Charcot-Marie-Tooth disease type 4A

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Charcot-Marie-Tooth disease type 4A. ORPHA:99948

Charcot-Marie-Tooth disease type 4A (CMT4A) is a subtype of Charcot-Marie-Tooth disease type 4 characterized by early-onset (infancy to early childhood) of severe, rapidly progressing demyelinating, axonal, or intermediate sensorimotor neuropathy usually affecting first, and more severely, the distal lower extremities and later the proximal muscles and upper extremities. Nerve conduction velocities range from very slow to normal. Apart from the typical CMT phenotype (distal muscle weakness and atrophy, sensory loss, frequent pes cavus foot deformity), patients commonly present delayed motor development, vocal cord paresis, mild sensory loss, abolished deep tendon reflexes, and skeletal deformities.