhelmed or distressed were supported during the jury process.

4.2 | Conclusion

A nationally representative citizens' jury provided recommendations on the use of genomics in the Australian newborn screening programmes. Although all members of the jury acknowledged the benefits of whole genome sequencing in newborn screening, they disagreed on how much genomic data should be extracted and retained. We conclude that this disagreement suggests, at least in the immediate term, that targeted rather than comprehensive extraction and retention of genomic data are more likely to ensure ongoing trust and participation in the programme.

Author Contributions

All authors meet the ICMJE criteria for authorship. Stacy M. Carter, Sarah Norris and Ainsley J. Newson led the funding acquisition; Stacy M. Carter, Sarah Norris, Ainsley J. Newso and Joanne Scarfe led the conceptualisation, with all authors contributing; Yves Saint James Aquino, Stacy M. Carter, Diana Popic, Lucy Carolan and Joanne Scarfe led the project administration; all authors contributed to the methodology and formal analysis; Yves Saint James Aquino and Stacy M. Carter led the formal analysis and writing of original draft, with all authors contributing to the formal analysis and writing of original draft.

Acknowledgements

Robin Banks (University of Tasmania) and Gabriel Watts (University of Sydney) provided valuable input into authors Margaret F.A. Otlowski and Ainsley J. Newson's expert package, and Olivia Ruhen and Belinda Burns (Western Australia Department of Health) provided valuable input into author Kristen Nowak's expert package. Open access publishing facilitated by University of Wollongong, as part of the Wiley - University of Wollongong agreement via the Council of Australasian University Librarians.

Funding

This study is funded through the gEnomics4newborns research project as part of a Medical Research Future Fund (MRFF) grant awarded in 2022 (MRF2015965). The funding source did not have any role in the planning, writing or publication, study design, data collection, analysis or interpretation and reporting or publication.

Disclosure

Not commissioned; externally peer reviewed.

Conflicts of Interest

Ainsley J. Newson is an unpaid member of the NSW Newborn Screening Expert Advisory Group and has acted (through her institution, with no additional salary) as a consultant to the Commonwealth of Australia on various matters of newborn screening ethics and policy. Yves Saint James Aquino, Joanne Scarfe, Diana Popic, Lucy Carolan, Kristen Nowak, Kathleen Prokopovich, Margaret F.A. Otlowski, Saniya Singh, Belinda Fabrianesi, Ainsley J. Newson, Patti Shih, Emma Frost, Sarah Norris, Stacy M. Carter are either investigators on or employed through the Medical Research Future Fund grant (MRF2015965).

Data Availability Statement

The data for this study (including transcripts of deliberations) will not be shared, as we do not have permission from the participants or ethics approval to do so.

References

1. C. Ji, M. A. Farrar, S. Norris, et al., "The Australian Landscape of Newborn Screening in the Genomics Era," *Rare Disease and Orphan Drugs Journal* 2 (2023): 1–19.

2. Y. E. Landau, U. Lichter-Konecki, and H. L. Levy, "Genomics in Newborn Screening," *Journal of Pediatrics* 164 (2014): 14–19.

3. S. White, T. Mossfield, J. Fleming, et al., "Expanding the Australian Newborn Blood Spot Screening Program Using Genomic Sequencing: Do We Want It and Are We Ready?," *European Journal of Human Genetics* 31 (2023): 703–711.

4. B. M. Knoppers, A. E. Bonilha, A.-M. Laberge, A. Ahmed, and A. J. Newson, "Genomic Sequencing in Newborn Screening: Balancing Consent With the Right of the Asymptomatic At-Risk Child To Be Found," *European Journal of Human Genetics* 33 (2025): 182–188.

5. A. J. Newson, "The Promise of Public Health Ethics for Precision Medicine: The Case of Newborn Preventive Genomic Sequencing," *Human Genetics* 141 (2022): 1035–1043.

6. E. L. Baple, R. H. Scott, S. Banka, et al., "Exploring the Benefits, Harms and Costs of Genomic Newborn Screening for Rare Diseases," *Nature Medicine* 30 (2024): 1823–1825.

7. Z. Stark and R. H. Scott, "Genomic Newborn Screening for Rare Diseases," *Nature Reviews. Genetics* 24 (2023): 755–766.

8. N. A. Garrison, M. Hudson, L. L. Ballantyne, et al., "Genomic Research Through an Indigenous Lens: Understanding the Expectations," *Annual Review of Genomics and Human Genetics* 20 (2019): 495–517.

9. G. Garvey and C. M. Bernardes, "Genetic Research in Indigenous Health: Significant Progress, Substantial Challenges," *Medical Journal of Australia* 197 (2012): 383–384, https://www.mja.com.au/journal/2012/197/7/genetic-research-indigenous-health-significant-progress-substantial-challenges-0?check_logged_in=1.

10. Department of Health, Disability and Ageing, "About Newborn Bloodspot Screening," 2025, https://www.health.gov.au/our-work/newborn-bloodspot-screening/about.

11. C. Degeling, S. M. Carter, and L. Rychetnik, "Which Public and Why Deliberate?–A Scoping Review of Public Deliberation in Public Health and Health Policy Research," *Social Science & Medicine* 131 (2015): 114–121.

12. C. Degeling, L. Rychetnik, J. Street, R. Thomas, and S. M. Carter, "Influencing Health Policy Through Public Deliberation: Lessons Learned From Two Decades of Citizens'/Community Juries," *Social Science & Medicine* 179 (2017): 166–171.

13. S. M. Carter, Y. S. J. Aquino, L. Carolan, et al., "How Should Artificial Intelligence Be Used in Australian Health Care? Recommendations From a Citizens' Jury," *Medical Journal of Australia* 220 (2024): 409–416, https://www.mja.com.au/journal/2024/220/8/how-should-artificial-intelligence-be-used-australian-health-care.

14. Organisation for Economic Co-operation and Development, "Good Practice Principles for Deliberative Processes for Public Decision Making," 2021, https://www.oecd.org/content/dam/oecd/en/topics/policy-issue-focus/innovative-citizen-participation/good-practice-principles-for-deliberative-processes-for-public-decision-making.pdf.

15. Sortition Foundation, "Democratic Lottery Services: Bespoke Recruitment for Citizens' Assemblies and Other Deliberative Processes," 2025, https://assets.nationbuilder.com/sortitionfoundation/pages/434/attachments/original/1734447137/SortitionFoundationServices-web.pdf?1734447137.

16. B. Flanigan, P. Gölz, A. Gupta, B. Hennig, and A. D. Procaccia, "Fair Algorithms for Selecting Citizens' Assemblies," *Nature* 596 (2021): 548–552.

17. VisionsLive, "Online Bulletin Boards & Communities," 2025, https://visionslive.com/online-bulletin-boards/.

18. K. White, N. Hunter, and K. Greaves, *Facilitating Deliberation: A Practical Guide* (MosaicLab, 2022), https://www.mosaiclab.com.au/the-big-book.

19. J. Brown and D. Isaacs, *The World Café: Shaping Our Futures Through Conversations That Matter* (Berrett–Koehler, 2005).

20. R. Thomas, R. Sims, C. Degeling, et al., "CJCheck Stage 1: Development and Testing of a Checklist for Reporting Community Juries–Delphi Process and Analysis of Studies Published in 1996–2015," *Health Expectations* 20 (2017): 626–637.

21. Australian Bureau of Statistics, "2021 Census All Persons QuickStats," 2023, https://www.abs.gov.au/census/find-census-data/quickstats/2021/AUS.

22. Australian Bureau of Statistics, "Disability, Ageing and Carers, Australia: Summary of Findings," 2023, https://www.abs.gov.au/statistics/health/disability/disability-ageing-and-carers-australia-summary-findings/latest-release.

23. E. Gray, "What Do We Know About Men's Fertility Levels in Australia?," *People Place* 10 (2002): 1–10.

24. Royal Australian College of General Practitioners, "SG16 Sex, Sexuality, Gender Diversity and Health Contextual Unit," 2023, https://www.racgp.org.au/FSDEDEV/media/documents/Education/Curriculum/sg16-sex-sexuality-gender-diversity-and-health.pdf.

25. Y. Bombard, F. A. Miller, R. Z. Hayeems, et al., "Public Views on Participating in Newborn Screening Using Genome Sequencing," *European Journal of Human Genetics* 22 (2014): 1248–1254.

26. G. T. Timmins, J. Wynn, A. M. Saami, A. Espinal, and W. K. Chung, "Diverse Parental Perspectives of the Social and Educational Needs for Expanding Newborn Screening Through Genomic Sequencing," *Public Health Genomics* 25 (2022): 185–192.

27. F. Lynch, S. Best, C. Gaff, et al., "Australian Public Perspectives on Genomic Newborn Screening: Which Conditions Should Be Included?," *Human Genomics* 18 (2024): 45.

28. M. Parfett, F. Johnson, R. Bennett, and F. Ulph, "Views of Children and Young Adults About Whole Genome Sequencing in Newborn Screening: A Qualitative Study," *European Journal of Human Genetics* 32 (2024): 1159–1165.

29. D. S. Kariyawasam, J. Scarfe, C. Meagher, et al., "Integrating Ethics and Equity With Economics and Effectiveness for Newborn Screening in the Genomic Age: A Qualitative Study Protocol of Stakeholder Perspectives," *PLoS One* 19 (2024): e0299336.

30. H. Van Mil, "Implications of Whole Genome Sequencing for Newborn Screening: A Public Dialogue," 2021, https://www.genomicsengland.co.uk/assets/documents/public-dialogue-wgs-for-nbs-final-report.pdf.

31. Department of Health, Disability and Ageing, "Newborn Bloodspot Screening: National Policy Framework," 2018, https://www.health.gov.au/sites/default/files/documents/2020/10/newborn-bloodspot-screening-national-policy-framework.pdf.

32. Department of Health, Disability and Ageing, "Newborn Bloodspot Screening (NBS) – Our National Decision-Making Pathway Fact Sheet," 2025, https://www.health.gov.au/resources/publications/newborn-bloodspot-screening-nbs-our-national-decision-making-pathway-fact-sheet?language=en.

33. Department of Health, Disability and Ageing, "Expansion of Newborn Bloodspot Screening," 2025, https://www.health.gov.au/our-work/newborn-bloodspot-screening/expansion.

34. Department of Health, Disability and Ageing, "Genomics Australia," 2025, https://www.health.gov.au/our-work/establishing-genomics-australia.

35. Department of Health, Disability and Ageing, "Cancer and Population Screening Committee," 2025, https://www.health.gov.au/committees-and-groups/cancer-and-population-screening-committee.

Supporting Information

Additional supporting information can be found online in the Supporting Information section. **Data S1:** mja270184-sup-0001-supinfo.pdf.