DISEASE:
Autosomal dominant Charcot-Marie-Tooth disease type 2I

NAME:	Autosomal dominant Charcot-Marie-Tooth disease type 2I
DESCRIPTION:	A form of axonal Charcot-Marie-Tooth disease, a peripheral sensorimotor neuropathy, characterized by a late onset with severe sensory loss (paresthesia and hypoesthesia) associated with distal weakness, mainly of the legs, and absent or reduced deep tendon reflexes.
ORPHACODE:	99942
SYNONYMS:	CMT2I
XREF(S):	Orphanet ICD-10 OMIM
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