DISEASE:
DNAJB2-related Charcot-Marie-Tooth disease type 2

NAME:	DNAJB2-related Charcot-Marie-Tooth disease type 2
DESCRIPTION:	A rare autosomal recessive axonal hereditary motor and sensory neuropathy characterized by adolescent or adult onset of slowly progressive muscle weakness and atrophy of the distal lower limbs progressing to involve also the upper limbs and proximal muscles, and sensory impairment. Patients present gait disturbances and loss of reflexes, at later stages loss of ambulation, dysarthria, dysphagia, facial weakness, and impairment of respiratory muscles requiring assisted ventilation.
ORPHACODE:	443950
SYNONYMS:	DNAJB2-related CMT2
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	DNAJB2
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RELATED CONTENT

Related Genetic Tests

- Neuromuscular disorders : congenital & distal myopathy, congenital muscle dystrophy / Limb-girdle muscular dystrophy / Rhabdomyolysis / Myopathy (with prominent contractures) / distal artrogryposis (gene panel)
- Neuropathy (gene panel)

Related Laboratories

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

Related Analytes

- DnaJ heat shock protein family (Hsp40) member B2

Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB
- Neuropathy (148 genes) - IPG