

LAMP2 Gene

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Lysosomal associated membrane protein 2

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1. Introduction

The *LAMP2* gene provides instructions for making a protein called lysosomal associated membrane protein-2 (LAMP-2), which, as its name suggests, is found in the membrane of cellular structures called lysosomes. Lysosomes are compartments in the cell that digest and recycle materials. The role the LAMP-2 protein plays in the lysosome is unclear. Some researchers think the LAMP-2 protein may help transport cellular materials or digestive enzymes into the lysosome. The transport of cellular materials into lysosomes requires the formation of cellular structures called autophagic vacuoles (or autophagosomes). Cellular material that will be degraded in a lysosome is first enclosed in an autophagic vacuole inside the cell. The autophagic vacuole attaches (fuses) to a lysosome to transfer the cellular material into the lysosome where it can be broken down. The LAMP-2 protein may be involved in the fusion between autophagic vacuoles and lysosomes.

Slightly different versions (isoforms) of the LAMP-2 protein are produced: LAMP-2A, LAMP-2B, and LAMP-2C. These isoforms are found in different tissues throughout the body. LAMP-2B is the main isoform found in the heart and the muscles used for movement (skeletal muscles).

2. Health Conditions Related to Genetic Changes

2.1. Danon Disease

There are many mutations in the *LAMP2* gene that can cause Danon disease. Danon disease is a condition characterized by weakening of the heart muscle (cardiomyopathy), weakening of skeletal muscles (myopathy), and intellectual disability. This condition affects men more severely than women.

The *LAMP2* gene mutations that cause Danon disease lead to the production of very little or no LAMP-2 protein. Most mutations affect all three isoforms of the LAMP-2 protein. However, a mutation that affects only the LAMP-2B isoform also causes Danon disease, suggesting that the condition is caused by defects in the LAMP-2B protein.

Some studies have shown that in cells without the LAMP-2 protein, fusion between autophagic vacuoles and lysosomes occurs more slowly, which may lead to the accumulation of autophagic vacuoles. People with Danon disease have an abnormally large number of autophagic vacuoles in their heart and skeletal muscle cells. It is possible that this accumulation leads to breakdown of the muscle cells, causing the muscle weakness seen in Danon disease.

Some people with *LAMP2* gene mutations develop hypertrophic cardiomyopathy without the other characteristic features of Danon disease. Hypertrophic cardiomyopathy is a thickening of the heart muscle that may make it harder for the heart to pump blood. In people with *LAMP2* gene mutations, the lower left chamber of the heart, called the left ventricle, is usually the affected region. People with hypertrophic cardiomyopathy caused by *LAMP2* mutations often have a particular abnormality of the electrical signals that control the heartbeat called cardiac preexcitation. It is unclear whether this is a separate condition or a milder form of Danon disease. Furthermore, it is unknown why some people with *LAMP2* mutations develop hypertrophic cardiomyopathy but not the other features of Danon disease.

3. Other Names for This Gene

- CD107 antigen-like family member B

- CD107b
- LAMP-2
- LAMP2_HUMAN
- LAMPB
- LGP110
- lysosomal-associated membrane protein 2
- lysosome-associated membrane glycoprotein 2
- lysosome-associated membrane protein 2

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