



## Myotonia congenita

Myotonia congenita is a disorder that affects muscles used for movement (skeletal muscles). Beginning in childhood, people with this condition experience bouts of sustained muscle tensing (myotonia) that prevent muscles from relaxing normally. Although myotonia can affect any skeletal muscles, including muscles of the face and tongue, it occurs most often in the legs. Myotonia causes muscle stiffness that can interfere with movement. In some people the stiffness is very mild, while in other cases it may be severe enough to interfere with walking, running, and other activities of daily life. These muscle problems are particularly noticeable during movement following a period of rest. Many affected individuals find that repeated movements can temporarily alleviate their muscle stiffness, a phenomenon known as the warm-up effect.

The two major types of myotonia congenita are known as Thomsen disease and Becker disease. These conditions are distinguished by the severity of their symptoms and their patterns of inheritance. Becker disease usually appears later in childhood than Thomsen disease and causes more severe muscle stiffness, particularly in males. People with Becker disease often experience temporary attacks of muscle weakness, particularly in the arms and hands, brought on by movement after periods of rest. They may also develop mild, permanent muscle weakness over time. This muscle weakness is not seen in people with Thomsen disease.

### Frequency

Myotonia congenita is estimated to affect 1 in 100,000 people worldwide. This condition is more common in northern Scandinavia, where it occurs in approximately 1 in 10,000 people.

### Causes

Mutations in the *CLCN1* gene cause myotonia congenita.

The *CLCN1* gene provides instructions for making a protein that is critical for the normal function of skeletal muscle cells. For the body to move normally, skeletal muscles must tense (contract) and relax in a coordinated way. Muscle contraction and relaxation are controlled by the flow of charged atoms (ions) into and out of muscle cells. Specifically, the protein produced from the *CLCN1* gene forms a channel that controls the flow of negatively charged chlorine atoms (chloride ions) into these cells. The main function of this channel is to stabilize the cells' electrical charge, which prevents muscles from contracting abnormally.

Mutations in the *CLCN1* gene alter the usual structure or function of chloride channels. The altered channels cannot properly regulate ion flow, reducing the movement of

chloride ions into skeletal muscle cells. This disruption in chloride ion flow triggers prolonged muscle contractions, which are the hallmark of myotonia.

## **Inheritance Pattern**

The two forms of myotonia congenita have different patterns of inheritance. Thomsen disease is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. In most cases, an affected person has one parent with the condition.

Becker disease is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. Most often, the parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but do not show signs and symptoms of the condition.

Because several *CLCN1* mutations can cause either Becker disease or Thomsen disease, doctors usually rely on characteristic signs and symptoms to distinguish the two forms of myotonia congenita.

## **Other Names for This Condition**

- Congenital myotonia

## **Diagnosis & Management**

### Genetic Testing Information

- What is genetic testing?  
[/primer/testing/genetictesting](#)
- Genetic Testing Registry: Congenital myotonia, autosomal dominant form  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2936781/>
- Genetic Testing Registry: Congenital myotonia, autosomal recessive form  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0751360/>
- Genetic Testing Registry: Myotonia congenita  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0027127/>

### Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?cond=%22myotonia+congenita%22>

### Other Diagnosis and Management Resources

- GeneReview: Myotonia Congenita  
<https://www.ncbi.nlm.nih.gov/books/NBK1355>
- MedlinePlus Encyclopedia: Myotonia congenita  
<https://medlineplus.gov/ency/article/001424.htm>

## **Additional Information & Resources**

### Health Information from MedlinePlus

- Encyclopedia: Myotonia congenita  
<https://medlineplus.gov/ency/article/001424.htm>
- Health Topic: Muscle Disorders  
<https://medlineplus.gov/muscledisorders.html>

### Genetic and Rare Diseases Information Center

- Myotonia congenita  
<https://rarediseases.info.nih.gov/diseases/12301/myotonia-congenita>

### Additional NIH Resources

- National Institute of Neurological Disorders and Stroke  
<https://www.ninds.nih.gov/Disorders/All-Disorders/Myotonia-Information-Page>

### Educational Resources

- MalaCards: myotonia congenita  
[https://www.malacards.org/card/myotonia\\_congenita](https://www.malacards.org/card/myotonia_congenita)
- Merck Manual Consumer Version: Congenital Myopathies  
<https://www.merckmanuals.com/home/children-s-health-issues/muscular-dystrophies-and-related-disorders/congenital-myopathies>
- Orphanet: Thomsen and Becker disease  
[https://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=614](https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=614)

### Patient Support and Advocacy Resources

- Muscular Dystrophy Association  
<https://www.mda.org/disease/endocrine-myopathies>
- National Organization for Rare Disorders (NORD)  
<https://rarediseases.org/rare-diseases/myotonia-congenita/>
- Resource list from the University of Kansas Medical Center  
<http://www.kumc.edu/gec/support/muscular.html>

### Clinical Information from GeneReviews

- Myotonia Congenita  
<https://www.ncbi.nlm.nih.gov/books/NBK1355>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Myotonia+Congenita%5BMAJR%5D%29+AND+%28%28myotonia+congenita%5BTIAB%5D%29+OR+%28Becker's+myotonia%5BTIAB%5D%29+OR+%28Thomsen's+disease%5BTIAB%5D%29+OR+%28Thomsen+disease%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### Catalog of Genes and Diseases from OMIM

- MYOTONIA CONGENITA, AUTOSOMAL DOMINANT  
<http://omim.org/entry/160800>
- MYOTONIA CONGENITA, AUTOSOMAL RECESSIVE  
<http://omim.org/entry/255700>

### Medical Genetics Database from MedGen

- Congenital myotonia, autosomal dominant form  
<https://www.ncbi.nlm.nih.gov/medgen/422446>
- Congenital myotonia, autosomal recessive form  
<https://www.ncbi.nlm.nih.gov/medgen/155852>
- Myotonia congenita  
<https://www.ncbi.nlm.nih.gov/medgen/10158>

### **Sources for This Summary**

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*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/8857733>
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