Autosomal dominant Charcot-Marie-Tooth disease type 2Q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal dominant Charcot-Marie-Tooth disease type 2Q. ORPHA:329258

Autosomal dominant Charcot-Marie-Tooth disease type 2Q is a rare subtype of autosomal dominant Charcot-Marie-Tooth disease type 2 characterized by adolescent to adulthood-onset of symmetrical, slowly progressive distal muscle weakness and atrophy (with a predominant weakness of the distal lower limbs) associated with reduced or absent deep tendon reflexes, pes cavus and mild to moderated deep sensory impairment.