Charcot-Marie Tooth Disorder (CMT)

What is Charcot-Marie Tooth Disorder?

Charcot-Marie-Tooth Disorder, or CMT, is the most common inherited neurological disorder, affecting approximately 125,000 Americans. CMT is found worldwide, in all races and ethnic groups. Although discovered in 1886 by three physicians, Jean-Marie Charcot, Pierre Marie and Howard Henry Tooth, the disorder has remained a mystery to the general public and medical community. CMT patients slowly lose normal use of their feet/legs and hands/arms as nerves to the extremities degenerate. The muscles in the extremities become weakened because of the loss of stimulation by affected nerves. There is a corresponding loss of sensory nerve function. Unlike muscular dystrophy in which the defect is in the muscles, CMT is a disorder in which the defect is in the nerves that control the muscles. CMT is not a fatal disorder, and patients enjoy a normal life expectancy.

What are the characteristics?

A high arched foot is one of the first signs of this disorder. As the disease progresses, structural foot deformities take place. The patient develops a pes cavus foot, characterized by foot drop and hammer toes. Ankle sprains are frequent. The progressive muscle wasting leads to problems with walking, running and balance. The knees have to be raised higher off the ground and the patient develops high steppage gait. In some patients, muscle weakness may also occur in the upper legs.

Hand function also becomes affected because of progressive muscle atrophy, making fine manipulatory acts like writing more difficult.

The loss of nerve function in the limbs and extremities leads to sensory loss as well. The ability to distinguish hot and cold food is diminished as well as the sense of touch.

The degree of severity can vary greatly from patient to patient, even within the same family. A child may or may not be more severely disabled than his/her parent.

How is it inherited?

CMT is generally inherited in an autosomal dominant pattern. This means that if one parent has the disease (either the father or the mother) there is a 50% chance of passing it on to each child.

CMT can also be inherited in a recessive or in an X-linked pattern. To determine the pattern of inheritance, each CMT patient should consult a genetic counselor, neurologist or other medical authority familiar with the disease to carefully define the pattern of inheritance in his/her family.

How is it diagnosed?

Careful diagnosis of CMT involves clinical evaluation of muscle atrophy, testing of muscle and sensory responses, nerve conduction and electromyographic studies, as well as a thorough review of the patient's history.

Even within the same family group, some CMT patients may be more disabled than others. Some people who carry the CMT genetic trait show no apparent physical symptoms. This variation in degree of physical disability, together with a lack of physician awareness, has often led to misdiagnosis.

Today, the Charcot-Marie-Tooth Association is educating both medical specialists and patients about CMT.

How is it treated?

At present there is no cure for CMT, although physical therapy and moderate activity are often recommended to maintain muscle strength and endurance.

Leg braces and custom made shoes can help improve the quality of life for most patients. Corrective orthopaedic foot surgery is available to help maintain mobility when medically indicated.

Splinting, specific exercises, adaptive devices and sometimes surgery will help maintain hand function.

What is the Charcot-Marie Tooth Assoc.?

The CMTA is a non-profit organization founded in 1983 whose goals are patient support, public education, promotion of research and the ultimate treatment and cure of CMT.

The CMTA is governed by a Board of Directors comprised of CMT patients, CMT family members, medical professionals, and other persons whose expertise facilitate the work of the CMTA.

The Medical Advisory Board of the CMTA consists of over forty clinical and research medical professionals. This international group of men and women are noted CMT experts in neurology, genetics, orthopaedic surgery, physiatry, physical therapy and podiatry.

The membership of the CMTA is comprised of CMT patients/families, supportive friends, and medical professionals.

What is the CMTA doing to help?

The CMTA...

- Offers help and information to the CMT patient/family and the medical community through its quarterly newsletter, The CMTA Report.
- Conducts patient/family and professional conferences throughout the country.
- Establishes and sponsors support groups throughout the country.
- Has a videotape program, which is the only source of videotaped CMT educational lectures.
- Has an international referral list of physicians and other medical professionals knowledgeable about CMT.
- Has available a national list of suppliers and fitters of footwear for the CMT foot.
- Has a list of suppliers of adaptive "gadgets" to aid in daily living tasks.
- Assists the CMT research community by maintaining the CMTA patient registry.

CMT...a summary

- Is the most common inherited neurological disease, affecting approximately 125,000 Americans.
- Is slowly progressive, causing deterioration of peripheral nerves which control sensory information and muscle function of foot/lower leg and hand/forearm muscles.
- Causes degeneration of foot and lower leg muscles and hand and forearm muscles.
- Causes foot drop walking gait, foot bone abnormalities (high arches and hammer toes), problems with hand function, balance problems, occasional lower leg and forearm muscle cramping, loss of some normal reflexes, occasional partial sight and/or hearing loss problems and in some patients may cause scoliosis (curvature of the spine).
- May produce chronic pain and fatigue.
- Does not affect life expectancy.
- Has no effective treatment, although physical therapy and moderate physical activity are beneficial.
- Is sometimes treated surgically.
- Is usually inherited in a autosomal dominant pattern, which means if one parent has CMT there is a 50% chance of passing it on to each child.
- Is present in the world-wide population.
- May become worse if certain neurotoxic drugs are taken.
- Can vary greatly in its severity, even within the same family.
- Is the focus of significant genetic research bringing us closer to answering the CMT enigma.

How to contact CMTA:

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