

Rare Disease Experts Support Newborn Genome Sequencing

May 8, 2023



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Research led by Mass General Hospital for Children suggests that almost 90% of rare disease experts are in favor of newborn genome sequencing for monogenic treatable disorders.

As reported in *JAMA Network Open*

([https://jama.jamanetwork.com/article.aspx?](https://jama.jamanetwork.com/article.aspx?doi=10.1001/jamanetworkopen.2023.12231&utm_campaign=articlePDF%26utm_medium=articlePDFlink%26utm_source=articlePDF%26utm_content=jamanetworkopen.2023.12231)

[doi=10.1001/jamanetworkopen.2023.12231&utm_campaign=articlePDF%26utm_medium=articlePDFlink%26utm_source=articlePDF%26utm_content=jamanetworkopen.2023.12231](https://jama.jamanetwork.com/article.aspx?doi=10.1001/jamanetworkopen.2023.12231&utm_campaign=articlePDF%26utm_medium=articlePDFlink%26utm_source=articlePDF%26utm_content=jamanetworkopen.2023.12231)), the researchers also asked about newborn screening for actionable adult-onset conditions and screening for conditions with no established therapies or management guidelines. Support for these options was present, but only in around a third of those surveyed.

“Newborn screening is a successful, 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,” write the authors.

Currently, this screening takes the form of heel prick testing. The number of conditions that are included in the test varies depending on country and also state within the U.S. A core panel of 34 conditions is recommended for all newborns in the U.S., with 26 secondary conditions that can be added depending on the area.

The continuing improvements in pricing and accessibility of genome sequencing have led to a number of studies and programs (<https://www.insideprecisionmedicine.com/topics/patient-care/pediatric-diseases/the-changing-face-of-newborn-screening/>) testing the benefits of this type of more extensive screening both in sick and healthy children with good results. However, newborn sequencing is still far from mainstream and is not something most families currently have access to.

An important part of introducing this form of screening more widely is acceptance by healthcare providers. This study sought the opinions of rare disease experts, as newborn sequencing has the potential to be very beneficial for rare disease diagnostics, on introducing newborn sequencing for various purposes.

Overall, 238 experts were surveyed (average age 53 years, 53% female) as part of the study. A significant majority (88%) agreed that newborn sequencing for monogenic treatable disorders should be made available to all newborns, with 59% agreeing that genetic variants linked to treatable disorders should be included, even if the conditions have low penetrance.

Around 37% of experts thought actionable adult-onset conditions should also be tested for to help set up cascade testing in parents and 28% thought conditions with no established therapies or management strategies should be included. The researchers also asked the experts to state the genetic variants and conditions they supported including, and 42 gene-disease pairs were endorsed by more than 80% of those surveyed.

“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, a medical geneticist at Massachusetts General Hospital for Children, a member of Mass General Brigham, in a press statement.

“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.”
