DISEASE:
Autosomal dominant Charcot-Marie-Tooth disease type 2A1

NAME:	Autosomal dominant Charcot-Marie-Tooth disease type 2A1
DESCRIPTION:	A form of axonal Charcot-Marie-Tooth disease, a peripheral sensorimotor neuropathy, presenting with a more prominent muscle weakness in lower than upper limbs and frequent postural tremor.
ORPHACODE:	99946
SYNONYMS:	CMT2A1
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	KIF1B
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Neuropathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- kinesin family member 1B

Related Gene Panels

- Neuropathy (148 genes) - IPG