11 November 2022

NEWBORN BLOODSPOT SCREENING CONSULTATION PAPER

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Part A: Background and information about the programs

Section 1 Overview

1.1 Purpose of this consultation paper

The Australian Government has committed $39.0 million over four years to expand newborn bloodspot screening (NBS) to include more conditions and ensure consistency of programs across Australia.

This consultation process will give the Department of Health and Aged Care (the department) important information about the range of issues that should be considered during implementation of expansion.

This consultation seeks to ensure that that people's views are included, heard and recognised, and used to inform the expansion of NBS.

1.2 Structure, what to expect as you read on

Part A: Background and information about the programs, provides a high-level overview of NBS, including why it is so important, the success of the programs so far, and why steps are being taken to further expand the programs.

Part B: Consultation Questions, seeks your input, providing you with information and discussion prompts to respond to a series of questions. There is also a free text section where you can tell us anything additional.

1.3 Responding to the paper

Responses to the consultation paper will commence from Friday 11 November 2022, closing 11:59PM AEDT Monday 12 December 2022.

Responses are captured through the consultation survey. It has 11 questions with an additional set of questions based on what group of people are responding (consumers or clinicians or other groups), and should take between 15 and 30 minutes to complete. Please contact NBS@health.gov.au if you have any questions or concerns.

Section 2: About newborn bloodspot screening

In this section, you can find out more about NBS, why it is so important, and who does what in the program.

2.1 Why is NBS so important?

Newborn bloodspot screening is an essential preventative public health success story. Through small drops of blood taken from a baby's heel, the programs have saved and improved the lives of thousands of Australian babies since newborn bloodspot screening began in the 1960s.

The value of these programs is a clear testament to the hard work of those who implement and support the programs, including the state and territory governments, scientists, laboratory staff, medical staff, hospital staff and health care providers. It is also acknowledging the families and parents who have helped to shape the programs.

The benefit of the NBS test is that, if a baby is found to have a condition, they can then receive an intervention or management earlier than would have otherwise been possible. This earlier intervention leads to better health outcomes for the baby and the family.

In Australia, more than 99 per cent of babies receive newborn bloodspot screening. This means that more than 300,000 babies are screened each year. Of the babies screened, around one in every thousand has a condition that would otherwise have not been detected within the first few days of life, prior to the onset of clinical presentation. This early intervention allowing for targeted treatment while the baby is asymptomatic.

Newborn blood screening tests identify babies at risk for rare, but serious medical conditions. Click here to watch a video about the life saving impacts of Australia's newborn bloodspot screening programs.

2.2 How does NBS work for families?

Through the programs, an NBS test is offered to all families for their baby within the first few days after the birth. The bloodspot is generally taken in a hospital (or at home or in the community), by a midwife or health care professional. The NBS test is provided at no cost.

Information about NBS is provided to families usually at prenatal classes and again at time of collecting the sample. A midwife or health care professional provides the NBS information to families, where the family can decide whether to undergo screening. If they agree, as is almost always the case, the baby's heel is pricked and bloodspots are collected on a special filter paper card.

The bloodspot card, which includes the collected blood sample, the baby of (mother's name) and other details, is sent to one of five specialist NBS laboratories around Australia. A specific bloodspot test is then used to determine whether the baby is at risk of a range of conditions. More information about the conditions screened, and why, is provided under section 3.2.

If the results of this test suggest the baby is at increased risk of having one of these conditions, immediate notifications are made to health care providers, such as a specialist doctor with expertise in the treatment of these conditions. Through the specialist clinical team, the family is recalled to have more definitive diagnostic testing that will confirm whether the baby does have the condition and determine if urgent care required.

2.3 What is a newborn bloodspot screening *program*?

A newborn bloodspot screening *program* refers to the people, processes, tests and information-flows that are essential to offer screening to families

A program starts at the point a parent or guardian is first given information about screening and includes the steps that follow to support testing in the laboratory.

While the program does not include follow-up testing and care, it does include the processes and pathways to ensure that follow up testing and care occurs in a timely manner, to support the best outcomes for the baby.

2.4 Who does what in the programs?

Newborn bloodspot screening programs in Australia are operated by state and territory governments, through a paediatric specialist hospital and clinical pathology networks.

The bloodspot is taken by a midwife, nurse, phlebotomist (someone who collects blood), or health care professional.

The testing of dried bloodspots is undertaken by five major laboratories, which are located in New South Wales, Queensland, South Australia, Victoria and Western Australia. Dried bloodspots from the other jurisdictions are sent interstate for testing.

Follow up testing and care is managed by health care programs, external to the NBS program. Follow up care can include; medical support, medicines, dietary modifications, surgery and care, and/or regular appointments with a metabolic specialist, endocrinologist, GP or other health care providers.

The Australian Government has a role in assessing current evidence to consider the harms and benefits, and recommend on which conditions to screen.

More information:

The Newborn Bloodspot Screening National Policy Framework provides more detailed background about the programs. The framework provides:

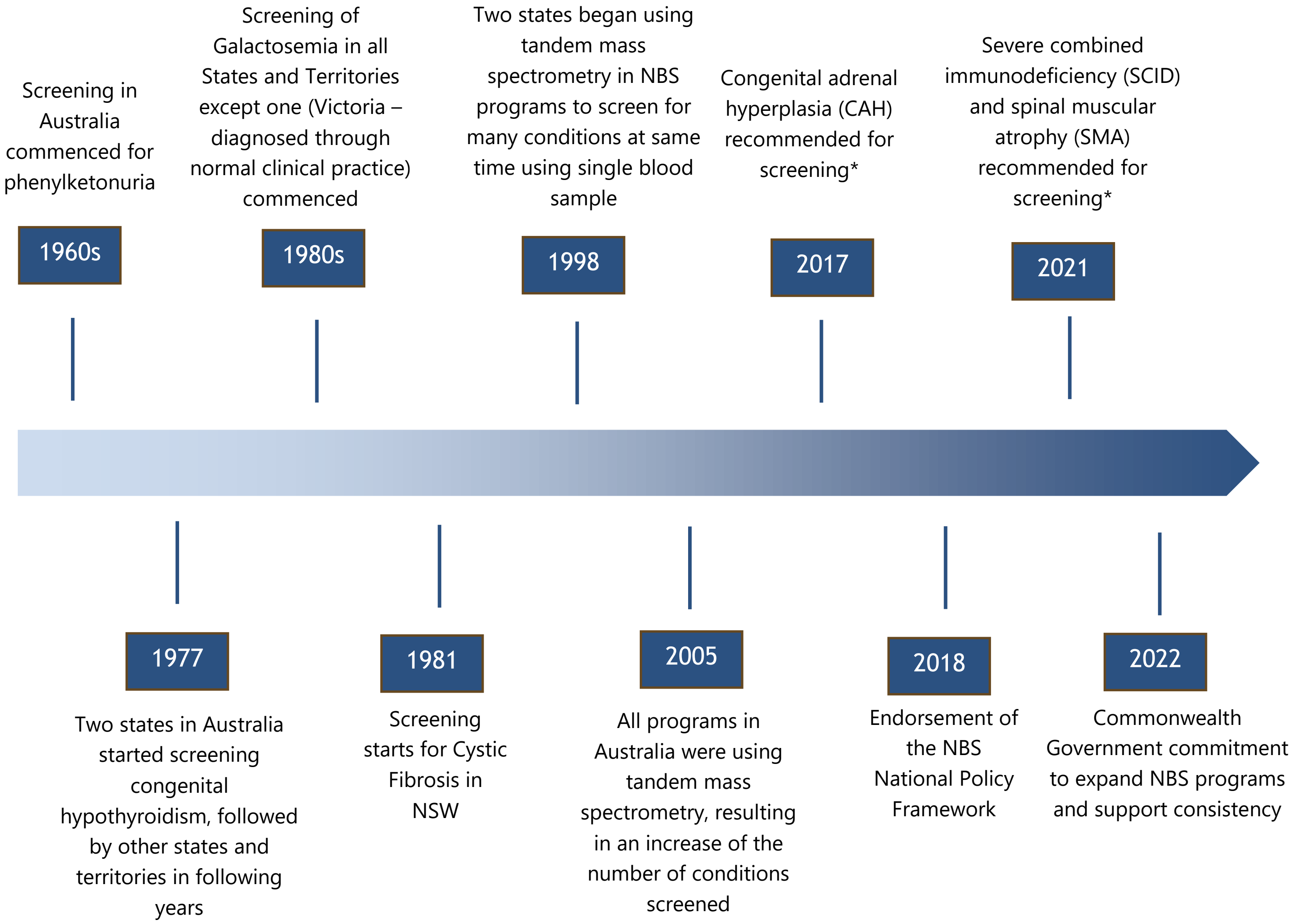
- An overview of the programs
- More information about how the programs are implemented
- Approaches to quality and safety in the programs
- Guidance for monitoring and evaluating the programs, and
- A robust, transparent process for shared national decision-making regarding conditions screened as part of NBS. This process identifies criteria that must considered before a condition is included in NBS programs.
    - These criteria ensure the full benefits and harms of screening are considered. The criteria explore the condition, the way the condition would be tested in the laboratory, what interventions are available and the broader impact on the program and healthcare system.

Section 3: Why expand NBS?

This section reflects on the history of the programs, their success and why steps are being taken to expand them into a unified initiative.

3.1 Where have the programs come from?

Australian newborn bloodspot screening programs have evolved over time, with significant progress in their 60 year history (see figure 1).



1960s
Screening in Australia commenced for phenylketonuria

1977
Two states in Australia started screening congenital hypothyroidism, followed by other states and territories in following years

1980s
Screening of Galactosemia in all States and Territories except one (Victoria – diagnosed through normal clinical practice) commenced

1981
Screening starts for Cystic Fibrosis in NSW

1998
Two states began using tandem mass spectrometry in NBS programs to screen for many conditions at same time using single blood sample

2005
All programs in Australia were using tandem mass spectrometry, resulting in an increase of the number of conditions screened

2017
Congenital adrenal hyperplasia (CAH) recommended for screening*

2018
Endorsement of the NBS National Policy Framework

2021
Severe combined immunodeficiency (SCID) and spinal muscular atrophy (SMA) recommended for screening*

2022
Commonwealth Government commitment to expand NBS programs and support consistency

* Note, following a recommendation to add a condition, states and territories have taken individual steps to add CAH, SCID and SMA, the timing of which has differed across jurisdictions. The Commonwealth and state and territory governments will work together to increase consistency and the conditions screened through the program.

Figure 1: Progress of NBS programs in Australia.

3.2 What we currently screen and why

The conditions screened across the programs in Australia are listed here.

There are thousands of conditions that may affect newborns. While newborn bloodspot screening is highly effective for selected conditions, it cannot identify all conditions. Sometimes, early intervention can be achieved without screening by having effective clinical pathways in place. It is important to note that screening has historically been prioritised for conditions where intervention is urgent, and delays in intervention or intervention after symptoms arise can cause significant preventable harm. This is why careful consideration is needed when deciding which conditions to screen, to make sure that screening continues to deliver health benefits to babies, without causing avoidable harm.

The Australian Government, through the Department of Health and Aged Care, leads a national process to consider the benefits and harms of adding a condition to NBS programs. This process uses an expert committee to assess the evidence, against criteria within the *NBS National Policy Framework* (see Policy Area 4, Part II of the Policy Framework*).*

Following this outcome, the skilled experts in individual states and territories undertake important work to ensure the safe and careful expansion of NBS. This includes within the laboratory, to ensure that the tests and machines are accurate, and clear processes are in place. It also includes ensuring that the doctors, midwives, hospitals and other services all have a clear understanding of their roles to ensure that any baby identified with a new condition receives the right care in the shortest timeframe possible.

3.3 Why we are expanding the programs?

The NBS programs have operated successfully for the past 60 years. Work is now needed to ensure the NBS programs' continued success into the future. This includes work to add more conditions to the programs, and ensure consistency of the conditions screened and the health outcomes delivered through this screening across Australia. It will also be important to monitor screening of these conditions, to ensure they deliver the expected benefits and health improvements to Australian babies.

While new conditions have been added to the programs in recent years, the process can be slow. As a result, this has led to some differences and inequities in the conditions screened in Australia.

The Australian Government has recognised the need for action and funding to support expansion to ensure timely expansion of the programs, while maintaining their quality, and to facilitate consistency across Australia.

3.4 How will the programs be expanded?

The Australian Government is working closely with state and territory governments, scientists and laboratory staff, clinicians, and importantly families and advocates, to support the expansion process for the introduction of new condition into routine practice.

It has also commenced working with First Nations Australians, culturally and linguistically and diverse Australians, and those in rural and remote regions to ensure that their needs inform our way forward.

The expansion will be informed by:

- Looking at the evidence, to understand the benefits and harms of adding conditions
- Understanding what is needed to safely expand the programs, including what is needed within laboratories, counselling and confirmatory diagnostic testing, and within the broader health care setting
- Reflecting on what is needed to support ensure screening is equitable and culturally safe, and ensuring high participation in the program
- Understanding what families need to ensure they are supported when they have a test result requiring additional action, or are diagnosed with one of the conditions tested
- Providing funds to states and territories to support the expansion of their programs.

Importantly, all steps will be underpinned by consultation. So far, the department has met with a wide range of different stakeholders to get early input into the process.

Your response to this paper will build on what we have heard through our initial consultation (outlined below) and provide more information to inform the next steps of the expansion process.

3.5 What we have heard so far

In the past few months, in addition to the states and territories, the department has met with groups who are experts in the field of NBS to formulate ideas and develop consensus in moving forward. This has included consumers and advocacy groups, NBS program scientists and staff, clinicians, First Nations representatives, culturally and linguistically diverse community groups, and other experts, to learn more about the programs.

The consultation has provided overwhelming support for the Australian Government’s commitment to expand NBS. It has also identified strong trust in the programs, and the programs’ ability to save and improve the lives of the newborn population in Australia. Those who engaged in the discussions highlighted their excitement about the ability to further build on these successes to expand the programs.

Part B: Consultation questions

This section of the paper seeks your thoughts on what is important to think about when expanding NBS programs, against five key areas:

1) The aims and objectives of the programs
2) National consistency
3) Assessment of conditions
4) Readiness
5) Measuring success.

Information about you

First, we are keen to learn a little bit about you.

Through this consultation, we are seeking to understand the support that various experts, as well as consumers, need along the screening pathway.

Understanding information about you and your area of interest and/or expertise will help us interpret the responses. It will also help us to use the information you provide to develop appropriate materials and information for people with a similar lived experience, similar cultural background or who are from many of the First Nations groups across Australia, should you wish to share your Aboriginal or Torres Strait Islander Status. You can opt not to provide some information.

Your information will be stored securely.

You can get more information about the way in which the Department will manage your personal information, including our privacy policy, at: https://www.health.gov.au/resources/publications/privacy-policy

It is intended that a report of findings from this consultation will be developed and shared publicly. Do you consent to your responses to these questions being shared?

- Yes
- Yes, if steps are taken to anonymise content
- No.

Please select the category that best describes you:

- Technical expert (e.g., clinician, paediatrician, laboratory worker)
    - Midwife
    - Scientist
    - Pathologist
    - Nurse
    - General Practitioner
    - Community health worker
    - Researcher
    - Medical Specialist (specialty area optional)
    - Public health practitioner
    - Ethicist

  - Academic
  - Laboratory scientist
  - Other – please include details

  If so, where do you work or practice?

- General public
    - Parent
    - Guardian
    - Family member
    - Other
    - If so,
        - Are you of Aboriginal or Torres Strait Islander origin
        - Are you from a culturally or linguistically diverse background
- Consumer peak body or advocacy group
        - If so, which? Please name the group.
- Rare disease group
        - If so, which? Please name the group.
- Professional peak body
    - If so, which? Please name the organisation.
- State or Territory Government Health Department
- Other
    - Please provide further details

1. Where we are going (two questions)

This section describes the commitment to expand the programs, and then reflects on the existing aim and objectives of the programs.

Australia's newborn bloodspot screening (NBS) programs are well regarded and trusted by the Australian communities they also have a strong international reputation. Like all screening programs, NBS programs need regular updating to keep up with new science and evidence.

The Australian Government is committed to increasing the number and consistency of conditions screened across NBS programs. This expansion project aims to:

- ensure every baby born in Australia will be screened for the same conditions and have access to the same high-quality care no matter where they live
- ensure Australian babies are screened for more conditions in a safe and high-quality matter where recommended
- assess new conditions in a more streamlined and efficient way
- monitor emerging interventions, and technologies to test for conditions that could be screened through the programs
- help states and territories implement NBS for additional conditions that have been recommended for inclusion
- ensure that equity of access and outcomes are considered and incorporated into the design of the programs.

Aim and objectives of NBS

We are keen to understand if you have any comments or reflections on the current aim and objectives for the programs

Box 1: Nationally agreed aim and objectives for newborn bloodspot screening[1]

The aim of newborn bloodspot screening is to improve the health of babies by identifying those at risk of developing a serious condition early, generally before symptoms present, thereby enabling earlier intervention.

The objectives are that newborn bloodspot screening programs:

- provide quality, timely and evidence-informed screening to all newborns in Australia
- enable early detection of individuals at risk of conditions screened to reduce the morbidity and mortality associated with the relevant conditions
- support referral to enable early diagnosis and management of conditions identified through screening
- maximise program participation and public trust
- encourage strong partnerships across health systems that are focused on the delivery of high-quality newborn bloodspot screening
- support timely communication of high-quality information to families on all aspects of newborn bloodspot screening

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[1] From the NBS National Policy Framework

- develop and learn through continuous improvement processes that assess short- and long-term program performance.

Questions

We are keen to understand if the existing aim and objectives of the program **(Box 1)** remain current

1.1 Do you have any comments or reflections on the aim of NBS above?

(free text)

1.2 Do you have any comments or reflections on the objectives?

(free text)

2. Achieving national consistency (two questions)

This relates to commitments to achieve national consistency, and then how this consistency is maintained throughout the expansion process.

What have we heard through our consultation so far

- There has been a strong positive response to the intention to achieve national consistency in the conditions screened.
- There was recognition that all governments have agreed to the NBS National Policy Framework, which looks to support a level of national consistency across the program. However, more can be done to support consistency across Australia of conditions screened.
- The Australian Government's commitment responds to this need and in collaboration with the efforts of states and territories, provides a unique opportunity to ensure consistency in the programs.
- NBS programs across Australia should add conditions at the same time, to ensure consistency of screening.

Questions

2.1. Noting the above, please provide any reflections, suggestions or comments on progress to achieve national consistency.

(free text)

2.2. What do you consider to be the key issues when considering national consistency, for example equity of access to screening, follow-up services, consumer information and education?

(free text)

3. Assessment (seven questions)

'Assessment' refers to the process by which a condition is identified and assessed prior to being included in NBS programs. The NBS National Policy Framework includes criteria for assessing conditions (section 6).

What we have heard from consultation so far

- There is ongoing support for the criteria within the NBS – National Policy Framework to be used to assess conditions for inclusion in the program.
- Feedback has been clear that there is a continued need for a detailed assessment process, that considers evidence, and the benefits and harms of screening.
- It was recognised that there may not always be the level of evidence or local data to comprehensively assess the full range of benefits and harms, with a lack of Australian data for specific conditions or evidence relating to cost effectiveness provided as examples.

Consultation so far has suggested several opportunities to enhance, improve and streamline the assessment process. These include:

- Instituting an ongoing review of evidence to identify new conditions for assessment
- Removing the reliance on families and civil society to drive the nomination and assessment process, to ensure that no undue burden is placed on families
- Ensuring a consumer input at relevant stages of the assessment process
- Ensuring that the assessment process is fit for purpose, timely and ensures robust assessment.

Questions

3.1. What do you view as the benefits of NBS?

(free text)

3.2. What do you view as the harms of NBS?

(free text)

3.3. How important do you think it is to consider the:

- ○ Benefits of screening – very important to not important scale
- ○ Harms of screening – very important to not important scale

3.4 The NBS National Policy Framework decision-making criteria are designed to assess the benefits and harms of screening. They focus on:

1) what we know about the condition,
2) how good the screening test is at finding the condition in a newborn,
3) what we know about how to best manage a condition, and
4) how screening can best be implemented in the health system.

The Policy Framework talks about ‘intervention’ rather than ‘treatment’ since some conditions that may be considered for NBS may respond to pharmaceutical as well as other (non-pharmaceutical) interventions.

In your view, how important is it that there is an intervention that can significantly improve the outcomes for the baby? What does an accepted intervention look like?

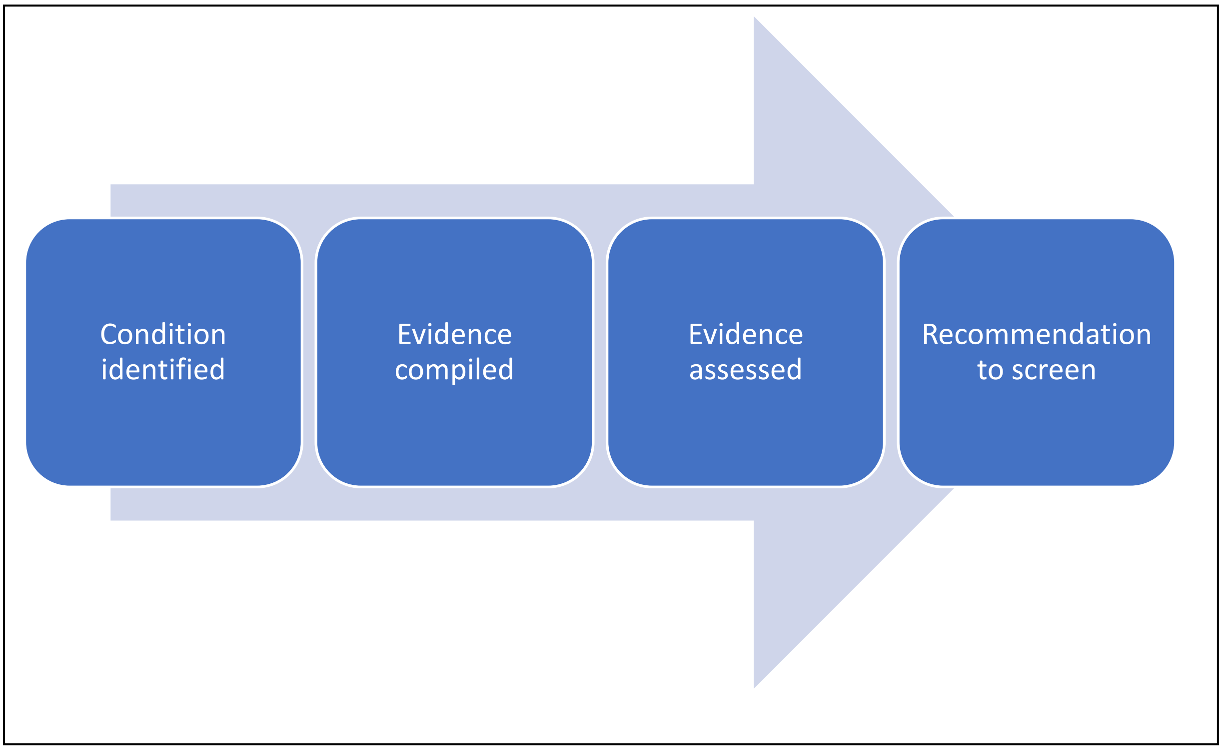
(free text)

3.5. Please describe any other areas that should be a focus when considering the evidence for a new condition. You may wish to reflect on the NBS National Policy Framework criteria, although it is not essential for you to do so.

(free text)

3.6 Any assessment process will provide all stakeholders the opportunity to comment or seek information as conditions are considered for the programs. Noting the below simplified assessment process for illustrative purposes, are there any specific points along the assessment process at which the consumer input should be sought?

(free text)



3.7 In your view, what would be the most appropriate way for you to be involved?

- Consultation forums

- Online approaches
- Surveys
- Consultation paper
- Other – please specify
- Not interested in being involved in consultation

3.8. How should the department best advise on consultation opportunities for conditions going through an assessment process?

- Medical Services Advisory Committee website
- Health website
- Other – please specify

4. Readiness (number of questions [tailored])

'Readiness' relates to what is needed 'on the ground' to ensure that the NBS programs and clinicians and services are best able to provide expanded screening and support families following an abnormal result

What we have heard through consultation so far

- When expanding NBS, it is important not just think about the condition, or the screening test, but also any further testing needed, and the clinical and support services that families need to ensure the best possible outcomes following an abnormal result.
- Given this, expansion requires careful planning and engagement to ensure that all relevant sections of the health system are able to support expanded screening.
- Efforts are needed to ensure that follow up testing, services and care can be provided to the same degree of quality across Australia, regardless of where a person lives. This will ensure equity of outcomes and support across rural and remote regions, and metropolitan regions.
- There is a need for information that supports consumers throughout the screening process, and also information to support them during any further testing or care, focused on the needs of consumers.
- These materials should also be tailored to provide information to First Nations' families and culturally and linguistically diverse families. More work is needed to also understand what is needed to support these families throughout the screening and follow-up pathways.
- There may be the need for materials and information for clinicians to enable them to quickly understand the conditions added and clinical pathways.

The following material and questions seek to explore what is needed to support the expansion, building on what we have heard from the consultation.

Box 4.1. The screening pathway is supported by the following key activities

1. Information is provided to families, with an opportunity to discuss this information
2. All families are offered screening for their baby
3. Dried bloodspots are provided to the laboratory in a timely way
4. Accurate and timely testing of the bloodspot occurs
5. Every baby has a recorded screening result or refusal
6. The family of a baby with an abnormal result is contacted by the appropriate health care provider for diagnostic testing in a timely manner, and/or have further samples or testing
7. Diagnostic testing occurs and for those babies identified as being at increased risk of having a condition, results are provided to the newborn bloodspot screening laboratory for data collection
8. The family is offered care and intervention (specialist care, medications and other clinical support)
9. Families access other supportive services (consumer groups, counselling).

Note, further information on how the programs operate can be found in the NBS National Policy Framework.

Questions

4.1. From your experience, which activities from Box 4.1 do you think will be most impacted by an expansion of the NBS programs, and why?

(free text)

4.2. Thinking about your experience with NBS, or with maternal hospital services more broadly, please select any issues that may impact the likelihood of accessing screening services:

Select all that apply

Coding note: Rotate/Randomise order of responses

- Language or communication barriers
- Cultural or religious beliefs
- Previous experiences with medical staff
- Staff who reflect the consumer’s cultural background
- Staff who respect the consumer’s cultural background
- Clear information about the screening process
- Clear information about the need for screening
- Fear of medical procedures
- Other, please specify
- Not applicable

Follow up question for those who select any of the options above:

4.3. Thinking about the issues you selected above, please highlight how important you think they are for making a decision to access screening services, or discussing them with consumers:

Present options selected

Response frame: Not at all important, slightly important, moderately important, very important, extremely important

Public/Consumer question 1.

4.4 Thinking about the consumer experience of care outside the hospital setting (this might include at a pathology service, with GPs, or specialist services) please select any issues that may impact the likelihood of attending these services if a baby had a positive NBS diagnosis:

Select all that apply

Coding note: Rotate/Randomise order of responses

- Language or communication barriers
- Transport
- Location of services
- Cultural or religious beliefs
- Previous experiences with medical staff
- Staff who reflect the consumer’s cultural background
- Staff who respect the consumer’s cultural background
- Clear information about the screening process
- Clear information about the need for screening
- Fear of medical procedures
- Privacy
- Knowing where to go for screening
- Other, please specify
- N/A

Follow up question for those who select any of the options above:

4.5 Thinking about the issues you selected above, please highlight how important you think they are for making a decision to accessing follow up:

Present options selected

Response frame: Not at all important, slightly important, moderately important, very important, extremely important

Public/Consumer question 2.

4.6 What else would you like to tell us about what is needed to make NBS and follow-up testing care accessible, and culturally safe?

(free text)

The following questions help us to understand where program information is currently accessed and what further information is needed to support families and clinicians for the expanded programs.

4.7 Where do you currently access information about the NBS program? Select all that apply

- Department of Health and Aged Care website
- State based websites
- Local GP
- Health Care Provider (website, fact sheets, pamphlets, in person advice)
- Facebook or social media
- Word of mouth
- Google/search engine
- Friends or family
- Other health professional
- Other please specify
- N/A

Public/Consumer question 3.
4.8 What national information is needed to support consumers, such as parents, families and carers, along the NBS pathway?

- Printed materials
- Information modules
- One stop shop for information
- National phone line
- Publicly available clinical care guidelines or pathways
- Other
- N/A

Technical expert question 1.
4.9 What could be done **nationally** to support clinicians to help provide informed and timely support to families following NBS?

- Modules
- One stop shop for information
- National phone line
- Clinical care pathways
- Clinical guidelines
- Other
- N/A

Technical expert question 2.
4.10 What could be done at a **state or territory level** to support clinicians to help provide informed and timely support to families following NBS?

- Modules
- One stop shop for information

- National phone line
- Clinical care pathways
- Clinical guidelines
- Other
- N/A

Technical expert question 3.
4.11 What else can be done to support clinicians to deliver safe and effective care at the following levels

- National

(free text)

- State

(free text)

- Local/clinical level

(free text)

5. What does success look like? (one question)

5.1 In your view, once the programs are successfully expanded, what do you consider will be the three most important signs of success and how should they be measured?

- Measures
- Measures
- Measures

6. Next Steps

Thank you for responding to this consultation paper. Stakeholder input is and will continue to be essential to expanding NBS programs.

The information collected will be analysed and inform the next steps for the national expansion project.