MYBPC3 Gene

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myosin binding protein C, cardiac

genes

1. Introduction

The *MYBPC3* gene provides instructions for making cardiac myosin binding protein C (cardiac MyBP-C), which is found in heart (cardiac) muscle cells. In these cells, cardiac MyBP-C is associated with a structure called the sarcomere, which is the basic unit of muscle contraction. Sarcomeres are made up of thick and thin filaments. The overlapping thick and thin filaments attach to each other and release, which allows the filaments to move relative to one another so that muscles can contract. Regular contractions of cardiac muscle pump blood to the rest of the body.

In cardiac muscle sarcomeres, cardiac MyBP-C attaches to thick filaments and keeps them from being broken down prematurely. Cardiac MyBP-C has molecules called phosphate groups attached to it; when the phosphate groups are removed, cardiac MyBP-C is broken down, followed by the breakdown of proteins of the thick filament. Cardiac MyBP-C also regulates how fast muscles contract, although the mechanism is not fully understood.

2. Health Conditions Related to Genetic Changes

2.1. Familial hypertrophic cardiomyopathy

Mutations in the *MYBPC3* gene are a common cause of familial hypertrophic cardiomyopathy, accounting for up to 30 percent of all cases. This condition is characterized by thickening (hypertrophy) of the cardiac muscle. Although some people with familial hypertrophic cardiomyopathy have no obvious health effects, all affected individuals have an increased risk of heart failure and sudden death.

MYBPC3 gene mutations that cause familial hypertrophic cardiomyopathy lead to an abnormally short or otherwise altered cardiac MyBP-C protein. It is unknown how these changes cause hypertrophy of the heart muscle.

2.2. Left ventricular noncompaction

At least four mutations in the *MYBPC3* gene have been found to cause left ventricular noncompaction, which occurs when the lower left chamber of the heart (left ventricle) does not develop correctly. The heart muscle is

weakened and cannot pump blood efficiently. These cardiac abnormalities can result in a wide range of outcomes from a complete lack of symptoms to sudden cardiac death. Other signs and symptoms include an irregular heart rhythm (arrhythmia), shortness of breath (dyspnea), and heart failure.

It is unclear how *MYBPC3* gene mutations cause left ventricular noncompaction. During normal development before birth, cardiac muscle gets compacted, becoming smooth and firm. *MYBPC3* gene mutations likely lead to changes in this process, resulting in a left ventricular cardiac muscle that is not compacted but is thick and spongy. This abnormal cardiac muscle is weak and cannot contract effectively, causing the varied signs and symptoms of left ventricular noncompaction.

3. Other Names for This Gene

- C-protein, cardiac muscle isoform
- MYBP-C
- myosin-binding protein C, cardiac-type
- MYPC3_HUMAN

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