

Whole Exome Sequencing

Gene package Primary Immunodeficiency Disorders, version 10, 30-9-2021



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/>). Sequence variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: <http://www.broadinstitute.org/gatk/>). The detected sequence variants are filtered and annotated with Alissa Interpret software and classified with Alamut Visual. Copy 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 filtered and annotated with the NexusClinical software and classified using UCSC Genome Browser (NCBI37/hg19). It is not excluded that pathogenic variants 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).



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 $\geq 50x$
ACD	609377	100	100	100	99.57
ACP5	171640	100	100	100	100
ACTB	102630	100	100	100	99.59
ADA	608958	100	100	100	94.61
ADA2	607575	100	100	100	100
ADAM17	603639	100	99.37	97.32	91.29
ADAMTS3	605011	100	100	100	99.73
ADAR	146920	98.45	98.45	98.45	98.30
ADGRE2	606100	100	100	99.09	95.05
AGA	613228	100	100	100	97.37
AICDA	605257	100	100	99.86	80.84
AIRE	607358	100	100	97.60	86.34
AK2	103020	100	100	96.18	88.71
ALG13	300776	100	100	100	95.01
ALPI	171740	100	100	100	98.28
ANGPT1	601667	100	100	100	99.79
AP1S3	615781	100	100	95.91	95.91
AP3B1	603401	97.72	95.97	94.31	85.98
AP3D1	607246	97.71	97.71	96.35	90.76

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
APOL1	603743	100	100	100	99.93
ARHGEF1	601855	100	99.60	97.56	86.77
ARPC1B	604223	100	100	100	98.53
ATG4A	300663	100	100	100	97.86
ATM	607585	100	100	82.83	52.02
ATP6AP1	300197	100	98.66	89.10	84.13
B2M	109700	100	100	100	100
BACH2	605394	100	100	100	99.54
BCL10	603517	100	100	94.80	87.94
BCL11B	606558	100	99.08	97.43	90.55
BLK	191305	100	99.17	97.04	87.32
BLM	604610	100	99.64	98.63	94.07
BLNK	604515	99.94	96.78	93.33	88.41
BLOC1S6	604310	100	99.45	92.16	78.29
BTK	300300	100	100	100	97.97
C1QA	120550	100	100	100	100
C1QB	120570	100	100	100	97.43
C1QC	120575	100	100	100	93.55
C1R	613785	100	100	99.79	96.81
C1S	120580	100	100	100	96.76
C2	613927	100	100	100	97.52
C3	120700	100	100	99.61	96.57
C5	120900	100	99.46	97.40	91.16
C6	217050	100	100	100	100
C7	217070	100	100	100	100
C8A	120950	100	100	100	99.09
C8B	120960	100	100	100	100
C8G	120930	100	100	96.18	74.66
C9	120940	100	100	100	100
CA2	611492	100	100	100	100
CARD11	607210	100	99.85	97.03	94.88
CARD14	607211	100	99.33	97.88	90.84
CARD9	607212	100	99.13	97.19	90.15
CARMIL2	610859	96.75	94.72	93.26	87.43
CASP10	601762	100	100	100	100
CASP8	601763	100	100	98.57	93.79
CAVIN1	603198	100	100	100	97.27
CCBE1	612753	100	100	100	96.49
CD19	107265	100	100	98.74	93.52
CD247	186780	85.23	85.23	85.23	85.23

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CD27	186711	100	100	100	99.44
CD3D	186790	100	100	100	100
CD3E	186830	100	100	100	99.94
CD3G	186740	100	100	100	100
CD40	109535	100	100	100	100
CD40LG	300386	100	100	100	100
CD46	120920	100	100	100	96.67
CD55	125240	100	97.14	94.22	82.31
CD59	107271	100	100	100	100
CD70	602840	100	100	100	98.79
CD79A	112205	100	92.03	85.22	67.99
CD79B	147245	100	100	100	100
CD81	186845	100	100	97.41	82.89
CD8A	186910	100	100	99.15	87.58
CDC42	116952	100	100	100	100
CDCA7	609937	100	98.55	95.55	89.42
CDKN2B	600431	100	100	99.45	90.31
CEBPE	600749	100	100	100	99.89
CFB	138470	100	100	100	99.51
CFD	134350	100	99.89	89.26	67.95
CFH	134370	99.81	98.52	95.51	92.17
CFHR1	134371	100	91.77	91.77	91.77
CFHR3	605336	100	100	100	88.42
CFHR5	608593	100	99.48	97.17	91.87
CFI	217030	86.45	83.17	81.24	78.74
CFP	300383	100	99.21	96.28	82.23
CFTR	602421	100	100	100	97.15
CHD7	608892	100	100	99.90	98.10
CIB1	602293	100	100	97.12	89.80
CIITA	600005	100	99.56	97.15	88.42
CLCN7	602727	100	100	99.90	94.44
CLEC4D	609964	100	100	100	96.73
CLEC7A	606264	100	100	100	100
CLPB	616254	100	100	100	98.66
COG6	606977	100	100	100	94.13
COPA	601924	100	100	100	99.01
CORO1A	605000	92.29	92.10	90.97	87.68
CR2	120650	100	100	100	98.25
CREBBP	600140	100	99.23	97.31	92.60
CSF2RA	306250	100	100	100	99.38

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CSF2RB	425000	100	99.02	96.44	91.07
CSF3R	138981	100	99.07	95.95	91.73
CTC1	138971	100	100	99.94	95.90
CTLA4	613129	100	100	100	100
CTNBL1	123890	100	100	99.40	93.38
CTPS1	611537	100	100	98.72	92.36
CTSC	123860	100	100	100	100
CXCR4	602365	100	100	99.77	93.79
CYBA	162643	94.18	84.18	69.36	51.70
CYBB	608508	100	100	99.37	90.15
CYBC1	300481	100	99.85	93.76	84.58
DBR1	618334	100	100	100	97.82
DCLRE1B	607024	100	100	100	98.37
DCLRE1C	609683	100	100	99.94	92.61
DDX58	605988	100	100	99.44	97.27
DEF6	609631	100	98.56	95.58	90.53
DGAT1	610094	94.65	87.80	87.80	87.80
DHFR	604900	100	100	99.93	87.44
DKC1	126060	100	99.10	97.39	86.54
DNAJC21	300126	100	99.90	98.39	88.33
DNASE1	617048	100	100	100	99.38
DNASE1L3	125505	100	100	100	98.45
DNASE2	602244	100	100	98	89.25
DNMT3B	126350	100	100	99.60	94.18
DOCK2	602900	100	99.64	99.04	97.67
DOCK8	603122	100	99.90	99.32	96.02
EFL1	611432	100	100	100	99.24
ELANE	617538	100	100	100	96.34
ELF4	130130	100	98.98	95.21	87.69
EPG5	300775	100	99.91	99.15	95.77
ERCC2	615068	100	100	100	95.44
ERCC3	126340	100	100	100	97.49
ERCC6L2	133510	100	100	99.91	96.00
EXTL3	615667	100	100	100	100
F12	605744	100	100	100	98.07
FAAP24	610619	100	100	100	99.17
FADD	610884	100	100	100	100
FAS	602457	100	100	100	87.44
FASLG	134637	100	100	97.83	91.50
FAT4	134638	100	100	99.98	99.68

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FCGR1A	612411	100	100	95.77	95.77
FCGR2A	146760	100	100	100	100
FCGR2B	146790	86.32	72.63	72.63	70.47
FCGR3A	604590	100	100	100	100
FCGR3B	146740	100	100	100	100
FCHO1	610665	99.97	97.04	91.07	81.80
FCN3	613437	100	100	100	90.07
FERMT3	604973	100	98.55	95.07	89.17
FNIP1	607901	100	100	99.56	97.10
FOXN1	610594	100	100	99.90	96.22
FOXP3	600838	100	100	97.39	71.38
FPR1	300292	100	100	100	100
G6PC1	136537	No coverage data			
G6PC3	613742	100	100	100	100
G6PD	611045	100	99.87	96.31	89.68
GATA2	305900	100	100	100	88.90
GFI1	137295	100	100	99.17	96.10
GINS1	600871	100	100	100	100
GJC2	610608	99.36	94.35	86.24	62.68
GRHL2	608803	100	100	100	97.04
GTF2H5	608576	100	100	100	100
HAVCR2	608780	100	100	100	99.19
HAX1	606652	100	100	100	99.54
HELLS	605998	100	99.42	95.58	89.60
HMOX1	603946	100	100	99.69	92.06
HYOU1	141250	100	100	99.30	96.30
ICOS	601746	100	100	100	100
ICOSLG	604558	100	100	100	100
IFIH1	605717	100	100	100	99.12
IFNAR1	606951	98.12	98.12	97.10	90.15
IFNAR2	107450	100	100	100	99.30
IFNG	602376	100	99.83	99.83	98.10
IFNGR1	147570	100	100	97.92	90.79
IFNGR2	107470	92.43	92.43	92.43	90.15
IGLL1	147020	100	100	100	91.03
IKBKB	146770	100	100	99.63	95.13
IKBKG	603258	38.62	26.61	25.50	20.50
IKZF1	300248	100	100	100	96.22
IL10	603023	100	100	100	100
IL10RA	124092	100	100	98.69	95.09

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IL10RB	146933	100	93.99	93.81	90.67
IL12B	123889	100	100	100	93.39
IL12RB1	161561	100	100	97.63	83.62
IL17F	601604	100	100	100	100
IL17RA	606496	100	98.23	94.47	90.73
IL17RC	605461	100	98.95	95.88	83.73
IL18BP	610925	100	100	100	97.95
IL1RN	604113	100	100	100	99.77
IL2	147679	100	100	99.26	92.86
IL21	147680	100	100	95.90	85.41
IL21R	605384	100	100	100	97.47
IL2RA	605383	100	100	100	100
IL2RB	147730	100	100	99.45	94.65
IL2RG	146710	100	100	100	95.50
IL36RN	308380	100	100	99.82	89.72
IL6R	605507	93.09	91.96	87.00	81.63
IL6ST	147880	100	100	97.92	92.67
IL7R	600694	100	100	100	100
INO80	146661	100	99.48	99.00	96.68
INSR	610169	99.97	98.56	97.38	96.76
IRAK1	147670	99.21	94.99	83.20	52.94
IRAK4	300283	100	100	100	94.95
IRF2BP2	606883	100	99.89	88.12	64.77
IRF3	615332	100	100	100	94.47
IRF4	603734	100	99.67	93.23	92.56
IRF7	601900	100	98.78	96.99	90.74
IRF8	605047	100	99.17	93.46	74.66
IRF9	601565	100	100	100	96.79
ISG15	147574	100	100	100	95.89
ITCH	147571	98.68	96.77	94.95	91.42
ITGB2	606409	100	98.62	95.82	84.75
ITK	600065	100	100	100	99.41
IVNS1ABP	186973	100	100	100	100
JAGN1	609209	100	100	100	100
JAK1	616012	100	100	99.99	98.11
JAK2	147795	100	99.69	98.72	94.92
JAK3	147796	100	100	99.14	94.30
KDM6A	600173	100	99.78	97.98	86.21
KMT2A	300128	99.92	99.22	98.31	95.93
KMT2D	159555	100	100	99.94	98.49

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
KRAS	602113	100	100	100	100
LACC1	190070	100	100	100	100
LAMTOR2	613409	100	100	100	98.68
LAT	610389	100	100	99.29	82.40
LCK	602354	100	100	98.05	90.63
LCP2	153390	100	94.03	90.84	89.87
LIG1	601603	100	98.83	95.10	81.54
LIG4	126391	96.16	96.16	96.16	96.16
LPIN2	601837	100	100	100	98.86
LRBA	605519	99.46	99.46	98.88	95.72
LRRC8A	606453	100	100	100	100
LSM11	608360	100	89.61	73.79	59.66
LYST	617910	100	99.93	99.20	94.88
MAGT1	606897	100	97.92	91.47	86.70
MAL	300715	100	100	100	94.81
MALT1	188860	95.00	91.36	90.18	89.31
MAN2B1	604860	100	99.90	97.81	88.61
MANBA	609458	100	100	99.97	99.16
MAP1LC3B2	609489	100	100	100	100
MAP3K14	604655	100	98.81	94.70	92.62
MAPK8	601158	100	100	100	99.13
MASP2	605102	100	100	97.94	95.42
MBL2	154545	100	100	100	100
MC2R	607397	100	100	100	100
MCM10	609357	100	100	99.53	93.71
MCM4	602638	100	99.43	97.66	94.59
MEFV	608107	100	100	99.96	95.89
MOGS	601336	100	100	100	98.42
MRE11	600814	100	100	99.92	95.71
MRTFA	606078	92.22	90.96	88.80	79.21
MS4A1	112210	100	100	95.80	83.28
MSN	309845	100	100	100	96.33
MTHFD1	172460	100	100	100	98.82
MVK	251170	100	100	100	97.30
MYD88	602170	100	100	100	100
MYSM1	612176	99.83	96.98	93.20	90.22
NBAS	608025	100	100	99.93	97.07
NBN	602667	100	100	100	97.35
NCF1	608512	58.99	53.96	41.33	24.75
NCF2	608515	100	100	100	100

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NCF4	601488	100	100	99.63	87.67
NCKAP1L	141180	100	100	99.36	95.29
NCSTN	605254	100	100	100	99.23
NFAT5	604708	100	99.96	98.71	95.84
NFE2L2	600492	100	100	100	96.61
NFKB1	164011	100	100	99.97	96.97
NFKB2	164012	100	100	98.71	91.15
NFKBIA	164008	100	100	99.72	93.88
NHEJ1	611290	100	100	100	98.72
NHP2	606470	100	100	100	100
NLRC4	606831	100	100	100	100
NLRP1	606636	100	100	100	98.22
NLRP12	609648	100	100	99.44	95.88
NLRP3	606416	100	100	100	99.67
NOD2	605956	100	100	100	96.90
NOP10	606471	100	100	100	100
NOS2	163730	100	100	99.71	93.35
NRAS	164790	100	100	100	99.85
NSMCE3	608243	100	100	100	95.60
OAS1	164350	100	98.60	96.62	89.21
ORAI1	610277	99.37	95.52	92.83	88.94
OSTM1	607649	100	95.94	82.58	65.29
OTULIN	615712	90.38	85.62	85.62	83.36
PARN	604212	100	100	100	96.98
PAX1	167411	100	95.30	80.70	68.74
PAX5	167414	100	100	100	99.74
PBX1	176310	100	98.91	92.04	79.97
PCCA	232000	100	99.02	95.93	90.63
PCCB	232050	100	100	100	98.60
PEPD	613230	100	100	99.80	91.29
PGM3	172100	100	100	100	99.37
PIGA	311770	100	100	96.12	95.35
PIK3CD	602839	100	100	99.72	98.82
PIK3CG	601232	100	100	100	99.74
PIK3R1	171833	100	100	100	98.58
PLCG2	600220	100	100	100	97.94
PLEKHM1	611466	99.28	97.99	96.81	90.73
PLG	173350	100	100	98.57	85.86
PMM2	601785	100	100	100	98.16
PNP	164050	100	100	100	100

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POLA1	312040	100	99.72	96.89	86.82
POLE2	602670	100	97.93	91.56	84.19
POMP	613386	99.45	85.08	85.08	80.85
POT1	606478	100	100	99.77	93.78
POU2AF1	601206	100	99.37	94.64	80.36
PRF1	170280	100	100	100	100
PRKCD	176977	97.63	97.63	97.63	97.40
PRKDC	600899	99.87	99.09	98.87	96.61
PRPS1	311850	100	100	100	98.76
PSENN1	607632	100	100	100	91.46
PSMA3	176843	100	100	100	95.63
PSMB4	602177	100	100	100	94.69
PSMB8	177046	100	100	100	100
PSMB9	177045	100	100	100	89.70
PSMG2	609702	100	100	100	99.03
PSTPIP1	606347	100	100	100	93.25
PTPN22	600716	98.20	98.20	98.20	97.80
PTPRC	151460	100	98.51	94.99	89.36
RAB27A	603868	100	100	100	100
RAC2	602049	100	100	100	98.71
RAG1	179615	100	100	100	100
RAG2	179616	100	100	100	100
RANBP2	601181	99.55	98.83	97.67	94.69
RASGRP1	603962	100	100	100	99.60
RASGRP2	605577	100	100	99.11	91.54
RBCK1	610924	100	100	100	89.60
RC3H1	609424	100	99.72	98.72	94.10
RECQL4	603780	100	99.95	99.36	94.91
RELB	604758	100	98.79	93.37	73.27
RFX5	601863	100	100	100	99.56
RFXANK	603200	100	100	100	89.84
RFXAP	601861	100	100	97.26	93.49
RHOG	179505	100	100	100	100
RHOH	602037	100	100	100	100
RIPK1	603453	100	100	99.59	95.26
RMRP	157660	No coverage data			
RNASEH2A	606034	100	100	100	95.93
RNASEH2B	610326	100	91.35	91.35	91.35
RNASEH2C	610330	100	100	100	100
RNF168	612688	100	100	100	99.45

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RNF31	612487	100	100	99.16	96.40
RNU4ATAC	601428	No coverage data			
RORC	602943	100	99.94	95.80	80.86
RPSA	150370	100	100	100	100
RSPH9	612648	100	100	100	99.52
RTEL1	608833	100	100	99.83	95.32
SAMD9	610456	100	100	100	99.84
SAMD9L	611170	100	100	100	100
SAMHD1	606754	100	100	100	100
SASH3	300441	100	100	98.54	79.73
SBDS	607444	100	100	100	100
SEC61A1	609213	100	100	100	99.16
SEMA3E	608166	100	98.59	95.80	92.61
SERAC1	614725	100	100	100	98.51
SERPING1	606860	100	98.02	95.67	92.10
SH2B3	605093	100	100	96.89	77.27
SH2D1A	300490	100	100	100	100
SH3BP2	602104	91.15	91.15	91.15	89.14
SH3KBP1	300374	100	99.02	97.43	90.71
SKIV2L	600478	100	100	99.84	96.66
SLC29A3	612373	100	97.60	97.60	97.60
SLC35A1	605634	100	98.77	96.30	95.28
SLC35C1	605881	100	100	100	100
SLC37A4	602671	99.87	99.87	99.87	97.46
SLC39A4	607059	100	100	100	100
SLC39A7	601416	100	100	100	100
SLC46A1	611672	99.87	99.87	99.87	99.77
SLC7A7	603593	100	100	100	99.30
SMARCAL1	606622	100	100	100	97.91
SMARCD2	601736	87.26	87.26	87.26	86.40
SNORA31	619378	No coverage data			
SNX10	614780	100	100	100	100
SOCS1	603597	100	100	90.74	62.02
SOCS4	616337	100	100	100	97.24
SP110	604457	100	100	98.59	92.37
SPINK5	605010	100	99.14	96.90	84.44
SPPL2A	608238	99.06	95.32	89.46	84.35
SRP54	604857	99.67	98.23	96.66	90.12
SRP72	602122	100	100	100	99.81
STAT1	600555	97.25	97.09	97.09	96.12

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
STAT2	600556	100	100	100	99.34
STAT3	102582	100	100	100	96.39
STAT4	600558	100	97.04	96.78	95.38
STAT5B	604260	100	99.49	97.08	89.89
STAT6	601512	100	100	98.13	85.39
STIM1	605921	96.76	96.76	96.76	96.76
STING1	612374	100	100	100	89.67
STK4	604965	100	100	98.51	95.00
STN1	613128	100	100	100	100
STX11	605014	100	100	100	100
STXBP2	601717	100	96.88	91.83	79.08
TAP1	170260	100	100	98.60	92.45
TAP2	170261	100	98.84	95.83	89.77
TAPBP	601962	100	99.18	95.91	85.38
TFAZZIN	300394	No coverage data			
TBX1	602054	89.95	75.58	63.04	42.43
TBX21	604895	100	99.97	94.78	79.22
TCF3	147141	100	100	96.06	76.03
TCIRG1	604592	100	99.23	95.33	87.67
TCN2	613441	100	100	100	96.44
TERC	602322	No coverage data			
TERT	187270	100	98.76	95.16	90.50
TET2	612839	100	100	99.89	98.38
TFRC	190010	100	100	99.60	97.54
TGFB1	190180	100	100	100	99.39
THBD	188040	100	100	100	98.68
TICAM1	607601	100	100	100	92.58
TINF2	604319	100	100	100	100
TIRAP	606252	100	100	100	100
TLR3	603029	100	100	100	99.55
TLR4	603030	100	100	100	100
TLR7	300365	100	100	100	100
TLR8	300366	100	100	100	100
TMC6	605828	100	100	99.73	90.55
TMC8	605829	100	99.80	94.47	66.63
TNFAIP3	191163	100	100	100	98.26
TNFRSF11A	603499	90.20	90.20	88.03	82.66
TNFRSF13B	604907	100	100	99.54	91.32
TNFRSF13C	606269	91.01	65.44	58.50	47.14
TNFRSF1A	191190	100	100	100	97.25

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
TNFRSF4	600315	100	100	96.96	59.94
TNFRSF9	602250	100	100	99.28	86.96
TNFSF11	602642	100	100	100	100
TNFSF12	602695	94.64	86.13	73.45	58.51
TNFSF13	604472	100	100	100	99.60
TOM1	604700	100	100	100	99.07
TOP2B	126431	99.84	99.20	96.28	86.19
TPP2	190470	96.74	96.12	92.96	86.68
TRAF3	601896	100	100	99.84	91.96
TRAF3IP2	607043	100	100	100	100
TREX1	606609	100	100	100	100
TRIM22	606559	100	100	100	94.86
TRNT1	612907	100	95.77	89.81	87.03
TTC37	614589	100	100	100	98.33
TTC7A	609332	100	100	99.97	92.78
TYK2	176941	100	100	100	98.39
UBA1	314370	100	100	97.89	87.89
UNC13D	608897	100	100	99.27	92.38
UNC93B1	608204	75.39	68.32	63.56	59.49
UNG	191525	100	100	100	97.08
USB1	613276	100	100	90.43	75.35
USP18	607057	95.21	95.21	95.21	93.58
VAV1	164875	100	100	99.90	95.66
VPS13B	607817	99.37	98.22	97.00	90.85
VPS45	610035	100	100	100	98.46
WAS	300392	100	99.26	90.35	70.02
WDR1	604734	100	98.70	98.30	97.80
WIPF1	602357	100	99.11	97.46	89.23
WRAP53	612661	100	100	100	98.51
XIAP	300079	100	100	99.35	89.29
ZAP70	176947	100	100	98.88	94.95
ZBTB24	614064	100	100	100	100
ZNF341	618269	98.22	98.22	96.28	84.94

- OMIM release used: 23-9-2021

- The statistics above are based on a set of 150 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

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
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