DISEASE:
Distal hereditary motor neuropathy type 7

NAME:	Distal hereditary motor neuropathy type 7
DESCRIPTION:	A rare, slowly progressive genetic peripheral neuropathy characterized by distal atrophy and weakness affecting the upper limbs (with a predilection for the thenar eminence) and subsequently the lower limbs, associated with uni- or bilateral vocal cord paresis leading to hoarse voice and breathing difficulties, and facial weakness.
ORPHACODE:	139589
SYNONYMS:	Distal spinal muscular atrophy with vocal cord paralysis dHMN7
XREF(S):	Orphanet OMIM OMIM ICD-10
ANALYTE(S):	DCTN1 SLC5A7
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RELATED CONTENT

Related Genetic Tests

- Neurodegeneration (gene panel)
- 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

- dynactin subunit 1
- solute carrier family 5 member 7

Related Gene Panels

- Neurodegeneration (99 genes) - IPG
- Neuromuscular disorders (166 genes) - VUB
- Neuropathy (148 genes) - IPG