DISEASE:
Muscle-eye-brain disease

NAME:	Muscle-eye-brain disease
DESCRIPTION:	A rare, congenital muscular dystrophy due to dystroglycanopathy characterized by early onset muscular dystrophy, severe muscular hypotonia, severe intellectual disability and typical brain and eye malformations including pachygyria, polymicrogyria, agyria, brainstem and cerebellar structural anomalies, severe myopia, glaucoma, optic nerve and retinal hypoplasia. A broad clinical spectrum is observed with variable involvement of each organ system.
ORPHACODE:	588
SYNONYMS:	MEB syndrome Muscle-eye-brain syndrome Santavuori congenital muscular dystrophy

XREF(S):	Orphanet ICD-10 OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM
ANALYTE(S):	CRPPA LARGE1 POMGNT1 POMT1 POMT2 FKTN FKRP B3GALNT2 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

- beta-1,3-N-acetylgalactosaminyltransferase 2
- CDP-L-ribitol pyrophosphorylase A
- fukutin related protein
- fukutin
- GDP-mannose pyrophosphorylase B
- LARGE xylosyl- and glucuronyltransferase 1
- protein O-linked mannose N-acetylglucosaminyltransferase 1 (beta 1,2-)
- protein O-mannosyltransferase 1
- protein O-mannosyltransferase 2

Related Gene Panels

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