Charcot-Marie-Tooth disease-deafness-intellectual disability syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Charcot-Marie-Tooth disease-deafness-intellectual disability syndrome. ORPHA:90103

Charcot-Marie-Tooth disease-deafness-intellectual disability syndrome is a rare demyelinating hereditary motor and sensory neuropathy characterized by early-onset, slowly progressive, distal muscular weakness and atrophy with no sensory impairment, congenital sensorineural deafness and mild intellectual disability (with absence of normal speech development). The absence of large myelinated fibers on sural nerve biopsy is equally characteristic of the disease.