Newborn screening—progress and challenges

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Four years have passed since the International Symposium of the International Society for Neonatal Screening (ISNS) on Awaji Island in Japan provided the scientific material for the first and very successful special issue of the *Journal of Inherited Metabolic Disease* on this topic. Newborn screening, especially for inherited metabolic diseases, has always been a focal point for the Society for the Study of Inborn Errors of Metabolism (SSIEM) and its journal. Powered by developments in molecular medicine, reliable high-throughput multianalyte technologies, and new treatments, recent years have seen incredible progress in this field. The ISNS is a relatively new society, formed in 1991, and traditionally centred on the dissemination of information and networking among colleagues working directly in the field. It is developing into a broader platform, not only for the exchange of rapidly growing technical knowledge but with a central focus on providing care for the individual as well as the general population amidst the increasingly

complex interplay among scientists, physicians, ethicists, patient groups, and advocates as well as public and political organisations.

The more than 20 papers in this second special issue of the journal on the topic of newborn screening encapsulate the science of the 13th International Symposium of the ISNS and the 7th International Congress of the Latin-American Society of Inborn Error of Metabolism and Neonatal Screening (SLEIMPN) held in Cancun, Mexico, in December 2009. This issue reflects the rapidly enlarging field, from technological advances to complex ethical issues for societies with different backgrounds. The disorders considered go beyond inherited metabolic and endocrinological diseases as documented by papers not only on screening for cystic fibrosis but for congenital cytomegalovirus and severe combined immune deficiency.

We hope that this special issue will provide important information and stimulus to the numerous initiatives in countries all over the world. It illustrates what was always true even though possibly not so obvious to everyone: newborn screening is one of the most complex scientific and medical endeavours. All its different aspects were there from the beginning of the “crusade” of the late Robert Guthrie, who transformed his seminal work on bacterial inhibition assays using dried blood spots on filter paper into population-based newborn screening, starting with phenylketonuria in the 1960s. Guthrie and his colleagues around the world developed newborn screening as a powerful tool of preventive medicine for rare inborn errors of metabolism with the vision of improving the health and quality of life of affected individuals. Developments in technology have led to improved analytical sensitivity and accuracy and have greatly increased the range of metabolites that can be assayed. However, advances are hampered by significant regional variations in screening programmes, diagnostic

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processes, and the organisation of screening centres. Currently these are presenting a major challenge, especially for the optimisation of mass spectrometry MS/MS-based screening. In view of the very low incidence of almost all disorders, the optimal use, continuous improvement, harmonisation, and further expansion of MS/MS-based programmes, as well as other new high-throughput technologies under consideration, can only be achieved through concerted transnational actions. A major challenge is to balance the different and often well-justified approaches. The best answers are likely to come from the continuous evaluation of different programmes including a careful follow-up of patients and their families and economic assessment. Process times for taking screening samples, establishment of a screening diagnosis, confirmation of

suspect diagnoses and start of treatment as well as outcome variables, e.g. metabolic decompensations, clinical status, and intellectual development, all need to be evaluated. Currently most of these aspects have been addressed in only a small number of research publications. We can add to these the varied conclusions on diverse scientific questions drawn together in this issue. Even screening for congenital hypothyroidism still has several important issues to be resolved, and it is important to remember that the majority of babies born in the world still lack screening for this or indeed any other disorder. Scientific work and evidence-based evaluation of the essential issues in newborn screening are indispensable and will help with decisions on how to proceed in different countries. There will not be an optimal screening model for the entire world but optimized strategies for each country.