Harvard Heart Letter

Hypertrophic cardiomyopathy: Who has an inherited risk?

Genetic testing can help doctors guide care for families with a history of heart disease.

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Having a family member with heart disease—especially when it shows up at a young age—is a warning sign that you too may be at risk. The more common maladies such as high blood pressure and coronary artery disease are influenced by an array of different genes and compounded by lifestyle choices and environmental factors. However, certain relatively rare conditions stem from only one or a few faulty genes with powerful disease-causing effects. The most common of these inherited heart conditions is hypertrophic cardiomyopathy (HCM), which affects up to one in every 500 people.

What is HCM?

Best known for cutting down young athletes in their prime, HCM causes an abnormal thickening of the heart wall. In rare cases, the first inkling of disease may be fainting or sudden death due to an abnormal heart rhythm. More often, symptoms such as shortness of breath or chest pain with exercise develop over time.

The key feature of inherited HCM is that the thickening of the heart muscle is unexplained, says Dr. Carolyn Y. Ho, medical director of the cardiovascular genetics center at Harvard-affiliated Brigham and Women's Hospital. "We wouldn't leap to the diagnosis of HCM if we saw these changes in an older person with a history of high blood pressure. But if it occurs early in life without the presence of other triggering factors, we would think about HCM and look for a familial pattern," she says.

The detective work

The genetic risk for HCM is passed from one generation to the next by way of dominant-acting mutations in genes governing the structure of the heart muscle. That means that first-degree relatives (parents, siblings, and children) of an affected person have a 50% chance of having inherited the same mutation. Before genetic testing became widely available, doctors had only this information to help guide families. Now, it's possible to determine if a family member is at risk for developing HCM even before the disease can be clinically diagnosed.

"When you decide do genetic testing, the implications go beyond just the individual being tested," says Dr. Ho. The most efficient approach is first to do comprehensive gene sequencing on the person with the most serious manifestation of disease. This gives the best chance of uncovering the most important disease-causing mutations. If an HCM mutation is found, other family members can undergo more limited—and less costly—screening to look for that specific mutation.

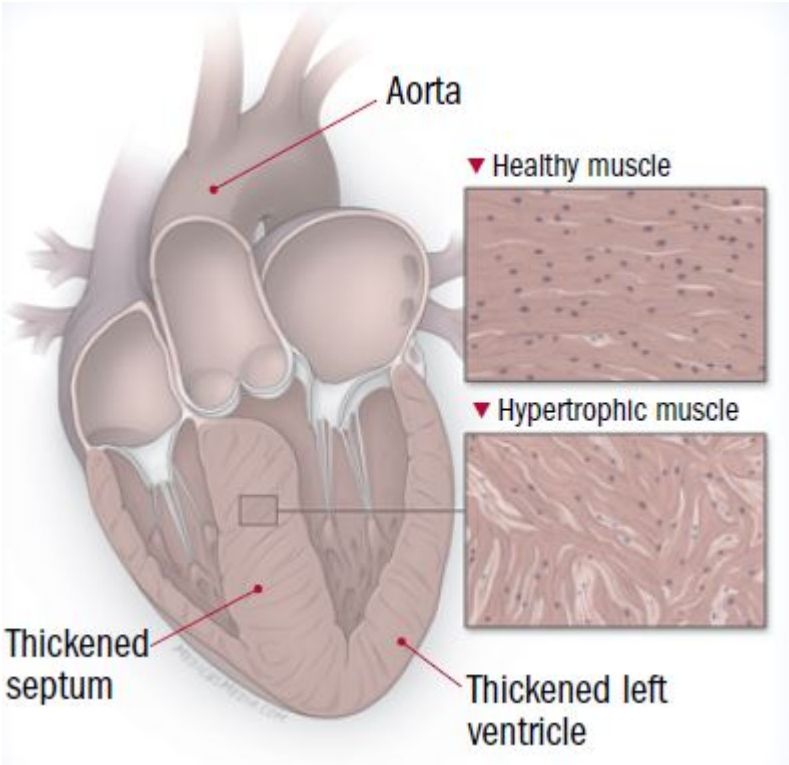
Decision making in the family

In that scenario, if you have a negative result showing that you do not carry the problem mutation, you can feel relatively reassured that you are not at increased risk for developing the disease. Nor do you need to worry about passing the trait to your offspring. However, Dr. Ho cautions, anyone with a strong family history of HCM should get checked out by a cardiologist, regardless of the results of a genetic screening, if a heart murmur or other unexplained heart symptom develops.

A positive result indicates that the relative has inherited the mutation and is at risk for developing HCM. But, as of now, we cannot predict when, or even if, HCM will develop or how severe it will be, says Dr. Ho. Therefore, relatives who have inherited the mutation are advised to undergo clinical screening on a regular basis to detect the earliest signs of heart muscle changes. This includes having an echocardiogram (heart ultrasound) every year during adolescence and young adulthood, when HCM most commonly develops. Even a person who has remained symptom-free into middle age should be screened occasionally because the disease has been known to crop up later in life.

Although there currently aren't any therapies to prevent or delay HCM, much can be done to help individuals live comfortably with the disease and treat potential problems as early as possible. That is why Dr. Ho stresses that families at risk should be followed by a team of doctors experienced with HCM. She and others are investigating how gene mutations cause HCM with the goal of discovering ways to delay or prevent the disease.

Hypertrophic cardiomyopathy



Classic signs of HCM include thickening of the muscular wall separating the right and left sides of the heart (septum) and the heart's main pumping chamber (left ventricle). Instead of neatly lining up, muscle cells are irregular and disorganized. The thickened septum can restrict blood flow out of the left ventricle.

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