HEARING IMPAIRMENT GENE PANEL DG 2.14 (168 genes)

Gene	Median Coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ACTB	129	99.1	94.2	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTG1	149.4	100	100	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717
ADCY1	145	94.7	93.3	?Deafness, autosomal recessive 44, 610154
ADGRV1	140.3	99.5	97	?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
AIFM1	106.2	100	99.7	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614
APOPT1	63.8	81.4	78.1	Mitochondrial complex IV deficiency, 220110
ATP1A3	177.3	100	100	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235
ATP2B2	186.5	100	100	{Deafness, autosomal recessive 12, modifier of}, 601386
ATP6V1B1	176.6	100	100	Renal tubular acidosis with deafness, 267300
BCS1L	182.3	100	100	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BDP1	125.5	95.6	90	No OMIM phenotype ?Hearing loss (Girotto (2013) PLoS One 8,e80323)
BSND	137.1	100	100	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
CABP2	67.1	74.6	65.5	Deafness, autosomal recessive 93, 614899
CACNA1D	149.9	98	97.8	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CCDC50	135.1	99.9	98.9	?Deafness, autosomal dominant 44, 607453

CD164	130.4	98	93.9	?Deafness, autosomal dominant 66, 616969
CDC14A	161.4	98.3	93.5	Deafness, autosomal recessive 35, with or without immotile sperm, 608653
CDH23	197.2	100	100	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067 {Pituitary adenoma 5, multiple types}, 617540
CEACAM16	144.9	100	99.7	Deafness, autosomal dominant 4B, 614614
CEP78	112.1	97	93.9	Cone-rod dystrophy and hearing loss, 617236
CIB2	229.9	99.9	99.6	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CLDN14	130.6	100	99.9	Deafness, autosomal recessive 29, 614035
CLIC5	118.3	100	99.9	?Deafness, autosomal recessive 103, 616042
CLPP	115.4	99.8	96.9	Perrault syndrome 3, 614129
CLRN1	157.2	100	99.8	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
COCH	194.4	99.9	99.6	Deafness, autosomal dominant 9, 601369
COL11A1	90.8	94.9	89.6	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932
COL11A2	92.2	99.9	98.3	Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150
COL2A1	103.4	99.9	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 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, 0
COL4A3	89.6	97.8	95.5	Alport syndrome, autosomal dominant, 104200 Alport syndrome, autosomal recessive, 203780 Hematuria, benign familial, 141200
COL4A4	85	97.6	93.5	Alport syndrome, autosomal recessive, 203780 Hematuria, familial benign, 0
COL4A5	52.3	92.1	77.5	Alport syndrome, 301050
COL4A6	81.2	96	89.4	?Deafness, X-linked 6, 300914
COL9A1	121.2	99.5	96.9	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	65.1	98.3	88.8	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
CRYM	97.4	99.9	98.3	Deafness, autosomal dominant 40, 616357
DCDC2	150.5	99.9	99.6	?Deafness, autosomal recessive 66, 610212 Nephronophthisis 19, 616217 Sclerosing cholangitis, neonatal, 617394
DFNA5	112.9	99.9	99.4	Deafness, autosomal dominant 5, 600994
DFNB59	123.6	100	99.2	Deafness, autosomal recessive 59, 610220
DIABLO	206	100	99.5	Deafness, autosomal dominant 64, 614152
DIAPH1	120.7	99.3	97.8	Deafness, autosomal dominant 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DIAPH3	73.9	97.9	90.1	Auditory neuropathy, autosomal dominant, 1, 609129
DMXL2	181.4	98.6	96	?Deafness, autosomal dominant 71, 617605 ?Polyendocrine-polyneuropathy syndrome, 616113
DSPP	155.7	99.9	99.3	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	134.4	100	99.5	Central hypoventilation syndrome, congenital, 209880 Waardenburg syndrome, type 4B, 613265

				{Hirschsprung disease, susceptibility to, 4}, 613712
EDNRB	131	95.6	90.9	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580 {Hirschsprung disease, susceptibility to, 2}, 600155
ELMOD3	156.2	100	99.7	?Deafness, autosomal recessive 88, 615429
EPS8	122.8	96.9	91.4	?Deafness, autosomal recessive 102, 615974
EPS8L2	115.1	94.4	90.9	Deafness autosomal recessive 106, 617637
ERAL1	181.5	100	100	Perrault syndrome 6, 617565
ESPN	28.6	44.2	35.3	Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant, 0
ESRP1	106.5	99.9	98.5	?Deafness, autosomal recessive 109, 618013
ESRRB	119.7	100	99.2	Deafness, autosomal recessive 35, 608565
EXOSC2	142	100	100	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EYA1	144.2	100	99.7	?Otofaciocervical syndrome, 166780 Anterior segment anomalies with or without cataract, 602588 Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650
EYA4	160.6	100	99.5	?Cardiomyopathy, dilated, 1J, 605362 Deafness, autosomal dominant 10, 601316
FGF3	73.9	92	75.7	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FOXI1	152.5	100	100	Enlarged vestibular aqueduct, 600791
FRMPD4	114.1	99.7	98	Mental retardation, X-linked 104, 300983
GAB1	170	100	99.7	?Deafness, autosomal recessive 26, 605428
GATA3	186.8	100	100	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GIPC3	103.3	91.8	85.5	Deafness, autosomal recessive 15, 601869
GJB2	205.1	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	308.9	100	100	Deafness, autosomal dominant 2B, 612644 Deafness, autosomal dominant, with peripheral neuropathy, 0

				Deafness, autosomal recessive, 0 Deafness, digenic, GJB2/GJB3, 220290 Erythrokeratodermia variabilis et progressiva 1, 133200
GJB6	185.4	100	100	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GPSM2	112.9	99.8	97	Chudley-McCullough syndrome, 604213
GRHL2	134.6	100	100	Corneal dystrophy, posterior polymorphous, 4, 618031 Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
GRXCR1	183.7	100	99.8	Deafness, autosomal recessive 25, 613285
GRXCR2	114.9	100	100	?Deafness, autosomal recessive 101, 615837
HARS	159.4	100	100	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HARS2	169.7	99.9	99.2	?Perrault syndrome 2, 614926
HGF	146.9	99.4	96.9	Deafness, autosomal recessive 39, 608265
HOMER2	133.5	99.5	99.4	?Deafness, autosomal dominant 68, 616707
HSD17B4	95.1	93.9	90.8	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
IFNLR1	90.6	97.6	96.5	No OMIM phenotype
ILDR1	107.4	100	99.9	Deafness, autosomal recessive 42, 609646
KARS	122.6	100	99.3	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, autosomal recessive 89, 613916
KCNE1	462.6	100	100	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695
KCNJ10	213.4	89.3	89.1	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNQ1	114.7	93	90.3	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	135.2	93.7	89.1	Deafness, autosomal dominant 2A, 600101

KITLG	81.8	97	91.6	Deafness, autosomal dominant 69, unilateral or asymmetric, 616697 Hyperpigmentation with or without hypopigmentation, 145250 [Skin/hair/eye pigmentation 7, blond/brown hair], 611664
LARS2	143	100	100	?Hydrops, lactic acidosis, and sideroblastic anemia, 617021 Perrault syndrome 4, 615300
LHFPL5	287.7	100	100	Deafness, autosomal recessive 67, 610265
LMX1A	109.2	100	99.9	No OMIM phenotype
LOXHD1	136.5	100	99.6	Deafness, autosomal recessive 77, 613079
LRP5	189.8	98.2	97.9	Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteopetrosis, autosomal dominant 1, 607634 Osteoporosis-pseudoglioma syndrome, 259770 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 van Buchem disease, type 2, 607636 [Bone mineral density variability 1], 601884 {Osteoporosis}, 166710
LRTOMT	125.9	99.8	96.6	Deafness, autosomal recessive 63, 611451
MARVELD2	159.4	97.5	94	Deafness, autosomal recessive 49, 610153
MCM2	176.8	100	100	?Deafness, autosomal dominant 70, 616968
MET	184.7	100	99.6	?Deafness, autosomal recessive 97, 616705 Hepatocellular carcinoma, childhood type, somatic, 114550 Renal cell carcinoma, papillary, 1, familial and somatic, 605074 {Osteofibrous dysplasia, susceptibility to}, 607278
MIR96				Deafness, autosomal dominant 50, 613074
MITF	155.5	100	99.9	COMMAD syndrome, 617306 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456
MPZL2	96.9	100	100	No OMIM phenotype
MSRB3	155.6	99.7	98.5	Deafness, autosomal recessive 74, 613718
MYH14	102	97.7	91.5	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 Deafness, autosomal dominant 4A, 600652
MYH9	130.5	99.4	98.1	Deafness, autosomal dominant 17, 603622

				Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYO15A	116.5	97.3	94	Deafness, autosomal recessive 3, 600316
MYO3A	119.8	98.5	93	Deafness, autosomal recessive 30, 607101
MYO6	89.7	98.1	92.3	Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic Cardiomyopathy, 606346 Deafness, autosomal recessive 37, 607821
MYO7A	134.1	99.7	98.1	Deafness, autosomal dominant 11, 601317 Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900
NARS2	120	97.4	97.1	Combined oxidative phosphorylation deficiency 24, 616239
NLRP3	150.4	100	100	CINCA syndrome, 607115 Deafness, autosomal dominant 34, with or without inflammation, 617772 Familial cold inflammatory syndrome 1, 120100 Keratoendothelitis fugax hereditaria, 148200 Muckle-Wells syndrome, 191900
OPA1	122.5	99.1	94.1	?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 Behr syndrome, 210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 {Glaucoma, normal tension, susceptibility to}, 606657
OSBPL2	145.8	100	100	Deafness, autosomal dominant 67, 616340
OTOA	117.3	99	96.6	Deafness, autosomal recessive 22, 607039
OTOF	131.2	100	99.7	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071
OTOG	145.1	99.4	98.2	Deafness, autosomal recessive 18B, 614945
OTOGL	113.3	98.4	93.9	Deafness, autosomal recessive 84B, 614944
P2RX2	132.8	99	95.5	Deafness, autosomal dominant 41, 608224
PAX3	118.5	100	100	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PCDH15	153.9	99	98	Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083

PDE1C	124	100	99.7	No OMIM phenotype Hearing loss, non-syndromic (Wang (2018) Hum Genet 137,437)
PDZD7	80.9	98.4	93.9	Deafness, autosomal recessive 57, 618003 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 {Retinal disease in Usher syndrome type IIA, modifier of}, 276901
PET100	94.5	88.8	74.8	Mitochondrial complex IV deficiency, 220110
PEX1	115.8	97.7	95.4	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX6	94.5	90.4	86.1	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PNPT1	53.7	93.3	80.9	Combined oxidative phosphorylation deficiency 13, 614932 Deafness, autosomal recessive 70, 614934
POU3F4	151	100	99.9	Deafness, X-linked 2, 304400
POU4F3	298.1	100	100	Deafness, autosomal dominant 15, 602459
PPIP5K2	74.7	96.6	86.6	No OMIM phenotype Hearing loss (Yousaf (2018) PLoS Genet 14)
PRKCB	164.5	100	99.8	No OMIM phenotype Hearing loss (Martin-Sierra (2016) Hum Mol Genet epub,epub)
PRPS1	149.5	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	104.7	93.3	89.1	Deafness, autosomal dominant 73, 617663 Deafness, autosomal recessive 84A, 613391
RAI1	146.3	100	99.7	Smith-Magenis syndrome, 182290
RDX	43.2	84.7	64.8	Deafness, autosomal recessive 24, 611022
RIPOR2	121.4	100	99.9	?Deafness, autosomal recessive 104, 616515
ROR1	177.5	96.8	96.8	?Deafness, autosomal recessive 108, 617654
S1PR2	278	97.4	92.7	Deafness, autosomal recessive 68, 610419
SERPINB6	164.1	95.9	95.9	?Deafness, autosomal recessive 91, 613453
SIX1	117.3	99.7	97.6	Branchiootic syndrome 3, 608389

				Deafness, autosomal dominant 23, 605192
SIX5	43.8	88.3	76.1	Branchiootorenal syndrome 2, 610896
SLC17A8	132.7	100	100	Deafness, autosomal dominant 25, 605583
SLC22A4	119.1	99.8	98.2	{Rheumatoid arthritis, susceptibility to}, 180300
SLC25A2	219.9	100	100	No OMIM phenotype
SLC26A4	123.3	99.9	99.1	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 Pendred syndrome, 274600
SLC26A5	150.9	98.7	95.8	?Deafness, autosomal recessive 61, 613865
SLC29A3	203.6	99.9	99.5	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC33A1	140.9	96.8	90.1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC44A4	122.9	100	99.9	?Deafness, autosomal dominant 72, 617606
SLITRK6	206.9	100	100	Deafness and myopia, 221200
SMPX	68.5	99.9	95.5	Deafness, X-linked 4, 300066
SNAI2	129.8	100	99.8	Piebaldism, 172800 Waardenburg syndrome, type 2D, 608890
SOX10	65.8	98.2	91.3	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SPATA5	132	99.9	99.2	Epilepsy, hearing loss, and mental retardation syndrome, 616577
STRC	119	99.9	98.4	Deafness, autosomal recessive 16, 603720
SYNE4	73.9	98.3	91.6	Deafness, autosomal recessive 76, 615540
TBC1D24	179.2	100	100	Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044 DOORS syndrome, 220500 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021
TECTA	208	100	100	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TIMM8A	46	94.5	78.8	Mohr-Tranebjaerg syndrome, 304700
TJP2	111.1	93.8	92.2	Cholestasis, progressive familial intrahepatic 4, 615878 Hypercholanemia, familial, 607748
TMC1	122.8	98.2	93.8	Deafness, autosomal dominant 36, 606705 Deafness, autosomal recessive 7, 600974

TMEM132E	115.9	94.8	91.8	No OMIM phenotype Deafness,autosomal dominant 99 (Li et al. Hum Mutat 2015 36(1) 98-105)
TMIE	109.6	98.8	92.1	Deafness, autosomal recessive 6, 600971
TMPRSS3	125.5	100	99.9	Deafness, autosomal recessive 8/10, 601072
TMTC2	146.9	97.5	97.4	No OMIM phenotype
TNC	187.5	100	99.7	Deafness, autosomal dominant 56, 615629
TPRN	62.7	74.7	65.4	Deafness, autosomal recessive 79, 613307
TRIOBP	135.6	97	94.9	Deafness, autosomal recessive 28, 609823
TSPEAR	141.5	100	99	?Deafness, autosomal recessive 98, 614861
TYR	185.3	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	97.5	100	99.4	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
USH1G	195.3	98.4	96.3	Usher syndrome, type 1G, 606943
USH2A	148.5	100	99.7	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
WBP2	93.7	100	100	Deafness, autosomal recessive 107, 617639
WFS1	251.4	100	99.7	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 Wolfram syndrome 1, 222300 Wolfram-like syndrome, autosomal dominant, 614296 {Diabetes mellitus, noninsulin-dependent, association with}, 125853
WHRN	114	99.8	98.8	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
YAP1	95.6	87.8	81.6	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 : September 11th, 2018.
This list is accurate for panel version DG 2.14
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors