

particular technology or treatment works—*can* it be done? This type of assessment usually involves major input by technical experts, including physicians, and rests on the ability to evaluate objectively specific expectations of a new laboratory test or a new therapy. It has become codified in 'evidence-based medicine', and it has become critically important in decision-making regarding health care resource allocation. However, public values also play a central role in the process—*should* it be done? This is a much more difficult area of public policy development, and it requires input from a much wider group of stakeholders, including the general public.

### Concluding Remarks

Paediatricians are faced with a number of challenges directly related to the management of inborn errors of metabolism. Staying abreast of the rapid expansion of knowledge in this area, along with the emergence of new diagnostic and screening technologies, is a familiar, though daunting, task. Despite the advances in knowledge, however, there is a pressing need for the development of new and effective treatments for many inherited metabolic diseases, especially those affecting the brain. One of the most formidable and difficult challenges is the removal of barriers to access to existing diagnostic and treatment services. Facing the public policy challenge is generally unfamiliar territory for practicing paediatricians. However, it is one in which we must be prepared to participate in a more active way than in the past if the advances in medicine that are occurring now are to reach our patients in the future.

### Public Health Approach of Inborn Errors of Metabolism in Hong Kong

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The commonest inherited metabolic disease in Hong Kong is glucose-6-phosphate dehydrogenase (G6PD) deficiency. When a disease is of significant incidence and severity, the public health approach would be considered for its prevention and management. In Hong Kong, G6PD deficiency was recognised as one such condition that required massive screening and early intervention. A combined neonatal screening programme for G6PD deficiency and congenital hypothyroidism (CHT) was started in Hong Kong by the Department of Health (DH) since 1984. The screening system included activities in public education, sampling, laboratory assays, follow up intervention and evaluation. As a public health programme, it was provided free of charge on a totally voluntary basis. Although all newborns in Hong Kong were entitled to this service, only about 70% of life birth had their blood samples directed to the central neonatal screening laboratory under this programme, the remaining 30% received screening from laboratories in private hospitals. Overall, more than 99% of all newborns in Hong Kong received screening for these disorders. Cord blood was used universally as the screening sample for both conditions. The decision of employing cord blood, instead of filter paper blood spot, was based on two reasons. Firstly, it was considered important that any deficiency of this enzyme in a newborn need be notified within the first couple of days for effective counselling and intervention. The use of cord blood certainly offered distinct advantage. Secondly, transport of these samples was not a problem in a geographically compact place like Hong Kong. Screening for G6PD enzyme activity was performed by colorimetric assay. Up to the end of December 2001, a total of 679,241 neonates had been screened in the public hospitals. It was found that 4.53% of the males and 0.32% of the females were affected. Compared with 1970s, there was a tremendous decrease in the morbidity and mortality resulting from hyperbilirubinaemia due to G6PD deficiency. There are occasional cases of mishap as a result of failure to inform individual families of G6PD deficiency during the long holidays. Counselling for this condition was normally conducted by phone by genetic counselors, and this method had been shown to be effective.