Autosomal dominant Charcot-Marie-Tooth disease type 2M

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal dominant Charcot-Marie-Tooth disease type 2M. ORPHA:228179

Autosomal dominant Charcot-Marie-Tooth disease type 2M (CMT2M) is a form of axonal Charcot-Marie-Tooth disease, a peripheral motor and sensory neuropathy. CMT2M is characterized by congenital pstosis and early cataract associated to a mildly progressive peripheral neuropathy of variable onset from birth to the 6th decade, pes cavus, reduced to absent ankles tendon reflexes and sometimes neutropenia.