

WES VISION DISORDERS DG 2.17

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
ABCA4	109.9	99.9	99.1	248200;601718;153800;604116
ABCC6	116.6	93.7	93.1	264800;177850
ABHD12	96.9	100.0	99.5	612674
ACBD5	144.7	99.8	98.1	-
ACO2	125.5	95.6	90.3	616289
ADAM9	140.7	99.9	98.1	612775
ADAMTS18	134.6	100.0	99.8	615458
ADAMTSL4	137.7	100.0	99.8	225200
ADGRV1	126.0	99.8	98.3	605472
ADIPOR1	91.6	99.5	95.6	-
AGBL1	109.9	98.5	98.3	615523
AGBL5	111.4	100.0	99.4	617023
AGK	109.6	99.6	95.5	614691
AHI1	125.5	99.9	97.6	608629
AHR	181.2	100.0	99.5	618345
AIPL1	124.2	100.0	100.0	604393
ALDH1A3	110.3	100.0	98.4	615113
ALMS1	178.0	100.0	99.8	203800
AP3B1	108.2	99.4	95.7	608233
AP3D1	135.1	98.5	97.9	617050
APOPT1	80.4	82.1	82.1	220110
ARHGEF18	153.5	99.9	98.7	617433
ARL13B	98.7	100.0	99.7	612291
ARL2BP	66.5	92.9	83.8	615434
ARL3	76.1	99.9	97.6	618173

ARL6	91.8	99.9	97.7	613575;209900
ARR3	87.8	99.9	99.6	301010
ARSG	120.4	100.0	98.7	618144
ASPH	109.3	100.0	98.9	601552
ASRGL1	129.9	100.0	100.0	-
ATF6	125.0	100.0	99.4	616517
ATOH7	210.8	99.8	99.2	221900
B3GLCT	97.3	100.0	99.7	261540
BBIP1	116.1	98.5	91.4	615995
BBS1	156.1	100.0	100.0	209900
BBS10	156.7	100.0	100.0	615987
BBS12	193.6	100.0	100.0	615989
BBS2	153.3	100.0	99.7	616562;615981
BBS4	113.2	100.0	98.4	615982
BBS5	94.9	98.4	92.3	615983
BBS7	136.8	99.0	95.3	615984
BBS9	113.2	98.8	94.8	615986
BCOR	109.0	99.2	96.2	300166
BEST1	137.3	99.9	98.2	611809;613194;153700;608161;193220
BFSP1	105.8	100.0	99.3	611391
BFSP2	106.9	100.0	99.2	611597
BLOC1S3	79.6	100.0	100.0	614077
BLOC1S6	103.0	99.3	92.1	614171
BMP4	192.0	100.0	100.0	607932
C12orf65	112.4	100.0	99.8	613559;615035
C19orf12	117.5	100.0	99.9	614298
C1QTNF5	173.0	97.8	91.6	605670
C21orf2	146.9	100.0	99.4	617547
C2orf71	128.5	100.0	99.3	613428
C5orf42	122.3	99.7	97.4	614615

C8orf37	146.4	100.0	99.9	614500;617406
CA4	175.5	100.0	100.0	600852
CABP4	168.9	100.0	100.0	610427
CACNA1F	91.4	99.8	97.7	300600;300476;300071
CACNA2D4	103.4	99.2	97.4	610478
CAPN5	167.1	100.0	100.0	193235
CC2D2A	112.6	99.0	97.0	612285;216360;612284
CCT2	145.5	100.0	99.9	-
CDH23	186.7	100.0	100.0	601067
CDH3	148.2	100.0	99.9	601553;225280
CDHR1	153.8	99.9	99.2	613660
CDK10	141.9	100.0	100.0	-
CEP164	96.7	99.9	98.4	614845
CEP250	108.0	99.9	99.2	618358
CEP290	77.6	96.9	88.7	610188;611134;610189;615991;611755
CEP41	79.1	98.7	94.4	614464
CEP78	123.1	99.8	96.9	617236
CEP83	103.3	99.8	96.2	615862
CERKL	115.2	99.5	96.8	608380
CFH	148.8	99.4	97.4	126700
CHD7	143.6	100.0	99.5	214800
CHM	98.7	98.3	91.4	303100
CHMP4B	150.6	100.0	98.5	605387
CHRD1	89.5	100.0	99.1	309300
CHST6	322.9	100.0	100.0	217800
CIB2	218.3	100.0	99.5	614869
CISD2	116.7	83.4	83.4	604928
CLCC1	100.3	99.9	98.5	-
CLN3	123.4	92.5	92.2	204200
CLN5	139.4	100.0	99.5	256731

CLN6	141.7	100.0	100.0	601780
CLN8	156.2	83.5	83.5	600143
CLRN1	140.6	100.0	99.5	614180;276902
CLUAP1	138.4	100.0	99.7	204000
CNGA1	110.6	93.0	86.9	613756
CNGA3	160.9	100.0	99.8	216900
CNGB1	116.9	99.7	98.3	613767
CNGB3	101.6	98.4	93.7	248200;262300
CNNM4	177.3	100.0	99.9	217080
COL11A1	94.6	98.0	93.6	604841;154780
COL18A1	153.2	99.7	97.9	267750
COL25A1	134.2	99.3	98.7	616219
COL2A1	121.1	100.0	99.8	108300
COL8A2	140.0	100.0	100.0	136800;609140
COL9A1	132.9	100.0	99.5	614134
COL9A2	104.9	100.0	99.6	614284
CRB1	158.9	100.0	100.0	600105;613835;172870
CRX	216.9	100.0	100.0	120970;613829
CRYAA	145.4	96.7	91.1	604219
CRYAB	96.3	99.9	97.9	613763
CRYBA1	108.4	100.0	99.8	600881
CRYBA2	183.6	100.0	100.0	115900
CRYBA4	126.6	100.0	100.0	610425
CRYBB1	132.7	100.0	99.9	611544
CRYBB2	148.3	100.0	100.0	601547
CRYBB3	149.6	100.0	100.0	609741
CRYGB	111.5	100.0	98.3	615188
CRYGC	138.2	100.0	99.7	604307
CRYGD	112.7	100.0	99.7	115700
CRYGS	84.2	94.4	85.4	116100

CSPP1	117.4	100.0	99.4	615636
CTDP1	141.7	96.2	88.2	604168
CTNNA1	113.7	99.4	97.7	608970
CTNNB1	129.5	100.0	100.0	617272
CTSD	187.3	100.0	99.0	610127
CWC27	82.5	99.8	97.3	250410
CYP1B1	153.6	100.0	100.0	231300;604229
CYP4V2	140.6	99.8	98.4	210370
DCN	131.5	95.7	95.3	610048
DDHD1	166.9	100.0	99.1	-
DHDDS	84.5	97.3	94.0	613861
DHX38	112.7	100.0	99.5	618220
DKC1	93.9	99.7	98.0	305000
DNM1L	120.8	99.9	97.7	610708
DRAM2	127.1	100.0	100.0	616502
DTNBP1	118.8	99.8	97.5	614076
EFEMP1	152.0	100.0	99.5	126600
ELOVL1	90.9	99.7	96.6	-
ELOVL4	103.3	100.0	99.6	600110
EMC1	111.0	100.0	99.2	616875
EPG5	111.4	99.5	98.3	242840
EPHA2	173.3	100.0	99.9	116600
EXOSC2	114.1	100.0	99.9	617763
EYA1	121.6	100.0	99.9	113650;602588
EYS	130.0	99.8	97.3	602772
FA2H	101.5	99.3	95.1	612319
FAM161A	142.0	99.9	99.3	606068
FLVCR1	154.8	100.0	99.4	609033
FOXC1	96.1	99.9	99.2	602482
FOXE3	111.7	93.8	87.3	107250

FREM1	112.5	99.8	98.8	248450
FRMD7	101.2	99.9	97.9	310700
FTL	164.3	100.0	98.4	600886
FYCO1	135.2	100.0	100.0	610019
FZD4	192.1	100.0	100.0	133780
GALK1	186.1	100.0	99.9	230200
GALT	165.3	100.0	100.0	230400
GCNT2	158.5	99.5	99.5	116700
GDF3	132.5	100.0	100.0	613703;613704
GDF6	180.6	100.0	100.0	615360;613094
GFER	103.0	100.0	99.9	613076
GJA1	162.4	100.0	100.0	257850;164200
GJA3	185.2	100.0	100.0	601885
GJA8	173.2	100.0	100.0	116200
GNAT1	197.4	100.0	100.0	610444;-
GNAT2	106.3	100.0	98.5	613856
GNB3	159.3	100.0	100.0	617024
GNPTG	199.0	99.9	99.4	252605
GPR143	60.8	91.8	81.6	300500;300814
GPR179	163.2	100.0	100.0	614515
GRHL2	119.8	100.0	100.0	618031
GRK1	149.0	100.0	100.0	613411
GRM6	152.8	99.2	94.6	257270
GSN	123.5	95.6	93.8	105120
GUCA1A	189.2	100.0	100.0	602093
GUCA1B	135.9	100.0	100.0	613827
GUCY2D	120.6	100.0	100.0	601777;204000
HARS	142.4	100.0	100.0	614504
HCCS	90.4	99.6	96.6	309801
HGSNAT	99.9	88.2	86.3	252930;616544

HK1	123.7	100.0	99.6	617460
HMX1	47.2	89.3	70.6	612109
HPS1	125.7	100.0	100.0	203300
HPS3	133.9	99.9	98.2	614072
HPS4	135.2	100.0	100.0	614073
HPS5	122.5	99.9	98.9	614074
HPS6	183.5	100.0	99.2	614075
HRAS	196.0	100.0	100.0	218040
HSF4	166.2	99.9	99.3	116800
HSPG2	132.7	99.5	99.2	255800
IDH3B	136.5	95.8	95.4	612572
IFT140	127.6	100.0	99.6	266920;617781
IFT172	98.4	100.0	99.5	616394;615630
IFT27	122.9	100.0	99.9	615996
IFT43	119.5	100.0	100.0	617871
IFT74	81.5	99.4	93.7	617119
IFT81	90.5	93.0	88.0	-
IMPDH1	57.4	97.6	87.3	613837;180105
IMPG1	92.5	99.8	98.2	616151
IMPG2	127.1	99.5	98.0	616152;613581
INPP5E	131.1	100.0	99.3	213300;610156
INVS	147.7	100.0	100.0	-
IQCB1	90.6	91.0	79.0	609254
IRX1	175.4	99.1	93.2	-
ITPR1	136.4	100.0	99.8	206700
JAG1	143.4	99.4	97.6	118450
JAM3	132.0	100.0	100.0	613730
KCNJ13	142.5	100.0	100.0	614186;193230
KCNV2	154.7	100.0	100.0	610356
KERA	174.8	100.0	100.0	217300

KIAA1549	125.5	98.7	97.6	-
KIF11	89.7	97.6	94.9	152950
KIF21A	118.6	99.9	98.8	135700
KIF7	120.4	99.3	96.6	200990
KIZ	151.3	99.8	97.7	615780
KLHL7	116.8	100.0	99.9	612943
KRT12	142.5	99.8	97.9	122100
KRT3	125.0	100.0	99.9	122100
LAMA1	119.9	100.0	99.6	615960
LAMB2	182.1	100.0	99.7	609049
LCA5	137.1	99.9	98.9	604537
LEMD2	117.7	100.0	99.5	212500
LIM2	122.5	100.0	99.9	615277
LRAT	252.8	100.0	100.0	613341
LRIT3	111.2	94.4	93.5	615058
LRMDA	119.8	99.4	98.0	615179
LRP2	140.5	100.0	99.9	222448
LRP5	183.1	99.9	99.4	601813;259770
LRPAP1	153.8	100.0	99.6	615431
LSS	138.5	100.0	99.9	616509
LTBP2	124.2	100.0	99.7	613086
LYST	135.6	99.3	97.1	214500
LZTFL1	116.5	99.9	99.2	615994
MAB21L2	265.2	100.0	100.0	615877
MAF	103.8	89.3	84.7	610202
MAK	134.1	99.2	96.5	614181
MAPKAPK3	94.7	100.0	99.6	617111
MERTK	133.5	99.5	99.0	613862
MFN2	130.9	100.0	99.9	601152;609260
MFRP	140.5	100.0	100.0	611040;609549

MFSD8	117.4	100.0	99.6	610951;616170
MIP	126.1	99.9	97.4	615274
MIR184				614303
MITF	145.6	100.0	100.0	103470
MKKS	161.5	83.2	83.2	209900
MKS1	98.8	99.9	98.5	249000;617121;615990
MVK	130.3	90.5	90.4	610377;260920
MYO7A	134.7	99.9	99.1	276900
MYOC	166.0	100.0	99.1	137750
NAA10	112.8	100.0	99.4	309800
NBAS	138.4	99.9	99.2	614800
NDP	96.2	100.0	99.6	310600;305390
NDUFS2	102.9	100.0	99.9	252010
NEK2	86.4	98.8	92.9	615565
NEUROD1	172.7	100.0	99.9	-
NHS	114.3	98.6	96.5	302350;302200
NMNAT1	118.9	99.9	98.3	608553
NPHP1	119.7	99.8	97.8	266900;609583
NPHP3	121.6	99.7	98.3	267010
NPHP4	134.2	100.0	99.8	606996
NR2E3	117.8	99.9	98.9	611131;268100
NR2F1	261.9	100.0	100.0	615722
NRL	129.8	100.0	99.0	613750
NYX	155.4	99.9	99.1	310500
OAT	69.1	80.2	69.8	258870
OCA2	123.6	99.8	97.4	203200
OCRL	106.2	99.9	98.6	309000
OFD1	52.3	85.5	70.0	300424;300804
OPA1	121.4	99.7	97.5	165500;125250
OPA3	171.9	100.0	99.9	258501;165300

OPN1LW	61.2	67.9	61.0	303700
OPN1MW	61.9	68.7	60.9	303700
OTX2	135.4	100.0	99.6	610125
OVOL2	130.8	99.9	98.7	122000
P3H2	102.6	100.0	99.1	614292
P4HA2	119.3	99.9	98.7	617238
PANK2	161.5	100.0	100.0	234200;607236
PAX2	198.0	100.0	100.0	120330
PAX6	122.8	100.0	99.9	106210;120430;120200;136520
PCDH15	139.4	99.2	98.9	602083;601067
PCYT1A	97.1	99.1	95.7	608940
PDE6A	105.5	100.0	99.7	613810
PDE6B	171.1	100.0	100.0	613801;163500
PDE6C	116.7	99.4	97.2	613093
PDE6D	121.7	100.0	100.0	615665
PDE6G	134.0	100.0	99.7	613582
PDE6H	60.3	96.8	76.1	610024
PDZD7	103.9	99.7	98.4	605472
PET100	95.2	99.7	90.6	220110
PEX1	126.3	100.0	99.1	214100;601539
PEX2	137.4	100.0	100.0	614867;614866
PEX26	105.1	100.0	100.0	614872
PEX7	108.8	91.3	91.0	614879;215100
PGK1	47.0	92.1	78.7	300653
PHOX2A	63.1	99.8	94.3	602078
PHYH	75.9	100.0	97.9	266500
PIKFYVE	136.7	99.9	99.3	121850
PITX2	186.2	100.0	99.6	604229
PITX3	103.0	100.0	99.8	610623
PLA2G5	111.2	100.0	100.0	228980

PLK4	145.8	99.9	98.1	616171
PNPLA6	153.1	100.0	99.6	275400;215470;245800
POC1B	77.8	99.7	97.8	615973
POC5	132.3	99.3	96.0	-
POMGNT1	123.6	100.0	99.8	617123
PPT1	140.2	90.3	89.2	256730
PRCD	107.5	100.0	100.0	610599
PRDM13	207.6	100.0	99.6	136550
PRDM5	136.5	99.9	98.2	614170
PRIMPOL	117.5	97.4	93.8	615421
PROM1	106.6	97.6	95.2	608051;612095;612657;603786
PRPF3	74.9	98.9	95.5	601414
PRPF31	127.5	99.8	97.9	600138
PRPF4	128.6	100.0	99.8	615922
PRPF6	120.0	100.0	99.8	613983
PRPF8	109.8	100.0	99.3	600059
PRPH2	217.4	100.0	100.0	608133;169150;136880;608161;613105
PRSS56	111.2	99.9	99.3	613517
PXDN	150.2	100.0	99.8	269400
RAB28	64.3	99.1	91.0	615374
RAB3GAP2	89.9	99.7	96.1	614225
RARB	93.5	100.0	100.0	615524
RAX	156.7	100.0	99.9	611038
RAX2	98.8	100.0	100.0	610381
RBP3	168.5	100.0	100.0	615233
RBP4	149.6	99.8	96.8	615147;616428
RCBTB1	98.4	99.9	99.0	617175
RD3	190.9	100.0	100.0	610612
RDH11	94.8	99.9	98.8	616108
RDH12	86.4	99.8	98.1	612712

RDH5	182.9	100.0	100.0	136880
REEP6	226.6	100.0	99.9	617304
RGS9	119.4	98.9	96.9	608415
RGS9BP	170.1	100.0	100.0	608415
RHO	180.4	100.0	100.0	613731;610445
RIMS1	129.8	99.6	98.1	603649
RLBP1	129.0	100.0	99.9	607476;607475;136880
ROM1	139.5	100.0	99.9	608133
RP1	112.2	91.5	91.0	180100
RP1L1	153.1	100.0	100.0	613587
RP2	159.7	100.0	99.2	312600
RP9	64.9	95.2	81.1	180104
RPE65	133.0	100.0	99.8	204100;613794
RPGR	80.2	83.1	74.6	300455;304020;300834;300029
RPGRIP1	132.7	100.0	99.9	613826;608194
RPGRIP1L	124.2	96.8	95.8	611561;611560;216360
RS1	54.6	99.2	89.0	312700
RTN4IP1	80.2	99.6	98.2	616732
SAG	131.3	100.0	99.9	613758;258100
SAMD11	110.0	96.5	90.5	-
SC5D	149.4	100.0	99.6	607330
SCAPER	137.7	97.8	96.0	618195
SCO2	134.9	100.0	100.0	608908
SDCCAG8	123.5	100.0	99.7	613615
SEMA4A	133.6	100.0	99.5	610283;610282
SGSH	152.5	98.1	94.9	252900
SHH	165.7	100.0	100.0	611638
SIPA1L3	192.1	100.0	99.8	616851
SIX6	303.9	100.0	100.0	212550
SLC16A12	134.4	100.0	99.9	612018

SLC24A1	175.4	100.0	100.0	613830
SLC24A5	102.2	100.0	99.6	113750
SLC25A46	175.2	99.8	97.2	616505
SLC33A1	135.7	99.8	97.0	614482
SLC38A8	77.1	99.3	95.7	609218
SLC39A5	143.6	100.0	99.7	615946
SLC45A2	119.4	100.0	99.8	606574
SLC4A11	173.6	100.0	100.0	217700;217400;613268
SLC52A2	213.2	100.0	100.0	614707
SLC7A14	155.0	100.0	100.0	615725
SMOC1	121.2	99.9	98.4	206920
SNRNP200	124.8	99.9	98.7	610359
SOX2	261.8	100.0	100.0	206900
SOX5	92.0	99.8	96.6	616803
SPATA7	120.5	99.7	97.1	604232
SPP2	113.1	100.0	100.0	-
STRA6	125.5	100.0	99.9	601186
TACSTD2	314.2	100.0	100.0	204870
TCTN1	96.0	95.7	92.6	614173
TCTN3	121.0	100.0	100.0	614815
TDRD7	139.3	99.9	99.0	613887
TEAD1	136.1	100.0	99.7	108985
TENM3	155.3	99.8	99.4	615145
TGFBI	118.3	99.9	98.8	121820;602082;122200;608471;607541;608470;121900
TIMM8A	50.3	95.4	80.0	304700;311150
TIMP3	148.2	100.0	100.0	136900
TMCO3	126.0	100.0	99.6	-
TMEM126A	100.7	95.6	79.5	612989
TMEM138	87.8	100.0	99.0	614465
TMEM216	92.0	99.9	96.9	608091

TMEM231	112.1	100.0	99.7	615397;614970
TMEM237	114.5	99.9	98.8	614424
TMEM67	80.6	99.3	93.5	607361;216360;610688
TOPORS	181.9	100.0	100.0	609923
TPP1	130.2	100.0	100.0	204500
TRAF3IP1	87.5	99.1	96.7	616629
TREX1	261.9	100.0	100.0	192315
TRIM32	132.8	100.0	100.0	615988
TRNT1	100.7	99.2	95.3	616959
TRPM1	134.7	100.0	99.3	613216
TSPAN12	132.9	100.0	99.8	613310
TTC8	116.8	99.7	97.8	615860;613464;615985
TTLL5	138.8	99.9	98.9	615860
TUB	112.3	100.0	99.4	616188
TUBA3D	109.2	100.0	97.4	617928
TUBB3	135.6	99.9	99.1	600638
TUBB4B	96.6	100.0	100.0	617879
TUBGCP4	108.4	97.8	95.1	616335
TULP1	128.8	100.0	99.7	600132;613843
TYR	153.5	100.0	100.0	103470;203100;606952
TYRP1	155.1	100.0	99.9	203290
UBIAD1	202.0	99.9	98.2	121800
UNC119	126.4	100.0	99.9	-
UNC45B	121.5	100.0	99.3	616279
USH1C	99.1	100.0	99.3	276904
USH1G	221.0	99.9	99.3	606943
USH2A	130.8	100.0	99.8	613809;276901
USP45	93.8	99.7	98.1	-
VAX1	108.2	100.0	99.4	614402
VCAN	155.3	100.0	100.0	143200

VIM	138.3	99.7	98.0	116300
VPS13B	135.9	99.4	97.8	216550
VSX1	73.0	99.0	92.6	148300
VSX2	134.2	100.0	100.0	610092;610093
WDPCP	105.7	97.1	93.6	615992
WDR19	125.3	100.0	99.4	614376;614378;616307
WFS1	210.0	100.0	99.8	614296;116400;222300
WHRN	145.9	100.0	99.3	611383
WRN	120.3	100.0	98.7	277700
YAP1	97.9	98.5	94.0	120433
YME1L1	102.3	98.1	92.4	617302
ZEB1	155.3	100.0	99.8	613270;609141
ZNF408	162.5	100.0	100.0	616468;616469
ZNF423	215.2	100.0	100.0	614844
ZNF469	180.5	100.0	100.0	229200
ZNF513	153.8	100.0	100.0	613617
ZNF644	153.0	100.0	99.9	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