X-linked Charcot-Marie-Tooth disease type 2

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. X-linked Charcot-Marie-Tooth disease type 2. ORPHA:101076

X-linked Charcot-Marie-Tooth disease type 2 is a rare, genetic, peripheral sensorimotor neuropathy characterized by an X-linked recessive inheritance pattern and the infantile- to childhood-onset of progressive, distal muscle weakness and atrophy (more prominent in the lower extremities than in the upper extremities), pes cavus, and absent tendon reflexes. Sensory impairment and intellectual disability has been reported in some individuals.