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# Advances in Newborn Screening

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## Message from the Guest Editor

More than 50 years have passed since the world's first newborn screening (NBS) program began. Initially, the NBS program was initiated for several diseases of inborn errors of metabolism, but it has gradually been expanded to include other diseases such as endocrine diseases. In the past two decades, as therapeutic options and diagnostic methods have evolved, the target diseases for NBS have become more diverse. The big change began with the expansion of NBS by MS/MS that began in the late 1990s. This method allowed us to simultaneously test for more than 20 kinds of amino acidemias, organic acidemias and fatty acid oxidation disorders. Subsequently, a method to simultaneously measure the activity of multiple enzymes in dried blood spots (DBS) was developed by utilizing the stability of lysosomal enzymes in the DBS, which allowed simultaneous screening for many lysozomal diseases such as mucopolysaccharidoses, Pompe disease, Fabry disease and Gaucher disease. The target of mass screening is expanding beyond classic metabolic disorders, and real-time PCR-based screening methods have been established for severe combined immunodeficiency (SCID) and spinal muscular atrophy (SMA).

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