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Experts Support DNA Sequencing in Newborns

- In a national survey, researchers found that rare disease experts support genomic sequencing in healthy newborns to detect risk for treatable conditions
- Report details confirmatory testing, surveillance, and treatment options for over 600 genetic conditions

Babies born in U.S. hospitals currently undergo routine newborn screening, a laboratory test to identify the risk of up to 60 treatable conditions. But hundreds of genetic disorders, including a growing number of devastating childhood diseases, now have targeted treatments, including gene and cell therapies, that can offer permanent cures. Despite these advances, the addition of genomic sequencing to newborn screening programs has remained controversial. Findings from a new study led by researchers at Mass General Brigham suggest that rare disease experts are now calling for more expansive newborn testing.

In a study published today in *JAMA Network Open*, the research team received responses from 238 experts, 88 percent of whom agreed that DNA sequencing to screen for treatable childhood disorders should be made available to all newborns. The study also identified 432 gene-disease pairs that are not currently screened for, but that were recommended for newborn screening by over 50 percent of the experts, ranked in order of those that had the greatest consensus. Among the genes that most experts recommended for newborn screening were those associated with a lethal liver and brain disorder, the severe bleeding disorders known as hemophilia A and B, and an increased risk for retinoblastoma, a rare and fatal eye tumor in young children.

“Early identification of infants who are at risk for genetic disorders can be lifesaving and screening has the potential to improve healthcare disparities for affected children,” said lead author Nina Gold, MD, a medical geneticist at Massachusetts General Hospital for Children. “Medical experts are now calling for more conditions to be included in newborn screening that can only be identified through DNA

sequencing. In our survey, they reached a striking consensus about the highest priority conditions to include.”

In addition to soliciting expert consensus, the authors consolidated recommendations for over 600 genetic conditions from rare disease sub-specialists in order to offer clinical guidance when DNA screening was positive. In many cases in which DNA sequencing identifies a child at risk, a blood test or imaging study can then determine whether the disease condition is already underway, enabling early treatment. In other cases, the child will be entirely healthy despite the positive DNA screen and can be followed for the appearance of symptoms and signs in the future. The researchers note that future studies will be needed to determine whether newborn sequencing is cost-effective and positively contributes to short- and long-term outcomes.

Over the past seven years, randomized controlled trials in the NIH/NCATS-funded [BabySeq Project](#), conducted by researchers at Mass General Brigham and Boston Children’s Hospital, have demonstrated that comprehensive DNA sequencing of newborns can identify life-threatening genetic risks, many of which are treatable, in some babies shortly after birth. Gene therapy for treating some of these conditions, including hemophilia and Duchenne muscular dystrophy, are currently in development or entering clinical trials. Mass General Brigham recently launched its [Gene and Cell Therapy Institute](#) to help translate scientific discoveries into first-in-human clinical trials and, ultimately, life-changing treatments for patients.

“It has been a longstanding dream to someday offer DNA sequencing to all newborns in order to detect their risk of disease,” said senior author Robert C. Green, MD, MPH, a physician-scientist at Brigham and Women’s Hospital, professor of genetics at Harvard Medical School, and lead investigator of the BabySeq Project. “Our empirical data from BabySeq have demonstrated that nearly 10 percent of infants carry actionable genetic variants. With the recent explosion of gene and cell therapies, some of which can fully prevent or cure a condition before symptoms appear, it is urgent that we move forward to provide this option to families who are interested.”

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Paper cited: Gold NB, et al. “Perspectives of Rare Disease Experts on Newborn Genome Sequencing” JAMA Network Open DOI: 10.1001/jamanetworkopen.2023.12231.



About Mass General Brigham

Mass General Hospital and Brigham and Women's Hospital are both founding members of Mass General Brigham, an integrated academic health care system uniting great minds in medicine to make life-changing impact for patients in our communities and people around the world. Mass General Brigham connects a full continuum of care across a system of academic medical centers, community and specialty hospitals, a health insurance plan, physician networks, community health centers, home care, and long-term care services. Mass General Brigham is a non-profit organization that is committed to patient care, research, teaching, and service to the community. In addition, Mass General Brigham is one of the nation's leading biomedical research organizations and a principal teaching affiliate of Harvard Medical School. For more information, see massgeneralbrigham.org.

About Genomes2People

The Genomes2People Research Program at Brigham and Women's Hospital, the Broad Institute and Harvard Medical School explores the medical, behavioral and economic outcomes of integrating genomic information into medicine and society see genomes2people.org.

About the BabySeq Project

The NIH/NCAT-funded BabySeq Project is the world's first project to introduce comprehensive DNA sequencing for apparently healthy infants and has measured medical benefits, psychosocial outcomes and medical costs associated with newborn sequencing, see genomes2people.org/research/babyseq.

About Precision Population Health at Ariadne Labs

The Precision Population Health program at Ariadne Labs aims to bring the power of genomic medicine to primary care settings, in order to prevent diseases before they occur, see ariadnelabs.org/precision-population-health.

