DISEASE:
Autosomal recessive Charcot-Marie-Tooth disease type 2X

NAME:	Autosomal recessive Charcot-Marie-Tooth disease type 2X
DESCRIPTION:	A rare autosomal recessive axonal hereditary motor and sensory neuropathy characterized by childhood to adult onset of slowly progressive, sometimes asymmetric distal muscle weakness and atrophy, as well as sensory impairment, predominantly of the lower limbs. Additional common features include pes cavus, kyphoscoliosis, ankle contractures, tremor, or urogenital dysfunction. Fasciculations and proximal involvement may be seen in some cases. Patients usually remain ambulatory.
ORPHACODE:	466775
SYNONYMS:	ARCMT2X Autosomal recessive Charcot-Marie-Tooth disease type 2 due to SPG11 mutation CMT2X
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	SPG11
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