HOW IS GENETIC TESTING DONE?

Genetic testing involves taking a sample of DNA from blood, saliva or tissue and analysing the genes that are most commonly known to cause HCM.

When testing a family, we begin by testing a person known to have HCM. If a mutation is identified in that individual, then testing can be performed on other family members to look for that specific mutation or mutations.

Genetic testing is complex; it takes place with genetic counselling and the process of informed consent.

If your family history suggests HCM, please talk to your doctor. A referral to Curie Genetics can help you find out more about HCM and genetic testing.

GENETIC COUNSELLING

The diagnosis of HCM has implications for other family members. A trained genetic counsellor will review your family history and discuss implications of the diagnosis with you and your family.

In addition, the genetic counsellor can serve as a resource to other family members who may have questions about the diagnosis. It is important to us that you understand all aspects of your condition and we are always available to answer questions.

REFERRAL INFORMATION

Address 📍: CURIE Genetics
(Mount Elizabeth Novena Hospital #10-37)
Singapore 329563

Telephone 📞: 6592 8615

Whatsapp : 8683 3767

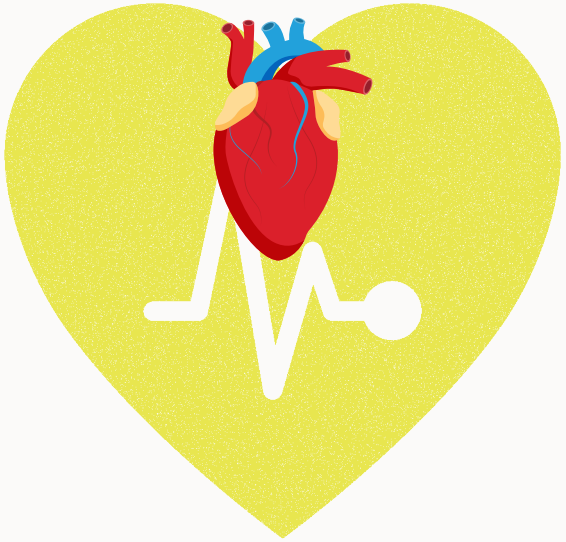
Website : www.curiegenetics.sg

Adapted content from Brigham and Women's Hospital Cardiovascular Genetics Center

ABOUT 30-60% OF

HYPERTROPHIC CARDIOMYOPATHY (HCM)

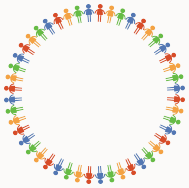
PATIENTS HAVE AN IDENTIFIABLE PATHOGENIC OR LIKELY-PATHOGENIC GENETIC VARIANT



CURIE GENETICS

BY CURIE ONCOLOGY

UNDERSTANDING HCM

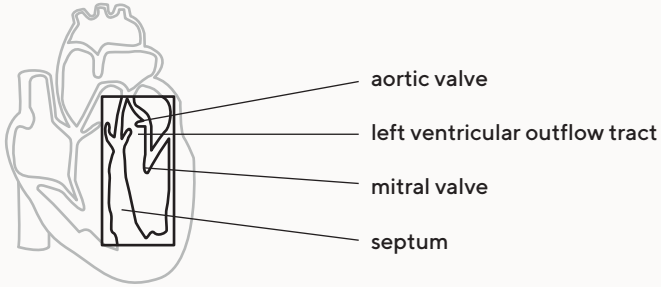


HCM is one of the most common inherited heart diseases, affecting about 1 in 500 to 1 in 1000 people.

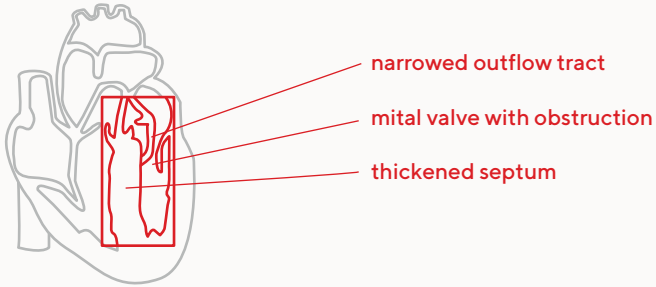
The disease is characterized by an abnormal increased thickening in the wall of the left ventricle (called left ventricular hypertrophy or LVH), the main pumping chamber of the heart.

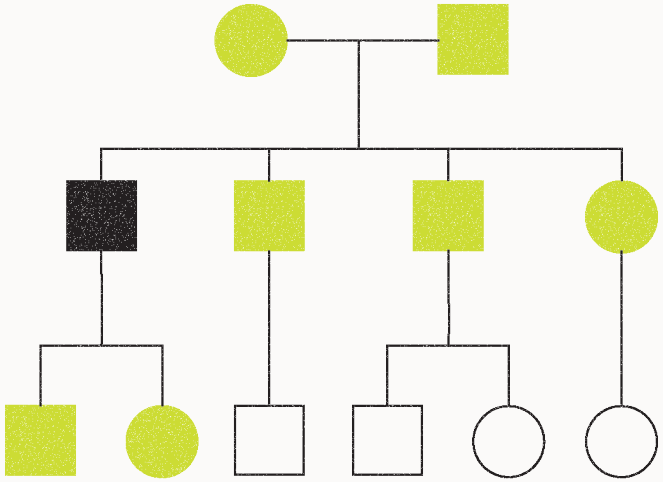
With HCM, the heart muscle may become stiff, making it difficult for it to relax. As a result, the heart requires higher pressure than normal to fill, which consequently can lead to shortness of breath, chest pain and dizziness.

NORMAL HEART



HCM HEART





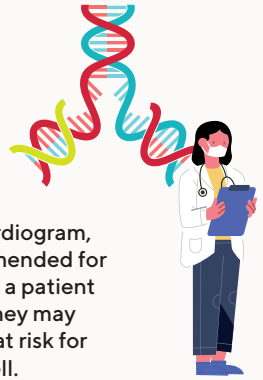
Genetic studies have identified HCM as a disease of the sarcomere, caused by mutations in any of 17 different genes.

To date, over 500 individual mutations have been identified. Identification of genes for HCM has made preclinical diagnosis possible in families with a known mutation.

Genetic testing and counseling services are available to symptomatic HCM patients and asymptomatic individuals with a known family history.

AM I OR MY FAMILY AT RISK OF HCM?

Apart from the treatment of your HCM, another important step in the management is to assess the risk in your family, including future generations. It is important to ask family members (parents, siblings, aunts, uncles, grandparents, cousins, children) if there are any cases of heart problems, sudden death, unexplained deaths and/or major health problems.

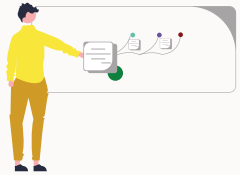


Physical examination, electrocardiogram, and echocardiogram are recommended for all immediate family members of a patient diagnosed with HCM because they may carry the gene mutation and be at risk for HCM, even if they are feeling well.

BENEFITS OF GENETIC TESTING

- Confirmation of a diagnosis

- Provide insights on underlying cause of heart disease



- To begin with risk reduction interventions by making informed medical decisions

- Identify at-risk family members for cascade screening