HEARING IMPAIRMENT GENE PANEL DGD20062014

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
ACTB	61,8	100%	95%	Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTG1	60,2	97%	87%	Deafness, autosomal dominant 20/26, 604717 Baraitser-Winter syndrome 2, 614583
ADCY1	97,4	94%	92%	?Deafness, autosomal recessive 44, 610154
BDP1	122,3	99%	98%	No OMIM phenotype Girotto (2013) PLoS One 8, e80323
BSND	105,3	100%	97%	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
CABP2	50,4	87%	70%	Deafness, autosomal recessive 93, 614899
CACNA1D	113,9	100%	98%	Sinoatrial node dysfunction and deafness, 614896
CCDC50	122,2	99%	96%	Deafness, autosomal dominant 44, 607453
CDH23	94,4	100%	98%	Usher syndrome, type 1D, 601067 Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067
CEACAM16	92,9	99%	93%	Deafness, autosomal dominant 4B, 614614
CIB2	130,2	100%	100%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CLDN14	69,2	100%	94%	Deafness, autosomal recessive 29, 614035
CLIC5	89,8	95%	89%	No OMIM phenotype

CLPP	76,1	93%	88%	Perrault syndrome 3, 614129

CLRN1	155,5	100%	100%	Usher syndrome, type 3A, 276902 Retinitis pigmentosa 61, 614180 -3
COCH	103,2	99%	97%	Deafness, autosomal dominant 9, 601369
COL11A1	92,1	98%	97%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis, 228520
COL11A2	13,4	55%	16%	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	80,5	99%	95%	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	70	97%	93%	Alport syndrome, autosomal recessive, 203780 Hematuria, benign familial, 141200 Alport syndrome, autosomal dominant, 104200

COL4A4	83,8	99%	97%	Alport syndrome, autosomal recessive, 203780 Hematuria,familial benign
COL4A5	69,5	100%	96%	diffuse leiomyomatosis with Alport syndrome = contiguous gene syndrome with COL4A6 Alport syndrome, 301050
COL4A6	81,8	99%	97%	diffuse leiomyomatosis with Alport syndrome = contiguous gene with COL4A5 Leiomyomatosis, diffuse, with Alport syndrome, 308940 (4)
COL9A1	96,9	99%	93%	Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	75,1	95%	93%	Epiphyseal dysplasia, multiple, 2, 600204 {Intervertebral disc disease, susceptibility to}, 603932 Stickler syndrome, type V, 614284
CRYM	70,4	100%	99%	Deafness, autosomal dominant 40
DFNA5	96,6	97%	93%	Deafness, autosomal dominant 5, 600994
DFNB31	78,7	99%	94%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
DFNB59	112,2	100%	100%	Deafness, autosomal recessive 59, 610220
DIABLO	103,4	100%	90%	Deafness, autosomal dominant 64, 614152
DIAPH1	81,4	99%	89%	Deafness, autosomal dominant 1, 124900
DIAPH3	104,5	99%	96%	Auditory neuropathy, autosomal dominant, 1, 609129
DSPP	138,4	98%	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 -3

EDN3	91,7	100%	100%	Waardenburg syndrome, type 4B, 613265 Central hypoventilation syndrome, congenital, 209880 {Hirschsprung disease, susceptibility to, 4}, 613712
EDNRB	148	100%	99%	?{Hirschsprung disease, susceptibility to, 2}, 600155 ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580
ELMOD3	103,4	100%	100%	?Deafness, autosomal recessive 88, 615429
EPS8	80,2	100%	96%	?Deafness, autosomal recessive 102, 615974
ESPN	41,3	78%	57%	Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant
ESRRB	54,1	87%	73%	Deafness, autosomal recessive 35, 608565
EYA1	104,9	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	116,9	100%	100%	Deafness, autosomal dominant 10, 601316 Cardiomyopathy, dilated, 1J, 605362
FGF3	88,2	100%	98%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FOXI1	94,3	100%	100%	Enlarged vestibular aqueduct, 600791
GIPC3	111,9	98%	91%	Deafness, autosomal recessive 15, 601869
GJB2	174,6	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	128,3	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	141,9	100%	100%	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GPR98	108,3	100%	98%	Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
GPSM2	131,1	100%	100%	Chudley-McCullough syndrome, 604213
GRHL2	102	100%	99%	Deafness, autosomal dominant 28, 608641
GRXCR1	182,9	100%	100%	Deafness, autosomal recessive 25, 613285
HARS	121,1	100%	99%	Usher syndrome type 3B, 614504
HARS2	132,5	100%	100%	Perrault syndrome 2, 614926
HGF	103,3	100%	100%	Deafness, autosomal recessive 39, 608265
HSD17B4	91,8	100%	98%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
ILDR1	54,3	100%	99%	Deafness, autosomal recessive 42, 609646
KARS	114,1	100%	100%	Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, autosomal recessive 89, 613916
KCNE1	203,1	100%	100%	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome-5, 613695
KCNJ10	148,5	100%	100%	SESAME syndrome, 612780 Enlarged vestibular aqueduct, digenic, 600791

KCNQ1	64	91%	82%	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	89,4	88%	83%	Deafness, autosomal dominant 2A, 600101
LARS2	108,5	100%	100%	Perrault syndrome 4, 615300
LHFPL5	160,3	100%	100%	Deafness, autosomal recessive 67, 610265
LOXHD1	99,6	100%	99%	Deafness, autosomal recessive 77, 613079
LRTOMT	107,3	93%	90%	Deafness, autosomal recessive 63, 611451
MARVELD2	157,5	98%	95%	Deafness, autosomal recessive 49, 610153
MITF	145,9	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	117,1	100%	100%	Deafness, autosomal recessive 74, 613718
MYH14	61,5	92%	81%	Deafness, autosomal dominant 4A, 600652 Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369
MYH9	93,7	99%	97%	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	89,4	97%	92%	Deafness, autosomal recessive 3, 600316
MYO3A	104,4	99%	96%	Deafness, autosomal recessive 30, 607101

MYO6	99,5	99%	98%	Deafness, autosomal dominant 22, 606346 Deafness, autosomal recessive 37, 607821 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346
MYO7A	76,4	97%	91%	Usher syndrome, type 1B, 276900 Deafness, autosomal recessive 2, 600060 Deafness, autosomal dominant 11, 601317

NLRP3	117,3	100%	99%	Cold-induced autoinflammatory syndrome, familial, 120100 Muckle-Wells syndrome, 191900 CINCA syndrome, 607115
OPA1	124,9	100%	99%	Optic atrophy 1, 165500 {Glaucoma, normal tension, susceptibility to}, 606657 Optic atrophy plus syndrome, 125250
OTOA	69,9	67%	66%	Deafness, autosomal recessive 22, 607039
OTOF	92	99%	97%	Deafness, autosomal recessive 9, 601071 Auditory neuropathy, autosomal recessive, 1, 601071
OTOG	88,3	97%	93%	Deafness, autosomal recessive 18B, 614945
OTOGL	111,3	100%	99%	Deafness, autosomal recessive 84B, 614944
P2RX2	96,3	100%	98%	Deafness, autosomal dominant 41, 608224
PAX3	105,6	99%	97%	Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820 Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220
PCDH15	126,6	100%	100%	Usher syndrome, type 1F, 602083 Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067
PDZD7	75,3	95%	87%	{Retinal disease in Usher syndrome type IIA, modifier of}, 276901 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472

PNPT1	99,1	100%	99%	Combined oxidative phosphorylation deficiency 13, 614932 Deafness, autosomal recessive 70, 614934

POU3F4	137,5	100%	100%	Deafness, X-linked 2, 304400
POU4F3	160,4	100%	100%	Deafness, autosomal dominant 15, 602459

PRPS1	131,1	100%	100%	Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Arts syndrome, 301835 Deafness, X-linked 1, 304500
PTPRQ	110,9	99%	98%	Deafness, autosomal recessive 84A, 613391
RDX	48,8	84%	71%	Deafness, autosomal recessive 24, 611022
SERPINB6	130,6	99%	99%	Deafness, autosomal recessive 91, 613453
SIX1	84,4	100%	100%	Brachiootic syndrome 3, 608389 Deafness, autosomal dominant 23, 605192
SIX5	39	95%	79%	Branchiootorenal syndrome 2, 610896
SLC17A8	122,2	100%	100%	Deafness, autosomal dominant 25, 605583
SLC26A4	94,1	99%	97%	Pendred syndrome, 274600 Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791
SLC26A5	89,1	100%	97%	Deafness, autosomal recessive 61, 613865
SLC33A1	92,1	100%	99%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLITRK6	156,2	100%	100%	Deafness and myopia, 221200
SMPX	106,6	100%	99%	Deafness, X-linked 4, 300066

SNAI2	79,3	100%	100%	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SOX10	67,6	100%	100%	Waardenburg syndrome, type 4C, 613266 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136
STRC	17,5	18%	16%	Deafness, autosomal recessive 16, 603720

TBC1D24	109,5	100%	100%	Myoclonic epilepsy, infantile, familial, 605021 Epileptic encephalopathy, early infantile, 16, 615338
TECTA	114,6	99%	97%	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TIMM8A	57,3	94%	85%	Deafness, X-linked 1, progressive Mohr-Tranebjaerg syndrome, 304700 Jensen syndrome, 311150
TMC1	106,6	100%	100%	Deafness, autosomal recessive 7, 600974 Deafness, autosomal dominant 36, 606705
TMIE	58,1	99%	91%	Deafness, autosomal recessive 6, 600971
TMPRSS3	91,1	100%	96%	Deafness, autosomal recessive 8/10, 601072
TNC	126,5	100%	98%	Deafness, autosomal dominant 56, 615629
TPRN	42,6	82%	74%	Deafness, autosomal recessive 79, 613307
TRIOBP	95	96%	92%	Deafness, autosomal recessive 28, 609823
TSPEAR	108,5	100%	99%	Deafness, autosomal recessive 98, 614861
USH1C	75,6	98%	93%	Acadian and Samaritan variety Usher syndrome, type 1C, 276904 Deafness, autosomal recessive 18A, 602092
USH1G	104,9	94%	88%	Usher syndrome, type 1G, 606943

USH2A	109,7	99%	98%	Usher syndrome, type 2A, 276901 Retinitis pigmentosa 39, 613809 -3
WFS1	146,8	100%	99%	Wolfram syndrome, 222300 Deafness, autosomal dominant 6/14/38, 600965 Wolfram-like syndrome, autosomal dominant, 614296 {Diabetes mellitus, noninsulin-dependent, association with}, 125853

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
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 : 15 october 2013
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors