

# Whole Exome Sequencing

## Gene package Ciliopathy, version 10, 25-2-2022



### Technical information

DNA was enriched using the Agilent SureSelectXT Human All Exon V7 capture kit and paired-end sequenced on the Illumina platform (outsourced). Sequencing data are demultiplexed with bcl2fastq2 Conversion Software from Illumina. Illumina DRAGEN Bio-IT Platform is used for read mapping to the hg19 genome and sequence variant detection. The detected sequence variants are annotated and filtered with Alissa Interpret software and classified with Alamut Visual. Copy number variant detection is performed using the BAM multiscale reference method using depth of coverage analysis and dynamical bins in NexusClinical. The detected copy number variants are annotated and filtered with the NexusClinical software and classified using UCSC Genome Browser (NCBI37/hg19). The sensitivity to detect variants using this technology is not 100%; pathogenic variants could be missed. 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).



**Dept. Clinical Genetics**

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
ACVR2B	602730	100	98.69	95.90	95.90
ADAMTS9	605421	99.85	99.35	97.95	96.53
AHI1	608894	100	100	100	99.41
AK7	615364	100	100	100	100
ALMS1	606844	99.89	99.40	98.78	96.05
ANKS6	615370	95.91	94.32	93.46	90.10
ARL13B	608922	100	99.46	98.38	87.50
ARL3	604695	100	99.40	89.34	85.89
ARL6	608845	100	100	100	95.81
ARMC9	617612	100	99.93	98.64	90.94
B9D1	614144	100	100	100	97.19
B9D2	611951	100	100	100	100
BBIP1	613605	100	100	100	100
BBS1	209901	100	100	100	98.56
BBS10	610148	100	100	99.41	97.96
BBS12	610683	100	100	100	100
BBS2	606151	100	100	100	100
BBS4	600374	100	100	100	96.00

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BBS5	603650	100	100	100	99.01
BBS7	607590	100	100	99.68	97.21
BBS9	607968	99.97	99.87	99.08	94.25
C2CD3	615944	100	100	99.96	98.51
CC2D2A	612013	100	99.84	98.65	91.30
CCDC103	614677	100	100	100	100
CCDC28B	610162	100	100	99.13	85.06
CCDC39	613798	100	99.71	98.09	87.36
CCDC40	613799	100	99.45	96.22	89.22
CCDC65	611088	100	100	99.69	93.92
CCNO	607752	100	100	100	95.50
CENPF	600236	100	100	99.89	97.58
CEP104	616690	100	100	99.53	96.98
CEP120	613446	100	100	100	97.66
CEP164	614848	100	99.52	97.04	86.58
CEP290	610142	95.56	90.87	87.25	80.13
CEP41	610523	100	100	100	99.55
CEP55	610000	100	100	97.20	83.44
CEP83	615847	100	97.51	90.22	80.44
CFAP221	618704	100	100	99.97	97.42
CFAP251	618146	100	100	99.73	92.58
CFAP298	615494	100	100	100	98.92
CFAP300	618058	100	100	100	89.80
CFAP410	603191	100	100	99.67	86.95
CFAP418	614477	No coverage data			
CFAP43	617558	100	100	100	96.94
CFAP44	617559	100	100	99.86	96.70
CFAP45	605152	100	100	99.08	91.57
CFAP52	609804	100	100	100	99.11
CFAP53	614759	99.38	96.74	95.12	87.46
CFAP69	617949	100	98.51	96.92	87.19
CFC1	605194	33.46	33.46	32.70	27.88
CPLANE1	614571	100	99.80	98.38	95.82
CSPP1	611654	100	99.29	96.27	91.41
DCDC2	605755	100	100	97.97	88.76
DDX59	615464	100	100	100	99.35
DNAAF1	613190	100	100	100	96.17
DNAAF11	614930	No coverage data			
DNAAF2	612517	100	100	100	100

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DNAAF3	614566	100	99.98	97.12	86.12
DNAAF4	608706	100	96.77	92.99	82.50
DNAAF5	614864	94.87	84.62	79.17	73.54
DNAAF6	300933	100	100	100	99.54
DNAH1	603332	100	99.94	99.31	95.37
DNAH11	603339	100	100	99.94	98.42
DNAH17	610063	100	99.90	99.54	96.15
DNAH5	603335	100	99.85	99.34	97.23
DNAH8	603337	100	99.96	99.48	96.00
DNAH9	603330	100	100	99.62	97.55
DNAI1	604366	100	100	99.28	94.36
DNAI2	605483	93.11	93.11	92.91	91.19
DNAJB13	610263	100	100	100	100
DNHD1	617277	100	100	100	100
DRC1	615288	100	100	99.82	95.25
DYNC2H1	603297	99.09	98.87	97.49	91.46
DYNC2I1	615462	99.92	96.05	87.67	76.33
DYNC2I2	613363	100	100	98.01	84.76
DYNC2LI1	617083	100	100	100	98.89
DYNLT2B	617353	100	100	100	100
ENKUR	611025	100	100	100	100
EVC	604831	94.29	94.26	93.73	87.77
EVC2	607261	100	99.86	99.03	95.16
EXOC8	615283	100	100	100	100
FAM149B1	618413	100	100	100	96.84
FOXJ1	602291	100	100	97.16	90.48
FSIP2	615796	100	100	99.70	97.18
FUZ	610622	100	99.02	96.51	81.21
GAS2L2	611398	100	100	100	97.57
GAS8	605178	100	100	100	94.33
GDF1	602880	100	87.20	74.01	59.86
GLIS2	608539	100	100	99.38	92.32
HYDIN	610812	98.12	90.57	83.20	69.56
HYLS1	610693	100	100	100	97.16
IFT122	606045	100	100	99.77	95.61
IFT140	614620	100	100	99.07	94.18
IFT172	607386	100	100	99.58	97.91
IFT27	615870	100	100	100	98.35
IFT43	614068	100	100	100	98.69

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IFT52	617094	100	100	100	100
IFT74	608040	100	100	95.28	79.01
IFT80	611177	100	100	99.70	91.35
IFT81	605489	100	100	99.87	94.26
INTU	610621	100	100	100	98.22
INVS	243305	100	99.60	98.89	96.61
IQCB1	609237	100	100	100	99.98
KATNIP	616650	100	99.80	98.31	93.37
KCTD3	613272	100	100	99.14	95.22
KIAA0586	610178	100	100	98.83	91.18
KIAA0753	617112	100	100	99.77	98.11
KIF14	611279	100	98.48	97.88	93.58
KIF7	611254	99.38	95.78	90.74	79.67
LBR	600024	100	100	100	94.80
LCA5	611408	100	100	100	99.33
LEFTY2	601877	100	100	100	97.54
LRRC56	618227	100	100	99.35	82.50
LZTFL1	606568	100	100	92.66	80.73
MAPKBP1	616786	100	99.55	98.37	93.82
MCIDAS	614086	100	100	100	98.26
MKKS	604896	100	100	100	100
MKS1	609883	100	99.95	98.94	93.72
MMP23A	603320	No coverage data			
NCAPG2	608532	100	100	99.16	92.27
NEK1	604588	100	99.49	98.10	94.33
NEK10	618726	100	100	99.39	96.91
NEK9	609798	100	100	99.41	96.32
NME5	603575	100	100	100	100
NME7	613465	100	100	99.93	94.23
NME8	607421	100	100	98.69	89.41
NODAL	601265	100	100	100	100
NPHP1	607100	99.79	96.88	96.88	95.81
NPHP3	608002	100	100	99.82	95.51
NPHP4	607215	100	100	99.14	92.28
OCRL	300535	98.15	98.15	98.15	97.22
ODAD1	615038	96.25	96.08	93.98	84.17
ODAD2	615408	100	98.60	96.70	92.34
ODAD3	615956	100	99.34	97.48	90.24
ODAD4	617095	100	100	99.75	91.36

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OFD1	300170	100	99.97	99.28	94.92
PDE6D	602676	100	100	100	100
PIBF1	607532	100	100	98.79	81.48
PIK3C2A	603601	100	100	100	98.02
PKD1	601313	95.97	91.51	85.12	70.96
PRKD2	606702	100	99.33	98.33	95.61
PKHD1	613036	100	100	99.98	98.54
PMPCA	614783	100	100	99.24	93.13
POC1A	607074	100	100	100	97.36
RPGR	312610	77.06	72.91	70.98	65.44
RPGRIP1	605446	100	100	99.78	98.61
RPGRIP1L	610937	99.73	98.44	96.86	92.30
RSPH1	609314	100	100	99.08	87.36
RSPH3	615876	100	100	100	96.83
RSPH4A	612647	100	100	100	100
RSPH9	612648	100	100	100	99.46
SCLT1	611399	95.52	84.60	76.02	67.06
SDCCAG8	613524	100	100	99.08	92.68
SPAG1	603395	100	98.04	93.83	80.54
SPATA7	609868	100	99.90	95.24	86.60
SPEF2	610172	100	100	98.57	95.14
STK36	607652	100	99.94	99.43	98.30
SUFU	607035	100	100	99.70	96.51
TBC1D32	615867	100	97.60	93.76	86.40
TCTN1	609863	100	100	100	100
TCTN2	613846	100	100	99.43	95.27
TCTN3	613847	100	100	100	98.55
TMEM107	616183	100	100	100	95.82
TMEM138	614459	100	100	100	100
TMEM216	613277	100	100	100	99.45
TMEM218	619285	100	100	100	96.22
TMEM231	614949	100	100	100	95.56
TMEM237	614423	100	100	99.26	94.88
TMEM260	617449	100	100	99.26	95.84
TMEM67	609884	100	100	99.87	95.43
TOGARAM1	617618	100	100	99.65	96.38
TRAF3IP1	607380	100	99.52	95.15	79.98
TRIM32	602290	100	100	100	100
TTBK2	611695	100	100	100	99.53

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TTC12	610732	100	100	100	97.88
TTC21B	612014	100	100	100	99.65
TTC26	617453	100	100	100	100
TTC8	608132	100	100	100	100
TULP1	602280	100	96.21	86.42	61.71
WDPCP	613580	100	100	99.54	95.61
WDR19	608151	100	100	99.75	97.05
WDR35	613602	100	99.78	99.23	96.28
XPNPEP3	613553	100	100	100	92.20
ZIC3	300265	100	100	100	98.56
ZMYND10	607070	100	100	100	100
ZNF423	604557	98.51	98.51	98.51	98.09

- OMIM release used: 23-9-2021

- The statistics above are based on a set of 104 samples

- % Covered 10x , 20x, 30x and 50x describes the percentage of a gene's coding sequence ( $\pm 10bp$  flanking introns) that is covered at least 10x, 20x, 30x or 50x