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

NAME:	Autosomal dominant Charcot-Marie-Tooth disease type 2F
DESCRIPTION:	A form of axonal Charcot-Marie-Tooth disease, a peripheral sensorimotor neuropathy, characterized by symmetric weakness primarily occurring in the lower limbs (distal muscles in a majority of cases) and reaching the arms only after 5 to 10 years, occasional and predominantly distal sensory loss and reduced tendon reflexes. It presents with gait anomaly between the 1st and 6th decade and early onset is generally associated to a more severe phenotype which may include foot drop.
ORPHACODE:	99940
SYNONYMS:	CMT2F
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	HSPB1
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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

- heat shock protein family B (small) member 1

Related Gene Panels

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