HEARING IMPAIRMENT GENE PANEL DG 2.3.x

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM ID*
ACTB	68,9	100%	94%	Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTG1	57,8	100%	94%	Deafness, autosomal dominant 20/26, 604717 Baraitser-Winter syndrome 2, 614583
ADCY1	102,4	92%	92%	?Deafness, autosomal recessive 44, 610154
APOPT1	90,9	87%	80%	Mitochondrial Complex IV Deficiency, 220110
ATP1A2	107,9	100%	99%	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP6V1B1	108,7	100%	99%	Renal tubular acidosis with deafness, 267300
BDP1	134,1	100%	99%	No OMIM phenotype Hearing loss (Girotto (2013) PLoS One 8,e80323)
BSND	119,6	100%	99%	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
CABP2	54,2	85%	67%	Deafness, autosomal recessive 93, 614899
CACNA1D	118,2	98%	98%	Sinoatrial node dysfunction and deafness, 614896
CCDC50	133,9	97%	96%	Deafness, autosomal dominant 44, 607453
CDH23	100,3	98%	96%	Usher syndrome, type 1D, 601067 Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067
CEACAM16	86,8	100%	98%	Deafness, autosomal dominant 4B, 614614
CIB2	115,4	100%	100%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CLDN14	65,5	100%	100%	Deafness, autosomal recessive 29, 614035
CLIC5	106,8	96%	89%	?Deafness, autosomal recessive 103, 616042
CLPP	85,9	99%	93%	Perrault syndrome 3, 614129

CLRN1	166,3	100%	100%	Usher syndrome, type 3A, 276902 Retinitis pigmentosa 61, 614180
COCH	122,9	100%	99%	Deafness, autosomal dominant 9, 601369
COL11A1	102,1	98%	98%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis, 228520
COL11A2	14,6	56%	21%	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	91,7	100%	97%	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 Spondyloperipheral dysplasia, 271700 SED, Namaqualand type Osteoarthritis with mild chondrodysplasia, 604864 Vitreoretinopathy with phalangeal epiphyseal dysplasia Platyspondylic skeletal dysplasia, Torrance type, 151210 Otospondylomegaepiphyseal dysplasia, 215150 Avascular necrosis of the femoral head, 608805 Legg-Calve-Perthes disease, 150600 Stickler sydrome, type I, nonsyndromic ocular, 609508 Czech dysplasia, 609162
COL4A3	76,6	97%	93%	Alport syndrome, autosomal recessive, 203780 Hematuria, benign familial, 141200 Alport syndrome, autosomal dominant, 104200
COL4A4	91,7	99%	98%	Alport syndrome, autosomal recessive, 203780 Hematuria,familial benign
COL4A5	73,2	99%	96%	Alport syndrome, 301050

COL4A6	84,5	97%	94%	?Deafness,X-linked 6,300914
COL9A1	108,9	100%	95%	Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	70,6	96%	90%	Epiphyseal dysplasia, multiple, 2, 600204 {Intervertebral disc disease, susceptibility to}, 603932 Stickler syndrome, type V, 614284
CRYM	72,5	99%	97%	Deafness, autosomal dominant 40
DCDC2	172,8	100%	100%	?Deafness,autosomal recessive 66,610212 Nephronophthisis 19,616217
DFNA5	102,3	100%	97%	Deafness, autosomal dominant 5, 600994
DFNB31	90,6	98%	96%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
DFNB59	129,9	100%	100%	Deafness, autosomal recessive 59, 610220
DIABLO	115,3	100%	87%	Deafness, autosomal dominant 64, 614152
DIAPH1	88,6	99%	92%	Deafness, autosomal dominant 1, 124900
DIAPH3	117,3	98%	97%	Auditory neuropathy, autosomal dominant, 1, 609129
DSPP	154,6	99%	96%	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	88,6	100%	100%	Waardenburg syndrome, type 4B, 613265 Central hypoventilation syndrome, congenital, 209880 {Hirschsprung disease, susceptibility to}, 613712
EDNRB	146,4	100%	99%	?{Hirschsprung disease, susceptibility to}, 600155 ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580
ELMOD3	117,6	100%	100%	?Deafness, autosomal recessive 88, 615429
EPS8	91,8	100%	99%	?Deafness, autosomal recessive 102, 615974
ESPN	44,9	78%	55%	Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant
ESRRB	59,6	95%	80%	Deafness, autosomal recessive 35, 608565

EYA1	109,6	100%	98%	Branchiootorenal syndrome 1, with or without cataracts, 113650 Anterior segment anomalies with or without cataract, 113650 Branchiootic syndrome 1, 602588 Otofaciocervical syndrome, 166780
EYA4	126,3	100%	100%	Deafness, autosomal dominant 10, 601316 Cardiomyopathy, dilated, 1J, 605362
FAM65B	96,7	100%	99%	?Deafness,autosomal recessive 104,616515
FGF3	76,7	100%	96%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FOXI1	92,4	100%	100%	Enlarged vestibular aqueduct, 600791
GIPC3	102,7	100%	92%	Deafness, autosomal recessive 15, 601869
GJB2	178,9	100%	100%	Deafness, autosomal recessive 1A, 220290 Deafness, autosomal dominant 3A, 601544 Vohwinkel syndrome, 124500 Keratoderma, palmoplantar, with deafness, 148350 Keratitis-ichthyosis-deafness syndrome, 148210 Hystrix-like ichthyosis with deafness, 602540 Bart-Pumphrey syndrome, 149200
GJB3	153	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	172,9	100%	100%	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GPR98	121,4	100%	99%	Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
GPSM2	150,6	100%	99%	Chudley-McCullough syndrome, 604213
GRHL2	108,8	100%	99%	Deafness, autosomal dominant 28, 608641
GRXCR1	204,8	100%	100%	Deafness, autosomal recessive 25, 613285
GRXCR2	144,5	100%	100%	?Deafness, autosomal recessive 101, 615837
HARS	133,7	100%	100%	Usher syndrome type 3B, 614504

HARS2	148,8	100%	100%	Perrault syndrome 2, 614926
HGF	106,8	96%	96%	Deafness, autosomal recessive 39, 608265
HSD17B4	96	96%	96%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
ILDR1	61,8	100%	99%	Deafness, autosomal recessive 42, 609646
KARS	124	100%	100%	Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, autosomal recessive 89, 613916
KCNE1	226,6	100%	100%	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome-5, 613695
KCNJ10	162,2	100%	100%	SESAME syndrome, 612780 Enlarged vestibular aqueduct, digenic, 600791
KCNQ1	64,3	93%	83%	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	100,3	92%	84%	ness, autosomal dominant 2A, 600101
KITLG	84,3	100%	98%	Hyperpigmentation with or without hypopigmentation,145250 [Skin/hair/eye pigmentation 7],611664
LARS2	122,7	100%	100%	Perrault syndrome 4, 615300
LHFPL5	195,8	100%	100%	Deafness, autosomal recessive 67, 610265
LOXHD1	110,3	100%	99%	Deafness, autosomal recessive 77, 613079
LRTOMT	111,3	86%	84%	Deafness, autosomal recessive 63, 611451
MARVELD2	169,9	100%	95%	Deafness, autosomal recessive 49, 610153
MITF	123,9	100%	99%	Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 Tietz albinism-deafness syndrome, 103500 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456
MSRB3	132,2	100%	100%	Deafness, autosomal recessive 74, 613718
MYH14	63,6	92%	81%	Deafness, autosomal dominant 4A, 600652 Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369

MYH9	99,9	100%	98%	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	89,6	97%	92%	Deafness, autosomal recessive 3, 600316
MYO3A	113,4	99%	98%	Deafness, autosomal recessive 30, 607101
MYO6	106,3	100%	98%	Deafness,autosomal dominant 22,606346 Deafness,autosomal dominant 22,with hypertrophic cardiomyopathy,606346 Deafness,autosomal recessive 37,607821
MYO7A	82,9	97%	93%	Usher syndrome, type 1B, 276900 Deafness,autosomal dominant 11,601317 Deafness,autosomal recessive 2,600060
NARS2	117,6	100%	99%	Combined oxidative phosphorylation deficiency 24,616239 DFNB94, Simon, PLoS Genet. 2015 Mar 25;11
NLRP3	122,9	100%	99%	Cold-induced autoinflammatory syndrome, familial, 120100
OPA1	130,6	100%	99%	Optic atrophy 1, 165500
OSBPL2	90,1	100%	99%	Deafness,autosomal dominant 67,616340
OTOA	73,4	69%	68%	Deafness, autosomal recessive 22, 607039
OTOF	96,6	98%	95%	Deafness, autosomal recessive 9, 601071
OTOG	96,9	99%	95%	Deafness, autosomal recessive 18B, 614945
OTOGL	121,5	100%	99%	Deafness, autosomal recessive 84B, 614944
P2RX2	94	100%	98%	Deafness, autosomal dominant 41, 608224
PAX3	128,7	100%	98%	Waardenburg syndrome, type 1, 193500 Craniofacial-deafness-hand syndrome,122880 Rhabdomyosarcoma 2,alveolar,268220 Waardenburg syndrome,type 3,148820
PCDH15	135,4	99%	99%	Usher syndrome, type 1F, 602083 Deafness,autosomal recessive 23,609533 Usher syndrome, type 1D/F digenic,601067
PDZD7	83,3	97%	89%	{Retinal disease in Usher syndrome type IIA, modifier of}, 276901
PET100	81,2	100%	99%	Mitochondrial complex IV deficiency, 220110
PNPT1	110,2	100%	100%	Combined oxidative phosphorylation deficiency 13, 614932

POU3F4	149,2	100%	100%	Deafness, X-linked 2, 304400
POU4F3	183,6	100%	100%	Deafness, autosomal dominant 15, 602459
PRPS1	135	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	114,4	94%	93%	Deafness, autosomal recessive 84A, 613391
RDX	56,8	90%	79%	Deafness, autosomal recessive 24, 611022
SERPINB6	133,9	100%	100%	Deafness, autosomal recessive 91, 613453
SIX1	77,3	95%	95%	Brachiootic syndrome 3, 608389 Deafness,autosomal dominant 23,605192
SIX5	36,4	87%	77%	Branchiootorenal syndrome 2, 610896
SLC17A8	131,2	100%	100%	Deafness, autosomal dominant 25, 605583
SLC26A4	99,8	99%	97%	Pendred syndrome, 274600 Deafness,autosomal recessive 4,with enlarged vestibular aqueduct,600791
SLC26A5	95,6	100%	100%	Deafness, autosomal recessive 61, 613865
SLC33A1	96,6	100%	100%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts,hearing loss,and neurodegeneration,614482
SLITRK6	170,1	100%	100%	Deafness and myopia, 221200
SMPX	124,2	100%	99%	Deafness, X-linked 4, 300066
SNAI2	86,6	100%	100%	Waardenburg syndrome, type 2D, 608890 Piebaldism,172800
SOX10	72,3	100%	100%	Waardenburg syndrome, type 4C, 613266 PCWH syndrome,609136 Waardenburg syndrome,type 2E,with/without neurologic involvement,611584
STRC	12,8	18%	14%	Deafness, autosomal recessive 16, 603720
SYNE4	88,4	100%	100%	Deafness, autosomal recessive 76, 615540
TBC1D24	112,2	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	126	100%	99%	Deafness, autosomal dominant 8/12, 601543 Deafness,autosomal recessive 21,603629
TIMM8A	52,8	99%	82%	Deafness, X-linked 1, progressive
TJP2	89,7	100%	98%	Cholestasis, progressive familial intrahepatic 4, 615878 Hypercholanemia, familial, 607748
TMC1	116,9	100%	100%	Deafness, autosomal recessive 7, 600974 Deafness,autosomal dominant 36,606705
TMEM132E	75,5	99%	96%	Deafness,autosomal dominant 99 (Li et al. Hum Mutat 2015 36(1) 98-105)
TMIE	59,2	99%	83%	Deafness, autosomal recessive 6, 600971
TMPRSS3	92,6	100%	96%	Deafness, autosomal recessive 8/10, 601072
TNC	134,1	96%	95%	Deafness, autosomal dominant 56, 615629
TPRN	40,9	79%	63%	Deafness, autosomal recessive 79, 613307
TRIOBP	95,4	95%	91%	Deafness, autosomal recessive 28, 609823
TSPEAR	114,2	100%	99%	Deafness, autosomal recessive 98, 614861
TYR	146,5	74%	74%	Albinism,oculocutaneous,type IA,203100 Albinism,oculocutaneous,type IB,606952 Waardenburg syndrome/albinism, digenic,103470 [Skin/hair/eye pigmentation 3],601800
USH1C	76,7	99%	92%	Deafness,autosomal recessive 18A,602092 Usher syndrome,type 1C,276904
USH1G	110,5	98%	88%	Usher syndrome, type 1G, 606943
USH2A	122,7	100%	99%	Usher syndrome, type 2A, 276901
WFS1	155,3	100%	100%	Deafness,autosomal dominant 6/14/38,600965 Wolfram syndrome,222300 Wolfram-like syndrome,autosomal dominant,614296 {Diabetes mellitus,noninsulin-dependent,association with},125853
YAP1	76,7	99%	90%	Coloboma, ocular, 120433 Coloboma, ocular, with/without hearing impairment, cleft lip/palate, mental retardation, 120433

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
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
OMIM release used for OMIM disease identifiers and descriptions : June 30th, 2015

This list is accurate for all panel versions starting with DG 2.3. (where x is a random number signifying a minor analysis patch without consequences for the panel composition or coverage information)

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