

RESTRICTIVE CARDIOMYOPATHY

Restrictive cardiomyopathy (RCM) is a rare form of heart muscle disease that is characterized by restrictive filling of the ventricles. In this disease the contractile function (squeeze) of the heart and wall thicknesses are usually normal, but the relaxation or filling phase of the heart is very abnormal. This occurs because the heart muscle is stiff and poorly compliant and does not allow the ventricular chambers to fill with blood normally. This inability to relax and fill with blood results in a “back up” of blood into the atria (top chambers of the heart), lungs and body causing the symptoms and signs of heart failure.

Within the broad category of cardiomyopathy, RCM is the least common in children, accounting for 2.5–5% of the diagnosed cardiomyopathies (occurs in less than one per million children). The average age at diagnosis is 5 to 6 years. RCM appears to affect girls somewhat more often than boys. There is a family history of cardiomyopathy in approximately 30% of cases. In most cases the cause of the disease is unknown (idiopathic), although a genetic cause is suspected in most cases of pediatric RCM.

Signs and Symptoms of RCM

In children the first symptoms of RCM often seem related to problems other than the heart. The most common symptoms at first may appear to be lung related. Children with RCM frequently have a history of “repeated lung infections” or “asthma.” In these cases, referral to a cardiologist eventually occurs when a large heart is seen on chest x-ray. The second most common reason for referral is an abnormal physical finding during a doctor’s examination. Children who have ascites (fluid in the abdomen), hepatomegaly (enlarged liver) and edema (fluid causing puffy looking feet, legs, hands or face) are often sent to see a gastroenterologist first. Referral to a cardiologist is made when additional cardiac signs or symptoms occur, a chest x-ray is found to be abnormal or no specific gastrointestinal cause is found for the edema or enlarged liver. When the first sign of the disease is an abnormal heart sound, or signs of heart failure are recognized, then earlier referral to a cardiologist occurs. In approximately 10% of cases, fainting is the first symptom causing concern. Unfortunately, sudden death has been the initial presentation in some patients.



Diagnosis of RCM

Restrictive cardiomyopathy is among the rarest of childhood cardiomyopathies. Its diagnosis is difficult to establish early in the clinical course due to the lack of symptoms. Therefore, in many cases, this diagnosis is made only after presentation with symptoms such as decreased exercise tolerance, new heart sound (gallop), syncope (passing out) or chest pain with exercise.

Once suspected, there are certain tests that can help confirm this diagnosis. An electrocardiogram, or EKG, which records the electrical conduction through the heart, can be very helpful. This can show abnormally large electrical forces from enlargement of the atria (upper chambers) of the heart. An echocardiogram, or ultrasound of the heart, can provide additional clues to help make this diagnosis. Generally, in children with RCM, the echocardiogram shows marked enlargement of the atria (upper chambers), normal sized ventricles (lower chambers) and normal heart function. In more advanced disease states, pulmonary artery pressure (blood pressure in the lungs) will be increased and can often be estimated during the echocardiogram.

Cardiac catheterization is usually the next procedure done to confirm the diagnosis. During this procedure, a catheter (thin plastic tube) will be slowly advanced through an artery or vein into the heart (while watching its course on a TV monitor) so that pressures within the heart chambers can be measured. These measurements often show significantly elevated pressures during the relaxation period of the heart (when it fills with blood before the next beat) and varying degrees of increased pulmonary artery pressure (which can confirm the echo estimates) in the absence of any other structural heart disease. In very rare cases, based on clinical symptoms and prior laboratory evaluation, a cardiac biopsy may be performed. This involves removing tiny pieces of heart muscle for inspection under the microscope to search for potential causes of this condition (such as amyloidosis or sarcoidosis, which are common causes of RCM in adults but rarely in pediatric patients).

Finally, since childhood RCM is often genetic and in many cases will be inherited, once this diagnosis is established, your doctor will likely request that parents, siblings of the patient and sometimes other close relatives be screened with an echocardiogram to rule out the presence of this disease in other family members.

Causes of RCM

Although the cause of RCM is not known in most pediatric cases, there is some scientific evidence suggesting that individual genetic “mutations” may be a cause in some cases of RCM in children. For a greater understanding of the basics of human inheritance patterns and a more detailed discussion of the potential genetic causes of RCM, the reader is encouraged to read separate sections entitled “Overview of Inheritance” and “Genetics of Cardiomyopathies” printed elsewhere in this brochure.

Current Treatment

Currently, there are no therapies that can “cure” RCM; however, some treatments are available that can improve symptoms in children with RCM. The choice of a specific therapy depends on the clinical condition of the child, the risk of dangerous events and the ability of the child to tolerate the therapy.

Medical Therapies to Treat RCM and Associated Heart Failure

Some children with RCM have signs and symptoms of heart failure due to the abnormal relaxation properties of the heart muscle. The most common types of medications used to treat heart failure under these circumstances include diuretics, beta-blockers and occasionally afterload reducing agents.

Diuretics, sometimes called “water pills,” reduce excess fluid in the lungs or other organs by increasing urine production. Diuretics can be given either orally or intravenously. Common diuretics include furosemide, spironolactone, bumetanide and metolazone. Common side effects of diuretics include dehydration and abnormalities in the blood chemistries (particularly potassium loss). In patients with RCM, diuretics must be used very carefully and given only in doses to treat extra lung and abdominal fluid without inducing excessive fluid loss as this may cause symptomatically low blood pressure.

Beta-blockers slow the heartbeat and increase the relaxation time of the heart. This may allow the heart to fill better with blood before each heart beat and decrease some of the symptoms created by the stiff pumping chambers. Common beta-blockers (taken by mouth) include carvedilol, metoprolol, propranolol and atenolol. Side effects include dizziness, low heart rate, low blood pressure, and, in some cases, fluid retention, fatigue, impaired school performance and depression.

Anticoagulation Medications

In children with a heart that does not relax well, there is a risk of blood clots forming inside the heart possibly leading to a stroke. Anticoagulation medications, also known as blood thinners, are often used in these situations. The choice of anticoagulation drug depends on how likely it is that a blood clot will form. Less strong anticoagulation medications include aspirin and dipyridamole. Stronger anticoagulation drugs are warfarin, heparin, and enoxaparin; these drugs require careful monitoring with regular blood testing. While variable, common side effects of anticoagulants include excessive bruising or bleeding from otherwise minor skin injuries, interaction with other medications and, for warfarin, fluctuations in anticoagulation blood levels caused by changes in daily dietary intake. Information regarding which food groups can significantly affect warfarin levels can be obtained from your cardiologist.

Surgery for Restrictive Cardiomyopathy

No surgery has been effective in improving the heart function in restrictive cardiomyopathy. Heart transplantation is the only effective surgery offered for patients with RCM, particularly those who already have symptoms at the time of diagnosis or in whom reactive pulmonary hypertension exists.

HEART TRANSPLANTATION

Since there are no proven effective therapies for children with RCM, transplantation is the only known intervention for this disease. This is especially true in cases where evaluation has demonstrated the presence of pulmonary hypertension, which can be fatal if not treated. For children with RCM, heart transplantation can address both the abnormal heart function as well as associated pulmonary hypertension. A heart transplant offers the child with RCM the chance to return to a normal lifestyle. While a donor heart can cure the symptoms of heart failure and greatly improve survival, it is a major operation with considerable risks and long-term complications. Once a transplant is done, other concerns arise, such as infection, organ rejection, coronary artery disease, and the side effects of medications.

Prognosis

The long-term prognosis for children with RCM varies depending on the symptoms at the time of diagnosis and the presence of pulmonary hypertension. Children with RCM should be watched closely for the development of excess fluid retention, abnormal heart rhythms, blood clots inside the heart or evidence of progressive pulmonary hypertension.

As discussed previously, RCM is rare, and there is limited information on this disease in children. Once diagnosed, approximately 20% develop thrombotic or embolic events (blood clots). The average two-year survival rate for children with RCM is 45–70% including those treated with heart transplantation. Survival increases significantly for children who are transplanted. Irreversible pulmonary hypertension (high blood pressure in the lung vessels) has been the only major risk factor associated with poor outcome among these patients. Therefore, it is important that transplant be considered sooner than later to optimize long-term survival, as the waiting time for a donor heart can be unpredictable. Finally, a child with RCM should be closely monitored at a center with expertise in pediatric heart failure, cardiomyopathy, arrhythmias and transplantation in order to ensure rapid response to any worsening of his or her condition.

Living with RCM

The diagnosis of RCM affects many areas of a child's life. The following sections outline the general approaches to living with RCM. It is important that specific recommendations are developed by the team caring for the child with restrictive cardiomyopathy.

Physical Activity

Children with RCM are not allowed to play competitive sports because of the possibility of a sudden collapse or increased heart failure. A competitive sport is an organized team activity for which training is required.

A child with restrictive cardiomyopathy and no heart failure symptoms can be allowed to perform recreational athletic activities, also known as low-dynamic or low-static sports, in a non-competitive situation. Specific activity recommendations should be individualized by the treating cardiologist.

School

The intellectual, psychological and social benefits of attending school cannot be overestimated in the child with RCM. Adjusting medication schedules so they do not interfere with school activities, discussing safe activity levels with school personnel, and providing tutoring to maintain academic performance are important interventions that can help a child to stay in school and keep up with their peers. Often close communication between the parents, medical care team, and the school nurse can help to keep a child up to date in school.

Friends

Every effort should be made to allow a child with RCM to spend time with friends. The child should also be allowed to participate in recreational activities whenever possible. However, an effort should be made to avoid contact with those who are acutely ill with fever, even though many children with RCM are able to tolerate upper respiratory tract illnesses (common colds) well.

Psychological Issues

Adjusting to having a chronic illness is stressful for the child and the family. The child's reaction to having RCM often depends on the stage of the child's development. Discussions about the disease should be tailored to the specific concerns of the child. Child-life professionals and pediatric psychiatrists are important resources to help children cope, and their services are often available through the treating center.

Family

The impact of a diagnosis of RCM is felt throughout the child's immediate and extended family. It is important for parents and other caregivers to realize that they are not alone in feeling the weight of responsibility that comes with taking care of a child with a chronic illness. Anticipating and/or preventing the stress imposed by an illness is an important part of caring for the child and family and personnel at the cardiomyopathy center can help identify issues that can lead to increased stress.

Practical solutions to problems giving medications, keeping track of appointments, and maintaining normal family life can often be found through discussions with nurse clinicians, the social worker, psychiatrist, and other parents of children with RCM.

Diet

All children with RCM should follow a healthy diet. The recommendations published in 2005 by the United States Department of Agriculture (USDA) can be found at the following website address: <http://www.mypyramid.gov/>. In children with RCM and extra lung and abdominal fluid, a low-salt diet is recommended to avoid excessive fluid retention.

Some children with heart failure may not grow well. In these cases, a diet that increases calories is recommended. Children who are taking some medications may have low levels of magnesium or potassium and a diet that has a higher amount of one or both of these two electrolytes may be recommended. Some children with severe heart failure can retain extra body fluid, and it may be necessary to limit the amount that a child can drink to prevent fluid from accumulating in the lungs.

Health Maintenance

Routine pediatric care is important for children with RCM. Regular well child visits and standard childhood immunizations should be performed. The influenza vaccine should be administered on a yearly basis. Children under age 2 should receive Synagis for protection against respiratory syncytial virus.

A medical alert bracelet is an important safety measure for children with RCM. In the event of an emergency, these bracelets allow medical personnel to know details about a child's illness, especially if a family member is not available.

What Does the Future Hold for RCM?

Slowly, progress is being made in our ability to diagnose RCM in both the clinical and molecular arenas. However, much additional research is needed in this field. Areas of research to be highlighted over the next decade include: 1) better understanding of RCM as a disease process and the characteristics of the disease as they relate to outcome, which will lead to better management strategies; 2) increased clinical trials which will lead to new drug development and more effective therapies; and 3) molecular identification of novel genetic mutations as well as more precise diagnostic genetic testing/screening which will result in more accurate diagnosis.

It is the expectation of the medical community that the data derived from exploring these avenues of scientific research will translate into a clinician's ability to tailor medical therapy based on a given child's precise diagnosis. Achieving this goal over the next couple of decades will represent a large milestone in the field of pediatric cardiomyopathy and will, hopefully, improve the ongoing care and prognosis of children afflicted with these heart muscle diseases.