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NEWS

Can gene sequencing at birth prevent terrible diseases? Researchers hope so.

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Every baby born in the United States is pricked in the heel shortly after birth. A blood sample is then analyzed to look for one of 20 to 30 inherited diseases.

Early identification of a particular disease means treatment can start right away, potentially saving or extending the child's life.

Now, doctors want to go even further: They want to look not just at blood, but at genes.

A new effort announced Wednesday by a genetic testing company paired with researchers at NewYork-Presbyterian/Columbia University aims to sequence 100,000 newborns in New York City over the next five years.

The sequencing would look for about 250 diseases that strike before age 5 and for which there are treatments or approaches that can make a difference in a child's life.

A similar effort in the United Kingdom is also examining the genes of 100,000 newborns, looking for diseases for which there is a treatment or a cure.

The programs promise to bring treatments to babies before symptoms become obvious and at a time when something can be done to help them.

"The appetite for this is growing. The awareness of this is growing. We all see it as inevitable," said Dr. Robert Green, a medical geneticist at Brigham and Women's Hospital

and Harvard Medical School, both in Boston. "We are grossly underutilizing the life-saving benefits of genetics and we have to get past that."

This week, Green is hosting a conference in Boston, bringing together researchers and industry representatives from the U.S., U.K., European Union and Australia to set standards and discuss the challenges and opportunities presented by scaling up newborn genetic sequencing.

This kind of early sequencing and treatment is possible now for the first time because of dramatic advances in diagnostics, therapies and digital data storage, as well as a reduction in the cost of sequencing, said Dr. Paul Kruszka, a clinical geneticist and chief medical officer of GeneDx at Sema4, which is leading the new program.

"We're entering the therapeutic era and leaving the diagnostic era," Kruszka said. "This potentially has the opportunity to change the way we practice medicine especially in rare disease."

Right now, families with rare diseases often search for a diagnosis for 5, 10 or even 20 years. If the child could be diagnosed at birth, he said, it would short-circuit that process and treatment could begin much earlier – hopefully before the child suffers irreversible damage.

Before deciding whether every family should get access to genetic sequencing for their newborn, large studies like Sema4's are needed to justify the cost, Kruszka said.

The price of gene sequencing has dropped precipitiously, with one company, Illumina. announcing last week that its newest-generation sequencing machines can run a complete sequence for about $200. Kruszka said Sema4 expects to still pay about $1,000 for each sequence of all 20,000 genes.

Gene sequencing at birth should be able to save money over the child's lifetime by preventing illness, Green said. The costs of sequencing are limited, he said, but the benefits will build up over the child's lifetime and may help family members, too.

Green and his team began analyzing the genetic sequences of newborns in 2013, and has found lots of useful information among the first 320 babies sequenced, he said. He now has funding to expand his sequencing research to 1,000 newborns.

Large numbers are essential because most of the diseases being diagnosed are extremely rare.

Convincing parents to participate in a sequencing research trial "is not easy," Green said. Many are concerned about privacy and the discrimination their child might face if their genome were made public. And it can be a unpleasant for parents to consider the horrible diseases their perfect newborn might be harboring, he said.

"You've gone through all this pregancy and you're sitting there with a healthy baby (and I'm) offering you the opportunity to find out something that's devastating and terrifying," he said. "How fun is that?"

He doesn't think privacy needs to be a major parental concern. Companies can learn more useful information by tracking someone's cell phone or credit card than their genome and most common diseases are the result of many combinations of genes.

"Many people hear 'genetics' and worry somehow that that is a special kind of privacy threat," he said, adding that he doesn't think there is. "We haven't been paying attention to the medical benefits of genetic testing, particularly predictive genetic testing."

if people don't want to know, that's okay, too, Green said. "We can respect people who don't want to know, but as also respect people who do want to know," he said. "Some families will say 'I treasure the precious ignorance.' Others will say 'If I could have known, I would have poured my heart and soul into clinical trials or spent more time with the child when she was healthy."

In a five-year review of their research, Green and his colleagues found that "terrible things didn't happen" when they sequenced newborn genomes.

Families, he said, "did not in fact have downstream distress," he said. "They did have appropriate medical follow-up and that there were amazing benefits to the babies and the

families as a result of the surveillance and treatment."

The baby sequencing identified several parents who had inherited illnesses and received risk-reducing surgery, he said, as well as a baby who had a narrowed aorta that wouldn't have been detected if its genetics hadn't indicated the need for an echocardiogram.

"Even in a small sample we found much to act on," he said.

At Rady Children's Hospital in San Diego, they're trying to rapidly sequence the genomes of babies who already have problems and are being treated in one of 83 children's hospitals across Canada and the U.S.

Every morning, samples arrive by Fedex. In some cases, the baby is in such dire shape than an answer is needed immediately. For those children, "we've got to drop what we're doing and go," said Dr. Stephen Kingsmore, the president and CEO of Rady's Institute for Genomic Medicine. "Even a day can cost a child's life or brain function."

For babies who are stable, sequencing still happens rapidly, but a little less so. "Every sample gets onto a sequencer the same day," he said.

So far, the institute, which is also collaborating on a newborn sequencing study in Greece, has been able to provide a 1,500 children with a diagnosis in the first weeks of life in addition to a life-saving treatment.

"That idea, that future is where a child never experiences a sick day, even though they have a fatal condition," the institute's former director of marketing, Graciela Sevilla, said earlier this year. "We'd love to see that on a regular basis."

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