

VISION DISORDERS GENE PANEL DG 2.18 (446 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
<i>ABCA4</i>	99,90%	99,30%	100%	100%	Retinal dystrophy, early-onset severe, 248200 Stargardt disease 1, 248200 Fundus flavimaculatus, 248200 Cone-rod dystrophy 3, 604116 Retinitis pigmentosa 19, 601718
<i>ABCC6</i>	93,60%	92,40%	100%	100%	Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850 Arterial calcification, generalized, of infancy, 2, 614473
<i>ABHD12</i>	98,70%	92,30%	100%	99,30%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
<i>ACBD5</i>	100%	99,20%	100%	100%	Retinal dystrophy with leukodystrophy, 618863
<i>ACO2</i>	96,30%	90,30%	100%	100%	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
<i>ADAM9</i>	99,80%	99,10%	100%	100%	Cone-rod dystrophy 9, 612775
<i>ADAMTS18</i>	100%	99,70%	100%	100%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
<i>ADAMTSL4</i>	100%	99,20%	100%	100%	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
<i>ADGRV1</i>	99,60%	98,60%	100%	100%	Usher syndrome, type 2C, 605472 ?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
<i>ADIPOR1</i>	99,90%	97,80%	100%	100%	No OMIM disease ID
<i>AGBL1</i>	98,50%	98,50%	100%	100%	Corneal dystrophy, Fuchs endothelial, 8, 615523
<i>AGBL5</i>	99,90%	99,30%	100%	100%	Retinitis pigmentosa 75, 617023
<i>AGK</i>	99,90%	97,60%	100%	100%	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691
<i>AHI1</i>	99,70%	97,90%	100%	100%	Joubert syndrome 3, 608629
<i>AHR</i>	100%	99,60%	100%	100%	?Retinitis pigmentosa 85, 618345
<i>AIPL1</i>	100%	99,80%	100%	100%	Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393 Cone-rod dystrophy, 604393
<i>ALDH1A3</i>	97,20%	94,50%	100%	99,90%	Microphthalmia, isolated 8, 615113

<i>ALMS1</i>	99,80%	99,50%	100%	100%	Alstrom syndrome, 203800
<i>AP3B1</i>	99,20%	95,80%	100%	100%	Hermansky-Pudlak syndrome 2, 608233
<i>AP3D1</i>	99,80%	98,60%	100%	100%	?Hermansky-Pudlak syndrome 10, 617050
<i>APOPT1</i>	81,90%	80,70%	93,50%	93,40%	Mitochondrial complex IV deficiency, 220110
<i>ARHGEF18</i>	95,40%	92,30%	100%	100%	Retinitis pigmentosa 78, 617433
<i>ARL13B</i>	100%	99,20%	100%	100%	Joubert syndrome 8, 612291
<i>ARL2BP</i>	95,90%	88,30%	100%	100%	Retinitis pigmentosa with or without situs inversus, 615434
<i>ARL3</i>	100%	98,40%	100%	100%	Joubert syndrome 35, 618161 Retinitis pigmentosa 83, 618173
<i>ARL6</i>	99,90%	98,60%	100%	100%	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151
<i>ARR3</i>	100%	99,80%	100%	100%	Myopia 26, X-linked, female-limited, 301010
<i>ARSG</i>	100%	99,50%	100%	100%	Usher syndrome, type IV, 618144
<i>ASB10</i>	99,40%	95,70%	100%	100%	Glaucoma 1, open angle, F, 603383
<i>ASPH</i>	99,90%	98,80%	100%	100%	Traboulsi syndrome, 601552
<i>ASRGL1</i>	100%	100%	100%	100%	No OMIM disease ID
<i>ATF6</i>	100%	99,90%	100%	100%	Achromatopsia 7, 616517
<i>ATOH7</i>	96,00%	91,20%	99,10%	94,40%	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
<i>B3GLCT</i>	99,60%	96,30%	99,90%	99,20%	Peters-plus syndrome, 261540
<i>BBIP1</i>	98,60%	92,40%	100%	100%	?Bardet-Biedl syndrome 18, 615995
<i>BBS1</i>	100%	100%	100%	100%	Bardet-Biedl syndrome 1, 209900
<i>BBS10</i>	100%	99,80%	100%	100%	Bardet-Biedl syndrome 10, 615987
<i>BBS12</i>	100%	100%	100%	100%	Bardet-Biedl syndrome 12, 615989
<i>BBS2</i>	100%	99,50%	100%	100%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
<i>BBS4</i>	99,90%	99,30%	100%	100%	Bardet-Biedl syndrome 4, 615982
<i>BBS5</i>	99,00%	93,90%	100%	100%	Bardet-Biedl syndrome 5, 615983
<i>BBS7</i>	98,70%	95,50%	100%	100%	Bardet-Biedl syndrome 7, 615984
<i>BBS9</i>	99,70%	97,60%	100%	100%	Bardet-Biedl syndrome 9, 615986
<i>BCOR</i>	99,60%	97,40%	100%	99,90%	Microphthalmia, syndromic 2, 300166
<i>BEST1</i>	99,40%	96,40%	99,90%	99,40%	Retinitis pigmentosa-50, 613194 Bestrophinopathy, autosomal recessive, 611809 Retinitis pigmentosa, concentric, 613194 Vitreoretinopathopathy, 193220 Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 Macular dystrophy, vitelliform, 2, 153700
<i>BFSP1</i>	99,00%	89,90%	100%	99,90%	Cataract 33, multiple types, 611391

<i>BFSP2</i>	99,80%	97,60%	100%	100%	Cataract 12, multiple types, 611597
<i>BLOC1S3</i>	98,50%	81,30%	100%	100%	Hermansky-Pudlak syndrome 8, 614077
<i>BLOC1S6</i>	99,90%	97,10%	94,90%	94,90%	?Hermansky-pudlak syndrome 9, 614171
<i>BMP4</i>	100%	100%	100%	100%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
<i>C12orf65</i>	99,80%	98,50%	100%	100%	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
<i>C19orf12</i>	100%	99,80%	100%	100%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
<i>C1QTNF5</i>	90,90%	78,50%	100%	100%	Retinal degeneration, late-onset, autosomal dominant, 605670
<i>C21orf2</i>	100%	99,30%	100%	100%	Spondylometaphyseal dysplasia, axial, 602271 Retinal dystrophy with macular staphyloma, 617547
<i>C2orf71</i>	99,60%	98,50%	100%	100%	Retinitis pigmentosa 54, 613428
<i>C5orf42</i>	99,70%	98,40%	100%	100%	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
<i>C8orf37</i>	100%	99,40%	100%	100%	Retinitis pigmentosa 64, 614500 Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500
<i>CA4</i>	100%	100%	100%	100%	Retinitis pigmentosa 17, 600852
<i>CABP4</i>	100%	99,90%	100%	100%	Cone-rod synaptic disorder, congenital nonprogressive, 610427
<i>CACNA1F</i>	99,70%	97,50%	100%	100%	Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 Aland Island eye disease, 300600
<i>CACNA2D4</i>	98,90%	97,70%	100%	100%	Retinal cone dystrophy 4, 610478
<i>CAPN5</i>	100%	100%	100%	100%	Vitreoretinopathy, neovascular inflammatory, 193235
<i>CC2D2A</i>	99,70%	97,70%	98,20%	98,20%	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome, 216360
<i>CCT2</i>	100%	100%	100%	100%	No OMIM disease ID
<i>CDH23</i>	100%	100%	100%	100%	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1D, 601067
<i>CDH3</i>	100%	99,50%	100%	100%	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
<i>CDHR1</i>	99,20%	98,10%	100%	100%	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
<i>CDK10</i>	100%	99,90%	100%	100%	Al Kaissi syndrome, 617694
<i>CEP164</i>	99,90%	98,30%	100%	100%	Nephronophthisis 15, 614845

<i>CEP250</i>	100%	99,20%	100%	100%	Cone-rod dystrophy and hearing loss 2, 618358
<i>CEP290</i>	96,10%	90,00%	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
<i>CEP41</i>	99,80%	97,40%	100%	100%	Joubert syndrome 15, 614464
<i>CEP78</i>	99,70%	97,60%	100%	100%	Cone-rod dystrophy and hearing loss, 617236
<i>CEP83</i>	99,80%	97,40%	100%	100%	Nephronophthisis 18, 615862
<i>CERKL</i>	99,50%	96,90%	100%	100%	Retinitis pigmentosa 26, 608380
<i>CFH</i>	99,90%	99,00%	100%	99,90%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
<i>CHD7</i>	100%	99,50%	100%	100%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
<i>CHM</i>	98,50%	94,50%	98,80%	97,40%	Choroideremia, 303100
<i>CHMP4B</i>	100%	99,30%	100%	100%	Cataract 31, multiple types, 605387
<i>CHRD1</i>	100%	99,80%	100%	100%	Megalocornea 1, X-linked, 309300
<i>CHST6</i>	100%	100%	100%	100%	Macular corneal dystrophy, 217800
<i>CIB2</i>	99,70%	97,00%	100%	99,90%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
<i>CISD2</i>	83,40%	83,40%	100%	100%	Wolfram syndrome 2, 604928
<i>CLCC1</i>	99,80%	98,00%	100%	100%	No OMIM disease ID
<i>CLN3</i>	92,50%	91,80%	92,50%	92,50%	Ceroid lipofuscinosis, neuronal, 3, 204200
<i>CLN5</i>	99,40%	95,50%	100%	100%	Ceroid lipofuscinosis, neuronal, 5, 256731
<i>CLN6</i>	99,90%	97,10%	100%	100%	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 Ceroid lipofuscinosis, neuronal, 6, 601780
<i>CLN8</i>	83,50%	83,50%	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
<i>CLRN1</i>	100%	99,80%	100%	100%	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
<i>CLUAP1</i>	100%	99,80%	100%	100%	No OMIM disease ID
<i>CNGA1</i>	91,70%	86,30%	91,00%	91,00%	Retinitis pigmentosa 49, 613756
<i>CNGA3</i>	100%	99,70%	100%	100%	Achromatopsia 2, 216900
<i>CNGB1</i>	99,40%	97,50%	100%	100%	Retinitis pigmentosa 45, 613767
<i>CNGB3</i>	99,40%	95,90%	100%	100%	Macular degeneration, juvenile, 248200 Achromatopsia 3, 262300
<i>CNNM4</i>	99,80%	98,90%	99,70%	98,80%	Jalili syndrome, 217080

<i>COL11A1</i>	99,20%	95,70%	100%	100%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 ?Deafness, autosomal dominant 37, 618533 Fibrochondrogenesis 1, 228520
<i>COL18A1</i>	98,10%	95,60%	100%	100%	Knobloch syndrome, type 1, 267750
<i>COL25A1</i>	99,50%	99,10%	99,90%	99,90%	Fibrosis of extraocular muscles, congenital, 5, 616219
<i>COL2A1</i>	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 syndrome, type I, nonsyndromic ocular, 609508 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Vitreoretinopathy with phalangeal epiphyseal dysplasia, 0
<i>COL4A1</i>	98,70%	97,40%	100%	100%	?Retinal arteries, tortuosity of, 180000 Brain small vessel disease with or without ocular anomalies, 175780 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564
<i>COL8A2</i>	99,90%	97,90%	100%	100%	Corneal dystrophy, posterior polymorphous 2, 609140 Corneal dystrophy, Fuchs endothelial, 1, 136800
<i>COL9A1</i>	100%	99,20%	100%	100%	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
<i>COL9A2</i>	99,90%	99,00%	100%	100%	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
<i>COL9A3</i>	98,70%	95,50%	99,70%	98,60%	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969
<i>CPAMD8</i>	95,80%	92,80%	99,90%	99,60%	Anterior segment dysgenesis 8, 617319
<i>CPSF1</i>	98,20%	96,50%	100%	100%	Myopia 27, 618827
<i>CRB1</i>	100%	99,90%	100%	100%	Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, 600105 Leber congenital amaurosis 8, 613835

<i>CRX</i>	100%	100%	100%	100%	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
<i>CRYAA</i>	99,90%	97,50%	100%	100%	Cataract 9, multiple types, 604219
<i>CRYAB</i>	100%	99,20%	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
<i>CRYBA1</i>	100%	99,40%	100%	100%	Cataract 10, multiple types, 600881
<i>CRYBA2</i>	100%	100%	100%	100%	?Cataract 42, 115900
<i>CRYBA4</i>	100%	100%	100%	100%	Cataract 23, 610425
<i>CRYBB1</i>	100%	100%	100%	100%	Cataract 17, multiple types, 611544
<i>CRYBB2</i>	100%	100%	100%	100%	Cataract 3, multiple types, 601547
<i>CRYBB3</i>	100%	100%	100%	100%	Cataract 22, 609741
<i>CRYGB</i>	100%	99,60%	100%	100%	Cataract 39, multiple types, autosomal dominant, 615188
<i>CRYGC</i>	99,80%	96,90%	100%	100%	Cataract 2, multiple types, 604307
<i>CRYGD</i>	100%	98,90%	100%	100%	Cataract 4, multiple types, 115700
<i>CRYGS</i>	94,10%	86,60%	100%	100%	Cataract 20, multiple types, 116100
<i>CSPP1</i>	99,80%	98,70%	100%	100%	Joubert syndrome 21, 615636
<i>CTDP1</i>	88,40%	84,30%	100%	99,40%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
<i>CTNNA1</i>	99,30%	98,10%	100%	100%	Macular dystrophy, patterned, 2, 608970
<i>CTNNB1</i>	100%	100%	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
<i>CTSD</i>	98,40%	95,00%	100%	100%	Ceroid lipofuscinosis, neuronal, 10, 610127
<i>CWC27</i>	99,30%	96,50%	100%	100%	Retinitis pigmentosa with or without skeletal anomalies, 250410
<i>CYP1B1</i>	100%	100%	100%	100%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Anterior segment dysgenesis 6, multiple subtypes, 617315
<i>CYP4V2</i>	99,90%	98,40%	100%	100%	Bietti crystalline corneoretinal dystrophy, 210370
<i>DCN</i>	95,70%	95,60%	95,70%	95,70%	Corneal dystrophy, congenital stromal, 610048
<i>DDHD1</i>	97,90%	95,80%	100%	100%	Spastic paraplegia 28, autosomal recessive, 609340
<i>DHDDS</i>	99,00%	95,00%	95,20%	95,20%	Retinitis pigmentosa 59, 613861 ?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836
<i>DHX38</i>	100%	99,30%	100%	100%	Retinitis pigmentosa 84, 618220

<i>DKC1</i>	99,80%	98,70%	100%	99,70%	Dyskeratosis congenita, X-linked, 305000
<i>DNM1L</i>	99,90%	98,50%	100%	100%	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
<i>DNMBP</i>	100%	99,70%	100%	100%	Cataract 48, 618415
<i>DRAM2</i>	100%	99,90%	100%	100%	Cone-rod dystrophy 21, 616502
<i>DTNBP1</i>	99,80%	98,70%	100%	100%	Hermansky-Pudlak syndrome 7, 614076
<i>EFEMP1</i>	100%	99,90%	100%	100%	Doyme honeycomb degeneration of retina, 126600
<i>ELOVL1</i>	99,80%	97,60%	100%	100%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
<i>ELOVL4</i>	100%	99,50%	100%	100%	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
<i>EMC1</i>	100%	99,30%	100%	100%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
<i>EPG5</i>	99,50%	98,50%	100%	100%	Vici syndrome, 242840
<i>EPHA2</i>	100%	99,50%	100%	100%	Cataract 6, multiple types, 116600
<i>EXOSC2</i>	100%	100%	100%	100%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
<i>EYA1</i>	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
<i>EYS</i>	99,70%	98,20%	100%	100%	Retinitis pigmentosa 25, 602772
<i>FA2H</i>	92,00%	83,10%	100%	100%	Spastic paraplegia 35, autosomal recessive, 612319
<i>FAM161A</i>	100%	99,70%	100%	100%	Retinitis pigmentosa 28, 606068
<i>FLVCR1</i>	100%	98,90%	100%	100%	Ataxia, posterior column, with retinitis pigmentosa, 609033
<i>FOXC1</i>	98,00%	89,60%	99,90%	98,50%	Axenfeld-Rieger syndrome, type 3, 602482 Anterior segment dysgenesis 3, multiple subtypes, 601631
<i>FOXE3</i>	82,60%	72,00%	94,40%	87,80%	Cataract 34, multiple types, 612968 Anterior segment dysgenesis 2, multiple subtypes, 610256
<i>FREM1</i>	99,90%	99,10%	100%	100%	Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485 Bifid nose with or without anorectal and renal anomalies, 608980
<i>FRMD7</i>	99,90%	99,10%	100%	99,60%	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
<i>FTL</i>	98,50%	89,40%	100%	100%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159 L-ferritin deficiency, dominant and recessive, 615604
<i>FYCO1</i>	100%	99,90%	100%	100%	Cataract 18, autosomal recessive, 610019
<i>FZD4</i>	100%	100%	100%	100%	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780

<i>GALK1</i>	100%	99,10%	100%	100%	Galactokinase deficiency with cataracts, 230200
<i>GALM</i>	100%	99,90%	100%	100%	No OMIM disease ID
<i>GALT</i>	100%	99,70%	100%	100%	Galactosemia, 230400
<i>GCNT2</i>	99,50%	99,50%	100%	100%	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700
<i>GDF3</i>	100%	100%	100%	100%	Microphthalmia, isolated 7, 613704 Microphthalmia with coloboma 6, 613703 Klippel-Feil syndrome 3, autosomal dominant, 613702
<i>GDF6</i>	100%	99,90%	100%	99,40%	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
<i>GFER</i>	99,60%	93,90%	100%	100%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
<i>GJA1</i>	100%	100%	100%	100%	Erythrokeratoderma 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
<i>GJA3</i>	100%	99,70%	100%	100%	Cataract 14, multiple types, 601885
<i>GJA8</i>	100%	100%	100%	100%	Cataract 1, multiple types, 116200
<i>GNAT1</i>	100%	100%	100%	100%	Night blindness, congenital stationary, type 1G, 616389 Night blindness, congenital stationary, autosomal dominant 3, 610444
<i>GNAT2</i>	99,90%	99,00%	100%	100%	Achromatopsia 4, 613856
<i>GNB3</i>	100%	100%	100%	100%	Night blindness, congenital stationary, type 1H, 617024
<i>GNPTG</i>	99,10%	94,30%	100%	99,90%	Mucopolipidosis III gamma, 252605
<i>GPR143</i>	85,80%	76,40%	99,80%	97,90%	Ocular albinism, type I, Nettleship-Falls type, 300500 Nystagmus 6, congenital, X-linked, 300814
<i>GPR179</i>	100%	100%	100%	100%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
<i>GRHL2</i>	100%	100%	100%	100%	Deafness, autosomal dominant 28, 608641 Corneal dystrophy, posterior polymorphous, 4, 618031 Ectodermal dysplasia/short stature syndrome, 616029
<i>GRK1</i>	100%	100%	100%	100%	Oguchi disease-2, 613411
<i>GRM6</i>	90,20%	80,60%	98,30%	96,30%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270

<i>GSN</i>	95,80%	93,50%	99,90%	99,30%	Amyloidosis, Finnish type, 105120
<i>GUCA1A</i>	100%	100%	100%	100%	Cone-rod dystrophy 14, 602093 Cone dystrophy-3, 602093
<i>GUCA1B</i>	100%	100%	100%	100%	Retinitis pigmentosa 48, 613827
<i>GUCY2D</i>	99,60%	96,20%	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
<i>HARS</i>	100%	100%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
<i>HCCS</i>	99,80%	97,60%	100%	100%	Linear skin defects with multiple congenital anomalies 1, 309801
<i>HGSNAT</i>	86,40%	86,30%	91,20%	89,30%	Retinitis pigmentosa 73, 616544 Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
<i>HK1</i>	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
<i>HMX1</i>	62,40%	42,90%	99,70%	96,10%	Oculoauricular syndrome, 612109
<i>HPS1</i>	100%	100%	100%	100%	Hermansky-Pudlak syndrome 1, 203300
<i>HPS3</i>	99,70%	97,50%	100%	100%	Hermansky-Pudlak syndrome 3, 614072
<i>HPS4</i>	100%	100%	100%	100%	Hermansky-Pudlak syndrome 4, 614073
<i>HPS5</i>	100%	99,70%	100%	100%	Hermansky-Pudlak syndrome 5, 614074
<i>HPS6</i>	97,10%	88,90%	100%	100%	Hermansky-Pudlak syndrome 6, 614075
<i>HRAS</i>	100%	100%	100%	100%	Nevus sebaceous or woolly hair nevus, somatic, 162900 Congenital myopathy with excess of muscle spindles, 218040 Bladder cancer, somatic, 109800 Thyroid carcinoma, follicular, somatic, 188470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550 Costello syndrome, 218040
<i>HSF4</i>	99,60%	97,20%	100%	100%	Cataract 5, multiple types, 116800
<i>HSPG2</i>	99,20%	97,70%	100%	99,90%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
<i>IDH3B</i>	95,40%	95,40%	100%	100%	Retinitis pigmentosa 46, 612572
<i>IFT140</i>	99,80%	98,80%	100%	100%	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
<i>IFT172</i>	99,90%	99,10%	100%	100%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630

<i>IFT27</i>	100%	100%	100%	100%	?Bardet-Biedl syndrome 19, 615996
<i>IFT43</i>	100%	100%	100%	100%	?Cranioectodermal dysplasia 3, 614099 Short-rib thoracic dysplasia 18 with polydactyly, 617866 ?Retinitis pigmentosa 81, 617871
<i>IFT74</i>	98,40%	93,90%	100%	100%	?Bardet-Biedl syndrome 20, 617119
<i>IFT81</i>	93,50%	90,10%	95,00%	94,90%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
<i>IMPDH1</i>	87,90%	80,20%	100%	100%	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
<i>IMPG1</i>	99,70%	98,50%	100%	100%	Macular dystrophy, vitelliform, 4, 616151
<i>IMPG2</i>	99,80%	98,40%	100%	100%	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581
<i>INPP5E</i>	97,10%	92,70%	100%	100%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
<i>INVS</i>	100%	100%	100%	100%	Nephronophthisis 2, infantile, 602088
<i>IQCB1</i>	93,90%	85,00%	100%	100%	Senior-Loken syndrome 5, 609254
<i>IRX1</i>	87,40%	81,30%	97,70%	94,00%	No OMIM disease ID
<i>ITPR1</i>	100%	99,90%	100%	100%	Spinocerebellar ataxia 15, 606658 Gillespie syndrome, 206700 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
<i>JAG1</i>	97,70%	96,80%	100%	100%	Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon, 617992
<i>JAM3</i>	100%	99,90%	100%	100%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
<i>KCNJ13</i>	100%	100%	100%	100%	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230
<i>KCNV2</i>	100%	99,90%	100%	100%	Retinal cone dystrophy 3B, 610356
<i>KERA</i>	100%	100%	100%	100%	Cornea plana 2, autosomal recessive, 217300
<i>KIAA1549</i>	97,90%	96,40%	98,80%	98,00%	Retinitis pigmentosa 86, 618613
<i>KIF11</i>	97,60%	94,80%	100%	100%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
<i>KIF21A</i>	99,90%	99,30%	100%	100%	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700
<i>KIF7</i>	93,60%	90,60%	99,10%	97,80%	?Hydrolethalmus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
<i>KIZ</i>	100%	99,20%	100%	100%	Retinitis pigmentosa 69, 615780
<i>KLHL7</i>	99,90%	99,80%	100%	100%	Retinitis pigmentosa 42, 612943 PERCHING syndrome, 617055

<i>KRT12</i>	99,70%	97,80%	100%	100%	Meesmann corneal dystrophy 1, 122100
<i>KRT3</i>	100%	100%	100%	100%	Meesmann corneal dystrophy 2, 618767
<i>LAMA1</i>	100%	99,70%	100%	100%	Poretti-Boltshauser syndrome, 615960
<i>LAMB2</i>	100%	99,70%	100%	100%	Pierson syndrome, 609049 Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199
<i>LAMP2</i>	99,20%	95,60%	100%	100%	Danon disease, 300257
<i>LCA5</i>	99,90%	99,20%	100%	100%	Leber congenital amaurosis 5, 604537
<i>LEMD2</i>	98,70%	92,00%	100%	100%	Cataract 46, juvenile-onset, 212500
<i>LIM2</i>	100%	100%	100%	100%	Cataract 19, multiple types, 615277
<i>LRAT</i>	100%	100%	100%	100%	Retinal dystrophy, early-onset severe, 613341 Leber congenital amaurosis 14, 613341 Retinitis pigmentosa, juvenile, 613341
<i>LRIT3</i>	93,90%	91,90%	100%	100%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
<i>LRMDA</i>	96,80%	95,60%	99,60%	99,60%	Albinism, oculocutaneous, type VII, 615179
<i>LRP2</i>	100%	99,90%	100%	100%	Donnai-Barrow syndrome, 222448
<i>LRP5</i>	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
<i>LRPAP1</i>	100%	100%	100%	100%	Myopia 23, autosomal recessive, 615431
<i>LSS</i>	100%	99,90%	100%	100%	Alopecia-mental retardation syndrome 4, 618840 Cataract 44, 616509 Hypotrichosis 14, 618275
<i>LTBP2</i>	99,90%	99,00%	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
<i>LYST</i>	99,60%	98,30%	100%	100%	Chediak-Higashi syndrome, 214500
<i>LZTFL1</i>	99,90%	99,20%	100%	100%	Bardet-Biedl syndrome 17, 615994
<i>MAB21L2</i>	100%	100%	100%	100%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
<i>MAF</i>	83,50%	78,00%	88,60%	82,20%	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
<i>MAK</i>	98,70%	96,80%	100%	100%	Retinitis pigmentosa 62, 614181
<i>MAPKAPK3</i>	100%	99,60%	100%	100%	?Macular dystrophy, patterned, 3, 617111
<i>MERTK</i>	99,50%	98,80%	99,10%	99,10%	Retinitis pigmentosa 38, 613862

<i>MFN2</i>	100%	99,90%	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
<i>MFRP</i>	100%	100%	100%	100%	Nanophthalmos 2, 609549 Microphthalmia, isolated 5, 611040
<i>MFSD8</i>	100%	99,70%	100%	100%	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
<i>MIP</i>	100%	98,90%	100%	100%	Cataract 15, multiple types, 615274
<i>MIR184</i>	NC	NC	NC	NC	EDICT syndrome, 614303
<i>MITF</i>	100%	99,90%	100%	100%	COMMAD syndrome, 617306 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 Tietz albinism-deafness syndrome, 103500
<i>MKKS</i>	83,20%	83,20%	90,70%	90,70%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
<i>MKS1</i>	99,80%	97,90%	100%	100%	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
<i>MVK</i>	90,90%	90,50%	90,50%	90,50%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
<i>MYO7A</i>	99,30%	97,40%	100%	100%	Deafness, autosomal recessive 2, 600060 Deafness, autosomal dominant 11, 601317 Usher syndrome, type 1B, 276900
<i>MYOC</i>	100%	98,60%	100%	100%	Glaucoma 1A, primary open angle, 137750
<i>NAA10</i>	99,70%	98,50%	99,90%	99,90%	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800
<i>NBAS</i>	100%	99,60%	100%	100%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
<i>NDP</i>	100%	99,70%	100%	100%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
<i>NDUFS2</i>	100%	100%	100%	100%	Mitochondrial complex I deficiency, nuclear type 6, 618228
<i>NEK2</i>	99,70%	95,50%	96,10%	96,10%	?Retinitis pigmentosa 67, 615565
<i>NEUROD1</i>	100%	99,10%	100%	100%	Maturity-onset diabetes of the young 6, 606394
<i>NHS</i>	95,40%	93,90%	100%	99,80%	Nance-Horan syndrome, 302350 Cataract 40, X-linked, 302200
<i>NMNAT1</i>	100%	99,20%	98,30%	95,60%	Leber congenital amaurosis 9, 608553

<i>NPHP1</i>	100%	99,00%	100%	100%	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
<i>NPHP3</i>	99,70%	98,40%	100%	100%	Meckel syndrome 7, 267010 Renal-hepatic-pancreatic dysplasia 1, 208540 Nephronophthisis 3, 604387
<i>NPHP4</i>	100%	99,80%	100%	100%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
<i>NR2E3</i>	100%	99,60%	100%	100%	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131
<i>NR2F1</i>	100%	100%	99,10%	95,10%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
<i>NRL</i>	99,50%	94,80%	100%	100%	Retinitis pigmentosa 27, 613750 Retinal degeneration, autosomal recessive, clumped pigment type, 0
<i>NYX</i>	96,30%	94,10%	99,70%	98,80%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
<i>OAT</i>	85,20%	76,30%	100%	100%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
<i>OCA2</i>	99,90%	98,70%	100%	100%	Albinism, oculocutaneous, type II, 203200 Albinism, brown oculocutaneous, 203200
<i>OCRL</i>	99,90%	98,60%	100%	99,90%	Lowe syndrome, 309000 Dent disease 2, 300555
<i>OFD1</i>	88,00%	73,70%	100%	99,90%	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
<i>OPA1</i>	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
<i>OPA3</i>	100%	99,00%	100%	100%	Optic atrophy 3 with cataract, 165300 3-methylglutaconic aciduria, type III, 258501
<i>OPN1LW</i>	67,20%	60,60%	98,30%	98,10%	Blue cone monochromacy, 303700 Colorblindness, protan, 303900
<i>OPN1MW</i>	66,30%	58,50%	98,90%	97,50%	Colorblindness, deutan, 303800 Blue cone monochromacy, 303700
<i>OPTN</i>	100%	99,90%	100%	100%	Glaucoma 1, open angle, E, 137760 Amyotrophic lateral sclerosis 12, 613435
<i>OTX2</i>	100%	99,70%	100%	100%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125

<i>OVOL2</i>	95,70%	89,50%	100%	100%	Corneal dystrophy, posterior polymorphous, 1, 122000
<i>P3H2</i>	99,80%	98,00%	100%	100%	Myopia, high, with cataract and vitreoretinal degeneration, 614292
<i>P4HA2</i>	100%	99,20%	100%	100%	Myopia 25, autosomal dominant, 617238
<i>PANK2</i>	100%	99,30%	100%	100%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
<i>PAX2</i>	100%	99,90%	100%	100%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
<i>PAX6</i>	100%	100%	100%	100%	Optic nerve hypoplasia, 165550 ?Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Aniridia, 106210 Keratitis, 148190 ?Coloboma of optic nerve, 120430 ?Morning glory disc anomaly, 120430 Cataract with late-onset corneal dystrophy, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229
<i>PCDH15</i>	98,60%	97,50%	100%	100%	Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083 Deafness, autosomal recessive 23, 609533
<i>PCYT1A</i>	98,90%	95,50%	100%	100%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
<i>PDE6A</i>	100%	99,60%	100%	100%	Retinitis pigmentosa 43, 613810
<i>PDE6B</i>	100%	99,90%	100%	100%	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
<i>PDE6C</i>	99,90%	97,80%	100%	100%	Cone dystrophy 4, 613093
<i>PDE6D</i>	100%	100%	100%	100%	?Joubert syndrome 22, 615665
<i>PDE6G</i>	100%	99,70%	100%	100%	Retinitis pigmentosa 57, 613582
<i>PDE6H</i>	100%	97,90%	100%	100%	Retinal cone dystrophy 3, 610024 Achromatopsia 6, 610024
<i>PDZD7</i>	98,80%	96,30%	100%	100%	Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 Deafness, autosomal recessive 57, 618003
<i>PET100</i>	100%	99,60%	100%	100%	Mitochondrial complex IV deficiency, 220110
<i>PEX1</i>	99,90%	99,40%	100%	100%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
<i>PEX2</i>	100%	100%	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
<i>PEX26</i>	100%	100%	100%	100%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873

<i>PEX6</i>	94,50%	86,70%	100%	100%	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862
<i>PEX7</i>	87,80%	80,70%	91,30%	91,30%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
<i>PGK1</i>	92,80%	79,30%	100%	100%	Phosphoglycerate kinase 1 deficiency, 300653
<i>PHOX2A</i>	91,60%	72,70%	100%	99,80%	Fibrosis of extraocular muscles, congenital, 2, 602078
<i>PHYH</i>	100%	99,60%	100%	100%	Refsum disease, 266500
<i>TACSTD2</i>	99,00%	96,40%	100%	100%	Corneal dystrophy, gelatinous drop-like, 204870
<i>PIKFYVE</i>	99,90%	99,40%	100%	100%	Corneal fleck dystrophy, 121850
<i>PITX2</i>	99,90%	97,70%	100%	100%	Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550 Anterior segment dysgenesis 4, 137600
<i>PITX3</i>	100%	98,40%	100%	100%	Anterior segment dysgenesis 1, multiple subtypes, 107250 Cataract 11, syndromic, autosomal recessive, 610623 Cataract 11, multiple types, 610623
<i>PLA2G5</i>	100%	100%	100%	100%	No OMIM disease ID
<i>PLK4</i>	99,90%	98,20%	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
<i>PNPLA6</i>	100%	99,70%	100%	100%	Spastic paraplegia 39, autosomal recessive, 612020 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800
<i>POC1B</i>	99,80%	98,80%	100%	100%	Cone-rod dystrophy 20, 615973
<i>POC5</i>	99,70%	97,60%	100%	100%	No OMIM disease ID
<i>POMGNT1</i>	100%	99,90%	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
<i>PPT1</i>	90,30%	90,30%	82,50%	82,50%	Ceroid lipofuscinosis, neuronal, 1, 256730
<i>PRCD</i>	100%	100%	100%	100%	Retinitis pigmentosa 36, 610599
<i>PRDM13</i>	99,20%	94,10%	100%	100%	No OMIM disease ID
<i>PRDM5</i>	99,90%	99,20%	100%	100%	Brittle cornea syndrome 2, 614170
<i>PRIMPOL</i>	97,50%	94,60%	100%	100%	Myopia 22, autosomal dominant, 615420
<i>PROM1</i>	97,20%	96,10%	100%	100%	Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786 Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051
<i>PRPF3</i>	98,80%	95,30%	100%	100%	Retinitis pigmentosa 18, 601414

<i>PRPF31</i>	100%	98,70%	100%	100%	Retinitis pigmentosa 11, 600138
<i>PRPF4</i>	100%	99,80%	100%	100%	Retinitis pigmentosa 70, 615922
<i>PRPF6</i>	100%	99,80%	100%	100%	Retinitis pigmentosa 60, 613983
<i>PRPF8</i>	100%	99,30%	100%	100%	Retinitis pigmentosa 13, 600059
<i>PRPH2</i>	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
<i>PRSS56</i>	99,90%	96,40%	100%	100%	Microphthalmia, isolated 6, 613517
<i>PXDN</i>	100%	99,60%	100%	100%	Anterior segment dysgenesis 7, with sclerocornea, 269400
<i>RAB28</i>	99,70%	96,00%	100%	100%	Cone-rod dystrophy 18, 615374
<i>RAB3GAP2</i>	99,50%	97,00%	100%	100%	Warburg micro syndrome 2, 614225 Martsolf syndrome, 212720
<i>RARB</i>	100%	100%	100%	100%	Microphthalmia, syndromic 12, 615524
<i>RAX</i>	96,00%	87,00%	100%	98,40%	Microphthalmia, isolated 3, 611038
<i>RAX2</i>	100%	92,30%	100%	100%	?Macular degeneration, age-related, 6, 613757 Cone-rod dystrophy 11, 610381
<i>RBP3</i>	100%	100%	100%	100%	?Retinitis pigmentosa 66, 615233
<i>RBP4</i>	99,90%	97,70%	100%	100%	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
<i>RCBTB1</i>	99,90%	99,50%	100%	100%	Retinal dystrophy with or without extraocular anomalies, 617175
<i>RD3</i>	100%	100%	100%	100%	Leber congenital amaurosis 12, 610612
<i>RDH11</i>	100%	99,00%	100%	100%	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
<i>RDH12</i>	100%	98,60%	100%	100%	Leber congenital amaurosis 13, 612712
<i>RDH5</i>	100%	99,90%	100%	100%	Fundus albipunctatus, 136880
<i>REEP6</i>	100%	100%	100%	100%	Retinitis pigmentosa 77, 617304
<i>RGS9</i>	98,50%	97,10%	100%	100%	Bradyopsia, 608415
<i>RGS9BP</i>	99,60%	95,00%	100%	100%	Bradyopsia, 608415
<i>RHO</i>	100%	100%	100%	100%	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis punctata albescens, 136880 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731
<i>RIMS1</i>	99,80%	97,70%	100%	100%	Cone-rod dystrophy 7, 603649
<i>RLBP1</i>	100%	99,90%	100%	100%	Fundus albipunctatus, 136880 Retinitis punctata albescens, 136880 Bothnia retinal dystrophy, 607475 Newfoundland rod-cone dystrophy, 607476

<i>ROM1</i>	100%	99,90%	100%	100%	Retinitis pigmentosa 7, digenic form, 608133
<i>RP1</i>	91,50%	90,60%	100%	100%	Retinitis pigmentosa 1, 180100
<i>RP1L1</i>	100%	100%	100%	100%	Retinitis pigmentosa 88, 618826 Occult macular dystrophy, 613587
<i>RP2</i>	100%	99,80%	100%	100%	Retinitis pigmentosa 2, 312600
<i>RP9</i>	80,80%	75,90%	100%	99,30%	?Retinitis pigmentosa 9, 180104
<i>RPE65</i>	99,80%	97,80%	100%	100%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 87 with choroidal involvement, 618697 Retinitis pigmentosa 20, 613794
<i>RPGR</i>	77,30%	72,70%	100%	99,90%	Cone-rod dystrophy, X-linked, 1, 304020 Retinitis pigmentosa 3, 300029 Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 Macular degeneration, X-linked atrophic, 300834
<i>RPGRIP1</i>	100%	99,90%	100%	100%	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
<i>RPGRIP1L</i>	96,70%	95,70%	100%	99,50%	COACH syndrome, 216360 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
<i>RS1</i>	99,80%	93,90%	100%	100%	Retinoschisis, 312700
<i>RTN4IP1</i>	99,90%	98,70%	100%	100%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
<i>SAG</i>	100%	100%	100%	100%	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
<i>SAMD11</i>	90,20%	81,60%	100%	100%	No OMIM disease ID
<i>SC5D</i>	100%	99,50%	100%	100%	Lathosterolosis, 607330
<i>SCAPER</i>	99,70%	98,20%	100%	100%	Intellectual developmental disorder and retinitis pigmentosa, 618195
<i>SCO2</i>	100%	100%	100%	100%	Myopia 6, 608908 Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377
<i>SDCCAG8</i>	100%	99,90%	100%	100%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
<i>SEMA4A</i>	100%	99,80%	100%	100%	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
<i>SGSH</i>	94,40%	94,10%	100%	100%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
<i>SHH</i>	100%	99,50%	100%	100%	Schizencephaly, 269160 Microphthalmia with coloboma 5, 611638 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
<i>SIPA1L3</i>	99,90%	98,90%	100%	100%	?Cataract 45, 616851
<i>SIX6</i>	100%	100%	100%	100%	Optic disc anomalies with retinal and/or macular dystrophy, 212550

<i>SLC16A12</i>	100%	99,90%	100%	100%	Cataract 47, juvenile, with microcornea, 612018
<i>SLC24A1</i>	100%	99,90%	100%	100%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
<i>SLC24A5</i>	99,90%	99,10%	100%	100%	Albinism, oculocutaneous, type VI, 113750
<i>SLC25A46</i>	99,70%	97,30%	100%	100%	Neuropathy, hereditary motor and sensory, type VIB, 616505
<i>SLC33A1</i>	99,90%	98,90%	100%	100%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
<i>SLC38A8</i>	99,90%	97,30%	100%	100%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
<i>SLC39A5</i>	99,90%	99,00%	100%	100%	Myopia 24, autosomal dominant, 615946
<i>SLC45A2</i>	100%	99,90%	100%	100%	Albinism, oculocutaneous, type IV, 606574
<i>SLC4A11</i>	100%	99,90%	100%	100%	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy, autosomal recessive, 217700 Corneal endothelial dystrophy and perceptive deafness, 217400
<i>SLC52A2</i>	100%	100%	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
<i>SLC7A14</i>	100%	100%	100%	100%	Retinitis pigmentosa 68, 615725
<i>SMOC1</i>	99,90%	98,40%	100%	100%	Microphthalmia with limb anomalies, 206920
<i>SNRNP200</i>	99,90%	99,10%	100%	100%	Retinitis pigmentosa 33, 610359
<i>SOX2</i>	100%	100%	100%	100%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
<i>SOX5</i>	99,90%	98,90%	100%	100%	Lamb-Shaffer syndrome, 616803
<i>SPATA7</i>	99,80%	98,20%	100%	100%	Retinitis pigmentosa, juvenile, autosomal recessive, 604232 Leber congenital amaurosis 3, 604232
<i>SPP2</i>	100%	99,90%	100%	100%	No OMIM disease ID
<i>STRA6</i>	100%	99,80%	100%	100%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
<i>TCF4</i>	100%	99,80%	100%	100%	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
<i>TCTN1</i>	96,70%	93,00%	94,70%	94,70%	Joubert syndrome 13, 614173
<i>TCTN3</i>	100%	100%	100%	100%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
<i>TDRD7</i>	99,90%	99,10%	100%	100%	Cataract 36, 613887
<i>TEAD1</i>	100%	99,90%	100%	100%	Sveinsson chorioretinal atrophy, 108985
<i>TEK</i>	100%	100%	100%	100%	Glaucoma 3, primary congenital, E, 617272 Venous malformations, multiple cutaneous and mucosal, 600195
<i>TENM3</i>	100%	99,70%	100%	100%	Microphthalmia, syndromic 15, 615145 ?Microphthalmia, isolated, with coloboma 9, 615145
<i>TGFBI</i>	99,50%	94,60%	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
<i>TIMM8A</i>	98,00%	90,10%	100%	100%	Mohr-Tranebjaerg syndrome, 304700
<i>TIMP3</i>	100%	100%	100%	100%	Sorsby fundus dystrophy, 136900
<i>TMCO3</i>	100%	99,40%	100%	100%	No OMIM disease ID
<i>TMEM126A</i>	96,30%	84,40%	100%	100%	Optic atrophy 7, 612989
<i>TMEM138</i>	100%	99,10%	100%	100%	Joubert syndrome 16, 614465
<i>TMEM216</i>	99,90%	98,10%	100%	100%	Meckel syndrome 2, 603194 Joubert syndrome 2, 608091
<i>TMEM231</i>	100%	99,60%	100%	100%	Meckel syndrome 11, 615397 Joubert syndrome 20, 614970
<i>TMEM237</i>	100%	99,90%	100%	100%	Joubert syndrome 14, 614424
<i>TMEM67</i>	99,50%	95,00%	100%	99,90%	Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550 COACH syndrome, 216360 Joubert syndrome 6, 610688
<i>TMEM98</i>	99,30%	97,80%	100%	100%	Nanophthalmos 4, 615972
<i>TOPORS</i>	100%	100%	100%	100%	Retinitis pigmentosa 31, 609923
<i>TPP1</i>	100%	100%	100%	100%	Spinocerebellar ataxia, autosomal recessive 7, 609270 Ceroid lipofuscinosis, neuronal, 2, 204500
<i>TRAF3IP1</i>	99,60%	97,60%	100%	100%	Senior-Loken syndrome 9, 616629
<i>TREX1</i>	100%	100%	100%	100%	Vasculopathy, retinal, with cerebral leukodystrophy, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
<i>TRIM32</i>	100%	100%	100%	100%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
<i>TRNT1</i>	99,50%	96,50%	100%	100%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
<i>TRPM1</i>	100%	99,80%	100%	100%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
<i>TSPAN12</i>	100%	99,80%	100%	100%	Exudative vitreoretinopathy 5, 613310
<i>TTC8</i>	99,60%	98,10%	100%	100%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
<i>TTL5</i>	100%	99,70%	100%	100%	Cone-rod dystrophy 19, 615860
<i>TUB</i>	99,40%	97,10%	100%	100%	?Retinal dystrophy and obesity, 616188
<i>TUBA3D</i>	100%	99,20%	100%	100%	Keratoconus 9, 617928

<i>TUBB3</i>	98,30%	96,90%	100%	100%	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
<i>TUBB4B</i>	99,90%	96,90%	100%	100%	Leber congenital amaurosis with early-onset deafness, 617879
<i>TUBGCP4</i>	99,20%	96,40%	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
<i>TUBGCP6</i>	100%	99,30%	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
<i>TULP1</i>	100%	99,50%	100%	100%	Retinitis pigmentosa 14, 600132 Leber congenital amaurosis 15, 613843
<i>TYR</i>	100%	100%	100%	100%	Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IB, 606952 Albinism, oculocutaneous, type IA, 203100
<i>TYRP1</i>	100%	99,80%	100%	100%	Albinism, oculocutaneous, type III, 203290
<i>UBIAD1</i>	99,50%	96,00%	100%	100%	Corneal dystrophy, Schnyder type, 121800
<i>UNC45B</i>	99,30%	98,00%	100%	100%	?Cataract 43, 616279
<i>USH1C</i>	100%	99,80%	100%	100%	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
<i>USH1G</i>	99,60%	97,90%	100%	100%	Usher syndrome, type 1G, 606943
<i>USH2A</i>	100%	99,80%	100%	100%	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
<i>USP45</i>	99,60%	98,10%	100%	100%	?Leber congenital amaurosis 19, 618513
<i>VAX1</i>	97,50%	91,50%	95,70%	91,70%	?Microphthalmia, syndromic 11, 614402
<i>VCAN</i>	100%	100%	100%	100%	Wagner syndrome 1, 143200
<i>VIM</i>	99,30%	97,00%	100%	100%	Cataract 30, pulverulent, 116300
<i>VPS13B</i>	99,50%	98,20%	99,50%	99,40%	Cohen syndrome, 216550
<i>VSX1</i>	84,70%	80,50%	100%	100%	Keratoconus 1, 148300 ?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195
<i>VSX2</i>	100%	99,30%	100%	100%	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
<i>WDPCP</i>	98,20%	94,40%	98,10%	98,10%	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
<i>WDR19</i>	100%	99,40%	100%	100%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
<i>WDR36</i>	99,70%	97,60%	100%	100%	Glaucoma 1, open angle, G, 609887
<i>WFS1</i>	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
WRN	99,90%	98,80%	100%	100%	Werner syndrome, 277700
YAP1	96,40%	89,40%	100%	100%	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
YME1L1	99,00%	95,20%	100%	100%	?Optic atrophy 11, 617302
ZEB1	100%	99,40%	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 HGNC 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