Nomination forms for requesting the assessment of a condition for addition to or removal from newborn bloodspot screening

Overview

This document contains the nomination forms provided in the *Newborn Bloodspot Screening National Policy Framework* (the Framework) and described in Policy Area 5: Decision-making process. These forms must be completed in order to request the assessment of a condition for addition to or removal from newborn bloodspot screening.

Before submitting a nomination form, please consider the following information.

Completing a nomination form

The Standing Committee on Screening (SCoS) will consider conditions for assessment once a year. Nomination forms must be completed and submitted to SCoS@health.gov.au by **no later than 1 November** each year. Nominations received after this date will be considered as part of the following year's submissions.

Anyone in Australia can nominate a condition (either for addition to screening or removal from screening) by completing the appropriate nomination form. It is recommended that nominees seek the advice and guidance of an Australian newborn bloodspot screening program prior to completing the form. This will help to ensure that conditions appropriate for nomination and assessment have not already been considered for inclusion, and reduce the chance of duplication if multiple groups or jurisdictions are working on similar applications. Contact details for the newborn bloodspot screening programs may be sought from the national Newborn Bloodspot Screening Program Management Committee, via the SCoS email address listed above.

Consideration of the nomination form

Once the completed nomination forms have been received, the Program Management Committee will make an initial assessment of all applications and provide a recommendation to SCoS as to which of the nominated conditions merit more detailed assessment. The recommendation reached by the Program Management Committee is primarily based on the information provided in the nomination forms. As such, it is important that the nomination form is as complete, comprehensive and accurate in its responses as possible. If insufficient evidence is provided in the nomination form, applicants will be advised after this first assessment and may wish to resubmit once all the required information is obtained.

Recommendations for further assessment

After considering the recommendation from the Program Management Committee, SCoS will determine which conditions merit detailed review. A detailed review involves an assessment of all available evidence on screening for the condition in question, in line with the decision-making criteria in the Framework.

Progression to and timing of the detailed review are dependent on a number of considerations including: availability of staff and resources to support the review; the level of evidence available in Australia and internationally; the complexity of the issues being considered; and whether an economic analysis is conducted.

Final recommendation

Based on the outcome of the detailed review, SCoS will arrive at one of the final recommendations shown in the box below. If SCoS recommends screening (or ceasing screening) for a condition, the relevant recommendation, accompanied by preliminary cost implications where necessary, will be submitted to the Australian Health Ministers' Advisory Council (AHMAC) for consideration, via the relevant Principal Committee.

If the recommendation is supported by AHMAC, state and territory governments are then responsible for funding and establishing any other requirements around adding conditions, taking into account local contexts. It may not be appropriate for all states and territories to screen for all conditions due to differences in local populations, priorities and/or feasibility.

Recommendations that can be made following assessment of the evidence for screening a condition

1. **When considering including a condition in newborn bloodspot screening**, possible recommendations include:
    - Screening is recommended.
    - A pilot is recommended and specific issues flagged for investigation.
    - Based on the current evidence and understanding of a condition, screening is not recommended at this time. However, there may be merit in revisiting this condition in the future if further evidence emerges.
    - Screening is not recommended.

2. **When considering removing a condition currently screened**, possible recommendations include:
    - Continue screening.
    - Cease screening.

More information

For more information regarding the decision-making process for adding or removing conditions from newborn bloodspot screening programs, see Policy Area 5: Decision-making process in the *Newborn Bloodspot Screening National Policy Framework*.

Any specific questions regarding the nomination or assessment process can be submitted to the Program Management Committee, via SCoS@health.gov.au.

Nomination form requesting assessment of a condition for *addition to* newborn bloodspot screening

Please submit to the Newborn Bloodspot Screening Program Management Committee via SCoS@health.gov.au

Date received: (to be completed by secretariat)

Questions	Response
Name of nominator(s)	
Organisation(s) (if applicable)	
Contact details (address, phone, email)	
Role(s) (for example, clinician, researcher, parent, advocate etc.)	
Condition nominated for assessment (specifying form(s), if applicable)	
OMIM* or other names for the condition	

*Online Mendelian Inheritance in Man: http://www.omim.org/

Instructions for completion

- Please complete as many of the 'response' sections within this form as possible, citing relevant references within the text by number, then list and attach all references at section 6.
- It is recommended that a nominee who is not from a newborn bloodspot screening program seeks the advice and guidance of their jurisdiction's newborn bloodspot screening program regarding the required documentation and evidence in order to make a submission for the addition or removal of a condition.
- When the nomination form is complete, it should be submitted to the Newborn Bloodspot Screening Program Management Committee.

1. The condition

The condition should be a serious health problem that leads to significant morbidity or mortality. There should be a benefit to conducting screening in the newborn period; and the natural history of the condition, including development from latent to declared disease, should be adequately understood.

Guiding questions	Response
What is the incidence of the condition in Australia? Is this determined clinically or through screening studies in other countries?	

Guiding questions	Response
What is the burden of disease associated with the condition, including morbidity and mortality? What is the spectrum of disease—in particular, are there mild or late-onset forms?	
At what age would the condition usually be detected clinically?	
What are the benefits of early diagnosis and intervention/treatment? (Consider such benefits as early intervention, prevention of symptoms, reduction of disease severity, provision of a definitive diagnosis, emotional and social benefits and provision of information that would assist families with reproductive decision making.)	
What are the possible harms of screening and/or early diagnosis?	

2. The test

There should be a suitable test protocol to identify the presence of the condition, and the test protocol should be socially and ethically acceptable to health professionals and the public.

Guiding questions	Nominator's response
Describe a detailed methodology for the test (for example, tandem mass spectrometry, immunoassay, molecular), including any second-tier testing required. Provide reference to a published methodology and describe any modifications required.	
Can the test be performed on the same dried bloodspot specimen that is used currently? If not, what additional sample would be required?	
For the proposed testing protocol, comment on the:	
clinical and analytic validity	
sensitivity	
specificity	
false positive rate	
false negative rate	

Guiding questions	Nominator's response
positive predictive value	
negative predictive value	
Can the test be multiplexed?	
What other conditions may be detected (clinical or of unknown significance)?	
What would be the cost of the test?	
If DNA analysis is required, would testing include common mutations, a panel or full sequencing?	
What are the potential harms associated with the test protocol?	

3. The intervention

There should be an accepted intervention for patients with recognised disease, and facilities for diagnosis and management should be available so that these services can be offered if there is a positive screening result.

Guiding questions	Nominator's response
What diagnostic testing is necessary? Is it available and reliable? What is its associated cost?	
What is the established intervention/treatment for this condition?	
Do all patients require an intervention or treatment upon diagnosis? If not, can those who require treatment be distinguished from those who do not?	
How effective is the intervention/treatment? (Does it alleviate symptoms, slow/halt progression?)	
What are the impacts on quality of life?	
How urgent is the intervention/treatment? Must it be initiated before symptoms present?	
What are the potential harms of the intervention/treatment?	
What is the cost of the intervention/treatment?	
What facilities are required to deliver the intervention/treatment? Do current health care facilities in each state and	

Guiding questions	Nominator's response
territory have capacity, and are they of sufficient quality, to support the intervention/treatment? Is there equitable access to the intervention/treatment?	

4. Cost-effectiveness

Guiding questions	Nominator's response
Provide any available evidence for the cost-effectiveness of screening for this condition, either from Australia or internationally.	

5. Any other comments

6. References

Please list and attach relevant references.

Nomination form requesting assessment of a condition for *removal from* newborn bloodspot screening

Please submit to the Newborn Bloodspot Screening Program Management Committee via SCoS@health.gov.au

Date received: (to be completed by secretariat)

Questions	Response
Name of nominator(s)	
Organisation(s) (if applicable)	
Contact details (address, phone, email)	
Role(s) (for example, clinician, researcher, parent, advocate etc.)	
Condition nominated for assessment (specifying form(s) if applicable)	
Screening method	
OMIM* or other names for the condition	

*Online Mendelian Inheritance in Man: http://www.omim.org/

Instructions for completion

- Please complete as many of the 'response' sections within this form as possible, citing relevant references within the text by number, then list and attach all references at the end of the form.
- It is recommended that a nominee who is not from a newborn bloodspot screening program seeks the advice and guidance of their jurisdiction's newborn bloodspot screening program regarding the required documentation and evidence in order to make a submission for the addition or removal of a condition.
- When the nomination form is complete, it should be submitted to the Newborn Bloodspot Screening Program Management Committee.

Guiding questions	Response
When was screening initiated for this condition and why?	
What is the rationale for proposing to remove the condition from screening? Provide relevant information drawing on current screening experience and a review of literature to support removal.	

Guiding questions	Response
What is the incidence in Australia? Is this determined clinically or through screening studies in Australia or other countries?	
What positive impacts would removing this condition have on the program (for example, in terms of the impact on families, on the laboratory, on maternity service providers etc.)?	
What would be the clinical implications of removing the condition from screening? Include reference to the burden of disease associated with the condition, including morbidity and mortality, and the spectrum of disease.	
Are there other risks of removing this condition from screening (for example, impact on the ability to detect other conditions; impact on the family, including future reproductive risk; community concern etc.)?	
Is the condition screened internationally?	
Would removal of this condition from screening have any other implications for the quality of the program?	
Are there any alternatives to removal (for example, alterations to cut-offs, further follow-up testing etc.)?	
For the current testing protocol, comment on the:	
clinical and analytic validity	
sensitivity	
specificity	
false positive rate	
false negative rate	
positive predictive value	
negative predictive value	
Is the test multiplexed?	
Does testing identify other conditions (clinical or of unknown significance)?	
What would be the cost implications of removing the test?	

Any other comments

References

Please list and attach relevant references.