

WES VISION DISORDERS DG 3.00

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
ABCA4	125.5	99.9	99.3	604116;601718;248200;153800
ABCC6	117.4	93.6	92.4	177850;264800
ABHD12	94.8	91.2	85.3	612674
ACBD5	181.0	100.0	99.1	-
ACO2	123.4	96.4	90.4	616289
ADAM9	174.4	99.8	99.0	612775
ADAMTS18	163.5	100.0	99.7	615458
ADAMTSL4	134.8	100.0	99.2	225200
ADGRV1	154.9	99.6	98.6	605472
ADIPOR1	111.9	99.9	97.8	-
AFG3L2	115.4	95.0	91.1	618977
AGBL1	121.6	98.5	98.4	615523
AGBL5	124.7	99.9	99.3	617023
AGK	123.5	90.6	88.5	614691
AHI1	154.0	99.7	97.9	608629
AHR	203.6	99.2	98.8	618345
AIPL1	108.2	100.0	99.8	604393
ALDH1A3	112.4	97.2	94.5	615113
ALMS1	203.3	99.8	99.5	203800
AP3B1	140.2	99.2	95.7	608233
AP3D1	125.6	99.8	98.6	617050
ARHGEF18	152.3	95.5	92.4	617433
ARL13B	108.1	100.0	99.2	612291
ARL2BP	81.8	95.9	88.2	615434
ARL3	89.5	100.0	98.5	618173

ARL6	127.5	99.9	98.6	613575;209900
ARR3	103.0	100.0	99.8	301010
ARSG	133.2	100.0	99.6	618144
ASB10	97.8	99.4	95.8	603383
ASPH	129.8	99.9	98.8	601552
ASRGL1	141.2	100.0	100.0	-
ATF6	158.6	100.0	99.9	616517
ATOH7	103.8	96.0	91.2	221900
B3GLCT	116.2	99.6	96.3	261540
BBIP1	152.2	98.6	92.4	615995
BBS1	173.0	100.0	100.0	209900
BBS10	168.0	100.0	99.8	615987
BBS12	213.0	100.0	100.0	615989
BBS2	179.9	100.0	99.5	615981;616562
BBS4	127.4	99.9	99.3	615982
BBS5	112.8	99.0	93.8	615983
BBS7	173.6	98.7	95.5	615984
BBS9	121.7	92.3	90.4	615986
BCOR	117.9	99.6	97.4	300166
BEST1	148.0	99.4	96.4	608161;613194;193220;611809;153700
BFSP1	111.8	99.0	89.9	611391
BFSP2	94.9	99.8	97.7	611597
BLOC1S3	40.3	98.6	81.6	614077
BLOC1S6	121.6	99.9	97.7	614171
BMP4	179.3	100.0	100.0	607932
C12orf65	122.8	99.8	98.5	613559;615035
C19orf12	122.8	100.0	99.8	614298
C1QTNF5	118.4	90.9	78.5	605670
C8orf37	151.9	100.0	99.4	617406;614500
CA4	171.2	100.0	100.0	600852

CABP4	165.9	100.0	99.9	610427
CACNA1F	99.2	99.7	97.6	300071;300476;300600
CACNA2D4	107.4	98.9	97.7	610478
CAPN5	156.2	100.0	100.0	193235
CC2D2A	131.4	98.5	96.5	612284;216360;612285
CCT2	172.3	100.0	100.0	-
CDH2	141.5	99.3	97.7	618929
CDH23	188.4	100.0	100.0	601067
CDH3	158.9	100.0	99.5	601553;225280
CDHR1	162.4	99.2	98.1	613660
CDK10	125.2	100.0	99.9	-
CEP120	170.5	100.0	99.5	616300
CEP164	103.0	99.9	98.3	614845
CEP250	103.1	100.0	99.2	618358
CEP290	96.8	96.1	89.9	611134;615991;611755;610189;610188
CEP41	92.8	99.8	97.4	614464
CEP78	129.5	98.9	96.8	617236
CEP83	132.9	99.8	97.3	615862
CERKL	123.9	99.5	96.9	608380
CFAP410	107.6	100.0	99.3	617547
CFH	193.7	99.9	99.0	126700
CHD7	158.9	100.0	99.5	214800
CHM	114.6	98.4	94.4	303100
CHMP4B	147.3	100.0	99.3	605387
CHRD1	112.2	100.0	99.8	309300
CHST6	198.7	100.0	100.0	217800
CIB2	203.5	99.7	97.0	614869
CISD2	135.5	83.4	83.4	604928
CLCC1	121.0	99.8	98.0	-
CLDN19	128.9	98.6	93.2	248190

CLN3	133.3	92.5	91.8	204200
CLN5	125.9	69.3	66.3	256731
CLN6	141.7	100.0	97.2	601780
CLN8	174.3	83.5	83.5	600143
CLRN1	167.9	100.0	99.8	276902;614180
CLUAP1	155.7	100.0	99.8	204000
CNGA1	129.2	91.8	86.4	613756
CNGA3	175.6	100.0	99.7	216900
CNGB1	121.2	99.4	97.5	613767
CNGB3	133.5	99.4	95.9	262300;248200
CNNM4	176.8	99.8	99.0	217080
COA8	92.5	81.9	80.8	220110
COL11A1	113.4	96.3	92.8	154780;604841
COL18A1	123.6	98.1	95.7	267750
COL25A1	149.5	95.8	95.3	616219
COL2A1	126.2	100.0	99.7	108300
COL4A1	115.8	98.8	97.4	175780
COL8A2	89.3	100.0	98.0	136800;609140
COL9A1	159.8	100.0	99.2	614134
COL9A2	103.7	99.9	99.0	614284
COL9A3	109.4	98.8	95.6	-
CPAMD8	105.7	95.8	92.8	617319
CPLANE1	148.7	99.7	98.4	614615
CPSF1	152.9	98.2	96.5	618827
CRB1	194.4	100.0	99.9	613835;172870;600105
CRX	215.5	100.0	100.0	613829;120970
CRYAA	122.4	99.9	97.5	604219
CRYAB	108.3	100.0	99.2	613763
CRYBA1	134.4	100.0	99.4	600881
CRYBA2	163.5	100.0	100.0	115900

CRYBA4	127.2	100.0	100.0	610425
CRYBB1	130.3	100.0	100.0	611544
CRYBB2	150.2	100.0	100.0	601547
CRYBB3	169.3	100.0	100.0	609741
CRYGB	112.0	100.0	99.6	615188
CRYGC	137.8	99.8	96.9	604307
CRYGD	112.3	100.0	98.8	115700
CRYGS	109.9	94.2	86.7	116100
CSPP1	132.3	99.8	98.7	615636
CTDP1	129.8	88.4	84.3	604168
CTNNA1	126.0	99.3	98.1	608970
CTNNB1	147.0	100.0	99.9	617272
CTSD	174.9	98.5	95.0	610127
CTSH	120.0	100.0	100.0	-
CWC27	105.4	99.3	96.4	250410
CYP1B1	97.5	100.0	100.0	231300
CYP4V2	163.8	99.9	98.4	210370
DCN	162.6	95.7	95.6	610048
DDHD1	164.0	97.9	95.9	-
DHDDS	93.2	99.0	95.0	613861
DHX38	120.5	100.0	99.3	618220
DKC1	109.3	99.8	98.7	305000
DNM1L	139.1	99.9	98.5	610708
DNMBP	163.8	100.0	99.7	618415
DRAM2	158.5	100.0	99.9	616502
DTNBP1	126.9	99.8	98.7	614076
EFEMP1	163.8	100.0	99.9	126600
ELOVL1	94.1	99.8	97.7	618527
ELOVL4	122.6	100.0	99.5	600110
EMC1	133.8	100.0	99.2	616875

EPG5	137.9	99.5	98.5	242840
EPHA2	159.1	100.0	99.5	116600
ERCC2	135.9	100.0	99.7	601675
EXOSC2	117.8	100.0	100.0	617763
EYA1	145.8	99.9	99.7	602588;113650
EYS	158.8	99.7	98.1	602772
FA2H	96.6	92.1	83.2	612319
FAM161A	167.3	100.0	99.7	606068
FBN1	169.7	100.0	99.9	154700
FDXR	126.7	100.0	99.3	617717
FLVCR1	153.3	100.0	98.9	609033
FOXC1	55.5	98.0	89.6	602482;601631
FOXE3	46.2	82.8	72.1	612968;610256
FREM1	135.1	99.9	99.1	248450
FRMD7	116.5	99.9	99.1	310700
FTL	144.3	98.5	89.5	600886
FYCO1	124.9	100.0	99.9	610019
FZD4	188.8	100.0	100.0	133780
GALK1	142.8	100.0	99.2	230200
GALM	106.1	100.0	99.9	-
GALT	168.0	100.0	99.7	230400
GCNT2	180.3	99.5	99.5	116700
GDF3	139.0	100.0	100.0	613704;613703
GDF6	117.3	100.0	99.9	613094;615360
GFER	91.7	99.6	93.9	613076
GJA1	188.0	100.0	100.0	257850;164200
GJA3	148.2	100.0	99.7	601885
GJA8	174.0	100.0	100.0	116200
GNAT1	162.3	100.0	100.0	616389;610444
GNAT2	119.8	99.9	99.0	613856

GNB3	154.4	100.0	100.0	617024
GNPTG	156.7	99.2	94.4	252605
GPR143	65.8	85.9	76.5	300814;300500
GPR179	158.2	100.0	100.0	614515
GRHL2	139.7	100.0	100.0	618031
GRK1	160.1	100.0	100.0	613411
GRM6	138.3	90.2	80.6	257270
GSN	117.5	95.8	93.5	105120
GUCA1A	177.5	100.0	100.0	602093
GUCA1B	139.6	100.0	100.0	613827
GUCY2D	100.3	99.6	96.2	204000;601777
HARS1	152.2	100.0	100.0	614504
HCCS	111.3	99.8	97.6	309801
HGSNAT	125.5	86.4	86.3	252930;616544
HK1	140.5	100.0	100.0	617460
HMX1	22.5	62.5	42.8	612109
HPS1	123.4	100.0	100.0	203300
HPS3	165.7	99.8	97.6	614072
HPS4	142.4	100.0	100.0	614073
HPS5	142.4	100.0	99.7	614074
HPS6	172.8	97.2	89.1	614075
HRAS	178.4	100.0	100.0	218040
HSF4	131.7	99.7	97.2	116800
HSPG2	122.6	99.2	97.7	255800
IDH3A	159.6	99.4	97.3	619007
IDH3B	150.4	95.4	95.4	612572
IFT140	124.8	99.8	98.8	266920;617781
IFT172	107.8	99.9	99.1	615630;616394
IFT27	135.7	100.0	100.0	615996
IFT43	134.3	100.0	100.0	617871

IFT52	148.5	100.0	99.9	617102
IFT74	95.1	98.4	93.8	617119
IFT81	108.1	93.4	90.0	-
IMPDH1	57.6	88.0	80.2	180105;613837
IMPG1	110.5	99.7	98.4	616151
IMPG2	150.2	99.8	98.4	613581;616152
INPP5E	107.9	97.1	92.8	610156;213300
INVS	163.7	100.0	100.0	-
IQCB1	113.4	93.9	84.9	609254
IRX1	135.3	87.5	81.3	-
ITPR1	152.6	100.0	99.9	206700
JAG1	147.7	97.7	96.9	118450
JAM3	150.0	100.0	99.9	613730
KCNJ13	155.1	100.0	100.0	614186;193230
KCNV2	111.7	100.0	99.9	610356
KERA	204.1	100.0	100.0	217300
KIAA0586	143.6	97.3	93.0	616546
KIAA1549	133.8	97.9	96.4	618613
KIF11	105.0	97.6	94.8	152950
KIF21A	147.5	99.9	99.3	135700
KIF3B	114.9	100.0	99.7	618955
KIF7	100.4	93.6	90.6	200990
KIZ	170.0	100.0	99.2	615780
KLHL7	137.3	99.9	99.8	612943
KRT12	166.9	99.7	97.9	122100
KRT3	129.4	100.0	100.0	122100
LAMA1	142.2	100.0	99.7	615960
LAMB2	172.2	100.0	99.7	609049
LAMP2	109.5	99.2	95.7	300257
LCA5	162.7	99.9	99.2	604537

LEMD2	89.9	98.6	92.1	212500
LIM2	120.7	100.0	100.0	615277
LRAT	284.3	100.0	100.0	613341
LRIT3	116.2	93.9	91.9	615058
LRMDA	143.5	96.8	95.6	615179
LRP2	171.0	100.0	99.9	222448
LRP5	180.4	98.5	98.1	259770;601813
LRPAP1	121.7	100.0	100.0	615431
LSS	138.9	100.0	99.9	616509
LTBP2	113.2	99.9	99.0	613086
LYST	169.3	99.6	98.2	214500
LZTFL1	131.8	99.9	99.2	615994
MAB21L2	181.9	100.0	100.0	615877
MAF	79.4	83.5	78.0	610202
MAK	174.4	98.7	96.8	614181
MAPKAPK3	97.3	100.0	99.6	617111
MERTK	159.7	99.5	98.8	613862
MFN2	138.4	100.0	99.9	601152;609260
MFRP	139.7	100.0	100.0	611040;609549
MFSD8	143.1	100.0	99.7	610951;616170
MIP	114.5	100.0	98.9	615274
MIR184				614303
MITF	160.8	100.0	99.9	103470
MKKS	232.7	100.0	100.0	209900
MKS1	104.9	99.8	97.8	615990;249000;617121
MVK	121.1	91.0	90.5	260920;610377
MYO7A	137.5	99.3	97.4	276900
MYOC	162.2	100.0	98.6	137750
NAA10	117.7	99.7	98.5	309800
NBAS	169.2	100.0	99.6	614800

NDP	89.7	100.0	99.7	305390;310600
NDUFS2	118.6	100.0	100.0	252010
NEK2	96.6	99.7	95.5	615565
NEUROD1	153.2	100.0	99.0	-
NHS	129.1	95.4	93.9	302350;302200
NMNAT1	131.8	100.0	99.2	608553
NPHP1	151.9	100.0	99.0	266900;609583
NPHP3	143.2	99.7	98.4	267010
NPHP4	138.2	100.0	99.8	606996
NR2E3	104.1	100.0	99.6	268100;611131
NR2F1	183.1	100.0	100.0	615722
NRL	93.3	99.5	94.9	613750
NYX	96.7	96.4	94.1	310500
OAT	87.0	85.3	76.2	258870
OCA2	128.8	99.9	98.7	203200
OCRL	128.1	100.0	98.6	309000
OFD1	61.6	88.0	73.8	300804;300424
OPA1	155.0	99.6	97.5	125250;165500
OPA3	125.1	100.0	99.0	165300;258501
OPN1LW	77.9	67.5	61.0	303700
OPN1MW	68.7	66.3	58.6	303700
OPTN	120.0	100.0	99.9	137760
OTX2	152.7	100.0	99.7	610125
OVOL2	111.4	95.8	89.6	122000
P3H2	111.0	99.8	98.0	614292
P4HA2	138.6	100.0	99.3	617238
PANK2	185.7	100.0	99.3	234200;607236
PAX2	202.2	100.0	99.9	120330
PAX6	141.6	100.0	100.0	136520;120200;604229;120430;106210
PCARE	124.7	99.5	98.5	613428

PCDH15	167.9	97.8	96.7	602083;601067
PCYT1A	113.1	98.9	95.5	608940
PDE6A	120.5	100.0	99.6	613810
PDE6B	156.3	100.0	99.9	613801;163500
PDE6C	136.6	99.9	97.8	613093
PDE6D	128.7	100.0	100.0	615665
PDE6G	158.4	100.0	100.0	613582
PDE6H	73.7	100.0	97.9	610024
PDZD7	86.9	97.0	93.0	605472
PET100	107.3	100.0	99.6	220110
PEX1	155.3	99.9	99.4	601539;214100
PEX2	158.8	100.0	100.0	614866;614867
PEX26	94.2	100.0	100.0	614872
PEX6	113.1	94.6	86.8	614862
PEX7	135.8	87.9	80.7	215100;614879
PGK1	51.1	92.9	79.5	300653
PHOX2A	37.1	91.8	72.9	602078
PHYH	89.2	100.0	99.6	266500
PIKFYVE	164.2	99.9	99.4	121850
PITX2	158.9	99.9	97.9	137600
PITX3	59.2	100.0	98.4	107250;610623
PLA2G5	145.1	100.0	100.0	228980
PLK4	177.8	99.9	98.1	616171
PNPLA6	140.1	100.0	99.7	215470;275400;245800
POC1B	96.6	99.8	98.9	615973
POC5	168.6	99.7	97.6	-
POMGNT1	131.8	100.0	99.9	617123
PPT1	164.7	90.3	90.3	256730
PRCD	125.3	100.0	100.0	610599
PRDM13	133.5	99.2	94.1	136550

PRDM5	162.4	99.9	99.2	614170
PRIMPOL	138.1	97.5	94.6	615421
PROM1	133.4	97.2	96.1	608051;612657;603786;612095
PRPF3	90.4	98.8	95.3	601414
PRPF31	133.8	100.0	98.8	600138
PRPF4	148.3	100.0	99.8	615922
PRPF6	123.6	100.0	99.9	613983
PRPF8	122.7	100.0	99.3	600059
PRPH2	243.3	100.0	100.0	608161;136880;169150;608133;613105
PRSS56	77.4	99.9	96.6	613517
PXDN	150.9	100.0	99.6	269400
RAB28	80.0	99.6	96.0	615374
RAB3GAP2	108.0	99.5	97.0	614225
RARB	109.7	100.0	100.0	615524
RAX	96.3	96.1	87.1	611038
RAX2	76.7	100.0	92.8	610381
RBP3	150.7	100.0	100.0	615233
RBP4	140.9	99.9	97.7	615147;616428
RCBTB1	120.0	99.9	99.5	617175
RD3	148.1	100.0	100.0	610612
RDH11	107.9	100.0	99.0	616108
RDH12	102.2	100.0	98.6	612712
RDH5	163.4	100.0	100.0	136880
REEP6	188.7	100.0	100.0	617304
RGS9	121.2	98.5	97.1	608415
RGS9BP	94.9	99.6	95.2	608415
RHO	173.1	100.0	100.0	610445;613731
RIMS1	138.2	99.8	97.7	603649
RIMS2	148.9	96.7	95.3	618970
RLBP1	133.7	100.0	99.9	607476;607475;136880

ROM1	115.9	100.0	99.9	608133
RP1	123.8	91.5	90.6	180100
RP1L1	135.6	100.0	100.0	613587;618826
RP2	179.1	100.0	99.8	312600
RP9	67.9	80.8	75.9	180104
RPE65	153.2	99.8	97.8	204100;613794;618697
RPGR	75.8	76.6	72.0	304020;300455;300834;300029
RPGRIP1	146.4	100.0	99.9	613826;608194
RPGRIP1L	155.4	96.7	95.6	216360;611560;611561
RS1	61.0	99.9	94.2	312700
RTN4IP1	96.9	99.9	98.6	616732
SAG	150.2	100.0	100.0	613758;258100
SAMD11	95.3	90.3	81.7	-
SC5D	192.0	100.0	99.5	607330
SCAPER	177.7	99.7	98.2	618195
SCO2	107.8	100.0	100.0	608908
SDCCAG8	137.8	100.0	99.9	613615
SEMA4A	135.1	100.0	99.8	610282;610283
SGSH	149.5	94.5	94.1	252900
SHH	129.3	100.0	99.6	611638
SIPA1L3	172.4	99.9	98.9	616851
SIX6	264.3	100.0	100.0	212550
SLC16A12	152.3	100.0	99.9	612018
SLC24A1	169.4	100.0	99.9	613830
SLC24A5	113.7	99.9	99.1	113750
SLC25A46	199.4	99.7	97.4	616505
SLC33A1	156.0	99.9	98.9	614482
SLC38A8	84.6	99.9	97.3	609218
SLC39A5	145.8	99.9	99.1	615946
SLC45A2	137.8	100.0	99.9	606574

SLC4A11	174.6	100.0	99.9	217700;613268;217400
SLC52A2	171.3	100.0	100.0	614707
SLC7A14	157.8	100.0	100.0	615725
SMOC1	132.3	99.9	98.4	206920
SNRNP200	139.2	99.9	99.1	610359
SOX2	208.1	100.0	100.0	206900
SOX5	113.1	99.9	98.9	616803
SPATA7	138.3	99.8	98.1	604232
SPP2	123.7	100.0	99.9	-
SSBP1	68.2	99.8	97.6	165510
STRA6	130.9	100.0	99.8	601186
TACSTD2	215.4	99.0	96.5	204870
TCF4	132.6	100.0	99.8	613267
TCTN1	114.8	96.8	93.0	614173
TCTN3	136.5	100.0	100.0	614815
TDRD7	162.3	99.9	99.1	613887
TEAD1	157.6	100.0	99.9	108985
TEK	191.9	100.0	100.0	617272
TENM3	175.5	100.0	99.6	615145
TGFBI	135.0	99.5	94.6	122200;121900;602082;608470;607541;608471;121820
TIMM8A	63.1	98.0	90.6	304700;311150
TIMP3	145.4	100.0	100.0	136900
TMCO3	151.2	100.0	99.4	-
TMEM126A	125.1	96.2	84.4	612989
TMEM138	95.0	100.0	99.1	614465
TMEM216	114.5	99.9	98.2	608091
TMEM231	106.7	100.0	99.6	615397;614970
TMEM237	141.6	100.0	99.9	614424
TMEM67	96.7	99.4	94.9	607361;610688;216360
TMEM98	141.3	99.3	98.0	615972

TOPORS	202.2	100.0	100.0	609923
TPP1	146.6	100.0	100.0	204500
TRAF3IP1	91.2	99.5	97.5	616629
TREX1	249.3	100.0	100.0	192315
TRIM32	131.5	100.0	100.0	615988
TRNT1	108.2	99.5	96.4	616959
TRPM1	151.2	100.0	99.8	613216
TSPAN12	163.7	100.0	99.8	613310
TTC8	135.0	99.6	98.0	613464;615985
TTLL5	167.0	100.0	99.6	615860
TUB	112.0	99.4	97.2	616188
TUBA3D	115.1	100.0	99.2	617928
TUBB3	114.1	98.2	96.9	600638
TUBB4B	81.3	99.9	97.0	617879
TUBGCP4	123.5	99.2	96.5	616335
TUBGCP6	158.3	100.0	99.3	251270
TULP1	134.6	100.0	99.5	600132;613843
TYR	164.6	100.0	100.0	203100;103470;606952
TYRP1	183.6	100.0	99.8	203290
UBIAD1	250.0	99.5	96.0	121800
UNC45B	126.3	99.3	98.0	616279
USH1C	109.8	100.0	99.8	276904
USH1G	161.7	99.6	97.9	606943
USH2A	156.8	100.0	99.8	613809;276901
USP45	120.1	99.5	98.1	618513
VAX1	97.5	97.5	91.5	614402
VCAN	180.5	100.0	100.0	143200
VIM	122.0	99.3	97.0	116300
VPS13B	163.9	99.5	98.2	216550
VSX1	69.0	84.7	80.5	148300

VSX2	100.7	100.0	99.4	610092;610093
WDPCP	122.2	98.2	94.3	615992
WDR19	153.0	100.0	99.3	616307;614378;614376
WDR36	151.0	99.7	97.6	609887
WFS1	193.8	100.0	99.9	222300;116400;614296
WHRN	127.1	99.8	98.0	611383
WRN	147.6	99.9	98.8	277700
YAP1	106.6	96.4	89.4	120433
YME1L1	133.4	99.0	95.2	617302
ZEB1	170.4	100.0	99.4	609141;613270
ZNF408	147.4	100.0	100.0	616469;616468
ZNF423	196.5	100.0	100.0	614844
ZNF469	155.2	100.0	100.0	229200
ZNF513	142.9	100.0	100.0	613617
ZNF644	178.3	100.0	100.0	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.

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