MTM1 Gene

Subjects: Genetics & Heredity
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myotubularin 1

genes

1. Introduction

The *MTM1* gene provides instructions for producing an enzyme called myotubularin. Myotubularin is thought to be involved in the development and maintenance of muscle cells. This enzyme acts as a phosphatase, which means that it removes clusters of oxygen and phosphorus atoms (phosphate groups) from other molecules. Myotubularin removes phosphate groups from two molecules called phosphatidylinositol 3-phosphate and phosphatidylinositol 3,5-biphosphate. These molecules are found within cell membranes and are likely involved in transporting molecules within cells.

2. Health Conditions Related to Genetic Changes

2.1. X-linked myotubular myopathy

More than 200 mutations in the *MTM1* gene have been found to cause X-linked myotubular myopathy. Some *MTM1* gene mutations change one of the protein building blocks (amino acids) in myotubularin, while other mutations result in an abnormally short, nonfunctional enzyme. The *MTM1* gene mutations that prevent the production of any functional myotubularin tend to result in a more severe disease. Individuals who are mildly affected tend to have an *MTM1* mutation that allows some functional myotubularin to be produced.

Mutations in the *MTM1* gene are thought to disrupt myotubularin's role in muscle cell development and maintenance, causing muscle weakness and other signs and symptoms of X-linked myotubular myopathy.

3. Other Names for This Gene

- CNM
- MTM1_HUMAN
- MTMX

- myotubularin
- XLMTM

References

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