What is Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy (ARVD/C)?

ARVD/C is an inherited cardiac disease that can lead to sudden death in young individuals.

ARVD/C is the second most common cause of sudden cardiac death (SCD) in young adults, including trained athletes.1,2 Affecting as many as 1 in 1000 individuals,3 ARVD/C arises when the muscle tissue that surrounds the right pumping chamber of the heart (the right ventricle) is replaced by fatty tissue, disrupting the normal function of the ventricle. This can lead to disturbances in the regular beating of the heart (arrhythmia) and, sometimes, to a complete breakdown of the heart beat resulting in sudden cardiac death.

What causes ARVD/C?

About half of all cases of ARVD/C are caused by a gene defect.

Up to 50% of all cases of ARVD/C are caused by a genetic defect, which can occur in any one of several different genes (reviewed in 4). Of those individuals who harbor such an ARVD/C-associated defect in their genes, about half will develop ARVD/C. If an individual harbors an ARVD/C-associated genetic defect, each of his or her children has a 50% chance of inheriting this defect, and thus, a high risk of developing the condition.

What are signs of ARVD/C?

The first signs of ARVD/C may be fainting, heart palpitations, or cardiac arrest.

ARVD/C symptoms most commonly first occur in individuals in their thirties, but can appear earlier. The first signs may be fainting, heart palpitations (the sensation that the heart is beating irregularly or “fluttering”), or cardiac arrest.6

How is ARVD/C diagnosed?

ARVD/C is usually diagnosed by an electrocardiogram and/or echocardiogram.

ARVD/C is usually diagnosed based on a number of characteristic findings.7,8 These findings include (1) specific electrical abnormalities that can be detected in a test called an electrocardiogram (EKG), which measures the electrical activity controlling the rhythm of the heart, and (2) stretching (dilation) of the right heart chamber that can be detected by a test called an echocardiogram (ECHO), which images the heart.

How is ARVD/C treated?

ARVD/C is treated with medication and preventive surgery.

ARVD/C is often treated with medications that prevent disturbances in the rhythm of the heart. Risk of SCD may be reduced by certain lifestyle adjustments, such as avoiding intense exercise. In patients at high risk of SCD, a device called an implantable cardioverter-defibrillator (ICD) may be surgically implanted. This device automatically “resets” the heart rhythm if dangerous arrhythmias are detected. In rare cases of ARVD/C, heart transplantation may be necessary.

How can genetic testing help families with ARVD/C?

Genetic testing can help to identify family members who are at increased risk of ARVD/C and family members who are not.

Sudden death due to ARVD/C most commonly occurs in young adults who are often not aware of their condition. It is therefore important to find out if an adolescent (or young child or young
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Adult) is genetically predisposed to ARVD/C, so that regular exams can be performed. Family history of ARVD/C is a strong indicator of risk even before symptoms develop.

Genetic testing can identify the genetic defect that leads to ARVD/C in a particular family. Other members of the same family can then easily be tested for the presence of the genetic defect (called "familial mutation"). Family members who harbor the familial mutation should undergo regular heart exams, consider preventative lifestyle changes, and seek treatment early. Family members who do not harbor the familial mutation no longer have to worry about developing the disease or passing it on to their children. Neither they nor their children need to undergo the rigorous and costly exams recommended for ARVD/C patients.

More information is available from:

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Children’s Cardiomyopathy Foundation (CCF)
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C.A.R.E. Foundation
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References