Researchers study costs of integrating genetic sequencing into clinical care

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Integrating whole genome sequencing into primary care and heart disease care is unlikely to substantially increase the costs of health care utilization and follow-up tests, according to research presented at the American Society of Human Genetics (ASHG) 2015 Annual Meeting in Baltimore.

"This finding helps to allay the worry that patients whose genomes are sequenced will run out and spend thousands of dollars on follow-up tests and care, which may or may not improve their eventual health outcomes," said Jason L. Vassy, MD, MPH, a primary care physician and co-investigator on the study at Brigham and Women's Hospital.

The cost analysis was conducted as part of the MedSeq Project, a broader study of the best ways to integrate genome sequencing into clinical medicine and how these might impact health systems and individuals. The project has enrolled a total of 200 patients: 100 healthy adults in primary care settings and 100 adults receiving care for complex cardiomyopathy, a type of heart disease. Half of the patients in each group were randomly assigned to receive the standard of care, which included a discussion of family history of disease, and the other half received the standard of care plus whole genome sequencing, a personalized Genome Report, and a discussion of the results. Results from 108 of the 200 patients, 70 in the primary care group and 38 in the cardiology group, were included in the preliminary cost analysis presented at the meeting.

On average, patients whose genomes were sequenced incurred a cost of $719 in follow-up tests and care over the following year, including out-of-pocket expenses, while standard treatment and follow-up averaged $430 per patient.

"This difference was not statistically significant in our study, but more importantly, it was far smaller than some may have feared," said Dr. Vassy. Costs were calculated using Medicare reimbursement schedules for the care received, and did not include the laboratory costs of sequencing patients' genomes and assembling a report of the results. On average, the cost of sequencing itself was $6,923 per patient, which included the initial test, confirmation and interpretation of results, and time spent conveying them to the patient.

"Genome sequencing and analysis is not inexpensive, but by identifying risk factors for health conditions that enable prevention or early treatment, it may offset future costs," said Dmitry Dukhovny, MD, MPH, Assistant Professor of Pediatrics at Oregon Health & Science University and first author on the study. "However, this study cannot yet address genome sequencing's benefits to cost and health, which will take place over the course of a lifetime," he added.

Beyond the laboratory costs of sequencing, sorting through genetic data to isolate the most important results adds to its cost. Laboratories do not currently follow a standard format for reporting genetic test results, which means that the development of a Genome Report for each patient is not an automated process and may entail a search through the scientific literature. However, as more genetic variants are added to the Genome Report's database, the incremental cost per patient is expected to decrease.

"Regardless of our findings," Dr. Vassy said, "the fact remains that health care costs money. A more meaningful question is what you get for

that price and whether it's worth it."

"This study gets at the definition of cost-effectiveness," Dr. Dukhovny added. "In this case, we will want to assess the total value gained by spending on sequencing, analysis, and counseling patients, which includes future cost savings as well as less tangible improvements to health and quality of life."

Provided by American Society of Human Genetics

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