STAT

Sequencing patients' genomes might not break the health care bank, study finds

By Sharon Begley @sxbegle

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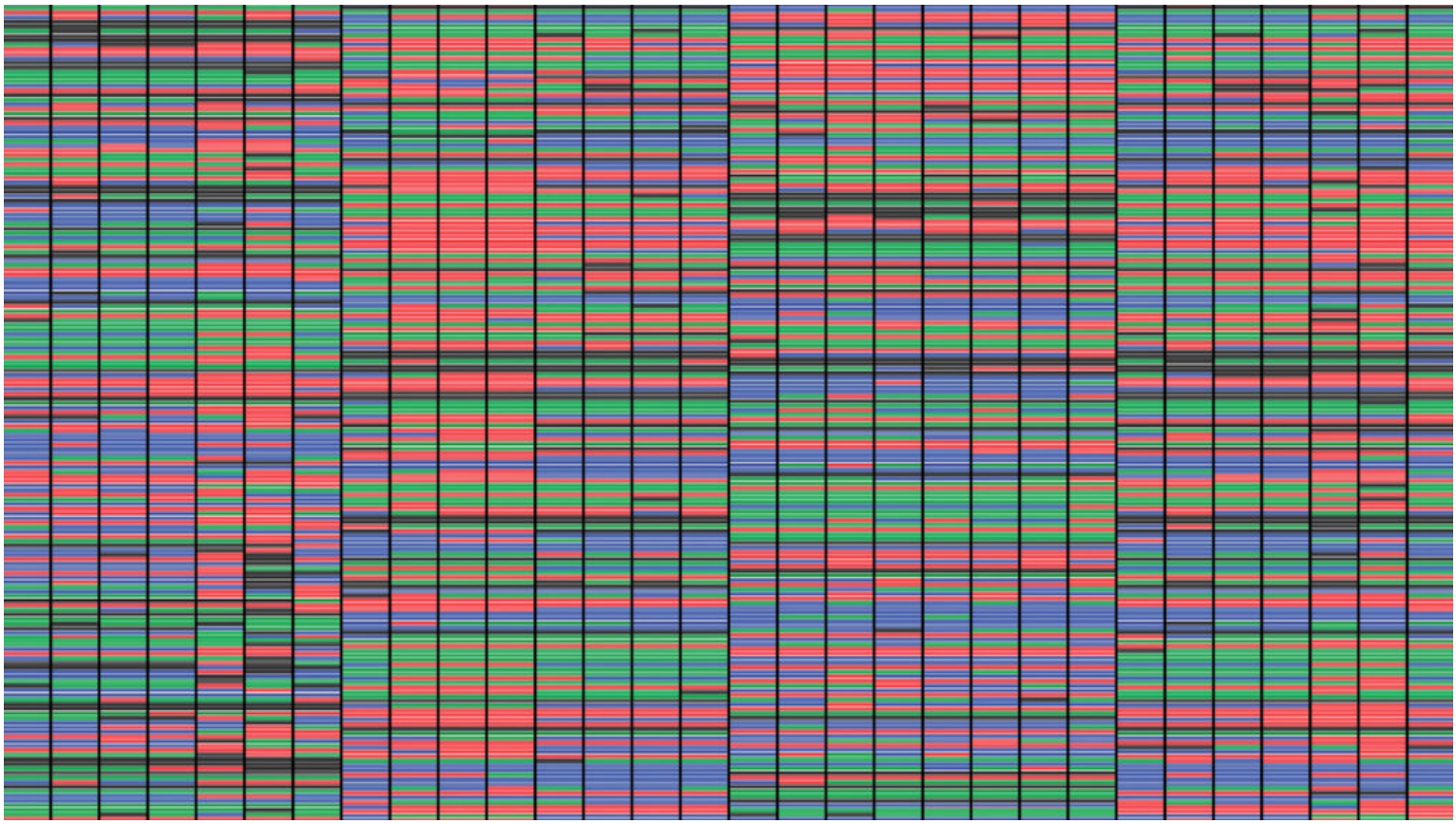

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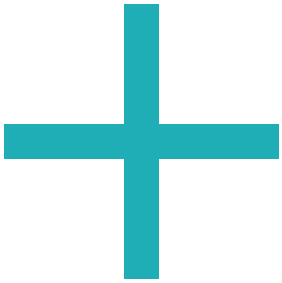
The first rigorous study of its kind finds that sequencing people's genomes[1] might not lead to extensive and expensive follow-up care, researchers reported on Thursday.

"That's been the critical question that has the field [of medical genetics] very concerned: Will whole-genome sequencing drive up downstream costs?" said Kurt Christensen of Brigham and Women's Hospital in Boston, lead author of the study[2] in Genetics in Medicine.

To find out, he and his colleagues recruited 100 healthy adults and 100 patients with cardiomyopathy, assigning roughly half of each group to undergo whole-genome sequencing and the other half to have a review of their family medical history. The researchers followed the volunteers for six months after they and their physicians received the sequencing or family history results, reasoning that if people were told they had a genetic variant that increased the chance of having, say, a rare liver ailment, then they would

seek further testing right away.

Over that period, the healthy volunteers who had genome sequencing incurred slightly higher medical costs of $3,670, on average, compared to $2,989 for those who had just a basic family medical history. The two groups had fairly similar numbers of outpatient lab tests (5.5 vs. 4.4) and doctor visits (8.4 vs. 6.9).



Medical costs for the cardiology patients who had whole-genome sequencing averaged $8,109 for the six months that the researchers followed them, which was less than the $9,670 costs for those who had a family medical history.

That counterintuitive finding was probably the result of a couple of the heart patients in the family medical history group being hospitalized for reasons unrelated to their participation in the study, Christensen said. Excluding hospitalizations, costs for the sequencing group averaged $5,392 compared to $4,692 for the family history group. Their number of doctor visits (7.8 vs. 7.2) was comparable, though those who were sequenced averaged 9.5 outpatient lab tests compared to 6.9 for the family history group.

The spending difference isn't nothing, of course, and multiplied by hundreds of millions of people (if genome sequencing becomes as routine as, say, cholesterol tests) would add up to billions of dollars. But sequencing is a one-time cost, not a recurring one. In the study the sequencing, including interpreting the results, cost about $5,000, and that has been falling quickly[4].

Overall, said medical geneticist and co-author Dr. Robert Green of Brigham and Women's, "downstream medical costs of sequencing may be far more modest than the common narrative suggests." Green leads the MedSeq[5] project, which studies how genome sequencing can be integrated into clinical practice.

The modest downstream costs cannot be explained by the sequencing not finding any red flags: 18 percent of the healthy patients had a genetic variant that was known to cause disease or likely to do so. Of the cardiology patients, half had a variant associated with heart disease and 16 percent had one or

more associated with another disorder.

The physicians in the study "seem to be responding responsibly" to the sequencing results, Christensen said, not ordering countless follow-up tests or frequent screening out of fear that they'll miss signs of the disease the patient is at risk for.

Whether every physician would react that way is unclear, however. Surveys show that doctors are not well-versed in genetics or statistics. That has spurred concern that they'll assume the worst of genetic variants that experts classify as possibly (but far from definitely) pathogenic or of unknown significance, and order endless, costly screening and other tests out of an abundance of caution or defensive medicine.

"If someone has whole-genome sequencing and it finds low-penetrant mutations," those that might or might not cause disease, "all the problems of endless testing can occur," said molecular geneticist Madhuri Hegde, chief scientific officer of global lab testing at PerkinElmer Inc. and an expert on genetic testing, who was not involved in the study. It's particularly critical, she said, not to order tests, or panic, when a patient has what's called a variant of unknown significance, meaning studies are ambiguous about whether it causes disease.

"That's why genetic counseling is so important," Hegde said. "Physicians need to stick to giving medical advice and have genetic counselors interpret genetic results."

That's easier said than done, however, since the U.S. has a shortage of genetic counselors. In the new study, the physicians all had access to a genome resource center, as well as knowledgeable academic colleagues, to help them interpret sequencing results.

Only a study that enrolls more patients and follows them longer, and that uses community physicians rather than academic ones, who don't have special access to genome expertise, can tell whether the encouraging results are likely to be true in general. Christensen and his colleagues plan to follow more patients for at least five years.

It's even possible that widespread genome sequencing could save money for the health care system. "If you can identify individuals who are asymptomatic but have an actionable mutation," such as one causing breast or colorectal cancer, Hegde said, "you can do prophylactic surgery or increased screening, and probably save a lot of dollars" if the person never develops cancer.

About the Author

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