HEARING IMPAIRMENT GENE PANEL DG 2.18 (206 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
ABCC1	98,90%	97,90%	100%	100%	No OMIM disease ID
ABHD12	98,70%	92,30%	100%	99,30%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACTB	99,70%	96,10%	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	95,20%	93,80%	98,50%	97,90%	?Deafness, autosomal recessive 44, 610154
ADGRV1	99,60%	98,60%	100%	100%	Usher syndrome, type 2C, 605472 ?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
AIFM1	99,90%	98,80%	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	99,80%	99,50%	100%	100%	Alstrom syndrome, 203800
AP1B1	100%	99,50%	100%	100%	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150
APOPT1	81,90%	80,70%	93,50%	93,40%	Mitochondrial complex IV deficiency, 220110
ARSG	100%	99,50%	100%	100%	Usher syndrome, type IV, 618144
ATP1A3	100%	99,90%	100%	100%	CAPOS syndrome, 601338 Alternating hemiplegia of childhood 2, 614820 Dystonia-12, 128235
ATP2B2	100%	99,90%	100%	100%	No OMIM disease ID
ATP6V0A4	100%	99,90%	100%	100%	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1B1	100%	100%	100%	100%	Renal tubular acidosis with deafness, 267300
ATP6V1B2	100%	99,30%	100%	100%	Zimmermann-Laband syndrome 2, 616455 Deafness, congenital, with onychodystrophy, autosomal dominant, 124480
BCAP31	92,60%	83,20%	100%	99,90%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCS1L	100%	100%	100%	100%	Leigh syndrome, 256000 GRACILE syndrome, 603358

					Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BDP1	98,80%	95,30%	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%	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
CABP2	75,90%	68,00%	100%	100%	Deafness, autosomal recessive 93, 614899
CACNA1D	98,00%	97,90%	100%	100%	Sinoatrial node dysfunction and deafness, 614896 Primary aldosteronism, seizures, and neurologic abnormalities, 615474
CCDC50	100%	99,70%	100%	100%	?Deafness, autosomal dominant 44, 607453
CD164	99,10%	94,80%	100%	100%	?Deafness, autosomal dominant 66, 616969
CDC14A	100%	98,90%	100%	100%	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDH23	100%	100%	100%	100%	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1D, 601067
CEACAM16	100%	99,50%	100%	100%	Deafness, autosomal recessive 113, 618410 Deafness, autosomal dominant 4B, 614614
CEP250	100%	99,20%	100%	100%	Cone-rod dystrophy and hearing loss 2, 618358
CEP78	99,70%	97,60%	100%	100%	Cone-rod dystrophy and hearing loss, 617236
CHD7	100%	99,50%	100%	100%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CIB2	99,70%	97,00%	100%	99,90%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CISD2	83,40%	83,40%	100%	100%	Wolfram syndrome 2, 604928
CLDN14	100%	99,70%	100%	100%	Deafness, autosomal recessive 29, 614035
CLDN9	100%	100%	100%	100%	No OMIM disease ID
CLIC5	100%	99,90%	100%	100%	?Deafness, autosomal recessive 103, 616042
CLPP	100%	99,10%	100%	100%	Perrault syndrome 3, 614129
CLRN1	100%	99,80%	100%	100%	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
COCH	100%	99,90%	100%	100%	?Deafness, autosomal recessive 110, 618094 Deafness, autosomal dominant 9, 601369
COL11A1	99,20%	95,70%	100%	100%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 ?Deafness, autosomal dominant 37, 618533 Fibrochondrogenesis 1, 228520

COL11A2	100%	99,50%	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%	99,70%	100%	100%	Achondrogenesis, type II or hypochondrogenesis, 200610 Spondyloperipheral dysplasia, 271700 Kniest dysplasia, 156550 Stickler syndrome, type I, 108300 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Osteoarthritis with mild chondrodysplasia, 604864 Platyspondylic skeletal dysplasia, Torrance type, 151210 Avascular necrosis of the femoral head, 608805 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	98,70%	98,00%	100%	100%	Hematuria, benign familial, 141200 Alport syndrome 2, autosomal recessive, 203780 Alport syndrome 3, autosomal dominant, 104200
COL4A4	99,90%	98,20%	100%	100%	Alport syndrome 2, autosomal recessive, 203780 Hematuria, familial benign, 141200
COL4A5	97,80%	89,00%	100%	99,80%	Alport syndrome 1, X-linked, 301050
COL4A6	97,50%	93,30%	100%	99,90%	?Deafness, X-linked 6, 300914
COL9A1	100%	99,20%	100%	100%	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	99,90%	99,00%	100%	100%	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
COL9A3	98,70%	95,50%	99,70%	98,60%	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969
CRYL1	100%	99,90%	100%	100%	No OMIM disease ID
CRYM	100%	99,60%	100%	100%	Deafness, autosomal dominant 40, 616357
DCDC2	100%	99,90%	100%	100%	Sclerosing cholangitis, neonatal, 617394 Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DFNA5	100%	99,20%	100%	100%	Deafness, autosomal dominant 5, 600994

DFNB59	100%	99,70%	100%	100%	Deafness, autosomal recessive 59, 610220
DIABLO	100%	99,80%	100%	100%	Deafness, autosomal dominant 64, 614152
DIAPH1	99,80%	99,00%	99,50%	97,90%	Seizures, cortical blindness, microcephaly syndrome, 616632 Deafness, autosomal dominant 1, 124900
DIAPH3	99,60%	97,00%	100%	100%	Auditory neuropathy, autosomal dominant, 1, 609129
DMXL2	99,90%	99,10%	100%	100%	?Deafness, autosomal dominant 71, 617605 ?Polyendocrine-polyneuropathy syndrome, 616113 Epileptic encephalopathy, early infantile, 81, 618663
DSPP	96,80%	86,10%	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	100%	99,90%	100%	100%	Waardenburg syndrome, type 4B, 613265 Central hypoventilation syndrome, congenital, 209880
EDNRB	98,00%	93,80%	100%	100%	Waardenburg syndrome, type 4A, 277580 ABCD syndrome, 600501
EFNB2	100%	99,80%	100%	100%	No OMIM disease ID
ELMOD3	100%	99,80%	100%	100%	?Deafness, autosomal recessive 88, 615429
EPS8	100%	99,10%	100%	100%	?Deafness, autosomal recessive 102, 615974
EPS8L2	96,10%	93,70%	100%	100%	Deafness autosomal recessive 106, 617637
ERAL1	100%	99,70%	100%	100%	Perrault syndrome 6, 617565
ESPN	44,60%	35,80%	100%	99,80%	?Usher syndrome, type 1M, 618632 Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006
ESRP1	99,90%	98,90%	100%	100%	?Deafness, autosomal recessive 109, 618013
ESRRB	99,90%	98,00%	100%	100%	Deafness, autosomal recessive 35, 608565
EXOSC2	100%	100%	100%	100%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EYA1	99,90%	99,70%	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%	99,70%	100%	100%	Deafness, autosomal dominant 10, 601316 ?Cardiomyopathy, dilated, 1J, 605362
FGF3	99,80%	95,10%	100%	100%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FOXF2	93,60%	86,60%	96,40%	94,80%	No OMIM disease ID
FOXI1	100%	100%	100%	100%	Enlarged vestibular aqueduct, 600791
GAB1	100%	99,40%	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	97,50%	94,20%	99,60%	98,10%	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
GPSM2	99,90%	99,20%	100%	100%	Chudley-McCullough syndrome, 604213
GRAP	82,80%	78,30%	100%	100%	Deafness, autosomal recessive 114, 618456
GREB1L	100%	99,90%	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%	99,80%	100%	100%	Deafness, autosomal recessive 25, 613285
GRXCR2	100%	100%	100%	100%	?Deafness, autosomal recessive 101, 615837
HARS	100%	100%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HARS2	100%	100%	100%	100%	?Perrault syndrome 2, 614926
HGF	100%	99,40%	100%	100%	Deafness, autosomal recessive 39, 608265
HOMER2	99,50%	99,40%	100%	100%	?Deafness, autosomal dominant 68, 616707
HSD17B4	96,00%	93,70%	96,60%	96,60%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
IFNLR1	99,40%	97,10%	100%	100%	No OMIM disease ID
ILDR1	99,90%	98,50%	100%	100%	Deafness, autosomal recessive 42, 609646
KARS	100%	99,90%	100%	100%	Deafness, autosomal recessive 89, 613916 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641

KCNE1	100%	100%	100%	100%	Long QT syndrome 5, 613695 Jervell and Lange-Nielsen syndrome 2, 612347
KCNJ10	89,30%	89,00%	100%	100%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNQ1	95,50%	94,20%	100%	99,80%	Long QT syndrome 1, 192500 Jervell and Lange-Nielsen syndrome, 220400 Short QT syndrome 2, 609621 Atrial fibrillation, familial, 3, 607554
KCNQ4	97,00%	95,70%	96,40%	93,90%	Deafness, autosomal dominant 2A, 600101
KITLG	100%	98,50%	100%	100%	Hyperpigmentation with or without hypopigmentation, 145250 Deafness, autosomal dominant 69, unilateral or asymmetric, 616697
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%	99,70%	100%	100%	Deafness, autosomal recessive 77, 613079
LOXL3	100%	99,20%	100%	100%	No OMIM disease ID
LRP5	98,50%	98,10%	100%	99,70%	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
LRTOMT	99,30%	94,20%	93,30%	93,10%	Deafness, autosomal recessive 63, 611451
MARVELD2	99,20%	96,10%	100%	100%	Deafness, autosomal recessive 49, 610153
MCM2	100%	100%	100%	100%	?Deafness, autosomal dominant 70, 616968
MET	100%	99,50%	100%	100%	Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705 Renal cell carcinoma, papillary, 1, familial and somatic, 605074
MIA3	99,80%	99,10%	100%	100%	No OMIM disease ID
MIR96	NC	NC	NC	NC	Deafness, autosomal dominant 50, 613074
MITF	100%	99,90%	100%	100%	COMMAD syndrome, 617306 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 Tietz albinism-deafness syndrome, 103500
MPZL2	100%	99,90%	100%	100%	Deafness, autosomal recessive 111, 618145
MSRB3	100%	99,40%	100%	100%	Deafness, autosomal recessive 74, 613718

MYH14	98,40%	94,00%	100%	100%	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 Deafness, autosomal dominant 4A, 600652
MYH9	100%	99,30%	100%	100%	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYO15A	98,80%	97,00%	100%	99,90%	Deafness, autosomal recessive 3, 600316
MYO3A	99,60%	96,60%	100%	100%	Deafness, autosomal recessive 30, 607101
MYO6	99,50%	96,60%	100%	100%	Deafness, autosomal recessive 37, 607821 Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346
MYO7A	99,30%	97,40%	100%	100%	Deafness, autosomal recessive 2, 600060 Deafness, autosomal dominant 11, 601317 Usher syndrome, type 1B, 276900
NARS2	98,30%	97,40%	100%	100%	?Deafness, autosomal recessive 94, 618434 Combined oxidative phosphorylation deficiency 24, 616239
NLRP3	100%	99,90%	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	99,70%	97,60%	100%	100%	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	99,40%	97,60%	100%	99,90%	Deafness, autosomal recessive 22, 607039
OTOF	100%	99,90%	100%	100%	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071
OTOG	99,40%	98,60%	100%	99,90%	Deafness, autosomal recessive 18B, 614945
OTOGL	99,50%	97,40%	100%	100%	Deafness, autosomal recessive 84B, 614944
P2RX2	100%	100%	100%	100%	Deafness, autosomal dominant 41, 608224
PAX3	100%	99,90%	100%	100%	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220

					Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PCDH15	98,60%	97,50%	100%	100%	Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083 Deafness, autosomal recessive 23, 609533
PDE1C	100%	99,60%	100%	100%	?Deafness, autosomal dominant 74, 618140
PDZD7	98,80%	96,30%	100%	100%	Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 Deafness, autosomal recessive 57, 618003
PET100	100%	99,60%	100%	100%	Mitochondrial complex IV deficiency, 220110
PEX1	99,90%	99,40%	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	94,50%	86,70%	100%	100%	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862
PLOD3	99,80%	98,00%	100%	100%	Lysyl hydroxylase 3 deficiency, 612394
PLS1	100%	99,10%	100%	100%	Deafness, autosomal dominant 76, 618787
PNPT1	97,70%	89,70%	100%	100%	Deafness, autosomal recessive 70, 614934 Combined oxidative phosphorylation deficiency 13, 614932
POLD1	98,50%	95,20%	100%	100%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381
POLR1C	99,30%	95,50%	90,70%	90,70%	Treacher Collins syndrome 3, 248390 Leukodystrophy, hypomyelinating, 11, 616494
POLR1D	91,60%	91,60%	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	98,90%	95,20%	100%	100%	Deafness, autosomal recessive 100, 618422
PRKCB	100%	99,90%	100%	100%	No OMIM disease ID
PRPS1	100%	100%	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
PTPRQ	94,60%	92,50%	92,80%	92,70%	Deafness, autosomal dominant 73, 617663 Deafness, autosomal recessive 84A, 613391
RAI1	100%	100%	100%	100%	Smith-Magenis syndrome, 182290
RDX	89,10%	71,50%	100%	100%	Deafness, autosomal recessive 24, 611022

REST	98,50%	98,20%	98,60%	98,60%	Fibromatosis, gingival, 5, 617626 ?Deafness, autosomal dominant 27, 612431
RIPOR2	100%	99,80%	100%	100%	?Deafness, autosomal recessive 104, 616515
ROBO1	100%	99,90%	100%	100%	No OMIM disease ID
ROR1	97,00%	96,80%	99,90%	99,30%	?Deafness, autosomal recessive 108, 617654
S1PR2	99,40%	96,90%	100%	100%	Deafness, autosomal recessive 68, 610419
SCD5	100%	99,80%	100%	100%	No OMIM disease ID
SERPINB6	95,90%	95,90%	100%	100%	?Deafness, autosomal recessive 91, 613453
SIX1	100%	99,20%	100%	100%	Deafness, autosomal dominant 23, 605192 Branchiootic syndrome 3, 608389
SIX5	95,40%	88,20%	100%	100%	Branchiootorenal syndrome 2, 610896
SLC12A1	100%	99,90%	100%	100%	Bartter syndrome, type 1, 601678
SLC17A8	100%	100%	100%	100%	Deafness, autosomal dominant 25, 605583
SLC19A2	100%	99,70%	100%	100%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC22A4	100%	99,60%	100%	100%	No OMIM disease ID
SLC25A2	100%	100%	100%	100%	No OMIM disease ID
SLC26A4	100%	99,70%	100%	100%	Pendred syndrome, 274600 Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791
SLC26A5	99,10%	96,80%	100%	100%	?Deafness, autosomal recessive 61, 613865
SLC29A3	100%	99,60%	100%	100%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC33A1	99,90%	98,90%	100%	100%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC44A4	100%	99,50%	100%	100%	?Deafness, autosomal dominant 72, 617606
SLC9A3R1	100%	98,70%	100%	100%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLITRK6	100%	100%	100%	100%	Deafness and myopia, 221200
SMPX	100%	97,60%	100%	100%	Deafness, X-linked 4, 300066
SNAI2	100%	99,10%	100%	100%	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SOX10	99,90%	97,90%	100%	100%	Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136 Waardenburg syndrome, type 4C, 613266
SPATA5	100%	99,70%	100%	100%	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPNS2	92,10%	89,30%	97,60%	95,70%	?Deafness, autosomal recessive 115, 618457
STRC	99,90%	98,00%	100%	100%	Deafness, autosomal recessive 16, 603720
SYNE4	99,70%	97,00%	100%	100%	Deafness, autosomal recessive 76, 615540
TBC1D24	100%	100%	100%	100%	Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 DOORS syndrome, 220500

					Deafness, autosomal dominant 65, 616044 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021 Deafness , autosomal recessive 86, 614617
TCOF1	99,70%	98,60%	100%	100%	Treacher Collins syndrome 1, 154500
TECTA	100%	99,90%	100%	100%	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TIMM8A	98,00%	90,10%	100%	100%	Mohr-Tranebjaerg syndrome, 304700
TJP2	94,00%	93,60%	100%	100%	Hypercholanemia, familial, 607748 Cholestasis, progressive familial intrahepatic 4, 615878
TMC1	99,70%	97,10%	100%	100%	Deafness, autosomal recessive 7, 600974 Deafness, autosomal dominant 36, 606705
TMEM132E	96,90%	93,50%	100%	100%	?Deafness, autosomal recessive 99, 618481
TMIE	99,20%	95,10%	100%	100%	Deafness, autosomal recessive 6, 600971
TMPRSS3	100%	99,90%	100%	100%	Deafness, autosomal recessive 8/10, 601072
TMTC2	97,50%	97,50%	97,50%	97,50%	No OMIM disease ID
TNC	100%	99,80%	100%	100%	Deafness, autosomal dominant 56, 615629
TPRN	87,90%	79,30%	94,40%	89,80%	Deafness, autosomal recessive 79, 613307
TRIOBP	97,80%	96,10%	99,90%	99,60%	Deafness, autosomal recessive 28, 609823
TRRAP	99,90%	99,50%	100%	100%	Developmental delay with or without dysmorphic facies and autism, 618454 ?Deafness, autosomal dominant 75, 618778
TSPEAR	100%	99,20%	97,90%	97,90%	Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180 ?Deafness, autosomal recessive 98, 614861
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 Albinism, oculocutaneous, type IA, 203100
USH1C	100%	99,80%	100%	100%	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
USH1G	99,60%	97,90%	100%	100%	Usher syndrome, type 1G, 606943
USH2A	100%	99,80%	100%	100%	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
WBP2	100%	99,70%	100%	100%	Deafness, autosomal recessive 107, 617639
WFS1	100%	99,90%	100%	100%	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965

					Wolfram-like syndrome, autosomal dominant, 614296 Wolfram syndrome 1, 222300
WHRN	99,80%	98,10%	100%	100%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
YAP1	96,40%	89,40%	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 : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors