Rare genetic variants identified in sudden cardiac deaths associated with obesity and hypertension

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The University of Oulu's Cardiology Research Group identified several rare genetic variants in victims of sudden death caused by hypertension-

or obesity-related cardiac hypertrophy. The findings suggest that, in addition to lifestyle, genes may also play a role in hypertension- and obesity-related cardiac hypertrophy.

Especially in the case of middle-aged and older people, sudden cardiac death is thought to be due to acquired, i.e. non-inherited cardiac diseases such as cardiac hypertrophy due to obesity or high blood pressure. On the other hand, genetic predisposition to sudden death has also been identified in this age group, but the mechanism of the predisposition and the genetic link have been unclear.

The genetic study focused on 151 victims of sudden death between 20 and 80 years of age with hypertension- or obesity-related cardiac hypertrophy and net accumulation of extracellular matrix in the myocardium, or fibrosis. The control group consisted of people living with long-term hypertensive cardiac hypertrophy.

The study revealed changes mainly in genes involved in regulating the structure or contractility of the heart muscle. No significant changes were observed in the genes involved in the electrical activity of cardiac muscle cells.

The observation suggests that the genetic background of sudden cardiac death may be at least partly related to a wide range of different genetic variants affecting the myocardial structure. "We believe that the role of multifactorial myocardial disease in sudden deaths related to cardiac diseases is greater than currently recognized," says head researcher Lauri Holmström from the University of Oulu, Finland.

A better understanding of the genetic background of cardiac disease leading to sudden death can contribute to identifying persons at high risk and benefiting from closer monitoring. While understanding the genetic background can increase individual knowledge of the risk and nature of

cardiac disease, prevention is largely down to a healthy lifestyle.

"The focus of prevention of sudden cardiac death is still on healthy lifestyle, including sufficient physical activity, weight management, non-smoking and avoidance of excessive alcohol consumption. If high blood pressure, diabetes, or high cholesterol have developed regardless of the above, their appropriate treatment is crucial," says Holmström.

Genetic studies increase understanding of the development of myocardial diseases and the mechanism of sudden death. The results obtained are among the first to provide new information on the role of myocardial diseases in multifactorial cardiac diseases. However, further research is still needed to determine the significance of the observed genetic variants and genes more closely. Also, examining the clinical characteristics of myocardial diseases related to genetic vulnerability would, according to Holmström, be important in order to identify these diseases in people and refer them to further examination.

The study is based on the FinGesture data, which consists of all the deceased shown to have died from sudden cardiac death in a forensic autopsy in Northern Finland in 1998–2017. In total, the FinGesture study has collected data on nearly 6,000 deaths from sudden cardiac death. Approximately one in five of the deaths were due to acquired myocardial disease.

The study was published in *Scientific Reports*, a part of Nature Publishing Group.

More information: Lauri Holmström et al, Genetic contributions to the expression of acquired causes of cardiac hypertrophy in non-ischemic sudden cardiac death victims, *Scientific Reports* (2021). DOI: 10.1038/s41598-021-90693-7

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