DISEASE:
X-linked distal spinal muscular atrophy type 3

NAME:	X-linked distal spinal muscular atrophy type 3
DESCRIPTION:	X-linked distal spinal muscular atrophy type 3 is a rare distal hereditary motor neuropathy characterized by slowly progressive atrophy and weakness of distal muscles of hands and feet with normal deep tendon reflexes or absent ankle reflexes and minimal or no sensory loss, sometimes mild proximal weakness in the legs and feet and hand deformities in males.
ORPHACODE:	139557
SYNONYMS:	ATP7A-related distal motor neuropathy DSMAX SMAX3 X-linked dHMN3 X-linked dSMA3 X-linked distal hereditary motor neuropathy type 3
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	ATP7A
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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)
- Occipital horn syndrome / Distal Spinal Muscular atrophy

Related Laboratories

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

Related Analytes

- ATPase copper transporting alpha

Related Gene Panels

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

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