DISEASE:
Congenital muscular dystrophy without intellectual disability

NAME:	Congenital muscular dystrophy without intellectual disability
DESCRIPTION:	Congenital muscular dystrophy without intellectual disability is a rare, genetic, congenital muscular dystrophy due to dystroglycanopathy disorder characterized by a wide phenotypic spectrum which includes hypotonia and muscular weakness present at birth or early infancy, delayed or arrested motor development, and normal intellectual abilities with normal (or only mild abnormalities) neuroimaging studies. Feeding difficulties, joint and spinal deformities, and respiratory insufficiency may be associated. Decreased alpha-dystroglycan on immunohistochemical muscle staining and elevated serum creatine kinase are observed.
ORPHACODE:	370980
SYNONYMS:	CMD without intellectual disability CMD-no MR Congenital muscular dystrophy-dystroglycanopathy without intellectual disability
XREF(S):	Orphanet ICD-10 OMIM OMIM

ANALYTE(S):	POMT1 FKTN FKRP CRPPA
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/1803

RELATED CONTENT

Related Genetic Tests

- Congenital disorders of glycosylation (79 genes)
- 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 Menselijke Erfelijkheid - KUL

Related Analytes

- CDP-L-ribitol pyrophosphorylase A
- fukutin related protein
- fukutin
- protein O-mannosyltransferase 1

Related Gene Panels

- Congenital disorders of glycosylation (79 genes) - KUL
- Neuromuscular disorders (166 genes) - VUB