What is Charcot-Marie-Tooth (CMT)?

Charcot-Marie-Tooth disease is a progressive neuromuscular disease that causes deterioration of the peripheral nerves—the long nerves that go from the brain to the hands and feet, controlling sensory information and muscle function. As the nerves deteriorate, the muscles around them atrophy. Charcot-Marie-Tooth, pronounced Shark-O-Marie-Tooth, is one of the most commonly inherited neuropathies, affecting an estimated 150,000 people in the United States and 2.8 million people worldwide. More people are affected by CMT than ALS, yet few people have ever heard of it! There is no cure for CMT.

The Charcot-Marie-Tooth Association, a not-for-profit organization (EIN #22-2480896) representing people with CMT, is dedicated to providing advocacy, education and research to improve the lives of everyone touched by this disease. We are funded solely through private donations and grassroots fundraising. We provide educational materials, sponsor 22 CMT clinics, hold several Patient/Family conferences each year and much more. With more than 70 community branches, the CMTA is the world leader in providing those affected by CMT help for today and hope for tomorrow. Our mission is to support the development of new drugs to treat CMT, to improve the quality of life for people with CMT, and, ultimately, to find a cure. Our vision is a world without CMT.

How is Charcot-Marie-Tooth Diagnosed?

Although CMT is one of the most common hereditary neuropathies, its symptoms are similar to other types of neuropathy. Not all doctors are familiar with CMT and diagnosing it can challenging for some physicians. Often, a person who has difficulty walking will first see an orthopedic doctor, who may recognize the CMT's characteristic high arches, hammer toes and gait abnormality. In making a diagnosis, a 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.

Early Symptoms of Charcot-Marie-Tooth

- Clumsiness
- Slight difficulty in walking because of trouble picking up feet or toe walking
- Weak leg muscles
- Fatigue

Common Symptoms of Charcot-Marie-Tooth

- Foot deformity (very high arch foot/feet)
- Difficulty lifting foot at the ankle (foot drop)
- Curled toes (known as hammer toes)
- Loss of lower leg muscle
- Numbness or burning sensation in the feet or hands
- "Slapping" when walking (feet hit the floor hard when walking)
- Weakness of the hips (hip dysplasia), legs, or feet
- Leg and hand cramps
- Loss of balance, tripping, and falling
- Difficulty grasping and holding objects and opening jars and bottles
- Clawed fingers, hands turning inward
- Curvature of the spine (scoliosis) or forward rounding of the back (kyphosis)

Rare Symptoms of Charcot-Marie-Tooth

- Speech and swallowing difficulties
- Breathing difficulties, especially when lying flat
- Hearing loss and vision difficulties
- Vocal cord paralysis

CMT usually gets slowly worse over time. Problems with weakness, numbness, balance and orthopedic problems can progress to the point of disability. Pain can be an issue, either as a direct result of the neuropathy (neuropathic pain) or as consequence of orthopedic problems. Other potential complications include:

- Progressive inability to walk from weakness, balance problems, and/or orthopedic problems
- Progressive inability to use hands effectively
- Injury to areas of the body that have decreased sensation.

Most of these symptoms can be managed very effectively with medication, rehabilitation and other strategies. Effective symptom management by an interdisciplinary team of healthcare professionals is one of the key components of comprehensive CMT care.

Developing a Care Team

In addition to the neurologist, a comprehensive CMT care team typically includes:

- An orthopedist for bracing, in-shoe orthotics, surgery, scoliosis and kyphosis
- A rehabilitation specialist such as a physiatrist, physical therapist or occupational therapist
- A mental health specialist like a psychologist or social worker
- A nutritionist
- A genetic counselor to determine the type of CMT
- A primary care physician, who plays a pivotal role attending to overall health and wellness

Sometimes this team works within a single center; more often people with CMT are referred (by their local CMTA branch or physician) to specialists in the community with whom they build their own team for comprehensive, coordinated care.

Register with the Inherited Neuropathy Consortium (INC)

INC's main goal is to measure how various forms of CMT progress over time so that clinical trials can determine whether potential therapeutics alter disease progression. INC is currently building the infrastructure to test these treatments. Anyone who has been seen at a CMT Center of Excellence can help by returning for a follow-up visit as part of INC's natural history studies, which depend on knowing how different types of CMT progress over time.

The consortium also works to develop standards of care for patients with CMT in areas like surgery, exercise and genetic testing, and to train the next generation of CMT scientists and physicians.

INC also identifies new causes of CMT and works with patients in its Patient Contact Registry to ensure that measures of disease severity and progression are relevant to those who actually have the disease, not just the investigators studying it. Anyone who hasn't yet done so is urged to join the Contact Registry online (www.rarediseasesnetwork.org/registry).