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Can parents handle finding out their newborn is at higher risk of certain diseases? A small study suggests they can

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By Andrew Joseph Aug. 23, 2021

A row of newborn infants. *APStock*

There's a heated debate over whether to expand genomic sequencing to more newborns — especially about whether to sequence healthy babies. How will parents react to the results? The tests can turn out uncertain information — perhaps this child is at higher risk for a disease, but there's no guarantee she'll

actually develop it — and families could worry, subjecting their child and the health system to unnecessary tests, appointments, and other care.

A new study has found some encouraging signs. When parents were surveyed about measures like how well they bonded with their children, their levels of distress, and even the parents' relationships with one another, there were no meaningful differences for parents whose children had their genomes sequenced compared to those whose children underwent standard newborn screening.

"We did not find evidence of any sustained negative psychosocial impacts," said Amy McGuire, a bioethicist at Baylor College of Medicine and one of the authors of the paper. "We did not find that in this study at all, and we were very reassured by that."

The authors noted that the study looked at family dynamics for just 10 months and included a limited participant group. But the researchers say that it's a first step toward understanding how families will absorb the information provided by genomic sequencing and that it can be used as another factor as experts consider the risks and benefits of making sequencing more widely available.

The new paper, published Monday in JAMA Pediatrics, is the latest from the BabySeq project, one of four federally funded endeavors looking at the question of using genomic sequencing more commonly for newborns and whether doing so is merited.

Already, newborns are screened for a range of genetic diseases for which early detection and treatment are crucial. Genomic sequencing would take that many steps further, potentially uncovering additional health risks lurking in babies' DNA.

Whereas newborn screening is essentially a diagnostic — yes or no, do these children have beta thalassemia or cystic fibrosis? — the results of genomic sequencing aren't nearly as specific. The tests often identify genetic variants that raise someone's risk of developing a certain condition, but don't necessarily cause the disease. Experts are divided about how helpful that information is: perhaps some families get their children to the right specialists early, but other families might subject their children to extra doctors' visits they don't ultimately need. More sequencing could add costs and burdens to the health care system. There are also concerns about false positives and false negatives with sequencing.

The BabySeq study is a clinical trial involving several hundred families, some with sick babies, some with healthy; half the babies received standard newborn screening, and half received screening plus sequencing focused on genetic variants implicated in a range of childhood-onset conditions, plus a few adult-onset conditions. In 2019, the researchers published data showing that 9.4% of babies in the group had genetic variants that raised their risk of certain childhood conditions, and the new paper is based on survey data with the same families.

The researchers wanted to go beyond just asking parents if the results caused them anxiety or depression. So in addition to inquiring about distress, the survey asked parents how they were bonding with their children and about their relationship with their partners. Parents filled out the surveys when they found out the results of their children's tests, and then at three months and 10 months.

The researchers found some variation on some of the measures at certain points during the study among the different groups of parents, but those discrepancies went away over time. Overall, the study reported, there were no

negative impacts on families whose children's genomes were sequenced, even if the results suggested the children were at higher risk for certain conditions.

Jeffrey Botkin, a pediatrician and ethicist at the University of Utah, who was not involved with the new study, has been more skeptical of the utility of expanding sequencing to healthy newborns without family histories of genetic disease. He said that the new study "is useful as a piece in a much larger puzzle" and that it "provides important information about the psychological and family impacts of genomic sequencing in newborns," but that it did not address broader concerns with expanding sequencing.

Understanding the impact of sequencing healthy newborns "will require a determination of whether families and providers use this information effectively to prevent or ameliorate disease in at-risk children," Botkin wrote in an email. "This study is helping us understand that the risks are probably low (in this context), but we know the costs of sequencing would be high if used on a broad scale and the benefits for healthy children remain to be determined."

McGuire noted that the group in the study is not representative. The families were recruited at three Boston hospitals and had higher educational levels than the U.S. population overall. Three in four of the parents surveyed were white. The BabySeq team hopes to expand the study in terms of diversity and geography, and also to follow families for longer. One outstanding question: if families know their child is at a higher risk for a certain heart condition, does that affect what kind of activities they let their child participate in or how cautious they are in keeping their kids home from school during flu season? Experts have wondered, McGuire said, if this type of information will lead to a culture of "vulnerable child syndrome."

The new study “reassures us that, for a select group of parents who undertake [sequencing] for their newborn in a controlled environment with the best resources and care that money can buy, substantial personal harms do not appear in the first 10 months of life,” Beth Tarini, the associate director of Children’s National Hospital’s Center for Translational Research, wrote in an accompanying editorial also published Monday. “What happens in the U.S. health care landscape is beyond the scope of the larger BabySeq study but is a critical future step both in this area of research and for decision-making about the widespread use” of newborn sequencing.

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