Autosomal dominant Charcot-Marie-Tooth disease type 2Y

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal dominant Charcot-Marie-Tooth disease type 2Y. ORPHA:435387

A rare, axonal hereditary motor and sensory neuropathy characterized by progressive distal muscle weakness and atrophy of variable onset and severity. Patients present with postural instability, gait and running difficulties, decreased deep tendon reflexes, foot deformities, fine motor impairment, and distal sensory impairment. Dysarthria, dysphagia, and mild cognitive and behavioral abnormalities have also been reported.