Autosomal dominant intermediate Charcot-Marie-Tooth disease type D

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal dominant intermediate Charcot-Marie-Tooth disease type D. ORPHA:100046

Autosomal dominant intermediate Charcot-Marie-Tooth disease type D is a rare hereditary motor and sensory neuropathy characterized by intermediate motor median nerve conduction velocities (usually between 25 and 45 m/s) and signs of both axonal degeneration and demyelination without onion bulbs in nerve biopsies. It presents with usual Charcot-Marie-Tooth disease clinical features of variable severity (progressive muscle weakness and atrophy of the distal extremities, distal sensory loss, reduced or absent deep tendon reflexes, and feet deformities). Other findings in some of the families include debilitating neuropathic pain and mild postural/kinetic upper limb tremor.