

CHARCOT-MARIE-TOOTH DISEASE

Charcot-Marie-Tooth disease (CMT) is a genetic disorder characterised by slowly progressive muscular weakness. The onset of the condition is frequently first apparent in childhood. Onset is in the lower limbs, causing weakness around the ankles and often an abnormality in the shape of the feet (high in-step) which doctors call 'pes cavus'. Mild loss of sensation may be present in the feet and hands. After many years, weakness may develop in the hands and spread upwards in the lower limbs to affect the knees and thighs.

The underlying cause is an abnormality (called a mutation) in one of the genes responsible for producing a protein that is essential for normal nerve function. Each nerve is made up of thousands of proteins. CMT can be caused by mutations in any one of about 30 genes - each protein that is affected is different, the end result is similar in terms of the overall symptoms experienced by an individual with CMT.

CMT affects peripheral nerves - these nerves are responsible for passing on commands from the brain to the muscles (motor nerves) and for passing information to the brain about sensations, such as pain, heat, cold, touch and balance, where your joints are in space (sensory nerves). When these nerves are damaged, children are said to have a neuropathy.

CMT is thought to be the most common inherited neurological condition in the UK. There are two main forms: CMT 1 Demyelinating - affecting the myelin sheath insulating and nourishing the nerve's axon. CMT2 Axonal - directly affecting the axon.

*Children's needs are **individual** - some children may require additional support:*

- Aspects of personal care, e.g. toileting, changing for PE
- Completing tasks: tiring during the day, due to increased effort
- Fine motor skills affecting handwriting and practical tasks
- Gross motor skills, mobility around the school environment (which may gradually reduce requiring a wheelchair to aid locomotion)
- Organising and arranging equipment and materials
- PE and School Sports may require alternatives / differentiation
- Potential emotional outbursts related to the deteriorating condition
- Self esteem (loosing mobility), requiring pastoral support
- Task completion, focusing and concentrating on activities

FURTHER INFORMATION: Charcot-Marie-Tooth UK <http://www.cmt.org.uk>