DISEASE:
Congenital muscular dystrophy with intellectual disability

NAME:	Congenital muscular dystrophy with intellectual disability
DESCRIPTION:	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 and delayed or arrested motor development, associated with mild to severe intellectual disability and variable brain abnormalities on neuroimaging studies. Feeding difficulties, joint and spinal deformities, respiratory insufficiency, and ocular anomalies (e.g. strabismus, retinal dystrophy, oculomotor apraxia) may be associated. Decreased or absent alpha-dystroglycan on immunohistochemical muscle staining and elevated serum creatine kinase are observed.
ORPHACODE:	370968
SYNONYMS:	CMD with intellectual disability CMD-MR

XREF(S):	Orphanet OMIM OMIM OMIM OMIM OMIM OMIM ICD-10
ANALYTE(S):	POMT1 POMT2 FKRP LARGE1 GMPPB
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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

- fukutin related protein
- GDP-mannose pyrophosphorylase B
- LARGE xylosyl- and glucuronyltransferase 1
- protein O-mannosyltransferase 1
- protein O-mannosyltransferase 2

Related Gene Panels

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

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