Gene variant raises risk for aortic tear and rupture

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Researchers from Yale School of Medicine and Celera Diagnostics have confirmed the significance of a genetic variant that substantially increases the risk of a frequently fatal thoracic aortic dissection or full rupture. The study appears online in *PLOS ONE*.

Thoracic aortic aneurysms, or bulges in the artery wall, can develop without pain or other symptoms. If they lead to a tear—dissection—or full rupture, the patient will often die without immediate treatment. Therefore, better identification of patients at risk for aortic aneurysm and dissection is considered essential.

The research team, following up on a previous genome-wide association study by researchers at Baylor College of Medicine, investigated genetic variations in a protein called FBN-1, which is essential for a strong arterial wall. After studying hundreds of patients at Yale, they confirmed what was found in the Baylor study: that one variation, known as rs2118181, put patients at significantly increased risk of aortic tear and rupture.

"Although surgical therapy is remarkable and effective, it is incumbent on us to move to a higher genetic level of understanding of these diseases," said senior author John Elefteriades, M.D., the William W. L. Glenn Professor of Surgery (Section of Cardiac Surgery) at Yale School of Medicine, and director of the Aortic Institute at Yale-New Haven Hospital. "Such studies represent important steps along that path."

The researchers hope their confirmation of the earlier study may help lead to better clinical care of patients who may be at high risk of this fatal condition. "Patients with this mutation may merit earlier surgical therapy, before aortic dissection has a chance to occur," Elefteriades says. Yale cardiothoracic surgeons will now begin assessing this gene in clinical patients with aneurysm disease.

More information: dx.plos.org/10.1371/journal.pone.0091437

Provided by Yale University

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