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

NAME:	Autosomal dominant Charcot-Marie-Tooth disease type 2L
DESCRIPTION:	A form of axonal Charcot-Marie-Tooth disease, a peripheral sensorimotor neuropathy. In the single family reported to date, CMT2L onset is between 15 and 33 years. Patients present with a symmetric distal weakness of legs and occasionally of the hands, absent or reduced tendon reflexes, distal legs sensory loss and frequently a pes cavus. Progression is slow.
ORPHACODE:	99945
SYNONYMS:	CMT2L
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	HSPB8
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RELATED CONTENT

Related Genetic Tests

- Neuropathy (gene panel)
- Peripheral neuropathy (gene panel)

Related Laboratories

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

Related Analytes

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Related Gene Panels

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- Neuropathy (genepanel) - UZA