

# Newborn genomic sequencing detects unanticipated disease risk factors

January 3 2019

---



Credit: CC0 Public Domain

As genomic sequencing becomes increasingly commonplace in the clinic, questions remain about its use and role among newborns. Can sequencing provide actionable insights? How common is it to find something important to a child's future health? What benefits or consequences will sequencing have for families? The BabySeq Project, a

joint endeavor led by investigators at Brigham and Women's Hospital and Boston Children's Hospital, with collaborators at the Baylor College of Medicine, is revealing the answers to these questions and more. In a paper published in the *American Journal of Human Genetics*, the research team reports that genomic sequencing can identify risk for a wide range of disorders that may not be detected otherwise. Importantly, early knowledge about several of these conditions can lead to surveillance and interventions that could improve health outcomes for newborns and their families.

"The BabySeq Project is the first randomized trial of sequencing in newborns and the first study to fully examine the wealth of unanticipated genetic risk information in children," said Robert Green, MD, MPH, co-director of the study at the Brigham and a professor at Harvard Medical School. "We were stunned by the number of babies with unanticipated genetic findings that could lead to disease prevention in the future."

The BabySeq Project enrolled healthy newborns from the Brigham's well baby nursery and ill newborns from BCH, the Brigham and Massachusetts General Hospital's neonatal and pediatric intensive care units. Family histories were collected for all enrolled participants. Half of the families from each group were randomized to receive standard care, including "heel prick" newborn screening which tests for about 30 genetic conditions, and genetic counseling based on family history; the other half received whole exome sequencing in addition to standard care and genetic counseling. Sequencing results were disclosed to families during the genetic counseling session. The team monitored health outcomes and collected medical, behavioral and [economic data](#) at the time of disclosure, three months later and 10 months later to measure the impact of genomic sequencing on the infant's clinical care, parent and clinician behaviors, and economic outcomes.

A total of 159 newborns (128 healthy newborns and 31 ill newborns)

were randomized to receive [genomic sequencing](#). Of those, 15 (9.4 percent) were found to have a genetic variant for which there was strong evidence of increased risk of a disorder that presents or is clinically manageable during childhood, or a variant in a gene for which there was moderate evidence of risk but for which an intervention during childhood might prevent devastating outcomes later in life.

The team found variants associated with several heart conditions, including six newborns with variants associated with dilated or hypertrophic cardiomyopathy and another newborn with a variant associated with supra-aortic stenosis. These conditions can be monitored over time, and families have been referred to cardiac specialists. Another newborn was found to have a risk variant for biotinidase deficiency. Further testing determined that the infant had partial biotinidase deficiency, a condition that can cause skin rash, hair loss and seizures. The child's diet is now being supplemented with biotin, which is expected to prevent any disease manifestations.

"Sequencing results have potential to raise questions that may be upsetting for parents but could also lead to helpful or even lifesaving interventions," said senior author and BabySeq co-director Alan Beggs, Ph.D., director of The Manton Center of Orphan Disease Research at Boston Children's Hospital. "Only time will tell how the costs—both financial and in terms of extra medical testing and [family](#) stress—balance out against the benefits. That's what we're really trying to find out."

The team also offered to provide information regarding their child's risk for medically actionable, adult onset conditions. Three of 85 infants of parents who agreed to receive this information carried such variants. These variants were also identified in the mothers of the three children.

"Disclosing genetic risk for adult onset conditions in children has been

discouraged in traditional genetics in order to protect the child's right 'not to know,' but our results demonstrate that many parents want access to this information about their child," said Green. "Our findings suggest that sequencing newborns thoroughly reveals potentially life-saving information in both infants and their parents far more commonly than was previously thought, and should encourage our entire field to re-evaluate the value of comprehensively analyzing and disclosing genomic information at any age."

**More information:** Ceyhan-Birsoy, O et al. "Interpretation of genomic sequencing results in healthy and ill newborns: Results from the BabySeq Project" *American Journal of Human Genetics* [DOI: 10.1016/j.ajhg.2018.11.016](https://doi.org/10.1016/j.ajhg.2018.11.016)

Provided by Brigham and Women's Hospital

Citation: Newborn genomic sequencing detects unanticipated disease risk factors (2019, January 3) retrieved 27 April 2024 from <https://medicalxpress.com/news/2019-01-newborn-genomic-sequencing-unanticipated-disease.html>

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.