MYL3

The *MYL3* gene is one of many genes that helps muscles tense up (contract). The *MYL3* gene makes a protein which plays a key role in allowing muscles in the heart to contract. When this protein doesn't work properly, it decreases the heart's ability to pump blood to the rest of the body.

Impact of *MYL3* mutations
Individuals with a mutation in the *MYL3* gene are at an increased risk for developing different hereditary cardiovascular (heart and blood vessel) disorders called cardiomyopathies, which can affect the heart's ability to pump blood (cardiomyopathy). These include hypertrophic cardiomyopathy and restrictive cardiomyopathy.

Disorders associated with the *MYL3* gene

Mutations in the *MYL2* gene have been associated with the following disorders:

Hypertrophic Cardiomyopathy
Hypertrophic cardiomyopathy (HCM) is associated with an abnormal thickening of the heart muscle, which can make it hard for the heart to pump blood.

HCM is associated with an abnormal thickening (hypertrophy) of the heart muscle in the major pumping chamber of the heart, called the left ventricle. This means blood is pumped out of the heart less efficiently and blood flow may even be blocked in some individuals. Symptoms of HCM may include fatigue, shortness of breath with exertion, pounding sensations in the heart (palpitations), light-headedness, dizziness or fainting.

The majority of people with HCM will have mild symptoms and a normal life expectancy. However, in some cases, symptoms of HCM can be life-threatening. People with HCM can have a problem with the electrical system of the heart that controls the heartbeat's regular rhythm (arrhythmias), which can increase the risk of sudden cardiac death. Individuals can also develop heart failure that is potentially fatal if untreated. Age of onset and severity of symptoms may vary, even within the same family.

Diagnosing HCM 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 HCM are advised to make certain lifestyle changes, such as staying well-hydrated and avoiding strenuous exercise and certain medications. Depending on whether

HCM symptoms are present, medications that help control blood pressure may be prescribed. Antibiotics may be prescribed before certain medical and dental procedures to guard against infections in the heart. 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.

Regular visits to a cardiologist specializing in HCM are recommended in order to check that treatment is effective.

Restrictive Cardiomyopathy
Restrictive cardiomyopathy (RCM) is associated with an abnormal stiffness of the heart muscle, which can affect the heart's ability to pump blood.

RCM is associated with a replacement of normal tissue with scar tissue in the heart's pumping chambers (ventricles). When this happens, the ventricles are not able to fill with blood normally, which reduces the blood flow in the heart. This can lead to problems such as heart failure and sudden cardiac death. People can also have a problem with the electrical system of the heart that controls the heartbeat's regular rhythm (arrhythmias), including a type called heart block which causes the heart to beat too slowly. Symptoms include shortness of breath, persistent cough, strong or irregular heartbeats (heart palpitations), tiredness, dizziness, fainting due to a fall in blood pressure (syncope), chest pain, buildup of fluid in the body (edema), or nausea, bloating, and poor appetite. Blood clots may also occur. Age of onset and severity of symptoms may vary, even within the same family. Some individuals with RCM experience no noticeable symptoms, but may still be at risk for heart failure or sudden cardiac arrest.

Diagnosing RCM 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) with an experienced cardiologist. Additional screening and diagnostic tests may be ordered, including an MRI.

Treatment typically involves taking medications to reduce the risk of blood clots. 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.

Regular visits to a cardiologist specializing in RCM are recommended in order to check that treatment is effective.

Useful resources

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

Hypertrophic Cardiomyopathy Association
Provides support, education, and advocacy as well as advancing research, understanding and care to those with hypertrophic cardiomyopathy.
www.4hcm.org

SHARE Registry
Advancing the understanding of cardiomyopathy by increasing community awareness and supporting research.
https://thesharegistry.org

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