Charcot-Marie-Tooth disease type 2R

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Charcot-Marie-Tooth disease type 2R. ORPHA:397968

Charcot-Marie-Tooth disease type 2R is a rare subtype of axonal hereditary motor and sensory neuropathy characterized by early-onset axial hypotonia, generalized muscle weakness, absent deep tendon reflexes and decreased muscle mass. Electromyography reveals decreased motor nerve conduction velocities with markedly reduced sensory and motor amplitudes.