Genetic testing pinpoints cardiovascular risk in patients with aortic disorders

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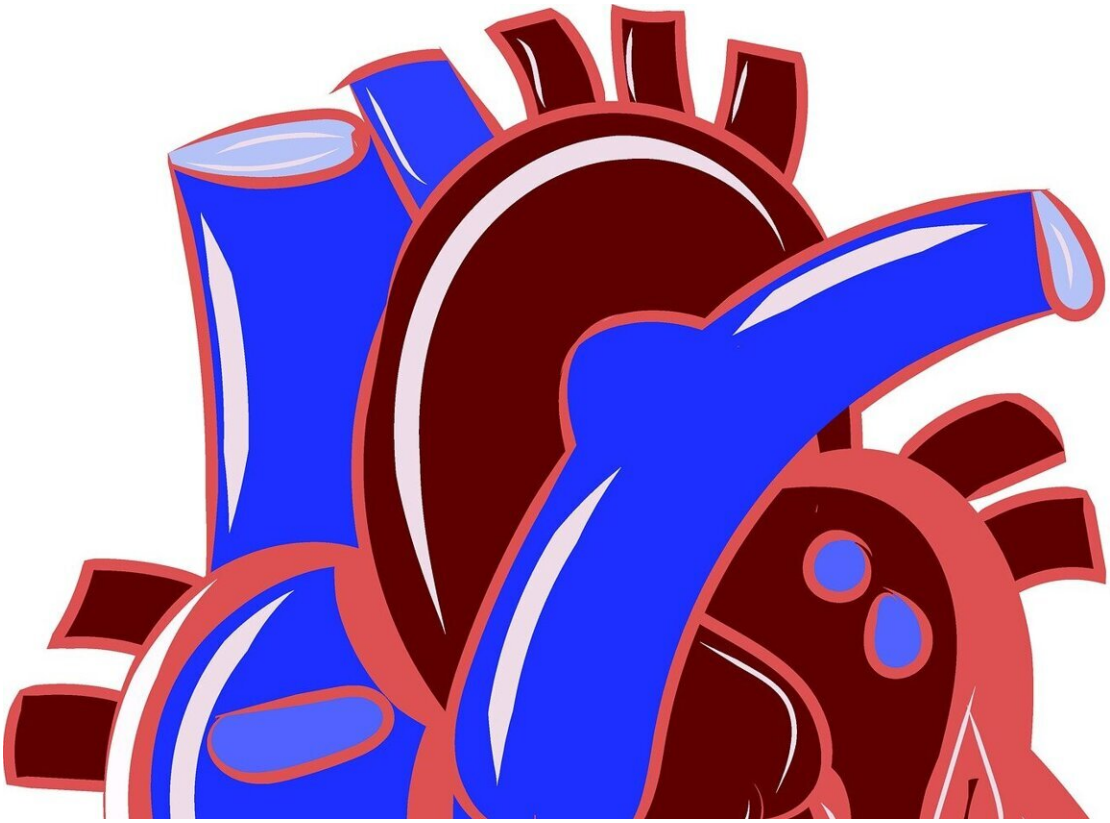

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A new Mayo Clinic systematic review, published in the *Journal of Vascular Surgery* has shed light on genetically inherited aortic disorders, highlighting the importance of early diagnosis and treatment for those

who have inherited cardiovascular risk.

The researchers identified that a procedure known as Thoracic Endovascular Aortic Repair (TEVAR) provided a high survival rate for young adult patients with type B aortic dissections. Type B aortic dissections consist of a tear in the descending part of the aorta that may extend into the abdominal aorta.

"We studied type B aortic dissections and aortic aneurysms and their management, especially in people born with genetic complications or syndromes," says Mohammed M. Firwana, M.B.B.S., research fellow and lead author of the study.

Notably, in more than one-third of patients, genetic testing identified new aortic events within 15 years. Patients with genetic risk factors exhibited a 62% rate of recurring aortic events, compared with 11% in patients without genetic risk factors, signifying the potential for more severe complications in the presence of such disorders.

Dr. Firwana adds that some of the findings varied by race, underlining the critical need for awareness and early treatment across all demographics.

"One of the more interesting findings was that Black patients with genetic risk factors had lower mortality than White patients, and that's mostly explained by how they presented at a younger age and their condition was likely managed earlier," says Dr. Firwana.

Researchers underscore that aortic aneurysms, dissections, and ruptures are thought to be the cause of 43,000 to 47,000 fatalities in the U.S. each year and that approximately 20% of thoracic aortic aneurysms are related to the genetic condition, also known as heritable thoracic aortic disease. These conditions are often silent but can lead to aortic dissection

or rupture with often fatal outcomes.

Dr. Firwana emphasizes that identifying certain genetic mutations provides important knowledge and understanding of genetically inherited aortic disorders in patients.

He adds that there are multiple risk factors that can lead to a higher likelihood of aortic disorders, for example, Marfan syndrome, a connective tissue disease, that affects 1 in 5,000 people in the U.S.

Dr. Firwana notes that another condition that can leave people prone to aortic issues is hypertension.

"Hypertension stands out as an important risk factor," says Dr. Firwana. "It contributes to the weakening of the walls of the aorta over time, making them more susceptible to tearing or dissection."

Researchers note that the systematic review is being used to help support guideline development when managing genetically inherited aortic disorders in patients and that more research is needed to identify the role of Thoracic Endovascular Aortic Repair in patients with connective tissue disease.

Dr. Firwana emphasizes that quitting smoking, managing hypertension, and addressing diabetes are crucial preventive strategies for those with these genetic predispositions.

"A panel of experts from the Society for Vascular Surgery will use this systematic review to develop a clinical practice guideline that aims to help patients make decisions about their care," says M. Hassan Murad, M.D., senior author of the study. "Further research on this topic can guide tailored interventions for improved patient outcomes."

More information: Mohammed Firwana et al, A systematic review supporting the Society for Vascular Surgery guidelines on the management of heritable aortopathies, *Journal of Vascular Surgery* (2023). DOI: 10.1016/j.jvs.2023.06.004

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