Hypertrophic cardiomyopathy (HCM) is a disease in which the heart muscle (myocardium) becomes abnormally thick — or hypertrophied. This thickened heart muscle can make it harder for the heart to pump blood. Hypertrophic cardiomyopathy may also affect the heart's electrical system.

Hypertrophic cardiomyopathy often goes undiagnosed, because many of those with hypertrophic cardiomyopathy have few, if any, symptoms. In a small number of people with this condition, the thickened heart muscle can cause signs and symptoms, such as shortness of breath and problems in the heart's electrical system resulting in life-threatening abnormal heart rhythms (arrhythmias).

Fortunately, people with hypertrophic cardiomyopathy often lead normal lives with no significant problems.

Symptoms

Hypertrophic cardiomyopathy symptoms include:

- Shortness of breath, especially during exercise or exertion
- Chest pain, especially during exercise or exertion
- Fainting, especially during exercise or exertion
- Dizziness
- Fatigue
- Heart palpitations — the sensation of rapid, fluttering or pounding heartbeats

Causes

Hypertrophic cardiomyopathy is usually caused by gene mutations. It's thought these mutations cause the heart muscle to grow abnormally thick. People with hypertrophic cardiomyopathy also have an abnormal arrangement of heart muscle fibers. The heart muscle cells become jumbled, known as myofiber disarray. This disarray can contribute to an irregular heartbeat (arrhythmia) in some people.

The severity of hypertrophic cardiomyopathy varies widely. Most people with hypertrophic cardiomyopathy have a form of the disease in which the wall (septum) between the two bottom chambers of the heart (the ventricles) becomes enlarged and obstructs blood flow. This is sometimes referred to as hypertrophic cardiomyopathy with obstruction or hypertrophic obstructive cardiomyopathy.

Sometimes hypertrophic cardiomyopathy occurs without significant obstruction of blood flow. However, the heart's main pumping chamber (the left ventricle) may become stiff, which reduces how much blood the ventricle can hold and how much blood gets pumped out to the body with each contraction. Doctors sometimes refer to this as hypertrophic cardiomyopathy without obstruction or nonobstructive hypertrophic cardiomyopathy.

Risk factors

Hypertrophic cardiomyopathy affects men and women equally.

The condition is usually inherited. There's a 50 percent chance that the children of those with hypertrophic cardiomyopathy will inherit the genetic mutation for the disorder. Siblings of those with hypertrophic cardiomyopathy also are at risk. As a result, close relatives of someone with hypertrophic cardiomyopathy are urged to talk to their doctors about getting screened for the disease.

Complications

In many people, hypertrophic cardiomyopathy doesn't cause significant health problems. However, in some people, hypertrophic cardiomyopathy can cause severe signs and symptoms, such as shortness of breath, chest pain or fainting.

People with hypertrophic cardiomyopathy are at risk of dangerous abnormal heart rhythms (arrhythmias), such as ventricular tachycardia or ventricular fibrillation. These abnormal heart rhythms can cause sudden cardiac death. Hypertrophic cardiomyopathy is the leading cause of heart-related sudden death in people under 30. Fortunately, such deaths are rare.

Possible complications of hypertrophic cardiomyopathy include:

- **Arrhythmias.** Thickened heart muscle, as well as the abnormal structure of heart cells (disarray), can disrupt the normal functioning of the heart's electrical system, resulting in fast or irregular heartbeats. Atrial fibrillation, ventricular tachycardia and ventricular fibrillation are among the arrhythmias that may be caused by hypertrophic cardiomyopathy.

  The most dreaded risk of hypertrophic cardiomyopathy is sudden cardiac death due to ventricular tachycardia or ventricular fibrillation. Unfortunately, it can be difficult to predict which people with hypertrophic cardiomyopathy are most prone to these life-threatening, abnormal heart rhythms. If you experience fainting spells, extreme dizziness or prolonged palpitations, you should seek immediate medical care.

- **Obstructed blood flow.** In many people, the thickened heart muscle causes obstruction to blood flow leaving the heart. This can lead to shortness of breath with exertion, chest pain, dizziness and fainting spells.
- **Mitral valve problems.** The thickened heart muscle can leave a smaller space for blood to flow, which in turn causes blood to rush through your heart valves more quickly and more forcefully. This increased force can prevent your mitral valve — the valve between your heart's left atrium and left ventricle — from closing properly. As a result, blood can leak backward into the left atrium. This is called mitral valve regurgitation. Mitral valve regurgitation can lead to other complications, such as heart failure or arrhythmias.
- **Heart failure.** Heart failure means your heart can't pump enough blood to meet your body's needs. The thickened heart muscle of hypertrophic cardiomyopathy can eventually become too stiff to fill effectively, which can lead to shortness of breath and heart failure.
- **Dilated cardiomyopathy.** Over time, thickened heart muscle may become weak and ineffective. The ventricle becomes enlarged (dilated), and its pumping ability becomes less forceful.

Prevention

Because hypertrophic cardiomyopathy is inherited, it can't be prevented. However, doctors and scientists are learning more about the genetic mutations that cause the disorder. Though the condition itself can't be prevented, it's important to identify this condition as early as possible to guide treatment and prevent complications.

Preventing sudden death

The use of an implantable cardioverter-defibrillator has been shown to help prevent sudden cardiac death, which occurs rarely in those with hypertrophic cardiomyopathy.

Unfortunately, because many people with hypertrophic cardiomyopathy don't realize they have it, there are instances where the first sign of a problem is sudden cardiac death. These cases can happen in seemingly healthy young people, including high school athletes and other young, active adults. News of these types of deaths generates understandable attention because they're so unexpected, but parents should be aware these deaths are quite rare.

Still, experts in heart abnormalities generally recommend that those with hypertrophic cardiomyopathy not participate in most competitive sports, with the possible exception of some low-intensity sports. You should talk with your cardiologist about specific recommendations. The use of an implantable cardioverter-defibrillator should not be viewed as a substitute for these recommendations.

Sudden Cardiac Arrest Risk Assessment Form Below

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Sudden Cardiac Arrest Risk in the Young Assessment Form

To assess the risk of sudden cardiac arrest complete this form for each person under the age of 50, including children, periodically at suggested intervals including neonatal, preschool, before and during middle school, before and during high school before college and every few years through adulthood. If you answer "YES" or "UNSURE" to any questions please refer to the back of this form.

Name:_____ Age:_____ Date:_____

Individual History Questions:	**Yes**	**No**	**Unsure**
Has this person fainted or passed out DURING exercise, emotion or startle?			
Has this person fainted or passed out AFTER exercise?			
Has this person had extreme fatigue associated with exercise? (different from others of similar age)			
Has this person ever had unusual or extreme shortness of breath during exercise?			
Has this person ever had discomfort, pain or pressure in his chest during exercise, or complained of his heart "racing or skipping beats"?			
Has a doctor ever told this person they have: □ high blood pressure □ high cholesterol □ a heart murmur or □ a heart infection? (Check which one, if any "yes" answer.)			
Has a doctor ever ordered a test for this person's heart? If yes, what test and when?			
Has this person ever been diagnosed with an unexplained seizure disorder or exercise-induced asthma? If yes, which one and when?			
Has this person ever been diagnosed with any form of heart/cardiovascular disease? If yes, what was the diagnosis:			
Is this person adopted or was an egg or sperm donor used for conception?			
Family History Questions: (think of Grandparents, Parents, Aunts, Uncles, Cousins and Siblings)			
Are there any family members who had a sudden, unexpected, unexplained death before age 50? (including SIDS, car accident, drowning, in their sleep, or other)			
Are there any family members who died suddenly of "heart problems" before age 50?			
Are there any family members who have had unexplained fainting or seizures?			
Are there any family members who are disabled due to "heart problems" under the age of 50?			
Are there <u>any</u> relatives with certain conditions such as:			
Check the appropriate box: □ Hypertrophic cardiomyopathy (HCM) , □Dilated cardiomyopathy (DCM), □Arrhythmogenic right ventricular cardiomyopathy (ARVC), □ Long QT syndrome (LQTS), □Short QT syndrome, □Brugada syndrome, □Catecholaminergic ventricular tachycardia			
Coronary artery atherosclerotic disease (Heart attack, age 50 years or younger)			
Aortic rupture or Marfan syndrome or Ehlers-Danlos syndrome			
Primary pulmonary hypertension			
Congenital deafness (deaf at birth)			
□Pacemaker or □implanted cardiac defibrillator (if yes, whom and at what age was it implanted?)			
Other form of heart/cardiovascular disease or mitochondrial disease			
Has anyone in the family had genetic testing for a heart/cardiovascular disease? If yes, what was tested for?_____ Was a gene mutation found: YES/NO			
Please explain more about any "yes" answers here:			
Physical Exam from Physician should include: **(to be performed by a physician – made available here for the purpose of parent/patient education to ensure all evaluations have been completed)**			
Evaluation for heart murmur in both supine and standing position and during valsalva			
Femoral pulse			
Brachial artery blood pressure – taken in both arms			
Evaluation for Marfan syndrome stigmata			

This form includes all items suggested in the American Heart Associations Recommendations for Preparticipation Screening for Cardiovascular Abnormalities in Competitive Athletes– 2007 Update Circulation 2007:115

<u>For more information visit the HCMA at www.4hcm.org</u>

At this point you may have answered YES or UNSURE to one or more questions on the front of this form and you may be wondering what to do next. The first thing we can tell you is don't worry – just act!

It is as easy as 1-2-3!

Step One – Contact your health care provider, normally your General Physician, Family Practitioner or Pediatrician and discuss the form including areas of risk you have identified and discuss having a full cardiac exam by a cardiac professional. Some general physicians/family practice or pediatricians may be comfortable ordering cardiac testing and interpreting the results and some may not, therefore a referral may be needed to a cardiologist.

Step Two – Based upon your insurance provider either ask your doctor for a referral for a complete cardiac evaluation by a cardiologist or seek the appointment on your own. This appointment should include basic cardiac testing based on the individual's history but normally includes a consult with the cardiologist, an electrocardiogram (ECG), echocardiogram (echo) and in some cases stress testing and additional cardiac imaging such as CT Scanning or Cardiovascular magnetic resonance CMR.

Step Three – Communicate YOUR history to the rest of your family so they can seek appropriate screening.

Things you should know about additional testing for sudden cardiac arrest risks (SCA):

1. Nearly all tests are painless, non invasive and require no needles.
2. Tests are an evaluation of the heart at that moment in time and things may change over time, therefore you may need to repeat the testing on yourself or your child at intervals through out life.
3. The knowledge of cardiac diseases that causes sudden cardiac arrest are an evolving field and testing may change over time or the definition of normal or abnormal may also change.
4. If you and/or your loved one are found to be at risk for SCA there are things you can do to help prevent SCA including:
    a. Taking medication
    b. Having an implantable cardioverter defibrillator,ICD, implanted (a pacemaker like device that can provide a lifesaving shock should you experience SCA)
    c. Making lifestyle modifications to reduce risk (refrain from *competitive* sports for some)

Special note: If you have answered UNSURE to matters of health history the details should be discussed with complete candor with your health care provider. Cases of adoption, egg or sperm donation or uncertain paternity are areas of specific concern as the health information that may have been available at the time of adoption, donation or last contact with the father may have changed and you may be unaware – therefore the HCMA suggests to err on the side of caution and seek base line cardiac testing in these cases.

If you have any questions or need additional information please feel free to contact the HCMA at 973-983-7429 or visit us online at www.4hcm.org

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