# Whole Exome Sequencing
# Gene package Ciliopathy, version 6, 21-2-2020

**Technical information**
DNA was enriched using Agilent SureSelect DNA + SureSelect OneSeq 300kb CNV Backbone + Human All Exon V7 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 10 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate and non-unique reads are excluded. Data are demultiplexed with bcl2fastq Conversion Software from Illumina. Reads are mapped to the genome using the BWA-MEM algorithm (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected variants are filtered and annotated with Cartagenia software and classified with Alamut Visual. It is not excluded that pathogenic mutations are being missed using this technology. At this moment, there is not enough information about the sensitivity of this technique with respect to the detection of deletions and duplications of more than 5 nucleotides and of somatic mosaic mutations (all types of sequence changes).

Erasmus MC
University Medical Center Rotterdam

**Dept. Clinical Genetics**

| HGNC approved gene symbol | OMIM gene ID (active link to omim.org) | median depth | % covered >10x | % covered >20x | % covered >30x |
|---|---|---|---|---|---|
| ACVR2B | 602730 | 105 | 100 | 100 | 98 |
| ADAMTS9 | 605421 | 67 | 99 | 96 | 90 |
| AHI1 | 608894 | 67 | 100 | 98 | 90 |
| ALMS1 | 606844 | 100 | 100 | 100 | 99 |
| ANKS6 | 615370 | 89 | 95 | 93 | 90 |
| ARL13B | 608922 | 63 | 100 | 100 | 94 |
| ARL6 | 608845 | 43 | 96 | 95 | 83 |
| ARMC4 | 615408 | 67 | 98 | 96 | 92 |
| ARMC9 | 617612 | 76 | 100 | 98 | 93 |
| B9D1 | 614144 | 90 | 100 | 100 | 100 |
| B9D2 | 611951 | 82 | 100 | 100 | 100 |
| BBIP1 | 613605 | 45 | 100 | 96 | 86 |
| BBS1 | 209901 | 126 | 100 | 100 | 100 |
| BBS10 | 610148 | 66 | 100 | 100 | 100 |
| BBS12 | 610683 | 59 | 100 | 100 | 99 |
| BBS2 | 606151 | 77 | 100 | 100 | 97 |
| BBS4 | 600374 | 76 | 100 | 100 | 95 |

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|---|---|---|---|---|---|
| BBS5 | 603650 | 65 | 100 | 98 | 89 |
| BBS7 | 607590 | 64 | 100 | 99 | 95 |
| BBS9 | 607968 | 59 | 96 | 95 | 90 |
| C2CD3 | 615944 | 80 | 100 | 100 | 99 |
| C8orf37 | 614477 | 78 | 100 | 100 | 96 |
| CC2D2A | 612013 | 66 | 100 | 100 | 95 |
| CCDC103 | 614677 | 83 | 100 | 100 | 100 |
| CCDC114 | 615038 | 125 | 100 | 100 | 100 |
| CCDC151 | 615956 | 104 | 100 | 100 | 100 |
| CCDC28B | 610162 | 83 | 100 | 100 | 100 |
| CCDC39 | 613798 | 80 | 100 | 99 | 94 |
| CCDC40 | 613799 | 100 | 100 | 100 | 100 |
| CCDC65 | 611088 | 57 | 100 | 100 | 94 |
| CCNO | 607752 | 113 | 100 | 100 | 100 |
| CENPF | 600236 | 70 | 100 | 99 | 95 |
| CEP104 | 616690 | 71 | 100 | 98 | 93 |
| CEP120 | 613446 | 75 | 100 | 100 | 96 |
| CEP164 | 614848 | 93 | 100 | 100 | 98 |
| CEP290 | 610142 | 71 | 100 | 98 | 89 |
| CEP41 | 610523 | 69 | 100 | 100 | 94 |
| CEP55 | 610000 | 71 | 100 | 100 | 97 |
| CEP83 | 615847 | 60 | 100 | 96 | 81 |
| CFAP298 | 615494 | 75 | 100 | 100 | 96 |
| CFAP300 | 618058 | 53 | 100 | 98 | 87 |
| CFAP410 | 603191 | 141 | 100 | 100 | 100 |
| CFAP53 | 614759 | 98 | 100 | 100 | 98 |
| CFC1 | 605194 | 70 | 41 | 34 | 28 |
| CPLANE1 | 614571 | 71 | 100 | 99 | 95 |
| CSPP1 | 611654 | 83 | 100 | 100 | 97 |
| DCDC2 | 605755 | 96 | 100 | 100 | 97 |
| DDX59 | 615464 | 62 | 100 | 98 | 92 |
| DNAAF1 | 613190 | 131 | 100 | 100 | 99 |
| DNAAF2 | 612517 | 124 | 100 | 100 | 100 |
| DNAAF3 | 614566 | 98 | 100 | 100 | 99 |
| DNAAF4 | 608706 | 54 | 100 | 99 | 89 |

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|---|---|---|---|---|---|
| DNAAF5 | 614864 | 104 | 96 | 86 | 79 |
| DNAH1 | 603332 | 127 | 100 | 100 | 100 |
| DNAH11 | 603339 | 74 | 100 | 100 | 96 |
| DNAH17 | 610063 | 94 | 100 | 99 | 96 |
| DNAH5 | 603335 | 74 | 100 | 100 | 96 |
| DNAH9 | 603330 | 93 | 100 | 100 | 98 |
| DNAI1 | 604366 | 91 | 100 | 98 | 97 |
| DNAI2 | 605483 | 131 | 100 | 100 | 99 |
| DNAJB13 | 610263 | 88 | 100 | 100 | 100 |
| DNAL1 | 610062 | 77 | 100 | 100 | 95 |
| DNHD1 | 617277 | 117 | 100 | 100 | 100 |
| DRC1 | 615288 | 81 | 100 | 100 | 98 |
| DYNC2H1 | 603297 | 71 | 100 | 99 | 91 |
| DYNC2LI1 | 617083 | 53 | 100 | 100 | 91 |
| EVC | 604831 | 98 | 96 | 94 | 94 |
| EVC2 | 607261 | 87 | 100 | 99 | 95 |
| EXOC8 | 615283 | 118 | 100 | 100 | 100 |
| FAM149B1 | 618413 | 61 | 100 | 93 | 83 |
| FOXJ1 | 602291 | 116 | 100 | 100 | 100 |
| FUZ | 610622 | 109 | 100 | 100 | 100 |
| GAS2L2 | 611398 | 154 | 100 | 100 | 100 |
| GAS8 | 605178 | 85 | 99 | 97 | 93 |
| GDF1 | 602880 | 69 | 100 | 93 | 81 |
| GLIS2 | 608539 | 117 | 100 | 100 | 100 |
| HYDIN | 610812 | 46 | 82 | 69 | 57 |
| HYLS1 | 610693 | 74 | 100 | 100 | 100 |
| IFT122 | 606045 | 111 | 100 | 100 | 99 |
| IFT140 | 614620 | 113 | 100 | 100 | 98 |
| IFT172 | 607386 | 75 | 100 | 100 | 96 |
| IFT27 | 615870 | 84 | 100 | 100 | 99 |
| IFT43 | 614068 | 75 | 100 | 100 | 100 |
| IFT52 | 617094 | 50 | 100 | 99 | 91 |
| IFT80 | 611177 | 62 | 100 | 98 | 87 |
| IFT81 | 605489 | 47 | 99 | 94 | 82 |
| INPP5E | 613037 | 113 | 100 | 100 | 97 |

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|---|---|---|---|---|---|
| INTU | 610621 | 59 | 100 | 99 | 92 |
| INVS | 243305 | 81 | 100 | 100 | 97 |
| IQCB1 | 609237 | 54 | 100 | 94 | 78 |
| KCTD3 | 613272 | 61 | 100 | 100 | 96 |
| KIAA0556 | 616650 | 97 | 100 | 99 | 97 |
| KIAA0586 | 610178 | 72 | 100 | 98 | 94 |
| KIAA0753 | 617112 | 52 | 100 | 98 | 91 |
| KIF14 | 611279 | 74 | 100 | 99 | 91 |
| KIF7 | 611254 | 107 | 98 | 96 | 93 |
| LCA5 | 611408 | 74 | 100 | 100 | 99 |
| LEFTY2 | 601877 | 144 | 100 | 100 | 99 |
| LRRC56 | 618227 | 118 | 100 | 100 | 98 |
| LRRC6 | 614930 | 79 | 100 | 99 | 93 |
| LZTFL1 | 606568 | 79 | 100 | 100 | 93 |
| MAPKBP1 | 616786 | 108 | 100 | 100 | 98 |
| MCIDAS | 614086 | 97 | 100 | 100 | 92 |
| MKKS | 604896 | 80 | 100 | 100 | 100 |
| MKS1 | 609883 | 117 | 100 | 100 | 99 |
| MMP21 | 608416 | 81 | 100 | 96 | 88 |
| NCAPG2 | 608532 | 51 | 100 | 94 | 79 |
| NEK1 | 604588 | 56 | 100 | 98 | 89 |
| NEK8 | 609799 | 132 | 100 | 100 | 100 |
| NME8 | 607421 | 65 | 100 | 100 | 93 |
| NODAL | 601265 | 119 | 100 | 100 | 100 |
| NPHP1 | 607100 | 58 | 100 | 98 | 88 |
| NPHP3 | 608002 | 66 | 100 | 99 | 94 |
| NPHP4 | 607215 | 110 | 100 | 100 | 100 |
| OCRL | 300535 | 43 | 100 | 95 | 80 |
| OFD1 | 300170 | 43 | 100 | 95 | 75 |
| PDE6D | 602676 | 86 | 100 | 100 | 100 |
| PIBF1 | 607532 | 42 | 100 | 93 | 72 |
| PIH1D3 | 300933 | 40 | 100 | 92 | 66 |
| PIK3C2A | 603601 | 53 | 100 | 98 | 91 |
| PKD1 | 601313 | 93 | 97 | 95 | 90 |
| PKD2 | 173910 | 62 | 100 | 97 | 89 |

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|---|---|---|---|---|---|
| PKHD1 | 606702 | 82 | 100 | 100 | 98 |
| POC1A | 614783 | 90 | 100 | 100 | 100 |
| RPGRIP1 | 605446 | 88 | 100 | 100 | 96 |
| RPGRIP1L | 610937 | 60 | 98 | 96 | 89 |
| RSPH1 | 609314 | 80 | 100 | 100 | 95 |
| RSPH3 | 615876 | 89 | 100 | 99 | 94 |
| RSPH4A | 612647 | 100 | 100 | 100 | 100 |
| RSPH9 | 612648 | 125 | 100 | 100 | 100 |
| SCLT1 | 611399 | 51 | 100 | 97 | 87 |
| SDCCAG8 | 613524 | 92 | 100 | 100 | 96 |
| SPAG1 | 603395 | 64 | 100 | 99 | 91 |
| SPATA7 | 609868 | 62 | 100 | 99 | 94 |
| SUFU | 607035 | 104 | 100 | 100 | 100 |
| TBC1D32 | 615867 | 61 | 100 | 98 | 90 |
| TCTEX1D2 | 617353 | 89 | 100 | 100 | 93 |
| TCTN1 | 609863 | 94 | 100 | 100 | 98 |
| TCTN2 | 613846 | 84 | 100 | 100 | 97 |
| TCTN3 | 613847 | 66 | 100 | 100 | 97 |
| TMEM107 | 616183 | 100 | 100 | 100 | 94 |
| TMEM138 | 614459 | 54 | 100 | 100 | 100 |
| TMEM216 | 613277 | 87 | 100 | 100 | 95 |
| TMEM231 | 614949 | 96 | 100 | 95 | 90 |
| TMEM237 | 614423 | 57 | 100 | 98 | 87 |
| TMEM260 | 617449 | 58 | 100 | 98 | 93 |
| TMEM67 | 609884 | 77 | 100 | 100 | 94 |
| TRAF3IP1 | 607380 | 55 | 100 | 97 | 86 |
| TRIM32 | 602290 | 107 | 100 | 100 | 100 |
| TTBK2 | 611695 | 74 | 100 | 100 | 99 |
| TTC21B | 612014 | 75 | 100 | 99 | 94 |
| TTC25 | 617095 | 71 | 100 | 99 | 97 |
| TTC26 | 617453 | 55 | 100 | 99 | 93 |
| TTC8 | 608132 | 66 | 100 | 100 | 93 |
| TULP1 | 602280 | 108 | 100 | 100 | 100 |
| WDPCP | 613580 | 72 | 100 | 99 | 93 |
| WDR19 | 608151 | 71 | 100 | 100 | 97 |

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|---|---|---|---|---|---|
| WDR34 | 613363 | 116 | 100 | 100 | 100 |
| WDR35 | 613602 | 62 | 100 | 100 | 94 |
| WDR60 | 615462 | 80 | 100 | 99 | 94 |
| XPNPEP3 | 613553 | 63 | 100 | 100 | 96 |
| ZIC3 | 300265 | 101 | 100 | 100 | 97 |
| ZMYND10 | 607070 | 130 | 100 | 100 | 100 |
| ZNF423 | 604557 | 166 | 100 | 100 | 100 |

- OMIM release used: 8-9-2019
- The statistics above are based on a set of 100 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (±10bp flanking introns) of the longest transcript
- % Covered 10x , 20x and 30 x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x, 20x or 30x