VISION DISORDERS PANEL DG-5.1.0 (634 GENES)

Gene	Twist X2 covered 10x	Twist X2 covered 20x	srWGS covered 10x	srWGS covered 15x	srWGS covered 20x	Associated Phenotype description and OMIM disease ID
ABCA4	100%	100%	100%	100%	99.7%	Retinal dystrophy, early-onset severe, 248200;Retinitis pigmentosa 19, 601718;{Macular degeneration, age-related, 2}, 153800;Cone-rod dystrophy 3, 604116;Fundus flavimaculatus, 248200;Stargardt disease 1, 248200
ABCB6	100%	100%	100%	100%	99.5%	Dyschromatosis universalis hereditaria 3, 615402;[Blood group, Langereis system], 111600;Pseudohyperkalemia, familial, 2, due to red cell leak, 609153;Microphthalmia/coloboma 7, 614497
ABCC6	94.4%	94.4%	100%	100%	99.5%	Pseudoxanthoma elasticum, 264800;Arterial calcification, generalized, of infancy, 2, 614473;Pseudoxanthoma elasticum, forme fruste, 177850
ABCD1	89.3%	86.3%	98.7%	89.2%	70.9%	Adrenoleukodystrophy, 300100;Adrenomyeloneuropathy, adult, 300100
ABHD12	100%	100%	100%	100%	99.6%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674

ACBD5	85.6%	85.6%	100%	100%	99.5%	Retinal dystrophy with leukodystrophy, 618863
ACO2	90.7%	86.8%	100%	100%	99.7%	Optic atrophy 9, 616289;Infantile cerebellar-retinal degeneration, 614559
ADAM9	95%	95%	100%	100%	99.6%	Cone-rod dystrophy 9, 612775
ADAMTS10	100%	100%	100%	100%	99.6%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS17	100%	100%	100%	100%	99.3%	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS18	100%	100%	100%	100%	99.7%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTSL4	100%	100%	100%	100%	99.4%	Ectopia lentis et pupillae, 225200;Ectopia lentis, isolated, autosomal recessive, 225100
ADGRV1	100%	100%	100%	100%	99.8%	Usher syndrome, type 2C, 605472;Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472;?Febrile seizures, familial, 4, 604352
ADIPOR1	100%	100%	100%	100%	99.8%	
AFG3L2	94.2%	92.2%	100%	100%	99.5%	Spastic ataxia 5, autosomal recessive, 614487;Optic atrophy 12, 618977;Spinocerebellar ataxia 28, 610246
AGBL1	100%	100%	100%	100%	99.8%	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGBL5	100%	100%	100%	100%	99.3%	Retinitis pigmentosa 75, 617023

AGK	91.6%	88.9%	100%	100%	100%	Cataract 38, autosomal recessive, 614691;Sengers syndrome, 212350
AGXT	97.4%	96.4%	100%	100%	99.5%	Hyperoxaluria, primary, type 1, 259900
AHI1	98.7%	98.7%	100%	99.9%	99.6%	Joubert syndrome 3, 608629
AHR	100%	100%	100%	100%	99.8%	Foveal hypoplasia 3, 620958;?Retinitis pigmentosa 85, 618345
AIPL1	100%	100%	100%	99.9%	98.9%	Leber congenital amaurosis 4, 604393;Retinitis pigmentosa, juvenile, 604393;Cone-rod dystrophy, 604393
ALDH18A1	100%	100%	100%	100%	99.6%	Spastic paraplegia 9A, autosomal dominant, 601162;Cutis laxa, autosomal recessive, type IIIA, 219150;Spastic paraplegia 9B, autosomal recessive, 616586;Cutis laxa, autosomal dominant 3, 616603
ALDH1A3	94.4%	94.4%	100%	99.9%	99.4%	Microphthalmia, isolated 8, 615113
ALDH3A2	93.5%	93.5%	100%	100%	99.7%	Sjogren-Larsson syndrome, 270200
ALMS1	100%	100%	100%	100%	99.8%	Alstrom syndrome, 203800
ALPK1	100%	100%	100%	100%	99.7%	ROSAH syndrome, 614979
AMACR	100%	100%	100%	99.8%	99.4%	Alpha-methylacyl-CoA racemase deficiency, 614307;Bile acid synthesis defect, congenital, 4, 214950

ANK3	99.7%	99.7%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 37, 615493
AP3B1	100%	100%	100%	100%	99.8%	Hermansky-Pudlak syndrome 2, 608233
AP3D1	100%	100%	100%	100%	99.7%	?Hermansky-Pudlak syndrome 10, 617050
AP5B1	100%	100%	100%	100%	99.4%	
AP5M1	100%	100%	100%	100%	99.9%	
AP5Z1	91.2%	91%	100%	100%	99.5%	Spastic paraplegia 48, autosomal recessive, 613647
ARHGAP35	100%	100%	100%	100%	99.5%	
ARHGEF18	100%	100%	100%	100%	99.3%	Retinitis pigmentosa 78, 617433
ARL13B	93.4%	93.3%	100%	100%	99.7%	Joubert syndrome 8, 612291
ARL2	100%	100%	100%	100%	99%	?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 1, 619082
ARL2BP	100%	100%	100%	100%	99.9%	Retinitis pigmentosa 82 with or without situs inversus, 615434
ARL3	86.8%	86.8%	100%	100%	99.8%	Retinitis pigmentosa 83, 618173;Joubert syndrome 35, 618161
ARL6	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 55, 613575;{Bardet-Biedl syndrome 1, modifier of}, 209900;Bardet-Biedl syndrome 3, 600151
ARR3	100%	100%	99.5%	90.6%	69.9%	Myopia 26, X-linked, female-limited, 301010
ARSG	100%	100%	100%	100%	99.1%	Usher syndrome, type IV, 618144
ASB10	100%	100%	100%	100%	99.7%	Glaucoma 1, open angle, F, 603383

ASPH	100%	100%	100%	100%	99.7%	Traboulsi syndrome, 601552
ASRGL1	100%	100%	100%	100%	100%	
ATF6	90.9%	90.9%	100%	100%	99.7%	Achromatopsia 7, 616517
ATOH7	100%	100%	100%	100%	99.6%	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
AUH	100%	100%	100%	100%	99.7%	3-methylglutaconic aciduria, type I, 250950
B3GLCT	100%	100%	100%	100%	99.9%	Peters-plus syndrome, 261540
BAIAP3	100%	100%	100%	99.9%	99.4%	
BBIP1	100%	100%	100%	100%	99.7%	Bardet-Biedl syndrome 18, 615995
BBS1	93.7%	93.7%	100%	100%	99.3%	Bardet-Biedl syndrome 1, 209900
BBS10	100%	100%	100%	100%	99.8%	Bardet-Biedl syndrome 10, 615987
BBS12	100%	100%	100%	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	98%	98%	100%	100%	99.8%	Retinitis pigmentosa 74, 616562;Bardet-Biedl syndrome 2, 615981
BBS4	100%	100%	100%	100%	99.7%	Bardet-Biedl syndrome 4, 615982
BBS5	100%	100%	100%	100%	99.9%	Bardet-Biedl syndrome 5, 615983
BBS7	100%	100%	100%	100%	99.9%	Bardet-Biedl syndrome 7, 615984
BBS9	91.9%	91.9%	100%	100%	99.7%	Bardet-Biedl syndrome 9, 615986
BCOR	100%	100%	98.9%	89.4%	69.8%	Microphthalmia, syndromic 2, 300166

BEST1	100%	100%	100%	99.9%	98.6%	Macular dystrophy, vitelliform, 2, 153700;Retinitis pigmentosa 50, 613194;?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 2, 193220;Retinitis pigmentosa, concentric, 613194;Vitreoretinochoroidopathy, 193220;Bestrophinopathy, autosomal recessive, 611809
BFSP1	100%	100%	100%	99.9%	99.4%	Cataract 33, multiple types, 611391
BFSP2	100%	100%	100%	99.9%	99.1%	Cataract 12, multiple types, 611597
BLOC1S3	100%	100%	100%	100%	98.5%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S5	100%	100%	100%	100%	99.7%	Hermansky-Pudlak syndrome 11, 619172
BLOC1S6	100%	100%	100%	100%	99.9%	Hermansky-Pudlak syndrome 9, 614171
BMP4	100%	100%	100%	100%	99.5%	Orofacial cleft 11, 600625;Microphthalmia, syndromic 6, 607932
BMPR1B	95.6%	94.1%	100%	100%	99.7%	Acromesomelic dysplasia 3, 609441;Brachydactyly, type A2, 112600;Brachydactyly, type A1, D, 616849
BTD	94.2%	94.2%	100%	100%	99.9%	Biotinidase deficiency, 253260
C19orf12	100%	100%	100%	99.9%	98.7%	Neurodegeneration with brain iron accumulation 4, 614298;?Spastic paraplegia 43, autosomal recessive, 615043
C19orf44	100%	100%	100%	100%	99.4%	

C1QTNF5	100%	100%	100%	100%	99.6%	Retinal degeneration, late-onset, autosomal dominant, 605670
CABP4	100%	100%	100%	100%	99.5%	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACNA1F	100%	100%	98%	86.9%	67.2%	Cone-rod dystrophy, X-linked, 3, 300476;Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071;Aland Island eye disease, 300600
CACNA2D4	100%	100%	100%	100%	99.5%	Retinal cone dystrophy 4, 610478
CAPN5	100%	99.9%	100%	100%	99.4%	Vitreoretinopathy, neovascular inflammatory, 193235
CBS	87.7%	85.5%	100%	100%	99.5%	Thrombosis, hyperhomocysteinemic, 236200;Homocystinuria, B6-responsive and nonresponsive types, 236200
CC2D2A	98.2%	98.2%	100%	100%	99.8%	COACH syndrome 2, 619111;Retinitis pigmentosa 93, 619845;Meckel syndrome 6, 612284;Joubert syndrome 9, 612285
CCT2	97.1%	94.6%	100%	100%	99.9%	
CDH2	100%	100%	100%	100%	99.7%	Arrhythmogenic right ventricular dysplasia 14, 618920;?Attention deficit-hyperactivity disorder 8, 619957;Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929

CDH23	100%	100%	100%	100%	99.4%	Usher syndrome, type 1D, 601067;{Pituitary adenoma 5, multiple types}, 617540;Usher syndrome, type 1D/F digenic, 601067;Deafness, autosomal recessive 12, 601386
CDH3	100%	100%	100%	100%	99.6%	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553;Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280
CDH4	100%	100%	100%	100%	99.6%	
CDHR1	100%	100%	100%	100%	99.7%	Macular dystrophy, retinal, 613660;Cone-rod dystrophy 15, 613660;Retinitis pigmentosa 65, 613660
CDK10	100%	100%	100%	100%	99.6%	Al Kaissi syndrome, 617694
CDON	100%	100%	100%	100%	99.9%	Holoprosencephaly 11, 614226
CEP120	100%	100%	100%	100%	99.6%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300;Joubert syndrome 31, 617761
CEP162	100%	100%	100%	100%	99.8%	
CEP164	100%	100%	100%	100%	99.5%	Nephronophthisis 15, 614845
CEP250	100%	100%	100%	100%	99.5%	Cone-rod dystrophy and hearing loss 2, 618358

CEP290	100%	100%	100%	100%	99.8%	Leber congenital amaurosis 10, 611755;Joubert syndrome 5, 610188;Senior-Loken syndrome 6, 610189;?Bardet-Biedl syndrome 14, 615991;Meckel syndrome 4, 611134
CEP41	100%	100%	100%	100%	99.8%	Joubert syndrome 15, 614464
CEP78	100%	100%	100%	100%	99.6%	Cone-rod dystrophy and hearing loss, 617236
CEP83	100%	100%	100%	100%	99.8%	Nephronophthisis 18, 615862
CERKL	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 26, 608380
CFAP410	100%	100%	100%	100%	99.3%	Retinal dystrophy with macular staphyloma, 617547;Spondylometaphyseal dysplasia, axial, 602271
CFAP418	100%	100%	100%	100%	99.5%	Retinitis pigmentosa 64, 614500;Cone-rod dystrophy 16, 614500;Bardet-Biedl syndrome 21, 617406
CFH	97.5%	97.5%	100%	100%	99.9%	{Macular degeneration, age-related, 4}, 610698;Basal laminar drusen, 126700;Complement factor H deficiency, 609814;{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400
CHD7	100%	100%	100%	100%	99.8%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800
CHM	99.5%	98.5%	98.8%	92.3%	75.2%	Choroideremia, 303100

CHMP4B	100%	100%	100%	100%	99.4%	Cataract 31, multiple types, 605387
CHN1	96.5%	96.5%	100%	100%	99.7%	Duane retraction syndrome 2, 604356
CHRDL1	100%	100%	99.3%	91.9%	74.5%	Megalocornea 1, X-linked, 309300
CHST6	100%	100%	100%	100%	99.6%	Macular corneal dystrophy, 217800
CIB2	100%	100%	100%	100%	99.9%	Deafness, autosomal recessive 48, 609439
CISD2	100%	100%	100%	99.9%	99.4%	Wolfram syndrome 2, 604928
CLCC1	98.6%	98.6%	100%	100%	99.7%	Retinitis pigmentosa 32, 609913
CLDN19	100%	100%	100%	100%	99.5%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLEC3B	100%	100%	100%	100%	99.9%	Macular dystrophy, retinal, 4, 619977
CLN3	93.1%	93.1%	100%	100%	99.4%	Ceroid lipofuscinosis, neuronal, 3, 204200;Retinitis pigmentosa 101, 621548
CLN5	83%	83%	100%	100%	99.8%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	95.1%	95.1%	100%	100%	99.4%	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300;Ceroid lipofuscinosis, neuronal, 6A, 601780
CLN8	100%	100%	100%	100%	99.5%	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003;Ceroid lipofuscinosis, neuronal, 8, 600143
CLRN1	100%	100%	100%	99.9%	99.1%	Usher syndrome, type 3A, 276902;Retinitis pigmentosa 61, 614180
CLUAP1	100%	100%	100%	100%	99.5%	

CNGA1	100%	100%	100%	100%	99.7%	Retinitis pigmentosa 49, 613756
CNGA3	100%	100%	100%	100%	99.2%	Achromatopsia 2, 216900
CNGB1	100%	100%	100%	100%	99.4%	Retinitis pigmentosa 45, 613767
CNGB3	100%	100%	100%	99.9%	99.5%	Achromatopsia 3, 262300
CNNM4	100%	100%	100%	100%	99.4%	Jalili syndrome, 217080
COA8	100%	100%	100%	100%	99.7%	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COL11A1	100%	100%	100%	100%	99.8%	Fibrochondrogenesis 1, 228520;Stickler syndrome, type II, 604841;Marshall syndrome, 154780;Deafness, autosomal dominant 37, 618533;{Lumbar disc herniation, susceptibility to}, 603932
COL17A1	100%	100%	100%	100%	99.4%	Epithelial recurrent erosion dystrophy, 122400;Epidermolysis bullosa, junctional 4, intermediate, 619787
COL18A1	99.2%	98%	100%	99.9%	99.2%	Knobloch syndrome, type 1, 267750;Glaucoma, primary closed-angle, 618880

COL1A1	100%	100%	100%	100%	99.5%	Osteogenesis imperfecta, type II, 166210;Caffey disease, 114000;Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060;Osteogenesis imperfecta, type I, 166200;{Bone mineral density variation QTL, osteoporosis}, 166710;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115;Osteogenesis imperfecta, type IV, 166220;Osteogenesis imperfecta, type III, 259420
COL25A1	100%	100%	100%	100%	99.7%	Fibrosis of extraocular muscles, congenital, 5, 616219

COL2A1	100%	100%	100%	100%	99.5%	?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248;Czech dysplasia, 609162;Achondrogenesis, type II or hypochondrogenesis, 200610;Spondyloperipheral dysplasia, 271700;SMED Strudwick type, 184250;?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450;Spondylometaphyseal dysplasia, Algerian type, 184253;SED congenita, 183900;Kniest dysplasia, 156550;Stickler syndrome, type I, nonsyndromic ocular, 609508;Osteoarthritis with mild chondrodysplasia, 604864;Stickler syndrome, type I, 108300;Platyspondylic skeletal dysplasia, Torrance type, 151210;Spondyloepiphyseal dysplasia, Stanescu type, 616583;Avascular necrosis of the femoral head, 608805;Legg-Calve-Perthes disease, 150600
COL3A1	100%	100%	100%	100%	99.7%	Ehlers-Danlos syndrome, vascular type, 130050;Polymicrogyria with or without vascular-type EDS, 618343

COL4A1	100%	100%	100%	100%	99.7%	?Retinal arteries, tortuosity of, 180000;{Hemorrhage, intracerebral, susceptibility to}, 614519;Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773;Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564;Brain small vessel disease with or without ocular anomalies, 175780
COL5A1	100%	100%	100%	100%	99.4%	Ehlers-Danlos syndrome, classic type, 1, 130000;Fibromuscular dysplasia, multifocal, 619329
COL8A2	100%	100%	100%	100%	98.7%	Corneal dystrophy, posterior polymorphous 2, 609140;Corneal dystrophy, Fuchs endothelial, 1, 136800
COL9A1	100%	100%	100%	100%	99.7%	Stickler syndrome, type IV, 614134;?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	100%	100%	100%	100%	99.3%	Epiphyseal dysplasia, multiple, 2, 600204;?Stickler syndrome, type V, 614284
COL9A3	100%	100%	100%	100%	99.4%	{Intervertebral disc disease, susceptibility to}, 603932;Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969;Stickler syndrome, type VI, 620022

COQ2	96.3%	96.3%	100%	100%	99.5%	{Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426
COQ8B	92.1%	92.1%	100%	100%	99.6%	Nephrotic syndrome, type 9, 615573
COX7B	100%	100%	98.7%	86.9%	70%	Linear skin defects with multiple congenital anomalies 2, 300887
CPAMD8	100%	100%	100%	99.9%	99.3%	Anterior segment dysgenesis 8, 617319
CPLANE1	100%	100%	100%	100%	99.8%	Orofaciodigital syndrome VI, 277170;Joubert syndrome 17, 614615
CPSF1	100%	100%	100%	99.9%	99%	Myopia 27, 618827
CRB1	98.6%	98.6%	100%	100%	99.9%	Leber congenital amaurosis 8, 613835;Retinitis pigmentosa-12, 600105;Pigmented paravenous chorioretinal atrophy, 172870
CREB3	100%	100%	100%	99.9%	99.4%	
CREBBP	100%	100%	100%	99.9%	99.5%	Menke-Hennekam syndrome 1, 618332;Rubinstein-Taybi syndrome 1, 180849
CRX	100%	100%	100%	100%	99.1%	Leber congenital amaurosis 7, 613829;Cone-rod retinal dystrophy-2, 120970
CRYAA	100%	100%	100%	100%	99.3%	Cataract 9, multiple types, 604219

CRYAB	100%	100%	100%	100%	99.7%	Myopathy, myofibrillar, 2B, infantile-onset, 613869;Myopathy, myofibrillar, 2A, adult-onset, 608810;Cataract 16, multiple types, 613763;Cardiomyopathy, dilated, 1II, 615184
CRYBA1	100%	100%	100%	100%	99.7%	Cataract 10, multiple types, 600881
CRYBA2	100%	100%	100%	99.9%	98.7%	?Cataract 42, 115900
CRYBA4	100%	100%	100%	100%	99.8%	Cataract 23, 610425
CRYBB1	100%	100%	100%	100%	99.5%	Cataract 17, multiple types, 611544
CRYBB2	100%	100%	100%	100%	99.9%	Cataract 3, multiple types, 601547
CRYBB3	100%	100%	100%	100%	99.6%	Cataract 22, 609741
CRYGB	100%	100%	100%	100%	99.6%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	100%	100%	100%	100%	99.3%	Cataract 2, multiple types, 604307
CRYGD	100%	100%	100%	99.9%	99.3%	Cataract 4, multiple types, 115700
CRYGS	100%	100%	100%	100%	99.8%	Cataract 20, multiple types, 116100
CSPP1	96.9%	96.9%	100%	100%	99.6%	Joubert syndrome 21, 615636
CTC1	100%	100%	100%	100%	99.4%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTDP1	90.8%	90.5%	100%	100%	99.2%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNA1	94.8%	94.8%	100%	100%	99.8%	Macular dystrophy, patterned, 2, 608970

CTNNB1	100%	100%	100%	100%	99.9%	Exudative vitreoretinopathy 7, 617572;Pilomatricoma, somatic, 132600;Colorectal cancer, somatic, 114500;Neurodevelopmental disorder with spastic diplegia and visual defects, 615075;Medulloblastoma, somatic, 155255;Ovarian cancer, somatic, 167000;Hepatocellular carcinoma, somatic, 114550
CTSD	86%	86%	100%	100%	99.5%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSH	93.4%	92.1%	100%	100%	99.3%	
CWC27	82.6%	82.6%	100%	100%	99.8%	Retinitis pigmentosa with or without skeletal anomalies, 250410
CYP1B1	100%	100%	100%	100%	99.5%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300;Anterior segment dysgenesis 6, multiple subtypes, 617315
CYP27A1	100%	100%	100%	100%	99.5%	Cerebrotendinous xanthomatosis, 213700
CYP4V2	100%	100%	100%	100%	99.8%	Bietti crystalline corneoretinal dystrophy, 210370
DCDC1	100%	100%	100%	100%	99.8%	
DCN	89%	89%	100%	100%	99.8%	Corneal dystrophy, congenital stromal, 610048
DCT	100%	100%	100%	99.8%	98.9%	Oculocutaneous albinism, type VIII, 619165
DDHD1	100%	100%	100%	100%	99.3%	Spastic paraplegia 28, autosomal recessive, 609340

DHDDS	74.4%	74.4%	100%	100%	99.8%	Developmental delay and seizures with or without movement abnormalities, 617836;?Congenital disorder of glycosylation, type 1bb, 621567;Retinitis pigmentosa 59, 613861
DHX38	100%	100%	100%	100%	99.6%	Retinitis pigmentosa 84, 618220
DKC1	100%	100%	99%	89.5%	70.6%	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000
DNAJC30	100%	100%	100%	100%	99.7%	Leber-like hereditary optic neuropathy, autosomal recessive 1, 619382
DNM1L	100%	100%	100%	100%	99.9%	Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DNMBP	100%	100%	100%	100%	99.6%	Cataract 48, 618415
DRAM2	98.4%	93.3%	100%	100%	99.7%	Cone-rod dystrophy 21, 616502
DTNBP1	96.4%	96.4%	100%	100%	99.7%	Hermansky-Pudlak syndrome 7, 614076
DYNC2H1	100%	100%	100%	100%	99.8%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2I2	100%	100%	100%	100%	99.4%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633

EFEMP1	84.9%	84.9%	100%	100%	99.8%	Doyne honeycomb degeneration of retina, 126600;Cutis laxa, autosomal recessive, type ID, 620780;Glaucoma 1, open angle, H, 611276
EGFLAM	100%	100%	100%	100%	99.8%	
ELOVL1	100%	100%	100%	100%	99.9%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
ELOVL4	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia 34, 133190;Stargardt disease 3, 600110;Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457
ELP4	87.8%	87.8%	100%	100%	99.7%	?Aniridia 2, 617141
EMC1	99.6%	98.2%	100%	100%	99.6%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EPG5	100%	100%	100%	100%	99.8%	Vici syndrome, 242840;Neurodevelopmental disorder with parkinsonism or other movement abnormalities, 621506
EPHA2	100%	100%	100%	100%	99.5%	Cataract 6, multiple types, 116600
ERCC2	96.3%	94.6%	100%	100%	99.3%	Xeroderma pigmentosum, group D, 278730;Trichothiodystrophy 1, photosensitive, 601675;?Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	100%	100%	100%	100%	99.7%	Trichothiodystrophy 2, photosensitive, 616390;Xeroderma pigmentosum, group B, 610651

ERCC6	100%	100%	100%	100%	99.6%	UV-sensitive syndrome 1, 600630;Cerebrooculofacioskeletal syndrome 1, 214150;?De Sanctis-Cacchione syndrome, 278800;Cockayne syndrome, type B, 133540;{Macular degeneration, age-related, susceptibility to, 5}, 613761;Premature ovarian failure 11, 616946;{Lung cancer, susceptibility to}, 211980
ERCC8	95.2%	95.2%	100%	99.9%	98.8%	UV-sensitive syndrome 2, 614621;Cockayne syndrome, type A, 216400
EXOSC2	91.1%	91%	100%	100%	99.7%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EYA1	100%	100%	100%	100%	100%	Branchiootic syndrome 1, 602588;Branchiootorenal syndrome 1, with or without cataracts, 113650;Anterior segment anomalies with or without cataract, 602588;?Otofaciocervical syndrome, 166780
EYS	100%	100%	100%	100%	99.9%	Retinitis pigmentosa 25, 602772
FA2H	100%	100%	100%	100%	99.7%	Spastic paraplegia 35, autosomal recessive, 612319
FAM161A	100%	100%	100%	100%	99.5%	Retinitis pigmentosa 28, 606068

FBN1	100%	100%	100%	100%	99.8%	Geleophysic dysplasia 2, 614185;Weill-Marchesani syndrome 2, dominant, 608328;Ectopia lentis, familial, 129600;MASS syndrome, 604308;Marfan lipodystrophy syndrome, 616914;Acromicric dysplasia, 102370;Marfan syndrome, 154700;Stiff skin syndrome, 184900
FBN2	99.2%	99.2%	100%	100%	99.6%	Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050
FBXW11	100%	100%	100%	100%	99.4%	Neurodevelopmental, jaw, eye, and digital syndrome, 618914
FDXR	100%	100%	100%	100%	99.4%	Multiple mitochondrial dysfunctions syndrome 9B, 620887;Auditory neuropathy and optic atrophy, 617717
FLVCR1	95%	94.6%	100%	100%	99.7%	Retinopathy-sensory neuropathy syndrome, 609033;Neurodevelopmental disorder with microcephaly, absent speech, and hypotonia, 621060
FOXC1	100%	100%	100%	99.4%	94.5%	Axenfeld-Rieger syndrome, type 3, 602482;Anterior segment dysgenesis 3, multiple subtypes, 601631

FOXE3	100%	100%	100%	99.8%	97.4%	Anterior segment dysgenesis 2, multiple subtypes, 610256;{Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349;Cataract 34, multiple types, 612968
FREM1	100%	100%	100%	100%	99.6%	Manitoba oculotrichoanal syndrome, 248450;Bifid nose with or without anorectal and renal anomalies, 608980;Trigonocephaly 2, 614485
FRMD7	100%	100%	99.5%	93.2%	75.5%	Nystagmus, infantile periodic alternating, X-linked, 310700;Nystagmus 1, congenital, X-linked, 310700
FSD1L	100%	100%	100%	99.9%	99.6%	
FTL	100%	100%	100%	100%	99.3%	Hyperferritinemia-cataract syndrome, 600886;L-ferritin deficiency, dominant and recessive, 615604;Neurodegeneration with brain iron accumulation 3, 606159
FYCO1	100%	100%	100%	100%	99.5%	Cataract 18, autosomal recessive, 610019
FZD4	100%	100%	100%	99.8%	99%	Retinopathy of prematurity, 133780;Exudative vitreoretinopathy 1, 133780
FZD5	100%	100%	100%	100%	98.9%	Microphthalmia/coloboma 11, 620731
GALK1	92.3%	92.3%	100%	100%	99.8%	Galactokinase deficiency with cataracts, 230200

GALM	84%	84%	100%	100%	99.7%	Galactosemia IV, 618881
GALT	100%	100%	100%	100%	99.5%	Galactosemia, 230400
GCNT2	100%	100%	100%	100%	99.6%	[Blood group, Ii], 110800;Adult i phenotype without cataract, 110800;Cataract 13 with adult i phenotype, 116700
GDF3	100%	100%	100%	100%	99.6%	Klippel-Feil syndrome 3, autosomal dominant, 613702;Microphthalmia, isolated, with coloboma 6, 613703;Microphthalmia, isolated 7, 613704
GDF6	100%	100%	100%	99.9%	98.2%	Microphthalmia with coloboma 6, digenic, 613703;Microphthalmia, isolated 4, 613094;Leber congenital amaurosis 17, 615360;Multiple synostoses syndrome 4, 617898;Klippel-Feil syndrome 1, autosomal dominant, 118100
GDPD1	100%	100%	100%	100%	99.8%	
GFER	100%	100%	100%	99.9%	99.4%	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076

GJA1	100%	100%	100%	100%	99.7%	Erythrokeratodermia variabilis et progressiva 3, 617525;Craniometaphyseal dysplasia, autosomal recessive, 218400;Oculodentodigital dysplasia, 164200;Palmoplantar keratoderma with congenital alopecia, 104100;Syndactyly, type III, 186100;Oculodentodigital dysplasia, autosomal recessive, 257850
GJA3	100%	100%	100%	99.9%	98.8%	Cataract 14, multiple types, 601885
GJA8	100%	100%	100%	100%	99.6%	Cataract 1, multiple types, 116200
GLIS3-AS1						
GNAT1	100%	100%	100%	100%	98.9%	Night blindness, congenital stationary, autosomal dominant 3, 610444;Night blindness, congenital stationary, type 1G, 616389
GNAT2	100%	100%	100%	100%	99.8%	Achromatopsia 4, 613856
GNB3	100%	100%	100%	100%	99.1%	Night blindness, congenital stationary, type 1H, 617024;{Hypertension, essential, susceptibility to}, 145500
GNPTG	90.1%	90.1%	100%	99.8%	98.5%	Mucolipidosis III gamma, 252605
GPATCH11	100%	100%	100%	100%	99.8%	
GPR143	93.1%	93.1%	98.6%	89.3%	71.2%	Ocular albinism, type I, Nettleship-Falls type, 300500;Nystagmus 6, congenital, X-linked, 300814

GPR179	100%	100%	100%	100%	99.5%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GRHL2	100%	100%	100%	100%	99.8%	Deafness, autosomal dominant 28, 608641;Ectodermal dysplasia/short stature syndrome, 616029;Corneal dystrophy, posterior polymorphous, 4, 618031
GRK1	100%	100%	100%	100%	98.7%	Oguchi disease-2, 613411
GRM6	100%	100%	100%	99.9%	99.2%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GSN	92.1%	90%	100%	99.8%	98.9%	Amyloidosis, Finnish type, 105120
GUCA1A	100%	100%	100%	100%	98.7%	Cone-rod dystrophy 14, 602093;Cone dystrophy-3, 602093
GUCA1B	100%	100%	100%	99.8%	98.5%	Retinitis pigmentosa 48, 613827
GUCY2D	100%	100%	100%	100%	99.5%	Cone-rod dystrophy 6, 601777;?Choroidal dystrophy, central areolar 1, 215500;Leber congenital amaurosis 1, 204000;Night blindness, congenital stationary, type 1I, 618555
GZF1	99.3%	97.6%	100%	100%	99.7%	Joint laxity, short stature, and myopia, 617662
HARS1	93.2%	93.2%	100%	99.9%	99.9%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625;Usher syndrome type 3B, 614504

HCCS	100%	100%	99.3%	91.9%	75.5%	Linear skin defects with multiple congenital anomalies 1, 309801
HESX1	100%	100%	100%	100%	99.7%	Pituitary hormone deficiency, combined, 5, 182230;Septooptic dysplasia, 182230;Growth hormone deficiency with pituitary anomalies, 182230
HGSNAT	83.5%	83.5%	100%	100%	99.8%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930;Retinitis pigmentosa 73, 616544
HK1	98.2%	96.5%	100%	99.9%	99.5%	Anemia, congenital, nonspherocytic hemolytic, 5, hexokinase deficient, 235700;Retinitis pigmentosa 79, 617460;Neuropathy, hereditary motor and sensory, Russe type, 605285;Neurodevelop mental disorder with visual defects and brain anomalies, 618547
HKDC1	100%	100%	100%	99.9%	99.3%	Retinitis pigmentosa 92, 619614
HMGB3	100%	100%	99.1%	89.3%	70.4%	?Microphthalmia, syndromic 13, 300915
HMX1	100%	100%	99.9%	99.5%	97.7%	Oculoauricular syndrome, 612109
HPS1	100%	100%	100%	100%	99.7%	Hermansky-Pudlak syndrome 1, 203300
HPS3	100%	100%	100%	100%	99.8%	Hermansky-Pudlak syndrome 3, 614072
HPS4	100%	100%	100%	100%	99.8%	Hermansky-Pudlak syndrome 4, 614073
HPS5	100%	100%	100%	100%	99.8%	Hermansky-Pudlak syndrome 5, 614074
HPS6	100%	100%	100%	100%	99.4%	Hermansky-Pudlak syndrome 6, 614075

HRAS	100%	100%	100%	99.9%	99.5%	Bladder cancer, somatic, 109800;Thyroid carcinoma, follicular, somatic, 188470;Congenital myopathy with excess of muscle spindles, 218040;Nevus sebaceous or woolly hair nevus, somatic, 162900;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Spitz nevus or nevus spilus, somatic, 137550;Costello syndrome, 218040
HSF4	100%	100%	100%	99.9%	99.2%	Cataract 5, multiple types, 116800
HSPG2	100%	100%	100%	100%	99.4%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410;Schwartz-Jampel syndrome, type 1, 255800
HYCC1	100%	100%	100%	100%	100%	Leukodystrophy, hypomyelinating, 5, 610532
IBA57	100%	100%	100%	100%	99.5%	Multiple mitochondrial dysfunctions syndrome 3, 615330;?Spastic paraplegia 74, autosomal recessive, 616451
IDH3A	100%	100%	100%	100%	99.9%	Retinitis pigmentosa 90, 619007
IDH3B	100%	100%	100%	100%	99%	Retinitis pigmentosa 46, 612572
IDH3G	100%	100%	98.6%	88%	69.7%	Retinitis pigmentosa 99, 301148

IDUA	96%	96%	100%	99.8%	98.7%	Mucopolysaccharidosis Is, 607016;Mucopolysaccharidosis Ih/s, 607015;Mucopolysaccharidosis Ih, 607014
IFIH1	100%	100%	100%	100%	99.8%	Immunodeficiency 95, 619773;Aicardi-Goutieres syndrome 7, 615846;Singleton-Merten syndrome 1, 182250
IFT140	100%	100%	100%	100%	99.6%	{Polycystic kidney disease 9, susceptibility to}, 621164;Short-rib thoracic dysplasia 9 with or without polydactyly, 266920;Retinitis pigmentosa 80, 617781;Cranioectodermal dysplasia 5, 621180
IFT172	100%	100%	100%	100%	99.7%	Retinitis pigmentosa 71, 616394;Bardet-Biedl syndrome 20, 619471;Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	99.8%	96.5%	100%	100%	99.9%	Bardet-Biedl syndrome 19, 615996
IFT43	93.5%	93.5%	100%	100%	99.7%	?Cranioectodermal dysplasia 3, 614099;?Retinitis pigmentosa 81, 617871;Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT52	92.9%	92.9%	100%	100%	99.8%	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT57	92.4%	92.4%	100%	100%	99.9%	?Orofaciodigital syndrome XVIII, 617927

IFT74	100%	100%	100%	100%	100%	Bardet-Biedl syndrome 22, 617119;Spermatogenic failure 58, 619585;Joubert syndrome 40, 619582
IFT81	92.5%	92.5%	100%	100%	99.7%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGBP1	100%	100%	98.8%	91.9%	72.1%	?Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia, 300472
IGSF3	100%	100%	100%	100%	99.6%	?Lacrimal duct defect, 149700
IKBKG	94.1%	93.5%	99%	90.5%	73.6%	Incontinentia pigmenti, 308300;Ectodermal dysplasia and immunodeficiency 1, 300291;Immunodeficiency 33, 300636;Autoinflammatory disease, systemic, X-linked, 301081
IMPDH1	100%	100%	100%	100%	99.5%	Retinitis pigmentosa 10, 180105;Leber congenital amaurosis 11, 613837
IMPG1	100%	100%	100%	100%	99.7%	Macular dystrophy, vitelliform, 4, 616151;Retinitis pigmentosa 91, 153870
IMPG2	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 56, 613581;Macular dystrophy, vitelliform, 5, 616152
INPP5E	100%	100%	100%	99.8%	98.1%	Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome, 610156;Joubert syndrome 1, 213300

INTS11	100%	100%	100%	99.9%	98.6%	Neurodevelopmental disorder with motor and language delay, ocular defects, and brain abnormalities, 620428
INVS	100%	100%	100%	100%	99.9%	Nephronophthisis 2, infantile, 602088
IQCB1	95.8%	95.8%	100%	100%	99.7%	Senior-Loken syndrome 5, 609254
IRX1	100%	100%	100%	99.9%	98%	
ITM2B	91.5%	91.5%	100%	100%	99.8%	?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079;Dementia, familial British, 176500;Dementia, familial Danish, 117300
ITPR1	100%	100%	100%	100%	99.6%	Gillespie syndrome, 206700;Spinocerebellar ataxia 29, congenital nonprogressive, 117360;Spinocerebellar ataxia 15, 606658
JAG1	100%	100%	100%	100%	99.6%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992;Charcot-Marie-Tooth disease, axonal, type 2HH, 619574;Alagille syndrome 1, 118450;Tetralogy of Fallot, 187500
JAM3	98.1%	93.6%	100%	100%	99.5%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KCNJ13	100%	100%	100%	100%	99.5%	Snowflake vitreoretinal degeneration, 193230;Leber congenital amaurosis 16, 614186

KCNV2	100%	100%	100%	100%	99.7%	Cone dystrophy with supernormal rod responses, 610356
KERA	100%	100%	100%	99.7%	98.6%	Cornea plana 2, autosomal recessive, 217300
KIAA0586	95.6%	95.6%	100%	100%	99.7%	Short-rib thoracic dysplasia 14 with polydactyly, 616546;Joubert syndrome 23, 616490
KIAA0753	100%	100%	100%	100%	99.8%	?Orofaciodigital syndrome XV, 617127;?Joubert syndrome 38, 619476;Short-rib thoracic dysplasia 21 without polydactyly, 619479
KIAA1549	99.5%	99.3%	100%	100%	99.3%	Retinitis pigmentosa 86, 618613
KIF11	100%	100%	100%	100%	99.8%	Microcephaly with or without chorioretinopathy, lymphedema, or impaired intellectual development, 152950
KIF21A	100%	100%	100%	100%	99.6%	Fibrosis of extraocular muscles, congenital, 3B, 135700;Fibrosis of extraocular muscles, congenital, 1, 135700
KIF3B	100%	100%	100%	100%	99.6%	Retinitis pigmentosa 89, 618955
KIF7	100%	100%	100%	100%	99.2%	Joubert syndrome 12, 200990;Acrocallosal syndrome, 200990;?Hydrolethalus syndrome 2, 614120;?Al-Gazali-Bakalinova syndrome, 607131
KIZ	100%	100%	100%	100%	99.7%	Retinitis pigmentosa 69, 615780

KLHL7	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 42, 612943;PERCHING syndrome, 617055
KRT12	100%	100%	100%	100%	99.7%	Meesmann corneal dystrophy 1, 122100
KRT3	100%	100%	100%	100%	99.5%	Meesmann corneal dystrophy 2, 618767
LAMA1	98.9%	98.9%	100%	100%	99.6%	Poretti-Boltshauser syndrome, 615960
LAMB2	100%	100%	100%	100%	99.7%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199;Pierson syndrome, 609049
LAMP2	70.7%	70.7%	98.9%	91%	71.6%	Danon disease, 300257
LCA5	100%	100%	100%	100%	99.5%	Leber congenital amaurosis 5, 604537
LCAT	100%	100%	100%	99.9%	99%	Fish-eye disease, 136120;Norum disease, 245900
LEMD2	100%	100%	100%	100%	99.8%	Marbach-Rustad progeroid syndrome, 619322;Cataract 46, juvenile-onset, 212500
LIM2	100%	100%	100%	100%	98.6%	Cataract 19, multiple types, 615277
LMX1B	100%	100%	100%	100%	99%	Focal segmental glomerulosclerosis 10, 256020;Nail-patella syndrome, 161200
LONP1	99.3%	97%	100%	100%	99.5%	CODAS syndrome, 600373
LOXL1	100%	100%	100%	99.8%	98.5%	{Exfoliation syndrome, susceptibility to}, 177650
LOXL3	100%	100%	100%	100%	99.3%	Myopia 28, autosomal recessive, 619781

LRAT	100%	100%	100%	100%	99.4%	Leber congenital amaurosis 14, 613341;Retinal dystrophy, early-onset severe, 613341;Retinitis pigmentosa, juvenile, 613341
LRIT3	100%	100%	100%	100%	99.9%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRMDA	97.8%	97.8%	100%	100%	99.4%	Albinism, oculocutaneous, type VII, 615179
LRP2	100%	100%	100%	100%	99.8%	Donnai-Barrow syndrome, 222448
LRP5	100%	100%	100%	100%	99%	Osteopetrosis, autosomal dominant 1, 607634;Polycystic liver disease 4 with or without kidney cysts, 617875;Endosteal hyperostosis, 144750;Osteoporosis-pseudoglioma syndrome, 259770;[Bone mineral density variability 1, high bone mass], 601884;Exudative vitreoretinopathy 4, 601813
LRPAP1	84.8%	84.8%	100%	100%	99.4%	Myopia 23, autosomal recessive, 615431
LSS	100%	100%	100%	100%	99.8%	Hypotrichosis 14, 618275;Cataract 44, 616509;Alopecia-intellectual disability syndrome 4, 618840

LTBP2	100%	100%	100%	100%	99.7%	Glaucoma 3, primary congenital, D, 613086;Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750;?Weill-Marchesani syndrome 3, recessive, 614819
LYST	99.5%	99.5%	100%	100%	99.9%	Chediak-Higashi syndrome, 214500
LZTFL1	100%	100%	100%	100%	99.9%	Bardet-Biedl syndrome 17, 615994
MAB21L2	100%	100%	100%	100%	99.2%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MAF	95.1%	91.7%	100%	99.4%	96.8%	Cataract 21, multiple types, 610202;Ayme-Gripp syndrome, 601088
MAFB	100%	100%	100%	99.9%	98.8%	Duane retraction syndrome 3, 617041;Multicentric carpotarsal osteolysis syndrome, 166300
MAK	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 62, 614181
MAPKAPK3	92.7%	89.5%	100%	100%	99.7%	?Macular dystrophy, patterned, 3, 617111
MCAT	95.9%	95.9%	100%	100%	99.6%	Optic atrophy 15, 620583
MCOLN1	98.3%	95.7%	100%	100%	99.7%	Lisch epithelial corneal dystrophy, 620763;Mucolipidosis IV, 252650
MDM1	100%	100%	100%	100%	99.6%	

MECR	93.5%	93.5%	100%	100%	99.6%	?Optic atrophy 16, 620629;Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MERTK	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 38, 613862
MFN2	100%	100%	100%	100%	99.7%	Lipomatosis, multiple symmetric, with or without peripheral neuropathy, 151800;Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260;Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087;Hereditary motor and sensory neuropathy VIA, 601152
MFRP	100%	100%	100%	100%	99.4%	Microphthalmia, isolated 5, 611040;Nanophthalmos 2, 609549
MFSD8	100%	100%	100%	100%	100%	Macular dystrophy with central cone involvement, 616170;Ceroid lipofuscinosis, neuronal, 7, 610951
MIEF1	100%	100%	100%	100%	99.3%	Optic atrophy 14, 620550
MIP	100%	100%	100%	100%	99.7%	Cataract 15, multiple types, 615274
MIR184						EDICT syndrome, 614303
MIR204						Retinal dystrophy and iris coloboma with or without cataract, 616722

MITF	94.7%	94.7%	100%	100%	99.9%	Waardenburg syndrome, type 2A, 193510;{Melanoma, cutaneous malignant, susceptibility to, 8}, 614456;Tietz albinism-deafness syndrome, 103500;COMMAD syndrome, 617306
MKKS	100%	100%	100%	100%	100%	McKusick-Kaufman syndrome, 236700;Bardet-Biedl syndrome 6, 605231
MKS1	99.1%	99%	100%	100%	99.7%	Bardet-Biedl syndrome 13, 615990;Meckel syndrome 1, 249000;Joubert syndrome 28, 617121
MPDZ	100%	100%	100%	100%	99.7%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MTFMT	91.8%	91.8%	100%	99.9%	98.6%	Combined oxidative phosphorylation deficiency 15, 614947;Mitochondrial complex I deficiency, nuclear type 27, 618248
MTRFR	100%	100%	100%	100%	99.7%	Spastic paraplegia 55, autosomal recessive, 615035;Combined oxidative phosphorylation deficiency 7, 613559
MVK	100%	100%	100%	100%	99.8%	Hyper-IgD syndrome, 260920;Porokeratosis 3, multiple types, 175900;Mevalonic aciduria, 610377
MYCBP2	100%	99.8%	100%	100%	99.8%	

MYH9	97.3%	97%	100%	100%	99.4%	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100;Deafness, autosomal dominant 17, 603622
MYO5A	99%	99%	100%	100%	99.7%	Griscelli syndrome, type 1, 214450
MYO7A	100%	100%	100%	100%	99.5%	Deafness, autosomal recessive 2, 600060;Usher syndrome, type 1B, 276900;Deafness, autosomal dominant 11, 601317
MYOC	95.3%	95.2%	100%	100%	99.6%	Glaucoma 1A, primary open angle, 137750
MYRF	100%	99.8%	100%	99.9%	99.2%	Nanophthalmos 1, 600165;Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113;Cardiac-urogenital syndrome, 618280
NAA10	100%	100%	99%	89%	68.4%	Microphthalmia, syndromic 1, 309800;Ogden syndrome, 300855
NADK2	100%	100%	100%	99.9%	99.2%	2,4-dienoyl-CoA reductase deficiency, 616034
NBAS	100%	100%	100%	100%	99.7%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800;Infantile liver failure syndrome 2, 616483
NDP	100%	100%	98.2%	90.3%	76.4%	Exudative vitreoretinopathy 2, X-linked, 305390;Norrie disease, 310600
NDUFA10	82.9%	80.5%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 22, 618243

NDUFA12	80.4%	80.4%	100%	99.9%	99.7%	Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA7	78.2%	78.2%	100%	100%	98.6%	
NDUFAF2	67.4%	67.4%	100%	100%	99.6%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF4	100%	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	90.9%	90.9%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF8	100%	100%	100%	100%	99.4%	Mitochondrial complex I deficiency, nuclear type 34, 618776
NDUFB11	99.9%	99.3%	93.6%	79.3%	59.7%	Linear skin defects with multiple congenital anomalies 3, 300952;?Mitochondrial complex I deficiency, nuclear type 30, 301021
NDUFS1	100%	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	99.9%	98.3%	100%	100%	99.6%	?Leber-like hereditary optic neuropathy, autosomal recessive 2, 620569;Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS7	89.1%	85%	100%	100%	99.4%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFV1	100%	100%	100%	100%	98.7%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	100%	100%	100%	100%	99.5%	Mitochondrial complex I deficiency, nuclear type 7, 618229

NEK1	100%	100%	100%	100%	99.8%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520;?Orofaciodigital syndrome II, 252100;{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
NEK2	95.9%	95.9%	100%	100%	100%	?Retinitis pigmentosa 67, 615565
NEUROD1	100%	100%	100%	100%	99%	{Type 2 diabetes mellitus, susceptibility to}, 125853;Maturity-onset diabetes of the young 6, 606394
NHEJ1	92.9%	92.9%	100%	100%	99.7%	Microphthalmia/coloboma 13, 620968;Immunodeficiency 124, severe combined, 611291
NHS	100%	100%	98.9%	90.1%	71.8%	Cataract 40, X-linked, 302200;Nance-Horan syndrome, 302350
NMNAT1	100%	99.5%	100%	100%	99.4%	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260;Leber congenital amaurosis 9, 608553
NOP10	92.5%	92.5%	100%	100%	99.9%	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 9, 620400;?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 2, 620425;?Dyskeratosis congenita, autosomal recessive 1, 224230

NPHP1	100%	100%	100%	100%	99.8%	Joubert syndrome 4, 609583;Nephronophthisis 1, juvenile, 256100;Senior-Loken syndrome-1, 266900
NPHP3	100%	100%	100%	100%	99.6%	Nephronophthisis 3, 604387;Renal-hepatic-pancreatic dysplasia 1, 208540;Meckel syndrome 7, 267010
NPHP4	100%	100%	100%	100%	99.5%	Senior-Loken syndrome 4, 606996;Nephronophthisis 4, 606966
NR2E3	100%	100%	100%	100%	99.5%	Retinitis pigmentosa 37, 611131;Enhanced S-cone syndrome 1, 268100
NR2F1	100%	100%	100%	99.9%	98.5%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR6A1	96.3%	96.3%	100%	100%	99.4%	Oculovertebral syndrome, 621277
NRL	100%	100%	100%	100%	99.4%	Enhanced S-cone syndrome 2, 621371;Retinitis pigmentosa 27, 613750
NSUN3	100%	100%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 48, 619012
NTF4	100%	100%	100%	99.9%	98.8%	Glaucoma 1, open angle, 1O, 613100
NYX	100%	100%	98.3%	86.8%	66.3%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	100%	100%	100%	99.9%	99.3%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870

OCA2	93.4%	93.4%	100%	100%	99.7%	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220;[Skin/hair/eye pigmentation 1, blond/brown hair], 227220;Albinism, brown oculocutaneous, 203200;Albinism, oculocutaneous, type II, 203200
OCRL	100%	100%	99.3%	92.4%	74.8%	Dent disease 2, 300555;Lowe syndrome, 309000
OFD1	100%	100%	99.1%	91%	73.2%	Simpson-Golabi-Behmel syndrome, type 2, 300209;?Retinitis pigmentosa 23, 300424;Orofaciodigital syndrome I, 311200;Joubert syndrome 10, 300804
OPA1	94.7%	94.7%	100%	100%	99.7%	Optic atrophy plus syndrome, 125250;{Glaucoma, normal tension, susceptibility to}, 606657;Optic atrophy 1, 165500;Behr syndrome, 210000;Mitochondrial DNA depletion syndrome 14A (encephalomyopathic type), 621481;?Mitochondrial DNA depletion syndrome 14B (cardioencephalomyopathic type), 616896
OPA3	100%	100%	100%	100%	99.6%	3-methylglutaconic aciduria, type III, 258501;Optic atrophy 3 with cataract, 165300
OPN1LW	99.5%	99.1%	94.3%	83.2%	66%	Blue cone monochromacy, 303700;Colorblindness, protan, 303900

OPN1MW	99.1%	97.5%	83.4%	70.4%	55.4%	Colorblindness, deutan, 303800;Blue cone monochromacy, 303700
OTX2	100%	100%	100%	100%	99.9%	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125;Pituitary hormone deficiency, combined, 6, 613986;Microphthalmia, syndromic 5, 610125
OVOL2	100%	100%	100%	99.9%	99.2%	Corneal dystrophy, posterior polymorphous, 1, 122000
P3H1	95.3%	93.2%	100%	99.9%	99.1%	Osteogenesis imperfecta, type VIII, 610915
P3H2	100%	100%	100%	99.8%	99.2%	Myopia, high, with cataract and vitreoretinal degeneration, 614292
P4HA2	100%	100%	100%	100%	99.6%	Myopia 25, autosomal dominant, 617238
PAK2	100%	100%	100%	100%	99.8%	?Knobloch syndrome 2, 618458
PANK2	100%	100%	100%	100%	99.6%	Neurodegeneration with brain iron accumulation 1, 234200
PANK4	100%	100%	100%	100%	99.1%	?Cataract 49, 619593
PAX2	100%	100%	100%	100%	99.2%	Glomerulosclerosis, focal segmental, 7, 616002;Papillorenal syndrome, 120330

PAX6	100%	100%	100%	100%	99.5%	Optic nerve hypoplasia, 165550;Cataract with late-onset corneal dystrophy, 106210;Microphthalmia /coloboma 12, 120200;?Coloboma of optic nerve, 120430;Aniridia, 106210;Anterior segment dysgenesis 5, multiple subtypes, 604229;?Morning glory disc anomaly, 120430;Foveal hypoplasia 1, 136520;Keratitis, 148190
PCARE	100%	100%	100%	100%	99.6%	Retinitis pigmentosa 54, 613428
PCDH15	100%	100%	100%	100%	99.6%	Usher syndrome, type 1D/F digenic, 601067;Deafness, autosomal recessive 23, 609533;Usher syndrome, type 1F, 602083
PCYT1A	100%	100%	100%	100%	99.7%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940;Lipodystrophy, congenital generalized, type 5, 620680
PDE6A	100%	100%	100%	100%	99.6%	Retinitis pigmentosa 43, 613810
PDE6B	100%	100%	100%	100%	99.2%	Retinitis pigmentosa-40, 613801;Night blindness, congenital stationary, autosomal dominant 2, 163500
PDE6C	100%	100%	100%	100%	99.9%	Cone dystrophy 4, 613093
PDE6D	100%	100%	100%	100%	99.1%	Joubert syndrome 22, 615665
PDE6G	100%	100%	100%	100%	99.2%	Retinitis pigmentosa 57, 613582

PDE6H	100%	100%	100%	100%	99.9%	Achromatopsia 6, 610024
PDGFRA	100%	100%	100%	100%	99.6%	Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial, 175510;Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685
PDSS1	100%	100%	100%	100%	99.7%	Coenzyme Q10 deficiency, primary, 2, 614651
PDZD7	100%	99.8%	100%	100%	99.3%	Deafness, autosomal recessive 57, 618003;{Retinal disease in Usher syndrome type IIA, modifier of}, 276901;Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472
PET100	100%	100%	100%	100%	99.2%	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PEX1	100%	100%	100%	100%	99.9%	Heimler syndrome 1, 234580;Peroxisome biogenesis disorder 1B (NALD/IRD), 601539;Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX11B	100%	100%	100%	99.9%	99.1%	Peroxisome biogenesis disorder 14B, 614920
PEX2	100%	100%	100%	100%	99.7%	Peroxisome biogenesis disorder 5A (Zellweger), 614866;Peroxisome biogenesis disorder 5B, 614867
PEX26	91.9%	91.7%	100%	99.9%	99.1%	Peroxisome biogenesis disorder 7B, 614873;Peroxisome biogenesis disorder 7A (Zellweger), 614872

PEX6	100%	100%	100%	100%	99.1%	Peroxisome biogenesis disorder 4B, 614863;Peroxisome biogenesis disorder 4A (Zellweger), 614862;Heimler syndrome 2, 616617
PEX7	97.9%	97.9%	100%	100%	99.6%	Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879
PGK1	92.7%	92.7%	99.3%	90.9%	71.5%	Phosphoglycerate kinase 1 deficiency, 300653
PHOX2A	100%	100%	100%	99.9%	99.2%	Fibrosis of extraocular muscles, congenital, 2, 602078
PHYH	100%	100%	100%	99.8%	99.1%	Refsum disease, 266500
PIK3R1	100%	100%	100%	100%	99.7%	Immunodeficiency 36, 616005;SHORT syndrome, 269880;Agammaglobulinemia 7, autosomal recessive, 615214
PIKFYVE	100%	100%	100%	100%	99.8%	Corneal fleck dystrophy, 121850
PITX2	100%	100%	100%	100%	99.1%	Ring dermoid of cornea, 180550;Axenfeld-Rieger syndrome, type 1, 180500;Anterior segment dysgenesis 4, 137600
PITX3	100%	100%	100%	99.9%	99.3%	Cataract 11, multiple types, 610623;Anterior segment dysgenesis 1, multiple subtypes, 107250;Cataract 11, syndromic, autosomal recessive, 610623
PLA2G5	100%	100%	100%	100%	99.3%	[Fleck retina, familial benign], 228980

PLK4	100%	100%	100%	100%	99.9%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLOD3	98.5%	96.8%	100%	99.8%	98.9%	BCARD syndrome (lysyl hydroxylase 3 deficiency), 612394
PMPCA	96.2%	96.2%	100%	100%	99.1%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PNPLA6	100%	100%	100%	100%	99.3%	Spastic paraplegia 39, autosomal recessive, 612020;Oliver-McFarlane syndrome, 275400;?Laurence-Moon syndrome, 245800;Boucher-Neuhauser syndrome, 215470
POC1B	100%	100%	100%	100%	99.7%	Cone-rod dystrophy 20, 615973
POC5	93.4%	93.4%	100%	100%	99.9%	
POLG	100%	100%	100%	100%	99.8%	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459;Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662;Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700;Progressive external ophthalmoplegia, autosomal dominant 1, 157640;Progressive external ophthalmoplegia, autosomal recessive 1, 258450

POLG2	95.8%	95.8%	100%	100%	99.5%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131;?Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528;?Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), 619425
POMGNT1	100%	100%	100%	100%	99.6%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3, 613151;Retinitis pigmentosa 76, 617123;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280
PPIB	100%	100%	100%	100%	99.6%	Osteogenesis imperfecta, type IX, 259440
PPT1	90.3%	90.3%	100%	100%	99.7%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRCD	100%	100%	100%	99.8%	99.7%	Retinitis pigmentosa 36, 610599
PRDM13	100%	100%	100%	99.9%	98.7%	Pontocerebellar hypoplasia, type 17, 619909;Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761
PRDM5	100%	100%	100%	100%	99.8%	Brittle cornea syndrome 2, 614170

PRDX3	100%	98.8%	100%	100%	99.7%	Spinocerebellar ataxia, autosomal recessive 32, 619862;Corneal dystrophy, punctiform and polychromatic pre-Descemet, 619871
PRIMPOL	93.5%	93.5%	100%	100%	99.7%	Myopia 22, autosomal dominant, 615420
PRKCZ	100%	100%	100%	100%	99.1%	
PROM1	100%	100%	100%	100%	99.7%	Macular dystrophy, retinal, 2, 608051;Retinitis pigmentosa 41, 612095;Stargardt disease 4, 603786;Cone-rod dystrophy 12, 612657
PRPF3	100%	100%	100%	100%	99.7%	Retinitis pigmentosa 18, 601414
PRPF31	93.1%	90.1%	100%	100%	99.4%	Retinitis pigmentosa 11, 600138
PRPF4	90.5%	87.4%	100%	100%	99.6%	Retinitis pigmentosa 70, 615922
PRPF6	96.2%	96.2%	100%	100%	99.4%	Retinitis pigmentosa 60, 613983
PRPF8	100%	100%	100%	100%	99.3%	Retinitis pigmentosa 13, 600059
PRPH2	100%	100%	100%	100%	99.4%	Macular dystrophy, patterned, 1, 169150;Choroidal dystrophy, central areolar 2, 613105;Retinitis punctata albescens, 136880;Leber congenital amaurosis 18, 608133;Macular dystrophy, vitelliform, 3, 608161;Retinitis pigmentosa 7 and digenic form, 608133

PRPS1	100%	100%	99%	89.8%	71.8%	Arts syndrome, 301835;Phosphoribosyl pyrophosphate synthetase superactivity, 300661;Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070;Deafness, X-linked 1, 304500;Gout, PRPS-related, 300661
PRR11	100%	100%	100%	99.9%	99.3%	
PRR12	100%	100%	100%	99.9%	98.7%	Neuroocular syndrome, 619539
PRSS56	100%	100%	100%	100%	99.5%	Microphthalmia, isolated 6, 613517
PTCHD1	100%	100%	98.9%	89.1%	69.9%	{Autism, susceptibility to, X-linked 4}, 300830
PXDN	100%	99.6%	100%	99.9%	99.3%	Anterior segment dysgenesis 7, with sclerocornea, 269400
RAB28	100%	100%	100%	100%	99.6%	Cone-rod dystrophy 18, 615374
RAB3GAP2	94.4%	94.4%	100%	100%	99.8%	Martsolf syndrome 1, 212720;Warburg micro syndrome 2, 614225
RARB	98%	98%	100%	100%	99.7%	Microphthalmia, syndromic 12, 615524
RAX	100%	100%	100%	100%	99.1%	Microphthalmia, syndromic 16, 611038
RAX2	100%	100%	100%	100%	99.6%	Retinitis pigmentosa 95, 620102;Cone-rod dystrophy 11, 610381;?Macular degeneration, age-related, 6, 613757
RBP3	100%	100%	100%	100%	99.5%	?Retinitis pigmentosa 66, 615233

RBP4	100%	100%	100%	100%	98.8%	Microphthalmia/coloboma 10, 616428;Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RCBTB1	100%	100%	100%	100%	99.7%	Retinal dystrophy with or without extraocular anomalies, 617175
RD3	100%	100%	100%	100%	99.4%	Leber congenital amaurosis 12, 610612
RDH11	94.9%	94%	100%	100%	99.5%	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	100%	100%	100%	100%	99.6%	Leber congenital amaurosis 13, 612712
RDH5	100%	100%	100%	100%	99.5%	Fundus albipunctatus, 136880
REEP6	100%	100%	100%	100%	99.5%	Retinitis pigmentosa 77, 617304
RGS9	95.9%	95.9%	100%	100%	99.6%	Prolonged electroretinal response suppression 1, 608415
RGS9BP	100%	100%	100%	100%	99.4%	Prolonged electroretinal response suppression 2, 620344
RHO	100%	100%	100%	100%	99.8%	Night blindness, congenital stationary, autosomal dominant 1, 610445;Retinitis pigmentosa 4, autosomal dominant or recessive, 613731;Retinitis punctata albescens, 136880
RIGI	98.6%	98.6%	100%	100%	99.9%	Singleton-Merten syndrome 2, 616298
RIMS2	99.2%	99.2%	100%	100%	99.8%	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970

RLBP1	100%	100%	100%	100%	99.8%	Bothnia retinal dystrophy, 607475;Newfoundland rod-cone dystrophy, 607476;Retinitis punctata albescens, 136880;Fundus albipunctatus, 136880
RNU4-2						Retinitis pigmentosa 102, 621560;ReNU syndrome, 620851
RNU4ATAC						Roifman syndrome, 616651;Lowry-Wood syndrome, 226960;Microcephalic osteodysplastic primordial dwarfism, type I, 210710
RNU6-1						Retinitis pigmentosa 103, 621561
RNU6-2						Retinitis pigmentosa 104, 621562
RNU6-8						Retinitis pigmentosa 105, 621563
RNU6-9						Retinitis pigmentosa 106, 621564
ROM1	100%	100%	100%	100%	99.4%	Retinitis pigmentosa 7, digenic form, 608133
RP1	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 1, 180100
RP1L1	100%	100%	100%	99.9%	99%	Occult macular dystrophy, 613587;Retinitis pigmentosa 88, 618826
RP2	100%	100%	98.2%	89.2%	73.9%	Retinitis pigmentosa 2, 312600
RP9	100%	100%	100%	99.8%	98%	?Retinitis pigmentosa 9, 180104

RPE65	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 20, 613794;Retinitis pigmentosa 87 with choroidal involvement, 618697;Leber congenital amaurosis 2, 204100
RPGR	100%	99.7%	92.5%	79.2%	61.2%	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455;Cone-rod dystrophy, X-linked, 1, 304020;Retinitis pigmentosa 3, 300029;Macular degeneration, X-linked atrophic, 300834
RPGRIP1	100%	100%	100%	99.9%	99.1%	Cone-rod dystrophy 13, 608194;Leber congenital amaurosis 6, 613826
RPGRIP1L	100%	100%	100%	100%	99.7%	Joubert syndrome 7, 611560;Meckel syndrome 5, 611561;?COACH syndrome 3, 619113
RS1	100%	100%	98.6%	87.3%	69.1%	Retinoschisis, 312700
RTN4IP1	100%	100%	100%	100%	99.8%	Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732
SAG	100%	100%	100%	100%	99.6%	Retinitis pigmentosa 47, autosomal recessive, 613758;Retinitis pigmentosa 96, autosomal dominant, 620228;Oguchi disease-1, 258100
SALL2	100%	100%	100%	100%	99.7%	?Coloboma, ocular, autosomal recessive, 216820
SAMD11	100%	100%	100%	100%	98.9%	

SAMD7	100%	100%	100%	100%	99.2%	Macular dystrophy with or without cone dysfunction, 620762
SBF2	93.7%	93.7%	100%	100%	99.7%	Charcot-Marie-Tooth disease, type 4B2, 604563
SC5D	100%	100%	100%	100%	99.9%	Lathosterolosis, 607330
SCAPER	97.9%	97.9%	100%	100%	99.8%	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCLT1	95.2%	95.2%	100%	100%	99.8%	
SCO2	100%	100%	100%	100%	99.8%	Myopia 6, 608908;Mitochondrial complex IV deficiency, nuclear type 2, 604377
SDCCAG8	100%	100%	100%	100%	99.9%	Senior-Loken syndrome 7, 613615;Bardet-Biedl syndrome 16, 615993
SEMA4A	100%	100%	100%	100%	98.9%	Retinitis pigmentosa 35, 610282;Cone-rod dystrophy 10, 610283
SGSH	100%	100%	100%	100%	99.5%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH3PXD2B	99.9%	98.6%	100%	100%	99.2%	Frank-ter Haar syndrome, 249420
SHH	100%	100%	100%	99.8%	98.4%	Single median maxillary central incisor, 147250;Holoprosencephaly 3, 142945;Microphthalmia/coloboma 5, 611638
SIL1	91.5%	91.5%	100%	100%	99.5%	Marinesco-Sjogren syndrome, 248800
SIPA1L3	100%	100%	100%	100%	99.4%	?Cataract 45, 616851
SIX6	100%	100%	100%	100%	99.3%	Optic disc anomalies with retinal and/or macular dystrophy, 212550

SLC16A12	100%	100%	100%	99.9%	99.8%	Cataract 47, juvenile, with microcornea, 612018
SLC19A2	100%	100%	100%	100%	99.7%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC24A1	100%	100%	100%	100%	99.4%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A5	100%	100%	100%	100%	99.7%	[Skin/hair/eye pigmentation 4, fair/dark skin], 113750;Albinism, oculocutaneous, type VI, 113750
SLC25A46	100%	100%	100%	100%	99.7%	Neuropathy, hereditary motor and sensory, type VIB, 616505;Pontocerebellar hypoplasia, type 1E, 619303
SLC33A1	92.6%	92.6%	100%	100%	99.7%	Spastic paraplegia 42, autosomal dominant, 612539;Huppke-Brendel syndrome, 614482
SLC37A3	100%	100%	100%	100%	99.8%	
SLC38A8	100%	100%	100%	100%	99.5%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A12	100%	100%	100%	100%	99.8%	
SLC39A5	100%	100%	100%	100%	99.3%	Myopia 24, autosomal dominant, 615946

SLC45A2	100%	100%	100%	100%	99.6%	[Skin/hair/eye pigmentation 5, dark/light eyes], 227240;[Skin/hair/eye pigmentation 5, black/nonblack hair], 227240;Albinism, oculocutaneous, type IV, 606574;[Skin/hair/eye pigmentation 5, dark/fair skin], 227240
SLC4A11	100%	100%	100%	100%	99.6%	Corneal endothelial dystrophy, autosomal recessive, 217700;Corneal dystrophy, Fuchs endothelial, 4, 613268;Corneal endothelial dystrophy and perceptive deafness, 217400
SLC4A4	95%	95%	100%	100%	99.9%	Proximal renal tubular acidosis-ocular anomaly syndrome, 604278
SLC4A7	100%	100%	100%	100%	99.8%	
SLC52A2	100%	100%	100%	99.9%	99.7%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC66A1	100%	100%	100%	100%	99.6%	
SLC7A14	100%	100%	100%	99.9%	99.6%	Retinitis pigmentosa 68, 615725
SLITRK6	100%	100%	100%	100%	99.7%	Deafness and myopia, 221200
SMG8	100%	100%	100%	100%	99.7%	Alzahrani-Kuwahara syndrome, 619268
SMOC1	96%	96%	100%	100%	99.7%	Microphthalmia with limb anomalies, 206920
SNF8	94.6%	94.6%	100%	100%	99.6%	Developmental and epileptic encephalopathy 115, 620783;Neurodevelopmental disorder plus optic atrophy, 620784

SNRNP200	100%	100%	100%	100%	99.4%	Retinitis pigmentosa 33, 610359
SNX10	89.3%	89.3%	100%	100%	99.6%	Osteopetrosis, autosomal recessive 8, 615085
SOX2	100%	100%	100%	99.9%	97.8%	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900;Microphthalmia, syndromic 3, 206900
SOX5	100%	100%	100%	100%	99.8%	Lamb-Shaffer syndrome, 616803
SPARCL1	100%	100%	100%	100%	99.9%	
SPATA7	100%	100%	100%	100%	99.9%	Leber congenital amaurosis 3, 604232;Retinitis pigmentosa 94, variable age at onset, autosomal recessive, 604232
SPG7	100%	100%	100%	99.9%	99.3%	Spastic paraplegia 7, autosomal recessive, 607259
SPP2	100%	100%	100%	100%	100%	
SSBP1	100%	100%	100%	100%	100%	Optic atrophy 13 with retinal and foveal abnormalities, 165510
STRA6	100%	100%	100%	100%	99.3%	Microphthalmia, syndromic 9, 601186;Microphthalmia, isolated, with coloboma 8, 601186
STS	97.1%	97.1%	98.9%	90.5%	73.5%	Ichthyosis, X-linked, 308100
STX3	100%	100%	100%	100%	99.7%	Retinal dystrophy and microvillus inclusion disease, 619446;Diarrhea 12, with microvillus atrophy, 619445
SUMF1	100%	100%	100%	100%	99.9%	Multiple sulfatase deficiency, 272200

TACSTD2	100%	100%	100%	99.6%	97.6%	Corneal dystrophy, gelatinous drop-like, 204870
TBC1D2B	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with seizures and gingival overgrowth, 619323
TBC1D32	100%	100%	100%	100%	99.8%	Orofaciodigital syndrome IX, 258865;Alsahan-Harris syndrome, 621307;Retinitis pigmentosa 100, 621280
TCF4	97.1%	97.1%	100%	100%	99.6%	Pitt-Hopkins syndrome, 610954;Corneal dystrophy, Fuchs endothelial, 3, 613267
TCTN1	92.6%	90.8%	100%	100%	99.5%	Joubert syndrome 13, 614173
TCTN2	98.5%	98.5%	100%	100%	99.5%	Joubert syndrome 24, 616654;?Meckel syndrome 8, 613885
TCTN3	100%	100%	100%	100%	99.5%	Joubert syndrome 18, 614815;Orofaciodigital syndrome IV, 258860
TDRD7	100%	100%	100%	100%	99.8%	Cataract 36, 613887
TEAD1	100%	100%	100%	99.9%	99.5%	Sveinsson chorioretinal atrophy, 108985
TEK	100%	100%	100%	100%	99.9%	Venous malformations, multiple cutaneous and mucosal, 600195;Glaucoma 3, primary congenital, E, 617272
TENM3	99.6%	99.6%	100%	100%	99.6%	Microphthalmia, syndromic 15, 615145;?Microphthalmia/coloboma 9, 615145
TFAP2A	100%	100%	100%	100%	98.8%	Branchiooculofacial syndrome, 113620
TFPT	100%	100%	100%	100%	99%	

TGFBI	100%	100%	100%	100%	99.7%	Corneal dystrophy, Avellino type, 607541;Corneal dystrophy, Reis-Bucklers type, 608470;Corneal dystrophy, Thiel-Behnke type, 602082;Corneal dystrophy, Groenouw type I, 121900;Corneal dystrophy, epithelial basement membrane, 121820;Corneal dystrophy, lattice type I, 122200;Corneal dystrophy, lattice type IIIA, 608471
THRB	100%	100%	100%	100%	99.9%	Thyroid hormone resistance, autosomal recessive, 274300;Thyroid hormone resistance, 188570;Thyroid hormone resistance, selective pituitary, 145650
TIMM8A	100%	100%	98.2%	87.9%	71.6%	Mohr-Tranebjaerg syndrome, 304700
TIMP3	84%	84%	100%	100%	99.4%	Sorsby fundus dystrophy, 136900
TINF2	100%	100%	100%	100%	99.7%	Dyskeratosis congenita, autosomal dominant 3, 613990;Revesz syndrome, 268130
TLCD3B	100%	100%	100%	99.8%	99%	Cone-rod dystrophy 22, 619531
TMCO3	94.6%	94.6%	100%	99.9%	99.1%	
TMEM126A	100%	100%	100%	100%	100%	Optic atrophy 7, 612989
TMEM138	100%	98.8%	100%	99.9%	99.7%	Joubert syndrome 16, 614465

TMEM216	100%	100%	100%	100%	99.8%	Joubert syndrome 2, 608091;Retinitis pigmentosa 98, 620996;Meckel syndrome 2, 603194
TMEM218	100%	100%	100%	100%	99.6%	Joubert syndrome 39, 619562
TMEM231	93.5%	93.5%	100%	100%	99.5%	Joubert syndrome 20, 614970;Meckel syndrome 11, 615397
TMEM237	98.2%	98.2%	100%	100%	99.7%	Joubert syndrome 14, 614424
TMEM67	96.1%	96.1%	100%	100%	99.7%	Nephronophthisis 11, 613550;{Bardet-Biedl syndrome 14, modifier of}, 615991;Joubert syndrome 6, 610688;Meckel syndrome 3, 607361;?RHYNS syndrome, 602152;COACH syndrome 1, 216360
TMEM98	100%	100%	100%	100%	99.9%	Nanophthalmos 4, 615972
TOGARAM1	100%	100%	100%	100%	99.7%	Joubert syndrome 37, 619185
TOPORS	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 31, 609923
TPP1	100%	100%	100%	100%	99.3%	Ceroid lipofuscinosis, neuronal, 2, 204500;Spinocerebellar ataxia, autosomal recessive 7, 609270
TRAF3IP1	100%	100%	100%	100%	99.6%	

TREX1	100%	100%	100%	100%	99.5%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315;Aicardi-Goutieres syndrome 1, dominant and recessive, 225750;{Systemic lupus erythematosus, susceptibility to}, 152700;Chilblain lupus, 610448
TRIM32	100%	100%	100%	100%	99.6%	?Bardet-Biedl syndrome 11, 615988;Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRNT1	91.9%	91.9%	100%	100%	99.9%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084;Retinitis pigmentosa and erythrocytic microcytosis, 616959
TRPM1	100%	100%	100%	100%	99.8%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM3	97.8%	97.8%	100%	100%	99.6%	?Cataract 50 with or without glaucoma, 620253;Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures, 620224
TSFM	91.5%	91.1%	100%	100%	99.3%	Combined oxidative phosphorylation deficiency 3, 610505
TSPAN12	100%	100%	100%	100%	99.7%	Exudative vitreoretinopathy 5, 613310

TTC8	91.4%	91.4%	100%	100%	99.9%	Bardet-Biedl syndrome 8, 615985;?Retinitis pigmentosa 51, 613464
TTLL5	100%	100%	100%	100%	99.7%	Cone-rod dystrophy 19, 615860
TUB	100%	100%	100%	100%	99.5%	?Retinal dystrophy and obesity, 616188
TUBA1A	100%	100%	100%	100%	99.6%	Lissencephaly 3, 611603
TUBA3D	100%	99.9%	100%	100%	99.6%	Keratoconus 9, 617928
TUBB3	100%	100%	100%	100%	99.5%	Fibrosis of extraocular muscles, congenital, 3A, 600638;Cortical dysplasia, complex, with other brain malformations 1, 614039
TUBB4B	100%	100%	100%	100%	99.3%	Leber congenital amaurosis with early-onset deafness, 617879
TUBGCP4	100%	100%	100%	99.8%	98.7%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	100%	100%	100%	99.9%	99.2%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TULP1	100%	100%	100%	100%	99.5%	Leber congenital amaurosis 15, 613843;Retinitis pigmentosa 14, 600132
TWNK	100%	100%	100%	100%	99.6%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286;Perrault syndrome 5, 616138

TYR	100%	100%	100%	100%	99.7%	[Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800;[Skin/hair/eye pigmentation 3, blue/green eyes], 601800;{Melanoma, cutaneous malignant, susceptibility to, 8}, 601800;Albinism, oculocutaneous, type IB, 606952;Albinism, oculocutaneous, type IA, 203100
TYRP1	100%	100%	100%	100%	99.7%	[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271;Albinism, oculocutaneous, type III, 203290
UBAP1L	100%	100%	100%	100%	99.5%	Retinal dystrophy, Zeitz-Han type, 621558
UBIAD1	100%	100%	100%	100%	99.7%	Corneal dystrophy, Schnyder type, 121800
UCHL1	100%	100%	100%	100%	99.8%	{?Parkinson disease 5, susceptibility to}, 613643;Spastic paraplegia 79A, autosomal dominant, 620221;Spastic paraplegia 79B, autosomal recessive, 615491
UNC119	100%	100%	100%	100%	99.6%	Cone-rod dystrophy 24, 620342;?Immunodeficiency 13, 615518
UNC45B	96.1%	95.9%	100%	100%	99.7%	?Cataract 43, 616279;Myofibrillar myopathy 11, 619178
USH1C	100%	100%	100%	100%	99.6%	Usher syndrome, type 1C, 276904;Deafness, autosomal recessive 18A, 602092
USH1G	100%	100%	100%	100%	99.1%	Usher syndrome, type 1G, 606943

USH2A	100%	100%	100%	100%	99.7%	Usher syndrome, type 2A, 276901;Retinitis pigmentosa 39, 613809
USP45	100%	100%	100%	100%	99.8%	?Leber congenital amaurosis 19, 618513
VAX1	100%	100%	100%	100%	98.5%	?Microphthalmia, syndromic 11, 614402
VCAN	100%	100%	100%	100%	99.8%	Wagner syndrome 1, 143200
VHL	87.7%	87.7%	100%	99.9%	98.8%	Hemangioblastoma, cerebellar, somatic;Erythrocytosis, familial, 2, 263400;von Hippel-Lindau syndrome, 193300;Renal cell carcinoma, somatic, 144700;Pheochromocytoma, 171300
VIM	100%	100%	100%	99.9%	99%	Cataract 30, pulverulent, 116300
VPS13B	100%	100%	100%	100%	99.7%	Cohen syndrome, 216550
VSX1	100%	100%	100%	100%	99.6%	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195;Keratoconus 1, 148300
VSX2	100%	100%	100%	99.9%	99.1%	Microphthalmia, isolated 2, 610093;Microphthalmia/coloboma 3, 610092
VWA8	100%	100%	100%	100%	99.8%	?Retinitis pigmentosa 97, 620422
WDPCP	92.1%	92.1%	100%	100%	99.8%	Bardet-Biedl syndrome 15, 615992;Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085

WDR19	100%	100%	100%	100%	99.8%	Nephronophthisis 13, 614377;Cranioectodermal dysplasia 4, 614378;Senior-Loken syndrome 8, 616307;Short-rib thoracic dysplasia 5 with or without polydactyly, 614376;?Spermatogenic failure 72, 619867
WDR36	100%	100%	100%	99.8%	99.4%	Glaucoma 1, open angle, G, 609887
WDR73	100%	100%	100%	100%	99.3%	Galloway-Mowat syndrome 1, 251300
WFS1	91.5%	90.5%	100%	100%	99.4%	Deafness, autosomal dominant 6/14/38, 600965;?Cataract 41, 116400;Wolfram-like syndrome, autosomal dominant, 614296;{Diabetes mellitus, noninsulin-dependent, association with}, 125853;Wolfram syndrome 1, 222300
WHRN	100%	100%	100%	100%	99.4%	Deafness, autosomal recessive 31, 607084;Usher syndrome, type 2D, 611383
WRN	100%	100%	100%	100%	99.7%	Werner syndrome, 277700
YAP1	92.3%	92.3%	100%	100%	99.6%	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or impaired intellectual development, 120433
YARS1	100%	98.5%	100%	100%	99.5%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418;Charcot-Marie-Tooth disease, dominant intermediate C, 608323

YME1L1	96.7%	96.7%	100%	100%	99.7%	?Optic atrophy 11, 617302
YPEL2	100%	100%	100%	100%	100%	
ZEB1	100%	100%	100%	100%	99.8%	Corneal dystrophy, posterior polymorphous, 3, 609141;Corneal dystrophy, Fuchs endothelial, 6, 613270
ZNF124	100%	100%	100%	100%	99.7%	
ZNF408	96.2%	96.2%	100%	100%	99.6%	Retinitis pigmentosa 72, 616469;?Exudative vitreoretinopathy 6, 616468
ZNF423	100%	100%	100%	100%	99.4%	Nephronophthisis 14, 614844;Joubert syndrome 19, 614844
ZNF469	100%	100%	100%	99.9%	99.1%	Brittle cornea syndrome 1, 229200
ZNF513	100%	100%	100%	100%	99.4%	?Retinitis pigmentosa 58, 613617
ZNF644	100%	100%	100%	100%	99.9%	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.

TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
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 : November 25th, 2024.
This list is accurate for panel version DG 5.1.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors