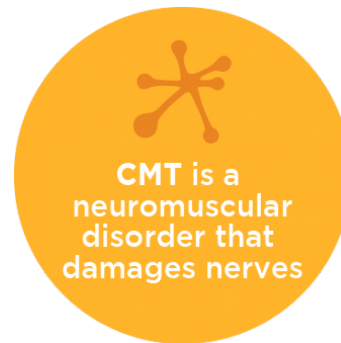


Charcot-Marie-Tooth Disease

What Is Charcot-Marie-Tooth Disease (CMT)?

Charcot-Marie-Tooth Disease (CMT) is a group of neuromuscular disorders that damage nerves in the legs and arms (also known as the peripheral nerves). CMT disease usually affects the nerves that control movement and sensation. It also affects cells that support and protect nerves. As a result, muscle tissue begins to waste and weaken.



What Causes Charcot-Marie-Tooth Disease?

Pronounced “Sharko Marie Tooth disease,” CMT is the most commonly inherited disorder that damages the nerves. It affects one in every 2,500 U.S. children. More than 30 genes are associated with the condition.

CMT Types

CMT disorder is caused by changes (mutations) in different genes. Based on which gene it affects, CMT usually gets categorized into the following types.

CMT1

This form progresses slowly and causes muscle weakness, decreased muscle mass (also known as atrophy) and loss of sensation. CMT1 typically develops in the first or second decade of life.

CMT1 is the most common form of CMT. Only one parent has to have the abnormal gene to pass the condition to a child.

CMT2

Like CMT1, CMT2 causes damage to the peripheral nerves. However, CMT2 is usually less severe. Charcot-Marie-Tooth Type 2 typically develops later in life than CMT1 does.

Congenital Hypomyelinating Neuropathy (CHN), also known as Type 3 CMT or Dejerin-Sottas Disease

This form of CMT progresses slowly and usually develops before age 3. It affects the nerve fibers' protective coating (also known as myelin). CHN is usually quite severe. It can cause profound disability, loss of sensation, scoliosis and mild hearing loss.

CMT4

This type is often more severe, and starts earlier in a child's life. It causes muscle weakness, decreased muscle mass (also known as atrophy) in the lower leg and/or hand and loss of sensation in the lower limbs. Many children who have CMT4 also have hammertoes (or other foot deformities), develop scoliosis or have difficulty walking.

CMTX

This form of CMT causes moderate to severe impairments to movement and sensation. It occurs more often in males than in females.

Charcot-Marie-Tooth Disease Symptoms and Effects

The symptoms of CMT can range from mild to severe. They usually begin in adolescence or early adulthood.

Some of the most common symptoms are:

- Muscle weakness and decreased muscle mass (also known as atrophy) in the feet and lower legs, and/or in the hands and arms as the disease progresses.
- High-stepped walking pattern.
- Frequent tripping or falling.
- Foot deformities, such as high arches or hammertoes.
- Mild hearing loss in more severe forms of CMT.

Charcot-Marie-Tooth Disease Treatment

It's very important for children who have CMT to get tested as soon as you or your pediatrician suspect CMT. Early

treatment can help children avoid some secondary effects of the condition.

CMT diagnoses usually require the following tests:

- **Physical exam** of muscle mass, reflexes, sensory abilities and foot deformities.
- **Nerve tests** that measure electrical signals produced by the nerves.
- **Tests that measure electrical activity** while muscles are relaxed and contracted (**electromyography**).
- **Lab tests** of a small piece of the nerve (nerve biopsy).
- **Genetic and DNA** tests of blood samples that look for the genetic abnormalities responsible for CMT.

The following are the most common methods of treatment for CMT:

Medication

Some people who have CMT experience pain caused by muscle cramps and nerve damage. Prescription medicines can help control pain.

Physical Therapy

Physical therapy can help prevent muscle loss and strengthen muscles. It can also help prevent further nerve damage and deterioration.

Occupational Therapy

CMT usually begins with muscle weakness in the feet and legs, but often moves later into the arms and hands. This progression can make writing, grasping or other tasks more difficult. **Occupational therapy** helps strengthen the muscles used for these tasks.

Orthopedic Devices

Orthopedic devices—such as special shoes, grips or braces—can provide stability and help make daily tasks easier.

Surgery

Although surgery can't help with loss of sensation or muscle weakness, it can:

- Help stabilize foot deformities.
- Reduce pain.
- Improve walking ability for people who have severe foot deformities caused by CMT.

Integrated Care

If your child has Charcot-Marie-Tooth disease, the Gillette Children's Specialty Healthcare team of experts will work

closely with you to create a customized treatment plan. Ours is one of the region's top neuromuscular programs. Here, your child might receive treatment from experts in several areas—we'll help you navigate the services you need:

Medical Specialties

- [Neurology](#).
- [Rehabilitation medicine](#).
- [Medical genetics and genetic counseling](#).
- [Cardiology](#).
- [Pulmonology and respiratory care](#).
- [Endocrinology](#).
- [Orthopedics](#).

Rehabilitation Therapies and Assistive Technology Services

- [Speech and language therapy](#).
- [Occupational therapy](#).
- [Physical therapy](#).
- [Orthotics](#).
- [Seating and mobility](#).
- [Assistive technology](#).

Family Support

- [Child life](#) specialists.
- [Social workers](#).
- [Therapeutic recreation](#) specialists.
- Dietitians.
- [Psychologists](#).
- [Neuropsychologists](#).

Throughout your child's treatment, our internationally recognized team of specialists will offer support, answer your questions and provide family-centered care.

Explore Charcot-Marie-Tooth Disease Resources

- Learn more about CMT and available research on this condition at the [Charcot-Marie-Tooth Association](#).

[Make An Appointment](#) [651-290-8707](#)

This information is for educational purposes only. It is not intended to replace the advice of your health care providers. If you have any questions, talk with your doctor or others on your health care team.

If you are a Gillette patient with urgent questions or concerns, please contact Telehealth Nursing at [651-229-3890](#).