DISEASE:
Autosomal recessive intermediate Charcot-Marie-Tooth disease type B

NAME:	Autosomal recessive intermediate Charcot-Marie-Tooth disease type B
DESCRIPTION:	An extremely rare subtype of autosomal recessive intermediate Charcot-Marie-Tooth (CMT) disease characterized by a CMT neuropathy associated with developmental delay, self-abusive behavior, dysmorphic features and vestibular Schwannoma. Motor nerve conduction velocities demonstrate features of both demyelinating and axonal pathology.
ORPHACODE:	254334
SYNONYMS:	RI-CMT type B
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	KARS1
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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

- lysyl-tRNA synthetase 1

Related Gene Panels

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