DISEASE:
Myosclerosis

NAME:	Myosclerosis
DESCRIPTION:	Myosclerosis is a rare, genetic, non-dystrophic myopathy characterized by early, diffuse, progressive muscle and joint contractures that result in severe limitation of movement of axial, proximal, and distal joints, walking difficulties in early childhood and toe walking. Patients typically present thin, sclerotic muscles with a woody consistency, mild girdle and proximal limb weakness with moderate distal weakness and scoliosis. Muscle biopsy shows partial collagen VI deficiency at the myofiber basement membrane and absent collagen VI around most endomysial/perimysial capillaries.
ORPHACODE:	289380
SYNONYMS:	Congenital myosclerosis, Löwenthal type
XREF(S):	Orphanet MedDRA OMIM ICD-10
ANALYTE(S):	COL6A2
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RELATED CONTENT

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- Bethlem myopathy / Ullrich congenital muscular dystrophy / Myosclerosis Myopathy
- 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
- Centrum Medische Genetica - UZ Gent

Related Analytes

- collagen type VI alpha 2 chain

Related Gene Panels

- Bethlem myopathy / Ullrich / Myosclerosis Myopathy - UGent
- Neuromuscular disorders (166 genes) - VUB

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