

Genetic testing for inherited cardiac conditions is 'patchy' in Europe

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While genetic inheritance is known to play a role in the multifactorial development of most diseases of the heart, there are also a number of clearly diagnosed cardiac conditions which owe their development to quite specific genetic abnormalities. When these genetic disorders affect the integrity of the heart's muscle they are known as a "cardiomyopathy"; when the disorder affects the heart's "excitability", it is known as a "channelopathy".

Both conditions predispose to arrhythmias and [sudden cardiac death](#) - often in the young. A reliable genetic test for the presence of [DNA changes](#) in the genes which encode for ion channels and relevant proteins would not only help identify affected patients and reduce these serious risks, but also provide information for personalised treatment.

An expert [consensus statement](#) on the value of diagnostic [genetic testing](#) for these inherited [cardiac conditions](#) will be unveiled today at the EHRA EUROPACE 2011 congress in Madrid. The report, the HRS/EHRA Expert Consensus Statement on the State of Genetic Testing for the Channelopathies and Cardiomyopathies, is a joint development of the Heart Rhythm Society and the European Heart Rhythm Association (EHRA). The latter is the organiser of EHRA EUROPACE 2011.

According to Dr Silvia Priori, who will present details of the consensus statement today, its aim is to provide recommendations on how each of 13 inherited conditions might be tested for and diagnosed using genetic analysis. The guidance makes clear that these recommendations deal with uncommon diseases and are based on the results of studies which are much smaller than those available for more [common diseases](#), such as [myocardial infarction](#) or [heart failure](#). But, says Dr Priori, the field is evolving rapidly. In deed, the genetics of inherited arrhythmogenic diseases is a recent sub-specialty of cardiology and it's only in

the past 25 years that the first causative genes for channelopathies and cardiomyopathies were discovered.

Dr Priori, who is director of Molecular Cardiology at the Fondazione Salvatore Maugeri and University in Pavia, and Director of Cardiovascular Genetics at New York University, describes the penetration of use of genetic testing in Europe as "patchy", with some countries still without even a limited framework for their application.

"The document is intended to provide guidance to cardiologists in the use of genetic testing among patients and family members," she explains. Results may be useful for both the diagnosis and treatment of affected individuals. The appropriate use of these tests, she adds, is critical because they are expensive, and should, therefore, be used in patients with a clinical diagnosis (or high suspicion) of these diseases.

The recommendations focus on testing for 13 inherited conditions, including hypertrophic cardiomyopathy, long QT syndrome, Brugada syndrome, and dilated cardiomyopathy. In addition, the statement includes guidance on the use of genetic testing for out-of-hospital cardiac arrest survivors and post-mortem testing in cases of sudden death, the most dramatic consequence of these conditions.

Dr Priori describes the prevalence of these conditions among the general population as ranging from one in 500 to one in 10,000 - with an average prevalence of around one in 2000. Based on current knowledge, it is still not possible to find [genetic abnormalities](#) in all patients affected by these conditions; however, in some - such as hypertrophic cardiomyopathy or long QT syndrome - genetic testing may identify a causative mutation in as many as 70% of cases. In other diseases, however, the yield of testing is much lower, and improvements will depend on the discovery of more

genes.

"So genetic testing cannot be viewed as a one-size fits all solution, but its contribution to family screening and management in affected patients should be defined for each disease," says Dr Priori, "and results should defined in the context of a comprehensive clinical evaluation." Counselling, particularly among family members, is essential for reassurance about disease risk and surveillance.

The consensus provides an assessment of the strength of indication for genetic testing in different conditions. In some diseases, such as hypertrophic cardiomyopathy or long QT syndrome, the recommendations to test all individuals with a clinical diagnosis are strong. In other diseases, such as Brugada syndrome or dilated cardiomyopathy, there is a value in performing the test, but the strength of recommendation is lower. And there are some instances - such as atrial fibrillation - where genetic testing cannot yet be indicated.

Similarly, the report does not recommend genetic testing in all cases of out-of-hospital cardiac arrest, but recommends that testing should be performed if there is a clinical sign or suspicion of an inherited arrhythmogenic disease. However, genetic testing "may be considered" in all cases of sudden unexpected death, including sudden infant deaths (SIDS), where autopsy yields negative results. Studies suggest that genetic mutations can explain an underlying cause of sudden unexpected death in up to 35% of cases. Even events such as drowning or motor accidents in the young may in fact be attributed to cardiac arrhythmias of genetic cause.

Ultimately, says Dr Priori, the report hopes to lower the risk of sudden cardiac death by promoting the appropriate use of [genetic testing](#) - and to ensure their reimbursement from insurance and health care systems.

Provided by European Society of Cardiology

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