Whole Exome Sequencing
Gene package Oncogenetics, version 3, 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
A2ML1	610627	66	100	100	96
ACD	609377	139	100	100	100
ACTRT1	300487	78	100	100	100
ACVRL1	601284	102	100	100	95
AIP	605555	134	100	100	100
AKT1	164730	147	100	100	100
ALK	105590	113	100	100	99
ANKRD26	610855	70	100	98	90
APC	611731	190	100	100	100
ARMC5	615549	142	100	98	95
ATG2B	616226	55	100	98	88
ATM	607585	75	100	99	95
ATR	601215	91	100	99	92
AXIN2	604025	108	100	100	98
BAP1	603089	121	100	100	100
BARD1	601593	78	100	100	98
BLM	604610	82	100	100	96

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BMPR1A	601299	76	100	100	100
BRAF	164757	68	100	100	94
BRCA1	113705	239	100	100	99
BRCA2	600185	202	100	100	100
BRIP1	605882	55	100	99	91
BUB1	602452	79	100	100	97
BUB1B	602860	70	100	99	94
BUB3	603719	74	100	98	93
CASR	601199	146	100	100	100
CBL	165360	78	100	100	100
CDC73	607393	75	100	100	97
CDH1	192090	92	100	100	99
CDK4	123829	88	100	100	100
CDKN1A	116899	170	100	100	100
CDKN1B	600778	163	100	100	100
CDKN1C	600856	74	90	83	76
CDKN2A	600160	139	100	100	100
CDKN2B	600431	105	100	100	100
CDKN2C	603369	92	100	100	100
CEBPA	116897	130	100	93	75
CFTR	602421	93	100	100	97
CHEK2	604373	81	100	97	87
CHRNA3	118503	141	100	100	100
CHRNA5	118505	69	100	97	93
COL17A1	113811	98	100	100	99
CREBBP	600140	85	100	99	94
CSF3R	138971	98	100	100	100
CTC1	613129	100	100	100	100
CTNNA1	116805	93	100	100	96
CTRC	601405	111	100	100	100
CYLD	605018	56	100	98	89
DDB2	600811	113	100	100	99
DDX41	608170	135	100	100	100
DICER1	606241	77	100	99	95
DIS3L2	614184	100	100	100	97
DKC1	300126	53	100	98	89

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DNAJC21	617048	62	100	96	89
DOCK8	611432	74	100	99	94
EGFR	131550	87	100	100	98
EGLN1	606425	114	100	92	83
ELANE	130130	127	100	100	100
ENG	131195	98	100	100	99
ERCC1	126380	71	100	100	94
ERCC2	126340	97	100	99	98
ERCC3	133510	82	100	100	99
ERCC4	133520	83	100	100	97
ERCC5	133530	85	100	100	98
ERCC6	609413	88	100	100	97
ETV6	600618	131	100	100	99
EXO1	606063	70	100	100	97
EXT1	608177	70	100	100	96
EXT2	608210	98	100	100	97
FAN1	613534	75	100	100	98
FANCA	607139	102	100	100	97
FANCB	300515	45	100	97	83
FANCC	613899	69	100	99	91
FANCD2	613984	71	100	100	96
FANCE	613976	123	100	99	90
FANCF	613897	170	100	100	100
FANCG	602956	126	100	100	100
FANCI	611360	72	100	100	98
FANCL	608111	57	100	99	88
FANCM	609644	75	100	99	95
FAS	134637	264	100	100	96
FH	136850	82	99	94	87
FLCN	607273	127	100	100	100
FOCAD	614606	64	100	98	93
G6PC3	611045	103	100	100	100
GATA1	305371	75	100	100	96
GATA2	137295	100	100	100	100
GDNF	600837	81	100	100	100
GFI1	600871	88	100	100	100

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GPC3	300037	136	100	100	100
GREM1	603054	53	100	99	91
GSKIP	616605	101	100	100	100
HABP2	603924	69	100	100	100
HAX1	605998	92	100	100	96
HNF1A	142410	99	100	100	99
HNF1B	189907	157	100	100	100
HOXB13	604607	112	100	100	100
HRAS	190020	121	100	100	100
ITK	186973	179	100	100	100
KIF1B	605995	76	100	100	95
KIT	164920	71	100	100	97
KLLN	612105	79	100	100	99
KRAS	190070	98	100	100	100
LZTR1	600574	83	100	100	87
MAP2K1	176872	117	100	100	100
MAP2K2	601263	79	100	100	96
MAX	154950	117	100	100	95
MBD4	603574	72	100	100	100
MC1R	155555	83	100	100	98
MEN1	613733	237	100	100	100
MET	164860	182	100	100	100
MITF	156845	64	100	100	96
MLH1	120436	87	100	100	99
MLH3	604395	203	100	100	100
MPL	159530	62	100	100	99
MRE11	600814	109	100	100	100
MSH2	609309	55	100	97	83
MSH3	600887	191	100	100	100
MSH6	600678	81	100	100	97
MTAP	156540	213	100	100	100
MUTYH	604933	72	100	99	90
NBN	602667	175	100	100	100
NF1	613113	68	100	99	88
NF2	607379	55	97	90	79
NHP2	606470	141	100	100	100

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NOP10	606471	122	100	100	100
NOTCH2	600275	147	100	100	100
NRAS	164790	98	100	99	98
NSD1	606681	63	100	100	98
NTHL1	602656	77	100	100	98
NTRK1	191315	115	100	100	100
OGG1	601982	119	100	100	99
PALB2	610355	101	100	100	100
PALLD	608092	69	100	100	97
PARN	604212	66	100	100	96
PAX5	167414	59	100	99	92
PDGFRA	173490	106	100	100	95
PHOX2B	603851	79	100	98	95
PIK3CA	171834	149	100	100	100
PMS2	600259	85	100	99	96
POLD1	174761	148	100	96	93
POLE	174762	114	100	99	96
POLH	603968	117	100	100	100
POT1	606478	74	100	100	98
PRF1	170280	71	100	99	94
PRKAR1A	188830	120	100	100	100
PRSS1	276000	86	100	100	99
PRSS2	601564	239	100	100	100
PTCH1	601309	91	100	98	96
PTCH2	603673	108	100	100	100
PTEN	601728	115	85	78	76
PTPN11	176876	77	100	99	90
RAD50	604040	98	100	100	98
RAD51B	602948	70	100	100	98
RAD51C	602774	64	100	100	93
RAD51D	602954	117	100	100	100
RAF1	164760	78	100	99	95
RASAL1	604118	104	100	99	97
RB1	614041	163	100	100	98
RBBP6	600938	61	100	98	89
RBM8A	605313	99	100	100	100

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RECQL	600537	93	100	100	99
RECQL4	603780	148	100	100	99
REST	600571	69	100	100	97
RET	164761	173	100	100	100
RGS17	607191	47	98	87	77
RHBDF2	614404	100	100	99	96
RINT1	610089	55	100	97	90
RIT1	609591	72	100	100	100
RMRP	157660	No coverage data			
RNF43	612482	110	100	100	99
RPL11	604175	62	100	100	100
RPL15	604174	58	100	100	92
RPL35A	180468	86	100	100	100
RPL5	603634	54	99	93	85
RPS10	603632	61	100	100	97
RPS17	180472	2	5	5	5
RPS19	603474	97	100	100	92
RPS24	602412	105	100	100	100
RPS26	603701	81	100	100	100
RPS29	603633	69	100	100	100
RPS7	603658	54	100	100	89
RTEL1	608833	131	100	100	99
RUNX1	151385	76	100	100	98
SAMD9	610456	68	100	100	100
SAMD9L	611170	63	100	100	99
SBDS	607444	79	100	100	99
SDHA	600857	134	100	97	91
SDHAF2	613019	140	100	100	100
SDHB	185470	149	100	100	100
SDHC	602413	170	100	100	100
SDHD	602690	164	100	100	100
SERPINA1	107400	90	100	100	100
SFTPA1	178630	207	100	100	100
SFTPA2	178642	191	100	100	100
SH2D1A	300490	56	100	98	83
SHOC2	602775	61	100	99	95

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SLX4	613278	118	100	100	99
SMAD4	600993	79	100	100	98
SMAD9	603295	85	100	100	99
SMARCA4	603254	127	100	100	100
SMARCB1	601607	130	100	100	99
SMARCE1	603111	57	100	100	93
SOS1	182530	71	100	100	95
SPINK1	167790	79	100	100	97
SPRED1	609291	57	100	98	92
SRP72	602122	56	100	99	89
SRY	480000	159	100	100	100
STAT3	102582	90	100	100	99
STK11	602216	161	100	100	100
SUFU	607035	104	100	100	100
TERC	602322	No coverage data			
TERF1	600951	46	94	81	64
TERF2IP	605061	109	100	100	95
TERT	187270	136	100	100	99
TGFBR1	190181	171	94	93	93
TGFBR2	190182	210	100	100	100
TINF2	604319	155	100	100	100
TMEM127	613403	99	100	100	99
TNFRSF11A	603499	97	95	95	95
TP53	191170	167	100	100	100
TP63	603273	115	100	100	98
TRIM37	605073	61	100	100	94
TSC1	605284	167	100	100	100
TSC2	191092	184	100	100	100
UROD	613521	87	100	100	97
USB1	613276	93	100	100	99
VHL	608537	183	100	100	100
WAS	300392	66	97	84	76
WRAP53	612661	148	100	100	100
WRN	604611	63	100	100	95
WT1	607102	117	100	100	98
XPA	611153	71	100	100	93

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
XPC	613208	108	100	100	98
XRCC1	194360	97	100	100	100
XRCC2	600375	79	100	100	100
XRCC3	600675	101	100	100	99
XRCC4	194363	54	100	99	90

- 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