Whole Exome Sequencing
Gene package Primary Immunodeficiency Disorders, version 7, 21-2-2020

Technical information
DNA was enriched using Agilent SureSelect DNA + SureSelect OneSeq 300kb CNV Backbone + 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 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
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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	139	100	100	100
ACP5	171640	143	100	100	100
ACTB	102630	192	100	100	100
ADA	608958	85	100	100	96
ADA2	607575	96	100	100	99
ADAM17	603639	67	100	99	94
ADAMTS3	605011	62	100	99	95
ADAR	146920	90	100	100	100
AGA	613228	73	100	100	96
AICDA	605257	134	100	100	95
AIRE	607358	100	100	100	100
AK2	103020	68	100	100	97
ALG13	300776	50	100	97	83
ALPI	171740	200	100	100	100
AP1S3	615781	49	100	100	93
AP3B1	603401	66	100	98	87
AP3D1	607246	103	99	98	97

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered 30x
APOL1	603743	113	100	100	100
ARHGEF1	601855	97	100	100	98
ARPC1B	604223	107	100	100	100
ATM	607585	75	100	99	95
ATP6AP1	300197	76	100	99	95
B2M	109700	94	100	100	100
BACH2	605394	118	100	100	100
BCL10	603517	66	100	100	96
BCL11B	606558	106	100	97	92
BLK	191305	113	100	100	100
BLM	604610	82	100	100	96
BLNK	604515	60	100	98	88
BLOC1S6	604310	46	100	100	84
BTK	300300	50	100	98	83
C1QA	120550	155	100	100	100
C1QB	120570	121	100	100	100
C1QC	120575	150	100	100	100
C1R	613785	125	100	100	100
C1S	120580	96	100	100	98
C2	613927	106	100	100	100
C3	120700	127	100	100	99
C5	120900	72	100	98	94
C6	217050	65	100	99	94
C7	217070	79	100	99	96
C8A	120950	69	100	100	98
C8B	120960	100	100	100	98
C8G	120930	151	100	100	100
C9	120940	66	100	100	97
CA2	611492	99	100	100	100
CARD11	607210	108	100	100	99
CARD14	607211	112	100	99	99
CARD9	607212	122	100	100	99
CARMIL2	610859	125	100	96	94
CASP10	601762	75	100	100	97
CASP8	601763	87	100	100	100
CAVIN1	603198	172	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered 30x
CCBE1	612753	87	100	99	95
CD19	107265	107	100	100	99
CD247	186780	74	100	100	100
CD27	186711	95	100	100	100
CD3D	186790	100	100	100	99
CD3E	186830	113	100	100	100
CD3G	186740	76	100	100	99
CD40	109535	104	100	100	100
CD40LG	300386	67	100	100	97
CD46	120920	88	100	100	95
CD55	125240	55	91	85	79
CD59	107271	86	100	100	100
CD70	602840	100	100	100	100
CD79A	112205	83	100	91	85
CD79B	147245	146	100	100	100
CD81	186845	146	97	90	90
CD8A	186910	100	100	100	97
CDCA7	609937	61	100	99	92
CDKN2B	600431	105	100	100	100
CEBPE	600749	57	100	100	99
CFB	138470	106	100	100	100
CFD	134350	99	100	98	90
CFH	134370	78	100	100	96
CFHR1	134371	65	93	93	83
CFHR3	605336	55	100	93	87
CFHR5	608593	75	100	100	97
CFI	217030	67	100	99	93
CFP	300383	68	100	95	86
CFTR	602421	93	100	100	97
CHD7	608892	87	100	100	97
CIB1	602293	100	100	94	92
CIITA	600005	117	100	100	99
CLCN7	602727	128	100	100	99
CLEC4D	609964	52	100	100	93
CLEC7A	606264	64	100	100	91
CLPB	616254	114	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered 30x
COG6	606977	65	100	99	90
COPA	601924	75	100	100	99
CORO1A	605000	122	92	92	91
CR2	120650	63	100	100	97
CREBBP	600140	85	100	99	94
CSF2RA	306250	0	0	0	0
CSF2RB	425000	132	100	100	100
CSF3R	138981	98	100	100	100
CTC1	138971	100	100	100	100
CTLA4	613129	90	100	100	100
CTPS1	123890	70	100	100	97
CTSC	123860	75	100	100	97
CXCR4	602365	82	83	83	82
CYBA	162643	106	94	81	66
CYBB	608508	49	100	98	91
CYBC1	300481	119	100	100	98
DBR1	607024	56	99	95	85
DCLRE1B	609683	95	100	100	98
DCLRE1C	605988	64	100	100	92
DDX58	609631	68	100	100	96
DEF6	610094	112	100	100	96
DGAT1	604900	116	96	91	85
DHFR	126060	76	100	100	98
DKC1	300126	53	100	98	89
DNAJC21	617048	62	100	96	89
DNASE1	125505	132	100	100	100
DNASE1L3	602244	67	100	100	98
DNASE2	126350	95	100	100	100
DNMT3B	602900	115	100	100	99
DOCK2	603122	82	100	100	97
DOCK8	611432	74	100	99	94
EFL1	617538	60	99	97	92
ELANE	130130	127	100	100	100
ELF4	300775	67	100	100	100
EPG5	615068	69	100	100	97
ERCC2	126340	97	100	99	98

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered 30x
ERCC3	133510	82	100	100	99
ERCC6L2	615667	57	100	98	93
EXTL3	605744	150	100	100	100
F12	610619	135	100	100	100
FAAP24	610884	97	100	100	100
FADD	602457	143	100	100	100
FAS	134637	264	100	100	96
FASLG	134638	65	100	100	100
FAT4	612411	90	100	100	99
FCGR1A	146760	83	97	97	93
FCGR2A	146790	150	100	100	100
FCGR2B	604590	99	84	82	78
FCGR3A	146740	244	100	100	98
FCGR3B	610665	210	100	100	100
FCHO1	613437	86	100	97	92
FCN3	604973	103	100	100	97
FERMT3	607901	114	100	100	97
FNIP1	610594	59	100	99	94
FOXN1	600838	136	100	100	100
FOXP3	300292	69	100	99	88
FPR1	136537	129	100	100	100
G6PC	613742	113	100	100	100
G6PC3	611045	103	100	100	100
G6PD	305900	96	100	100	100
GATA2	137295	100	100	100	100
GFI1	600871	136	100	100	100
GINS1	610608	65	100	100	91
GJC2	608803	85	96	86	76
GRHL2	608576	81	100	100	99
GTF2H5	608780	53	100	100	100
HAVCR2	606652	58	100	99	92
HAX1	605998	99	100	100	99
HELLS	603946	61	100	96	84
HMOX1	141250	112	100	100	99
HYOU1	601746	112	100	100	98
ICOS	604558	57	100	100	95

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ICOSLG	605717	93	100	97	95
IFIH1	606951	86	100	100	96
IFNAR1	107450	61	100	97	88
IFNAR2	602376	93	100	100	97
IFNGR1	107470	68	100	100	95
IFNGR2	147569	64	97	92	90
IGLL1	146770	132	100	100	100
IKBKB	603258	86	100	100	97
IKBKG	300248	24	38	33	31
IKZF1	603023	166	100	100	100
IL10	124092	120	100	100	99
IL10RA	146933	125	100	100	99
IL10RB	123889	69	100	100	97
IL12B	161561	77	100	100	98
IL12RB1	601604	116	100	100	100
IL17F	606496	79	100	100	100
IL17RA	605461	125	100	100	100
IL17RC	610925	109	100	100	99
IL18BP	604113	149	100	100	100
IL1RN	147679	76	100	100	90
IL2	147680	42	100	88	77
IL21	605384	69	100	99	89
IL21R	605383	113	100	100	100
IL2RA	147730	96	100	100	100
IL2RB	146710	84	100	100	99
IL2RG	308380	54	100	99	91
IL36RN	605507	82	100	100	100
IL6R	147880	132	100	100	99
IL6ST	600694	64	100	96	83
IL7R	146661	60	100	100	99
INO80	610169	73	100	99	92
INSR	147670	115	100	98	95
IRAK1	300283	62	100	97	86
IRAK4	606883	72	100	99	91
IRF2BP2	615332	82	100	100	91
IRF3	603734	132	100	100	97

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered 30x
IRF4	601900	147	100	100	100
IRF7	605047	128	100	99	98
IRF8	601565	136	100	100	99
IRF9	147574	123	100	100	99
ISG15	147571	144	100	100	100
ITCH	606409	61	96	95	90
ITGB2	600065	140	100	100	100
ITK	186973	76	100	100	95
JAGN1	616012	91	100	100	100
JAK1	147795	71	100	98	93
JAK2	147796	63	100	99	93
JAK3	600173	104	100	100	99
KDM6A	300128	52	100	95	80
KMT2A	159555	71	100	100	98
KMT2D	602113	114	100	100	99
LACC1	613409	62	100	98	93
LAMTOR2	610389	108	100	100	100
LAT	602354	91	100	100	97
LCK	153390	160	100	100	100
LIG1	126391	94	100	100	99
LIG4	601837	71	100	100	100
LPIN2	605519	77	100	100	98
LRBA	606453	65	100	99	94
LRRC8A	608360	174	100	100	100
LTBP3	602090	130	100	99	96
LYST	606897	72	100	99	95
MAGT1	300715	47	100	99	85
MAL	188860	102	100	100	98
MALT1	604860	68	97	93	89
MAN2B1	609458	115	100	100	100
MANBA	609489	85	100	100	95
MAP3K14	604655	113	100	100	100
MASP2	605102	85	100	100	97
MBL2	154545	122	100	100	100
MC2R	607397	102	100	100	100
MCM4	602638	88	100	100	97

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered 30x
MEFV	608107	105	100	100	100
MOGS	601336	130	100	100	100
MRE11	600814	55	100	97	83
MRTFA	606078	94	98	97	92
MS4A1	112210	80	100	100	98
MSN	309845	51	99	92	78
MTHFD1	172460	76	100	100	96
MVK	251170	100	100	100	100
MYD88	602170	157	100	100	100
MYSM1	612176	55	100	98	90
NBAS	608025	66	100	99	94
NBN	602667	68	100	99	88
NCF1	608512	108	68	65	61
NCF2	608515	94	100	100	98
NCF4	601488	121	100	100	100
NCSTN	605254	91	100	100	98
NFAT5	604708	64	100	99	96
NFE2L2	600492	67	100	99	94
NFKB1	164011	56	100	98	89
NFKB2	164012	143	100	100	96
NFKBIA	164008	131	100	100	100
NHEJ1	611290	55	100	100	96
NHP2	606470	122	100	100	100
NLRC4	606831	83	100	100	97
NLRP1	606636	103	100	100	100
NLRP12	609648	135	100	100	100
NLRP3	606416	125	100	100	100
NOD2	605956	113	100	100	99
NOP10	606471	147	100	100	100
NRAS	164790	63	100	100	98
NSMCE3	608243	161	100	100	100
OAS1	164350	108	100	100	100
ORAI1	610277	175	99	99	97
OSTM1	607649	79	100	100	97
OTULIN	615712	70	97	91	82
PARN	604212	59	100	99	92

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered 30x
PAX5	167414	106	100	100	95
PBX1	176310	72	100	100	98
PCCA	232000	68	100	100	95
PCCB	232050	72	100	98	95
PEPD	613230	100	100	100	98
PGM3	172100	88	100	100	98
PIGA	311770	67	100	100	97
PIK3CD	602839	137	100	99	98
PIK3R1	171833	80	100	100	97
PLCG2	600220	107	100	100	99
PLEKHM1	611466	174	100	100	99
PLG	173350	81	100	100	98
PMM2	601785	72	100	100	97
PNP	164050	70	100	100	97
POLA1	312040	44	99	92	77
POLE2	602670	40	99	92	69
POMP	613386	45	100	100	87
POT1	606478	71	100	99	94
PRF1	170280	120	100	100	100
PRKCD	176977	115	100	100	100
PRKDC	600899	73	100	99	94
PRPS1	311850	50	100	99	90
PSENEN	607632	88	100	100	100
PSMA3	176843	53	100	99	87
PSMB4	602177	73	100	100	100
PSMB8	177046	124	100	100	100
PSMB9	177045	59	100	90	90
PSMG2	609702	58	100	99	86
PSTPIP1	606347	109	100	100	99
PTPN22	600716	54	100	99	92
PTPRC	151460	73	100	99	95
RAB27A	603868	54	100	100	91
RAC2	602049	99	100	100	100
RAG1	179615	101	100	100	100
RAG2	179616	71	100	100	100
RANBP2	601181	112	100	100	99

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered 30x
RASGRP1	603962	70	100	100	98
RASGRP2	605577	93	100	100	100
RBCK1	610924	135	100	100	99
RECQL4	603780	148	100	100	99
RELB	604758	88	100	98	93
RFX5	601863	100	100	100	98
RFXANK	603200	118	100	100	100
RFXAP	601861	148	100	100	100
RHOH	602037	101	100	100	100
RIPK1	603453	54	100	97	91
RMRP	157660	No coverage data			
RNASEH2A	606034	109	100	100	100
RNASEH2B	610326	62	100	98	88
RNASEH2C	610330	323	100	100	100
RNF168	612688	93	100	100	99
RNF31	612487	121	100	100	99
RNU4ATAC	601428	No coverage data			
RORC	602943	97	100	100	99
RPSA	150370	84	100	100	100
RSPH9	612648	125	100	100	100
RTEL1	608833	131	100	100	99
SAMD9	610456	68	100	100	100
SAMD9L	611170	63	100	100	99
SAMHD1	606754	64	100	99	88
SBDS	607444	79	100	100	99
SEC61A1	609213	76	100	100	96
SEMA3E	608166	64	100	100	94
SERAC1	614725	65	100	99	86
SERPING1	606860	112	100	100	99
SH2B3	605093	115	100	100	100
SH2D1A	300490	56	100	98	83
SH3BP2	602104	127	91	91	91
SH3KBP1	300374	51	99	89	69
SKIV2L	600478	124	100	100	100
SLC29A3	612373	152	100	99	99
SLC35A1	605634	66	100	100	97

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered 30x
SLC35C1	605881	146	100	100	100
SLC37A4	602671	96	100	100	95
SLC39A4	607059	125	100	100	100
SLC39A7	601416	110	100	100	100
SLC46A1	611672	121	100	100	98
SLC7A7	603593	79	100	100	97
SMARCAL1	606622	88	100	100	98
SMARCD2	601736	77	87	86	84
SNX10	614780	77	100	100	98
SOCS4	616337	78	100	100	100
SP110	604457	61	100	100	96
SPINK5	605010	66	100	99	95
SPPL2A	608238	32	92	71	46
SRP54	604857	56	100	97	89
SRP72	602122	56	100	99	89
STAT1	600555	64	100	99	94
STAT2	600556	87	100	100	97
STAT3	102582	90	100	100	99
STAT4	600558	63	100	99	93
STAT5B	604260	107	100	100	97
STAT6	601512	95	100	100	98
STIM1	605921	95	100	100	99
STING1	612374	122	100	100	100
STK4	604965	82	100	100	99
STN1	613128	62	100	97	90
STX11	605014	308	100	100	100
STXBP2	601717	121	100	100	100
TAP1	170260	125	100	100	100
TAP2	170261	106	100	100	100
TAPBP	601962	106	100	100	100
TAZ	300394	97	100	98	90
TBX1	602054	94	94	85	80
TCF3	147141	81	100	100	96
TCIRG1	604592	117	100	100	97
TCN2	613441	118	100	100	100
TERC	602322	No coverage data			

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered 30x
TERT	187270	136	100	100	99
TFRC	190010	58	100	99	93
TGFB1	190180	111	100	99	95
THBD	188040	195	100	100	100
TICAM1	607601	127	100	100	98
TINF2	604319	155	100	100	100
TIRAP	606252	158	100	100	100
TLR3	603029	66	100	100	99
TLR4	603030	61	100	100	99
TMC6	605828	86	100	100	97
TMC8	605829	118	100	100	100
TNFAIP3	191163	106	100	99	96
TNFRSF11A	603499	97	95	95	95
TNFRSF13B	604907	106	100	100	100
TNFRSF13C	606269	78	100	91	73
TNFRSF1A	191190	97	100	98	94
TNFRSF4	600315	61	99	89	82
TNFRSF9	602250	70	100	100	99
TNFSF11	602642	56	100	100	94
TNFSF12	602695	68	100	97	92
TOM1	604700	115	100	100	100
TOP2B	126431	49	100	95	82
TPP2	190470	58	100	99	91
TRAC	186880	76	100	100	100
TRAF3	601896	102	100	100	100
TRAF3IP2	607043	94	100	100	99
TREX1	606609	234	100	100	100
TRIM22	606559	86	100	100	98
TRNT1	612907	68	100	99	92
TTC37	614589	58	100	99	93
TTC7A	609332	110	100	100	99
TYK2	176941	128	100	100	99
UNC13D	608897	100	100	100	100
UNC93B1	608204	69	96	87	76
UNG	191525	104	100	100	100
USB1	613276	93	100	100	99

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered 30x
USP18	607057	117	95	95	93
VAV1	164875	98	100	100	95
VPS13B	607817	75	100	99	96
VPS45	610035	74	100	100	98
WAS	300392	66	97	84	76
WDR1	604734	77	100	99	94
WIPF1	602357	73	100	100	96
WRAP53	612661	148	100	100	100
XIAP	300079	47	100	95	80
ZAP70	176947	151	100	100	100
ZBTB24	614064	73	100	100	100
ZNF341	618269	111	100	99	96

- 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