Whole Exome Sequencing
Gene package Primary Immunodeficiency Disorders, version 8, 30-9-2020

Technical information
DNA was enriched using Agilent SureSelect DNA + Human All Exon V7capture 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
ACD	609377	237	100	100	100
ACP5	171640	280	100	100	100
ACTB	102630	253	100	100	100
ADA	608958	161	100	100	100
ADA2	607575	157	100	100	100
ADAM17	603639	98	99	94	88
ADAMTS3	605011	130	100	100	100
ADAR	146920	163	100	100	100
AGA	613228	83	100	98	90
AICDA	605257	191	100	100	100
AIRE	607358	248	100	100	100
AK2	103020	99	100	100	98
ALG13	300776	67	98	92	81
ALPI	171740	390	100	100	100
AP1S3	615781	101	100	100	99
AP3B1	603401	58	93	79	66
AP3D1	607246	241	99	98	97
APOL1	603743	170	100	100	100
ARHGEF1	601855	215	100	100	100
ARPC1B	604223	266	100	100	100
ATM	607585	61	96	86	75
ATP6AP1	300197	134	100	100	99
B2M	109700	131	100	100	100
BACH2	605394	212	100	100	100
BCL10	603517	82	96	85	77

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered 30x
BCL11B	606558	242	100	100	99
BLK	191305	167	100	100	100
BLM	604610	64	99	90	76
BLNK	604515	89	95	93	88
BLOC1S6	604310	65	100	78	57
BTK	300300	77	100	99	95
C1QA	120550	288	100	100	100
C1QB	120570	230	100	100	100
C1QC	120575	262	100	100	100
C1R	613785	219	100	100	100
C1S	120580	126	100	100	97
C2	613927	174	100	100	100
C3	120700	210	100	100	100
C5	120900	70	92	83	78
C6	217050	96	100	100	99
C7	217070	107	100	99	93
C8A	120950	141	100	100	100
C8B	120960	125	100	98	97
C8G	120930	278	100	100	100
C9	120940	78	100	96	93
CA2	611492	116	100	100	100
CARD11	607210	214	100	100	100
CARD14	607211	291	100	100	100
CARD9	607212	265	100	100	100
CARMIL2	610859	217	100	98	96
CASP10	601762	115	100	100	98
CASP8	601763	117	100	97	92
CAVIN1	603198	225	100	100	100
CCBE1	612753	163	95	95	95
CD19	107265	226	100	100	99
CD247	186780	180	100	100	100
CD27	186711	205	100	100	100
CD3D	186790	150	100	100	100
CD3E	186830	103	100	95	90
CD3G	186740	143	100	100	100
CD40	109535	158	100	100	100
CD40LG	300386	61	100	92	77
CD46	120920	75	99	95	85
CD55	125240	79	100	95	90
CD59	107271	136	100	100	100
CD70	602840	165	100	100	99
CD79A	112205	222	100	99	96
CD79B	147245	197	100	100	100
CD81	186845	187	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered 30x
CD8A	186910	221	100	100	100
CDCA7	609937	113	100	98	92
CDKN2B	600431	260	100	100	100
CEBPE	600749	294	100	100	100
CFB	138470	158	100	100	100
CFD	134350	226	100	100	100
CFH	134370	78	95	89	81
CFHR1	134371	58	93	86	76
CFHR3	605336	37	74	65	53
CFHR5	608593	65	98	90	80
CFI	217030	78	96	92	89
CFP	300383	156	100	100	100
CFTR	602421	124	98	94	88
CHD7	608892	167	100	99	97
CIB1	602293	235	100	100	100
CIITA	600005	286	100	100	100
CLCN7	602727	244	100	100	100
CLEC4D	609964	74	100	99	85
CLEC7A	606264	76	100	100	97
CLPB	616254	209	100	100	100
COG6	606977	66	94	83	74
COPA	601924	119	100	100	99
CORO1A	605000	287	92	92	92
CR2	120650	122	99	99	96
CREBBP	600140	275	99	99	98
CSF2RA	306250	47	46	45	40
CSF2RB	425000	226	100	100	100
CSF3R	138981	236	100	100	100
CTC1	138971	174	100	100	100
CTLA4	613129	153	100	100	100
CTPS1	123890	96	100	100	99
CTSC	123860	182	100	100	100
CXCR4	602365	190	100	100	100
CYBA	162643	164	100	98	96
CYBB	608508	65	100	98	91
CYBC1	300481	243	100	100	100
DBR1	607024	94	99	94	87
DCLRE1B	609683	184	100	100	100
DCLRE1C	605988	151	100	97	91
DDX58	609631	74	97	90	83
DEF6	610094	209	100	100	99
DGAT1	604900	341	99	96	94
DHFR	126060	57	93	71	43
DKC1	300126	73	100	93	88

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered 30x
DNAJC21	617048	64	100	96	86
DNASE1	125505	290	100	100	100
DNASE1L3	602244	108	100	100	99
DNASE2	126350	178	100	100	100
DNMT3B	602900	192	100	98	94
DOCK2	603122	118	100	100	100
DOCK8	611432	118	100	99	96
EFL1	617538	99	98	94	89
ELANE	130130	211	100	100	100
ELF4	300775	128	100	100	99
EPG5	615068	106	100	99	98
ERCC2	126340	262	100	100	100
ERCC3	133510	178	100	100	100
ERCC6L2	615667	66	99	93	86
EXTL3	605744	223	100	100	100
F12	610619	312	100	100	100
FAAP24	610884	100	100	100	100
FADD	602457	277	100	100	100
FAS	134637	114	91	83	74
FASLG	134638	106	100	100	99
FAT4	612411	170	100	99	98
FCGR1A	146760	158	97	97	97
FCGR2A	146790	239	100	100	100
FCGR2B	604590	184	76	72	71
FCGR3A	146740	215	100	100	100
FCGR3B	610665	160	100	100	100
FCHO1	613437	208	100	100	100
FCN3	604973	191	100	100	100
FERMT3	607901	243	100	100	100
FNIP1	610594	95	99	94	90
FOXN1	600838	201	100	100	100
FOXP3	300292	157	100	100	100
FPR1	136537	187	100	100	100
G6PC	613742	178	100	100	100
G6PC3	611045	161	100	99	95
G6PD	305900	211	100	100	100
GATA2	137295	235	100	100	100
GFI1	600871	221	100	100	99
GINS1	610608	115	100	100	96
GJC2	608803	283	100	100	98
GRHL2	608576	120	100	100	98
GTF2H5	608780	81	100	100	100
HAVCR2	606652	115	100	100	97
HAX1	605998	120	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered 30x
HELLS	603946	42	92	79	62
HMOX1	141250	187	100	100	100
HYOU1	601746	169	100	100	100
ICOS	604558	70	100	95	87
ICOSLG	605717	360	100	100	100
IFIH1	606951	70	99	91	80
IFNAR1	107450	43	95	81	62
IFNAR2	602376	113	100	95	84
IFNG	147570	52	100	89	73
IFNGR1	107470	103	92	91	91
IFNGR2	147569	101	100	100	92
IGLL1	146770	157	100	100	100
IKBKB	603258	159	100	100	100
IKBKG	300248	35	36	26	26
IKZF1	603023	257	100	100	99
IL10	124092	156	100	100	100
IL10RA	146933	181	100	100	100
IL10RB	123889	86	100	98	96
IL12B	161561	123	100	100	99
IL12RB1	601604	204	100	98	98
IL17F	606496	142	100	100	100
IL17RA	605461	346	100	100	100
IL17RC	610925	192	100	100	100
IL18BP	604113	304	100	100	100
IL1RN	147679	138	100	100	100
IL2	147680	58	100	95	76
IL21	605384	53	93	80	68
IL21R	605383	271	100	100	100
IL2RA	147730	139	100	100	100
IL2RB	146710	229	100	100	100
IL2RG	308380	88	100	100	97
IL36RN	605507	192	100	100	100
IL6R	147880	154	100	100	100
IL6ST	600694	79	99	92	81
IL7R	146661	105	100	100	98
INO80	610169	107	100	99	96
INSR	147670	243	100	100	99
IRAK1	300283	124	100	100	98
IRAK4	606883	51	96	85	74
IRF2BP2	615332	161	100	100	100
IRF3	603734	235	100	100	100
IRF4	601900	204	100	100	100
IRF7	605047	236	100	100	100
IRF8	601565	157	100	100	97

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IRF9	147574	197	100	100	100
ISG15	147571	418	100	100	100
ITCH	606409	61	96	90	80
ITGB2	600065	241	100	100	100
ITK	186973	104	100	100	96
IVNS1ABP	609209	93	100	99	95
JAGN1	616012	191	100	100	100
JAK1	147795	110	100	98	94
JAK2	147796	55	98	91	79
JAK3	600173	266	100	100	100
KDM6A	300128	57	95	86	73
KMT2A	159555	118	100	100	98
KMT2D	602113	341	100	100	100
LACC1	613409	72	100	98	91
LAMTOR2	610389	239	100	100	100
LAT	602354	181	100	100	100
LCK	153390	183	100	100	100
LIG1	126391	172	100	100	100
LIG4	601837	86	100	97	93
LPIN2	605519	125	100	100	99
LRBA	606453	90	99	96	91
LRRC8A	608360	319	100	100	100
LYST	606897	87	97	92	89
MAGT1	300715	69	93	87	78
MAL	188860	148	100	100	100
MALT1	604860	75	97	88	78
MAN2B1	609458	213	100	100	100
MANBA	609489	93	100	99	94
MAP3K14	604655	196	100	100	100
MASP2	605102	160	100	100	100
MBL2	154545	162	100	100	99
MC2R	607397	210	100	100	100
MCM4	602638	154	97	96	92
MEFV	608107	158	100	100	100
MOGS	601336	229	100	100	100
MRE11	600814	61	97	88	76
MRTFA	606078	212	100	100	100
MS4A1	112210	59	95	82	67
MSN	309845	99	100	100	97
MTHFD1	172460	118	100	99	96
MVK	251170	207	100	100	100
MYD88	602170	228	100	100	100
MYSM1	612176	55	90	82	72
NBAS	608025	103	100	98	94

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered 30x
NBN	602667	57	98	83	71
NCF1	608512	62	63	58	56
NCF2	608515	213	100	100	100
NCF4	601488	170	100	100	100
NCKAP1L	141180	112	100	99	96
NCSTN	605254	140	100	100	100
NFAT5	604708	96	99	97	92
NFE2L2	600492	137	100	100	99
NFKB1	164011	112	100	99	95
NFKB2	164012	245	100	100	100
NFKBIA	164008	168	100	100	100
NHEJ1	611290	108	100	100	96
NHP2	606470	210	100	100	100
NLRC4	606831	127	100	97	94
NLRP1	606636	173	100	100	100
NLRP12	609648	246	100	100	100
NLRP3	606416	404	100	100	100
NOD2	605956	268	100	100	100
NOP10	606471	174	100	100	100
NRAS	164790	107	100	100	99
NSMCE3	608243	164	100	100	100
OAS1	164350	131	100	100	100
ORAI1	610277	276	100	99	99
OSTM1	607649	118	93	70	64
OTULIN	615712	81	98	91	78
PARN	604212	79	100	98	93
PAX5	167414	207	100	100	100
PBX1	176310	97	100	100	96
PCCA	232000	71	98	93	87
PCCB	232050	159	100	98	95
PEPD	613230	216	100	100	100
PGM3	172100	97	100	97	91
PIGA	311770	62	100	99	86
PIK3CD	602839	284	100	100	100
PIK3R1	171833	65	100	93	82
PLCG2	600220	146	100	100	98
PLEKHM1	611466	191	99	98	98
PLG	173350	118	100	99	97
PMM2	601785	115	100	100	99
PNP	164050	329	100	100	98
POLA1	312040	47	94	84	70
POLE2	602670	45	92	79	65
POMP	613386	76	85	85	85
POT1	606478	66	99	95	83

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered 30x
PRF1	170280	383	100	100	100
PRKCD	176977	201	100	100	100
PRKDC	600899	99	97	94	89
PRPS1	311850	70	100	98	92
PSENEN	607632	150	100	100	100
PSMA3	176843	57	93	86	71
PSMB4	602177	141	100	100	100
PSMB8	177046	205	100	100	100
PSMB9	177045	206	100	100	100
PSMG2	609702	87	100	96	90
PSTPIP1	606347	257	100	100	100
PTPN22	600716	74	98	96	88
PTPRC	151460	62	93	84	76
RAB27A	603868	88	100	100	97
RAC2	602049	233	100	100	100
RAG1	179615	199	100	100	100
RAG2	179616	130	100	100	100
RANBP2	601181	91	95	90	85
RASGRP1	603962	138	100	100	100
RASGRP2	605577	310	100	100	100
RBCK1	610924	200	100	100	100
RECQL4	603780	312	100	100	100
RELB	604758	191	100	100	100
RFX5	601863	180	100	100	100
RFXANK	603200	218	100	100	100
RFXAP	601861	136	100	100	100
RHOH	602037	299	100	100	100
RIPK1	603453	112	100	96	95
RMRP	157660	No coverage data			
RNASEH2A	606034	265	100	100	100
RNASEH2B	610326	60	100	99	89
RNASEH2C	610330	261	100	100	100
RNF168	612688	137	99	97	93
RNF31	612487	216	100	100	100
RNU4ATAC	601428	No coverage data			
RORC	602943	171	100	100	100
RPSA	150370	110	100	100	95
RSPH9	612648	215	100	100	100
RTEL1	608833	260	100	100	100
SAMD9	610456	73	99	94	89
SAMD9L	611170	85	100	100	99
SAMHD1	606754	97	100	99	93
SBDS	607444	131	100	100	99
SEC61A1	609213	142	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered 30x
SEMA3E	608166	91	93	91	87
SERAC1	614725	98	98	95	89
SERPING1	606860	113	100	100	99
SH2B3	605093	211	100	100	100
SH2D1A	300490	81	100	95	85
SH3BP2	602104	248	94	91	91
SH3KBP1	300374	85	100	97	92
SKIV2L	600478	190	100	100	100
SLC29A3	612373	233	100	100	100
SLC35A1	605634	69	100	99	95
SLC35C1	605881	329	100	100	100
SLC37A4	602671	193	100	100	100
SLC39A4	607059	284	100	100	100
SLC39A7	601416	196	100	100	100
SLC46A1	611672	271	100	100	100
SLC7A7	603593	146	100	100	100
SMARCAL1	606622	146	100	100	98
SMARCD2	601736	191	87	87	87
SNX10	614780	71	100	100	98
SOCS4	616337	120	100	99	94
SP110	604457	117	96	93	92
SPINK5	605010	65	96	87	77
SPPL2A	608238	53	94	84	69
SRP54	604857	43	95	82	64
SRP72	602122	79	100	93	87
STAT1	600555	91	97	95	90
STAT2	600556	144	100	100	100
STAT3	102582	135	100	100	99
STAT4	600558	75	97	93	87
STAT5B	604260	157	100	100	100
STAT6	601512	162	100	100	99
STIM1	605921	202	100	100	100
STING1	612374	214	100	100	100
STK4	604965	97	100	95	81
STN1	613128	112	100	100	98
STX11	605014	478	100	100	100
STXBP2	601717	220	100	100	100
TAP1	170260	172	100	100	100
TAP2	170261	166	100	100	100
TAPBP	601962	233	100	100	100
TAZ	300394	145	100	100	100
TBX1	602054	131	94	90	87
TCF3	147141	205	100	100	100
TCIRG1	604592	242	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered 30x
TCN2	613441	190	100	100	100
TERC	602322	No coverage data			
TERT	187270	276	100	100	99
TFRC	190010	68	100	99	92
TGFB1	190180	274	100	100	100
THBD	188040	398	100	100	100
TICAM1	607601	256	100	100	100
TINF2	604319	229	100	100	100
TIRAP	606252	353	100	100	100
TLR3	603029	87	100	97	91
TLR4	603030	130	100	100	100
TLR7	300365	68	100	99	92
TMC6	605828	223	100	100	100
TMC8	605829	156	100	100	100
TNFAIP3	191163	169	100	99	97
TNFRSF11A	603499	178	95	95	95
TNFRSF13B	604907	241	100	100	100
TNFRSF13C	606269	135	100	98	90
TNFRSF1A	191190	204	100	100	100
TNFRSF4	600315	201	100	100	100
TNFRSF9	602250	83	100	99	92
TNFSF11	602642	119	100	100	100
TNFSF12	602695	123	100	100	98
TOM1	604700	215	100	100	100
TOP2B	126431	60	92	79	68
TPP2	190470	72	96	91	87
TRAF3	601896	156	98	92	91
TRAF3IP2	607043	157	100	100	100
TREX1	606609	313	100	100	100
TRIM22	606559	119	100	100	99
TRNT1	612907	47	90	86	74
TTC37	614589	65	99	96	91
TTC7A	609332	192	100	100	100
TYK2	176941	276	100	100	100
UNC13D	608897	248	100	100	100
UNC93B1	608204	159	83	75	69
UNG	191525	123	100	100	99
USB1	613276	145	100	100	93
USP18	607057	132	95	95	95
VAV1	164875	176	100	100	100
VPS13B	607817	98	99	95	89
VPS45	610035	72	100	93	92
WAS	300392	113	100	100	100
WDR1	604734	239	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered 30x
WIPF1	602357	186	100	100	99
WRAP53	612661	213	100	100	100
XIAP	300079	63	94	89	81
ZAP70	176947	312	100	100	100
ZBTB24	614064	155	100	100	100
ZNF341	618269	178	100	100	100

- 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