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Nutritional Treatment and Screening for Hereditary Metabolic Disorders

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Message from the Guest Editors

Neonatal screening for metabolic disorders has expanded significantly since its inception in the early 1960s. The introduction of tandem mass spectrometry in the late 1990s has enabled the detection of approximately 40 conditions. Nutritional management is crucial in these conditions to prevent mortality, metabolic crises, and long-term complications. This management often involves restricting precursors of toxic metabolites and supplementing deficient precursors while ensuring adequate caloric intake. Additionally, supplementation with cofactors, vitamins, or specialized metabolic formulas is often required.

Despite significant advancements over the past two decades, numerous challenges remain. These include differentiating between severe and mild phenotypes, identifying sensitive and specific markers for the presymptomatic diagnosis of treatable conditions such as certain urea cycle disorders, harmonizing newborn screening programs, implementing advanced therapies, and providing recommendations for breastfeeding in infants with inherited metabolic diseases. These advancements are all aimed at ultimately improving patients' quality of life and clinical outcomes.

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Message from the Editorial Board

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