DISEASE:
Charcot-Marie-Tooth disease type 2B2

NAME:	Charcot-Marie-Tooth disease type 2B2
DESCRIPTION:	Charcot-Marie-Tooth disease, type 2B2 (CMT2B2, also referred to as CMT4C3) is an axonal CMT peripheral sensorimotor polyneuropathy that has been described in a large consanguineous Costa Rican family of Spanish ancestry.
ORPHACODE:	101101
SYNONYMS:	AR-CMT2B2 Autosomal recessive axonal CMT4C3 Autosomal recessive axonal Charcot-Marie-Tooth disease type 2B2
XREF(S):	Orphanet MeSH OMIM ICD-10
ANALYTE(S):	PNKP
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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

- polynucleotide kinase 3'-phosphatase

Related Gene Panels

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

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