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

NAME:	Autosomal dominant Charcot-Marie-Tooth disease type 2M
DESCRIPTION:	A form of axonal Charcot-Marie-Tooth disease, a peripheral motor and sensory neuropathy, characterized by congenital pstosis and early cataract associated to a mildly progressive peripheral neuropathy of variable onset from birth to the 6th decade, pes cavus, reduced to absent ankles tendon reflexes and sometimes neutropenia.
ORPHACODE:	228179
SYNONYMS:	CMT2M
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	DNM2
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RELATED CONTENT

Related Genetic Tests

- Charcot-Marie-Tooth (other than type 1A) (gene panel, IPN panel)
- Neuromuscular disorders : congenital & distal myopathy, congenital muscle dystrophy / Limb-girdle muscular dystrophy / Rhabdomyolysis / Myopathy (with prominent contractures) / distal artrogryposis (gene panel)
- Neuropathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Brussel VUB
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- dynamin 2

Related Gene Panels

- Inherited Peripheral Neuropathies gene panel (139 genes) - KUL
- Neuromuscular disorders (166 genes) - VUB
- Neuropathy (148 genes) - IPG