DISEASE:
Congenital myasthenic syndromes with glycosylation defect

NAME:	Congenital myasthenic syndromes with glycosylation defect
ORPHACODE:	353327
XREF(S):	Orphanet OMIM OMIM OMIM OMIM ICD-10
ANALYTE(S):	ALG2 DPAGT1 GFPT1 ALG14 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

- ALG14 UDP-N-acetylglucosaminyltransferase subunit
- ALG2 alpha-1,3/1,6-mannosyltransferase
- dolichyl-phosphate N-acetylglucosaminephosphotransferase 1
- glutamine--fructose-6-phosphate transaminase 1
- GDP-mannose pyrophosphorylase B

Related Gene Panels

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

Source URL: http://gentest.healthdata.be/disease/1910