TMEM43

The *TMEM43* gene is one of many genes that helps provide strength and stability to tissues in the body. The *TMEM43* gene makes a protein which is found in the heart muscle. When this protein doesn't work properly, the heart can be weakened.

Impact of *TMEM43* mutations
Individuals with a mutation in the *TMEM43* gene are at an increased risk for developing arrhythmogenic cardiomyopathy, a hereditary cardiovascular (heart and blood vessel) disorder, which can affect the heart's ability to pump blood.

Disorders associated with the *TMEM43* gene

Mutations in the *TMEM43* gene have been associated with the following disorder:

Arrhythmogenic Cardiomyopathy
Arrhythmogenic cardiomyopathy (AC) is associated with a replacement of heart tissue with fat and/or fibrous tissue, which can make it hard for the heart to pump blood.

AC, also referred to as arrhythmogenic right ventricular cardiomyopathy or dysplasia (ARVC or ARVD), is associated with a replacement of the muscle with fat and/or fibrous tissue in the minor pumping chamber of the heart, called the right ventricle. As a result, the right ventricle is enlarged (dilated) and the heart has a difficult time pumping blood, which can cause heart failure. In some cases, the major pumping chamber of the heart, called the left ventricle, can also be affected. The most common symptoms are strong or irregular heartbeats (heart palpitations), lightheadedness, chest pain or fainting due to a fall in blood pressure (syncope). People with AC can have a problem with the electrical system of the heart that controls the heartbeat's regular rhythm (arrhythmia), which can increase the risk of sudden cardiac death. Sudden cardiac death can occur, even in individuals who have no other symptoms. Most people are diagnosed between their 20s and 40s.

Diagnosing AC typically involves evaluating an individual's medical and family histories, as well as a regular physical exam, an imaging test used to see whether the heart muscle is abnormally thick (echocardiogram), and a test of the heart's electrical system called an electrocardiogram (EKG or ECG). Additional screening and diagnostic tests may be ordered, including an MRI.

Individuals with AC are advised to make certain lifestyle changes, such as avoiding strenuous exercise. Depending on whether AC symptoms are present, medications may be prescribed. Some individuals may also need a device that detects a dangerously fast heart rhythm and delivers a shock to correct it called an implantable cardioverter defibrillator (ICD) or other surgical procedures. If medications and surgical procedures are not working to manage heart failure, a heart transplantation may be considered.

Useful resources

American Heart Association
Focused on building lives free of heart disease by providing accessible education and funding innovative research.
www.heart.org

ARVD/C Patient Registry (The Johns Hopkins Hospital)
The goal of the registry is to clinically characterize AC patients and learn more about the natural history of the disorder, range of severity and the genes that cause AC.
www.hopkinsmedicine.org

Sudden Arrhythmia Death syndromes (SADS)
SADS advocates for nondiscriminatory treatment for people who are diagnosed with a SADS disorder. SADS is committed to supporting efforts that will improve the quality of life for patients with heart rhythm abnormalities.
www.sads.org

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