DISEASE:
Congenital fiber-type disproportion myopathy

NAME:	Congenital fiber-type disproportion myopathy
DESCRIPTION:	A rare genetic, congenital, non-dystrophic myopathy characterized by neonatal or infantile-onset hypotonia and mild to severe generalized muscle weakness.
ORPHACODE:	2020
SYNONYMS:	CFTDM
XREF(S):	Orphanet ICD-10 OMIM OMIM OMIM
ANALYTE(S):	MAP3K20 ACTA1 SELENON TPM2 TPM3 MYL2 ITGA7 HACD1

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RELATED CONTENT

Related Genetic Tests

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

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Brussel VUB
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- actin alpha 1, skeletal muscle
- 3-hydroxyacyl-CoA dehydratase 1
- integrin subunit alpha 7
- mitogen-activated protein kinase kinase kinase 20
- myosin light chain 2
- selenoprotein N
- tropomyosin 2
- tropomyosin 3

Related Gene Panels

- Cardiomyopathy, hereditary (208 genes) - VUB
- Cardiopathies, hereditary (102 genes) - KUL
- Hypertrophic cardiomyopathy (75 genes) - IPG
- Neuromuscular disorders (166 genes) - VUB
