Charcot-Marie-Tooth disease type 4H

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Charcot-Marie-Tooth disease type 4H. ORPHA:99954

Charcot-Marie-Tooth disease type 4H is a subtype of Charcot-Marie-Tooth disease type 4 characterized by onset before two years of age of severe, slowly progressive, demyelinating sensorimotor neuropathy manifesting with delayed motor development (walking), unsteady gait, distal muscle weakness and atrophy (more prominent in the lower limbs), areflexia, mild symmetrical stocking-distribution hypoesthesia, and skeletal malformations (incl. kyphoscoliosis, short neck, pes cavus and pes equinus). Severely reduced nerve conduction velocities are associated.