Condition: Hereditary motor and sensory neuropathy (Charcot-Marie-Tooth disease)

Inheritance:
Genetically heterogeneous.

Genetic etiology:
Multiple genes have been identified as being responsible for hereditary motor and sensory neuropathy. The most common form is due to duplication of the *PMP22* gene on chromosome 17. This gene encodes a protein involved in myelin formation. Deletion of the same gene results in hereditary neuropathy with predisposition to pressure palsies.

Frequency:
Overall frequency of hereditary neuropathies approximately 1/3,000.

Clinical features:
The hereditary neuropathies are a diverse group of disorders characterized by dysfunction of peripheral nerves. HSMN1 (CMT1) is the most common, and represents a demyelinating neuropathy. It presents as progressive weakness and sensory loss, initially affecting long nerves to the feet. There is loss of deep tendon reflexes and development of high arches. Nerve conduction velocities are slowed. Other forms of hereditary neuropathy are distinguished by involvement of axons instead of myelin, and specific nerves affected.

Management:
Supportive care.

Genetic counseling:
Depends on specific genetic type and mode of inheritance; genetic testing available for CMT1, and for many other forms.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.