

Full name:	Heterotaxie PCD - UGent
Description:	
Version number:	V3 (148 genen)
Laboratory:	<u>Centrum Medische Genetica - UZ Gent</u>
Created:	15 Dec 2020 - 16:24
Changed:	25 Mar 2024 - 12:21

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ACTC1</u>	98.57	0	
<u>ACVR2B</u>	99.99	0	
<u>AK7</u>	99.93	0	
<u>ALMS1</u>	99.90	0	
<u>ANKS6</u>	100.00	0	
<u>BBS1</u>	100.00	0	
<u>BBS10</u>	99.98	0	
<u>BBS2</u>	99.90	0	
<u>BCL9L</u>	99.99	0	
<u>BCOR</u>	99.97	0	
<u>BRAF</u>	99.78	0	
<u>C1orf127</u>	99.98	0	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>CBL</u>	99.95	0	
<u>CCDC103</u>	99.68	0	
<u>CCDC39</u>	99.74	0	
<u>CCDC40</u>	100.00	0	
<u>CCDC65</u>	99.80	0	
<u>CCNO</u>	100.00	0	
<u>CENPF</u>	99.97	0	
<u>CEP164</u>	99.99	0	
<u>CEP290</u>	98.10	0	
<u>CFAP298</u>	99.96	0	
<u>CFAP300</u>	99.47	0	
<u>CFAP45</u>	99.97	0	
<u>CFAP52</u>	99.92	0	
<u>CFAP53</u>	99.95	0	
<u>CFAP58</u>	99.96	0	
<u>CFC1</u>	21.93	0	
<u>CFTR</u>	99.45	0	
<u>CHD7</u>	99.99	0	
<u>CIROP</u>	91.91	0	
<u>CITED2</u>	100.00	0	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>CRELD1</u>	99.99	0	
<u>DAW1</u>	99.90	0	
<u>DGAT2L6</u>	99.97	0	
<u>DNAAF1</u>	99.99	0	
<u>DNAAF11</u>	99.80	0	
<u>DNAAF2</u>	99.91	0	
<u>DNAAF3</u>	99.99	0	
<u>DNAAF4</u>	99.78	0	
<u>DNAAF5</u>	99.99	0	
<u>DNAAF6</u>	98.73	0	
<u>DNAH1</u>	99.98	0	
<u>DNAH11</u>	99.93	0	
<u>DNAH12</u>	99.36	0	
<u>DNAH14</u>	99.64	0	
<u>DNAH17</u>	99.98	0	
<u>DNAH5</u>	99.98	0	
<u>DNAH8</u>	99.84	0	
<u>DNAH9</u>	99.79	0	
<u>DNAI1</u>	99.92	0	
<u>DNAI2</u>	99.86	0	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>DNAJB13</u>	99.91	0	
<u>DRC1</u>	99.93	0	
<u>DYNC1H1</u>	99.99	0	
<u>DYNLT2</u>	99.95	0	
<u>ELN</u>	99.86	0	
<u>EVC</u>	99.95	0	
<u>EVC2</u>	99.97	0	
<u>FOXF1</u>	99.99	0	
<u>FOXH1</u>	100.00	0	
<u>FOXJ1</u>	100.00	0	
<u>GAS2L2</u>	100.00	0	
<u>GAS8</u>	100.00	0	
<u>GATA4</u>	99.99	0	
<u>GATA6</u>	99.90	0	
<u>GDF1</u>	100.00	0	
<u>GJA1</u>	100.00	0	
<u>GPC3</u>	99.60	0	
<u>HAND1</u>	99.99	0	
<u>HES7</u>	100.00	0	
<u>HYDIN</u>	81.28	0	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>IFT46</u>	99.58	0	
<u>INVS</u>	99.94	0	
<u>JAG1</u>	100.00	0	
<u>KIF7</u>	100.00	0	
<u>LEFTY1</u>	99.98	0	
<u>LEFTY2</u>	100.00	0	
<u>LMBRD1</u>	99.67	0	
<u>LMNA</u>	99.96	0	
<u>LRRC56</u>	99.98	0	
<u>LZTFL1</u>	100.00	0	
<u>MAP2K1</u>	99.98	0	
<u>MAP2K2</u>	99.99	0	
<u>MCIDAS</u>	100.00	0	
<u>MED13L</u>	99.99	0	
<u>MEGF8</u>	99.90	0	
<u>MEIS2</u>	99.97	0	
<u>MKS1</u>	99.92	0	
<u>MMP21</u>	99.99	0	
<u>MNS1</u>	99.86	0	
<u>MRE11</u>	99.93	0	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>MYH6</u>	100.00	0	
<u>NAT10</u>	99.97	0	
<u>NEK10</u>	96.92	0	
<u>NEK9</u>	99.99	0	
<u>NKX2-5</u>	99.75	0	
<u>NKX2-6</u>	100.00	0	
<u>NME5</u>	99.81	0	
<u>NME8</u>	99.83	0	
<u>NODAL</u>	99.98	0	
<u>NOTCH1</u>	99.98	0	
<u>NOTCH2</u>	99.03	0	
<u>NPHP3</u>	99.89	0	
<u>NR2F2</u>	100.00	0	
<u>NSD1</u>	99.98	0	
<u>ODAD1</u>	96.04	0	
<u>ODAD2</u>	98.19	0	
<u>ODAD3</u>	99.96	0	
<u>ODAD4</u>	99.85	0	
<u>OFD1</u>	99.68	0	
<u>PITRM1</u>	99.89	0	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>PKD1L1</u>	99.84	0	
<u>PKD2</u>	99.91	0	
<u>PQBP1</u>	99.99	0	
<u>PRRX1</u>	99.56	0	
<u>PTPN11</u>	99.98	0	
<u>RAF1</u>	99.97	0	
<u>RIT1</u>	99.78	0	
<u>RPGR</u>	94.45	0	
<u>RSPH1</u>	99.87	0	
<u>RSPH3</u>	99.94	0	
<u>RSPH4A</u>	99.95	0	
<u>RSPH9</u>	99.99	0	
<u>SERPINE2</u>	99.97	0	
<u>SHOC2</u>	99.96	0	
<u>SHROOM3</u>	99.99	0	
<u>SMAD2</u>	99.92	0	
<u>SMAD6</u>	100.00	0	
<u>SOS1</u>	99.68	0	
<u>SPAG1</u>	99.78	0	
<u>SPEF2</u>	99.93	0	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>STK36</u>	99.98	0	
<u>TBX1</u>	99.95	0	
<u>TBX5</u>	99.98	0	
<u>TCTN2</u>	99.99	0	
<u>TP73</u>	100.00	0	
<u>TTC12</u>	99.97	0	
<u>TTC8</u>	99.67	0	
<u>UBR1</u>	99.93	0	
<u>WDR35</u>	99.92	0	
<u>WDR47</u>	95.08	0	
<u>ZFPM2</u>	100.00	0	
<u>ZIC3</u>	99.90	0	
<u>ZMPSTE24</u>	98.70	0	
<u>ZMYND10</u>	99.99	0	
<u>ZNF423</u>	98.94	0	