Australian Genetic Heart Disease Registry
Information Sheet

Last updated 27 January 2016

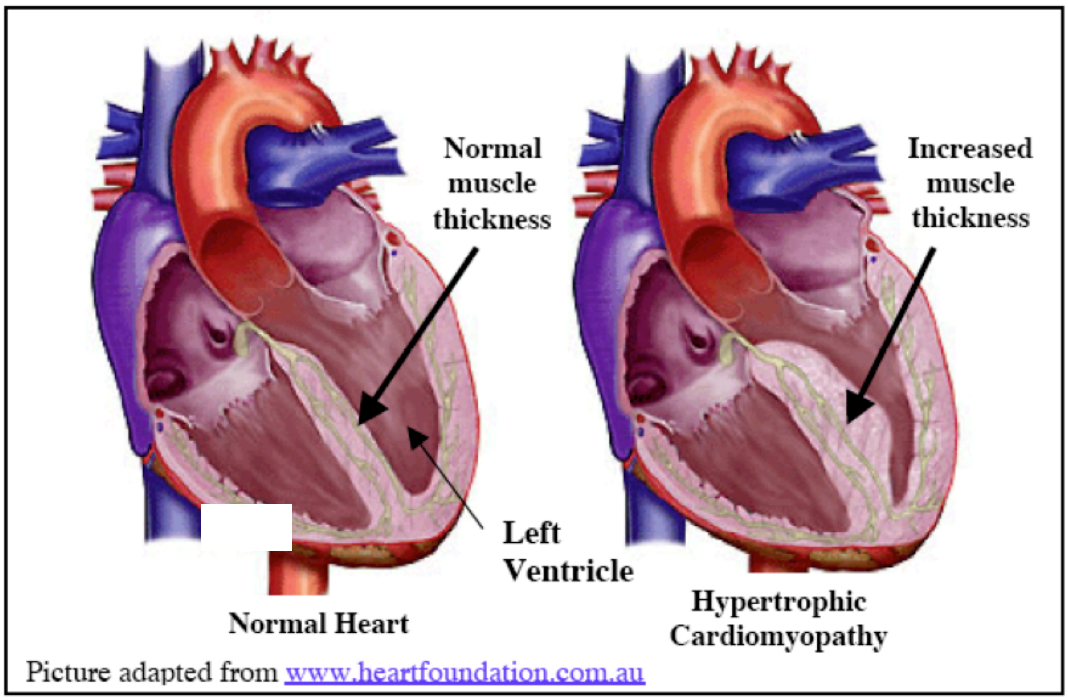
AUSTRALIAN GENETIC HEART DISEASE REGISTRY

Hypertrophic Cardiomyopathy (HCM)

What is hypertrophic cardiomyopathy?

Hypertrophic cardiomyopathy (often shortened to HCM) is an inherited condition. It leads to abnormal thickening of the heart muscle, most often of the left ventricle (the main pumping chamber of the heart). The thickened muscle creates problems because it causes the heart to work less efficiently. Therefore, some people may experience chest pain, shortness of breath, dizziness, fainting episodes or palpitations. Occasionally, the condition may cause sudden death.

In adults, the walls of the left ventricle are usually 7 to 10 mm thick. To be diagnosed with hypertrophic cardiomyopathy the walls need to measure 15 mm or more. If you have a family member with HCM then to walls need to measure 13mm or more. The wall thickening is often unevenly distributed, unlike in people with high blood pressure. This is illustrated in the diagram below. In about 25% (1 in 4) of people with hypertrophic cardiomyopathy there is obstruction to the blood being pumped out of the heart because of the thickened muscle. This is known as the obstructive form of hypertrophic cardiomyopathy (sometimes referred to as HOCM).



Picture adapted from www.heartfoundation.com.au

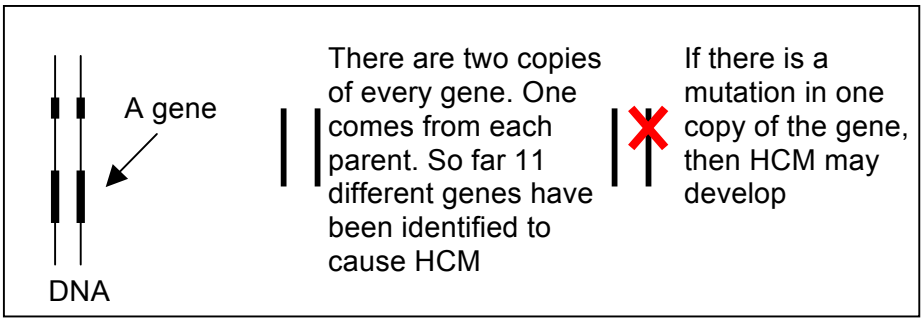
It is thought that up to 1 in 200 people have hypertrophic cardiomyopathy, and many never have any symptoms. Tragically, in some cases the first sign of this condition is sudden death. ***Therefore, it is very important that everyone with a family history of hypertrophic cardiomyopathy be screened by a cardiologist.***

What causes hypertrophic cardiomyopathy?

Our body is made up of millions of cells. There are many different types of cells, including brain cells, liver cells and heart cells to name a few. Each cell contains 46 chromosomes, which hold the genetic material that decides features such as the colour of our eyes and whether we are tall or short. These 46 chromosomes are grouped into 23 pairs, one of each pair coming from mum and the other from dad. One of these pairs is known as the sex chromosomes, and these decide whether we are male or female.

If you imagine a chromosome as being like a ball of wool, you could stretch it out into one long strand, which is known as the DNA. Along the length of DNA there are regions called genes. As there are two copies of every chromosome, there are also two copies of every gene (as mentioned, one from each parent).

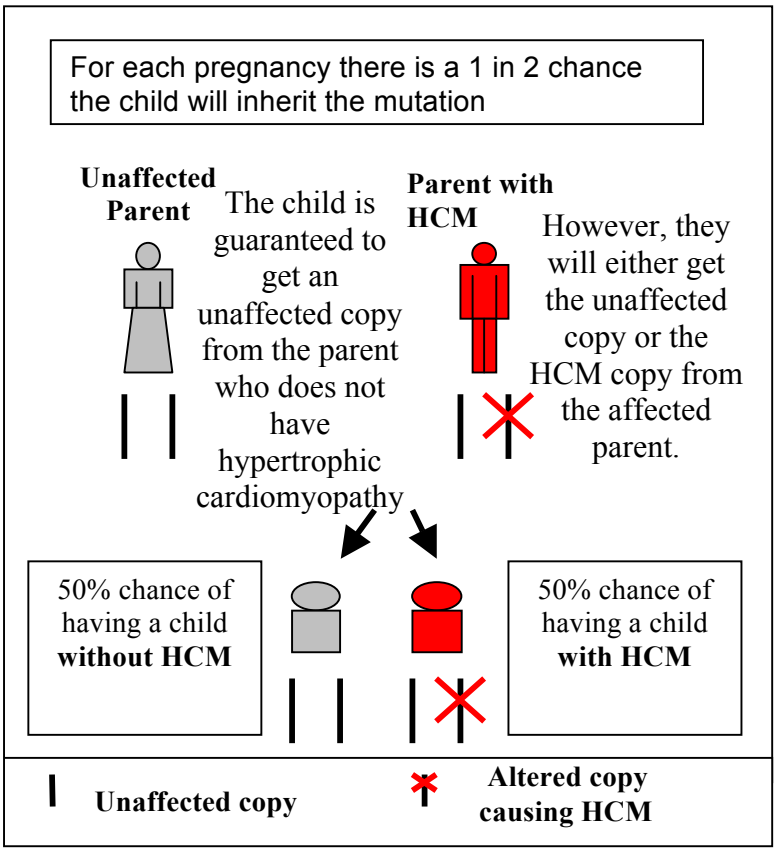
Genes act as recipes to make certain things in the body, and each recipe is unique based on the order of units it is made up of. If there is a mistake in one of these genes (a mix-up in the order of these units) it may lead to the development of hypertrophic cardiomyopathy. This mistake is known as a gene alteration.



In the case of hypertrophic cardiomyopathy, we know of alterations in at least 11 genes can cause the disease. If there is an alteration in one of these genes hypertrophic cardiomyopathy may be the result.

How is HCM inherited?

Hypertrophic cardiomyopathy is known as an autosomal dominant condition, whereby the gene with the alteration "dominates" over the unaffected gene, causing the disease. This means that an affected person has inherited one altered HCM copy of the gene from one parent, and one unaffected copy from the other parent. Likewise, if a person who carries this HCM copy of the gene has a child, then **the chance that the child will be affected is 50%, or 1 in 2**. This is because they are guaranteed to get a unaffected copy of the gene from the unaffected parent, and they can either get a altered (HCM) copy or a unaffected copy from the parent with the condition. This is illustrated in the diagram.



Is it possible to have a gene alteration that's not inherited?

In rare instances, a person may have a gene alteration that has not been inherited from their parents. Instead, the alteration has arisen some time after conception. This means that their brothers and sisters are not likely to develop the hypertrophic cardiomyopathy, however, the gene alteration can still be passed down to any children of the affected person.

What is genetic testing?

Genetic testing involves looking for a mistake in the genes known to cause hypertrophic cardiomyopathy. It is a long and difficult task that has been likened to looking for a single spelling mistake in an encyclopaedia set or very large instruction manual. This is because most families have their own unique gene alteration that may exist in one of the genes we have already identified, or even in a gene not yet discovered to be involved in this condition. A genetic result may take more than 3 months.

If genetic testing does not reveal a gene alteration then we call it an inconclusive test result. This means that a gene alteration most likely exists, but either our current technology is not yet good enough to detect the alteration, or the alteration is in a gene which has not yet been identified. At present, alterations are identified in about 50% of cases.

Once the gene alteration is identified in the affected person we can then offer a gene test to other family members. This test takes 2-4 weeks for a result, and will quickly determine if the individual has inherited the HCM copy or the unaffected copy of the gene. This is called preclinical genetic testing, and allows a person to know if they are likely to develop the condition even before symptoms are present. If they do not carry the gene alteration then we can say that they will not develop hypertrophic cardiomyopathy, and future ECG's and ultrasounds are not necessary. If they are shown to have the HCM gene alteration then there is a good chance they will develop the condition, and close follow-up would be recommended.

Genetic testing of the known HCM genes has recently become a commercial test. This means that there is a cost involved for the patient and testing needs to be organised on their behalf by a doctor. If you would like to know more about centres offering commercial genetic testing, please look at our Information Sheet on Cardiac Genetic Services or contact the Registry Coordinator on registry@centenary.org.au or (02) 9565 6185 or speak to your regular cardiologist or doctor.

Genetic testing can affect applications for life insurance. If you would like to know about how genetic test results could affect your application for Life Insurance, please visit http://www.genetics.edu.au/Information/Genetics-Fact-Sheets (fact sheet #23A).

Research into hypertrophic cardiomyopathy (HCM)

In addition, the Agnes Ginges Centre for Molecular Cardiology, Sydney is continuing HCM research to identify new genes and modifying factors (e.g. understanding why some people die suddenly while others live a normal life with minimal symptoms). If you would like to find out more about this research and potentially provide a blood sample for this research, please contact Ms Laura Yeates Cardiovascular Genetics Coordinator on 02 9565 6187.

Will a diagnosis of hypertrophic cardiomyopathy change my lifestyle?

There are key recommendations we advise people about when they are diagnosed with hypertrophic cardiomyopathy (HCM). These guidelines help to reduce the risk of the most devastating outcome – sudden cardiac death.

- **Avoid competitive sports and strenuous physical activity:** Competitive sports include those that require significant exertion, for example: Touch football, Basketball, Netball, Squash, running and even social games that you may not think are too strenuous. We recommend people avoid competitive sports because it has been observed that a large proportion of the young people who die suddenly due to hypertrophic cardiomyopathy, die during or just after exercise.
- **Regular light exercise:** Although there is a lot of emphasis on avoiding sport, it is still important to maintain a healthy lifestyle. Regular light exercise such as walking is not only good for your heart, but also good for your general health.
- **Regular check-ups with your cardiologist:** It is important to monitor the progression of your condition, and you should also report any new symptoms. This is because we use many different factors to determine whether you are at an increased risk of sudden death. Your risk of sudden death determines what mode of treatment will most benefit you.
- **Surgical procedures:** You should inform the attending doctor of your condition before any surgical or dental procedures.
- **Consult with your GP or cardiologist before taking any new medications:** It is important to talk to your doctor before taking any new medications, including over-the-counter (OTC) medications, complementary medicines, natural products and herbal remedies.
- **Avoid excessive amounts of alcohol:** Alcohol can have adverse effects on the body and can worsen the symptoms of hypertrophic cardiomyopathy.
- **Encourage relatives to be screened:** As mentioned, siblings and children of an affected person have a 1 in 2 (50%) chance of being affected. Screening of relatives involves seeing a cardiologist to have a physical examination, an ECG and an Echocardiogram (ultrasound of the heart). The relative must inform the doctor there is a family history of hypertrophic cardiomyopathy to ensure no signs are missed. Find a specialist cardiac genetic service like the Genetic Heart Disease Clinics held at RPAH's Sydney Heart Centre (tel 02 9517 2011). See our Information Sheet on Cardiac Genetic Services to find your local service or speak to your regular cardiologist about screening family members.

Further information:

If you have any further questions, please contact:

Your local GP or cardiologist

The Cardiomyopathy Association of Australia www.cmaa.org.au
For your nearest contact person, contact the Heart Foundation's Heartline on 1300 362 787

Australian Genetic Heart Disease Registry www.heartregistry.org.au
Tel: 02 9565 6185 or email registry@centenary.org.au
Visit the Registry website today to find out how to enrol. Your participation can help save and improve lives by improving our knowledge of genetic heart conditions in Australia.

Other helpful websites:
www.4hcm.org/home.php Hypertrophic Cardiomyopathy Association (USA)
www.americanheart.org American Heart Association
www.cardiomyopathy.org Cardiomyopathy Association (UK)