HEARING IMPAIRMENT PANEL (INCLUDING GJB2) DG-5.1.0 (300 GENES)

Gene	*Twist X2 covered 10x*	*Twist X2 covered 20x*	*srWGS covered 10x*	*srWGS covered 15x*	*srWGS covered 20x*	*Associated Phenotype description and OMIM disease ID*
ABCC1	96.8%	96.8%	100%	100%	99.4%	?Deafness, autosomal dominant 77, 618915
ABHD12	100%	100%	100%	100%	99.6%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	100%	100%	100%	100%	99.9%	Chanarin-Dorfman syndrome, 275630
ACOX1	97.9%	97.9%	100%	100%	99.7%	Mitchell syndrome, 618960;Peroxisomal acyl-CoA oxidase deficiency, 264470
ACTB	100%	100%	100%	99.9%	99.4%	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, 620470
ACTG1	100%	100%	100%	99.9%	98.9%	Deafness, autosomal dominant 20/26, 604717;Baraitser-Winter syndrome 2, 614583
ADAMTS1	100%	100%	100%	100%	99.7%	
ADCY1	99.2%	98.6%	100%	99.9%	98.8%	?Deafness, autosomal recessive 44, 610154

ADGRV1	100%	100%	100%	100%	99.8%	Usher syndrome, type 2C, 605472;Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472;?Febrile seizures, familial, 4, 604352
AFG2A	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577
AFG2B	100%	100%	100%	100%	99.8%	Deafness, autosomal recessive 119, 619615;Neurodevelopmental disorder with hearing loss and spasticity, 619616
AGBL5	100%	100%	100%	100%	99.3%	Retinitis pigmentosa 75, 617023
AIFM1	100%	100%	99.3%	90.3%	72.6%	Combined oxidative phosphorylation deficiency 6, 300816;Cowchock syndrome, 310490;Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232;Deafness, X-linked 5, 300614
ALMS1	100%	100%	100%	100%	99.8%	Alstrom syndrome, 203800
AMMECR1	99.9%	97.7%	99.1%	88.8%	71.2%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
ANLN	98.2%	98.2%	100%	100%	99.7%	Focal segmental glomerulosclerosis 8, 616032
AP1B1	93.7%	91.9%	100%	100%	99.6%	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150

ARSG	100%	100%	100%	100%	99.1%	Usher syndrome, type IV, 618144
ATOH1	100%	100%	100%	100%	99.6%	?Deafness, autosomal dominant 89, 620284
ATP11A	100%	100%	100%	100%	99.7%	?Auditory neuropathy, autosomal dominant 2, 620384;?Leukodystrophy, hypomyelinating, 24, 619851;Deafness, autosomal dominant 84, 619810
ATP1A1	97.2%	95.4%	100%	100%	99.6%	Hypomagnesemia, seizures, and impaired intellectual development 2, 618314;Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP1A3	100%	100%	100%	100%	99.2%	Alternating hemiplegia of childhood 2, 614820;Dystonia-12, 128235;CAPOS syndrome, 601338;Developmental and epileptic encephalopathy 99, 619606
ATP2B2	100%	100%	100%	100%	99.2%	Deafness, autosomal dominant 82, 619804
ATP6V0A4	100%	100%	100%	99.9%	99%	Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722
ATP6V1B1	100%	100%	100%	100%	99.5%	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300
ATP6V1B2	94.3%	94.3%	100%	100%	99.8%	Zimmermann-Laband syndrome 2, 616455;Deafness, congenital, with onychodystrophy, autosomal dominant, 124480
ATP6V1C1	100%	100%	100%	100%	100%	

BCAP31	91.5%	86.7%	98.7%	88.7%	70.4%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCS1L	100%	100%	100%	100%	99.6%	GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Bjornstad syndrome, 262000
BDP1	100%	100%	100%	100%	99.8%	?Deafness, autosomal recessive 112, 618257
BICD1	100%	100%	100%	100%	99.4%	
BMP4	100%	100%	100%	100%	99.5%	Orofacial cleft 11, 600625;Microphthalmia, syndromic 6, 607932
BSND	100%	100%	100%	100%	99.8%	Sensorineural deafness with mild renal dysfunction, 602522;Bartter syndrome, type 4a, 602522
BTD	94.2%	94.2%	100%	100%	99.9%	Biotinidase deficiency, 253260
CABP2	100%	100%	100%	99.7%	97.3%	Deafness, autosomal recessive 93, 614899
CACNA1D	100%	100%	100%	100%	99.7%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474;Sinoatrial node dysfunction and deafness, 614896
CCDC50	94.8%	94.8%	100%	100%	99.7%	?Deafness, autosomal dominant 44, 607453
CD151	100%	100%	100%	100%	99.7%	[Blood group, Raph], 179620;Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057
CD164	100%	100%	100%	100%	100%	?Deafness, autosomal dominant 66, 616969

CDC14A	96.9%	96.9%	100%	100%	99.8%	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDC42	100%	100%	100%	100%	99.6%	Takenouchi-Kosaki syndrome, 616737
CDH23	100%	100%	100%	100%	99.4%	Usher syndrome, type 1D, 601067;{Pituitary adenoma 5, multiple types}, 617540;Usher syndrome, type 1D/F digenic, 601067;Deafness, autosomal recessive 12, 601386
CDK9	100%	100%	100%	100%	99.2%	
CEACAM16	100%	100%	100%	100%	99.6%	Deafness, autosomal dominant 4B, 614614;Deafness, autosomal recessive 113, 618410
CEP250	100%	100%	100%	100%	99.5%	Cone-rod dystrophy and hearing loss 2, 618358
CEP78	100%	100%	100%	100%	99.6%	Cone-rod dystrophy and hearing loss, 617236
CGN	100%	100%	100%	100%	99.5%	
CHD4	100%	100%	100%	100%	99.5%	Sifrim-Hitz-Weiss syndrome, 617159
CHD7	100%	100%	100%	100%	99.8%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800
CHSY1	100%	99.9%	100%	99.8%	99.1%	Temtamy preaxial brachydactyly syndrome, 605282
CIB2	100%	100%	100%	100%	99.9%	Deafness, autosomal recessive 48, 609439
CISD2	100%	100%	100%	99.9%	99.4%	Wolfram syndrome 2, 604928

CLDN14	100%	100%	100%	100%	99.4%	Deafness, autosomal recessive 29, 614035
CLDN9	100%	100%	100%	100%	99.2%	Deafness, autosomal recessive 116, 619093
CLIC5	100%	100%	100%	100%	99.9%	?Deafness, autosomal recessive 103, 616042
CLPP	100%	100%	100%	100%	98.9%	Perrault syndrome 3, 614129
CLRN1	100%	100%	100%	99.9%	99.1%	Usher syndrome, type 3A, 276902;Retinitis pigmentosa 61, 614180
CLRN2	100%	100%	100%	100%	99.6%	Deafness, autosomal recessive 117, 619174
COA8	100%	100%	100%	100%	99.7%	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COCH	100%	100%	100%	100%	99.9%	Deafness, autosomal dominant 9, 601369;?Deafness, autosomal recessive 110, 618094
COL11A1	100%	100%	100%	100%	99.8%	Fibrochondrogenesis 1, 228520;Stickler syndrome, type II, 604841;Marshall syndrome, 154780;Deafness, autosomal dominant 37, 618533;{Lumbar disc herniation, susceptibility to}, 603932

COL11A2	100%	100%	100%	100%	99.1%	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%	100%	100%	100%	99.5%	?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;Spondylometaphyseal dysplasia, Algerian type, 184253;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%	100%	100%	100%	99.6%	Alport syndrome 3A, autosomal dominant, 104200;Hematuria, benign familial, 2, 620320;Alport syndrome 3B, autosomal recessive, 620536

COL4A4	99.7%	98.9%	100%	100%	99.7%	Hematuria, familial benign, 1, 141200;Alport syndrome 2, autosomal recessive, 203780
COL4A5	100%	100%	99.2%	91.7%	73%	Alport syndrome 1, X-linked, 301050
COL4A6	100%	100%	98.9%	90%	71.4%	?Deafness, X-linked 6, 300914
COL9A1	100%	100%	100%	100%	99.7%	Stickler syndrome, type IV, 614134;?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	100%	100%	100%	100%	99.3%	Epiphyseal dysplasia, multiple, 2, 600204;?Stickler syndrome, type V, 614284
COL9A3	100%	100%	100%	100%	99.4%	{Intervertebral disc disease, susceptibility to}, 603932;Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969;Stickler syndrome, type VI, 620022
CRYL1	100%	100%	100%	100%	99%	
CRYM	96.3%	93.1%	100%	100%	99.3%	Deafness, autosomal dominant 40, 616357
DAP3	91.7%	90.8%	100%	100%	99.6%	Perrault syndrome 7, 621101
DCAF17	96.5%	96.5%	100%	99.9%	99.7%	Woodhouse-Sakati syndrome, 241080
DCDC2	100%	100%	100%	100%	99.8%	Nephronophthisis 19, 616217;?Deafness, autosomal recessive 66, 610212;Sclerosing cholangitis, neonatal, 617394

DHX16	100%	100%	100%	100%	99.4%	Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733
DIABLO	90.8%	90.8%	100%	100%	99.4%	Deafness, autosomal dominant 64, 614152
DIAPH1	100%	100%	100%	100%	99.5%	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900;Seizures, cortical blindness, microcephaly syndrome, 616632
DIAPH3	100%	100%	100%	100%	99.8%	Auditory neuropathy, autosomal dominant 1, 609129
DKC1	100%	100%	99%	89.5%	70.6%	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000
DLL1	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709
DLX5	100%	100%	100%	100%	99.2%	Split-hand/foot malformation 1, 183600;?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DMXL2	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 81, 618663;?Deafness, autosomal dominant 71, 617605;?Polyendocrine-polyneuropathy syndrome, 616113

DSPP	100%	100%	99.1%	98.3%	97%	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%	100%	100%	99.9%	99.4%	Waardenburg syndrome, type 4B, 613265;{Hirschsprung disease, susceptibility to, 4}, 613712
EDNRB	100%	100%	100%	100%	99.5%	{Hirschsprung disease, susceptibility to, 2}, 600155;?ABCD syndrome, 600501;Waardenburg syndrome, type 4A, 277580
EFNB2	100%	100%	100%	100%	99.6%	
EHD1	100%	100%	100%	100%	99.2%	
ELMOD3	100%	100%	100%	100%	99.8%	?Deafness, autosomal recessive 88, 615429;?Deafness, autosomal dominant 81, 619500
ELOVL1	100%	100%	100%	100%	99.9%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
EPHA10	100%	100%	100%	99.9%	99%	?Deafness, autosomal dominant 88, 620283
EPS8	95.3%	95.3%	100%	100%	99.9%	?Deafness, autosomal recessive 102, 615974
EPS8L2	100%	100%	100%	99.9%	99.1%	Deafness autosomal recessive 106, 617637
ERAL1	100%	100%	100%	99.9%	99.3%	Perrault syndrome 6, 617565

ESPN	100%	100%	100%	99.8%	98.2%	Deafness, autosomal recessive 36, 609006;Deafness, autosomal dominant 91, 621556;?Usher syndrome, type 1M, 618632
ESRP1	100%	100%	100%	100%	99.7%	?Deafness, autosomal recessive 109, 618013
ESRRB	100%	100%	100%	100%	99.7%	Deafness, autosomal recessive 35, 608565
EXOSC2	91.1%	91%	100%	100%	99.7%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EYA1	100%	100%	100%	100%	100%	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%	100%	100%	100%	99.9%	?Cardiomyopathy, dilated, 1J, 605362;Deafness, autosomal dominant 10, 601316
FDXR	100%	100%	100%	100%	99.4%	Multiple mitochondrial dysfunctions syndrome 9B, 620887;Auditory neuropathy and optic atrophy, 617717
FGF3	100%	100%	100%	99.8%	97.8%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706

FGFR3	100%	100%	100%	99.9%	99.1%	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%	100%	100%	100%	99.2%	Siddiqi syndrome, 618635
FOXC1	100%	100%	100%	99.4%	94.5%	Axenfeld-Rieger syndrome, type 3, 602482;Anterior segment dysgenesis 3, multiple subtypes, 601631
FOXF2	99.9%	99.4%	100%	99.7%	97.4%	
FOXI1	100%	100%	100%	100%	99.8%	Enlarged vestibular aqueduct, 600791
FRMPD4	100%	100%	98.7%	88.4%	70.1%	Intellectual developmental disorder, X-linked 104, 300983
GAB1	100%	100%	100%	100%	99.8%	?Deafness, autosomal recessive 26, 605428
GAS2	100%	100%	100%	100%	99.9%	?Deafness, autosomal recessive 125, 620877

GATA3	100%	100%	100%	100%	98.6%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GIPC3	100%	100%	100%	100%	99%	Deafness, autosomal recessive 15, 601869
GJB2	100%	100%	100%	100%	99.6%	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%	100%	100%	100%	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%	100%	100%	99.9%	99.8%	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%	99.3%	91.7%	72.9%	Fabry disease, cardiac variant, 301500;Fabry disease, 301500

GOSR2	100%	100%	100%	100%	99.5%	Epilepsy, progressive myoclonic 6, 614018;Muscular dystrophy, congenital, with or without seizures, 620166
GPR156	100%	100%	100%	100%	99.6%	Deafness, autosomal recessive 121, 620551
GPRASP2	100%	100%	98.8%	89.2%	72.5%	?Deafness, X-linked 7, 301018
GPSM2	95.5%	95.5%	100%	100%	99.8%	Chudley-McCullough syndrome, 604213
GRAP	100%	100%	100%	99.7%	99.5%	Deafness, autosomal recessive 114, 618456
GREB1L	100%	100%	100%	100%	99.7%	Deafness, autosomal dominant 80, 619274;Renal hypodysplasia/aplasia 3, 617805
GRHL2	100%	100%	100%	100%	99.8%	Deafness, autosomal dominant 28, 608641;Ectodermal dysplasia/short stature syndrome, 616029;Corneal dystrophy, posterior polymorphous, 4, 618031
GRXCR1	100%	100%	100%	100%	99.5%	Deafness, autosomal recessive 25, 613285
GRXCR2	100%	100%	100%	100%	99.6%	?Deafness, autosomal recessive 101, 615837
GSDME	100%	100%	100%	100%	99.7%	Deafness, autosomal dominant 5, 600994
HARS1	93.2%	93.2%	100%	99.9%	99.9%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625;Usher syndrome type 3B, 614504
HARS2	100%	100%	100%	99.9%	98.9%	Perrault syndrome 2, 614926
HGF	100%	100%	100%	100%	99.4%	Deafness, autosomal recessive 39, 608265

HOMER2	93.8%	93.8%	100%	100%	99.8%	?Deafness, autosomal dominant 68, 616707
HOXA2	100%	100%	100%	99.6%	98%	Microtia with or without hearing impairment (AD), 612290;?Microtia, hearing impairment, and cleft palate (AR), 612290
HSD17B4	100%	100%	100%	100%	99.9%	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400
IFNLR1	100%	100%	100%	99.9%	99.5%	
IKZF2	100%	100%	100%	100%	99.9%	Immunodysregulation, craniofacial anomalies, hearing impairment, athelia, and developmental delay, 621234;Immunodysregulation with variable immunodeficiency and autoimmunity, 621233
ILDR1	100%	100%	100%	100%	99.8%	Deafness, autosomal recessive 42, 609646
JAG1	100%	100%	100%	100%	99.6%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992;Charcot-Marie-Tooth disease, axonal, type 2HH, 619574;Alagille syndrome 1, 118450;Tetralogy of Fallot, 187500
KARS1	94.3%	94.3%	100%	100%	99.8%	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%	100%	100%	100%	100%	Jervell and Lange-Nielsen syndrome 2, 612347;Long QT syndrome 5, 613695
KCNJ10	100%	100%	100%	100%	99.7%	Enlarged vestibular aqueduct, digenic, 600791;SESAME syndrome, 612780
KCNQ1	100%	100%	100%	99.9%	99.3%	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	95.3%	93%	100%	99.9%	98.9%	Deafness, autosomal dominant 2A, 600101
KITLG	100%	100%	100%	100%	99.9%	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	96.2%	96.2%	100%	100%	99.8%	Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LHFPL5	100%	100%	100%	99.8%	99.2%	Deafness, autosomal recessive 67, 610265
LMX1A	100%	100%	100%	99.9%	99.6%	Deafness, autosomal dominant 7, 601412

LMX1B	100%	100%	100%	100%	99%	Focal segmental glomerulosclerosis 10, 256020;Nail-patella syndrome, 161200
LOXHD1	100%	100%	100%	100%	99.7%	Deafness, autosomal recessive 77, 613079
LOXL3	100%	100%	100%	100%	99.3%	Myopia 28, autosomal recessive, 619781
LRP2	100%	100%	100%	100%	99.8%	Donnai-Barrow syndrome, 222448
LRP5	100%	100%	100%	100%	99%	Osteopetrosis, autosomal dominant 1, 607634;Polycystic liver disease 4 with or without kidney cysts, 617875;Endosteal hyperostosis, 144750;Osteoporosis-pseudoglioma syndrome, 259770;[Bone mineral density variability 1, high bone mass], 601884;Exudative vitreoretinopathy 4, 601813
LRTOMT	100%	100%	100%	100%	99.7%	Deafness, autosomal recessive 63, 611451
MAN2B1	100%	100%	100%	100%	99.1%	Mannosidosis, alpha-, types I and II, 248500
MANBA	99.4%	97.5%	100%	100%	99.5%	Mannosidosis, beta, 248510
MAP1B	100%	100%	100%	100%	99.6%	?Deafness, autosomal dominant 83, 619808;Periventricular nodular heterotopia 9, 618918
MAP3K1	100%	100%	100%	99.9%	99.5%	46XY sex reversal 6, 613762
MARVELD2	100%	100%	100%	100%	99.8%	Deafness, autosomal recessive 49, 610153
MCM2	100%	100%	100%	100%	99.4%	?Deafness, autosomal dominant 70, 616968

MET	100%	100%	100%	100%	99.8%	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%	100%	100%	100%	99.7%	Keutel syndrome, 245150
MIA3	100%	100%	100%	100%	99.7%	?Odontochondrodysplasia 2 with hearing loss and diabetes, 619269
MINAR2	100%	100%	100%	100%	99.8%	Deafness, autosomal recessive 120, 620238
MIR96						Deafness, autosomal dominant 50, 613074
MITF	94.7%	94.7%	100%	100%	99.9%	Waardenburg syndrome, type 2A, 193510;{Melanoma, cutaneous malignant, susceptibility to, 8}, 614456;Tietz albinism-deafness syndrome, 103500;COMMAD syndrome, 617306
MPDZ	100%	100%	100%	100%	99.7%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219

MPZ	100%	100%	100%	100%	98.9%	Charcot-Marie-Tooth disease, type 2I, 607677;Dejerine-Sottas disease, 145900;Charcot-Marie-Tooth disease, type 1B, 118200;Roussy-Levy syndrome, 180800;Charcot-Marie-Tooth disease, dominant intermediate D, 607791;Hypomyelinating neuropathy, congenital, 2, 618184;Charcot-Marie-Tooth disease, type 2J, 607736
MPZL2	87.9%	87.9%	100%	100%	99.4%	Deafness, autosomal recessive 111, 618145
MRPL49	100%	100%	100%	100%	99.3%	Combined oxidative phosphorylation deficiency 60, 621195
MRPL50	100%	100%	100%	99.6%	98.7%	
MSRB3	100%	100%	100%	100%	99.9%	Deafness, autosomal recessive 74, 613718
MYH14	100%	100%	100%	99.9%	98.8%	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369;Deafness, autosomal dominant 4A, 600652
MYH7B	100%	100%	100%	100%	99.2%	
MYH9	97.3%	97%	100%	100%	99.4%	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100;Deafness, autosomal dominant 17, 603622
MYO15A	100%	100%	100%	100%	99.3%	Deafness, autosomal recessive 3, 600316

MYO3A	100%	100%	100%	100%	99.8%	Deafness, autosomal recessive 30, 607101;Deafness, autosomal dominant 90, 620722
MYO6	100%	100%	100%	100%	99.9%	Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346;Deafness, autosomal dominant 22, 606346;Deafness, autosomal recessive 37, 607821
MYO7A	100%	100%	100%	100%	99.5%	Deafness, autosomal recessive 2, 600060;Usher syndrome, type 1B, 276900;Deafness, autosomal dominant 11, 601317
NARS2	92.3%	92.3%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 24, 616239;?Deafness, autosomal recessive 94, 618434
NCOA3	100%	100%	100%	100%	99.8%	
NDP	100%	100%	98.2%	90.3%	76.4%	Exudative vitreoretinopathy 2, X-linked, 305390;Norrie disease, 310600
NHERF1	85.8%	85.8%	100%	100%	99.2%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
NLRP3	100%	100%	100%	100%	99.6%	CINCA syndrome, 607115;Familial cold inflammatory syndrome 1, 120100;Keratoendotheliitis fugax hereditaria, 148200;Deafness, autosomal dominant 34, with or without inflammation, 617772;Muckle-Wells syndrome, 191900

NOG	100%	100%	100%	99.7%	98.3%	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
NOP10	92.5%	92.5%	100%	100%	99.9%	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 9, 620400;?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 2, 620425;?Dyskeratosis congenita, autosomal recessive 1, 224230
OPA1	94.7%	94.7%	100%	100%	99.7%	Optic atrophy plus syndrome, 125250;{Glaucoma, normal tension, susceptibility to}, 606657;Optic atrophy 1, 165500;Behr syndrome, 210000;Mitochondrial DNA depletion syndrome 14A (encephalomyopathic type), 621481;?Mitochondrial DNA depletion syndrome 14B (cardioencephalomyopathic type), 616896
OSBPL2	100%	100%	100%	100%	99.4%	Dyschromatosis, ichthyosis, deafness, and atopic disease, 621400;Deafness, autosomal dominant 67, 616340
OTOA	100%	100%	100%	99.9%	99.3%	Deafness, autosomal recessive 22, 607039

OTOF	100%	100%	100%	100%	99.3%	Auditory neuropathy, autosomal recessive, 1, 601071;Deafness, autosomal recessive 9, 601071
OTOG	100%	100%	100%	99.9%	99.3%	Deafness, autosomal recessive 18B, 614945
OTOGL	100%	100%	100%	100%	99.7%	Deafness, autosomal recessive 84B, 614944
P2RX2	100%	100%	100%	99.9%	98.4%	Deafness, autosomal dominant 41, 608224
PAX3	100%	100%	100%	100%	99.6%	Craniofacial-deafness-hand syndrome, 122880;Waardenburg syndrome, type 3, 148820;Waardenburg syndrome, type 1, 193500;Rhabdomyosarcoma 2, alveolar, 268220
PBX1	100%	100%	100%	100%	99.7%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCDH15	100%	100%	100%	100%	99.6%	Usher syndrome, type 1D/F digenic, 601067;Deafness, autosomal recessive 23, 609533;Usher syndrome, type 1F, 602083
PDE1C	100%	100%	100%	100%	99.8%	?Deafness, autosomal dominant 74, 618140
PDZD7	100%	99.8%	100%	100%	99.3%	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%	100%	100%	100%	99.2%	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PEX1	100%	100%	100%	100%	99.9%	Heimler syndrome 1, 234580;Peroxisome biogenesis disorder 1B (NALD/IRD), 601539;Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX11B	100%	100%	100%	99.9%	99.1%	Peroxisome biogenesis disorder 14B, 614920
PEX26	91.9%	91.7%	100%	99.9%	99.1%	Peroxisome biogenesis disorder 7B, 614873;Peroxisome biogenesis disorder 7A (Zellweger), 614872
PEX6	100%	100%	100%	100%	99.1%	Peroxisome biogenesis disorder 4B, 614863;Peroxisome biogenesis disorder 4A (Zellweger), 614862;Heimler syndrome 2, 616617
PI4KB	100%	100%	100%	100%	99.3%	Deafness, autosomal dominant 87, 620281
PISD	100%	100%	100%	99.9%	99.5%	Liberfarb syndrome, 618889
PJVK	100%	100%	100%	100%	99.9%	Deafness, autosomal recessive 59, 610220
PKHD1L1	100%	100%	100%	100%	99.8%	Deafness, autosomal recessive 124, 620794
PLCG1	100%	100%	100%	100%	99.6%	?Immune dysregulation, autoimmunity, and autoinflammation, 620514
PLOD3	98.5%	96.8%	100%	99.8%	98.9%	BCARD syndrome (lysyl hydroxylase 3 deficiency), 612394
PLS1	100%	100%	100%	100%	99.7%	Deafness, autosomal dominant 76, 618787
PLXNB2	100%	100%	100%	100%	99.1%	

PNPT1	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia 25, 608703;Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934;Combined oxidative phosphorylation deficiency 13, 614932
POLD1	100%	100%	100%	99.9%	99%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381;Immunodeficiency 120, 620836;{Colorectal cancer, susceptibility to, 10}, 612591
POLR1C	83.6%	83.5%	100%	100%	99.2%	Leukodystrophy, hypomyelinating, 11, 616494;Treacher Collins syndrome 3, 248390
POLR1D	100%	100%	100%	100%	99.8%	Treacher Collins syndrome 2, 613717
POU3F4	100%	100%	98%	86%	66.8%	Deafness, X-linked 2, 304400
POU4F3	100%	100%	100%	100%	99.6%	Deafness, autosomal dominant 15/52, 602459
PPIP5K2	100%	100%	100%	100%	99.6%	Deafness, autosomal recessive 100, 618422
PRKCB	96.4%	96.4%	100%	100%	99.7%	
PRORP	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 54, 619737

PRPS1	100%	100%	99%	89.8%	71.8%	Arts syndrome, 301835;Phosphoribosyl pyrophosphate synthetase superactivity, 300661;Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070;Deafness, X-linked 1, 304500;Gout, PRPS-related, 300661
PSIP1	100%	100%	100%	100%	99.9%	
PTPRQ	100%	100%	100%	99.9%	99.7%	Deafness, autosomal dominant 73, 617663;Deafness, autosomal recessive 84A, 613391
PTRH2	100%	100%	100%	100%	99.9%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
RAI1	100%	100%	100%	100%	99%	Smith-Magenis syndrome, 182290
RDX	100%	100%	100%	100%	99.9%	Deafness, autosomal recessive 24, 611022
REST	100%	100%	100%	99.9%	99.5%	Deafness, autosomal dominant 27, 612431;{Wilms tumor 6, susceptibility to}, 616806;Fibromatosis, gingival, 5, 617626
RIPOR2	100%	100%	100%	100%	99.8%	Deafness, autosomal dominant 21, 607017;?Deafness, autosomal recessive 104, 616515
RMND1	85.8%	85.8%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 11, 614922

ROBO1	100%	100%	100%	100%	99.6%	Pituitary hormone deficiency, combined or isolated, 8, 620303;Neurooculorenal syndrome, 620305;?Nystagmus 8, congenital, autosomal recessive, 257400
ROR1	100%	100%	100%	100%	99.8%	?Deafness, autosomal recessive 108, 617654
RRM2B	100%	100%	100%	100%	99.9%	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%	100%	100%	100%	99.7%	Deafness, autosomal recessive 68, 610419
SALL1	100%	100%	100%	100%	99.2%	Townes-Brocks syndrome 1, 107480;Townes-Brocks branchiootorenal-like syndrome, 107480
SCD5	100%	100%	100%	100%	99.5%	?Deafness, autosomal dominant 79, 619086
SEMA3F	100%	100%	100%	99.9%	99.1%	
SERAC1	100%	100%	100%	100%	99.9%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739

SERPINB6	91%	90.7%	100%	100%	99.5%	?Deafness, autosomal recessive 91, 613453
SEZ6	100%	100%	100%	99.9%	99.3%	
SIX1	100%	100%	100%	100%	98.8%	Deafness, autosomal dominant 23, 605192;Branchiootic syndrome 3, 608389
SIX5	100%	100%	100%	100%	99.4%	Branchiootorenal syndrome 2, 610896
SLC12A1	96.3%	96.3%	100%	100%	99.8%	Bartter syndrome, type 1, 601678
SLC12A2	100%	100%	100%	100%	99.6%	Kilquist syndrome, 619080;Delpire-McNeill syndrome, 619083;Deafness, autosomal dominant 78, 619081
SLC17A8	100%	100%	100%	100%	99.8%	Deafness, autosomal dominant 25, 605583
SLC19A2	100%	100%	100%	100%	99.7%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC22A4	100%	100%	100%	100%	99.5%	{Rheumatoid arthritis, susceptibility to}, 180300
SLC26A4	100%	100%	100%	100%	99.8%	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791;Pendred syndrome, 274600
SLC26A5	100%	100%	100%	100%	99.9%	?Deafness, autosomal recessive 61, 613865
SLC29A3	100%	100%	100%	100%	99.5%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC30A9	100%	100%	100%	100%	99.9%	Birk-Landau-Perez syndrome, 617595
SLC33A1	92.6%	92.6%	100%	100%	99.7%	Spastic paraplegia 42, autosomal dominant, 612539;Huppke-Brendel syndrome, 614482

SLC44A4	100%	100%	100%	99.9%	99.2%	?Deafness, autosomal dominant 72, 617606
SLC4A11	100%	100%	100%	100%	99.6%	Corneal endothelial dystrophy, autosomal recessive, 217700;Corneal dystrophy, Fuchs endothelial, 4, 613268;Corneal endothelial dystrophy and perceptive deafness, 217400
SLC52A2	100%	100%	100%	99.9%	99.7%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100%	100%	100%	100%	99.4%	?Fazio-Londe disease, 211500;Brown-Vialetto-Van Laere syndrome 1, 211530
SLC7A14	100%	100%	100%	99.9%	99.6%	Retinitis pigmentosa 68, 615725
SLC9A1	100%	100%	100%	99.9%	99.1%	Lichtenstein-Knorr syndrome, 616291
SLITRK6	100%	100%	100%	100%	99.7%	Deafness and myopia, 221200
SMPX	100%	100%	98.9%	89.7%	69.6%	Myopathy, distal, 7, adult-onset, X-linked, 301075;Deafness, X-linked 4, 300066
SNAI2	100%	100%	100%	100%	99.7%	
SOX10	97.8%	97.8%	100%	100%	99.2%	Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SPNS2	100%	100%	100%	99.8%	99%	?Deafness, autosomal recessive 115, 618457

STAG2	100%	100%	99.3%	91.5%	74%	Holoprosencephaly 13, X-linked, 301043;Mullegama-Klein-Martinez syndrome, 301022
STRC	100%	100%	100%	99.9%	99.2%	Deafness, autosomal recessive 16, 603720
STX4	100%	100%	100%	99.9%	99%	?Deafness, autosomal recessive 123, 620745
SUCLA2	96.3%	96.3%	100%	100%	99.9%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SYNE4	100%	99.7%	100%	100%	99.7%	Deafness, autosomal recessive 76, 615540
TBC1D24	100%	100%	100%	100%	99.4%	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%	100%	98.6%	88.7%	70.4%	Hypothyroidism, congenital, nongoitrous, 8, 301033
TBL1Y	50%	50%	48.1%	37.9%	20.5%	?Deafness, Y-linked 2, 400047
TCOF1	97.9%	97.9%	100%	100%	99.6%	Treacher Collins syndrome 1, 154500

TECTA	100%	100%	100%	100%	99.7%	Deafness, autosomal dominant 8/12, 601543;Deafness, autosomal recessive 21, 603629
TFAP2A	100%	100%	100%	100%	98.8%	Branchiooculofacial syndrome, 113620
THBS1	100%	100%	100%	100%	99.6%	
THOC1	100%	100%	100%	100%	99.8%	?Deafness, autosomal dominant 86, 620280
TIMM8A	100%	100%	98.2%	87.9%	71.6%	Mohr-Tranebjaerg syndrome, 304700
TJP2	97.1%	95.8%	100%	100%	99.8%	Hypercholanemia, familial 1, 607748;Cholestasis, progressive familial intrahepatic 4, 615878
TMC1	100%	100%	100%	100%	99.7%	Deafness, autosomal dominant 36, 606705;Deafness, autosomal recessive 7, 600974
TMEM132E	100%	100%	100%	100%	99.6%	Deafness, autosomal recessive 99, 618481
TMEM43	100%	100%	100%	100%	99.5%	Arrhythmogenic right ventricular dysplasia 5, 604400;Auditory neuropathy, autosomal dominant 3, 619832;Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMIE	100%	100%	100%	99.9%	99.4%	Deafness, autosomal recessive 6, 600971
TMPRSS3	100%	100%	100%	100%	99.5%	Deafness, autosomal recessive 8/10, 601072
TMTC2	97.3%	97.1%	100%	100%	99.7%	
TMTC4	100%	100%	100%	100%	99.9%	?Deafness, autosomal recessive 122, 620714
TNC	100%	100%	100%	100%	99.6%	Deafness, autosomal dominant 56, 615629

TPRN	97.4%	95.3%	100%	99.7%	96.6%	Deafness, autosomal recessive 79, 613307
TRIM71	100%	100%	100%	100%	98.9%	Hydrocephalus, congenital, 4, 618667
TRIOBP	100%	100%	100%	99.9%	99.1%	Deafness, autosomal recessive 28, 609823
TRRAP	100%	100%	100%	100%	99.5%	?Deafness, autosomal dominant 75, 618778;Developmental delay with or without dysmorphic facies and autism, 618454
TSHZ1	100%	100%	100%	99.9%	99.4%	Aural atresia, congenital, 607842
TUBB4B	100%	100%	100%	100%	99.3%	Leber congenital amaurosis with early-onset deafness, 617879
TWNK	100%	100%	100%	100%	99.6%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286;Perrault syndrome 5, 616138
USH1C	100%	100%	100%	100%	99.6%	Usher syndrome, type 1C, 276904;Deafness, autosomal recessive 18A, 602092
USH1G	100%	100%	100%	100%	99.1%	Usher syndrome, type 1G, 606943
USH2A	100%	100%	100%	100%	99.7%	Usher syndrome, type 2A, 276901;Retinitis pigmentosa 39, 613809
USP48	100%	100%	100%	100%	99.9%	Deafness, autosomal dominant 85, 620227
WBP2	100%	100%	100%	100%	99.3%	Deafness, autosomal recessive 107, 617639

WFS1	91.5%	90.5%	100%	100%	99.4%	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%	100%	100%	100%	99.4%	Deafness, autosomal recessive 31, 607084;Usher syndrome, type 2D, 611383
XKR8	100%	100%	100%	100%	99.6%	
XYLT2	99.9%	98.8%	100%	99.9%	99.5%	{Pseudoxanthoma elasticum, modifier of severity of}, 264800;Spondyloocular syndrome, 605822
YAP1	92.3%	92.3%	100%	100%	99.6%	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or impaired intellectual development, 120433
YARS1	100%	98.5%	100%	100%	99.5%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418;Charcot-Marie-Tooth disease, dominant intermediate C, 608323

Gene symbols used follow HGNC 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 mapped against GRCh38.
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 mapped against GRCh38.
srWGS 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 covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS 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 : November 25th, 2024.
This list is accurate for panel version DG 5.1.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors