DISEASE:
Proximal spinal muscular atrophy type 3

NAME:	Proximal spinal muscular atrophy type 3
DESCRIPTION:	A rare, genetic proximal spinal muscular atrophy characterized by degeneration of alpha motor neurons in the anterior horns of the spinal cord and lower brain stem manifesting with onset of progressive proximal muscle weakness (legs greater than arms) between 18 months and adulthood. Motor development is heterogeneous but walking is typically acquired.
ORPHACODE:	83419
SYNONYMS:	Juvenile spinal muscular atrophy Kugelberg-Welander disease SMA type 3 SMA type III SMA-III SMA3
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	SMN2 NAIP SMN1

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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)
- Spinal muscular atrophy (SMA) type 1 (Werdnig-Hoffmann), type 2, type 3 (Kugelberg-Welander) and type 4
- Spinal muscular atrophy (SMA) type 1 (Werdnig-Hoffmann), type 2, type 3 (Kugelberg-Welander) and type 4
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- Spinal muscular atrophy (SMA) type 1 (Werdnig-Hoffmann), type 2, type 3 (Kugelberg-Welander) and type 4 (Full sequencing)
- Spinal muscular atrophy (SMA) type 1 (Werdnig-Hoffmann), type 2, type 3 (Kugelberg-Welander) and type 4 (SMN1 & SMN2 genes)

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique Humaine - Erasme ULB
- Centre de Génétique Médicale UCL
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen
- Centrum Medische Genetica - UZ Brussel VUB
- Centrum Medische Genetica - UZ Gent
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- NLR family apoptosis inhibitory protein
- survival of motor neuron 1, telomeric
- survival of motor neuron 2, centromeric

Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB
