Four in ten cardiomyopathies—a major cause of sudden death in young people—are genetic

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Four in ten cardiomyopathies - a major cause of sudden cardiac death and heart failure in young people - are genetic, according to a European Society of Cardiology (ESC) study published today in *European Heart Journal*.1 Family screening is urgently needed to prevent early death in apparently healthy relatives, the paper says.

Cardiomyopathy is where the heart muscle becomes enlarged, thick or rigid. As the condition worsens, the heart becomes weaker and less able to pump blood through the body and maintain a normal electrical rhythm. Around one in 300 people in Europe has a cardiomyopathy.

This is the first European registry on cardiomyopathies, and was conducted as part of the EURObservational Research Programme (EORP) of the ESC. The study included 3,208 patients enrolled by 69 centres in 18 countries. Data was collected on patient characteristics and treatments.

"We were surprised to see how frequently the disease is inherited," said first author Cardiology Professor Philippe Charron of the Pitié-Salpêtrière Hospital in Paris, France. "About 40% of patients had familial disease. As a consequence it is very important to improve screening to detect the disease in the relatives who are apparently healthy."

Two-thirds of relatives in the study were diagnosed through family screening, which includes echocardiography and an electrocardiogram (ECG). In many cases their disease was as severe as in the first family member identified, with the same frequency of symptoms and requirement for an implantable cardioverter defibrillator (ICD) to stop life threatening arrhythmias.

The study also suggests that recommendations should be changed so that family screening starts earlier than ten years of age and extends beyond the current threshold of 50 to 60 years. The EORP study found that some relatives were diagnosed much younger than ten while others were older than 70.

"The cardiac expression of these genetic diseases starts early in some patients," said Professor Charron. "In others it is delayed for many decades - longer than previously thought."

Genetic testing was performed in 36% of patients in the study - an increase from ten years ago - but not as good as it should be, the paper said. When a mutation is identified, genetic testing can then be performed in first degree relatives. Those with the mutation should have regular cardiac follow up, including an ECG and echocardiogram, to check for changes to heart muscle and function.

"The earlier we identify relatives with the mutation, the better we can manage them and prevent complications including sudden cardiac death," said Professor Charron.

Sudden cardiac deaths from cardiomyopathies are caused by heart rhythm disorders, called arrhythmias. An important part of managing patients with cardiomyopathies is to diagnose arrhythmias early. However, the study found that one in three patients did not receive the necessary diagnostic tests.

Professor Charron said: "The frequency of arrhythmias in the study was high. In one cardiomyopathy subtype, 39% of patients had potentially deadly arrhythmias. But diagnostic testing for arrhythmias was suboptimal and patients may be missing out on lifesaving treatment with an

ICD or pacemaker."

The authors said expert centres were needed at European and national levels to diagnose and manage patients with cardiomyopathies. Expert centres should meet the following criteria:

- Cardiac tests conducted to identify life threatening arrhythmias
- Tests to check heart function including magnetic resonance imaging when appropriate
- Specific investigations performed to identify rare causes of a cardiomyopathy
- Genetic tests performed in patients with cardiomyopathies, then relatives screened for the genetic mutation with subsequent cardiac monitoring if a mutation is found.

Professor Charron said: "Expert centres for cardiomyopathies would improve the diagnosis and management of these diseases in patients and their family members, and help to prevent heart failure and sudden deaths."

More information: The Cardiomyopathy Registry of the EURObservational Research Programme of the European Society of Cardiology: baseline data and contemporary management of adult patients with cardiomyopathies. *European Heart Journal* (2018). DOI: 10.1093/eurheartj/ehx819

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