DISEASE:
Charcot-Marie-Tooth disease type 2R

NAME:	Charcot-Marie-Tooth disease type 2R
DESCRIPTION:	Charcot-Marie-Tooth disease type 2R is a rare subtype of axonal hereditary motor and sensory neuropathy characterized by early-onset axial hypotonia, generalized muscle weakness, absent deep tendon reflexes and decreased muscle mass. Electromyography reveals decreased motor nerve conduction velocities with markedly reduced sensory and motor amplitudes.
ORPHACODE:	397968
SYNONYMS:	CMT2R
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	TRIM2
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RELATED CONTENT

Related Genetic Tests

- Neuropathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- tripartite motif containing 2

Related Gene Panels

- Neuropathy (148 genes) - IPG