



## How is Hypertrophic Cardiomyopathy Diagnosed?

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Hypertrophic cardiomyopathy may be suspected because of symptoms, family history, a murmur or an abnormal ECG/EKG. An individual with the condition may present with varied symptom including, shortness of breath (with or without exercise), palpitations, lightheadedness, fainting, nearly fainting, chest pain/pressure/discomfort, fatigue, cardiac arrest, stroke or there may be no outward symptoms at all. Because such symptoms could be caused by a large number of other conditions, further tests are necessary.

### Electrocardiogram or ECG (EKG)

An ECG records the electrical signals from the heart and is performed by placing electrodes on the chest, wrist and ankles. ECG's are very safe and non-invasive. In hypertrophic cardiomyopathy the ECG usually shows an abnormal electrical signal due to muscle thickening and disorganization of the muscle structure. In a minority of patients (approximately 5% to 10%) the ECG may be normal or show only minor changes. ECG abnormalities are also not specific to hypertrophic cardiomyopathy and may be found in other heart conditions.

### Echocardiogram or ECHO

Currently, the diagnosis of hypertrophic cardiomyopathy is made by an ultrasound scan of the heart called an "echocardiogram" or ECHO. Like the ECG this is an entirely safe and non-invasive test. An ECHO produces a picture of the heart where excessive thickness of the muscle can be measured and valves assessed. Additional equipment called "Doppler" ultrasound can produce a color image of blood flow within the heart and measure the heart's contraction and filling. Turbulent flow can be detected. Therefore ECHO provides a very thorough assessment of hypertrophic cardiomyopathy. Echocardiograms should be a complete evaluation of the entire heart; quick screens of only regions of the heart may yield misleading results. Echocardiograms should be completed by a cardiologist in a clinical environment to completely assess cardiac structure.

### Physical Examination

In the majority of patients with hypertrophic cardiomyopathy, the physical examination is unremarkable and the abnormalities may be subtle. Some patients have forceful or jerky pulse and a forceful heartbeat, which can be felt on the left side of the chest. Both of these reflect the thickened, strongly contracting heart. However, the most obvious abnormality on physical examination is a heart murmur, which is present in only 25-30% of patients.

### Genetic Testing

The role of genetic testing is evolving in HCM. It is now possible to screen for the genetic mutations that cause HCM. At this time, a family member with confirmed HCM may receive testing to see if they carry one of the known genetic mutations associated with HCM. This patient is called the "index patient" for the family. There is a 70% chance that the "index patient" would have a mutation that has already been associated with HCM. There are still mutations yet to be discovered therefore this testing may yield inconclusive results. Once a mutation is found in the "index patient", the remainder of the family can have genetic testing performed to see if they carry the familial HCM mutation.

**\*\***In some patients the line between HCM and "athletes heart" can be rather difficult to assess. In these situations it is advised to consult with a physician specializing in HCM for a more in-depth assessment.

Maron, BJ et al American College of Cardiology/European Society of Cardiology clinical expert consensus document on hypertrophic cardiomyopathy. J Am Coll Cardiol. 2003 Nov 5; 42(9):1687-713