sciensano **Belgian Genetic Tests database**

DISEASE:
Young adult-onset distal hereditary motor neuropathy

NAME:	Young adult-onset distal hereditary motor neuropathy
DESCRIPTION:	Young adult-onset distal hereditary motor neuropathy is a rare autosomal recessive distal hereditary motor neuropathy characterized by slowly progressive muscular weakness, hypotonia and atrophy of the lower limbs, more pronounced distally, leading to paralysis, and loss of tendon reflexes. Additional features may include pes cavus and mild dysphonia. The upper limbs are relatively spared.
ORPHACODE:	314485
SYNONYMS:	Autosomal recessive distal spinal muscular atrophy type 5 Young adult-onset dHMN dSMA5
XREF(S):	Orphanet OMIM ICD-10 OMIM
ANALYTE(S):	DNAJB2 VWA1
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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
- von Willebrand factor A domain containing 1

Related Gene Panels

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