DISEASE:
Congenital muscular dystrophy with cerebellar involvement

NAME:	Congenital muscular dystrophy with cerebellar involvement
DESCRIPTION:	A rare, congenital muscular dystrophy due to dystroglycanopathy characterized by proximal muscle weakness with a tendency for muscle hypertrophy and pseudohypertrophy, variable cognitive impairment, microcephaly, cerebellar hypoplasia with or without cysts, and other structural brain anomalies.
ORPHACODE:	370959
SYNONYMS:	CMD with cerebellar involvement CMD-CRB
XREF(S):	Orphanet ICD-10 OMIM OMIM OMIM OMIM OMIM

ANALYTE(S):	POMGNT1 POMT1 POMT2 FKRP POMK GMPPB
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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

- fukutin related protein
- GDP-mannose pyrophosphorylase B
- protein O-linked mannose N-acetylglucosaminyltransferase 1 (beta 1,2-)
- protein O-mannose kinase
- protein O-mannosyltransferase 1
- protein O-mannosyltransferase 2

Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB