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

NAME:	Autosomal dominant Charcot-Marie-Tooth disease type 2Z
DESCRIPTION:	A rare autosomal dominant hereditary axonal motor and sensory neuropathy characterized by early onset of generalized hypotonia and weakness, or later onset of distal lower limb muscle weakness and atrophy, cramps, and sensory impairment. Weakness and atrophy progress in an asymmetric fashion to involve also the proximal and upper limbs in the course of the disease. Additional features are pyramidal signs like increased muscle tone and extensor plantar reflexes, as well as learning difficulties.
ORPHACODE:	466768
SYNONYMS:	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to MORC2 mutation CMT2Z
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	MORC2
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