

### **Introduction**

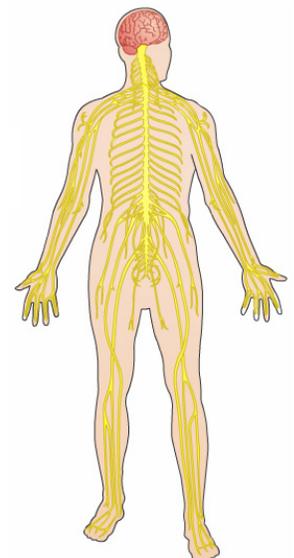
Charcot-Marie-Tooth disease, or CMT, is a group of genetic nerve disorders. It is named after the three doctors who first identified it. CMT affects the nerves that carry movement and sensation signals between the brain and spinal cord and the rest of the body. There is no cure, but many people with CMT lead active lives.

This reference summary will help you understand Charcot-Marie-Tooth disease, including the symptoms and causes. Also covered are diagnosis and treatment options.

### **The Peripheral Nerves**

Together, the brain and spinal cord are called the “central nervous system.” The nerves in the rest of the body are called the “peripheral nervous system.” The brain is the command center of the body. In addition to thinking and feeling, it receives information and sends orders to different parts of the body. Orders from the brain travel through the spinal cord. From the spinal cord, orders travel to the rest of the body through the peripheral nerves.

The main cells that make up the nervous system are called neurons. Each neuron has a body and an axon. Axons are long fibers that are similar to electrical wires. Neurons communicate with each other by sending electrical signals through the axons. A special material called myelin covers axons. Myelin improves the conduction of the electrical current and communication between neurons.



The Nervous System

Some neurons send messages about movement. These neurons are known as motor neurons. For example, if you want to raise your hand, your brain tells your arm and hand to do it. Other neurons also receive messages about sensation, or feeling. These neurons are known as sensory neurons. If you burn your hand while cooking, the nerves in the skin send pain signals to your brain.

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If nerves become damaged, they cannot send and receive messages properly. This can interfere with movement and sensation.

## Symptoms

CMT symptoms can be so mild you don't realize you have the disease. In other people, CMT may be more severe. CMT affects both motor and sensory nerves. This can cause muscle weakness, as well as decreased sensations in the affected limbs. Symptoms of CMT usually start around the teen years or early adulthood. The disease progresses slowly.

A common symptom of CMT is weakness of the foot and lower leg muscles. This can cause frequent tripping or falling and a high-stepped gait, or walk. CMT can also cause "footdrop." Footdrop is when a person has difficulty lifting the foot up by bending at the ankle.

Weakness in the small muscles of the feet is also common in people with CMT. This can cause high arches in the foot and hammer toe syndrome. Hammer toe syndrome is a condition in which the middle joint of a toe bends upwards. Sometimes it is simply called hammer toes. The lower legs may also change appearance due to CMT. This is due to the lower part of the legs losing muscle in people with the disease.



People with CMT often have decreased sensation in the legs and feet. The legs and feet may even become numb. Later in the disease, the muscles in the hands may become weak and lose sensation. This makes it difficult to use the fingers, hands, and wrists for everyday tasks such as writing. In severe cases, the thighs and arms may also be affected late in the disease.

## Causes

CMT is a group of genetic disorders that can be passed from parents to children. It is caused by defective genes. This section explains what genes are and how defective genes can cause CMT.

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Each person has 23 pairs of chromosomes. Genes are found on chromosomes. Genes tell the body to make certain substances. Every person has thousands of genes. Our genes make us look the way we do. They also have a lot to do with our health. There are many genes responsible for producing proteins that make peripheral nerves work correctly. These genes may produce substances for the axon or for the myelin around the axon. If one or more of these genes become defective, the person develops CMT.

CMT is caused by defective genes that are responsible for producing proteins that affect the peripheral nerves. There are several types of CMT. Some types of CMT need only one defective gene to cause the disorder. Other types of CMT need two defective genes. Often the gene mutations that cause CMT are inherited. This means that the defective gene can be passed from parent to child.

## Diagnosis

Your healthcare provider will ask about you and your family's medical history. He or she will also perform a physical exam. During the physical exam, your healthcare provider will look for signs of CMT, such as:

- Muscle weakness in the legs, feet, arms, or hands
- Physical changes in the feet, such as high arches or hammer toes
- Reduced feeling in the feet or hands
- Slow reflexes

Your healthcare provider may also use some tests to diagnose CMT, including:

- Nerve conduction studies
- Electromyography, or EMG
- Nerve biopsy
- Genetic testing



A nerve biopsy or genetic testing can confirm a diagnosis of CMT. Nerve conduction studies can show how well your nerves conduct electric currents. Electrodes are placed on the skin over a peripheral nerve. The electrodes stimulate the nerve with small electric shocks. If the responses to the shocks are slow or weak, it may indicate a nerve disorder like CMT.

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Electromyography, or EMG, also uses an electrode to measure electrical activity. However, for this test a needle electrode is inserted into the skin. As you tense and relax your muscles, the electrical activity of the muscles is recorded. An EMG can tell doctors which muscles are affected by CMT. To determine if your symptoms are caused by CMT or another nerve disorder, a nerve biopsy may be done. During a nerve biopsy, a small incision is made in the skin of the lower leg. Then, a small piece of peripheral nerve is taken and examined.

Genetic testing uses a sample of blood to look for defective genes that are known to cause CMT. It can also be used to help people with CMT know the risks of passing on the genes to their children.

## **Treatment**

There is no cure for CMT. However, there are many treatment options to manage the symptoms and help patients cope with the condition.

Physical therapy can help people with CMT strengthen their muscles with exercises and stretching. This can prevent further muscle loss.

Some people with CMT experience muscle weakness in the arms and hands, interfering with certain tasks. In these cases, occupational therapy may be needed. Occupational therapy teaches people how to perform everyday tasks such as writing or buttoning up a shirt.



Orthopedic devices, such as leg and ankle braces or splints, may also be used. These devices help people with CMT move without getting hurt. Thumb splints may also improve your ability to grip and hold objects. Pain medication can be prescribed for some people with CMT to treat muscle cramps or nerve damage. However, these conditions do not commonly cause pain in people with CMT.

Surgery may be done to fix severe foot problems that cause pain and make walking difficult. However, surgery cannot help improve muscle weakness in the foot or loss of sensation.

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You can also take steps to manage your symptoms at home by:

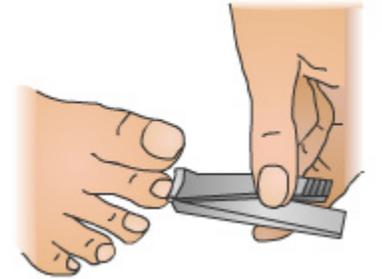
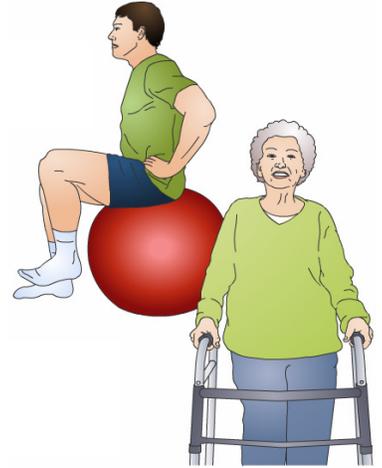
- Exercising regularly
- Stretching daily
- Using a walker or cane to improve your stability

If you have CMT, it is also important to care for your feet. CMT can cause a loss of sensation in the feet. You need to check your feet daily for any problems such as infections, wounds, or calluses.

If you do not check your feet daily, a foot sore may develop and you may not be able to feel any pain. If the sore grows, gangrene can develop and your foot may have to be amputated, or removed from the body.

You can prevent problems with your feet by:

- Cutting your nails regularly and straight across to avoid infections and ingrown toenails
- Soaking and moisturizing your feet with lotion
- Wearing shoes that fit properly



## Summary

Charcot-Marie-Tooth disease, or CMT, is a group of nerve disorders. It is named after the three doctors who first identified it. CMT affects the nerves that carry movement and sensation signals between the brain and spinal cord and the rest of the body.

CMT is caused by changes in the genetic material responsible for making proteins needed by nerves. These changes are known as gene mutations. The defective genes cause messages between the brain and limbs to be interrupted or slowed in people with CMT.

Several treatment options exist to manage the symptoms of CMT. They include: physical therapy, occupational therapy, and braces and other devices. Sometimes surgery can help patients. CMT can be inherited. It can also be mild or severe. There is no cure for CMT. However, with treatment many people with CMT can lead active lives.



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