

Whole Exome Sequencing

Gene package Ciliopathy, version 8, 26-2-2021



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/>). Sequence variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: <http://www.broadinstitute.org/gatk/>). The detected sequence variants are filtered and annotated with Alissa Interpret software and classified with Alamut Visual. Copy 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 filtered and annotated with the NexusClinical software and classified using UCSC Genome Browser (NCBI37/hg19). It is not excluded that pathogenic variants 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).



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	97.35	95.90	95.64
ADAMTS9	605421	99.86	99.20	97.93	95.50
AHI1	608894	100	100	99.93	98.69
ALMS1	606844	99.78	99.08	97.76	93.88
ANKS6	615370	97.68	93.82	92.04	87.68
ARL13B	608922	100	98.18	93.87	78.87
ARL3	604695	100	95.65	89.34	85.74
ARL6	608845	100	100	99.39	95.53
ARMC4	615408	100	96.70	96.06	88.62
ARMC9	617612	100	98.64	94.58	85.33
B9D1	614144	100	100	100	96.69
B9D2	611951	100	100	100	100
BBIP1	613605	100	100	100	100
BBS1	209901	100	100	100	97.85
BBS10	610148	100	100	99.09	97.44
BBS12	610683	100	100	100	100
BBS2	606151	100	100	100	98.62
BBS4	600374	100	100	98.72	89.85

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BBS5	603650	100	100	100	96.04
BBS7	607590	100	99.49	98.48	95.17
BBS9	607968	99.97	99.27	96.37	92.04
C2CD3	615944	100	100	99.55	96.55
C8orf37	614477	100	100	100	100
CC2D2A	612013	100	98.73	95.51	85.85
CCDC103	614677	100	100	100	97.07
CCDC114	615038	96.25	96.25	94.15	83.11
CCDC151	615956	100	99.58	97.80	90.54
CCDC28B	610162	100	100	99.50	83.31
CCDC39	613798	100	98.57	93.89	81.68
CCDC40	613799	100	98.63	94.15	86.45
CCDC65	611088	100	100	98.08	87.16
CCNO	607752	100	100	100	90.99
CENPF	600236	100	99.92	99.14	91.32
CEP104	616690	100	100	99.33	95.15
CEP120	613446	100	100	99.96	95.96
CEP164	614848	100	99.42	96.54	84.31
CEP290	610142	92.98	88.53	84.16	75.57
CEP41	610523	100	100	100	63.51
CEP55	610000	100	97.91	91.88	73.84
CEP83	615847	99.88	91.20	82.06	73.67
CFAP298	615494	100	100	100	91.69
CFAP300	618058	100	100	96.33	85.23
CFAP410	603191	100	100	97.85	76.65
CFAP53	614759	98.91	95.59	93.89	84.72
CFC1	605194	33.46	33.46	30.16	24.59
CPLANE1	614571	99.99	98.94	97.28	93.86
CSPP1	611654	99.57	95.75	93.87	87.67
DCDC2	605755	100	99.36	95.76	87.13
DDX59	615464	100	100	99.97	98.02
DNAAF1	613190	100	100	98.43	90.48
DNAAF2	612517	100	100	98.06	89.87
DNAAF3	614566	100	99.27	95.20	81.71
DNAAF4	608706	99.93	94.58	88.65	81.81
DNAAF5	614864	94.96	84.27	78.27	71.35
DNAH1	603332	100	99.78	98.65	93.56
DNAH11	603339	100	99.97	99.48	95.73
DNAH17	610063	100	99.90	99.49	95.07

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DNAH5	603335	100	99.57	98.78	95.30
DNAH9	603330	100	100	99.27	95.93
DNAI1	604366	100	99.52	97.59	87.05
DNAI2	605483	93.11	93.11	92.43	88.99
DNAJB13	610263	100	100	100	99.86
DNAL1	610062	100	91.44	89.59	88.29
DNHD1	617277	100	100	100	98.65
DRC1	615288	100	99.86	98.05	85.66
DYNC2H1	603297	99.06	97.98	95.50	87.54
DYNC2LI1	617083	100	100	99.52	96.47
EVC	604831	94.29	93.49	91.71	82.76
EVC2	607261	100	99.70	98.53	93.34
EXOC8	615283	100	100	100	100
FAM149B1	618413	100	100	99.11	96.67
FOXJ1	602291	100	99.50	95.82	79.36
FUZ	610622	100	98.24	94.79	77.79
GAS2L2	611398	100	100	100	95.11
GAS8	605178	100	100	98.72	89.78
GDF1	602880	100	84.78	73.92	57.87
GLIS2	608539	100	100	99.20	91.70
HYDIN	610812	94.93	87.09	78.18	67.63
HYLS1	610693	100	100	100	96.29
IFT122	606045	100	99.83	98.59	91.64
IFT140	614620	100	100	98.91	92.50
IFT172	607386	100	100	99.30	96.91
IFT27	615870	100	100	100	92.91
IFT43	614068	100	100	100	97.33
IFT52	617094	100	100	100	100
IFT74	608040	100	96.51	89.89	65.78
IFT80	611177	100	99.93	97.09	85.61
IFT81	605489	100	99.56	97.09	82.18
INPP5E	613037	99.81	97.42	94.23	87.57
INTU	610621	100	100	99.49	96.54
INVS	243305	100	99.29	98.31	94.13
IQCB1	609237	100	100	100	99.03
KCTD3	613272	100	98.31	96.33	91.64
KIAA0556	616650	100	99.74	98.06	92.87
KIAA0586	610178	100	99.66	96.58	86.09
KIAA0753	617112	100	99.88	98.91	95.88

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KIF14	611279	99.27	98.19	96.42	91.26
KIF7	611254	99.65	96.04	90.61	78.96
LBR	600024	100	100	97.36	90.52
LCA5	611408	100	100	100	97.98
LEFTY2	601877	100	100	99.53	96.86
LRRC56	618227	100	100	97.18	78.79
LRRC6	614930	94.02	94.02	93.47	90.72
LZTFL1	606568	100	96.36	87.71	77.48
MAPKBP1	616786	100	99.55	98.30	93.62
MCIDAS	614086	100	100	100	96.80
MKKS	604896	100	100	100	100
MKS1	609883	100	99.90	98.82	92.05
MMP21	608416	100	97.08	92.61	82.27
NCAPG2	608532	100	99.21	95.85	87.64
NEK1	604588	100	98.60	97.14	89.44
NEK10	618726	100	99.59	98.46	93.13
NEK8	609799	100	100	99.58	92.21
NME5	603575	100	100	100	99.32
NME8	607421	100	99.03	93.41	84.33
NODAL	601265	100	100	100	100
NPHP1	607100	96.88	96.88	96.88	92.57
NPHP3	608002	100	99.97	98.05	91.53
NPHP4	607215	100	99.94	98.34	89.49
OCRL	300535	98.15	98.15	98.15	96.50
OFD1	300170	100	98.67	94.05	75.14
PDE6D	602676	100	100	100	98.36
PIBF1	607532	100	99.58	94.15	67.48
PIH1D3	300933	100	100	100	91.67
PIK3C2A	603601	100	100	99.66	95.62
PKD1	601313	95.38	89.50	82.53	66.79
PKD2	173910	99.19	93.13	87.92	78.28
PKHD1	606702	100	100	99.63	96.86
POC1A	614783	100	100	100	97.36
RPGRIP1	605446	100	99.86	99.45	95.94
RPGRIP1L	610937	99.01	97.45	94.84	90.66
RSPH1	609314	100	100	97.89	87.98
RSPH3	615876	100	100	98.53	94.18
RSPH4A	612647	100	100	100	98.24
RSPH9	612648	100	100	100	98.71

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SCLT1	611399	90.35	76.02	71.73	57.89
SDCCAG8	613524	100	99.54	96.41	81.03
SPAG1	603395	99.36	94.58	88.75	72.66
SPATA7	609868	100	96.34	91.02	81.74
STK36	607652	100	99.90	99.47	98.01
SUFU	607035	100	100	99.73	96.13
TBC1D32	615867	99.41	95.02	90.66	81.60
TCTEX1D2	617353	100	100	100	100
TCTN1	609863	100	100	100	100
TCTN2	613846	100	100	99.08	93.90
TCTN3	613847	100	100	100	95.60
TMEM107	616183	100	100	100	93.49
TMEM138	614459	100	100	100	100
TMEM216	613277	100	100	100	96.69
TMEM218	No ID	100	100	100	84.43
TMEM231	614949	100	100	99.19	94.38
TMEM237	614423	100	100	98.32	92.86
TMEM260	617449	100	99.06	97.75	91.95
TMEM67	609884	100	100	99.55	88.82
TOGARAM1	617618	100	99.56	98.38	94.48
TRAF3IP1	607380	100	94.07	84.73	71.11
TRIM32	602290	100	100	100	100
TTBK2	611695	100	100	100	99.21
TTC12	610732	100	100	98.86	92.95
TTC21B	612014	100	100	100	98.62
TTC25	617095	100	100	97.74	77.70
TTC26	617453	100	100	100	96.92
TTC8	608132	100	100	100	98.76
TULP1	602280	100	96.29	87.18	60.93
WDPCP	613580	100	99.65	97.17	90.36
WDR19	608151	100	99.81	98.99	94.93
WDR34	613363	100	99.78	96.90	76.51
WDR35	613602	99.99	99.40	97.68	94.88
WDR60	615462	98.65	91.18	82.59	72.35
XPNPEP3	613553	100	100	100	93.66
ZIC3	300265	100	100	100	94.18
ZMYND10	607070	100	100	100	100
ZNF423	604557	100	100	100	98.31

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- OMIM release used: 18-2-2021
- The statistics above are based on a set of 100 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