DISEASE:
Charcot-Marie-Tooth disease type 2B5

NAME:	Charcot-Marie-Tooth disease type 2B5
DESCRIPTION:	A rare axonal hereditary motor and sensory neuropathy characterized by infantile onset of slowly progressive distal motor weakness and atrophy (more severe in legs and moderate in arms) with mildly delayed motor development, hypotonia, and distal sensory impairment of all sensory modalities.
ORPHACODE:	228374
SYNONYMS:	AR-CMT2B5 Autosomal recessive Charcot-Marie-Tooth disease type 2B5 SEOAN due to NEFL deficiency Severe early-onset axonal neuropathy due to NEFL deficiency Severe early-onset axonal neuropathy due to light neurofilament subunit deficiency
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	NEFL
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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

- neurofilament light chain

Related Gene Panels

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