Autosomal dominant Charcot-Marie-Tooth disease type 2U

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal dominant Charcot-Marie-Tooth disease type 2U. ORPHA:397735

Autosomal dominant Charcot-Marie-Tooth disease type 2U is a subtype of autosomal dominant Charcot-Marie-Tooth disease type 2 characterized by late adult-onset (50-60 years of age) of slowly progressive, axonal, peripheral sensorimotor neuropathy resulting in distal upper limb and proximal and distal lower limb muscle weakness and atrophy, in conjunction with distal, panmodal sensory impairment in upper and lower limbs. Tendon reflexes are reduced and nerve conduction velocities range from reduced to absent. Neuropathic pain has also been associated.