DISEASE:
Proximal spinal muscular atrophy type 1

NAME:	Proximal spinal muscular atrophy type 1
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 severe and progressive muscle weakness in the first 6 months of life and presenting with severe, generalized hypotonia and weakness,. Dysphagia and respiratory impairment may also be present at presentation or appear at a later stage. Classically, before the advent of recent therapies, type 1 patients never achieved sitting without support.
ORPHACODE:	83330
SYNONYMS:	Infantile spinal muscular atrophy Infantile-onset spinal muscular atrophy SMA type 1 SMA type I SMA-I SMA1 Werdnig-Hoffmann disease
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
