Autosomal dominant Charcot-Marie-Tooth disease type 2O

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal dominant Charcot-Marie-Tooth disease type 2O. ORPHA:284232

Autosomal dominant Charcot-Marie-Tooth disease type 2O is a rare, genetic, subtype of autosomal dominant Charcot-Marie-Tooth disease type 2 characterized by early childhood-onset of slowly progressive, predominantly distal, lower limb muscle weakness and atrophy, delayed motor development, variable sensory loss, and pes cavus in the presence of normal or near-normal nerve conduction velocities. Additional variable features may include proximal muscle weakness, abnormal gait, arthrogryposis, scoliosis, cognitive impairment, and spasticity.