CHARCOT-MARIE-TOOTH DISEASE





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WHAT IS CHARCOT-MARIE-TOOTH DISEASE?

Charcot-Marie-Tooth (CMT) disease, also known as hereditary motor and sensory neuropathy, is an umbrella term for a range of inherited genetic disorders that affect the peripheral nerves in the body. It is named after the three physicians who first described the condition in 1886. CMT causes muscle weakness, loss of sensation, and impaired reflexes in the arms and legs. Orthopaedic issues such as foot drop, high arches in the feet, scoliosis, and joint deformities are common in CMT and can impact mobility and quality of life.

SYMPTOMS

Symptoms of CMT can vary widely depending on the type and severity of the condition. Some common symptoms include:

- Muscle weakness and atrophy, especially in the feet, legs, hands, and forearms
- Loss of sensation or numbness in the feet and hands
- Impaired reflexes

- Foot deformities such as high arches
- Foot drop, or difficulty lifting the foot when walking.
- Scoliosis or other spinal deformities
- Hip instability

DIAGNOSIS

A combination of medical history, physical examination, nerve conduction studies, and genetic testing is commonly used to make a diagnosis. Early diagnosis is important for optimising outcomes and addressing orthopaedic issues before they become more severe.

TREATMENT

Treatment for CMT focuses on managing symptoms, improving function, and maximising quality of life. Depending on the individual's needs and goals, treatment may involve a combination of regular exercise and stretching, physiotherapy and occupational therapy.

Orthopaedic issues such as foot drop, foot deformity, scoliosis, and hip problems may require surgical intervention or treatment with physiotherapy, bracing, casting, or orthotics to improve mobility and quality of life. CMT is a lifelong and often progressive condition.

USEFUL WEBSITES

Health Direct

https://www.healthdirect.gov.au/charcot-marie-tooth-disease