Whole Exome Sequencing
Gene package Oncogenetics, version 3.1, 30-9-2020

Technical information
DNA was enriched using Agilent SureSelect DNA + 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 Alissa Interpret 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	128	100	100	100
ACD	609377	237	100	100	100
ACTRT1	300487	105	100	100	100
ACVRL1	601284	298	100	100	100
AIP	605555	251	100	100	100
AKT1	164730	284	100	100	100
ALK	105590	174	100	100	99
ANKRD26	610855	43	74	58	48
APC	611731	99	100	99	97
ARMC5	615549	281	100	100	100
ATG2B	616226	75	99	95	87
ATM	607585	61	96	86	75
ATR	601215	76	98	92	83
AXIN2	604025	193	100	100	100
BAP1	603089	213	100	100	100
BARD1	601593	107	100	99	96
BLM	604610	64	99	90	76
BMPR1A	601299	103	100	100	96
BRAF	164757	84	100	99	94
BRCA1	113705	110	100	100	97
BRCA2	600185	74	99	96	90
BRIP1	605882	73	97	91	84
BUB1	602452	100	99	97	93
BUB1B	602860	99	98	95	89
BUB3	603719	130	100	99	96
CASR	601199	210	100	100	100

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CBL	165360	130	100	100	99
CDC73	607393	53	100	92	76
CDH1	192090	120	100	100	98
CDK4	123829	134	100	100	100
CDKN1A	116899	397	100	100	100
CDKN1B	600778	257	100	100	100
CDKN1C	600856	156	92	87	83
CDKN2A	600160	272	100	100	100
CDKN2B	600431	260	100	100	100
CDKN2C	603369	136	100	100	100
CEBPA	116897	177	100	100	94
CFTR	602421	124	98	94	88
CHEK2	604373	105	91	91	88
CHRNA3	118503	165	100	100	98
CHRNA5	118505	73	100	96	90
COL17A1	113811	157	100	98	97
CREBBP	600140	275	99	99	98
CSF3R	138971	236	100	100	100
CTC1	613129	174	100	100	100
CTNNA1	116805	109	100	100	97
CTRC	601405	271	100	100	100
CYLD	605018	81	97	88	74
DDB2	600811	179	100	100	100
DDX41	608170	236	100	99	98
DICER1	606241	95	100	99	96
DIS3L2	614184	153	100	97	96
DKC1	300126	73	100	93	88
DNAJC21	617048	64	100	96	86
DOCK8	611432	118	100	99	96
EGFR	131550	156	100	100	99
EGLN1	606425	181	100	100	98
ELANE	130130	211	100	100	100
ENG	131195	281	100	100	100
ERCC1	126380	143	100	100	100
ERCC2	126340	262	100	100	100
ERCC3	133510	178	100	100	100
ERCC4	133520	101	100	98	92
ERCC5	133530	87	99	96	89
ERCC6	609413	128	100	100	98
ETV6	600618	186	100	100	100
EXO1	606063	74	96	90	82
EXT1	608177	298	100	100	100
EXT2	608210	142	100	100	100
FAN1	613534	134	100	100	99

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FANCA	607139	163	100	98	96
FANCB	300515	38	94	88	69
FANCC	613899	115	100	97	94
FANCD2	613984	92	100	96	92
FANCE	613976	166	100	100	100
FANCF	613897	432	100	100	100
FANCG	602956	171	100	100	100
FANCI	611360	83	100	98	93
FANCL	608111	59	100	92	74
FANCM	609644	66	95	88	78
FAS	134637	114	91	83	74
FH	136850	83	100	100	98
FLCN	607273	218	100	100	100
FOCAD	614606	99	98	95	92
G6PC3	611045	161	100	99	95
GATA1	305371	112	100	100	99
GATA2	137295	235	100	100	100
GDNF	600837	114	100	100	100
GFI1	600871	221	100	100	99
GPC3	300037	86	100	98	92
GREM1	603054	158	100	100	100
GSKIP	616605	56	100	97	89
HABP2	603924	138	100	100	98
HAX1	605998	120	100	100	100
HNF1A	142410	285	100	100	100
HNF1B	189907	231	100	100	100
HOXB13	604607	302	100	100	100
HRAS	190020	426	100	100	100
ITK	186973	104	100	100	96
KIF1B	605995	104	100	100	98
KIT	164920	103	100	99	97
KLLN	612105	367	100	100	100
KRAS	190070	59	90	78	77
LZTR1	600574	242	100	100	100
MAP2K1	176872	148	100	100	99
MAP2K2	601263	276	100	100	100
MAX	154950	127	100	99	92
MBD4	603574	75	100	99	93
MC1R	155555	264	100	100	100
MEN1	613733	267	100	100	100
MET	164860	111	100	99	97
MITF	156845	119	100	100	93
MLH1	120436	140	100	100	99
MLH3	604395	100	100	99	97

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MPL	159530	229	100	100	100
MRE11	600814	61	97	88	76
MSH2	609309	88	96	91	84
MSH3	600887	75	99	92	80
MSH6	600678	137	100	100	98
MTAP	156540	109	100	100	97
MUTYH	604933	248	100	100	100
NBN	602667	57	98	83	71
NF1	613113	88	97	91	85
NF2	607379	165	100	100	100
NHP2	606470	210	100	100	100
NOP10	606471	174	100	100	100
NOTCH2	600275	188	100	98	97
NRAS	164790	107	100	100	99
NSD1	606681	208	100	99	98
NTHL1	602656	217	100	100	100
NTRK1	191315	272	100	100	100
OGG1	601982	201	100	100	100
PALB2	610355	103	100	99	97
PALLD	608092	112	100	100	98
PARN	604212	79	100	98	93
PAX5	167414	207	100	100	100
PDGFRA	173490	115	100	100	99
PHOX2B	603851	140	100	95	89
PIK3CA	171834	69	100	95	89
PMS2	600259	108	99	94	87
POLD1	174761	291	100	100	100
POLE	174762	195	100	100	100
POLH	603968	117	100	100	100
POT1	606478	66	99	95	83
PRF1	170280	383	100	100	100
PRKAR1A	188830	98	100	100	99
PRSS1	276000	209	100	100	100
PRSS2	601564	No coverage data			
PTCH1	601309	201	100	100	98
PTCH2	603673	280	100	100	100
PTEN	601728	100	99	94	87
PTPN11	176876	80	98	94	89
RAD50	604040	51	94	76	57
RAD51B	602948	88	99	96	90
RAD51C	602774	98	100	90	88
RAD51D	602954	134	100	100	100
RAF1	164760	124	100	98	97
RASAL1	604118	234	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
RB1	614041	54	90	74	62
RBBP6	600938	58	97	91	82
RBM8A	605313	82	100	100	95
RECQL	600537	47	95	80	65
RECQL4	603780	312	100	100	100
REST	600571	141	100	100	99
RET	164761	217	100	100	100
RGS17	607191	85	100	95	84
RHBDF2	614404	259	100	100	100
RINT1	610089	79	100	98	91
RIT1	609591	123	100	100	100
RMRP	157660	No coverage data			
RNF43	612482	226	100	100	100
RPL11	604175	155	100	100	100
RPL15	604174	90	100	100	99
RPL35A	180468	109	89	89	89
RPL5	603634	69	89	85	74
RPS10	603632	115	100	100	96
RPS17	180472	1	5	5	0
RPS19	603474	178	100	100	100
RPS24	602412	138	100	100	100
RPS26	603701	118	100	100	100
RPS29	603633	123	100	100	100
RPS7	603658	93	100	97	88
RTEL1	608833	260	100	100	100
RUNX1	151385	200	100	96	96
SAMD9	610456	73	99	94	89
SAMD9L	611170	85	100	100	99
SBDS	607444	131	100	100	99
SDHA	600857	155	100	100	100
SDHAF2	613019	76	100	100	98
SDHB	185470	123	100	100	98
SDHC	602413	143	100	100	100
SDHD	602690	123	100	100	100
SERPINA1	107400	176	100	100	100
SFTPA1	178630	298	100	100	100
SFTPA2	178642	255	100	100	100
SH2D1A	300490	81	100	95	85
SHOC2	602775	74	100	100	96
SLX4	613278	192	100	100	100
SMAD4	600993	78	100	97	95
SMAD9	603295	117	100	99	97
SMARCA4	603254	225	100	100	100
SMARCB1	601607	216	100	100	100

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SMARCE1	603111	102	100	94	83
SOS1	182530	74	94	89	79
SPINK1	167790	70	100	100	100
SPRED1	609291	108	100	100	98
SRP72	602122	79	100	93	87
SRY	480000	382	100	100	100
STAT3	102582	135	100	100	99
STK11	602216	310	100	100	100
SUFU	607035	206	100	100	100
TERC	602322	No coverage data			
TERF1	600951	61	57	50	45
TERF2IP	605061	184	100	100	99
TERT	187270	276	100	100	99
TGFBR1	190181	110	97	94	92
TGFBR2	190182	214	100	100	99
TINF2	604319	229	100	100	100
TMEM127	613403	194	100	100	100
TNFRSF11A	603499	178	95	95	95
TP53	191170	246	100	100	100
TP63	603273	134	100	100	99
TRIM37	605073	69	100	97	91
TSC1	605284	153	100	100	100
TSC2	191092	314	100	100	100
UROD	613521	191	100	100	100
USB1	613276	145	100	100	93
VHL	608537	136	100	100	100
WAS	300392	113	100	100	100
WRAP53	612661	213	100	100	100
WRN	604611	62	95	87	77
WT1	607102	175	100	100	100
XPA	611153	49	99	92	75
XPC	613208	138	100	100	99
XRCC1	194360	200	100	100	100
XRCC2	600375	109	100	89	86
XRCC3	600675	302	100	100	100
XRCC4	194363	39	95	77	54

- 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

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