

# WES VISION DISORDERS DG 3.2

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt;10x</i>	<i>% covered &gt;20x</i>	<i>OMIM disease ID</i>
ABCA4	110.4	99.9	99.0	604116;601718;248200;153800
ABCC6	110.8	93.6	92.5	177850;264800
ABHD12	81.7	91.7	86.0	612674
ACBD5	150.7	100.0	98.4	-
ACO2	128.7	94.1	86.3	616289
ADAM9	149.9	99.7	98.0	612775
ADAMTS18	141.0	100.0	99.6	615458
ADAMTSL4	138.1	99.8	98.4	225200
ADGRV1	134.4	99.5	98.4	605472
ADIPOR1	92.8	99.2	94.4	-
AFG3L2	98.3	94.6	86.3	618977
AGBL1	109.0	98.5	98.1	615523
AGBL5	107.3	99.6	98.4	617023
AGK	104.4	90.4	87.9	614691
AHI1	132.9	99.4	97.4	608629
AHR	171.8	99.2	98.6	618345
AIPL1	106.0	100.0	99.2	604393
ALDH18A1	120.0	100.0	99.9	616603;219150
ALDH1A3	100.0	97.6	94.6	615113
ALMS1	174.7	99.7	99.5	203800
AP3B1	114.7	99.2	96.4	608233
AP3D1	124.5	99.6	98.4	617050
ARHGEF18	151.0	98.8	93.9	617433
ARL13B	85.8	100.0	99.3	612291
ARL2	136.0	100.0	99.7	619082

ARL2BP	67.3	94.9	87.3	615434
ARL3	80.1	99.7	96.5	618173
ARL6	112.4	99.1	98.4	613575;209900
ARR3	90.7	100.0	99.6	301010
ARSG	119.0	99.9	98.0	618144
ASB10	106.1	99.4	95.7	603383
ASPH	115.2	99.6	98.6	601552
ASRGL1	111.7	100.0	99.9	-
ATF6	134.6	99.9	99.5	616517
ATOH7	101.7	97.7	92.6	221900
B3GLCT	100.4	99.7	98.2	261540
BBIP1	142.6	95.7	87.4	615995
BBS1	160.2	100.0	100.0	209900
BBS10	151.0	100.0	99.9	615987
BBS12	167.8	100.0	100.0	615989
BBS2	148.8	99.4	98.0	615981;616562
BBS4	108.1	99.9	98.9	615982
BBS5	95.0	98.4	94.7	615983
BBS7	151.4	99.0	96.5	615984
BBS9	104.9	92.0	89.0	615986
BCOR	99.9	99.2	95.8	300166
BEST1	131.3	98.7	95.0	608161;613194;193220;611809;153700
BFSP1	94.4	99.8	94.0	611391
BFSP2	89.3	99.7	97.2	611597
BLOC1S3	42.1	99.9	90.3	614077
BLOC1S5	129.6	99.7	98.7	619172
BLOC1S6	106.9	99.3	97.6	614171
BMP4	175.3	100.0	100.0	607932
C12orf65	78.9	99.0	94.5	613559;615035
C19orf12	122.8	100.0	99.8	614298

C1QTNF5	122.4	91.1	78.9	605670
C8orf37	135.1	99.7	99.6	617406;614500
CABP4	173.0	100.0	99.6	610427
CACNA1F	93.4	99.7	97.2	300071;300476;300600
CACNA2D4	97.7	98.6	96.9	610478
CAPN5	153.0	100.0	99.9	193235
CC2D2A	109.3	98.3	96.6	612284;216360;612285
CCT2	146.9	100.0	99.6	-
CDH2	119.0	99.4	97.1	618929
CDH23	189.6	100.0	100.0	601067
CDH3	151.5	100.0	99.1	601553;225280
CDHR1	145.7	99.5	98.1	613660
CDK10	122.7	100.0	99.5	-
CEP120	140.6	99.9	99.6	616300
CEP164	95.5	99.8	98.2	614845
CEP250	95.4	99.9	98.4	618358
CEP290	86.2	96.2	90.8	611134;615991;611755;610189;610188
CEP41	77.1	98.8	93.4	614464
CEP78	114.1	98.8	95.6	617236
CEP83	107.1	99.0	96.6	615862
CERKL	115.1	99.3	96.5	608380
CFAP410	129.3	100.0	99.6	617547
CFH	178.9	99.8	98.5	126700
CHD7	137.1	100.0	99.2	214800
CHM	103.2	98.2	91.6	303100
CHMP4B	142.7	100.0	99.2	605387
CHRD1	98.8	100.0	99.5	309300
CHST6	216.4	100.0	100.0	217800
CIB2	218.0	99.3	96.2	614869
CISD2	112.9	83.4	83.4	604928

CLCC1	100.1	99.5	95.6	-
CLDN19	136.6	98.3	92.9	248190
CLN3	128.6	92.5	92.4	204200
CLN5	106.5	69.0	66.3	256731
CLN6	136.4	99.9	98.9	601780
CLN8	152.5	83.5	83.5	600143
CLRN1	127.0	100.0	99.7	276902;614180
CLUAP1	126.7	99.9	99.7	204000
CNGA1	113.3	91.0	85.6	613756
CNGA3	164.1	100.0	99.8	216900
CNGB1	112.9	99.0	97.1	613767
CNGB3	111.0	97.8	90.6	262300;248200
CNNM4	164.9	99.9	99.0	217080
COA8	71.5	81.9	80.8	220110
COL11A1	100.9	96.0	92.7	154780;604841
COL17A1	108.1	99.0	96.3	122400
COL18A1	132.1	98.2	95.7	267750
COL25A1	126.3	95.6	95.3	616219
COL2A1	122.0	100.0	99.8	108300
COL4A1	103.1	99.0	97.0	175780
COL8A2	107.4	100.0	99.8	136800;609140
COL9A1	129.1	99.9	98.6	614134
COL9A2	105.0	99.9	98.9	614284
COL9A3	106.6	98.9	95.5	-
CPAMD8	103.6	95.9	92.6	617319
CPLANE1	129.6	99.4	98.2	614615
CPSF1	157.2	98.5	97.1	618827
CRB1	163.0	100.0	99.9	613835;172870;600105
CRX	216.9	100.0	100.0	613829;120970
CRYAA	128.3	99.9	98.0	604219

CRYAB	95.9	100.0	98.2	613763
CRYBA1	113.3	100.0	99.6	600881
CRYBA2	162.2	100.0	100.0	115900
CRYBA4	121.2	100.0	100.0	610425
CRYBB1	127.3	100.0	99.9	611544
CRYBB2	144.9	100.0	99.9	601547
CRYBB3	150.9	100.0	99.9	609741
CRYGB	101.5	100.0	98.7	615188
CRYGC	130.0	99.0	95.2	604307
CRYGD	111.7	100.0	98.9	115700
CRYGS	87.5	92.8	82.1	116100
CSPP1	116.4	99.7	98.1	615636
CTDP1	125.4	88.7	85.0	604168
CTNNA1	113.3	98.8	97.2	608970
CTNNB1	127.1	100.0	100.0	617272
CTSD	174.4	98.4	95.0	610127
CTSH	109.3	100.0	100.0	-
CWC27	87.4	99.5	95.9	250410
CYP1B1	96.4	100.0	100.0	231300
CYP4V2	140.5	99.6	97.0	210370
DCN	136.1	95.7	95.0	610048
DCT	142.6	99.9	99.9	619165
DDHD1	142.3	98.5	96.5	-
DHDDS	80.8	99.4	95.6	613861
DHX38	109.0	100.0	99.1	618220
DKC1	90.7	99.7	97.2	305000
DNAJC30	149.9	100.0	100.0	619382
DNM1L	118.8	99.6	98.3	610708
DNMBP	134.7	99.9	98.9	618415
DRAM2	128.0	100.0	99.9	616502

DTNBP1	109.6	99.7	98.0	614076
DYNC2H1	107.7	98.6	95.2	613091
EFEMP1	146.4	100.0	100.0	126600
ELOVL1	87.5	99.6	96.5	618527
ELOVL4	104.8	99.7	98.9	600110
EMC1	112.1	99.9	98.0	616875
EPG5	113.6	99.2	97.8	242840
EPHA2	167.2	100.0	99.7	116600
ERCC2	130.5	100.0	99.4	601675
EXOSC2	109.0	100.0	99.9	617763
EYA1	120.9	99.9	99.5	602588;113650
EYS	138.7	99.5	97.5	602772
FA2H	86.5	92.4	82.6	612319
FAM161A	137.4	99.8	99.5	606068
FBN1	149.0	100.0	99.7	154700
FDXR	131.6	100.0	98.6	617717
FLVCR1	142.6	99.7	98.3	609033
FOXC1	56.3	98.7	93.7	602482;601631
FOXE3	52.2	87.6	79.0	612968;610256
FREM1	116.9	99.8	98.4	248450
FRMD7	99.7	99.9	98.1	310700
FTL	134.8	98.6	88.5	600886
FYCO1	117.7	100.0	100.0	610019
FZD4	169.6	100.0	100.0	133780
GALK1	156.6	100.0	99.2	230200
GALM	93.5	100.0	99.5	-
GALT	160.1	100.0	99.6	230400
GCNT2	152.2	99.5	99.5	116700
GDF3	114.9	100.0	100.0	613704;613703
GDF6	113.8	100.0	100.0	613094;615360

GDPD1	133.0	99.3	96.2	600852
GFER	89.3	99.8	97.6	613076
GJA1	151.1	100.0	100.0	257850;164200
GJA3	146.4	100.0	99.8	601885
GJA8	170.3	100.0	100.0	116200
GNAT1	157.4	100.0	100.0	616389;610444
GNAT2	108.1	99.9	97.9	613856
GNB3	147.5	100.0	100.0	617024
GNPTG	144.9	99.8	96.6	252605
GPR143	60.2	86.3	74.8	300814;300500
GPR179	145.5	100.0	100.0	614515
GRHL2	125.6	100.0	99.9	618031
GRK1	159.1	100.0	100.0	613411
GRM6	137.8	93.1	83.6	257270
GSN	113.6	95.7	93.5	105120
GUCA1A	163.6	100.0	100.0	602093
GUCA1B	119.6	100.0	100.0	613827
GUCY2D	103.9	100.0	98.7	204000;601777
HARS1	135.7	100.0	100.0	614504
HCCS	99.1	99.3	96.1	309801
HGSNAT	109.4	86.4	86.2	252930;616544
HK1	120.9	100.0	99.9	617460
HMX1	23.5	64.0	43.1	612109
HPS1	121.5	100.0	100.0	203300
HPS3	137.0	99.8	97.1	614072
HPS4	126.3	100.0	100.0	614073
HPS5	119.5	99.9	99.3	614074
HPS6	173.8	97.7	88.7	614075
HRAS	179.8	100.0	100.0	218040
HSF4	138.4	99.0	96.3	116800

HSPG2	123.6	99.2	97.5	255800
IDH3A	133.6	98.9	97.3	619007
IDH3B	127.4	95.4	95.4	612572
IFT140	120.4	99.9	99.2	266920;617781
IFT172	94.8	99.6	98.6	615630;616394
IFT27	119.6	100.0	100.0	615996
IFT43	116.4	100.0	100.0	617871
IFT52	130.6	100.0	99.9	617102
IFT74	81.3	98.6	96.2	617119
IFT81	91.9	92.9	89.6	-
IMPDH1	56.2	89.0	81.7	180105;613837
IMPG1	102.5	99.6	98.3	616151
IMPG2	130.6	99.4	97.9	613581;616152
INPP5E	113.1	96.9	93.2	610156;213300
INVS	145.6	100.0	99.8	-
IQCB1	96.5	92.8	82.8	609254
IRX1	134.0	87.1	81.1	-
ITPR1	131.7	100.0	99.5	206700
JAG1	127.4	97.8	96.7	118450
JAM3	134.7	100.0	100.0	613730
KCNJ13	141.3	100.0	100.0	614186;193230
KCNV2	120.1	100.0	99.7	610356
KERA	186.3	100.0	100.0	217300
KIAA0586	126.1	97.1	92.0	616546
KIAA1549	112.6	97.8	96.3	618613
KIF11	87.8	96.8	93.1	152950
KIF21A	127.5	99.7	98.9	135700
KIF3B	99.2	99.9	98.9	618955
KIF7	105.7	93.6	91.9	200990
KIZ	141.5	99.8	98.4	615780



KLHL7	112.8	99.8	99.7	612943
KRT12	143.9	99.9	98.8	122100
KRT3	129.7	100.0	99.7	122100
LAMA1	123.5	99.9	99.3	615960
LAMB2	174.2	99.9	99.3	609049
LAMP2	88.2	99.3	96.0	300257
LCA5	136.1	99.6	97.9	604537
LEMD2	91.7	99.9	96.1	212500
LIM2	103.7	100.0	99.8	615277
LRAT	288.9	100.0	100.0	613341
LRIT3	108.5	94.1	92.2	615058
LRMDA	120.5	97.4	95.5	615179
LRP2	147.6	100.0	99.8	222448
LRP5	183.2	99.2	98.2	259770;601813
LRPAP1	110.3	100.0	99.9	615431
LSS	138.7	100.0	99.4	616509
LTBP2	115.3	99.8	98.9	613086
LYST	140.4	99.4	97.8	214500
LZTFL1	112.9	99.7	99.4	615994
MAB21L2	193.2	100.0	100.0	615877
MAF	71.4	84.3	78.9	610202
MAK	134.1	99.7	97.9	614181
MAPKAPK3	93.3	100.0	98.6	617111
MERTK	136.9	99.4	98.6	613862
MFN2	118.1	100.0	99.8	601152;609260
MFRP	136.5	100.0	100.0	611040;609549
MFSD8	121.3	99.6	99.4	610951;616170
MIP	125.4	99.7	94.1	615274
MIR184				614303
MITF	135.0	100.0	99.9	103470

MKKS	204.9	100.0	100.0	209900
MKS1	93.4	99.4	96.3	615990;249000;617121
MVK	117.9	91.4	90.5	260920;610377
MYO7A	137.3	99.7	98.3	276900
MYOC	136.1	99.9	98.0	137750
NAA10	110.0	99.8	97.9	309800
NBAS	141.0	99.9	99.3	614800
NDP	89.6	100.0	99.7	305390;310600
NDUFS2	98.4	100.0	100.0	252010
NEK2	82.0	99.3	93.4	615565
NEUROD1	135.1	100.0	99.4	-
NHS	113.5	96.1	94.1	302350;302200
NMNAT1	110.5	100.0	99.2	608553
NPHP1	135.6	99.8	99.1	266900;609583
NPHP3	122.6	99.6	98.5	267010
NPHP4	130.3	100.0	99.8	606996
NR2E3	111.5	100.0	99.6	268100;611131
NR2F1	188.1	100.0	100.0	615722
NRL	106.0	99.6	96.4	613750
NYX	111.1	97.4	96.1	310500
OAT	72.2	82.0	73.0	258870
OCA2	118.9	99.9	98.3	203200
OCRL	107.2	99.4	97.6	309000
OFD1	50.3	87.1	71.3	300804;300424
OPA1	128.7	99.5	96.7	125250;165500
OPA3	134.8	100.0	99.5	165300;258501
OPN1LW	76.5	66.3	59.8	303700
OPN1MW	68.2	69.8	62.9	303700
OPTN	107.3	99.9	99.9	137760
OTX2	124.0	100.0	99.0	610125

OVOL2	102.3	97.8	91.8	122000
P3H2	95.0	99.9	98.0	614292
P4HA2	123.7	99.9	98.7	617238
PANK2	159.3	100.0	99.7	234200;607236
PAX2	193.6	100.0	100.0	120330
PAX6	117.9	100.0	99.9	136520;120200;604229;120430;106210
PCARE	113.0	99.6	98.1	613428
PCDH15	144.5	97.9	96.8	602083;601067
PCYT1A	102.5	99.2	95.7	608940
PDE6A	105.6	100.0	99.5	613810
PDE6B	152.8	100.0	99.8	613801;163500
PDE6C	116.0	99.7	97.0	613093
PDE6D	108.3	100.0	99.9	615665
PDE6G	141.6	100.0	100.0	613582
PDE6H	60.0	99.8	86.1	610024
PDZD7	84.5	96.0	91.5	605472
PET100	88.7	100.0	99.2	220110
PEX1	135.8	99.8	99.4	601539;214100
PEX2	133.2	100.0	100.0	614866;614867
PEX26	86.7	100.0	99.8	614872
PEX6	109.2	96.4	88.0	614862
PEX7	113.8	88.0	81.0	215100;614879
PGK1	43.3	90.3	73.2	300653
PHOX2A	36.3	92.8	74.4	602078
PHYH	71.1	100.0	98.9	266500
PIKFYVE	143.0	99.9	99.3	121850
PITX2	147.2	99.8	97.2	137600
PITX3	61.6	100.0	98.0	107250;610623
PLA2G5	120.0	100.0	100.0	228980
PLK4	145.5	99.4	98.4	616171

PNPLA6	144.9	100.0	99.8	215470;275400;245800
POC1B	80.3	99.5	97.3	615973
POC5	155.7	99.4	97.1	-
POMGNT1	116.4	100.0	99.8	617123
PPT1	137.3	90.3	89.9	256730
PRCD	128.1	100.0	100.0	610599
PRDM13	132.6	98.5	93.5	136550
PRDM5	132.3	99.8	98.4	614170
PRIMPOL	118.1	97.5	94.6	615421
PROM1	111.9	97.2	96.5	608051;612657;603786;612095
PRPF3	72.8	97.8	92.9	601414
PRPF31	138.2	99.3	95.1	600138
PRPF4	124.8	99.9	99.8	615922
PRPF6	110.3	100.0	99.7	613983
PRPF8	108.7	99.9	98.6	600059
PRPH2	207.6	100.0	100.0	608161;136880;169150;608133;613105
PRR11	178.8	100.0	99.9	600852
PRSS56	85.3	100.0	98.0	613517
PXDN	134.4	99.9	99.6	269400
RAB28	65.9	99.2	97.2	615374
RAB3GAP2	90.7	99.1	96.3	614225
RARB	93.0	100.0	99.8	615524
RAX	106.2	98.1	89.3	611038
RAX2	78.8	100.0	95.5	610381
RBP3	152.4	100.0	100.0	615233
RBP4	126.9	99.6	96.3	615147;616428
RCBTB1	106.1	99.7	98.6	617175
RD3	171.3	100.0	100.0	610612
RDH11	89.0	99.6	96.9	616108
RDH12	82.4	99.3	95.4	612712

RDH5	178.1	100.0	100.0	136880
REEP6	195.5	100.0	99.9	617304
RGS9	111.7	97.9	96.5	608415
RGS9BP	114.7	100.0	99.4	608415
RHO	171.4	100.0	100.0	610445;613731
RIMS1	123.5	99.7	97.3	603649
RIMS2	130.3	96.6	94.5	618970
RLBP1	120.4	100.0	99.8	607476;607475;136880
ROM1	112.5	100.0	99.9	608133
RP1	109.2	91.2	90.7	180100
RP1L1	139.1	100.0	100.0	613587;618826
RP2	161.5	99.8	99.8	312600
RP9	63.8	82.5	72.7	180104
RPE65	125.6	99.9	98.7	204100;613794;618697
RPGR	68.9	76.4	70.8	304020;300455;300834;300029
RPGRIP1	124.2	100.0	99.7	613826;608194
RPGRIP1L	129.9	96.5	95.3	216360;611560;611561
RS1	54.2	98.4	89.6	312700
RTN4IP1	78.6	99.6	97.3	616732
SAG	130.6	100.0	100.0	613758;258100
SAMD11	102.0	92.2	85.0	-
SC5D	178.6	99.9	99.1	607330
SCAPER	150.7	99.5	97.1	618195
SCO2	117.1	100.0	100.0	608908
SDCCAG8	115.0	99.8	99.8	613615
SEMA4A	127.6	100.0	99.4	610282;610283
SGSH	143.6	94.8	94.1	252900
SHH	125.7	100.0	100.0	611638
SIL1	148.1	98.7	96.0	248800
SIPA1L3	181.9	100.0	99.3	616851

SIX6	274.1	100.0	100.0	212550
SLC16A12	133.5	100.0	99.9	612018
SLC24A1	158.5	100.0	99.9	613830
SLC24A5	97.0	99.6	98.5	113750
SLC25A46	184.6	99.7	98.6	616505
SLC33A1	120.6	99.8	98.5	614482
SLC38A8	83.4	99.9	97.9	609218
SLC39A5	135.8	99.9	99.0	615946
SLC45A2	125.6	100.0	99.9	606574
SLC4A11	173.0	100.0	99.9	217700;613268;217400
SLC52A2	171.9	100.0	100.0	614707
SLC7A14	139.4	100.0	100.0	615725
SMG8	151.1	100.0	100.0	600852
SMOC1	116.5	99.8	98.2	206920
SNRNP200	120.8	99.8	98.3	610359
SOX2	194.7	100.0	99.8	206900
SOX5	93.7	99.8	97.6	616803
SPATA7	122.1	99.6	98.2	604232
SPP2	115.2	99.8	99.8	-
SSBP1	56.3	99.1	94.1	165510
STRA6	131.1	100.0	99.9	601186
STX3	109.7	93.0	92.5	619446
TACSTD2	219.5	99.1	96.6	204870
TCF4	106.4	100.0	99.9	613267
TCTN1	102.5	96.8	92.8	614173
TCTN2	127.2	99.9	99.1	613885;616654
TCTN3	118.9	100.0	100.0	614815
TDRD7	139.9	99.9	99.3	613887
TEAD1	136.0	100.0	99.7	108985
TEK	155.7	100.0	99.9	617272

TENM3	153.2	100.0	99.7	615145
TGFBI	117.7	99.9	96.3	122200;121900;602082;608470;607541;608471;121820
TIMM8A	50.9	96.2	83.1	304700;311150
TIMP3	136.8	100.0	100.0	136900
TMCO3	125.6	99.9	98.6	-
TMEM126A	105.6	95.4	80.0	612989
TMEM138	87.3	99.8	93.1	614465
TMEM216	89.9	98.5	92.8	608091
TMEM231	99.0	100.0	99.3	615397;614970
TMEM237	121.4	99.8	99.3	614424
TMEM67	84.8	98.6	93.5	607361;610688;216360
TMEM98	126.2	99.2	95.8	615972
TOGARAM1	127.1	99.6	97.5	619185
TOPORS	183.9	100.0	100.0	609923
TPP1	130.4	100.0	100.0	204500
TRAF3IP1	82.2	98.7	95.4	616629
TREX1	288.9	100.0	100.0	192315
TRIM32	119.5	100.0	99.9	615988
TRNT1	84.7	99.7	97.4	616959
TRPM1	131.4	100.0	99.4	613216
TSPAN12	134.8	100.0	99.9	613310
TTC8	125.6	99.5	98.0	613464;615985
TTLL5	135.5	99.9	98.1	615860
TUB	108.6	99.8	97.1	616188
TUBA3D	109.7	99.9	96.5	617928
TUBB3	118.3	98.5	96.8	600638
TUBB4B	83.7	100.0	98.7	617879
TUBGCP4	109.1	98.9	94.7	616335
TUBGCP6	147.7	100.0	99.4	251270
TULP1	128.6	99.8	98.2	600132;613843

TYR	147.1	100.0	100.0	203100;103470;606952
TYRP1	164.9	100.0	99.9	203290
UBIAD1	227.9	99.5	96.6	121800
UNC45B	116.7	99.4	98.0	616279
USH1C	99.1	99.9	99.2	276904
USH1G	172.0	99.7	96.6	606943
USH2A	133.7	100.0	99.7	613809;276901
USP45	109.7	99.4	97.6	618513
VAX1	97.1	98.9	93.9	614402
VCAN	147.9	100.0	99.9	143200
VIM	104.2	98.8	96.8	116300
VPS13B	139.7	99.4	97.8	216550
VSX1	64.0	85.4	81.3	148300
VSX2	104.3	100.0	99.2	610092;610093
WDPCP	101.6	98.0	94.1	615992
WDR19	128.4	99.8	98.6	616307;614378;614376
WDR34	119.3	100.0	99.8	615633
WDR36	134.5	99.7	97.8	609887
WFS1	192.3	100.0	99.8	222300;116400;614296
WHRN	125.1	99.6	97.6	611383
WRN	133.2	99.3	98.2	277700
YAP1	92.8	96.6	90.3	120433
YME1L1	109.3	98.9	93.7	617302
YPEL2	97.3	96.6	90.8	600852
ZEB1	153.1	100.0	99.5	609141;613270
ZNF408	149.5	100.0	100.0	616469;616468
ZNF423	189.1	100.0	100.0	614844
ZNF469	166.5	100.0	100.0	229200
ZNF513	132.9	100.0	100.0	613617
ZNF644	156.6	100.0	99.8	614167



*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.*

*TWIST is the default chemistry for all WES samples. Agilent V5 was the default chemistry until Q3 2021.*

*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 no value for coverage are non protein coding genes.*

*Non protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.*

*OMIM release used for OMIM disease identifiers and descriptions: September 1st, 2021.*

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