

Molecular imaging detects signs of genetic heart disease before symptoms arise

June 6 2011

Research being presented at SNM's 58th Annual Meeting shows that molecular imaging is helping to determine the cause and expand treatment for a silent killer. A study focusing on hypertrophic cardiomyopathy (HCM)—a cardiovascular disorder that causes a thickening of the heart muscle—is proving that the effects of a genetic mutation may be an important key to understanding the disease. In related research, a treatment called alcohol septal ablation is being revealed as an effective treatment for severe cases of HCM.

"These studies show that molecular imaging has great potential for clinical use in the field of cardiology," says Stefan Timmer, MD, Free University Medical Centre, Amsterdam, The Netherlands. "HCM is usually asymptomatic, and one of the first symptoms is acute death due to arrhythmia, which demonstrates how essential it is to detect this disease. We used [molecular imaging](#) to gauge the heart's efficiency in subjects with a pathogenic mutation linked to HCM. Unexpectedly, we found that carriers of the mutation already have a less efficient heart despite the absence of any symptoms. In addition, the research provides a suggestion why a frequently used therapy, alcohol septal ablation, is an effective treatment for symptomatic patients who remain symptomatic despite optimal medical treatment."

The abnormal thickening of the muscle, or myocardium, involved in HCM makes it harder for the heart to contract and relax, forcing the heart to work harder to pump blood. HCM is often asymptomatic and can cause sudden cardiac death at any age. The pathophysiology of HCM

is not well understood. However, preclinical research points toward genetic mutations, such as MYBPC3, which are exclusive proteins in the heart's contractile tissue.

In one study, researchers imaged 16 subjects with the MYBPC3 mutation using positron emission tomography (PET) in order to image the heart's oxygen usage. This was coupled with cardiovascular magnetic resonance imaging to gauge the heart's ability to use energy efficiently. To their surprise, patients carrying the MYBPC3 mutation were shown to have less efficient hearts even prior to the thickening and subsequent symptoms associated with HCM, which makes it a prime causal candidate for the development of the disease.

For HCM patients who remain symptomatic despite medication, alcohol septal ablation is a popular treatment. It is essentially a miniature, controlled heart attack triggered by an absolute alcohol solution administered to the septal artery that provides blood flow to the thickened heart muscle. Interventional cardiologists perform the non-invasive therapy using wires and balloons guided to the artery. There are few complications associated with the procedure, and it is the preferred treatment over open-heart surgery. In the study researching alcohol septal ablation for obstructive HCM, 15 patients received the procedure, and results were analyzed. Researchers concluded that obstructive HCM was at least in part reversible with alcohol septal ablation due to its ability to improve oxygen delivery to the [heart](#) and improve myocardial energetics, which is the heart's use of energy in relation to its ability to pump blood.

Further research could potentially lead to better screening for asymptomatic HCM carriers and the development of early treatments that could halt or at least postpone full onset of the disease.

More information: Scientific Paper 2: S. Timmer, T. Germans, W.

Brouwer, M. Lubberink, A. Lammertsma, P. Knaapen, A. van Rossum, Vrije Universiteit Medical Center, Amsterdam, The Netherlands; "Impaired myocardial energetics precede coronary microvascular dysfunction and ventricular hypertrophy in carriers of the hypertrophic cardiomyopathy MYBPC3 mutation," SNM's 58th Annual Meeting, June 4-8, 2011, San Antonio, TX.

Scientific Paper 109: S. Timmer, P. Knaapen, T. Germans, P. Dijkmans, M. Lubberink, J. ten Berg, F. ten Cate, M. Götte, A. Lammertsma, A. van Rossum, Vrije Universiteit Medical Center, Amsterdam, The Netherlands; St. Antonius Hospital, Nieuwegein, The Netherlands; Thoraxcenter Erasmus Medical Center, Rotterdam, The Netherlands; "Effects of alcohol septal ablation on coronary microvascular function and myocardial energetics in hypertrophic obstructive cardiomyopathy," SNM's 58th Annual Meeting, June 4-8, 2011, San Antonio, TX.

Provided by Society of Nuclear Medicine

Citation: Molecular imaging detects signs of genetic heart disease before symptoms arise (2011, June 6) retrieved 10 April 2024 from

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