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HEALTH Genetics Add Topic

Should a baby's genes be sequenced at birth? Study finds potential life-saving benefits

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What would happen if every newborn's genes were sequenced at birth? That's the question the BabySeq study has been trying to answer for a decade.

Its newest results suggest the genetic information could be used to save lives. And not just the baby's.

Several years ago, the research showed that out of 159 seemingly healthy babies whose genetic information was sequenced at birth, 17 were found to have "actionable" mutations – their genes predicted or increased the likelihood they would fall ill – and knowing that ahead of time could change the course of their lives.

This follow-up, 3 to 5 years later, shows the information was useful, both for the babies and three of their mothers. Most genetic diseases are inherited from either parent or the combination of the two, so the baby's mutation indicated one parent's genes carried a similar disease risk.

The exact details were not revealed, but all three mothers took action to prevent a serious condition, such as a double mastectomy for a genetic finding that indicated a high risk for breast cancer.

Right now, newborns in every state get a heel-stick blood test to screen for as many as 60 diseases that strike early in childhood. But there are already more than 700 treatable conditions not included in screenings, with many more treatments under development, said

Dr. Robert Green, a geneticist at Brigham and Women's Hospital and Harvard Medical School, who helped lead the research. Few American adults have had their genes sequenced.

"In the future, imagine identifying a risk for a devastating illness in a healthy newborn baby," Green said. "Imagine then being able to find the biomarkers for the ones who are going to develop the disease and even preventing it. Imagine how thrilling that would be."

With treatment options expanding, now is the right time to expand genetic testing, said Fyodor Urnov, an expert in gene editing at the University of California, Berkeley.

"For the first time, our ability to diagnose genetic disease is finally being matched by being able to do something about it," he said.

What should newborn genetic testing look for?

Initially, the BabySeq study team wasn't sure if they should include conditions that would only impact the baby later in life, such as the BRCA1 and BRCA2 genes known to dramatically increase risk for a number of cancers, including breast, ovarian prostate and pancreatic cancers.

The team decided if they found important mutations they had an ethical responsibility to inform the families. "We were stunned in BabySeq to find that once we opened the door to those adult-onset conditions, we found three babies who were carrying mutations in one of those conditions" that increases cancer risk, Green said.

Three mothers, who had no idea they were carrying this increased risk, had risk-reducing surgery after learning of their baby's status.

Previous research by the team showed families were not upset by this type of information. "We did not disrupt parent-child bonding," Green said.

It's unclear precisely how much genetic sequencing of newborns would cost. Now, the BabySeq team pays well over $1,000 per baby, including the sequencing and returning understandable results, but that cost would come down with widespread use, Green said.

While privacy is a concern, he thinks it's been overblown. "We've overindexed on the risk and underindexed on the benefits" of genetic sequencing, Green said. "We're going to come to a better balance as people's lives start getting saved."

The BabySeq team is currently recruiting for a larger study, including more diverse participants to better understand the impact of this kind of genetic sequencing. "We'll need

several thousand babies before we get a good read on what's representative and what's not," Green said.

Parents who don't want to know the information are free not to participate, so "we're just not getting a lot of families who are distressed" by the findings, Green said.

Putting newborn genetic testing to good use

Although some people might take issue with the idea of knowing someone's genetic destiny at birth, Green said the information can be extremely useful – and isn't fundamentally different from the heel sticks now done on newborns. "It doesn't seem very far away from something we've already decided is a public good," he said.

Pediatricians and parents will need training to understand the implications of genetic findings. Not every mutation is bad and many just indicate an increased risk, not a certain future, Green said.

But understanding genetic risk can enable people to take better care of their health, he said. "I believe that our entire medical system can benefit from integrating genomic information at every level."

Many genetic diseases strike early in life and often aren't diagnosed until after symptoms emerge and permanent damage has occurred, Urnov said. Sequencing at birth would allow treatments to start before that damage.

"For many of these diseases, giving early access to the therapy is the difference between a lifetime of disability or frankly, death, and health," he said.

Since these mutations can now be identified and treated or prevented it's unethical not to provide people with that information, Urnov said. "That's a moral wrong."

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