

Study finds certain genetic tests not useful in predicting heart disease risk

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A Polygenic Risk Score—a genetic assessment that doctors have hoped could predict coronary heart disease (CHD) in patients—has been found not to be a useful predictive biomarker for disease risk, according to a Vanderbilt study published in the *Journal of the American Medical Association*.

Lead author Jonathan Mosley, MD, Ph.D., assistant professor of Medicine and Biomedical Informatics, and senior author Thomas Wang, MD, former director of the Division of Cardiovascular Medicine and now chair of the Department of Internal Medicine at the University of Texas Southwestern Medical Center in Dallas, conducted a retrospective cohort study of the predictive accuracy of polygenic risk scores in 7,306 adults of European ancestry ages 45-79. The patients were taken from two large cohort studies, the Atherosclerosis Risk in Communities (ARIC) study and the Multi-Ethnic Study of Atherosclerosis (MESA).

Mosley found that the polygenic risk score didn't significantly improve prediction of CHD risk in this generally white, middle-age population. It was no more useful than the conventional method of determining CHD Risk—assigning a patient a clinical risk score based on factors including age, gender, <u>cholesterol levels</u> and tobacco use, Mosley said.

Physicians have long sought to reduce cardiovascular mortality by early identification of CHD.

Though his findings suggest a genetic biomarker has not been discovered for this general population of CHD patients, Mosley said further study is needed to determine whether special populations may benefit from a polygenic risk score.



"Genetic methods are getting better and people are developing these risk scores that are performing better than they have in the past," he said. "There are a lot of calls to see whether these can be converted to clinical tools. A critical piece of this process is to conduct a rigorous evaluation of any risk score, as has been done for other proposed cardiac biomarkers."

Mosley said the study suggests that polygenic risk scores should not be added to the standard of care for identifying high-risk CHD patients at this time. "Essentially, they are more likely to add costs than benefits at this point," he said.

Wang emphasized, "This study does not minimize the importance of genetic contributors to cardiovascular risk. What it does show is that <u>genetic factors</u> provide limited information about who will actually have a cardiovascular disease event."

"I think it's possible that we could develop better-performing scores," Mosley added. "I think you could develop more rigorous methodologies that may enhance the degree to which these predict."

More information: Sadiya S. Khan et al, Do Polygenic Risk Scores Improve Patient Selection for Prevention of Coronary Artery Disease?, *JAMA* (2020). DOI: 10.1001/jama.2019.21667

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