DISEASE:
Charcot-Marie-Tooth disease type 4D

NAME:	Charcot-Marie-Tooth disease type 4D
DESCRIPTION:	Charcot-Marie-Tooth disease type 4D (CMT4D) is a subtype of Charcot-Marie-Tooth disease type 4 characterized by a childhood-onset of severe, progressive, demyelinating sensorimotor neuropathy manifesting with distal muscle weakness and atrophy, sensorineural hearing impairment leading to deafness (usually in third decade), severely reduced nerve conduction velocities, and skeletal, especially foot, deformities. Tongue atrophy has also been reported.
ORPHACODE:	99950
SYNONYMS:	CMT4D HMSN, Lom type HMSN-Lom Hereditary motor and sensory neuropathy, Lom type
XREF(S):	Orphanet MeSH OMIM ICD-10
ANALYTE(S):	NDRG1
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