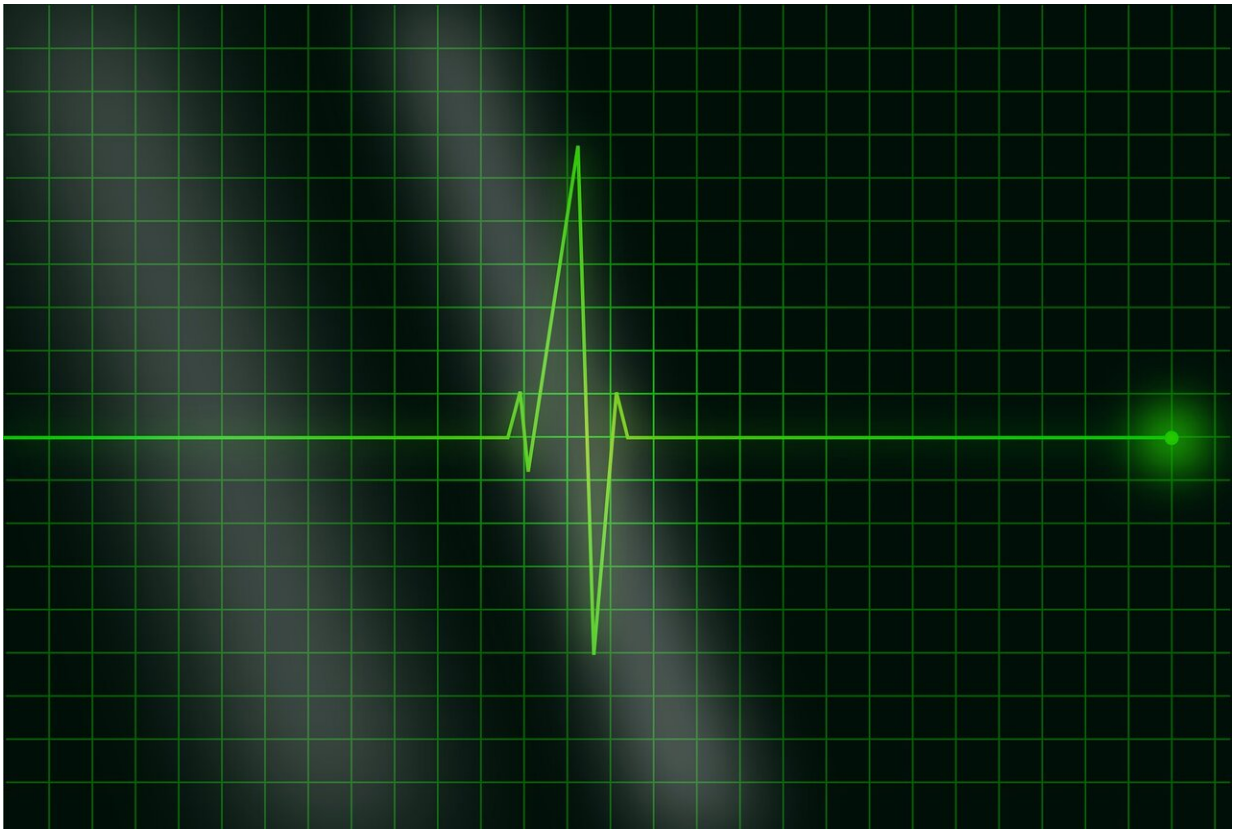
New study finds the prealbumin gene alone is insufficient for diagnosis of heart failure

July 28 2023



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A new multi-center study led by doctors at Boston Medical Center and Columbia University found that having a genetic variant in the prealbumin gene alone is not sufficient for diagnosis of transthyretin

amyloid cardiomyopathy in older Black patients. Published in the Journal of the American Heart Association, researchers discovered that a blood test that measures the transthyretin or prealbumin protein might also be helpful in diagnosing transthyretin amyloid cardiomyopathy and could be used to trigger more definitive imaging testing.

Transthyretin amyloid cardiomyopathy (ATTR-CM) is an underdiagnosed cause of congestive heart failure among patients 60+ years of age. There is a common genetic variant, V122I (or Val122Ile), in a protein called transthyretin (TTR) or prealbumin that is associated with ATTR-CM and present in 3.4% of Black individuals, or 1.5 million people. V122I is so common because when present, the variant is passed genetically from parent to child 50% of the time. Importantly, of those who have this variant, it is unknown who will develop ATTR.

Researchers note that since more people are getting their genes tested using commercial services, some of which return the V122I test result, it is important for people to know the association between a positive genetic result and the disease with which it is associated.

"Cardiac amyloidosis is a serious heart condition that can be caused by a common genetic variant carried by 1.5 million people," said senior author Frederick L. Ruberg, MD, a cardiologist at Boston Medical Center and Associate Professor of Cardiovascular Medicine and Radiology at Boston University Chobanian & Avedisian School of Medicine. "Our study shows that of those who have inherited this variant, only 39% developed cardiac amyloidosis, so not everyone who inherits the variant will necessarily develop this serious condition."

Researchers enrolled 278 self-identified Black heart failure patients from the Screening for Cardiac Amyloidosis with Nuclear Imaging in Minority Populations (SCAN-MP) study, funded by the National Institutes of Health. Study participants live in Boston and New York City

and were tested for the genetic variant. Participants were also scanned with a special nuclear heart-imaging test to determine whether they have ATTR-CM.

With 1.5 million people carrying the V122I variant in the US, there are a great number people at risk for ATTR-CM. This study shows that though carriers may have the gene, they will not necessarily develop the disease. The study also shows that just testing for and identifying the V122I variant is not enough to infer that that heart failure is due to cardiac amyloidosis.

"Our study suggests that a widely available blood test to measure prealbumin levels may also be useful in identifying patients that should have more sensitive imaging testing for ATTR-CM," said co-senior author Mathew Maurer, MD, Arnold and Arlene Goldstein Professor of Cardiology at Columbia University Irving Medical Center and Director of the Cardiac Amyloidosi Program.. "Our results also help better understand how heart failure from ATTR-CM impacts older Black individuals."

More information: Clinical Penetrance of the Transthyretin V122I Variant in Older Black Patients With Heart Failure: The SCAN-MP (Screening for Cardiac Amyloidosis With Nuclear Imaging in Minority Populations) Study, *Journal of the American Heart Association* (2023). DOI: 10.1161/JAHA

Provided by Boston Medical Center