DISEASE:
Walker-Warburg syndrome

NAME:	Walker-Warburg syndrome
DESCRIPTION:	A rare form of congenital muscular dystrophy (CMD) associated with severe brain and eye abnormalities. It is the most severe form of CMD.
ORPHACODE:	899
SYNONYMS:	HARD syndrome Hydrocephalus-agyria-retinal dysplasia syndrome WWS

XREF(S):	Orphanet OMIM MeSH OMIM OMIM OMIM OMIM OMIM OMIM ICD-10 OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM
ANALYTE(S):	POMGNT1 POMT1 POMT2 COL4A1 FKTN FKRP LARGE1 DAG1 CRPPA POMGNT2 RXYLT1 B3GALNT2 B4GAT1 POMK

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RELATED CONTENT

Related Genetic Tests

- Congenital disorders of glycosylation (79 genes)
- Epilepsy (gene panel)
- Glaucoma (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

- Centrum Medische Genetica - UZ Antwerpen
- Centrum Medische Genetica - UZ Brussel VUB
- Centrum Medische Genetica - UZ Gent
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- beta-1,3-N-acetylgalactosaminyltransferase 2
- beta-1,4-glucuronyltransferase 1
- collagen type IV alpha 1 chain
- CDP-L-ribitol pyrophosphorylase A
- dystroglycan 1
- fukutin related protein
- fukutin
- LARGE xylosyl- and glucuronyltransferase 1

- protein O-linked mannose N-acetylglucosaminyltransferase 1 (beta 1,2-)
- protein O-linked mannose N-acetylglucosaminyltransferase 2 (beta 1,4-)
- protein O-mannose kinase
- protein O-mannosyltransferase 1
- protein O-mannosyltransferase 2
- ribitol xylosyltransferase 1

Related Gene Panels

- Congenital disorders of glycosylation (79 genes) - KUL
- Glaucoma - UGent
- Neuromuscular disorders (166 genes) - VUB
- Rare epilepsy with developmental delay (> 240 genes) - UZA
