HEARING IMPAIRMENT GENE PANEL DGD141114

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
ACTB	60.1	97%	90%	Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTG1	56.9	99%	93%	Deafness, autosomal dominant 20/26, 604717 Baraitser-Winter syndrome 2, 614583
ADCY1	97.4	93%	92%	?Deafness, autosomal recessive 44, 610154
APOPT1	100.9	100%	99%	Mitochondrial Complex IV Deficiency, 220110
ATP1A2	100.1	100%	98%	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP6V1B1	98.0	100%	98%	Renal tubular acidosis with deafness, 267300
BDP1	126.7	99%	97%	No OMIM phenotype Hearing loss (Girotto (2013) PLoS One 8,e80323)
BSND	116.3	100%	98%	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
CABP2	47.4	84%	72%	Deafness, autosomal recessive 93, 614899
CACNA1D	115.7	100%	98%	Sinoatrial node dysfunction and deafness, 614896
CCDC50	132.9	99%	96%	Deafness, autosomal dominant 44, 607453
CDH23	93.7	99%	98%	Usher syndrome, type 1D, 601067 Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067
CEACAM16	92.3	97%	90%	Deafness, autosomal dominant 4B, 614614
CIB2	115.9	100%	100%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CLDN14	66.2	100%	99%	Deafness, autosomal recessive 29, 614035
CLIC5	93.8	97%	89%	?Deafness, autosomal recessive 103, 616042
CLPP	90.1	97%	91%	Perrault syndrome 3, 614129
CLRN1	165.9	100%	100%	Usher syndrome, type 3A, 276902 Retinitis pigmentosa 61, 614180
COCH	117.8	100%	98%	Deafness, autosomal dominant 9, 601369

COL11A1	98.0	98%	97%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis, 228520
COL11A2	14.4	61%	17%	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	84.6	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	74.2	97%	94%	Alport syndrome, autosomal recessive, 203780 Hematuria, benign familial, 141200 Alport syndrome, autosomal dominant, 104200
COL4A4	92.0	100%	97%	Alport syndrome, autosomal recessive, 203780 Hematuria,familial benign
COL4A5	38.4	94%	78%	diffuse leiomyomatosis with Alport syndrome = contiguous gene syndrome with COL4A6 Alport syndrome, 301050
COL4A6	44.3	96%	83%	diffuse leiomyomatosis with Alport syndrome = contiguous gene with COL4A5 Leiomyomatosis, diffuse, with Alport syndrome, 308940 (4)
COL9A1	108.1	100%	96%	Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134

COL9A2	72.9	98%	93%	Epiphyseal dysplasia, multiple, 2, 600204 {Intervertebral disc disease, susceptibility to}, 603932 Stickler syndrome, type V, 614284
CRYM	75.1	100%	99%	Deafness, autosomal dominant 40
DFNA5	100.4	99%	95%	Deafness, autosomal dominant 5, 600994
DFNB31	85.0	100%	98%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
DFNB59	127.4	100%	100%	Deafness, autosomal recessive 59, 610220
DIABLO	114.9	97%	90%	Deafness, autosomal dominant 64, 614152
DIAPH1	82.5	99%	92%	Deafness, autosomal dominant 1, 124900
DIAPH3	114.9	98%	96%	Auditory neuropathy, autosomal dominant, 1, 609129
DSPP	139.2	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
EDN3	91.8	100%	100%	Waardenburg syndrome, type 4B, 613265 Central hypoventilation syndrome, congenital, 209880 {Hirschsprung disease, susceptibility to}, 613712
EDNRB	178.1	100%	99%	?{Hirschsprung disease, susceptibility to}, 600155 ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580
ELMOD3	110.4	100%	100%	?Deafness, autosomal recessive 88, 615429
EPS8	88.2	100%	98%	?Deafness, autosomal recessive 102, 615974
ESPN	42.7	77%	52%	Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant
ESRRB	63.0	84%	75%	Deafness, autosomal recessive 35, 608565
EYA1	113.1	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	126.8	100%	100%	Deafness, autosomal dominant 10, 601316 Cardiomyopathy, dilated, 1J, 605362
FAM65B	95.4	100%	98%	No OMIM phenotype Hearing loss, non-syndromic, autosomal recessive (Diaz-Horta (2014) Proc Natl Acad Sci USA 111,9864)
FGF3	72.2	100%	93%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FOXI1	93.8	100%	100%	Enlarged vestibular aqueduct, 600791

GIPC3	107.2	95%	92%	Deafness, autosomal recessive 15, 601869
GJB2	172.7	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	149.1	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	157.2	100%	100%	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GPR98	115.4	99%	98%	Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
GPSM2	146.5	100%	100%	Chudley-McCullough syndrome, 604213
GRHL2	100.4	100%	98%	Deafness, autosomal dominant 28, 608641
GRXCR1	189.0	100%	100%	Deafness, autosomal recessive 25, 613285
GRXCR2	140.8	100%	100%	?Deafness, autosomal recessive 101, 615837
HARS	123.3	100%	100%	Usher syndrome type 3B, 614504
HARS2	136.3	100%	100%	Perrault syndrome 2, 614926
HGF	116.0	100%	100%	Deafness, autosomal recessive 39, 608265
HSD17B4	98.2	100%	99%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
ILDR1	58.1	100%	99%	Deafness, autosomal recessive 42, 609646
KARS	117.9	100%	100%	Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, autosomal recessive 89, 613916
KCNE1	206.9	100%	100%	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome-5, 613695
KCNJ10	151.4	100%	100%	SESAME syndrome, 612780 Enlarged vestibular aqueduct, digenic, 600791

KCNQ1	62.5	93%	81%	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	93.7	94%	89%	ness, autosomal dominant 2A, 600101
LARS2	116.2	100%	99%	Perrault syndrome 4, 615300
LHFPL5	176.4	100%	100%	Deafness, autosomal recessive 67, 610265
LOXHD1	103.8	100%	99%	Deafness, autosomal recessive 77, 613079
LRTOMT	99.4	92%	88%	Deafness, autosomal recessive 63, 611451
MARVELD2	153.4	98%	95%	Deafness, autosomal recessive 49, 610153
MITF	131.8	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	115.5	100%	98%	Deafness, autosomal recessive 74, 613718
MYH14	62.6	96%	85%	Deafness, autosomal dominant 4A, 600652 Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369
MYH9	96.5	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	89.7	98%	92%	Deafness, autosomal recessive 3, 600316
MYO3A	109.8	99%	97%	Deafness, autosomal recessive 30, 607101
MYO6	108.0	100%	99%	Deafness, autosomal dominant 22, 606346
MYO7A	77.0	95%	91%	Usher syndrome, type 1B, 276900
NLRP3	124.2	100%	99%	Cold-induced autoinflammatory syndrome, familial, 120100
OPA1	131.1	99%	99%	Optic atrophy 1, 165500
OSBPL2	92.6	98%	95%	No OMIM phenotype
OTOA	65.3	67%	66%	Deafness, autosomal recessive 22, 607039
OTOF	92.6	99%	96%	Deafness, autosomal recessive 9, 601071
OTOG	88.9	99%	94%	Deafness, autosomal recessive 18B, 614945
OTOGL	119.0	100%	99%	Deafness, autosomal recessive 84B, 614944
P2RX2	92.1	100%	98%	Deafness, autosomal dominant 41, 608224

PAX3	111.9	99%	96%	Waardenburg syndrome, type 1, 193500
PCDH15	137.6	100%	100%	Usher syndrome, type 1F, 602083
PDZD7	74.0	96%	86%	{Retinal disease in Usher syndrome type IIA, modifier of}, 276901
PET100	70.5	100%	99%	Mitochondrial complex IV deficiency, 220110
PNPT1	103.6	100%	100%	Combined oxidative phosphorylation deficiency 13, 614932
POU3F4	68.1	100%	100%	Deafness, X-linked 2, 304400
POU4F3	173.5	100%	100%	Deafness, autosomal dominant 15, 602459
PRPS1	62.8	99%	97%	Gout, PRPS-related, 300661
PTPRQ	118.2	100%	98%	Deafness, autosomal recessive 84A, 613391
RDX	56.6	91%	81%	Deafness, autosomal recessive 24, 611022
SERPINB6	142.3	99%	99%	Deafness, autosomal recessive 91, 613453
SIX1	80.7	100%	100%	Brachiootic syndrome 3, 608389
SIX5	44.7	94%	81%	Branchiootorenal syndrome 2, 610896
SLC17A8	122.6	100%	100%	Deafness, autosomal dominant 25, 605583
SLC26A4	95.4	99%	95%	Pendred syndrome, 274600
SLC26A5	93.4	100%	99%	Deafness, autosomal recessive 61, 613865
SLC33A1	100.8	100%	100%	Spastic paraplegia 42, autosomal dominant, 612539
SLITRK6	162.2	100%	100%	Deafness and myopia, 221200
SMPX	61.5	100%	89%	Deafness, X-linked 4, 300066
SNAI2	84.3	100%	100%	Waardenburg syndrome, type 2D, 608890
SOX10	85.0	100%	100%	Waardenburg syndrome, type 4C, 613266
STRC	11.6	17%	15%	Deafness, autosomal recessive 16, 603720
SYNE4	81.5	100%	100%	Deafness, autosomal recessive 76, 615540
TBC1D24	106.0	100%	100%	Myoclonic epilepsy, infantile, familial, 605021
TECTA	122.4	99%	98%	Deafness, autosomal dominant 8/12, 601543
TIMM8A	26.0	75%	67%	Deafness, X-linked 1, progressive
TJP2	84.5	100%	97%	Cholestasis, progressive familial intrahepatic 4, 615878 Hypercholanemia, familial, 607748
TMC1	111.4	100%	100%	Deafness, autosomal recessive 7, 600974
TMIE	57.4	99%	87%	Deafness, autosomal recessive 6, 600971
TMPRSS3	91.3	100%	96%	Deafness, autosomal recessive 8/10, 601072
TNC	130.3	100%	99%	Deafness, autosomal dominant 56, 615629
TPRN	48.1	82%	78%	Deafness, autosomal recessive 79, 613307
TRIOBP	93.9	95%	91%	Deafness, autosomal recessive 28, 609823
TSPEAR	120.9	100%	99%	Deafness, autosomal recessive 98, 614861
USH1C	79.4	99%	92%	Acadian and Samaritan variety Usher syndrome, type 1C, 276904

USH1G	98.7	95%	88%	Usher syndrome, type 1G, 606943
USH2A	116.3	100%	99%	Usher syndrome, type 2A, 276901
WFS1	145.6	100%	98%	Wolfram syndrome, 222300
YAP1	76.6	96%	88%	Coloboma, ocular, 120433 Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or 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 : 31 october 2014
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors