What is restrictive cardiomyopathy?

Restrictive cardiomyopathy (RCM) is less common affecting 3 to 5 percent of children with cardiomyopathy. With RCM, the walls of the lower **chambers of the heart (ventricles) are abnormally stiff** but not necessarily thickened or enlarged. The heart's rigid walls fail to relax and expand adequately, "restricting" the ability of the heart to fill with blood.

While the heart pumps normally, it is still unable to supply enough blood to the body. This puts pressure on the upper chambers of the heart (atria), and they become enlarged and out of proportion to the size of the lower chambers of the heart (ventricles). In advanced stages of the disease, the heart may not pump blood efficiently and blood may back up into the liver and lungs as a result of congestive heart failure.

How many children are affected?

According to the Pediatric Cardiomyopathy Registry, RCM occurs at a rate of **less than 1 per million children**.

Diagnosis

What is the prognosis?

RCM is a rare disease and there is **limited information** on the disease in children. Long-term survival increases for children who receive heart transplants. Irreversible and severe pulmonary hypertension has been the only risk factor associated with poor outcome for children with RCM.

This booklet was created to provide families and caregivers with a broad overview of cardiomyopathy and is for general information only. The material presented is not intended to be complete or serve as medical advice. The information should not be a substitute for consultation with a qualified health care professional who is more familiar with individual medical conditions and needs.

The Children's Cardiomyopathy Foundation (CCF) is dedicated to finding causes and cures for pediatric cardiomyopathy through the support of research, education, and increased awareness and advocacy.

Register with CCF for additional resources: childrenscardiomyopathy.org

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Understanding Restrictive Cardiomyopathy

What causes restrictive cardiomyopathy?

In most cases, the cause of RCM in children is unknown (idiopathic). Many RCM cases result from **sporadic gene mutations** when changes in the DNA of a gene occur spontaneously during fetal development for unknown reasons. Occasionally, RCM runs in a family and is inherited in an autosomal dominant manner in which one parent contributes the defective gene and there is a 50 percent chance their child will inherit the condition.

RCM also can be **secondary to a number of rare cardiac and systemic disorders** that lead to a build-up of fats, proteins or iron in the heart. This includes endomyocardial fibrosis, infiltrative disorders (amyloidosis, sarcoidosis, hemochromoatosis), connective tissue diseases (scleroderma) and rare metabolic disorders (Gaucher or Fabry disease). RCM caused by the infiltration of the heart muscle is more likely to be inherited in an autosomal recessive manner in which both parents contribute a defective gene and there is a 25 percent chance their child will inherit the condition.

What are the common symptoms?

The onset of symptoms in RCM is often very subtle. Symptoms arise from the decreased filling of the heart and insufficient blood flow to the body. Infants and young children may show irritability, poor appetite and slow weight gain. Older children may experience fatigue, **difficulty exercising or breathing** (dyspnea), a persistent cough or wheezing, fainting (syncope), abnormal heart beat (palpitations), chest pain (angina) and an upset stomach. As RCM advances, fluid builds up in the lungs as well as in the veins that carry blood back toward the heart. This may cause neck veins to bulge; an enlarged liver; and fluid in the abdomen, face and legs.

Some children may develop abnormal heart rhythms (arrhythmia) where the heart beats too slow (bradycardia) or too fast (tachycardia). With some forms of RCM, a slow heart rate may develop from **heart block**. This is due to the abnormal conduction of signals to the heart's pumping chambers.

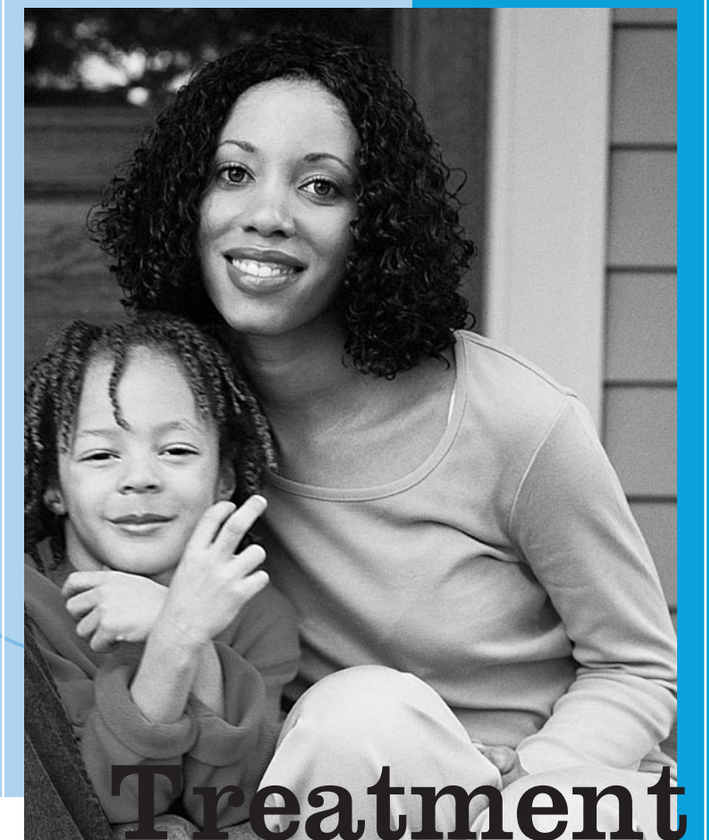
Pulmonary hypertension and blood clots are common complications of RCM. **Pulmonary hypertension,** defined as high blood pressure in the lungs, may occur when the arteries in the lungs are restricted and the heart must work harder to pump blood through the lungs. **Blood clots** may also form in the enlarged areas of the heart and travel to the brain or other parts of the body.

How is restrictive cardiomyopathy diagnosed?

In the early stages, RCM may be difficult to detect in a physical examination because of the absence of symptoms. Therefore, cardiologists rely on noninvasive cardiac tests such as the **echocardiogram** and the **electrocardiogram** (EKG) to diagnose the condition. An echocardiogram measures the size of the heart, how well the heart pumps and the severity of pulmonary hypertension if an issue. An ejection fraction can be calculated by measuring the percentage of blood ejected from the heart with each beat. Unlike other forms of cardiomyopathy, children with RCM typically have normal ejection fractions of 50 to 70 percent because the heart's pumping function is unaffected until the later stages of the disease. An EKG provides information on the heart's electrical activity and whether there is heart block or irregular heart rhythms. This test nearly always shows abnormal patterns associated with the enlargement of the heart's upper chamber (atria).

Other tests may be ordered to access the heart's condition and determine a treatment plan. These tests include a **chest x-ray** to check the heart's shape and size and to look for fluid in the lungs, **computed tomography** (CT scan) to observe the structure and function of the heart and blood vessels, **magnetic resonance imaging** (MRI) to evaluate heart and blood vessel function, and a **Holter monitor** to look for abnormal heartbeats.

In order to get a more precise RCM diagnosis, more invasive tests may be necessary. A **cardiac catheterization** is used to measure heart and lung pressures, which are usually elevated with RCM. A heart (endomyocardial) biopsy, which involves removing a small piece of heart muscle for microscopic examination, is usually performed at the same time. The laboratory checks for infiltrating substances or abnormal deposits in the heart. It also looks for any indication of constrictive pericarditis, which resembles RCM in its signs and symptoms



Treatment

What are the treatment options?

For children with RCM, medical therapy aims to improve symptoms of heart failure, control arrhythmias and prevent blood clots. Because the cause of cardiac dysfunction is different from DCM and HCM, **medications** such as angiotensin converting enzyme (ACE) inhibitors, calcium channel blockers and beta-blockers are not commonly used to treat children with RCM. Low doses of diuretics (bumetanide, chlorothiazide, furosemide, spironolactone) may be used to alleviate symptoms related to excess fluid in the lungs and body. Anti-coagulation medication or blood thinners (aspirin, dipyridamole, enoxaparin, heparin, warfarin) may also be prescribed to prevent the development of blood clots.

Special diets and anti-inflammatory medications may be used to treat RCM caused by excess deposits in the heart. For children with heart rhythm problems, anti-arrhythmic medications (amiodarone, digoxin, procaineamide) may be prescribed to keep the heart beating at a regular rate. A **pacemaker** or **automatic implantable cardioverter defibrillator** (AICD) may be surgically inserted to control arrhythmias that do not respond to medication.

Close monitoring is important since a child with RCM can be stable for years and then suddenly deteriorate rapidly. A **heart transplant** may be necessary when a child does not respond to medical treatment and begins to show signs of severe heart failure. High blood pressure in the lungs (pulmonary hypertension) is more likely to develop in children with RCM than in other forms of cardiomyopathy, and it can negatively affect the outcome of a heart transplant. Therefore, children with RCM need to be listed for transplant earlier if they develop symptoms of pulmonary hypertension. Unfortunately, a heart transplant may not be an option when RCM is related to a disease that causes abnormal deposits in the heart and other organs.