Autosomal dominant Charcot-Marie-Tooth disease type 2L

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal dominant Charcot-Marie-Tooth disease type 2L. ORPHA:99945

Autosomal dominant Charcot-Marie-Tooth disease type 2L (CMT2L) is a form of axonal Charcot-Marie-Tooth disease, a peripheral sensorimotor neuropathy. In the single family reported to date, CMT2L onset is between 15 and 33 years. Patients present with a symmetric distal weakness of legs and occasionally of the hands, absent or reduced tendon reflexes, distal legs sensory loss and frequently a pes cavus. Progression is slow.