HEARING IMPAIRMENT GENE PANEL DG 2.5/2.6

Gene name	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
ACTB	106.7	100%	93%	Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTG1	110	100%	100%	Deafness, autosomal dominant 20/26, 604717 Baraitser-Winter syndrome 2, 614583
ADCY1	135	92%	86%	?Deafness, autosomal recessive 44, 610154
AIFM1	76.8	100%	100%	Combined oxidative phosphorylation deficiency 6,300816 Cowchock syndrome,310490 Deafness,X-linked 5,300614
APOPT1	62.9	87%	82%	Mitochondrial Complex IV Deficiency, 220110
ATP1A2	176.3	100%	100%	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP6V1B1	164.5	100%	100%	Renal tubular acidosis with deafness, 267300
BDP1	119.2	93%	88%	No OMIM phenotype Hearing loss (Girotto (2013) PLoS One 8,e80323)
BSND	131.6	100%	100%	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
CABP2	73.1	97%	91%	Deafness, autosomal recessive 93, 614899
CACNA1D	143.9	100%	99%	Sinoatrial node dysfunction and deafness, 614896
CCDC50	128.4	100%	99%	Deafness, autosomal dominant 44, 607453
CD164	117.4	94%	94%	No OMIM phenotype Hearing impairment,nonsyndromic (Nyegaard (2015) PLoS Genet 11,e1005386)
CDH23	183	100%	99%	Usher syndrome, type 1D, 601067 Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067
CEACAM16	131.5	100%	100%	Deafness, autosomal dominant 4B, 614614
CIB2	188.6	100%	100%	Deafness, autosomal recessive 48, 609439

				Usher syndrome, type IJ, 614869
CLDN14	120.7	100%	99%	Deafness, autosomal recessive 29, 614035
CLIC5	121	100%	100%	?Deafness, autosomal recessive 103, 616042
CLPP	106.7	98%	92%	Perrault syndrome 3, 614129
CLRN1	141.9	100%	100%	Usher syndrome, type 3A, 276902 Retinitis pigmentosa 61, 614180
COCH	197.6	100%	99%	Deafness, autosomal dominant 9, 601369
COL11A1	85.5	92%	83%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis, 228520
COL11A2	11	46%	13%	Stickler syndrome, type III, 184840 Otospondylomegaepiphyseal dysplasia, 215150 Weissenbacher-Zweymuller syndrome, 277610 Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524
COL2A1	93.5	99%	96%	Stickler syndrome, type I, 108300 Kniest dysplasia, 156550 Achondrogenesis, type II or hypochondrogenesis, 200610 SED congenita, 183900 SMED Strudwick type, 184250 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Spondylo
COL4A3	81.2	97%	90%	Alport syndrome, autosomal recessive, 203780 Hematuria, benign familial, 141200 Alport syndrome, autosomal dominant, 104200
COL4A4	78.4	98%	93%	Alport syndrome, autosomal recessive, 203780 Hematuria,familial benign
COL4A5	32.2	76%	50%	Alport syndrome, 301050
COL4A6	49.9	92%	82%	?Deafness,X-linked 6,300914
COL9A1	107.6	99%	95%	Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134

COL9A2	55.5	98%	86%	Epiphyseal dysplasia, multiple, 2, 600204 {Intervertebral disc disease, susceptibility to}, 603932 Stickler syndrome, type V, 614284
CRYM	88	99%	92%	Deafness, autosomal dominant 40
DCDC2	132	100%	100%	?Deafness,autosomal recessive 66,610212 Nephronophthisis 19,616217
DFNA5	102.2	99%	98%	Deafness, autosomal dominant 5, 600994
DFNB31	102.8	100%	97%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
DFNB59	116.4	100%	98%	Deafness, autosomal recessive 59, 610220
DIABLO	244.1	100%	100%	Deafness, autosomal dominant 64, 614152
DIAPH1	114.3	99%	97%	Deafness, autosomal dominant 1, 124900
DIAPH3	77.4	97%	88%	Auditory neuropathy, autosomal dominant, 1, 609129
DSPP	182.5	100%	98%	Dentinogenesis imperfecta, Shields type II, 125490 Deafness, autosomal dominant 36, with dentinogenesis, 605594 Dentinogenesis imperfecta, Shields type III, 125500 Dentin dysplasia, type II, 125420
EDN3	106.9	100%	95%	Waardenburg syndrome, type 4B, 613265 Central hypoventilation syndrome, congenital, 209880 {Hirschsprung disease, susceptibility to}, 613712
EDNRB	124.7	95%	89%	?{Hirschsprung disease, susceptibility to}, 600155 ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580
ELMOD3	138.4	100%	100%	?Deafness, autosomal recessive 88, 615429
EPS8	135.6	98%	91%	?Deafness, autosomal recessive 102, 615974
ESPN	39.8	70%	56%	Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant
ESRRB	107.8	98%	94%	Deafness, autosomal recessive 35, 608565
EYA1	139.1	100%	99%	Branchiootorenal syndrome 1, with or without cataracts, 113650 Anterior segment anomalies with or without cataract, 113650 Branchiootic syndrome 1, 602588 Otofaciocervical syndrome, 166780
EYA4	153.1	99%	97%	Deafness, autosomal dominant 10, 601316 Cardiomyopathy, dilated, 1J, 605362

FAM65B	107.1	100%	99%	?Deafness,autosomal recessive 104,616515
FGF3	49.5	88%	75%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FOXI1	131.3	100%	100%	Enlarged vestibular aqueduct, 600791
GIPC3	89.3	88%	84%	Deafness, autosomal recessive 15, 601869
GJB2	182.1	100%	100%	Bart-Pumphrey syndrome,149200 Deafness,autosomal recessive 1A,220290 Hystrix-like ichthyosis with deafness,602540 Keratitis,ichthyosis-deafness syndrome,148210 Keratoderma,palmoplantar,with deafness,148350 Vohwinkel syndrome,124500
GJB3	300.8	100%	100%	Erythrokeratodermia variabilis et progressiva, 133200 Deafness, autosomal dominant 2B, 612644 Deafness, autosomal recessive Deafness, autosomal dominant, with peripheral neuropathy Deafness, digenic, GJB2/GJB3, 220290
GJB6	191.1	100%	100%	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GPR98	145.8	99%	94%	Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
GPSM2	109.2	99%	94%	Chudley-McCullough syndrome, 604213
GRHL2	127.4	100%	100%	Deafness, autosomal dominant 28, 608641
GRXCR1	189.1	100%	100%	Deafness, autosomal recessive 25, 613285
GRXCR2	101.3	100%	100%	?Deafness, autosomal recessive 101, 615837
HARS	143.6	100%	100%	Usher syndrome type 3B, 614504
HARS2	166.6	100%	100%	Perrault syndrome 2, 614926
HGF	139.5	98%	97%	Deafness, autosomal recessive 39, 608265
HOMER2	123.8	99%	99%	?Deafness,autosomal dominant 68,616707
HSD17B4	100.2	93%	92%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
ILDR1	93.5	100%	99%	Deafness, autosomal recessive 42, 609646

KARS	116.5	100%	98%	Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, autosomal recessive 89, 613916
KCNE1	443.9	100%	100%	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome-5, 613695
KCNJ10	199.6	100%	100%	SESAME syndrome, 612780 Enlarged vestibular aqueduct, digenic, 600791
KCNQ1	96.7	88%	85%	Long QT syndrome-1, 192500 Jervell and Lange-Nielsen syndrome, 220400 Atrial fibrillation, familial, 3, 607554 Short QT syndrome-2, 609621 {Long QT syndrome 1, acquired, susceptibility to}, 192500
KCNQ4	118.3	93%	91%	Deafness, autosomal dominant 2A, 600101
KITLG	76.3	91%	88%	Hyperpigmentation with or without hypopigmentation,145250 [Skin/hair/eye pigmentation 7],611664
LARS2	126.7	100%	100%	Perrault syndrome 4, 615300
LHFPL5	289.3	100%	100%	Deafness, autosomal recessive 67, 610265
LOXHD1	128.4	99%	98%	Deafness, autosomal recessive 77, 613079
LRTOMT	105.8	98%	90%	Deafness, autosomal recessive 63, 611451
MARVELD2	146	93%	92%	Deafness, autosomal recessive 49, 610153
MCM2	155.7	100%	100%	No OMIM phenotype Hearing loss,nonsyndromic,autosomal dominant (Gao (2015) PLoS One 10)
MIR96				Deafness,autosomal dominant 50,613074
MITF	134.3	100%	100%	Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 Tietz albinism-deafness syndrome, 103500 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456
MSRB3	147.3	96%	96%	Deafness, autosomal recessive 74, 613718
MYH14	92.2	95%	84%	Deafness, autosomal dominant 4A, 600652 Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369
MYH9	123.8	99%	95%	May-Hegglin anomaly, 155100 Fechtner syndrome, 153640 Sebastian syndrome, 605249 Deafness, autosomal dominant 17, 603622 Epstein syndrome, 153650

				Macrothrombocytopenia and progressive sensorineural deafness, 600208
MYO15A	102.3	94%	90%	Deafness, autosomal recessive 3, 600316
MYO3A	116.8	98%	87%	Deafness, autosomal recessive 30, 607101
MYO6	86.8	96%	86%	Deafness,autosomal dominant 22,606346 Deafness,autosomal dominant 22,with hypertrophic cardiomyopathy,606346 Deafness,autosomal recessive 37,607821
MYO7A	127	98%	95%	Usher syndrome, type 1B, 276900 Deafness,autosomal dominant 11,601317 Deafness,autosomal recessive 2,600060
NARS2	125.8	97%	97%	Combined oxidative phosphorylation deficiency 24,616239 DFNB94, Simon, PLoS Genet. 2015 Mar 25;11
NLRP3	130.4	100%	100%	Cold-induced autoinflammatory syndrome, familial, 120100
OPA1	115.4	98%	89%	Optic atrophy 1, 165500
OSBPL2	137.3	100%	100%	Deafness,autosomal dominant 67,616340
OTOA	112	98%	95%	Deafness, autosomal recessive 22, 607039
OTOF	121.5	100%	99%	Deafness, autosomal recessive 9, 601071
OTOG	124.6	100%	98%	Deafness, autosomal recessive 18B, 614945
OTOGL	123.3	96%	93%	Deafness, autosomal recessive 84B, 614944
P2RX2	110.6	98%	95%	Deafness, autosomal dominant 41, 608224
PAX3	108.8	100%	99%	Waardenburg syndrome, type 1, 193500 Craniofacial-deafness-hand syndrome,122880 Rhabdomyosarcoma 2,alveolar,268220 Waardenburg syndrome,type 3,148820
PCDH15	164.2	99%	98%	Usher syndrome, type 1F, 602083 Deafness,autosomal recessive 23,609533 Usher syndrome, type 1D/F digenic,601067
PDZD7	77.7	100%	89%	{Retinal disease in Usher syndrome type IIA, modifier of}, 276901
PET100	110.5	89%	72%	Mitochondrial complex IV deficiency, 220110
PNPT1	48.8	94%	77%	Combined oxidative phosphorylation deficiency 13, 614932
POU3F4	90.3	100%	100%	Deafness, X-linked 2, 304400

POU4F3	239.8	100%	100%	Deafness, autosomal dominant 15, 602459
PRPS1	113.7	100%	100%	Arts syndrome,301835 Charcot-Marie-Tooth disease,X-linked recessive,5,311070 Deafness,X-linked 1,304500 Gout,PRPS-related,300661 Phosphoribosylpyrophosphate synthetase superactivity,300661
PTPRQ	105	92%	87%	Deafness, autosomal recessive 84A, 613391
RDX	36.8	74%	58%	Deafness, autosomal recessive 24, 611022
S1PR2	217.1	100%	91%	Deafness,autosomal recessive 68,610419
SERPINB6	167.2	100%	100%	Deafness, autosomal recessive 91, 613453
SIX1	100.8	95%	94%	Brachiootic syndrome 3, 608389 Deafness,autosomal dominant 23,605192
SIX5	32.9	82%	61%	Branchiootorenal syndrome 2, 610896
SLC17A8	128.8	100%	97%	Deafness, autosomal dominant 25, 605583
SLC26A4	121.4	99%	97%	Pendred syndrome, 274600 Deafness,autosomal recessive 4,with enlarged vestibular aqueduct,600791
SLC26A5	151.2	100%	99%	Deafness, autosomal recessive 61, 613865
SLC33A1	133.8	98%	89%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts,hearing loss,and neurodegeneration,614482
SLITRK6	232.1	100%	100%	Deafness and myopia, 221200
SMPX	50.2	100%	97%	Deafness, X-linked 4, 300066
SNAI2	125.1	100%	98%	Waardenburg syndrome, type 2D, 608890 Piebaldism,172800
SOX10	65.7	92%	88%	Waardenburg syndrome, type 4C, 613266 PCWH syndrome,609136 Waardenburg syndrome,type 2E,with/without neurologic involvement,611584
STRC	88	99%	95%	Deafness, autosomal recessive 16, 603720
SYNE4	58.9	96%	90%	Deafness, autosomal recessive 76, 615540
TBC1D24	149.2	100%	100%	Deafness,autosomal recessive 86,614617 Deafness,autosomal dominant 65,616044 DOOR syndrome,220500 Epileptic encephalopathy,early infantile,16,615338 Myoclonic epilepsy, infantile, familial, 605021

TECTA	192.7	100%	100%	Deafness, autosomal dominant 8/12, 601543 Deafness,autosomal recessive 21,603629
TIMM8A	17.5	64%	30%	Deafness, X-linked 1, progressive
TJP2	108.5	99%	99%	Cholestasis, progressive familial intrahepatic 4, 615878 Hypercholanemia, familial, 607748
TMC1	136.6	95%	94%	Deafness, autosomal recessive 7, 600974 Deafness,autosomal dominant 36,606705
TMEM132E	108.5	97%	93%	No OMIM phenotype Deafness,autosomal dominant 99 (Li et al. Hum Mutat 2015 36(1) 98-105)
TMIE	100.5	96%	87%	Deafness, autosomal recessive 6, 600971
TMPRSS3	111.4	99%	97%	Deafness, autosomal recessive 8/10, 601072
TNC	166.6	99%	99%	Deafness, autosomal dominant 56, 615629
TPRN	55.4	81%	74%	Deafness, autosomal recessive 79, 613307
TRIOBP	104.5	96%	94%	Deafness, autosomal recessive 28, 609823
TSPEAR	128.2	100%	99%	Deafness, autosomal recessive 98, 614861
TYR	189.7	100%	100%	Albinism,oculocutaneous,type IA,203100 Albinism,oculocutaneous,type IB,606952 Waardenburg syndrome/albinism, digenic,103470 [Skin/hair/eye pigmentation 3],601800
USH1C	99.1	98%	95%	Deafness,autosomal recessive 18A,602092 Usher syndrome,type 1C,276904
USH1G	153.6	95%	93%	Usher syndrome, type 1G, 606943
USH2A	156.8	99%	98%	Usher syndrome, type 2A, 276901
WBP2	93.8	100%	100%	No OMIM phenotype progressive high-frequency hearing loss (Buniello (2016) EMBO Molecular Medicine 8,191-207
WFS1	218.6	98%	97%	?Cataract 41,116400 Deafness,autosomal dominant 6/14/38,600965 Wolfram syndrome,222300 Wolfram-like syndrome,autosomal dominant,614296 {Diabetes mellitus,noninsulin-dependent,association with},125853
YAP1	95.5	85%	79%	Coloboma, ocular, 120433 Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433

Gene symbols used follow HGCN 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 : April 10th, 2016.
This list is accurate for panel versions DG 2.5 and DG 2.6. From DG 2.5 to DG 2.6 no changes were made to the content of the gene panels.

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