
DISEASE:
Charcot-Marie-Tooth disease type 1C

NAME:	Charcot-Marie-Tooth disease type 1C
DESCRIPTION:	A rare, autosomal dominant, hereditary, demyelinating motor and sensory neuropathy which may present either as a classic Charcot-Marie-Tooth disease phenotype with distal motor weakness and wasting, gait difficulties, paresthasias, decreased vibration and pain sensation, or as a milder, predominantly sensory form with transient paresthasias, decreased sensation and distal pain in upper or lower limbs, without significant motor weakness. Pes cavus is a common feature, and additional symptoms may include hand tremor and decreased or absent deep tendon reflexes.
ORPHACODE:	101083
SYNONYMS:	CMT1C
XREF(S):	Orphanet ICD-10 MeSH OMIM
ANALYTE(S):	LITAF
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