DISEASE:
Autosomal dominant Charcot-Marie-Tooth disease type 2 due to KIF5A mutation

NAME:	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to KIF5A mutation
DESCRIPTION:	A rare form of axonal peripheral sensorimotor neuropathy characterized by classical CMT2 signs and symptoms (progressive weakness and atrophy of distal limb muscles, mild sensory deficits of position, vibration and pain/temperature, pes cavus, and symmetrically absent or reduced muscle and sensory action potentials with relatively preserved nerve conduction velocities in neurophysiological studies) as well as pyramidal tract involvement (spasticity, hyperreflexia). Spasticity and pain may be the presenting symptoms.
ORPHACODE:	324611
SYNONYMS:	CMT2 due to KIF5A mutation
XREF(S):	Orphanet
ANALYTE(S):	KIF5A
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RELATED CONTENT

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