

Experts Endorse Universal Genomic Screening of Monogenic, Treatable Diseases in Newborns

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NEW YORK — An overwhelming majority of rare disease experts agree that a genomic sequencing test for monogenic treatable conditions should be available to all newborns, a new survey shows.

Currently, newborn screening is a state-mandated public health program that primarily uses mass spectrometry to identify and direct the initial treatment of infants at risk for rare, childhood-onset disorders that are amenable to early treatment, the authors noted.



While newborn genome sequencing has the potential to simultaneously evaluate risk for thousands of genetic disorders and has received a lot of interest from parents, the authors said that opinions of medical geneticists and other rare disease experts have largely been undocumented.

For this survey, published in [JAMA](#) on Monday, a team led by researchers at the Massachusetts General Hospital for Children and Harvard Medical School reached out to 386 rare disease experts, out of which 238 responded. The respondents were asked six questions to understand their perspectives on newborn genome sequencing and the answers were statistically analyzed.

The findings showed that 87.9 percent (161) respondents agreed that genome sequencing for monogenic treatable disorders should be made available to all newborns.

Younger experts were more likely to agree that genomic sequencing for treatable genetic diseases be made available for all newborns, compared to older experts. This suggested that clinical experts who trained more recently are more open to the use of molecular screening tools in apparently healthy newborns, the authors said.

Meanwhile, the respondents were also presented with a list of 651 gene-disease pairs and were asked whether they would recommend those for a newborn sequencing test. These genes belong to various clinical areas such cardiovascular, endocrinology, gastroenterology, hematology, immunology, metabolic, nephrology, neurology, oncology, ophthalmology, and pulmonology.

The survey revealed that 58.5 percent (107) of respondents agreed that newborn sequencing should include genes associated with treatable disorders even if those conditions are low penetrance, while 37.2 percent (68) agreed that actionable adult-onset conditions should be sequenced in newborns to facilitate cascade.

Overall, a list of 25 gene-disease pairs were endorsed by 85 percent or more of the experts. Many of these gene-disease pairs are clinically similar to disorders currently included on the Recommended Uniform Screening Panel (RUSP), the authors said.

There was also a strong concordance (98.4 percent) to incorporate screening for OTC deficiency, a condition with high morbidity and mortality in male infants that is currently not included in RUSP. Meanwhile, none of the cardiovascular, ophthalmology, or pulmonary genes had 80 percent or higher concordance among experts surveyed.

Experts also included many gene-disease pairs in clinical areas that have not previously been included in screening, such as genes for childhood-onset cancer predisposition conditions and bleeding disorders, the authors noted. They cited the example of RB1, a gene associated with hereditary retinoblastoma that was endorsed for screening by 89.3 percent of experts. Genes related to disorders with newly developed and emerging pharmacologic therapies, such as Niemann-Pick disease, were also recommended for screening.

The findings are one small step toward making a change at the policy level to offer these tests to newborns on a large scale, lead and corresponding author, Nina Gold, a physician in the medical genetics and metabolism division of Mass General Hospital for Children, told *GenomeWeb*.

"I am hoping that this list will provide a roadmap to incorporate genomic sequencing in newborns and help in identifying some of these treatable conditions," she added.

Highlighting one of the limitations of the survey, the authors noted that survey respondents were not asked about practical considerations about genomic testing such as cost and consent, as well as the relative scarcity of medical geneticists and other rare disease experts.

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