Toward National Newborn Screening in Australia

Veronica Wiley, PhD, FHGSA, FFSc(RCPA)

NSW Newborn Screening Programme,
The Children's Hospital at Westmead

Disciplines of Genetic Medicine and
Paediatrics and Child Health
University of Sydney

Members of SCOS NBSWG

This talk

- Overview of newborn screening practice

- Development of a national framework

NBS in Australia

- Began in NSW in 1964 with PKU
- Other states soon offered screening
- Screening was offered to all by mid 1970s
- Screening governance state based

- Program leaders collaborated

HGSA

- Founded in 1976; incorporated 1977
- provide a forum for the various disciplines collected under the title of Human Genetics
- “members” have a recognised qualification and are employed in that discipline
- Branches: 6
- SIGs: 4
- Committees: 7

HGSA/RACP Newborn Screening Committee

- Founded in 1985
- Advises HGSA on all aspects relevant to newborn screening
- Representatives from each program
  - Clinical representative
  - Scientific representative
- Policies include
  - Newborn bloodspot testing
  - Retention and storage of bloodspot cards

Newborn Bloodspot Policy

- Australasian policy
- Voluntary and publically funded
- Public health initiative for early identification
- Timely intervention – regardless of external event
- Local differences exist
  - State decisions
- 6 laboratories (5 Aust 1 NZ)

http://www.hgsa.org.au/

Newborn Screening in Australia

COVERAGE	BIRTHS ABS 2013	DISORDERS SCREENED
NSW + ACT	106,007	pku, ch, cf, gal MSMS
Victoria	73,969	pku, ch, cf, MSMS
Queensland + upper NT	65,379	pku, ch, cf, gal MSMS
S. Australia+lower NT	22115	pku, ch, cf, gal MSMS
Tasmania	6,049	
W. Australia	34,516	pku, ch, cf, gal MSMS

HGSA Recommendations for screening for specific disorders – Cat 1

- When assessing whether a particular disorder should be added to the screening program of a region, the appropriate cost comparison is the cost of adding the disorder versus not adding it.
- Screening is highly recommended for the following conditions because there is a demonstrated benefit from early diagnosis, the benefit is balanced against financial and other costs, there are suitable tests and follow-up services are available.
  - Congenital adrenal hyperplasia (CAH)
  - Primary congenital hypothyroidism (CH)
  - Cystic fibrosis (CF)
  - Disorders of amino acid, organic acid and fatty acid metabolism covered by analysis of aminoacids and acylcarnitines by tandem mass spectrometry. Note that disorders not being sought but using markers for other disorders may be found and followed up.

  - Conditions not screened by all jurisdictions - CAH (currently screened in NZ, APEG recommended) and SCID (currently under consideration for screening by NZ Ministry) and supported by ICSA. Disoders in MSMS panel may differ eg NZ not screen for 3MCCC and soon CUD

Recommendations for screening for specific disorders –cat 2

- Screening is recommended for the following conditions depending on local circumstances. There is lack of consensus at the international level on the benefit of screening for these conditions.

  - Biotinidase deficiency
  - Galactosaemias
  - Haemoglobinopathies

Recommendations for screening for specific disorders –cat 3

- Screening is currently not recommended for the following conditions where screening tests are not available, or, tests are available but proof of advantage from early diagnosis is absent or uncertain, or the test is unsuitable or does not detect those cases in which there might be an advantage. New knowledge about screening and screening outcome in these conditions should be monitored regularly.
  - Bile acid disorders
  - Cytomegalovirus
  - Duchenne muscular dystrophy
  - Familial hypercholesterolemia II
  - Fragile X
  - G6PD deficiency
  - Haemochromatosis
  - Lysosomal storage disorders
  - SCID
  - Toxoplasmosis

Expanding/changing screening protocol

- Each program approach relevant ministry
- No formal govt process to oversee NBS
  - If it is not broken don't fix it
- HGSA wrote to Federal Health Minister to request inclusion of CAH screening in Australia
- SCOS
  - NBS working group

SCOS NBS WG

- Formed in 2013
- Tasked with using a consultative approach
- Chaired by independent health policy expert
- Secretariat support
- Representatives:
  - Commonwealth government
  - SCOS
  - Jurisdictional screening policy
  - NBS program management
  - NBS scientist
  - Consumer
  - Bioethicist
  - Professional societies: ACM, HGSA, RACP, RCPA, ISNS

Project Objectives

- To provide clear policy guidance, based on the best available evidence, to support the continued success of NBS.

- To develop a national process to recommend to governments the conditions for which screening should be offered.

Consultation

- Contact to professionals and consumers
- Communiques after each meeting
- Website for easy update
  - http://www.genomics.health.wa.gov.au/nbspf/index.cfm
- Survey –online public consultation survey
- Focused open consultation workshops

Communique

Newborn Bloodspot Screening Working Group

Communique: 16 and 17 September 2015

Purpose

The purpose of this communique is to provide a timely overview of progress on the development of a national newborn bloodspot screening (NBS) policy framework. More information can be found on the *project website*.

Background

The Newborn Bloodspot Screening Working Group (NBSWG) was convened in March 2014 by the Standing Committee on Screening (SCoS), to develop a national policy framework for NBS in Australia. Communiques are released following each NBSWG meeting, and other significant milestones.

Meeting overview

The NBSWG held its 12th meeting in Sydney on 16 and 17 September 2015, where it reviewed the draft policy framework in full and considered outstanding issues.

Context

Early in the project, it was agreed that to support the continued success and development of NBS, the policy framework would include the following six chapters:

- *introduction*
- *program overview*
- *program implementation*
- *quality and safety*
- *monitoring, evaluation and review, and*

1. decision making framework.

Website

http://www.genomics.health.wa.gov.au/nbspf/index.cfm

- About NBS in Australia
- What is a policy framework?
- Why is a policy framework needed for NBS in Australia?
- How will the policy framework affect me?
- Who is responsible for the project?
- How will the project proceed?
- Project updates
- Stakeholder involvement
- Past stakeholder consultations
- How can I find out more?
- Useful links

Consultation workshops

Discussion of:

- Needs and expectations of stakeholders from newborn screening
- Content of the framework
- Beneficiary of newborn screening
- Decision making criteria that may be used to assess new conditions for newborn screening.

Framework

- builds on the strengths and the successes of current programs
- is applicable in all jurisdictions
- provides clear policy guidance
- supports consistency of programs
- increases transparency of the programs
- supports a safe and effective screening environment
- enables assessment of conditions for screening
- is consumer-focused
- maximises benefits to the community, and
- supports Australian services to lead international best practice.

NBS Policy framework

- supports those working in the programs and families through providing clear policy direction and information on NBS.
- includes policies:
  - Program overview
  - Program implementation
  - Quality and safety
  - Monitoring, evaluation and review
  - decision making framework to enable assessment of disorders for screening.

Nomination form for assessment of a condition for Newborn Bloodspot Screening - Adding or removing a condition

Date received:	(to be completed by Secretariat)

Questions	**Nominator's response**
Name of nominator/s:	
Organisation/s (if applicable):	
Role/s (eg clinician, researcher, parent, advocate etc)	
Condition nominated for assessment:	
Condition nominated for recommendation of *addition to* or *removal from* screening:	
Type of condition:	
Screening method:	
Gene/s (if applicable):	
Locus:	
OMIM or other names for the disorder:	

The condition

The condition should be serious health problem which 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	Nominator's response
What is the incidence in Australia? Is this determined clinically or through screening?	
What is the burden of disease associated with the condition, including morbidity and mortality? What is the spectrum of disease?	
When would the condition usually be detected clinically?	
What are the benefits of early diagnosis and intervention? (Consider benefits including early intervention, prevention of symptoms, reduction of disease severity, provision of a definitive diagnosis, emotional and social benefits, or provision of actionable reproductive information to parents).	
What are the possible harms of screening/ early diagnosis?	

The test

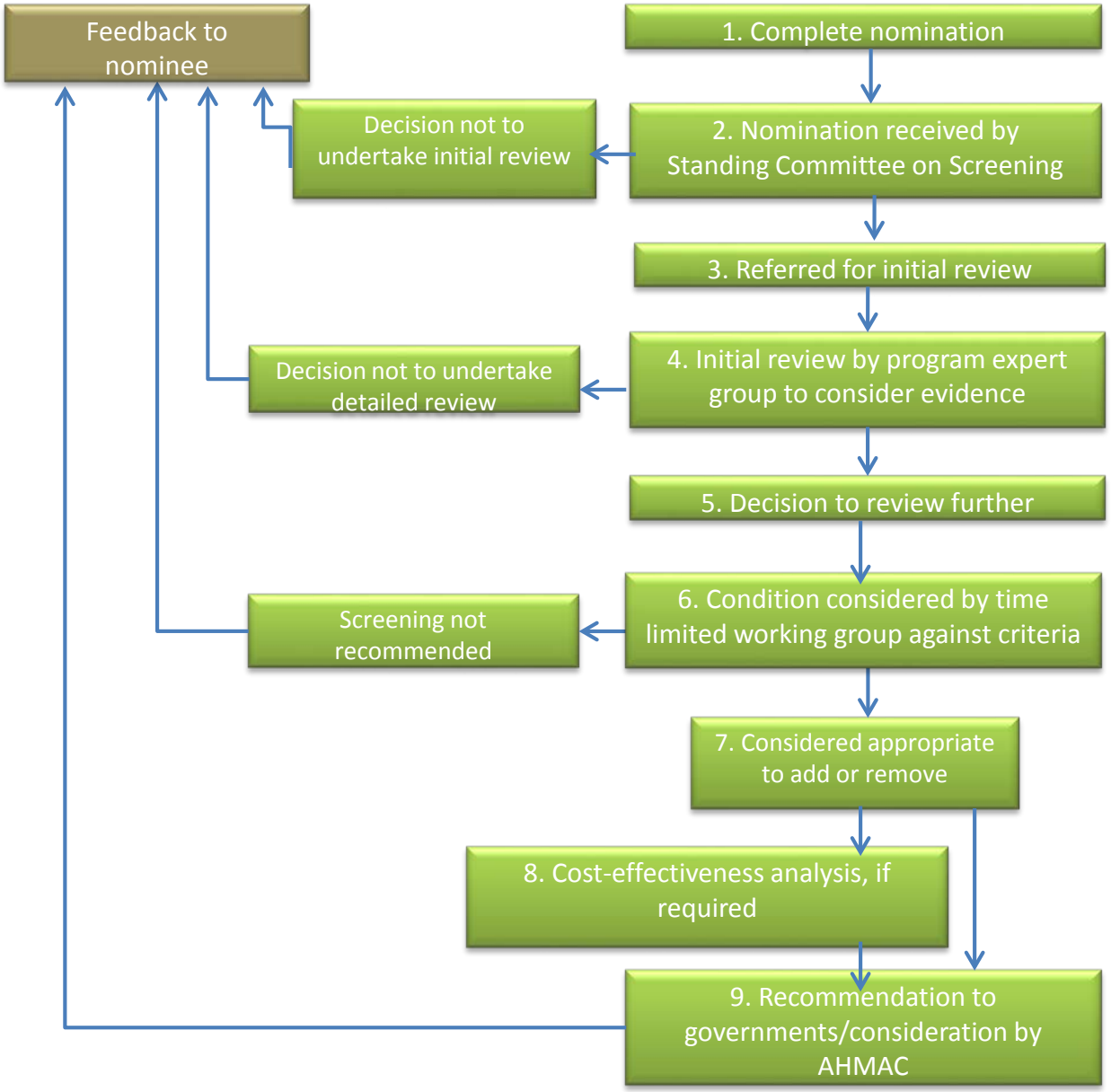
There should be a suitable test protocol to identify the presence of the condition, and the test protocol should be socially and ethically acceptable to health professionals and the public.

Guiding questions	Nominator's response
Can the test be performed on the same dried bloodspots as are used currently? If not, what sample would be required?	
For the proposed testing protocol, comment on the:	
clinical and analytic validity	
sensitivity	
specificity	
false positive rate	
false negative rate	
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?	
What confirmatory and diagnostic testing is necessary? Is it available and reliable?	
If DNA analysis is required, would testing include common mutations only or full screening?	
What are the potential harms associated with the test protocol?	

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 is the established intervention for this condition?	
Do all patients require intervention?	
How effective is the intervention? (Does it alleviate symptoms, slow/halt progression?) What are the impacts on quality of life?	
How urgent is the intervention? Must it be initiated before symptoms present?	
What are the potential harms of intervention?	
What is the cost of intervention?	
Do current facilities have capacity, and are they of sufficient quality, to support intervention? Is there equitable access to intervention?	


Feedback to nominee
1. Complete nomination
Decision not to undertake initial review
2. Nomination received by Standing Committee on Screening
3. Referred for initial review
Decision not to undertake detailed review
4. Initial review by program expert group to consider evidence
5. Decision to review further
Screening not recommended
6. Condition considered by time limited working group against criteria
7. Considered appropriate to add or remove
8. Cost-effectiveness analysis, if required
9. Recommendation to governments/consideration by AHMAC

Government review of framework

SCoS	• Standing Committee on Screening • Health officials from Commonwealth, state and territory health departments with expertise in screening
CCPHPC	• Community Care and Population Health Principal Committee • Health executives from each health department across Australia
AHMAC	• Australian Health Ministers' Advisory Council • Directors General, Secretaries, Chief Executives
SCoH	• Standing Council on Health/ aka COAG Health Council • Commonwealth and state and territory Health Ministers

Conclusion

SCOS NBSWG developed a policy built on the strengths and successes of the last 50 years of screening in Australia to provide clear policy guidance, support consistency, with increased transparency as well as enable structured assessment of disorders.

The policy was submitted in November 2015 and is going through the hierarchy approval process – anticipated approval in mid 2016

Thank you

Questions?/Comments?