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My46 Trait Profile

Hypertrophic cardiomyopathy

Hypertrophic cardiomyopathy is a condition in which the heart muscle becomes thickened and may not pump blood effectively. It can be caused by mutations in any one of many different genes.

Characteristics of Hypertrophic cardiomyopathy

Hypertrophic cardiomyopathy (HCM) is the most common inherited cardiovascular condition (i.e., a disorder involving the heart and blood vessels) affecting about 1 in 500 to 1 in 1000 people. It is also the most common cause of sudden cardiac death in athletes. HCM is characterized by abnormally increased thickening of the muscular walls of the left lower chamber of the heart (also known as left ventricular hypertrophy or LVH). In HCM, LVH occurs in the absence of other known causes of LVH, including long standing high blood pressure and valve disease.

HCM is very variable and can range from no symptoms to progressive heart failure, even within the same family. Common symptoms may include shortness of breath, particularly with exertion, chest pain and dizziness. In some people, there may be obstruction to the flow of blood out of the heart. This obstruction may lead to the symptoms described above and produce a heart murmur. Most people with HCM will have normal life expectancy and symptoms that can be readily controlled. However, HCM can cause important problems, including heart failure and an increased risk of dangerous heart rhythm disturbances which may result in sudden death.

Diagnosis/Testing

The diagnosis of HCM is most often established by clinical tests such as EKGs (tests that checks for problems with the electrical activity of the heart), echocardiograms (ultrasounds of the heart) and/or MRI.

Approximately 50% of individuals with HCM have at least one relative with HCM. In approximately 50-60% of these familial cases, a change or mutation in a gene known to make different components of the cardiac sarcomere (the basic unit of muscle contraction in the heart) is found. The cardiac sarcomere genes are a group of at least 14 genes that are responsible for coordinating the contraction and relaxation of the heart muscle. Identifying a mutation can confirm a diagnosis of HCM in an affected individual and identify currently unaffected family members who are at risk for disease development. However, knowing the gene mutation in a person does not provide any information about when the condition will develop or how mild or severe it will be for that person.

Management/Surveillance

Management of HCM includes regular surveillance by a cardiologist. This surveillance often includes cardiac imaging, exercise testing, and assessment for the risk of sudden cardiac death. Medical therapy may be used to manage symptoms. More advanced therapies may be necessary in cases of heart failure, heart rhythm disturbances and/or obstruction to blood flow.

Management of patients with HCM also includes screening recommendations for their at-risk relatives. These individuals should get periodic evaluation by a cardiologist including an EKG and echocardiogram to look for any signs of HCM. Because HCM is variable it is possible that family members have HCM and are not aware because they have no symptoms.

Mode of inheritance

HCM is inherited in an autosomal dominant pattern. This means inheriting one mutation increases the chance to develop HCM. The mutation can be inherited from an affected parent or it can occur brand new (de novo) in an affected child.

Risk to family members

The risk to family members depends on whether or not the individual with HCM has a parent affected with HCM. If a parent also has the condition, the risk of having a child with HCM is 50% with each pregnancy. If a parent does not have HCM, the risk of other siblings being affected is very low.

Special considerations

None

Resources

Hypertrophic Cardiomyopathy Association
http://www.4hcm.org
The Cardiomyopathy Association
http://www.cardiomyopathy.org
Children's Cardiomyopathy Foundation
http://www.childrenscardiomyopathy.org
Cardiac Inherited Diseases Group
http://www.cidg.org/webcontent/cidg/Home/tabid/53/Default.aspx
Genetics Home Reference: Familial hypertrophic cardiomyopathy
http://ghr.nlm.nih.gov/condition/familial-hypertrophic-cardiomyopathy

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Updated: mm/yyyy **Edited by:** Seema Jamal, MSc, LCGC