DISEASE:
Distal hereditary motor neuropathy, Jerash type

NAME:	Distal hereditary motor neuropathy, Jerash type
DESCRIPTION:	A rare, genetic, neuromuscular disease characterized by progressive, symmetrical, moderate to severe, distal muscle weakness and atrophy, without sensory involvement, first affecting the lower limbs (towards the end of the first decade) and then involving (within two years) the upper extremities. Patients typically develop foot drop, pes varus, hammer toes and claw hands. Pyramidal tract signs (such as brisk knee reflexes and positive Babinski sign) with absent ankle reflexes are initially associated but regress as disease stabilizes (~10 years after onset).
ORPHACODE:	139552
SYNONYMS:	Autosomal recessive distal spinal muscular atrophy type 2 dHMNJ
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	SIGMAR1
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- Neuropathy (gene panel)

Related Laboratories

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

Related Analytes

- sigma non-opioid intracellular receptor 1

Related Gene Panels

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