



What is left ventricular non-compaction cardiomyopathy?

Left ventricular non-compaction cardiomyopathy (LVNC) is a newly recognized form of cardiomyopathy still in the early phases of medical understanding. Also known as “isolated non-compaction of the left ventricular myocardium” or “spongiform cardiomyopathy,” it is primarily caused by the abnormal embryonic development of the heart muscle.

In LVNC, the lower left chamber of the heart contains bundles or pieces of muscle that extend into the chamber called trabeculations. Typically, trabeculations are located at the bottom tip or apex of the heart, but they can be seen anywhere in the left ventricle.

There are different subgroups of LVNC based on heart function, thickness of the ventricles, heart chamber sizes and presence of abnormal heart rhythms (arrhythmias). LVNC can occur in the presence of normal heart function or in association with heart muscle dysfunction where the heart does not squeeze normally. This can lead to arrhythmias and heart failure.

When LVNC occurs with normal left ventricular thickness, size and function, it is called isolated LVNC. LVNC can occur in combination with abnormalities in heart squeeze (dilated form of LVNC) or in heart relaxation (hypertrophic or restrictive form of LVNC). The course of the disease and outcomes will vary based upon the LVNC subtype and prescribed treatment.

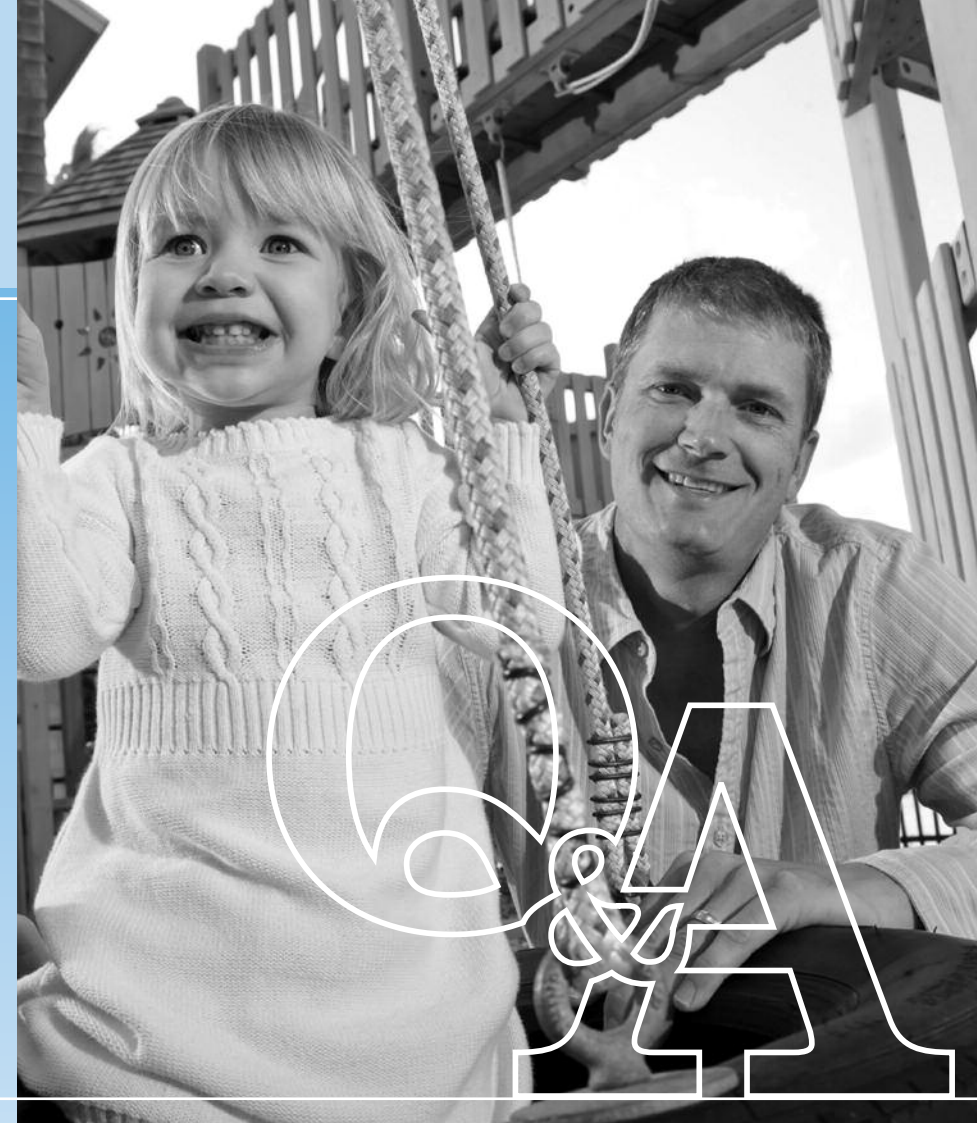
What is the prognosis?

Prognosis, or the likely course of a disease, in LVNC depends on many factors, such as a patient’s age, existing conditions or health problems, the subtype of LVNC, and the presence or absence of arrhythmias. Recent studies suggest that outcomes may not be as severe as was thought in the past, but more research needs to be done before any conclusions can be made.

In cases of more severe LVNC, prognosis may be similar to other types of cardiomyopathy advancing to heart failure. Increased awareness, early detection, careful surveillance and appropriate treatment by a cardiologist familiar with LVNC can greatly improve a child’s outcome.

The Children’s Cardiomyopathy Foundation (CCF) is a national non-profit organization dedicated to finding causes and cures for pediatric cardiomyopathy through the support of research, education, and increased awareness and advocacy.

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Education



Children’s Cardiomyopathy Foundation
P.O. Box 547 | Tenafly, NJ 07670
Tel: 866.808.CURE | Fax: 201.227.7016
Email: info@childrenscardiomyopathy.org

Understanding Left Ventricular Non-Compaction Cardiomyopathy



How many children are affected?

LVNC is a rare condition that affects less than 0.3 percent of the population. According to the Centers for Disease Control and Prevention, LVNC is estimated to affect about **1.2 per million children** between 0-10 years of age. However, it is likely that many cases are overlooked and mistaken for dilated or hypertrophic cardiomyopathy. As new imaging techniques with higher resolution are being utilized, trabeculations in the heart are being detected more often, resulting in more children and adults being diagnosed with LVNC.

What causes LVNC?

LVNC is caused by problems during the development of the heart in the embryo. In early fetal life, the inside of the heart contains pieces of muscles or trabeculations that extend into the heart chamber. During normal heart development this sponge-like network of muscle fibers becomes compacted, transforming the trabeculations from sponge-like to smooth and solid. LVNC occurs when the compaction process does not occur and the inside of the heart muscle remains trabeculated and spongy.

LVNC is considered a **genetic condition** that can be inherited (familial origin)

or occur spontaneously for unknown reasons (sporadic origin). Approximately 40 percent of individuals diagnosed with isolated LVNC have a family history of cardiomyopathy, which can include LVNC, dilated, hypertrophic or restrictive cardiomyopathy. LVNC is typically inherited in an **autosomal dominant pattern** in which a parent with a disease-causing mutation has a 50 percent chance of passing the mutation to each child. Although less common, LVNC can be inherited in an X-linked manner in which the mother passes the disease-causing mutation.

Once diagnosed with LVNC, there is a 20 to 40 percent chance that a genetic cause of the disease will be identified. Research has shown that mutations of the same genes known to cause dilated, hypertrophic and restrictive cardiomyopathy may be found in LVNC individuals, which suggests some overlap among these conditions.

The majority of cases are caused by **mutations in genes** coding for sarcomeric proteins that are responsible for proper cardiac muscle contraction. There are other genetic mutations responsible for LVNC associated with genetic syndromes, metabolic or mitochondrial disorders, and neuromuscular diseases. There are not any known acquired causes of LVNC.



Information

What are the common symptoms?

Symptoms can vary considerably and is determined by how the heart function is affected. Based on the subtype of LVNC that is diagnosed, symptoms usually overlap with those associated with dilated, hypertrophic or restrictive cardiomyopathy. Children who have the structural features of LVNC but normal heart function may have no symptoms.

Those with a more severe form of LVNC may present with symptoms of **heart failure or abnormal heart rhythm** (arrhythmia). This may include shortness of breath (dyspnea), fatigue, unexplained weight gain or swelling (edema), dizziness or light-headedness (presyncope), fainting or passing out (syncope), abnormal heartbeat (palpitations), and limited physical capacity or exercise intolerance. Infants may experience excessive sweating during activity, difficulty feeding and poor growth.

There may be other complications related to LVNC, such as an increased risk of **blood clots** (thrombosis), fast heart rhythms and **sudden cardiac arrest**. Although the risk for sudden cardiac arrest is low, it is best to be closely monitored by a cardiologist familiar with LVNC.

How do you diagnose LVNC?

The diagnosis of LVNC is based on cardiac testing and physical exam as well as family and medical history. During **cardiac testing**, an echocardiogram is used to diagnose LVNC. It can reveal trabeculations and deep recesses within the heart wall, as well as measure the overall heart function, size of the trabeculations and thickness of the compacted heart muscle. Multiple subgroups of LVNC exist and symptoms may overlap with other cardiomyopathy forms. In some cases, the heart may unexpectedly change its appearance from one form to another form over time. This is referred to as an "undulating phenotype" that is usually related to LVNC. A **physical exam** also will be done to look for evidence of heart failure and skeletal muscle weakness, which has been linked to certain forms of LVNC.

The evaluation will include a **medical history** taken of the child to look for any indication of heart failure. Signs would include feeding and growth issues, heart rhythm problems and exercise intolerance. A detailed **family history** is done to determine if other family members have a history of cardiomyopathy, heart rhythm problems, sudden cardiac arrest or unexplained death, and any other heart disease or surgery.

All first-degree relatives (parents, siblings and children) of an individual who has LVNC should undergo routine screening. Because LVNC can vary in presentation, it is possible to have LVNC without symptoms. A cardiologist and geneticist experienced in managing patients with cardiomyopathy can advise on the frequency of screening and the need for further genetic testing.

What are the treatment options?

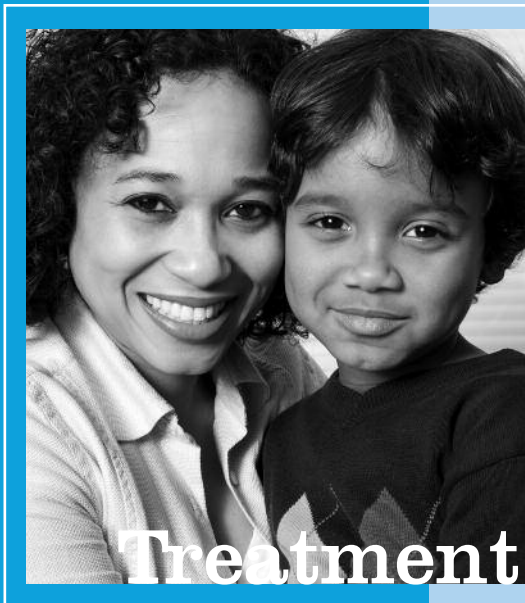
The goal of LVNC treatment is to improve heart function and prevent symptoms. The management of LVNC is influenced by its subgroup and the resulting symptoms and diagnostic evaluation. Factors such as heart function, ventricle thickness, heart chamber size and presence of arrhythmia will impact the treatment plan.

Because there are different forms of LVNC, treatment will address either a thick heart muscle (hypertrophic cardiomyopathy) or a poorly squeezing heart muscle (dilated cardiomyopathy). **Medication** is often used to treat symptoms, reverse heart damage and prevent ongoing damage to the heart muscle. In patients with heart failure, anti-congestive therapy similar to that used in patients with dilated cardiomyopathy may be recommended. This would include ACE inhibitors such as captopril and enalapril, as well as beta-blockers such as metoprolol or carvedilol. Diuretics may also be needed.

In patients with symptoms more consistent with hypertrophic cardiomyopathy, beta-blocker therapy with propranolol or atenolol may be recommended. In LVNC patients with mitochondrial or metabolic causes, a "vitamin cocktail" might be added that includes coenzyme Q10, carnitine, riboflavin and thiamine used alone or in combination.

If heart function is low, there is a greater chance for a blood clot to form in between the trabeculations and lead to stroke or other organ damage. A blood thinning medication such as aspirin may be recommended to prevent blood clots.

Lifestyle changes like limiting physical activity and sports may be necessary for some children. For those with an increased risk for sudden cardiac arrest, an **implantable cardioverter-defibrillator (ICD)** may be placed, or in some patients, a **cardiac resynchronization therapy (CRT)** heart device may be recommended. This involves implanting a combination pacemaker and ICD to coordinate the heart's pumping action and improve blood flow to the body. If heart function continues to weaken in spite of treatment, a mechanical pump or ventricular assist device may need to be surgically inserted to help the heart pump and supply blood to the body. In advanced cases, a heart transplant may be necessary.



Treatment