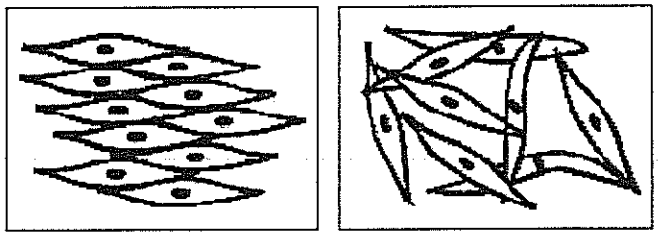
What is Hypertrophic Cardiomyopathy (HCM)?

LAST UPDATED: 2007/03/31 20:00

What is Hypertrophic Cardiomyopathy (HCM)?

Cardiomyopathy is a condition in which the muscle of the heart is abnormal in the absence of an apparent cause. This terminology is purely descriptive and is based on the Latin deviation. HCM is a primary and usually familial cardiac disorder with heterogeneous expression, unique pathophysiology, and a diverse clinical course, for which several disease causing mutations in the genes encoding proteins of the cardiac sacomere have been reported. While HCM has typically been recognized by its structure ie., hypertrophy, the electrical function of the heart are also adversely affected. There are three types of cardiomyopathy: **"hypertrophic"**, **"dilated"** and **"restrictive"**. The main feature of hypertrophic cardiomyopathy is an excessive thickening of the heart muscle (hypertrophy literally means to thicken). Thickening is seen in the ventricular septal measurement (normal range .08-1.2cm), and in weight. In HCM, septal measurements may be in the range of 1.3cm to 6.0+cm. Heart muscle may also thicken in normal individuals as a result of high blood pressure or prolonged athletic training. Furthermore, there is a fine line between and athletic heart and a heart with HCM.

In Hypertrophic Cardiomyopathy (HCM), the muscle thickening occurs without an obvious cause. In addition, microscopic examination of the heart muscle in HCM is abnormal. The normal alignment of muscle cells is absent and this abnormality is called **"myocardial disarray"**.



Normal Muscle Structure Myocardial Disarray

Myocardial Disarray

These diagrams contrast the regular, parallel alignment of muscle cells in a normal heart with the irregular, disorganized alignment of muscle cells or "myocardial disarray" found in some parts of the heart in hypertrophic cardiomyopathy.

History and Other Names

What's in a Name?

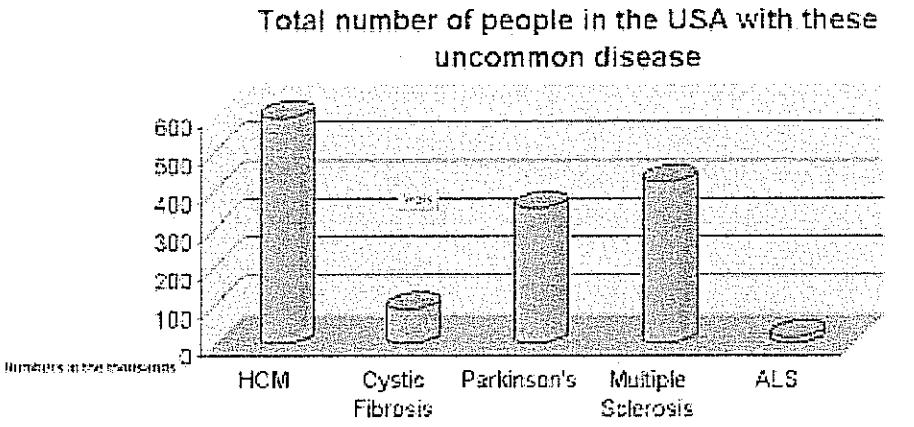
It is confusing. Remarkably, this disease (hypertrophic cardiomyopathy) has been given 75 separate names by individual investigators over the last 40 years. Literally, no other disease can make that claim. Why has this occurred? The principal reason for the proliferation of names has undoubtedly been the heterogeneity and diversity in which the disease is expressed. Few individual investigators have seen large numbers of patients with hypertrophic cardiomyopathy, and therefore individuals have come to regard the overall disease based on their own (sometimes limited) experience. Many of the names are somewhat misleading since they emphasize obstruction to left ventricular outflow which is a highly visible feature of the disease but is probably present in no more than about 25% of all patients. These names include IHSS (or idiopathic hypertrophic subaortic stenosis) which was the first popular term used in the United States; "stenosis" means obstruction. The same can be said for HOCM (hypertrophic obstructive cardiomyopathy) which is still used in the United Kingdom...largely out of habit and convenience.

Nevertheless, virtually all HCM experts and other cardiovascular specialists now regard as the best single name for this broad disease spectrum --- hypertrophic cardiomyopathy or HCM. This term emphasizes the hypertrophy which is the diagnostic marker in most patients and the fact that this disease is a form of cardiomyopathy -- or heart muscle disorder... without mentioning obstruction. Therefore, it is preferable to describe the disease as either "HCM with obstruction" or "HCM without obstruction."

"Cardiomyopathy" itself is a very general term referring to any condition (and there are many) importantly affecting the heart muscle itself while "hypertrophic cardiomyopathy" refers to a specific and genetic condition which usually shows a familial pattern. The most characteristic feature of HCM is a hypertrophied left ventricle (asymmetric thickening of the wall usually most prominently involving the ventricular septum) without abnormal enlargement of the ventricular cavities.

How Common is Hypertrophic Cardiomyopathy?

Hypertrophic Cardiomyopathy is a relatively uncommon heart disease. Its exact frequency is unknown. A paper by Dr. Barry Maron of the Minneapolis Heart Institute, August 1995, estimated that between 1 in 500 and 1 in 1000 births could be affected by HCM. Based on these data we may estimate that as many as 300,000 people in the United States have HCM. In comparison Cystic Fibrosis has a prevalence of 1 in 3300. This means HCM is nearly 7 times more common than Cystic Fibrosis. If you would like to help the HCMA spread the word about this condition, please contact our offices.



How Does Hypertrophic Cardiomyopathy Affect the Heart?

The Normal Heart

It is helpful to be familiar with the structure and function of the normal heart in order to understand the abnormalities in HCM. Fig. 3 shows a normal heart and indicates the heart chambers, valves and the direction of blood flow. The walls of the heart are composed of specialized muscle known as the myocardium. It is this part of the heart which is abnormal in HCM.

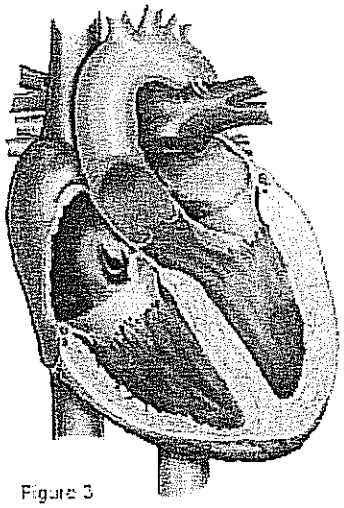


Figure 3

Structure and Function

This is a picture of the internal structure of a normal heart. The four chambers and four one-way valves are indicated. The right atrium receives blood from the body, transfers it to the lungs to the left atrium. It is transferred to the left ventricle which pumps it around the body for another cycle. Figure 4 again shows a normal heart but in this diagram the electrical activity of the heart is shown. Every heartbeat results from an electrical signal starting at the top and passing down through the heart. The abnormality of the heart muscle in Hypertrophic Cardiomyopathy can sometimes interfere with this normal electrical signal.

The Normal Electrical Impulse in the Heart

The normal electrical impulse starts in the right atrium as shown by the arrows. It travels by special conducting tissue down through the heart and into the muscle to start a contraction.

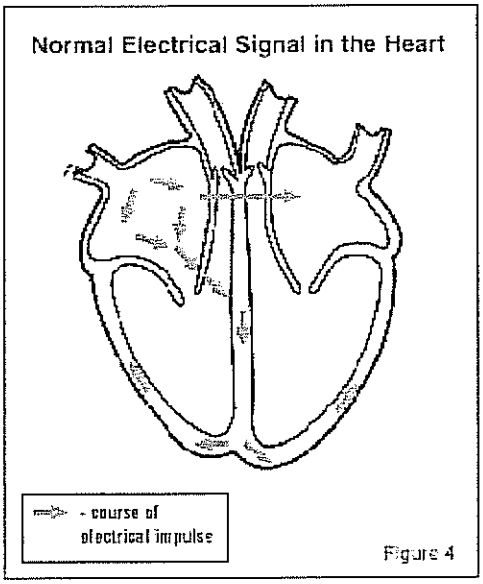


Figure 4

The Heart in Hypertrophic Cardiomyopathy

The major abnormality of the heart in Hypertrophic Cardiomyopathy is an excessive thickening of the muscle. The distribution of muscle thickening or hypertrophy is variable. The left ventricle is almost always affected and in some patients the muscle of the right ventricle also thickens.

Asymmetric Septal Hypertrophy

Figure 5 shows the most common form of HCM where the muscle thickening occurs predominantly in the "septum" or the dividing wall between the right and left sides of the heart. This form is called *"asymmetric septal hypertrophy".*

Figure 6 shows the most common form of HCM where the muscle thickening occurs mainly in the upper part of the septum. Note that the mitral valve maintains a normal position.

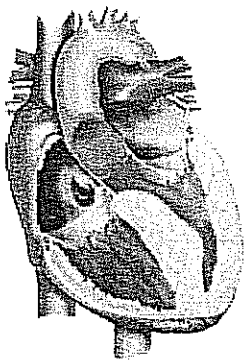


Figure 5

Asymmetric Septal Hypertrophy without obstruction

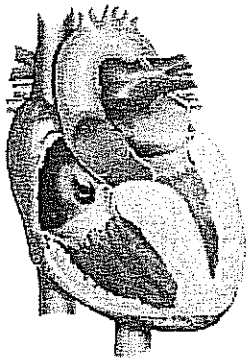


Figure 6

Asymmetric Septal Hypertrophy with obstruction

It can be seen from figure 5 that the hypertrophy is usually greatest in the upper septum, in the area where blood flows out of the heart into the aorta or "outflow tract". The muscle thickening in this region may be sufficient to narrow the outflow tract, Figure 6. In such cases during the ejection of the blood flow from the heart, the mitral valve touches the septum (there should normally be a considerable gap between these structures, Figure 5). This narrowing of the outflow tract interferes with the normal ejection of blood. It causes turbulent blood flow and sometimes obstruction to flow. The turbulent flow produces a murmur which is audible with a stethoscope. In such patients, the abnormal position of the mitral valve may cause it to leak. This is called "mitral regurgitation" and may also cause a murmur, Figure 6.

In some cases of asymmetric septal hypertrophy obstruction to the outflow of blood from the heart may occur as shown here. Note that the mitral valve now touches the septum blocking the outflow tract ("systolic anterior motion of the mitral valve" or "SAM"). Some blood is leaking back through the mitral valve ("mitral valve regurgitation").

Other Patterns of Muscle Thickening

In approximately 25% of patients the muscle thickening is evenly distributed throughout the ventricle. This is known as "symmetric" or "concentric" ventricular hypertrophy.

In a small proportion of patients (approximately 10%), myocardial thickening is predominantly at the tip or "apex" of the heart. This appears to be more common pattern of hypertrophy in Japan than in the West. Patients with Concentric and Apical Hypertrophic Cardiomyopathy usually do not have a murmur.

Function of the heart in Hypertrophic Cardiomyopathy

The thickened muscle usually contracts well and ejects most of the blood from the heart. However the muscle in HCM is often stiff and relaxes poorly. This requires higher pressures than normal to expand with the inflow of blood. The amount of blood which the heart can hold is therefore reduced and this in turn will limit the amount of blood which can be ejected with the next contraction.

Occasionally patients present with minimal to no hypertrophy, but sever restrictions to the normal inflow of blood into the ventricles. The differentiation from Restrictive Cardiomyopathy may be different and accurate diagnosis relies on the presence of other features of the two conditions.

Muscle Cells under the Microscope

Examination of the heart muscle in Hypertrophic Cardiomyopathy under a microscope shows that the normal parallel alignment of muscle cells has been lost. The cells appear disorganized. This abnormality is called "myocardial disarray". It is probable that myocardial disarray interferes with normal electrical transmission and predisposes to irregularities of the heart beat.

Normal parts of the Heart in Hypertrophic Cardiomyopathy

Finally, it is important to note that parts of the heart commonly affected in other conditions e.g. the heart valves and main coronary arteries (blood vessels that supply the heart) are normal in Hypertrophic Cardiomyopathy. In this case the muscle thickening is of equal severity throughout the whole left ventricle. In this form of HCM the muscle thickening occurs predominantly at the tip (apex) of the left ventricle. Only a small slit-like cavity remains.

What Symptoms Does Hypertrophic Cardiomyopathy Cause?

There is no particular symptom or complaint which is unique to Hypertrophic Cardiomyopathy. Symptoms may occur at any stage in a person's life even though the condition may have been present for some time. The reason for the onset of symptoms is often not clear.

Shortness of Breath

Exercise capacity may be limited by breathlessness and fatigue. Most individuals experience only mild exercise limitations, but occasionally limitation is severe and a minority may have shortness of breath at rest.

Chest Pain

Chest pain is a common symptom. It is usually brought on by exertion and relieved by rest, but pain may occur at rest or during sleep and may persist. The cause of the pain is thought to be insufficient oxygen supply to the myocardium. In Hypertrophic Cardiomyopathy the main coronary arteries are usually normal, but the greatly thickened muscle demands an increased oxygen supply which cannot be met in some circumstances.

Palpitation

Palpitation is an uncomfortable awareness of the heart beat. People may occasionally feel an extra beat or a skipped beat and this is usually normal. Sometimes an awareness of the heart beating does suggest an irregular heart rhythm. In this case, palpitation may start suddenly, appear to be very fast and may be associated with sweating or light-headedness. The cause of such episodes should be determined and treated.

Light-Headedness and Blackouts

Persons with the condition may experience light-headedness, dizziness and more seriously, blackouts. Episodes may occur in association with exercise, with palpitations or without any apparent provocation. The reasons for these episodes are not always clear. They may be due to an irregularity of the heart beat, or fall in blood pressure. Episodes of light-headedness and certainly a blackout should be reported to one's doctor and investigated.

Most commonly seen "misdiagnosis" associated with HCM?

Most commonly seen is asthma, specifically 'athletically induced asthma" as a first sign or symptom. This is likely due to transient shortness of breath often seen in HCM. It is also common to have a diagnosis of "mitral valve prolapse" prior to a proper diagnosis of HCM. This often happens upon an audible murmur that is thought to be simply mitral valve prolapse a common condition. It is not uncommon to see people diagnosed with anxiety attacks, panic attacks or some forms of depression only to find that the underlying cause of the symptoms is HCM.

When Does Hypertrophic Cardiomyopathy Develop?

Although hypertrophy may be present at birth or in childhood, it is much more common for the heart to appear normal at this time. Occasionally, Hypertrophic Cardiomyopathy is the cause of a stillbirth or develops during infancy, with heart failure, which may be fatal. However, hypertrophy more commonly develops in association with growth and is usually apparent by the late teens or early twenties. After this time it appears that there is no significant change in muscle thickness in the years of adult life. Children and adolescents with the condition usually come to attention when a family screening is performed after an adult in the family is found to be affected. Approximately 50% of adults with the condition present with symptoms. In the remainder the diagnosis is made during family screening or following the detection of a murmur or abnormality on routine electrocardiogram (ECG). A growing number of HCM patients are being identified later in life and are referred to as "adult onset". In the early literature the occurrence of adult onset appeared rare, now we know it to be far more common. Therefore in recent years those in the HCM community have encouraged those

with a family history of HCM to continue to be screened every 5 years, or if symptoms occur, after the age of 25 for the remainder of life. For screening information see Family Screenings on this web site.

How is Hypertrophic Cardiomyopathy Diagnosed?

Hypertrophic Cardiomyopathy may be suspected because of symptoms, a murmur or an abnormal ECG/EKG. An individual with the condition may present with any of the symptoms described previously. Because such symptoms could be caused by a large number of other conditions, further tests are necessary.

Electrocardiogram or ECG (EKG)

An ECG records the electrical signals from the heart and is performed by placing electrodes on the chest, wrist and ankles, see figure 9. In Hypertrophic Cardiomyopathy the ECG usually shows an abnormal electrical signal due to muscle thickening and disorganization of the muscle structure. In a minority of patients (approximately 10%) the ECG may be normal or show only minor changes. ECG abnormalities are also not specific to Hypertrophic Cardiomyopathy and may be found in other heart conditions.

Echocardiogram or ECHO

Currently, the diagnosis of Hypertrophic Cardiomyopathy is made by an ultrasound scan of the heart called an "echocardiogram" or ECHO. Like the ECG this is an entirely safe test, see figure 10. An ECHO produces a picture of the heart. Excessive thickness of the muscle can be easily measured. Additional equipment called "Doppler" ultrasound can produce a color image of blood flow within the heart and measure the heart's contraction and filling. Turbulent flow can be detected. Therefore ECHO provides a very thorough assessment of Hypertrophic Cardiomyopathy.

Other Investigations Which May Be Necessary

Cardiac Catheterization

Patients with breathlessness, which does not respond to therapy may require cardiac catheterization. In this test a fine tube is passed from a blood vessel (usually in the groin) to the heart using x-ray guidance. Pressures inside the heart are then measured and an x-ray of the heart is taken (angiography) to assess mitral regurgitation and overall function.

Coronary Angiography

Patients who experience chest pain which does not respond to therapy, may require coronary angiography. This is an x-ray of the coronary arteries to determine if they are diseased and it is performed during cardiac catheterization.

Electrophysiological Studies

These are a special form of catheterization performed to define the risk of electrical instability which may predispose to sudden death. This test involves the passage of fine wires from the veins in the groin, arm or shoulder to the heart under x-ray guidance. These wires are then used to apply electrical stimuli to record the response of the electrical system of the heart.

Exercise Test

The severity of the exercise limitation and the effect of therapy can be assessed with bicycle or treadmill exercise testing. Exercise testing also provides and objective measurement of improvement, stability or deterioration over time.

Holter Monitor

This test is a continuous recording of the heart beat over 24 to 48 hours. A Holter monitor is a simple and safe test which will detect irregularity of the heart beat (otherwise known as arrhythmia).

Radionuclide Studies

In these tests, substance producing very tiny (safe) amount of radioactivity are given by injection. These tests may be used to assess the contraction and filling of the heart and also to estimate its blood supply at rest and on exercise.

Complications

In a minority of cases a number of specific complications can occur:

Arrhythmias

Arrhythmias, irregularities of the heart beat, are a common complication. Symptoms such as palpitation may occur, but not often. Exercise testing or Holter monitoring may detect them. The arrhthymias called ventricular tachycardia (arising from the ventricles) or atrial fibrillation (described below) are particularly important and may require treatment. Atrial Fibrillation, the normal regular rhythm of the heart beat is lost and replaced by an irregular rhythm which may be episodic or persistent. The loss of normal atrial (the top of the heart) contraction produces a risk of clot formation in the atria. Anticoagulants and drugs to slow the heart rate are required.

Endocarditis

This is an infection of the heart which occurs rarely in Hypertrophic Cardiomyopathy. Bacteria in the bloodstream can stick to the inside of the heart where it has been roughened by turbulent blood flow. **As uncommon as this is, the President of the HCMA fell victim to this. She had been MISINFORMED by a dentist who said "Genetic disorders do not need to be premedicated". The HCMA advises extreme caution when undergoing any "invasive" medical procedure**

Heart Block

The normal electrical signal may travel down to the ventricles slowly or may even be completely blocked, "heart block". If this occurs, a pacemaker is implanted (see "other forms of therapy").

Sudden Death

Overall, in patients with Hypertrophic Cardiomyopathy there is an increased risk of premature death, which can occur with little or no warning. Sudden death can strike at any age. In the past the risk of sudden death was thought to be much higher than we believe it to be today. It is estimated that the risk of sudden death is between 1 and 2 % in the HCM population. There are members of the HCM population at a higher risk for sudden death and for those at higher risk it is advised they consult with their health care provider about receiving an implantable defibrillator.

"Burnt out" or "End Stage" HCM

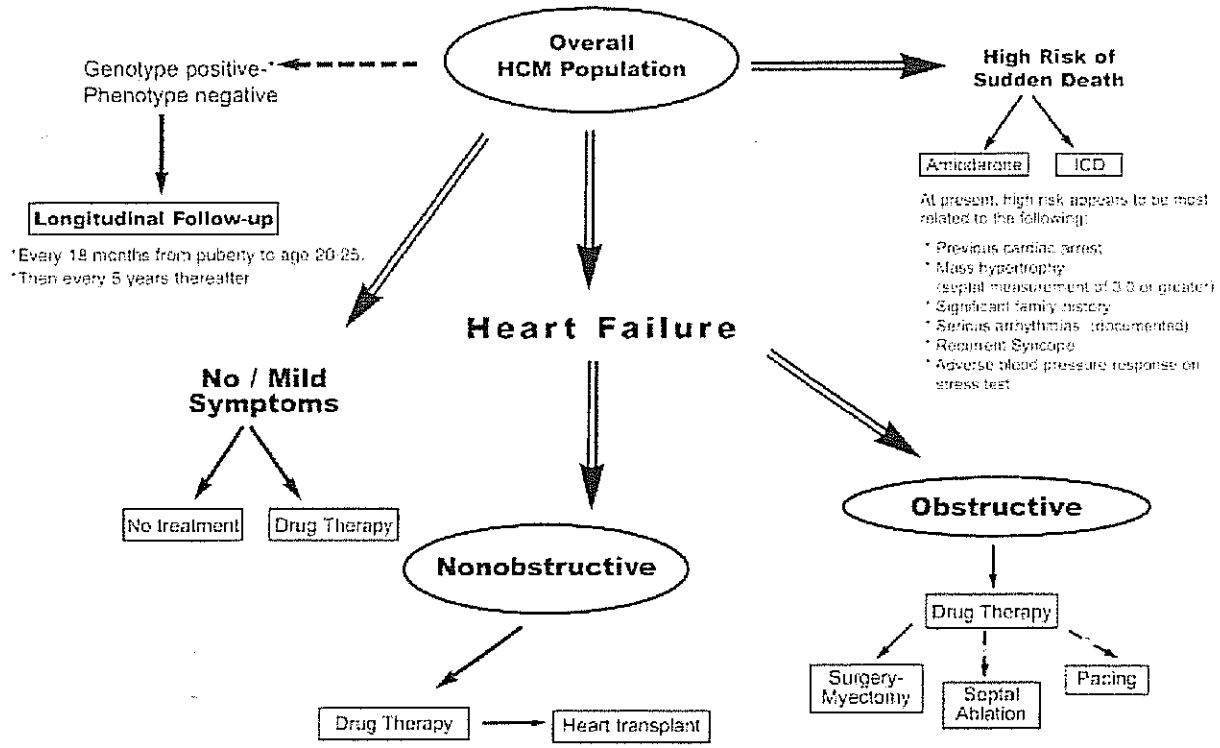
The terms associated with this part of the HCM disease process are misleading and rather depressing. It is our goal to help readers gain a better understand this rare but important potential consequence of HCM, occurring in under 3% of the total HCM population. It is poorly understood at this time why this occurs. Some theories suggest it is the individual genetic mutation that determines the progression of the disease.

While "End stage"/"Burnt out" is not precisely the same it is remarkably similar to the dilated form of cardiomyopathy. In *dilated cardiomyopathy* (often referred to as congestive cardiomyopathy), the heart reaches a point of generalized weakening and thinning of the muscle walls with dilated chambers, particularly the left ventricle. While the walls may have previously been normal or thickened from overwork through trying to compensate for an inadequate ability to pump, (or in the case of HCM), previously "thick" muscle now changes to a different form of damaged tissue. The dilation and thinning of the cardiac chambers, especially of the left ventricle, is often referred to as "remodeling". The weakening and dilation of the heart muscle eventually leads to heart failure. previously normal or in the case of HCM, previously "thick" heart muscle changes to a different form of damaged tissue, leading to a generalized weakening of the walls of the cardiac chambers. To compensate for the weakening of their muscular walls, the cardiac chambers dilate. (The dilation of the cardiac chambers, especially of the left ventricle, is often referred to as "remodeling.") The weakening and the dilation of the heart muscle eventually leads to heart failure.

Is a cure available?

At present there is no cure for Hypertrophic Cardiomyopathy, HCM. There is a slight possibility that some drugs may decrease the degree of muscle thickening. However, no treatment has been shown to return the heart to normal. Research continues in this area. Developments are most likely to come from the early detection of persons carrying the gene for Hypertrophic Cardiomyopathy and treating them to prevent the development of hypertrophy. The achievement of this goal will require much further research and is many years away from reality.

HCM Treatment Overview



Drug Treatment

Drug treatment or medication is primarily given when a person has some or all of the symptoms described earlier. The choice of treatment will vary from individual to individual but the common groups of drugs used are as follows:

Beta-Blockers
Calcium Antagonists
Anti-Arrhythmic Drugs
Diuretics
Antibiotics

Summary of Drug Treatment

In summary, a variety of drug treatments are currently used in hypertrophic cardiomyopathy (HCM) and of course new drugs may be discovered in the future. The need for any treatment and choice of that treatment has to be made on an individual basis and may change in any one individual over the years. It is very important to discuss your symptoms with your doctor and plan a treatment plan for you, as each patient with hypertrophic cardiomyopathy (HCM) is different.

Surgical Options

Surgical "myectomy" (removing a small amount of muscle from the ventricular wall) is successful in the relief of symptoms. It is considered in individuals with severe symptoms despite drug treatment, in whom the left ventricular outflow tract narrowing causes obstruction of the blood flow (See Figure 6). Typically, if a person has an obstuctive gradient greater than 50mm of mercury and is symptomatic and has failed drug therapy it is only then advised they consider this procedure. In this operation the surgeon removes a portion of the thickened muscle from the septum, thereby widening the outflow tract and relieving the obstruction. In rare cases, instead of a myectomy, the mitral valve is replaced with an artificial valve. As described earlier, during obstruction to outflow from the heart, the mitral valve touches the septum and blood leaks back through the valve. If this mitral valve regurgitation is severe, then the valve may be replaced (this is a rare occurrence). These are major operations which carry a definite risk and therefore are reserved for patients with severe symptoms and certain forms of HCM. It is imperative that the patient choose a center that is experienced in the surgical procedure and a surgeon with ample experience. In the most experienced centers this operation has a success rate of 99% for survival and more than 70% of patients have significant improvement in quality of life that persists many years later. In very rare cases the surgery must be done a second time.

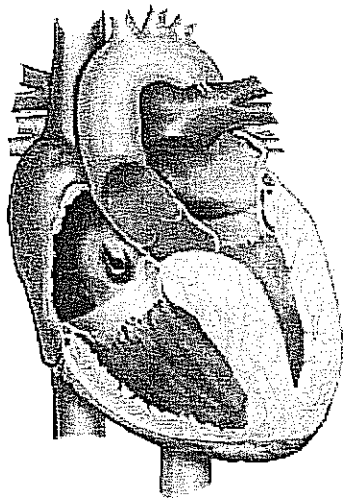


Figure 6

Heart Transplant

Rarely, heart transplantation is necessary for those individuals who have a severe impairment of pumping action of the heart without outflow obstruction. Those with end stage HCM may consider transplant.

Surgery in the Pediatric Population

According to an article published December 1996 in The Journal of Thoracic and Cardiovascular Surgery, "Hypertrophic cardiomyopathy (HCM) in Pediatric Patients: Results of surgical treatment" by Dr.'s D. Theodoro, G. Danielson, R. Feldt, and B. Anderson RN of the Mayo Clinic, Rochester, MN. This paper reports "96% of the young patients (ages between 2 months and 20 years, mean age of 11.2 years) have had significant improvement in preoperative symptoms. There was no early or late mortality."

Alternative treatments for Atrial Fibrillation

Are there alternative treatments for Atrial Fibrillation for those with HCM?

Yes there are. Pulmonary vein ablation – or PVAI and MAZE procedures have been used with success in patients with persistent atrial fibriallation. The information below has been taken in part from the Cleveland Clinics website.

What is pulmonary vein antrum isolation?

Research has shown that almost all atrial fibrillation signals come from the four pulmonary veins. Pulmonary vein antrum isolation (PVAI), also called pulmonary vein ablation, is a treatment for atrial fibrillation. During PVAI, a doctor inserts catheters into the blood vessels of the atrium.

A special machine delivers energy through the catheters to the area of the atria that connects to the pulmonary vein (ostia). This energy (ablation) produces a circular scar that blocks any impulses firing from within the pulmonary vein, thereby "disconnecting" the pathway of the abnormal rhythm and preventing atrial fibrillation. In some cases, ablation also may be performed in other parts of the heart such as the superior vena cava.

How successful is PVAI in treating atrial fibrillation?

Success rates for PVAI are defined as restoring a patient's normal sinus rhythm while not being dependent on medications to control the heart rhythm. In the general population pulmonary vein isolation has an 80 to 85 percent success rate with the first ablation. For those who have returned for further ablation, the success rate has been 95 percent. The success rate in the HCM population is less clear.

What is a MAZE procedure and how is it done?

An incision is made along the sternum (breast bone). This may be a traditional incision, or in some cases, a minimally invasive incision may be used. The heart-lung machine oxygenates the blood and circulates it throughout the body during surgery.

Certain patients with isolated atrial fibrillation, especially continuous atrial fibrillation and/or enlarged atria, are candidates for the Maze procedure. This procedure, developed by Dr. Jim Cox, can treat the atrial fibrillation and restore the atria to a more normal size.

During the Maze procedure, a series of precise incisions are made in the right and left atria to interrupt the conduction of abnormal impulses. This allows sinus impulses to travel to the atrioventricular node (AV node) as they normally should.

The Maze procedure has been very successful with a 98% success rate in "lone atrial fibrillation" patients and a 90% success rate overall. Post -procedure freedom from stroke has been over 99%.

For patients who require other forms of heart surgery, surgeons may perform either a classic Maze procedure or a modified Maze procedure. The classic Maze procedure cures atrial fibrillation in more than 90 percent of patients, but requires about one hour to complete. In most patients who are having additional heart surgery, the surgeon chooses to perform a modified Maze procedure.

Outcome:

Improvements in surgical techniques over the years have produced successful results in most patients:

- Long-term freedom from atrial fibrillation
- Decreased symptoms
- Greatly reduced embolic events (such as blood clots or stroke)
- Decreased atrial (top chamber of the heart) size in those with enlarged atria pre-surgery, particularly those who undergo Maze procedure with mitral valve repair procedure.

Success with the MAZE procedure in HCM patients has been seen however the numbers are very small in any one center. Both the Cleveland Clinic and the Mayo Clinic have had successful MAZE in HCM.

Alternatives to Surgery

Alcohol Septal Ablation

In the mid 1990s a new procedure emerged for the treatment of outflow obstruction in HCM patients. A catheterization is performed; alcohol is injected into the septum through a small coronary artery. This causes a 'controlled' myocardial infarction, (a small controlled heart attack). This procedure is still very new and long term outcomes are yet to be clearly understood. Some patients have been rendered pacemaker-dependent after this procedure (based on how the procedure is done this number varies greatly from 8 to 25%). There have been deaths associated with this procedure in the United States, which makes the risk comparable to surgery.

Alcohol septal ablation has several names including PTSMA -Percutaneous Alcohol Septal Ablation and TASH- Transcoronary ablation of septal hypertrophy. What is interesting to note is that since the creation of this procedure there has been a large jump in the number of patients requiring intervention for obstruction. This has lead many to wonder why? Since the creation of the procedure known as myectomy nearly 40 years ago approximately 4000 procedures have been preformed around the world. In stark comparison in the past 8 -10 years there have been well over 1000 ablations done world wide with no significant change in the number of myectomies performed. One area of concern to the HCMA is that many ablations are being performed at hospitals with limited experience with HCM and patient selection has been less then optimal. Many of these patients require multiple procedures or need to have myectomy performed after the ablation.

Patients who wish to consider this treatment option would benefit from a consult from a center with a complete HCM program and learning about ALL options and their various pro's and con's. Patient selection for a successful alcohol septal ablation is the most important step to ensure a positive

outcome. Patients must have a septal measurement of greater than 1.8 but less than 3.0, the mitral valve must not be a contributing factor to obstruction and the patient must have the proper anatomy to allow access to the area creating obstruction (a good 1st septal perforator located in the proper position). Patients need to be aware they are having a procedure that induces a myocardial infarction, a heart attack, and this will create a scar in the heart that may create a pro-arrhythmic situation in your heart, which due to your diagnosis of HCM your heart already has a propensity for arrhythmia. Also it is not recommended for younger patients to utilize this treatment modality. Patients over the age 55 or those with serious co-existing diseases should discuss with their doctors the potential benefits of alcohol ablation should all other factors point to this as a treatment option.

It should be clearly noted that according to the ACC/ESC consensus document on the treatment and management of HCM myectomy remains the gold standard treatment for obstruction.

If you doctor has recommended to you that you should have an alcohol septal ablation here are some questions to ask him/her:

- How many patients with HCM do you see annually, how many are obstructed and how many non-obstructed?
- How many times have you (or your center) preformed alcohol septal ablation?
- Where did you receive your training for alcohol septal ablation?
- How many myectomies do you perform annually?
- How many patients have you referred to other doctors for second options or treatment of their HCM?

If your doctor has not seen many patients, has only done a few procedures, has not received training from a center that also performs myectomies, they do not offer myectomy or have only done a handful and/or your doctor does not refer to specialty centers it would be advisable to seek a second opinion. At any point should you have questions please feel free to contact the HCMA office for more information.

As this is a newer procedure, the HCMA suggests caution and careful planning when reviewing this option. We have had a number of members of the HCMA undergo this procedure with varied outcomes. It is important to note that the variable nature of long and short term outcome is far different than the results we have seen from myectomy which provides a much more consistent positive outcome.

Is a Cure Available?

At present there is no cure for Hypertrophic Cardiomyopathy. While we remain very hopeful that new treatments will continue to improve our understanding and the management of HCM it is not likely that there will be an outright cure within the next 20 years. We have made significant advances in risk stratification for sudden death, learned that implantable defibrillators can save lives and made marked advancements in our understanding of the genetics behind HCM.
In the coming months and years we will learn more about the use of MRI in HCM, the role of genetic testing, how to screen more effectively. There are many exciting research efforts underway at many facilities around the world and with continued efforts on behalf of our wonderful HCM researcher we will have more information as to how we can improve our quality of life, minimize risks and live longer lives with HCM.

Screening for HCM

The Hypertrophic Cardiomyopathy Association is a not for profit organization providing information and support to those affected by HCM, their families, and the medical community. The following is the HCMA's views on screenings for HCM and basic management of the condition. In the event of the diagnosis of HCM in a family or an HCM related death of a family member, we strongly recommend that all blood relatives be screened. This screening MUST include an EKG/ECG, Echocardiogram, and a cardiac check up by a cardiologist. Ideally the cardiologist should have specialized knowledge of hypertrophic cardiomyopathy. Due to the fact HCM is a rather 'rare' condition it may require the patient seek out a specialist in the field of HCM. In many cases this will require travel out of state and in some cases out of the country.

Hypertrophic cardiomyopathy may be suspected because of family history, symptoms, a murmur or an abnormal EKG/ECG. Many symptoms or signs of hypertrophic cardiomyopathy are similar to various other conditions; therefore, it is important to follow doctor's instructions on complete testing to assure accurate results. An EKG/ECG records the electrical signals of the heart. In hypertrophic cardiomyopathy the EKG/ECG usually shows an abnormal electrical signal due to muscle thickening and disorganization of the muscle structure. In a minority of patients (approximately 10%) the ECG/ECG may be normal or show only minor abnormalities. EKG/ECG abnormalities are also not specific to hypertrophic cardiomyopathy and will be found in many other conditions.

Presently, the diagnosis of hypertrophic cardiomyopathy is made by an ultrasound scan of the heart called "echocardiogram" or "Echo" for short. Like the EKG/ECG, this is an entirely safe test. Excessive thickness of the muscle can be easily measured. Additional equipment called "Doppler" ultrasound can produce a color image of the blood flow within the heart and measure the contractions and filling of the heart. Turbulent flow can be detected using the doppler. Therefore, Echo/Doppler provides a very through assessment of hypertrophic cardiomyopathy.

After a diagnosis is made several other tests may be required. Holtor monitoring is a continuous recording of the heartbeat over a period of 24 to 48 hours. A holtor monitor is a simple and safe test that will detect irregularities of the heart beat (otherwise known as arrhythmia). Stress tests or Stress Echo's may be used to monitor the heartbeat during exercise as well as blood pressure response and then the echo to see if there have been any structural changes due to the exercise. MRI has been used in more frequent years to gain a more detailed image of the heart. Cardiac catherization may follow but it is not used on all patients with HCM.

↑TOP

The following article was written by Dr. Barry Maron of the Minneapolis Heart Institute Foundation and was in a past HCMA newsletter:

Since HCM is usually a genetic disease, we are often asked, in the context of the family evaluation, how screening for the disease should be carried out. Laboratory genotyping (using 10 cc of blood) would, of course, be the most definitive approach to identifying any individual with the disease (i.e. gene defect)--- theoretically. However, such laboratory techniques are expensive, laborious, not routinely available, and do not guarantee a positive answer. Genetic testing is presently confined to a very small number of research laboratories that work on highly selected pedigrees (families) only for investigational purposes. Therefore,, such family screening is still best carried out as it has been for many years --- with two dimensional echocardiography often performed serially (i.e., more than once) in growing children from families with HCM. The purpose of echocardiography (which is non-invasive, painless and has a "retail cost" in the range of $600.00) is to identify what clinicians refer to as the phenotype or overt expression of HCM --- i.e., the thickening of the left ventricular wall, usually of the ventricular septum (the portion of the wall separating the left and right ventricular chambers).

In general terms, the heart wall thickening is nor usually evident before the age of 10 and is most likely to be detected after the age of 12. Wall thickness usually increases as the child progresses through puberty with accelerated growth. The change in thickness can be abrupt and striking and therefore the appearance of the heart can change completely between 12 and 14 years of age --- often progressing from completely normal to very thick. Experts believe these changes of hypertrophy, while often alarming to the family and even some physicians, represent the normal pattern with which the heart forms in HCM and does not present either deterioration, an alarming clinical sign, or a warning of imminent danger. If the wall thickening becomes evident on the echocardiogram --- and is unexplained in other ways, such as by athletic training, other diseases, etc. --- then it may be assumed to represent a gene mutation responsible for HCM. Based on the available

growth and maturation is achieved (about age 17-20 years) then it would be less common for it to happen later. Therefore, if an individual is an HCM family is "echo-negative" by the time of adulthood, and then there is a chance of not being affected by the gene. However it is possible to develop HCM at any point in life, therefore the recommendation to continue screening every 5 years throughout life.

But when should echocardiograms be performed in children in families with HCM? Screening echocardiograms before the age of 10 or 12 are optional since these studies are rarely positive at this time, even in the presence of an HCM gene mutation, and recognition of the disease at this age would not necessitate intervention. Exceptions to this would be in selected families with multiple occurrences of premature death due to HCM or if the child is truly a competitive trained athlete. In such instances echocardiographic studies would appear obligatory at young ages, since HCM is the most important cause of sudden death during sports in young people, and a reason for disqualification from training and competition.

Otherwise, we recommend serial echocardiograms about every 18 months or so (every 12 months if a trained athlete) throughout adolescence or until the echocardiogram "converts" from normal to abnormal. Strangely enough, the standard electrocardiogram (EKG) may be abnormal in a genetically affected child long before the echocardiogram changes from normal to abnormal. Recent data does show the possibility of HCM appearing later in life if far more common then previously thought. With this new knowledge it is now suggested that children / young adults continue screening until age 25 on the every 12-18 monthly basis then switch to every 5 years through adulthood.

↑TOP

What to tell your family members about screenings for HCM

This can often be a difficult and stressful topic for discussion.A first great step is to print out this portion of the website and give it to family members.What they need to know is rather simple; all first-degree relatives of the affected person should get screened as soon as possible.Example: if your husband is diagnosed with HCM his parents, siblings and children should all be screened. Screenings must be done annually from age 12-20 and every 5 years age 25 and over. Screenings must include an ECG (EKG), echocardiogram and a visit to a cardiologist.