UNSW SYDNEY

i2i UNSW Implementation to Impact

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.

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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

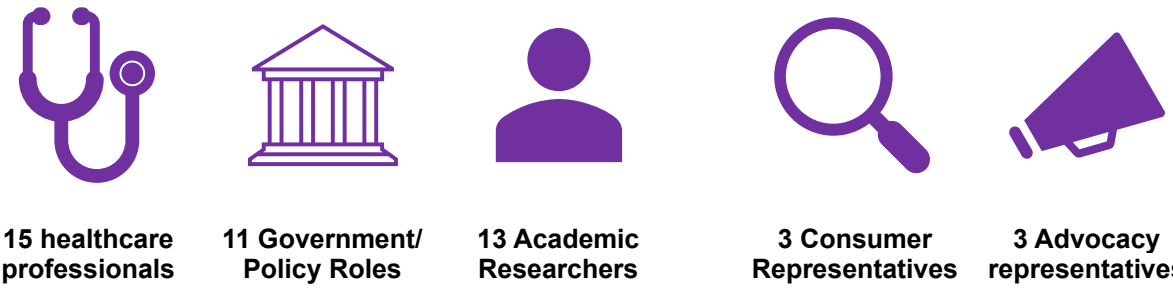

Figure 1. Professional titles held by participants in Workshop One (with some individuals holding dual roles)

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 Guthrie Card

- Consent statement "I have been fully informed of the benefits of newborn screening" may be misleading with no mention of potential risks or second screens, with the term 'fully informed' potentially problematic.
- Does not capture reasons for declining consent.
- No mention of where to turn for further information.
- Does not mention any future research activities to consent to.

Domain 2: Information Resources

- Despite publicly available online NBS information resources for all jurisdictions except the Northern Territory, participants were not aware of these.
- Despite translated information pamphlets available for non-English speakers in some states and territories, participants were not aware of these.
- Limited information provided pre-delivery during antenatal care.
- Information pamphlets vary across sites due to the absence of a standard approach.

- 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 publicly available resources detailing how and where NBS data are stored.
- Participants were unaware of consent data storage beyond Guthrie Cards themselves.
- Participants were not aware of data storage processes (including consent) across jurisdictions, other than on the Guthrie Card itself.
- Participants were not aware of the data storage plans or protocols in place for each jurisdiction.

Domain 5: Timing of Consent

- Lack of standardised NBS information being provided antenatally.
- Need to consider if shortly after birth is the best time to introduce NBS.
- Current consent is sought following childbirth, a time when new mothers may have impaired ability to provide informed consent.

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 collaborate to ensure consistent information, leading to varied consent processes across Australia. For example, based on our review of publicly available resources, there is limited information to confirm whether Queensland, Western Australia, South Australia and Tasmania use a separate consent option for testing, storage of data, or research. Participants confirmed inconsistencies in the consent process and information resources across states and identified additional gaps specific to Queensland.

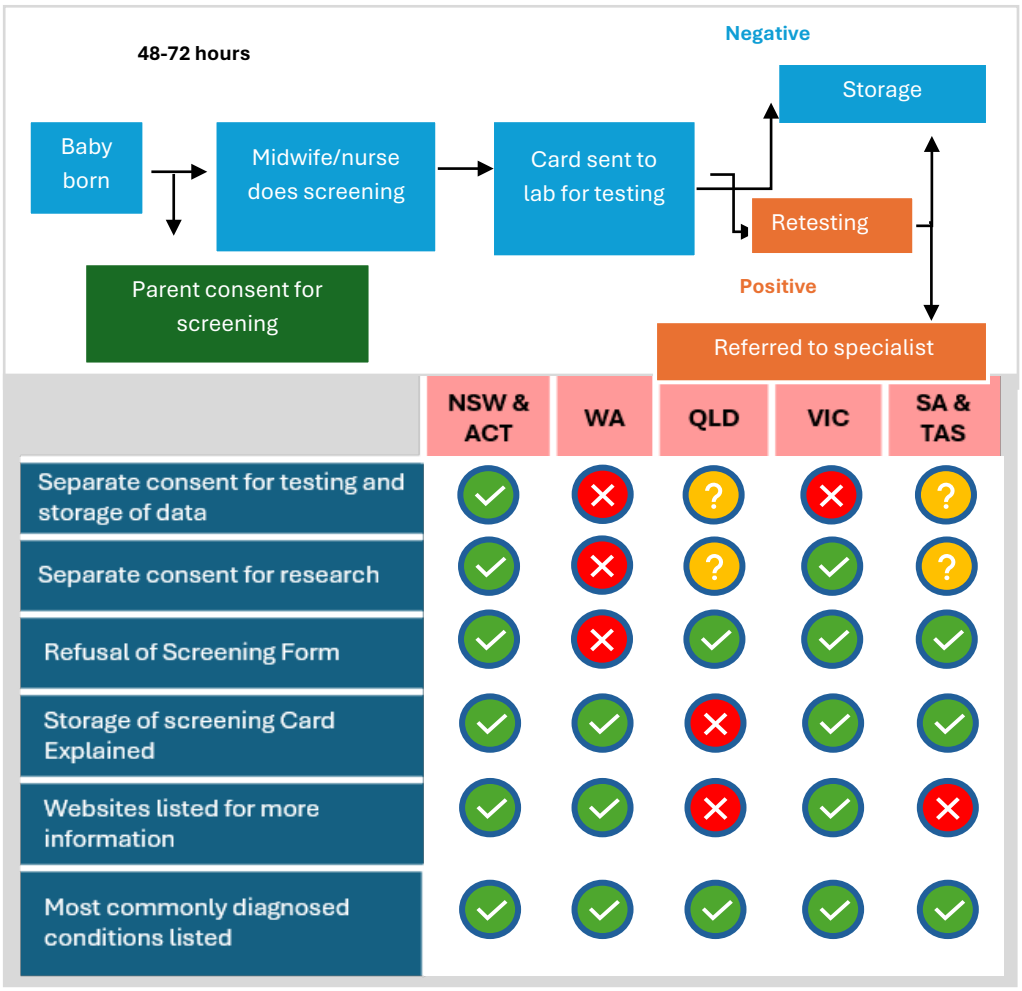

Figure 2. Summary of Newborn Bloodspot Screening participant information and consent processes

Resources for parents

The website for each Hospital and Health Service (HSS) in Queensland was searched for the terms “heel prick” and “bloodspot”, with the results presented in **Table 1**. A state-wide factsheet and website are publicly available, with a handful of HHSs presenting information of their websites in the form of a brief overview. There was inconsistent information presented across the brochures such as consent (verbal versus signature) and length of sample storage. This research was undertaken following the two workshops, and this information was not provided at the time of the two workshops.

Table 1. Summary of publicly available newborn bloodspot screening information for parents

Queensland Hospital and Health Service	Parent factsheet	Online factsheet
State-wide	Brochure	Website
Cairns and Hinterland	-	-
Central Queensland	-	-
Central West	-	-
Children’s Health Service	-	-
Darling Downs	-	-
Gold Coast	Brochure	-
MacKay	-	Website
Metro North	Brochure	Website
Metro South	-	-
North West	-	Website
South West	-	-
Torres and Cape	-	-
Townsville	-	Website
West Moreton	-	-
Wide Bay	-	-

Workshop Two

Objectives

- Identify specific gaps in practice and knowledge that are feasible for short term improvements, using three priority domains (Guthrie card, Information Resources, and Training for consent personnel).
- Obtain recommended solutions across three prioritised domains: The Guthrie Card, Information resources, Training for consent personnel.
- Facilitate collaborative discussions to improve the consent process in Queensland for both routine screening and research integration.

Participants

Figure 3. Professional titles held by participants in Workshop Two (with some individuals holding dual roles)

Topic selection

Workshop 2 focused on developing feasible solutions to address gaps in three priority domains identified in Workshop 1: the Guthrie Card, Information Resources, and Training for consent personnel. Participants were asked to rank specific issues within the domains in terms of impact and feasibility to determine discussion topics. Using a live ranking tool, the topics in **Table 2** were selected. Participants were then broken into smaller groups to discuss ways to address the prioritised issues including what an optimal consent process would look like.

Table 2. Priority topics selected by participants

Domain	Priority issues ranked for discussion
Guthrie Card	Consent statement may be misleading
	No mention of where to turn for further information
Information Resources	No statewide Queensland NBS website or brochure for parent information
	No information provided pre-delivery during antenatal care
Training for consent personnel	No formal educational sessions on how to obtain consent
	Lack of standard operating procedures to refer to
	No monitoring or assessment of if consent practices are occurring successfully

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: *No mention of where to turn for further information*

- Potential accessibility issues were raised such as potentially limited access to technology and internet availability in rural areas.
- **Recommendation**: The Guthrie card could provide a link/QR card to a website. The card itself needs easy-read essential information only, including a brief description of newborn bloodspot screening, potential outcomes and links to additional information.
- **Recommendation**: Queensland Health could host an information website, which would include links to program pathways for families. The website could be easily updated with no need to keep updating the Guthrie card in the case that new conditions get added.

Domain 2: Information Resources

Priority Area: *No statewide Queensland NBS website or brochure for parent information*

- While there are some state-wide NBS resources available for parents in QLD (see **Table 1**), the workshop participants were not aware of these at the time of meeting.
- **Recommendation**: A Queensland Health hosted website could help families and clinicians be aware of where they can find reliable and accessible information that is standardised across providers.
- **Recommendation**: The website should use standard, generic, visual information, and should be tailored and targeted to ensure accessibility for different population groups.

- **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*

- The current extent of 'informed consent' depends on the health professional who draws the blood.
- Formal education sessions, monitoring and assessments, and a regularly updated standard operation procedure (SOP) would be required.
- **Recommendation**: Developing a standardised approach on how much information to give, which could involve developing a script for obtaining consent.
- **Recommendation**: A SOP could be developed at higher levels of Queensland Health but be coordinated by a governing body such as Queensland Clinical Guidelines.

Priority Area: *No formal educational sessions on how to obtain consent & No monitoring or assessment of if consent practices are occurring successfully*

- More psychological support options are needed if families get an abnormal newborn screening.
- **Recommendation**: More staff training is required such as holistic training programs that looks at the whole pipeline of NBS from consent to collection.
- **Recommendation**: Employ a spoken hub model for staff upskilling; with experts from Pathology Queensland that people can go to for information and support, or employing a manager at each hospital that goes through extensive training then conducts training at local sites. These methods would need day to day quality assurance.

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 1:

A nurse is undertaking research for her Masters project. Their research questions are:

1. How do parents of newborns with positive screening results perceive the newborn screening process?
2. What suggestions do parents have for improvement?

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.

Hypothetical Case Study 2:

A pathology lab is undertaking research into using new screening technology for the NBS program. Their research question is:

1. How does a retrospective analysis using new screening technology compare to the current standard screening program in identifying genetic conditions?

Hypothetical case study two: key findings

- There would be a logistical burden on lab staff for reprocessing Guthrie Cards in a retrospective study, noting that additional funding and support staff would be required to handle this workload.
- It appears unethical for research to begin before the necessary infrastructure for genetic screening is established.
- Testing the same conditions with new technology could ethically justify a waiver of consent. New consent procedures would be necessary if introducing new tests, particularly when contacting parents or using different treatment pathways.
- Even when testing for the same conditions, re- consent might be required to allow for the possibility of genetic testing identifying something not detected through biochemical screening.
- The ethical justification of testing for additional conditions is a concern, particularly in cases where rare conditions lack established treatment pathways.
- Establishing effective communication channels with parents is important in the event of positive screening results using the new technology.

Hypothetical Case Study 3:

A university research team is undertaking research into parental attitudes towards newborn screening. Their research question is:

1. What are the outcomes and parental perspectives of implementing targeted genetic sequencing as a new screening technology for newborns?

Hypothetical case study three: key findings and recommendations

- There are different approaches to developing consent for this research including the same consent process as for newborn screening, or a more layered approach (general consent for newborn screening, specific consent for a pilot study, and providing options for consent personnel to access additional information to better support families with questions). Layered consent could be facilitated by using a research protocol and consent script for clear communication.

- 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.

Conclusions & Next Steps

Discussions from the workshops on addressing prioritised gaps in the consent process and conducting research ethically within the NBS program will guide strategic efforts to enhance consent practices. These efforts will focus on involving stakeholders in designing procedures that better support parents and families. The next step is to develop a roadmap for implementing these recommendations in Queensland, ensuring that NBS consent procedures remain adaptable to new technologies, such as genetic and genomic sequencing.

The plans below outline strategies to implement recommendations suggested for each priority area from Workshop 2 to optimise current consent processes. The Implementation to Impact team at UNSW will work with Pathology Queensland and Queensland Health to implement these actions.

1. The Guthrie Card

Objective: Expand the existing consent statement, design a Guthrie Card with a QR code, and ensure interaction with a new dedicated QLD NBS website.

Steps:

1. **Review Informed Consent Statements:**
    - Collect informed consent statements from other jurisdictions.
    - Identify key elements and differences, focusing on standard NBS consent and consent for NBS research activities.
    - Draft revisions to the existing consent statement to include broader aspects including health and digital literacy.
2. **Design and Test Guthrie Card:**
    - Work with QLD Health to create a QR code linking to the Queensland NBS website.
    - Design versions of the Guthrie Card incorporating the QR code.
3. **User Testing:**
    - Conduct user testing with a sample group to gather feedback on the card design and QR code functionality.
    - Make necessary adjustments based on feedback.
4. **Finalisation and Distribution:**
    - Finalise the Guthrie Card design.
    - Work with QLD Health to distribute the cards to relevant healthcare providers (along with training – see part 3).

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 (e.g., Healthy Hearing) as a basis for developing NBS material that is fit for purpose and accessible.
    - Provide a formal pathway for feedback and input on all resources developed to ensure all stakeholder views are considered e.g. consumers, clinicians, scientists
3. **Consider Presentation of Information Antenatally:**
    - Initiate discussions with QLD NBS Unit, midwives, GPs, and nurses regarding presenting NBS information antenatally.
    - Establish the level of information appropriate to share and the extent to which consent for NBS could be collected antenatally
    - Develop a plan to integrate this information into antenatal care programs.

3. Training for Consent Personnel

Objective: Standardise the consent process and develop a training program for NBS.

Steps:

1. **Standardise Consent Approach:**
    - Work with relevant NBS stakeholders to co-design a standardised approach to obtaining consent.
    - Confirm the need for a consent script for health professionals to ensure consistency.
2. **Develop Training Program:**
    - Identify the most effective methods for conducting and disseminating training.
    - Develop training materials and resources for healthcare professionals involved in or impacted by NBS practices.
    - Conduct training sessions and gather feedback to improve the program.

References

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