

Charcot-Marie-Tooth Disease

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

CMT refers to a group of disorders that damage peripheral nerves. These nerves branch out from your spinal cord and go to your arms and legs.

There are two types of peripheral nerves:

- Motor nerves. These nerves carry signals from your brain to your muscles. These signals move the muscles in your arms and legs.
- Sensory nerves. These nerves carry signals from the skin and muscles to your brain. These signals let you feel heat, cold, touch and pain.

CMT causes nerves to break down (degenerate) and lose their ability to send signals.

- When CMT affects motor nerves, your muscles become weak. You have difficulty with movement, usually starting in the feet and lower legs. As the disease slowly progresses, it can affect movement in your lower thigh, hands and arms. Despite difficulties, most people with CMT remain able to walk, although some need help with foot or leg braces.
- When CMT affects sensory nerves you have difficulty with feeling (sensation), usually in your feet and hands. You may be less able to feel heat, cold, touch and pain.

CMT is usually inherited (passed down from one or both parents). There is no cure for CMT, but supportive treatment can help you manage the symptoms. Most forms of CMT do not shorten life expectancy.

CMT is also called Hereditary Motor and Sensory Neuropathy (HMSN)

Here's what these words mean:

Hereditary = Inherited. Passed down from parent to child in genes.

Motor = Affecting movement

Sensory = Affecting sensation (feeling)

Neuropathy = A problem with nerves

What are the symptoms of CMT?

The symptoms of CMT include:

- Muscles weakness in your feet, ankles, legs or hands.
- Loss of muscle (muscle wasting or atrophy) in your lower legs.
- Loss of feeling in your feet, legs, arms or hands. You may be less able to feel heat, cold, touch and pain.
- Mild to severe pain from stress on muscles and joints, often in the feet, lower legs and lower back.
- Changes in your feet. You may develop high arches or hammertoes (toes that bend or curl downwards, causing the middle joint to rise up).
- Changes in the way you walk. You may have difficulty lifting your foot up (foot drop), which causes you to drag your foot or lift it higher (high-stepping).
- Difficulty walking or running. Your ankles may twist and you may trip or fall.
- Difficulty with small movements of your fingers and hands.

Symptoms usually begin before age 20, but some people develop symptoms in middle age or later.

Symptoms slowly worsen over time. The severity of symptoms varies, even among family members with the disease.

What causes CMT?

CMT is caused by changes (mutations) in the genes that control the development of peripheral nerves. The mutations create abnormal proteins, which cause the nerves to slowly break down and lose their ability to send the signals needed for feeling and movement.

Different abnormal proteins cause different forms of CMT.

- Some abnormal proteins affect the nerve fibres (axons), causing 'Axonal' forms of CMT.
- Some abnormal proteins affect myelin, the insulating coating on the nerve fibres, causing 'Demyelinating' forms of CMT (CMT1 and CMT4). CMT1a is the most common form of CMT.

The gene mutations that cause CMT are usually inherited.

CMT can be inherited in several ways

You have 2 copies of every gene, one inherited from each parent.

CMT can be inherited by getting:

- 1 abnormal copy of the gene from one parent (autosomal dominant)
- 2 abnormal copies of the gene – one from each parent (autosomal recessive)
- an abnormal gene on the X chromosome from one parent (X-linked).

Although it is rare, the gene mutation causing CMT can occur spontaneously. This means your genes change unexpectedly and you did not inherit CMT.

How do you know I have CMT?

Your doctor or specialist can tell if you have CMT from:

- Talking to you about your symptoms and family history
- A physical exam to see how well your muscles and nerves are working
- Reviewing the results of tests

Tests for CMT include:

- Nerve conduction studies and Electromyography. These tests measure the electrical activity in your muscles and nerves. Knowing how fast your nerves send signals to your muscles can help determine which type of CMT you have.
- Blood/genetic tests. These blood tests check for abnormal genes in your white blood cells that cause different types of CMT.

How is CMT treated?

CMT cannot be cured or treated with medications. Supportive treatment can help you move and cope with symptoms. This includes physiotherapy and the use of orthopedic equipment. You will work closely with the health care team to make a treatment plan that meets your needs.

Physiotherapy

- A Physiotherapist can design a program of exercises and stretches to suit your needs.
- Exercise can strengthen your legs, improve your ability to walk, build endurance, and improve your posture and balance. Exercise also benefits your heart and overall health. Low impact exercise is best, such as biking or swimming. These activities do not put stress on your muscles and joints.
- Stretching may prevent or reduce problems (such as hammertoes) from the uneven pull of muscles on bones.

If started early, physiotherapy can delay or reduce nerve damage and muscle weakness.

Orthopedic equipment

- Ankle braces and orthotics can help with foot drop, provide support and prevent injury during activities such as walking and climbing stairs.
- High-top shoes or boots can provide support if you have weak ankles.

How can I learn more about CMT genes?

A genetic counsellor can help you understand the results of your genetic tests and how your type of CMT could affect any children you may have.

**If you have questions about CMT or your care,
please speak with your health care team
at the Neuromuscular Clinic.**

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