

## RVA Education: Expansion of Newborn Bloodspot Screening Public Consultation

### Background

On 11 November 2022, at Rare Voices Australia's (RVA) National Rare Disease Summit, the Hon Mark Butler MP, Minister for Health and Aged Care, announced [a public consultation on the expansion of newborn bloodspot screening \(NBS\)](#). This announcement followed the Australian Government's commitment to invest \$39 million over four years to expand NBS to include more conditions and ensure consistency of programs across Australia. Historically, NBS programs have been funded and implemented at a state government level. RVA welcomes the commitment and investment by the Commonwealth into this critical program.

Australia's NBS program is important for the rare disease community as the majority of conditions currently screened, and being considered for screening, are rare conditions. NBS is an important program that supports the earliest possible diagnosis of some rare diseases. As outlined in the [National Strategic Action Plan for Rare Diseases](#) (the Action Plan), early diagnosis is critical for better outcomes as it enables the best immediate treatment and care. The rare disease community has been calling for increased equity, timeliness and consistency of screening for many years. As the national peak body for Australians living with a rare disease, RVA has been highlighting the need for increased sustainability, equity, transparency and consistency of screening for NBS for some time. RVA has conducted targeted advocacy at both the Commonwealth and state levels over the last two years. We also acknowledge the advocacy of our RVA Partner organisations and the broader rare disease sector.

This consultation is an opportunity for stakeholders to lodge their input into NBS policy decisions. RVA has developed this resource to provide RVA Partner organisations, and the broader rare disease sector, with prompts that may assist in the development of a submission. Not all prompts will be relevant for all submissions.

### Consultation Details

The consultation is a survey available on the [Department of Health and Aged Care's Consultation Hub](#).

The survey covers five areas:

1. Where are we heading? Aims and objectives of NBS programs.
2. Achieving national consistency. Considering how this can be achieved and barriers to achieving it.
3. Assessments. The assessment process, ethical and social complexity, types of interventions to be considered.
4. Readiness. What needs to be in place for effective screening and follow up?
5. What does success look like?

The Department of Health has developed this [Consultation Paper](#), which provides useful background for those considering making a submission.

**The consultation closes 12 December 2022.**

## Key Messages

As the national peak body for Australians living with a rare disease, RVA will be making a submission to this consultation. We recommend those considering making a submission review the [Newborn Bloodspot Screening National Policy Framework](#) as it provides insight into the types of conditions that may be considered for inclusion in the NBS program. RVA's submission will be guided by the Action Plan. Specific aspects of the Action Plan relevant to this consultation include:

### ***Priority 2.2: Ensure diagnosis of a rare disease is timely and accurate.***

#### **Action 2.2.1: Ensure all Australians have equitable access to a range of diagnostic tools and tests, providing the best chance of early and accurate diagnosis.**

*2.2.1.2. Ensure all existing screening and testing programs are sustainable and evolve in line with innovation over time*

#### **Action 2.2.2: Develop policy that supports the implementation of diagnostic tools and tests.**

*2.2.2.2. Address urgent funding gaps associated with the effective implementation and sustained success of the Newborn Bloodspot Screening (NBS) National Policy Framework.*

### ***Priority 2.1: Provide rare disease care and support that is integrated and appropriate for all Australians living with a rare disease, while being both person and family-centred.***

#### **Action 2.1.5: Embed the voice of people living with a rare disease and their families and carers throughout structures and systems that impact rare diseases.**

### ***Priority 2.3: Facilitate increased reproductive confidence.***

NBS also aligns with aspects of the [National Preventative Health Strategy 2021-2030](#). In particular, ensuring that Australian children have the best start in life and ensuring “groups that experience poorer health outcomes compared to the rest of the population have greater improvements in health – addressing inequity in health”. Australians living with a rare disease often experience inequity in health and other support systems.

The following dot points draw on key government policy positions and may provide some prompts when developing your submission.

- 1. Where are we heading? Aims and objectives of NBS programs. The consultation survey provides background information and details about the aims and objectives of the program.**
  - Measurement of, and accountability for, meeting the aims and objectives of NBS programs. In particular, timeliness, equity and the ability to evolve in line with innovation over time.
  - Processes and timeframes for reviews of aims and objectives to ensure they meet community expectations.
  - How we ensure programs are person and family-centred.

- Consideration of the value of knowing, the impact of an extended diagnostic odyssey and the importance of timely information to facilitate increased reproductive confidence for families whose child is seriously unwell as part of the aims and objectives of NBS.
  - Strategies to maintain the high level of public trust and acceptability are important when expanding the program to meet aims and objectives.
- 2. Achieving national consistency. The consultation survey provides background information about consistency between programs and asks for reflections and thoughts on issues related to programs achieving consistency.**
- Consistency needs to address both the conditions tested for and time of implementation. Inconsistencies in either area create inequities for Australian families with babies being tested for a serious condition in some jurisdictions but not others.
  - Consistency should also consider access to support, clinical care and education for those who have a confirmed diagnosis.
  - Policy and budget mechanisms in state programs need to be considered to ensure conditions recommended for inclusion in NBS programs are implemented in a timely and consistent manner throughout Australia.
  - State NBS policymakers to be engaged in and informed about assessments of new conditions for NBS.
- 3. Assessment. This section covers assessment criteria, assessment processes, types of interventions that should be considered and ethical and social complexity associated with NBS. The consultation survey provides background information to consider when responding.**
- Evidence-based decision making informed by expertise that ensures public trust in NBS programs.
  - Process completed in a timely manner with assessments for conditions triaged against a predetermined set of criteria, including clinical need and evidence available. Horizon scanning to drive a more proactive assessment that addresses innovation, including emerging science and medical technologies.
  - Transparent processes for embedding the voice of the community in decision-making processes to be considered.
  - Interventions that could be considered may include promising clinical trials, symptom management and care that inhibits disease progression.
  - Consideration of ethical and social complexities, including how these complexities may be managed.

**4. Readiness. This section asks you to consider how expansion of NBS could impact on readiness to provide appropriate intervention, care and support to families that receive a positive diagnosis. The consultation survey provides some background information for you to consider when responding.**

- Consideration of providing high quality, specialist care to families regardless of where they live.
- The role of, and access to, genetic counselling and mental health supports for families who receive a diagnosis.
- Specialist care and support provided at time of diagnosis.
- Appropriately resourced care teams.

**5. What does success look like? This section asks you to consider the most important measures of a successful expansion of NBS.**

- Diagnosis is timely and accurate.
- Rare disease care and support that is integrated and appropriate for all Australians living with a rare disease, while being both person and family-centred.
- NBS programs are sustainable and evolve in line with innovation over time.
- Consistency and equity of access.
- Strategies to manage complexities associated with population-wide screening programs.
- Consideration of potential benefits such as the value of knowing, reducing diagnostic odyssey and increasing reproductive confidence.

### **Additional Support and Resources for RVA Partners**

RVA hosted *The Changing Face of Newborn Bloodspot Screening – A Rare Disease Sector Forum* on 16 June 2022. The forum brought together RVA Partners, NBS clinical and research experts, NBS policy experts and industry to discuss the importance of a consistent and equitable program, the role of national leadership, how the NBS program is changing and what these changes mean for the rare disease sector. Other topics included how Australia's NBS program compares to international programs, emerging research in the NBS space, the role of genomics and the link between NBS, care and policy. [Watch the replay](#) on RVA's Online Education Portal and lookout for a follow-up forum that will be published soon.

Additionally, RVA has just released a new course, *Writing Effective Government Submissions*, which is also available on the Online Education Portal. [Click here](#) to access the course, which is available exclusively to RVA Partners.

**Note:** you will need to [register on RVA's Online Education Portal](#) to access the forum and the course if you haven't already registered on the site.

RVA Partners can also access customised mentoring support by contacting Louise Healy, RVA's Education and Advocacy Manager: [education@rarevoices.org.au](mailto:education@rarevoices.org.au).