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

NAME:	Autosomal dominant intermediate Charcot-Marie-Tooth disease type D
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 axonal degeneration and demyelination without onion bulbs in nerve biopsies. It presents with usual Charcot-Marie-Tooth disease clinical features of variable severity (progressive muscle weakness and atrophy of the distal extremities, distal sensory loss, reduced or absent deep tendon reflexes, and feet deformities). Other findings in some of the families include debilitating neuropathic pain and mild postural/kinetic upper limb tremor.
ORPHACODE:	100046
SYNONYMS:	CMTDID
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	MPZ
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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

- myelin protein zero

Related Gene Panels

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