Charcot-Marie-Tooth disease

CMT is managed symptomatically via a number of therapies including podiatry, orthopaedic surgery, physiotherapy and occupational therapy.

Charcot-Marie-Tooth disease (CMT) also known as hereditary motor and sensory neuropathy (HMSN) comprises a family of genetic conditions that mainly affect the motor and sensory nerves, which run from the spinal cord down the arms and legs.

It is caused by alterations in genes that produce proteins involved in the structure and function of either the peripheral nerve cell (axon), or the myelin sheath that wraps around the axon to insulate it and in normal circumstances allows the signals to travel faster and better.

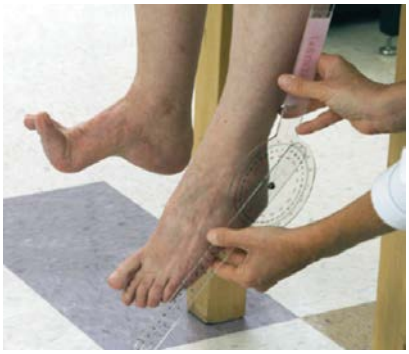
They are many types of CMT based on the specific gene abnormalities involved in the structure and function of axons or myelin sheath.

The principal types include CMT1, CMT2, CMT3, and CMT4. Age of onset varies between types of CMT, with symptoms becoming apparent between the ages of 5-15 years for those with CMT1, and between 10 -20 years for those with CMT2.

Onset of CMT3 and 4 will usually occur before three years of age. The severity of symptoms will vary greatly from person to person even within the same family.

Features of CMT include:

- Foot problems – high arches, hammer toes, foot drop.



- Muscle weakness in lower legs causing problems with running, walking and balance.
- Hand function can be affected resulting in problems holding pens, grasping, and performing fine motor tasks.
- Tingling and burning sensations in the hands and feet due to the loss of nerve function.
- The sense of touch is diminished, as is the ability to perceive changes in temperature due to sensory loss.
- Sensitive to the cold due to loss of insulating muscle mass, which can leave people with chronically cold hands and feet and lead to swelling of the feet and ankles.
- Loss of deep-tendon reflexes, such as the knee jerk reaction.
- Scoliosis or mild curvature of the spine.

Management of CMT

CMT is managed symptomatically via a number of therapies depending on the needs of the individual. These include:

- Podiatry: Care and monitoring of foot problems.
- Orthotics clinics: Manufacture and fitting of braces and customised footware.
- Orthopaedic surgery: Straighten toes, lengthen heel cords, or lower arches.
- Physiotherapy and occupational therapy: Design exercise programmes to strengthen muscles; learn about energy conservation; provide assistive devices.
- Dietitian: To maintain a healthy balanced diet and weight.

CMT: A practical guide published and updated in 2014 by CMT United Kingdom is tailored for those affected by CMT and health professionals. It covers theoretical and practical issues – understanding the genetics and mechanics of CMT, diagnosis, advice on coping and managing the condition day to day. It can be downloaded on the MDANZ website.

Medications

There is a list of medications that are potentially toxic to people with CMT. This list is available on the MDANZ website. Please share this with your GP and relevant health providers. Ⓡ