Discovery could improve screening for sudden cardiac death

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Unfortunately, newspaper articles about young athletes dying suddenly on the field are not unheard of. Such reports fuel discussions about compulsory screening, for example of young footballers, for heart failure. Research by scientists from Ghent (VIB/UGent) and Italy will benefit these screening methods. They have discovered a link between mutations in a certain gene and the heart condition Arrhythmogenic Right Ventricular Cardiomyopathy.

ARVC is a hereditary heart condition in which the heart muscle (particularly the right ventricle) is partly replaced by fatty tissue and connective tissue. Cardiac arrhythmias can occur as a result of the changes in the heart muscle. Severe arrhythmias can cause dizziness or even lead to fainting or an acute cardiac arrest (= sudden death). ARVC is a progressive disease that usually presents during the teenage years.

Known mutations in desmosomal proteins

Mutations in various genes have already been linked to ARVC. These are primarily genes that are responsible for the production of proteins in the desmosomes. Desmosomes are structures in the heart that ensure that the heart muscle cells remain connected to each other. Therefore, it was assumed that defects in the desmosomes were the most important factors in developing ARVC.

Identification of new mutations

Together with Italian scientists, Jolanda van Hengel, studied patients with ARVC who did not exhibit mutations in the desmosomal genes. The scientists identified mutations in the CTNNA3 gene in these patients, which codes for the protein αT-catenin – a component of the area composita. The area composita is a structure specifically modified to the heart, where extra strong connections between cardiac muscle cells occur.

The scientists' findings indicate that there is a link between mutations in the CTNNA3 gene and ARVC. It was demonstrated for the first time that – in addition to desmosomal genes – an area composita gene also plays a role in the development of ARVC. Future genetic screening tests for ARVC should include the CTNNA3 gene as a standard part of the test. This would increase the value of the screening.

More information: The research is published in the leading journal *European Heart Journal*, van Hengel et al., Mutations in the area composita protein alpha-T-catenin are associated with arrhythmogenic right ventricular cardiomyopathy. eurheartj.oxfordjournals.org/c … artj.ehs373.abstract

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