VISION DISORDERS GENE PANEL DG 3.1.0 (466 genes)

Releasedate: 23-03-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
ABCA4	99,9	99,3	96,5	96,5	Retinal dystrophy, early-onset severe, 248200 Stargardt disease 1, 248200 Fundus flavimaculatus, 248200 {Macular degeneration, age-related, 2}, 153800 Cone-rod dystrophy 3, 604116 Retinitis pigmentosa 19, 601718
ABCC6	93,6	92,4	100	100	Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850 Arterial calcification, generalized, of infancy, 2, 614473
ABHD12	91,2	85,2	100	99,4	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACBD5	100	99,2	100	100	Retinal dystrophy with leukodystrophy, 618863
ACO2	96,3	90,3	100	100	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ADAM9	99,8	99,1	100	100	Cone-rod dystrophy 9, 612775
ADAMTS18	100	99,7	100	100	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTSL4	100	99,2	100	100	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
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
ADIPOR1	99,9	97,8	100	100	No OMIM disease ID
AFG3L2	95	91,1	100	99,9	Spastic ataxia 5, autosomal recessive, 614487 Optic atrophy 12, 618977 Spinocerebellar ataxia 28, 610246
AGBL1	98,5	98,4	100	100	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGBL5	99,9	99,3	100	100	Retinitis pigmentosa 75, 617023
AGK	90,6	88,6	91,2	91,2	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691
AHI1	99,7	97,9	100	100	Joubert syndrome 3, 608629

AHR	99,2	98,8	100	100	?Retinitis pigmentosa 85, 618345
AIPL1	100	99,8	100	100	Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393 Cone-rod dystrophy, 604393
ALDH18A1	100	99,9	100	100	Cutis laxa, autosomal recessive, type IIIA, 219150 Cutis laxa, autosomal dominant 3, 616603 Spastic paraplegia 9B, autosomal recessive, 616586 Spastic paraplegia 9A, autosomal dominant, 601162
ALDH1A3	97,2	94,5	100	99,9	Microphthalmia, isolated 8, 615113
ALMS1	99,8	99,5	100	100	Alstrom syndrome, 203800
AP3B1	99,2	95,8	100	100	Hermansky-Pudlak syndrome 2, 608233
AP3D1	99,8	98,6	100	100	?Hermansky-Pudlak syndrome 10, 617050
ARHGEF18	95,4	92,3	100	100	Retinitis pigmentosa 78, 617433
ARL13B	100	99,2	100	100	Joubert syndrome 8, 612291
ARL2	100	100	100	100	?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 1, 619082
ARL2BP	95,9	88,3	100	100	Retinitis pigmentosa with or without situs inversus, 615434
ARL3	100	98,4	100	100	Joubert syndrome 35, 618161 Retinitis pigmentosa 83, 618173
ARL6	99,9	98,6	100	100	Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARR3	100	99,8	100	100	Myopia 26, X-linked, female-limited, 301010
ARSG	100	99,5	100	100	Usher syndrome, type IV, 618144
ASB10	99,4	95,7	100	100	Glaucoma 1, open angle, F, 603383
ASPH	99,9	98,8	100	100	Traboulsi syndrome, 601552
ASRGL1	100	100	100	100	No OMIM disease ID
ATF6	100	99,9	100	100	Achromatopsia 7, 616517
ATOH7	96	91,2	99,1	94,4	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
B3GLCT	99,6	96,3	99,9	99,2	Peters-plus syndrome, 261540
BBIP1	98,6	92,4	100	100	?Bardet-Biedl syndrome 18, 615995
BBS1	100	100	100	100	Bardet-Biedl syndrome 1, 209900
BBS10	100	99,8	100	100	Bardet-Biedl syndrome 10, 615987
BBS12	100	100	100	100	Bardet-Biedl syndrome 12, 615989
BBS2	100	99,5	100	100	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	99,9	99,3	100	100	Bardet-Biedl syndrome 4, 615982
BBS5	99	93,9	100	100	Bardet-Biedl syndrome 5, 615983

BBS7	98,7	95,5	100	100	Bardet-Biedl syndrome 7, 615984
BBS9	92,3	90,4	95,8	95,8	Bardet-Biedl syndrome 9, 615986
BCOR	99,6	97,4	100	99,9	Microphthalmia, syndromic 2, 300166
BEST1	99,4	96,4	99,9	99,4	Retinitis pigmentosa-50, 613194 Bestrophinopathy, autosomal recessive, 611809 Retinitis pigmentosa, concentric, 613194 Vitreoretinochoroidopathy, 193220 ?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 2, 193220 Macular dystrophy, vitelliform, 2, 153700
BFSP1	99	89,9	100	99,9	Cataract 33, multiple types, 611391
BFSP2	99,8	97,6	100	100	Cataract 12, multiple types, 611597
BLOC1S3	98,5	81,3	100	100	Hermansky-Pudlak syndrome 8, 614077
BLOC1S5	100	99,2	100	100	Hermansky-Pudlak syndrome 11, 619172
BLOC1S6	99,9	97,8	100	100	?Hermansky-pudlak syndrome 9, 614171
BMP4	100	100	100	100	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
C12orf65	99,8	98,5	100	100	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
C19orf12	100	99,8	100	100	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C1QTNF5	90,9	78,5	100	100	Retinal degeneration, late-onset, autosomal dominant, 605670
C8orf37	100	99,4	100	100	Retinitis pigmentosa 64, 614500 Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500
CABP4	100	99,9	100	100	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACNA1F	99,7	97,5	100	100	Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 Aland Island eye disease, 300600
CACNA2D4	98,9	97,7	100	100	Retinal cone dystrophy 4, 610478
CAPN5	100	100	100	100	Vitreoretinopathy, neovascular inflammatory, 193235
CC2D2A	98,5	96,5	97,1	97,1	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome 2, 619111
CCT2	100	100	100	100	No OMIM disease ID
CDH2	99,3	97,7	100	100	Arrhythmogenic right ventricular dysplasia, familial, 14, 618920 Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929

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
CDH3	100	99,5	100	100	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDHR1	99,2	98,1	100	100	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
CDK10	100	99,9	100	100	Al Kaissi syndrome, 617694
CEP120	100	99,5	100	100	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP164	99,9	98,3	100	100	Nephronophthisis 15, 614845
CEP250	100	99,2	100	100	Cone-rod dystrophy and hearing loss 2, 618358
CEP290	96,1	90	100	100	?Bardet-Biedl syndrome 14, 615991 Leber congenital amaurosis 10, 611755 Senior-Loken syndrome 6, 610189 Meckel syndrome 4, 611134 Joubert syndrome 5, 610188
CEP41	99,8	97,4	100	100	Joubert syndrome 15, 614464
CEP78	98,9	96,8	100	100	Cone-rod dystrophy and hearing loss, 617236
CEP83	99,8	97,4	100	100	Nephronophthisis 18, 615862
CERKL	99,5	96,9	100	100	Retinitis pigmentosa 26, 608380
CFAP410	100	99,3	100	100	Spondylometaphyseal dysplasia, axial, 602271 Retinal dystrophy with macular staphyloma, 617547
CFH	99,9	99	100	99,9	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CHD7	100	99,5	100	100	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CHM	98,5	94,5	98,8	97,4	Choroideremia, 303100
CHMP4B	100	99,3	100	100	Cataract 31, multiple types, 605387
CHRDL1	100	99,8	100	100	Megalocornea 1, X-linked, 309300
CHST6	100	100	100	100	Macular corneal dystrophy, 217800
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

CLCC1	99,8	98	100	100	Retinitis pigmentosa 32, 609913
CLDN19	98,5	93,1	100	100	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLN3	92,5	91,8	92,5	92,5	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	69,3	66,3	72,1	71,6	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	99,9	97,1	100	100	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 Ceroid lipofuscinosis, neuronal, 6, 601780
CLN8	83,5	83,5	100	100	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLRN1	100	99,8	100	100	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
CLUAP1	100	99,8	100	100	No OMIM disease ID
CNGA1	91,7	86,3	91	91	Retinitis pigmentosa 49, 613756
CNGA3	100	99,7	100	100	Achromatopsia 2, 216900
CNGB1	99,4	97,5	100	100	Retinitis pigmentosa 45, 613767
CNGB3	99,4	95,9	100	100	Achromatopsia 3, 262300
CNNM4	99,8	98,9	99,7	98,8	Jalili syndrome, 217080
COA8	81,9	80,7	93,5	93,4	Mitochondrial complex IV deficiency, nuclear type 17, 619061
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
COL18A1	98,1	95,6	100	100	Knobloch syndrome, type 1, 267750 Glaucoma, primary closed-angle, 618880
COL25A1	95,8	95,3	99,9	99,9	Fibrosis of extraocular muscles, congenital, 5, 616219
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
COL4A1	98,7	97,4	100	100	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 175780 {Hemorrhage, intracerebral, susceptibility to}, 614519 ?Retinal arteries, tortuosity of, 180000 Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564
COL8A2	99,9	97,9	100	100	Corneal dystrophy, posterior polymorphous 2, 609140 Corneal dystrophy, Fuchs endothelial, 1, 136800
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
CPAMD8	95,8	92,8	99,9	99,6	Anterior segment dysgenesis 8, 617319
CPLANE1	99,7	98,4	100	100	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CPSF1	98,2	96,5	100	100	Myopia 27, 618827
CRB1	100	99,9	100	100	Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, 600105 Leber congenital amaurosis 8, 613835
CRX	100	100	100	100	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
CRYAA	99,9	97,5	100	100	Cataract 9, multiple types, 604219
CRYAB	100	99,2	100	100	Myopathy, myofibrillar, 2, 608810 Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869
CRYBA1	100	99,4	100	100	Cataract 10, multiple types, 600881
CRYBA2	100	100	100	100	?Cataract 42, 115900
CRYBA4	100	100	100	100	Cataract 23, 610425
CRYBB1	100	100	100	100	Cataract 17, multiple types, 611544
CRYBB2	100	100	100	100	Cataract 3, multiple types, 601547
CRYBB3	100	100	100	100	Cataract 22, 609741
CRYGB	100	99,6	100	100	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	99,8	96,9	100	100	Cataract 2, multiple types, 604307

CRYGD	100	98,9	100	100	Cataract 4, multiple types, 115700
CRYGS	94,1	86,6	100	100	Cataract 20, multiple types, 116100
CSPP1	99,8	98,7	100	100	Joubert syndrome 21, 615636
CTDP1	88,4	84,3	100	99,4	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNA1	99,3	98,1	100	100	Macular dystrophy, patterned, 2, 608970
CTNNB1	100	99,9	100	100	Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 Medulloblastoma, somatic, 155255 Hepatocellular carcinoma, somatic, 114550 Pilomatricoma, somatic, 132600 Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 Exudative vitreoretinopathy 7, 617572
CTSD	98,4	95	100	100	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSH	100	100	100	100	No OMIM disease ID
CWC27	99,3	96,5	100	100	Retinitis pigmentosa with or without skeletal anomalies, 250410
CYP1B1	100	100	100	100	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Anterior segment dysgenesis 6, multiple subtypes, 617315
CYP4V2	99,9	98,4	100	100	Bietti crystalline corneoretinal dystrophy, 210370
DCN	95,7	95,6	95,7	95,7	Corneal dystrophy, congenital stromal, 610048
DDHD1	97,9	95,8	100	100	Spastic paraplegia 28, autosomal recessive, 609340
DHDDS	99	95	95,2	95,2	Retinitis pigmentosa 59, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 ?Congenital disorder of glycosylation, type 1bb, 613861
DHX38	100	99,3	100	100	Retinitis pigmentosa 84, 618220
DKC1	99,8	98,7	100	99,7	Dyskeratosis congenita, X-linked, 305000
DNAJC30	100	100	100	100	No OMIM disease ID
DNM1L	99,9	98,5	100	100	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DNMBP	100	99,7	100	100	Cataract 48, 618415
DRAM2	100	99,9	100	100	Cone-rod dystrophy 21, 616502
DTNBP1	99,8	98,7	100	100	Hermansky-Pudlak syndrome 7, 614076
EFEMP1	100	99,9	100	100	Doyne honeycomb degeneration of retina, 126600
ELOVL1	99,8	97,6	100	100	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
ELOVL4	100	99,5	100	100	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
EMC1	100	99,3	100	100	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875

EPG5	99,5	98,5	100	100	Vici syndrome, 242840
EPHA2	100	99,5	100	100	Cataract 6, multiple types, 116600
ERCC2	100	99,7	100	100	Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756 Xeroderma pigmentosum, group D, 278730
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 Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650
EYS	99,7	98,2	100	100	Retinitis pigmentosa 25, 602772
FA2H	92	83,1	100	100	Spastic paraplegia 35, autosomal recessive, 612319
FAM161A	100	99,7	100	100	Retinitis pigmentosa 28, 606068
FBN1	100	99,9	100	100	Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 MASS syndrome, 604308 Ectopia lentis, familial, 129600 Acromicric dysplasia, 102370 Weill-Marchesani syndrome 2, dominant, 608328 Geleophysic dysplasia 2, 614185 Stiff skin syndrome, 184900
FDXR	100	99,3	100	100	Auditory neuropathy and optic atrophy, 617717
FLVCR1	100	98,9	100	100	Ataxia, posterior column, with retinitis pigmentosa, 609033
FOXC1	98	89,6	99,9	98,5	Axenfeld-Rieger syndrome, type 3, 602482 Anterior segment dysgenesis 3, multiple subtypes, 601631
FOXE3	82,6	72	94,4	87,8	Cataract 34, multiple types, 612968 Anterior segment dysgenesis 2, multiple subtypes, 610256 {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349
FREM1	99,9	99,1	100	100	Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485 Bifid nose with or without anorectal and renal anomalies, 608980
FRMD7	99,9	99,1	100	99,6	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
FTL	98,5	89,4	100	100	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159 L-ferritin deficiency, dominant and recessive, 615604
FYCO1	100	99,9	100	100	Cataract 18, autosomal recessive, 610019

FZD4	100	100	100	100	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780
GALK1	100	99,1	100	100	Galactokinase deficiency with cataracts, 230200
GALM	100	99,9	100	100	Galactosemia IV, 618881
GALT	100	99,7	100	100	Galactosemia, 230400
GCNT2	99,5	99,5	100	100	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700 [Blood group, Ii], 110800
GDF3	100	100	100	100	Microphthalmia, isolated 7, 613704 Microphthalmia with coloboma 6, 613703 Klippel-Feil syndrome 3, autosomal dominant, 613702
GDF6	100	99,9	100	99,4	Leber congenital amaurosis 17, 615360 Klippel-Feil syndrome 1, autosomal dominant, 118100 Multiple synostoses syndrome 4, 617898 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094
GDPD1	100	98,7	100	100	No OMIM disease ID
GFER	99,6	93,9	100	100	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076
GJA1	100	100	100	100	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Atrioventricular septal defect 3, 600309 Oculodentodigital dysplasia, 164200 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100
GJA3	100	99,7	100	100	Cataract 14, multiple types, 601885
GJA8	100	100	100	100	Cataract 1, multiple types, 116200
GNAT1	100	100	100	100	Night blindness, congenital stationary, type 1G, 616389 Night blindness, congenital stationary, autosomal dominant 3, 610444
GNAT2	99,9	99	100	100	Achromatopsia 4, 613856
GNB3	100	100	100	100	{Hypertension, essential, susceptibility to}, 145500 Night blindness, congenital stationary, type 1H, 617024
GNPTG	99,1	94,3	100	99,9	Mucolipidosis III gamma, 252605
GPR143	85,8	76,4	99,8	97,9	Ocular albinism, type I, Nettleship-Falls type, 300500 Nystagmus 6, congenital, X-linked, 300814
GPR179	100	100	100	100	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565

GRHL2	100	100	100	100	Deafness, autosomal dominant 28, 608641 Corneal dystrophy, posterior polymorphous, 4, 618031 Ectodermal dysplasia/short stature syndrome, 616029
GRK1	100	100	100	100	Oguchi disease-2, 613411
GRM6	90,2	80,6	98,3	96,3	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GSN	95,8	93,5	99,9	99,3	Amyloidosis, Finnish type, 105120
GUCA1A	100	100	100	100	Cone-rod dystrophy 14, 602093 Cone dystrophy-3, 602093
GUCA1B	100	100	100	100	Retinitis pigmentosa 48, 613827
GUCY2D	99,6	96,2	100	100	Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000 Night blindness, congenital stationary, type 1I, 618555 ?Choroidal dystrophy, central areolar 1, 215500
HARS1	100	100	100	100	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HCCS	99,8	97,6	100	100	Linear skin defects with multiple congenital anomalies 1, 309801
HGSNAT	86,4	86,3	91,2	89,3	Retinitis pigmentosa 73, 616544 Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HK1	100	100	100	100	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Neurodevelopmental disorder with visual defects and brain anomalies, 618547 Retinitis pigmentosa 79, 617460
HMX1	62,4	42,9	99,7	96,1	Oculoauricular syndrome, 612109
HPS1	100	100	100	100	Hermansky-Pudlak syndrome 1, 203300
HPS3	99,7	97,5	100	100	Hermansky-Pudlak syndrome 3, 614072
HPS4	100	100	100	100	Hermansky-Pudlak syndrome 4, 614073
HPS5	100	99,7	100	100	Hermansky-Pudlak syndrome 5, 614074
HPS6	97,1	88,9	100	100	Hermansky-Pudlak syndrome 6, 614075
HRAS	100	100	100	100	Costello syndrome, 218040 Bladder cancer, somatic, 109800 Nevus sebaceous or woolly hair nevus, somatic, 162900 Congenital myopathy with excess of muscle spindles, 218040 Thyroid carcinoma, follicular, somatic, 188470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550
HSF4	99,6	97,2	100	100	Cataract 5, multiple types, 116800
HSPG2	99,2	97,7	100	99,9	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800

IDH3A	99,4	97,3	100	100	Retinitis pigmentosa 90, 619007
IDH3B	95,4	95,4	100	100	Retinitis pigmentosa 46, 612572
IFT140	99,8	98,8	100	100	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	99,9	99,1	100	100	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	100	100	100	100	?Bardet-Biedl syndrome 19, 615996
IFT43	100	100	100	100	?Cranioectodermal dysplasia 3, 614099 Short-rib thoracic dysplasia 18 with polydactyly, 617866 ?Retinitis pigmentosa 81, 617871
IFT52	100	99,9	100	100	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT74	98,4	93,9	100	100	?Bardet-Biedl syndrome 20, 617119
IFT81	93,5	90,1	95	94,9	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IMPDH1	87,9	80,2	100	100	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
IMPG1	99,7	98,5	100	100	Macular dystrophy, vitelliform, 4, 616151
IMPG2	99,8	98,4	100	100	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581
INPP5E	97,1	92,7	100	100	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INVS	100	100	100	100	Nephronophthisis 2, infantile, 602088
IQCB1	93,9	85	100	100	Senior-Loken syndrome 5, 609254
IRX1	87,4	81,3	97,7	94	No OMIM disease ID
ITPR1	100	99,9	100	100	Spinocerebellar ataxia 29, congenital nonprogressive, 117360 Spinocerebellar ataxia 15, 606658 Gillespie syndrome, 206700
JAG1	97,7	96,8	100	100	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
JAM3	100	99,9	100	100	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KCNJ13	100	100	100	100	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230
KCNV2	100	99,9	100	100	Retinal cone dystrophy 3B, 610356
KERA	100	100	100	100	Cornea plana 2, autosomal recessive, 217300
KIAA0586	97,3	93,1	95,8	95,8	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA1549	97,9	96,4	98,8	98	Retinitis pigmentosa 86, 618613

KIF11	97,6	94,8	100	100	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF21A	99,9	99,3	100	100	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700
KIF3B	100	99,7	100	100	Retinitis pigmentosa 89, 618955
KIF7	93,6	90,6	99,1	97,8	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
KIZ	100	99,2	100	100	Retinitis pigmentosa 69, 615780
KLHL7	99,9	99,8	100	100	Retinitis pigmentosa 42, 612943 PERCHING syndrome, 617055
KRT12	99,7	97,8	100	100	Meesmann corneal dystrophy 1, 122100
KRT3	100	100	100	100	Meesmann corneal dystrophy 2, 618767
LAMA1	100	99,7	100	100	Poretti-Boltshauser syndrome, 615960
LAMB2	100	99,7	100	100	Pierson syndrome, 609049 Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199
LAMP2	99,2	95,6	100	100	Danon disease, 300257
LCA5	99,9	99,2	100	100	Leber congenital amaurosis 5, 604537
LEMD2	98,7	92	100	100	Cataract 46, juvenile-onset, 212500
LIM2	100	100	100	100	Cataract 19, multiple types, 615277
LRAT	100	100	100	100	Retinal dystrophy, early-onset severe, 613341 Leber congenital amaurosis 14, 613341 Retinitis pigmentosa, juvenile, 613341
LRIT3	93,9	91,9	100	100	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRMDA	96,8	95,6	99,6	99,6	Albinism, oculocutaneous, type VII, 615179
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
LRPAP1	100	100	100	100	Myopia 23, autosomal recessive, 615431

LSS	100	99,9	100	100	Alopecia-mental retardation syndrome 4, 618840 Cataract 44, 616509 Hypotrichosis 14, 618275
LTBP2	99,9	99	100	100	Glaucoma 3, primary congenital, D, 613086 ?Weill-Marchesani syndrome 3, recessive, 614819 Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750
LYST	99,6	98,3	100	100	Chediak-Higashi syndrome, 214500
LZTFL1	99,9	99,2	100	100	Bardet-Biedl syndrome 17, 615994
MAB21L2	100	100	100	100	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MAF	83,5	78	88,6	82,2	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
MAK	98,7	96,8	100	100	Retinitis pigmentosa 62, 614181
MAPKAPK3	100	99,6	100	100	?Macular dystrophy, patterned, 3, 617111
MERTK	99,5	98,8	99,1	99,1	Retinitis pigmentosa 38, 613862
MFN2	100	99,9	100	100	Hereditary motor and sensory neuropathy VIA, 601152 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260
MFRP	100	100	100	100	Nanophthalmos 2, 609549 Microphthalmia, isolated 5, 611040
MFSD8	100	99,7	100	100	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MIP	100	98,9	100	100	Cataract 15, multiple types, 615274
MIR184	NC	NC	NC	NC	EDICT syndrome, 614303
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
MKKS	100	100	100	100	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	99,8	97,9	100	100	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MVK	90,9	90,5	90,5	90,5	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377

MYO7A	99,3	97,3	100	100	Deafness, autosomal recessive 2, 600060 Deafness, autosomal dominant 11, 601317 Usher syndrome, type 1B, 276900
MYOC	100	98,6	100	100	Glaucoma 1A, primary open angle, 137750
NAA10	99,7	98,5	99,9	99,9	Ogden syndrome, 300855 Microphthalmia, syndromic 1, 309800
NBAS	100	99,6	100	100	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NDP	100	99,7	100	100	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDUFS2	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 6, 618228
NEK2	99,7	95,5	96,1	96,1	?Retinitis pigmentosa 67, 615565
NEUROD1	100	99,1	100	100	Maturity-onset diabetes of the young 6, 606394 {Type 2 diabetes mellitus, susceptibility to}, 125853
NHS	95,4	93,9	100	99,8	Nance-Horan syndrome, 302350 Cataract 40, X-linked, 302200
NMNAT1	100	99,2	98,3	95,6	Leber congenital amaurosis 9, 608553
NPHP1	100	99	100	100	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NPHP3	99,7	98,4	100	100	Meckel syndrome 7, 267010 Renal-hepatic-pancreatic dysplasia 1, 208540 Nephronophthisis 3, 604387
NPHP4	100	99,8	100	100	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NR2E3	100	99,6	100	100	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131
NR2F1	100	100	99,1	95,1	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NRL	99,5	94,8	100	100	Retinitis pigmentosa 27, 613750 Retinal degeneration, autosomal recessive, clumped pigment type, 0
NYX	96,3	94,1	99,7	98,8	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	85,2	76,3	100	100	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCA2	99,9	98,7	100	100	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 Albinism, oculocutaneous, type II, 203200 Albinism, brown oculocutaneous, 203200
OCRL	99,9	98,6	100	99,9	Lowe syndrome, 309000 Dent disease 2, 300555

OFD1	88	73,7	100	99,9	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
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
OPA3	100	99	100	100	Optic atrophy 3 with cataract, 165300 3-methylglutaconic aciduria, type III, 258501
OPN1LW	67,2	60,6	98,3	98,1	Blue cone monochromacy, 303700 Colorblindness, protan, 303900
OPN1MW	66,3	58,5	98,9	97,5	Colorblindness, deutan, 303800 Blue cone monochromacy, 303700
OPTN	100	99,9	100	100	{Glaucoma, normal tension, susceptibility to}, 606657 Glaucoma 1, open angle, E, 137760 Amyotrophic lateral sclerosis 12 with or without frontotemporal dementia, 613435
OTX2	100	99,7	100	100	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
OVOL2	95,7	89,5	100	100	Corneal dystrophy, posterior polymorphous, 1, 122000
P3H2	99,8	98	100	100	Myopia, high, with cataract and vitreoretinal degeneration, 614292
P4HA2	100	99,2	100	100	Myopia 25, autosomal dominant, 617238
PANK2	100	99,3	100	100	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAX2	100	99,9	100	100	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
PAX6	100	100	100	100	Optic nerve hypoplasia, 165550 ?Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Aniridia, 106210 Keratitis, 148190 Anterior segment dysgenesis 5, multiple subtypes, 604229 Cataract with late-onset corneal dystrophy, 106210 ?Coloboma of optic nerve, 120430 ?Morning glory disc anomaly, 120430
PCARE	99,6	98,5	100	100	Retinitis pigmentosa 54, 613428

PCDH15	97,8	96,7	100	100	Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083 Deafness, autosomal recessive 23, 609533
PCYT1A	98,9	95,5	100	100	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE6A	100	99,6	100	100	Retinitis pigmentosa 43, 613810
PDE6B	100	99,9	100	100	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	99,9	97,8	100	100	Cone dystrophy 4, 613093
PDE6D	100	100	100	100	Joubert syndrome 22, 615665
PDE6G	100	100	100	100	Retinitis pigmentosa 57, 613582
PDE6H	100	97,9	100	100	Retinal cone dystrophy 3, 610024 Achromatopsia 6, 610024
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
PEX2	100	100	100	100	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
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
PEX7	87,8	80,7	91,3	91,3	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PGK1	92,8	79,3	100	100	Phosphoglycerate kinase 1 deficiency, 300653
PHOX2A	91,6	72,7	100	99,8	Fibrosis of extraocular muscles, congenital, 2, 602078
PHYH	100	99,6	100	100	Refsum disease, 266500
PIKFYVE	99,9	99,4	100	100	Corneal fleck dystrophy, 121850
PITX2	99,9	97,7	100	100	Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550 Anterior segment dysgenesis 4, 137600
PITX3	100	98,4	100	100	Anterior segment dysgenesis 1, multiple subtypes, 107250 Cataract 11, syndromic, autosomal recessive, 610623 Cataract 11, multiple types, 610623

PLA2G5	100	100	100	100	[Fleck retina, familial benign], 228980
PLK4	99,9	98,2	100	100	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PNPLA6	100	99,7	100	100	Spastic paraplegia 39, autosomal recessive, 612020 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800
POC1B	99,8	98,8	100	100	Cone-rod dystrophy 20, 615973
POC5	99,7	97,6	100	100	No OMIM disease ID
POMGNT1	100	99,9	100	100	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280
PPT1	90,3	90,3	82,5	82,5	Ceroid lipofuscinosis, neuronal, 1, 256730
PRCD	100	100	100	100	Retinitis pigmentosa 36, 610599
PRDM13	99,2	94,1	100	100	No OMIM disease ID
PRDM5	99,9	99,2	100	100	Brittle cornea syndrome 2, 614170
PRIMPOL	97,5	94,6	100	100	Myopia 22, autosomal dominant, 615420
PROM1	97,2	96,1	100	100	Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786 Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051
PRPF3	98,8	95,3	100	100	Retinitis pigmentosa 18, 601414
PRPF31	100	98,7	100	100	Retinitis pigmentosa 11, 600138
PRPF4	100	99,8	100	100	Retinitis pigmentosa 70, 615922
PRPF6	100	99,8	100	100	Retinitis pigmentosa 60, 613983
PRPF8	100	99,3	100	100	Retinitis pigmentosa 13, 600059
PRPH2	100	100	100	100	Macular dystrophy, patterned, 1, 169150 Retinitis punctata albescens, 136880 Choroidal dystrophy, central areolar 2, 613105 Retinitis pigmentosa 7 and digenic form, 608133 Leber congenital amaurosis 18, 608133 Macular dystrophy, vitelliform, 3, 608161
PRSS56	99,9	96,5	100	100	Microphthalmia, isolated 6, 613517
PXDN	100	99,6	100	100	Anterior segment dysgenesis 7, with sclerocornea, 269400
RAB28	99,7	96	100	100	Cone-rod dystrophy 18, 615374
RAB3GAP2	99,5	97	100	100	Warburg micro syndrome 2, 614225 Martsolf syndrome, 212720

RARB	100	100	100	100	Microphthalmia, syndromic 12, 615524
RAX	96	87	100	98,4	Microphthalmia, isolated 3, 611038
RAX2	100	92,3	100	100	?Macular degeneration, age-related, 6, 613757 Cone-rod dystrophy 11, 610381
RBP3	100	100	100	100	?Retinitis pigmentosa 66, 615233
RBP4	99,9	97,7	100	100	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RCBTB1	99,9	99,5	100	100	Retinal dystrophy with or without extraocular anomalies, 617175
RD3	100	100	100	100	Leber congenital amaurosis 12, 610612
RDH11	100	99	100	100	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	100	98,6	100	100	Leber congenital amaurosis 13, 612712
RDH5	100	99,9	100	100	Fundus albipunctatus, 136880
REEP6	100	100	91,5	87,4	Retinitis pigmentosa 77, 617304
RGS9	98,5	97,1	100	100	Bradyopsia, 608415
RGS9BP	99,6	95	100	100	Bradyopsia, 608415
RHO	100	100	100	100	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis punctata albescens, 136880 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731
RIMS1	99,8	97,7	100	100	Cone-rod dystrophy 7, 603649
RIMS2	96,7	95,3	97,8	97,7	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970
RLBP1	100	99,9	100	100	Retinitis punctata albescens, 136880 Bothnia retinal dystrophy, 607475 Newfoundland rod-cone dystrophy, 607476 Fundus albipunctatus, 136880
ROM1	100	99,9	100	100	Retinitis pigmentosa 7, digenic form, 608133
RP1	91,5	90,6	100	100	Retinitis pigmentosa 1, 180100
RP1L1	100	100	100	100	Retinitis pigmentosa 88, 618826 Occult macular dystrophy, 613587
RP2	100	99,8	100	100	Retinitis pigmentosa 2, 312600
RP9	80,8	75,9	100	99,3	?Retinitis pigmentosa 9, 180104
RPE65	99,8	97,8	100	100	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 87 with choroidal involvement, 618697 Retinitis pigmentosa 20, 613794
RPGR	76,5	72	100	99,9	Cone-rod dystrophy, X-linked, 1, 304020 Retinitis pigmentosa 3, 300029 Macular degeneration, X-linked atrophic, 300834 Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455

RPGRIP1	100	99,9	100	100	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
RPGRIP1L	96,7	95,7	100	99,5	?COACH syndrome 3, 619113 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
RS1	99,8	93,9	100	100	Retinoschisis, 312700
RTN4IP1	99,9	98,7	100	100	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
SAG	100	100	100	100	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SAMD11	90,2	81,6	100	100	No OMIM disease ID
SC5D	100	99,5	100	100	Lathosterolosis, 607330
SCAPER	99,7	98,2	100	100	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCO2	100	100	100	100	Myopia 6, 608908 Mitochondrial complex IV deficiency, nuclear type 2, 604377
SDCCAG8	100	99,9	100	100	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SEMA4A	100	99,8	100	100	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SGSH	94,4	94,1	100	100	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SHH	100	99,5	100	100	Schizencephaly, 269160 Microphthalmia with coloboma 5, 611638 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SIPA1L3	99,9	98,9	100	100	?Cataract 45, 616851
SIX6	100	100	100	100	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SLC16A12	100	99,9	100	100	Cataract 47, juvenile, with microcornea, 612018
SLC24A1	100	99,9	100	100	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A5	99,9	99,1	100	100	[Skin/hair/eye pigmentation 4, fair/dark skin], 113750 Albinism, oculocutaneous, type VI, 113750
SLC25A46	99,7	97,3	100	100	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC33A1	99,9	98,9	100	100	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC38A8	99,9	97,3	100	100	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A5	99,9	99	100	100	Myopia 24, autosomal dominant, 615946
SLC45A2	100	99,9	100	100	[Skin/hair/eye pigmentation 5, dark/fair skin], 227240 [Skin/hair/eye pigmentation 5, black/nonblack hair], 227240

					Albinism, oculocutaneous, type IV, 606574 [Skin/hair/eye pigmentation 5, dark/light eyes], 227240
SLC4A11	100	99,9	100	100	Corneal endothelial dystrophy, autosomal recessive, 217700 Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy and perceptive deafness, 217400
SLC52A2	100	100	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC7A14	100	100	100	100	Retinitis pigmentosa 68, 615725
SMOC1	99,9	98,4	100	100	Microphthalmia with limb anomalies, 206920
SNRNP200	99,9	99,1	100	100	Retinitis pigmentosa 33, 610359
SOX2	100	100	100	100	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX5	99,9	98,9	100	100	Lamb-Shaffer syndrome, 616803
SPATA7	99,8	98,2	100	100	Retinitis pigmentosa, juvenile, autosomal recessive, 604232 Leber congenital amaurosis 3, 604232
SPP2	100	99,9	100	100	No OMIM disease ID
SSBP1	99,8	97,6	100	100	Optic atrophy 13 with retinal and foveal abnormalities, 165510
STRA6	100	99,8	100	100	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
TACSTD2	99	96,4	100	100	Corneal dystrophy, gelatinous drop-like, 204870
TCF4	100	99,8	100	100	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TCTN1	96,7	93	94,7	94,7	Joubert syndrome 13, 614173
TCTN3	100	100	100	100	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TDRD7	99,9	99,1	100	100	Cataract 36, 613887
TEAD1	100	99,9	100	100	Sveinsson chorioretinal atrophy, 108985
TEK	100	100	100	100	Glaucoma 3, primary congenital, E, 617272 Venous malformations, multiple cutaneous and mucosal, 600195
TENM3	100	99,7	100	100	Microphthalmia, syndromic 15, 615145 ?Microphthalmia, isolated, with coloboma 9, 615145
TGFBI	99,5	94,6	100	100	Corneal dystrophy, lattice type IIIA, 608471 Corneal dystrophy, Groenouw type I, 121900 Corneal dystrophy, lattice type I, 122200 Corneal dystrophy, Reis-Bucklers type, 608470 Corneal dystrophy, Thiel-Behnke type, 602082 Corneal dystrophy, epithelial basement membrane, 121820 Corneal dystrophy, Avellino type, 607541

TIMM8A	98,1	90,6	100	100	Mohr-Tranebjaerg syndrome, 304700
TIMP3	100	100	100	100	Sorsby fundus dystrophy, 136900
TMCO3	100	99,4	100	100	No OMIM disease ID
TMEM126A	96,3	84,4	100	100	Optic atrophy 7, 612989
TMEM138	100	99,1	100	100	Joubert syndrome 16, 614465
TMEM216	99,9	98,1	100	100	Meckel syndrome 2, 603194 Joubert syndrome 2, 608091
TMEM231	100	99,6	100	100	Meckel syndrome 11, 615397 Joubert syndrome 20, 614970
TMEM237	100	99,9	100	100	Joubert syndrome 14, 614424
TMEM67	99,5	95	100	99,9	Meckel syndrome 3, 607361 COACH syndrome 1, 216360 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991 Joubert syndrome 6, 610688
TMEM98	99,3	97,8	100	100	Nanophthalmos 4, 615972
TOGARAM1	99,6	98,1	100	100	Joubert syndrome 37, 619185
TOPORS	100	100	100	100	Retinitis pigmentosa 31, 609923
TPP1	100	100	100	100	Spinocerebellar ataxia, autosomal recessive 7, 609270 Ceroid lipofuscinosis, neuronal, 2, 204500
TRAF3IP1	99,6	97,6	100	100	Senior-Loken syndrome 9, 616629
TREX1	100	100	100	100	{Systemic lupus erythematosus, susceptibility to}, 152700 Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRIM32	100	100	100	100	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRNT1	99,5	96,5	100	100	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TRPM1	100	99,8	100	100	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TSPAN12	100	99,8	100	100	Exudative vitreoretinopathy 5, 613310
TTC8	99,6	98,1	100	100	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TTLL5	100	99,7	100	100	Cone-rod dystrophy 19, 615860
TUB	99,4	97,1	100	100	?Retinal dystrophy and obesity, 616188
TUBA3D	100	99,2	100	100	Keratoconus 9, 617928

TUBB3	98,3	96,9	100	100	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
TUBB4B	99,9	96,9	100	100	Leber congenital amaurosis with early-onset deafness, 617879
TUBGCP4	99,2	96,4	100	100	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	100	99,3	100	100	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TULP1	100	99,5	100	100	Retinitis pigmentosa 14, 600132 Leber congenital amaurosis 15, 613843
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
TYRP1	100	99,8	100	100	[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271 Albinism, oculocutaneous, type III, 203290
UBIAD1	99,5	96	100	100	Corneal dystrophy, Schnyder type, 121800
UNC45B	99,3	98	100	100	?Cataract 43, 616279 Myofibrillar myopathy 11, 619178
USH1C	100	99,8	100	100	Usher syndrome, type 1C, 276904 Deafness, autosomal recessive 18A, 602092
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
USP45	99,6	98,1	100	100	?Leber congenital amaurosis 19, 618513
VAX1	97,5	91,5	95,7	91,7	?Microphthalmia, syndromic 11, 614402
VCAN	100	100	100	100	Wagner syndrome 1, 143200
VIM	99,3	97	100	100	Cataract 30, pulverulent, 116300
VPS13B	99,5	98,2	99,5	99,4	Cohen syndrome, 216550
VSX1	84,7	80,5	100	100	Keratoconus 1, 148300 ?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195
VSX2	100	99,3	100	100	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
WDPCP	98,2	94,4	98,1	98,1	?Bardet-Biedl syndrome 15, 615992 Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	100	99,4	100	100	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376

WDR34	100	99,6	100	100	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR36	99,7	97,6	100	100	Glaucoma 1, open angle, G, 609887
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
WRN	99,9	98,8	100	100	Werner syndrome, 277700
YAP1	96,4	89,4	100	100	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
YME1L1	99	95,2	100	100	?Optic atrophy 11, 617302
ZEB1	100	99,4	100	100	Corneal dystrophy, posterior polymorphous, 3, 609141 Corneal dystrophy, Fuchs endothelial, 6, 613270
ZNF408	100	100	100	100	?Exudative vitreoretinopathy 6, 616468 Retinitis pigmentosa 72, 616469
ZNF423	100	100	100	100	Nephronophthisis 14, 614844 Joubert syndrome 19, 614844
ZNF469	100	100	100	100	Brittle cornea syndrome 1, 229200
ZNF513	100	100	100	100	?Retinitis pigmentosa 58, 613617
ZNF644	100	100	100	100	Myopia 21, autosomal dominant, 614167

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-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 : March 23rd , 2021.
This list is accurate for panel version DG 3.1.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors