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Cardiac Inherited Disease Registry N.Z

Hypertrophic Cardiomyopathy (HCM)

Information for Families

Background

Hypertrophic cardiomyopathy (HCM) is an inherited condition affecting the muscles of the pumping chambers of the heart (the ventricles). It can be very severe, causing sudden death or heart failure at a young age. However in most people it is relatively mild; many people with the condition may have no symptoms at all and live a long life.

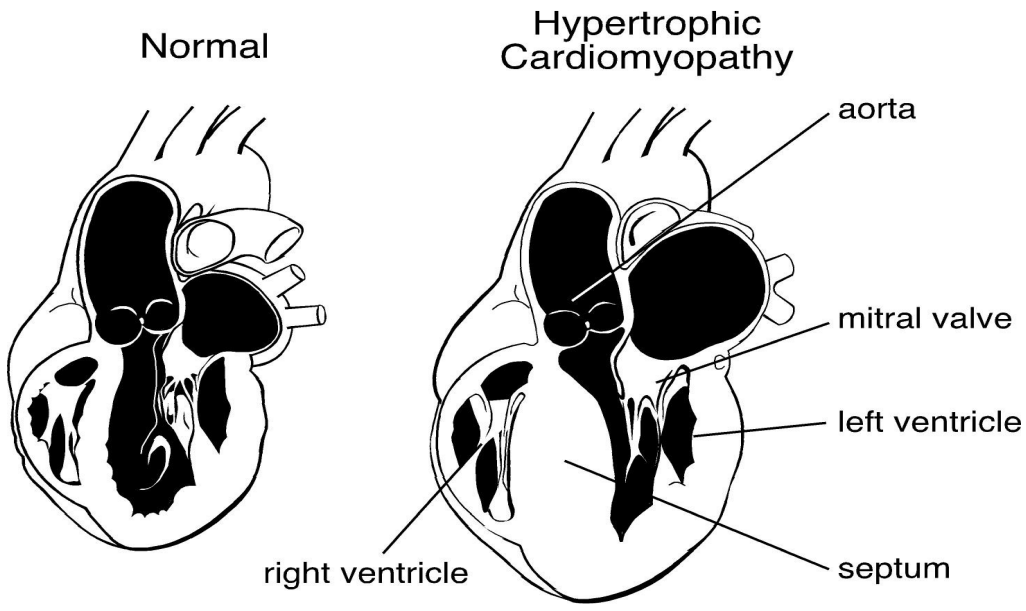
Each child born to a parent affected by, or carrying the condition, has a 50% chance of inheriting the gene which puts them at risk of developing HCM. Doctors do not yet know why some people get more severely affected than others, so it is important for people at risk to have heart tests looking for signs of the condition every few years. Lives of those severely affected can be saved by the use of defibrillator-pacemakers, and the avoidance of high-impact sporting/athletic activities and or heavy manual labour.

Familial Hypertrophic Cardiomyopathy is one of the more common cardiac inherited diseases with the at-risk genes occurring in approximately one in 500 people in the general population.

1) What is Hypertrophic Cardiomyopathy?

In hypertrophic cardiomyopathy (HCM), the growth and arrangement of the heart muscle fibres are abnormal. There is abnormal thickening of the left ventricular wall and septum (the septum is the wall which separates the right and left ventricles) and the muscle fibres are disorganised. Some individuals with hypertrophic cardiomyopathy develop an obstruction to blood flow as blood is pushed out of the heart.

The altered shape of the left ventricle can also cause the inlet valve (mitral valve) to leak.



2) What are the symptoms of Hypertrophic Cardiomyopathy?

Many individuals with HCM have no symptoms. Symptoms can appear in childhood or adulthood. Symptoms include breathlessness, fatigue or excessive tiredness, dizziness, fainting or collapse during physical activity, strong rapid heart beats which feel like a pounding sensation in the chest (palpitations) and chest discomfort that feels like heaviness in the chest (commonly occurring with exercise). In some cases the first symptom of the disease may be collapse, typically during exercise, which can be fatal.

3) How is Hypertrophic Cardiomyopathy diagnosed?

HCM is usually diagnosed with echocardiography. This is an ultrasound scan of the heart. Measurements of the thickness of the heart wall can be made. ECGs (Electrocardiograms) are also used. In some cases cardiac MRI (Magnetic Resonance Imaging) is also now be utilised especially where it is difficult to view the heart well with echocardiography.

4) Are genetic tests valuable?

In up to half of families affected by HCM, a genetic variation (mutation) can be found which is linked to the development of HCM. The first genetic test (by a simple blood sample) is done on someone in the family who definitely has HCM. Families tend to have their own unique genetic variation within 11 currently identified genes linked to HCM (more will be discovered in the future). If this variation is found in a person known to have HCM in a family; then this information can be used to test for that abnormality in other family members if they would like to know whether they carry the genetic variation or not.

If no genetic change is found in the affected family member then this does not necessarily mean that the HCM is not familial (running in the family), rather it is more likely to be due to limitations in our knowledge of the genetic cause of HCM at this time. Other family members should still be seen by a cardiologist to discuss appropriate surveillance, and those people without symptoms should still be evaluated by a Cardiologist since it is possible to carry the condition but have no symptoms.

The diagnosis of HCM itself does not rely on the genetic test – rather it is a clinical diagnosis, made by the heart specialist on review of the heart tests (ECG and Echocardiogram).

5) Who should have a genetic test?

This is a choice made by the patient after taking proper medical advice.

Absence of the genetic change which has been linked to HCM in a family will mean that person has no increased risk of developing HCM than the general population; repeated heart tests are then not usually necessary. They may then take part in sporting activities and they will know that their children are not at increased risk of developing HCM.

Presence of the genetic change linked to HCM in the family means that they might develop the disease during their life. The test gives no indication of likely severity of the disease, should it develop, or the risk of sudden death. Screening tests (heart ultrasound and ECG) are then recommended every few years. Presence of the genetic change also means that each of their children will have a 50% risk of carrying the same at-risk gene.

In the end, it is the choice of the individual. Each family member will need to consider the pros and cons carefully, after receiving appropriate genetic counselling.

6) The role of family registries in Hypertrophic Cardiomyopathy (HCM)

A registry of patients and families with HCM allows appropriate clinical and genetic screening for HCM throughout New Zealand. It helps to ensure all the relevant family members undergo appropriate screening. A centralised registry allows us to contact patients with medical updates and/or offers of research participation. Collecting anonymised data about the families (eg. how many people in New Zealand have this condition) allows informed decision-making by health funding bodies.

7) Treatment for Hypertrophic Cardiomyopathy

There is no cure currently available for Hypertrophic Cardiomyopathy, but many people need no treatment. For those that do, treatment options include medications that help to relieve the symptoms of exercise intolerance, palpitations and chest discomfort, Implantable defibrillators (ICDs) to help regulate the heart beat and prevent sudden death due to rapid chaotic heart rhythms, and operations to relieve obstruction to blood flow. These treatment options are very specific to the individual; your specialist will discuss these options with you and recommend the best options for dealing with your particular case.

8) Further Information

The Cardiomyopathy Association of Australia Ltd is a non-government organisation providing information, support and advocacy for people living with cardiomyopathy and their families and supporting research into Cardiomyopathy. www.cmaa.org.au.

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9) Participation in Cardiac inherited disease registry:

If you would like more information prior to deciding whether you would like to join the Cardiac inherited disease registry, please feel free to contact us.

If you have any queries or concerns regarding your rights as a participant in the Cardiac Inherited disease registry you can contact an independent Health and Disability Advocate. This is a free service provided under the Health & Disability Act; through the offices of the Health and Disability Commissioner.

Telephone (NZ wide): 0800 555 050
Free Fax (NZ wide): 0800 2787 7678 (0800 2 SUPPORT)
Email: advocacy@hdc.org.nz

This registry has received Ethical Approval from the N.Z. Multi-centre Ethics Committee: AKX/02/00/107