DISEASE:
Spinal muscular atrophy-progressive myoclonic epilepsy syndrome

NAME:	Spinal muscular atrophy-progressive myoclonic epilepsy syndrome
DESCRIPTION:	Spinal muscular atrophy-progressive myoclonic epilepsy syndrome is characterized by hereditary myoclonus and progressive distal muscular atrophy. Less than 10 cases have been reported. Treatment with clonazepam results in complete and lasting improvement of the myoclonus.
ORPHACODE:	2590
SYNONYMS:	Hereditary myoclonus-progressive distal muscular atrophy syndrome Jankovic-Rivera syndrome SMA-PME
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ASAH1
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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)

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- N-acylsphingosine amidohydrolase 1

Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB