Autosomal dominant Charcot-Marie-Tooth disease type 2E

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal dominant Charcot-Marie-Tooth disease type 2E. ORPHA:99939

Autosomal dominant Charcot-Marie-Tooth disease type 2E (CMT2E) is a form of axonal Charcot-Marie-Tooth disease, a peripheral sensorimotor neuropathy. CMT2E onset is in the first to 6th decade with a gait anomaly and a leg weakness that reaches the arms secondarily. Tendon reflexes are reduced or absent and, after years, all patients have a pes cavus. Other signs may be present, including hearing loss and postural tremor.