HEARING IMPAIRMENT GENE PANEL DG 3.00 (239 genes)

Releasedate: 02-12-2020

Gene	*Agilent V5 covered > 10x*	*Agilent V5 covered > 20x*	*TWIST covered > 10x*	*TWIST covered 20x*	*Associated Phenotype description and OMIM disease ID*
ABCC1	98,9	97,9	100	100	?Deafness, autosomal dominant 77, 618915
ABHD12	91,2	85,2	100	99,4	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACTB	99,7	96,1	100	100	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTG1	100	100	100	100	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717
ADCY1	95,2	93,8	98,5	97,9	?Deafness, autosomal recessive 44, 610154
ADGRV1	99,6	98,6	100	100	Usher syndrome, type 2C, 605472 ?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
AIFM1	99,9	98,8	100	100	Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Combined oxidative phosphorylation deficiency 6, 300816 Deafness, X-linked 5, 300614
ALMS1	99,8	99,5	100	100	Alstrom syndrome, 203800
ANLN	98,7	97,5	100	100	Focal segmental glomerulosclerosis 8, 616032
AP1B1	100	99,5	100	100	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150
ARSG	100	99,5	100	100	Usher syndrome, type IV, 618144
ATOH1	100	100	100	100	No OMIM disease ID
ATP1A3	100	99,9	100	100	CAPOS syndrome, 601338 Alternating hemiplegia of childhood 2, 614820 Dystonia-12, 128235
ATP2B2	100	99,9	100	100	{Deafness, autosomal recessive 12, modifier of}, 601386
ATP6V0A4	100	99,9	100	100	Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722

ATP6V1B1	100	100	100	100	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300
ATP6V1B2	100	99,3	100	100	Zimmermann-Laband syndrome 2, 616455 Deafness, congenital, with onychodystrophy, autosomal dominant, 124480
BCAP31	92,6	83,2	100	99,9	Deafness, dystonia, and cerebral hypomyelination, 300475
BCS1L	100	100	100	100	GRACILE syndrome, 603358 Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BDP1	98,8	95,3	100	100	?Deafness, autosomal recessive 112, 618257
BMP4	100	100	100	100	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
BSND	100	100	100	100	Sensorineural deafness with mild renal dysfunction, 602522 Bartter syndrome, type 4a, 602522
BTD	83,1	83	83,1	83,1	Biotinidase deficiency, 253260
CABP2	75,9	68	100	100	Deafness, autosomal recessive 93, 614899
CACNA1D	98	97,9	100	100	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CCDC50	100	99,7	100	100	?Deafness, autosomal dominant 44, 607453
CD151	100	100	100	100	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 [Blood group, Raph], 179620
CD164	99,1	94,8	100	100	?Deafness, autosomal dominant 66, 616969
CDC14A	100	99	100	100	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDC42	97,9	90,9	100	100	Takenouchi-Kosaki syndrome, 616737
CDH23	100	100	100	100	{Pituitary adenoma 5, multiple types}, 617540 Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1D, 601067
CEACAM16	100	99,5	100	100	Deafness, autosomal recessive 113, 618410 Deafness, autosomal dominant 4B, 614614
CEP250	100	99,2	100	100	Cone-rod dystrophy and hearing loss 2, 618358
CEP78	98,9	96,8	100	100	Cone-rod dystrophy and hearing loss, 617236
CHD7	100	99,5	100	100	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800

CHSY1	97,2	95,7	99,7	98	Temtamy preaxial brachydactyly syndrome, 605282
CIB2	99,7	97	100	99,9	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CISD2	83,4	83,4	100	100	Wolfram syndrome 2, 604928
CLDN14	100	99,7	100	100	Deafness, autosomal recessive 29, 614035
CLDN9	100	100	100	100	?Deafness, autosomal recessive 116, 619093
CLIC5	89,9	88	100	100	?Deafness, autosomal recessive 103, 616042
CLPP	100	99,1	100	100	Perrault syndrome 3, 614129
CLRN1	100	99,8	100	100	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
CLRN2	99,7	96,6	100	100	No OMIM disease ID
COA8	81,9	80,7	93,5	93,4	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COCH	95,2	93,2	100	100	?Deafness, autosomal recessive 110, 618094 Deafness, autosomal dominant 9, 601369
COL11A1	96,2	92,8	100	100	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 ?Deafness, autosomal dominant 37, 618533 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis 1, 228520
COL11A2	100	99,7	100	100	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	99,7	100	100	Achondrogenesis, type II or hypochondrogenesis, 200610 Spondyloperipheral dysplasia, 271700 Kniest dysplasia, 156550 Stickler syndrome, type I, 108300 Osteoarthritis with mild chondrodysplasia, 604864 Platyspondylic skeletal dysplasia, Torrance type, 151210 Avascular necrosis of the femoral head, 608805 ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 SED congenita, 183900 Legg-Calve-Perthes disease, 150600

					SMED Strudwick type, 184250 Czech dysplasia, 609162 Stickler sydrome, type I, nonsyndromic ocular, 609508 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Vitreoretinopathy with phalangeal epiphyseal dysplasia, 0
COL4A3	98,7	98	100	100	Alport syndrome 2, autosomal recessive, 203780 Alport syndrome 3, autosomal dominant, 104200 Hematuria, benign familial, 141200
COL4A4	99,9	98,2	100	100	Alport syndrome 2, autosomal recessive, 203780 Hematuria, familial benign, 141200
COL4A5	97,8	89,1	100	99,8	Alport syndrome 1, X-linked, 301050
COL4A6	97,5	93,3	100	99,9	?Deafness, X-linked 6, 300914
COL9A1	100	99,2	100	100	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	99,9	99	100	100	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
COL9A3	98,7	95,5	99,7	98,6	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 {Intervertebral disc disease, susceptibility to}, 603932
CRYL1	100	99,9	100	100	No OMIM disease ID
CRYM	100	99,6	100	100	Deafness, autosomal dominant 40, 616357
DCAF17	98,9	93,3	100	100	Woodhouse-Sakati syndrome, 241080
DCDC2	100	99,9	100	100	Sclerosing cholangitis, neonatal, 617394 Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DIABLO	100	99,9	100	100	Deafness, autosomal dominant 64, 614152
DIAPH1	99,8	99	99,5	98	Seizures, cortical blindness, microcephaly syndrome, 616632 Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900
DIAPH3	99,6	97	100	100	Auditory neuropathy, autosomal dominant, 1, 609129
DLX5	100	99,9	100	100	Split-hand/foot malformation 1, 183600 ?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DMXL2	99,9	99,1	100	100	Developmental and epileptic encephalopathy 81, 618663 ?Deafness, autosomal dominant 71, 617605 ?Polyendocrine-polyneuropathy syndrome, 616113

DSPP	96,8	86,1	100	100	Dentin dysplasia, type II, 125420 Deafness, autosomal dominant 39, with dentinogenesis, 605594 Dentinogenesis imperfecta, Shields type II, 125490 Dentinogenesis imperfecta, Shields type III, 125500
EDN3	98,8	98,8	100	100	Waardenburg syndrome, type 4B, 613265 Central hypoventilation syndrome, congenital, 209880 {Hirschsprung disease, susceptibility to, 4}, 613712
EDNRB	98	93,8	100	100	{Hirschsprung disease, susceptibility to, 2}, 600155 Waardenburg syndrome, type 4A, 277580 ABCD syndrome, 600501
EFNB2	100	99,8	100	100	No OMIM disease ID
ELMOD3	100	99,8	100	100	?Deafness, autosomal recessive 88, 615429
EPS8	97	96,2	100	100	?Deafness, autosomal recessive 102, 615974
EPS8L2	84,5	82,5	88	88	Deafness autosomal recessive 106, 617637
ERAL1	100	99,7	100	100	Perrault syndrome 6, 617565
ESPN	44,6	35,8	100	99,8	?Usher syndrome, type 1M, 618632 Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006
ESRP1	99,9	98,9	100	100	?Deafness, autosomal recessive 109, 618013
ESRRB	96,8	95	100	100	Deafness, autosomal recessive 35, 608565
EXOSC2	100	100	100	100	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EYA1	99,9	99,7	100	100	?Otofaciocervical syndrome, 166780 Anterior segment anomalies with or without cataract, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 Branchiootic syndrome 1, 602588
EYA4	100	99,7	100	100	Deafness, autosomal dominant 10, 601316 ?Cardiomyopathy, dilated, 1J, 605362
FDXR	100	99,3	100	100	Auditory neuropathy and optic atrophy, 617717
FGF3	99,8	95,1	100	100	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGFR3	99,8	97,7	100	99,8	Muenke syndrome, 602849 Nevus, epidermal, somatic, 162900 Thanatophoric dysplasia, type II, 187601 Bladder cancer, somatic, 109800

					CATSHL syndrome, 610474 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000 LADD syndrome, 149730 Achondroplasia, 100800 Thanatophoric dysplasia, type I, 187600 Colorectal cancer, somatic, 114500 Spermatocytic seminoma, somatic, 273300 Cervical cancer, somatic, 603956 SADDAN, 616482
FITM2	100	100	100	100	Siddiqi syndrome, 618635
FOXF2	93,6	86,6	96,4	94,8	No OMIM disease ID
FOXI1	100	100	100	100	Enlarged vestibular aqueduct, 600791
GAB1	100	99,4	100	100	?Deafness, autosomal recessive 26, 605428
GAS2	100	100	100	100	No OMIM disease ID
GATA3	100	100	100	100	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GIPC3	24,8	23	99,9	99,6	Deafness, autosomal recessive 15, 601869
GJB2	100	100	100	100	Deafness, autosomal dominant 3A, 601544 Deafness, autosomal recessive 1A, 220290 Bart-Pumphrey syndrome, 149200 Vohwinkel syndrome, 124500 Keratoderma, palmoplantar, with deafness, 148350 Keratitis-ichthyosis-deafness syndrome, 148210 Hystrix-like ichthyosis with deafness, 602540
GJB3	100	100	100	100	Deafness, autosomal dominant 2B, 612644 Deafness, digenic, GJB2/GJB3, 220290 Erythrokeratodermia variabilis et progressiva 1, 133200 Deafness, autosomal recessive, 0 Deafness, autosomal dominant, with peripheral neuropathy, 0
GJB6	100	100	100	100	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500

GLA	91,1	88,2	91,3	91,3	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GPSM2	99,9	99,2	100	100	Chudley-McCullough syndrome, 604213
GRAP	82,8	78,3	100	100	Deafness, autosomal recessive 114, 618456
GREB1L	100	99,9	100	100	Renal hypodysplasia/aplasia 3, 617805
GRHL2	100	100	100	100	Deafness, autosomal dominant 28, 608641 Corneal dystrophy, posterior polymorphous, 4, 618031 Ectodermal dysplasia/short stature syndrome, 616029
GRXCR1	100	99,8	100	100	Deafness, autosomal recessive 25, 613285
GRXCR2	100	100	100	100	?Deafness, autosomal recessive 101, 615837
GSDME	100	99,2	100	100	Deafness, autosomal dominant 5, 600994
HARS1	100	100	100	100	Usher syndrome type 3B, 614504 Charcot-Marie-Tooth disease, axonal, type 2W, 616625
HARS2	100	100	100	100	Perrault syndrome 2, 614926
HGF	100	99,4	100	100	Deafness, autosomal recessive 39, 608265
HOMER2	99,5	99,4	100	100	?Deafness, autosomal dominant 68, 616707
HSD17B4	95,4	93,1	96,6	96,6	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
IFNLR1	99,4	97,1	100	100	No OMIM disease ID
ILDR1	99,9	98,4	100	100	Deafness, autosomal recessive 42, 609646
KARS1	100	99,9	100	100	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, autosomal recessive 89, 613916
KCNE1	100	100	100	100	Long QT syndrome 5, 613695 Jervell and Lange-Nielsen syndrome 2, 612347
KCNJ10	89,3	89	100	100	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNQ1	93,3	90,6	100	99,8	Long QT syndrome 1, 192500 Jervell and Lange-Nielsen syndrome, 220400 Short QT syndrome 2, 609621 {Long QT syndrome 1, acquired, susceptibility to}, 192500 Atrial fibrillation, familial, 3, 607554
KCNQ4	97	95,7	96,4	93,9	Deafness, autosomal dominant 2A, 600101

KITLG	100	98,5	100	100	Deafness, autosomal dominant 69, unilateral or asymmetric, 616697 Hyperpigmentation with or without hypopigmentation, 145250 [Skin/hair/eye pigmentation 7, blond/brown hair], 611664
LARS2	100	100	100	100	Perrault syndrome 4, 615300 ?Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LHFPL5	100	100	100	100	Deafness, autosomal recessive 67, 610265
LMX1A	100	100	100	100	Deafness, autosomal dominant 7, 601412
LOXHD1	100	99,7	100	100	Deafness, autosomal recessive 77, 613079
LOXL3	100	99,2	100	100	No OMIM disease ID
LRP2	100	99,9	100	100	Donnai-Barrow syndrome, 222448
LRP5	98,5	98,1	100	99,7	van Buchem disease, type 2, 607636 Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 Osteoporosis-pseudoglioma syndrome, 259770 Osteopetrosis, autosomal dominant 1, 607634 {Osteoporosis}, 166710 [Bone mineral density variability 1], 601884
LRTOMT	100	99,2	100	100	Deafness, autosomal recessive 63, 611451
MAN2B1	99,8	97,9	100	100	Mannosidosis, alpha-, types I and II, 248500
MARVELD2	99,2	96,1	100	100	Deafness, autosomal recessive 49, 610153
MCM2	100	100	100	100	?Deafness, autosomal dominant 70, 616968
MET	100	99,5	100	100	{Osteofibrous dysplasia, susceptibility to}, 607278 Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705 Renal cell carcinoma, papillary, 1, familial and somatic, 605074
MGP	98,7	95,1	100	100	Keutel syndrome, 245150
MIA3	99,8	99,1	100	100	No OMIM disease ID
MIR96					Deafness, autosomal dominant 50, 613074
MITF	100	99,9	100	100	COMMAD syndrome, 617306 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456

					Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 Tietz albinism-deafness syndrome, 103500
MPZL2	100	99,9	100	100	Deafness, autosomal recessive 111, 618145
MSRB3	100	99,4	100	100	Deafness, autosomal recessive 74, 613718
MYH14	98,4	94	100	100	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 Deafness, autosomal dominant 4A, 600652
MYH9	100	99,3	100	100	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYO15A	98,8	97	100	99,9	Deafness, autosomal recessive 3, 600316
MYO3A	99,6	96,6	100	100	Deafness, autosomal recessive 30, 607101
MYO6	99,5	96,6	100	100	Deafness, autosomal recessive 37, 607821 Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346
MYO7A	99,3	97,3	100	100	Deafness, autosomal recessive 2, 600060 Deafness, autosomal dominant 11, 601317 Usher syndrome, type 1B, 276900
NARS2	98,3	97,4	100	100	?Deafness, autosomal recessive 94, 618434 Combined oxidative phosphorylation deficiency 24, 616239
NCOA3	99,6	97,4	100	100	No OMIM disease ID
NDP	100	99,7	100	100	Norrie disease, 310600 Exudative vitreoretinopathy 2, X-linked, 305390
NLRP3	100	99,9	100	100	Familial cold inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900 CINCA syndrome, 607115 Deafness, autosomal dominant 34, with or without inflammation, 617772 Keratoendothelitis fugax hereditaria, 148200
NOG	100	100	100	100	Tarsal-carpal coalition syndrome, 186570 Symphalangism, proximal, 1A, 185800 Stapes ankylosis with broad thumbs and toes, 184460 Multiple synostoses syndrome 1, 186500 Brachydactyly, type B2, 611377
OPA1	99,6	97,6	100	100	{Glaucoma, normal tension, susceptibility to}, 606657 Behr syndrome, 210000

					Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
OSBPL2	100	100	100	100	Deafness, autosomal dominant 67, 616340
OTOA	99,4	97,6	100	99,9	Deafness, autosomal recessive 22, 607039
OTOF	100	99,9	100	100	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071
OTOG	99,4	98,6	100	100	Deafness, autosomal recessive 18B, 614945
OTOGL	99,5	97,4	100	100	Deafness, autosomal recessive 84B, 614944
P2RX2	100	100	100	100	Deafness, autosomal dominant 41, 608224
PAX3	100	99,9	100	100	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PCDH15	97,8	96,7	100	100	Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083 Deafness, autosomal recessive 23, 609533
PDE1C	100	99,6	100	100	?Deafness, autosomal dominant 74, 618140
PDZD7	97	93	100	99,8	Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 Deafness, autosomal recessive 57, 618003 {Retinal disease in Usher syndrome type IIA, modifier of}, 276901
PET100	100	99,6	100	100	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PEX1	99,9	99,4	100	100	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX26	100	100	100	100	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX6	94,5	86,7	100	100	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862
PISD	100	100	100	100	Liberfarb syndrome, 618889
PJVK	100	99,7	100	100	Deafness, autosomal recessive 59, 610220
PLOD3	99,8	98	100	100	Lysyl hydroxylase 3 deficiency, 612394

PLS1	100	99,1	100	100	Deafness, autosomal dominant 76, 618787
PNPT1	97,7	89,7	100	100	Deafness, autosomal recessive 70, 614934 Combined oxidative phosphorylation deficiency 13, 614932
POLD1	98,5	95,2	100	100	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 {Colorectal cancer, susceptibility to, 10}, 612591
POLR1C	90,5	87	82,8	82,8	Treacher Collins syndrome 3, 248390 Leukodystrophy, hypomyelinating, 11, 616494
POLR1D	91,6	91,6	100	100	Treacher Collins syndrome 2, 613717
POU3F4	100	100	100	100	Deafness, X-linked 2, 304400
POU4F3	100	100	100	100	Deafness, autosomal dominant 15, 602459
PPIP5K2	98,9	95,2	100	100	Deafness, autosomal recessive 100, 618422
PRKCB	100	100	100	100	No OMIM disease ID
PRORP	100	99,5	100	100	No OMIM disease ID
PRPS1	86,4	86,4	100	100	Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Deafness, X-linked 1, 304500 Arts syndrome, 301835 Gout, PRPS-related, 300661
PSIP1	98,8	93,5	100	100	No OMIM disease ID
PTPRQ	94,6	92,5	92,8	92,7	Deafness, autosomal dominant 73, 617663 Deafness, autosomal recessive 84A, 613391
PTRH2	100	100	100	100	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
RAI1	100	100	100	100	Smith-Magenis syndrome, 182290
RDX	89,1	71,5	100	100	Deafness, autosomal recessive 24, 611022
REST	98,5	98,2	98,6	98,6	{Wilms tumor 6, susceptibility to}, 616806 Fibromatosis, gingival, 5, 617626 ?Deafness, autosomal dominant 27, 612431
RIPOR2	100	99,8	100	100	?Deafness, autosomal recessive 104, 616515
RMND1	100	98,6	100	100	Combined oxidative phosphorylation deficiency 11, 614922
ROBO1	100	99,9	100	100	No OMIM disease ID

ROR1	97	96,8	99,9	99,3	?Deafness, autosomal recessive 108, 617654
RRM2B	100	99,7	100	100	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075
S1PR2	99,4	96,9	100	100	Deafness, autosomal recessive 68, 610419
SCD5	100	99,8	100	100	?Deafness, autosomal dominant 79, 619086
SERPINB6	93,4	93,4	100	100	?Deafness, autosomal recessive 91, 613453
SIX1	100	99,2	100	100	Deafness, autosomal dominant 23, 605192 Branchiootic syndrome 3, 608389
SIX5	95,4	88,2	100	100	Branchiootorenal syndrome 2, 610896
SLC12A1	96,2	96,1	96,2	96,2	Bartter syndrome, type 1, 601678
SLC12A2	94	91,4	100	99,8	Delpire-McNeill syndrome, 619083 Kilquist syndrome, 619080 Deafness, autosomal dominant 78, 619081
SLC17A8	100	100	100	100	Deafness, autosomal dominant 25, 605583
SLC19A2	100	99,7	100	100	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC22A4	100	99,6	100	100	{Rheumatoid arthritis, susceptibility to}, 180300
SLC26A4	100	99,7	100	100	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 Pendred syndrome, 274600
SLC26A5	99,1	96,8	100	100	?Deafness, autosomal recessive 61, 613865
SLC29A3	100	99,6	100	100	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC33A1	99,9	98,9	100	100	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC44A4	100	99,5	100	100	?Deafness, autosomal dominant 72, 617606
SLC52A2	100	100	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100	100	100	100	Brown-Vialetto-Van Laere syndrome 1, 211530 ?Fazio-Londe disease, 211500
SLC9A3R1	100	98,7	100	100	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLITRK6	100	100	100	100	Deafness and myopia, 221200
SMPX	100	97,6	100	100	Deafness, X-linked 4, 300066

SNAI2	100	99,1	100	100	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SOX10	99,9	97,9	100	100	Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136 Waardenburg syndrome, type 4C, 613266
SPATA5	100	99,7	100	100	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPNS2	92,1	89,3	97,6	95,7	?Deafness, autosomal recessive 115, 618457
STRC	99,9	98	100	100	Deafness, autosomal recessive 16, 603720
SUCLA2	89,5	82,2	99,9	99,8	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SYNE4	99,7	97	100	100	Deafness, autosomal recessive 76, 615540
TBC1D24	100	100	100	100	Developmental and epileptic encephalopathy 16, 615338 Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 DOORS syndrome, 220500 Deafness, autosomal dominant 65, 616044 Myoclonic epilepsy, infantile, familial, 605021 Deafness , autosomal recessive 86, 614617
TBL1Y	49,4	45,3	60	59,9	?Deafness, Y-linked 2, 400047
TCOF1	99,7	98,6	100	100	Treacher Collins syndrome 1, 154500
TECTA	100	99,9	100	100	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
THOC1	99,7	97,8	100	100	No OMIM disease ID
TIMM8A	98,1	90,6	100	100	Mohr-Tranebjaerg syndrome, 304700
TJP2	92,8	92,5	98,8	98,8	Cholestasis, progressive familial intrahepatic 4, 615878 Hypercholanemia, familial, 607748
TMC1	99,7	97,3	100	100	Deafness, autosomal recessive 7, 600974 Deafness, autosomal dominant 36, 606705
TMEM132E	96,9	93,5	100	100	Deafness, autosomal recessive 99, 618481
TMIE	99,2	95,1	100	100	Deafness, autosomal recessive 6, 600971
TMPRSS3	100	99,9	100	100	Deafness, autosomal recessive 8/10, 601072
TMTC2	97,5	97,5	97,5	97,5	No OMIM disease ID
TNC	100	99,8	100	100	Deafness, autosomal dominant 56, 615629

TPRN	87,9	79,3	94,4	89,8	Deafness, autosomal recessive 79, 613307
TRIOBP	97,8	96,1	99,9	99,6	Deafness, autosomal recessive 28, 609823
TRRAP	99,9	99,5	100	100	Developmental delay with or without dysmorphic facies and autism, 618454 ?Deafness, autosomal dominant 75, 618778
TSHZ1	98,8	98,8	100	100	Aural atresia, congenital, 607842
TSPEAR	100	99,2	100	100	Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180 ?Deafness, autosomal recessive 98, 614861
TUBB4B	99,9	96,9	100	100	Leber congenital amaurosis with early-onset deafness, 617879
TWNK	100	100	100	100	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	100	100	100	Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IB, 606952 [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800 Albinism, oculocutaneous, type IA, 203100 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800
USH1C	100	99,8	100	100	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
USH1G	99,6	97,9	100	100	Usher syndrome, type 1G, 606943
USH2A	100	99,8	99,5	99,5	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
USP48	99,9	99,3	100	100	No OMIM disease ID
WBP2	100	99,7	100	100	Deafness, autosomal recessive 107, 617639
WFS1	100	99,9	100	100	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 {Diabetes mellitus, noninsulin-dependent, association with}, 125853 Wolfram-like syndrome, autosomal dominant, 614296 Wolfram syndrome 1, 222300
WHRN	99,8	98	100	100	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
XYLT2	100	98,3	96,7	96,7	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 Spondyloocular syndrome, 605822
YAP1	96,4	89,4	100	100	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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID 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-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 20th , 2020.
This list is accurate for panel version DG 3.0.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors