What is restrictive cardiomyopathy?

Restrictive cardiomyopathy (RCM) is less common, affecting 3 to 5 percent of children with cardiomyopathy. In 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), which 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.


GAVIN

Gavin, diagnosed with RCM and treated with multiple surgeries and a pacemaker as an infant, is now thriving on medication, enjoying time with his twin sister, friends, and dogs, and spreading joy with his laughter.

What causes RCM?

In most cases, the cause of RCM in children is unknown (idiopathic). However, some cases will result from a genetic mutation. 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 that their child will inherit the condition.

RCM can also be secondary to a number of rare cardiac and systemic disorders that lead to a buildup of fats, proteins, or iron in the heart. This includes endomyocardial fibrosis, infiltrative disorders (amyloidosis, sarcoidosis, hemochromatosis), 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 that their child will inherit the condition. If a genetic cause is found, ask your child's doctor whether close family members (like parents or siblings) should also be tested.

What is the prognosis?

RCM is a rare disease, and there is limited information on the disease in children. There are no medications that are known to improve survival in children with RCM, and some children will need a heart transplant. Additionally, pulmonary hypertension is associated with poor outcomes for children with RCM.

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This booklet, featuring real children with cardiomyopathy, 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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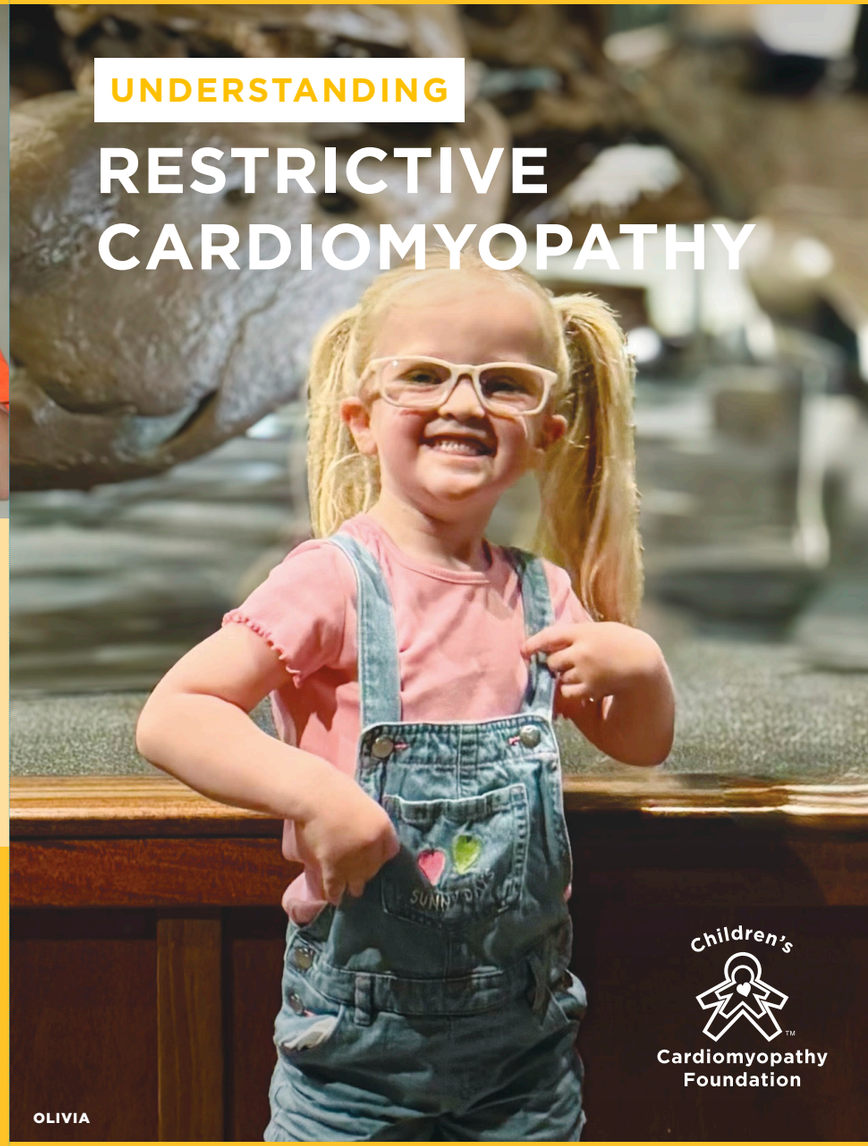
UNDERSTANDING RESTRICTIVE CARDIOMYOPATHY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How is RCM diagnosed?

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 echocardiograms and electrocardiograms (ECGs/EKGs) 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 (EF) 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 chambers (atria).

OWEN

Other tests may be ordered to assess 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, a 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. 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, may be performed at the same time. The laboratory checks for infiltrating substances or abnormal deposits in the heart.

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 heartbeat (palpitations), chest pain (angina), fluid retention (edema), 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 (arrhythmias) where the heart beats too slow (bradycardia) or too fast (tachycardia). In some cases 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 (thromboses) 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 many children are affected?

According to the CCF-supported Pediatric Cardiomyopathy Registry (PCMR), RCM occurs at a rate of less than 1 per 1-million children.

What are treatment options?

For children with RCM, medical therapy aims to improve symptoms of heart failure, control arrhythmias, and prevent blood clots. The cause of cardiac dysfunction in RCM is different from that of DCM and HCM. Therefore, 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. Anticoagulation 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, antiarrhythmic medications (amiodarone, digoxin, procainamide) 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. 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.

JACOB