Are You and Your Primary Care Doc Ready to Talk About Your DNA?

The test will look for mutations that are associated with medical conditions ranging from heart disease to cancer.

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If you have a genetic mutation that increases your risk for a treatable medical condition, would you want to know? For many people the answer is yes. But such information is not commonly part of routine primary care.

For patients at Geisinger Health System, that could soon change. Starting in the next month or so, the Pennsylvania-based system will offer DNA sequencing to 1,000 patients, with the goal to eventually extend the offer to all 3 million Geisinger patients.

The test will look for mutations in at least 77 genes that are associated with dozens of medical conditions ranging from heart disease to cancer, as well as variability in how people respond to pharmaceuticals based on heredity.

“We’re giving more precision to the very important decisions that people need to make,” said Dr. David Feinberg, Geisinger’s president and CEO. In the same way that primary care providers currently suggest checking someone’s cholesterol, “we would have that discussion with patients,” he said. “‘It looks like we haven’t done your genome. Why don’t we do that?’”

Some physicians and health policy analysts question whether such genetic information is necessary to provide good primary care — or feasible for many primary care physicians.

The new clinical program builds on a research biobank and genome-sequencing initiative called MyCode that Geisinger started in 2007 to collect and analyze its patients’ DNA. That effort has enrolled more than 200,000 people.

Like MyCode, the new clinical program is based on whole “exome” sequencing,analyzing the roughly 1 percent of the genome that provides instructions for making proteins, where most known disease-causing mutations occur.

Using this analysis, clinicians might be able to tell Geisinger patients that they have a genetic variant associated with Lynch syndrome, for example, which leads to increased risk of colon and other cancers, or familial hypercholesterolemia, which can result in high cholesterol levels and

heart disease at a young age. Some people might learn they have increased susceptibility to malignant hyperthermia, a hereditary mutation that can be fatal since it causes a severe reaction to certain medications used during anesthesia.

Samples of a patient's blood or spit are used to provide a DNA sample. After analysis, the results are sent to the patient's primary care doctor.

Before speaking with the patient, the doctor takes a 30-minute online continuing education tutorial to review details about genetic testing and the disorder. Then the patient is informed and invited to meet with the primary care provider, along with a genetic counselor if desired. At that point, doctor and patient can discuss treatment and prevention options, including lifestyle changes like diet and exercise that can reduce the risk of disease.

About 3.5 percent of the people who've been tested through Geisinger's research program had a genetic variant that could result in a medical problem for which clinicians can recommend steps to influence their health, Feinberg said. Only actionable mutations are communicated to patients. Geisinger won't inform them if they have a variant of the APOE gene that increases their risk for Alzheimer's disease, for example, because there's no clinical treatment. (Geisinger is working toward developing a policy for how to handle these results if patients ask for them.)

Wendy Wilson, a Geisinger spokeswoman, said that what they're doing is very different from direct-to-consumer services like 23andMe, which tests customers' saliva to determine their genetic risk for several diseases and traits and makes the results available in an online report.

"Geisinger is prescribing DNA sequencing to patients and putting DNA results in electronic health records and actually creating an action plan to prevent that predisposition from occurring. We are preventing disease from happening," she said.

Geisinger will absorb the estimated $300 to $500 cost of the sequencing test. Insurance companies typically don't cover DNA sequencing and limit coverage for adult genetic tests for specific mutations, such as those related to the breast cancer susceptibility genes BRCA1 or BRCA2, unless the patient has a family history of the condition or other indications they're at high risk.

"Most of the medical spending in America is done after people have gotten sick," said Feinberg. "We think this will decrease spending on a lot of care."

Some clinicians aren't so sure. Dr. H. Gilbert Welch is a professor at the Dartmouth Institute for Health Policy and Clinical Practice who has authored books about overdiagnosis and overscreening, including "Less Medicine, More Health."

He credited Geisinger with carefully targeting the genes in which it looks for actionable mutations instead of taking an all-encompassing approach. He acknowledged that for some conditions, like Lynch syndrome, people with genetic mutations would benefit from being followed closely. But he questioned the value of DNA sequencing to identify other conditions, such as some related to heart disease.

“What are we really going to do differently for those patients?” he asked. “We should all be concerned about heart disease. We should all exercise, we should eat real food.”

Welch said he was also concerned about the cascading effect of expensive and potentially harmful medical treatment when a genetic risk is identified.

“Doctors will feel the pressure to do something: start a medication, order a test, make a referral. You have to be careful. Bad things happen,” he said.

Other clinicians question primary care physicians’ comfort with and time for incorporating DNA sequencing into their practices.

A survey of nearly 500 primary care providers in the New York City area published in Health Affairs this month found that only a third of them had ordered a genetic test, given patients a genetic test result or referred one for genetic counseling in the past year.

Only a quarter of survey respondents said they felt prepared to work with patients who had genetic testing for common diseases or were at high risk for genetic conditions. Just 14 percent reported they were confident they could interpret genetic test results.

“Even though they had training, they felt unprepared to incorporate genomics into their practice,” said Dr. Carol Horowitz, a professor at the Icahn School of Medicine at Mount Sinai in New York, who co-authored the study.

Speaking as a busy primary care practitioner, she questioned the feasibility of adding genomic medicine to regular visits.

“Geisinger is a very well-resourced health system and they’ve made a decision to incorporate that into their practices,” she said. In Harlem, where Horowitz works as an internist, it could be a daunting challenge. “Our plates are already overflowing, and now you’re going to dump a lot more on our plate.”

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