DISEASE:
Lethal multiple pterygium syndrome

NAME:	Lethal multiple pterygium syndrome
DESCRIPTION:	A rare genetic multiple pterygium syndrome characterized by intrauterine growth retardation, fetal akinesia, multiple joint contractures causing severe arthrogryposis and pterygia (webbing) across multiple joints. Cystic hygroma and/or fetal hydrops are almost invariably present.
ORPHACODE:	33108
SYNONYMS:	Autosomal recessive lethal multiple pterygium syndrome LMPS
XREF(S):	Orphanet ICD-10 OMIM MeSH
ANALYTE(S):	NEB RAPSN RYR1 CHRNA1 CHRND CHRNG
CREATED:	13 May 2019 - 01:02

CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/index.php/index.php/disease/2961

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

- cholinergic receptor nicotinic alpha 1 subunit
- cholinergic receptor nicotinic delta subunit
- cholinergic receptor nicotinic gamma subunit
- nebulin
- receptor associated protein of the synapse
- ryanodine receptor 1

Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB

Source URL: http://gentest.healthdata.be/index.php/index.php/disease/2961