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

NAME:	Autosomal dominant Charcot-Marie-Tooth disease type 2N
DESCRIPTION:	A mild form of axonal Charcot-Marie-Tooth disease, a peripheral sensorimotor neuropathy, characterized by distal legs sensory loss and weakness that can be asymmetric. Tendon reflexes are reduced in the knees and absent in ankles. Progression is slow.
ORPHACODE:	228174
SYNONYMS:	CMT2N
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	AARS1
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)
- Peripheral neuropathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- alanyl-tRNA synthetase 1

Related Gene Panels

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