MedSeq Study Finds Whole-Genome Sequencing Does Not Increase Medical Costs in Short Term

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NEW YORK (GenomeWeb) – Whole-genome sequencing does not dramatically increase follow-up clinical costs for healthy individuals or cardiology patients, at least over the first six months after results are returned, according to research by the MedSeq Project, led by Brigham and Women's Hospital and Harvard Medical School.

"Our data provide reassurance that physicians seem to be responding responsibly and that we're not seeing evidence of dramatically increased downstream spending," first author Kurt Christensen, a genetics researcher at BWH and Harvard, said in a statement.

As part of a pilot study, Christensen and colleagues turned to medical record data, patient surveys, and other approaches to tally up participants' healthcare costs in the six months after their whole-genome sequencing results were returned, along with a family history report. Compared with individuals randomized to receive just a family history review and no sequencing, they did not see a statistically significant increase in healthcare costs for genome-sequenced individuals.

Christensen presented similar results at the American Society of Human Genetics annual meeting in 2015. The latest findings were published online today in Genetics in Medicine.

"Though there are limitations to our pilot study, our work provides novel and much-needed data to help decision makers begin to understand the short-term cost implications of integrating whole-genome sequencing into clinical care," Christensen said, "and provides insight about what data are needed to provide more clarity about the economic implications of this technology."

For several years, MedSeq investigators have been comparing consequences, costs, clinical outcomes, and more in 200 participants randomized to receive a family history review alone or in combination with whole-genome sequence data. Half of the participants belong to a seemingly healthy primary care cohort and the other half includes cardiology patients diagnosed with cardiomyopathy.

Past results from MedSeq and other efforts to sequence symptom-free individuals suggest that a significant subset of healthy individuals carry clinically actionable mutations in disease-related genes, highlighting the potential need for follow-up testing and healthcare visits in the post-sequencing period.

In the new study, the researchers put the initial cost of whole-genome sequencing at $5,222 per individual in the primary care cohort, based on reagent use, personnel time, and the like. The price per genome sequence was similar in the cardiology group, they reported, coming in at $5,268 apiece.

When it came to post-sequencing costs, meanwhile, the team reported, the overall cost of downstream testing and healthcare services over six months post-disclosure was comparable in the control and genome-sequenced individuals.

On the primary care side, for example, follow-up health spending increased by an average of $681 per person in the genome-sequenced group relative to the control group but differed a lot between individuals. The cost difference between cardiology cases and controls was more pronounced. There, the cost per person was reportedly $1,560 lower in the genome sequencing arm on average, again differing a lot between patients. Cost differences did not reach statistical significance in either cohort.

In both the primary care and cardiology groups, the team saw an uptick in healthcare visits and related costs in the post-disclosure period compared with the months that preceded disclosures. Again, though, these differences were deemed non-significant.

"This study demonstrates the power of a randomized trial where outcomes can be compared between those who were sequenced and those who were not sequenced but were followed in exactly the same way," BWH and Harvard researcher Robert Green, a MedSeq leader and the study's senior author, said in a statement. "Our challenge now is to replicate these findings in a larger sample size and over a longer period of time."

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