DISEASE:
Charcot-Marie-Tooth disease type 4C

NAME:	Charcot-Marie-Tooth disease type 4C
DESCRIPTION:	Charcot-Marie-Tooth disease type 4C (CMT4C) is a subtype of Charcot-Marie-Tooth type 4 characterized by childhood or adolescent-onset of a relatively mild, demyelinating sensorimotor neuropathy that contrasts with a severe, rapidly progressing, early-onset scoliosis, and the typical CMT phenotype (i.e. distal muscle weakness and atrophy, sensory loss, and often foot deformity). A wide spectrum of nerve conduction velocities are observed and cranial nerve involvement and kyphoscoliosis have also been reported.
ORPHACODE:	99949
SYNONYMS:	CMT4C
XREF(S):	Orphanet ICD-10 MeSH OMIM
ANALYTE(S):	SH3TC2
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RELATED CONTENT

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

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