



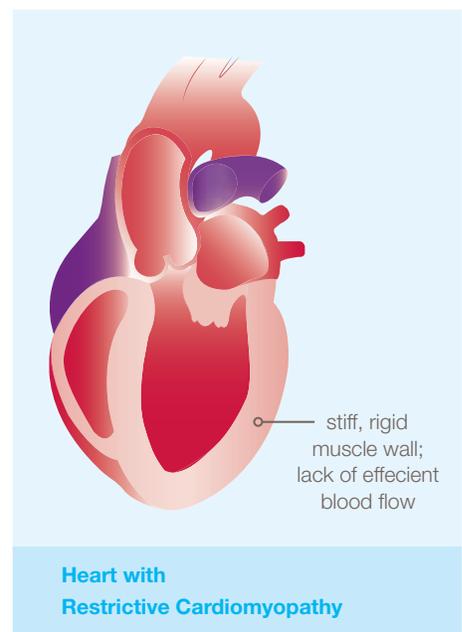
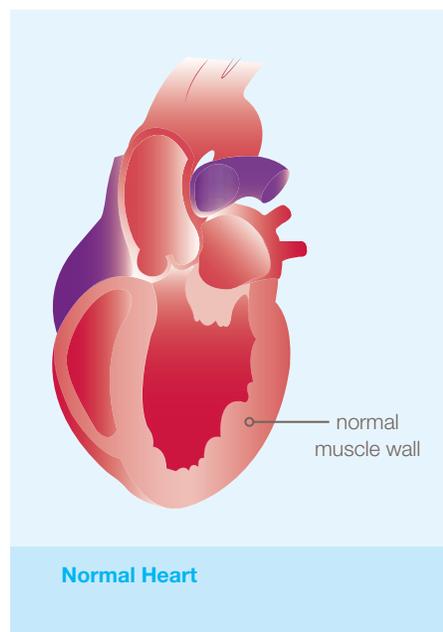
Restrictive Cardiomyopathy (RCM)

CONTACT US

To learn more about treatment of Restrictive Cardiomyopathy at the Heart Institute at Cincinnati Children's Hospital Medical Center, please call **513-803-1746**.

Definition

Cardiomyopathy is a disease that affects the heart muscle. In restrictive cardiomyopathy (RCM) the heart muscle becomes rigid and unable to relax and fill with blood. The function or squeeze of the heart may be normal, but the relaxation is abnormal. When the lower left chamber of the heart, called the left ventricle, is unable to stretch and fill with blood, pressure builds up causing abnormal heart rhythms and symptoms of heart failure.



Symptoms

Many individuals with RCM experience no symptoms. Symptoms can develop at any age and are primarily associated with heart failure.

Older children and adults

- Shortness of breath and fatigue
- Feeling dizzy or light-headed
- Fainting or passing out (syncope)
- Persistent cough
- Feeling abnormal heart beats (palpitations)

Newborns and babies

- Difficulty feeding or poor growth
- Excessive sweating with feeds or activity
- Excessive fussiness
- Fast breathing

Even though some people can have RCM and experience no symptoms, they may still be at risk for complications of the condition. The most serious complications of RCM is sudden cardiac arrest. Sudden cardiac arrest is a sudden loss of heart function caused by a dangerously fast heart rhythm. Unless emergency treatments, including CPR and defibrillation, are initiated immediately, sudden cardiac death can occur. For this reason, it is important for anyone at risk to be followed by a cardiologist with experience in caring for individuals with RCM.

Causes

RCM can be inherited. When an individual is diagnosed with RCM, there is a 25 to 30 percent chance that an underlying genetic cause will be identified. Most commonly, the genetic cause results in a condition that runs in a family and affects only the heart. It is possible that parents, siblings, and even aunts/uncles or cousins of an individual diagnosed with RCM may also have a heart muscle condition. If they haven't had cardiac screening, family members may not know they are affected.

There are other genetic causes of RCM which can be associated with other health problems. These include genetic syndromes. In some people, the cause of RCM is unknown. Other health conditions can cause build-up of substances in the heart muscle which cause it to be rigid resulting in abnormal relaxation. Some of these conditions include:

- Build-up of abnormal proteins (amyloidosis) in the heart muscle
- Excess iron (hemosiderosis) in the heart
- Other systemic diseases (sarcoidosis)
- Inherited diseases (such as Fabry disease)

Diagnosis

The diagnosis of RCM is based on family history, medical history, a physical exam, and cardiac testing.

- **Family history:** A family tree should be constructed with specific attention to cardiomyopathy, rhythm problems, sudden cardiac or unexplained death, cardiac surgery or presence of other cardiac disease in relatives.
- **Medical history:** A history of heart failure symptoms and rhythm problems is important.
- **Physical exam:** Special attention should be paid to the cardiac and skeletal muscle systems.
- **Cardiac testing:** An echocardiogram is the most common test used to diagnose RCM, as the enlargement of the upper chambers of the heart (atria) is usually visible. The atria become enlarged because of increasing pressure which results from abnormal relaxation of the ventricles. Cardiac catheterization may be used to measure the pressures in the heart. Other tests may include blood tests, electrocardiogram (EKG), exercise stress echo test, and magnetic resonance imaging (MRI).

Management and Treatment

The treatment for individuals with RCM can be challenging. Treatment is focused on minimizing or preventing symptoms, protecting the patient from abnormal heart rhythms and delaying disease progression.

Individuals with RCM need to be followed by a cardiologist on a regular basis. Medications are used to treat symptoms but currently there are not any medications that improve the heart's ability to relax. The most important treatment is the prevention of abnormal heart rates, or arrhythmias.

Lifestyle changes are recommended for some individuals. Some individuals will be discouraged from exercise, especially competitive sports.

Some individuals with RCM are at increased risk for a sudden cardiac arrest. For individuals determined to be at increased risk of a sudden cardiac arrest, preventive treatments such as medication to control heart rate (antiarrhythmic medications) or an implantable cardioverter defibrillator (ICD) may be recommended.

If the condition is severe, a heart transplant may be considered.

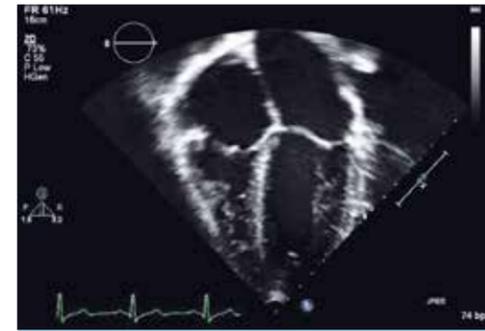


Fig 1.1

Normal Heart

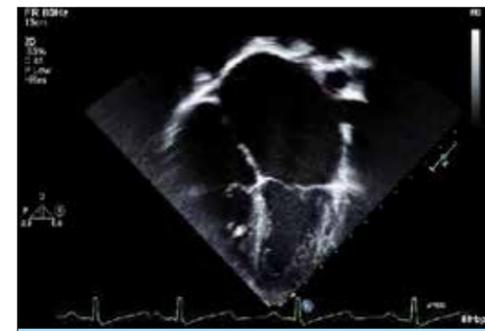


Fig 1.2

Heart with Restrictive Cardiomyopathy

*Note that the atria are enlarged.

RESOURCES FOR FAMILIES

The Cardiomyopathy Association:

www.cardiomyopathy.org

Children's Cardiomyopathy Foundation:

www.childrenscardiomyopathy.org



Cincinnati Children's is ranked #8 in Cardiology and Heart Surgery, and third among all Honor Roll hospitals in the 2012–13 *U.S. News & World Report* listing of Best Children's Hospitals.

Genetic Testing

Genes are packages of genetic information that tell our bodies how to develop and function. RCM can be caused by a change or mutation in a gene that encodes proteins important for the contraction or squeeze of the heart muscle. Most often, RCM is inherited. A parent that carries a gene mutation will have a 50 percent chance of passing it on to each child.

Genetic testing is available to look for mutations in the genes known to cause RCM. This type of testing is done on a blood sample. It is best to begin genetic testing in a family with an individual who is known to have RCM. If a gene mutation is found in someone with RCM, genetic testing for the same mutation can be offered to close family members. Finding a gene mutation in other family members will help identify relatives who may be at risk to develop RCM and who should have cardiac screening.

A genetic counselor and/or geneticist can assess the family history and provide information about the chance that there is a genetic predisposition to cardiomyopathy. A genetics professional can also explain and facilitate genetic testing and interpret results for patients and families.

Screening for Family Members

All first degree relatives of an individual who has RCM should undergo routine cardiac evaluation. This includes parents, brothers, sisters and children. If a gene mutation causing RCM in an individual is known, cardiac screening is recommended for those family members proven to also carry the gene mutation. Routine cardiac screening would not be recommended for family members who did not inherit the gene mutation.

The timing of routine cardiac screening for family members should be discussed with a cardiologist and genetics professional who have familiarity in caring for individuals with cardiomyopathy. How frequently an individual should be evaluated is based on published guidelines and the individual's age, personal medical and family history.

For immediate consultations, physician referrals and patient care follow-up, call the National Physician Priority Link at **888-987-7997**.

www.cincinnatichildrens.org