
DISEASE:
Charcot-Marie-Tooth disease type 2T

NAME:	Charcot-Marie-Tooth disease type 2T
DESCRIPTION:	A rare autosomal recessive axonal hereditary motor and sensory neuropathy characterized by adult onset of slowly progressive distal muscle weakness and atrophy, sensory impairment, and decreased or absent deep tendon reflexes predominantly in the lower extremities. Patients present gait disturbances but remain ambulatory. Mild involvement of the upper limbs may be seen.
ORPHACODE:	495274
SYNONYMS:	AR-CMT2T Autosomal recessive axonal Charcot-Marie-Tooth disease type 2T CMT2T
XREF(S):	<u>Orphanet</u> <u>OMIM</u> <u>ICD-10</u>
ANALYTE(S):	<u>MME</u>
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

- [membrane metalloendopeptidase](#)

Related Gene Panels

- [Neuropathy \(148 genes\) - IPG](#)

Source URL: <http://gentest.healthdata.be/disease/2598>