DISEASE:
Autosomal dominant intermediate Charcot-Marie-Tooth disease type B

NAME:	Autosomal dominant intermediate Charcot-Marie-Tooth disease type B
DESCRIPTION:	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.
ORPHACODE:	100044
SYNONYMS:	CMTDIB
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	DNM2
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RELATED CONTENT

Related Genetic Tests

- Charcot-Marie-Tooth (other than type 1A) (gene panel, IPN panel)
- Neuropathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- dynamin 2

Related Gene Panels

- Inherited Peripheral Neuropathies gene panel (139 genes) - KUL
- Neuropathy (148 genes) - IPG