

Hypertrophic Cardiomyopathy

IMPORTANT

For the majority of affected individuals, Hypertrophic Cardiomyopathy is a condition which will not limit the quality or duration of life. A minority, however experience significant symptoms and are at risk of sudden death. Evaluation by a cardiologist is recommended to confirm the diagnosis and to assess the outlook and particularly the risk of complications.

INTRODUCTION

This site is intended for anyone interested in learning more about the heart condition Hypertrophic Cardiomyopathy. It has been produced in consultation with doctors, other medical personnel and those with the condition. The content seeks to address the major questions and concerns of patients and their relatives about the condition.

Medical terms which describe the condition and which might be encountered in conversations with doctors are included and explained in the text. In addition, words in ***bold italics*** are explained in a glossary at the end.

The Cardiomyopathy Association encourages correspondence and comments to improve this site.

WHAT IS HYPERTROPHIC CARDIOMYOPATHY?

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 derivation. There are four types of cardiomyopathy: **Hypertrophic (HCM)**, **Dilated (DCM)**, **Restrictive (RCM)** and **Arrhythmogenic Right Ventricular (ARVC)**. The main feature of Hypertrophic Cardiomyopathy is an excessive thickening of the heart muscle (hypertrophy literally means to thicken). Heart muscle may thicken in normal individuals as a result of high blood pressure or prolonged athletic training. In Hypertrophic Cardiomyopathy, however, the muscle thickening occurs without an obvious cause. In addition, microscopic examination of the heart muscle in Hypertrophic Cardiomyopathy shows that it is abnormal. The normal alignment of muscle cells is absent and this abnormality is called ***myocardial disarray*** (See **Figure 1**).

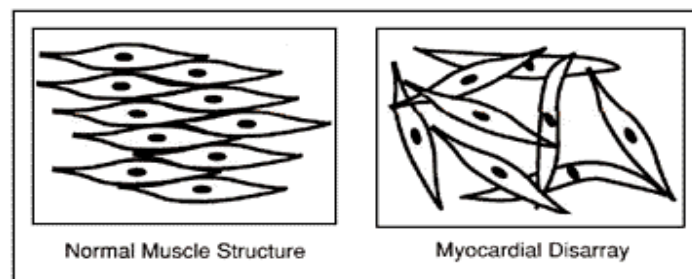


FIGURE 1

Myocardial Disarray

These diagrams contrast the regular, parallel alignment of muscle cells in a normal heart with the irregular, disorganised alignment of muscle cells or myocardial disarray found in some parts of the heart in Hypertrophic Cardiomyopathy.

HISTORY AND OTHER NAMES

Hypertrophic Cardiomyopathy was first recognised in the late 1950s. The condition has been known by a number of names including Hypertrophic Obstructive Cardiomyopathy (HOCM), Idiopathic Hypertrophic Sub-aortic Stenosis (**IHSS**) and Muscular Sub-aortic Stenosis. The general term **Hypertrophic Cardiomyopathy (HCM)** is now most widely used.

HOW COMMON IS HYPERTROPHIC CARDIOMYOPATHY?

Recent studies in the USA have suggested that Hypertrophic Cardiomyopathy is more common than previously reported. It is now estimated that approximately 1 in every 500 of the UK population suffers from the disease. An important objective of the Cardiomyopathy Association is to establish exactly how many people are affected in the UK.

WHAT IS THE CAUSE OF HYPERTROPHIC CARDIOMYOPATHY?

The cause of Hypertrophic Cardiomyopathy is not yet known. In the majority of cases the condition is inherited. In others there is either no evidence of inheritance or there is insufficient information about the individual's family to assess inheritance. In affected families the condition usually passes from one generation to the next and generations are not skipped. This pattern of inheritance is called **dominant** see **Figure 2**, and the condition may be passed on from affected males and females.

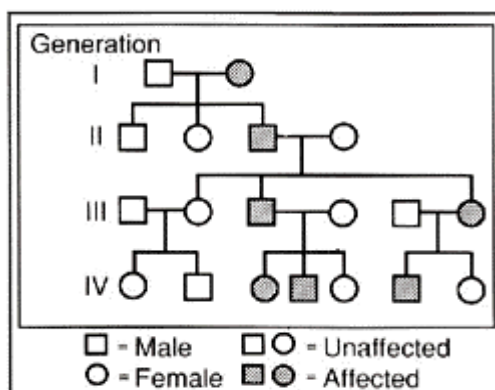


FIGURE 2

A Family Tree

This family tree shows four generations affected by Hypertrophic Cardiomyopathy. The condition is transmitted from one generation to the next without skipping a generation. This is called dominant inheritance. Each child of an affected person has a 50:50 chance of inheriting the condition.

NEW DISCOVERIES

Recently research has identified abnormalities in at least 6 related genes that are important in the

development of heart muscle cells. The abnormality is known as a **mutation** and may be likened to a spelling mistake in the genetic code make up of DNA. In approximately 50-60% of families, affected individuals are found to have a mutation in the gene for **myosin**, troponin T, alpha tropomyosin, cardiac myosin binding protein-C, or the essential and regulatory light chains. These are important proteins for the contraction of the heart.

Our bodies are made of millions of cells

Each cell has a nucleus

Each nucleus has 46 chromosomes

Each chromosome is made up of a long spiral of DNA

The DNA spiral is divided into genes

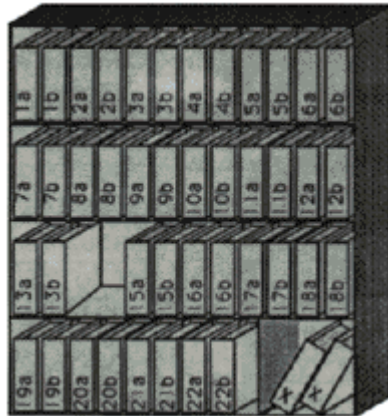
FIGURE 3



From Chromosome to Protein

Look at this book case!

We are searching for a specific word with a spelling mistake!



The cell nucleus contains 22 pairs of chromosomes plus one pair of sex chromosomes. A chromosome can be likened to a volume of an encyclopedia. Each person has two copies of the same volume, one copy from each parent.



FIGURE 4

----- DNA -----

DNA is a very long molecule composed of four different types of units.

The units are called **nucleotides**, and they are Adenine, Thymine, Guanine and Cytosine.

The nucleotides pair specifically to each other, in the manner A to T and C to G. This bonding keeps the two DNA strands together.

The order and length of the nucleotides determines which protein will be produced from the DNA sequence.

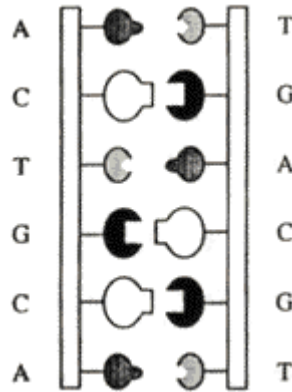


FIGURE 5

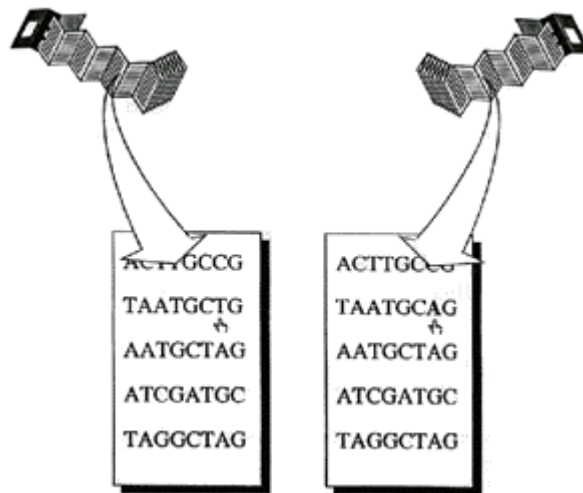


FIGURE 6

A gene is a sequence of "letters"; the length and order of the characters define the nature of the protein it produces

A mutation occurs where the sequence varies from the norm. That difference could be as minute as a single "letter" mismatch.

The discovery of these gene abnormalities is a major step towards understanding the cause of Hypertrophic Cardiomyopathy. Ultimately this should allow diagnosis from a blood or saliva test which will be particularly useful in children and adolescents.

Ongoing research aims to identify the other gene(s) which cause Hypertrophic Cardiomyopathy and to understand how these gene abnormalities result in the heart abnormalities which we recognise.

Such work is taking place worldwide, and in the UK, predominately at St George's Hospital Medical School in London, alongside research aimed at discovering other genes which may modify the disease and hence explain how people within the same family can have very different forms of Hypertrophic Cardiomyopathy.

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 Hypertrophic Cardiomyopathy. **Figure 7** shows a normal heart and indicates the heart chambers, valves and the direction of blood flow. The walls of the heart are composed of specialised muscle known as the **myocardium**. It is this part of the heart which is abnormal in Hypertrophic Cardiomyopathy.

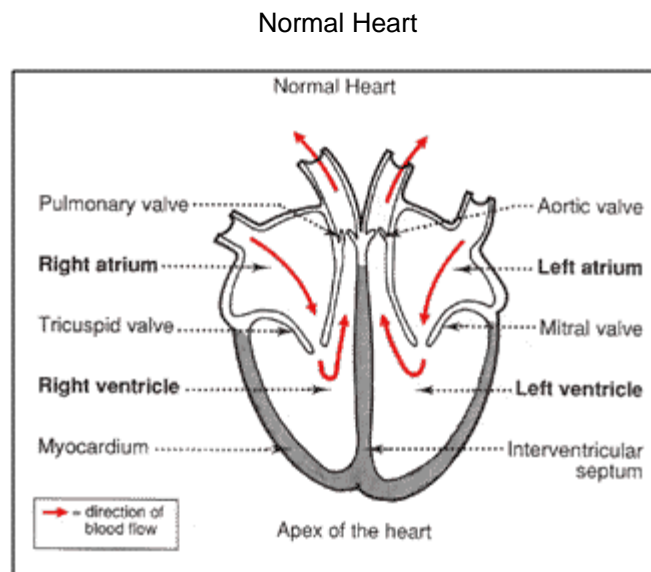


FIGURE 7

The Normal Heart Structure and Function

This is a diagrammatic representation of the internal structure of a normal heart. The four chambers and four one-way valves are indicated. The arrows show the direction of blood flow through the heart: the right atrium receives blood from the body, transfers it to the right ventricle which pumps it into the lungs to receive oxygen. Blood returns from the lungs to the left atrium. It is transferred to the left ventricle which pumps it around the body for another cycle.

Figure 8 shows a normal heart and 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 contraction of the heart follows the same course. The abnormality of the heart muscle in Hypertrophic Cardiomyopathy can sometimes interfere with this normal electrical activity. In abnormal segments of the heart the electrical signal may become unstable as it crosses the areas of fibrosis (scarring) and disarrayed cells. This in turn can lead to disorganised electrical impulses that generate fast or erratic heart rhythms.

Normal Electrical Signal in the Heart

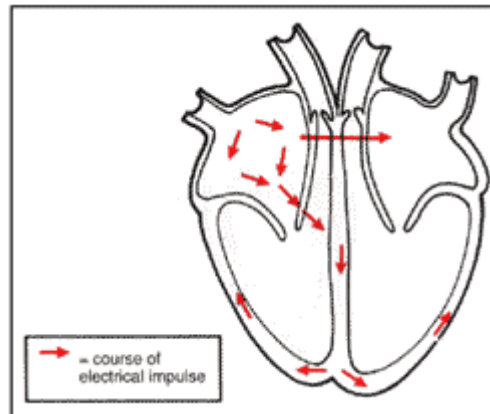


FIGURE 8

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.

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 9 shows the commonest form of Hypertrophic Cardiomyopathy where the muscle thickening occurs predominantly in the **septum** or dividing wall between the right and left sides of the heart.

Hypertrophic Cardiomyopathy ***Asymmetric septal hypertrophy without obstruction***

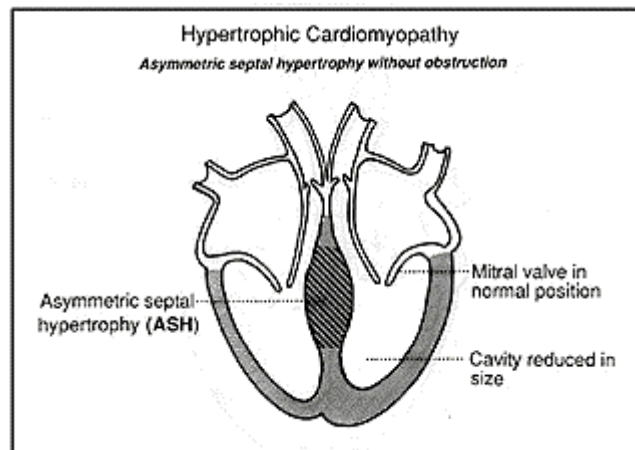


FIGURE 9

Hypertrophic Cardiomyopathy: Asymmetric Septal Hypertrophy without Obstruction

This diagram shows the commonest form of Hypertrophic Cardiomyopathy where the muscle thickening occurs mainly in the upper part of the septum. Note that the mitral valve maintains a normal position.

It can be seen from **Figure 9** 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 10**. 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 9**). 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 10**).

Hypertrophic Cardiomyopathy

Asymmetric septal hypertrophy with obstruction

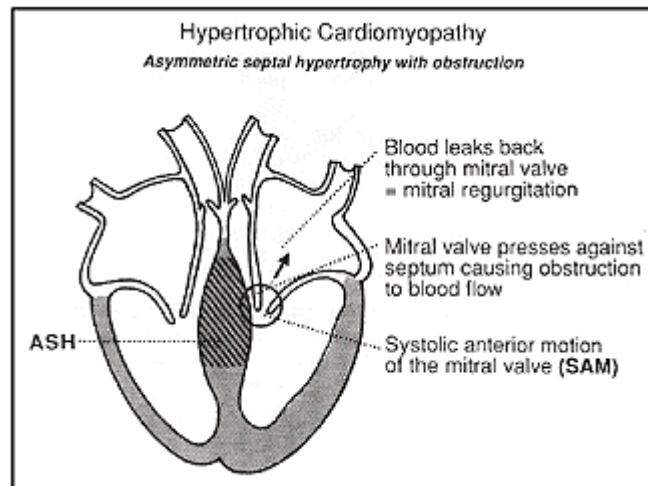


FIGURE 10

Hypertrophic Cardiomyopathy: Asymmetric Septal Hypertrophy with Obstruction

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 regurgitation).

Other patterns of muscle thickening

In approximately 20-25% of patients the muscle thickening is evenly distributed throughout the ventricle. This is known as symmetric or **concentric** left ventricular hypertrophy (**Figure 11**). In a small proportion of patients (approximately 10%), myocardial thickening is predominantly at the tip or **apex** of the heart, (**Figure 12**). This appears to be a 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 Hypertrophic Cardiomyopathy is often stiff and relaxes poorly, requiring 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 or no hypertrophy, but severe restriction to the normal inflow of blood into the ventricles. The differentiation from **Restrictive Cardiomyopathy** may be difficult 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 disorganised. This abnormality is called myocardial disarray (**Figure 1**). 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 the main coronary arteries (blood vessels that supply the heart) are normal in Hypertrophic Cardiomyopathy.

Hypertrophic Cardiomyopathy
Symmetric hypertrophy

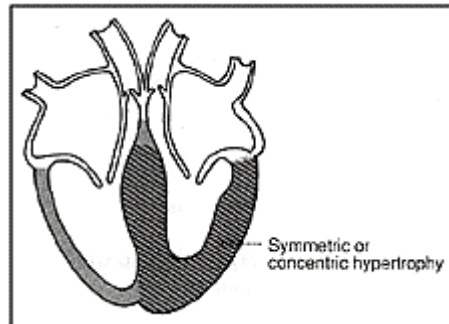


FIGURE 11

Hypertrophic Cardiomyopathy: Symmetric Hypertrophy

In this case the muscle thickening is of equal severity throughout the whole left ventricle.

Hypertrophic Cardiomyopathy
A typical hypertrophy

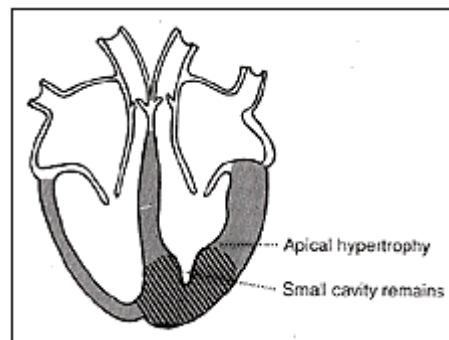


FIGURE 12

Hypertrophic Cardiomyopathy: Apical Hypertrophy

In this form of Hypertrophic Cardiomyopathy the muscle thickening occurs predominantly at the tip (apex of the left ventricle) Only a small slit-like cavity remains.

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. The condition can also develop during infancy, and if this is present with **congestive heart failure** it 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 are usually identified when family screening is performed after an adult in the family is found to be affected. Of these adults approximately 50% will have experienced symptoms. In the remainder the diagnosis is made during family screening or following the detection of a murmur or an abnormality on routine **electrocardiogram** (ECG) and **echocardiogram** (ECHO).

WHAT SYMPTOMS DOES HYPERTROPHIC CARDIOMYOPATHY CAUSE?

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

Shortness of breath

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

Chest pain

Chest pain (sometimes called **angina**) 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

People may occasionally feel an extra beat or a skipped beat but this is usually normal. Sometimes, however, 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 palpitation, 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 a fall in blood pressure. Episodes of light-headedness and certainly a blackout should be reported to one's doctor and investigated.

PHYSICAL EXAMINATION

In the majority of patients with Hypertrophic Cardiomyopathy, the physical examination is unremarkable and the abnormalities detected may be subtle. Most patients have a forceful or jerky pulse and a forceful heart beat, which can be felt on the left side of the chest. Both of these reflect the thickened, strongly contracting heart. However the most obvious abnormality on physical examination is a heart murmur,

which is present in 30-40% of patients.

HOW IS HYPERTROPHIC CARDIOMYOPATHY DIAGNOSED?

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

Electrocardiogram or ECG

An ECG records the electrical signal from the heart and is performed by placing electrodes on the chest, wrists and ankles (**Figure 13**). 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 (5-10%) 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.

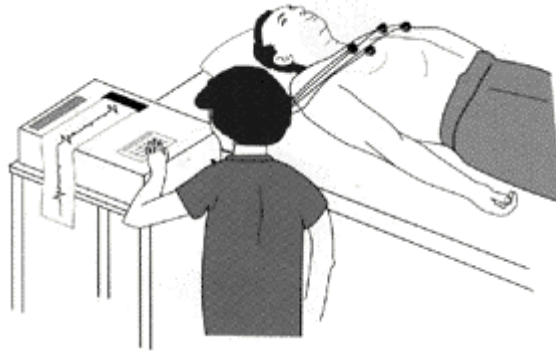


FIGURE 13

Echocardiogram or ECHO

Nowadays the diagnosis of Hypertrophic Cardiomyopathy is made by an ultrasound scan of the heart called an echocardiogram or **ECHO** for short. Like the ECG this is an entirely safe test and produces a picture of the heart similar to those in **Figures 9-12**, where excessive thickness of the muscle can be easily measured.

Additional equipment called **Doppler** ultrasound can produce a colour 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.



FIGURE 14

OTHER INVESTIGATIONS WHICH MAY BE NECESSARY

Additional investigations may be required to assess symptoms, to assess the risk of complications, particularly sudden death, and to select the best treatment.

Cardiac Catheterisation

Patients with breathlessness, which does not respond to therapy may require **cardiac catheterisation**. 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 **angiography** (x-ray) of the heart is taken 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 is performed during cardiac catheterisation.

Electrophysiological Studies (EPS)

These are a specialised form of catheterisation performed to define the risk of electrical instability which may predispose to sudden death. **Electrophysiological studies** involve 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 testing

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

Holter monitor

This test is a continuous **ambulatory** recording of the heart beat over 24 to 48 hours, see **Figure 15**. 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, substances producing very tiny (safe) amounts of radioactivity are given by injection. These tests may be used to assess the contraction, filling, structure and function of the heart and also to estimate its blood supply at rest and on exercise.

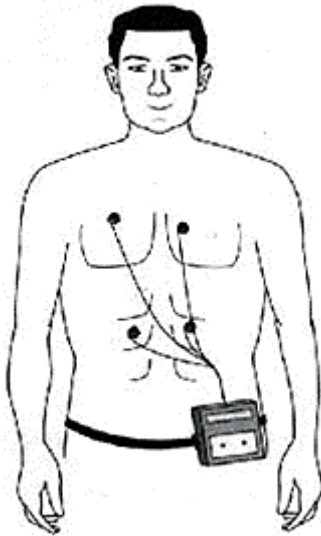


FIGURE 15

WHAT IS THE OUTLOOK FOR AFFECTED PERSONS?

The severity of symptoms and risk of complications varies greatly between patients but it should be emphasised that many people never have any serious problems related to their condition. Each person, however, must be assessed and advised individually and this website can only give a broad overview of the outlook for affected persons.

Pattern of Symptoms

In general, symptoms, whether mild or considerable, tend to be stable throughout adult life. Some people experience a worsening of symptoms in later life and this may be due to a progressive stiffening of the heart muscle or, in rare cases, to a reduction in the force of contraction.

COMPLICATIONS OF HYPERTROPHIC CARDIOMYOPATHY

In a minority of cases a number of specific complications can occur and may include:

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 arrhythmias called **ventricular tachycardia** (arising from the ventricles) or **atrial fibrillation** 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 (paroxysmal atrial fibrillation) or persistent. The loss of normal atrial contraction produces a risk of clot formation in the **atria**. **Anticoagulation** and drugs to slow the heart rate are required. Sometimes **Electrical Cardioversion** may be used to shock the heart back into normal rhythm. If this procedure is necessary, it is carried out under general anaesthesia.

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.

Heart Block

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

Sudden Death

A small number of patients with Hypertrophic Cardiomyopathy have an increased risk of premature death, which may occur with little or no warning. Systematic evaluation can identify the majority of patients at particular risk, with potential for targeted therapy i.e. drugs, pacemaker, ICD or surgery as appropriate.

IS THERE A CURE AVAILABLE?

At present there is no cure for Hypertrophic Cardiomyopathy although there is a slight possibility that some drugs may decrease the degree of muscle thickening. Regrettably, no treatment has yet been shown to return the heart to normal but research is continuing in this area. Developments are most likely to come from the early detection of persons carrying the gene for Hypertrophic Cardiomyopathy and from treating them to prevent the development of hypertrophy.

TREATMENT

Treatment aims to improve symptoms and prevent complications. Although the condition cannot be cured, there are many forms of treatment available which may improve the function of the heart and relieve symptoms. Many individuals who have few or no symptoms do not require treatment. For those who do, the treatment available is best considered under three headings:

- 1) **Drug Treatment**
- 2) **Surgery**
- 3) **Other Forms of Treatment**

1. 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

Beta-blockers are drugs which slow the heart beat and reduce its force of contraction. These drugs usually relieve chest pain, breathlessness and palpitation. Beta-blockers are widely used in medical practice for other types of heart disease and for high blood pressure. Occasionally excessive heart rate slowing can cause fatigue. There are many beta-blockers: the most commonly used are propranolol, atenolol, sotalol and nadolol.

Calcium Antagonists

The second major group of drugs used are the calcium antagonists or calcium channel blockers. Within this group verapamil is the drug which has been most used in Hypertrophic Cardiomyopathy. It improves the filling of the heart by reducing the stiffness of the myocardium and, like beta blockers, reduces symptoms such as chest pain, breathlessness and palpitations. Also, like beta-blockers, verapamil can cause excessive slowing of the heart rate and lower blood pressure.

Anti-Arrhythmic Drugs

These drugs might be used when an arrhythmia such as ventricular tachycardia is detected and felt to be important in an individual case. Of these anti-arrhythmic drugs amiodarone is the most commonly used in Hypertrophic Cardiomyopathy in the UK. It is an extremely effective drug and is most commonly used to reduce the risk of sudden death. However it does have several potential side effects, especially sensitivity to the sunlight (which can be avoided with use of barrier creams) and effects on the thyroid gland, which are reversible, but require regular testing.

Other Drugs

There are a number of specific complications described earlier which are rare but which require the use of additional drugs.

Anticoagulants

Patients with episodic or persistent atrial fibrillation should take anticoagulants (blood thinners) to prevent clot formation in the atria. Warfarin is the tablet commonly used. It requires monitoring with a blood test, approximately on a monthly basis.

Diuretics

Occasionally patients develop fluid retention and in this situation **diuretics** (water tablets) which increase urine flow are administered.

Antibiotics

Although **endocarditis** is rare, persons who have turbulent blood flow in the left ventricular outflow tract or across the mitral valve should receive **antibiotic prophylaxis** prior to dental procedures and any other situations where there is an increased risk of bacteria entering the bloodstream.

Summary of Drug Treatment

In summary, a variety of drug treatments are currently used in Hypertrophic Cardiomyopathy 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.

2. Surgery

Surgical myectomy (removal of muscle) 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 (**Figure 10**).

In this operation the surgeon removes a portion of the thickened muscle from the septum, thereby widening the outflow tract and relieving the obstruction.

Sometimes, together with 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 (**Figure 10**) if this mitral valve regurgitation is severe, then the valve may be replaced.

These are major operations which carry a definite risk and are therefore reserved for patients with severe symptoms and certain forms of Hypertrophic Cardiomyopathy.

Non surgical myectomy is a new procedure, pioneered in the UK, which has been devised to reduce hypertrophy of the upper septum without the need for open heart surgery. This technique involves injecting a small amount of alcohol solution down a minor branch of the coronary artery that supplies the upper septum, thus destroying this part of the myocardium. This is performed during a cardiac catheterisation and can be done under a local anaesthetic. Although only in the early stages of development, this strategy should provide a useful addition to the non surgical therapies available to patients with symptoms associated with Hypertrophic Cardiomyopathy.

Heart Transplantation

For a small minority, heart transplantation is necessary for those individuals who have a severe impairment of the pumping action of the heart.

3. Other Forms of Treatment

There are a number of other forms of treatment which may occasionally be recommended in Hypertrophic Cardiomyopathy. These are mentioned briefly:

Electrical Cardioversion

Atrial fibrillation may be stopped by the application of a small electrical shock to the chest. This procedure is carried out under general anaesthetic. Such cardioversion is often worthwhile as it may relieve symptoms and avoid the need for anticoagulation.

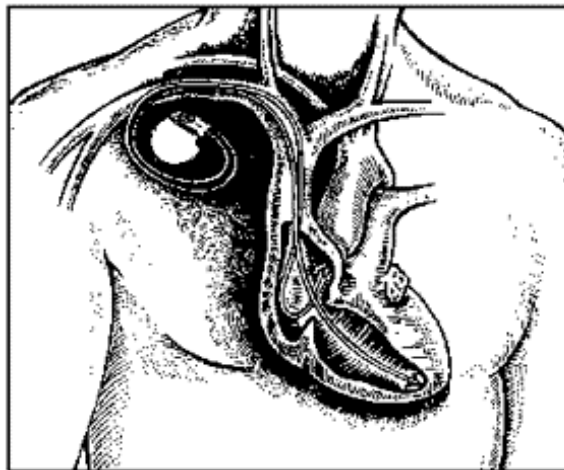


FIGURE 16

Pacemaker

As described earlier the normal electrical signal may fail to travel down the ventricles and in this situation a **pacemaker** is implanted. This involves putting a small box containing a battery under the skin of the chest, with a fine wire going through the veins to the heart to deliver the necessary electrical signal.

Dual Chamber Pacemaker

Pacemakers may also be used to reverse the normal electrical activation of the heart, preferentially activating the apex first. This allows the apex to contract before the septum, reducing the obstruction across the outlet of the heart and allowing more blood to be ejected from the left ventricle. However only a subset of patients may benefit from this technique. This form of treatment only applies to patients with Hypertrophic Obstructive Cardiomyopathy.

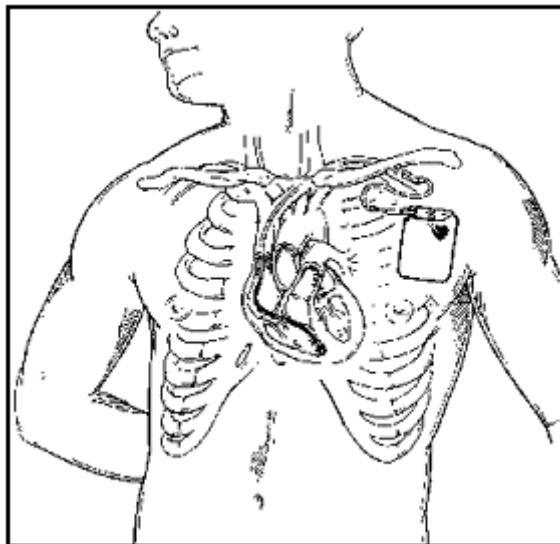


FIGURE 17

ICD (Implantable Cardioverter Defibrillator)

Recurrent episodes of rapid heart beat which cannot be controlled by drugs may require treatment with an **Implantable Cardioverter Defibrillator**. This requires implantation of a box which is slightly larger than a pacemaker under the skin in the chest and occasionally the abdomen. The box is attached with fine wires to the heart to record and deliver electrical impulses when normal electrical action is absent.

HOW IS LIFESTYLE AFFECTED BY HYPERTROPHIC CARDIOMYOPATHY?

Exercise

For many people the condition should not interfere with their lifestyle in any way. Some individuals may have symptoms related to exertion and find that they cannot undertake as much physical work or recreation as other people of their age. Medical advice should be sought before undertaking physically demanding activities. Some persons may be advised not to take part in competitive sports or other strenuous physical effort.

Life Insurance

As with any heart condition, difficulties may be experienced in obtaining life insurance and/or premiums may reflect an insurer's position. As of February 1997, the Association of British Insurers (ABI) position was that they will continue to take account of family history and of other medical information.

On genetic testing, the ABI have stated that "**any genetic tests which are to the detriment of applications for life insurance up to a total of &#pound;100,000 will be ignored where the insurance is linked to a new mortgage for a home.**"

The ABI also state that they will not ask people to take genetic tests when applying for life insurance but "people wishing to take out new life insurance policies will continue to be required to report the results of any genetic tests undertaken unless otherwise indicated by the life insurance company."

FAMILY SCREENING

The majority of patients with Hypertrophic Cardiomyopathy have at least one other affected first degree relative, i.e. a parent, brother, sister or child. When a person is diagnosed as having Hypertrophic Cardiomyopathy it is advised that all first degree relatives are screened for the condition. It is important to remember that family evaluation is recommended because Hypertrophic Cardiomyopathy may be present without any symptoms. Evaluation involves a physical examination, an ECG and an ECHO.

In most adults where the results of the above tests show no abnormality a diagnosis of Hypertrophic Cardiomyopathy may be excluded. In a small number of patients where there is no physical evidence of the disease, the abnormal gene may still be present. In this situation, the gene carrier will be normal, but may still pass on the gene for Hypertrophic Cardiomyopathy to the next generation.

In children, however, regular clinical evaluation is required until full growth spurt is completed at around 20 years of age, as symptoms may not show until adulthood. While undiagnosed, children of an affected parent are advised to be screened every 3 years until puberty and then on an annual basis until around 20 years. ECG and ECHO should be used each time.

WHAT ABOUT HAVING CHILDREN?

Inheritance

Clearly, the main concern of an affected parent is the risk of passing on the condition to their children. It is not possible to make a general statement about the risk and every case will need to be considered individually. For instance, in a family which has shown a **dominant** pattern of transmission from one generation to the next, (**see Figure 2**) the likelihood of a parent passing on the abnormal gene to a child is one in two. In other cases where there is no inheritance pattern in the family, the offspring of an affected individual probably also have a one in two risk of inheriting the condition.

Even if a child inherits the abnormal gene, the degree to which he or she might be affected is very variable. There is no definite method of predicting how severe the condition might be. In general it is recommended that children are screened regularly until adulthood. If there is no evidence of Hypertrophic Cardiomyopathy by early adulthood it is extremely unlikely that the condition will develop in later life.

Pregnancy and Childbirth

Pregnancy in Hypertrophic Cardiomyopathy is usually well tolerated and safe and for the majority of women with Hypertrophic Cardiomyopathy, pregnancy and delivery should be entirely normal.

However, as for women with any heart condition, pregnancy carries a slight additional risk for women with Hypertrophic Cardiomyopathy. This is because pregnancy imposes an increased demand on the heart.

Women may find that they develop symptoms for the first time in pregnancy or that their usual symptoms are increased. Also, the question of taking drug treatment around the time of conception or during pregnancy arises in many cases. For all these reasons it is best to plan pregnancy in advance and discuss all aspects at an early stage with your doctor.

It is best to avoid epidural anaesthesia at delivery as this may cause an excessive fall in blood pressure.

WHICH DOCTORS SHOULD BE INVOLVED IN ROUTINE MEDICAL CARE?

The family doctor or general practitioner should be involved at all times. The diagnosis of Hypertrophic Cardiomyopathy will usually be made by a cardiologist who should also be involved in follow-up. The exact arrangements will vary from place to place and depend on the individual patient. In general, all persons with the condition should have an annual medical check-up.

For some who are more symptomatic and need drug treatment, more frequent follow-up is required. After the initial diagnostic tests, the need for further investigations will depend on individual and local circumstances and on new developments.

GENERAL ADVICE

Diet

If an individual is overweight, it places an extra strain on the heart. Sensible eating habits are encouraged to maintain weight within the normal range for height and age. Any rapid increase in weight is likely to be due to fluid retention and your doctor needs to be notified.

Alcohol

There is no strong evidence on this in HCM, but moderation is advised, as alcohol has a depressant effect on heart muscle.

Driving (United Kingdom)

Group 1 Entitlement - Driving may continue **unless** there are distracting or disabling symptoms, in which case the DVLA should be notified.

Group 2 Entitlement - Recommended permanent refusal or revocation.

Flu vaccination

This may be recommended by your doctor to help prevent a severe bout of the flu. Unfortunately this vaccination is not always successful.

Holidays and Travel

It is advisable to see your GP for medical approval before travelling. If travelling abroad, you should also obtain information from your travel agent regarding insurance and the health care policy of the country you are visiting. An E1 11 form gives advice of health care in E.C. countries and is available at most UK post offices.

Allowances (United Kingdom)

Allowances are available for those people whose symptoms cause severe restrictions.

Disability Living Allowance is a Social Security benefit for those with an illness or disability who need help getting around or help with personal care.

Disability Working Allowance is a Social Security benefit to help with getting a job for those who are disadvantaged at work by their condition.

Details of these benefits can be obtained by calling Freephone number 0800 882 2000 (Benefit Enquiry Line) or by visiting your Social Security Office, Citizens Advice Bureau or Social Services Department.

WHAT RESEARCH IS BEING CONDUCTED?

Research is being conducted into Hypertrophic Cardiomyopathy on many levels and in many countries. As outlined earlier, genetic research aims to identify the remaining gene(s) which cause Hypertrophic Cardiomyopathy and to examine how these abnormalities resolve in disease. In particular, why do these gene abnormalities cause muscle cell disorganisation and overall thickening of the myocardium? What effect do they have on the way a single muscle cell contracts? Clinical research attempts to determine the reasons for symptoms and sudden collapse and to find new forms of treatment.

GLOSSARY

AMBULATORY: Refers to tests performed when a person is walking around or going about their normal activities.

ANGINA: A chest pain or discomfort usually brought on by exertion and relieved by rest. Angina results from insufficient oxygen supply to the heart muscle.

ANGIOGRAPHY: An internal x-ray of the heart and blood vessels which may be taken at the time of cardiac catheterisation (see below). In particular this test assesses the coronary arteries (the blood vessels which supply the heart muscle).

ANTIBIOTIC PROPHYLAXIS (Prevention): Persons with Hypertrophic Cardiomyopathy should usually take an antibiotic prior to dental procedures and other surgery. This is recommended because, during such procedures, bacteria may enter the bloodstream and could cause infection in the heart (see **ENDOCARDITIS**).

ANTICOAGULATION: Treatment (e.g. heparin or warfarin) to reduce the clotting ability of the blood. Such treatment is used when there is a risk of clot formation in the heart, e.g. in atrial fibrillation (see below).

AORTA: The main blood vessel which arises from the left ventricle and carries the blood from the heart to the rest of the body, see **Figure 7**.

APEX/APICAL FORM OF HYPERTROPHIC CARDIOMYOPATHY: The tip of the left ventricle is called the apex. This region may be the main part of the heart affected in which case the condition is then called Apical Hypertrophic Cardiomyopathy, see **Figure 12**.

ARRHYTHMIA: An abnormal rhythm or irregularity of the heart beat. The heart beat may be either too fast (**tachycardia**) or too slow (**bradycardia**). Arrhythmias may cause symptoms such as palpitation or light-headedness.

ATRIA: The filling chambers of the heart, one on the right side and one on the left. Blood is collected in the atria while the ventricles are contracting. This blood is then released into the ventricles when they are ready to fill, see **Figure 7**.

ATRIAL FIBRILLATION: A common type of arrhythmia. It begins in the atria and may be transient or persistent. The heart rhythm is irregular.

CARDIAC CATHETERISATION: A special test used for many forms of heart disease which is sometimes performed in Hypertrophic Cardiomyopathy. At cardiac catheterisation a fine tube is passed from a blood vessel (usually in the groin) to the heart, using x-ray guidance. The structure and function of the heart can then be assessed.

CARDIOMYOPATHY: Any disease of the heart muscle; cardia refers to the heart and myopathy means an abnormality of muscle.

CHROMOSOME: see **GENE**.

CONCENTRIC HYPERTROPHY: Where the walls of the left ventricle are thickened uniformly, also called symmetric hypertrophy, see **Figure 11**.

DIURETICS: Occasionally patients may develop fluid retention in which case they may be given tablets called diuretics which increase the production of urine by the kidneys.

DOMINANT PATTERN OF INHERITANCE: Where a condition is transmitted in a family from one generation to the next without skipping any generations.

DOPPLER ULTRASOUND: A test usually combined with **ECHO** (see below). Doppler can produce a **colour coded image** of blood flow within the heart and detect areas of **turbulent flow**. The pattern of filling and contraction of the heart in Hypertrophic Cardiomyopathy can also be assessed.

ECHOCARDIOGRAM (commonly abbreviated to **ECHO**): This is an ultrasound scan of the heart, similar to the type of scan performed during pregnancy. An ECHO can produce a picture of the heart similar to those in **Figures 9-12**. **Doppler** ultrasound (see above) is usually combined with ECHO. Echocardiography is the single most important test in the assessment of Hypertrophic Cardiomyopathy.

ELECTRICAL CARIOVERSION: An arrhythmia, such as atrial fibrillation, may be stopped by the application of an electric shock to the chest. If this procedure is necessary, it is carried out under general anaesthesia.

ELECTROCARDIOGRAM or ECG: A very common test for all forms of heart disease. In this test, electrodes placed on the chest, wrists and ankles detect an electrical signal from the heart. In Hypertrophic Cardiomyopathy the ECG often shows an increased electrical signal due to the muscle thickening.

ELECTROPHYSIOLOGICAL STUDY or EPS: In this test catheters are introduced into the heart as in cardiac catheterisation (see above). These catheters can record and stimulate the electrical activity of the heart.

ENDOCARDITIS: An infection of the heart which can occur in Hypertrophic Cardiomyopathy, but is very rare. Bacteria in the bloodstream can stick to the internal surface of the heart or heart valves where they have been roughened by turbulent blood flow.

EXERCISE (STRESS) TESTING: Exercise capability may be tested using either a treadmill or a stationary bicycle. During an exercise test a doctor and technician will monitor a patient's symptoms, ECG, blood pressure and, sometimes, breathing.

GENES AND CHROMOSOMES: Genes are the code or blueprint which build all the tissues in the body. Each individual has thousands of genes and they are all present in every cell of the body. Genes come in pairs, one inherited from one's mother and the other from one's father. In each cell the genes are grouped together in tiny, thread like structures called chromosomes. Each individual has 23 pairs of chromosomes.

HEART BLOCK: Occasionally the normal electrical signal does not travel down to the ventricles resulting in a slow heart rate or heart block. This situation can be diagnosed on ECG. If it occurs, a pacemaker is implanted.

HEART FAILURE (CONGESTIVE): A condition where weakness of the beating action of the heart causes fluid retention and symptoms of shortness of breath and tiredness on exercise. It can be associated with heart beat irregularities, fluid in the lungs or swollen legs.

HOLTER MONITOR: A continuous recording of the heart beat over 24 to 45 hours. Adhesive electrodes are placed on the chest, wires from these go to a special cassette recorder which is worn on a belt. A Holter monitor detects irregularity of the heart beat, otherwise known as arrhythmia.

HYPERTROPHY: A thickening of any body tissue, usually a muscle. In Hypertrophic Cardiomyopathy it refers to an abnormal or excessive thickening and of the heart muscle.

IHSS: Abbreviation of Idiopathic Hypertrophic Sub-aortic Stenosis which was an older name for Hypertrophic Cardiomyopathy, used particularly in the USA.

IMPLANTABLE CARDIOVERTER DEFIBRILLATOR (commonly abbreviated to ICD): A specialised pacemaker which recognises when a heart rate is excessively fast and responds by either pacing the heart or delivering a small electrical shock to restore the normal heart rhythm. The ICD can also serve as a conventional pacemaker to deliver the necessary impulses when the heart rate is too slow.

MITRAL REGURGITATION: Refers to blood leaking back through the mitral valve. This may occur in cases of Hypertrophic Cardiomyopathy, where there is outflow tract obstruction.

MURMUR: A murmur is caused by turbulent blood flow within the heart. In Hypertrophic Cardiomyopathy a murmur may be due to increased force of contraction and/or narrowing of the outflow tract. By analogy, increased pressure in a water hosepipe and/or narrowing of the outlet will cause turbulence.

MUTATION: A genetic defect that causes a change in the normal DNA code.

MYECTOMY: An operation which may be performed in Hypertrophic Cardiomyopathy to remove thickened muscle and therefore relieve the outflow tract obstruction. When successful, this operation is usually associated with improvement of symptoms.

MYOCARDIAL DISARRAY: Where under a microscope it may be seen that the normal parallel alignment of the muscle cells is absent. The muscle cell alignment appears disorganised or in disarray.

MYOCARDIUM: The specialised muscle which makes up the walls of the heart. It is this part of the heart which is abnormal in Hypertrophic Cardiomyopathy.

MYOSIN: A protein within each muscle cell which is involved in the normal contraction of the muscle. It has been discovered that the gene which produces myosin is abnormal in some families with Hypertrophic Cardiomyopathy.

OUTFLOW TRACT: The short channel in the heart through which blood passes from the ventricle into the aorta.

PACEMAKER: When the normal electrical impulse fails to be transmitted to the ventricles a pacemaker is implanted. This involves inserting a small box containing a battery under the skin of the chest, with a fine wire going through the veins to the heart, to deliver the necessary impulses.

PALPITATION: An uncomfortable awareness of the heart beat. Palpitation may be due to a normal heart beat made more prominent by anxiety or exercise or may be caused by an arrhythmia.

RESTRICTIVE CARDIOMYOPATHY: Restrictive Cardiomyopathy is characterised by a restriction to the inflow of blood into one or both ventricles of the heart. The thickness of the heart muscle and the size of the ventricular cavities are usually normal.

SEPTUM: The dividing wall between the right and left ventricles. In Hypertrophic Cardiomyopathy the muscle thickening is often most marked in the septum and this is called asymmetric septal hypertrophy,

see **Figure 9**.

SYSTOLIC ANTERIOR MOTION OF THE MITRAL VALVE or SAM: In some cases of Hypertrophic Cardiomyopathy, during the ejection of blood from the heart, the mitral valve moves forward and touches the septum (there should normally be a considerable gap between these structures) thus blocking the outflow tract. This is called SAM, see **Figure 10**.

VENTRICLES: The main pumping chambers of the heart, one on the right side and one on the left. The left ventricle is the part of the heart most commonly affected in Hypertrophic Cardiomyopathy.

VENTRICULAR TACHYCARDIA: A type of arrhythmia which takes the form of a fast heart beat arising in the ventricles.

GUIDELINES FOR PATIENTS AND RELATIVES ON THE MEDICAL ASPECTS OF HYPERTROPHIC CARDIOMYOPATHY (HCM)

By the Medical Advisory-Committee of the Cardiomyopathy Association

1. All patients diagnosed as having HCM should ask their general practitioner and cardiologist about the complications of the disease, the purposes and methods of treatment and the benefits, disadvantages and risks of the proposed medication or suggested operations.

2. Although HCM is not strictly curable, much can now be done by attention to appropriate lifestyle and administration of suitable drugs and, occasionally, surgical treatment. Ongoing research is opening up promising new possibilities.

3. It is important that relatives of patients with HCM should be examined by expert cardiologists to detect the disease if present.

4. Situations to avoid.

Violent exercise: always seek the advice of your consultant and general practitioner on how much exercise you should take

Acute severe loss of blood or body fluid: haemorrhage, diarrhoea, vomiting

Prolonged standing in hot conditions that might predispose to fainting which can be dangerous

Very hot baths/showers

During anaesthesia: including an epidural, special attention is required to avoid a sudden drop in blood pressure

5. Warning symptoms:

- a) Sudden loss of consciousness
- b) Episodes of rapid palpitation
- c) Onset of central chest pain
- d) Unexplained breathlessness

If any of these occur, see your doctor as soon as possible.

LATEST NEWS! A twenty minute video is now available for purchase on the diagnosis, treatment and hereditary effects of HCM.

