Newborn bloodspot screening: setting the Australian national policy agenda

Australia lags behind other countries in developing a national policy framework for newborn screening

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Since the late 1960s, newborn bloodspot screening (NBS) programs have successfully integrated public health and clinical medicine to identify and treat rare disease early, preventing disability and death. The management of these programs is increasingly being challenged by opportunities offered by rapidly evolving technologies, and by different perspectives on the role of screening. For NBS programs to respond effectively and consistently, a national mechanism is required to define and monitor standards and guide the introduction of new tests and technologies. This mechanism exists (or is in development) in many countries, but not in Australia.

Despite recommendations made by the Australian Law Reform Commission (ALRC) in 2003, NBS programs, managed and funded by state health departments, continue to exist in the absence of "nationally consistent rules" or "standards".[1] In response to the ALRC Report 96, the Australian Health Ministers' Advisory Council Advisory Group on Human Gene Patents and Genetic Testing took on the task of developing national guidelines; however, the policy, which had successfully worked through the controversial issues of consent, storage and use, failed to achieve national endorsement and has since been abandoned. The reasons for this are varied, but fundamental problems are likely to be the lack of conviction that government involvement was required and the absence of broader stakeholder engagement. As a result, in the face of overlapping and often contradictory legal frameworks,[2] each state has been left to make its own decisions regarding what to screen for and how to manage the technical and regulatory coordination of the program.

Meanwhile, the critical issue of how conditions should be assessed for inclusion remains unresolved. In this void, the Human Genetics Society of Australasia (HGSA) with the Division of Paediatrics of the Royal Australasian College of Physicians (RACP) has coordinated policy positions for health professionals engaged directly in NBS. The policy, which includes a template for assessing new conditions,[3] recommends screening for 33 conditions including congenital adrenal hyperplasia (CAH). For many, but not all conditions, evidence supports screening; however, there remains no national policy framework to analyse evidence and prioritise future decision making.

By the early 2000s, tandem mass spectrometry was incorporated into all Australian NBS programs, marking a new era in NBS.[4] This technology, along with the identification of new disease markers and genes, and a growing knowledge of disease aetiology and treatment, has led to the expansion of NBS to most of the conditions covered by the HGSA/RACP policy — but not CAH. The traditional criteria for conditions screened include an understanding of the disease, and the availability of a cost-effective, suitable and acceptable test and treatment. In the absence of clear evidence — a hallmark of rare disease — these criteria can be difficult to meet. To further complicate the matter, the concept of the benefits of screening is now evolving past health benefits for the child to benefits for the family, such as reproductive choice and the avoidance of "diagnostic odyssey", and even enhanced research opportunities.[5]

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The effectiveness of NBS is compromised by the introduction of tests that do not warrant screening (as seen with NBS for Krabbe disease in New York State),[6,7] but also by the failure to introduce those tests that do. The latter is perhaps of greater concern in Australia, where there is no mechanism to add tests for such conditions, evident in the failure to at least assess the proposals for CAH screening.[8,9] So far, despite strong advocacy for NBS for CAH and calls for a systematic evaluation, all initiatives have floundered. In an *MJA InSight* article, eminent Australian endocrinologist Garry Warne described being bounced between state and federal bodies while trying to have the test introduced.[8] A spokesperson for the Department of Health noted that there had not been an application to the Medical Services Advisory Committee (required before the listing of any service on the Medicare Benefits Schedule [MBS]).[8] It would be clear to anyone involved that the states manage NBS as a service program, and tests are not covered by the MBS, nor were they intended to be.

Internationally, the response has been more productive. In 2009, the European Commission established a network of multidisciplinary experts including representatives from professional and scientific organisations and patient groups to guide NBS programs. The resulting document, endorsed in 2011, includes a system to evaluate the quality and ethical aspects of screening, a decision matrix to systematically expand (or contract) programs and a recommendation for a European Union NBS committee for ongoing monitoring and support.[10] In the United States, an advisory committee on heritable disorders in newborns and children has been created to take on this role.[11] Similarly, in New Zealand, the Ministry of Health's National Screening Unit has established expert groups and recommendations for the expansion of NBS to ensure the program is evidence-based and achieves quality standards.[12] In the United Kingdom, the Fetal, Maternal and Child Health Coordinating Group of the National Screening Committee's policy framework is supported by independent systematic reviews. These groups and policies differ in many aspects, including the extent of their jurisdictional power, but all present some mechanism to

encourage effective and consistent management of NBS programs.

Without a national framework, Australian NBS programs remain ill prepared to meet challenges, expand screening, and ensure equitable and consistent services. Similar to other countries, Australia needs a mechanism to consider evidence-based policy, funding and stakeholder engagement to lead effective planning and implementation of NBS programs. An initial step would be to establish a national NBS advisory body responsible for monitoring performance and evaluating — and recommending — new and existing tests for NBS. This body should lay the foundations for the development of a policy that represents the views of consumers and experts in health policy, NBS, paediatrics, law and bioethics, and which is agreed between jurisdictions, with each state retaining responsibility for funding and managing its NBS program. Issues requiring national consensus include managing children, program monitoring and evaluation, consent, storage and retention, legislative or regulatory oversight and evidence-based program expansion. The responsibility rests firmly with jurisdictional policy advisers working with those who manage the delivery of NBS programs.

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