

# WES PRIMARY IMMUNODEFICIENCIES DG 3.1

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt;10x</i>	<i>% covered &gt;20x</i>	<i>OMIM disease ID</i>
ACD	148.6	100.0	99.9	616553
ACP5	186.4	99.8	98.3	607944
ACTB	74.3	99.7	96.1	243310
ADA	105.4	100.0	99.7	102700
ADA2	90.5	100.0	99.0	615688
ADAM17	151.6	99.9	99.0	614328
ADAR	122.4	100.0	99.8	615010
AGA	191.3	100.0	100.0	208400
AICDA	148.6	100.0	100.0	605258
AIRE	91.0	100.0	99.8	240300
AK2	115.9	98.9	94.9	267500
ALG13	92.1	98.4	92.6	300884
ALPI	138.6	100.0	99.5	-
ANGPT1	180.1	99.8	98.7	No OMIM phenotype
AP1S3	146.7	90.4	90.1	616106
AP3B1	140.4	99.2	95.8	608233
AP3D1	125.3	99.8	98.6	617050
APOL1	171.5	100.0	100.0	-
ARHGEF1	101.7	99.9	98.4	618459
ARPC1B	131.7	100.0	100.0	617718
ATG4A	80.1	99.5	96.7	No OMIM phenotype
ATM	133.4	99.8	98.1	208900
ATP6AP1	104.8	98.2	92.1	300972
B2M	237.4	100.0	100.0	241600
BACH2	179.8	100.0	100.0	618394

BCL10	149.9	100.0	100.0	616098
BCL11B	97.4	99.1	95.6	617237
BLK	136.1	100.0	100.0	613375
BLM	133.4	99.8	98.3	210900
BLNK	105.4	97.1	95.5	613502
BLOC1S6	122.0	99.9	97.8	614171
BTK	126.7	100.0	99.9	307200
C1QA	194.6	100.0	100.0	613652
C1QB	193.3	100.0	100.0	613652
C1QC	200.1	100.0	99.2	613652
C1R	207.1	100.0	100.0	216950
C1S	113.0	99.9	99.0	613783
C2	140.1	100.0	100.0	217000
C3	153.5	99.9	99.2	613779
C5	151.8	99.9	98.5	609536
C6	174.7	100.0	99.7	612446
C7	138.5	100.0	98.9	610102
C8A	124.3	100.0	99.6	613790
C8B	136.9	100.0	99.2	613789
C8G	144.9	100.0	100.0	-
C9	145.6	99.9	99.5	613825
CA2	172.2	100.0	100.0	259730
CARD11	150.3	100.0	99.9	616452;615206;617638
CARD14	125.1	100.0	98.9	173200;602723
CARD9	112.5	99.9	98.4	212050
CARMIL2	144.2	96.3	94.5	618131
CASP10	129.9	99.5	97.3	603909
CASP8	154.8	95.6	95.4	607271
CAVIN1	158.7	100.0	100.0	613327
CCBE1	81.2	99.8	98.8	235510

CD19	127.3	100.0	100.0	613493
CD247	119.5	100.0	100.0	610163
CD27	103.3	99.9	96.9	615122
CD3D	176.6	100.0	100.0	608971
CD3E	151.7	100.0	99.5	608971
CD3G	169.4	100.0	100.0	615607
CD40	181.5	100.0	100.0	606843
CD40LG	132.0	97.3	88.1	308230
CD46	151.5	99.9	99.4	612922
CD55	152.1	92.9	85.4	613793
CD59	175.5	80.0	71.6	612300
CD70	126.0	99.8	97.7	618261
CD79A	158.5	100.0	100.0	613501
CD79B	202.1	100.0	100.0	612692
CD81	145.2	100.0	99.9	613496
CD8A	146.6	100.0	99.8	608957
CDC42	121.8	97.9	90.9	616737
CDCA7	138.0	100.0	99.6	616910
CDKN2B	88.3	100.0	99.9	-
CEBPE	100.3	100.0	100.0	245480
CFB	139.7	100.0	100.0	612924
CFD	87.7	89.3	83.7	613912
CFH	194.1	99.9	99.0	609814
CFHR1	201.6	96.4	94.9	235400
CFHR2	116.7	76.0	74.3	-
CFHR3	111.8	94.0	92.2	235400
CFHR4	152.1	100.0	99.9	-
CFHR5	115.2	99.6	98.4	614809
CFI	169.4	99.2	96.8	610984
CFP	108.8	100.0	99.0	312060

CFTR	140.6	99.6	97.9	211400;167800;219700
CHD7	158.7	100.0	99.5	214800
CIB1	131.3	97.3	93.6	618267
CIITA	160.2	100.0	99.5	209920
CLCN7	142.0	99.7	98.4	166600;611490
CLEC4D	144.6	100.0	99.8	-
CLEC7A	170.5	100.0	100.0	613108
CLPB	135.7	94.9	94.9	616271
COLEC11	178.1	100.0	100.0	265050
COPA	133.6	100.0	99.2	616414
CORO1A	144.9	100.0	98.6	615401
CR2	165.8	100.0	99.8	614699
CREBBP	124.1	99.7	98.5	180849
CSF2RA	60.8	89.9	87.5	300770
CSF2RB	139.8	100.0	99.0	614370
CSF3R	108.5	99.6	98.2	617014;162830
CTC1	128.0	100.0	99.6	612199
CTLA4	162.1	100.0	100.0	616100
CTNBL1	116.2	99.8	99.5	-
CTPS1	121.8	93.0	93.0	615897
CTSC	137.1	100.0	100.0	170650
CXCR4	151.1	100.0	100.0	193670
CYBA	115.6	95.0	82.4	233690
CYBB	121.9	99.9	99.3	300645
CYBC1	142.3	99.6	97.0	618935
DBF4	99.1	96.6	89.6	No OMIM phenotype
DBR1	142.0	100.0	99.3	-
DCLRE1C	163.4	100.0	99.4	602450
DDX58	149.1	99.9	99.0	616298
DEF6	113.0	96.7	93.8	-

DHFR	51.8	92.1	78.9	613839
DIAPH1	119.5	99.8	99.0	616632;124900
DKC1	108.6	99.8	98.7	305000
DNASE1	167.2	100.0	99.9	152700
DNASE1L3	140.2	100.0	100.0	614420
DNASE2	96.7	99.7	97.1	-
DNMT3B	133.6	100.0	100.0	242860
DOCK2	140.8	100.0	99.6	616433
DOCK8	135.8	100.0	99.6	243700
ELANE	143.2	99.7	97.4	202700;162800
ELF4	96.0	100.0	99.7	307200
EPG5	137.8	99.5	98.5	242840
ERCC2	135.5	100.0	99.7	601675
ERCC3	107.8	96.9	96.3	601675
EXTL3	199.3	100.0	100.0	617425
F12	127.6	99.9	98.8	610618
FAAP24	128.5	99.3	96.7	-
FADD	147.2	100.0	100.0	613759
FAS	262.8	100.0	99.6	601859
FASLG	98.0	100.0	99.6	601859
FAT4	227.0	100.0	100.0	616006
FCGR1A	54.9	46.8	44.1	-
FCGR2A	195.0	100.0	100.0	152700;219700
FCGR2B	135.8	99.5	95.4	152700
FCGR3A	196.4	99.0	97.1	152700
FCGR3B	149.2	99.3	97.9	-
FCHO1	111.1	98.9	97.7	619164
FCN3	143.9	100.0	100.0	613860
FERMT3	142.1	100.0	100.0	612840
FNIP1	186.0	100.0	99.8	No OMIM phenotype

FOXN1	149.9	100.0	99.6	601705
FOXP3	129.5	99.2	95.5	304790
FPR1	183.0	100.0	100.0	-
G6PC	185.9	100.0	100.0	232200
G6PC3	113.2	100.0	99.9	612541
G6PD	114.5	99.3	98.0	305900
GATA2	119.1	100.0	98.3	614172
GFI1	88.3	100.0	99.2	607847;613107
GIMAP5	208.7	100.0	100.0	-
GINS1	140.2	99.3	94.9	617827
GJC2	37.0	78.2	58.8	613480
GRHL2	139.4	100.0	100.0	616029
GTF2H5	75.9	72.5	72.2	601675
HAVCR2	141.9	100.0	100.0	618398
HAX1	166.6	100.0	100.0	610738
HELLS	131.6	97.8	92.1	616911
HMOX1	137.4	98.4	89.9	614034
HYOU1	153.0	100.0	99.5	233600
ICOS	184.9	99.9	99.8	607594
ICOSLG	161.7	99.5	98.8	-
IFIH1	137.3	99.7	98.4	615846;182250
IFNAR1	101.9	97.7	97.1	-
IFNAR2	160.2	100.0	99.7	616669
IFNG	190.2	100.0	100.0	No OMIM phenotype
IFNGR1	162.4	98.2	97.5	615978;209950
IFNGR2	164.4	93.3	93.2	614889
IGHM	170.9	100.0	100.0	601495
IGLL1	91.1	99.9	96.9	613500
IKBKB	133.8	99.8	97.4	615592;618204
IKBKG	62.9	84.1	77.2	300291;300291;308300

IKZF1	188.0	99.3	99.3	616873
IL10	109.7	99.8	98.2	609423
IL10RA	151.8	100.0	100.0	613148
IL10RB	157.2	99.8	98.0	612567
IL12B	118.2	100.0	99.3	209950
IL12RB1	121.8	98.9	96.3	209950
IL17F	88.7	99.9	97.2	613956
IL17RA	129.6	100.0	99.4	613953
IL17RC	121.0	100.0	99.9	616445
IL18BP	175.8	100.0	100.0	618549
IL1RN	154.6	100.0	100.0	612852
IL2	84.5	94.5	88.0	613011
IL21	77.0	99.4	95.7	615767
IL21R	145.0	100.0	100.0	615207
IL2RA	122.3	100.0	99.7	606367
IL2RB	117.9	100.0	99.7	618495
IL2RG	71.1	99.8	97.1	312863
IL36RN	104.4	100.0	100.0	614204
IL6R	153.1	98.4	94.2	614752;614689
IL6ST	112.4	96.4	90.3	618523
IL7R	147.7	100.0	99.8	608971
INO80	114.7	100.0	99.1	-
INSR	138.2	97.8	94.7	246200
IRAK1	76.4	99.3	94.9	-
IRAK4	116.9	99.8	97.7	610799
IRF2BP2	79.7	93.9	77.7	617765
IRF3	140.7	100.0	99.8	616532
IRF4	220.9	100.0	100.0	611724
IRF7	126.8	100.0	99.9	616345
IRF8	113.2	99.0	95.7	614893;614894

IRF9	138.5	100.0	100.0	618648
IRGM	124.7	100.0	100.0	612278;607948
ISG15	150.7	100.0	100.0	616126
ITCH	133.7	91.6	91.3	613385
ITGB2	160.3	97.2	97.2	116920
ITK	126.2	100.0	98.9	613011
ITPR3	152.6	100.0	99.7	No OMIM phenotype
IVNS1ABP	154.1	99.9	98.4	618969
JAGN1	119.7	100.0	100.0	616022
JAK1	132.4	100.0	99.8	618999
JAK2	127.4	98.1	95.8	614521
JAK3	127.9	99.9	98.7	600802
KDM6A	114.8	96.1	88.7	300867
KMT2D	142.1	100.0	99.4	147920
KRAS	84.3	99.5	96.9	614470
LACC1	174.2	100.0	99.4	618795
LAMTOR2	189.2	100.0	99.7	610798
LAT	120.5	100.0	99.2	617514
LCK	166.5	98.9	96.6	615758
LIG1	117.7	100.0	99.7	-
LIG4	222.9	100.0	99.9	602450;606593
LPIN2	119.0	100.0	100.0	609628
LRBA	154.3	99.9	99.6	614700
LRRC8A	247.1	100.0	99.8	613506
LSM11	136.5	99.9	97.6	No OMIM phenotype
LYST	169.3	99.6	98.3	214500
MAGT1	124.5	98.5	96.5	300853
MAL2	192.3	99.9	98.0	-
MALT1	154.0	91.2	89.4	615468
MAN2B1	142.3	99.8	97.9	248500



MANBA	128.8	87.8	86.5	248510
MAP1LC3B2	163.4	100.0	100.0	-
MAP3K14	130.6	100.0	99.9	-
MAPK8	165.9	100.0	99.8	-
MASP2	137.2	100.0	99.6	613791
MBL2	127.1	100.0	99.8	614372
MC2R	183.7	99.9	98.3	202200
MCM10	137.5	100.0	99.9	-
MCM4	140.8	95.5	95.0	609981
MEFV	141.6	99.9	98.6	134610
MOGS	146.7	100.0	99.9	606056
MRE11	64.1	98.9	93.3	604391
MRTFA	106.5	91.4	90.2	618847
MS4A1	140.8	99.8	98.8	613495
MSN	80.9	99.0	95.7	300988
MTHFD1	144.1	100.0	99.5	617780
MVK	120.8	90.9	90.5	260920
MYD88	179.6	100.0	100.0	612260
MYSM1	130.4	96.4	95.5	618116
NBAS	169.2	100.0	99.6	616483
NBN	109.6	99.9	98.6	251260
NCF1	25.8	26.0	25.8	233700
NCF2	145.7	99.9	98.3	233710
NCF4	168.1	100.0	100.0	613960
NCKAP1L	146.5	100.0	99.9	618982
NCSTN	107.5	100.0	99.8	142690
NFAT5	203.4	99.8	99.1	-
NFE2L2	199.9	100.0	99.9	617744
NFKB1	113.7	100.0	99.4	616576
NFKB2	134.6	98.8	95.6	615577

NFKBIA	132.6	95.2	88.0	612132
NHEJ1	65.6	100.0	96.2	611291
NHP2	146.1	100.0	100.0	613987
NLRC4	191.9	100.0	100.0	616115;616050
NLRP1	131.6	99.6	98.0	617388;606579
NLRP12	176.9	100.0	99.9	611762
NLRP3	153.4	100.0	99.9	120100;617772;191900;607115
NLRP6	106.4	98.7	95.9	No OMIM phenotype
NLRP7	128.2	100.0	99.6	-
NOD2	126.1	100.0	99.9	186580
NOP10	147.4	100.0	99.8	224230
NOS2	111.8	96.6	92.4	-
NRAS	185.6	100.0	100.0	614470
NSMCE3	213.0	100.0	100.0	617241
OAS1	139.8	100.0	100.0	-
ORAI1	183.1	99.1	96.4	612782
OSTM1	95.7	98.6	94.0	259720
OTULIN	148.9	92.6	86.5	617099
PARN	134.2	81.2	81.1	616353;616371
PAX1	165.8	92.4	87.9	615560
PAX5	115.0	98.7	96.1	615545
PBX1	146.9	100.0	99.4	617641
PCCA	115.9	99.5	96.7	606054
PCCB	133.2	97.9	96.0	606054
PEPD	109.7	100.0	98.8	170100
PEX16	158.6	97.9	94.2	No OMIM phenotype
PGM3	177.0	100.0	99.8	615816
PIGA	85.7	93.8	86.7	300818
PIK3CD	160.8	98.8	96.9	615513
PIK3CG	194.6	100.0	100.0	No OMIM phenotype

PIK3R1	148.2	99.8	99.0	616005
PLCG2	125.6	100.0	99.8	614878;614468
PLEKHM1	129.9	100.0	99.8	611497
PLG	117.3	87.8	87.5	217090
PMM2	145.4	100.0	100.0	212065
PNP	124.6	99.8	98.9	613179
POLA1	123.1	99.3	95.4	301220
POLE2	82.0	97.3	89.8	-
POMP	160.3	100.0	99.1	618048;601952
POT1	120.7	99.9	99.0	615848
POU2AF1	115.0	100.0	99.3	ESID2020: POU2AF1 (BOB1), B-cell differentiation, Kury et al. in preparation
PRF1	114.3	91.2	90.8	603553
PRKCD	175.0	100.0	100.0	615559
PRKDC	118.8	99.7	98.0	615966
PRPS1	118.4	86.4	86.4	301835
PSEKEN	103.3	100.0	100.0	613736
PSMA3	87.4	99.8	97.2	-
PSMB4	127.4	100.0	100.0	617591
PSMB8	132.9	99.9	98.5	256040
PSMB9	102.1	99.9	97.7	617591
PSMG2	142.0	100.0	98.9	619183
PSTPIP1	98.5	100.0	99.1	604416
PTPN22	148.3	99.5	97.1	152700
PTPRC	122.1	99.0	95.1	608971
RAB27A	163.4	100.0	100.0	607624
RAC2	104.2	99.9	98.3	608203;618986;618987
RAG1	169.0	100.0	100.0	609889
RAG2	223.3	100.0	100.0	233650
RANBP2	122.8	49.7	49.3	608033
RASGRP1	129.8	100.0	99.6	618534

RASGRP2	105.0	99.7	97.3	615888
RBCK1	105.6	99.9	98.2	615895
RC3H1	144.0	100.0	99.4	618998
RECQL4	149.8	99.8	98.1	268400
RELB	115.2	98.8	88.7	617585
RFX5	119.8	99.7	98.1	209920
RFXANK	130.0	100.0	99.5	209920
RFXAP	116.2	99.3	97.0	209920
RHOG	264.8	100.0	100.0	No OMIM phenotype
RHOH	148.8	100.0	100.0	618307
RIPK1	118.2	100.0	99.0	618108
RMRP				250250
RNASEH2A	150.5	100.0	100.0	610333
RNASEH2B	116.5	80.6	78.1	610181
RNASEH2C	279.8	100.0	99.5	610329
RNF168	202.2	100.0	99.8	611943
RNF31	157.3	99.9	99.0	-
RNU4ATAC				210710
RORC	123.9	100.0	100.0	616622
RPSA	75.7	100.0	99.8	271400
RSPH9	135.9	99.9	97.9	612650
RTEL1	127.7	99.5	96.8	615190
SAMD9	185.7	100.0	99.8	610455;617053
SAMD9L	192.5	100.0	100.0	159550
SAMHD1	162.2	98.7	98.4	612952
SASH3	103.0	99.9	97.6	No OMIM phenotype
SBDS	197.8	100.0	100.0	260400
SCIMP	113.1	81.0	79.9	-
SEC61A1	148.0	100.0	100.0	617056
SEMA3E	158.1	99.2	98.9	214800

SERAC1	134.6	99.9	99.5	614739
SERPING1	98.8	99.7	97.5	120790
SH2B3	106.6	99.4	95.1	-
SH2D1A	122.6	97.2	94.0	308240
SH3BP2	152.8	91.4	91.2	118400
SH3KBP1	106.6	99.7	95.9	300310
SKIV2L	155.8	100.0	99.8	614602
SLC29A3	192.3	100.0	99.6	602782
SLC35A1	150.9	100.0	99.7	603585
SLC35C1	162.1	99.9	98.7	266265
SLC37A4	124.2	100.0	99.2	232220
SLC39A4	100.1	99.5	95.5	201100
SLC39A7	149.0	100.0	100.0	-
SLC46A1	116.3	99.9	98.5	229050
SLC7A7	126.7	100.0	99.9	-
SMARCAL1	130.9	100.0	99.9	242900
SMARCD2	102.9	87.0	85.9	617475
SNORA31				-
SNX10	159.9	96.2	95.7	615085
SOCS1	101.3	100.0	100.0	No OMIM phenotype
SOCS4	271.8	99.9	99.2	-
SP110	134.6	100.0	100.0	235550
SPINK5	142.1	99.9	99.5	256500
SPPL2A	77.1	85.9	74.6	-
SRP54	133.2	99.5	96.5	618752
SRP72	81.8	97.6	89.7	614675
STAT1	134.8	93.7	91.7	614162
STAT2	115.3	100.0	99.9	616636
STAT3	129.0	100.0	99.8	147060
STAT4	174.6	99.9	99.6	612253

STAT5B	136.2	100.0	98.5	245590
STAT6	128.0	100.0	99.9	-
STIM1	126.2	99.8	98.0	612783
STING1	115.6	99.7	95.3	615934
STK4	138.1	100.0	99.8	614868
STX11	275.0	100.0	100.0	603552
STXBP2	104.1	82.1	79.7	613101
TAP1	108.6	100.0	99.2	604571
TAP2	108.9	99.9	99.3	604571
TAPBP	121.8	96.5	95.5	604571
TAZ	101.2	99.1	95.5	302060
TBX1	89.7	87.0	77.5	602054
TBX21	108.2	95.4	86.6	208550
TCF3	96.2	97.1	94.0	616941
TCIRG1	116.3	97.6	90.1	259700
TCN2	178.3	100.0	100.0	275350
TERC				127550
TERT	132.4	96.2	94.5	613989
TET2	201.7	100.0	100.0	619126;614286
TFRC	166.7	100.0	99.8	616740
TGFB1	89.2	100.0	99.9	618213
THBD	139.3	100.0	99.7	612926
TICAM1	118.3	100.0	99.9	614850
TINF2	187.5	100.0	100.0	613990
TIRAP	117.4	100.0	100.0	607948;614382;610799
TLR3	221.7	100.0	99.6	613002
TLR4	159.0	100.0	99.8	-
TLR7	128.6	100.0	99.9	301051
TLR8	176.8	100.0	99.8	No OMIM phenotype
TMC6	90.0	100.0	99.3	226400

TMC8	125.7	100.0	98.7	226400
TNFAIP3	176.4	100.0	100.0	616744
TNFRSF11A	141.5	94.6	93.3	612301
TNFRSF13B	106.3	100.0	100.0	240500
TNFRSF13C	97.5	80.1	75.4	613494
TNFRSF1A	95.0	90.6	87.6	142680
TNFRSF4	66.3	99.4	95.4	615593
TNFRSF9	157.2	100.0	100.0	-
TNFSF11	156.7	100.0	99.9	259710
TNFSF12	89.6	98.0	93.6	-
TNFSF13	89.3	98.2	92.4	-
TOP2B	140.2	99.4	96.3	-
TPP2	136.7	99.2	96.8	-
TRAC	142.3	100.0	100.0	615387
TRAF3	129.6	100.0	99.9	614849
TRAF3IP2	127.7	100.0	99.3	615527
TREX1	248.4	100.0	100.0	225750
TRIM22	152.8	100.0	100.0	-
TRNT1	108.4	99.5	96.5	616084
TTC37	165.8	100.0	99.3	222470
TTC7A	121.3	99.3	95.4	243150
TYK2	133.3	99.9	99.0	611521
UBA1	142.9	99.4	98.2	301054
UNC13D	106.2	99.7	98.1	608898
UNC93B1	60.3	60.6	58.8	610551
UNG	124.8	100.0	98.8	608106
USB1	139.6	100.0	99.4	604173
USP18	185.6	95.9	95.9	617397
VAV1	105.9	98.5	97.1	-
VPS13B	163.9	99.5	98.2	216550

VPS45	157.5	99.2	95.7	615285
WAS	81.1	95.9	85.3	300299
WDR1	116.9	100.0	99.6	150550
WIPF1	111.8	100.0	99.9	614493
WRAP53	187.5	100.0	100.0	613988
XIAP	100.4	93.0	88.8	300635
ZAP70	166.6	100.0	99.3	269840;617006
ZBTB24	168.0	100.0	100.0	614069
ZNF341	129.4	97.2	95.0	618282

*Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.*

*Median Coverage describes the average number of reads seen across 50 exomes.*

*% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.*

*% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.*

*Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.*

*OMIM release used for OMIM disease identifiers and descriptions : October 1st, 2016.*

*Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*