



The Royal Australasian
College of Physicians

NEWBORN SCREENING IN AUSTRALIA

Position Statement

May 2015

Preamble

Newborn screening is an important child health issue, since early identification of infants who are affected by certain congenital disorders and timely intervention significantly reduce morbidity, mortality and associated disabilities. Newborn screening can be used to identify specific health conditions which, untreated, lead to intellectual disability, other significant morbidity or child death. It is a public health issue which currently lacks a mechanism for advancement in Australia. A framework for improving and expanding newborn screening across Australia is important, as it will improve health outcomes from screening and ensure equitable access.

The RACP advocates for an effective, equitable and fully funded national newborn screening program for Australia that includes ongoing scientific evaluation of conditions that could be considered for inclusion in, or removal from, the screening program.

Recommendations

The RACP recommends that the Federal and State Governments:

1. Develop a national strategy for newborn screening.
2. Establish an expert panel as part of this strategy and task it with the evaluation of additional health conditions for inclusion in the national screening program, the technologies used for newborn screening and related issues.
3. Establish an equitable national funding model in order to fully implement the recommendations of the panel.

Principles

The criteria underlying a newborn screening policy framework should recognise:

1. Newborn screening and early management of congenital health conditions has the potential to reduce child morbidity and mortality, thereby improving population health.
2. Evidence shows that newborn blood spot and hearing screening is cost effective in Australasia.ⁱ
3. The need for planning, implementing and monitoring newborn screening for health conditions as scientific evidence becomes available, in line with international best practice.
4. Criteria for inclusion should address the feasibility of testing and the expected improvement to health outcomes.
5. A standardised national approach to newborn screening, including evaluation of outcomes, would address a range of issues, such as equity, informed consent and ethical factors, and deliver optimum health benefits to the maximum possible number of newborns.

Background

Historical context

Newborn blood spot screening programs were established in most Australian states in the 1960s to screen for phenylketonuria, a genetic disorder that leads to accumulation of phenylalanine in the body. Untreated, it causes significant intellectual disability. These programs were established in an ad hoc manner, funded by individual states. The subsequent addition of congenital hypothyroidism screening in the 1970s prevented intellectual disability in even more children. Other programs were added (such as tandem mass spectrometry expanded screeningⁱⁱ), often because a need was highlighted by a pilot program and funding found by individual states. Programs were occasionally removed because of ineffectiveness, both here and overseas (for example, screening for histidinaemia).ⁱⁱⁱ

Current population screening of newborns in Australia

Currently, all Australian-born newborns are screened for congenital hypothyroidism, cystic fibrosis, amino acid disorders including phenylketonuria, organic acidemias and fatty acid oxidation defects. All states and territories except Victoria also screen for galactosemia.

Universal newborn hearing screening was proposed at the July 2002 Australian Health Ministers' Conference in Darwin. The Ministers requested that the Medical Services Advisory Committee conduct a review of the feasibility of hearing screening in an Australian context. A 2007 report by that body found that, over the long term, a universal national hearing screening program would be "cost saving for Australian society".^{iv} Newborn screening for hearing deficit has proven effective in early detection of congenital deafness in New South Wales and Victoria.

The Australian Standing Committee on Screening had not been involved in considerations of newborn screening until 2014, when it set up a screening working group to develop a newborn blood spot screening policy framework. Managed by the Western Australian Department of Health, this Committee's work will continue through 2015.

Concerns with current approach to population screening of newborns in Australia

There is a lack of consistency within Australia in consent for screening and subsequent storage of the residual dried blood spot cards. Victoria introduced signed consent for screening and separate consent for the use of anonymised samples for research in 2011 following privacy concerns in that state. Retention of cards following completion of screening varies from two years, such as in Western Australia, to permanent storage, such as in South Australia.

Despite the lack of formal national systems, the Australian state-based newborn screening laboratories have maintained a strong collaborative focus. They have provided an internationally respected level of evidence of the effectiveness of newborn screening for cystic fibrosis^{v, vi}, medium-chain acyl-coenzyme A dehydrogenase deficiency (MCAD) and expanded screening using tandem mass spectrometry.^{vii, viii, ix, x, xi}

Establishing greater consistency across Australia's jurisdictions would enable more effective sharing of research, and better service provision. Beyond the States and Territories, Australia could move to implement beneficial screening programs already in play in New Zealand.

International models that may inform the development in Australia of a comprehensive and national newborn screening program

New Zealand has a policy framework and funding underpinning a national program of newborn screening, as well as an implementation framework.^{xii} New Zealand introduced congenital adrenal hyperplasia (CAH) screening in 1984, with demonstrated effectiveness.^{xiii, xiv} CAH screening has been implemented in most first world countries for more than 30 years. Over the past 10 years, the Australian Paediatric Endocrine Group (APEG) and the Human Genetics Society of Australasia (HGSA) have unsuccessfully applied to state health departments to introduce CAH screening. New Zealand infants are also screened for biotinidase deficiency, and plans are underway to screen for Severe Combined Immunodeficiency (SCID).

The USA has a model for consideration of additional health conditions, developed after the Maternal and Child Health Bureau (MCHB) of the Health Resources and Services Administration (HRSA) of the United States Department of Health and Human Services (DHHS) commissioned the American College of Medical Genetics (ACMG) in 2002 to:

1. Conduct an analysis of the scientific literature on the effectiveness of newborn screening.
2. Gather expert opinion to delineate the best evidence for screening for specified conditions and develop recommendations focused on newborn screening, including but not limited to the development of a uniform condition panel.
3. Consider other components of the newborn screening system that are critical to achieving the expected outcomes in those screened.^{xv}

Similarly to New Zealand, the USA's system of review contains elements that could be emulated in creating a newborn screening body in Australia.

Additional health conditions that could be considered for inclusion in newborn screening programs

There are a growing number of conditions that could be considered for inclusion in newborn screening programs. These include, among others, congenital adrenal hyperplasia, severe combined immunodeficiency, Fragile X and lysosomal storage diseases. The potential benefits of newborn screening for congenital heart and lung conditions with an electrocardiogram (for prolonged QT Syndrome) and pulse oximetry (for cyanotic congenital heart disease) have also been raised. The emerging evidence in relation to these conditions increases the imperative to develop a more consistent approach to their evaluation of the potential benefits of their inclusion in screening programs.

The natural history and variation in presentation of rare conditions can usually only be understood by screening large populations. Newborn screening programs may have a legitimate research component as part of their public health warrant.^{xvi}

Conclusion

A unified national system of recommendations, advice and information is important. It will help to enhance, expand and improve the ability to reduce mortality and morbidity from congenital disorders in newborns and children, and will bring Australia in to line with other first world health systems. The health and financial benefits of implementing such a system are significant.

Document details

Publication name:	Newborn Screening Position Statement
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Approval date:	7 May 2015
Review date:	May 2020

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