

# Whole Exome Sequencing

## Gene package Oncogenetics, version 5, 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 $\geq 50x$
A2ML1	610627	100	99.96	99.39	97.99
ACD	609377	100	100	100	97.43
ACTRT1	300487	100	100	100	100
ACVRL1	601284	100	100	100	88.28
AIP	605555	100	100	100	97.94
AKT1	164730	100	100	100	97.50
ALK	105590	100	99.01	95.43	88.36
ANKRD26	610855	97.20	93.08	88.47	78.35
APC	611731	100	100	100	99.62
ARMC5	615549	100	100	99.92	89.72
ATG2B	616226	100	100	99.77	95.43
ATM	607585	100	100	93.43	54.67
ATR	601215	100	100	99.89	97.02
AXIN2	604025	100	100	99.63	97.62
BAP1	603089	100	99.94	98.16	93.00
BARD1	601593	100	100	96.64	84.53
BLM	604610	100	99.87	99.08	96.13
BMPR1A	601299	100	100	100	100
BRAF	164757	95.04	94.58	93.81	88.27
BRCA1	113705	100	100	100	100
BRCA2	600185	100	100	100	99.89
BRIP1	605882	100	100	100	99.92
BUB1	602452	100	100	100	98.76
BUB1B	602860	100	99.74	98.05	95.43

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BUB3	603719	100	100	100	99.38
CASR	601199	100	100	100	99.88
CBL	165360	100	100	100	99.03
CDC73	607393	100	100	99.02	94.96
CDH1	192090	100	100	100	99.74
CDK4	123829	100	100	97.52	90.80
CDKN1A	116899	100	100	100	100
CDKN1B	600778	100	100	100	100
CDKN1C	600856	84.56	72.52	60.78	51.01
CDKN2A	600160	100	100	96.17	86.99
CDKN2B	600431	100	100	98.68	88.99
CDKN2C	603369	100	100	99.36	91.82
CEBPA	116897	96.07	68.79	52.04	48.30
CHEK2	604373	100	99.39	95.04	83.58
CHRNA3	118503	99.91	97.34	93.76	93.76
CHRNA5	118505	92.83	79	79	74
COL17A1	113811	100	98.77	97.44	91.78
CREBBP	600140	100	99.21	97.19	91.73
CSF3R	138971	100	98.13	95.07	88.84
CTC1	613129	100	100	99.71	94.64
CTNNA1	116805	100	99.92	98.92	94.80
CTRC	601405	100	100	100	96.01
CYLD	605018	100	100	99.30	95.34
DDB2	600811	100	100	100	98.68
DDX41	608170	99.12	96.49	94.72	90.41
DICER1	606241	100	100	100	99.04
DIS3L2	614184	100	100	100	95.21
DKC1	300126	100	100	98.05	93.43
DNAJC21	617048	100	100	98.94	92.66
DOCK8	611432	100	99.94	99.37	96.42
EGFR	131550	100	100	98.97	96.26
EGLN1	606425	100	96.59	88.61	74.75
ELANE	130130	100	100	100	95.95
ENG	131195	100	100	98.31	90.90
ERCC1	126380	100	98.77	94.13	63.79
ERCC2	126340	100	100	100	94.45
ERCC3	133510	100	100	100	97.60
ERCC4	133520	100	100	100	96.66
ERCC5	133530	100	100	98.83	92.83
ERCC6	609413	100	99.84	99.20	98.06
ETV6	600618	100	100	100	99.01
EXO1	606063	99.25	98.09	96.68	91.76
EXT1	608177	100	100	100	99.43
EXT2	608210	100	100	100	99.64

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FAN1	613534	100	100	100	98.22
FANCA	607139	100	100	100	97.23
FANCB	300515	100	100	99.67	93.46
FANCC	613899	100	100	100	97.90
FANCD2	613984	97.60	97.60	97.60	94.35
FANCE	613976	98.34	92.17	88.97	83.08
FANCF	613897	100	100	100	99.39
FANCG	602956	100	100	98.65	93.29
FANCI	611360	100	100	100	99.61
FANCL	608111	100	100	100	99.05
FANCM	609644	100	99.89	98.86	94.44
FAS	134637	100	100	100	92.44
FH	136850	100	97.54	94.62	91.21
FLCN	607273	96.36	96.36	96.36	93.23
FOCAD	614606	100	99.70	99.23	95.35
G6PC3	611045	100	100	100	100
GATA1	305371	100	98.81	94.25	79.42
GATA2	137295	100	100	100	85.78
GDNF	600837	100	100	96.24	84.34
GFI1	600871	100	99.78	98.70	96.32
GPC3	300037	100	100	99.55	95.55
GREM1	603054	100	100	98.25	93.01
GSKIP	616605	100	100	100	93.87
HABP2	603924	100	100	100	98.89
HAX1	605998	100	100	100	98.77
HNF1A	142410	100	100	100	97.09
HNF1B	189907	100	100	100	97.49
HOXB13	604607	100	100	100	97.14
HRAS	190020	100	100	100	100
ITK	186973	100	100	100	98.77
KIF1B	605995	100	100	100	98.30
KIT	164920	100	100	100	99.82
KLLN	612105	100	100	100	100
KRAS	190070	100	100	100	100
LZTR1	600574	100	100	100	98.04
MAP2K1	176872	100	100	100	97.96
MAP2K2	601263	100	100	98.27	87.68
MAX	154950	100	100	92.07	72.93
MBD4	603574	100	100	100	98.16
MC1R	155555	100	100	98.35	94.44
MEN1	613733	98.96	94.25	93.23	91.67
MET	164860	100	100	99.48	97.53
MITF	156845	100	100	100	100
MLH1	120436	100	100	100	99.43

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MLH3	604395	100	100	99.93	99.36
MPL	159530	100	100	100	98.09
MRE11	600814	100	100	100	97.46
MSH2	609309	100	99.97	99.74	96.52
MSH3	600887	100	100	99.41	93.25
MSH6	600678	100	100	100	99.23
MTAP	156540	100	100	100	100
MUTYH	604933	100	100	100	98.53
NBN	602667	100	100	100	99.11
NF1	613113	100	99.95	99.13	94.91
NF2	607379	100	100	100	100
NHP2	606470	100	100	100	100
NOP10	606471	100	100	100	100
NOTCH2	600275	99.86	98.85	98.02	95.09
NRAS	164790	100	100	100	99.54
NSD1	606681	100	99.87	99.57	97.75
NTHL1	602656	100	100	100	93.70
NTRK1	191315	100	99.62	96.55	85.15
OGG1	601982	100	100	100	96.13
PALB2	610355	100	100	100	98.66
PALLD	608092	100	100	99.16	96.63
PARN	604212	100	100	100	98.33
PAX5	167414	100	100	100	99.62
PDGFRA	173490	100	100	100	99.83
PHOX2B	603851	93.16	83.78	74.85	55.94
PIK3CA	171834	100	100	100	99.11
PMS2	600259	100	99.69	98.46	93.71
POLD1	174761	100	99.85	98.25	90.01
POLE	174762	100	100	99.53	94.72
POLH	603968	100	100	100	100
POT1	606478	100	100	100	95.16
PRF1	170280	100	100	100	100
PRKAR1A	188830	100	100	100	100
PTCH1	601309	96.46	96.46	96.46	96.46
PTCH2	603673	100	100	99.33	96.47
PTEN	601728	100	100	100	100
PTPN11	176876	98.37	98.37	98.37	98.37
RAD50	604040	100	99.92	96.90	84.75
RAD51B	602948	100	100	98.80	93.82
RAD51C	602774	100	100	100	99.55
RAD51D	602954	100	96.32	92.45	74.37
RAF1	164760	100	100	100	96.29
RASAL1	604118	100	99.96	98.70	90.12
RB1	614041	100	98.07	95.73	89.05

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RBBP6	600938	100	98.72	95.82	92.99
RBM8A	605313	100	100	100	94.55
RECQL	600537	100	100	99.87	95.29
RECQL4	603780	100	99.95	99.35	94.89
REST	600571	100	100	100	100
RET	164761	100	99.85	97.07	91.06
RGS17	<b>607191</b>	100	100	100	100
RHBDF2	614404	100	100	99.59	96.08
RINT1	610089	100	100	100	98.41
RIT1	609591	100	100	100	100
RNF43	612482	100	100	99.78	95.75
RPL11	604175	100	100	100	100
RPL15	604174	100	100	95.98	91.00
RPL35A	180468	100	96.34	90	90
RPL5	603634	99.14	97.15	94.96	90.87
RPS10	603632	100	100	100	96.64
RPS17	180472	No coverage			
RPS19	603474	100	100	100	97.27
RPS24	602412	100	100	100	97.78
RPS26	603701	100	100	100	100
RPS29	603633	100	100	100	100
RPS7	603658	97.51	86.97	86.47	83.33
RTEL1	608833	100	100	99.77	94.04
RUNX1	151385	100	100	100	92.76
SAMD9	610456	100	100	100	99.87
SAMD9L	611170	100	100	100	100
SBDS	607444	100	100	100	100
SDHA	600857	100	97.10	96.38	94.50
SDHAF2	613019	100	100	100	91.00
SDHB	185470	100	100	100	99.60
SDHC	602413	100	100	100	100
SDHD	602690	100	100	100	100
SERPINA1	107400	100	100	100	100
SFTPA1	178630	100	100	100	99.83
SFTPA2	178642	100	100	100	100
SH2D1A	300490	100	100	100	100
SHOC2	602775	100	100	100	98.53
SLX4	613278	100	99.93	99.03	92.91
SMAD4	600993	100	100	100	99.47
SMAD9	603295	100	99.38	96.78	91.72
SMARCA4	603254	100	100	99.61	94.47
SMARCB1	601607	100	100	100	97.91
SMARCE1	603111	100	100	100	98.95
SOS1	182530	100	99.96	97.71	96.03

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SPINK1	167790	100	100	100	100
SPRED1	609291	100	100	100	100
SRP72	602122	100	100	100	100
SRY	480000	No coverage			
STAT3	102582	100	100	100	95.99
STK11	602216	100	100	100	100
SUFU	607035	100	100	99.70	96.51
TERF1	600951	99.47	86.39	80.92	68.75
TERF2IP	605061	100	100	100	98.25
TERT	187270	100	99.15	96.14	90.14
TGFBR1	190181	93.12	93.12	93.12	93.12
TGFBR2	190182	100	100	100	98.71
TINF2	604319	100	100	100	100
TMEM127	613403	100	100	100	88.31
TNFRSF11A	603499	90.20	90.20	87.10	81.42
TP53	191170	100	100	100	100
TP63	603273	100	100	100	96.68
TRIM37	605073	100	100	100	97.76
TSC1	605284	100	100	100	99.86
TSC2	191092	100	100	99.90	96.43
UROD	613521	100	100	100	99.77
USB1	613276	100	99.20	88.66	75.29
VHL	608537	100	100	100	77.83
WAS	300392	100	99.86	94.70	77.23
WRAP53	612661	100	100	100	97.04
WRN	604611	100	100	99.77	94.20
WT1	607102	100	100	100	100
XPA	611153	100	100	96.27	79.55
XPC	613208	100	100	98.61	88.95
XRCC1	194360	100	99.29	96.14	82.83
XRCC2	600375	100	100	100	100
XRCC3	600675	100	100	95.42	81.71
XRCC4	194363	100	100	100	95.91

- OMIM release used: 18-2-2021

- 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