



Charcot-Marie-Tooth Disease: The Basics

Charcot-Marie-Tooth disease (CMT), named after the three doctors who first identified it, is one of the most common inherited nerve disorders. CMT affects an estimated 1 in 2,500 people in the United States and 2.6 million people worldwide, although experts believe the number could be much higher.

Understanding what the disease is, what causes it, how doctors diagnose it, and the different treatment options available can help you or someone you know to manage the disease and lead an active and fulfilling life.

What is CMT?

CMT is also commonly called **hereditary sensory and motor neuropathy**. This means that the disease runs in families and causes problems with the sensory and motor nerves, the nerves that run from the arms and legs to the spinal cord and brain.

When the parts of the nerves—the axons and the myelin—become damaged, messages that run along the nerves move more slowly or have a weak signal. Over time, this causes muscles in the feet, legs, and hands to lose strength. Often, the muscle loss happens unevenly, which causes deformity as muscles waste away (atrophy) at different rates.

Researchers recognize five main types of CMT and many subtypes. Research is still under way to define all of the different types fully. CMT type 1 is the most common form of the disease.

What causes CMT?

Researchers know that faulty genes cause CMT disease. Although many of these genes have been identified, ongoing research continues to discover additional genes involved in CMT. The disease is usually passed on from parent to child, with a 50 percent chance of the child inheriting the condition. However, recent testing has found that some people have the faulty genes, but their parents do not.

What are the symptoms of CMT?

Symptoms differ from person to person and even between members of the same family. Here is a list of



some common symptoms of CMT you may experience.

Early symptoms

- ⌘ clumsiness
- ⌘ slight difficulty in walking because of trouble picking up the feet
- ⌘ weak leg muscles
- ⌘ fatigue

(continued)

Common symptoms

- ⚡ foot deformity (very high arched foot/feet)
- ⚡ difficulty lifting foot at the ankle (foot drop)
- ⚡ curled toes (known as hammer toes)
- ⚡ loss of lower leg muscle, which leads to skinny calves
- ⚡ numbness or burning sensation in the feet or hands
- ⚡ “slapping” when walking (feet hit the floor hard when walking)
- ⚡ weakness of the hips, legs, or feet
- ⚡ leg and hand cramps
- ⚡ loss of balance, tripping, and falling
- ⚡ difficulty grasping and holding objects and opening jars and bottles

Later symptoms

- ⚡ similar symptoms in the arms and hands
- ⚡ curvature of the spine (scoliosis)

Very rare symptoms

- ⚡ speech and swallowing difficulties
- ⚡ breathing difficulties, especially when lying flat
- ⚡ hearing loss
- ⚡ vocal cord paralysis

How is CMT diagnosed?

Although CMT is one of the more common hereditary neuropathies, its symptoms are similar to other types of neuropathy. In addition, not all doctors are familiar with CMT. Therefore, diagnosing CMT can be challenging. To help in the diagnosis, your doctor or neurologist may perform the following examinations and tests:

- ⚡ **Medical and family history.** Your doctor asks questions about your health and your family’s health.
- ⚡ **Physical examination.** Your doctor examines you to look for symptoms of CMT and other health problems.
- ⚡ **Nerve conduction velocity test.** Electrodes are placed on the skin over the nerves on your legs and arms to measure how quickly the nerves carry electrical signals. This test can be uncomfortable.
- ⚡ **Electromyography (EMG).** A needle electrode is inserted through the skin on your legs and arms to

measure the electrical signals received by your muscles. This test can be uncomfortable and, for some people, painful.

- ⚡ **Genetic test.** A blood test may tell whether you have CMT, although it may not provide all the answers.

What is the future outlook for people with CMT?

Most people with CMT learn how to manage their symptoms and go on to live fulfilling lives. Generally, the progression of the disease is slow. In many cases, the symptoms are mild, and properly fitted orthotics can help with difficulty in walking. In more severe cases, people with CMT must wear braces or use a wheelchair to get around. In rare cases where CMT causes difficulty with breathing, additional medical consultation is recommended to manage these symptoms..

How is CMT treated?

Although there is no cure for CMT, therapies can treat the symptoms. The development of a comprehensive treatment plan with a health care team can help people with CMT lead an active life. Treatment plans generally involve one or more of the following:

- ⚡ **Physical and occupational therapy.** Therapists work to improve muscle strength and stamina and to help people complete tasks of daily living.
- ⚡ **Braces and other orthopedic devices.** Custom shoes or shoe inserts (foot orthoses) help to improve walking ability. Leg braces prevent ankle sprains and improve walking ability.
- ⚡ **Surgery.** For some people, surgery can help prevent or reverse foot and joint deformities.
- ⚡ **Pain management.** Pain-killing drugs may be prescribed for people who have severe pain.
- ⚡ **Complementary therapies.** Some people with CMT have found that alternative therapies—such as acupuncture, biofeedback, capsaicin, alpha-lipoic acid, or meditation—can help relieve symptoms.