



Understanding Pediatric Cardiomyopathy



What is pediatric cardiomyopathy?

Cardiomyopathy means “a **disease of the heart muscle.**” It is a chronic and sometimes progressive disease in which the heart muscle (myocardium) becomes abnormally enlarged, thickened and/or stiffened such that the heart can no longer contract or relax normally. Eventually, the heart loses its ability to pump blood effectively and irregular heartbeats (arrhythmias) and heart failure may occur.

Cardiomyopathy can affect anyone regardless of age, race, gender or socioeconomic background. While cardiomyopathy predominantly affects adults, in rare instances it does affect infants and children, in which case it is called **pediatric cardiomyopathy.**

According to the Pediatric Cardiomyopathy Registry, 1 in every 100,000 children in the U.S. under the age of 18 is diagnosed with cardiomyopathy. This conservative estimate does not include children that are undiagnosed or have secondary cardiomyopathy where the heart condition is caused by infection, metabolic irregularities or another systemic disease.



Information

There are four types of cardiomyopathy:

Dilated Cardiomyopathy (DCM)

DCM is the most common form, and it occurs when the muscle fibers of one or both pumping chambers are weak and unable to contract normally. As the heart pumps less strongly, it enlarges to help deliver more blood with each beat. Eventually the weakened heart fails, causing fluid to build up in other organs.

Hypertrophic Cardiomyopathy (HCM)

The second most common form, HCM, is characterized by the abnormal growth and arrangement of muscle fibers in the heart. The thickened and stiffer heart muscle reduces the capacity of the heart to fill and contract properly. This may lead to arrhythmia or obstruction of blood outflow from the heart. In advanced HCM, the heart muscle may thin and stretch to compensate for the poor pumping action.

Restrictive Cardiomyopathy (RCM)

RCM is a less common condition, and it accounts for 3 to 5 percent of patients with cardiomyopathy. With this form, the heart has normal contraction but abnormal relaxation. The walls of the ventricles become stiff, preventing the heart from filling with blood. The low amount of blood in the heart leads to exercise intolerance and heart failure.

Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)

ARVC is the least common form and extremely rare in young children. It usually presents in teens or young adults instead. ARVC is caused by the death of healthy heart muscle and its replacement with scar tissue and fat. This results in a disorganized structure of heart muscle tissues causing abnormal electrical activities (arrhythmia) and problems with the heart's contraction.

There are also new forms of cardiomyopathy that are being recognized. **Left ventricular non-compaction cardiomyopathy (LVNC)** is a rare form of cardiomyopathy that can manifest as DCM, HCM or RCM. With this condition, the developing heart muscle fails to become compacted, resulting in a spongiform appearance of the chamber wall, increased muscle thickness and weak pumping action.

Each type of cardiomyopathy is characterized by a slightly different disease process and set of symptoms. For specific information on the different forms of cardiomyopathy, please refer to the inserts accompanying this booklet.

What causes cardiomyopathy?

The cause of cardiomyopathy in children is currently poorly understood. Approximately two-thirds of reported cases do not have a known cause. Of those cases where a cause is identified, myocarditis (27 percent) and familial inheritance (25 percent) are the leading reasons according to the Pediatric Cardiomyopathy Registry. This is followed by neuromuscular disorders (22 percent), metabolic disorders (16 percent) and genetic malformation syndromes (10 percent).

Cardiomyopathy can be **inherited or acquired**. Some cardiomyopathies (primary cardiomyopathies) are isolated and affect only the heart while other cardiomyopathies (secondary cardiomyopathies) are associated with non-heart related medical problems. Both can be caused by genetic or unknown (idiopathic) reasons. Genetic conditions are usually caused by changes in specific genes (mutations) that are inherited from one or both parents. In some instances, cardiomyopathy can occur for the first time in a family due to a sporadic genetic mutation in the child that is not inherited from either parent. This happens when changes in the DNA of a gene occur spontaneously during fetal development for unknown reasons.

When the disease is inherited, there are several ways that cardiomyopathy can be passed onto the child. With **autosomal dominant genetic conditions**, one parent carries a genetic mutation for cardiomyopathy and has a 50 percent chance of passing it on to their child. Usually this parent also has cardiomyopathy and may or may not show symptoms of the disease. With **autosomal recessive genetic conditions**, such as fatty acid oxidation defects or glycogen storage diseases, both parents are unaffected carriers and have a 25 percent chance of having an affected child. Some forms of cardiomyopathy caused by Barth syndrome or Duchene muscular dystrophy can occur sporadically in boys or may be passed on from mother to son (**X-linked transmission**). In the latter, sons will be at a 50 percent risk of having the condition while daughters will not have the condition but may be carriers like their mothers.

Some cardiomyopathies are acquired due to exposures to infection, toxins or medications that can weaken the heart muscle. The most common cause of acquired cardiomyopathy is **myocarditis**, a viral infection that causes inflammation of the heart. Viruses that can affect the heart include coxsackie A & B, echovirus, adenovirus, HIV and mumps. Less common causes of acquired cardiomyopathy include nutritional deficiencies and exposure to excessive alcohol, radiation, heavy metals and cancer chemotherapy drugs.

Cardiomyopathy can also develop in response to an underlying medical condition that affects other organs or the entire body system. These include **metabolic and mitochondrial abnormalities**, build up of proteins in the heart (amyloidosis), excess iron in the heart (hemochromatosis), thyroid disorders, severe anemia, complications from other cardiovascular conditions

(congenital heart defect, hypertension or surgery), auto-immune disease (lupus) and pregnancy related complications.

There are more than 100 specific causes of cardiomyopathy in children that can be classified into one of four forms (DCM, HCM, RCM and ARVC). Determining the root cause of the disease can be a complex and involved process. Most of the rare disorders associated with cardiomyopathy can only be confirmed by diagnostic testing and evaluation from other medical specialists. Your child's cardiologist and team of specialists (geneticists, neurologists) will work together to determine the cause of the disease in your child.

How is cardiomyopathy diagnosed?

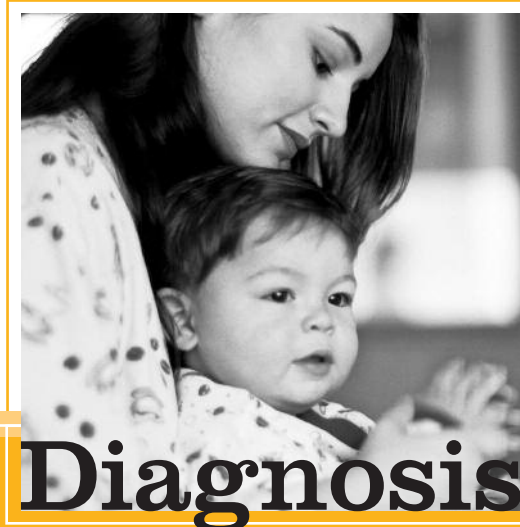
Since pediatric cardiomyopathy is rare, it can be missed or misdiagnosed. Many times the disease goes undetected in young children because there is no family history of cardiomyopathy or the child shows no symptoms (asymptomatic). Many children with cardiomyopathy are active, appear healthy and meet their developmental milestones.

An accurate and thorough diagnosis involves determining the type of cardiomyopathy, its severity and cause. The more specific the diagnosis, the more tailored and effective the treatment can be. If a child is suspected of having cardiomyopathy, an evaluation will begin with a visit to a pediatric cardiologist for a **complete history review** and **physical examination**. During the evaluation, the cardiologist will ask about the child's and family's

medical history, symptoms and prior medical tests. The physician will also examine the child and listen to the heart through a stethoscope. Specific cardiac tests will follow to determine the heart size and whether there is thickening, blood flow obstruction or valve leakage.

Diagnosis is confirmed by an **electrocardiogram (EKG)** and two-dimensional **echocardiogram** with Doppler ultrasound. An EKG records the heart's electrical activity (rate and rhythm) using electrodes placed on the child's arms, legs and chest wall. An echocardiogram uses ultrasound waves to produce moving pictures of the beating heart on a video screen. These cross-sectional views of the heart allow the cardiologist to measure the heart's size, muscle thickness, pumping ability, degree of obstruction and potential leakage (regurgitation).

Other **noninvasive procedures** may be performed, including magnetic resonance imaging (MRI), a Holter monitor and an exercise stress test for older children. In some cases, an **invasive procedure** under anesthesia may be necessary for further



investigation of the heart function or to determine possible causes. This includes cardiac catheterization, radionuclide ventriculogram, heart (endomyocardial) biopsy, skeletal muscle biopsy or an electrophysiology (EP) study. These procedures are explained in more detail on CCF's website.

If necessary, the diagnostic visit also may involve blood or urine testing, assessment of a child's neurological development and muscle tone, review of any unusual physical features associated with other syndromes, and evaluation of any organs that might be affected. Some forms of cardiomyopathy are further defined by **genetic testing**.

During the evaluation process, you may work with a pediatric cardiologist, geneticist, heart surgeon and possibly specialists from neurology, immunology, nutrition, endocrinology and infectious disease. Whenever possible, it is best to work with a **team of experts** at a medical center that treats many children with cardiomyopathy and engages in research on the disease. These are usually large teaching hospitals with a pediatric cardiomyopathy clinic and a heart failure or heart transplant program.

What are the common symptoms?

Cardiomyopathy is an **extremely variable disease**, and it may manifest differently in adults and children. Symptoms can vary with different forms of cardiomyopathy and even among family members with the same form of cardiomyopathy. Symptoms can be absent, mild or severe.

Children may be diagnosed following the detection of a heart murmur or evidence of heart enlargement during a routine visit to the pediatrician. However, the **signs of cardiomyopathy are not always so obvious** in children. Symptoms may be mistaken to be a common cold, flu, asthma or stomachache. In babies and young children it is more difficult to assess the severity of the disease because they are not able to clearly communicate how they feel.

Many children are only referred to a heart specialist once more serious symptoms appear. These symptoms include shortness of breath, rapid breathing, fainting spells, irregular or rapid heart rate, chest pain and extreme fatigue. In infants, symptoms include poor weight gain, difficulty feeding, excessive sweating, and unusual fussiness or lethargy. For information about symptoms associated with a particular form of cardiomyopathy, please refer to the DCM, HCM or RCM inserts available with this booklet.

What are available treatment options?

Cardiomyopathy is a chronic disease of the heart muscle. Unfortunately, there is no treatment that can repair the structural damage of the heart. If detected in the early stages, cardiomyopathy can be controlled with medications and certain surgical procedures. In general, the goal is to restore a child to the best possible health with the least amount of intervention.

A child's treatment plan should be done in consultation with a pediatric cardiologist, and if necessary a cardiothoracic surgeon, electrophysiologist and geneticist. Treating cardiomyopathy in a standardized manner is difficult because of its variability in manifestation and outcome. Each type of cardiomyopathy has slightly different symptoms and therefore requires an **individualized treatment plan** to address varying short- and long-term medical issues. Several factors will determine the appropriate therapy such as the child's age, overall health, medical history, underlying cause, disease severity, tolerance for certain medications and procedures, and the expected outcome for the disease. The risk for arrhythmia, sudden cardiac arrest and progressive heart failure will also determine how aggressive treatment should be.

Medication or medical therapy is used for various reasons: to help the heart beat more effectively, control symptoms related to heart failure or blood flow obstruction, prevent the formation of blood clots, reduce inflammation, and regulate abnormal heart rhythms.

Medications commonly prescribed include angiotensin converting enzyme (ACE) inhibitors, beta-blockers, calcium-channel blockers, digoxin, diuretics, anti-arrhythmics, antibiotics and anti-coagulants. These medications require careful monitoring for possible side effects and changes in heart function. The medications and dosages will depend on each child's heart condition and may change as the child grows and responds to therapy.

If a child has an irregular heartbeat, a **pacemaker** or **automatic implantable cardioverter defibrillator (AICD)** may be recommended. This is a small mechanical device surgically placed under the skin of the chest or abdomen with wire leads threaded through veins into the heart or attached to the surface of the heart. This is a self-regulating sensing device that activates when a patient's heartbeat reaches dangerously high or low levels. In some DCM cases, a biventricular pacemaker may be used to synchronize the contraction of the left and right chambers of the heart, even without the presence of arrhythmia.



Treatment

Surgery may be an additional treatment option for HCM patients. A **septal myectomy** involves removing a part of the thickened heart muscle. When there is mitral regurgitation, a valve repair or replacement is performed to stop leakage of blood from the left ventricle into the left atrium (mitral regurgitation). These surgical procedures help to control symptoms of heart failure, but they do not stop the progression of the disease. For DCM and RCM, there are no surgical options for improving heart failure.

When a child no longer responds to treatment and experiences end-stage heart failure, a **heart transplant** may be necessary. While waiting for a donor heart, intravenous medications and cardiac assist devices may be used to extend the life of a child's heart.

What is the likely prognosis for my child?

The long-term outlook of pediatric cardiomyopathy continues to be unpredictable because of its multiple causes and wide spectrum of disease presentation and outcomes. It is not unusual for one child to have a more severe form of the disease and his or her sibling or parent to have a mild or asymptomatic form of the same disease. The overall prognosis for a child will depend on the type of cardiomyopathy, cause of the disease, family history and the severity of cardiac dysfunction when diagnosed.

Although there is no cure for the disease, symptoms and complications can be managed and controlled with regular monitoring. Some children will stabilize with treatment and lead a relatively normal life with few physical activity restrictions. Other children with a more serious form of cardiomyopathy may face more limitations, need specialized care and encounter minor developmental delays. Occasionally children with certain types of cardiomyopathy do improve, but the majority do not show any recovery in heart function.

For the most severe cases, a heart transplant may be necessary. Children diagnosed with DCM or RCM are more likely to require a transplant; it is less common with HCM. Post transplant survival continues to improve with two-



Outcome

year survival rates at 80 percent and ten-year survival rates near 60 percent. Longer-term survival remains to be determined but is expected to improve with more medical progress and research.

Should my family undergo screening or genetic testing?

Since **cardiomyopathy can be inherited**, other family members may be at risk even if they have no symptoms. Therefore, it is recommended that parents and siblings of a diagnosed child undergo an **echocardiogram** and **EKG screening**. This is especially important for at risk individuals where there is a family history of sudden infant death, sudden cardiac arrest or heart attack under the age of 40. Even if the initial exam results are normal, family members may need periodic re-screening. Screening frequency will depend on the type of cardiomyopathy, its cause and the age of the individual. Your cardiologist and geneticist will advise what is appropriate for your family's medical situation.

Clinical genetic testing may also be an option for certain types of cardiomyopathy. These diagnostic tests can determine whether an individual has a known genetic mutation that may cause the disease. Early detection can lead to **treatments** that can control or limit the damage caused by the disease. Parents discovered to carry a gene associated with cardiomyopathy should seek genetic counseling to determine who else in the extended family may be at risk and to discuss

the possibility of recurrence in future pregnancies.

How will the disease impact my child and family?

While pediatric cardiomyopathy is a serious heart condition, many children can lead a normal life with proper medical management. The degree of impact on lifestyle will depend on whether a child improves, stabilizes or progresses to an advanced stage of heart failure. It will also depend on the type of cardiomyopathy and its cause. A diagnosis will mean **more frequent visits to the cardiologist** to monitor changes in heart function, evaluate treatment effectiveness and discuss any new complications. It may also mean daily intake of **various medications** and some activity restrictions. If surgical procedures are required and there is prolonged hospitalization, a child may temporarily fall behind developmentally or academically.

Daily challenges that might arise include dealing with the **psychological issues** of living with a chronic disease and adjusting to modified routines related to medication intake, diet and restrictions on activity level. There may be other issues as a child gets older, which may include handling school and social situations, obtaining special child services, ensuring medication compliance, and securing health and life insurance.

Poor appetite and slow weight gain are common in children with cardiomyopathy. This is due to a child's inability to take in sufficient calories while the body tries to

compensate for the heart's increased workload. Symptoms such as rapid breathing, fatigue, respiratory infections and abnormal absorption of nutrients may also contribute to poor appetite. In this situation, a nutritionist may recommend smaller, more frequent feedings, a higher calorie diet or supplements to encourage weight gain. With metabolic based cardiomyopathies, a low fat diet, special formulas or supplements may be prescribed to help regulate body function. In addition, children taking certain types of medication may require higher levels of magnesium or potassium in their diet, whereas children with DCM and heart failure may need a low sodium diet to prevent fluid retention.

Typically, children with cardiomyopathy do not have any serious limitations that would prevent them from attending school. However, some children do have **physical activity restrictions**, and they are unable to participate in gym activities that involve vigorous exertion. For children at risk of sudden cardiac arrest, they may be advised to refrain from weight lifting, competitive team sports and strenuous activities that could put strain on the heart. Excessive sweating or exposure to extreme temperatures may also aggravate existing symptoms. Moderate exercises, walking and less intense recreational sports are acceptable as long as they do not lead to dehydration or exhaustion. Guidelines for physical activity restrictions should be set by your child's physician based on your child's type of cardiomyopathy, family medical

history, disease severity and medical management.

All school personal, including the principal, school counselor, nurse, classroom teachers, gym instructor and special education coordinator should be made aware of your child's diagnosis. Depending on your child's condition, **school modifications and accommodations** may be necessary. It is advisable to schedule a meeting at the beginning of the school year to discuss academic or social issues that might arise. Practical issues such as common symptoms, medication side effects, special dietary needs, precautions related to implantable devices, and exercise and sports restrictions should be discussed. The need for external defibrillators (AED) and CPR-trained personnel on school premises also should be covered.

Parents can work with the school staff to develop three essential plans: health plan, education plan and emergency plan. An **individualized education plan** (IEP) states what special educational services or modifications a child needs to function at his or her maximum potential in school. This plan may detail a place and time to take medication, request special test-taking or homework modifications, require adaptive physical education, and specify special arrangements to reduce fatigue during the school day. The health needs of your child are addressed by the **health plan**, which includes basic information about the disease, contact information, medication administration and possible side effects, physical activity restrictions, and emergency care. The **emergency care plan** provides details about how to

recognize and respond to a cardiac emergency. This may include information about your child's treatment, warning signs, appropriate interventions and emergency contact information. More information about these plans and how to best work with the school system can be found in CCF's *Ensuring a Good Learning Environment: A Cardiomyopathy Resource Kit for School Personnel and Parents*.

Aside from diet and school concerns, other decisions will need to be made — whether your child should wear a medical alert bracelet, whether to purchase an automatic external defibrillator (AED) for home and travel use, and whether to schedule annual flu shots to protect your child's heart from influenza. Your child's doctor should be consulted on whether antibiotics are needed before dental or surgical procedures and which over-the-counter decongestants or herbal remedies should be avoided.

Consideration should be given to how the disease is explained to your child, his or her siblings and others close to him or her. A parent can help siblings, friends and classmates understand and accept your child's condition by talking about the disease in simple terms using age-appropriate books and role-play. Child life professionals and child psychiatrists can help you address any further concerns that you or your child has.

How do I cope with a chronic disease?

As with any chronic and potentially life-threatening disease, the entire family is affected. The possibility of deterioration or premature death creates unique social and emotional issues for families. The anxiety of an uncertain future combined with the isolation of dealing with a rare disorder can make raising a child with cardiomyopathy stressful at times. Some families feel responsible for passing the disease on to their child while others mourn the lost dream of raising a healthy child. As a parent, it is natural to feel a mixture of intense emotions such as fear, anger, guilt, shame and sadness. Keeping a journal, maintaining open communication with family members and developing a **support network** — whether it is relatives, friends, a faith-based group or other cardiomyopathy families — will help you accept the challenges of living with the disease.



Knowledge



Research

Being knowledgeable about the disease can also alleviate feelings of anxiety and helplessness. **Educate yourself** and your family about pediatric cardiomyopathy and be prepared for a cardiac emergency should it occur. Some parents may become more protective of their child's safety, but it is important to try to maintain a "normal" lifestyle and allow your child to interact with other children and engage in routine play. As long as your child is being monitored regularly and receiving appropriate treatment, the likelihood of a sudden cardiac event is low.

As your child gets older, he or she may face different social and emotional issues related to the disease. While your child passes through these stages, you should try to manage your own feelings so that your fears and disappointments do not interfere with your child's coping process or outlook on life. With many children, restrictions in activities can make them feel different or left out. Therefore, it is important to introduce your child to new activities that are less physically demanding, yet are rewarding and enjoyable. Be positive about what your child can do rather than focus on what he or she cannot do. Work together to find **special-interest clubs and activities** that provide a sense of inclusion as well as self-assurance.

You can help your child adjust by educating him/her about the disease and about what to expect. Encourage your child to become involved in his or her own care, to recognize his or her physical limitations and to be aware of certain warning signs that require immediate medical attention. Your child should also feel comfortable talking about his or her condition with others. With this approach, your child will more likely be confident and positive about living with cardiomyopathy.

Where do I go for more information and support?

Cardiomyopathy is a complex disease, and it can be difficult to understand completely. Healthcare professionals, support groups and other resources

can provide you with medical information, practical advice and emotional support. The **Children's Cardiomyopathy Foundation (CCF)** (childrenscardiomyopathy.org) provides detailed information on pediatric cardiomyopathy as well as helpful resources. Additional resources include the **American Heart Association** (americanheart.org), the **National Organization for Rare Disease** (rarediseases.org) and the **National Heart, Lung and Blood Institute** (nhlbi.org). For more specific answers to your child's treatment or care, you should contact your child's pediatric cardiologist or nurse.

Talking to families with similar circumstances can also be comforting. Group support can be especially helpful at the beginning stages and on a long-term basis. The Children's Cardio-myopathy Foundation (CCF) can help put you in touch with **other affected families** who understand the issues that you are facing.

What research is underway?

Most research to date has been focused on identifying **the genes that cause cardiomyopathy** and on understanding how these genetic abnormalities contribute to the disease. Some studies are on **mouse or other animal models** to mimic the disease found in humans. Others are **genetic studies** of extended families with a strong history of the disease. While researchers continue to try to identify new genes that cause different forms of cardiomyopathy, they are also studying why the disease manifests and progresses so

differently among individuals with the same affected gene. CCF has established a **Pediatric Cardiomyopathy Repository** to collect DNA and tissue samples for scientists to accelerate research in these areas.

Molecular based research is necessary before any new therapies can be developed. This kind of research also helps in the development of comprehensive and affordable **genetic tests** specific to pediatric cardiomyopathy. These diagnostic blood tests, run in a clinical laboratory, are used to identify the disease in asymptomatic individuals, guide family-planning decisions, and aid at risk family members in making informed medical care decisions.

In addition to basic research, epidemiological and clinical research are also taking place. The National Institute of Health (NIH) funded **North America Pediatric Cardiomyopathy Registry (PCMR)** is tracking, recording and analyzing clinical data on the features and outcomes of children with various forms of cardiomyopathy. Multi-center studies focused on factors affecting disease progression and long-term outcome have also been initiated by the **Pediatric Heart Network** and **Pediatric Heart Transplant Study Group**.

Getting more families involved in research is important to advancing medical knowledge on pediatric cardiomyopathy. If your family is interested in participating in research, your child's pediatric cardiologist or geneticist can guide you in selecting the most appropriate studies.

What else should I keep in mind?

It is important to remember that cardiomyopathy in children is rare. Information on the disease is limited, and knowledge in the field is still evolving. When reviewing published medical literature on the disease, parents should keep in mind that there is a **wide spectrum of outcomes** and studies may be based on the most serious cases or on a small sample group.

With any rare condition such as pediatric cardiomyopathy, parents and caregivers need to take an active role in their child's disease management. Try to learn as much as possible about your child's disease so that you can make informed decisions. Prior to any appointment, prepare a list of questions to discuss all relevant concerns. Remember that no question is too simple or unimportant to ask. Also, work closely with your child's medical team to provide feedback that might assist in their evaluation and treatment.

When necessary, do not be afraid to seek a **second opinion** or to ask your physician to consult with a cardiomyopathy and heart failure center. Specialists at these medical centers will likely have more experience managing children with cardiomyopathy and be aware of the latest treatment developments. These specialty centers will also have expertise in arrhythmia, heart failure, cardiac assist devices and transplantation. Educating yourself about the disease, taking a **proactive approach** and consulting with the top specialists will ensure that your child gets the best possible medical care.

The information provided in this booklet is meant to assist families and caregivers by providing a broad overview of the disease. The material presented is not intended to be complete or to replace the advice given by a medical professional. Only a physician can offer guidelines on treatment options and lifestyle restrictions that are right for your child.



Add HCM, DCM or RCM
insert here.

Insert Physician Card

For answers to any additional questions you may have,
please contact your child's physician.

About the Children's Cardiomyopathy Foundation

The Children's Cardiomyopathy Foundation (CCF) is a national non-profit organization dedicated to finding causes and cures for pediatric cardiomyopathy. CCF started in 2002 with one family's determination to call attention to this chronic heart disease and to take action on the lack of medical progress and public awareness. Since then, CCF has grown into a global community of families, physicians and scientists focused on improving diagnosis, treatment and quality of life for children with cardiomyopathy.

CCF is involved in all aspects of the disease ranging from research and education to patient support, aware-

ness, and advocacy. More than \$1.6 million has been committed to research and treatment initiatives, and CCF has led the way in establishing the first biologic specimen repository and planning the first international scientific conference on pediatric cardiomyopathy.

CCF continues to be an invaluable lifeline to thousands of affected families worldwide providing information, resources and guidance. Currently CCF's educational materials are distributed to more than 70 hospitals in the U.S. and Canada. CCF's hope for the future is that more lives will be saved, and every child with cardiomyopathy will have a chance to live a full and active life.

**Register with CCF:
childrenscardiomyopathy.org**



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