

# Queensland Newborn Bloodspot Screening Strategic Framework



## **Queensland Newborn Bloodspot Screening Strategic Framework**

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# Contents

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<b>Introduction</b>	<b>4</b>
Purpose, scope and intended audience	4
Associated documents	4
Stakeholder consultation	4
What is newborn bloodspot screening?	5
How NBS is organised	5
Opportunities to strengthen NBS in Queensland	6
The vision for NBS in Queensland	8
Guiding principles	8
<b>Strategic Outcome 1</b>	<b>9</b>
Priority Area 1.1 Program delivery framework	9
<b>Strategic Outcome 2</b>	<b>11</b>
Priority Area 2.1 Access and Participation	11
Priority Area 2.2 Roles and responsibilities of those involved in NBS delivery	13
Priority Area 2.3 Program operations	15
<b>Strategic Outcome 3</b>	<b>17</b>
Priority Area 3.1 Governance	17
Priority Area 3.2 Quality and safety	18
Priority Area 3.3 Risk management	19
Priority Area 3.4 Best practice	20
<b>Strategic Outcome 4</b>	<b>21</b>
Priority Area 4.1 Program monitoring	21
Priority Area 4.2 Data collection and analysis	23
<b>Strategic Outcome 5</b>	<b>24</b>
Priority Area 5.1 Addition or removal of conditions	24
<b>Appendix 1: Queensland Newborn Bloodspot Screening conditions tested</b>	<b>25</b>
<b>Appendix 2: Stakeholder consultation list</b>	<b>26</b>
<b>Appendix 3: National NBS Screening Pathway</b>	<b>27</b>
<b>References</b>	<b>28</b>

# Introduction

## Purpose, scope and intended audience

The Queensland Newborn Bloodspot Screening Strategic Framework (Queensland NBS Framework) outlines the aims of a high-functioning newborn bloodspot screening (NBS) program for Queensland.

It provides strategic direction for Queensland NBS policy and guideline development, governance, quality, monitoring, and evaluation, as well as a guide for all other elements of program delivery. The framework was developed in consideration of the recommendations of the Newborn Bloodspot Screening National Policy Framework (National NBS Framework) and the unique aspects of NBS delivery in Queensland.

The intended audience is all Queensland NBS stakeholders in both public and private sectors.

## Associated documents

The Queensland NBS Framework reflects the guiding principles and components of population screening programs identified by the World Health Organisation (WHO) (Wilson & Jungner, 1968) and reflects the principles for implementing and managing a screening program outlined in the Australian Population-Based Screening Framework (Australian Department of Health, 2018b).

The structure of the Queensland NBS Framework aligns with the National NBS Framework. Each policy area identified in the National NBS Framework has been defined as a strategic outcome in the Queensland context, and national recommendations have been used to inform local objectives.

An associated '**Next Steps**' document will identify the immediate actions required to progress towards the strategic outcomes. The Next Steps will be released following the endorsement of this framework. It is anticipated that a detailed implementation plan will follow.

## Stakeholder consultation

The Queensland NBS Framework was developed in consultation with a wide range of stakeholders from across the state, including women whose babies recently participated in NBS, and a broad range of public and private health care practitioners involved in NBS. Additionally, NBS leads in other Australian States and Territories were consulted. A full list of stakeholders involved in the development of the Queensland NBS Framework can be found in Appendix 2: Stakeholder consultation list.

# What is newborn bloodspot screening?

NBS is a population screening program provided at no direct cost to participants, that aims to detect a number of rare but serious conditions in neonates. Early diagnosis of these conditions allows for early intervention which can improve morbidity and mortality, resulting in better outcomes for babies and their families, and reduced costs to the health system and society as a whole. NBS has been offered in Australia and Queensland since the 1960s, initially testing for only one condition, and now currently screening for over 25 conditions, with an agreed national process for adding nominated conditions to the suite (see Appendix 1: Queensland Newborn Bloodspot Screening conditions tested for details of current conditions tested) as further tests are developed and validated.

Ninety-nine per cent of the 60,000 babies born in Queensland each year participate in NBS (Pathology Queensland, 2022). Similar overall participation rates are reported by all States and Territories. Approximately 1 in 1000 babies screened nationally receive a diagnosis for one of the conditions included in the current NBS suite.

Taking an NBS sample involves pricking a neonate's heel with a lancet and applying blood to a specially designed collection card; the Guthrie Card. The Guthrie Card is then dried before being transported to a laboratory for testing. It is critical that the card is received by the laboratory in a timely manner to allow for testing, notification of results, and the commencement of treatment, before babies become symptomatic. In Queensland, the only laboratory that performs testing for NBS is the Brisbane laboratory of Pathology Queensland (PQ), part of Queensland Health.

## How NBS is organised

NBS delivery is complex. It requires input from numerous arms of the workforce and organisations across the health system. Those involved in NBS delivery include midwives, nurses, phlebotomists, other clinicians, pathologists, scientists, and transport providers. Program governance requires collaboration between clinicians, education providers, health communications experts, program managers, data managers, information technology experts, public health policy makers, and quality experts.

NBS programs in Australia operate independently from each other and are jointly funded by the Australian Government and States and Territories, which are responsible for all aspects of delivery. In most jurisdictions, including Queensland, NBS delivery has historically been led by pathology laboratories.

In 2018, States and Territories came together to create a shared vision for NBS and an agreed way of working. The resultant National NBS Framework established responsibilities at the national and state level and provided guidance for state-level NBS delivery and management. It also established an agreed process for the review of conditions nominated for addition to and/or removal from the NBS panel.

A National NBS Program Management Committee established in 2017 consists of representatives from all States and Territories and has a remit to coordinate and lead NBS in Australia. The Committee provides a forum to discuss and consider program operational and

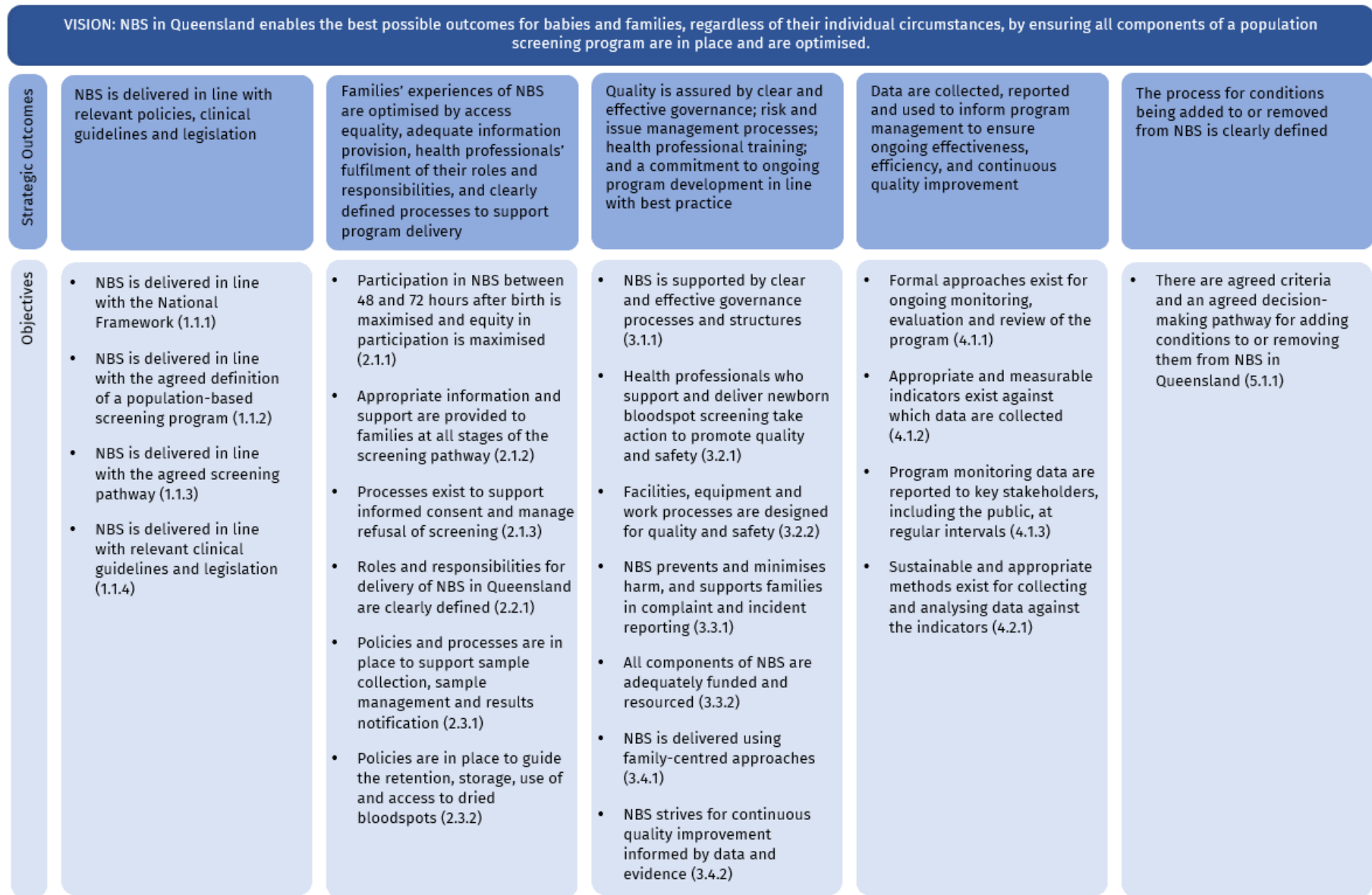
quality issues, share information, and undertake forward planning at a national level (Newborn Bloodspot Screening National Program Management Committee, 2017). In June 2022, the Australian Government announced a commitment of \$38.4 million over 4 years to expand and support standardisation of newborn screening across Australia.

## Opportunities to strengthen NBS in Queensland

The main areas identified to strengthen NBS in Queensland are:

- ensuring samples are collected within the critical period of 48 to 72 hours after birth;
- ensuring transport times are within the recommended limit, particularly in regional and remote areas;
- clarity and agreement about the responsibilities of those involved in NBS delivery;
- introducing a new information system that meets monitoring and clinical needs;
- standardising sample collection techniques, training and competency checks;
- standardising methods for transporting samples to PQ;
- ensuring current and relevant plain language information is available for families, including priority populations;
- improving understanding by families regarding the purpose of NBS;
- ensuring families provide informed consent for NBS screening;
- developing state-wide guidelines, quality standards and measures; and
- securing sustained funding for all components of the screening pathway.

# Figure 1: Queensland NBS Strategic Framework Overview



# The vision for NBS in Queensland

**NBS in Queensland enables the best possible outcomes for babies and families, regardless of their individual circumstances, by ensuring all components of a population screening program are in place and are optimised.**

## Guiding principles

The development of the Queensland NBS Framework was guided by the following key principles which should also underpin its implementation:

- Queensland babies and families are the central focus of NBS: meeting their needs is paramount.
- The benefits of a population-based screening program are maximised, and harms are minimised, when all components are in place and adequately and sustainably funded.
- Inequities are minimised when the unique geography and demography of local contexts are considered in all aspects of screening delivery and management.
- Quality, safety, and optimised consumer experience are achieved when all those who deliver and manage screening fulfil their roles and responsibilities.
- Trust in screening is engendered when developments are undertaken in line with agreed policies and processes as well as technological advances.



# Strategic Outcome 1

## NBS is delivered in line with relevant policies, clinical guidelines and legislation

### Priority Area 1.1 Program delivery framework

#### Objective 1.1.1 NBS is delivered in line with the National NBS Framework

The Queensland NBS Framework is built upon similar principles to those used to guide the development of the National NBS Framework. It places families at the centre of the program and provides guidance for delivering high-quality and safe NBS to protect babies from the effects of serious conditions. It acknowledges the changing environment in which NBS is delivered and proposes that future developments in NBS in Queensland should be focused on conditions and not driven by technology per se.

Whilst informed by the National NBS Framework, the strategic approach outlined in the Queensland NBS Framework was developed with consideration of local geography and demography, as well as the structure of the Queensland health system.

#### Objective 1.1.2 NBS is delivered in line with the agreed definition of a population-based screening program

Population-based screening is defined as:

*A screening test [that] is offered systematically to all individuals in the defined target group within a framework of agreed policy, protocols, quality management, monitoring, evaluation and review. All of these activities must be resourced adequately to ensure benefits are maximised [...]* (Australian Department of Health, 2018b)

The target group for NBS in Queensland is defined as all newborn babies in the state. NBS in the case of neonatal death should be defined in state-wide policies.

The Queensland NBS Framework provides guidance for the development of state-wide policies, processes, governance, and quality mechanisms. Implementation plans arising from the Queensland NBS Framework will detail the actions and indicative funding that may be required to achieve the strategic outcomes of NBS in Queensland and to maintain optimal delivery in the long term. To maximise effectiveness, adequate funding is required for all aspects of screening delivery and management (World Health Organisation, 2020) (see Objective 3.3.2).

#### Objective 1.1.3 NBS is delivered in line with the agreed screening pathway

The screening pathway is the cornerstone of a screening program. If sufficiently detailed, it can be used to plan the personnel, training, information technology, and expenditure requirements of the screening program, as well as diagnostic and treatment capacity and

expenditure (World Health Organisation, 2020). The nationally agreed screening pathway for NBS, which is also applicable in Queensland, may be found in Appendix 3: National NBS Screening Pathway. This details the path that families should follow from the time they are made aware of the NBS program to the time the laboratory confirms with the family's maternity service provider that the sample has been tested (for a negative result) or the family is referred for diagnostic testing (for a positive result). Treatment and management for babies diagnosed with a condition is not part of the screening pathway.

### Objective 1.1.4 NBS is delivered in line with relevant clinical guidelines and legislation

The National NBS Framework recommends that NBS screening pathways in each State or Territory should be delivered in line with evidence-based best practice, in the form of clinical guidelines, and the relevant legislation, to ensure standardised practice and quality, and to provide protection for families and other NBS stakeholders. It notes that clinical guidelines relevant to supporting NBS delivery include those relating to neonatal care, obstetrics and midwifery care, paediatrics, pathology, general practice and information sharing.

In November 2021, the Queensland Clinical Guideline for Newborn Baby Assessment (Routine) was amended to include NBS (Queensland Clinical Guidelines, 2021). Elements from this Guideline and PQ's Newborn Screening Guidelines (Pathology Queensland, 2021) should be incorporated into a state-wide clinical guideline for the NBS program for all NBS stakeholders, including those in the private sector.

The development of a specific clinical guideline for information provision and informed consent for NBS would ensure that this crucial aspect of a population-based screening program is consistently delivered in Queensland. Any guideline that addresses NBS in Queensland should specify how best to address the needs of priority populations, such as families living in regional and remote areas, culturally and linguistically diverse families, and Aboriginal and Torres Strait Islander families.

Any new guidelines or additions to existing guidelines should be developed in line with best practice and informed by the most recent evidence. Updates to guidelines should be explicitly communicated to all NBS stakeholders, including those in the private sector. Any guidelines developed in response to the Queensland Framework should also adhere to relevant legislation regarding, for example, privacy, informed consent, open disclosure, incident management, sample storage, and the ethical conduct of research.

# Strategic Outcome 2

**Families' experiences of NBS are optimised by access equality, adequate information provision, health professionals' fulfilment of their roles and responsibilities, and clearly defined processes to support program delivery**

## Priority Area 2.1 Access and Participation

**Objective 2.1.1 Participation in NBS between 48 and 72 hours after birth is maximised and equity in participation is maximised**

The maximum benefits of NBS will be realised when participation in the critical period of 48 to 72 hours after birth is maximised. Participation in this period ensures that babies can be reliably tested for every condition in the NBS suite, resulting in the greatest reductions in morbidity, mortality, and costs associated with delayed diagnosis and treatment. There is also less potential for families to be harmed by false negative or false positive test results (Australian Department of Health, 2018b).

In 2021, of the 99% of Queensland newborns who participated in NBS, 88% participated between 48 and 72 hours after birth. About 2% of babies who participated had samples collected prior to 48 hours, while about 10% had samples collected after 72 hours (Pathology Queensland, 2022). As is typical of screening participation for priority populations, Queensland babies who participate in NBS outside the critical period are more likely to be socially, culturally or situationally disadvantaged (World Health Organisation, 2020). Priority populations are therefore more likely to be disproportionately impacted by the inability to test for all conditions in the NBS suite, and by false negative and false positive notifications. Consideration must be given to actions aimed at ensuring equity of access to and participation in NBS, regardless of demographics.

**Objective 2.1.2 Appropriate information and support are provided to families at all stages of the screening pathway**

It is essential that screening programs provide quality information to their target groups at all stages of the screening pathway. Information that meets consumers' needs and is provided at the appropriate time can aid decision-making at critical points along the pathway, such as when deciding whether to participate (see Objective 2.1.3), as well as help participants to feel engaged and supported on their screening journey (World Health Organisation, 2020; World Health Organisation, 2017). To ensure consumers' needs are met, resources must be co-designed and consider language, culture, and levels of English literacy and health literacy (World Health Organisation, 2020).

There are numerous NBS consumer information resources available in Queensland, mostly developed by individual maternity service providers. There are also resources intended for state-wide use (developed by PQ and Clinical Excellence Queensland).

Standardised and state-wide plain language information resources should be developed and reviewed periodically to support NBS in Queensland. These should be co-designed with stakeholders, including consumers, and cover topics including the conditions screened; risks and benefits of screening; results notification; and retention, storage, secondary use, release and destruction of personal information and dried bloodspots (see Objective 2.3.2). They should also be made available in different languages and formats, which could include Easy English, text to voice, online or multi-media formats to maximise opportunities for access.

### Objective 2.1.3 Processes exist to support informed consent and manage refusal of screening

In Australia, participation in all population-based screening programs is voluntary (Australian Department of Health, 2018a). The World Health Organisation (2020) advises that screening programs should provide unbiased and easy-to-understand information to the target group to enable them to make an informed decision on whether to participate in screening (see Objective 2.1.2). They should also be provided with the opportunity to ask questions of a knowledgeable health professional to facilitate decision-making.

For NBS, consent is provided by families on behalf of babies. The National NBS Framework asserts that each program must have a policy outlining its consent process and that this policy must adhere to legal requirements (Australian Department of Health, 2018a). It recommends that the policy provides for information to be given to families both prior to and following birth and that a family's understanding of that information is confirmed prior to consent being sought and a sample being taken.

As participation in NBS is voluntary, families have the right to decline screening for their baby. If families have been provided with information that details the risks and benefits of screening and their understanding of this information has been confirmed, their decision to refuse screening must be accepted and respected. The National NBS Framework recommends that state policies cover screening refusal, including advice to families regarding what to do if a baby becomes unwell and the importance of informing relevant healthcare providers that the baby was not screened (Australian Department of Health, 2018a).

In Queensland, consent is currently sought usually from the mother by the sample collector immediately prior to the sample being taken. Consent or refusal are recorded on the Guthrie Card, which the mother may complete and sign, although consent is often provided verbally.

Given some mothers report difficulty in effectively processing information in the immediate post-birth period, consideration could be given to seeking consent for NBS in the antenatal period, at the same time and in the same way that consent is sought for Vitamin K injections and Hepatitis B vaccines. For these two postnatal injections, a succinct one-page information sheet is provided by the midwife at an antenatal visit, with an opportunity to discuss and ask questions. Women then take home the consent form and return it signed at their next antenatal appointment.

## Priority Area 2.2 Roles and responsibilities of those involved in NBS delivery

**Objective 2.2.1** Roles and responsibilities for delivery of NBS in Queensland are clearly defined.

To ensure program quality, it is crucial that those responsible for NBS delivery are familiar with the agreed screening pathway and their role in it, and that it is embedded in practice. State responsibilities for program management are addressed in Objective 3.1.1. This section details the recommended specific responsibilities of those involved in NBS delivery in both private and public sectors in Queensland.

### **Maternity service providers**

Maternity service providers are responsible for enabling effective NBS frontline delivery by:

- nominating an NBS Liaison Officer for each health care setting delivering maternity services (see below)
- providing appropriate education and/or information on NBS to families
- facilitating collection of bloodspot samples for all newborns at 48-72 hours post birth
- developing service-based operational policies and procedures based on state-wide clinical guidelines and standards to support a safe and quality program
- collecting data required to support program monitoring, evaluation and review
- supporting program-wide quality improvement.

### **Sample collectors**

NBS sample collectors include midwives, nurses and phlebotomists. Their responsibilities may include:

- ensuring all families are offered screening for their babies
- ensuring appropriate information is provided to families about NBS, including that participation is optional and how results will be provided
- ensuring that NBS consent or refusal is documented, as well as collecting all other data relevant to sample collection
- ensuring the correct timing of sample collection (between 48 and 72 hours after birth) wherever possible and following policies for collection outside of this period if necessary
- ensuring that samples are collected, dried, and arranged for transport in line with clinical guidelines
- ensuring the Guthrie card is correctly filled out with all appropriate clinical information.

## **Pathology Queensland – Newborn Screening Laboratory (NSL)**

Pathology Queensland NSL is responsible for dried bloodspot testing, sample management, and some elements of NBS program support. This includes:

- providing subject matter expertise to assist in the development of state-wide policies, protocols and guidelines; information resources for consumers; and education /training modules for sample collectors
- supplying and distributing Guthrie cards (and other resources where required) to maternity service providers
- providing support and information as needed for sample collectors, perhaps via the NBS Liaison Officer
- ensuring that NBS samples are processed in a timely manner and that information contained on the Guthrie cards is recorded promptly and accurately
- ensuring that all tests are recorded on the appropriate clinical and tracking systems (e.g, Auslab)
- ensuring the quality of NBS sample testing procedures for all disorders on the NBS screening panel, including an ongoing commitment to quality assurance and quality improvement
- managing NBS samples, including their retention, storage, secondary use, release and destruction
- developing comprehensive policies to support sample retention and making these available to all NBS stakeholders
- communicating in a timely manner with the family's maternity service provider that the sample has been tested
- promptly notifying an appropriate health care provider when the screening result is abnormal so the family can be notified as soon as possible
- issuing timely requests for repeat samples to maternity service providers
- managing a high-quality, secure laboratory information management system, including maintaining a record of all screening events and outcomes of diagnostic testing
- reporting program data for monitoring, evaluation and review purposes
- establishing and maintaining strong links with NBS Liaison Officers.

### **NBS Liaison Officer**

It is proposed that one role at each maternity service should be nominated to assume responsibility for newborn bloodspot screening at that service. This person would have responsibility to oversee the NBS day-to-day operations including:

- ensuring midwives, neonatal nurses and relevant staff are adequately trained and participate in continuing competency activities
- ensuring midwives, neonatal nurses and relevant staff receive ongoing support and opportunities for education
- ensuring timely transport of dried bloodspots to the laboratory
- ensuring the completeness of data sets provided to PQ
- ensuring every baby at the service has a recorded screening result or refusal, including babies who are transferred between hospitals

- assisting with timely follow-up for babies who have abnormal results or who require further samples to be taken
- ensuring diagnostic results have been received by PQ for those babies with an abnormal NBS result

## Priority Area 2.3 Program operations

### Objective 2.3.1 Policies and processes are in place to support sample collection, sample management and results notification

The National NBS Framework recommends the development of policies and protocols to guide sample collection, and timely sample transport, analysis, notification of results, and follow-up of abnormal results with families (Australian Department of Health, 2018a). All these aspects of NBS delivery impact families' experiences of screening, and any delays may impact the timing of the commencement of treatment for babies with an abnormal screening result.

Guidance for NBS delivery in Queensland is contained in PQ's Newborn Screening Guidelines (Pathology Queensland, 2021), and, since November 2021, NBS sample collection has been included in the Queensland Clinical Guideline for Newborn Assessment (Routine) (Queensland Clinical Guidelines, 2021). Whilst these guidelines address many aspects of NBS delivery, the Queensland NBS program could be strengthened by guidelines or work instructions regarding components such as sample transport and result notification/follow-up.

The development of standardised policies and protocols for these fundamental steps of the screening pathway should help to improve equity in families' experiences and outcomes for babies (see Objective 1.1.4). Any guidelines should also address the rationales for the timeframes for sample collection and transport recommended by the National NBS Framework, establish timeframes for abnormal results notification and follow up with families, and highlight the potential impact on babies when recommendations are not met. This may help to reinforce the importance of these aspects of NBS delivery to sample collectors, maternity service providers, and specialist clinicians. Involving the intended users of these policies and protocols in their development will ensure that they are comprehensive and are embedded in practice.

### Objective 2.3.2 Policies are in place to guide the retention, storage, use of and access to dried bloodspots

After being analysed, dried bloodspots are retained by laboratories for a minimum of two years and participants' personal information is retained indefinitely, in accordance with National Pathology Accreditation Advisory Council requirements. This is to allow the opportunity for subsequent investigations, quality assurance, and validation of tests. Dependent on jurisdiction, dried bloodspots may also be used for research purposes, and forensic and coronial investigations. The National NBS Framework recommends that jurisdictions develop policies that outline the potential uses of stored dried bloodspots, the length of time bloodspots are retained, the approvals required to access them, and how participants' confidentiality is maintained (Australian Department of Health, 2018b).



The National NBS Framework also states that bloodspot retention policies should be explicitly communicated to families, including at the time of consent (see Objective 2.1.3), and are easily accessible to stakeholders. A comprehensive state-wide policy should be developed regarding bloodspot retention and reference to this policy and its salient points included in a single, standardised information resource for families. Ideally, the policy and related consumer information resource would be made available online to permit ease of access.



# Strategic Outcome 3

**Quality is assured by clear and effective governance; risk and issue management processes; appropriate professional training; and a commitment to ongoing program development in line with best practice**

## Priority Area 3.1 Governance

**Objective 3.1.1 NBS is supported by clear and effective governance processes and structures**

Governance is essential for population-based screening programs to ensure that they are safe and effective (World Health Organisation, 2020). A clear governance structure provides delineation and demarcation of roles and responsibilities for those involved in service delivery and program management, leading to improved efficiency and reduced risks. When the structure is communicated publicly, it also assists the screening target group to better understand the program, know what to expect of participation, and identify where to seek further information. The roles and responsibilities of those involved in NBS delivery are outlined in Objective 2.2.1. This section addresses state-wide program governance.

As a priority, a 'policy home' for NBS in Queensland should be identified and appropriately resourced to fulfil the state responsibilities of NBS as outlined in the National NBS Framework and detailed below. State governance should also include a multidisciplinary governance committee to provide advice and recommendations regarding strategic and operational issues in Queensland. A formalised governance structure for NBS in Queensland that includes a state-wide NBS committee will align the state with other jurisdictions in Australia.

It is recommended that Queensland program management and coordination responsibilities include:

- developing, reviewing and maintaining state-wide policies
- coordinating the development and maintaining of NBS clinical guidelines and standards in collaboration with clinical stakeholders
- clarifying levels of authority
- coordinating the development and maintenance of NBS information for families in collaboration with a range of relevant stakeholders
- ensuring that NBS stakeholders have access to relevant resources, guidelines and standards
- regular monitoring program performance via data reports
- undertaking regular NBS evaluation, review and identifying emerging issues, and assisting with remediation
- horizon scanning and supporting strategic research
- assessment of new innovations or ideas
- advocating for adequate and ongoing program funding

- convening and maintaining a multidisciplinary governance committee to provide advice and recommendations regarding strategic and operational issues
- ensuring that comprehensive NBS education and training resources are available and accessible for relevant staff throughout Queensland
- representing Queensland on relevant national and state committees.

## Priority Area 3.2 Quality and safety

### Objective 3.2.1 Health professionals who support and deliver newborn bloodspot screening take action to promote quality and safety

The quality of a screening program is largely dependent on the knowledge, skills and experience of those who deliver it. All screening sample collectors must have relevant education and training to undertake their roles before commencing service delivery (World Health Organisation, 2020).

For NBS, the responsibility for ensuring that sample collectors are adequately trained lies primarily with maternity service providers but other stakeholders, such as state-wide program managers and the laboratory, also have roles to play (see Objective 2.2.1). There is a range of NBS sample collector training courses currently in Queensland. The content and scope of available training courses generally differs depending on the sample collector group at which they are aimed. It is acknowledged that different sample collector groups have different educational backgrounds and different role descriptions, however consistent NBS training is important to ensure equitable service delivery.

To optimise the experiences of families and babies, NBS sample collector training and education courses should cover the technical aspects of NBS sample collection and card drying, the information needs of families, and the interpersonal skills required when working with this population subgroup. The rationale for sample collection within the optimal period and transport to the laboratory within recommended timeframes should also be included to impress upon sample collectors the importance of these crucial elements in ensuring program quality.

Given the relative strengths of current training for different sample collector groups, standardised state-wide education and training courses should be developed in collaboration with nurses, midwives and phlebotomists. Input from other stakeholders such as the PQ laboratory would also be beneficial. Development of these courses should be managed by the 'policy home' of NBS in Queensland and they should be made available to all relevant education providers and maternity service providers in the state. To ensure continuous quality, there should also be a schedule of regular competency reviews for sample collectors.

### Objective 3.2.2 Facilities, equipment and work processes are designed for quality and safety

It is important that all facilities, equipment, and work processes involved in NBS are well organised and managed to meet the quality and safety requirements of the program. Facilities and equipment include those at point of sample collection, in transport of the sample, and at

the screening laboratory (Australian Department of Health, 2018a). Effective information management systems that support program delivery are also essential for quality assurance (World Health Organisation, 2020). The appropriate use of facilities and equipment should be defined in work processes to ensure all steps in the screening pathway are completed in a timely fashion.

All existing NBS work processes should be reviewed and standardised state-wide, ensuring that they align with the Queensland and National NBS Frameworks and included in the relevant guidelines and protocols,

NBS information management in Queensland is split between Laboratory Information Systems and standalone databases. A new information technology solution could integrate data from several existing platforms and sources to ensure high quality clinical management and reporting (see Objective 4.2.1) for all stakeholders/partners, especially as the NBS suite continues to expand in Queensland.

## Priority Area 3.3 Risk management

### Objective 3.3.1 NBS prevents and minimises harm, and supports families in complaint and incident reporting

The benefits of any population-based screening program must outweigh the harms (World Health Organisation, 2020). Whilst NBS fulfils the requirements of the Australian Population-Based Screening Framework (Australian Department of Health, 2018a) by delivering substantial benefits at a population level, there is always the potential for harm to occur.

There is a need to develop risk management and incident management plans for NBS in Queensland. The risk management plan should include strategies for minimising the potential of a baby missing NBS screening entirely, as well as reducing the risk of false negatives and false positives by ensuring sampling occurs between 48 and 72 hours after birth. The incident management plan should include guidance for management of complaints, missed cases, serious incidents or harms and should align with local critical incident reporting processes and the Australian Open Disclosure Framework (Australian Commission on Safety and Quality in Health Care, 2013). Both risks and incidents should be recorded and reported at a state-wide level.

There should be a transparent process in place for families should they wish to report a complaint or incident. Such information about reporting could be included in NBS resources and/or a relevant website.

### Objective 3.3.2 All components of NBS are adequately funded and resourced

To ensure that screening programs deliver anticipated benefits, they must be cost-effective (World Health Organisation, 2020). NBS is a cost-effective approach to reducing disease burden at the population level. To ensure its ongoing effectiveness, NBS should be assured of adequate and ongoing funding and resources (such as equipment and staffing) for all aspects of screening delivery and program management.

## Priority Area 3.4 Best practice

### Objective 3.4.1 NBS is delivered using family-centred approaches

NBS is family-centred. The input of families is valued and should inform all aspects of program delivery and management. Queensland mothers were engaged in the development of the Queensland NBS Framework and should continue to be involved in the development of state-wide policies, guidelines, education/training and information resources. Families' cultural, language, and literacy needs are particularly important to consider when developing information resources, as are their stated preferences for the timing of information provision (see Objective 2.1.2 and Objective 2.1.3). It would be beneficial for the state-wide 'policy home' of NBS to host NBS consumer and stakeholder groups that could be consulted on an ad hoc basis. It is also recommended that feedback from families is regularly sought regarding their experiences with NBS, for example via surveys, and used for quality improvement purposes.

### Objective 3.4.2 NBS strives for continuous quality improvement informed by data and evidence

To maintain high quality, a screening program should use both data generated by the program and regular scanning of the evidence base to identify areas for improvement and identify and extend excellence.

As outlined in Objective 4.1.1, the establishment of robust data analysis and reporting against performance measures will allow analysis of NBS performance at several levels. The provision of comprehensive data to maternity services should be used to support process and quality improvement at the local level and it will assist PQ in implementing its existing continuous quality improvement activities. The regular reporting of data to the proposed Queensland NBS committee will allow for opportunities for quality improvement at a state-wide level to be identified. Comparison of jurisdictional data contextualised by program components and experiences will allow underperformance to be identified and learnings to be shared between jurisdictions.

Furthermore, the 'policy home' of NBS in Queensland should have processes in place to identify and assess new ideas and innovative approaches and integrate them into practice. This could be assumed as a function of the state-wide NBS committee (see Objective 3.1.1). These activities will ensure that NBS continues to operate in line with the contemporary evidence base and reflects best practice.

# Strategic Outcome 4

## Data are collected, reported and used to inform program management to ensure ongoing effectiveness, efficiency, and continuous quality improvement

### Priority Area 4.1 Program monitoring

#### Objective 4.1.1 Formal approaches exist for ongoing monitoring, evaluation and review of the program

In order to ensure ongoing quality and safety, screening programs must develop approaches for data collection, reporting and use in program development. Each NBS program should articulate the specific local approaches that will enable the development of indicators, collection of data, reporting to relevant stakeholders, and how any issues identified will be addressed (Australian Department of Health, 2018a).

PQ regularly provides data reports to individual maternity service providers. Beyond this, there are opportunities to strengthen NBS monitoring, reporting, evaluation and review. It is recommended that a Queensland NBS data committee is formed to develop such approaches and to provide ongoing program support. This committee could be responsible for activities such as:

- developing program performance indicators, some of which should be developed in collaboration with other jurisdictions
- identifying the data needs of different NBS stakeholder groups
- determining a regular data reporting schedule, formats and methods
- determining where the NBS dataset will be housed
- undertaking regular analysis and reporting of data to maternity service providers, HHSs, relevant dataset collections (e.g. perinatal dataset) and the proposed state-wide NBS committee
- providing relevant advice to the Queensland representatives on the NBS National Program Management Committee
- assisting with the development of data-informed quality improvement
- undertaking wide-scale program evaluation or targeted review activities as required
- devising standardised tools to collect qualitative feedback from families to allow evaluation of the program from the perspective of the consumer.

### Objective 4.1.2 Appropriate and measurable indicators exist against which data are collected

Indicators are measures of performance that allow both short-term and long-term assessment against aims and objectives. Such assessment is essential for NBS monitoring, evaluation and review. Reporting data against indicators allows for local monitoring, as well as national and international benchmarking.

The National NBS Framework suggests that NBS programs should develop indicators that enable assessment of screening effectiveness. These would include diagnosis rates and the impact of screening on morbidity and mortality for the conditions in the NBS suite. Indicators could also be developed to measure participation, including by population sub-group; timeliness of sampling and testing; unsuitable samples; and false positives. Measures of family satisfaction are also crucial to program monitoring.

A full suite of indicators should be developed and reviewed to enable robust monitoring of program performance on a state-wide basis.

### Objective 4.1.3 Program monitoring data are reported to key stakeholders, including the public, at regular intervals

Regular reporting of data to relevant stakeholders ensures that emerging issues can be addressed and successes built upon. The National NBS Framework advises that consideration should be given to the needs and interests of stakeholders when selecting which data to provide and in which format.

Currently, Queensland NBS data reported are:

- regular reports provided by PQ to maternity service providers for clinical management purposes; and
- the overall participation rate, which is reported as part of the Queensland perinatal data set.

Queensland NBS data should continue to be provided to maternity service providers, in a format that facilitates local quality improvement as well as clinical management. Data reports should also be regularly provided to HHSs, relevant state-wide quality committees, and the proposed state-wide NBS committee in formats that meet their needs. The frequency of these reports should be determined to allow for timely responses to any issues identified (see Priority Area 3.3 and Objective 3.4.2). At a minimum, data should be reported on an annual basis, especially to the proposed state-wide NBS committee, the remit of which would include program monitoring and evaluation. Queensland should also work with the National NBS Program Management Committee to report key data sets at recommended intervals.

In addition, relevant Queensland NBS data should be made available to the public, reported in such a way that the information is easily accessible. This could be achieved by the use of plain language and online reporting via *The Health of Queenslanders: Report of the Chief Health Officer Queensland* or an alternate government or health website.

## Priority Area 4.2 Data collection and analysis

### Objective 4.2.1 Sustainable and appropriate methods exist for collecting and analysing data against the indicators

Due to the complexity of NBS, data to support monitoring, evaluation and review needs to be collected by different areas of the health system. While most data will be collected by the pathology laboratory, there is also a need for data to be collected by maternity service providers, general practitioners, specialist clinicians, nurses, midwives, and phlebotomists. It is therefore necessary for an NBS program to have an information system that supports data collection from multiple sources across the health system and for the responsibilities of those involved in data collection to be clearly defined (Australian Department of Health, 2018a).

The need to determine an information system solution for NBS in Queensland was identified as one of the most urgent recommendations, following the establishment of a robust governance structure. Any solution should be accompanied by policies that outline data collection and storage responsibilities and requirements, including the safeguarding of data.



# Strategic Outcome 5

## The process for conditions being added to or removed from NBS in Queensland is clearly defined

### Priority Area 5.1 Addition or removal of conditions

#### Objective 5.1.1 There are agreed criteria and an agreed decision-making pathway for adding conditions to or removing them from NBS in Queensland

Based on the World Health Organisation's screening principles (World Health Organisation, 2020), the Australian Population-Based Screening Framework outlines guidelines for ensuring that screening is provided only for those conditions where there is sufficient evidence to support screening and the benefits outweigh the harms (Australian Department of Health, 2018b). There must be a demonstrated benefit from early diagnosis and appropriate treatment must exist. It is not sufficient for a condition to meet only one or some of these guidelines. For example, the existence of a test is not in itself indicative of a condition's suitability for screening. Any changes to the evidence base may require removal of conditions or the consideration of the addition of conditions.

In 2018, the National NBS Framework established a single mechanism for assessing conditions for addition to or removal from NBS in Australia (Australian Department of Health, 2018a). Outcomes of assessments are then communicated to States and Territories to aid their decision-making. This is in line with international best practice. A national process for assessment removes duplication and ensures consistency by applying the same criteria to every condition. A robust evidence-based approach at the national level also minimises the extent to which individual NBS programs may be unduly influenced by external pressures.

The National NBS Framework includes a nomination form for adding conditions to or removing them from NBS (Australian Department of Health, 2018a). Since the publication of the National NBS Framework, the process for reviewing nominated conditions has changed, although is equally robust. The nomination of a condition is now followed by an initial review of evidence and consultation with the jurisdictional Newborn Bloodspot Screening Program Management Committee by the Australian Government's Chief Medical Officer (CMO), who will then decide whether a full review of evidence should be undertaken by the Medical Services Advisory Committee (MSAC). At the conclusion of a full review, MSAC will provide advice to the Minister for Health and the CMO. The CMO will brief the Health Chief Executives of the States and Territories on the basis of the MSAC advice. Once a recommendation has been made by the Commonwealth, the decision to include or not include a test in the NBS rests with individual States and Territories, which must consider local implementation requirements, including funding, and determine timings.

The National NBS Program Management Committee, which consists of representatives from every State and Territory, remains committed to supporting a single mechanism for assessing conditions in Australia.



# Appendix 1: Queensland Newborn Bloodspot Screening conditions tested

- congenital hypothyroidism
- cystic fibrosis (CF)
- galactosemia
- metabolic disorders:
  - aminoacidopathies:
    - phenylketonuria (PKU)
    - maple syrup urine disease
    - tyrosinemia (won't detect all cases)
    - homocystinuria
    - citrullinemia (argininosuccinic aciduria)
  - organic acidemias:
    - methylmalonic academia
    - propionic academia
    - isovaleric academia
    - glutaric aciduria type 1
    - cobalamin defects (most)
    - 3-methylcrotonyl coenzyme A carboxylase deficiency
    - beta-ketothiolase deficiency
    - Multiple Carboxylase Deficiency
    - HMG CoA Lyase Deficiency (3-hydroxy-3-methyl CoA Lyase Deficiency)
    - 2-methyl-3-hydroxybuteric aciduria
    - 2 methylbutyryl CoA dehydrogenase deficiency
    - Biotinidase deficiency (not all)
    - Malonyl CoA Decarboxylase Deficiency
  - fatty acid oxidation disorders:
    - medium chain acyl coenzyme A dehydrogenase deficiency
    - long chain 3 hydroxyacyl coenzyme A dehydrogenase deficiency (long chain acyl coenzyme A dehydrogenase deficiency)
    - very long chain acyl coenzyme A dehydrogenase deficiency
    - carnitine palmitoyl transferase I deficiency
    - carnitine palmitoyl transferase II deficiency
    - carnitine acylcarnitine translocase deficiency
    - carnitine transporter deficiency
    - multiple acyl coenzyme A dehydrogenase deficiency (glutaric aciduria Type 2)

*Spinal muscular atrophy and severe combined immunodeficiency disorder will be added by May 2023.*

# Appendix 2: Stakeholder consultation list

Stakeholders consulted in the development of the Queensland Framework included:

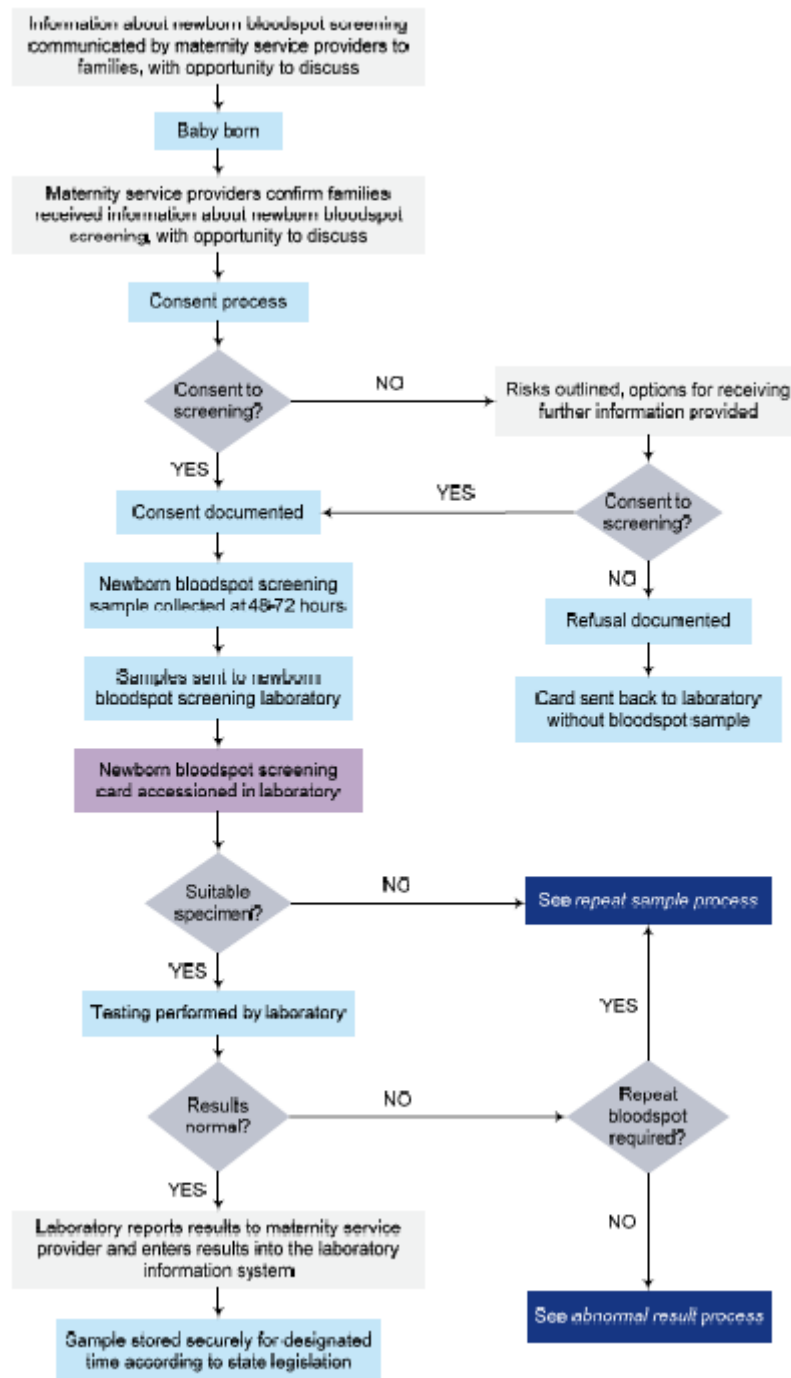
- Queensland mothers who recently gave birth, including those:
  - from metropolitan, regional and remote areas;
  - who gave birth in the public and private health systems;
  - who speak English as an additional language; and
  - who identified as Aboriginal and /or Torres Strait Islander.

These mothers were recruited by an expression of interest opportunity to discuss their experience with Newborn Bloodspot Screening, presented via the Maternity Consumer Network. This Australian network is a non-profit, non-political consumer network that provides advice to the Australian and State Government on policy issues relevant to the views and circumstances of maternity consumers.

- Interest groups for:
  - Aboriginal and Torres Strait Islander parents
  - culturally and linguistically diverse parents
  - parents with a disability or with low literacy and /or low health literacy
- Nurses in the public and private system and from metropolitan and regional areas
- Midwives in the public and private /homebirth system and from metropolitan and regional areas
- Aboriginal and Torres Strait Islander Health workers, nurses and midwives
- Specialist clinicians (paediatrics/metabolic diseases)
- Phlebotomists in the public and private system and from metropolitan and regional areas
- Public and private pathology services
- Information Technology experts
- State-wide maternal and neonatal specialist groups
- Population-based screening experts

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# Appendix 3: National NBS Screening Pathway



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