Recommendations on genetic testing for inherited cardiac diseases published today

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When should patients and family members undergo genetic assessment for a heart condition? Find out in an international consensus document published in EP Europace, a journal of the European Society of

Cardiology (ESC) and presented at EHRA 2022, a scientific congress of the ESC.

"This is now the reference document that all clinicians should use to decide whether genetic testing is indicated for patients with inherited cardiac diseases and their relatives," said lead author Professor Arthur Wilde of the Academic Medical Centre, Amsterdam University Medical Centres, the Netherlands. "We provide strict criteria on who should be assessed and recommend which genes should be examined."

The aim of genetic testing in patients with an inherited cardiac disease is to determine the cause. In some conditions this helps clinicians make a precise diagnosis, provides information about prognosis, and determines the treatment. For example in long QT syndrome, which is potentially lethal but treatable, a normal electrocardiogram (ECG) does not exclude the condition and genetic testing is required to clarify the diagnosis. The specific genetic variant impacts both prognosis and therapeutic choices.

Once a genetic cause is identified in the patient, family members, including children, can be screened. The document outlines in which conditions relatives should receiving genetic testing. In long QT syndrome, for example, family members should be tested. "Many of these conditions start with a cardiac arrest in a young individual who dies or almost dies," said Professor Wilde. "The way to avoid that happening in a family members is by genetic testing in conjunction with clinical screening. Those who are affected can be treated, for example with medications or with a defibrillator to correct a fatal heart rhythm, and those who are unaffected can be reassured."

Genetic counselling is essential and should start even before clinical and genetic testing are performed. A diagnosis can be life-changing as it "may provoke significant anxiety or aggressive treatment", states the document. Professor Wilde said that the consequences of a positive

diagnosis should be explained before any examinations. "For instance, if an individual has no symptoms but his or her sibling has a serious inherited cardiac disease, the first question should be 'do you want to know whether you have this condition, yes or no?'," he said. "A diagnosis may trigger difficulties with insurance, getting a mortgage, and so on. He or she needs to be informed before making any decisions."

The paper provides recommendations on genetic testing for four groups of heart conditions caused by genetic defects: inherited arrhythmia syndromes, cardiomyopathies, sudden cardiac death or survivors of unexplained cardiac arrest, and congenital heart disease. The most common of these is hypertrophic cardiomyopathy, which affects at least one in 500 individuals.

The chapter on congenital heart disease also provides detailed advice on genetic testing in pregnant women and offspring. Professor Wilde said: "This is a rapidly moving field and genetic testing is recommended for conditions in which there is a high likelihood of identifying the cause. As for all genetic cardiac conditions, testing for congenital heart defects should be coordinated by cardiologists and clinical genetics specialists with support from genetic counsellors."

In addition to conditions caused by single genetic defects, the authors describe how genetics can play a role in the manifestation of more common heart conditions such as coronary artery disease and heart failure. Professor Wilde explained: "These conditions are not caused by one genetic variant but in some patients there is a genetic component. Researchers are investigating how the combination of frequently occurring genetic variants may cause or influence susceptibility to disease. This is an emerging field and it is too early to make recommendations."

The international consensus statement on genetic testing for cardiac

diseases was developed by the European Heart Rhythm Association (EHRA), a branch of the ESC; the Heart Rhythm Society (HRS); the Asia Pacific Heart Rhythm Society (APHRS); and the Latin American Heart Rhythm Society (LAHRS).[3] It is also published in *Heart Rhythm*, the official journal of the HRS, *Journal of Arrhythmia*, the official journal of the APHRS, and *Journal of Interventional Cardiac Electrophysiology*, the official journal of the LAHRS.

More information: *EP Europace* (2022). DOI: 10.1093/europace/euac030