

Scottish Muscle Network

Charcot-Marie-Tooth (CMT) “at a glance” sheet

Diagnosis/ Genetics

- Autosomal dominant mutation in PMP22 is the commonest (CMT1A)
However, autosomal recessive and x-linked forms exist involving a variety of genes
- Genetic counselling of families is advised

Clinical features and outlook

Clinical onset usually in first or second decade and there is considerable variability in phenotype. Initial symptoms include ‘foot drop’ and ‘pes cavus’ which may be apparent from an early age. CMT causes slowly progressive weakness, atrophy and sensory loss in the distal muscles of the upper and lower limbs. Impaired balance is very common. Pain may be a feature of CMT and if present may be due to secondary effects on the joints or muscles. Pain should be treated along conventional lines. Sensory loss can be so severe as to lead to painless ulcers and secondary infections.

Associated features and anaesthetic risk

The neuropathy can be associated with enlarged nerves (hypertrophic neuropathy) which can lead to secondary compressive damage to peripheral nerves or spinal cord. There is not normally an anaesthetic risk.

Cardiac manifestations

There is normally no cardiac weakness associated with CMT.

Respiratory manifestations

In general, there is no respiratory muscle weakness in CMT. However, obstructive sleep apnoea and vocal cord palsy have both been described.

Supportive Treatment

Rehabilitation including orthotics and physiotherapy, surgical treatment of skeletal deformities and management of pain.

Patients should carry an alert bracelet or similar.

Sources of additional information

www.smn.scot.nhs.uk

www.musculardystrophyuk.org

www.cmt.org

www.neuromuscular.wustl.edu/index.html

NOTE

This guideline is not intended to be construed or to serve as a standard of care. Standards of care are determined on the basis of all clinical data available for an individual case and are subject to change as scientific knowledge and technology advance and patterns of care evolve. Adherence to guideline recommendations will not ensure a successful outcome in every case, nor should they be construed as including all proper methods of care or excluding other acceptable methods of care aimed at the same results. The ultimate judgement must be made by the appropriate healthcare professional(s) responsible for clinical decisions regarding a particular clinical procedure or treatment plan. This judgement should only be arrived at following discussion of the options with the patient, covering the diagnostic and treatment choices available. It is advised, however, that significant departures from the national guideline or any local guidelines derived from it should be fully documented in the patient's case notes at the time the relevant decision is taken.