Does genome sequencing increase downstream costs?

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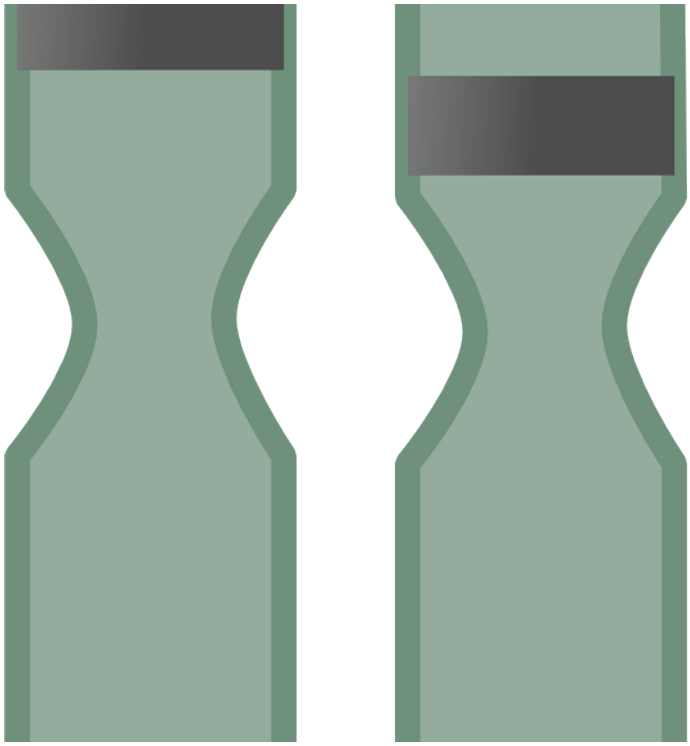

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As genome sequencing enters the clinic, fears have arisen about its potential to motivate follow-up testing and ongoing screening that could drastically increase health care spending. But few studies have quantified the downstream costs of returning genetic information to patients, especially ostensibly healthy patients. The MedSeq Project, led by

investigators at Brigham Women's Hospital, is the first randomized trial to provide whole genome sequencing to both presumably healthy patients as well as those with a known cardiology issue. As part of this pilot study, the project analyzed both the immediate costs of sequencing itself as well as downstream spending six months after genetic information was returned to physicians and their patients. The research team found that downstream costs did not significantly differ between patients who had received whole genome sequencing and those that did not. The team's findings are reported today in *Genetics in Medicine*, the peer-reviewed journal of the American College of Medical Genetics and Genomics.

"Whole genome sequencing is coming of age, but there's fear that with these advancements will come rocketing health care costs," said lead author Kurt Christensen, MPH, PhD, an instructor of medicine in the Division of Genetics at BWH. "Our pilot study is the first to provide insights into the cost of integrating whole genome sequencing into the everyday practice of medicine. Our data provide reassurance that physicians seem to be responding responsibly and that we're not seeing evidence of dramatically increased downstream spending."

The MedSeq Project includes 100 healthy primary care patients and 100 cardiology patients with cardiomyopathy diagnoses. Patients in the control arm of the study received a review of their family history; patients in the experimental arm of the trial received a review of their family history plus a whole genome sequencing analysis and report. For each participant, more than 4,600 genes associated with genetic disease were analyzed, and findings also included information about carrier status, drug implications and risk for diseases associated with multiple genetic markers.

Using data from patients' medical records as well as services reported in patient surveys, the team tracked health care costs incurred in the six

months after patients received their results. For both primary care and cardiology patients, there was no significant difference in the average downstream costs for patients in the control group versus patients who received whole genome sequencing. For cardiology patients, the average cost for the control group was $10,838 and $8,492 for the sequencing group. For primary care patients, the average costs for the control group was $3,175 and $3,566 for the sequencing group.

Sequencing did identify health-relevant genetic information in a large majority of participants, including diagnosis-related variants in half of sequenced cardiology patients and additional disease risks in eight cardiology patients and 13 primary care patients.

Robert Green, MD, MPH, professor of medicine at BWH and Harvard Medical School and a senior author on the paper noted: "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. Observational studies where sequencing is offered to anyone who wishes it cannot achieve the same methodological rigor. Our challenge now is to replicate these findings in a larger sample size and over a longer period of time."

The average cost per patient for sequencing itself was a little more than $5,000 for both cardiology and primary care patients, a total that includes the costs of sequencing, interpretation and disclosure.

While it's important to understand the short-term and immediate costs of sequencing, the team notes that six months may be too short a period to observe the full impact of sequencing on costs and health benefits. They plan to extend their study to follow patients for five years.

"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, and provides insight about what data are needed to provide more clarity about the economic implications of this technology," said Christensen.

Provided by Brigham and Women's Hospital