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Charcot-Marie-Tooth disease type 4E

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Charcot-Marie-Tooth disease type 4E. ORPHA:99951*

Charcot-Marie-Tooth disease type 4E (CMT4E) is a congenital, hypomyelinating subtype of Charcot-Marie-Tooth disease type 4 characterized by a Dejerine-Sottas syndrome-like phenotype (incl. hypotonia and/or delayed motor development in infancy), extremely slow nerve conduction velocities, potential respiratory dysfunction, cranial nerve involvement, and the typical CMT phenotype, i.e. distal muscle weakness and atrophy, sensory loss, and foot deformity.