# Autosomal dominant intermediate Charcot-Marie-Tooth disease type B

INSERM

## Source

*INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal dominant intermediate Charcot-Marie-Tooth disease type B. ORPHA:100044*

Autosomal dominant intermediate Charcot-Marie-Tooth disease type B is a rare hereditary motor and sensory neuropathy characterized by intermediate motor median nerve conduction velocities (usually between 25 and 45 m/s) and signs of both demyelination and axonal degeneration in nerve biopsies. It presents with mild to moderately severe, slowly progressive usual clinical features of Charcot-Marie-Tooth disease (muscle weakness and atrophy of the distal extremities, distal sensory loss, reduced or absent deep tendon reflexes, and feet deformities). Other findings include asymptomatic neutropenia and early-onset cataracts.