

Full name:	Hepatology panel - UGent
Version number:	v4
Laboratory:	<u>Centrum Medische Genetica - UZ Gent</u>
Created:	05 Dec 2022 - 14:08
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Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ABCB11</u>	99.86	1	
<u>ABCB4</u>	99.71	1	
<u>ABCC2</u>	99.94	1	
<u>ABCD3</u>	92.70	1	
<u>ABCG5</u>	99.96	1	
<u>ABCG8</u>	99.95	1	
<u>ACADM</u>	96.14	1	
<u>ACADVL</u>	100.00	1	
<u>ACAT1</u>	99.81	1	
<u>ACOX2</u>	99.81	1	
<u>ADK</u>	99.78	1	
<u>AGL</u>	97.67	1	
<u>AGPAT2</u>	100.00	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>AKR1D1</u>	99.91	1	
<u>ALAD</u>	99.99	1	
<u>ALDOA</u>	100.00	1	
<u>ALDOB</u>	100.00	1	
<u>ALG8</u>	95.49	1	
<u>ALG9</u>	99.73	1	
<u>ALMS1</u>	99.90	1	
<u>AMACR</u>	100.00	1	
<u>ANKS6</u>	100.00	1	
<u>AP1S1</u>	99.49	1	
<u>ARG1</u>	99.95	1	
<u>ASL</u>	99.98	1	
<u>ASS1</u>	77.52	1	
<u>ATP7B</u>	100.00	1	
<u>ATP8B1</u>	99.94	1	
<u>BAAT</u>	99.98	1	
<u>BCS1L</u>	99.99	1	
<u>BSCL2</u>	99.99	1	
<u>BTD</u>	100.00	1	
<u>CAVIN1</u>	100.00	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>CC2D2A</u>	99.95	1	
<u>CCDC115</u>	99.90	1	
<u>CFTR</u>	99.45	1	
<u>CLDN1</u>	99.99	1	
<u>COG6</u>	99.86	1	
<u>COG7</u>	99.74	1	
<u>CPT1A</u>	99.98	1	
<u>CPT2</u>	99.65	1	
<u>CREB3L3</u>	99.98	1	
<u>CYP21A2</u>	99.91	1	
<u>CYP27A1</u>	100.00	1	
<u>CYP7A1</u>	99.99	1	
<u>CYP7B1</u>	99.82	1	
<u>DCDC2</u>	99.96	1	
<u>DGUOK</u>	99.93	1	
<u>DHCR7</u>	99.97	1	
<u>DKC1</u>	99.59	1	
<u>DLD</u>	99.89	1	
<u>DNAJB11</u>	99.97	1	
<u>EHHADH</u>	99.99	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>EIF2AK3</u>	97.43	1	
<u>ENO3</u>	100.00	1	
<u>EPHX1</u>	99.97	1	
<u>EPM2A</u>	99.99	1	
<u>ETFA</u>	99.88	1	
<u>ETFB</u>	100.00	1	
<u>FANCA</u>	99.98	1	
<u>FARSA</u>	100.00	1	
<u>FARSB</u>	99.64	1	
<u>FBP1</u>	100.00	1	
<u>FTH1</u>	22.62	1	
<u>G6PC1</u>	99.93	1	
<u>GAA</u>	100.00	1	
<u>GALE</u>	99.90	1	
<u>GALK1</u>	100.00	1	
<u>GALM</u>	100.00	1	
<u>GALT</u>	100.00	1	
<u>GANAB</u>	99.97	1	
<u>GBA1</u>	96.92	1	
<u>GBE1</u>	99.73	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>GFM1</u>	99.95	1	
<u>GNAS</u>	100.00	1	
<u>GUSB</u>	95.07	1	
<u>GYS1</u>	99.98	1	
<u>GYS2</u>	99.86	1	
<u>HADHA</u>	99.98	1	
<u>HAMP</u>	99.99	1	
<u>HFE</u>	100.00	1	
<u>HJV</u>	99.99	1	
<u>HLCS</u>	99.97	1	
<u>HNF1B</u>	100.00	1	
<u>HSD17B4</u>	99.71	1	
<u>HSD3B7</u>	100.00	1	
<u>IARS1</u>	99.89	1	
<u>IFT140</u>	100.00	1	
<u>INVS</u>	99.94	1	
<u>IVD</u>	100.00	1	
<u>JAG1</u>	100.00	1	
<u>KIF12</u>	99.99	1	
<u>KRT18</u>	47.42	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>KRT8</u>	71.60	1	
<u>LAMP2</u>	98.95	1	
<u>LARS1</u>	99.87	1	
<u>LDHA</u>	99.94	1	
<u>LIPA</u>	99.96	1	
<u>LRP5</u>	99.95	1	
<u>LSR</u>	99.98	1	
<u>MARS1</u>	99.97	1	
<u>MCEE</u>	99.90	1	
<u>MKS1</u>	99.92	1	
<u>MMUT</u>	99.68	1	
<u>MPI</u>	99.95	1	
<u>MPV17</u>	99.98	1	
<u>MVK</u>	99.97	1	
<u>MYO5B</u>	100.00	1	
<u>NBAS</u>	99.86	1	
<u>NEK9</u>	99.99	1	
<u>NEU1</u>	99.98	1	
<u>NGLY1</u>	99.93	1	
<u>NHLRC1</u>	100.00	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>NOTCH2</u>	99.03	1	
<u>NPC1</u>	99.99	1	
<u>NPC2</u>	100.00	1	
<u>NPHP1</u>	99.05	1	
<u>NPHP3</u>	99.89	1	
<u>NPHP4</u>	99.98	1	
<u>NR1H4</u>	99.49	1	
<u>OXCT1</u>	99.82	1	
<u>PCCA</u>	99.90	1	
<u>PCCB</u>	99.97	1	
<u>PEX1</u>	98.80	1	
<u>PEX10</u>	100.00	1	
<u>PEX11B</u>	99.62	1	
<u>PEX12</u>	100.00	1	
<u>PEX13</u>	99.36	1	
<u>PEX14</u>	100.00	1	
<u>PEX16</u>	99.94	1	
<u>PEX19</u>	99.25	1	
<u>PEX2</u>	100.00	1	
<u>PEX26</u>	100.00	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>PEX3</u>	99.85	1	
<u>PEX5</u>	99.89	1	
<u>PEX6</u>	99.99	1	
<u>PEX7</u>	99.72	1	
<u>PFKM</u>	99.57	1	
<u>PGAM2</u>	100.00	1	
<u>PGK1</u>	99.93	1	
<u>PGM1</u>	96.77	1	
<u>PHKA1</u>	99.84	1	
<u>PHKA2</u>	99.92	1	
<u>PHKB</u>	99.69	1	
<u>PHKG2</u>	99.86	1	
<u>PKD1</u>	99.98	1	
<u>PKD2</u>	99.91	1	
<u>PKHD1</u>	99.95	1	
<u>PMM2</u>	99.93	1	
<u>PMP22</u>	99.99	1	
<u>PNPLA3</u>	100.00	1	
<u>PODXL</u>	99.99	1	
<u>POLG</u>	100.00	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>PPM1F</u>	100.00	1	
<u>PRKAG2</u>	99.96	1	
<u>PRKCSH</u>	99.99	1	
<u>PYGL</u>	99.99	1	
<u>PYGM</u>	99.96	1	
<u>RBCK1</u>	100.00	1	
<u>RINT1</u>	99.99	1	
<u>RPGRIP1L</u>	96.35	1	
<u>SBDS</u>	99.93	1	
<u>SCO1</u>	99.98	1	
<u>SEC63</u>	99.84	1	
<u>SERPINA1</u>	100.00	1	
<u>SKIC3</u>	99.82	1	
<u>SLC10A1</u>	99.99	1	
<u>SLC10A2</u>	99.99	1	
<u>SLC16A1</u>	99.26	1	
<u>SLC25A13</u>	99.67	1	
<u>SLC25A20</u>	100.00	1	
<u>SLC2A2</u>	99.96	1	
<u>SLC30A10</u>	99.99	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>SLC51A</u>	100.00	1	
<u>SLCO1B1</u>	98.69	1	
<u>SLCO1B3</u>	99.85	1	
<u>SMPD1</u>	100.00	1	
<u>STN1</u>	99.88	1	
<u>STT3B</u>	99.96	1	
<u>TALDO1</u>	100.00	1	
<u>TANGO2</u>	99.85	1	
<u>TFR2</u>	99.97	1	
<u>TJP2</u>	99.99	1	
<u>TMEM216</u>	99.98	1	
<u>TMEM67</u>	99.69	1	
<u>TRMU</u>	100.00	1	
<u>TWNK</u>	100.00	1	
<u>UGT1A1</u>	99.98	1	
<u>UNC45A</u>	100.00	1	
<u>UROS</u>	100.00	1	
<u>USP53</u>	99.92	1	
<u>UTP4</u>	99.91	1	
<u>VIPAS39</u>	99.87	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>VPS33B</u>	99.95	1	
<u>VPS50</u>	98.35	1	
<u>WDR83OS</u>	100.00	1	
<u>YARS1</u>	99.29	1	
<u>ZFYVE19</u>	99.99	1	