

PRESS BRIEF: RELEASED SEPTEMBER 12, 2018 7PM ET

Barriers to Entry: Study Explores Why Parents Declined Genome Sequencing of Their Newborns

The BabySeq Project reports that although more than 80 percent of approached mothers and fathers declined an offer for free genomic sequencing of their newborn, more than half of those parents were not interested in any research participation

Imagine that you are in the hospital the day after your child is born. If a researcher approached you and asked if your family would like to enroll in a study in which your newborn's genome might be sequenced, would you accept? Brigham and Women's Hospital and Boston Children's Hospital investigators for the NIH-funded BabySeq Project report that of the more than 3,800 families they approached, only 268 ultimately enrolled in their study to evaluate the medical, behavioral and economic outcomes of newborns provided an opportunity for free genomic sequencing prior to discharge from their birth hospital. In a paper published in *Genetics in Medicine*, the team reports on the reasons for the high rate of decline, many of which had less to do with genetics or privacy concerns than with a low interest in research in general as well as dealing with the logistical challenges of participating in a complex research study when offered immediately after the birth of a child.

“We found that families declined the opportunity to have their newborn sequenced for various reasons, many having nothing to do with genetics, but rather because of exhaustion, stress or simply not wanting to participate in any research,” said senior author Richard Parad, MD, MPH, director of the Newborn Genomic Medicine Program in Brigham's Department of Pediatric Newborn Medicine. “To continue advancing evidence-based care of newborns, it is critical to be able to enroll families in research studies around the time of birth. Of course, we understand that we are approaching them at a very challenging and overwhelming time.”

The BabySeq Project is the first randomized, controlled clinical trial of sequencing in both healthy and sick newborns, with half of enrolled families receiving standard newborn screening (including the state-mandated “heel prick” test that screens for a limited set of genetic conditions) and half receiving whole-exome sequencing. For the latter group, the BabySeq team returns genetic results related to childhood onset conditions, both carrier status and disease risk. A small group of actionable, adult onset conditions such as hereditary cancers that can be screened for early, are also assessed. Parents who enrolled had an additional blood sample drawn from their newborns, and agreed to return to the hospital to receive results as well as complete follow-up surveys over the first year of life.

“This study is the first of its kind to offer genomic sequencing to both healthy and sick newborns,” says Casie Genetti, MS, CGC, a genetic counselor in the Manton Center for Orphan Disease Research at Boston Children's Hospital and first author on the paper. “In speaking with parents, we have learned a great deal about what motivates some families to participate, but also documented the concerns, like genetic discrimination and uncertainty around the results, that make many of them hesitate to join such research.”

Of the 3,860 families approached, only 402 agreed to attend an enrollment session to hear more about participating in the project. The team surveyed more than 1,700 declining families to explore their reasoning. More than 58 percent responded that they were “not interested in any research.” Of parents who provided reasons for specifically not participating in the BabySeq Project, over 40 percent cited logistical concerns, such as the inconvenience of returning for the research visit, 15 percent reported feeling too overwhelmed to make a decision, 15 percent had concerns about privacy and insurance discrimination issues and 12 percent anticipated feeling uncomfortable with receiving either uncertain or bad news results.

Of the 402 families who agreed to hear about the details of the study, 67 percent enrolled. The enrollment session provided an overview of the study logistics, genomic testing, results to be returned and stored in the baby’s medical record and described protections for genetic privacy and against genetic discrimination. Interestingly, families that declined to enroll after this session cited different reasons for decline than those who declined on initial approach, more commonly citing discomfort about the potential results they could receive as well as concerns about privacy and insurance discrimination.

“The BabySeq Project is one of the rare genomics studies to begin its recruitment opportunity with a truly representative population of new parents,” said Robert Green, MD, MPH, Professor of Medicine in the Division of Genetics at BWH and Harvard Medical School, and co-director of the study with Alan Beggs, PhD, Professor of Pediatrics at Boston Children’s Hospital. “These results remind us how difficult it is to enroll truly representative samples into any form of research, and how especially difficult it is to enroll new parents who are stressed and fatigued immediately after birth. But it also reassures us that families were fully capable of asserting their consent in this vulnerable moment, and reminds us that genomics research cannot fully flourish until we address societal considerations like protecting participants from future life insurance discrimination.”

Early results around the medical, behavioral and economic outcomes of the BabySeq Project will be presented at the upcoming American Society for Human Genetics Annual Meeting in October.

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Paper cited: Genetti, C *et al.* “Parental interest in genomic sequencing of newborns: enrollment experience from the BabySeq Project.” *Genetics in Medicine*