Baby DNA: Boston Researchers Find Childhood Genetic Risks In 9 Percent Of Newborns

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Just after Cora Stetson was born two years ago, she tested positive on a standard newborn screening test for a rare enzyme deficiency that, in the worst cases, can cause seizures, deafness and blindness and cognitive delays.

When she was retested, with the same biochemical blood test, her results came up negative. Normal.

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But then came the call from BabySeq, a pioneering study that is exploring what happens when extensive DNA analysis searching for potential childhood disorders is performed on newborns.

"They said, 'Cora came back positive,' " says her mother, Lauren Stetson of Melrose. "'She has a genetic disorder that we can fix — but also that needs attention right now.' "

Cora, it turned out, had a partial version of the disorder — biotinidase deficiency — and the deep DNA testing her parents had signed her up for, known as whole-exome sequencing, had clinched the diagnosis.

The partial disorder could have left Cora struggling in school, needing glasses and having trouble hearing, Stetson says. While the family's pediatrician would have been vigilant for such symptoms, "we would have had to wait for those symptoms to arrive, and none of them are reversible," she says. "So if she had bad eyesight, you can't fix it."

Stetson compares her daughter's clear-cut case to her son's frustratingly mysterious eczema when he was an infant. It's a "freeing feeling," she says, to know what's happening with her child and what to do about it — just mix a cheap biotin supplement into her yogurt after dinner every day.

"It just seems crazy to me that in this world that we live in, that we're still on the defensive of our health care system," she says. "We should be on the offensive."

Getting out in front of disease is the ultimate goal of DNA screening like the

BabySeq project, says Dr. Robert Green, its joint director and a Harvard Medical School professor.

"We want to take a medical system that's mostly reactive and all about 'sick care,' and move it to be proactive and all about prevention," he says.

BabySeq, based mainly at Brigham and Women's Hospital and Boston Children's Hospital, is federally funded. It targets genes likely to lead to problems in childhood and that lend themselves to possible action.

In the latest findings, published Thursday in the American Journal of Human Genetics, those mutations turn out to be surprisingly common, turning up in more than 9 percent of babies tested.

That 9 percent of infants are at risk "for what we call monogenic diseases — or diseases that are caused by a change in a single gene — is really an unprecedented and pretty startling finding," Dr. Green says.

The study compares 159 babies who were randomly assigned to have their DNA sequenced, and 157 assigned only to standard newborn screening, a biochemical test performed on a drop of blood.

Among the 15 sequenced babies whose DNA raised a red flag, it was most often for a mutation linked to risky heart conditions or hearing loss. It is not clear whether the disorders linked to the genes will actually develop in each case.

About one in every 10 people has some sort of rare disease, says Dr. Alan Beggs, director of the Manton Center for Orphan Disease Research at Boston Children's Hospital and the paper's senior author.

"However, we don't expect one in 10 people to have a genetic disease with a mutation *that we can find*," he says. So one goal "is to try to understand which of these are actually going to potentially cause disease. And what we can do to help

mitigate the effects of that in the future."

As DNA sequencing becomes more common, BabySeq aims to answer an array of questions: How does it affect a child's care? How does it affect the family? How does it impact the costs of care? What do doctors do with the information?

Whether DNA screening should be done on healthy babies is a topic of lively debate, including concerns that genetic findings may cause anxiety and overtreatment.

The Hastings Center, a bio-ethics think tank, recently put out a report recommending against large-scale programs to test the DNA of healthy babies at this point, and advised doctors to discourage parents from using direct-to-consumer DNA tests on their children.

But the Hastings report acknowledged disagreement over its findings, and Dr. Green points out that the evidence from BabySeq and other studies is only beginning to roll in. He also notes that "early adopters" who volunteer for sequencing tend to be those who want information and can handle the uncertainty of genetic risk findings.

Dr. Beggs sees no need to sequence every healthy baby promptly at birth, he says, but he can imagine it as an early childhood routine like vaccination.

"I do think that sequencing early in life is very useful," he says, "simply because it can help inform on risks that might occur over the first few years of life," such as the development of muscular dystrophy.

In the future, he expects that most children will be sequenced. Dr. Green, too, expects widespread childhood sequencing, though he emphasizes that he does not expect it to be mandatory like the current newborn screening program.

One interesting twist in this latest study: Babies' DNA may help their parents,

too. In three cases, the study reports, infants were found to carry genes linked to breast or other cancer in adulthood. The findings alerted the parents who had passed on the genes.

As for 2-year-old Cora Stetson, when her parents signed up for the BabySeq study, they did discuss the complexities that could arise if the testing turned up a high-risk mutation with potentially serious consequences. They opted for maximal knowledge.

"Again, it goes back to offensive and defensive," Lauren Stetson says. "If I was a parent who knew that my kid was going to be sick, I would want to be on the offensive. I would want to be able to research and know what was coming for us, to be able to create the best life for our kid, which is what all parents want to do."

This segment aired on January 3, 2019.

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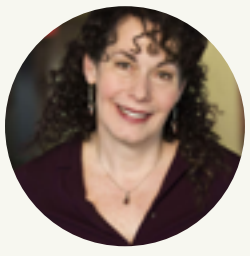

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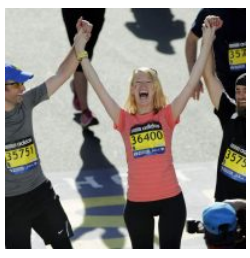

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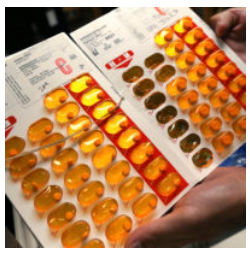

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