HEARING IMPAIRMENT GENE PANEL DG 2.9

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ACTB	118.8	98%	94%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACTG1	129.7	100%	100%	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717
ADCY1	160.9	95%	93%	?Deafness, autosomal recessive 44, 610154
AIFM1	126	100%	99%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614
APOPT1	90.9	87%	86%	Mitochondrial complex IV deficiency, 220110
ATP6V1B1	197.9	100%	100%	Renal tubular acidosis with deafness, 267300
BDP1	150.8	97%	92%	No OMIM phenotype Hearing loss (Girotto (2013) PLoS One 8,e80323)
BSND	180.6	100%	100%	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
CABP2	96.1	98%	92%	Deafness, autosomal recessive 93, 614899
CACNA1D	178.3	100%	99%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CCDC50	150.9	100%	99%	?Deafness, autosomal dominant 44, 607453
CD164	152.7	98%	94%	?Deafness,autosomal dominant 66, 616969
CDC14A	194.4	99%	96%	Deafness,autosomal recessive 105,616958
CDH23	225.8	99%	99%	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067
CEACAM16	160.2	100%	99%	Deafness, autosomal dominant 4B, 614614
CEP78	133.5	99%	96%	Cone-rod dystrophy and hearing loss, 617236
CIB2	235.2	100%	99%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CLDN14	132.4	100%	100%	Deafness, autosomal recessive 29, 614035

CLIC5	130	100%	100%	?Deafness, autosomal recessive 103, 616042
CLPP	140.5	99%	97%	Perrault syndrome 3, 614129
CLRN1	171.9	100%	99%	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
COCH	215.4	100%	99%	Deafness, autosomal dominant 9, 601369
COL11A1	112.4	96%	92%	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932
COL11A2	14.6	59%	23%	Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, 215150 Stickler syndrome, type III, 184840 Weissenbacher-Zweymuller syndrome, 277610
COL2A1	119.3	99%	99%	Achondrogenesis, type II or hypochondrogenesis, 200610 Avascular necrosis of the femoral head, 608805 Czech dysplasia, 609162 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Kniest dysplasia, 156550 Legg-Calve-Perthes disease, 150600 Osteoarthritis with mild chondrodysplasia, 604864 Otospondylomegaepiphyseal dysplasia, 215150 Platyspondylic skeletal dysplasia, Torrance type, 151210 SED congenita, 183900 SMED Strudwick type, 184250 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Spondyloperipheral dysplasia, 271700 Stickler sydrome, type I, nonsyndromic ocular, 609508 Stickler syndrome, type I, 108300 Vitreoretinopathy with phalangeal epiphyseal dysplasia
COL4A3	104.5	98%	95%	Alport syndrome, autosomal dominant, 104200 Alport syndrome, autosomal recessive, 203780 Hematuria, benign familial, 141200

COL4A4	97.9	98%	95%	Alport syndrome, autosomal recessive, 203780 Hematuria, familial benign
COL4A5	61	93%	81%	Alport syndrome, 301050
COL4A6	91	95%	90%	?Deafness, X-linked 6, 300914
COL9A1	143.9	99%	97%	Stickler syndrome, type IV, 614134 /?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	77.1	99%	94%	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284 {Intervertebral disc disease, susceptibility to}, 603932
CRYM	100	99%	97%	Deafness, autosomal dominant 40, 616357
DCDC2	180	100%	99%	Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DFNA5	123.3	99%	99%	Deafness, autosomal dominant 5, 600994
DFNB31	121.4	99%	98%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
DFNB59	168.9	100%	99%	Deafness, autosomal recessive 59, 610220
DIABLO	248.1	100%	99%	Deafness, autosomal dominant 64, 614152
DIAPH1	131.4	99%	98%	Deafness, autosomal dominant 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DIAPH3	91.2	99%	95%	Auditory neuropathy, autosomal dominant, 1, 609129
DSPP	150.9	99%	99%	Deafness, autosomal dominant 39, with dentinogenesis, 605594 Dentin dysplasia, type II, 125420 Dentinogenesis imperfecta, Shields type II, 125490 Dentinogenesis imperfecta, Shields type III, 125500
EDN3	145.1	100%	99%	Central hypoventilation syndrome, congenital, 209880 Waardenburg syndrome, type 4B, 613265 {Hirschsprung disease, susceptibility to, 4}, 613712
EDNRB	151.4	98%	93%	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580 {Hirschsprung disease, susceptibility to, 2}, 600155
ELMOD3	172.1	100%	99%	?Deafness, autosomal recessive 88, 615429
EPS8	149	98%	93%	?Deafness, autosomal recessive 102, 615974
ESPN	47.3	75%	59%	Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant

ESRRB	133	99%	97%	Deafness, autosomal recessive 35, 608565
EYA1	167.2	100%	99%	Anterior segment anomalies with or without cataract, 113650 Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 ?Otofaciocervical syndrome, 166780
EYA4	189.6	100%	99%	Cardiomyopathy, dilated, 1J, 605362 Deafness, autosomal dominant 10, 601316
FAM65B	135.6	100%	99%	?Deafness, autosomal recessive 104, 616515
FGF3	86.6	96%	87%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FOXI1	180.6	100%	100%	Enlarged vestibular aqueduct, 600791
GATA3	205.9	100%	100%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GIPC3	125.6	92%	88%	Deafness, autosomal recessive 15, 601869
GJB2	209.4	100%	100%	Bart-Pumphrey syndrome, 149200 Deafness, autosomal dominant 3A, 601544 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	343.4	100%	100%	Deafness, autosomal dominant 2B, 612644 Deafness, autosomal dominant, with peripheral neuropathy Deafness, autosomal recessive Deafness, digenic, GJB2/GJB3, 220290 Erythrokeratodermia variabilis et progressiva, 133200
GJB6	214	100%	100%	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GPR98	172.4	99%	98%	Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 ?Febrile seizures, familial, 4, 604352
GPSM2	135.2	99%	98%	Chudley-McCullough syndrome, 604213
GRHL2	152.8	100%	100%	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
GRXCR1	232.7	100%	100%	Deafness, autosomal recessive 25, 613285

GRXCR2	138.6	100%	100%	?Deafness, autosomal recessive 101, 615837
HARS	167	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HARS2	197.5	100%	99%	?Perrault syndrome 2, 614926
HGF	173.2	99%	98%	Deafness, autosomal recessive 39, 608265
HOMER2	165.9	99%	99%	?Deafness, autosomal dominant 68, 616707
HSD17B4	125.3	95%	93%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
ILDR1	111.6	100%	99%	Deafness, autosomal recessive 42, 609646
KARS	141.9	100%	99%	Deafness, autosomal recessive 89, 613916 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641
KCNE1	538.9	100%	100%	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695
KCNJ10	219.9	100%	99%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNQ1	127.6	92%	89%	Atrial fibrillation, familial, 3, 607554 Jervell and Lange-Nielsen syndrome, 220400 Long QT syndrome 1, 192500 Short QT syndrome 2, 609621 {Long QT syndrome 1, acquired, susceptibility to}, 192500
KCNQ4	155.5	94%	92%	Deafness, autosomal dominant 2A, 600101
KITLG	95.5	96%	92%	Deafness, congenital, unilateral or asymmetric, 616697 Hyperpigmentation with or without hypopigmentation, 145250 [Skin/hair/eye pigmentation 7, blond/brown hair], 611664
LARS2	157.5	100%	100%	Perrault syndrome 4, 615300 ?Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LHFPL5	306.5	100%	100%	Deafness, autosomal recessive 67, 610265
LOXHD1	146.9	100%	99%	Deafness, autosomal recessive 77, 613079
LRTOMT	147.8	99%	96%	Deafness, autosomal recessive 63, 611451
MARVELD2	184.2	98%	96%	Deafness, autosomal recessive 49, 610153
MCM2	189.5	100%	100%	?Deafness,autosomal dominant 70, 616968
MIR96	NC	NC	NC	Deafness, autosomal dominant 50, 613074

MITF	173.3	100%	100%	Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456
MPZL2	118.8	100%	99%	No OMIM phenotype
MSRB3	160	99%	98%	Deafness, autosomal recessive 74, 613718
MYH14	118.4	98%	93%	Deafness, autosomal dominant 4A, 600652 ?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369
MYH9	152.2	99%	98%	Deafness, autosomal dominant 17, 603622 Epstein syndrome, 153650 Fechtner syndrome, 153640 Macrothrombocytopenia and progressive sensorineural deafness, 600208 May-Hegglin anomaly, 155100 Sebastian syndrome, 605249
MYO15A	136.6	97%	94%	Deafness, autosomal recessive 3, 600316
MYO3A	133.3	98%	94%	Deafness, autosomal recessive 30, 607101
MYO6	108.6	98%	94%	Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 Deafness, autosomal recessive 37, 607821
MYO7A	149.2	99%	98%	Deafness, autosomal dominant 11, 601317 Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900
NARS2	153	97%	97%	Combined oxidative phosphorylation deficiency 24, 616239
NLRP3	162.9	100%	99%	CINCA syndrome, 607115 Familial cold-induced inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900
OPA1	146.6	99%	97%	Behr syndrome,210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type),616896 {Glaucoma, normal tension, susceptibility to}, 606657
OSBPL2	157	100%	99%	Deafness, autosomal dominant 67, 616340
OTOA	134.4	98%	96%	Deafness, autosomal recessive 22, 607039

OTOF	150.8	100%	99%	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071
OTOG	160.5	100%	99%	Deafness, autosomal recessive 18B, 614945
OTOGL	139.5	99%	96%	Deafness, autosomal recessive 84B, 614944
P2RX2	139.1	99%	99%	Deafness, autosomal dominant 41, 608224
PAX3	128.2	100%	99%	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PCDH15	179.5	99%	99%	Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083
PDZD7	113.2	99%	98%	Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 {Retinal disease in Usher syndrome type IIA, modifier of}, 276901
PET100	98.1	98%	86%	Mitochondrial complex IV deficiency, 220110
PNPT1	67.9	95%	87%	Combined oxidative phosphorylation deficiency 13, 614932 Deafness, autosomal recessive 70, 614934
POU3F4	159.7	100%	99%	Deafness, X-linked 2, 304400
POU4F3	295.2	100%	100%	Deafness, autosomal dominant 15, 602459
PRPS1	178.9	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	125.9	93%	91%	Deafness, autosomal recessive 84A, 613391
RDX	49.9	87%	69%	Deafness, autosomal recessive 24, 611022
S1PR2	244.8	98%	95%	Deafness, autosomal recessive 68, 610419
SERPINB6	205.3	100%	99%	?Deafness, autosomal recessive 91, 613453
SIX1	125.4	99%	96%	Brachiootic syndrome 3, 608389 Deafness, autosomal dominant 23, 605192
SIX5	57.7	93%	85%	Branchiootorenal syndrome 2, 610896
SLC17A8	161	100%	100%	Deafness, autosomal dominant 25, 605583
SLC22A4	144.2	99%	99%	{Rheumatoid arthritis, susceptibility to}, 180300

SLC26A4	160.6	100%	99%	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 Pendred syndrome, 274600
SLC26A5	183.7	99%	96%	?Deafness, autosomal recessive 61, 613865
SLC33A1	152.6	98%	94%	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLITRK6	263.8	100%	100%	Deafness and myopia, 221200
SMPX	84.6	100%	98%	Deafness, X-linked 4, 300066
SNAI2	128.7	100%	99%	Piebaldism, 172800 Waardenburg syndrome, type 2D, 608890
SOX10	74.6	98%	95%	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
STRC	111.3	99%	97%	Deafness, autosomal recessive 16, 603720
SYNE4	80.7	99%	95%	Deafness, autosomal recessive 76, 615540
TBC1D24	203.2	100%	99%	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	226.1	100%	100%	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TIMM8A	43.7	88%	71%	Jensen syndrome, 311150 Mohr-Tranebjaerg syndrome, 304700
TJP2	129.6	99%	99%	Cholestasis, progressive familial intrahepatic 4, 615878 Hypercholanemia, familial, 607748
TMC1	148.4	98%	96%	Deafness, autosomal dominant 36, 606705 Deafness, autosomal recessive 7, 600974
TMEM132E	140.4	99%	97%	No OMIM phenotype Deafness,autosomal dominant 99 (Li et al. Hum Mutat 2015 36(1) 98-105)
TMIE	129	99%	94%	Deafness, autosomal recessive 6, 600971
TMPRSS3	126.7	99%	99%	Deafness, autosomal recessive 8/10, 601072
TNC	199.3	100%	99%	Deafness, autosomal dominant 56, 615629

TPRN	82.9	83%	75%	Deafness, autosomal recessive 79, 613307
TRIOBP	150.3	97%	96%	Deafness, autosomal recessive 28, 609823
TSPEAR	153.1	100%	99%	Deafness, autosomal recessive 98, 614861 Ectodermal dysplasia (Peled et al. (2016) PLOS Genetics online)
TYR	205.9	100%	100%	Albinism, oculocutaneous, type IA, 203100 Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800
USH1C	119.4	100%	99%	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
USH1G	185.8	98%	96%	Usher syndrome, type 1G, 606943
USH2A	178.6	100%	99%	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
WBP2	103.5	100%	100%	No OMIM phenotype Deafness, progressive (Buniello (2016) EMBO Molecular Medicine 8,191-207
WFS1	260.1	99%	99%	Deafness, autosomal dominant 6/14/38, 600965 Wolfram syndrome, 222300 Wolfram-like syndrome, autosomal dominant, 614296 ?Cataract 41, 116400 {Diabetes mellitus, noninsulin-dependent, association with}, 125853
YAP1	112.2	90%	84%	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 14th 2017
This list is accurate for panel version DG 2.9

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