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Newborn Screening for Lysosomal Storage Disorders

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Message from the Guest Editors

Newborn screening for lysosomal storage disorders (LSDs) has gained significant momentum during the past two years, stimulated by the addition of Pompe Disease and Hurler Syndrome to the recommended universal screening panel (RUSP).

New therapies, especially enzyme replacement and hematopoietic stem cell transplantation, have given new hope to families with affected children. Also, the development of fluorometric enzymatic assays for LSDs in dried blood pioneered by Nestor Chamoles in 2001 and boosted by Michael Gelb's tandem mass spectrometry method (2004) enables early detection of newborns. Multiplexing of LSD enzyme assays using either MS/MS or digital microfluidic fluorometry has facilitated high-throughput prospective screening for multiple LSDs.

Many have argued against screening for LSDs, citing the expense and variable efficacy of current treatment options and the preponderance of cases with later onset forms of disease, pseudodeficiency alleles, genetic variants of unknown significance and carriers. Post-analytical methods including statistical analytical tools and second-tier biochemical tests should help clarify these challenges.



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Message from the Editorial Board

The *International Journal of Neonatal Screening (IJNS)* is owned by the International Society for Neonatal Screening. The journal is a publishing platform for all topics concerning newborn screening and covers clinical presentations, metabolite pattern detection, technical aspects, and “tips and tricks”. “Tips and tricks” will appear in the form of “Technical Notes” on the journal website, thus making *IJNS* the platform for NBS laboratories to share their valuable information as to how they have improved assays, workflow, and performance. Such improvements are sometimes achieved through tiny adaptations of existing methods, which normally remain in lab journals, but which might help improve the performance of NBS laboratories around the world.

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