



POSNA

The Core Curriculum

Hereditary motor sensory neuropathies (HMSN)

Objectives

1. Define hereditary motor sensory neuropathy and list several types
2. Describe presenting symptoms and signs of type 1 Charcot-Marie-Tooth disease
3. Discuss orthopaedic problems associated with HMSN, and an approach to management

Discussion

The hereditary motor sensory neuropathies (HMSN) are a heterogeneous group of variously inherited neuromuscular disorders. For practical purposes, the orthopaedically related HMSN's are confined to the first three HMSN's, which are otherwise known as Charcot-Marie-Tooth disease (CMT), types 1 and 2, and Déjérine-Sota disease. CMT1 is characterized by demyelinating of the peripheral nerves, and CMT2 by axonal degeneration. Patients with CMT1 have lowered motor and sensory nerve conduction; patients with CMT2 may have normal nerve conduction studies. Genetically, CMT is a very heterogeneous group of disorders, and this subject is being actively studied at present. Estimates of the overall incidence of CMT is 1/2500, so the disorder is quite common. The most common variety, CMT1 is described as being secondary to a presence of an extra copy of peripheral myelin protein 22, with an autosomal dominant pattern of inheritance. These patients usually have the onset of nerve conduction delays by age 2; active demyelination is limited to childhood. The most common clinical phenotype is distal muscle wasting and weakness, more severe in the lower than upper limbs, tendon areflexia, mild sensory loss, especially temperature, and foot deformity. Diagnosis is usually established with nerve conduction studies, sometimes nerve or muscle biopsies are helpful. Specific diagnosis can be established with DNA testing.

Of the orthopaedic problems, pes cavovarus is most common. The interossei and lumbricals are more severely involved, with relative sparing of the extrinsics, which undoubtedly contributes to the deformity. The general thought at present is to use soft tissue surgery and osteotomy rather than triple arthrodesis for management of pes cavovarus, as triple arthrodesis has received mixed reviews on follow-up studies. Acquired hip subluxation can occur in adolescence, and more active screening of this population has been recommended. Scoliosis occurs in a fairly large percentage but treatment is rarely necessary.

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