

## MTM1 gene

myotubularin 1

### Normal Function

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.

### Health Conditions Related to Genetic Changes

#### 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.

### Other Names for This Gene

- CNM
- MTM1\_HUMAN
- MTMX
- myotubularin
- XLMTM

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of MTM1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4534\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4534[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28MTM1%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>)

### Catalog of Genes and Diseases from OMIM

- MYOTUBULARIN; MTM1 (<https://omim.org/entry/300415>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/4534>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=MTM1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=MTM1[gene]))

## References

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## **Genomic Location**

The *MTM1* gene is found on the X chromosome (<https://medlineplus.gov/genetics/chromosome/x/>).

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