HEARING IMPAIRMENT PANEL (INCLUDING GJB2) DG-4.0.0 (275 GENES)

Gene	*Twist X2 covered >10x*	*Twist X2 covered >20x*	*WGS covered >10x*	*WGS covered >20x*	*Associated Phenotype description and OMIM disease ID*
ABCC1	100.0%	100.0%	100.0%	98.2%	?Deafness, autosomal dominant 77, 618915
ABHD12	100.0%	100.0%	99.9%	97.3%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACTB	100.0%	100.0%	100.0%	99.0%	Baraitser-Winter syndrome 1, 243310;Becker nevus, syndromic or isolated, somatic mosaic, 604919;Thrombocytopenia 8, with dysmorphic features and developmental delay, 620475;Dystonia-deafness syndrome 1, 607371;Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic, 620479
ACTG1	100.0%	100.0%	100.0%	98.2%	Deafness, autosomal dominant 20/26, 604717;Baraitser-Winter syndrome 2, 614583

ADAMTS1	100.0%	100.0%	100.0%	99.4%	
ADCY1	99.2%	98.7%	99.7%	95.3%	?Deafness, autosomal recessive 44, 610154
ADGRV1	100.0%	100.0%	100.0%	98.2%	Usher syndrome, type 2C, 605472;Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472;?Febrile seizures, familial, 4, 604352
AFG2A	100.0%	100.0%	100.0%	98.9%	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577
AFG2B	100.0%	100.0%	100.0%	97.6%	Deafness, autosomal recessive 119, 619615;Neurodevelopmental disorder with hearing loss and spasticity, 619616
AIFM1	100.0%	99.8%	97.6%	67.9%	Combined oxidative phosphorylation deficiency 6, 300816;Cowchock syndrome, 310490;Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232;Deafness, X-linked 5, 300614
ALMS1	100.0%	100.0%	100.0%	98.4%	Alstrom syndrome, 203800
AMMECR1	98.3%	94.8%	95.6%	66.0%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990

ANLN	100.0%	100.0%	100.0%	98.4%	Focal segmental glomerulosclerosis 8, 616032
AP1B1	100.0%	100.0%	100.0%	99.3%	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150
ARSG	100.0%	100.0%	99.9%	98.4%	Usher syndrome, type IV, 618144
ATOH1	100.0%	100.0%	100.0%	97.3%	?Deafness, autosomal dominant 89, 620284
ATP11A	100.0%	100.0%	100.0%	99.4%	?Auditory neuropathy, autosomal dominant 2, 620384;?Leukodystrophy, hypomyelinating, 24, 619851;Deafness, autosomal dominant 84, 619810
ATP1A1	100.0%	100.0%	100.0%	99.1%	Hypomagnesemia, seizures, and impaired intellectual development 2, 618314;Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP1A3	100.0%	100.0%	100.0%	98.9%	Alternating hemiplegia of childhood 2, 614820;Dystonia-12, 128235;CAPOS syndrome, 601338;Developmental and epileptic encephalopathy 99, 619606

ATP2B2	100.0%	100.0%	100.0%	98.3%	Deafness, autosomal dominant 82, 619804;{Deafness, autosomal recessive 12, modifier of}, 601386
ATP6V0A4	100.0%	100.0%	100.0%	98.1%	Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722
ATP6V1B1	100.0%	100.0%	100.0%	99.0%	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300
ATP6V1B2	100.0%	100.0%	100.0%	98.3%	Zimmermann-Laband syndrome 2, 616455;Deafness, congenital, with onychodystrophy, autosomal dominant, 124480
BCAP31	99.1%	92.8%	98.0%	69.1%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCS1L	100.0%	100.0%	100.0%	99.2%	GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Bjornstad syndrome, 262000
BDP1	100.0%	100.0%	100.0%	97.1%	?Deafness, autosomal recessive 112, 618257
BICD1	100.0%	100.0%	100.0%	98.5%	

BMP4	100.0%	100.0%	100.0%	99.6%	Orofacial cleft 11, 600625;Microphthalmia, syndromic 6, 607932
BSND	100.0%	100.0%	100.0%	99.1%	Sensorineural deafness with mild renal dysfunction, 602522;Bartter syndrome, type 4a, 602522
BTD	94.2%	94.2%	100.0%	99.5%	Biotinidase deficiency, 253260
CABP2	100.0%	100.0%	100.0%	99.2%	Deafness, autosomal recessive 93, 614899
CACNA1D	100.0%	100.0%	100.0%	98.5%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474;Sinoatrial node dysfunction and deafness, 614896
CCDC50	100.0%	100.0%	99.9%	97.5%	?Deafness, autosomal dominant 44, 607453
CD151	100.0%	100.0%	100.0%	99.8%	[Blood group, Raph], 179620;Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057
CD164	100.0%	100.0%	100.0%	97.0%	?Deafness, autosomal dominant 66, 616969
CDC14A	100.0%	100.0%	99.9%	96.8%	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDC42	100.0%	100.0%	100.0%	98.7%	Takenouchi-Kosaki syndrome, 616737

CDH23	100.0%	100.0%	100.0%	99.3%	Usher syndrome, type 1D, 601067;{Pituitary adenoma 5, multiple types}, 617540;Usher syndrome, type 1D/F digenic, 601067;Deafness, autosomal recessive 12, 601386
CEACAM16	100.0%	100.0%	100.0%	99.6%	Deafness, autosomal dominant 4B, 614614;Deafness, autosomal recessive 113, 618410
CEP250	100.0%	99.9%	100.0%	98.6%	Cone-rod dystrophy and hearing loss 2, 618358
CEP78	100.0%	100.0%	100.0%	98.5%	Cone-rod dystrophy and hearing loss, 617236
CGN	100.0%	100.0%	100.0%	99.0%	
CHD7	100.0%	100.0%	100.0%	98.6%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800
CHSY1	99.9%	99.7%	100.0%	97.5%	Temtamy preaxial brachydactyly syndrome, 605282
CIB2	100.0%	99.9%	99.9%	97.8%	Deafness, autosomal recessive 48, 609439;Usher syndrome, type IJ, 614869
CISD2	100.0%	100.0%	100.0%	98.0%	Wolfram syndrome 2, 604928

CLDN14	100.0%	100.0%	100.0%	99.8%	Deafness, autosomal recessive 29, 614035
CLDN9	100.0%	100.0%	100.0%	99.9%	Deafness, autosomal recessive 116, 619093
CLIC5	100.0%	100.0%	100.0%	96.9%	?Deafness, autosomal recessive 103, 616042
CLPP	100.0%	100.0%	100.0%	96.3%	Perrault syndrome 3, 614129
CLRN1	100.0%	100.0%	100.0%	98.0%	Usher syndrome, type 3A, 276902;Retinitis pigmentosa 61, 614180
CLRN2	100.0%	100.0%	100.0%	99.5%	Deafness, autosomal recessive 117, 619174
COA8	100.0%	99.9%	100.0%	97.0%	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COCH	100.0%	100.0%	100.0%	99.4%	Deafness, autosomal dominant 9, 601369;?Deafness, autosomal recessive 110, 618094
COL11A1	100.0%	100.0%	100.0%	97.9%	Fibrochondrogenesis 1, 228520;Stickler syndrome, type II, 604841;Marshall syndrome, 154780;Deafness, autosomal dominant 37, 618533;{Lumbar disc herniation, susceptibility to}, 603932

COL11A2	100.0%	100.0%	100.0%	98.6%	Deafness, autosomal dominant 13, 601868;Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150;Fibrochondrogenesis 2, 614524;Deafness, autosomal recessive 53, 609706;Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840

COL2A1	100.0%	100.0%	100.0%	99.1%	?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248;Czech dysplasia, 609162;Achondrogenesis, type II or hypochondrogenesis, 200610;Spondyloperipheral dysplasia, 271700;SMED Strudwick type, 184250;?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450;SED congenita, 183900;Kniest dysplasia, 156550;Stickler syndrome, type I, nonsyndromic ocular, 609508;Osteoarthritis with mild chondrodysplasia, 604864;Stickler syndrome, type I, 108300;Platyspondylic skeletal dysplasia, Torrance type, 151210;Spondyloepiphyseal dysplasia, Stanescu type, 616583;Avascular necrosis of the femoral head, 608805;Legg-Calve-Perthes disease, 150600

COL4A3	100.0%	100.0%	100.0%	98.1%	Alport syndrome 3A, autosomal dominant, 104200;Hematuria, benign familial, 2, 620320;Alport syndrome 3B, autosomal recessive, 620536
COL4A4	99.5%	98.6%	100.0%	98.5%	Hematuria, familial benign, 1, 141200;Alport syndrome 2, autosomal recessive, 203780
COL4A5	99.3%	98.7%	97.6%	68.1%	Alport syndrome 1, X-linked, 301050
COL4A6	99.4%	98.7%	97.1%	66.7%	?Deafness, X-linked 6, 300914
COL9A1	100.0%	100.0%	100.0%	97.8%	Stickler syndrome, type IV, 614134;?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	100.0%	100.0%	100.0%	97.9%	Epiphyseal dysplasia, multiple, 2, 600204;?Stickler syndrome, type V, 614284
COL9A3	100.0%	100.0%	100.0%	98.6%	{Intervertebral disc disease, susceptibility to}, 603932;Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969;Stickler syndrome, type VI, 620022
CRYL1	100.0%	100.0%	100.0%	98.4%	
CRYM	100.0%	100.0%	100.0%	97.6%	Deafness, autosomal dominant 40, 616357

DCAF17	100.0%	100.0%	99.9%	98.3%	Woodhouse-Sakati syndrome, 241080
DCDC2	100.0%	100.0%	100.0%	97.6%	Nephronophthisis 19, 616217;?Deafness, autosomal recessive 66, 610212;Sclerosing cholangitis, neonatal, 617394
DHX16	100.0%	100.0%	100.0%	98.8%	Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733
DIABLO	100.0%	100.0%	100.0%	98.8%	Deafness, autosomal dominant 64, 614152
DIAPH1	100.0%	100.0%	99.9%	95.3%	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900;Seizures, cortical blindness, microcephaly syndrome, 616632
DIAPH3	100.0%	99.8%	100.0%	98.4%	Auditory neuropathy, autosomal dominant 1, 609129
DLL1	100.0%	100.0%	100.0%	98.9%	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709
DLX5	100.0%	100.0%	100.0%	99.1%	Split-hand/foot malformation 1, 183600;?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600

DMXL2	100.0%	100.0%	100.0%	98.7%	Developmental and epileptic encephalopathy 81, 618663;?Deafness, autosomal dominant 71, 617605;?Polyendocrine-polyneuropathy syndrome, 616113
DSPP	100.0%	100.0%	97.9%	95.2%	Dentinogenesis imperfecta, Shields type III, 125500;Dentinogenesis imperfecta, Shields type II, 125490;Dentin dysplasia, type II, 125420;Deafness, autosomal dominant 39, with dentinogenesis, 605594
EDN3	100.0%	100.0%	100.0%	99.6%	Waardenburg syndrome, type 4B, 613265;{Hirschsprung disease, susceptibility to, 4}, 613712
EDNRB	100.0%	100.0%	100.0%	98.1%	{Hirschsprung disease, susceptibility to, 2}, 600155;?ABCD syndrome, 600501;Waardenburg syndrome, type 4A, 277580
EFNB2	100.0%	100.0%	100.0%	99.1%	
EHD1	100.0%	100.0%	100.0%	99.2%	
ELMOD3	100.0%	100.0%	100.0%	99.0%	?Deafness, autosomal recessive 88, 615429;?Deafness, autosomal dominant 81, 619500

ELOVL1	100.0%	100.0%	100.0%	99.5%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
EPS8	100.0%	100.0%	100.0%	98.4%	?Deafness, autosomal recessive 102, 615974
EPS8L2	100.0%	100.0%	100.0%	95.9%	Deafness autosomal recessive 106, 617637
ERAL1	100.0%	100.0%	100.0%	98.3%	Perrault syndrome 6, 617565
ESPN	100.0%	100.0%	99.9%	95.2%	Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006;Deafness, autosomal recessive 36, 609006;?Usher syndrome, type 1M, 618632
ESRP1	100.0%	100.0%	100.0%	98.6%	?Deafness, autosomal recessive 109, 618013
ESRRB	100.0%	100.0%	100.0%	99.7%	Deafness, autosomal recessive 35, 608565
EXOSC2	100.0%	99.2%	100.0%	97.9%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763

EYA1	100.0%	100.0%	100.0%	99.1%	Branchiootic syndrome 1, 602588;Branchiootorenal syndrome 1, with or without cataracts, 113650;Anterior segment anomalies with or without cataract, 602588;?Otofaciocervical syndrome, 166780
EYA4	100.0%	100.0%	100.0%	99.0%	?Cardiomyopathy, dilated, 1J, 605362;Deafness, autosomal dominant 10, 601316
FDXR	100.0%	100.0%	100.0%	99.4%	Multiple mitochondrial dysfunctions syndrome 9B, 620887;Auditory neuropathy and optic atrophy, 617717
FGF3	100.0%	100.0%	100.0%	95.9%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706

FGFR3	100.0%	100.0%	100.0%	99.8%	Muenke syndrome, 602849;SADDAN, 616482;Hypochondroplasia, 146000;Thanatophoric dysplasia, type II, 187601;Nevus, epidermal, somatic, 162900;CATSHL syndrome, 610474;Thanatophoric dysplasia, type I, 187600;Spermatocytic seminoma, somatic, 273300;Bladder cancer, somatic, 109800;LADD syndrome 2, 620192;Achondroplasia, 100800;Cervical cancer, somatic, 603956;Colorectal cancer, somatic, 114500;Crouzon syndrome with acanthosis nigricans, 612247
FITM2	100.0%	100.0%	100.0%	99.2%	Siddiqi syndrome, 618635
FOXF2	99.9%	99.3%	99.6%	85.4%	
FOXI1	100.0%	100.0%	100.0%	99.3%	Enlarged vestibular aqueduct, 600791
GAB1	100.0%	100.0%	100.0%	98.9%	?Deafness, autosomal recessive 26, 605428
GAS2	100.0%	100.0%	100.0%	98.1%	?Deafness, autosomal recessive 125, 620877
GATA3	100.0%	100.0%	100.0%	99.1%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255

GIPC3	100.0%	100.0%	100.0%	97.7%	Deafness, autosomal recessive 15, 601869
GJB2	100.0%	100.0%	100.0%	99.4%	Keratoderma, palmoplantar, with deafness, 148350;Deafness, autosomal recessive 1A, 220290;Deafness, autosomal dominant 3A, 601544;Hystrix-like ichthyosis with deafness, 602540;Bart-Pumphrey syndrome, 149200;Keratitis-ichthyosis-deafness syndrome, 148210;Vohwinkel syndrome, 124500
GJB3	100.0%	100.0%	100.0%	99.5%	Deafness, digenic, GJB2/GJB3, 220290;Erythrokeratodermia variabilis et progressiva 1, 133200;Deafness, autosomal dominant 2B, with or without peripheral neuropathy, 612644
GJB6	100.0%	100.0%	99.9%	97.9%	Ectodermal dysplasia 2, Clouston type, 129500;Deafness, autosomal dominant 3B, 612643;Deafness, autosomal recessive 1B, 612645;Deafness, digenic GJB2/GJB6, 220290

GLA	91.4%	91.4%	98.4%	73.6%	Fabry disease, cardiac variant, 301500;Fabry disease, 301500
GOSR2	100.0%	100.0%	100.0%	99.7%	Epilepsy, progressive myoclonic 6, 614018;Muscular dystrophy, congenital, with or without seizures, 620166
GPR156	100.0%	100.0%	100.0%	99.3%	Deafness, autosomal recessive 121, 620551
GPRASP2	100.0%	100.0%	98.2%	72.8%	?Deafness, X-linked 7, 301018
GPSM2	95.5%	95.5%	100.0%	98.5%	Chudley-McCullough syndrome, 604213
GRAP	100.0%	100.0%	99.8%	96.7%	Deafness, autosomal recessive 114, 618456
GREB1L	100.0%	100.0%	100.0%	98.8%	Deafness, autosomal dominant 80, 619274;Renal hypodysplasia/aplasia 3, 617805
GRHL2	100.0%	100.0%	100.0%	98.4%	Deafness, autosomal dominant 28, 608641;Ectodermal dysplasia/short stature syndrome, 616029;Corneal dystrophy, posterior polymorphous, 4, 618031
GRXCR1	99.9%	99.3%	100.0%	98.8%	Deafness, autosomal recessive 25, 613285
GRXCR2	100.0%	100.0%	100.0%	99.3%	?Deafness, autosomal recessive 101, 615837

GSDME	100.0%	100.0%	100.0%	99.1%	Deafness, autosomal dominant 5, 600994
HARS1	100.0%	100.0%	100.0%	98.6%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625;Usher syndrome type 3B, 614504
HARS2	100.0%	100.0%	100.0%	98.9%	Perrault syndrome 2, 614926
HGF	100.0%	100.0%	100.0%	98.4%	Deafness, autosomal recessive 39, 608265
HOMER2	100.0%	99.9%	100.0%	98.5%	?Deafness, autosomal dominant 68, 616707
HSD17B4	100.0%	100.0%	100.0%	98.2%	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400
IFNLR1	100.0%	100.0%	100.0%	97.8%	
IKZF2	100.0%	100.0%	100.0%	99.0%	
ILDR1	100.0%	100.0%	100.0%	99.7%	Deafness, autosomal recessive 42, 609646

KARS1	100.0%	100.0%	100.0%	98.4%	Deafness, autosomal recessive 89, 613916;Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147;?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641;Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196
KCNE1	100.0%	100.0%	100.0%	99.7%	Jervell and Lange-Nielsen syndrome 2, 612347;Long QT syndrome 5, 613695
KCNJ10	100.0%	100.0%	100.0%	99.5%	Enlarged vestibular aqueduct, digenic, 600791;SESAME syndrome, 612780
KCNQ1	100.0%	99.8%	99.7%	96.2%	Short QT syndrome 2, 609621;Atrial fibrillation, familial, 3, 607554;Long QT syndrome 1, 192500;{Long QT syndrome 1, acquired, susceptibility to}, 192500;Jervell and Lange-Nielsen syndrome, 220400
KCNQ4	100.0%	99.7%	100.0%	96.2%	Deafness, autosomal dominant 2A, 600101

KITLG	100.0%	99.3%	100.0%	98.2%	Hyperpigmentation with or without hypopigmentation, 145250;Waardenburg syndrome, type 2F, 619947;Deafness, autosomal dominant 69, unilateral or asymmetric, 616697;[Skin/hair/eye pigmentation 7, blond/brown hair], 611664
LARS2	100.0%	100.0%	100.0%	99.1%	Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LHFPL5	100.0%	100.0%	100.0%	99.3%	Deafness, autosomal recessive 67, 610265
LMX1A	100.0%	100.0%	100.0%	99.4%	Deafness, autosomal dominant 7, 601412
LOXHD1	100.0%	100.0%	100.0%	98.9%	Deafness, autosomal recessive 77, 613079
LOXL3	100.0%	100.0%	100.0%	99.3%	Myopia 28, autosomal recessive, 619781
LRP2	100.0%	100.0%	100.0%	99.0%	Donnai-Barrow syndrome, 222448

LRP5	100.0%	100.0%	99.8%	98.2%	Osteopetrosis, autosomal dominant 1, 607634;[Bone mineral density variability 1], 601884;Polycystic liver disease 4 with or without kidney cysts, 617875;Endosteal hyperostosis, 144750;Osteoporosis-pseudoglioma syndrome, 259770;Exudative vitreoretinopathy 4, 601813
LRTOMT	100.0%	100.0%	100.0%	98.7%	Deafness, autosomal recessive 63, 611451
MAN2B1	100.0%	100.0%	100.0%	99.1%	Mannosidosis, alpha-, types I and II, 248500
MAP1B	100.0%	100.0%	100.0%	96.5%	?Deafness, autosomal dominant 83, 619808;Periventricular nodular heterotopia 9, 618918
MARVELD2	100.0%	100.0%	100.0%	98.4%	Deafness, autosomal recessive 49, 610153
MCM2	100.0%	100.0%	100.0%	99.2%	?Deafness, autosomal dominant 70, 616968

MET	100.0%	100.0%	100.0%	98.7%	Renal cell carcinoma, papillary, 1, familial and somatic, 605074;?Arthrogryposis, distal, type 11, 620019;Hepatocellular carcinoma, childhood type, somatic, 114550;{Osteofibrous dysplasia, susceptibility to}, 607278;?Deafness, autosomal recessive 97, 616705
MGP	100.0%	100.0%	100.0%	97.1%	Keutel syndrome, 245150
MIA3	100.0%	100.0%	99.9%	97.4%	?Ondontochondrodysplasia 2 with hearing loss and diabetes, 619269
MINAR2	100.0%	100.0%	100.0%	96.4%	Deafness, autosomal recessive 120, 620238
MIR96					Deafness, autosomal dominant 50, 613074
MITF	99.9%	99.7%	100.0%	98.5%	Waardenburg syndrome, type 2A, 193510;{Melanoma, cutaneous malignant, susceptibility to, 8}, 614456;Tietz albinism-deafness syndrome, 103500;COMMAD syndrome, 617306
MPDZ	99.5%	99.1%	100.0%	98.8%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219

MPZL2	100.0%	100.0%	100.0%	99.2%	Deafness, autosomal recessive 111, 618145
MRPL50	100.0%	100.0%	100.0%	99.2%	
MSRB3	100.0%	100.0%	99.9%	96.3%	Deafness, autosomal recessive 74, 613718
MYH14	100.0%	100.0%	100.0%	98.5%	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369;Deafness, autosomal dominant 4A, 600652
MYH9	97.2%	97.2%	100.0%	98.8%	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100;Deafness, autosomal dominant 17, 603622
MYO15A	100.0%	100.0%	100.0%	99.3%	Deafness, autosomal recessive 3, 600316
MYO3A	100.0%	100.0%	100.0%	97.7%	Deafness, autosomal recessive 30, 607101;Deafness, autosomal dominant 90, 620722

MYO6	100.0%	100.0%	100.0%	97.9%	Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346;Deafness, autosomal dominant 22, 606346;Deafness, autosomal recessive 37, 607821
MYO7A	100.0%	100.0%	100.0%	98.8%	Deafness, autosomal recessive 2, 600060;Usher syndrome, type 1B, 276900;Deafness, autosomal dominant 11, 601317
NARS2	92.3%	92.3%	100.0%	98.5%	Combined oxidative phosphorylation deficiency 24, 616239;?Deafness, autosomal recessive 94, 618434
NCOA3	100.0%	100.0%	100.0%	99.0%	
NDP	100.0%	100.0%	98.0%	72.5%	Exudative vitreoretinopathy 2, X-linked, 305390;Norrie disease, 310600
NHERF1	100.0%	100.0%	100.0%	97.2%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287

NLRP3	100.0%	100.0%	100.0%	98.8%	CINCA syndrome, 607115;Familial cold inflammatory syndrome 1, 120100;Keratoendothelitis fugax hereditaria, 148200;Deafness, autosomal dominant 34, with or without inflammation, 617772;Muckle-Wells syndrome, 191900
NOG	100.0%	100.0%	100.0%	96.7%	Symphalangism, proximal, 1A, 185800;Brachydactyly, type B2, 611377;Stapes ankylosis with broad thumbs and toes, 184460;Tarsal-carpal coalition syndrome, 186570;Multiple synostoses syndrome 1, 186500
OPA1	100.0%	100.0%	100.0%	98.5%	Optic atrophy plus syndrome, 125250;{Glaucoma, normal tension, susceptibility to}, 606657;Optic atrophy 1, 165500;Behr syndrome, 210000;?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
OSBPL2	100.0%	100.0%	100.0%	99.3%	Deafness, autosomal dominant 67, 616340
OTOA	100.0%	100.0%	100.0%	98.8%	Deafness, autosomal recessive 22, 607039

OTOF	100.0%	100.0%	100.0%	98.7%	Auditory neuropathy, autosomal recessive, 1, 601071;Deafness, autosomal recessive 9, 601071
OTOG	100.0%	100.0%	100.0%	99.4%	Deafness, autosomal recessive 18B, 614945
OTOGL	100.0%	100.0%	100.0%	98.8%	Deafness, autosomal recessive 84B, 614944
P2RX2	100.0%	100.0%	99.9%	93.8%	Deafness, autosomal dominant 41, 608224
PAX3	100.0%	99.8%	100.0%	98.5%	Craniofacial-deafness-hand syndrome, 122880;Waardenburg syndrome, type 3, 148820;Waardenburg syndrome, type 1, 193500;Rhabdomyosarcoma 2, alveolar, 268220
PBX1	100.0%	99.9%	100.0%	98.4%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCDH15	100.0%	100.0%	100.0%	98.6%	Usher syndrome, type 1D/F digenic, 601067;Deafness, autosomal recessive 23, 609533;Usher syndrome, type 1F, 602083
PDE1C	99.4%	98.9%	100.0%	99.0%	?Deafness, autosomal dominant 74, 618140

PDZD7	100.0%	99.2%	100.0%	98.6%	Deafness, autosomal recessive 57, 618003;{Retinal disease in Usher syndrome type IIA, modifier of}, 276901;Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472
PET100	100.0%	100.0%	100.0%	99.3%	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PEX1	100.0%	100.0%	100.0%	98.5%	Heimler syndrome 1, 234580;Peroxisome biogenesis disorder 1B (NALD/IRD), 601539;Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX26	100.0%	100.0%	100.0%	98.0%	Peroxisome biogenesis disorder 7B, 614873;Peroxisome biogenesis disorder 7A (Zellweger), 614872
PEX6	100.0%	100.0%	100.0%	97.9%	Peroxisome biogenesis disorder 4B, 614863;Peroxisome biogenesis disorder 4A (Zellweger), 614862;Heimler syndrome 2, 616617
PI4KB	100.0%	100.0%	100.0%	99.4%	Deafness, autosomal dominant 87, 620281
PISD	100.0%	100.0%	100.0%	99.8%	Liberfarb syndrome, 618889

PJVK	100.0%	100.0%	100.0%	98.8%	Deafness, autosomal recessive 59, 610220
PKHD1L1	100.0%	100.0%	100.0%	98.5%	Deafness, autosomal recessive 124, 620794
PLCG1	100.0%	100.0%	100.0%	98.0%	?Immune dysregulation, autoimmunity, and autoinflammation, 620514
PLOD3	100.0%	100.0%	100.0%	98.0%	BCARD syndrome (lysyl hydroxylase 3 deficiency), 612394
PLS1	100.0%	99.9%	100.0%	98.4%	Deafness, autosomal dominant 76, 618787
PLXNB2	100.0%	100.0%	100.0%	99.3%	
PNPT1	100.0%	100.0%	100.0%	98.3%	Spinocerebellar ataxia 25, 608703;Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934;Combined oxidative phosphorylation deficiency 13, 614932
POLD1	100.0%	100.0%	100.0%	99.2%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381;Immunodeficiency 120, 620836;{Colorectal cancer, susceptibility to, 10}, 612591

POLR1C	83.3%	83.2%	100.0%	99.1%	Leukodystrophy, hypomyelinating, 11, 616494;Treacher Collins syndrome 3, 248390
POLR1D	100.0%	100.0%	100.0%	98.8%	Treacher Collins syndrome 2, 613717
POU3F4	100.0%	100.0%	97.8%	68.8%	Deafness, X-linked 2, 304400
POU4F3	100.0%	100.0%	100.0%	99.5%	Deafness, autosomal dominant 15/52, 602459
PPIP5K2	100.0%	100.0%	100.0%	98.7%	Deafness, autosomal recessive 100, 618422
PRKCB	100.0%	99.9%	100.0%	97.1%	
PRORP	100.0%	100.0%	100.0%	97.9%	Combined oxidative phosphorylation deficiency 54, 619737
PRPS1	100.0%	100.0%	96.3%	69.8%	Arts syndrome, 301835;Phosphoribosylpyrophosphate synthetase superactivity, 300661;Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070;Deafness, X-linked 1, 304500;Gout, PRPS-related, 300661
PSIP1	100.0%	100.0%	100.0%	95.7%	

PTPRQ	91.9%	91.9%	100.0%	98.3%	Deafness, autosomal dominant 73, 617663;Deafness, autosomal recessive 84A, 613391
PTRH2	100.0%	100.0%	100.0%	98.8%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
RAI1	100.0%	100.0%	100.0%	98.9%	Smith-Magenis syndrome, 182290
RDX	100.0%	100.0%	100.0%	98.2%	Deafness, autosomal recessive 24, 611022
REST	100.0%	100.0%	100.0%	99.1%	Deafness, autosomal dominant 27, 612431;{Wilms tumor 6, susceptibility to}, 616806;Fibromatosis, gingival, 5, 617626
RIPOR2	100.0%	100.0%	100.0%	98.6%	Deafness, autosomal dominant 21, 607017;?Deafness, autosomal recessive 104, 616515
RMND1	85.6%	85.6%	100.0%	97.6%	Combined oxidative phosphorylation deficiency 11, 614922

ROBO1	100.0%	99.9%	100.0%	99.3%	Pituitary hormone deficiency, combined or isolated, 8, 620303;Neurooculorenal syndrome, 620305;?Nystagmus 8, congenital, autosomal recessive, 257400
ROR1	100.0%	100.0%	100.0%	99.0%	?Deafness, autosomal recessive 108, 617654
RRM2B	100.0%	100.0%	100.0%	97.7%	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075;Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075;Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
S1PR2	100.0%	100.0%	100.0%	99.9%	Deafness, autosomal recessive 68, 610419
SCD5	100.0%	100.0%	100.0%	97.1%	?Deafness, autosomal dominant 79, 619086

SERAC1	100.0%	100.0%	100.0%	98.3%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINB6	100.0%	100.0%	100.0%	98.7%	?Deafness, autosomal recessive 91, 613453
SEZ6	100.0%	100.0%	100.0%	98.8%	
SIX1	100.0%	100.0%	100.0%	97.2%	Deafness, autosomal dominant 23, 605192;Branchiootic syndrome 3, 608389
SIX5	100.0%	100.0%	99.8%	95.0%	Branchiootorenal syndrome 2, 610896
SLC12A1	96.4%	96.3%	100.0%	98.4%	Bartter syndrome, type 1, 601678
SLC12A2	100.0%	100.0%	100.0%	97.7%	Kilquist syndrome, 619080;Delpire-McNeill syndrome, 619083;Deafness, autosomal dominant 78, 619081
SLC17A8	100.0%	100.0%	100.0%	97.7%	Deafness, autosomal dominant 25, 605583
SLC19A2	100.0%	100.0%	100.0%	99.5%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC22A4	100.0%	100.0%	100.0%	98.0%	{Rheumatoid arthritis, susceptibility to}, 180300

SLC26A4	100.0%	100.0%	100.0%	97.9%	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791;Pendred syndrome, 274600
SLC26A5	100.0%	100.0%	100.0%	98.7%	?Deafness, autosomal recessive 61, 613865
SLC29A3	100.0%	100.0%	100.0%	99.5%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC30A9	100.0%	100.0%	100.0%	98.4%	Birk-Landau-Perez syndrome, 617595
SLC33A1	100.0%	100.0%	100.0%	97.5%	Spastic paraplegia 42, autosomal dominant, 612539;Huppke-Brendel syndrome, 614482
SLC44A4	100.0%	100.0%	100.0%	98.9%	?Deafness, autosomal dominant 72, 617606
SLC4A11	100.0%	100.0%	100.0%	99.2%	Corneal endothelial dystrophy, autosomal recessive, 217700;Corneal dystrophy, Fuchs endothelial, 4, 613268;Corneal endothelial dystrophy and perceptive deafness, 217400
SLC52A2	100.0%	100.0%	100.0%	99.9%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100.0%	100.0%	100.0%	99.0%	?Fazio-Londe disease, 211500;Brown-Vialetto-Van Laere syndrome 1, 211530

SLC7A14	100.0%	100.0%	100.0%	99.3%	Retinitis pigmentosa 68, 615725
SLITRK6	100.0%	100.0%	100.0%	98.0%	Deafness and myopia, 221200
SMPX	100.0%	99.1%	97.8%	68.9%	Myopathy, distal, 7, adult-onset, X-linked, 301075;Deafness, X-linked 4, 300066
SNAI2	100.0%	100.0%	100.0%	99.1%	
SOX10	97.8%	97.8%	100.0%	97.9%	Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SPNS2	100.0%	99.7%	99.9%	95.5%	?Deafness, autosomal recessive 115, 618457
STAG2	100.0%	100.0%	97.8%	69.9%	Holoprosencephaly 13, X-linked, 301043;Mullegama-Klein-Martinez syndrome, 301022
STRC	100.0%	100.0%	100.0%	98.6%	Deafness, autosomal recessive 16, 603720
STX4	100.0%	100.0%	100.0%	98.4%	?Deafness, autosomal recessive 123, 620745
SUCLA2	100.0%	99.6%	100.0%	98.8%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073

SYNE4	100.0%	100.0%	100.0%	98.7%	Deafness, autosomal recessive 76, 615540
TBC1D24	100.0%	100.0%	100.0%	99.6%	Deafness, autosomal recessive 86, 614617;Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105;Myoclonic epilepsy, infantile, familial, 605021;Deafness, autosomal dominant 65, 616044;Developmental and epileptic encephalopathy 16, 615338;DOORS syndrome, 220500
TBL1X	100.0%	99.9%	98.4%	72.9%	Hypothyroidism, congenital, nongoitrous, 8, 301033
TBL1Y	50.0%	49.4%	47.3%	20.6%	?Deafness, Y-linked 2, 400047
TCOF1	100.0%	100.0%	100.0%	99.0%	Treacher Collins syndrome 1, 154500
TECTA	100.0%	100.0%	100.0%	99.3%	Deafness, autosomal dominant 8/12, 601543;Deafness, autosomal recessive 21, 603629
TFAP2A	100.0%	100.0%	99.8%	93.8%	Branchiooculofacial syndrome, 113620
THOC1	100.0%	100.0%	100.0%	96.6%	?Deafness, autosomal dominant 86, 620280
TIMM8A	100.0%	99.5%	97.6%	65.5%	Mohr-Tranebjaerg syndrome, 304700

TJP2	100.0%	100.0%	99.9%	98.7%	Hypercholanemia, familial 1, 607748;Cholestasis, progressive familial intrahepatic 4, 615878
TMC1	100.0%	100.0%	100.0%	96.6%	Deafness, autosomal dominant 36, 606705;Deafness, autosomal recessive 7, 600974
TMEM132E	100.0%	100.0%	100.0%	99.0%	Deafness, autosomal recessive 99, 618481
TMEM43	100.0%	100.0%	100.0%	98.7%	Arrhythmogenic right ventricular dysplasia 5, 604400;Auditory neuropathy, autosomal dominant 3, 619832;Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMIE	100.0%	100.0%	100.0%	99.2%	Deafness, autosomal recessive 6, 600971
TMPRSS3	100.0%	100.0%	100.0%	99.3%	Deafness, autosomal recessive 8/10, 601072
TMTC2	97.2%	97.1%	100.0%	99.1%	
TMTC4	100.0%	100.0%	100.0%	98.4%	?Deafness, autosomal recessive 122, 620714
TNC	100.0%	100.0%	100.0%	99.2%	Deafness, autosomal dominant 56, 615629
TPRN	97.1%	95.4%	97.6%	80.2%	Deafness, autosomal recessive 79, 613307
TRIOBP	100.0%	100.0%	100.0%	98.0%	Deafness, autosomal recessive 28, 609823

TRRAP	100.0%	100.0%	100.0%	98.8%	?Deafness, autosomal dominant 75, 618778;Developmental delay with or without dysmorphic facies and autism, 618454
TSHZ1	100.0%	100.0%	99.9%	98.5%	Aural atresia, congenital, 607842
TUBB4B	100.0%	100.0%	100.0%	98.1%	Leber congenital amaurosis with early-onset deafness, 617879
TWNK	100.0%	100.0%	100.0%	99.8%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286;Perrault syndrome 5, 616138
TYR	100.0%	99.9%	100.0%	98.8%	[Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800;[Skin/hair/eye pigmentation 3, blue/green eyes], 601800;{Melanoma, cutaneous malignant, susceptibility to, 8}, 601800;Albinism, oculocutaneous, type IB, 606952;Albinism, oculocutaneous, type IA, 203100

USH1C	100.0%	100.0%	100.0%	97.3%	Usher syndrome, type 1C, 276904;Deafness, autosomal recessive 18A, 602092
USH1G	100.0%	100.0%	100.0%	99.6%	Usher syndrome, type 1G, 606943
USH2A	99.9%	99.6%	100.0%	99.4%	Usher syndrome, type 2A, 276901;Retinitis pigmentosa 39, 613809
USP48	100.0%	100.0%	100.0%	97.9%	Deafness, autosomal dominant 85, 620227
WBP2	100.0%	100.0%	100.0%	97.4%	Deafness, autosomal recessive 107, 617639
WFS1	91.2%	91.2%	100.0%	99.6%	Deafness, autosomal dominant 6/14/38, 600965;?Cataract 41, 116400;Wolfram-like syndrome, autosomal dominant, 614296;{Diabetes mellitus, noninsulin-dependent, association with}, 125853;Wolfram syndrome 1, 222300
WHRN	100.0%	100.0%	100.0%	99.1%	Deafness, autosomal recessive 31, 607084;Usher syndrome, type 2D, 611383
XKR8	100.0%	100.0%	100.0%	98.2%	
XYLT2	99.9%	99.2%	100.0%	98.9%	{Pseudoxanthoma elasticum, modifier of severity of}, 264800;Spondyloocular syndrome, 605822

YAP1	100.0%	100.0%	99.9%	97.5%	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or impaired intellectual development, 120433
YARS1	100.0%	100.0%	100.0%	97.5%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418;Charcot-Marie-Tooth disease, dominant intermediate C, 608323

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.
TWIST X2 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry.
TWIST X2 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry.
srWGS GRCh38 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS GRCh38 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : March 17th, 2023.
This list is accurate for panel version DG 4.0.0

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