Charcot-Marie-Tooth disease type 1F

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Charcot-Marie-Tooth disease type 1F. ORPHA:101085

Charcot-Marie-Tooth disease type 1F (CMT1F) is a form of CMT1, with a variable clinical presentation that can range from severe impairment with onset in childhood to mild impairment appearing during adulthood. CMT1F is characterized by a progressive peripheral motor and sensory neuropathy with distal paresis in the lower limbs that varies from mild weakness to complete paralysis of the distal muscle groups, absent tendon reflexes and reduced nerve conduction. CMT1F represents the ''demyelinating'' form of CMT2E and is caused by mutations in the NEFL gene (8p21.2).