Pediatric Cardiomyopathy

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Pediatric Cardiomyopathy

Cardiomyopathy refers to diseases of the heart muscle in which the heart becomes enlarged or the muscle tissue thickens and becomes stiff and, in some cases, is replaced by scar tissue. The condition can lead to heart failure, arrhythmias or heart valve problems. According to the Pediatric Cardiomyopathy Registry, a project funded by the National Heart, Lung and Blood Institute, one in every 100,000 children in the United States under the age of 18 has been diagnosed with cardiomyopathy, although many more are likely at risk for developing the condition. The majority of these children are either younger than 12 months of age or between the ages of 12 to 18 years old.

Types of Cardiomyopathy

Cardiomyopathy may be acquired, inherited or idiopathic and is commonly divided into several categories based on phenotype or appearance:

- **Dilated cardiomyopathy (DCM)** is the most common type of cardiomyopathy in children. The underlying mechanism is loss of force transmission, caused by a wide variety of conditions affecting cardiomyocyte structure or function. Dilation is most commonly isolated to the left ventricle but may involve both the right and left heart. As the heart muscle dilates, the heart muscle is unable to contract normally and its ability to pump an adequate blood supply is inhibited. DCM can lead to heart failure, valve regurgitation, arrhythmias, intracardiac thrombus, and can be a cause of sudden cardiac arrest in young people. Approximately 1 in 2,500 people have been diagnosed with DCM, although more people probably have the disease and do not know it. The incidence of DCM in children is 0.58 cases per 100,000 children. Between 20 and 35 percent of people with DCM have at least one additional blood relative who also has either diagnosed or undiagnosed DCM.

- **Hypertrophic cardiomyopathy (HCM)** is a genetic derangement of contractile tissue leading to thickening of the myocardium. It may globally involve the left ventricle or can be isolated to a specific region of the left ventricular myocardium. Subaortic asymmetric septal hypertrophy may lead to obstruction of the left ventricular outflow tract. If this happens, the condition is called obstructive hypertrophic cardiomyopathy. HCM is also a common cause of arrhythmias and sudden cardiac arrest in young people, and may be the first symptom of an underlying cardiomyopathy. It affects an estimated 1 in 500 people, and the pediatric incidence is 0.47 cases per 100,000 children. These patients undergo risk stratification including genetic testing, magnetic resonance imaging, exercise stress testing, and family screening for history of sudden death.

- **Restrictive Cardiomyopathy (RCM)** is a disease in which the ventricles become stiff and diastolic function is impaired. As a result, this disease is marked by bi-atrial enlargement due to diastolic dysfunction. RCM may be associated with heart failure symptoms, arrhythmias, syncope, and pulmonary hypertension. Medical interventions for this disease are limited, and it is associated with a high mortality without heart transplantation. Pediatric incidence is 0.03 to 0.04 cases per 100,000 children.

- **Left ventricular non-compaction (LVNC)** is an emerging type of cardiomyopathy in children. This cardiomyopathy may be an isolated entity, but it has also been associated with some genetic syndromes (e.g. Barth syndrome) and certain subsets of congenital heart disease. The physiology can range from a benign form with no impact on left ventricular function to a severe form of cardiomyopathy. Currently, the incidence of LVNC is unknown in children and adults, but one large adult study noted 0.05 percent of echocardiograms had isolated LVNC.

Genetics and Cardiomyopathy

Even though the majority of cases of cardiomyopathy are idiopathic, some children may develop cardiomyopathy following myocarditis or in relation to a genetic etiology. Genetic causes have been identified for all types of cardiomyopathy including dilated, hypertrophic, restrictive and left ventricular non-compaction. Scientists have identified more than 80 genes that, when mutated, may cause DCM. To date, more than 1,000 variants have been identified in more than 30 genes causative of HCM, most of which affect the sarcomere. Many other genes are also involved in the onset of disease and the search for those genes is ongoing. It is important to note that individuals who are at risk for developing cardiomyopathy may be asymptomatic and have normal EKG and echo testing for many years. These individuals require life-long monitoring.

Genetic Testing for Patients with Cardiomyopathy and Their Families

Genetic testing is available for many of the different genes associated with cardiomyopathy. The underlying genetic cause can be identified in more than 80 percent of individuals with HCM and 50 percent of people with DCM. While genetic testing typically does not alter treatment of a patient with cardiomyopathy, given the fact that many cases are hereditary, genetic screening can identify other immediate family members who are at risk but have no signs, symptoms or abnormalities on echo or EKG. This is especially important as cardiomyopathy can often go undetected until serious damage to the heart muscle has occurred. Identifying cardiomyopathy in the families of patients diagnosed with the condition can allow physicians to more closely monitor the condition and potentially prevent irreparable injury to the heart.

How to Refer Patients and Families to the Cardiomyopathy Program at Nationwide Children's Hospital

The Cardiomyopathy Program in The Heart Center at Nationwide Children's offers genetic testing and treatment for patients with cardiomyopathy as well as provides screening services to those with a family history of cardiomyopathy. Services include clinical assessment, cardiac imaging, EKG, genetic counseling and genetic testing.

A family history, particularly of first-degree relatives, is critical in determining family members at risk for cardiomyopathy. The following patient family history will provide valuable information to physicians and in the referral process to Nationwide Children's.

Meet our Team

Lauren Fisher, RN, BSN
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Christina Phelps, MD
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Patient Family History

As cardiomyopathy can affect many people in a family, please provide the following information on your child and his or her biological relatives.

	First Name	Age	Heart Conditions	Age when diagnosed
Patient:				
Patient's siblings:				
Patient's Mother:				
Mother's siblings:				
Mother's mother:				
Mother's father:				
Patient's Father:				
Father's siblings:				
Father's mother:				
Father's father:				
Other affected family members:				

Referrals and Consultations

Online: NationwideChildrens.org/Heart

Phone: (614) 722-6200 or (877) 722-6220 | Fax: (614) 722-4000

Physician Direct Connect Line for 24-hour urgent physician consultations: (614) 355-0221 or (877) 355-0221.