MedicalXpress

Office-based test developed to identify amyloidosis-related heart failure

February 8 2017

Boston researchers have developed a new diagnostic test that may help doctors identify patients with a condition called cardiac amyloidosis. The findings appear in the journal *JAMA Cardiology*.

Cardiac amyloidosis is caused by abnormal folding of proteins that deposit in the heart. These protein deposits can also occur in other organ systems in the body and can cause life-threatening organ failure. Cardiac amyloidosis that results from the mis-folded protein transthyretin is called ATTR amyloidosis, and this form of the disease occurs in older patients.

ATTR amyloidosis can occur from a genetic mutation that causes the TTR protein to become unstable, mis-fold, and then build up in the heart. The most common genetic mutation that causes ATTR amyloidosis is seen in approximately 3.5 percent of African Americans. Amyloid deposition can cause electrical abnormalities and decrease the heart's ability to relax and contract, leading to congestive heart failure.

The diagnosis of ATTR amyloidosis can be challenging for doctors, and amyloidosis in many patients remains un-recognized, sometimes until the time of death. However, recent studies suggest that as many as 10 percent of older patients with certain types of congestive heart failure may have cardiac amyloidosis. This diagnostic challenge prompted the research team, led by Frederick L. Ruberg, MD, director of advanced imaging at Boston Medical Center and associate professor of medicine at Boston University School of Medicine, to develop new testing strategies

to improve diagnosis.

In this study, researchers identified that a specific blood protein named retinol-binding protein 4 (RBP4) can be used to determine the likelihood of ATTR amyloidosis in a patient with congestive heart failure. In addition, in work guided by Marios Arvanitis, MD, an internal medicine resident at BMC, the research team developed a mathematical calculator that incorporates RBP4 and other commonly ordered clinical tests that can be used to estimate the probability of ATTR amyloidosis in a given patient. An important advantage of this algorithm is that it can be used in the context of a doctor's office visit at the point-of-care.

According to the researchers, this discovery could guide clinical decision making and increase recognition of this disease. Since many new drug therapies are in various stages of development now for ATTR amyloidosis, recognition and accurate diagnosis is essential to get a patient on the correct treatment.

"Given that new targeted pharmacologic therapies are being developed that specifically treat ATTR, identifying patients with this disease and providing an early and accurate diagnosis is crucial to treatment," explained Ruberg, the study's corresponding author.

Ruberg believes there is also potential for this blood test or the entire algorithm to be used in ATTR cardiac amyloidosis monitoring as a marker of disease progression and prognosis.

Provided by Boston University Medical Center