DISEASE:
Charcot-Marie-Tooth disease type 4B2

NAME:	Charcot-Marie-Tooth disease type 4B2
DESCRIPTION:	Charcot-Marie-Tooth disease type 4B2 (CMT4B2) is a subtype of Charcot-Marie-Tooth type 4 characterized by a severe, early childhood-onset of demyelinating sensorimotor neuropathy, early-onset glaucoma, focally folded myelin sheaths in the peripheral nerves, severely reduced nerve conduction velocities, and the typical CMT phenotype (i.e. distal muscle weakness and atrophy, sensory loss, and frequent pes cavus). Severe visual impairment leading to visual loss has also been reported.
ORPHACODE:	99956
SYNONYMS:	CMT4B2
XREF(S):	Orphanet MeSH ICD-10 OMIM
ANALYTE(S):	SBF2
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Neuropathy (gene panel)

Related Laboratories

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

Related Analytes

- SET binding factor 2

Related Gene Panels

- Neuropathy (148 genes) - IPG