FACTS ABOUT

Cardiomyopathy

INTRODUCTION

Cardiomyopathy is a disease of the heart muscle. The heart loses its ability to pump blood and, in some instances, heart rhythm is disturbed, leading to irregular heartbeats, or arrhythmias. Usually, the exact cause of the muscle damage is never found.

Cardiomyopathy differs from many other heart disorders in a couple of ways. First, the types not related to coronary atherosclerosis are fairly uncommon. Cardiomyopathy affects about 50,000 Americans. However, the condition is a leading reason for heart transplantation.

Second, unlike many other forms of heart disease that affect middle-aged and older persons, certain types of cardiomopathies can, and often do, occur in the young. The condition tends to be progressive and sometimes worsens fairly quickly.

NONISCHEMIC CARDIOMYOPATHY

As noted, there are various types of cardiomyopathy. These fall into two major categories: "ischemic" and "nonischemic" cardiomyopathy.

- **Ischemic cardiomyopathy typically refers to heart muscle damage that results from coronary artery disease, such as heart attack, and will not be discussed here (see page 8 on how to get information on the disorder).**
- **Nonischemic cardiomyopathy includes several types. The three main types are covered in this fact sheet. They are: dilated, hypertrophic, and restrictive. The name of each describes the nature of its muscle damage.**

Dilated (Congestive) Cardiomyopathy

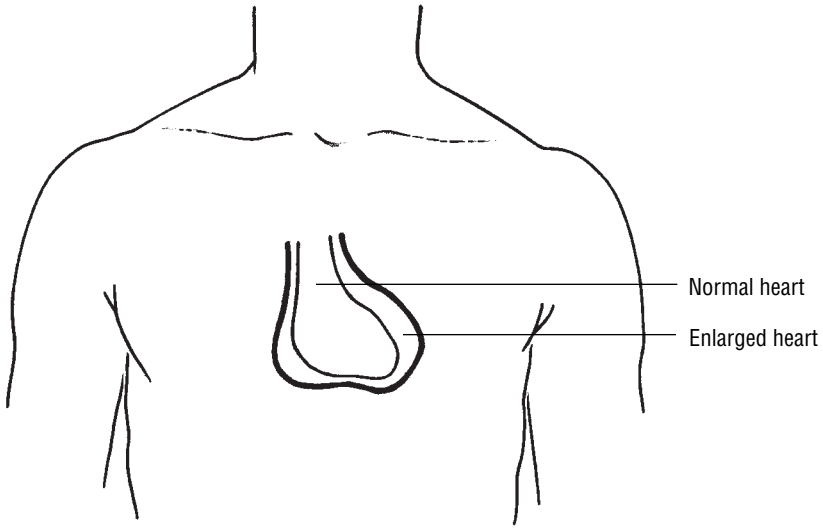
By far the most common type of nonischemic cardiomyopathy, the dilated (stretched) form occurs when disease- affected muscle fibers lead to enlargement, or dilation, of one or more chambers of the heart. This weakens the heart's pumping ability. The heart tries to cope with the pumping limitation by further enlarging and stretching—a process known as "compensation."

Dilated cardiomyopathy occurs most often in middle-aged people and more often in men than women. However, the disease has been diagnosed in people of all ages, including children.

In most cases, the disease is idiopathic—a specific cause for the damage is never identified.

The Heart in Cardiomyopathy

In cardiomyopathy, the heart muscle loses its ability to pump effectively. The heart becomes larger as it tries to compensate for its weakened condition. This illustration shows the change in size of the heart muscle.



But some factors have been linked to the disease's occurrence. For instance, alcohol has a direct suppressant effect on the heart. Dilated cardiomyopathy can be caused by chronic, excessive consumption of alcohol, particularly in combination with dietary deficiencies. Also, dilated cardiomyopathy occasionally occurs as a complication of pregnancy and childbirth. Other factors are: various infections, mostly viral, which lead to an inflammation of the heart muscle (myocarditis); toxins (such as cobalt, once used in beers, for instance); and, rarely, heredity.

Some drugs, used to treat a different medical condition, also can damage the heart and produce dilated cardiomyopathy. Such drugs include doxorubicin and daunorubicin, both used to treat cancer.

Whatever the cause, the clinical and pathological manifestations of dilated cardiomyopathy are usually the same.

Symptoms

Dilated cardiomyopathy can be present for several years without causing significant symptoms. With time, however, the enlarged heart gradually weakens.

This condition is commonly called "heart failure," and it is the hallmark of dilated cardiomyopathy. Typical signs and symptoms of heart failure include: fatigue; weakness; shortness of breath, sometimes severe and accompanied by a cough, particularly with exertion or when lying down; and swelling of the legs and feet, resulting from fluid accumulation that may also affect the lungs (congestion) and other parts of the body. It also produces abnormal weight gain. (The cough and congestion mimic and, therefore, can be misdiagnosed as pneumonia or acute bronchitis. Also, heart failure is often from heart disease other than cardiomyopathy.)

Because of the congestion, some physicians use the older term "congestive cardiomyopathy" to refer to dilated cardiomyopathy. In advanced stages of the disease, the congestion may cause pain in the chest or abdomen.

In advanced stages, some patients develop irregular heartbeats, which can be serious and even life threatening.

Diagnosis

Once symptoms appear, the condition may be tentatively diagnosed based on a physical examination and a patient's medical history. More often, though,

further examination is needed to differentiate dilated cardiomyopathy from other causes of heart failure.

A firm diagnosis typically requires a chest x ray to show whether the heart is enlarged, an electrocardiogram to reveal any abnormal electrical activity of the heart, and an echocardiogram, which uses sound waves to produce pictures of the heart.

Other, more specific tests may also be needed. These include:

- **A radionuclide ventriculogram. This involves injecting low-dose radioactive material (usually equal to that in a set of chest x rays) into a vein, through which it flows to the heart. Pictures are generated by a special camera to show how well the heart is functioning.**
- **A cardiac catheterization. In this procedure, a thin plastic tube is inserted through a blood vessel until it reaches the heart. A dye is injected and x rays taken to assess the heart's structure and function.**

Treatment

Since dilated cardiomyopathy is hard to diagnose early, it is rarely treated in its beginning stage.

The goal of treatment is to relieve any complicating factor, if known, control the symptoms, and stop the disease's progression. However, no cure now exists.

Therapy begins with the elimination of obvious risk factors, such as alcohol consumption. Weight loss and dietary changes, especially salt restriction, may also be advised.

Drugs used to treat the condition include:

- **Diuretics, which reduce excess fluid in the body;**
- **Vasodilators, such as angiotensin-converting enzyme (ACE) inhibitors, which relax blood vessels, helping to lower blood pressure and reducing the effort needed by the heart to pump blood through the body;**
- **Digitalis, which helps to improve pumping action and regulate heartbeat; and,**
- **Calcium blockers or beta blockers, which may be used in some patients to help regulate heartbeat and to alter the work of the heart muscle.**

Also, patients with irregular heartbeats may be put on any of various drugs to control the rhythm.

In critical cases where the condition is advanced and the patient does not sufficiently respond to other treatments, a heart transplantation may be needed. The patient's heart is replaced with a donor heart. Most heart transplant recipients are under age 60 and in good health other than their diseased heart.

Course of the disease

As the heart enlarges, it steadily decreases its efficiency in pumping blood and the amount of blood it can pump. As a result, some patients cannot perform even simple physical activities.

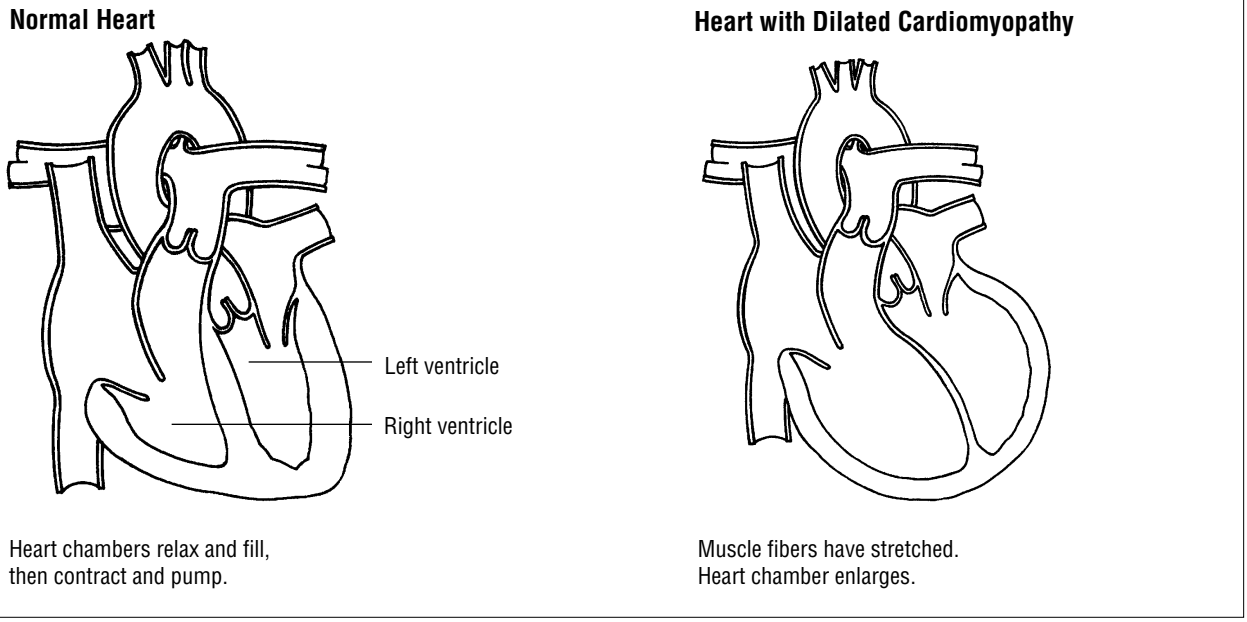
However, the disease also may remain fairly stable for years, especially with treatment and regular evaluation by a physician.

Unfortunately, by the time it is diagnosed, the disease often has reached an advanced stage and heart failure has occurred. Consequently, about 50 percent of patients with dilated cardiomyopathy live 5 years once heart failure is diagnosed; about 25 percent live 10 years after such a diagnosis.

Typically, patients die from a continued decline in heart muscle strength, but some die suddenly of irregular heartbeats.

For patients with advanced disease, heart transplantation greatly improves survival: 75 percent of patients live 5 years after a transplantation. However, in the United States, the scarcity of donor hearts limits the number of transplantations to about 2,000 persons a year. Those who qualify for heart transplantation often have to wait months, or even years, for a suitable donor heart. Some patients with dilated cardiomyopathy die awaiting a transplant but, according to recent studies, others improve enough from aggressive medical treatment to be taken off the waiting list.

Also, some critically ill cardiomyopathy patients with declining heart function use a small, implanted mechanical pump as a bridge to transplantation. Called left ventricular



assist devices (LVADs), these pumps take over part or virtually all of the heart's blood pumping activity. The devices provide only temporary assistance and are not now used as substitutes for heart transplantation.

Hypertrophic Cardiomyopathy

The second most common form of heart muscle disease is hypertrophic cardiomyopathy. Physicians sometimes call it by other names: idiopathic hypertrophic subaortic stenosis (IHSS), asymmetrical septal hypertrophy (ASH), or hypertrophic obstructive cardiomyopathy (HOCM).

In hypertrophic cardiomyopathy, the growth and arrangement of muscle fibers are abnormal, leading to thickened heart walls. The greatest thickening tends to occur in the left ventricle (the heart's main pumping chamber), especially in the septum, the wall that separates the left and right ventricles. The thickening reduces the size of the pumping chamber and obstructs blood flow. It also prevents the heart from properly relaxing between beats and so filling with blood. Eventually, this limits the pumping action.

Hypertrophic cardiomyopathy is a rare disease, occurring in no more than 0.2 percent of the U.S. population. It can affect men and women of all ages. Symptoms can appear in childhood or adulthood.

Most cases of hypertrophic cardiomyopathy are inherited. Because of this, a patient's family members often are checked for signs of the disease, although the signs may be much less evident or even absent in them. In other cases, there is no clear cause.

Symptoms

Many patients have no symptoms. For those who do, the most common are breathlessness and chest discomfort. Other signs are fainting during physical activity, strong rapid heartbeats that feel like a pounding in the chest, and fatigue, especially with physical exertion.

In some cases, the first and only manifestation of hypertrophic cardiomyopathy is sudden death, caused by a chaotic heartbeat. The heart's lower chambers beat so chaotically and fast that no blood is pumped. Instead of beating, the heart quivers.

In advanced stages of the disease, patients may have severe heart failure and its associated symptoms, including fluid accumulation or congestion.

Diagnosis

By listening through a stethoscope, a physician may hear the abnormal heart sounds characteristic of hypertrophic cardiomyopathy. The electrocardiogram (EKG, or ECG) may help diagnose the condition by detecting changes in the electrical activity of the heart as it beats.

Echocardiography is one of the best tools for diagnosing hypertrophic cardiomyopathy. It uses sound waves to detect the extent of muscle-wall thickening and to assess the status of the heart's functioning.

Physicians also may request radionuclide studies to gather added information about the disease's effect on how the heart is pumping blood.

Other tests that also may provide useful information are the chest x ray, cardiac catheterization, and a heart muscle biopsy.

Treatment

Treatments for hypertrophic cardiomyopathy vary but can include the following:

- **Lifestyle changes. Patients with serious electrical and blood-flow abnormalities must be less physically active.**
- **Medications. Various drugs are used to treat the disease. They include beta blockers (to ease symptoms by slowing the heart's pumping action), calcium channel blockers (to relax the heart and reduce the blood pressure in it), antiarrhythmic medications, and diuretics (to ease heart failure symptoms).**

  However, drugs do not work in all cases or may cause adverse side effects, such as fluid in the lungs, very low blood pressure, and sudden death. Then, other treatment, such as a pacemaker or surgery, may be needed.
- **Pacemakers. These change the pattern and decrease the force of the heart's contractions. The pacemaker can reduce the degree of obstruction and so relieve symptoms. A pacemaker needs to be carefully monitored after its insertion in order to properly adjust the electrical impulse. Some patients who have a pacemaker inserted feel no relief and go on to have heart surgery.**
- **Surgery. This usually calls for removal of part of the thickened septum (the muscle wall separating the chambers) that is blocking the blood flow. Sometimes, surgery also must replace a heart valve--the mitral valve, which connects the left ventricle and the left atrium, the upper chamber that receives oxygen-rich blood from the lungs.**

  Surgery to remove the thickening eases symptoms in about 70 percent of patients but results in death in about 1 to 3 percent of patients. Also, about 5 percent of those who have surgery develop a slow heartbeat, which is then corrected with a pacemaker.

Course of the disease

The course of the disease varies. Many patients remain stable; some improve; some worsen in symptoms and lead severely restricted lives. Patients may need drug treatment and careful medical supervision for the rest of their lives.

Hypertrophic cardiomyopathy patients also are at risk of sudden death. About 2 to 3 percent die each year because the heart suddenly stops beating. This cardiac arrest is brought on by an abnormal heartbeat. Over 10 years, the risk of sudden death can be 20 percent or more.

Restrictive Cardiomyopathy

Restrictive cardiomyopathy is rare in the United States and most other industrial nations. In this disease, the walls of the ventricles stiffen and lose their flexibility due to infiltration by abnormal tissue. As a result, the heart cannot fill adequately with blood and eventually loses its ability to pump properly.

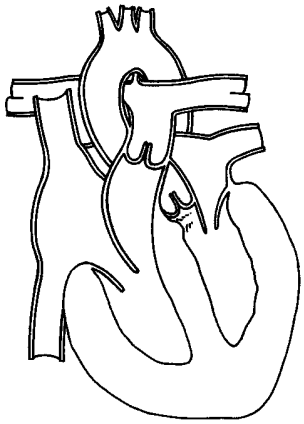
Restrictive cardiomyopathy typically results from another disease, which occurs elsewhere in the body. In the United States, restrictive cardiomyopathy is most commonly related to the following: amyloidosis, in which abnormal protein fibers (amyloid) accumulate in the heart's muscle; sarcoidosis, an inflammatory disease that causes the formation of small lumps in organs; and hemochromatosis, an iron overload of the body, usually due to a genetic disease.

In general, restrictive cardiomyopathy does not appear to be inherited; however, some of the diseases that lead to the condition are genetically transmitted.

Symptoms

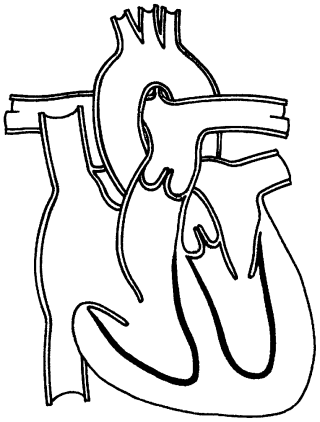
Typical signs of the condition include symptoms of congestive heart failure: weakness, fatigue, and breathlessness. Swelling of the legs, caused by fluid retention, occurs in a significant number of patients. Other symptoms include nausea, bloating, and poor appetite, probably because of the retention of fluid around the liver, stomach, and intestines.

Heart with Hypertrophic Cardiomyopathy



Growth and arrangement of muscle fibers are abnormal. Heart walls thicken, especially in the left ventricle.

Heart with Restrictive Cardiomyopathy



Ventricle walls stiffen and lose flexibility.

Diagnosis
A physician may suspect restrictive cardiomyopathy based on a patient's symptoms and the presence of another disease. Although symptoms of congestive heart failure may predominate, the size of the heart remains relatively small, unlike other cardiomyopathies.

Diagnostic information comes from an electrocardiogram or any of several imaging studies that provide pictures of the heart. These include echocardiography, magnetic resonance imaging, and computed tomography.

A definite diagnosis usually requires cardiac catheterization studies or a biopsy, in which a tiny piece of tissue--including heart muscle--is removed for laboratory analysis.

Treatment
Restrictive cardiomyopathy has no specific treatment. The underlying disease that leads to the heart problem also may not be treatable.

In general, the use of traditional heart drugs has been limited in this cardiomyopathy, although diuretics may help control fluid accumulation.

In rare cases, surgery is sometimes used to try to improve blood flow into the heart.

Course of the disease
The condition is similar to dilated cardiomyopathy and tends to worsen with time. Only about 30 percent of patients survive more than 5 years after diagnosis.

Future Directions

Future advances in the diagnosis and treatment of cardiomyopathy depend on a better understanding of the disease process and why heart muscle is damaged. A lot of research is under way to identify these processes and whether they can be halted or even reversed. Much of the research is conducted at or supported by the National Heart, Lung, and Blood Institute (NHLBI).

Promising clues came from investigators at and supported by the NHLBI who discovered some of the genes responsible for hypertrophic cardiomyopathy. Their work represents an important first step in understanding how the disease is transmitted and how it progresses.

Researchers also are trying to determine the best use of currently available treatments, especially drug therapies. Drugs useful for other conditions may help treat cardiomyopathy. For example, drugs effective in treating high blood pressure also help manage heart failure and irregular heartbeats.

Additionally, much work has been--and continues to be--done on identifying factors that increase or decrease the risk of death for persons with cardiomyopathy. Knowing which patients are at the greatest risk is very important in determining the best approach to evaluation and treat-

ment of their condition.

The development of improved treatments for cardiomyopathy, however, awaits still more research and a better understanding of the disease process.

GLOSSARY

Angiotensin converting enzyme (ACE) inhibitor—A drug used to decrease pressure inside blood vessels.

Arrhythmia—An irregular heartbeat.

Beta blocker—A drug used to slow the heart rate and reduce pressure inside blood vessels. It also can regulate heart rhythm.

Calcium channel blocker (or calcium blocker)—A drug used to relax the blood vessel and heart muscle, causing pressure inside blood vessels to drop. It also can regulate heart rhythm.

Cardiac arrest—A sudden stop of heart function. See also "sudden death."

Cardiac catheterization—A procedure in which a thin, hollow tube is inserted into a blood vessel. The tube is then advanced through the vessel into the heart, enabling a physician to study the heart and its pumping activity.

Cardiomyopathy—A disease of the heart muscle (myocardium).

Congestion—Abnormal fluid accumulation in the body, especially the lungs.

Digitalis—A drug used to increase the force of the heart's contraction and to regulate specific irregularities of heart rhythm.

Dilated cardiomyopathy—Heart muscle disease that leads to enlargement of the heart's chambers, robbing the heart of its pumping ability.

Diuretic—A drug that helps eliminate excess body fluid; usually used in the treatment of high blood pressure and heart failure.

Dyspnea—Shortness of breath.

Echocardiography—A test that bounces sound waves off the heart to produce pictures of its internal structures.

Edema—Abnormal fluid accumulation in body tissues.

Electrocardiogram (EKG or ECG)—Measurement of electrical activity during heartbeats.

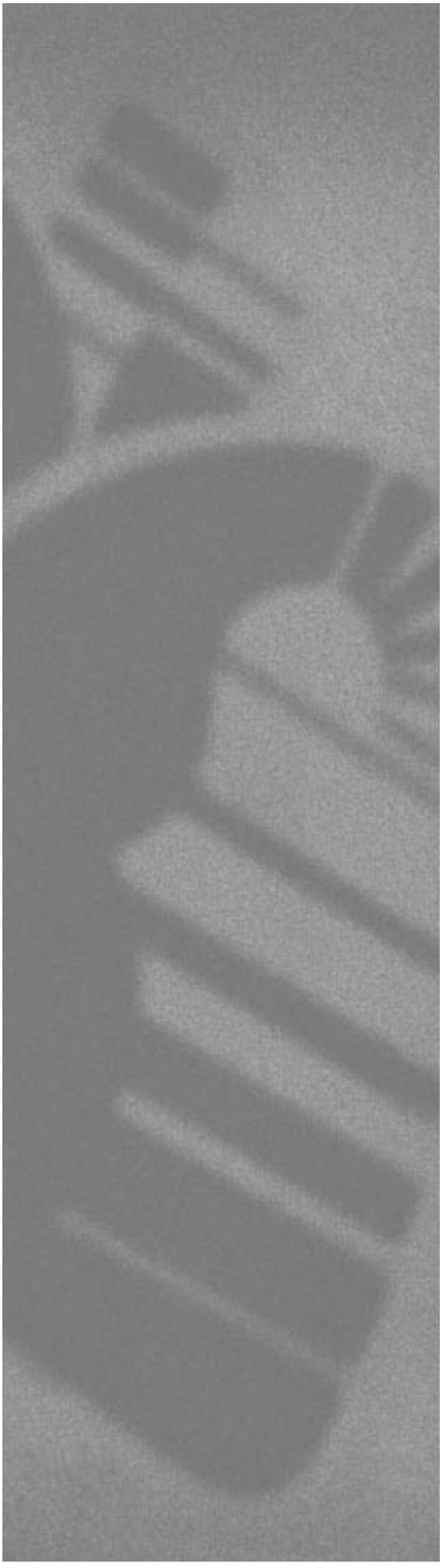
Heart failure—Loss of pumping ability by the heart, often accompanied by fatigue, breathlessness, and excess fluid accumulation in body tissues.

Hypertrophic cardiomyopathy— Heart muscle disease that leads to thickening of the heart walls, interfering with the heart's ability to fill with and pump blood.

Idiopathic—Results from an unknown cause.

Left ventricular assist device (LVAD)—A mechanical device used to increase the heart's pumping ability.

Pulmonary congestion (*or edema*)—Fluid accumulation in the lungs.



Restrictive cardiomyopathy—Heart muscle disease in which the muscle walls become stiff and lose their flexibility.

Septum—In the heart, a muscle wall separating the chambers.

Sudden death—Cardiac arrest caused by an irregular heartbeat. The term "death" is somewhat misleading, because some patients survive.

Ventricles—The two lower chambers of the heart. The left ventricle is the main pumping chamber in the heart.

Ventricular fibrillation—Rapid, irregular quivering of the heart's ventricles, with no effective heartbeat.

FOR MORE INFORMATION

For more information, contact the NHLBI Information Center, a service of the NHLBI and the National Institutes of Health. The Information Center provides information to health professionals, patients, and the public about the treatment, diagnosis, and prevention of heart, lung, and blood diseases.

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http://www.nhlbi.nih.gov/nhlbi/nhlbi.htm

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