DISEASE:
Autosomal dominant adult-onset proximal spinal muscular atrophy

NAME:	Autosomal dominant adult-onset proximal spinal muscular atrophy
DESCRIPTION:	A rare, genetic, motor neuron disease characterized by adulthood-onset of slowly progressive, proximal muscular weakness with fasciculations, amyotrophy, cramps, and absent/hypoactive reflexes, without bulbar or pyramidal involvement.
ORPHACODE:	209335
SYNONYMS:	Autosomal dominant adult-onset proximal SMA Autosomal dominant late-onset spinal muscular atrophy, Finkel type Finkel disease SMAFK
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	VAPB
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RELATED CONTENT

Related Genetic Tests

- Neurodegeneration (gene panel)
- 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

- VAMP associated protein B and C

Related Gene Panels

- Neurodegeneration (99 genes) - IPG
- Neuromuscular disorders (166 genes) - VUB
- Neuropathy (148 genes) - IPG