

WES VISION DISORDERS DG 2.15

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
ABCA4	127.1	100.0	99.5	248200;601718;153800;604116
ABCC6	116.4	93.6	92.6	264800;177850
ABHD12	107.0	97.3	88.0	612674
ACBD5	145.3	97.8	96.0	-
ACO2	129.3	95.8	91.8	616289
ADAM9	146.3	98.6	94.1	612775
ADAMTS18	147.5	99.9	98.9	615458
ADAMTSL4	90.6	99.9	98.8	225200
ADGRV1	140.3	99.5	97.0	605472
ADIPOR1	111.5	100.0	98.8	-
AGBL1	132.3	98.5	98.4	615523
AGBL5	111.0	100.0	99.8	617023
AGK	112.1	99.3	96.4	614691
AHI1	139.3	99.2	95.1	608629
AIPL1	116.0	100.0	99.5	604393
ALDH1A3	104.7	93.4	89.6	615113
ALMS1	179.8	99.9	99.7	203800
AP3B1	95.0	97.8	90.2	608233
AP3D1	121.0	98.1	97.8	617050
APOPT1	63.8	81.4	78.1	220110
ARHGEF18	114.0	97.7	93.0	617433
ARL13B	97.3	98.9	92.8	612291
ARL2BP	66.3	88.3	79.3	615434
ARL3	85.0	98.9	93.3	618173
ARL6	85.2	99.8	95.3	613575;209900

ARR3	98.1	100.0	99.9	301010
ARSG	141.1	100.0	99.4	618144
ASPH	117.3	98.8	93.9	601552
ASRGL1	131.0	100.0	99.6	-
ATF6	134.1	100.0	99.6	616517
ATOH7	102.8	95.8	89.6	221900
B3GLCT	101.2	97.4	93.4	261540
BBIP1	132.0	99.4	94.8	615995
BBS1	148.9	100.0	100.0	209900
BBS10	172.6	100.0	100.0	615987
BBS12	208.6	100.0	100.0	615989
BBS2	181.8	100.0	99.8	616562;615981
BBS4	135.9	99.7	97.3	615982
BBS5	81.1	95.8	84.1	615983
BBS7	120.7	98.1	91.7	615984
BBS9	112.9	96.0	93.8	615986
BCOR	109.7	99.3	96.8	300166
BEST1	144.6	99.6	97.5	611809;613194;153700;608161;193220
BFSP1	98.0	98.2	88.9	611391
BFSP2	89.5	99.8	97.6	611597
BLOC1S3	28.7	88.7	65.3	614077
BLOC1S6	97.2	98.7	91.3	614171
BMP4	151.7	100.0	99.9	607932
C12orf65	88.2	97.3	91.9	613559;615035
C19orf12	93.9	100.0	99.7	614298
C1QTNF5	151.2	79.7	65.5	605670
C21orf2	104.4	99.9	98.7	617547
C2orf71	124.6	99.7	98.8	613428
C5orf42	122.8	98.6	95.5	614615
C8orf37	126.4	100.0	99.0	614500;617406

CA4	142.4	100.0	99.9	600852
CABP4	98.4	99.7	97.7	610427
CACNA1F	99.4	99.8	97.9	300600;300476;300071
CACNA2D4	112.0	99.2	97.7	610478
CAPN5	166.1	100.0	99.9	193235
CC2D2A	127.4	99.5	97.1	612285;216360;612284
CCT2	150.4	99.9	98.7	-
CDH23	197.2	100.0	100.0	601067
CDH3	159.3	99.5	97.3	601553;225280
CDHR1	154.2	99.2	98.0	613660
CDK10	114.6	100.0	99.9	-
CEP164	94.2	99.9	98.0	614845
CEP250	105.0	99.9	98.8	-
CEP290	66.1	88.4	76.7	610188;611134;610189;615991;611755
CEP41	83.5	97.7	89.6	614464
CEP78	112.1	97.0	93.9	617236
CEP83	96.7	98.3	89.1	615862
CERKL	100.4	98.6	92.8	608380
CFH	183.2	98.7	95.3	126700
CHD7	150.7	99.9	98.9	214800
CHM	102.6	96.5	87.3	303100
CHMP4B	139.3	99.9	99.0	605387
CHRD1	109.7	100.0	99.4	309300
CHST6	334.0	100.0	100.0	217800
CIB2	229.9	99.9	99.6	614869
CISD2	127.9	83.4	83.4	604928
CLN3	114.9	92.5	90.7	204200
CLN5	146.1	98.2	92.2	256731
CLN6	131.6	98.9	95.3	601780
CLN8	163.9	83.5	83.5	600143

CLRN1	157.2	100.0	99.8	614180;276902
CLUAP1	152.6	99.9	99.5	204000
CNGA1	127.2	89.4	84.6	613756
CNGA3	167.7	100.0	99.9	216900
CNGB1	102.5	98.4	94.8	613767
CNGB3	101.4	97.7	93.0	248200;262300
CNNM4	190.5	98.7	97.8	217080
COL11A1	90.8	94.9	89.6	604841;154780
COL18A1	88.7	93.9	87.7	267750
COL25A1	126.8	98.4	95.8	616219
COL2A1	103.4	99.9	99.0	108300
COL8A2	37.5	84.6	69.4	136800;609140
COL9A1	121.2	99.5	96.9	614134
COL9A2	65.1	98.3	88.8	614284
CRB1	191.8	100.0	100.0	600105;613835;172870
CRX	114.2	99.9	98.7	120970;613829
CRYAA	135.3	92.7	86.2	604219
CRYAB	125.7	99.9	98.7	613763
CRYBA1	135.7	100.0	99.2	600881
CRYBA2	150.6	100.0	100.0	115900
CRYBA4	117.5	100.0	100.0	610425
CRYBB1	129.1	100.0	99.4	611544
CRYBB2	150.3	100.0	100.0	601547
CRYBB3	144.3	100.0	100.0	609741
CRYGB	97.4	99.8	97.4	615188
CRYGC	128.5	100.0	99.3	604307
CRYGD	100.4	100.0	99.8	115700
CRYGS	105.4	96.4	88.0	116100
CSPP1	112.0	99.8	97.8	615636
CTDP1	105.0	86.6	83.6	604168

CTNNA1	125.6	99.9	99.0	608970
CTNNB1	163.8	100.0	99.9	617272
CTSD	163.7	98.0	95.3	610127
CWC27	74.7	97.2	89.7	250410
CYP1B1	134.8	100.0	100.0	231300;604229
CYP4V2	147.5	99.8	98.5	210370
DCN	140.0	95.7	94.9	610048
DDHD1	141.8	97.1	94.8	-
DHDDS	93.5	97.8	94.8	613861
DHX38	130.3	99.9	99.0	618220
DKC1	111.9	99.6	98.1	305000
DNM1L	123.5	99.7	96.6	610708
DRAM2	131.8	100.0	99.7	616502
DTNBP1	115.2	99.3	95.1	614076
EFEMP1	167.9	100.0	99.6	126600
ELOVL4	91.9	99.9	98.0	600110
EPG5	126.0	99.3	97.7	242840
EPHA2	175.0	99.2	97.8	116600
EXOSC2	142.0	100.0	100.0	617763
EYA1	144.2	100.0	99.7	113650;602588
EYS	135.8	98.9	94.9	602772
FA2H	94.1	87.9	79.9	612319
FAM161A	115.2	98.5	95.0	606068
FLVCR1	139.5	99.2	95.8	609033
FOXC1	32.7	86.0	68.5	602482
FOXE3	20.6	69.0	47.8	107250
FREM1	138.4	99.9	99.1	248450
FRMD7	114.2	99.9	98.8	310700
FTL	147.7	99.0	93.2	600886
FYCO1	123.7	100.0	100.0	610019

FZD4	224.2	100.0	99.8	133780
GALK1	125.4	100.0	99.7	230200
GALT	168.7	100.0	100.0	230400
GCNT2	166.8	99.5	99.5	116700
GDF3	134.9	100.0	100.0	613703;613704
GDF6	75.2	98.7	89.0	615360;613094
GFER	76.1	92.9	75.4	613076
GJA1	246.4	100.0	100.0	257850;164200
GJA3	164.7	100.0	99.5	601885
GJA8	153.6	100.0	100.0	116200
GNAT1	153.9	100.0	100.0	610444;-
GNAT2	130.7	99.9	99.1	613856
GNB3	179.0	100.0	100.0	617024
GNPTG	151.6	96.1	89.7	252605
GPR143	61.5	85.3	75.5	300500;300814
GPR179	133.6	100.0	99.7	614515
GRHL2	134.6	100.0	100.0	618031
GRK1	126.4	100.0	99.9	613411
GRM6	151.7	93.3	86.7	257270
GSN	119.2	94.2	89.0	105120
GUCA1A	160.6	100.0	100.0	602093
GUCA1B	144.2	100.0	99.9	613827
GUCY2D	91.3	98.3	91.1	601777;204000
HARS	159.4	100.0	100.0	614504
HCCS	106.6	99.9	99.2	309801
HGSNAT	101.0	86.4	85.7	252930;616544
HK1	143.4	100.0	99.9	617460
HMX1	23.0	56.8	42.0	612109
HPS1	117.8	100.0	99.3	203300
HPS3	135.2	99.6	96.4	614072

HPS4	141.9	100.0	100.0	614073
HPS5	133.0	99.9	98.7	614074
HPS6	139.1	91.0	84.3	614075
HRAS	164.7	99.8	98.1	218040
HSF4	103.0	97.6	94.9	116800
HSPG2	121.3	99.4	98.2	255800
IDH3B	165.5	95.9	95.4	612572
IFT140	114.7	99.9	99.0	266920;617781
IFT172	116.5	100.0	99.6	616394;615630
IFT27	131.7	100.0	99.6	615996
IFT43	114.8	100.0	100.0	617871
IFT74	80.1	97.8	88.2	617119
IFT81	92.9	88.3	81.2	-
IMPDH1	61.3	87.8	83.5	613837;180105
IMPG1	103.4	99.6	97.3	616151
IMPG2	154.3	99.5	97.8	616152;613581
INPP5E	89.1	95.8	90.0	213300;610156
INVS	160.5	100.0	100.0	-
IQCB1	92.2	89.3	75.4	609254
IRX1	105.0	83.9	81.1	-
JAG1	148.4	98.1	97.5	118450
JAM3	158.6	100.0	100.0	613730
KCNJ13	210.4	100.0	99.9	614186;193230
KCNV2	137.7	100.0	100.0	610356
KERA	191.6	100.0	100.0	217300
KIAA1549	148.6	97.1	96.0	-
KIF11	83.8	97.2	94.2	152950
KIF21A	123.5	99.4	96.1	135700
KIF7	85.7	93.5	88.9	200990
KIZ	174.2	98.3	96.0	615780

KLHL7	123.5	100.0	99.5	612943
KRT12	126.1	98.6	95.4	122100
KRT3	105.1	100.0	99.7	122100
LAMA1	137.5	100.0	99.6	615960
LAMB2	201.7	100.0	99.8	609049
LCA5	127.8	97.3	95.7	604537
LEMD2	68.6	89.1	80.6	212500
LIM2	103.2	100.0	98.6	615277
LRAT	298.3	100.0	100.0	613341
LRIT3	142.4	94.4	94.1	615058
LRMDA	142.1	97.2	95.6	615179
LRP2	176.3	100.0	99.8	222448
LRP5	189.8	98.2	97.9	601813;259770
LRPAP1	138.3	99.5	97.2	615431
LSS	127.8	100.0	99.1	616509
LTBP2	104.6	99.6	97.1	613086
LYST	134.6	97.8	93.9	214500
LZTFL1	109.1	99.1	95.3	615994
MAB21L2	245.6	100.0	100.0	615877
MAF	60.2	77.5	72.7	610202
MAK	139.1	95.6	94.1	614181
MAPKAPK3	92.8	98.9	96.3	617111
MERTK	161.4	99.4	97.7	613862
MFN2	150.6	100.0	99.9	601152;609260
MFRP	121.3	100.0	100.0	611040;609549
MFSD8	125.1	99.9	98.4	610951;616170
MIP	132.2	99.8	96.1	615274
MIR184				614303
MITF	155.5	100.0	99.9	103470
MKKS	208.5	83.2	83.1	209900

MKS1	114.5	99.9	98.5	249000;617121;615990
MVK	124.3	92.1	90.4	610377;260920
MYO7A	134.1	99.7	98.1	276900
MYOC	172.5	100.0	99.3	137750
NAA10	102.4	98.7	96.7	309800
NBAS	145.3	99.5	97.6	614800
NDP	116.8	100.0	100.0	310600;305390
NDUFS2	117.8	100.0	100.0	252010
NEK2	110.4	98.8	93.3	615565
NEUROD1	166.5	100.0	100.0	-
NHS	127.1	94.3	93.3	302350;302200
NMNAT1	137.9	100.0	99.7	608553
NPHP1	117.6	98.8	96.4	266900;609583
NPHP3	115.6	99.4	96.1	267010
NPHP4	136.7	99.9	99.3	606996
NR2E3	93.4	99.9	98.9	611131;268100
NR2F1	201.6	99.9	98.4	615722
NRL	68.7	99.7	95.8	613750
NYX	94.9	98.1	96.0	310500
OAT	89.2	77.7	70.5	258870
OCA2	139.9	99.5	97.9	203200
OCRL	122.2	98.8	96.3	309000
OFD1	51.5	84.0	67.8	300424;300804
OPA1	122.5	99.1	94.1	165500;125250
OPA3	128.0	99.5	97.4	258501;165300
OPN1LW	76.6	67.4	61.9	303700
OPN1MW	67.2	68.2	60.4	303700
OTX2	154.8	100.0	99.8	610125
OVOL2	119.1	96.9	90.6	122000
P3H2	100.2	99.2	93.4	614292

P4HA2	158.7	100.0	99.5	617238
PANK2	146.6	99.3	93.1	234200;607236
PAX2	168.5	99.9	99.3	120330
PAX6	119.9	100.0	99.9	106210;120430;120200;136520
PCDH15	153.9	99.0	98.0	602083;601067
PCYT1A	113.5	98.3	94.7	608940
PDE6A	125.8	100.0	99.8	613810
PDE6B	147.9	100.0	100.0	613801;163500
PDE6C	137.0	99.0	96.7	613093
PDE6D	106.1	100.0	99.9	615665
PDE6G	95.3	99.5	96.3	613582
PDE6H	68.4	97.6	77.0	610024
PDZD7	80.9	98.4	93.9	605472
PET100	94.5	88.8	74.8	220110
PEX1	115.8	97.7	95.4	214100;601539
PEX2	147.1	100.0	100.0	614867;614866
PEX26	76.4	100.0	99.8	614872
PEX7	113.5	89.6	82.0	614879;215100
PGK1	54.5	93.3	81.6	300653
PHOX2A	29.9	59.9	32.6	602078
PHYH	74.6	97.5	90.8	266500
PIKFYVE	141.6	99.8	98.4	121850
PITX2	147.8	99.7	97.5	604229
PITX3	40.2	95.6	82.8	610623
PLA2G5	125.4	100.0	100.0	228980
PLK4	145.5	99.5	96.3	616171
PNPLA6	122.1	99.7	98.5	275400;215470;245800
POC1B	78.7	98.0	94.2	615973
POC5	123.3	97.5	91.3	-
POMGNT1	127.6	99.7	97.1	617123

PPT1	144.5	90.0	87.3	256730
PRCD	89.4	100.0	99.9	610599
PRDM13	122.6	92.7	87.8	136550
PRDM5	129.4	99.5	95.5	614170
PRIMPOL	110.1	95.7	90.1	615421
PROM1	112.3	95.4	92.8	608051;612095;612657;603786
PRPF3	86.0	98.8	96.0	601414
PRPF31	115.9	97.5	92.0	600138
PRPF4	149.4	100.0	99.4	615922
PRPF6	130.3	100.0	100.0	613983
PRPF8	139.0	99.9	99.0	600059
PRPH2	244.1	100.0	100.0	608133;169150;136880;608161;613105
PRSS56	51.5	96.6	83.8	613517
PXDN	163.8	99.8	98.5	269400
RAB28	52.1	96.5	87.1	615374
RARB	118.7	100.0	100.0	615524
RAX	82.5	88.7	77.3	611038
RAX2	52.2	91.0	67.8	610381
RBP3	155.2	100.0	100.0	615233
RBP4	99.6	99.4	96.1	615147;616428
RCBTB1	123.7	100.0	99.7	617175
RD3	161.6	100.0	99.9	610612
RDH11	119.2	100.0	99.9	616108
RDH12	94.4	100.0	98.4	612712
RDH5	160.2	100.0	99.7	136880
REEP6	171.8	99.4	96.4	617304
RGS9	101.4	98.5	97.2	608415
RGS9BP	93.9	100.0	99.6	608415
RHO	210.1	100.0	100.0	613731;610445
RIMS1	126.6	98.8	96.0	603649

RLBP1	144.8	100.0	100.0	607476;607475;136880
ROM1	115.0	100.0	99.4	608133
RP1	121.8	91.4	90.3	180100
RP1L1	94.8	100.0	99.8	613587
RP2	180.0	100.0	98.9	312600
RP9	62.9	77.8	76.0	180104
RPE65	130.3	100.0	99.3	204100;613794
RPGR	91.8	83.1	73.8	300455;304020;300834;300029
RPGRIP1	154.2	100.0	99.9	613826;608194
RPGRIP1L	126.2	96.4	93.9	611561;611560;216360
RS1	60.0	97.8	88.3	312700
RTN4IP1	98.1	99.9	99.1	616732
SAG	131.3	100.0	99.9	613758;258100
SAMD11	63.2	82.8	75.2	-
SC5D	198.4	100.0	99.2	6607330
SCAPER	135.9	96.0	93.6	618195
SCO2	113.1	100.0	100.0	608908
SDCCAG8	123.9	99.8	97.4	613615
SEMA4A	127.5	99.9	98.9	610283;610282
SGSH	129.0	95.1	93.6	252900
SHH	117.5	99.0	94.0	611638
SIPA1L3	140.8	99.7	98.6	616851
SIX6	228.6	100.0	100.0	212550
SLC16A12	164.4	100.0	99.9	612018
SLC24A1	218.5	100.0	100.0	613830
SLC24A5	114.5	99.6	97.7	113750
SLC25A46	205.7	95.9	87.3	616505
SLC33A1	140.9	96.8	90.1	614482
SLC38A8	76.4	99.4	95.8	609218
SLC39A5	119.3	100.0	99.2	615946

SLC45A2	148.1	100.0	99.9	606574
SLC4A11	153.0	100.0	99.7	217700;217400;613268
SLC52A2	177.6	100.0	100.0	614707
SLC7A14	191.9	100.0	100.0	615725
SMOC1	129.8	99.5	97.3	206920
SNRNP200	161.1	100.0	99.6	610359
SOX2	128.8	98.3	93.1	206900
SPATA7	119.6	97.8	90.8	604232
SPP2	144.1	100.0	100.0	-
STRA6	116.5	100.0	99.9	601186
TACSTD2	223.6	99.1	96.6	204870
TCTN1	98.8	95.7	92.8	614173
TCTN3	127.6	100.0	99.8	614815
TDRD7	168.3	99.0	97.7	613887
TEAD1	158.6	99.8	98.2	108985
TENM3	185.7	99.5	98.7	615145
TGFBI	130.9	99.0	94.5	121820;602082;122200;608471;607541;608470;121900
TIMM8A	46.0	94.5	78.8	304700;311150
TIMP3	147.0	100.0	100.0	136900
TMCO3	131.4	99.9	98.4	-
TMEM126A	120.3	98.4	86.2	612989
TMEM138	100.2	100.0	99.5	614465
TMEM216	111.9	100.0	98.7	608091
TMEM231	111.5	100.0	99.9	615397;614970
TMEM237	100.7	99.8	98.3	614424
TMEM67	72.9	93.3	83.4	607361;216360;610688
TOPORS	210.6	100.0	100.0	609923
TPP1	146.3	100.0	100.0	204500
TRAF3IP1	90.3	96.3	92.8	616629
TREX1	242.4	100.0	100.0	192315

TRIM32	141.2	100.0	100.0	615988
TRNT1	104.6	97.8	92.3	616959
TRPM1	161.0	100.0	99.6	613216
TSPAN12	129.4	100.0	99.5	613310
TTC8	106.9	97.9	92.0	615860;613464;615985
TTLL5	152.7	99.9	98.7	615860
TUB	103.0	97.3	95.2	616188
TUBA3D	144.5	100.0	99.6	617928
TUBB3	136.1	98.1	96.9	600638
TUBB4B	109.8	98.4	96.0	617879
TUBGCP4	130.8	99.1	96.2	616335
TULP1	97.8	96.8	91.7	600132;613843
TYR	185.3	100.0	100.0	103470;203100;606952
TYRP1	176.9	100.0	99.9	203290
UBIAD1	248.8	98.9	95.2	121800
UNC119	92.9	97.8	90.4	-
UNC45B	143.0	100.0	99.5	616279
USH1C	97.5	100.0	99.4	276904
USH1G	195.3	98.4	96.3	606943
USH2A	148.5	100.0	99.7	613809;276901
VAX1	52.2	88.4	78.0	614402
VCAN	186.5	100.0	100.0	143200
VIM	126.5	99.1	97.2	116300
VPS13B	143.8	98.6	96.8	216550
VSX1	52.2	85.5	76.1	148300
VSX2	77.7	99.8	97.3	610092;610093
WDPCP	107.3	93.9	88.9	615992
WDR19	132.1	99.8	98.1	614376;614378;616307
WFS1	251.4	100.0	99.7	614296;116400;222300
WHRN	114.0	99.8	98.8	611383

WRN	123.6	98.3	94.6	277700
YAP1	95.6	87.8	81.6	120433
YME1L1	105.3	97.7	91.9	617302
ZEB1	192.7	100.0	99.3	613270;609141
ZNF408	135.7	100.0	100.0	616468;616469
ZNF423	250.8	100.0	100.0	614844
ZNF469	93.1	98.7	96.3	229200
ZNF513	110.7	100.0	99.7	613617
ZNF644	156.3	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.

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