DISEASE:
X-linked Charcot-Marie-Tooth disease type 5

NAME:	X-linked Charcot-Marie-Tooth disease type 5
DESCRIPTION:	A rare form of X-linked Charcot-Marie-Tooth disease, a peripheral sensorimotor neuropathy, characterized by infancy- to childhood-onset of: 1) progressive distal muscle weakness and atrophy (first appearing and more prominent in the lower extremities than the upper) which usually manifests with foot drop and gait disturbance, 2) bilateral, profound, prelingual sensorineural hearing loss and 3) progressive optic neuropathy.
ORPHACODE:	99014
SYNONYMS:	CMT5X CMTX5
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	PRPS1
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