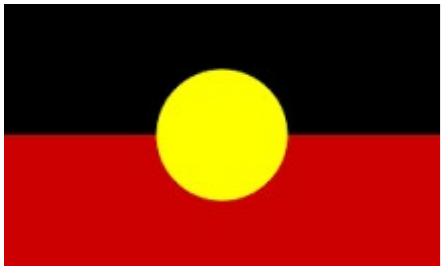
UNSW SYDNEY

Optimising Newborn Screening Consent in Queensland 2024

Summary report from two national workshops



Acknowledgment of Country

The Newborn Bloodspot Screening consent workshops were held on the lands of the Jagera people and the Turrbal people of Meanjin, and on the lands of the Bidjigal people of Eora. We would like to acknowledge these traditional custodians of the land on which we gathered and pay our respects to their elders past and present.

Citation

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EXECUTIVE SUMMARY

Overview

Newborn Bloodspot Screening (NBS) is a critical health intervention that identifies rare genetic diseases in pre-symptomatic children, allowing early identification and treatment. A key feature of the NBS Program is gaining informed consent from parents and so the University of New South Wales conducted two workshops to better understand and optimise the NBS consent process.

Workshop Aims: The primary workshop objectives were to a) Understand current NBS consent practice and challenges, b) Identify gaps and recommendations for improvement, c) Propose feasible short-term solutions.

Participants: A total of 86 stakeholders participated across the two workshops, including clinicians and subject matter experts, government and policy professionals, academic researchers, consumers, and advocacy group representatives.

Key findings

Process gaps

- Lack of clarity and consistency in consent process across jurisdictions, including inadequate presentation of information regarding data storage.
- Lack of formalised consent training, protocols, monitoring, and assessment.

Information gaps

- Inadequate information resources for parents, antenatal patients, and the public.
- Inadequate resources for culturally and linguistically diverse communities.

Key recommendations

The Guthrie Card

- Review and redesign consent statements to include future research and avoid coercive statements, including QR codes to dedicated information sources.
- User testing, finalisation, and distribution.

Information resources

- Develop Health website and hardcopy materials with standardised information.
- Investigate mechanisms for antenatal care period education and resources.

Staff training

- Develop a Standard Operating Procedure, scripts, and monitoring process.
- Develop and deliver training and education for relevant staff.

Introduction and background

Newborn Bloodspot Screening (NBS) is a highly effective health intervention, identifying multiple, rare genetic diseases in pre-symptomatic children to allow early treatment, which can be lifesaving (Waisbren et al. 2003). The Queensland NBS Program currently screens approximately 60,000 newborns every year for approximately 30 genetic diseases.

These workshops were part of a larger body of work investigating the implementation of new NBS screening technologies such as targeted adaptive genetic sequencing. Understanding the downstream implementation impacts of new technologies is needed to ensure that health system systems and processes can adequately adapt in ways that continue to provide safe and quality care, minimising risk.

One crucial component of NBS is parental consent, which was identified as requiring consideration and improvement in the 2023 Queensland NBS Framework (Queensland Health, 2023). The current Queensland consent guidelines and practices also need to be understood to enable the wider goal of developing an implementation framework for targeted adaptive newborn screening technologies.

Two workshops were held to better understand the current NBS consent practices and to identify ways to optimise the potential for incorporating new screening technologies. Due to a lack of nation-wide consensus on consent processes for NBS, key stakeholders were invited from Queensland, as well as all other states and territories. These workshops offered an important opportunity to rigorously gather and evaluate perspectives on enhancing the consent process for NBS through an implementation research lens, with implications for integrating targeted gene sequencing and future technologies to expand the conditions screened for newborns in Queensland and potentially other jurisdictions.

Workshop design and data analysis

Participants: A total of 86 stakeholders participated including 25 in Workshop 1, and 61 in Workshop 2. Key stakeholders involved or impacted by the NBS and paediatric care from Queensland and other states and territories were included, including genomic experts, clinicians and midwives, government agencies, researchers, consumer representatives, and policy and advocacy representatives.

Activities: A virtual workshop was held in March 2024 via Microsoft Teams (Workshop 1), followed by an in-person workshop at Queensland Health in Brisbane in May 2024 (Workshop 2). The format of the workshops used a combination of presentations from selected speakers and small group break-out discussions facilitated by the University of New South Wales (UNSW) School of Population Health Implementation to Impact team.

Ethical considerations: Ethics approval was granted by the UNSW Human Research Ethics Committee (iRECS5688) and participant consent was obtained via an online REDCap form.

Data analysis: Transcripts and notes from the workshops were consolidated and thematically analysed using a framework methodology (Ritchie & Spencer, 1994). Analysis included familiarisation with data, identifying and creating themes for a thematic framework, creating and applying categories (indexing and charting), and then mapping and interpreting the themes into findings and recommendations.

Aims: The workshops aims were to:

- Identify current gaps and opportunities for improving consent within the Queensland NBS program.
- Discuss solutions to gaps in the current processes that could be impactful and feasible to achieve in the next 12 months.
- Explore the requirements needed to facilitate genetic testing from a research perspective within the NBS program.

Workshop One

Objectives

- Understand challenges and concerns associated with the current routine NBS consent process in Queensland.
- Identify opportunities to enhance NBS transparency and information provision.
- Identify potential consent process improvements for Queensland (for both routine screening and research integration).

Participants

Figure 1. Professional titles held by participants in Workshop One (with some individuals holding dual roles)

Key findings

Five core domains were identified through thematic analysis of the data: the Guthrie Card, Information Resources, Training for consent personnel, Data Storage, and Timing of Consent. Insights were mapped across these five core domains as presented below.

Domain 1: The G

- There is no clear direction where to seek or direct parents with further questions.

Domain 3: Training for consent personnel

- No coordinated and structured approach to formal educational sessions on how to obtain consent.
- No monitoring or assessment of consent practices to ensure they are occurring correctly.
- Lack of standard operating procedures/guidelines to refer to.

Domain 4: Data Storage

- Participants were not aware of publ (ons) (.)]T8390 (of)]Tb2/cu aweTJ0 [a)10 (rvd)10 nwabar-0

Current consent process

Additional discussions were held regarding the current consent process of NBS in Queensland, including variations in process and the availability of information in other states and territories, is summarised in **Figure 2**. The National NBS Framework provides guidelines for obtaining consent, but individual states and territories must

Workshop Two

Objectives

- Identi

Key findings and recommendations

Domain 1: The Guthrie Card

Priority Area: *Consent Statement may be misleading*

- The statement “I have been fully informed of the benefits of newborn screening” was perceived as potentially coercive. The need to find a balance between coercion and informed consent was highlighted, where the consent statement could focus on information provision, or risks and advantages of NBS.
- **Recommendation**: A statement is needed about future research which needs to be comprehensive enough to accommodate genomic screening research. This could be a dual process, allowing parents to opt in or out of research but remain in the screening program.

Priority Area:

- **Recommendation**: Queensland Health could also develop flyers that provide parents with information and list new common tests instead of just saying ‘more than 25 tests’; e.g., a physical pamphlet with a QR code to the website.

Priority Area: *No information provided pre-delivery during antenatal care*

- Seeking consent shortly after giving birth could be overwhelming for the parent and cause information overload.
- The responsibility for education sits with the mother’s primary healthcare providers, and linking information to a mother’s pregnancy health record is valuable.
- **Recommendation**: Parents could have a consent primer, where they are made aware of available information and the process before birth (e.g. during antenatal care, or in the ‘pregnancy pathway packet’).
- **Recommendation**: Pregnancy is a time for various types of screening, so NBS could be discussed during this time in a broader approach to antenatal care.

Domain Area 3: Training for Consent Personnel

Priority Area: *Lack of SOP based documents to refer to*

- [illegible]

Research Case Studies – integrating research into the Newborn Bloodspot Screening consent process

In small groups, participants discussed logistical, ethical, and research planning concerns in relation to hypothetical case studies about undertaking research involving the newborn bloodspot screening program. Key points of the group discussions for each research case study are described below.

Hypothetical case study one: key findings and recommendations

- There are logistical concerns for both retrospective and prospective research.
- For retrospective studies, an intermediary group is needed to identify and contact parents of positive cases, raising ethical concerns about identifying parents.
- For prospective studies, obtaining enough positive cases and the timing of interviews, where undertaking a research interview with parents after an emotional period, could bias results.
- Focusing solely on positive cases could introduce bias but might indicate areas for improvement.
- **Recommendation**: Employ a multi-stage consent process with initial consent for screening and further consent for research after obtaining positive results.
- **Recommendation**: Ensure sensitive and ethical research by identifying the health status of the child before embarking on the study to ensure there is psychological support in place, and the training of research interviewers in psychology or genetic counselling.

- There are ethical concerns about the potential burden on parents being presented with genetic testing terminology, including possible negative connotations associated with the term "genetic," which might lead to feelings of parental blame.
- There are potential benefits of early targeted genetic screening for parents, such as cost savings through preventative care, but also potential risks that making the process too complex might cause parents to opt out.
- **Recommendation**: Conduct a formal trial to compare different consent methods or a retrospective study to gather parents' perspectives on screening, especially among those whose children have received positive or negative diagnoses.

2. Information Resources

Objective: Develop content for a single Queensland NBS website and create NBS resources.

Steps:

1. **Draft Website Content:**
    - Review existing NBS websites from other states to collate relevant transferable content and understand how they present to diverse populations.
    - Initiate discussions with stakeholders to gather requirements and expectations.
    - Propose a plan for the Queensland Health website based on best practices.
    - Draft content for the Queensland NBS website, ensuring it is accurate and comprehensive.
2. **Develop NBS Resources:**
    - Use existing information resources (0 (bs0 Tw 24d9 (P.e.39Wy[R)6 (e)1ex)o(o)-1 (p9004
        - Initiate discussions wi(he)L.5 0 (and)10 g NBU4 (nng[I)2 (n,7 (afm4 (fi)6 (dw (i)610 (n

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Acknowledgements

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