Charcot-Marie-Tooth disease type 4B1

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Charcot-Marie-Tooth disease type 4B1. ORPHA:99955

Charcot-Marie-Tooth disease type 4B1 (CMT4B1) is a subtype of Charcot-Marie-Tooth disease type 4 characterized by an early childhood-onset of severe, demyelinating sensorimotor neuropathy, various degrees of complex myelin outfoldings seen on peripheral nerve biopsy, very slow, and often undetectable, nerve conduction velocities, and the typical CMT phenotype (i.e. distal muscle weakness and atrophy, sensory loss, and frequent pes cavus). Other reported features include facial weakness, vocal cord paresis, respiratory difficulties, and skeletal deformities (e.g. chest deformities, claw hands, pes equinovarus).