HEARING IMPAIRMENT GENE PANEL DG 3.4.0 (253 genes)

Releasedate: 19-04-2022

Gene	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
ABCC1	100,0%	100,0%	?Deafness, autosomal dominant 77, 618915
ABHD12	100,0%	100,0%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACTB	100,0%	100,0%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACTG1	100,0%	100,0%	Deafness, autosomal dominant 20/26, 604717 Baraitser-Winter syndrome 2, 614583
ADAMTS1	100,0%	100,0%	No OMIM Disease ID
ADCY1	99,3%	98,9%	?Deafness, autosomal recessive 44, 610154
ADGRV1	100,0%	100,0%	Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 ?Febrile seizures, familial, 4, 604352
AIFM1	100,0%	100,0%	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%	Alstrom syndrome, 203800
AMMECR1	100,0%	100,0%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
ANLN	100,0%	100,0%	Focal segmental glomerulosclerosis 8, 616032
AP1B1	100,0%	100,0%	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150
ARSG	100,0%	100,0%	Usher syndrome, type IV, 618144
ATOH1	100,0%	100,0%	No OMIM Disease ID
ATP1A3	100,0%	100,0%	Alternating hemiplegia of childhood 2, 614820 Dystonia-12, 128235 CAPOS syndrome, 601338 Developmental and epileptic encephalopathy 99, 619606
ATP2B2	100,0%	100,0%	Deafness, autosomal dominant 82, 619804
ATP6V0A4	100,0%	100,0%	Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722
ATP6V1B1	100,0%	100,0%	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300

ATP6V1B2	100,0%	100,0%	Zimmermann-Laband syndrome 2, 616455 Deafness, congenital, with onychodystrophy, autosomal dominant, 124480
BCAP31	100,0%	100,0%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCS1L	100,0%	100,0%	GRACILE syndrome, 603358 Mitochondrial complex III deficiency, nuclear type 1, 124000 Bjornstad syndrome, 262000
BDP1	100,0%	100,0%	?Deafness, autosomal recessive 112, 618257
BMP4	100,0%	100,0%	Orofacial cleft 11, 600625 Microphthalmia, syndromic 6, 607932
BSND	100,0%	100,0%	Sensorineural deafness with mild renal dysfunction, 602522 Bartter syndrome, type 4a, 602522
BTD	83,1%	83,1%	Biotinidase deficiency, 253260
CABP2	100,0%	100,0%	Deafness, autosomal recessive 93, 614899
CACNA1D	100,0%	100,0%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CCDC50	100,0%	100,0%	?Deafness, autosomal dominant 44, 607453
CD151	100,0%	100,0%	Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057
CD164	100,0%	100,0%	?Deafness, autosomal dominant 66, 616969
CDC14A	100,0%	100,0%	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDC42	100,0%	100,0%	Takenouchi-Kosaki syndrome, 616737
CDH23	100,0%	100,0%	Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067 Deafness, autosomal recessive 12, 601386
CEACAM16	100,0%	100,0%	Deafness, autosomal dominant 4B, 614614 Deafness, autosomal recessive 113, 618410
CEP250	100,0%	100,0%	Cone-rod dystrophy and hearing loss 2, 618358
CEP78	100,0%	100,0%	Cone-rod dystrophy and hearing loss, 617236
CHD7	100,0%	100,0%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CHSY1	100,0%	99,9%	Temtamy preaxial brachydactyly syndrome, 605282
CIB2	100,0%	100,0%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CISD2	100,0%	100,0%	Wolfram syndrome 2, 604928
CLDN14	100,0%	100,0%	Deafness, autosomal recessive 29, 614035
CLDN9	100,0%	100,0%	?Deafness, autosomal recessive 116, 619093
CLIC5	100,0%	100,0%	?Deafness, autosomal recessive 103, 616042
CLPP	100,0%	100,0%	Perrault syndrome 3, 614129

CLRN1	100,0%	100,0%	Usher syndrome, type 3A, 276902 Retinitis pigmentosa 61, 614180
CLRN2	100,0%	100,0%	?Deafness, autosomal recessive 117, 619174
COA8	93,5%	93,5%	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COCH	100,0%	100,0%	Deafness, autosomal dominant 9, 601369 ?Deafness, autosomal recessive 110, 618094
COL11A1	100,0%	100,0%	Fibrochondrogenesis 1, 228520 Stickler syndrome, type II, 604841 Marshall syndrome, 154780 Deafness, autosomal dominant 37, 618533
COL11A2	100,0%	100,0%	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%	?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%	Hematuria, benign familial, 141200 Alport syndrome 3, autosomal dominant, 104200 Alport syndrome 2, autosomal recessive, 203780
COL4A4	100,0%	100,0%	Hematuria, familial benign, 141200 Alport syndrome 2, autosomal recessive, 203780
COL4A5	100,0%	100,0%	Alport syndrome 1, X-linked, 301050
COL4A6	100,0%	100,0%	?Deafness, X-linked 6, 300914

COL9A1	100,0%	100,0%	Stickler syndrome, type IV, 614134 ?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	100,0%	100,0%	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284
COL9A3	100,0%	100,0%	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969
CRYL1	100,0%	100,0%	No OMIM Disease ID
CRYM	100,0%	100,0%	Deafness, autosomal dominant 40, 616357
DCAF17	100,0%	100,0%	Woodhouse-Sakati syndrome, 241080
DCDC2	100,0%	100,0%	Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212 Sclerosing cholangitis, neonatal, 617394
DIABLO	100,0%	100,0%	Deafness, autosomal dominant 64, 614152
DIAPH1	100,0%	100,0%	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DIAPH3	100,0%	100,0%	Auditory neuropathy, autosomal dominant 1, 609129
DLL1	100,0%	100,0%	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709
DLX5	100,0%	100,0%	Split-hand/foot malformation 1, 183600 ?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DMXL2	100,0%	100,0%	Developmental and epileptic encephalopathy 81, 618663 ?Deafness, autosomal dominant 71, 617605 ?Polyendocrine-polyneuropathy syndrome, 616113
DSPP	100,0%	100,0%	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%	Waardenburg syndrome, type 4B, 613265
EDNRB	100,0%	100,0%	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580
EFNB2	100,0%	100,0%	No OMIM Disease ID
EHD1	100,0%	100,0%	No OMIM Disease ID
ELMOD3	100,0%	100,0%	?Deafness, autosomal recessive 88, 615429 ?Deafness, autosomal dominant 81, 619500
ELOVL1	100,0%	100,0%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
EPS8	100,0%	100,0%	?Deafness, autosomal recessive 102, 615974
EPS8L2	88,0%	88,0%	Deafness autosomal recessive 106, 617637
ERAL1	100,0%	100,0%	Perrault syndrome 6, 617565

ESPN	100,0%	100,0%	Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006 Deafness, autosomal recessive 36, 609006 ?Usher syndrome, type 1M, 618632
ESRP1	100,0%	100,0%	?Deafness, autosomal recessive 109, 618013
ESRRB	100,0%	100,0%	Deafness, autosomal recessive 35, 608565
EXOSC2	100,0%	100,0%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EYA1	100,0%	100,0%	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%	?Cardiomyopathy, dilated, 1J, 605362 Deafness, autosomal dominant 10, 601316
FDXR	100,0%	100,0%	Auditory neuropathy and optic atrophy, 617717
FGF3	100,0%	100,0%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGFR3	100,0%	100,0%	Muenke syndrome, 602849 SADDAN, 616482 Hypochondroplasia, 146000 LADD syndrome, 149730 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 Achondroplasia, 100800 Cervical cancer, somatic, 603956 Colorectal cancer, somatic, 114500 Crouzon syndrome with acanthosis nigricans, 612247
FITM2	100,0%	100,0%	Siddiqi syndrome, 618635
FOXF2	99,0%	97,7%	No OMIM Disease ID
FOXI1	100,0%	100,0%	Enlarged vestibular aqueduct, 600791
GAB1	100,0%	100,0%	?Deafness, autosomal recessive 26, 605428
GAS2	100,0%	100,0%	No OMIM Disease ID
GATA3	100,0%	100,0%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GIPC3	100,0%	100,0%	Deafness, autosomal recessive 15, 601869
GJB2	100,0%	100,0%	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%	Deafness, digenic, GJB2/GJB3, 220290 Deafness, autosomal dominant 2B, 612644 Erythrokeratodermia variabilis et progressiva 1, 133200 Deafness, autosomal recessive, Deafness, autosomal dominant, with peripheral neuropathy,
GJB6	100,0%	100,0%	Ectodermal dysplasia 2, Clouston type, 129500 Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290
GLA	91,3%	91,3%	Fabry disease, cardiac variant, 301500 Fabry disease, 301500
GPRASP2	100,0%	100,0%	?Deafness, X-linked 7, 301018
GPSM2	100,0%	100,0%	Chudley-McCullough syndrome, 604213
GRAP	100,0%	100,0%	Deafness, autosomal recessive 114, 618456
GREB1L	100,0%	100,0%	Deafness, autosomal dominant 80, 619274 Renal hypodysplasia/aplasia 3, 617805
GRHL2	100,0%	100,0%	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029 Corneal dystrophy, posterior polymorphous, 4, 618031
GRXCR1	100,0%	100,0%	Deafness, autosomal recessive 25, 613285
GRXCR2	100,0%	100,0%	?Deafness, autosomal recessive 101, 615837
GSDME	100,0%	100,0%	Deafness, autosomal dominant 5, 600994
HARS1	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HARS2	100,0%	100,0%	Perrault syndrome 2, 614926
HGF	100,0%	100,0%	Deafness, autosomal recessive 39, 608265
HOMER2	100,0%	100,0%	?Deafness, autosomal dominant 68, 616707
HSD17B4	96,6%	96,6%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
IFNLR1	100,0%	100,0%	No OMIM Disease ID
ILDR1	100,0%	100,0%	Deafness, autosomal recessive 42, 609646
KARS1	100,0%	100,0%	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%	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695
KCNJ10	100,0%	100,0%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNQ1	100,0%	100,0%	Short QT syndrome 2, 609621 Atrial fibrillation, familial, 3, 607554 Long QT syndrome 1, 192500 Jervell and Lange-Nielsen syndrome, 220400
KCNQ4	99,9%	99,3%	Deafness, autosomal dominant 2A, 600101
KITLG	100,0%	100,0%	Hyperpigmentation with or without hypopigmentation, 145250 Deafness, autosomal dominant 69, unilateral or asymmetric, 616697
LARS2	100,0%	100,0%	Perrault syndrome 4, 615300 Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LHFPL5	100,0%	100,0%	Deafness, autosomal recessive 67, 610265
LMX1A	100,0%	100,0%	Deafness, autosomal dominant 7, 601412
LOXHD1	100,0%	100,0%	Deafness, autosomal recessive 77, 613079
LOXL3	100,0%	100,0%	Myopia 28, autosomal recessive, 619781
LRP2	100,0%	100,0%	Donnai-Barrow syndrome, 222448
LRP5	100,0%	100,0%	Osteopetrosis, autosomal dominant 1, 607634 Hyperostosis, endosteal, 144750 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 Osteoporosis-pseudoglioma syndrome, 259770 Exudative vitreoretinopathy 4, 601813 van Buchem disease, type 2, 607636
LRTOMT	100,0%	100,0%	Deafness, autosomal recessive 63, 611451
MAN2B1	100,0%	100,0%	Mannosidosis, alpha-, types I and II, 248500
MARVELD2	100,0%	100,0%	Deafness, autosomal recessive 49, 610153
MCM2	100,0%	100,0%	?Deafness, autosomal dominant 70, 616968
MET	100,0%	100,0%	Renal cell carcinoma, papillary, 1, familial and somatic, 605074 Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705
MGP	100,0%	100,0%	Keutel syndrome, 245150
MIA3	100,0%	100,0%	?Ondontochondrodysplasia 2 with hearing loss and diabetes, 619269
MIR96	NC	NC	Deafness, autosomal dominant 50, 613074

MITF	100,0%	100,0%	Waardenburg syndrome, type 2A, 193510 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome/ocular albinism, digenic, 103470 COMMAD syndrome, 617306
MPDZ	100,0%	100,0%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPZL2	100,0%	100,0%	Deafness, autosomal recessive 111, 618145
MSRB3	100,0%	100,0%	Deafness, autosomal recessive 74, 613718
MVD	100,0%	100,0%	Porokeratosis 7, multiple types, 614714
MYH14	100,0%	100,0%	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 Deafness, autosomal dominant 4A, 600652
MYH9	100,0%	100,0%	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100 Deafness, autosomal dominant 17, 603622
MYO15A	100,0%	100,0%	Deafness, autosomal recessive 3, 600316
MYO3A	100,0%	100,0%	Deafness, autosomal recessive 30, 607101
MYO6	100,0%	100,0%	Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 Deafness, autosomal dominant 22, 606346 Deafness, autosomal recessive 37, 607821
MYO7A	100,0%	100,0%	Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900 Deafness, autosomal dominant 11, 601317
NARS2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 24, 616239 ?Deafness, autosomal recessive 94, 618434
NCOA3	100,0%	100,0%	No OMIM Disease ID
NDP	100,0%	100,0%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NLRP3	100,0%	100,0%	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%	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%	Optic atrophy plus syndrome, 125250 Optic atrophy 1, 165500

			Behr syndrome, 210000 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
OSBPL2	100,0%	100,0%	Deafness, autosomal dominant 67, 616340
OTOA	100,0%	100,0%	Deafness, autosomal recessive 22, 607039
OTOF	100,0%	100,0%	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071
OTOG	100,0%	100,0%	Deafness, autosomal recessive 18B, 614945
OTOGL	100,0%	100,0%	Deafness, autosomal recessive 84B, 614944
P2RX2	100,0%	100,0%	Deafness, autosomal dominant 41, 608224
PAX3	100,0%	100,0%	Craniofacial-deafness-hand syndrome, 122880 Waardenburg syndrome, type 3, 148820 Waardenburg syndrome, type 1, 193500 Rhabdomyosarcoma 2, alveolar, 268220
PCDH15	100,0%	100,0%	Usher syndrome, type 1D/F digenic, 601067 Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1F, 602083
PDE1C	100,0%	100,0%	?Deafness, autosomal dominant 74, 618140
PDZD7	100,0%	100,0%	Deafness, autosomal recessive 57, 618003 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472
PET100	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PEX1	100,0%	100,0%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX26	100,0%	100,0%	Peroxisome biogenesis disorder 7B, 614873 Peroxisome biogenesis disorder 7A (Zellweger), 614872
PEX6	100,0%	100,0%	Peroxisome biogenesis disorder 4B, 614863 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Heimler syndrome 2, 616617
PI4KB	100,0%	100,0%	No OMIM Disease ID
PISD	100,0%	100,0%	Liberfarb syndrome, 618889
PJVK	100,0%	100,0%	Deafness, autosomal recessive 59, 610220
PLOD3	100,0%	100,0%	Lysyl hydroxylase 3 deficiency, 612394
PLS1	100,0%	100,0%	Deafness, autosomal dominant 76, 618787
PNPT1	100,0%	100,0%	Deafness, autosomal recessive 70, 614934 Combined oxidative phosphorylation deficiency 13, 614932
POLD1	100,0%	100,0%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381

POLR1C	83,0%	82,8%	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	100,0%	100,0%	Treacher Collins syndrome 2, 613717
POU3F4	100,0%	100,0%	Deafness, X-linked 2, 304400
POU4F3	100,0%	100,0%	Deafness, autosomal dominant 15, 602459
PPIP5K2	100,0%	100,0%	Deafness, autosomal recessive 100, 618422
PRKCB	100,0%	100,0%	No OMIM Disease ID
PRORP	100,0%	100,0%	Combined oxidative phosphorylation deficiency 54, 619737
PRPS1	100,0%	100,0%	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%	No OMIM Disease ID
PTPRQ	92,8%	92,6%	Deafness, autosomal dominant 73, 617663 Deafness, autosomal recessive 84A, 613391
PTRH2	100,0%	100,0%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
RAI1	100,0%	100,0%	Smith-Magenis syndrome, 182290
RDX	100,0%	100,0%	Deafness, autosomal recessive 24, 611022
REST	98,6%	98,6%	?Deafness, autosomal dominant 27, 612431 Fibromatosis, gingival, 5, 617626
RIPOR2	100,0%	100,0%	Deafness, autosomal dominant 21, 607017 ?Deafness, autosomal recessive 104, 616515
RMND1	100,0%	100,0%	Combined oxidative phosphorylation deficiency 11, 614922
ROBO1	100,0%	100,0%	No OMIM Disease ID
ROR1	100,0%	100,0%	?Deafness, autosomal recessive 108, 617654
RRM2B	100,0%	100,0%	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%	Deafness, autosomal recessive 68, 610419
SCD5	100,0%	100,0%	?Deafness, autosomal dominant 79, 619086
SERAC1	100,0%	100,0%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINB6	100,0%	100,0%	?Deafness, autosomal recessive 91, 613453
SEZ6	100,0%	100,0%	No OMIM Disease ID
SIX1	100,0%	100,0%	Deafness, autosomal dominant 23, 605192 Branchiootic syndrome 3, 608389

SIX5	100,0%	100,0%	Branchiootorenal syndrome 2, 610896
SLC12A1	96,2%	96,2%	Bartter syndrome, type 1, 601678
SLC12A2	100,0%	100,0%	Kilquist syndrome, 619080 Delpire-McNeill syndrome, 619083 Deafness, autosomal dominant 78, 619081
SLC17A8	100,0%	100,0%	Deafness, autosomal dominant 25, 605583
SLC19A2	100,0%	100,0%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC22A4	100,0%	100,0%	No OMIM Disease ID
SLC26A4	100,0%	100,0%	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 Pendred syndrome, 274600
SLC26A5	100,0%	100,0%	?Deafness, autosomal recessive 61, 613865
SLC29A3	100,0%	100,0%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC33A1	100,0%	100,0%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC44A4	100,0%	100,0%	?Deafness, autosomal dominant 72, 617606
SLC4A11	100,0%	100,0%	Corneal endothelial dystrophy, autosomal recessive, 217700 Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy and perceptive deafness, 217400
SLC52A2	100,0%	100,0%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100,0%	100,0%	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SLC9A3R1	100,0%	100,0%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLITRK6	100,0%	100,0%	Deafness and myopia, 221200
SMPX	100,0%	100,0%	Myopathy, distal, 7, adult-onset, X-linked, 301075 Deafness, X-linked 4, 300066
SNAI2	100,0%	100,0%	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SOX10	100,0%	100,0%	Waardenburg syndrome, type 4C, 613266 PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SPATA5	100,0%	100,0%	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577
SPNS2	99,9%	99,6%	?Deafness, autosomal recessive 115, 618457
STRC	100,0%	100,0%	Deafness, autosomal recessive 16, 603720
SUCLA2	100,0%	99,9%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SYNE4	100,0%	100,0%	Deafness, autosomal recessive 76, 615540
TBC1D24	100,0%	100,0%	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%	100,0%	Hypothyroidism, congenital, nongoitrous, 8, 301033
TBL1Y	50,0%	50,0%	?Deafness, Y-linked 2, 400047
TCOF1	100,0%	100,0%	Treacher Collins syndrome 1, 154500
TECTA	100,0%	100,0%	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TFAP2A	100,0%	100,0%	Branchiooculofacial syndrome, 113620
THOC1	100,0%	100,0%	No OMIM Disease ID
TIMM8A	100,0%	100,0%	Mohr-Tranebjaerg syndrome, 304700
TJP2	98,8%	98,8%	Hypercholanemia, familial 1, 607748 Cholestasis, progressive familial intrahepatic 4, 615878
TMC1	100,0%	100,0%	Deafness, autosomal dominant 36, 606705 Deafness, autosomal recessive 7, 600974
TMEM132E	100,0%	100,0%	Deafness, autosomal recessive 99, 618481
TMEM43	100,0%	100,0%	Arrhythmogenic right ventricular dysplasia 5, 604400 Auditory neuropathy, autosomal dominant 3, 619832 Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMIE	100,0%	100,0%	Deafness, autosomal recessive 6, 600971
TMPRSS3	100,0%	100,0%	Deafness, autosomal recessive 8/10, 601072
TMTC2	97,5%	97,5%	No OMIM Disease ID
TNC	100,0%	100,0%	Deafness, autosomal dominant 56, 615629
TPRN	97,6%	96,0%	Deafness, autosomal recessive 79, 613307
TRIOBP	100,0%	100,0%	Deafness, autosomal recessive 28, 609823
TRRAP	100,0%	100,0%	?Deafness, autosomal dominant 75, 618778 Developmental delay with or without dysmorphic facies and autism, 618454
TSHZ1	100,0%	100,0%	Aural atresia, congenital, 607842
TUBB4B	100,0%	100,0%	Leber congenital amaurosis with early-onset deafness, 617879
TWNK	100,0%	100,0%	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%	100,0%	Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IA, 203100

USH1C	100,0%	100,0%	Usher syndrome, type 1C, 276904 Deafness, autosomal recessive 18A, 602092
USH1G	100,0%	100,0%	Usher syndrome, type 1G, 606943
USH2A	99,5%	99,5%	Usher syndrome, type 2A, 276901 Retinitis pigmentosa 39, 613809
USP48	100,0%	100,0%	No OMIM Disease ID
WBP2	100,0%	100,0%	Deafness, autosomal recessive 107, 617639
WFS1	100,0%	100,0%	Deafness, autosomal dominant 6/14/38, 600965 ?Cataract 41, 116400 Wolfram-like syndrome, autosomal dominant, 614296 Wolfram syndrome 1, 222300
WHRN	100,0%	100,0%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
XYLT2	96,7%	96,7%	Spondyloocular syndrome, 605822
YAP1	100,0%	100,0%	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433

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.
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 is the chemistry used for WES analysis.
Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with coverage denoting NC are non-protein coding genes.
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 : April 19th , 2022.
This list is accurate for panel version DG 3.4.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors