Response

Expansion into Newborn Screening

From the National Newborn Bloodspot Screening Policy Framework
Target Questions for response

Welcome to the Newborn Bloodspot Screening Consultation

This consultation 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.

1. Where are we going?

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

Nationally agreed aim and objectives for newborn bloodspot screening^
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 blood spot screening
• develop and learn through continuous improvement processes that assess short- and long-term program performance.
^*From the NBS National Policy Framework*

We are keen to understand if the existing aim and objectives of the program shown above remain current.

Question 1.1

1.1 Please provide any comments or reflections on the aim of NBS above.

Australian Genomics is working towards the ethical, appropriate, and evidence-based application of clinical genomics into health practice in Australia. This includes supporting research into the potential use of genomics in newborn bloodspot screening (NBS).

Australian Genomics supports the current aim of NBS, 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.

As we know more about the outcomes of the current genomics research projects and consider the outcomes with an ethical lens informed by societal acceptability, there may be the potential need to revisit the aim of the NBS.

One of the GenSCAN genomics research projects is exploring the concept of data captured at birth through NBS being available for the lifespan of the baby from newborn to beyond. In addition, some conditions identify parents at risk or treatment that will involve future family planning. Whilst the current aim is a strong overarching statement, there is the potential to expand the target audience through the aim to include newborns and their families.

Examples of this are:

- Familial Hypercholesteremia where the initial risk is for the parents of the newborn, and subsequently the newborn in time.
- Duchene Muscular Dystrophy (DMD) where there will be benefit to the newborn and also benefit to the family in terms of reproductive decision making for the future.
- A role for pharmacogenomics also needs to be considered.

Questions 1.2

1.2 Please provide any comments or reflections on the objectives.

Australian Genomics echo the values of nationally consistent approaches that are informed by international best practice, whilst addressing the needs of the Australian public and the health sector.

Whilst the current objectives of the National Newborn Bloodspot Screening Policy Framework are valid, it is critical that a consistent national approach to the conditions which are screened is undertaken.

We have seen with the recent pilot and implementation of SMA into the NSW Newborn Screening Program how critical the bloodspot programs are for early detection of disease and appropriate intervention. Through current and future research new conditions such as SMA will be identified, and equally new therapies and interventions will be become available. Implementation considerations for the expansion project 3 and 4 (3. Assess new conditions in a more streamlined and efficient way, monitor emerging interventions; and 4. Technologies to test for conditions that could be screened through the programs) will need to be considered in a rapid and agile manner to support the changing landscape.

Lastly, point 5 of the expansion project implementation considerations (providing funds to states and territories to support the expansion of their programs) is critical in ensuring a consistent and equitable national approach, however, does imply resourcing requirements.

2. Achieving National Consistency

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.

Question 2.1

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

Australian Genomics coordinates, conducts and enables genomic research through a national network of more than 100 partners and collaborators, including hospitals, universities, research institutes and centres, sequencing laboratories and community organisations across all states and territories. We acknowledge the challenges that this coordination role can bring.

We also acknowledge the work that had gone into the Nation Newborn Bloodspot Screening Policy Framework to facilitate national consistency through federal and state and territory partnerships.

Following the Genomic Health Futures Mission (GHFM) investment into five genomic research projects for newborn bloodspot screening, Australian Genomics formed the Genomic Screening Consortium for Australian Newborns (GenSCAN), which includes the lead investigators of each of the five projects. GenSCAN was developed for the purpose of enabling improved efficiency and impact of the MRFF GHFM investment through complementary and collaborative research, as well as a cohesive national approach to the exploration of genomics into newborn screening. Through GenSCAN, the GHFM funded projects are also exploring the opportunities to expand nationally the existing gene lists current screened.

The GenSCAN Steering Committee meet monthly to ensure a nationally consistent approach into the exploration of genomics in NBS. Under the Steering Committee there are five working groups collectively exploring:

- Technical Platforms
- Bioinformatics and data
- Clinical matters and conditions to be screened

- Ethics, legal and social issues
- Health policy and economic feasibility.

GenSCAN endeavour to present as one voice when engaging with stakeholders so as not to lead to consumer fatigue or to be seen as fragmented. The five project leads of GenSCAN recently presented at HGSA 2022 in a panel session with representatives from the Commonwealth Department of Health and community advocacy group Rare Voices Australia further emphasising the collaborative approach of GenSCAN in the aim of achieving national consistency.

Australian Genomics supported research has a strong emphasis on bioethics as well as exploring questions of implementation in a national setting.

It is worth noting the existing newborn screening laboratories have developed systems based on state funded infrastructure. Whilst there has been the objective by Australian Society of Inborn Metabolism to govern and harmonise systems, this can only be achieved through agreements by the Health Departments of each state and territory. Several states still do not have formal governance structures to coordinate NBS and the governance systems need to be agreed upon nationally engaging state health departments.

Furthermore, the National Newborn Bloodspot Screening Policy Framework itself implies on page 18 that “Ideally, screening programs should be managed nationally and implemented the same way across the country”.

A key factor for the programs moving forward will be data; data security, data access and the ability to aggregate data nationally.

A number of Australian Genomics projects are exploring national consistency in genomic health practices across Australia, including consistency between clinical and laboratory services, consistent approaches to consent for genomic testing, how data is captured and consistent terminology.

Question 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?

Key considerations for achieving national consistency include:

- National consistency in the conditions screened
- Equity of access regardless of location or cultural background to all stages of the screening journey
- Consumer voice and meeting the needs of the Australian population

    - Federated screening system operated by States and Territories
    - Data sovereignty and access
    - Data storage and the system/solution for storage

    - Data storage policies and consistency nationally (including timeframe for storage)

    - Accessibility versus feasibility and consistent economic models i.e. testing prices vary state by state
    - Contractual agreements and MoUs
        - some states undertake testing for other states and territories based on a tender system
        - some states have MoUs with legal entities such as NSW Police (https://www.health.nsw.gov.au/legislation/Documents/mou.pdf)

- Consistent and standardised models of testing and reporting
- Standardising national engagement with industry and setting an agreed and nationally consistent pricing structure for testing materials and requirements. Within the policy framework, section 3.5 regarding funding for the programs is the briefest section.
- Standardising modes and method of test transport from testing centres to laboratory services.
- Standardising bloodspot cards across all programs
- Nationally consistent family information about the programs and consenting and the way consent is captured (paper based versus online).

In addition to the above considerations, Australian Genomics - through GenSCAN are looking to explore and gain a greater understanding of the specific questions raised in section 4.2 to 4.7 of the National Newborn Bloodspot Screening Policy Framework.

3. Assessment

'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 consumer input at relevant stages of the assessment
- Ensuring that the assessment process is fit for purpose, timely and ensures robust assessment.

Question 3.1

What you view as the benefits of NBS?

Australian Genomics agrees the current benefits of newborn screening programs include:

- early detection of conditions and early intervention
- improved health outcomes for newborns
- improved life outcomes for newborns and their families through referral to ongoing systems of care
- improved decision making regarding future family planning.

Exploration through current research, could potentially expand future benefits of newborn bloodspot screening to include:

- rapid analysis for acutely ill children through reanalysis of data obtained in newborn screening
- economic benefits of using the data generated in screening for later diagnostic testing.
- equity of access to services and conditions screened for nationally -access to life saving or life changing treatment
- supportive prospective management of families through difficult challenges.

- There is a need to have a screening system in place that can quickly adapt to new disorders as new life saving treatments become available. This is why through GenSCAN the MRFF projects are taking a whole genome approach to screening as additional disorders (and their genes) can be quickly added without the need to create new assays.

Australian Genomics would welcome the opportunity to further review and comment on the Assessment process of the National NBS Policy Framework.

Question 3.2

What you view as the harms of NBS?

The potential harms of the NBS programs include:

- false positive reporting and subsequent treatment or intervention
- false negative reporting and lack treatment or intervention
- inequity of access and resultant inequity to subsequent treatment or intervention
- lack of understanding or awareness of the benefits of NBS and refusal to participate
- there is a potentially workforce harm through lack of future proofing and national workforce readiness analysis, in the changing newborn screening landscape.

Question 3.3

How important do you think it is to consider the: (checkbox response)

	Not at all important	Slightly important	Moderately important	Very important	Extremely important
Benefit of NBS					x
Harm of NBS					x

Question 3.4

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

- what we know about the condition,
- how good the screening test is at finding the condition in a newborn,
- what we know about how to best manage a condition, and
- 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? (check box response)

Not at all important	Slightly important	Moderately important	Very important	Extremely important
				x

What does an accepted intervention look like?

Through existing conditions screened such as PKU or Maple Syrup Urine Disease, we know interventions can be lifelong diet modifications. Other conditions benefit from early intervention in the form of physiotherapy, occupational therapies, speech therapies (including feeding and assistive technologies) to improve the health outcomes of the newborn. Some conditions, such as DMD previously mentioned, the intervention is more targeted to the family and supporting decisions around future family planning.

Additional interventions include options for the family to access clinical trials.

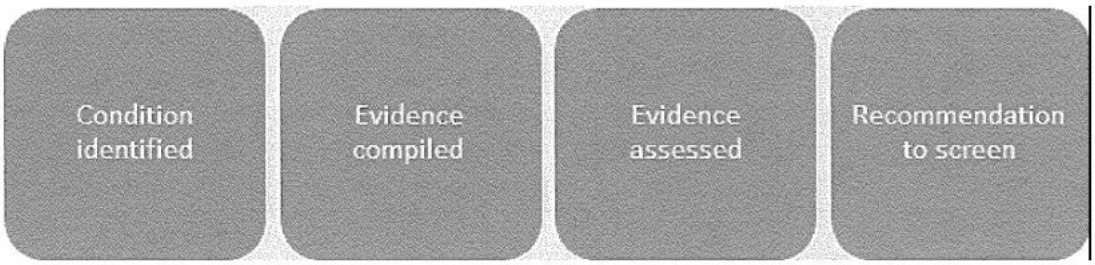
Question 3.5

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.

In light of research into genomics in newborn screening, further consideration needs to be given to conditions that onset beyond the newborn period, as well as criteria adjustments for conditions that do not have an associated intervention.

Question 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 above simplified assessment process for illustrative purposes, please highlight any specific points along the assessment process at which consumer input should be sought?

Best practice approaches tell us that consultation as early as possible is key to the successful outcomes. Consultation should take place at least by when evidence is being compiled.

Question 3.7

In your view, what would be the most appropriate way for you to be involved?(check box response)

Consultation forums	Online approaches	Surveys	Consultation paper	Not interested in being involved in consultation
x	x	x	x	

Other - please specify

Question 3.8

How should the department best advise on consultation opportunities for conditions going through an assessment process? (check box response)

Medical Services Advisory Committee website	x
Health website	x
Other - Please specify – GP clinics and/or Obstetricians	x

The Medical Services Advisory Committee may have a role to play, perhaps with the development of a broader specific Health Technology Assessment group that is able to assess each application on relative merits - this would be similar, to the Recommended Unified Screening Panel (RUSP) process in the USA.

The mention of the “Health Website” – would that be state or federal or both? This outward facing ongoing consultation opportunity brings into question the “consumer facing” component of national consistency. This equally involve consistency in messaging, language and imagery at all levels of newborn screening programs.

4. Readiness

'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 to 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.

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.

Question 4.1

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

Again, through GenSCAN and the associated MRFF projects, the possibility of using the dried bloodspot cards for multiple purposes is being explored. This will potentially impact of the timeliness and impact to families on diagnostic testing if the bloodspot samples already acquired are used for diagnostic purposes. Current investigations nationally and internationally are researching how (and whether) to introduce genomic testing to the NBS program. There are many issues to resolve before this can be done, including understanding how genomic tests will impact how consent is given. Considerable investments are being made to explore multiple touchpoints with parents throughout pregnancy to ensure information is understood prior to consenting. Additional considerations include ethical and privacy issues in relation to data storage, and data access from a national perspective.

Further, an ethical lens is required as considerations are made regarding which conditions should be included in NBS. Expansion of conditions tested will potentially lead to a created number of newborns identified with a condition which will have a further impact on workforce and support services if not adequately resources and assessed at a national scale. Further, whilst newborn bloodspot screening is a successful public health initiative, literacy about newborn screening is low. Existing research into parental views on informed consent for expanded NBS tells us that information delivered in a post-natal setting is often downplayed, misunderstood, or forgotten with competing priorities and potential parental fatigue (Louise Moody, 2013). **Error! Bookmark not defined.**An additional consideration of the current research is exploring the approach to expand mothers earlier in pregnancy to expand on the knowledge of newborn screening especially with the exploration of genomics in newborn screening.

Question 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: (checkbox response)

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

Question 5.1 - Measure of Success

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?

Measure of Success 1

Participation - high level of trust maintained in the program.

Measure of Success 2

Outcome measures should be considered a key measure of success (i.e. reduced morbidity/mortality, improved health/wellbeing or some other agreed measure of benefit).

Measure of Success 3

Economic assessment for good value healthcare.

Other:

- Reduced stigma to disability
- Effective management of true positive screens
- Effective counselling of false positive screens
- Identification of false negative screens
- Quality (highly accurate screening results)
- Safety (patient privacy and data securely maintained)
- Assessment process is streamlined – no long delays in reviewing evidence for new conditions to be included.

References

L Moody, K. C. (2013). Parental views on informed consent for expanded newborn screening. *Health Expectations*, 239-250.