

Genetic Testing Options for Charcot-Marie-Tooth Disease

Charcot-Marie-Tooth disease

(CMT) is a group of hereditary disorders that result in damage to the peripheral nerves, or the nerves that control the muscles in the arms and legs. CMT is a progressive disease. Usually, individuals with the disorder slowly lose normal function and/or feeling in their feet, legs, hands, and arms. CMT is the most common type of inherited peripheral neuropathy.



Receiving a CMT diagnosis

CMT is diagnosed by a clinical exam that evaluates muscle function and tests sensory responses. Nerve conduction studies and electromyography (EMG), which look at the health of the muscles and associated nerves, may provide additional information. If these tests lead to a diagnosis of CMT, your doctor might recommend genetic testing to determine the type of CMT.

MDA Care Centers are located at 150+ hospitals and healthcare institutions across the United States, providing expert care from clinicians specializing in neuromuscular diseases. To find an MDA Care Center near you, visit mda.org/care/mda-care-centers.

Benefits of genetic testing

Pursuing genetic testing is a personal decision that you should discuss with your doctor and a genetic counselor to determine if it is appropriate for you.

There are several reasons why genetic testing may be beneficial:

- Testing can help provide an accurate diagnosis and end the diagnostic odyssey.
- There may be opportunities to participate in clinical trials focused on specific genetic subtypes of CMT. A genetic diagnosis is a critical requirement to be able to participate in these studies.
- Having an accurate diagnosis through genetic testing can help guide management of the disease and ensure you get the most effective treatment.
- There are several subtypes of CMT, and symptoms can vary depending on the subtype.
- Testing can provide information that you can use to make informed decisions about family planning. It can also help identify other family members who could be at risk for developing symptoms of CMT.

It may not be possible to determine every genetic cause of CMT and some tests may come back as “inconclusive,” but as technology changes and new genetic causes are discovered, identification may be possible. It is important to check in with your doctor every few years to see if you should be retested.



To learn more about Charcot-Marie-Tooth disease, visit mda.org/disease/charcot-marie-tooth or contact the MDA Resource Center at 833-ASK-MDA1.



Cost of genetic testing

The cost of genetic testing varies depending on the specific test, laboratory, and insurance coverage.

A sponsored genetic testing program means a company sponsors the test so it is free of charge to the individual. There may be eligibility requirements for sponsored testing.

You can work with your healthcare provider and insurance company to determine potential costs and/or eligibility for sponsored genetic testing, including what information is obtained by the sponsor.

Many commercial genetic testing laboratories offer CMT genetic tests for a fee, which might be covered by health insurance. Talk with your neurologist and a genetic counselor to determine which type of genetic test is appropriate for you. To find a genetic counselor, visit findageneticcounselor.nsgc.org.

Healthcare providers can request a Charcot-Marie-Tooth Panel **specimen collection kit from GeneDx**. Billing options through GeneDx include insurance, Medicare, some Medicaid plans, private institutions, and self-pay.



Sponsored genetic testing program:

Invitae Detect Muscular Dystrophy