CMTA
Charcot-Marie-Tooth Association

What is CMT?

Charcot-Marie-Tooth

(shar-ko¯’ mä-re´ tooth)

CMT IS:

- A disease of the peripheral nerves that control the muscles (unlike the muscular dystrophies, which affect the muscles themselves).
- The most commonly inherited peripheral neuropathy, found in both genders and in all races and ethnic groups and affecting more than 3 million people worldwide. Although CMT is typically inherited from one's parents, it can also occur as a result of a new or spontaneous mutation. People who have these "de novo" mutations can then pass the condition on to their children.
- Usually slowly progressive, causing loss of normal function and/or sensation in the feet/legs and hands/arms.
- Currently incurable, but not usually fatal, though it can be severely disabling in some people.

What are the Symptoms of CMT?

First signs may include toe-walking, 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, but some people have flat feet.

Foot drop, poor balance and problems walking as muscle wasting in the lower legs and feet progresses.

Difficulty with tasks involving manual dexterity, such as writing and manipulating zippers and buttons, often accompanies muscular wasting in the hands.

Abnormal sensation, loss of ability to feel light touch, inability to differentiate between hot/cold, and the loss of proprioception or the inability to sense where one’s body is in space are also common, and many people experience neuropathic, muscle or joint pain.

Poor tolerance for cool, cold and/or hot temperatures is typical. Many people have chronically cold hands and feet.

Additional symptoms may include flexed fingers, contractures, tremor, knee and/or hip problems, cramps, thenar muscle atrophy (weakness of the muscles between the thumb and forefinger), muscle weakness and loss of hand strength, chronic fatigue, sleep apnea, breathing difficulties, swallowing difficulties, absent or reduced reflexes, poor circulation, scoliosis, kyphosis and hearing loss.

The psychological impact of having CMT can be devastating, leading to irritability, depression, anxiety, isolation, loss of pleasure, weight gain or loss, and feelings of hopelessness, worthlessness or guilt.

If you are having suicidal thoughts, immediately call your local suicide hotline or the National Suicide Prevention Lifeline: 1-800-273-TALK (1-800-273-8255), or seek help right away at a local emergency room.

A CMT diagnosis involves a 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.

Is there a Treatment for CMT?

Although there is no drug treatment for CMT, physical/occupational 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! 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 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?
Email askthexpert@cmtausa.org.

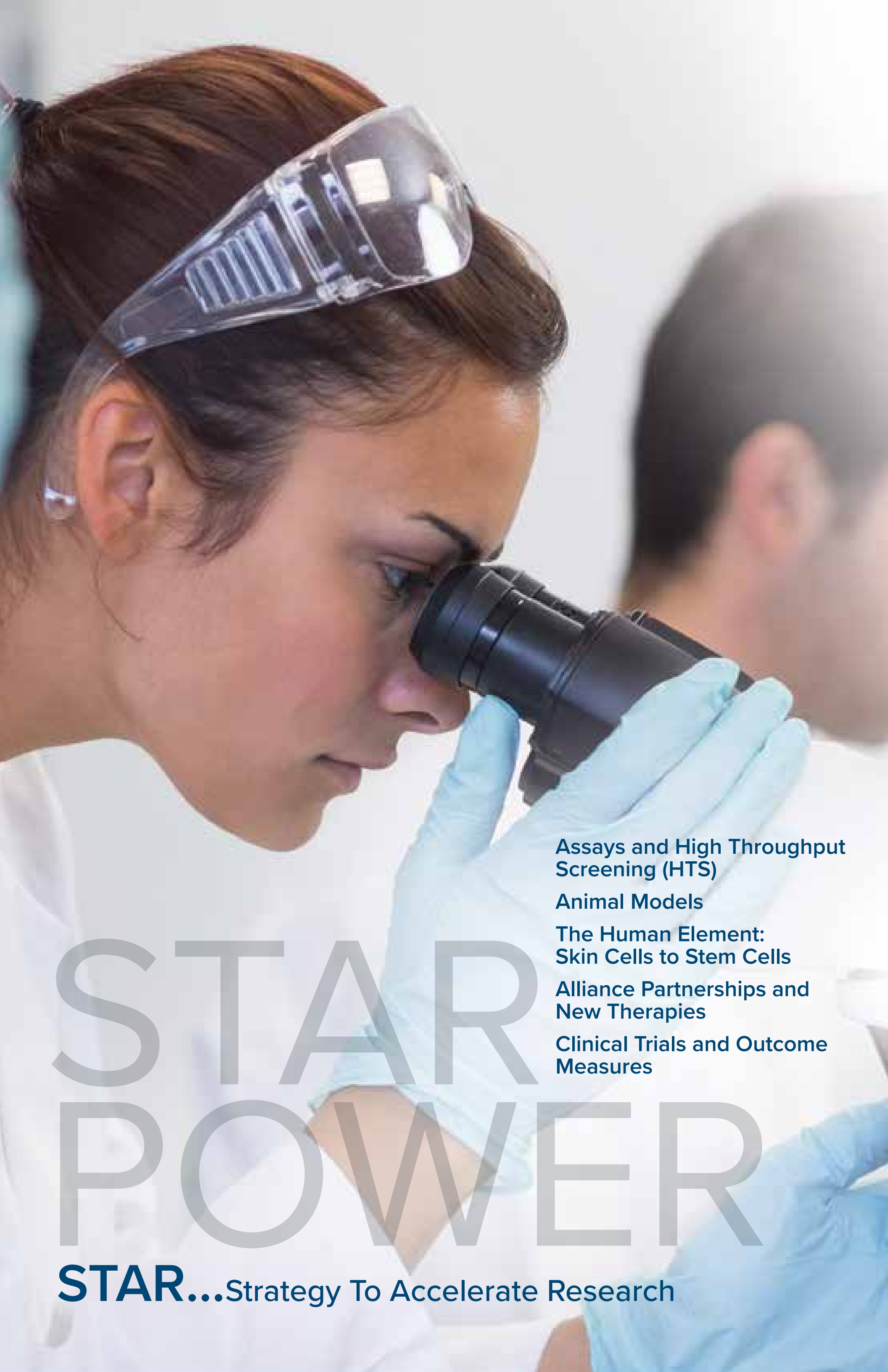
Where Can I Get Help?

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

The CMTA fulfills its mission with a wide variety of member services. Numerous publications are available, including The CMTA Report. Camp Footprint, a free, five-day sleepaway camp for youth (ages 10-18) with CMT is the only camp in the U.S. exclusively for kids with CMT. It's an unforgettable week of adventure and fun with friends! Our national network of CMTA branches provides members education and support while our Patient & Family Conferences offer them the chance to get firsthand information from the best clinicians and researchers in the world.

Many of those clinicians and researchers can also be seen at CMTA Centers of Excellence, where people with CMT are examined and treated in multidisciplinary clinics, all while helping the CMTA systematically gather information for use in clinical trials and other CMT research.

Visit cmtausa.org/coe for more information about a COE near you.

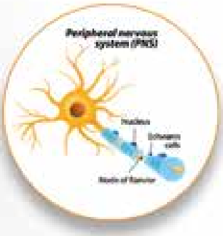


STAR POWER

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





The CMTA is The Driving Force Behind CMT Research

What is the CMTA Doing to Find a Cure?

The CMTA is aggressively pursuing treatments and a cure for all types of CMT. Since launching the Strategy to Accelerate Research (STAR) in 2008, the CMTA has been working with preeminent researchers, clinicians, patients and institutions in the global inherited neuropathy community to capitalize on breakthroughs in genetics.

Unlike other neuromuscular diseases, the genetic causes of CMT are known. In fact, more than 90 specific genes that cause the disease 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 using the latest pharmaceutical, biotech, and gene therapy technologies.

“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 unprecedented progress toward therapies for virtually almost all types of CMT. Since the inception of STAR, the CMTA has financed more than 100 projects and committed over $10 million in research. Thanks to support from our donors, STAR continues to gain momentum and the promise of breakthroughs is imminent.

To get the latest research updates and more information about STAR, visit cmtausa.org/STAR.

Stay Connected, Get Involved, Be Informed.

Living with CMT can be challenging, but you are not alone. The CMTA is with you every step of the way. Together, we can create a world without CMT!

Go to the CMTA website (cmtausa.org) to learn about CMT, connect with patient and scientific communities, and access the latest information and resources about all aspects of CMT, from genetic testing, bracing and exercise to clinical trials and CMTA research—STAR. While you are on the CMTA website be sure to:

Sign up for CMTA E-news (cmtausa.org/enews). Get breaking research updates and news about exciting awareness initiatives, fun community activities, fundraisers and more.

Look for a CMTA Branch near you (cmtausa.org/branch). At your local branch, 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.

"Like" us on Facebook (facebook.com/CMTAssociation) and join our CMTA Facebook Community-Focused Discussion Group (facebook.com/groups/CMTAssociation).

Become a Contributor or Fundraiser (cmtausa.org/donate). Whether you make an individual donation, sign on for monthly giving 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.

Receive the CMTA Report. When you give a donation of $100 or more to the CMTA, you will automatically be signed up to receive our quarterly newsletter, The CMTA Report—our gift to you!

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TREATMENT DRIVEN • COMMUNITY POWERED

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PO Box 105, Glenolden, PA 19036
cmtausa.org • 1-800-606-2682