Rewriting Life

Should Babies Have Their Genomes Sequenced?

The BabySeq project in Boston has begun collecting data to quantify the risks and benefits of DNA sequencing at birth.

by Anna Nowogrodzki July 2, 2015

Genomic sequencing is cheaper and faster than ever, and it helps doctors identify patients with family risk or rare diseases, but we have very little data on the benefits and harms of sequencing in healthy people.

healthy people.

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For 51 years, newborn babies have gotten a heel-prick test in which their blood is screened for dozens of congenital disorders. Routine newborn screening has basically eliminated the risk of death or irreversible brain damage that some of these disorders can pose if they are not identified right away.

Now some

researchers in Boston are trying to find out if genomic sequencing at birth would be as successful.

The BabySeq project is the first randomized, controlled trial to

measure the harms and benefits of newborn genomic sequencing. One of four NIH-funded projects granted a total of $25 million to examine genomic sequencing in newborns, BabySeq recently enrolled its first four subjects, three healthy babies and one baby from the neonatal ICU. The researchers got the first baby's genomic sequence data last week.

The central question for this project is what will come of giving genomic information to parents and their baby's doctor. Will doctors order more tests and interventions? Will those tests and interventions make babies healthier? Or will they just waste money, or even end up doing more harm than good?

As a randomized, controlled trial, BabySeq meets the gold standard for clinical study design. Eventually, it will enroll 240 healthy babies at Brigham and Women's Hospital and 240 babies in the neonatal ICU at Boston Children's Hospital. Half the babies in each group will be randomly assigned to have all their coding DNA

to have all their coding DNA sequenced and screened for 1,700

variants of genes that are associated with childhood-onset diseases. The other half will not have their DNA sequenced. The study will track the effects of the sequencing on the babies' health care, its costs, their parents' attitudes, and parent-child bonding.

"There is no scientific consensus that it is appropriate or useful to sequence healthy individuals," says Robert Green, co-leader of the trial. "Therefore, the only way this would be considered a public health mandate would be if we had a tremendous amount of evidence that sequencing a large number of people would be beneficial."

Whatever they find, positive or negative, will be important for the future of population-wide genomic sequencing. "I think it starts asking the right questions," says Muin Khoury, director of the CDC's Office of Public Health Genomics. "The whole ethical ramification of this is that newborns

ramification of this is that newborns have no choice to make, so parents have to make that decision for them."

BabySeq is funded through 2018, when the babies who have enrolled so far will be at most three years old, but if the team can secure longer-term funding, they will seek assent from the children when they turn 13, and consent at age 18.

For some of the diseases the study will examine, there are actions physicians and parents can take to reduce a child's risk, but for others there is little to be done. For example, BabySeq will screen for a gene variant associated with childhood-onset colon cancer; in that case, regular screening during childhood could allow early detection and successful treatment. On the other hand, BabySeq will also return results for a gene variant associated with Rett syndrome, which halts a child's development at six to 18 months and has no cure (treatments can help with some of the symptoms, such as seizures).

seizures).

BabySeq will only screen for gene variants that are linked to childhood-onset conditions, so if a baby had a gene variant associated with, for example, Alzheimer's disease, that information would remain unknown.

Green recognizes that there are many questions about whether and how to use genomic data in medicine, and he says BabySeq is an attempt "to grapple with as many of those questions as we can." For example, how accurate is the information revealed by a gene? How can we prevent doctors and families from misunderstanding genomic data? What about insurance discrimination? Will genomic information affect parent-child bonding—for example, would the knowledge that a child is predisposed to a developmental disability cause parents to underestimate that child's abilities?

Screening can have benefits, of course, but also risks. For example, screening for breast and prostate cancer can find lumps that are riskier to remove than

lumps that are riskier to remove than to leave alone, but once patients and

doctors know they're there, sometimes they are set on carrying out the risky interventions anyway.

For BabySeq's data to be broadly relevant, the participants should reflect the diversity of the general U.S. population. Because the study has only enrolled a few participants, however, it's too early to know if they are representative in terms of their racial, educational, and socioeconomic makeup.

Green says he would probably have his newborn's genome sequenced, if he had one (his children are in their 20s), "but I think the more important point is that we're very much talking about a model where people have that choice, and many people would choose differently for perfectly legitimate reasons."

Khoury doesn't believe newborn genomic sequencing will be generally

available soon, even as a choice. "I'd love to see more studies done first," he says. "I would see more sequencing done first in adults, and first in adults who are sick with genetic diseases. And pending the results of studies, I would see newborns sequenced last."

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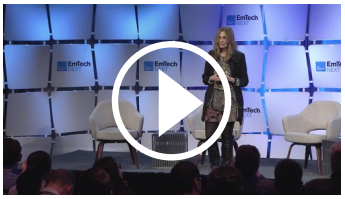

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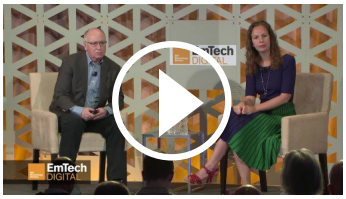

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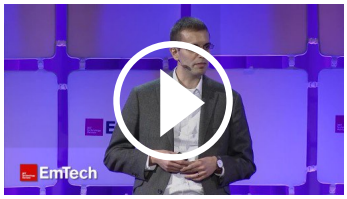

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