

Full name:	Primary Ciliary Dyskinesia (61 genes) - KUL
Description:	https://firebasestorage.googleapis.com/v0/b/uz-laboboeken.appspot.com/o/GHB_CME%2FPCD%20-%20NGS%20-%20full%20panel.pdf?alt=media&token=52c732f3-8502-4364-9d1b-1f16504426c6
Version number:	Primary_ciliary_dyskinesia_v4
Laboratory:	<u>Centrum Menselijke Erfelijkheid - KUL</u>
Created:	03 Jul 2019 - 11:16
Changed:	23 Aug 2023 - 11:02

Related Diseases

- Primary ciliary dyskinesia
- Primary ciliary dyskinesia-retinitis pigmentosa syndrome
- 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
<u>CCDC103</u>	100.00	0	NM_213607.3
<u>CCDC39</u>	100.00	0	NM_181426.2
<u>CCDC40</u>	100.00	0	NM_017950.4
<u>CCDC65</u>	100.00	0	NM_033124.5
<u>CCNO</u>	100.00	0	NM_021147.5
<u>CFAP221</u>	100.00	-2	
<u>CFAP251</u>	100.00	0	NM_144668.6
<u>CFAP298</u>	100.00	0	NM_021254.4

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>CFAP300</u>	100.00	0	NM_032930.3
<u>CFAP45</u>	100.00	0	NM_012337.3
<u>CFAP53</u>	100.00	0	NM_145020.5
<u>CFAP57</u>	100.00	0	NM_152498.3
<u>CFAP74</u>	100.00	0	NM_001304360.2
<u>DAW1</u>	100.00	-2	NM_178821.3
<u>DNAAF1</u>	100.00	0	NM_178452.6
<u>DNAAF11</u>	100.00	0	NM_012472.6
<u>DNAAF2</u>	100.00	0	NM_018139.2
<u>DNAAF3</u>	100.00	0	NM_001256714.1
<u>DNAAF4</u>	100.00	0	NM_130810.4
<u>DNAAF5</u>	100.00	0	NM_017802.4
<u>DNAAF6</u>	100.00	0	NM_001169154.2
<u>DNAH11</u>	100.00	0	NM_001277115.2
<u>DNAH17</u>	100.00	0	NM_173628.4
<u>DNAH2</u>	100.00	0	NM_020877.5
<u>DNAH5</u>	100.00	0	NM_001369.3
<u>DNAH6</u>	100.00	0	NM_001370.2
<u>DNAH8</u>	100.00	0	NM_001206927.2
<u>DNAH9</u>	100.00	0	NM_031427.4

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>DNAI1</u>	100.00	0	NM_012144.4
<u>DNAI2</u>	100.00	0	NM_023036.5
<u>DNAJB13</u>	100.00	0	NM_153614.4
<u>DNAL1</u>	100.00	-2	NM_031427.4
<u>DRC1</u>	100.00	0	NM_145038.5
<u>ENKUR</u>	100.00	0	NM_145010.4
<u>FOXJ1</u>	100.00	0	NM_001454.4
<u>GAS2L2</u>	100.00	0	NM_139285.4
<u>GAS8</u>	100.00	0	NM_001481.3
<u>HYDIN</u>	100.00	0	NM_001270974.2
<u>LRRC56</u>	100.00	0	NM_198075.4
<u>MCIDAS</u>	100.00	0	NM_001190787.3
<u>MNS1</u>	100.00	0	NM_018365.4
<u>NEK10</u>	100.00	0	NM_199347.4
<u>NME5</u>	100.00	0	NM_003551.3
<u>NME8</u>	100.00	0	NM_016616.5
<u>ODAD1</u>	100.00	0	NM_144577.4
<u>ODAD2</u>	100.00	0	NM_018076.5
<u>ODAD3</u>	100.00	0	NM_145045.5
<u>ODAD4</u>	100.00	0	NM_031421.5

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>OFD1</u>	100.00	0	NM_003611.3
<u>RPGR</u>	100.00	0	NM_000328.3
<u>RSPH1</u>	100.00	0	NM_080860.4
<u>RSPH3</u>	100.00	0	NM_031924.8
<u>RSPH4A</u>	100.00	0	NM_001010892.3
<u>RSPH9</u>	100.00	0	NM_152732.5
<u>SPAG1</u>	100.00	0	NM_172218.3
<u>SPEF2</u>	100.00	0	NM_024867.4
<u>STK36</u>	100.00	0	NM_015690.5
<u>TEKT1</u>	100.00	0	NM_053285.2
<u>TP73</u>	100.00	0	NM_005427.4
<u>TTC12</u>	100.00	0	NM_017868.4
<u>ZMYND10</u>	100.00	0	NM_015896.4