Left Ventricular Noncompaction

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Left ventricular noncompaction is a heart (cardiac) muscle disorder that occurs when the lower left chamber of the heart (left ventricle), which helps the heart pump blood, does not develop correctly.

Keywords: genetic conditions

1. Introduction

Instead of the muscle being smooth and firm, the cardiac muscle in the left ventricle is thick and appears spongy. The abnormal cardiac muscle is weak and has an impaired ability to pump blood because it either cannot completely contract or it cannot completely relax. For the heart to pump blood normally, cardiac muscle must contract and relax fully.

Some individuals with left ventricular noncompaction experience no symptoms at all; others have heart problems that can include sudden cardiac death. Additional signs and symptoms include abnormal blood clots, irregular heart rhythm (arrhythmia), a sensation of fluttering or pounding in the chest (palpitations), extreme fatigue during exercise (exercise intolerance), shortness of breath (dyspnea), fainting (syncope), swelling of the legs (lymphedema), and trouble laying down flat. Some affected individuals have features of other heart defects. Left ventricular noncompaction can be diagnosed at any age, from birth to late adulthood. Approximately two-thirds of individuals with left ventricular noncompaction develop heart failure.

2. Frequency

Left ventricular noncompaction is estimated to affect 8 to 12 per 1 million individuals per year. However, the condition is likely more common than this estimate because individuals who do not have any related signs or symptoms may not come to medical attention.

3. Causes

Mutations in several genes have been found to cause left ventricular noncompaction. Mutations in the *MYH7* and *MYBPC3* genes have been estimated to cause up to 30 percent of cases; mutations in other genes are each responsible for a small percentage of cases. However, the cause of the condition is often unknown.

Most of the genes associated with left ventricular noncompaction, including *MYH7* and *MYBPC3*, provide instructions for making proteins that play a role in the function of structures within muscle fibers called sarcomeres, which are necessary for muscles to tense (contract). Regular contractions of cardiac muscle pump blood to the rest of the body. Other genes associated with left ventricular noncompaction are involved in certain signaling pathways, the formation and maintenance of the structural framework (cytoskeleton) of cardiac muscle cells, or regulation of the electrical signals that control the heartbeat.

It is unclear how genetic mutations cause left ventricular noncompaction. During normal development before birth, cardiac muscle gets condensed (compacted), becoming smooth and firm. Mutations in certain genes likely lead to changes in this process, resulting in a left ventricular cardiac muscle that is not compacted but is thick and spongy, leading to left ventricular noncompaction.

Left ventricular noncompaction can also be part of syndromes that affect multiple parts of the body. When left ventricular noncompaction is part of a syndrome, it is caused by mutations in the gene that cause that syndrome. Additionally, left ventricular noncompaction has been found to develop in a small percentage of women during their first pregnancy. In these women, the cause of the condition is unknown but may be influenced by the increased stress that pregnancy puts on the cardiac muscle. In some cases, the cardiac muscle returns to normal following pregnancy and in other cases, the affected women have the varied signs and symptoms of left ventricular noncompaction throughout their lives.

3.1. The genes associated with Left ventricular noncompaction

- HCN4
- LDB3
- LMNA
- MYBPC3
- MYH7
- SCN5A
- TAZ
- TNNT2

4. Inheritance

Left ventricular noncompaction can have different inheritance patterns.

In most cases, including when the condition is caused by mutations in the *MYH7* or *MYBPC3* gene, left ventricular noncompaction is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. In most cases, an affected person inherits the mutation from one affected parent. Other cases result from new mutations in the gene and occur in people with no history of the disorder in their family.

Other cases of left ventricular noncompaction are inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Left ventricular noncompaction can also be inherited in an X-linked recessive pattern. Some genes associated with this condition are located on the X chromosome, which is one of the two sex chromosomes. In males (who have only one X chromosome), one altered copy of the gene in each cell is sufficient to cause the condition. In females (who have two X chromosomes), a mutation would have to occur in both copies of the gene to cause the disorder. Because it is unlikely that females will have two altered copies of this gene, males are affected by X-linked recessive disorders much more frequently than females. A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

5. Other Names for This Condition

- fetal myocardium
- honeycomb myocardium
- hypertrabeculation syndrome
- isolated noncompaction of the ventricular myocardium
- left ventricular hypertrabeculation
- left ventricular myocardial noncompaction cardiomyopathy
- left ventricular non-compaction
- LVHT
- non-compaction of the left ventricular myocardium
- noncompaction cardiomyopathy
- spongy myocardium

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