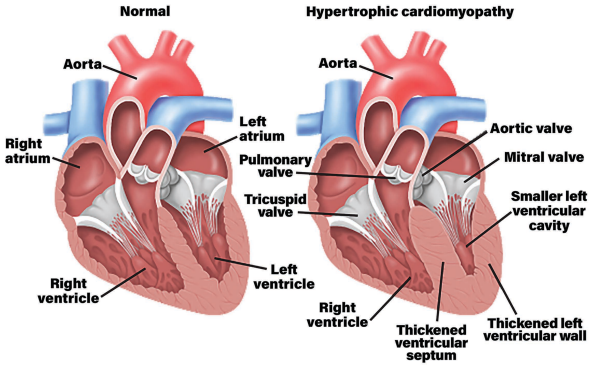
Hypertrophic Cardiomyopathy

Other names: HCM **Gene involved:** *MYBPC3*

What is Hypertrophic Cardiomyopathy?

The heart is a specialized muscle that pumps the blood through the lungs and the body. In Hypertrophic Cardiomyopathy (HCM) the heart muscle cells become enlarged. The word "hypertrophy" means enlargement. This enlargement of the heart muscle cells results in a thickening of the walls of the heart. The thickened walls make the heart stiff and prevent it from filling and pumping properly, especially with exertion. The thickening can also lead to abnormal and fast heartbeats, which can result in blackouts and in rare cases sudden death. The amount of hypertrophy that people with HCM develop varies greatly, even among members of the same family.



The heart on the left is normal. The heart on the right has hypertrophic cardiomyopathy. The condition causes the walls of the lower chambers of the heart (called ventricles) to thicken.

What are the signs and symptoms?

HCM varies widely in its initial symptoms and in how the disease progresses. People with one spelling change in MYBPC3 can have thickened heart muscle without symptoms. Others have chest pains, shortness of breath, irregular heartbeats, and blackouts. Some develop worsening heart failure and can have difficulty working and with day-to-day activity. Sudden death can be a rare presentation of HCM without prior chest pain or shortness of breath. It is important for anyone with a single MYBPC3 spelling change to have yearly checkups after age 10.

Management

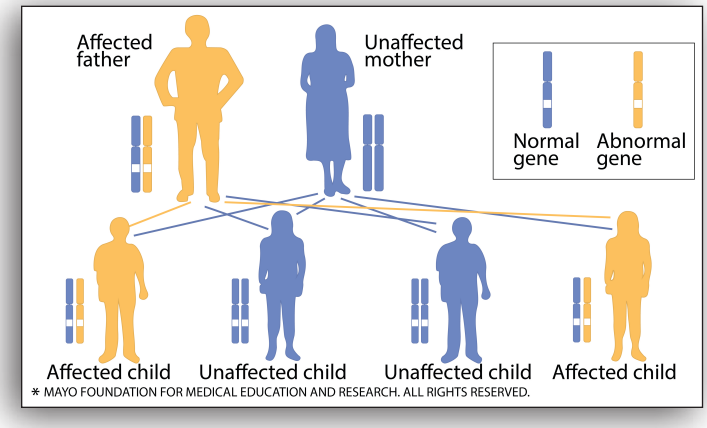
Although there is no cure for HCM, it can be managed. Early detection is important in trying to prevent severe symptoms. A gene test can determine if someone has the misspelled gene commonly causing HCM, even before the onset of symptoms. Echocardiograms (pictures of the heart) and EKGs (to look at the heart rhythm) are done regularly, usually at least once per year. Medications can treat symptoms by improving the function of the thickened heart muscle and preventing abnormal heart beats. In some patients, surgery may be used to remove portions of the thickened heart muscle. If necessary, a defibrillator (pacemaker-like device) may be implanted under the skin to treat dangerous heart rhythms.

Diagnosis

A gene test is required to make the diagnosis. To arrange this, please see the contact information below.

Autosomal Dominant Inheritance

Genes are instructions for growth and development. We have two copies of each gene. One copy is inherited from our mother, and one copy is inherited from our father. People with HCM have a spelling change in ONE copy of their MYBPC3 gene. If one parent has the MYBPC3 spelling change, there is a 1 in 2 (50%) chance that each of their children will inherit the spelling change and potentially develop HCM.



In some families both parents have the MYBPC3 spelling change. In that case, each of their children has a:

- 1 in 4 (25%) of inheriting the spelling change in the MYBPC3 gene from each parent. Without having a normal copy of the gene, these children develop severe HCM with serious heart problems and usually do not live beyond one year of age.
- 1 in 2 (50%) chance that each of their children will inherit one copy of the spelling change and potentially develop HCM.
- 1 in 4 (25%) chance they will inherit two normal copies of the MYBPC3 gene and will not be affected.

This informational pamphlet is a project of New Leaf Center - a member of the Plain Community Health Consortium (PCHC)

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Central Pennsylvania Clinic (Belleville, PA) **717-935-2065**
Clinic for Special Children (Strasburg, PA) **717-687-9407**
DDC Clinic (Middlefield, OH) **440-632-1668**
Midwest Genetics Network (Okemos, MI) **517-324-8300**
The Community Health Clinic (Topeka, IN) **260-593-0108**
UPMC Children's Hospital of Pittsburgh (Pittsburgh, PA) **412-692-5070**
WeCare Clinic - Medical Care for Special Needs (Pembroke, KY) **270-962-7383**