Study identifies patients' genetic risk for cardiovascular disease

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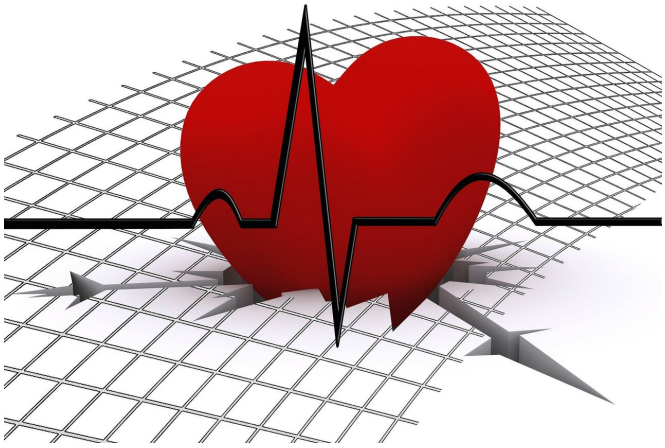

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A person's genetics may hold the key to early intervention in cardiovascular disease, leading to better outcomes in patient care. Researchers at Baylor College of Medicine's Human Genome Sequencing Center and Baylor cardiologists conducted a pilot study to determine if providing genetic testing for patients in cardiovascular clinics would benefit clinical care as part of a precision medicine initiative. They found that the test results did have implications on the course of treatment for approximately one-third of participants. Their results are published in the journal *Genetics in Medicine*.

The researchers developed a HeartCare panel that provided DNA sequence for 158 genes associated with medically actionable cardiovascular conditions along with a genetic risk score for developing cardiovascular disease and genetic data on drug interactions. A group of 709 patients was enrolled at Baylor College of Medicine cardiology clinics and received a free HeartCare panel test as part of their routine care. Results were returned to the patient's physicians and entered into their electronic medical records for ease of access.

After testing, 32% of participants received a genetic finding that impacted their clinical management. Of those participants, 11% were referred to a genetic specialist for further care. Out of all participants, 9% had an inherited pathogenic gene mutation associated with cardiovascular diseases like cardiomyopathy and high cholesterol, and 9% had a high overall genetic risk score for developing cardiovascular disease. High risk scores could be addressed with medication, diet and other lifestyle changes.

"This study shows that a large proportion of individuals in select ambulatory care clinics can benefit from genetic data," said Dr. Richard Gibbs, a senior author of the study and director of the Human Genome Sequencing Center and Wofford Cain Chair and professor of molecular and human genetics at Baylor. "There is tangible follow-up care for people who received a positive result, and in many cases for their family members."

"We have shown that genomic medicine can be integrated into cardiology clinical care and that we can find medically actionable issues," said Dr. Christie Ballantyne, a senior author of the study and professor of medicine and chief of the sections of cardiology and cardiovascular research at Baylor. "We treat a lot of cardiovascular diseases late in the process. If we can start our care earlier, we can prevent cardiovascular events and improve outcomes. Genetic testing may help us to identify not only who is at risk, but also allow us to screen and identify other family members who may be at risk."

The study also gathered feedback from physicians to determine how implementing genetic testing impacted their work. Among surveyed physicians, 84% said they changed the course of patient care based on test results, including referring patients to

a specialist, performing further cardiac tests and changing medications, and 60% of respondents believe the HeartCare study results improved clinical care.

"While the role of genetic testing in children has been known for some time, this study clearly demonstrates the benefits of comprehensive genetic screening in adults as well," said Dr. David Murdock, first author of the paper and assistant professor of molecular and human genetics at Baylor at the time of research.

Next, Baylor will expand the study by performing whole genome sequencing for participants to detect other genetic risk factors. The researchers also are working to replicate their study in other specialty areas and to determine if this testing can be effective on a larger scale.

More information: David R. Murdock et al, Genetic testing in ambulatory cardiology clinics reveals high rate of findings with clinical management implications, *Genetics in Medicine* (2021). DOI: 10.1038/s41436-021-01294-8

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