Autosomal recessive intermediate Charcot-Marie-Tooth disease type A

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive intermediate Charcot-Marie-Tooth disease type A. ORPHA:217055

Autosomal recessive intermediate Charcot-Marie-Tooth disease type A is a subtype of autosomal recessive intermediate Charcot-Marie-Tooth (CMT) disease characterized by severe, early childhood-onset CMT neuropathy with prominent pes equinovarus deformity and impairment of hand muscles. Nerve conduction velocities usually range between 25-35 m/s and both axonal and demyelinating changes are observed on peripheral nerve pathology.