DISEASE:
Autosomal dominant Charcot-Marie-Tooth disease type 2A2

NAME:	Autosomal dominant Charcot-Marie-Tooth disease type 2A2
DESCRIPTION:	A subtype of Autosomal dominant Charcot-Marie-Tooth disease type 2 characterized by the childhood onset of distal weakness and areflexia (with earlier and more severe involvement of the lower extremities), reduced sensory modalities (primarily pain and temperature sensation), foot deformities, postural tremor, scoliosis and contractures. Optic atrophy, vocal cord palsy with dysphonia, sensorineural hearing loss, spinal cord abnormalities and hydrocephalus have also been reported.
ORPHACODE:	99947
SYNONYMS:	CMT2A2
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	MFN2
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