Editorial

# Neonatal Screening – A Global Perspective

Jiun Lee,[1] *MBBS, M Med (Paed), FAMS*, Roy Joseph,[1,2] *MBBS, MMed (Paed)*, Victor Samuel Rajadurai,[3] *MBBS, MRCP (Paed) UK, FAMS*

This supplement issue of the *Annals Academy of Medicine Singapore* represents a collection of nearly two-thirds of the lectures presented at the 6th Asia-Pacific Regional Meeting of the International Society for Neonatal Screening, held in Singapore from 29 August to 1 September 2007. The theme for the meeting was "Improving Child Health Through Universal Neonatal Screening".

In her keynote lecture Professor Bridget Wilcken noted the success of neonatal screening programmes over the past 40 years in reducing mental retardation and other paediatric health problems, and emphasised the importance of congenital hypothyroidism screening for any country that is contemplating starting screening programmes, to reduce the devastating burden of this readily treatable condition. It is heartening to know that healthcare authorities in Bangladesh, China and Pakistan (Hasan, Gu, Afroze) are doing precisely that.

Hearing impairment is the commonest congenital disorder that can be detected and treated early through universal newborn hearing screening, resulting not only in improved speech, but also cognitive development for the affected children. Similar to many other human health problems, a multidisciplinary approach to its management is necessary for the holistic care of the hearing impaired (Lim, Hayes, Nie, Lim and Daniel, Reyes).

Since the pioneering work in the 1990s by Millington and Chace[1,2] in developing tandem mass spectrometry (MS/MS), this technique has gained acceptance in many developed countries for detecting inborn errors of metabolism (IEMs), represented classically by phenylketonuria. A simple blood spot could potentially screen for more than 25 IEMs. However, debate still surrounds issues like what conditions to include in the screening panel and its cost-effectiveness (Therrell and Wilcken, Padilla and Lam). Most centres would screen for phenylketonuria and medium-chain acyl-coenzyme A dehydrogenase deficiency. Success of screening programmes to a great extent depends on getting the processes right, and one prerequisite would be quality assurance through regular audits (Fletcher, Hsiao). For many countries, epidemiological studies of IEMs are few. National centres should therefore be created to reap economies of scale in view of the relative rarity of IEMs (Yamaguchi, Gu, Thong).

Neonatal screening has come a long way since Robert Guthrie first started phenylketonuria screening using the bacterial inhibition assay in the 1960s.[3] Screening for congenital hypothyroidism and glucose-6-phosphate dehydrogenase (G6PD) deficiency is efficacious because these two conditions are relatively prevalent (congenital hypo-thyroidism universally, G6PD deficiency in East Asia), the tests cheap and treatment fairly straightforward with good outcomes.

In contrast even as a group the incidence of IEMs is quite uncommon (about 1:4,000 to 1:5,000 from a full panel of acylcarnitines and amino acids using MS/MS).[4] The screening technique is expensive and management of affected infants is much more involved for the physician and the family. Hence, for developing countries IEM screening will probably have to come after immunisation and infectious diseases in terms of national healthcare priorities. For existing screening programmes the challenge must be to continually ensure adequate specialist follow-up for patients with these individually rare, lifelong disorders.

The Asia-Pacific region has a long history in neonatal screening. In 1965 the late Emeritus Professor Wong Hock Boon initiated cord blood G6PD screening in Singapore, and that virtually eliminated kernicterus which was then the commonest cause of first week deaths in newborn babies.[5] Professor Naruse (see Oration) began neonatal screening

[1] Department of Neonatology, National University Hospital, Singapore
[2] Department of Paediatrics, Yong Loo Lin School of Medicine, National University of Singapore, Singapore
[3] Department of Neonatology, KK Women's and Children's Hospital, Singapore
Address for Correspondence: Dr Lee Jiun, Department of Neonatology, National University Hospital, 5 Lower Kent Ridge Road, Singapore 119074.
Email: Lee_Jiun@nuhs.edu.sg

in Tokyo in 1966. His foresight and subsequent efforts have enabled neonatal screening meetings in the Asia-Pacific region to continue to flourish, with the aim of fostering regional cooperation and assistance.

We hope the articles in this issue, we hope, will give readers a global perspective surrounding neonatal screening, and also reinforce the importance of secondary prevention (identifying a disease in its earliest stages before symptoms appear) in improving the health of children.

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