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

NAME:	Autosomal dominant Charcot-Marie-Tooth disease type 2E
DESCRIPTION:	A form of axonal Charcot-Marie-Tooth disease, a peripheral sensorimotor neuropathy, with onset in the first to 6th decade with a gait anomaly and a leg weakness that reaches the arms secondarily. Tendon reflexes are reduced or absent and, after years, all patients have a pes cavus. Other signs may be present, including hearing loss and postural tremor.
ORPHACODE:	99939
SYNONYMS:	CMT2E
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	NEFL
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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

- neurofilament light chain

Related Gene Panels

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