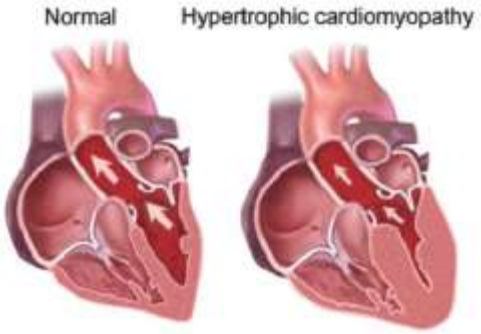

Hypertrophic Cardiomyopathy

Hypertrophic cardiomyopathy (HCM) is one of the most common heritable cardiac condition and is the most common cause of sudden cardiac arrest in the young, affecting about 1 in 500 people. Familial HCM is a heart condition characterized by thickening (hypertrophy) of the heart muscle, more specifically the ventricle. The thickened heart muscle can make it challenging to keep up with the oxygen demands of the body and of the heart muscle itself. Many people with HCM have few, if any, symptoms and can lead normal lives without significant symptoms. However, this condition can also have serious consequences. Life threatening arrhythmias resulting in cardiac arrest can sometimes be the first symptom.
Signs and symptoms of HCM may include one or more of the following:

- Shortness of breath, especially during exercise
- Chest pain, especially during exercise
- Fainting, especially during or just after exercise or exertion
- Sensation of rapid, fluttering or pounding heartbeats (palpitations)
- Heart murmur, which a doctor might detect while listening to your heart

Treatment and Management

Managing HCM requires lifelong visits with a cardiologist to screen for potential cardiac risks and obtain a careful family history. Routine testing can consist of echocardiograms, 12-lead ECGs, Holter monitoring, exercise stress tests, cardiac MRIs, or cardiac genetic testing.

Common information that may be attained from the tests:
Echocardiogram (Echo):

- The structure and function of the heart.
- The degree of thickening and shape of thickened area.
- Is the thickened area creating an obstruction of blood flow to the (aorta) artery that supplies oxygenated blood to the body from the heart?
- Is there any mitral valve involvement or mitral valve regurgitation?
- If the patient has a transvenous lead from an implanted cardiac rhythm device, how is this lead impacting the structures within the heart?

12-lead ECG (or EKG):

- Baseline rhythm
- The voltage force of the ventricle and the recovery of the heart muscle.
- If the patient is paced, the function of the device.

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Holter:

- Rhythm throughout the monitored period, including evidence of arrhythmias
- High, low, and average heart rates that may help determine medication dosing

Exercise Stress Test:

- Evidence of ischemia (decreased oxygenation to the heart muscle)
- Inducible arrhythmias throughout the test or in the post exercise period
- Heart rate and blood pressure response during exercise.

Cardiac MRI:

- Measurements of the ventricle chamber size, degree of hypertrophy, and areas of scar.

Genetic Testing:

Genetic testing identifies changes in the genes that are associated with HCM. Test results may be:

- Positive - a known genetic change that causes HCM is found,
- Negative - no known genetic change that causes HCM is found, or
- Variant of uncertain significance (VUS) - a change is found in a gene associated with HCM, but that change is not definitively known to cause HCM.

A positive test result confirms the diagnosis of HCM. A negative result indicates that a disease-causing mutation associated with HCM was not identified. Importantly, this does not rule out the possibility of having the disease. An individual may have a disease-causing mutation has not been recognized yet on the HCM panel. A variant of uncertain significance is an inconclusive finding. The genetic test results will be discussed with you by your medical team and may also be discussed by a genetic counselor.

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