Newborns' genomes shouldn't be tested for adult-onset conditions

November 13 2019

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In the last decade, genome sequencing has provided many people with a wealth of information about risks for certain diseases.

But should parents be able to get all of the sequence results of their children's genomes, especially if the variants would not cause illness or symptoms until adulthood?

No, says a University of Chicago medical ethicist, who recently examined the issue through the lens of one newborn genetic sequencing case study gone awry.

"The more we learn about genetics, the more we learn how non-

definitive it is," said Lainie Ross, MD, Ph.D., the Carolyn and Matthew Bucksbaum Professor of Clinical Medical Ethics and co-author of a new paper in the journal *Pediatrics*. "By sequencing newborns for adult conditions, you are creating 'patients in waiting.' They will just wait for the other shoe to drop, and it might not drop for decades, if ever."

At issue is a dispute that arose within the BabySeq Project (Genome Sequence-Based Screening for Childhood Risk and Newborn Illness). As part of the National Institutes of Health-funded study, a group of researchers wanted to sequence genomes of newborns to explore the medical, behavioral and economic impacts of integrating genomic sequencing into their care.

The researchers told parents they would screen for variants in genes that were linked to childhood-onset diseases, but one infant's screening showed a BRCA2 mutation, which is linked to an increased chance of breast and ovarian cancer. Because this is an adult-onset condition, the researchers returned to the Institutional Review Board and requested permission to give this information to the parents. Such information could benefit the child, they argued, because it might indicate that a parent also had this mutation. That information could potentially help keep the child's parent alive and well.

But that argument doesn't keep the best interest of the child at heart, Ross argues.

"Newborn screening should focus on what's best for the infant," she said. "You shouldn't use children as canaries in the coal mine for their parents. If you want to know what could affect the parents, test the mother and father."

In fact, there is a general consensus among the medical community that children should not be tested for adult-onset-only conditions, but the

American College of Medical Genetics and Genomics recommended in 2013 that all subjects who are clinically sequenced be tested for 56 gene variants linked to diseases. (The number has since increased to 59).

That included children, which brought up a whole host of issues: Do parents have a right to know or own their children's genetic data? And should children be tested for these genes, many of which are linked to adult-onset conditions?

In the case of the BabySeq Project study, which happened in 2014, Ross argues that the researchers shouldn't have tested for adult-onset conditions to begin with, and that all pediatric studies should avoid this conundrum by simply not testing for these conditions.

The American College of Medical Genetics and Genomics has since changed its guidelines to allow patients to opt out of this screening, and that's a step in the right direction, Ross said, since genetic medicine is still an evolving field. Recent studies show that some of the 59 genes identified by the American College of Medical Genetics and Genomics might not be associated with increased risk for people in some ethnic minority communities. That means genetic testing could lead to unnecessary anxiety and treatment, especially for a child who has decades before the disease could emerge.

And children often don't have the choice to opt out. That doesn't mean parents shouldn't allow genetic sequencing on their children altogether, Ross said, but it should be limited to conditions that affect them as children.

"Parents have a right to make health care decisions for their kids," she said. "But it stops when the child becomes an adult, or with health information that will affect them only as an adult."

More information: Lainie Friedman Ross et al. Ethical Issues in Newborn Sequencing Research: The Case Study of BabySeq, *Pediatrics* (2019). DOI: 10.1542/peds.2019-1031