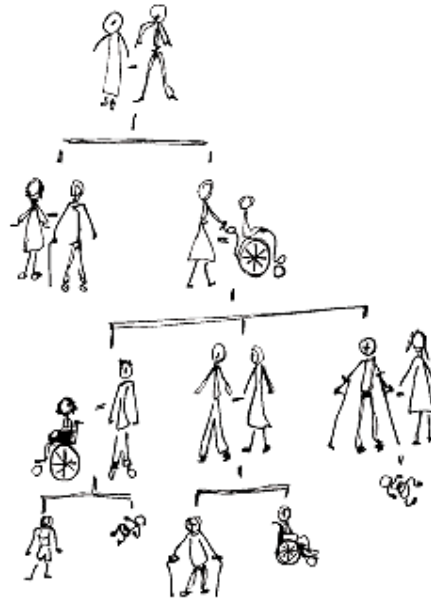


CROSS-CURRICULAR MATERIALS FOR KEY STAGE 4  
(SCOTTISH CERTIFICATE OF EDUCATION - STANDARD GRADE)



## Genes and You

Teaching about genetics from a human perspective

by Gill Mullinar

GENETIC INTEREST GROUP

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# GENETIC CHROMOSOMAL CONDITION CARDS

**GENETIC CHROMOSOMAL CONDITION CARDS ON THE FOLLOWING:**



**CHARCOT-MARIE-TOOTH DISEASE (CMT)**

## Charcot-Marie-Tooth Disease (CMT)

**WHAT IS CMT?** CMT is an inherited condition in which there is a defect in the peripheral nerves (to the arms and legs). CMT affects the transmission of messages along the nerves, either through deterioration of the Myelin (the insulating substance that wraps round the nerves) or of the axons, (depending on the type of CMT, though the axons are also affected in de-myelinating CMT later on). Myelin helps the electric signals (which tell the muscles to contract or relax) to travel along the nerves. When the myelin does not work well, as in CMT, these electric signals move very slowly. When the axons do not work properly, the electrical signals travel at near normal speed but are weaker. In both cases movement of the legs and arms is affected.

### HOW IS CMT PASSED ON?

There are several types of CMT. They are caused by defects in different genes and have different patterns of inheritance. The most common pattern of inheritance for CMT is autosomal dominant, which means that where one parent has CMT, there is a 50% (1 in 2) chance of each and every child being affected. N.B. Peter (see Genes and you: Unit 5) inherited CMT in this way. He received one faulty copy of the gene from his father, who has CMT, and one working copy from his mother. The faulty gene is dominant, so Peter is also affected by CMT.

A person with cmt may experience some, or all, of the following:

- Difficulty in walking because of problems picking up the feet
- Children may experience difficulty with running and general agility before any other noticeable symptoms – including being “clumsy”.
- Weakness in the hands and forearms, which can make certain activities such as doing up buttons, picking up small items, writing etc difficult.
- Mild loss of feeling in the limbs, fingers and toes, and loss of reflexes.
- Some muscle tremor.
- Chronic pain and fatigue because of the extra effort needed to do daily activities.
- Slight curve of the spine.
- Hip or knee problems.
- Increased difficulty walking – aids may be needed, such as orthoses and walking sticks.
- Around 10% - 20% of people with CMT have no symptoms at all.

### OTHER INFORMATION

CMT does not usually show up at birth. It tends to become evident some time between the ages of 5 and 15 and to become particularly noticeable during growth spurts at puberty. CMT is progressive (i.e. symptoms get worse over time), but some people find that it does not change much once they have stopped growing. It is not a life-threatening condition.

How severely a person is affected can vary greatly even within the same family. It is rare for a person with CMT to lose the ability to walk, although walking aids (sticks etc.) may be needed.

CMT Type 1a can now be detected by a blood test. This gives a more precise diagnosis than the electromyogram, a test which measures speed of conduction in the nerves (this is the test which Peter remembers having as a child - see Genes and you: Unit 5).

CMT is named after the three 19th century neurologists who first described the physical characteristics of the condition. Two were French (Charcot, pronounced Shar-koe, and Marie) and one was English (Tooth).

CMT has other names as well. Tooth discovered that one of the shin muscles (the peroneus muscle) wastes (or atrophies) in people with the condition, so CMT is also called Peroneal Muscular Atrophy. Another name for CMT (Hereditary Motor and Sensory Neuropathy or HMSN) shows that the condition is inherited (hereditary), and is a condition affecting two types (motor and sensory) of nerves (neuropathy).

If you are interested in finding out more about CMT, you can write (enclosing an A5 stamped addressed envelope) to: CMT UK, PO Box 5089, Christchurch, BH23 7ZX Telephone: 0870 7744 314. [www.cmt.org.uk](http://www.cmt.org.uk)