CONDITION

Myotonia congenita

Myotonia congenita is an inherited condition that makes skeletal muscles slow to relax after movement.

WHAT IT IS
Inherited muscle condition causing delayed relaxation after contraction

BODY SYSTEM
Skeletal muscles

MAIN SYMPTOMS
Muscle stiffness, delayed movement, temporary weakness, muscle enlargement

WHAT CAUSES IT
Inherited CLCN1 gene changes

HOW IT IS DIAGNOSED
Electromyography and genetic testing

MAIN TREATMENT
Activity planning, physical therapy, and anti-myotonia medicines

CAN IT BE CURED?
No - the inherited gene change remains

IS IT AN EMERGENCY?
Sometimes - if breathing or swallowing suddenly becomes difficult

LONG-TERM OUTLOOK
Usually normal lifespan with manageable, non-progressive symptoms

Overview

Myotonia congenita is an inherited condition affecting skeletal muscles, which are the muscles you control to move your body. It causes myotonia , meaning a muscle remains tight for longer than expected after it contracts.

For example, you may have trouble opening your hand after gripping an object. Your legs may feel stiff when you first stand up, but movement often becomes easier after repeating the action. This improvement is called the warm-up effect .

Myotonia congenita happens because muscle cells do not handle electrical signals normally. The problem involves chloride channels, which help muscles relax after they contract. It is caused by changes in a gene called CLCN1 .

The condition is sometimes divided into two main inherited forms. Thomsen disease is usually inherited in a dominant pattern, while Becker disease is usually inherited in a recessive pattern. These names describe inheritance and common clinical patterns, but symptoms can overlap.

Myotonia congenita is not a muscular dystrophy. It does not usually cause ongoing destruction of muscle tissue. Thinking, sensation, and life expectancy are generally unaffected. Symptoms can still interfere with walking, work, school, exercise, or daily tasks, especially when movement starts suddenly.

Symptoms

Symptoms often begin during infancy, childhood, or the teenage years. Some people are not diagnosed until adulthood, particularly when symptoms are mild.

Muscle stiffness is the main feature. It is often most noticeable after resting and may affect the legs, arms, hands, face, or jaw. A person may have trouble taking the first few steps after sitting, releasing a handshake, opening the eyes after closing them tightly, or starting to speak after a pause.

Common features include:

The enlarged appearance of muscles is called muscle hypertrophy . It results from the condition's effect on muscle activity and does not necessarily mean the muscles are stronger.

Temporary weakness is more common in recessively inherited myotonia congenita. It may appear when movement begins and improve within seconds or minutes. Fixed, steadily worsening weakness is less typical and may need further assessment.

Symptoms can be triggered by sudden movement, emotional surprise, cold weather, or a long period of rest. They may be more noticeable when getting out of a car, starting to climb stairs, or beginning physical activity without warming up.

Heart muscle and breathing muscles are not usually affected. Significant breathing trouble, fainting, or persistent swallowing difficulty should not automatically be assumed to be part of myotonia congenita.

- Stiffness when starting a movement
- Improvement after repeating the movement
- Worse stiffness in cold temperatures
- Difficulty releasing a grip
- Temporary weakness after rest

- Falls caused by sudden leg stiffness
- Muscles that look unusually large or well developed

Causes and risk factors

Myotonia congenita is caused by a change in the CLCN1 gene. This gene provides instructions for making chloride channels in skeletal muscle cells. When these channels do not work properly, muscle cells remain electrically active for too long. The muscle then takes longer to relax.

A person may inherit the condition in one of two patterns.

In autosomal dominant inheritance , one altered copy of the gene can cause the condition. A parent with the gene change has a chance of passing it to each child.

In autosomal recessive inheritance , both copies of the gene are altered. The parents are often carriers who do not have symptoms. If both parents carry a relevant CLCN1 change, each pregnancy can result in a child who is affected, a carrier, or neither.

Inheritance is not always straightforward. The same gene change may cause different levels of stiffness in different relatives. Some CLCN1 changes can act as dominant changes in one family and recessive changes in another.

The main risk factor is having a disease-causing CLCN1 change. Exercise habits, diet, stress, and parenting do not cause myotonia congenita.

Diagnosis

Diagnosis begins with your symptoms, when they started, what triggers them, and whether relatives have similar problems. A clinician may ask you to grip and release an object, close and open your eyes, or stand and walk after resting.

An examination may show delayed muscle relaxation. Tapping a muscle can sometimes produce a brief indentation or contraction called percussion myotonia .

An electromyogram , or EMG, records electrical activity in muscle through a small needle electrode. Myotonia produces a characteristic pattern of repeated electrical discharges. EMG supports the diagnosis but does not always identify which genetic muscle condition is responsible.

Genetic testing can find a disease-causing change in CLCN1 and confirm myotonia congenita. It can also clarify the likely inheritance pattern and help with testing relatives. Occasionally,

testing does not find a clear genetic cause even when the symptoms and EMG strongly suggest a chloride-channel disorder.

Other conditions can cause muscle stiffness or myotonia. These include sodium-channel myotonias, paramyotonia congenita, myotonic dystrophy, thyroid problems, and certain medicine effects. The pattern of symptoms and targeted tests help distinguish them.

A muscle biopsy, which removes a small piece of muscle for examination, is not usually needed when clinical, EMG, and genetic findings are clear.

Treatment and management

There is no treatment that removes the inherited gene change. Management aims to reduce stiffness, improve safe movement, and limit the effect on daily life.

Many people learn to use the warm-up effect. Repeating a movement gently before applying full force can help. A gradual warm-up before exercise, walking, or physically demanding work may reduce sudden stiffness. Regular activity can help maintain mobility, although it does not cure the condition.

Cold can make myotonia worse. Warm clothing, heated environments where practical, and extra time to warm up may help during cold weather. Avoiding all activity is not usually helpful and may reduce general fitness.

A physiotherapist or occupational therapist familiar with neuromuscular conditions can suggest safe ways to move, reduce falls, and adapt tasks at home, school, or work.

Medicine may be considered when stiffness causes substantial difficulty. Mexiletine, a medicine that changes electrical activity in muscle cells, has evidence for reducing myotonia in nondystrophic myotonic conditions. Other medicines may be considered in some circumstances.

These medicines are not suitable for everyone. The choice may require a review of heart health, other medicines, and possible side effects. Treatment is usually monitored by a clinician experienced in muscle channel disorders.

Tell the anesthesia team about myotonia congenita before any operation or procedure. Certain medicines, electrical stimulation, cold, and shivering can trigger severe muscle stiffness. The muscle relaxant succinylcholine is generally avoided because it can cause intense muscle contraction. This does not mean that anesthesia is impossible; it means the team should plan for the condition.

Complications

Falls and injuries can happen when leg muscles suddenly stiffen, especially after sitting or when responding quickly to a surprise. Stiffness may also affect driving, climbing stairs, or work involving rapid movements.

Jaw or throat stiffness may occasionally interfere with speaking, chewing, or swallowing. Persistent swallowing trouble is not a routine feature and should be assessed rather than assumed to be harmless.

Myotonia may complicate anesthesia or emergency treatment if the medical team does not know about the condition. Carrying medical information on a phone, card, or medical alert item can help.

The condition does not usually lead to progressive loss of muscle or heart disease. New fixed weakness, loss of muscle size, heart symptoms, or breathing problems may point to another issue and deserve medical review.

Medicines used to control myotonia can also cause side effects. Monitoring is based on the specific medicine and your health history.

Prevention

Myotonia congenita cannot be prevented through diet, exercise, or lifestyle changes because it is inherited.

Genetic counselling can help you understand how the condition may be passed through a family. If a specific CLCN1 change is known, testing may be available for relatives or during family planning. Options can include testing during pregnancy or testing embryos created through in vitro fertilisation.

These are personal choices. Genetic counselling provides information and support without assuming that testing or a particular reproductive option is right for everyone.

Practical management can help prevent some injuries. Warming up before sudden activity, taking care on slippery surfaces, and planning for cold conditions may reduce falls or stiffness-related accidents.

Living with it and what to expect

Myotonia congenita is lifelong, but it is usually not progressive in the way muscular dystrophies

are. Symptoms may become more noticeable during childhood and then remain fairly stable. Some people find that stiffness improves with age, while others continue to have regular symptoms.

Day-to-day severity can change. Cold, fatigue, stress, long periods of rest, and sudden movements may make stiffness more obvious. A difficult day does not necessarily mean the condition is getting worse.

Most people can take part in education, employment, family life, and physical activity. Some need extra time to begin movements, workplace adjustments, or protection from cold. Children may benefit from school staff understanding that stiffness after sitting is physical and improves with movement.

Regular follow-up may be useful when symptoms interfere with daily life, medicine needs monitoring, or new problems appear. People with mild, stable symptoms may need less frequent specialist review.

Family members can have different symptoms even when they carry the same gene change. One relative may have obvious stiffness, while another has few or no problems.

Life expectancy is generally normal. Myotonia congenita usually affects movement rather than the heart, intelligence, or sensation. However, the impact on quality of life can be meaningful, particularly when stiffness causes falls, embarrassment, pain, or limits work and social activities.

When to get medical help

Arrange a routine medical appointment if you have repeated muscle stiffness, trouble releasing your grip, falls after standing, or symptoms that improve with repeated movement. These features can have several causes and need proper assessment rather than self-diagnosis.

If you already have myotonia congenita, seek routine follow-up when:

Seek urgent medical help for new or rapidly worsening swallowing difficulty, repeated choking, severe weakness, or symptoms after starting a new medicine.

Call emergency services for severe breathing difficulty, inability to swallow saliva, loss of consciousness, or a serious injury from a fall. These are not usual everyday features of myotonia congenita and should be assessed promptly.

- Stiffness is increasingly limiting work, school, exercise, or self-care
- You are having falls or injuries

- Weakness becomes persistent rather than temporary
- Swallowing symptoms recur
- You are considering pregnancy or genetic testing
- You are planning surgery or anesthesia
- A medicine is not helping or may be causing side effects

Common questions

Is myotonia congenita a type of muscular dystrophy?

No. It is a muscle ion-channel condition, not a muscular dystrophy. Muscles are slow to relax, but they do not usually undergo the progressive breakdown seen in muscular dystrophies.

Does exercise help myotonia congenita?

Repeated movement often reduces stiffness for a time because of the warm-up effect. Gradual, regular activity may improve mobility and general fitness. Sudden intense movement after rest can be difficult, so warming up is important.

Does cold weather make it worse?

Cold commonly increases stiffness and may reduce the warm-up effect. Warm clothing, indoor warm-ups, and allowing extra time to start moving can help. The degree of cold sensitivity differs between people.

Will my child inherit myotonia congenita?

That depends on whether the CLCN1 change follows a dominant or recessive pattern and whether the other biological parent carries a relevant change. Genetic testing and counselling can clarify the chance for a particular family.

Can I have surgery or a general anesthetic?

Yes, but the anesthesia team needs to know about the condition in advance. They can avoid triggers and medicines that may cause severe muscle contraction and take steps to prevent shivering.

Can myotonia congenita go away?

The genetic cause remains for life. Symptoms may become milder, stay stable, or fluctuate over time. Warm-up strategies and medicine can reduce symptoms, but they do not remove the underlying gene change.

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