Autosomal dominant Charcot-Marie-Tooth disease type 2B

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal dominant Charcot-Marie-Tooth disease type 2B. ORPHA:99936

Autosomal dominant Charcot-Marie-Tooth disease type 2B (CMT2B) is a severe form of axonal Charcot-Marie-Tooth disease, a peripheral sensorimotor neuropathy. CMT2B onset, in the 2nd or 3rd decade, is characterized by ulcerations and infections of feet. Symmetric and distal weakness develops mostly in the legs together with a severe symmetric distal sensory loss, tendon reflexes are only reduced at ankles and foot deformities, including pes cavus or planus and hammer toes, appear in childhood.