

WES HEARING IMPAIRMENT (INCLUDING GJB2) DG 3.5

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
ABCC1	117.5	100.0	100.0	-
ABHD12	130.6	100.0	100.0	612674
ACTB	238.4	100.0	100.0	243310;607371
ACTG1	221.9	100.0	100.0	614583;604717
ADAMTS1	143.3	100.0	100.0	No OMIM phenotype
ADCY1	124.3	99.2	98.7	610154
ADGRV1	145.9	100.0	100.0	605472
AIFM1	101.3	100.0	99.9	300614
ALMS1	146.3	100.0	100.0	203800
AMMECR1	100.1	100.0	99.8	300990
ANLN	142.2	100.0	100.0	No OMIM phenotype
AP1B1	115.2	100.0	100.0	242150
ARSG	123.9	100.0	100.0	618144
ATOH1	134.7	100.0	100.0	No OMIM phenotype
ATP11A	131.7	100.0	100.0	619810
ATP1A3	138.8	100.0	100.0	-
ATP2B2	122.4	100.0	100.0	-
ATP6V0A4	123.1	100.0	100.0	602722
ATP6V1B1	130.1	100.0	100.0	267300
ATP6V1B2	149.0	100.0	100.0	124480
BCAP31	73.7	99.1	92.8	300475
BCS1L	133.1	100.0	100.0	262000
BDP1	147.1	100.0	100.0	618257
BMP4	129.1	100.0	100.0	-
BSND	118.0	100.0	100.0	602522

BTD	121.2	94.4	94.3	253260
CABP2	165.9	100.0	100.0	614899
CACNA1D	135.2	100.0	100.0	614896
CCDC50	143.0	100.0	100.0	607453
CD151	130.1	100.0	100.0	609057
CD164	156.1	100.0	100.0	616969
CDC14A	138.0	100.0	100.0	616958
CDC42	156.6	100.0	100.0	616737
CDH23	124.2	100.0	100.0	601067;601386
CEACAM16	118.4	100.0	100.0	614614
CEP250	111.1	100.0	100.0	618358
CEP78	143.0	100.0	100.0	617236
CHD7	133.9	100.0	100.0	214800
CHSY1	139.8	99.9	99.7	605282
CIB2	113.7	100.0	99.9	614869;609439
CISD2	162.2	100.0	100.0	604928
CLDN14	127.0	100.0	100.0	614035
CLDN9	151.0	100.0	100.0	-
CLIC5	137.4	100.0	100.0	616042
CLPP	112.2	100.0	100.0	614129
CLRN1	150.5	100.0	100.0	276902
CLRN2	120.3	100.0	100.0	618988
COA8	146.3	100.0	99.9	220110
COCH	147.3	100.0	100.0	601369
COL11A1	152.4	100.0	100.0	154780;604841
COL11A2	121.0	100.0	100.0	184840;609706;277610;601868;215150
COL2A1	126.1	100.0	100.0	215150;108300;132450;156550
COL4A3	132.2	100.0	100.0	203780;104200
COL4A4	129.6	100.0	100.0	203780
COL4A5	106.8	99.3	98.7	301050

COL4A6	94.7	99.4	98.7	300914
COL9A1	143.6	100.0	100.0	614134
COL9A2	130.7	100.0	100.0	614284
COL9A3	141.1	100.0	100.0	-
CRYL1	136.8	100.0	100.0	-
CRYM	123.3	100.0	100.0	616357
DCAF17	152.7	100.0	100.0	241080
DCDC2	140.0	100.0	100.0	610212
DIABLO	148.8	100.0	100.0	614152
DIAPH1	135.6	100.0	100.0	124900
DIAPH3	147.4	100.0	99.8	609129
DLL1	135.3	100.0	100.0	-
DLX5	129.1	100.0	100.0	183600;220600
DMXL2	149.3	100.0	100.0	617605
DSPP	249.2	100.0	100.0	605594
EDN3	164.5	100.0	100.0	613265
EDNRB	148.0	100.0	100.0	277580;600501
EFNB2	138.5	100.0	100.0	-
EHD1	144.4	100.0	100.0	No OMIM phenotype
ELMOD3	124.6	100.0	100.0	615429
ELOVL1	125.5	100.0	100.0	618527
EPS8	144.2	100.0	100.0	615974
EPS8L2	128.1	100.0	100.0	617637
ERAL1	120.6	100.0	100.0	617565
ESPN	166.1	100.0	100.0	609006
ESRP1	132.8	100.0	100.0	618013
ESRRB	137.8	100.0	100.0	608565
EXOSC2	120.4	100.0	100.0	617763
EYA1	145.7	100.0	100.0	602588;166780;113650
EYA4	149.3	100.0	100.0	605362;601316

FDXR	130.0	100.0	100.0	617717
FGF3	124.1	100.0	100.0	610706
FGFR3	140.9	100.0	100.0	602849
FITM2	133.6	100.0	100.0	618635
FOXF2	127.1	99.9	99.3	-
FOXI1	158.6	100.0	100.0	600791
GAB1	146.4	100.0	100.0	605428
GAS2	154.0	100.0	100.0	-
GATA3	147.3	100.0	100.0	146255
GIPC3	125.9	100.0	100.0	601869
GJB2	149.1	100.0	100.0	602540;149200;148350;601544;220290;148210;124500
GJB3	142.5	100.0	100.0	220290;612644
GJB6	137.1	100.0	100.0	612645;220290;612643
GLA	95.1	90.9	90.9	301500
GPRASP2	114.2	100.0	100.0	No OMIM phenotype
GPSM2	145.5	100.0	100.0	604213
GRAP	178.9	100.0	100.0	618456
GREB1L	126.2	100.0	100.0	-
GRHL2	131.5	100.0	100.0	608641
GRXCR1	133.6	99.9	99.3	613285
GRXCR2	129.1	100.0	100.0	615837
GSDME	126.7	100.0	100.0	600994
HARS1	132.1	100.0	100.0	614504
HARS2	140.0	100.0	100.0	614926
HGF	156.5	100.0	100.0	608265
HOMER2	120.4	100.0	99.9	616707
HSD17B4	142.8	96.6	96.6	233400
IFNLR1	111.3	100.0	100.0	-
ILDR1	134.4	100.0	100.0	609646
KARS1	127.9	100.0	100.0	613916

KCNE1	237.9	100.0	100.0	612347
KCNJ10	131.0	100.0	100.0	612780;600791
KCNQ1	123.7	100.0	100.0	220400
KCNQ4	123.0	100.0	99.7	600101
KITLG	158.1	100.0	99.3	616697
LARS2	127.2	100.0	100.0	615300
LHFPL5	121.6	100.0	100.0	610265
LMX1A	128.7	100.0	100.0	301412
LOXHD1	128.5	100.0	100.0	613079
LOXL3	124.3	100.0	100.0	-
LRP2	137.5	100.0	100.0	222448
LRP5	121.4	100.0	100.0	144750;607634
LRTOMT	106.5	100.0	100.0	611451
MAN2B1	121.4	100.0	100.0	No OMIM phenotype
MARVELD2	144.2	100.0	100.0	610153
MCM2	117.6	100.0	100.0	616968
MET	147.6	100.0	100.0	616705
MGP	152.5	100.0	100.0	245150
MIA3	145.6	100.0	100.0	-
MINAR2	150.9	100.0	100.0	No OMIM phenotype
MIR96				613074
MITF	137.1	99.9	99.7	103470;103500;193510
MPDZ	141.7	99.5	99.1	No OMIM phenotype
MPZL2	129.4	100.0	100.0	618145
MSRB3	147.2	100.0	100.0	613718
MVD	132.6	100.0	100.0	No OMIM phenotype
MYH14	127.8	100.0	100.0	614369;600652
MYH9	127.0	100.0	100.0	153640;600208;603622;153650
MYO15A	123.0	100.0	100.0	600316
MYO3A	151.3	100.0	100.0	607101

MYO6	154.1	100.0	100.0	606346;607821
MYO7A	129.2	100.0	100.0	600060;276900;601317
NARS2	139.9	100.0	100.0	618434
NCOA3	139.4	100.0	100.0	No OMIM phenotype
NDP	98.7	100.0	100.0	310600
NLRP3	124.2	100.0	100.0	191900
NOG	118.4	100.0	100.0	-
OPA1	148.0	100.0	100.0	125250
OSBPL2	130.2	100.0	100.0	616340
OTOA	119.4	100.0	100.0	607039
OTOF	121.6	100.0	100.0	601071
OTOG	126.8	100.0	100.0	614945
OTOGL	150.7	100.0	100.0	614944
P2RX2	130.5	100.0	100.0	608224
PAX3	127.0	100.0	99.8	148820;193500;122880
PCDH15	153.3	100.0	100.0	602083;601067;609533
PDE1C	131.7	99.4	98.9	618140
PDZD7	125.6	100.0	99.2	605472
PET100	154.9	100.0	100.0	220110
PEX1	145.2	100.0	100.0	601539;234580;214100
PEX26	126.8	100.0	100.0	614873
PEX6	133.9	100.0	100.0	616617;614863
PI4KB	118.0	100.0	100.0	No OMIM phenotype
PISD	127.8	100.0	100.0	618889
PJVK	139.9	100.0	100.0	610220
PLOD3	117.6	100.0	100.0	612394
PLS1	140.1	100.0	99.9	618787
PNPT1	150.5	100.0	100.0	614934
POLD1	133.4	100.0	100.0	615381
POLR1C	117.4	83.3	83.2	248390

POLR1D	139.2	100.0	100.0	613717
POU3F4	112.9	100.0	100.0	304400
POU4F3	133.2	100.0	100.0	602459
PPIP5K2	150.0	100.0	100.0	618422
PRKCB	132.5	100.0	99.9	-
PRORP	136.6	100.0	100.0	No OMIM phenotype
PRPS1	111.3	100.0	100.0	304500;301835;311070;300661
PSIP1	154.0	100.0	100.0	-
PTPRQ	142.9	91.9	91.9	613391
PTRH2	156.6	100.0	100.0	No OMIM phenotype
RAI1	122.6	100.0	100.0	182290
RDX	153.6	100.0	100.0	611022
REST	143.3	98.2	98.2	612431
RIPOR2	126.5	100.0	100.0	616515
RMND1	154.8	100.0	100.0	614922
ROBO1	140.9	100.0	99.9	-
ROR1	130.4	100.0	100.0	617654
RRM2B	144.5	100.0	100.0	No OMIM phenotype
S1PR2	124.2	100.0	100.0	610419
SCD5	132.8	100.0	100.0	-
SERAC1	147.5	100.0	100.0	614739
SERPIN6	134.1	100.0	100.0	613453
SEZ6	130.4	100.0	100.0	No OMIM phenotype
SIX1	122.9	100.0	100.0	605192;608389
SIX5	121.0	100.0	100.0	610896
SLC12A1	144.8	96.3	96.2	-
SLC12A2	158.4	100.0	100.0	619081;619083;619080
SLC17A8	140.9	100.0	100.0	605583
SLC19A2	146.0	100.0	100.0	249270
SLC22A4	137.6	100.0	100.0	-

SLC26A4	140.2	100.0	100.0	600791;274600
SLC26A5	139.2	100.0	100.0	613865
SLC29A3	116.6	100.0	100.0	602782
SLC33A1	133.1	100.0	100.0	614482
SLC44A4	121.6	100.0	100.0	617606
SLC4A11	135.4	100.0	100.0	217400
SLC52A2	172.5	100.0	100.0	614707
SLC52A3	133.0	100.0	100.0	211530
SLC7A14	130.1	100.0	100.0	-
SLC9A3R1	114.6	100.0	100.0	-
SLITRK6	167.7	100.0	100.0	221200
SMPX	110.2	100.0	99.1	300066
SNAI2	150.3	100.0	100.0	172800;608890
SOX10	142.9	100.0	100.0	613266;609136;611584
SPATA5	141.1	100.0	100.0	616577
SPNS2	127.7	100.0	99.7	618457
STRC	131.2	100.0	100.0	603720
SUCLA2	148.6	100.0	99.6	612073
SYNE4	111.3	100.0	100.0	615540
TBC1D24	137.9	100.0	100.0	220500;616044;614617
TBL1X	109.2	100.0	99.9	301033
TBL1Y	35.7	50.0	49.4	No OMIM phenotype
TCOF1	124.5	100.0	100.0	154500
TECTA	122.5	100.0	100.0	601543;603629
TFAP2A	131.0	100.0	100.0	113620
THOC1	159.8	100.0	100.0	No OMIM phenotype
TIMM8A	80.5	100.0	99.5	304700
TJP2	144.2	100.0	100.0	613558
TMC1	143.2	100.0	100.0	606705;600974
TMEM132E	131.6	100.0	100.0	618481

TMEM43	123.8	100.0	100.0	619832
TMIE	147.3	100.0	100.0	600971
TMPRSS3	126.0	100.0	100.0	601072
TMTC2	135.8	97.2	97.1	-
TNC	120.2	100.0	100.0	615629
TPRN	122.8	97.1	95.4	613307
TRIOBP	137.9	100.0	100.0	609823
TRRAP	126.6	100.0	100.0	618778
TSHZ1	115.8	100.0	100.0	607842
TUBB4B	237.0	100.0	100.0	617879
TWNK	117.5	100.0	100.0	616138
TYR	145.5	100.0	99.9	103470
USH1C	120.2	100.0	100.0	276904;602092
USH1G	117.8	100.0	100.0	606943
USH2A	144.6	99.9	99.6	613809;276901
USP48	145.2	100.0	100.0	No OMIM phenotype
WBP2	111.1	100.0	100.0	617639
WFS1	129.4	100.0	100.0	222300;600965;614296
WHRN	140.7	100.0	100.0	611383;607084
XYLT2	118.1	99.9	99.2	605822
YAP1	149.2	100.0	100.0	120433

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