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Should All U.S. Newborns Undergo Genomic Testing?

By [HealthDay](#)

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By [Cara Murez](#) HealthDay Reporter

TUESDAY, May 9, 2023 (HealthDay News) -- While newborns are only screened for about 60 treatable conditions, there are hundreds of genetic disorders that have targeted treatments.

Now, a national survey of experts in rare diseases found the vast majority support DNA sequencing in healthy newborns.

Testing, surveillance and treatment options exist for over 600 genetic conditions. This includes a growing number of devastating childhood diseases that now have targeted treatments that sometimes offer permanent cures.

“Early identification of infants who are at risk for genetic disorders can be lifesaving and screening has the potential to improve health care disparities for affected children,” said lead author Dr. Nina Gold, a medical geneticist at Massachusetts General Hospital for Children, in Boston.

“Medical experts are now calling for more conditions to be included in newborn screening that can only be identified through DNA sequencing,” Gold said in a hospital news release. “In our survey, they reached a striking consensus about the highest priority conditions to include.”



(HEALTHDAY)



The researchers conducted the survey between February and September 2022, receiving responses from 238 experts.

About 88% agreed that DNA sequencing to screen for treatable childhood disorders should be made available to all newborns. About 432 gene diseases that are not now screened for were recommended for newborn screening by more than 50% of the experts.

These include genes associated with a lethal liver and brain disorder, severe bleeding disorders known as hemophilia A and B, and an increased risk for a rare and fatal eye tumor in young children.

When DNA sequencing finds an infant at risk for a genetic disorder, often a blood test or imaging can determine if the condition is already ongoing. This makes early treatment possible.

In other cases, a child will be entirely healthy despite the positive DNA screen. In those circumstances, doctors can watch for potential future signs and symptoms.

Randomized, controlled trials in the U.S. National Institutes of Health (NIH)/National Center for Advancing Translational Sciences-funded [BabySeq Project](#) have found that comprehensive DNA sequencing of newborns can find treatable life-threatening risks.

The study, which was published online May 8 in *JAMA Network Open*, was funded by the NIH.

“It has been a longstanding dream to someday offer DNA sequencing to all newborns in order to detect their risk of disease,” said senior study author [Dr. Robert Green](#), a physician-scientist at Brigham and Women’s Hospital and lead investigator of the BabySeq Project.

“Our empirical data from BabySeq have demonstrated that nearly 10% 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,” Green said.

More information

The March of Dimes has more on [genetic and chromosomal conditions](#).

SOURCE: Mass General Brigham, news release, May 8, 2023

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