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Geneticists are studying the risks and benefits of sequencing the genome of every newborn. UZFOTO/SHUTTERSTOCK

Surprisingly few new parents enlist in study to have baby's genome sequenced

By **Jocelyn****Kaiser** | Oct. 19, 2016 , 12:45 PM

One of the first studies to

explore the idea of routinely sequencing the genes of newborns to help guide their health care has run into an unexpected road bump: Few

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parents approached are interested in having their baby's genome profiled.

When Robert Green, a geneticist at the Harvard University–affiliated Brigham and Women's Hospital in Boston, and co-workers began planning to sequence babies about 4 years ago, they surveyed more than 500 parents of healthy newborns. Nearly half declared they would be “very” or “extremely” interested and another 37% said “somewhat.” But since their actual

BabySeq Project began last year in May, only about 7% of more than 2400 couples approached so far have agreed to participate, says Green, who co-leads BabySeq with Alan Beggs of Boston Children's Hospital. That “very surprising” figure is the same both for parents of very sick infants and those with healthy babies, he adds.

BabySeq is one of four

projects funded by the National Institutes of Health (NIH) 3 years ago **to probe the risks and benefits of sequencing newborns' DNA**

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and compare the results to conventional newborn disease screening using biochemical analysis of blood spots. These studies got a slow start because the U.S. Food and Drug Administration decided some of the genome tests had to go through regulatory review.

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The BabySeq team is analyzing the newborn's protein-coding DNA for mutations in roughly 7000 genes implicated in childhood diseases or drug metabolism. Yet the parents of only 24 of 345 sick babies in neonatal intensive care, and 138 of 2062 healthy babies, have agreed to join BabySeq, Green is reporting today at the American Society of Human Genetics's (ASHG's) annual meeting in Vancouver, Canada.

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Many who declined cited logistics: They were apparently put off by having to return to the hospital with their newborns to discuss the sequencing results. Others who met with a genetic counselor before being invited to join the study had concerns such as privacy, receiving negative or unclear results, and insurance discrimination. Although U.S. federal law prohibits health insurers from denying coverage based on genetic data, the children could be denied life, disability, or long-term care insurance. "A lot of that is literally 50 years in the future and they are concerned," Green says.

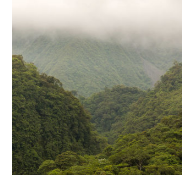
Yet another factor may be that the Boston group approaches parents soon after their babies are born, when they may be overwhelmed, says geneticist Cynthia Powell at the University of North Carolina in Chapel Hill. Her newborn sequencing

study, part of the NIH-funded quartet, enlists many parents during prenatal clinical visits. Although that study only began in April, so far more

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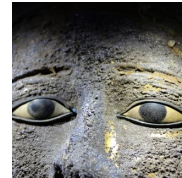
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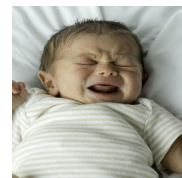
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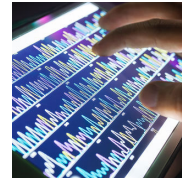
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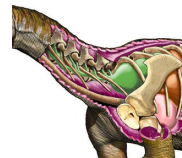
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than half the 50 or so families approached have agreed to participate, she says. (The other two NIH studies are further along—one focuses only on sequencing very sick newborns, whereas the second is mainly examining the DNA in archived bloodspots.)

Only a few newborns tested so far in BabySeq carry mutations in genes expected to make them sick, Green says. At least two have inherited mutations implicated in heart disease but no signs of illness in the parents or child; Green's team plans to study whether knowing about such mutations is beneficial for the family in the long run, or results in unnecessary anxiety and tests. Another baby had mutations that can cause an enzyme deficiency; and although asymptomatic, the baby has slightly below normal levels of the enzyme and is being treated. And a fifth child carried a mutation in the *BRCA2* gene, which is linked to breast cancer. Green's team initially did not plan to tell families about mutations linked to adult-

onset diseases but asked the study's ethics board whether they could disclose the *BRCA2* result. The mother "was obviously concerned but grateful," Green says.

ASHG has taken the position that only newborns with undiagnosed illnesses should undergo genome sequencing—and even then, analyzing only genes likely to explain the disorder may be preferable. Making genome sequencing part of routine newborn screening is "a dreadful idea," says pediatrician and ethicist Jeffrey Botkin of the University of Utah in Salt Lake City.

Genomics policy expert Misha Angrist of Duke University in Durham, North Carolina, thinks the appeal may grow. BabySeq is "an important proof of concept," he says. "These are still very early days—if more people do this and the discrimination and

confidentiality risks do not materialize, then presumably more people will choose newborn sequencing."

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