

Full name:	Congenital disorders of glycosylation (79 genes) - KUL
Version number:	CDG-v5
Laboratory:	<u>Centrum Menselijke Erfelijkheid - KUL</u>
Created:	18 Jul 2019 - 12:12
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Related Diseases

- ALG1-CDG
- ALG11-CDG
- ALG12-CDG
- ALG13-CDG
- ALG2-CDG
- ALG3-CDG
- ALG6-CDG
- ALG8-CDG
- ALG9-CDG
- Amish infantile epilepsy syndrome
- Autism spectrum disorder-epilepsy-arthrogryposis syndrome
- Autosomal recessive non-syndromic intellectual disability
- B4GALT1-CDG
- COG1-CDG
- COG2-CDG
- COG4-CDG
- COG5-CDG
- COG6-CGD
- COG7-CDG
- COG8-CDG
- Classic galactosemia
- Congenital dyserythropoietic anemia type III
- Congenital muscular dystrophy with intellectual disability

- Congenital muscular dystrophy with intellectual disability and severe epilepsy
- Congenital muscular dystrophy without intellectual disability
- Congenital muscular dystrophy, Fukuyama type
- Congenital myasthenic syndromes with glycosylation defect
- DDOST-CDG
- DK1-CDG
- DPAGT1-CDG
- DPM1-CDG
- DPM3-CDG
- Erythrocyte galactose epimerase deficiency
- Ferro-cerebro-cutaneous syndrome
- Fryns syndrome
- Galactokinase deficiency
- Generalized galactose epimerase deficiency
- Hereditary fructose intolerance
- Hypercoagulability syndrome due to glycosylphosphatidylinositol deficiency
- Hyperphosphatasia-intellectual disability syndrome
- Infantile spasms syndrome
- Leukocyte adhesion deficiency type II
- MAN1B1-CDG
- MGAT2-CDG
- MOGS-CDG
- MPDU1-CDG
- MPI-CDG
- Multiple congenital anomalies-hypotonia-seizures syndrome type 2
- Muscle-eye-brain disease
- Non-specific early-onset epileptic encephalopathy
- PGM1-CDG
- PGM3-CDG
- PMM2-CDG
- Peters plus syndrome
- RFT1-CDG
- SLC35A1-CDG
- SRD5A3-CDG
- STT3A-CDG
- STT3B-CDG

- [Salt-and-pepper syndrome](#)
- [Schneckenbecken dysplasia](#)
- [Sialuria](#)
- [TMEM165-CDG](#)
- [Triple A syndrome](#)
- [Walker-Warburg syndrome](#)
- [X-linked immunodeficiency with magnesium defect, Epstein-Barr virus infection and neoplasia](#)

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
ALDOB	95.00	0	NM_000035.3/ interpretable range CS1>95%
ALG1	95.00	0	NM_019109.4/ interpretable range CS1>95%
ALG10	95.00	0	NM_032834.3/ interpretable range CS1>95%
ALG11	95.00	0	NM_001004127.2/ interpretable range CS1>95%
ALG12	95.00	0	NM_024105.3/ interpretable range CS1>95%
ALG13	95.00	0	NM_001099922.2/ interpretable range CS1>95%
ALG14	95.00	0	NM_144988.3/ interpretable range CS1>95%
ALG2	95.00	0	NM_033087.3/ interpretable range CS1>95%
ALG3	95.00	0	NM_005787.5/ interpretable range CS1>95%
ALG5	95.00	0	NM_013338.4/ interpretable range CS1>95%
ALG6	95.00	0	NM_013339.3/ interpretable range CS1>95%
ALG8	95.00	0	NM_024079.4/ interpretable range CS1>95%
ALG9	95.00	0	NM_024740.2/ interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ATP6V0A2</u>	95.00	0	NM_012463.3/ interpretable range CS1>95%
<u>ATP9B</u>	95.00	0	NM_198531.4/ interpretable range CS1>95%
<u>B3GLCT</u>	95.00	0	NM_194318.3/ interpretable range CS1>95%
<u>B4GALT1</u>	95.00	0	NM_001497.3/ interpretable range CS1>95%
<u>COG1</u>	95.00	0	NM_018714.2/ interpretable range CS1>95%
<u>COG2</u>	95.00	0	NM_007357.2/ interpretable range CS1>95%
<u>COG3</u>	95.00	0	NM_031431.3/ interpretable range CS1>95%
<u>COG4</u>	95.00	0	NM_015386.2/ interpretable range CS1>95%
<u>COG5</u>	95.00	0	NM_006348.3/ interpretable range CS1>95%
<u>COG6</u>	95.00	0	NM_020751.2/ interpretable range CS1>95%
<u>COG7</u>	95.00	0	NM_153603.3/ interpretable range CS1>95%
<u>COG8</u>	95.00	0	NM_032382.4/ interpretable range CS1>95%
<u>DAD1</u>	95.00	0	NM_001344.3/ interpretable range CS1>95%
<u>DDOST</u>	95.00	0	NM_005216.4/ interpretable range CS1>95%
<u>DHDDS</u>	95.00	0	NM_024887.3/ interpretable range CS1>95%
<u>DOLK</u>	95.00	0	NM_014908.3/ interpretable range CS1>95%
<u>DPAGT1</u>	95.00	0	NM_001382.3/ interpretable range CS1>95%
<u>DPM1</u>	95.00	0	NM_003859.2/ interpretable range CS1>95%
<u>DPM2</u>	95.00	0	NM_003863.3/ interpretable range CS1>95%
<u>DPM3</u>	95.00	0	NM_153741.1/ interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>FKRP</u>	95.00	0	NM_024301.4/ interpretable range CS1>95%
<u>FKTN</u>	95.00	0	NM_001079802.1/ interpretable range CS1>95%
<u>FUT1</u>	95.00	0	NM_000148.3/ interpretable range CS1>95%
<u>GALE</u>	95.00	0	NM_000403.3/ interpretable range CS1>95%
<u>GALK1</u>	95.00	0	NM_000154.1/ interpretable range CS1>95%
<u>GALT</u>	95.00	0	NM_000155.3/ interpretable range CS1>95%
<u>GFPT1</u>	95.00	0	NM_002056.3/ interpretable range CS1>95%
<u>GMPPA</u>	95.00	0	NM_205847.2/ interpretable range CS1>95%
<u>GMPPB</u>	95.00	0	NM_013334.3/ interpretable range CS1>95%
<u>GNE</u>	95.00	0	NM_001128227.2/ interpretable range CS1>95%
<u>LARGE1</u>	95.00	0	NM_004737.6/ interpretable range CS1>95%
<u>MAGT1</u>	95.00	0	NM_032121.5/ interpretable range CS1>95%
<u>MAN1B1</u>	95.00	0	NM_016219.4/ interpretable range CS1>95%
<u>MGAT1</u>	95.00	0	NM_001114618.1/ interpretable range CS1>95%
<u>MGAT2</u>	95.00	0	NM_002408.3/ interpretable range CS1>95%
<u>MOGS</u>	95.00	0	NM_006302.2/ interpretable range CS1>95%
<u>MPDU1</u>	95.00	0	NM_004870.3/ interpretable range CS1>95%
<u>MPI</u>	95.00	0	NM_002435.2/ interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>OST4</u>	95.00	0	NM_001134693.1/ interpretable range CS1>95%
<u>PGM1</u>	95.00	0	NM_002633.2/ interpretable range CS1>95%
<u>PGM2</u>	95.00	0	NM_018290.3/ interpretable range CS1>95%
<u>PGM3</u>	95.00	0	NM_001199917.1/ interpretable range CS1>95%
<u>PIGA</u>	95.00	0	NM_002641.3/ interpretable range CS1>95%
<u>PIGL</u>	95.00	0	NM_004278.3/ interpretable range CS1>95%
<u>PIGM</u>	95.00	0	NM_145167.2/ interpretable range CS1>95%
<u>PIGN</u>	95.00	0	NM_176787.4/ interpretable range CS1>95%
<u>PIGV</u>	95.00	0	NM_017837.3/ interpretable range CS1>95%
<u>PMM2</u>	95.00	0	NM_000303.2/ interpretable range CS1>95%
<u>POMGNT1</u>	95.00	0	NM_017739.3/ interpretable range CS1>95%
<u>POMT1</u>	95.00	0	NM_007171.3/ interpretable range CS1>95%
<u>POMT2</u>	95.00	0	NM_013382.5/ interpretable range CS1>95%
<u>RFT1</u>	95.00	0	NM_052859.3/ interpretable range CS1>95%
<u>RPN1</u>	95.00	0	NM_002950.3/ interpretable range CS1>95%
<u>RPN2</u>	95.00	0	NM_002951.4/ interpretable range CS1>95%
<u>SEC23B</u>	95.00	0	NM_032985.5/ interpretable range CS1>95%
<u>SLC35A1</u>	95.00	0	NM_006416.4/ interpretable range CS1>95%
<u>SLC35A3</u>	95.00	0	NM_012243.2/ interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>SLC35C1</u>	95.00	0	NM_018389.4/ interpretable range CS1>95%
<u>SLC35D1</u>	95.00	0	NM_015139.2/ interpretable range CS1>95%
<u>SRD5A3</u>	95.00	0	NM_024592.4/ interpretable range CS1>95%
<u>ST3GAL3</u>	95.00	0	NM_174963.4/ interpretable range CS1>95%
<u>ST3GAL5</u>	95.00	0	NM_003896.3/ interpretable range CS1>95%
<u>STT3A</u>	95.00	0	NM_152713.4/ interpretable range CS1>95%
<u>STT3B</u>	95.00	0	NM_178862.2/ interpretable range CS1>95%
<u>TMEM165</u>	95.00	0	NM_018475.4/ interpretable range CS1>95%
<u>TUSC3</u>	95.00	0	NM_006765.3/ interpretable range CS1>95%