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BabySeq, MedSeq projects reveal how many people carry rare disease genetic risk variants

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Two projects in which healthy individuals have had their genomes sequenced have revealed that searching for unanticipated genetic results in newborns and adults can unearth far more variants associated with diseases than previously thought, and, importantly, reveal previously unrecognized but related clinical features of genetic conditions. Results from both the BabySeq Project, led by investigators at Brigham and Women's Hospital and Boston Children's Hospital, and the MedSeq Project, led by investigators at the Brigham, are being presented together at the 2018 American Society for Human Genetics meeting.

"These results are unexpected and exciting, suggesting that if we examine enough well-established, disease-associated genes, we will unearth monogenic risk variants in more than 10 percent of purportedly healthy individuals," said Robert Green, MD, MPH, a principal investigator on both projects and a clinical geneticist at BWH. "And if on the basis of these genetic clues, we carefully examine those individuals, we find that a quarter of them have previously unrecognized features of underlying disease—something that we might never have realized had we not performed genetic sequencing."

In the MedSeq and BabySeq Projects, Green and colleagues analyzed approximately 5,000 disease-associated genes to identify monogenic disease risk variants—mutations associated with diseases thought to be caused by variants in a single gene. Examples of such monogenic diseases include hereditary cancer syndromes, hereditary cardiac syndromes and metabolic disorders that can lead to life-threatening

complications.

Genome or exome sequencing was performed on a total of 269 individuals. Unanticipated risk variants were found in 16 of 110 adults (14.5 percent) and 18 of 159 infants (11.3 percent). When individuals were closely examined in the clinic, the team discovered clinical features suggestive of a previously unrecognized genetic condition in four of the adult cases and four of the newborn cases, including three cases with highly effective interventions. An additional newborn case provided an unsuspected molecular diagnosis for an observed clinical condition that had not been previously considered genetic.

The MedSeq and BabySeq Projects, both funded by the National Institutes of Health, explore the ramifications of using genome sequencing in apparently healthy adults and infants. Both are randomized clinical trials designed to explore the medical, behavioral and economic impacts of incorporating genome sequencing into everyday medicine.

Provided by Brigham and Women's Hospital

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