DISEASE:
Neurogenic scapuloperoneal syndrome, Kaeser type

NAME:	Neurogenic scapuloperoneal syndrome, Kaeser type
DESCRIPTION:	A rare, genetic, neuromuscular disease characterized by adult-onset muscle weakness and atrophy in a scapuloperoneal distribution, mild involvement of the facial muscles, dysphagia, and gynecomastia. Elevated serum CK levels and mixed myopathic and neurogenic abnormalities are associated clinical findings.
ORPHACODE:	85146
SYNONYMS:	Kaeser syndrome Stark-Kaeser syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	DES
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

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- Neuromuscular disorders : congenital & distal myopathy, congenital muscle dystrophy / Limb-girdle muscular dystrophy / Rhabdomyolysis / Myopathy (with prominent contractures) / distal artrogryposis (gene panel)

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- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

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