DISEASE:
Proximal spinal muscular atrophy type 2

NAME:	Proximal spinal muscular atrophy type 2
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 between 6 to 18 months of age with progressive, proximal muscle weakness, mild to moderate hypotonia and finger polymyoclonour tremor, with areflexia. Motor milestones are classically limited to independent sitting or standing.
ORPHACODE:	83418
SYNONYMS:	Intermediate spinal muscular atrophy SMA type 2 SMA type II SMA-II SMA2
XREF(S):	Orphanet OMIM ICD-10
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
