CONDITION

Charcot-marie-tooth disease

Learn how Charcot-Marie-Tooth disease affects peripheral nerves, including its symptoms, inheritance, diagnosis, treatment, complications, and follow-up care.

WHAT IT IS
Group of inherited peripheral nerve disorders

BODY SYSTEM
Nervous system

MAIN SYMPTOMS
Muscle weakness, foot drop, reduced sensation, poor balance

WHAT CAUSES IT
Inherited genetic changes

HOW IT IS DIAGNOSED
Nerve conduction studies, EMG, genetic testing

MAIN TREATMENT
Supportive rehabilitation and mobility aids

CAN IT BE CURED?
No, nerve damage cannot be reversed

IS IT AN EMERGENCY?
Sometimes, severe breathing difficulty or stroke signs

LONG-TERM OUTLOOK
Usually slow progression with many remaining independent

Overview

Charcot-Marie-Tooth disease, often shortened to CMT, is a group of inherited disorders that damage the peripheral nerves. These nerves carry messages between the brain and spinal cord and the muscles, skin, and other parts of the body.

CMT mainly affects nerves in the feet, lower legs, hands, and forearms. Over time, nerve damage can lead to muscle weakness, muscle loss, changes in foot shape, reduced sensation, and difficulty with balance or walking. Symptoms often begin in childhood or early adulthood, but they can appear in infancy or much later in life.

CMT is named after the three doctors who first described it. It is not related to tooth disease.

There are many genetic types of CMT. Some primarily damage myelin, the protective covering

around a nerve. Others damage the axon, which is the long part of the nerve that carries electrical signals. Both forms can interfere with communication between nerves and muscles.

CMT usually progresses slowly. Its effects vary greatly, even among relatives with the same genetic change. Many people remain active and independent, while others need braces, mobility aids, surgery, or help with certain daily tasks. CMT does not usually affect thinking or shorten life, although some uncommon genetic types can cause more complex medical problems.

Symptoms

CMT symptoms are usually gradual and affect both sides of the body. Problems often begin in the feet and lower legs because the longest nerves tend to be affected first. The hands may become involved later.

Early signs may include frequent ankle sprains, tripping, difficulty running, or trouble lifting the front of the foot. This lifting problem is called foot drop. A child may appear clumsy, walk on the toes, or have difficulty keeping up during physical activities.

Possible symptoms include:

Joint and bone changes may develop as weak muscles pull unevenly on the feet or spine. These changes can include stiff ankles, shortened tendons, scoliosis, or hip problems. Scoliosis is an abnormal side-to-side curve of the spine.

Less common features depend on the genetic subtype. They may include hearing loss, vocal cord weakness, vision problems, breathing muscle weakness, or difficulty swallowing. These symptoms are not typical of every form of CMT and may need evaluation for another cause.

- Weakness in the feet, ankles, and lower legs
- Foot drop or a high-stepping walk
- High arches, flat feet, curled toes, or hammertoes
- Loss of muscle bulk in the lower legs
- Reduced ability to feel touch, vibration, pain, or temperature
- Numbness, tingling, burning, or nerve pain
- Poor balance, especially in darkness or on uneven ground
- Hand weakness or difficulty with buttons, handwriting, jars, and small objects
- Muscle cramps, fatigue, tremor, or aching pain

- Reduced or absent reflexes, especially at the ankles

Causes and risk factors

CMT is caused by a disease-related change in a gene involved in the structure or function of peripheral nerves. Different genetic changes affect nerves in different ways, which is why CMT includes many distinct subtypes.

The condition may follow several inheritance patterns. In autosomal dominant CMT, a person can develop the condition after inheriting one altered gene copy from either parent. In autosomal recessive CMT, both copies of a gene are altered, usually with one inherited from each parent. X-linked CMT involves a gene on the X chromosome.

A person may also have a new genetic change that was not identified in either parent. This is called a de novo change. For that reason, having no known family history does not rule out CMT. Family history can also be unclear because some relatives have mild or unrecognized symptoms.

Doctors may group CMT according to nerve test findings and genetic cause. CMT1 generally describes forms that mainly affect myelin. CMT2 generally describes forms that primarily damage axons. Other categories include intermediate, X-linked, and recessive forms. Modern diagnosis increasingly uses the specific gene name because older category names do not capture the full range of disease.

The main risk factor is having a disease-causing genetic variant. CMT is not caused by diet, exercise, infection, or something a person did. It is not contagious.

Diagnosis

Diagnosis usually begins with a medical history, a family history, and a neurological examination. The clinician may check muscle strength, sensation, balance, walking pattern, reflexes, foot shape, and signs of muscle loss. Information about relatives with high arches, walking problems, unexplained weakness, or a neuropathy can be helpful.

Nerve conduction studies measure how quickly and strongly electrical signals travel through peripheral nerves. Slowed signals may suggest damage to myelin, while smaller signals may suggest axon loss. Electromyography, or EMG, uses a thin needle electrode to examine electrical activity in selected muscles. These tests can help identify the pattern and severity of nerve and muscle involvement.

Genetic testing may confirm the diagnosis and identify the subtype. Testing may look for extra

or missing copies of a gene, examine a panel of neuropathy-related genes, or use broader sequencing when earlier tests do not find a cause. Not every person with clinical features of CMT receives a definite genetic result because some disease-related variants are not yet known or are difficult to detect.

A genetic counselor can explain what a result means for the person and their relatives. Counseling is also useful before testing because results may reveal unexpected family information or a variant whose significance is uncertain.

Other conditions can cause peripheral neuropathy. Depending on the symptoms, a clinician may order blood tests or other studies to look for acquired causes, such as diabetes, vitamin deficiencies, thyroid disease, immune disorders, or medication effects. Sudden or rapidly progressive weakness is not the usual pattern of CMT and needs assessment for another cause.

Nerve biopsy is rarely needed now that nerve conduction studies and genetic testing are widely available. Imaging may be used to evaluate bone, joint, or spine problems rather than to diagnose CMT itself.

Treatment and management

There is currently no treatment that reverses the inherited nerve damage in most forms of CMT. Care focuses on maintaining movement, reducing pain, supporting independence, and preventing avoidable complications. Management may involve a neurologist, rehabilitation specialist, physical therapist, occupational therapist, orthotist, podiatrist, orthopedic surgeon, pain specialist, or genetic counselor.

Physical therapy can help preserve flexibility, balance, and safe movement. A therapist may recommend stretching for tight calf muscles and joints, along with appropriate strengthening and aerobic activity. Swimming, cycling, walking, and other low-impact activities may be suitable for some people. Exercise plans should account for weakness, joint alignment, pain, and fatigue rather than pushing through major loss of function.

An ankle-foot orthosis, or AFO, is a brace that supports the ankle and helps control foot drop. Shoe inserts, custom footwear, or other braces may improve alignment and reduce pressure on painful areas. Canes, walking poles, walkers, or wheelchairs can support safety and independence when needed.

Occupational therapy can help with hand weakness and daily activities. Adaptive pens, button hooks, jar openers, modified keyboards, and changes to a home or workplace may reduce strain. Energy-conservation techniques can help a person complete important tasks without

unnecessary fatigue.

Pain treatment depends on whether pain comes from nerves, joints, muscles, or poorly fitting footwear. Options may include rehabilitation, footwear changes, psychological approaches to chronic pain, and medicines selected by a clinician. Treatment should be individualized because medicines that help nerve pain differ from those used for joint or muscle pain.

Foot care is important when sensation is reduced. Feet should be checked regularly for blisters, cuts, redness, or pressure areas. Properly fitting shoes and professional care for difficult calluses or nail problems can reduce the chance of skin injury.

Surgery may be considered for severe or painful foot deformity, unstable ankles, tight tendons, or loss of function that does not improve with nonsurgical care. The procedure and timing depend on bone alignment, muscle balance, flexibility, age, and the person's goals.

Some medicines can worsen peripheral nerve injury. Vincristine, a chemotherapy medicine, is a particular concern in people with CMT. Patients should tell every prescribing clinician about their neuropathy before starting a new medicine. They should not stop prescribed treatment without discussing the risks and alternatives with the medical team.

Complications

Weakness, reduced sensation, and foot deformity can increase the risk of falls. Falls may lead to sprains, fractures, or reduced confidence with walking. Bracing, therapy, home safety changes, and an appropriate mobility aid can lower this risk.

Uneven pressure on the feet may cause calluses, blisters, wounds, or infection, especially when pain sensation is reduced. A wound that is not healing, becomes increasingly red, drains fluid, or is accompanied by fever needs prompt medical attention.

Muscle imbalance can gradually lead to joint stiffness, tendon shortening, arthritis, or fixed foot deformities. Hand weakness may make work, school, cooking, dressing, and personal care more difficult. Chronic pain and fatigue can also affect sleep, mood, relationships, and participation in valued activities.

Breathing or swallowing complications are uncommon in typical CMT but can occur in certain subtypes. New shortness of breath when lying down, repeated choking, a weak cough, or unexplained morning headaches should be discussed promptly with a clinician.

Follow up

Routine follow-up is used to track strength, sensation, balance, walking, pain, foot health, and the ability to complete daily activities. The frequency depends on age, subtype, symptoms, and the rate of change. Growing children may need repeated checks because braces, shoes, joints, and spinal alignment can change as they develop.

A follow-up visit is appropriate if there is a gradual increase in falls, foot pain, hand difficulty, fatigue, numbness, or trouble using a brace. New wounds, poorly fitting footwear, or changes in work and mobility needs should also be discussed.

Seek prompt medical assessment for weakness that develops suddenly or progresses much faster than usual. New one-sided weakness, facial drooping, severe dizziness, or difficulty speaking may indicate a condition other than CMT.

Call emergency services for severe breathing difficulty, blue or gray lips, inability to swallow saliva, loss of consciousness, or signs of stroke. These are not routine features of CMT and require immediate care.

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