

## X-linked myotubular myopathy

### Description

X-linked myotubular myopathy is a condition that primarily affects muscles used for movement (skeletal muscles) and occurs almost exclusively in males. People with this condition have muscle weakness (myopathy) and decreased muscle tone (hypotonia) that are usually evident at birth. When viewed under a microscope, the muscle fibers of affected individuals are typically small and underdeveloped.

The muscle problems in X-linked myotubular myopathy impair the development of motor skills such as sitting, standing, and walking. Affected infants may also have difficulties with feeding due to muscle weakness. Individuals with this condition often do not have the muscle strength to breathe regularly on their own and must be supported with a machine to help them get enough oxygen (mechanical ventilation). Some affected individuals need breathing assistance only periodically, typically during sleep, while others require it continuously. People with X-linked myotubular myopathy may also have weakness in the muscles that control eye movement (ophthalmoplegia), weakness in other muscles of the face, and absent reflexes (areflexia).

In X-linked myotubular myopathy, muscle weakness often disrupts normal bone development and can lead to fragile bones, an abnormal curvature of the spine (scoliosis), and joint deformities (contractures) of the hips and knees. People with X-linked myotubular myopathy may have a large head with a narrow and elongated face and a high, arched roof of the mouth (palate). They may also have recurrent ear and respiratory infections, seizures, or liver disease. Some affected individuals develop a serious liver condition called peliosis hepatitis, which can cause life-threatening bleeding (hemorrhage).

Because of their severe breathing problems, individuals with X-linked myotubular myopathy usually survive only into early childhood; however, some people with this condition have lived into adulthood.

X-linked myotubular myopathy is the most severe condition in a group of disorders called centronuclear myopathy. In centronuclear myopathy, the nucleus is found at the center of many rod-shaped muscle cells instead of at either end, where it is normally located.

## Frequency

The incidence of X-linked myotubular myopathy is estimated to be 1 in 50,000 newborn males worldwide.

## Causes

Mutations in the *MTM1* gene cause X-linked myotubular myopathy. 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.

*MTM1* gene mutations probably disrupt myotubularin's role in muscle cell development and maintenance, causing muscle weakness and other signs and symptoms of X-linked myotubular myopathy.

[Learn more about the gene associated with X-linked myotubular myopathy](#)

- MTM1

## Inheritance

X-linked myotubular myopathy is inherited in an X-linked recessive pattern. The gene associated with this condition is 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 must be present 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.

In X-linked myotubular myopathy, the affected male inherits one altered copy from his mother in 80 to 90 percent of cases. In the remaining 10 to 20 percent of cases, the disorder results from a new mutation in the gene that occurs during the formation of a parent's reproductive cells (eggs or sperm) or in early embryonic development. Females with one altered copy of the *MTM1* gene generally do not experience signs and symptoms of the disorder. In some cases, however, females who have one altered copy of the *MTM1* gene experience mild signs and symptoms of X-linked myotubular myopathy, including changes in the muscle fibers, muscle weakness, and breathing problems. Researchers are studying why some females are affected and others are not.

## Other Names for This Condition

- CNM
- MTMX
- X-linked centronuclear myopathy

- XLMTM
- XMTM

## **Additional Information & Resources**

### Genetic Testing Information

- Genetic Testing Registry: Severe X-linked myotubular myopathy (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0410203/>)

### Genetic and Rare Diseases Information Center

- X-linked centronuclear myopathy (<https://rarediseases.info.nih.gov/diseases/11925/index>)

### Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

### Clinical Trials

- ClinicalTrials.gov (<https://clinicaltrials.gov/search?cond=%22X-linked myotubular myopathy%22>)

### Catalog of Genes and Diseases from OMIM

- MYOPATHY, CENTRONUCLEAR, X-LINKED; CNMX (<https://omim.org/entry/310400>)

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28x-linked+myotubular+myopathy%5BTIAB%5D%29+OR+%28x-linked+centronuclear+myopathy%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>)

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