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

NAME:	Autosomal dominant intermediate Charcot-Marie-Tooth disease type C
DESCRIPTION:	A rare hereditary motor and sensory neuropathy characterized by intermediate motor median nerve conduction velocities (usually between 25 and 60 m/s). It presents with 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, feet deformities, extensor digitorum brevis atrophy). Findings in nerve biopsies include age-dependent axonal degeneration, reduced number of large myelinated fibres, segmental remyelination, and no onion bulbs.
ORPHACODE:	100045
SYNONYMS:	CMTDIC
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	YARS1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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

- tyrosyl-tRNA synthetase 1

Related Gene Panels

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

Source URL: http://gentest.healthdata.be/disease/3750