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STAT

## Rare-disease doctors support expanded newborn genomic screening, survey finds



By [Megan Molteni](#) May 8, 2023



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Plummeting costs of DNA sequencing technologies are injecting urgency into [the longstanding debate](#) over whether to dive deeper into the genomes of more infants — even apparently healthy ones.

Experts are divided about how helpful DNA sequencing data really are. The tests often identify mutations that raise someone’s risk of developing

a condition, but don't necessarily cause the disease. Uncertain results may confuse doctors and scare families — causing them to seek doctors' visits and treatments that most ultimately won't require, adding to unnecessary health care costs. There are also [concerns about false positives and false negatives](#) with sequencing. But [a new study](#) has found that among rare disease physicians — the ones to whom worried parents will turn — there is emerging consensus that these sorts of tests should be used on a broader scale.

When 238 rare-disease doctors across the U.S. were surveyed by a research team at Mass General Brigham in Boston, 88% of them agreed that DNA sequencing to screen for certain treatable childhood disorders should be made available to all newborns. The [study](#) was published Monday in JAMA Network Open.

“At a time when the health care system is quite burdened, and physician burnout is a real concern, we wanted to elicit the opinions of doctors who are going to be caring for these patients,” said Nina Gold, a medical geneticist who led the research. “We don't want to overburden them with a new type of clinical referral if they don't see value in it, so it's important that they think there's clinical utility in finding these patients early, diagnosing them and connecting them with available treatments.”

Expanded genomic newborn screening seeks to turn up hidden inherited diseases missed by current methods, which screen babies at birth for dozens of disorders by analyzing proteins and other molecules in a tiny drop of blood drawn from the heel. A small study published in February [found that 40% of infant deaths](#) were caused by a DNA glitch — compelling evidence that genetic disease may be the leading cause of infant mortality.

Gold is a part of the BabySeq Project, a clinical trial comparing standard newborn screening and genomic sequencing to determine the medical, economic, and social impacts of the latter. It is [one of four federally funded studies](#) investigating the merits of making sequencing available to infants more widely. Their team has reported that newborn sequencing revealed genetic variants that raised the risk of certain childhood conditions [in 9.4% of the babies tested](#).

In this survey, they asked rare-disease experts from around the country to weigh in on six statements regarding genomic newborn screening and the types of disorders that should be included. They also provided a more detailed list of 649 pairs of diseases and the gene that causes them, and asked experts which of these they would recommend being a part of newborn genomic screening programs. More than three-quarters of the physicians — who span many medical specialties including pediatric cardiovascular disease, endocrinology, hematology, neurology, and metabolism — agreed on 42 gene-disease pairs. They included diseases like hemophilia, Duchenne muscular dystrophy, and glycogen storage disease, illnesses for which treatments exist.

“In principle, there is no reason to screen for any disorder that has no effective treatment,” wrote one survey respondent. “Once an effective treatment becomes available and especially if treatment prior to symptoms is important to prevent irreversible damage, then screen.”

But for diseases without effective treatments, disorders that develop later in life, and conditions that are exceedingly rare, experts disagreed about how to handle them in a newborn screening program.

Mary Norton, a prenatal geneticist at the University of California, San

Francisco, who was not involved in the study, noted the variety in opinions as she cautioned reading too much into its results. “There is a difference between experts ‘calling for expansion’ — which implies it was instigated by a group of experts suggesting this is important to do — and a group responding to a survey,” Norton told STAT via email.

Among survey respondents, younger doctors tended to endorse more positive opinions about newborn genomic screening, an indication that attitudes in rare disease medicine appear to be changing. Gold thinks attitudes are shifting as an explosion of [cell and gene therapies](#) are becoming available, either through clinical trials or as approved products. Last year, the Food and Drug Administration’s undersized Office of Tissues and Advanced Therapies [began adding 100 new positions](#) to help manage more than 3,000 investigational new drug applications.

“Before, naming a genetic condition may not have led to any demonstrable change in symptoms,” Gold said. “We just couldn’t do anything about it. But now, as we’ve got more and more ways to treat some of these conditions, it’s an essential time to identify kids who could potentially benefit.”

Still, many questions remain about how providing families with DNA sequencing data might play out over time. At the heart of them is the big one: Do the benefits of having access to this information outweigh the costs? It might be a long time before there’s a definitive answer. But the picture is getting clearer as projects like BabySeq expand — it is currently recruiting a larger and more racially and economically diverse cohort — and more initiatives get underway; last year, England launched a pilot program [to sequence portions of DNA from 100,000 newborns](#).

## About the Author



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