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

NAME:	Autosomal dominant Charcot-Marie-Tooth disease type 2V
DESCRIPTION:	A rare, axonal hereditary motor and sensory neuropathy characterized by adult onset of recurrent pain in legs with or without cramps, progressive loss of deep tendon reflexes and vibration sense, paresthesias in the feet and later in the hands. Patients often experience sleep disturbances and mild sensory ataxia.
ORPHACODE:	447964
SYNONYMS:	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to NAGLU mutation CMT2V Hereditary adult-onset painful axonal polyneuropathy
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	NAGLU
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RELATED CONTENT

Related Genetic Tests

- Charcot-Marie-Tooth (other than type 1A) (gene panel, IPN panel)
- Neuropathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- N-acetyl-alpha-glucosaminidase

Related Gene Panels

- Inherited Peripheral Neuropathies gene panel (139 genes) - KUL
- Neuropathy (148 genes) - IPG