

Full name:	Ciliopathy (120 genes) - UGent
Version number:	Versie 3-Jul-2018 (H9.1-OP2-B16: Genpanel CilioPathy, 3-Jul-2018, in force on 4/07/2018)
Laboratory:	<u>Centrum Medische Genetica - UZ Gent</u>
Created:	11 Jun 2019 - 10:21
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Related Diseases

- Achromatopsia
- Acrocallosal syndrome
- Acrofacial dysostosis, Weyers type
- Ataxia-telangiectasia-like disorder
- Autosomal dominant polycystic kidney disease
- Autosomal recessive polycystic kidney disease
- Bardet-Biedl syndrome
- Caroli disease
- Cone rod dystrophy
- Cranioectodermal dysplasia
- Ellis Van Creveld syndrome
- Heart defect-tongue hamartoma-polysyndactyly syndrome
- Hydroletharus
- Infantile nephronophthisis
- Isolated neonatal sclerosing cholangitis
- Jeune syndrome
- Joubert syndrome
- Joubert syndrome with Jeune asphyxiating thoracic dystrophy
- Joubert syndrome with hepatic defect
- Joubert syndrome with oculorenal defect
- Joubert syndrome with renal defect
- Juvenile nephronophthisis
- Late-onset nephronophthisis

- [Leber congenital amaurosis](#)
- [MORM syndrome](#)
- [McKusick-Kaufman syndrome](#)
- [Meckel syndrome](#)
- [Multiple epiphyseal dysplasia, Al-Gazali type](#)
- [Non-syndromic male infertility due to sperm motility disorder](#)
- [Orofaciodigital syndrome type 1](#)
- [Orofaciodigital syndrome type 2](#)
- [Orofaciodigital syndrome type 3](#)
- [Orofaciodigital syndrome type 6](#)
- [Primary ciliary dyskinesia](#)
- [Primary ciliary dyskinesia-retinitis pigmentosa syndrome](#)
- [Renal-hepatic-pancreatic dysplasia](#)
- [Retinitis pigmentosa](#)
- [Saldino-Mainzer syndrome](#)
- [Senior-Boichis syndrome](#)
- [Senior-Loken syndrome](#)
- [Short rib-polydactyly syndrome type 5](#)
- [Short rib-polydactyly syndrome, Beemer-Langer type](#)
- [Short rib-polydactyly syndrome, Majewski type](#)
- [Short rib-polydactyly syndrome, Saldino-Noonan type](#)
- [Short rib-polydactyly syndrome, Verma-Naumoff type](#)
- [X-linked recessive intellectual disability-macrocephaly-ciliary dysfunction syndrome](#)

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
ACVR2B	99.99	0	
ADAMTS9	99.94	0	
AHI1	99.86	0	
ALMS1	99.90	0	
ANKS6	100.00	0	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ARL13B</u>	99.53	0	
<u>ARL3</u>	99.98	0	
<u>ARL6</u>	99.90	0	
<u>ARMC9</u>	99.77	0	
<u>B9D1</u>	99.80	0	
<u>B9D2</u>	99.88	0	
<u>BBIP1</u>	99.99	0	
<u>BBS1</u>	100.00	0	
<u>BBS10</u>	99.98	0	
<u>BBS12</u>	100.00	0	
<u>BBS2</u>	99.90	0	
<u>BBS4</u>	99.88	0	
<u>BBS5</u>	99.00	0	
<u>BBS7</u>	99.42	0	
<u>BBS9</u>	99.75	0	
<u>C2CD3</u>	99.88	0	
<u>CBY1</u>	100.00	0	
<u>CC2D2A</u>	99.95	0	
<u>CCDC103</u>	99.68	0	
<u>CCDC172</u>	99.85	0	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>CCDC28B</u>	99.99	0	
<u>CCDC32</u>	99.97	0	
<u>CCDC39</u>	99.74	0	
<u>CCDC40</u>	100.00	0	
<u>CCDC65</u>	99.80	0	
<u>CCDC96</u>	100.00	0	
<u>CCNO</u>	100.00	0	
<u>CENPF</u>	99.97	0	
<u>CEP104</u>	99.99	0	
<u>CEP120</u>	99.90	0	
<u>CEP164</u>	99.99	0	
<u>CEP290</u>	98.10	0	
<u>CEP295</u>	99.94	0	
<u>CEP41</u>	99.99	0	
<u>CEP55</u>	99.92	0	
<u>CEP83</u>	98.68	0	
<u>CFAP251</u>	99.98	0	
<u>CFAP298</u>	99.96	0	
<u>CFAP300</u>	99.47	0	
<u>CFAP410</u>	100.00	0	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>CFAP418</u>	100.00	0	
<u>CFAP44</u>	99.75	0	
<u>CFAP53</u>	99.95	0	
<u>CFAP69</u>	99.09	0	
<u>CFC1</u>	21.93	0	
<u>CILK1</u>	99.69	0	
<u>CPLANE1</u>	99.81	0	
<u>CRB2</u>	99.95	0	
<u>CSPP1</u>	98.31	0	
<u>DCDC2</u>	99.96	0	
<u>DDX59</u>	99.67	0	
<u>DEUP1</u>	99.86	0	
<u>DHCR7</u>	99.97	0	
<u>DLG5</u>	99.91	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	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>DNAAF6</u>	98.73	0	
<u>DNAH1</u>	99.98	0	
<u>DNAH11</u>	99.93	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	
<u>DNAJB13</u>	99.91	0	
<u>DRC1</u>	99.93	0	
<u>DYNC2H1</u>	99.66	0	
<u>DYNC2I1</u>	99.99	0	
<u>DYNC2I2</u>	99.98	0	
<u>DYNC2LI1</u>	99.94	0	
<u>DYNLT2B</u>	100.00	0	
<u>EVC</u>	99.95	0	
<u>EVC2</u>	99.97	0	
<u>EXOC3L2</u>	99.88	0	
<u>EXOC6B</u>	99.41	0	
<u>EXOC8</u>	99.99	0	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>EXTL3</u>	99.99	0	
<u>FAM149B1</u>	99.69	0	
<u>FAM166B</u>	99.99	0	
<u>FOXF1</u>	99.99	0	
<u>FUZ</u>	99.99	0	
<u>GAS2L2</u>	100.00	0	
<u>GAS8</u>	100.00	0	
<u>GLI3</u>	100.00	0	
<u>GLIS2</u>	100.00	0	
<u>HNF1B</u>	100.00	0	
<u>HYDIN</u>	81.28	0	
<u>HYLS1</u>	100.00	0	
<u>IFT122</u>	99.98	0	
<u>IFT140</u>	100.00	0	
<u>IFT172</u>	99.98	0	
<u>IFT27</u>	100.00	0	
<u>IFT43</u>	99.97	0	
<u>IFT52</u>	99.82	0	
<u>IFT74</u>	99.71	0	
<u>IFT80</u>	99.69	0	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>IFT81</u>	94.64	0	
<u>INPP5E</u>	99.85	0	
<u>INTS13</u>	98.20	0	
<u>INTU</u>	99.92	0	
<u>INVS</u>	99.94	0	
<u>IQCB1</u>	99.72	0	
<u>IQCE</u>	99.77	0	
<u>KATNIP</u>	99.13	0	
<u>KCTD3</u>	99.75	0	
<u>KIAA0586</u>	95.75	0	
<u>KIAA0753</u>	100.00	0	
<u>KIF14</u>	97.80	0	
<u>KIF3B</u>	99.99	0	
<u>KIF7</u>	100.00	0	
<u>LBR</u>	99.66	0	
<u>LCA5</u>	99.89	0	
<u>LRRC34</u>	99.52	0	
<u>LRRC45</u>	99.98	0	
<u>LRRC56</u>	99.98	0	
<u>LZTFL1</u>	100.00	0	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>MAPKBP1</u>	99.98	0	
<u>MCIDAS</u>	100.00	0	
<u>MKKS</u>	100.00	0	
<u>MKS1</u>	99.92	0	
<u>MMP21</u>	99.99	0	
<u>MRE11</u>	99.93	0	
<u>NCAPG2</u>	99.97	0	
<u>NEK1</u>	99.83	0	
<u>NEK9</u>	99.96	0	
<u>NME5</u>	99.81	0	
<u>NME8</u>	99.83	0	
<u>NPHP1</u>	99.05	0	
<u>NPHP3</u>	99.89	0	
<u>NPHP4</u>	99.98	0	
<u>OCRL</u>	99.89	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	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>PDE6D</u>	99.94	0	
<u>PIBF1</u>	99.90	0	
<u>PIK3C2A</u>	99.92	0	
<u>PKD1</u>	99.98	0	
<u>PKD2</u>	99.91	0	
<u>PKHD1</u>	99.95	0	
<u>PMFBP1</u>	99.89	0	
<u>PMM2</u>	99.93	0	
<u>POC1A</u>	99.98	0	
<u>POC1B</u>	100.00	0	
<u>RAB28</u>	99.97	0	
<u>RPGR</u>	94.45	0	
<u>RPGRIP1L</u>	96.35	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>SBDS</u>	99.93	0	
<u>SCLT1</u>	95.17	0	
<u>SCNM1</u>	99.47	0	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>SDCCAG8</u>	100.00	0	
<u>SLC30A7</u>	94.71	0	
<u>SPAG1</u>	99.78	0	
<u>STK36</u>	99.98	0	
<u>SUFU</u>	100.00	0	
<u>TBC1D32</u>	99.75	0	
<u>TCTN1</u>	99.92	0	
<u>TCTN2</u>	99.99	0	
<u>TCTN3</u>	99.92	0	
<u>TMEM107</u>	100.00	0	
<u>TMEM138</u>	100.00	0	
<u>TMEM17</u>	99.96	0	
<u>TMEM216</u>	99.98	0	
<u>TMEM218</u>	99.94	0	
<u>TMEM231</u>	88.88	0	
<u>TMEM237</u>	99.30	0	
<u>TMEM260</u>	99.90	0	
<u>TMEM67</u>	99.69	0	
<u>TOGARAM1</u>	99.93	0	
<u>TOPORS</u>	99.99	0	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>TRAF3IP1</u>	99.96	0	
<u>TRIM32</u>	100.00	0	
<u>TTC21B</u>	99.50	0	
<u>TTC23</u>	99.87	0	
<u>TTC26</u>	99.97	0	
<u>TTC6</u>	99.77	0	
<u>TTC8</u>	99.67	0	
<u>TUBGCP4</u>	99.80	0	
<u>TULP1</u>	99.99	0	
<u>TULP3</u>	99.91	0	
<u>TXNDC15</u>	99.78	0	
<u>VHL</u>	100.00	0	
<u>VPS13B</u>	99.90	0	
<u>WDPCP</u>	99.87	0	
<u>WDR19</u>	99.80	0	
<u>WDR35</u>	99.92	0	
<u>XPNPEP3</u>	99.99	0	
<u>ZFYVE19</u>	99.99	0	
<u>ZIC3</u>	99.90	0	
<u>ZMYND10</u>	99.99	0	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ZNF423</u>	98.94	0	
<u>ZSWIM6</u>	98.94	0	