DISEASE:
Autosomal recessive lower motor neuron disease with childhood onset

NAME:	Autosomal recessive lower motor neuron disease with childhood onset
DESCRIPTION:	A rare, genetic, neuromuscular disease characterized by proximal muscle weakness with an early involvement of foot and hand muscles following normal motor development in early childhood, a rapidly progressive disease course leading to generalized areflexic tetraplegia with contractures, severe scoliosis, hyperlordosis, and progressive respiratory insufficiency leading to assisted ventilation. Cranial nerve functions are normal and tongue wasting and fasciculations are absent. Milder phenotype with a moderate generalized weakness and slower disease progress was reported.
ORPHACODE:	206580
SYNONYMS:	Autosomal recessive distal spinal muscular atrophy type 4 Distal spinal muscular atrophy type 4 dSMA4
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	PLEKHG5
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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)

Related Laboratories

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

Related Analytes

- pleckstrin homology and RhoGEF domain containing G5

Related Gene Panels

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