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

NAME:	Autosomal dominant Charcot-Marie-Tooth disease type 2Y
DESCRIPTION:	A rare, axonal hereditary motor and sensory neuropathy characterized by progressive distal muscle weakness and atrophy of variable onset and severity. Patients present with postural instability, gait and running difficulties, decreased deep tendon reflexes, foot deformities, fine motor impairment, and distal sensory impairment. Dysarthria, dysphagia, and mild cognitive and behavioral abnormalities have also been reported.
ORPHACODE:	435387
SYNONYMS:	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to VCP mutation CMT2 due to VCP mutation CMT2Y
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	VCP
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/2543

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

- valosin containing protein

Related Gene Panels

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