Whole Exome Sequencing
Gene package Primary Immunodeficiency Disorders, version 11, 25-2-2022

Technical information
DNA was enriched using the Agilent SureSelectXT Human All Exon V7 capture kit and paired-end sequenced on the Illumina platform (outsourced). Sequencing data are demultiplexed with bcl2fastq2 Conversion Software from Illumina. Illumina DRAGEN Bio-IT Platform is used for read mapping to the hg19 genome and sequence variant detection. The detected sequence variants are annotated and filtered with Alissa Interpret software and classified with Alamut Visual. Copy number 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 annotated and filtered with the NexusClinical software and classified using UCSC Genome Browser (NCBI37/hg19). The sensitivity to detect variants using this technology is not 100%; pathogenic variants could be missed. 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)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
ACD	609377	100	100	100	97.43
ACP5	171640	100	100	100	100
ACTB	102630	100	100	100	99.18
ADA	608958	100	100	99.94	92.54
ADA2	607575	100	100	100	100
ADAM17	603639	100	99.70	98.07	91.74
ADAMTS3	605011	100	100	100	99.63
ADAR	146920	98.45	98.45	98.45	98.04
ADGRE2	606100	100	100	98.68	94.77
AGA	613228	100	100	100	97.41
AICDA	605257	100	100	100	88.83
AIRE	607358	100	99.53	96.71	88.36
AK2	103020	100	98.15	95.34	89.19
ALG13	300776	100	100	100	97.07
ALPI	171740	100	100	100	97.87
ANGPT1	601667	100	100	100	100
AP1S3	615781	100	100	97.78	95.91
AP3B1	603401	99.05	96.39	94.98	89.23
AP3D1	607246	97.71	97.64	95.96	89.40
APOL1	603743	100	100	100	99.34
ARHGEF1	601855	100	99.27	96.27	85.15
ARPC1B	604223	100	100	100	99.04
ATG4A	300663	100	100	100	99.69
ATM	607585	100	100	93.43	54.67

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ATP6AP1	300197	100	100	93.66	83.51
B2M	109700	100	100	100	100
BACH2	605394	100	100	100	99.02
BCL10	603517	100	100	97.43	88.34
BCL11B	606558	100	99.02	96.99	90.41
BLK	191305	100	99.12	96.35	87.07
BLM	604610	100	99.87	99.08	96.13
BLNK	604515	100	98.71	94.44	88.82
BLOC1S6	604310	100	100	96.91	78.29
BTK	300300	100	100	100	99.57
C1QA	120550	100	100	100	100
C1QB	120570	100	100	100	97.00
C1QC	120575	100	100	100	94.13
C1R	613785	100	100	99.33	95.05
C1S	120580	100	100	100	96.43
C2	613927	100	100	100	95.70
C3	120700	100	100	99.30	96.05
C5	120900	100	99.80	99.05	93.70
C6	217050	100	100	100	100
C7	217070	100	100	100	100
C8A	120950	100	100	100	98.66
C8B	120960	100	100	100	99.80
C8G	120930	100	100	94.84	74.66
C9	120940	100	99.84	99.53	98.79
CA2	611492	100	100	100	100
CARD11	607210	100	99.32	97.03	94.15
CARD14	607211	100	99.21	97.60	88.97
CARD9	607212	100	99.08	97.24	89.91
CARMIL2	610859	97.18	94.69	93.13	86.05
CASP10	601762	100	100	100	100
CASP8	601763	100	100	98.94	94.10
CAVIN1	603198	100	100	100	96.78
CCBE1	612753	100	100	100	98.23
CD19	107265	100	99.95	98.05	92.06
CD247	186780	85.23	85.23	85.23	83.73
CD27	186711	100	100	100	98.11
CD3D	186790	100	100	100	100
CD3E	186830	100	100	100	98.59
CD3G	186740	100	100	100	100
CD4	186940	100	100	100	100
CD40	109535	100	100	100	100
CD40LG	300386	100	100	100	100
CD46	120920	100	100	100	98.66
CD55	125240	100	97.01	94.12	84.45

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CD59	107271	100	100	100	100
CD70	602840	100	100	100	94.96
CD79A	112205	100	93.89	85.22	68.06
CD79B	147245	100	100	100	100
CD81	186845	100	100	98.16	85.89
CD8A	186910	100	100	100	91.64
CDC42	116952	100	100	100	100
CDCA7	609937	100	99.68	95.87	89.19
CDKN2B	600431	100	100	98.68	88.99
CEBPE	600749	100	100	100	99.89
CFB	138470	100	100	100	99.47
CFD	134350	100	100	94.72	68.18
CFH	134370	100	99.10	96.82	92.91
CFI	217030	86.45	84.36	82.41	78.74
CFP	300383	100	99.68	97.54	88.56
CFTR	602421	100	100	100	98.09
CHD7	608892	100	100	99.89	98.69
CIB1	602293	100	100	97.90	90.88
CIITA	600005	100	99.34	96.74	87.09
CLCN7	602727	100	100	99.97	94.84
CLEC4D	609964	100	100	100	97.39
CLEC7A	606264	100	100	100	100
CLPB	616254	100	100	100	98.48
COG6	606977	100	100	100	97.68
COPA	601924	100	100	100	98.67
CORO1A	605000	92.29	91.79	90.59	86.61
CR2	120650	100	100	100	98.43
CREBBP	600140	100	99.21	97.19	91.73
CSF2RA	306250	100	100	100	100
CSF2RB	138981	100	98.32	94.93	88.92
CSF3R	138971	100	98.13	95.07	88.84
CTC1	613129	100	100	99.71	94.64
CTLA4	123890	100	100	100	100
CTNNBL1	611537	100	100	98.95	92.86
CTPS1	123860	100	99.91	98.44	94.08
CTSC	602365	100	100	100	100
CXCR4	162643	100	100	99.55	95.37
CYBA	608508	95.74	84.68	71.77	50.92
CYBB	300481	100	100	99.95	95.66
CYBC1	618334	100	99.71	93.61	84.29
DBR1	607024	100	100	100	98.30
DCLRE1B	609683	100	100	100	95.69
DCLRE1C	605988	100	100	100	95.14
DDX58	609631	100	100	100	98.36

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DEF6	610094	100	98.20	94.82	89.23
DGAT1	604900	95.12	88.94	87.80	87.80
DHFR	126060	100	100	99.49	90.23
DKC1	300126	100	100	98.05	93.43
DNAJC21	617048	100	100	98.94	92.66
DNASE1	125505	100	100	100	97.31
DNASE1L3	602244	100	100	100	98.13
DNASE2	126350	100	100	97.25	88.21
DNMT3B	602900	100	100	99.17	93.33
DOCK2	603122	100	99.82	99.04	97.48
DOCK8	611432	100	99.94	99.37	96.42
EFL1	617538	100	100	100	99.69
ELANE	130130	100	100	100	95.95
ELF4	300775	100	99.46	96.84	87.62
EPG5	615068	100	99.90	99.27	95.85
ERCC2	126340	100	100	100	94.45
ERCC3	133510	100	100	100	97.60
ERCC6L2	615667	100	100	99.96	97.25
EXTL3	605744	100	100	100	100
F12	610619	100	100	100	98.02
FAAP24	610884	100	100	100	97.52
FADD	602457	100	100	100	100
FAS	134637	100	100	100	92.44
FASLG	134638	100	100	96.75	90.79
FAT4	612411	100	100	99.97	99.63
FCGR3A	146740	100	100	100	99.22
FCHO1	613437	100	97.57	91.61	80.80
FCN3	604973	100	100	100	88.41
FERMT1	607900	100	99.91	99.09	95.35
FERMT3	607901	100	99.08	94.02	88.34
FNIP1	610594	100	100	99.84	98.25
FOXN1	600838	100	100	99.29	94.96
FOXP3	300292	100	100	98.15	78.49
FPR1	136537	100	100	100	100
G6PC1	613742	No coverage data			
G6PC3	611045	100	100	100	100
G6PD	305900	100	100	98.26	92.43
GATA2	137295	100	100	100	85.78
GFI1	600871	100	99.78	98.70	96.32
GINS1	610608	100	100	100	100
GJC2	608803	98.99	92.11	85.49	65.22
GRHL2	608576	100	100	99.84	98.09
GTF2H5	608780	100	100	100	100
HAVCR2	606652	100	100	100	98.95

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HAX1	605998	100	100	100	98.77
HELLS	603946	100	100	97.20	91.60
HMOX1	141250	100	100	98.96	90.72
HS3ST6	619210	85.83	81.33	79.03	72.42
HYOU1	601746	100	100	98.74	94.57
ICOS	604558	100	100	100	100
ICOSLG	605717	100	100	100	100
IFIH1	606951	100	100	100	99.35
IFNAR1	107450	98.12	98.12	98.12	93.29
IFNAR2	602376	100	100	100	99.65
IFNG	147570	99.91	99.83	99.83	99.31
IFNGR1	107470	100	100	98.97	90.79
IFNGR2	147569	93.16	92.43	92.43	89.25
IGHM	147020	94.56	94.56	94.56	94.56
IGLL1	146770	100	100	100	92.31
IKBKB	603258	100	100	98.85	93.75
IKBKG	300248	36.54	25.95	25.35	20.68
IKZF1	603023	100	100	100	96.22
IKZF3	606221	100	100	100	97.14
IL10	124092	100	100	100	100
IL10RA	146933	100	100	98.77	94.77
IL10RB	123889	100	95.38	93.54	90.04
IL12B	161561	100	100	100	95.29
IL12RB1	601604	100	99.84	97.63	83.68
IL17F	606496	100	100	100	99.18
IL17RA	605461	100	99.91	95.36	89.59
IL17RC	610925	100	98.51	95.48	84.04
IL18BP	604113	100	100	100	97.36
IL1RN	147679	100	100	100	99.70
IL2	147680	100	100	100	95.73
IL21	605384	100	100	98.98	88.82
IL21R	605383	100	100	100	97.58
IL2RA	147730	100	100	100	100
IL2RB	146710	100	100	98.85	93.62
IL2RG	308380	100	100	100	97.91
IL36RN	605507	100	100	97.80	86.97
IL6R	147880	93.09	92.54	87.00	79.77
IL6ST	600694	100	100	98.87	93.58
IL7R	146661	100	100	100	100
INO80	610169	100	99.48	98.98	96.69
INSR	147670	100	99.08	97.94	96.53
IRAK1	300283	99.63	96.34	89.18	64.13
IRAK4	606883	100	100	100	96.94
IRF2BP2	615332	100	99.64	89.81	66.49

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IRF3	603734	100	100	99.35	91.93
IRF4	601900	100	99.04	92.80	92.60
IRF7	605047	100	98.67	96.82	89.87
IRF8	601565	100	97.73	91.80	71.35
IRF9	147574	100	100	99.55	95.39
ISG15	147571	100	100	100	93.46
ITCH	606409	98.24	96.53	94.90	91.64
ITGB2	600065	100	98.31	95.32	83.70
ITK	186973	100	100	100	98.77
IVNS1ABP	609209	100	100	100	100
JAGN1	616012	100	100	100	100
JAK1	147795	100	100	99.87	98.29
JAK2	147796	100	99.84	99.11	96.12
JAK3	600173	100	99.87	98.64	94.01
KDM6A	300128	100	100	99.30	92.95
KMT2A	159555	99.87	99.34	98.67	96.60
KMT2D	602113	100	100	99.75	97.30
KNG1	612358	100	100	100	98.36
KRAS	190070	100	100	100	100
LACC1	613409	100	100	100	100
LAMTOR2	610389	100	100	100	94.29
LAT	602354	100	100	96.91	76.37
LCK	153390	100	99.43	96.43	86.11
LCP2	601603	100	95.20	90.96	89.33
LIG1	126391	100	98.83	94.43	81.21
LIG4	601837	96.16	96.16	96.16	96.16
LPIN2	605519	100	100	100	98.57
LRBA	606453	100	99.46	99.40	97.12
LRRC8A	608360	100	100	100	99.47
LSM11	617910	100	93.06	77.89	64.05
LYST	606897	100	99.99	99.68	96.53
MAGT1	300715	100	98.65	94.16	90.58
MAL	188860	100	100	100	95.45
MALT1	604860	95.96	92.09	90.24	89.69
MAN2B1	609458	100	99.96	97.78	88.49
MANBA	609489	100	100	99.97	99.23
MAP1LC3B2	No OMIM id	100	100	100	100
MAP3K14	604655	100	98.84	95.06	91.78
MAPK8	601158	100	100	100	99.27
MASP2	605102	100	100	97.40	94.27
MBL2	154545	100	100	100	100
MC2R	607397	100	100	100	100
MCM10	609357	100	100	99.62	94.88
MCM4	602638	100	99.66	98.11	95.12

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MEFV	608107	100	100	99.57	94.81
MOGS	601336	100	100	100	98.42
MRTFA	606078	92.22	90.89	88.44	76.47
MS4A1	112210	100	100	97.33	86.99
MSN	309845	100	100	100	97.46
MTHFD1	172460	100	100	100	98.69
MVK	251170	100	100	100	96.15
MYD88	602170	100	100	100	99.65
MYSM1	612176	100	99.20	95.79	91.52
NBAS	608025	100	100	100	98.26
NBN	602667	100	100	100	99.11
NCF1	608512	58.99	50.72	35.79	21.19
NCF2	608515	100	100	100	98.03
NCF4	601488	100	100	99.22	84.51
NCKAP1L	141180	100	100	99.60	94.99
NCSTN	605254	100	100	100	98.42
NFAT5	604708	100	100	99.63	96.56
NFE2L2	600492	100	100	100	97.83
NFKB1	164011	100	100	100	97.45
NFKB2	164012	100	99.94	98.84	91.42
NFKBIA	164008	100	100	100	95.52
NHEJ1	611290	100	100	100	98.53
NHP2	606470	100	100	100	100
NLRC4	606831	100	100	100	100
NLRP1	606636	100	100	99.99	98.02
NLRP12	609648	100	100	99.26	94.99
NLRP3	606416	100	100	100	99.39
NOD2	605956	100	100	99.81	96.90
NOP10	606471	100	100	100	100
NOS2	163730	100	100	99.43	91.72
NRAS	164790	100	100	100	99.54
NSMCE3	608243	100	100	100	95.60
OAS1	164350	100	97.91	95.56	87.22
ORAI1	610277	99.37	95.26	92.94	89.73
OSTM1	607649	100	97.73	86.41	69.34
OTULIN	615712	91.60	85.62	85.62	82.65
PARN	604212	100	100	100	98.33
PAX1	167411	100	98.03	83.02	72.50
PAX5	167414	100	100	100	99.62
PBX1	176310	100	99.08	92.76	80.44
PCCA	232000	100	99.47	96.88	92.92
PCCB	232050	100	100	100	97.12
PEPD	613230	100	100	98.93	91.34
PGM3	172100	100	100	100	100

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PIGA	311770	100	100	99.88	96.12
PIK3CD	602839	100	100	99.61	98.52
PIK3CG	601232	100	100	100	99.59
PIK3R1	171833	100	100	100	98.75
PLCG2	600220	100	100	100	96.79
PLEKHM1	611466	98.79	97.89	96.21	85.01
PLG	173350	100	100	99.69	85.86
PMM2	601785	100	100	100	98.16
PNP	164050	100	100	100	100
POLA1	312040	100	100	99.02	92.76
POLE2	602670	100	98.55	94.19	86.77
POMP	613386	100	93.37	85.08	80.85
POT1	606478	100	100	100	95.16
POU2AF1	601206	100	97.58	93.15	76.15
PRF1	170280	100	100	100	100
PRKCD	176977	97.63	97.63	97.63	97.32
PRKDC	600899	100	99.09	98.86	97.00
PRPS1	311850	100	100	100	99.75
PSENEN	607632	100	100	98.35	89.94
PSMA3	176843	100	100	100	94.92
PSMB4	602177	100	100	99.57	92.86
PSMB8	177046	100	100	100	100
PSMB9	177045	100	100	99.94	89.70
PSMG2	609702	100	100	100	99.14
PSTPIP1	606347	100	100	100	92.34
PTPN22	600716	98.20	98.20	98.20	97.82
PTPRC	151460	100	99.32	96.86	90.29
RAB27A	603868	100	100	100	100
RAC2	602049	100	100	100	98.85
RAG1	179615	100	100	100	100
RAG2	179616	100	100	100	100
RANBP2	601181	99.96	98.98	97.95	95.83
RASGRP1	603962	100	100	100	99.95
RASGRP2	605577	100	99.98	98.54	89.89
RBCK1	610924	100	100	100	91.33
RC3H1	609424	100	99.46	98.51	93.85
RECQL4	603780	100	99.95	99.35	94.89
REL	164910	100	98.75	98.56	94.90
RELA	164014	100	97.73	96.10	92.45
RELB	604758	100	98.99	96.13	74.81
RFX5	601863	100	100	100	99.01
RFXANK	603200	100	100	98.72	86.17
RFXAP	601861	100	100	98.29	95.32
RHOG	179505	100	100	100	100

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RHOH	602037	100	100	100	100
RIPK1	603453	100	100	100	95.68
RNASEH2A	606034	100	100	99.81	92.38
RNASEH2B	610326	100	91.35	91.35	91.35
RNASEH2C	610330	100	100	100	100
RNF168	612688	100	100	100	99.65
RNF31	612487	100	100	99.20	95.41
RORC	602943	100	99.41	94.84	79.06
RPSA	150370	100	100	100	99.12
RSPH9	612648	100	100	100	99.46
RTEL1	608833	100	100	99.77	94.04
SAMD9	610456	100	100	100	99.87
SAMD9L	611170	100	100	100	100
SAMHD1	606754	100	100	100	100
SASH3	300441	100	100	98.92	87.50
SBDS	607444	100	100	100	100
SEC61A1	609213	100	100	100	99.46
SEMA3E	608166	100	100	97.39	92.50
SERAC1	614725	100	100	100	100
SERPING1	606860	100	98.38	95.43	91.46
SH2B3	605093	100	99.84	98.07	77.21
SH2D1A	300490	100	100	100	100
SH3BP2	602104	91.15	91.15	91.15	88.74
SH3KBP1	300374	100	99.02	97.43	93.18
SKIV2L	600478	100	100	99.13	93.93
SLC29A3	612373	100	97.60	97.60	97.48
SLC35A1	605634	100	100	96.30	95.28
SLC35C1	605881	100	100	100	99.65
SLC37A4	602671	99.87	99.87	99.42	96.62
SLC39A4	607059	100	100	100	100
SLC39A7	601416	100	100	100	99.35
SLC46A1	611672	99.87	99.87	99.87	99.77
SLC7A7	603593	100	100	100	99.01
SMARCAL1	606622	100	100	100	97.64
SMARCD2	601736	87.26	87.26	87.26	85.64
SNORA31	619378	No coverage data			
SNX10	614780	100	100	100	100
SOCS1	603597	100	100	92.88	65.39
SOCS4	616337	100	100	100	96.64
SP110	604457	100	99.62	98.53	93.59
SPINK5	605010	100	99.84	97.49	87.43
SPPL2A	608238	99.81	95.38	93.49	86.32
SRP54	604857	100	98.68	97.49	92.69
SRP72	602122	100	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
STAT1	600555	99.29	97.09	97.09	96.54
STAT2	600556	100	100	100	99.14
STAT3	102582	100	100	100	95.99
STAT4	600558	100	99.45	97.04	95.64
STAT5B	604260	100	99.03	96.66	89.84
STAT6	601512	100	99.85	97.01	83.08
STIM1	605921	96.76	96.76	96.76	95.20
STING1	612374	100	100	100	88.83
STK4	604965	100	100	98.04	95.21
STN1	613128	100	100	100	100
STX11	605014	100	100	100	100
STXBP2	601717	100	97.40	91.65	77.53
SYK	600085	100	100	98.82	95.57
TAP1	170260	100	100	98.17	90.89
TAP2	170261	100	98.11	94.82	88.28
TAPBP	601962	100	98.82	95.34	84.72
TAFAZZIN	300394	No coverage data			
TBX1	602054	91.65	78.18	67.92	47.40
TBX21	604895	100	100	98.38	80.49
TCF3	147141	100	100	96.64	76.34
TCIRG1	604592	100	98.97	94.86	86.76
TCN2	613441	100	100	100	95.04
TERT	187270	100	99.15	96.14	90.14
TET2	612839	100	100	99.84	98.41
TFRC	190010	100	99.96	99.36	97.54
TGFB1	190180	100	100	100	99.69
THBD	188040	100	100	100	99.00
TICAM1	607601	100	100	99.88	89.01
TINF2	604319	100	100	100	100
TIRAP	606252	100	100	100	100
TLR3	603029	100	100	100	99.70
TLR4	603030	100	100	100	100
TLR7	300365	100	100	100	100
TLR8	300366	100	100	100	100
TMC6	605828	100	100	99.75	89.52
TMC8	605829	100	99.62	94.45	66.99
TNFAIP3	191163	100	100	99.80	98.30
TNFRSF11A	603499	90.20	90.20	87.10	81.42
TNFRSF13B	604907	100	100	98.88	90.14
TNFRSF13C	606269	91.42	67.57	59.97	51.47
TNFRSF1A	191190	100	100	100	97.54
TNFRSF4	600315	100	100	95.62	59.58
TNFRSF9	602250	100	100	98.78	89.67
TNFSF11	602642	100	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
TNFSF12	602695	96.34	87.15	74.01	56.71
TNFSF13	604472	100	100	100	97.11
TOM1	604700	100	100	100	98.26
TOP2B	126431	99.95	99.59	97.93	90.42
TPP2	190470	96.74	96.49	94.70	88.11
TRAC	186880	100	100	100	100
TRAF3	601896	100	100	100	95.75
TRAF3IP2	607043	100	100	100	100
TREX1	606609	100	100	100	100
TRIM22	606559	100	100	99.60	93.15
TRNT1	612907	100	97.75	89.81	86.79
TTC37	614589	100	100	100	98.95
TTC7A	609332	100	100	99.67	92.91
TYK2	176941	100	100	99.81	96.75
UBA1	314370	100	99.93	97.88	90.57
UNC13D	608897	100	100	99.12	91.74
UNC93B1	608204	75.15	69.23	66.86	58.28
UNG	191525	100	100	100	97.68
USB1	613276	100	99.20	88.66	75.29
USP18	607057	95.21	95.21	95.21	92.59
VAV1	164875	100	100	99.28	94.88
VPS13B	607817	99.28	98.29	97.00	91.79
VPS45	610035	100	100	100	98.76
WAS	300392	100	99.86	94.70	77.23
WDR1	604734	100	99.91	98.30	97.36
WIPF1	602357	100	98.61	97.34	83.73
WRAP53	612661	100	100	100	97.04
XIAP	300079	100	100	100	93.11
ZAP70	176947	100	100	98.52	93.32
ZBTB24	614064	100	100	100	99.64
ZNF341	618269	98.67	98.22	95.28	84.49
ZNFX1	618931	100	100	99.51	97.82
0	0	0	0	0	0
0	0	0	0	0	0
0	0	0	0	0	0
0	0	0	0	0	0
0	0	0	0	0	0
0	0	0	0	0	0
0	0	0	0	0	0

- OMIM release used: 23-9-2021
- The statistics above are based on a set of 104 samples
- % Covered 10x , 20x, 30x and 50x describes the percentage of a gene's coding sequence (±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 ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x