

What is...

Charcot-Marie-Tooth Disease

Charcot-Marie-Tooth disease (CMT) is one of the most common **inherited neurological disorders**.

It is classified as a **peripheral neuropathy**, which means it affects the peripheral nerves (nerves that lie outside the brain and spinal cord).

Damage caused by CMT renders peripheral nerves **unable to activate muscles or relay sensory information** from the limbs back to the spinal cord and brain.

CMT is estimated to affect approximately **1 in 5,000 people** in the United States.

There are many different types of CMT disease, which may share some symptoms but vary by pattern of inheritance and age of onset. These include **CMT1, CMT2, CMT3, CMT4, and CMTX**.

Mutations (defects) in many different genes cause different forms of CMT.

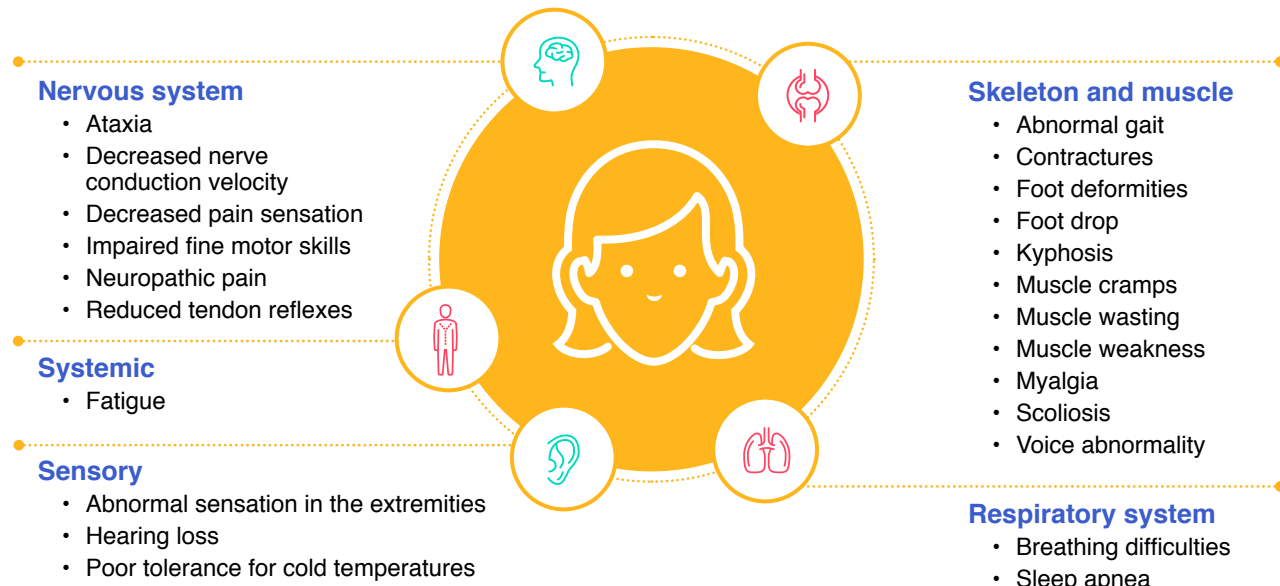
Most of the time, CMT is **inherited**. However, in some instances the disease may be **sporadic**, occurring in an individual who does not have a family history of the disease.

There currently is no cure for CMT, but physical therapy, occupational therapy, braces and other orthopedic devices, and even orthopedic surgery can help individuals manage and improve symptoms.



What are the symptoms of CMT?

Some of the most common symptoms of CMT include:



What should I know about CMT?

- 1 Onset of symptoms occurs most often in adolescence or early adulthood, but some individuals develop symptoms in mid-adulthood.
- 2 Severity of symptoms varies greatly among individuals and even among family members with the disease.
- 3 Progression of symptoms generally is gradual.
- 4 First signs include frequent tripping, ankle sprains, clumsiness, and "burning" or pins-and-needles sensations in the feet or hands.
- 5 Muscle wasting in the lower legs and feet may lead to foot drop, poor balance, and other gait problems. Foot deformities, such as high arches and hammertoes (a condition in which the middle joint of a toe bends upwards) are also characteristic due to weakness of the small muscles in the feet. In addition, the lower legs may take on an "inverted champagne bottle" appearance due to the loss of muscle bulk.
- 6 Muscular atrophy in the hands often causes people to have difficulty with tasks involving manual dexterity, such as writing and manipulating zippers and buttons.
- 7 As the disease progresses, weakness and decreased muscle mass may occur in the hands, arms, legs, or feet. People may lose the ability to feel heat, cold, and touch.
- 8 Chronic shortening of muscles or tendons around joints prevents the joints from moving freely, and muscle cramping is common.
- 9 Some people have pain that can range from mild to severe.
- 10 Although in rare cases, individuals may have respiratory muscle weakness, CMT is not considered a fatal disease and people with most forms of CMT have a normal life expectancy.
- 11 Some medications are potentially toxic to people with CMT. Before taking any medication or changing medications, individuals with CMT should always check with their physician.

How is CMT treated?

The goal of treatment in CMT is to help individuals cope with the disabling symptoms of the disease. Optimal treatment is multidisciplinary, with care provided by neurologists, genetic counselors, nurses, physical and occupational therapists, physiatrists, and orthopedic surgeons.

Pain-relieving medications can be prescribed for individuals who have severe pain.

Hearing aids can help with hearing loss.

Orthopedic surgery can fix foot and joint deformities.

Assisted ventilation may help those with breathing problems or sleep apnea.

Physical and occupational therapy for CMT is tailored to the individual and may involve muscle strength training to delay or reduce muscle atrophy, muscle and ligament stretching to prevent or reduce joint deformities that result from uneven muscle pull on bones, stamina training to increase endurance and help prevent fatigue, and moderate aerobic exercise to help maintain cardiovascular fitness and overall health.

Ankle braces can help prevent ankle sprains by providing support and stability during activities such as walking or climbing stairs. **High-top shoes or boots** can also provide support for weak ankles. **Thumb splints** can help with hand weakness and loss of fine motor skills.

Supportive aids such as braces, walkers, or wheelchairs may help those who experience problems with muscle weakness and fatigue to maintain everyday mobility and prevent injury.

Please talk to your medical provider to obtain more information on these treatments.



MDA Glossary

Ataxia

The inability to maintain balance and coordination

Contracture

A shortening of muscles or tendons around joints that can limit mobility

Foot drop

Difficulty in lifting the front part of the foot, so that the toes point downward during walking

Kyphosis

Abnormal front-to-back spine curvature

Mutation

A flaw in the DNA code

Myalgia

Muscle pain

Nerve conduction velocity

The speed at which electrical signals travel through the nerve

Peripheral neuropathy

A malfunction of the nerves that can lead to sensory impairment and muscle weakness

Scoliosis

An abnormal side-to-side curvature of the spine that occurs when weakened muscles are unable to hold the spine straight

Read more about CMT at mda.org. If you're looking for one-on-one support, you can also give our MDA Resource Center a call at 800-572-1717.



Designated a Top-Rated Charity by the American Institute of Philanthropy, MDA is the first nonprofit to receive a **Lifetime Achievement Award from the American Medical Association** for "significant and lasting contributions to the health and welfare of humanity."



Muscular Dystrophy Association

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