Autosomal dominant Charcot-Marie-Tooth disease type 2J

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal dominant Charcot-Marie-Tooth disease type 2J. ORPHA:99943

Autosomal dominant Charcot-Marie-Tooth disease type 2J (CMT2J) is a form of axonal Charcot-Marie-Tooth disease, a peripheral sensorimotor neuropathy, characterized by a relatively late onset, pupillary abnormalities and deafness, in most patients, associated with distal weakness and muscle atrophy.