HEARING IMPAIRMENT GENE PANEL DG 2.17 (173 genes)

Releasedate: 06-12-2019

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ACTB	92.6	100.0%	99.9%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTG1	131.2	100.0%	100.0%	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717
ADCY1	146.1	97.7%	96.3%	?Deafness, autosomal recessive 44, 610154
ADGRV1	126.0	99.8%	98.3%	Usher syndrome, type 2C, 605472 ?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
AIFM1	92.9	99.7%	96.5%	Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Combined oxidative phosphorylation deficiency 6, 300816 Deafness, X-linked 5, 300614
APOPT1	80.4	82.1%	82.1%	Mitochondrial complex IV deficiency, 220110
ATP1A3	173.9	100.0%	100.0%	CAPOS syndrome, 601338 Alternating hemiplegia of childhood 2, 614820 Dystonia-12, 128235
ATP2B2	178.7	100.0%	99.9%	No OMIM disease ID
ATP6V1B1	184.8	100.0%	100.0%	Renal tubular acidosis with deafness, 267300
BCS1L	160.0	100.0%	100.0%	Leigh syndrome, 256000 GRACILE syndrome, 603358 Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BDP1	136.2	98.4%	94.0%	?Deafness, autosomal recessive 112, 618257
BMP4	192.0	100.0%	100.0%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
BSND	150.8	100.0%	100.0%	Sensorineural deafness with mild renal dysfunction, 602522 Bartter syndrome, type 4a, 602522
CABP2	85.6	80.9%	74.6%	Deafness, autosomal recessive 93, 614899
CACNA1D	135.3	98.0%	97.8%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896

CCDC50	124.0	100.0%	99.8%	?Deafness, autosomal dominant 44, 607453
CD164	124.5	98.6%	93.9%	?Deafness, autosomal dominant 66, 616969
CDC14A	155.4	99.8%	97.3%	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDH23	186.7	100.0%	100.0%	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1D, 601067
CEACAM16	139.8	100.0%	100.0%	Deafness, autosomal recessive 113, 618410 Deafness, autosomal dominant 4B, 614614
CEP78	123.1	99.8%	96.9%	Cone-rod dystrophy and hearing loss, 617236
CIB2	218.3	100.0%	99.5%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CLDN14	121.5	100.0%	100.0%	Deafness, autosomal recessive 29, 614035
CLIC5	103.2	100.0%	99.9%	?Deafness, autosomal recessive 103, 616042
CLPP	152.0	100.0%	99.6%	Perrault syndrome 3, 614129
CLRN1	140.6	100.0%	99.5%	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
COCH	163.5	100.0%	100.0%	?Deafness, autosomal recessive 110, 618094 Deafness, autosomal dominant 9, 601369
COL11A1	94.6	98.0%	93.6%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 ?Deafness, autosomal dominant 37, 618533 Fibrochondrogenesis 1, 228520
COL11A2	122.3	100.0%	99.5%	Deafness, autosomal dominant 13, 601868 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150 Fibrochondrogenesis 2, 614524 Deafness, autosomal recessive 53, 609706 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840
COL2A1	121.1	100.0%	99.8%	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	93.8	99.6%	97.9%	Hematuria, benign familial, 141200 Alport syndrome 2, autosomal recessive, 203780 Alport syndrome 3, autosomal dominant, 104200
COL4A4	96.5	99.7%	98.0%	Alport syndrome 2, autosomal recessive, 203780 Hematuria, familial benign, 141200
COL4A5	57.3	97.4%	87.3%	Alport syndrome 1, X-linked, 301050
COL4A6	85.2	97.2%	92.1%	?Deafness, X-linked 6, 300914
COL9A1	132.9	100.0%	99.5%	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	104.9	100.0%	99.6%	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
CRYM	87.0	99.8%	97.5%	Deafness, autosomal dominant 40, 616357
DCDC2	158.0	100.0%	99.9%	Sclerosing cholangitis, neonatal, 617394 Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DFNA5	95.9	100.0%	99.4%	Deafness, autosomal dominant 5, 600994
DFNB59	116.0	100.0%	99.2%	Deafness, autosomal recessive 59, 610220
DIABLO	176.3	100.0%	99.8%	Deafness, autosomal dominant 64, 614152
DIAPH1	104.4	100.0%	99.8%	Seizures, cortical blindness, microcephaly syndrome, 616632 Deafness, autosomal dominant 1, 124900
DIAPH3	80.8	99.8%	96.0%	Auditory neuropathy, autosomal dominant, 1, 609129
DMXL2	151.5	99.8%	98.6%	?Deafness, autosomal dominant 71, 617605 ?Polyendocrine-polyneuropathy syndrome, 616113 Epileptic encephalopathy, early infantile, 81, 618663
DSPP	83.9	98.8%	95.5%	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	147.0	100.0%	100.0%	Waardenburg syndrome, type 4B, 613265 Central hypoventilation syndrome, congenital, 209880
EDNRB	126.2	96.4%	91.5%	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580
ELMOD3	144.7	100.0%	99.8%	?Deafness, autosomal recessive 88, 615429
EPS8	114.5	99.7%	96.2%	?Deafness, autosomal recessive 102, 615974

EPS8L2	180.6	99.8%	97.0%	Deafness autosomal recessive 106, 617637
ERAL1	168.0	100.0%	100.0%	Perrault syndrome 6, 617565
ESPN	35.1	55.8%	45.1%	?Usher syndrome, type 1M, 618632 Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006
ESRP1	96.9	99.9%	98.8%	?Deafness, autosomal recessive 109, 618013
ESRRB	131.0	100.0%	99.6%	Deafness, autosomal recessive 35, 608565
EXOSC2	114.1	100.0%	99.9%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EYA1	121.6	100.0%	99.9%	?Otofaciocervical syndrome, 166780 Anterior segment anomalies with or without cataract, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 Branchiootic syndrome 1, 602588
EYA4	134.5	100.0%	100.0%	Deafness, autosomal dominant 10, 601316 ?Cardiomyopathy, dilated, 1J, 605362
FGF3	160.1	100.0%	100.0%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FOXF2	128.2	96.8%	93.8%	No OMIM Disease ID
FOXI1	209.3	100.0%	100.0%	Enlarged vestibular aqueduct, 600791
GAB1	154.4	100.0%	99.8%	?Deafness, autosomal recessive 26, 605428
GATA3	247.4	100.0%	100.0%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GIPC3	141.7	99.4%	97.0%	Deafness, autosomal recessive 15, 601869
GJB2	151.0	100.0%	100.0%	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	245.5	100.0%	100.0%	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	146.2	100.0%	100.0%	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GPSM2	121.0	100.0%	99.4%	Chudley-McCullough syndrome, 604213
GRAP	89.5	87.5%	81.2%	Deafness, autosomal recessive 114, 618456

GREB1L	133.2	100.0%	99.4%	Renal hypodysplasia/aplasia 3, 617805
GRHL2	119.8	100.0%	100.0%	Deafness, autosomal dominant 28, 608641 Corneal dystrophy, posterior polymorphous, 4, 618031 Ectodermal dysplasia/short stature syndrome, 616029
GRXCR1	158.6	100.0%	99.9%	Deafness, autosomal recessive 25, 613285
GRXCR2	119.1	100.0%	100.0%	?Deafness, autosomal recessive 101, 615837
HARS	142.4	100.0%	100.0%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HARS2	141.0	100.0%	99.9%	?Perrault syndrome 2, 614926
HGF	134.3	99.9%	99.3%	Deafness, autosomal recessive 39, 608265
HOMER2	120.6	99.8%	98.5%	?Deafness, autosomal dominant 68, 616707
HSD17B4	106.4	95.5%	93.1%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
IFNLR1	109.4	99.9%	99.6%	No OMIM Disease ID
ILDR1	122.9	99.9%	98.6%	Deafness, autosomal recessive 42, 609646
KARS	109.9	100.0%	99.3%	Deafness, autosomal recessive 89, 613916 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641
KCNE1	398.8	100.0%	100.0%	Long QT syndrome 5, 613695 Jervell and Lange-Nielsen syndrome 2, 612347
KCNJ10	157.5	89.3%	88.6%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNQ1	150.5	98.9%	96.5%	Long QT syndrome 1, 192500 Jervell and Lange-Nielsen syndrome, 220400 Short QT syndrome 2, 609621 Atrial fibrillation, familial, 3, 607554
KCNQ4	182.4	99.8%	99.0%	Deafness, autosomal dominant 2A, 600101
KITLG	82.5	99.8%	96.5%	Hyperpigmentation with or without hypopigmentation, 145250 Deafness, autosomal dominant 69, unilateral or asymmetric, 616697
LARS2	128.3	100.0%	100.0%	Perrault syndrome 4, 615300 ?Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LHFPL5	228.8	100.0%	100.0%	Deafness, autosomal recessive 67, 610265
LMX1A	113.4	100.0%	99.9%	No OMIM Disease ID
LOXHD1	120.2	100.0%	99.6%	Deafness, autosomal recessive 77, 613079
LRP5	183.1	99.9%	99.4%	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	123.2	99.8%	96.2%	Deafness, autosomal recessive 63, 611451
MARVELD2	143.1	98.7%	95.2%	Deafness, autosomal recessive 49, 610153
MCM2	164.5	100.0%	100.0%	?Deafness, autosomal dominant 70, 616968
MET	153.7	100.0%	99.6%	Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705 Renal cell carcinoma, papillary, 1, familial and somatic, 605074
MIR96	NC	NC	NC	Deafness, autosomal dominant 50, 613074
MITF	145.6	100.0%	100.0%	COMMAD syndrome, 617306 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 Tietz albinism-deafness syndrome, 103500
MPZL2	92.7	100.0%	99.8%	Deafness, autosomal recessive 111, 618145
MSRB3	138.7	99.9%	99.3%	Deafness, autosomal recessive 74, 613718
MYH14	120.4	99.4%	96.3%	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 Deafness, autosomal dominant 4A, 600652
MYH9	140.9	99.7%	99.0%	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYO15A	159.8	100.0%	99.6%	Deafness, autosomal recessive 3, 600316
MYO3A	112.5	99.4%	95.4%	Deafness, autosomal recessive 30, 607101
MYO6	98.8	99.5%	95.6%	Deafness, autosomal recessive 37, 607821 Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346
MYO7A	134.7	99.9%	99.1%	Deafness, autosomal recessive 2, 600060 Deafness, autosomal dominant 11, 601317 Usher syndrome, type 1B, 276900
NARS2	121.5	97.6%	97.4%	?Deafness, autosomal recessive 94, 618434 Combined oxidative phosphorylation deficiency 24, 616239
NLRP3	146.0	100.0%	99.9%	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
OPA1	121.4	99.7%	97.5%	Behr syndrome, 210000 Optic atrophy 1, 165500

				Optic atrophy plus syndrome, 125250 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
OSBPL2	146.5	100.0%	100.0%	Deafness, autosomal dominant 67, 616340
OTOA	101.3	99.7%	98.4%	Deafness, autosomal recessive 22, 607039
OTOF	148.9	100.0%	99.8%	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071
OTOG	147.3	99.7%	99.1%	Deafness, autosomal recessive 18B, 614945
OTOGL	101.2	99.4%	96.7%	Deafness, autosomal recessive 84B, 614944
P2RX2	193.5	100.0%	100.0%	Deafness, autosomal dominant 41, 608224
PAX3	116.1	100.0%	99.9%	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PCDH15	139.4	99.2%	98.9%	Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083 Deafness, autosomal recessive 23, 609533
PDE1C	110.4	99.9%	99.4%	?Deafness, autosomal dominant 74, 618140
PDZD7	103.9	99.7%	98.4%	Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 Deafness, autosomal recessive 57, 618003
PET100	95.2	99.7%	90.6%	Mitochondrial complex IV deficiency, 220110
PEX1	126.3	100.0%	99.1%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX26	105.1	100.0%	100.0%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX6	117.6	99.1%	93.9%	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862
PNPT1	54.5	96.4%	83.0%	Deafness, autosomal recessive 70, 614934 Combined oxidative phosphorylation deficiency 13, 614932
POU3F4	148.1	100.0%	100.0%	Deafness, X-linked 2, 304400
POU4F3	297.4	100.0%	100.0%	Deafness, autosomal dominant 15, 602459
PPIP5K2	84.4	99.0%	93.7%	Deafness, autosomal recessive 100, 618422
PRKCB	145.0	100.0%	99.9%	No OMIM Disease ID
PRPS1	113.2	100.0%	99.9%	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	97.7	94.6%	91.6%	Deafness, autosomal dominant 73, 617663 Deafness, autosomal recessive 84A, 613391
RAI1	216.5	100.0%	100.0%	Smith-Magenis syndrome, 182290
RDX	37.2	87.1%	66.8%	Deafness, autosomal recessive 24, 611022
REST	123.0	98.5%	98.4%	Fibromatosis, gingival, 5, 617626 ?Deafness, autosomal dominant 27, 612431
RIPOR2	114.9	100.0%	99.9%	?Deafness, autosomal recessive 104, 616515
ROR1	154.0	98.9%	97.3%	?Deafness, autosomal recessive 108, 617654
S1PR2	225.1	99.7%	97.7%	Deafness, autosomal recessive 68, 610419
SERPINB6	142.3	95.9%	95.9%	?Deafness, autosomal recessive 91, 613453
SIX1	150.4	99.9%	99.2%	Deafness, autosomal dominant 23, 605192 Branchiootic syndrome 3, 608389
SIX5	87.2	100.0%	98.9%	Branchiootorenal syndrome 2, 610896
SLC17A8	123.8	100.0%	100.0%	Deafness, autosomal dominant 25, 605583
SLC22A4	123.4	99.9%	98.5%	No OMIM disease ID
SLC25A2	230.6	100.0%	100.0%	No OMIM Disease ID
SLC26A4	115.6	100.0%	99.7%	Pendred syndrome, 274600 Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791
SLC26A5	132.5	99.2%	96.0%	?Deafness, autosomal recessive 61, 613865
SLC29A3	190.1	100.0%	99.7%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC33A1	135.7	99.8%	97.0%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC44A4	122.2	100.0%	99.5%	?Deafness, autosomal dominant 72, 617606
SLITRK6	170.3	100.0%	100.0%	Deafness and myopia, 221200
SMPX	64.6	100.0%	96.6%	Deafness, X-linked 4, 300066
SNAI2	106.3	100.0%	99.1%	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SOX10	101.7	100.0%	99.8%	Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136 Waardenburg syndrome, type 4C, 613266
SPATA5	142.5	100.0%	99.8%	Epilepsy, hearing loss, and mental retardation syndrome, 616577
STRC	109.0	100.0%	99.1%	Deafness, autosomal recessive 16, 603720
SYNE4	92.6	100.0%	99.2%	Deafness, autosomal recessive 76, 615540
TBC1D24	199.9	100.0%	100.0%	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
TECTA	176.7	100.0%	99.9%	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TIMM8A	50.3	95.4%	80.0%	Mohr-Tranebjaerg syndrome, 304700
TJP2	114.9	94.0%	93.6%	Hypercholanemia, familial, 607748 Cholestasis, progressive familial intrahepatic 4, 615878
TMC1	111.5	99.9%	97.4%	Deafness, autosomal recessive 7, 600974 Deafness, autosomal dominant 36, 606705
TMEM132E	139.1	99.9%	98.0%	?Deafness, autosomal recessive 99, 618481
TMIE	112.7	100.0%	100.0%	Deafness, autosomal recessive 6, 600971
TMPRSS3	103.4	100.0%	99.6%	Deafness, autosomal recessive 8/10, 601072
TMTC2	143.2	97.5%	97.5%	No OMIM Disease ID
TNC	154.7	100.0%	99.9%	Deafness, autosomal dominant 56, 615629
TPRN	113.1	92.8%	88.3%	Deafness, autosomal recessive 79, 613307
TRIOBP	173.9	99.6%	98.1%	Deafness, autosomal recessive 28, 609823
TSPEAR	151.2	100.0%	99.9%	Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180 ?Deafness, autosomal recessive 98, 614861
TYR	153.5	100.0%	100.0%	Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IB, 606952 Albinism, oculocutaneous, type IA, 203100
USH1C	99.1	100.0%	99.3%	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
USH1G	221.0	99.9%	99.3%	Usher syndrome, type 1G, 606943
USH2A	130.8	100.0%	99.8%	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
WBP2	101.8	100.0%	99.5%	Deafness, autosomal recessive 107, 617639
WFS1	210.0	100.0%	99.8%	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 Wolfram-like syndrome, autosomal dominant, 614296 Wolfram syndrome 1, 222300
WHRN	145.9	100.0%	99.3%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
YAP1	97.9	98.5%	94.0%	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.
Median Coverage describes the average number of reads seen across 50 exomes.
% 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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : December 11th , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors