BUSM researchers part of multi-center study on cardiac amyloidosis

13 August 2012

Researchers at Boston University School of Medicine (BUSM) have been part of a multicenter observational study called TRACS (Transthyretin Amyloidosis Cardiac Study) to help determine the health significance of a particular gene mutation which is commonly found in Black Americans.

The gene, transthyretin (TTR) and the mutation V122I, is seen in about four percent of African Americans or roughly 1.5 million people. The mutation is strongly associated with a condition called cardiac amyloidosis - a disorder where the abnormally mutated TTR protein deposits in the heart of affected individuals, causing the heart to not pump adequately and ultimately results in death. Cardiac amyloidosis due to this mutation is seen in older people, over the age of 60 years. Cardiac amyloidosis can be also be due to genetically normal TTR as well, which occurs for unclear reasons but is strongly associated with age and male gender.

The researchers enrolled 29 patients with TTR cardiac amyloidosis due to either the V122I mutation or those with genetically normal TTR, and observed them for roughly 16 months. They found that Black patients with the mutation only survived for a median of 26 months after diagnosis, and that certain measured parameters including heart function by echocardiography, blood testing and exercise capacity fell during the study and were associated with survival. Quality of life determined by questionnaire also declined significantly. These findings currently appear online in the American Heart Journal.

"This study is important because it establishes that carriers of V122I are at increased risk of death from cardiac amyloidosis once they reach an age where the disease is expressed (generally over the age of 60)," explained lead author Frederick L. Ruberg, MD, assistant professor of medicine and radiology at BUSM. According to Ruberg these findings provide an important foundation on which future studies using new drugs can be based. "If patients on the new drug live longer or decline more slowly than those in TRACS, that would suggest a beneficial effect to treatment," he added.

Provided by Boston University Medical Center