

Newborn Screening

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Keywords

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Newborn screening (NBS) is evolving rapidly with advances in next-generation sequencing (NGS), creating opportunities to complement established biochemical screening and potentially expand the range of disorders detectable in the neonatal period. However, the concept of “genetics-first newborn screening” (gNBS) requires careful definition. Targeted molecular testing for a specific disorder, such as current genetic approaches for spinal muscular atrophy (SMA), should be distinguished from broad panel-based or genome-scale sequencing applied as a primary screening strategy.

Early genomic NBS studies have demonstrated that targeted analysis of sequencing data is technically feasible and can identify additional potentially actionable/treatable findings. However, technical feasibility and an increased screen-positive yield is not equivalent with demonstrated clinical utility or public-health benefit. For many disorders, available studies have not yet established condition-specific screening sensitivity, positive predictive value in unselected newborn populations, long-term clinical outcomes, cost-effectiveness, or the balance between benefits and potential harms associated with uncertain, incompletely penetrant, or late-onset findings.

Importantly, emerging evidence also illustrates the complementary nature of genomic and biochemical information. Studies such as BabyDetect have shown that biochemical data may help prevent inappropriate reporting or interpretation of genomic findings, including those involving *GALT* and complex *CFTR* alleles. These observations support consideration of genomic NBS not as a single uniform strategy, but within different screening options: sequencing as a stand-alone first-tier test; sequencing performed in parallel with conventional biochemical NBS; sequencing used as a second-tier investigation following an abnormal biochemical first-tier result to decrease the number of false positives and increase the positive prediction

rate. The latter approach is already incorporated into screening algorithms for selected disorders, including cystic fibrosis and severe combined immunodeficiency (SCID), and may currently provide the most pragmatic framework for integrating genomic technologies into established NBS programmes.

Ultimately, the optimal position of NGS is likely to be condition-dependent and should be determined by its incremental contribution to screening performance, diagnostic certainty, timely treatment, clinical outcomes, cost-effectiveness, and overall public-health benefit. A critical evaluation of genomic NBS should therefore clearly distinguish between technical feasibility, screen-positive rate, diagnostic yield, actionability, clinical utility, and demonstrated benefit at the population level.

The manuscript by Jacquemin et al. can form the base for further discussions involving all stakeholders on the newborn screening strategy of the future.

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