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Improved chances of discovering hereditary transthyretin amyloidosis

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New insights in hereditary transthyretin amyloidosis (ATTR) improve the chances of diagnosing the genetic disease using ECG and ultrasound. In a new doctoral dissertation by Sandra Arvidsson at Umeå University in Sweden, an explanation to the varying symptoms in patients with ATTR is explored. Of particular interest is why the disease only affects the heart in some patients—pinpointing age and gender as important explanations.

ATTR is a lethal disease characterised by pain, paraesthesia, muscular weakness and autonomic dysfunction. The disease is endemic in Portugal and northern Sweden where 200-300 patients are diagnosed with the disease, but affects people across the globe. ATTR is in many cases difficult to diagnose and can affect tissues throughout the body by changing the hereditary character of the protein transthyretin, which results in varying symptoms.

In some patients, most symptoms affect the stomach and intestines, and the nervous system, whilst others are almost only affected by a thickening of the myocardium—the heart muscle, which leads to heart failure. The study shows that the varying symptoms partly depend on age and gender, and that older male patients develop more severe effects on the heart.

ATTR can be discovered by heart screening, where a thickening of the myocardium can be a sign of the disease. However, other diseases can also result in the same symptom, which means distinction between

diseases can be difficult using ultrasound screening alone.

"I have discovered specific measurements that better identify a thickening of the myocardium caused by ATTR. By measuring the results of the ECG and the left ventricular thickness in the ultrasound, a more accurate suspicion of the disease can be confirmed," says Sandra Arvidsson, doctoral student at the Department of Public Health and Clinical Medicine at Umeå University and author of the dissertation.

"We found that patients with ATTR had reduced amplitudes on the ECG and a relatively symmetric wall thickening of the heart in comparison to patients where the thickening of the myocardium was due to other reasons," she continues.

In the study, ECG and ultrasound was used—two easily accessible and in general well-used screening methods to investigate suspected heart disease. The use of these two methods can increase the probability for patients with ATTR to receive correct diagnosis at an early stage of the disease.

Another decisive factor for the symptoms of ATTR can be found by looking at the composition of the protein transthyretin in the patient. Previous studies have shown that patients with ATTR have had two different kinds of protein composition. Many patients have full-length transthyretin molecules, whereas the proteins are fragmented in other patients.

The study shows that patients with partly fragmented transthyretin to a larger extent show signs of a thickening of the myocardium and also deteriorated in their heart disease as a consequence of liver transplant.

Liver transplant is a common treatment of ATTR. In many cases, the results are successful. However, in other cases, the disease continues to

develop first and foremost in the form of continued negative impacts on the heart.

More information:
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