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HEARING IMPAIRMENT PANEL (INCLUDING GJB2) DG-5.2.0 (307 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	n.t.b					?Deafness, autosomal dominant 77, 618915
ABHD12	n.t.b					Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	n.t.b					Chanarin-Dorfman syndrome, 275630
ACOX1	n.t.b					Mitchell syndrome, 618960;Peroxisomal acyl-CoA oxidase deficiency, 264470
ACTB	n.t.b					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	n.t.b					Deafness, autosomal dominant 20/26, 604717;Baraitser-Winter syndrome 2, 614583
ADAMTS1	n.t.b					
ADCY1	n.t.b					?Deafness, autosomal recessive 44, 610154

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ADGRV1	n.t.b					Usher syndrome, type 2C, 605472;Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472;?Febrile seizures, familial, 4, 604352
AFG2A	n.t.b					Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577
AFG2B	n.t.b					Deafness, autosomal recessive 119, 619615;Neurodevelopmental disorder with hearing loss and spasticity, 619616
AGBL5	n.t.b					Retinitis pigmentosa 75, 617023
AIFM1	n.t.b					Combined oxidative phosphorylation deficiency 6, 300816;Cowchock syndrome, 310490;Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232;Deafness, X-linked 5, 300614
ALMS1	n.t.b					Alstrom syndrome, 203800
AMMECR1	n.t.b					Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
ANKRD24	n.t.b					
ANLN	n.t.b					Focal segmental glomerulosclerosis 8, 616032

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AP1B1	n.t.b					Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150
ARSG	n.t.b					Usher syndrome, type IV, 618144
ATOH1	n.t.b					?Deafness, autosomal dominant 89, 620284
ATP11A	n.t.b					?Auditory neuropathy, autosomal dominant 2, 620384;?Leukodystrophy, hypomyelinating, 24, 619851;Deafness, autosomal dominant 84, 619810
ATP1A1	n.t.b					Hypomagnesemia, seizures, and impaired intellectual development 2, 618314;Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP1A3	n.t.b					Alternating hemiplegia of childhood 2, 614820;Dystonia-12, 128235;CAPOS syndrome, 601338;Developmental and epileptic encephalopathy 99, 619606
ATP2B2	n.t.b					Deafness, autosomal dominant 82, 619804
ATP6V0A4	n.t.b					Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722
ATP6V1B1	n.t.b					Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300

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ATP6V1B2	n.t.b					Zimmermann-Laband syndrome 2, 616455;Deafness, congenital, with onychodystrophy, autosomal dominant, 124480
ATP6V1C1	n.t.b					
BCAP31	n.t.b					Deafness, dystonia, and cerebral hypomyelination, 300475
BCS1L	n.t.b					GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Bjornstad syndrome, 262000
BDP1	n.t.b					?Deafness, autosomal recessive 112, 618257
BICD1	n.t.b					
BMP4	n.t.b					Orofacial cleft 11, 600625;Microphthalmia, syndromic 6, 607932
BSND	n.t.b					Sensorineural deafness with mild renal dysfunction, 602522;Bartter syndrome, type 4a, 602522
BTD	n.t.b					Biotinidase deficiency, 253260
CABP2	n.t.b					Deafness, autosomal recessive 93, 614899
CACNA1D	n.t.b					Primary aldosteronism, seizures, and neurologic abnormalities, 615474;Sinoatrial node dysfunction and deafness, 614896
CCDC50	n.t.b					?Deafness, autosomal dominant 44, 607453

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CD151	n.t.b					[Blood group, Raph], 179620;Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057
CD164	n.t.b					?Deafness, autosomal dominant 66, 616969
CDC14A	n.t.b					Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDC42	n.t.b					Takenouchi-Kosaki syndrome, 616737
CDH23	n.t.b					Usher syndrome, type 1D, 601067;{Pituitary adenoma 5, multiple types}, 617540;Usher syndrome, type 1D/F digenic, 601067;Deafness, autosomal recessive 12, 601386
CDK9	n.t.b					
CEACAM16	n.t.b					Deafness, autosomal dominant 4B, 614614;Deafness, autosomal recessive 113, 618410
CEP250	n.t.b					Cone-rod dystrophy and hearing loss 2, 618358
CEP78	n.t.b					Cone-rod dystrophy and hearing loss, 617236
CGN	n.t.b					
CHD4	n.t.b					Sifrim-Hitz-Weiss syndrome, 617159
CHD7	n.t.b					Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800

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CHSY1	n.t.b					Temtamy preaxial brachydactyly syndrome, 605282
CIB2	n.t.b					Deafness, autosomal recessive 48, 609439
CISD2	n.t.b					Wolfram syndrome 2, 604928
CLDN14	n.t.b					Deafness, autosomal recessive 29, 614035
CLDN9	n.t.b					Deafness, autosomal recessive 116, 619093
CLIC5	n.t.b					?Deafness, autosomal recessive 103, 616042
CLPP	n.t.b					Perrault syndrome 3, 614129
CLRN1	n.t.b					Usher syndrome, type 3A, 276902;Retinitis pigmentosa 61, 614180
CLRN2	n.t.b					Deafness, autosomal recessive 117, 619174
COA8	n.t.b					Mitochondrial complex IV deficiency, nuclear type 17, 619061
COCH	n.t.b					Deafness, autosomal dominant 9, 601369;?Deafness, autosomal recessive 110, 618094
COL11A1	n.t.b					Fibrochondrogenesis 1, 228520;Stickler syndrome, type II, 604841;Marshall syndrome, 154780;Deafness, autosomal dominant 37, 618533;{Lumbar disc herniation, susceptibility to}, 603932

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COL11A2	n.t.b					Deafness, autosomal dominant 13, 601868;Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150;Fibrochondrogenesis 2, 614524;Deafness, autosomal recessive 53, 609706;Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840

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COL2A1	n.t.b					?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248;Czech dysplasia, 609162;Achondrogenesis, type II or hypochondrogenesis, 200610;Spondyloperipheral dysplasia, 271700;SMED Strudwick type, 184250;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;Epiphyseal dysplasia, multiple, with myopia and deafness, 132450
COL4A3	n.t.b					Alport syndrome 3A, autosomal dominant, 104200;Hematuria, benign familial, 2, 620320;Alport syndrome 3B, autosomal recessive, 620536

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COL4A4	n.t.b					Hematuria, familial benign, 1, 141200;Alport syndrome 2, autosomal recessive, 203780
COL4A5	n.t.b					Alport syndrome 1, X-linked, 301050
COL4A6	n.t.b					?Deafness, X-linked 6, 300914
COL9A1	n.t.b					Stickler syndrome, type IV, 614134;?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	n.t.b					Epiphyseal dysplasia, multiple, 2, 600204;?Stickler syndrome, type V, 614284
COL9A3	n.t.b					{Intervertebral disc disease, susceptibility to}, 603932;Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969;Stickler syndrome, type VI, 620022
CRYL1	n.t.b					
CRYM	n.t.b					Deafness, autosomal dominant 40, 616357
DAP3	n.t.b					Perrault syndrome 7, 621101
DCAF17	n.t.b					Woodhouse-Sakati syndrome, 241080
DCDC2	n.t.b					Nephronophthisis 19, 616217;?Deafness, autosomal recessive 66, 610212;Sclerosing cholangitis, neonatal, 617394

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

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DSPP	n.t.b					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	n.t.b					Waardenburg syndrome, type 4B, 613265;{Hirschsprung disease, susceptibility to, 4}, 613712
EDNRB	n.t.b					{Hirschsprung disease, susceptibility to, 2}, 600155;?ABCD syndrome, 600501;Waardenburg syndrome, type 4A, 277580
EFNB2	n.t.b					
EHD1	n.t.b					
ELMOD3	n.t.b					?Deafness, autosomal recessive 88, 615429;?Deafness, autosomal dominant 81, 619500
ELOVL1	n.t.b					Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
EPHA10	n.t.b					?Deafness, autosomal dominant 88, 620283
EPS8	n.t.b					?Deafness, autosomal recessive 102, 615974
EPS8L2	n.t.b					Deafness autosomal recessive 106, 617637
ERAL1	n.t.b					Perrault syndrome 6, 617565

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ESPN	n.t.b					Deafness, autosomal recessive 36, 609006;Deafness, autosomal dominant 91, 621556;?Usher syndrome, type 1M, 618632
ESRP1	n.t.b					?Deafness, autosomal recessive 109, 618013
ESRRB	n.t.b					Deafness, autosomal recessive 35, 608565
EXOSC2	n.t.b					Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EYA1	n.t.b					Branchiootic syndrome 1, 602588;Branchiootorenal syndrome 1, with or without cataracts, 113650;Anterior segment anomalies with or without cataract, 602588;?Otofaciocervical syndrome, 166780
EYA4	n.t.b					?Cardiomyopathy, dilated, 1J, 605362;Deafness, autosomal dominant 10, 601316
FDXR	n.t.b					Multiple mitochondrial dysfunctions syndrome 9B, 620887;Auditory neuropathy and optic atrophy, 617717
FGF3	n.t.b					Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706

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FGFR3	n.t.b					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	n.t.b					Siddiqi syndrome, 618635
FMN1	n.t.b					
FOXC1	n.t.b					Axenfeld-Rieger syndrome, type 3, 602482;Anterior segment dysgenesis 3, multiple subtypes, 601631
FOXF2	n.t.b					
FOXI1	n.t.b					Enlarged vestibular aqueduct, 600791
FRMPD4	n.t.b					Intellectual developmental disorder, X-linked 104, 300983
GAB1	n.t.b					?Deafness, autosomal recessive 26, 605428
GAS2	n.t.b					?Deafness, autosomal recessive 125, 620877

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GATA3	n.t.b					Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GIPC3	n.t.b					Deafness, autosomal recessive 15, 601869
GJB2	n.t.b					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	n.t.b					Deafness, digenic, GJB2/GJB3, 220290;Erythrokeratodermia variabilis et progressiva 1, 133200;Deafness, autosomal dominant 2B, with or without peripheral neuropathy, 612644
GJB6	n.t.b					Ectodermal dysplasia 2, Clouston type, 129500;Deafness, autosomal dominant 3B, 612643;Deafness, autosomal recessive 1B, 612645;Deafness, digenic GJB2/GJB6, 220290
GLA	n.t.b					Fabry disease, cardiac variant, 301500;Fabry disease, 301500

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

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HOMER2	n.t.b					?Deafness, autosomal dominant 68, 616707
HOXA2	n.t.b					Microtia with or without hearing impairment (AD), 612290;?Microtia, hearing impairment, and cleft palate (AR), 612290
HSD17B4	n.t.b					D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400
IFNLR1	n.t.b					
IKZF2	n.t.b					Immunodysregulation, craniofacial anomalies, hearing impairment, athelia, and developmental delay, 621234;Immunodysregulation with variable immunodeficiency and autoimmunity, 621233
ILDR1	n.t.b					Deafness, autosomal recessive 42, 609646
IRF6	n.t.b					{Orofacial cleft 6}, 608864;Popliteal pterygium syndrome 1, 119500;van der Woude syndrome 1, 119300;Ectodermal dysplasia 19, hypohydrotic/tooth/nail type, 621749
JAG1	n.t.b					?Deafness, congenital heart defects, and posterior embryotoxon, 617992;Charcot-Marie-Tooth disease, axonal, type 2HH, 619574;Alagille syndrome 1, 118450;Tetralogy of Fallot, 187500

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KARS1	n.t.b					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	n.t.b					Jervell and Lange-Nielsen syndrome 2, 612347;Long QT syndrome 5, 613695
KCNJ10	n.t.b					Enlarged vestibular aqueduct, digenic, 600791;SESAME syndrome, 612780
KCNJ16	n.t.b					Hypokalemic tubulopathy and deafness, 619406
KCNQ1	n.t.b					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	n.t.b					Deafness, autosomal dominant 2A, 600101

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KITLG	n.t.b					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	n.t.b					Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LHFPL5	n.t.b					Deafness, autosomal recessive 67, 610265
LMX1A	n.t.b					Deafness, autosomal dominant 7, 601412
LMX1B	n.t.b					Focal segmental glomerulosclerosis 10, 256020;Nail-patella syndrome, 161200
LOXHD1	n.t.b					Deafness, autosomal recessive 77, 613079
LOXL3	n.t.b					Myopia 28, autosomal recessive, 619781
LRP2	n.t.b					Donnai-Barrow syndrome, 222448

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LRP5	n.t.b					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	n.t.b					Deafness, autosomal recessive 63, 611451
MAN2B1	n.t.b					Mannosidosis, alpha-, types I and II, 248500
MANBA	n.t.b					Mannosidosis, beta, 248510
MAP1B	n.t.b					?Deafness, autosomal dominant 83, 619808;Periventricular nodular heterotopia 9, 618918
MAP3K1	n.t.b					46XY sex reversal 6, 613762
MAP3K20	n.t.b					Centronuclear myopathy 6 with fiber-type disproportion, 617760;Split-foot malformation with mesoaxial polydactyly, 616890
MARVELD2	n.t.b					Deafness, autosomal recessive 49, 610153
MCM2	n.t.b					?Deafness, autosomal dominant 70, 616968

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

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MPZ	n.t.b					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	n.t.b					Deafness, autosomal recessive 111, 618145
MRPL49	n.t.b					Combined oxidative phosphorylation deficiency 60, 621195
MRPL50	n.t.b					
MSRB3	n.t.b					Deafness, autosomal recessive 74, 613718
MYH14	n.t.b					?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369;Deafness, autosomal dominant 4A, 600652
MYH7B	n.t.b					
MYH9	n.t.b					Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100;Deafness, autosomal dominant 17, 603622
MYO15A	n.t.b					Deafness, autosomal recessive 3, 600316

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MYO3A	n.t.b					Deafness, autosomal recessive 30, 607101;Deafness, autosomal dominant 90, 620722
MYO6	n.t.b					Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346;Deafness, autosomal dominant 22, 606346;Deafness, autosomal recessive 37, 607821
MYO7A	n.t.b					Deafness, autosomal recessive 2, 600060;Usher syndrome, type 1B, 276900;Deafness, autosomal dominant 11, 601317
NARS2	n.t.b					Combined oxidative phosphorylation deficiency 24, 616239;?Deafness, autosomal recessive 94, 618434
NCOA3	n.t.b					
NDP	n.t.b					Exudative vitreoretinopathy 2, X-linked, 305390;Norrie disease, 310600
NEU4	n.t.b					
NHERF1	n.t.b					Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287

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NLRP3	n.t.b					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	n.t.b					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	n.t.b					?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

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OPA1	n.t.b					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	n.t.b					Dyschromatosis, ichthyosis, deafness, and atopic disease, 621400;Deafness, autosomal dominant 67, 616340
OTOA	n.t.b					Deafness, autosomal recessive 22, 607039
OTOF	n.t.b					Auditory neuropathy, autosomal recessive, 1, 601071;Deafness, autosomal recessive 9, 601071
OTOG	n.t.b					Deafness, autosomal recessive 18B, 614945
OTOGL	n.t.b					Deafness, autosomal recessive 84B, 614944
P2RX2	n.t.b					Deafness, autosomal dominant 41, 608224
PAX3	n.t.b					Craniofacial-deafness-hand syndrome, 122880;Waardenburg syndrome, type 3, 148820;Waardenburg syndrome, type 1, 193500;Rhabdomyosarcoma 2, alveolar, 268220

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PBX1	n.t.b					Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCDH15	n.t.b					Usher syndrome, type 1D/F digenic, 601067;Deafness, autosomal recessive 23, 609533;Usher syndrome, type 1F, 602083
PDE1C	n.t.b					?Deafness, autosomal dominant 74, 618140
PDZD7	n.t.b					Deafness, autosomal recessive 57, 618003;{Retinal disease in Usher syndrome type IIA, modifier of}, 276901;Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472
PET100	n.t.b					Mitochondrial complex IV deficiency, nuclear type 12, 619055
PEX1	n.t.b					Heimler syndrome 1, 234580;Peroxisome biogenesis disorder 1B (NALD/IRD), 601539;Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX11B	n.t.b					Peroxisome biogenesis disorder 14B, 614920
PEX26	n.t.b					Peroxisome biogenesis disorder 7B, 614873;Peroxisome biogenesis disorder 7A (Zellweger), 614872

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PEX6	n.t.b					Peroxisome biogenesis disorder 4B, 614863;Peroxisome biogenesis disorder 4A (Zellweger), 614862;Heimler syndrome 2, 616617
PI4KB	n.t.b					Deafness, autosomal dominant 87, 620281
PISD	n.t.b					Liberfarb syndrome, 618889
PJVK	n.t.b					Deafness, autosomal recessive 59, 610220
PKHD1L1	n.t.b					Deafness, autosomal recessive 124, 620794
PLCG1	n.t.b					?Immune dysregulation, autoimmunity, and autoinflammation, 620514
PLOD3	n.t.b					BCARD syndrome (lysyl hydroxylase 3 deficiency), 612394
PLS1	n.t.b					Deafness, autosomal dominant 76, 618787
PLXNB2	n.t.b					
PNPT1	n.t.b					Spinocerebellar ataxia 25, 608703;Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934;Combined oxidative phosphorylation deficiency 13, 614932

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POLD1	n.t.b					Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381;Immunodeficiency 120, 620836;{Colorectal cancer, susceptibility to, 10}, 612591
POLR1C	n.t.b					Leukodystrophy, hypomyelinating, 11, 616494;Treacher Collins syndrome 3, 248390
POLR1D	n.t.b					Treacher Collins syndrome 2, 613717
POU3F4	n.t.b					Deafness, X-linked 2, 304400
POU4F3	n.t.b					Deafness, autosomal dominant 15/52, 602459
PPIP5K2	n.t.b					Deafness, autosomal recessive 100, 618422
PRKCB	n.t.b					
PRORP	n.t.b					Combined oxidative phosphorylation deficiency 54, 619737
PRPS1	n.t.b					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	n.t.b					

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PTPRQ	n.t.b					Deafness, autosomal dominant 73, 617663;Deafness, autosomal recessive 84A, 613391
PTRH2	n.t.b					Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
RAI1	n.t.b					Smith-Magenis syndrome, 182290
RDX	n.t.b					Deafness, autosomal recessive 24, 611022
REST	n.t.b					Deafness, autosomal dominant 27, 612431;{Wilms tumor 6, susceptibility to}, 616806;Fibromatosis, gingival, 5, 617626
RIPOR2	n.t.b					Deafness, autosomal dominant 21, 607017;?Deafness, autosomal recessive 104, 616515
RMND1	n.t.b					Combined oxidative phosphorylation deficiency 11, 614922
ROBO1	n.t.b					Pituitary hormone deficiency, combined or isolated, 8, 620303;Neurooculoren al syndrome, 620305;?Nystagmus 8, congenital, autosomal recessive, 257400
ROR1	n.t.b					?Deafness, autosomal recessive 108, 617654

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RRM2B	n.t.b					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	n.t.b					Deafness, autosomal recessive 68, 610419
SALL1	n.t.b					Townes-Brocks syndrome 1, 107480;Townes-Brocks branchiootorenal-like syndrome, 107480
SCD5	n.t.b					?Deafness, autosomal dominant 79, 619086
SEMA3F	n.t.b					
SERAC1	n.t.b					3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINB6	n.t.b					?Deafness, autosomal recessive 91, 613453
SEZ6	n.t.b					
SIX1	n.t.b					Deafness, autosomal dominant 23, 605192;Branchiootic syndrome 3, 608389
SIX5	n.t.b					Branchiootorenal syndrome 2, 610896

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SLC12A1	n.t.b					Bartter syndrome, type 1, 601678
SLC12A2	n.t.b					Kilquist syndrome, 619080;Delpire-McNeill syndrome, 619083;Deafness, autosomal dominant 78, 619081
SLC17A8	n.t.b					Deafness, autosomal dominant 25, 605583
SLC19A2	n.t.b					Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC22A4	n.t.b					{Rheumatoid arthritis, susceptibility to}, 180300
SLC26A4	n.t.b					Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791;Pendred syndrome, 274600
SLC26A5	n.t.b					?Deafness, autosomal recessive 61, 613865
SLC29A3	n.t.b					Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC30A9	n.t.b					Birk-Landau-Perez syndrome, 617595
SLC33A1	n.t.b					Spastic paraplegia 42, autosomal dominant, 612539;Huppke-Brendel syndrome, 614482
SLC44A4	n.t.b					?Deafness, autosomal dominant 72, 617606

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SLC4A11	n.t.b					Corneal endothelial dystrophy, autosomal recessive, 217700;Corneal dystrophy, Fuchs endothelial, 4, 613268;Corneal endothelial dystrophy and perceptive deafness, 217400
SLC52A2	n.t.b					Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	n.t.b					?Fazio-Londe disease, 211500;Brown-Vialetto-Van Laere syndrome 1, 211530
SLC7A14	n.t.b					Retinitis pigmentosa 68, 615725
SLC9A1	n.t.b					Lichtenstein-Knorr syndrome, 616291
SLITRK6	n.t.b					Deafness and myopia, 221200
SMPX	n.t.b					Myopathy, distal, 7, adult-onset, X-linked, 301075;Deafness, X-linked 4, 300066
SNAI2	n.t.b					
SOX10	n.t.b					Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SPNS2	n.t.b					?Deafness, autosomal recessive 115, 618457
STAG2	n.t.b					Holoprosencephaly 13, X-linked, 301043;Mullegama-Klein-Martinez syndrome, 301022

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STRC	n.t.b					Deafness, autosomal recessive 16, 603720
STX4	n.t.b					?Deafness, autosomal recessive 123, 620745
SUCLA2	n.t.b					Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SYNE4	n.t.b					Deafness, autosomal recessive 76, 615540
TBC1D24	n.t.b					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	n.t.b					Hypothyroidism, congenital, nongoitrous, 8, 301033
TBL1Y	n.t.b					?Deafness, Y-linked 2, 400047
TCOF1	n.t.b					Treacher Collins syndrome 1, 154500
TECTA	n.t.b					Deafness, autosomal dominant 8/12, 601543;Deafness, autosomal recessive 21, 603629
TECTB	n.t.b					Deafness, autosomal dominant 55, 621728

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TFAP2A	n.t.b					Branchiooculofacial syndrome, 113620
THBS1	n.t.b					
THOC1	n.t.b					?Deafness, autosomal dominant 86, 620280
TIMM8A	n.t.b					Mohr-Tranebjaerg syndrome, 304700
TJP2	n.t.b					Hypercholanemia, familial 1, 607748;Cholestasis, progressive familial intrahepatic 4, 615878
TMC1	n.t.b					Deafness, autosomal dominant 36, 606705;Deafness, autosomal recessive 7, 600974
TMEM132E	n.t.b					Deafness, autosomal recessive 99, 618481
TMEM43	n.t.b					Arrhythmogenic right ventricular dysplasia 5, 604400;Auditory neuropathy, autosomal dominant 3, 619832;Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMIE	n.t.b					Deafness, autosomal recessive 6, 600971
TMPRSS3	n.t.b					Deafness, autosomal recessive 8/10, 601072
TMTC2	n.t.b					
TMTC4	n.t.b					?Deafness, autosomal recessive 122, 620714
TNC	n.t.b					Deafness, autosomal dominant 56, 615629
TPRN	n.t.b					Deafness, autosomal recessive 79, 613307
TRIM71	n.t.b					Hydrocephalus, congenital, 4, 618667

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TRIOBP	n.t.b					Deafness, autosomal recessive 28, 609823
TRRAP	n.t.b					?Deafness, autosomal dominant 75, 618778;Developmental delay with or without dysmorphic facies and autism, 618454
TSHZ1	n.t.b					Aural atresia, congenital, 607842
TUBB4B	n.t.b					Leber congenital amaurosis with early-onset deafness, 617879
TWNK	n.t.b					Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286;Perrault syndrome 5, 616138
USH1C	n.t.b					Usher syndrome, type 1C, 276904;Deafness, autosomal recessive 18A, 602092
USH1G	n.t.b					Usher syndrome, type 1G, 606943
USH2A	n.t.b					Usher syndrome, type 2A, 276901;Retinitis pigmentosa 39, 613809
USP48	n.t.b					Deafness, autosomal dominant 85, 620227
WBP2	n.t.b					Deafness, autosomal recessive 107, 617639

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WFS1	n.t.b					Deafness, autosomal dominant 6/14/38, 600965;?Cataract 41, 116400;Wolfram-like syndrome, autosomal dominant, 614296;Wolfram syndrome 1, 222300
WHRN	n.t.b					Deafness, autosomal recessive 31, 607084;Usher syndrome, type 2D, 611383
XKR8	n.t.b					
XYLT2	n.t.b					{Pseudoxanthoma elasticum, modifier of severity of}, 264800;Spondyloocular syndrome, 605822
YAP1	n.t.b					Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or impaired intellectual development, 120433
YARS1	n.t.b					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