

Revvity study shows value of NGS in newborn screening

December 2023—A recent study by Revvity Omics has shown the clinical value of proactive, sequencing-based screening in apparently healthy newborns (Balciuniene J, et al. *JAMA Netw Open*. 2023;6[7]:e2326445). The objective of the study was to assess the clinical utility of genome sequencing versus a gene panel for a curated set of medically actionable pediatric-onset conditions in a large cohort of apparently healthy newborns and children tested at a clinical laboratory.

The case series study was conducted among 562 apparently healthy children screened by genome sequencing at the Revvity Omics laboratory in Pittsburgh. Forty-six children (8.2 percent) were found to be at risk for pediatric-onset diseases, including 22 (3.9 percent) who were at risk for high-penetrance disorders. In contrast, 2.1 percent of 606 children screened with an exome-based panel of 268 genes for well-known medically actionable pediatric conditions were found to be at risk.

The risks uncovered by genome sequencing involved a wide range of pediatric-onset conditions likely to be missed on limited gene panels. Many of these risks involve high-penetrance, often neurodevelopmental disorders that may benefit from early interventions.

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