CMTA

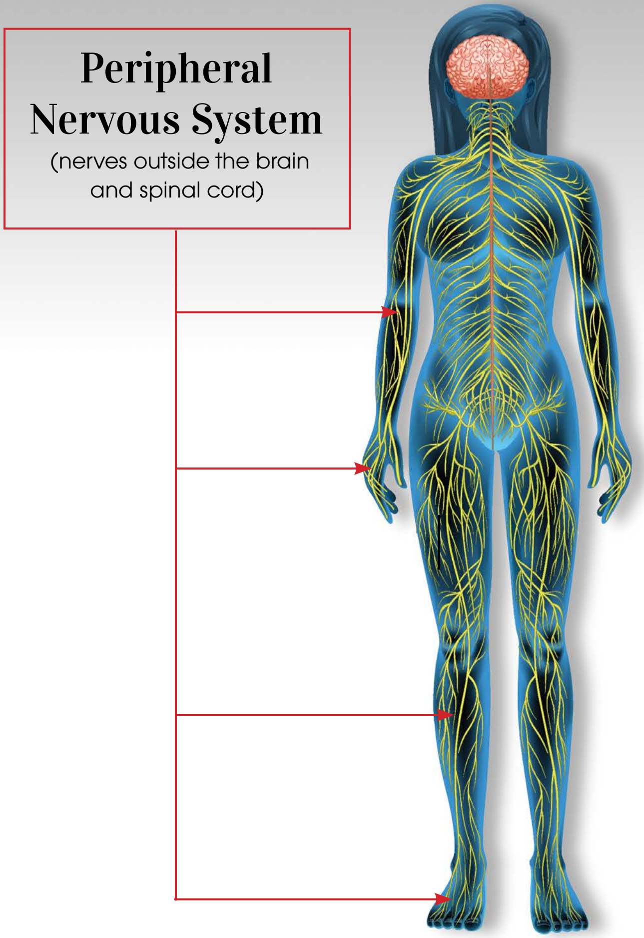
Charcot-Marie-Tooth Association

What is CMT?

Charcot-Marie-Tooth

(shar-ko¯´ mä-re´ tooth)

Charcot-Marie-Tooth is one of the most common inherited neuromuscular diseases, affecting the long (peripheral) nerves that go to the feet and hands of approximately one in 2,500 people worldwide. As the nerves die, the muscles around them atrophy.



Peripheral Nervous System

(nerves outside the brain and spinal cord)

CMT is:

A disease of the peripheral nerves that control muscles (unlike the muscular dystrophies, which affect the muscles themselves).
Found in both genders and in all races and ethnic groups, it is the most commonly inherited peripheral neuropathy, affecting 2.8 million people worldwide.
CMT is slowly progressive, causing loss of normal function and/or sensation in the lower legs/feet and hands/arms. CMT is currently incurable, but not usually fatal, though it is severely disabling in a small proportion of cases.

What are the Symptoms of CMT?

First signs include frequent tripping, ankle sprains, clumsiness and "burning" or pins-and-needles sensations in the feet or hands.

Structural foot deformities such as high arches and hammertoes are common.

Muscle wasting in the lower legs and feet may lead to foot drop, poor balance and other gait problems.

Muscular atrophy in the hands often causes people to have difficulty with tasks involving manual dexterity, such as writing and manipulating zippers and buttons.

Abnormal sensation in the extremities and an inability to sense where one's body is in space are also common, and many people experience neuropathi, muscle or joint pain.

Poor tolerance for cool or cold temperatures is typical and many people have chronically cold hands and feet.

Additional symptoms may include fatigue, sleep apnea, breathing difficulties, scoliosis, kyphosis and hearing loss.

A CMT diagnosis involves clinical evaluation of muscle function and atrophy, testing of sensory responses, and electromyographic and nerve conduction studies. Many types of CMT can also be diagnosed by genetic testing. Please visit www.cmtausa.org/diagnosis for more information.

Is there a Treatment for CMT?

Although there is no drug treatment for CMT, physical therapy and moderate activity (but not overexertion) can help maintain muscle strength, endurance and flexibility. Mechanical supports like AFOs (ankle-foot orthoses) and custom-made shoes can improve gait and balance. When medically indicated, orthopedic surgery can correct deformity and help maintain mobility and function. Occupational therapy and adaptive devices can help people perform activities of daily living.

Any medications people with CMT should avoid?

YES! Glad you asked! Some medications are potentially toxic to people with CMT. Vincristine has been proven hazardous and should be avoided by all CMT patients. Taxols also pose a high risk to people with CMT. The complete list of potentially neurotoxic medications can be viewed at www.cmtausa.org/medicationlist. Before taking any medication or changing medications, be sure your physician is fully aware of your medical condition, and discuss any possible side effects they may have on CMT. Have a drug/medication question? Visit www.cmtausa.org/medquest or email askthexpert@cmtausa.org.

Where Can I Get Help?

Founded in 1983, the Charcot-Marie-Tooth Association (CMTA) is a 501(c)(3) nonprofit organization whose mission is to support the development of drug therapies 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.

The CMTA fulfills its mission with a wide variety of member services. Numerous publications are available, including the long-running *CMTA Report* and *CMTeen*, an online magazine by, for and about teenagers with CMT. Branches provide members education and support while Patient/Family Conferences offer them the chance to get firsthand information from the best clinicians and researchers in the world.

Those clinicians and researchers can also be found at CMTA Centers of Excellence, where people with CMT are examined and treated in multidisciplinary clinics while helping the CMTA systematically gather information for use in clinical trials and other CMT research. Visit www.cmtausa.org/coe for more information about a COE near you.

University of Iowa
Center of Excellence

What is the CMTA Doing to Find a Cure?

STAR…

Strategy To Accelerate Research

Assays and High Throughput Screening (HTS)

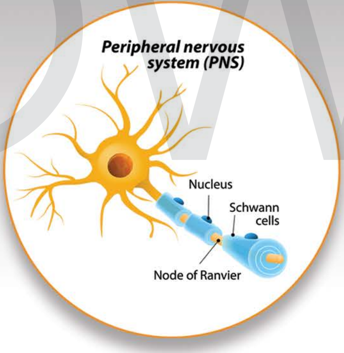
Animal Models

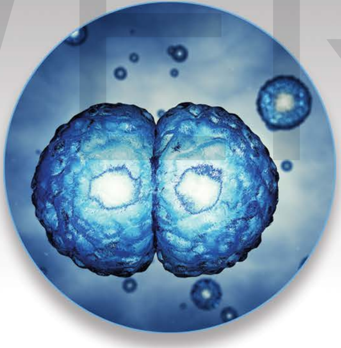
The Human Element: Skin Cells to Stem Cells

Alliance Partnerships and New Therapies

Clinical Trials and Outcome Measures

R.POWER





The CMTA is aggressively fighting to find a pharmaceutical treatment, and, ultimately, a cure for all types of CMT. Since 2008, when the CMTA launched its Strategy to Accelerate Research (STAR) to capitalize on breakthroughs in genetics, it has been working with the preeminent researchers, clinicians and institutions in the inherited neuropathy community.

CMT is unlike other neuromuscular disorders because at least 90 specific gene defects that cause it have been identified. More importantly, the fact that most of these genetic mutations can be replicated in laboratory models and grown as tissue cultures has opened an extraordinary window of opportunity to develop treatments and cures.

STAR "is the most important initiative ever undertaken by the Charcot-Marie-Tooth Association," according to Michael E. Shy, MD, Co-Chair of the Clinical Expert Board. "In addition to developing specific therapies for CMT patients, the translational science employed in the research could have major implications for the treatment of a host of related genetic disorders."

The CMTA has made amazing progress toward therapies for the most common types of CMT, which affect approximately 90 percent of diagnosed cases. In the past two years, the CMTA has financed 40 projects and spent $3.5 million on research. Thanks to the support from donors, there is huge momentum and promise. To get the latest research updates and more information about STAR, visit www.cmtausa.org/STAR.

CMTA

Charcot-Marie-Tooth Association

How Can I Get Involved?

Become a member of the CMTA today, and help make a difference. Together, we can create a world without CMT!

Join the CMTA Online Community

It's the best way to take advantage of everything our website has to offer and keep up with what's happening in the CMT patient and scientific communities. And it's free! Just visit www.cmtausa.org/join.

Become a Subscription or STAR Member

Once you've joined the CMTA Online Community, become a Subscription or STAR Member and you'll receive our quarterly magazine, The CMTA Report. You can read it online, get it delivered to your mailbox, or both.

Join a CMTA Branch

Visit www.cmtausa.org/branch or call 1-800-606-2682 x106 to find a branch near you. You'll learn how other people are coping with CMT and find out what they're doing to promote awareness, fund research and support the work of the CMTA.

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Become a Contributor or Fundraiser

Whether you make an individual donation or set up a fundraiser benefiting the CMTA, your contribution will help the CMTA fund research, provide information to patients and physicians, and promote awareness of CMT. Please visit www.cmtausa.org/fundraising to find out how easy we've made it to get involved. Thanks for your support!

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