

Full name:	Metabolic disorders (213 genes) - VUB
Abbreviation:	MD
Type of panel:	<u>Custom panel</u>
Version number:	V11
Laboratory:	<u>Centrum Medische Genetica - UZ Brussel VUB</u>
Created:	16 Jul 2019 - 11:41
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Related Diseases

- 3-hydroxy-3-methylglutaric aciduria
- Argininosuccinic aciduria
- Congenital disorder of glycosylation
- Glycogen storage disease due to glycogen debranching enzyme deficiency
- Hereditary orotic aciduria
- Malonic aciduria
- Methylmalonic aciduria due to transcobalamin receptor defect
- Mevalonic aciduria
- Oxoglutaric aciduria
- Peroxisomal acyl-CoA oxidase deficiency
- Urocanic aciduria
- Vitamin B12-responsive methylmalonic acidemia type cblB

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ABAT</u>	100.00	0	No comment
<u>ABCD1</u>	99.93	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ACAT1</u>	97.28	0	No comment
<u>ACOX1</u>	100.00	0	No comment
<u>AGL</u>	99.98	0	No comment
<u>AGPS</u>	100.00	0	No comment
<u>AGXT</u>	100.00	0	No comment
<u>ALDH18A1</u>	100.00	0	No comment
<u>ALDH7A1</u>	100.00	0	No comment
<u>ALDOA</u>	100.00	0	No comment
<u>ALDOB</u>	100.00	0	No comment
<u>ALG1</u>	92.65	0	No comment
<u>ALG10</u>	100.00	0	No comment
<u>ALG11</u>	100.00	0	No comment
<u>ALG12</u>	100.00	0	No comment
<u>ALG13</u>	99.96	0	No comment
<u>ALG14</u>	100.00	0	No comment
<u>ALG2</u>	99.94	0	No comment
<u>ALG3</u>	99.22	0	No comment
<u>ALG5</u>	99.99	0	No comment
<u>ALG6</u>	100.00	0	No comment
<u>ALG8</u>	95.91	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ALG9</u>	98.65	0	No comment
<u>AMACR</u>	99.83	0	No comment
<u>AMT</u>	99.94	0	No comment
<u>ASPA</u>	100.00	0	No comment
<u>ATP6AP1</u>	99.51	0	No comment
<u>ATP6V0A2</u>	100.00	0	No comment
<u>ATP6V1A</u>	100.00	0	No comment
<u>ATP6V1E1</u>	100.00	0	No comment
<u>ATP7A</u>	100.00	0	No comment
<u>ATP9B</u>	99.97	0	No comment
<u>B3GALNT2</u>	85.88	0	No comment
<u>B3GALT6</u>	56.12	0	No comment
<u>B3GAT3</u>	95.56	0	No comment
<u>B3GLCT</u>	94.89	0	No comment
<u>B4GALNT1</u>	100.00	0	No comment
<u>B4GALT1</u>	100.00	0	No comment
<u>B4GALT7</u>	93.23	0	No comment
<u>BCKDHA</u>	100.00	0	No comment
<u>BCKDHB</u>	100.00	0	No comment
<u>BTD</u>	100.00	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>CAD</u>	100.00	0	No comment
<u>CCDC115</u>	100.00	0	No comment
<u>CHST14</u>	98.57	0	No comment
<u>CHST3</u>	100.00	0	No comment
<u>CHST6</u>	100.00	0	No comment
<u>CHSY1</u>	91.67	0	No comment
<u>COG1</u>	99.24	0	No comment
<u>COG2</u>	100.00	0	No comment
<u>COG3</u>	100.00	0	No comment
<u>COG4</u>	100.00	0	No comment
<u>COG5</u>	100.00	0	No comment
<u>COG6</u>	100.00	0	No comment
<u>COG7</u>	100.00	0	No comment
<u>COG8</u>	100.00	0	No comment
<u>CRPPA</u>	95.75	0	No comment
<u>CTH</u>	100.00	0	No comment
<u>D2HGDH</u>	100.00	0	No comment
<u>DAD1</u>	100.00	0	No comment
<u>DBH</u>	100.00	0	No comment
<u>DBT</u>	100.00	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>DDC</u>	100.00	0	No comment
<u>DDOST</u>	100.00	0	No comment
<u>DHDDS</u>	100.00	0	No comment
<u>DNM1L</u>	100.00	0	No comment
<u>DOLK</u>	100.00	0	No comment
<u>DPAGT1</u>	100.00	0	No comment
<u>DPM1</u>	100.00	0	No comment
<u>DPM2</u>	100.00	0	No comment
<u>DPM3</u>	100.00	0	No comment
<u>ENO3</u>	100.00	0	No comment
<u>EOGT</u>	100.00	0	No comment
<u>EXT1</u>	100.00	0	No comment
<u>EXT2</u>	100.00	0	No comment
<u>FANCA</u>	100.00	0	No comment
<u>FKRP</u>	98.68	0	No comment
<u>FKTN</u>	100.00	0	No comment
<u>FUT1</u>	100.00	0	No comment
<u>G6PC1</u>	100.00	0	No comment
<u>GAA</u>	100.00	0	No comment
<u>GABRG2</u>	92.03	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>GALE</u>	100.00	0	No comment
<u>GALK1</u>	99.95	0	No comment
<u>GALNT3</u>	100.00	0	No comment
<u>GALT</u>	100.00	0	No comment
<u>GAMT</u>	98.60	0	No comment
<u>GANAB</u>	100.00	0	No comment
<u>GATM</u>	99.26	0	No comment
<u>GBE1</u>	100.00	0	No comment
<u>GCDH</u>	100.00	0	No comment
<u>GCH1</u>	96.30	0	No comment
<u>GCSH</u>	76.04	0	No comment
<u>GFPT1</u>	99.98	0	No comment
<u>GLDC</u>	96.53	0	No comment
<u>GMPPA</u>	100.00	0	No comment
<u>GMPPB</u>	100.00	0	No comment
<u>GNE</u>	100.00	0	No comment
<u>GNMT</u>	100.00	0	No comment
<u>GNPAT</u>	100.00	0	No comment
<u>GYS1</u>	100.00	0	No comment
<u>GYS2</u>	100.00	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>HGD</u>	100.00	0	No comment
<u>HLCS</u>	100.00	0	No comment
<u>HMGCL</u>	100.00	0	No comment
<u>HPRT1</u>	94.60	0	No comment
<u>HSD17B4</u>	100.00	0	No comment
<u>IVD</u>	100.00	0	No comment
<u>LAMP2</u>	100.00	0	No comment
<u>LARGE1</u>	100.00	0	No comment
<u>LDHA</u>	100.00	0	No comment
<u>LFNG</u>	82.55	0	No comment
<u>MAGT1</u>	100.00	0	No comment
<u>MAN1B1</u>	99.99	0	No comment
<u>MAOA</u>	99.95	0	No comment
<u>MCCC1</u>	100.00	0	No comment
<u>MCCC2</u>	99.99	0	No comment
<u>MGAT2</u>	100.00	0	No comment
<u>MMAA</u>	100.00	0	No comment
<u>MMAB</u>	100.00	0	No comment
<u>MMACHC</u>	100.00	0	No comment
<u>MMUT</u>	100.00	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>MOGS</u>	98.73	0	No comment
<u>MPDU1</u>	100.00	0	No comment
<u>MPI</u>	100.00	0	No comment
<u>NANS</u>	100.00	0	No comment
<u>NGLY1</u>	99.98	0	No comment
<u>NUS1</u>	100.00	0	No comment
<u>OAT</u>	100.00	0	No comment
<u>OST4</u>	100.00	0	No comment
<u>PCCA</u>	100.00	0	No comment
<u>PCCB</u>	99.98	0	No comment
<u>PEX1</u>	100.00	0	No comment
<u>PEX10</u>	88.47	0	No comment
<u>PEX11B</u>	100.00	0	No comment
<u>PEX12</u>	100.00	0	No comment
<u>PEX13</u>	100.00	0	No comment
<u>PEX14</u>	100.00	0	No comment
<u>PEX16</u>	100.00	0	No comment
<u>PEX19</u>	100.00	0	No comment
<u>PEX2</u>	100.00	0	No comment
<u>PEX26</u>	100.00	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>PEX3</u>	100.00	0	No comment
<u>PEX5</u>	100.00	0	No comment
<u>PEX6</u>	98.56	0	No comment
<u>PEX7</u>	91.13	0	No comment
<u>PFKM</u>	100.00	0	No comment
<u>PGAM2</u>	100.00	0	No comment
<u>PGAP1</u>	100.00	0	No comment
<u>PGAP2</u>	100.00	0	No comment
<u>PGAP3</u>	99.90	0	No comment
<u>PGM1</u>	100.00	0	No comment
<u>PGM2</u>	100.00	0	No comment
<u>PGM3</u>	99.99	0	No comment
<u>PHGDH</u>	100.00	0	No comment
<u>PHKA1</u>	100.00	0	No comment
<u>PHKA2</u>	100.00	0	No comment
<u>PHKB</u>	100.00	0	No comment
<u>PHKG2</u>	100.00	0	No comment
<u>PIGA</u>	100.00	0	No comment
<u>PIGC</u>	100.00	0	No comment
<u>PIGG</u>	100.00	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>PIGL</u>	100.00	0	No comment
<u>PIGM</u>	100.00	0	No comment
<u>PIGN</u>	100.00	0	No comment
<u>PIGO</u>	100.00	0	No comment
<u>PIGV</u>	100.00	0	No comment
<u>PIGW</u>	100.00	0	No comment
<u>PIGY</u>	100.00	0	No comment
<u>PMM2</u>	100.00	0	No comment
<u>PNPO</u>	100.00	0	No comment
<u>POFUT1</u>	100.00	0	No comment
<u>POGLUT1</u>	100.00	0	No comment
<u>POMGNT1</u>	100.00	0	No comment
<u>POMT1</u>	100.00	0	No comment
<u>POMT2</u>	99.90	0	No comment
<u>PRKCSH</u>	100.00	0	No comment
<u>PSAT1</u>	100.00	0	No comment
<u>PYGL</u>	100.00	0	No comment
<u>PYGM</u>	100.00	0	No comment
<u>RFT1</u>	100.00	0	No comment
<u>RPN1</u>	100.00	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>RPN2</u>	100.00	0	No comment
<u>RXYLT1</u>	99.84	0	No comment
<u>SCP2</u>	100.00	0	No comment
<u>SEC23B</u>	100.00	0	No comment
<u>SLC25A22</u>	100.00	0	No comment
<u>SLC2A1</u>	99.94	0	No comment
<u>SLC35A1</u>	100.00	0	No comment
<u>SLC35A2</u>	100.00	0	No comment
<u>SLC35A3</u>	100.00	0	No comment
<u>SLC35C1</u>	100.00	0	No comment
<u>SLC35D1</u>	99.99	0	No comment
<u>SLC39A8</u>	97.00	0	No comment
<u>SLC3A1</u>	100.00	0	No comment
<u>SLC6A19</u>	100.00	0	No comment
<u>SLC6A8</u>	98.78	0	No comment
<u>SLC7A9</u>	100.00	0	No comment
<u>SPR</u>	96.81	0	No comment
<u>SRD5A3</u>	99.42	0	No comment
<u>SSR3</u>	100.00	0	No comment
<u>SSR4</u>	99.93	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ST3GAL3</u>	100.00	0	No comment
<u>ST3GAL5</u>	92.64	0	No comment
<u>STT3A</u>	100.00	0	No comment
<u>STT3B</u>	99.25	0	No comment
<u>TMEM165</u>	89.91	0	No comment
<u>TMEM199</u>	100.00	0	No comment
<u>TRAPPC11</u>	100.00	0	No comment
<u>TUSC3</u>	100.00	0	No comment
<u>VPS13B</u>	99.25	0	No comment
<u>XYLT1</u>	88.83	0	No comment
<u>XYLT2</u>	98.70	0	No comment