RetNet panel

versie	V5 (290 genen)	Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
ABCA4	601691	Retinal dystrophy, early-onset severe, 248200 (3), Autosomal recessive; Stargardt disease 1, 248200 (3), Autosomal recessive; Fundus flavimaculatus, 248200 (3), Autosomal recessive; {Macular degeneration, age-related, 2}, 153800 (3), Autosomal dominant; Cone-rod dystrophy 3, 604116 (3); Retinitis pigmentosa 19, 601718 (3), Autosomal recessive
ABCC6	603234	Pseudoxanthoma elasticum, 264800 (3), Autosomal recessive; Pseudoxanthoma elasticum, forme fruste, 177850 (3), Autosomal dominant; Arterial calcification, generalized, of infancy, 2, 614473 (3), Autosomal recessive
ABHD12	613599	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 (3), Autosomal recessive
ACBD5	616618	Retinal dystrophy with leukodystrophy, 618863 (3), Autosomal recessive
ACO2	100850	Infantile cerebellar-retinal degeneration, 614559 (3), Autosomal recessive; ?Optic atrophy 9, 616289 (3), Autosomal recessive
ADAM9	602713	Cone-rod dystrophy 9, 612775 (3), Autosomal recessive
ADAMTS18	607512	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458 (3), Autosomal recessive
ADGRA3	612303	No OMIM phenotype
ADGRV1	602851	Usher syndrome, type 2C, 605472 (3), Autosomal recessive, Digenic dominant; ?Febrile seizures, familial, 4, 604352 (3), Autosomal dominant; Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 (3), Autosomal recessive, Digenic dominant
ADIPOR1	607945	No OMIM phenotype
AFG3L2	604581	Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Optic atrophy 12, 618977 (3); Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant
AGBL5	615900	Retinitis pigmentosa 75, 617023 (3), Autosomal recessive
AHI1	608894	Joubert syndrome 3, 608629 (3), Autosomal recessive
AHR	600253	?Retinitis pigmentosa 85, 618345 (3), Autosomal recessive
AIPL1	604392	Leber congenital amaurosis 4, 604393 (3), Autosomal recessive, Autosomal dominant; Retinitis pigmentosa, juvenile, 604393 (3), Autosomal recessive, Autosomal dominant; Cone-rod dystrophy, 604393 (3), Autosomal recessive, Autosomal dominant
ALMS1	606844	Alstrom syndrome, 203800 (3), Autosomal recessive
ARHGEF18	616432	Retinitis pigmentosa 78, 617433 (3), Autosomal recessive

ARL2BP	615407	Retinitis pigmentosa with or without situs inversus, 615434 (3), Autosomal recessive
ARL3	604695	Joubert syndrome 35, 618161 (3), Autosomal recessive; Retinitis pigmentosa 83, 618173 (3), Autosomal dominant
ARL6	608845	?Retinitis pigmentosa 55, 613575 (3); Bardet-Biedl syndrome 3, 600151 (3), Autosomal recessive; {Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Autosomal recessive, Digenic recessive
ARMS2	611313	{Macular degeneration, age-related, 8}, 613778 (3)
ARSG	610008	Usher syndrome, type IV, 618144 (3), Autosomal recessive
ASRGL1	No OMIM gene	No OMIM phenotype
ATF6	605537	Achromatopsia 7, 616517 (3), Autosomal recessive
ATXN7	607640	Spinocerebellar ataxia 7, 164500 (3), Autosomal dominant
BBIP1	613605	?Bardet-Biedl syndrome 18, 615995 (3), Autosomal recessive
BBS1	209901	Bardet-Biedl syndrome 1, 209900 (3), Autosomal recessive, Digenic recessive
BBS10	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
BBS12	610683	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive
BBS2	606151	Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive; Retinitis pigmentosa 74, 616562 (3), Autosomal recessive
BBS4	600374	Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive
BBS5	603650	Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive
BBS7	607590	Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive
BBS9	607968	Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive
BEST1	607854	Retinitis pigmentosa-50, 613194 (3); Bestrophinopathy, autosomal recessive, 611809 (3); Retinitis pigmentosa, concentric, 613194 (3); Vitreoretinochoroidopathy, 193220 (3), Autosomal dominant; Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 (3), Autosomal dominant; Macular dystrophy, vitelliform, 2, 153700 (3), Autosomal dominant
C12orf65	613541	Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive
C1QTNF5	608752	Retinal degeneration, late-onset, autosomal dominant, 605670 (3), Autosomal dominant
C2	613927	C2 deficiency, 217000 (3), Autosomal recessive; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3)
C3	120700	{Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 (3), Autosomal dominant; C3 deficiency, 613779 (3), Autosomal recessive; {Macular degeneration, age-related, 9}, 611378 (3)
C8orf37	614477	Retinitis pigmentosa 64, 614500 (3), Autosomal recessive; Bardet-Biedl syndrome 21, 617406 (3), Autosomal recessive; Cone-rod dystrophy 16, 614500 (3), Autosomal recessive
CA4	114760	Retinitis pigmentosa 17, 600852 (3), Autosomal dominant

CABP4	608965	Cone-rod synaptic disorder, congenital nonprogressive, 610427 (3), Autosomal recessive
CACNA1F	300110	Cone-rod dystrophy, X-linked, 3, 300476 (3), X-linked recessive; Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 (3), X-linked; Aland Island eye disease, 300600 (3), X-linked
CACNA2D4	608171	Retinal cone dystrophy 4, 610478 (3), Autosomal recessive
CAPN5	602537	Vitreoretinopathy, neovascular inflammatory, 193235 (3), Autosomal dominant
CC2D2A	612013	Meckel syndrome 6, 612284 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive; COACH syndrome, 216360 (3), Autosomal recessive
CCT2	605139	No OMIM phenotype
CDH23	605516	{Pituitary adenoma 5, multiple types}, 617540 (3), Autosomal dominant; Deafness, autosomal recessive 12, 601386 (3), Autosomal recessive; Usher syndrome, type 1D/F digenic, 601067 (3), Autosomal recessive, Digenic recessive; Usher syndrome, type 1D, 601067 (3), Autosomal recessive, Digenic recessive
CDH3	114021	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 (3), Autosomal recessive; Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 (3), Autosomal recessive
CDHR1	609502	Cone-rod dystrophy 15, 613660 (3), Autosomal recessive; Retinitis pigmentosa 65, 613660 (3), Autosomal recessive
CEP164	614848	Nephronophthisis 15, 614845 (3), Autosomal recessive
CEP19	615586	Morbid obesity and spermatogenic failure, 615703 (3), Autosomal recessive
CEP250	609689	Cone-rod dystrophy and hearing loss 2, 618358 (3), Autosomal recessive
CEP290	610142	?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Leber congenital amaurosis 10, 611755 (3); Senior-Loken syndrome 6, 610189 (3), Autosomal recessive; Meckel syndrome 4, 611134 (3), Autosomal recessive; Joubert syndrome 5, 610188 (3), Autosomal recessive
CEP78	617110	Cone-rod dystrophy and hearing loss, 617236 (3), Autosomal recessive
CERKL	608381	Retinitis pigmentosa 26, 608380 (3)
CFAP410	603191	Spondylometaphyseal dysplasia, axial, 602271 (3), Autosomal recessive; Retinal dystrophy with macular staphyloma, 617547 (3), Autosomal recessive
CFB	138470	?Complement factor B deficiency, 615561 (3), Autosomal recessive; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3); {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 (3), Autosomal dominant

CFH	134370	Complement factor H deficiency, 609814 (3), Autosomal recessive, Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 (3), Autosomal recessive, Autosomal dominant; Basal laminar drusen, 126700 (3), Autosomal dominant; {Macular degeneration, age-related, 4}, 610698 (3)
CHM	300390	Choroideremia, 303100 (3), X-linked dominant
CIB2	605564	Deafness, autosomal recessive 48, 609439 (3), Autosomal recessive; Usher syndrome, type IJ, 614869 (3), Autosomal recessive
CLCC1	617539	Retinitis pigmentosa 32, 609913 (3), Autosomal recessive
CLN3	607042	Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive
CLRN1	606397	Retinitis pigmentosa 61, 614180 (3); Usher syndrome, type 3A, 276902 (3), Autosomal recessive
CLUAP1	616787	No OMIM phenotype
CNGA1	123825	Retinitis pigmentosa 49, 613756 (3)
CNGA3	600053	Achromatopsia 2, 216900 (3), Autosomal recessive
CNGB1	600724	Retinitis pigmentosa 45, 613767 (3), Autosomal recessive
CNGB3	605080	Achromatopsia 3, 262300 (3), Autosomal recessive
CNNM4	607805	Jalili syndrome, 217080 (3), Autosomal recessive
COL11A1	120280	Stickler syndrome, type II, 604841 (3), Autosomal dominant; Marshall syndrome, 154780 (3), Autosomal dominant; ?Deafness, autosomal dominant 37, 618533 (3), Autosomal dominant; {Lumbar disc herniation, susceptibility to}, 603932 (3); Fibrochondrogenesis 1, 228520 (3), Autosomal recessive
COL2A1	120140	Vitreoretinopathy with phalangeal epiphyseal dysplasia (3); Achondrogenesis, type II or hypochondrogenesis, 200610 (3), Autosomal dominant; Spondyloperipheral dysplasia, 271700 (3), Autosomal dominant; Kniest dysplasia, 156550 (3), Autosomal dominant; Stickler syndrome, type I, 108300 (3), Autosomal dominant; Osteoarthritis with mild chondrodysplasia, 604864 (3), Autosomal dominant; Platyspondylic skeletal dysplasia, Torrance type, 151210 (3), Autosomal dominant; Avascular necrosis of the femoral head, 608805 (3), Autosomal dominant; ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 (3), Autosomal dominant; SED congenita, 183900 (3), Autosomal dominant; Legg-Calve-Perthes disease, 150600 (3), Autosomal dominant; SMED Strudwick type, 184250 (3), Autosomal dominant; Czech dysplasia, 609162 (3), Autosomal dominant; Stickler sydrome, type I, nonsyndromic ocular, 609508 (3), Autosomal dominant; Spondyloepiphyseal dysplasia, Stanescu type, 616583 (3), Autosomal dominant
COL9A1	120210	?Epiphyseal dysplasia, multiple, 6, 614135 (3), Autosomal dominant; Stickler syndrome, type IV, 614134 (3)
CRB1	604210	Pigmented paravenous chorioretinal atrophy, 172870 (3), Autosomal dominant; Retinitis pigmentosa-12, 600105 (3),

		Autosomal recessive; Leber congenital amaurosis 8, 613835 (3), Autosomal recessive
CRX	602225	Cone-rod retinal dystrophy-2, 120970 (3), Autosomal dominant; Leber congenital amaurosis 7, 613829 (3)
CSPP1	611654	Joubert syndrome 21, 615636 (3), Autosomal recessive
CTNNA1	116805	Macular dystrophy, patterned, 2, 608970 (3), Autosomal dominant
CWC27	617170	Retinitis pigmentosa with or without skeletal anomalies, 250410 (3), Autosomal recessive
CYP4V2	608614	Bietti crystalline corneoretinal dystrophy, 210370 (3), Autosomal recessive
DHDDS	608172	Retinitis pigmentosa 59, 613861 (3), Autosomal recessive; Developmental delay and seizures with or without movement abnormalities, 617836 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1bb, 613861 (3), Autosomal recessive
DHX38	605584	Retinitis pigmentosa 84, 618220 (3), Autosomal recessive
DMD	300377	Cardiomyopathy, dilated, 3B, 302045 (3), X-linked; Becker muscular dystrophy, 300376 (3), X-linked recessive; Duchenne muscular dystrophy, 310200 (3), X-linked recessive
DRAM2	613360	Cone-rod dystrophy 21, 616502 (3), Autosomal recessive
DTHD1	616979	No OMIM phenotype
DYNC2H1	603297	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091 (3), Autosomal recessive, Digenic recessive
EFEMP1	601548	Doyne honeycomb degeneration of retina, 126600 (3), Autosomal dominant
ELOVL1	611813	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527 (3), Autosomal dominant
ELOVL4	605512	Spinocerebellar ataxia 34, 133190 (3), Autosomal dominant; Stargardt disease 3, 600110 (3), Autosomal dominant; Ichthyosis, spastic quadriplegia, and mental retardation, 614457 (3), Autosomal recessive
EMC1	616846	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875 (3), Autosomal recessive
ERCC6	609413	{Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); {Lung cancer, susceptibility to}, 211980 (3), Autosomal dominant, Somatic mutation; Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; Premature ovarian failure 11, 616946 (3), Autosomal dominant; UV-sensitive syndrome 1, 600630 (3), Autosomal recessive; De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive
ESPN	606351	?Usher syndrome, type 1M, 618632 (3), Autosomal recessive; Deafness, autosomal recessive 36, 609006 (3), Autosomal recessive; Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006 (3), Autosomal recessive

EXOSC2	602238	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763 (3), Autosomal recessive
EYS	612424	Retinitis pigmentosa 25, 602772 (3), Autosomal recessive
FAM161A	613596	Retinitis pigmentosa 28, 606068 (3)
FBLN5	604580	Macular degeneration, age-related, 3, 608895 (3), Autosomal dominant; ?Cutis laxa, autosomal dominant 2, 614434 (3), Autosomal dominant; Neuropathy, hereditary, with or without age-related macular degeneration, 608895 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type IA, 219100 (3), Autosomal recessive
FBN2	612570	Contractural arachnodactyly, congenital, 121050 (3), Autosomal dominant; Macular degeneration, early-onset, 616118 (3), Autosomal dominant
FLVCR1	609144	Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive
FSCN2	607643	Retinitis pigmentosa 30, 607921 (3)
FZD4	604579	Exudative vitreoretinopathy 1, 133780 (3), Autosomal dominant; Retinopathy of prematurity, 133780 (3), Autosomal dominant
GDF6	601147	Leber congenital amaurosis 17, 615360 (3), Autosomal recessive; Klippel-Feil syndrome 1, autosomal dominant, 118100 (3), Autosomal dominant; Multiple synostoses syndrome 4, 617898 (3), Autosomal dominant; Microphthalmia with coloboma 6, digenic, 613703 (3), Autosomal dominant; Microphthalmia, isolated 4, 613094 (3)
GNAT1	139330	Night blindness, congenital stationary, type 1G, 616389 (3), Autosomal recessive; Night blindness, congenital stationary, autosomal dominant 3, 610444 (3), Autosomal dominant
GNAT2	139340	Achromatopsia 4, 613856 (3)
GNB3	139130	{Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial; Night blindness, congenital stationary, type 1H, 617024 (3), Autosomal recessive
GNPTG	607838	Mucolipidosis III gamma, 252605 (3), Autosomal recessive
GPR179	614515	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565 (3), Autosomal recessive
GRID2	602368	Spinocerebellar ataxia, autosomal recessive 18, 616204 (3), Autosomal recessive
GRK1	180381	Oguchi disease-2, 613411 (3)
GRM6	604096	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270 (3), Autosomal recessive
GUCA1A	600364	Cone-rod dystrophy 14, 602093 (3), Autosomal dominant; Cone dystrophy-3, 602093 (3), Autosomal dominant
GUCA1B	602275	Retinitis pigmentosa 48, 613827 (3)

GUCY2D	600179	Cone-rod dystrophy 6, 601777 (3), Autosomal recessive, Autosomal dominant; Leber congenital amaurosis 1, 204000 (3), Autosomal recessive; Night blindness, congenital stationary, type 1I, 618555 (3), Autosomal recessive; ?Choroidal dystrophy, central areolar 1, 215500 (3), Autosomal dominant
HARS1 (HARS)	142810	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 (3), Autosomal dominant; Usher syndrome type 3B, 614504 (3), Autosomal recessive
HGSNAT	610453	Retinitis pigmentosa 73, 616544 (3), Autosomal recessive; Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 (3), Autosomal recessive
HK1	142600	Hemolytic anemia due to hexokinase deficiency, 235700 (3), Autosomal recessive; Neuropathy, hereditary motor and sensory, Russe type, 605285 (3), Autosomal recessive; Neurodevelopmental disorder with visual defects and brain anomalies, 618547 (3), Autosomal dominant; Retinitis pigmentosa 79, 617460 (3), Autosomal dominant
HMCN1	608548	{Macular degeneration, age-related, 1}, 603075 (3), Autosomal dominant
HMX1	142992	Oculoauricular syndrome, 612109 (3), Autosomal recessive
HTRA1	602194	{Macular degeneration, age-related, neovascular type}, 610149 (3); {Macular degeneration, age-related, 7}, 610149 (3); Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 (3), Autosomal dominant; CARASIL syndrome, 600142 (3), Autosomal recessive
IDH3A	601149	No OMIM phenotype
IDH3B	604526	Retinitis pigmentosa 46, 612572 (3)
IFT140	614620	Retinitis pigmentosa 80, 617781 (3), Autosomal recessive; Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive
IFT172	607386	Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive
IFT27	615870	?Bardet-Biedl syndrome 19, 615996 (3), Autosomal recessive
IFT43	614068	?Cranioectodermal dysplasia 3, 614099 (3), Autosomal recessive; Short-rib thoracic dysplasia 18 with polydactyly, 617866 (3), Autosomal recessive; ?Retinitis pigmentosa 81, 617871 (3), Autosomal recessive
IFT81	605489	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895 (3), Autosomal recessive
IFT88	600595	No OMIM phenotype
IMPDH1	146690	Leber congenital amaurosis 11, 613837 (3), Autosomal dominant; Retinitis pigmentosa 10, 180105 (3), Autosomal dominant
IMPG1	602870	Macular dystrophy, vitelliform, 4, 616151 (3), Autosomal dominant
IMPG2	607056	Macular dystrophy, vitelliform, 5, 616152 (3), Autosomal dominant; Retinitis pigmentosa 56, 613581 (3), Autosomal recessive

INPP5E	613037	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 (3), Autosomal recessive; Joubert syndrome 1, 213300 (3), Autosomal recessive
INVS	243305	Nephronophthisis 2, infantile, 602088 (3), Autosomal recessive
IQCB1	609237	Senior-Loken syndrome 5, 609254 (3), Autosomal recessive
ITM2B	603904	Dementia, familial British, 176500 (3), Autosomal dominant; ?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 (3), Autosomal dominant; Dementia, familial Danish, 117300 (3), Autosomal dominant
JAG1	601920	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3); Alagille syndrome 1, 118450 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
KCNJ13	603208	Leber congenital amaurosis 16, 614186 (3), Autosomal recessive; Snowflake vitreoretinal degeneration, 193230 (3), Autosomal dominant
KCNV2	607604	Retinal cone dystrophy 3B, 610356 (3), Autosomal recessive
KIAA1549	613344	Retinitis pigmentosa 86, 618613 (3), Autosomal recessive
KIF11	148760	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950 (3), Autosomal dominant
KIF3B	603754	Retinitis pigmentosa 89, 618955 (3), Autosomal dominant
KIZ	615757	Retinitis pigmentosa 69, 615780 (3), Autosomal recessive
KLHL7	611119	Retinitis pigmentosa 42, 612943 (3), Autosomal dominant; PERCHING syndrome, 617055 (3), Autosomal recessive
LAMA1	150320	Poretti-Boltshauser syndrome, 615960 (3), Autosomal recessive
LAMB2	150325	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 (3); Pierson syndrome, 609049 (3), Autosomal recessive
LCA5	611408	Leber congenital amaurosis 5, 604537 (3), Autosomal recessive
LRAT	604863	Retinal dystrophy, early-onset severe, 613341 (3), Autosomal recessive; Leber congenital amaurosis 14, 613341 (3), Autosomal recessive; Retinitis pigmentosa, juvenile, 613341 (3), Autosomal recessive
LRIT3	615004	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058 (3), Autosomal recessive
LRP5	603506	van Buchem disease, type 2, 607636 (3), Autosomal dominant; Exudative vitreoretinopathy 4, 601813 (3), Autosomal recessive, Autosomal dominant; Hyperostosis, endosteal, 144750 (3), Autosomal dominant; Osteosclerosis, 144750 (3), Autosomal dominant; Polycystic liver disease 4 with or without kidney cysts, 617875 (3), Autosomal dominant; Osteoporosis-pseudoglioma syndrome, 259770 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 1, 607634 (3), Autosomal dominant; {Osteoporosis}, 166710 (3), Autosomal dominant; [Bone mineral density variability 1], 601884 (3), Autosomal dominant
LZTFL1	606568	Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive
MAK	154235	Retinitis pigmentosa 62, 614181 (3), Autosomal recessive

MAPKAPK3	602130	?Macular dystrophy, patterned, 3, 617111 (3), Autosomal dominant
MERTK	604705	Retinitis pigmentosa 38, 613862 (3), Autosomal recessive
MFN2	608507	Hereditary motor and sensory neuropathy VIA, 601152 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 (3), Autosomal dominant
MFRP	606227	Nanophthalmos 2, 609549 (3); Microphthalmia, isolated 5, 611040 (3), Autosomal recessive
MFSD8	611124	Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive
MIR204	610942	?Retinal dystrophy and iris coloboma with or without cataract, 616722 (3), Autosomal dominant
MKKS	604896	Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive; McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive
MKS1	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive
MTTP	157147	Abetalipoproteinemia, 200100 (3), Autosomal recessive; {Metabolic syndrome, protection against}, 605552 (3), Autosomal dominant
MVK	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant; Mevalonic aciduria, 610377 (3), Autosomal recessive
MYO7A	276903	Deafness, autosomal recessive 2, 600060 (3), Autosomal recessive; Deafness, autosomal dominant 11, 601317 (3), Autosomal dominant; Usher syndrome, type 1B, 276900 (3), Autosomal recessive
NBAS	608025	Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive; Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive
NDP	300658	Exudative vitreoretinopathy 2, X-linked, 305390 (3), X-linked recessive, X-linked dominant; Norrie disease, 310600 (3), X-linked recessive
NEK2	604043	?Retinitis pigmentosa 67, 615565 (3), Autosomal recessive
NEUROD1	601724	Maturity-onset diabetes of the young 6, 606394 (3); {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant
NMNAT1	608700	Leber congenital amaurosis 9, 608553 (3), Autosomal recessive
NPHP1	607100	Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive; Joubert syndrome 4, 609583 (3), Autosomal recessive
NPHP3	608002	Meckel syndrome 7, 267010 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive; Nephronophthisis 3, 604387 (3), Autosomal recessive
NPHP4	607215	Nephronophthisis 4, 606966 (3), Autosomal recessive; Senior-Loken syndrome 4, 606996 (3), Autosomal recessive

NR2E3	604485	Enhanced S-cone syndrome, 268100 (3), Autosomal recessive; Retinitis pigmentosa 37, 611131 (3), Autosomal recessive, Autosomal dominant
NR2F1	132890	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722 (3), Autosomal dominant
NRL	162080	Retinitis pigmentosa 27, 613750 (3), Autosomal dominant; Retinal degeneration, autosomal recessive, clumped pigment type (3)
NYX	300278	Night blindness, congenital stationary (complete), 1A, X-linked, 310500 (3), X-linked recessive
OAT	613349	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870 (3), Autosomal recessive
OFD1	300170	Orofaciodigital syndrome I, 311200 (3), X-linked dominant; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Joubert syndrome 10, 300804 (3), X-linked recessive; Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive
OPA1	605290	{Glaucoma, normal tension, susceptibility to}, 606657 (3); Behr syndrome, 210000 (3), Autosomal recessive; Optic atrophy 1, 165500 (3), Autosomal dominant; Optic atrophy plus syndrome, 125250 (3), Autosomal dominant; ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 (3), Autosomal recessive
OPA3	606580	3-methylglutaconic aciduria, type III, 258501 (3), Autosomal recessive; Optic atrophy 3 with cataract, 165300 (3), Autosomal dominant
OPN1LW	300822	Blue cone monochromacy, 303700 (3), X-linked recessive; Colorblindness, protan, 303900 (3), X-linked
OPN1MW	300821	Colorblindness, deutan, 303800 (3), X-linked; Blue cone monochromacy, 303700 (3), X-linked recessive
OPN1SW	613522	Colorblindness, tritan, 190900 (3), Autosomal dominant
OTX2	600037	Pituitary hormone deficiency, combined, 6, 613986 (3), Autosomal dominant; Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 (3), Autosomal dominant; Microphthalmia, syndromic 5, 610125 (3), Autosomal dominant
PANK2	606157	HARP syndrome, 607236 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 1, 234200 (3), Autosomal recessive
PAX2	167409	Glomerulosclerosis, focal segmental, 7, 616002 (3), Autosomal dominant; Papillorenal syndrome, 120330 (3), Autosomal dominant
PCARE	613425	Retinitis pigmentosa 54, 613428 (3)
PCDH15	605514	Usher syndrome, type 1D/F digenic, 601067 (3), Autosomal recessive, Digenic recessive; Usher syndrome, type 1F, 602083 (3), Autosomal recessive; Deafness, autosomal recessive 23, 609533 (3), Autosomal recessive
PCYT1A	123695	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940 (3), Autosomal recessive
PDE6A	180071	Retinitis pigmentosa 43, 613810 (3)

PDE6B	180072	Night blindness, congenital stationary, autosomal dominant 2, 163500 (3), Autosomal dominant; Retinitis pigmentosa-40, 613801 (3), Autosomal recessive
PDE6C	600827	Cone dystrophy 4, 613093 (3), Autosomal recessive
PDE6G	180073	Retinitis pigmentosa 57, 613582 (3), Autosomal recessive
PDE6H	601190	Retinal cone dystrophy 3, 610024 (3), Autosomal recessive, Autosomal dominant; Achromatopsia 6, 610024 (3), Autosomal recessive, Autosomal dominant
PDZD7	612971	Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 (3), Autosomal recessive, Digenic dominant; Deafness, autosomal recessive 57, 618003 (3), Autosomal recessive; {Retinal disease in Usher syndrome type IIA, modifier of}, 276901 (3), Autosomal recessive
PEX1	602136	Heimler syndrome 1, 234580 (3), Autosomal recessive; Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 (3), Autosomal recessive; Peroxisome biogenesis disorder 1A (Zellweger), 214100 (3), Autosomal recessive
PEX2	170993	Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive
PEX6	601498	Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal recessive, Autosomal dominant; Heimler syndrome 2, 616617 (3), Autosomal recessive; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive
PEX7	601757	Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive
PGK1	311800	Phosphoglycerate kinase 1 deficiency, 300653 (3), X-linked recessive
PHYH	602026	Refsum disease, 266500 (3), Autosomal recessive
PITPNM3	608921	Cone-rod dystrophy 5, 600977 (3), Autosomal dominant
PLA2G5	601192	[Fleck retina, familial benign], 228980 (3), Autosomal recessive
PLK4	605031	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171 (3), Autosomal recessive
PNPLA6	603197	Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive; Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive
POC1B	614784	Cone-rod dystrophy 20, 615973 (3), Autosomal recessive
POC5	617880	No OMIM phenotype

POMGNT1	606822	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 (3), Autosomal recessive; Retinitis pigmentosa 76, 617123 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 (3), Autosomal recessive
PRCD	610598	Retinitis pigmentosa 36, 610599 (3)
PRDM13	616741	No OMIM phenotype
PROM1	604365	Retinitis pigmentosa 41, 612095 (3), Autosomal recessive; Stargardt disease 4, 603786 (3), Autosomal dominant; Cone-rod dystrophy 12, 612657 (3), Autosomal recessive, Autosomal dominant; Macular dystrophy, retinal, 2, 608051 (3), Autosomal dominant
PRPF3	607301	Retinitis pigmentosa 18, 601414 (3), Autosomal dominant
PRPF31	606419	Retinitis pigmentosa 11, 600138 (3), Autosomal dominant
PRPF4	607795	Retinitis pigmentosa 70, 615922 (3), Autosomal dominant
PRPF6	613979	Retinitis pigmentosa 60, 613983 (3), Autosomal dominant
PRPF8	607300	Retinitis pigmentosa 13, 600059 (3), Autosomal dominant
PRPH2	179605	Macular dystrophy, patterned, 1, 169150 (3), Autosomal dominant; Retinitis punctata albescens, 136880 (3), Autosomal recessive, Autosomal dominant; Choroidal dystrophy, central areolar 2, 613105 (3), Autosomal dominant; Retinitis pigmentosa 7 and digenic form, 608133 (3), Autosomal recessive, Autosomal dominant; Leber congenital amaurosis 18, 608133 (3), Autosomal recessive, Autosomal dominant; Macular dystrophy, vitelliform, 3, 608161 (3), Autosomal dominant
PRPS1	311850	Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 (3), X-linked recessive; Phosphoribosylpyrophosphate synthetase superactivity, 300661 (3), X-linked recessive; Deafness, X-linked 1, 304500 (3), X-linked; Arts syndrome, 301835 (3), X-linked recessive; Gout, PRPS-related, 300661 (3), X-linked recessive
RAB28	612994	Cone-rod dystrophy 18, 615374 (3), Autosomal recessive
RAX2	610362	?Macular degeneration, age-related, 6, 613757 (3); Cone-rod dystrophy 11, 610381 (3), Autosomal dominant
RB1	614041	Small cell cancer of the lung, somatic, 182280 (3); Bladder cancer, somatic, 109800 (3); Retinoblastoma, trilateral, 180200 (3), Autosomal dominant, Somatic mutation; Osteosarcoma, somatic, 259500 (3); Retinoblastoma, 180200 (3), Autosomal dominant, Somatic mutation
RBP3	180290	?Retinitis pigmentosa 66, 615233 (3), Autosomal recessive
RBP4	180250	Microphthalmia, isolated, with coloboma 10, 616428 (3), Autosomal dominant; Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147 (3), Autosomal recessive
RCBTB1	607867	Retinal dystrophy with or without extraocular anomalies, 617175 (3), Autosomal recessive
RD3	180040	Leber congenital amaurosis 12, 610612 (3), Autosomal recessive

RDH11	607849	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108 (3), Autosomal recessive
RDH12	608830	Leber congenital amaurosis 13, 612712 (3), Autosomal recessive, Autosomal dominant
RDH5	601617	Fundus albipunctatus, 136880 (3), Autosomal recessive, Autosomal dominant
REEP6	609346	Retinitis pigmentosa 77, 617304 (3), Autosomal recessive
RGR	600342	Retinitis pigmentosa 44, 613769 (3)
RGS9	604067	Bradyopsia, 608415 (3)
RGS9BP	607814	Bradyopsia, 608415 (3)
RHO	180380	Night blindness, congenital stationary, autosomal dominant 1, 610445 (3); Retinitis punctata albescens, 136880 (3), Autosomal recessive, Autosomal dominant; Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 (3), Autosomal recessive, Autosomal dominant
RIMS1	606629	Cone-rod dystrophy 7, 603649 (3)
RIMS2	606630	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970 (3), Autosomal recessive
RLBP1	180090	Retinitis punctata albescens, 136880 (3), Autosomal recessive, Autosomal dominant; Bothnia retinal dystrophy, 607475 (3), Autosomal recessive; Newfoundland rod-cone dystrophy, 607476 (3); Fundus albipunctatus, 136880 (3), Autosomal recessive, Autosomal dominant
ROM1	180721	Retinitis pigmentosa 7, digenic form, 608133 (3), Autosomal recessive, Autosomal dominant
RP1	603937	Retinitis pigmentosa 1, 180100 (3), Autosomal recessive, Autosomal dominant
RP1L1	608581	Retinitis pigmentosa 88, 618826 (3), Autosomal recessive; Occult macular dystrophy, 613587 (3), Autosomal dominant
RP2	300757	Retinitis pigmentosa 2, 312600 (3), X-linked
RP9	607331	?Retinitis pigmentosa 9, 180104 (3), Autosomal dominant
RPE65	180069	Leber congenital amaurosis 2, 204100 (3), Autosomal recessive; Retinitis pigmentosa 87 with choroidal involvement, 618697 (3), Autosomal dominant; Retinitis pigmentosa 20, 613794 (3), Autosomal recessive
RPGR	312610	Cone-rod dystrophy, X-linked, 1, 304020 (3), X-linked; Retinitis pigmentosa 3, 300029 (3); Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 (3); Macular degeneration, X-linked atrophic, 300834 (3), X-linked recessive
RPGRIP1	605446	Cone-rod dystrophy 13, 608194 (3); Leber congenital amaurosis 6, 613826 (3), Autosomal recessive
RPGRIP1L	610937	COACH syndrome, 216360 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; Joubert syndrome 7, 611560 (3), Autosomal recessive
RS1	300839	Retinoschisis, 312700 (3), X-linked recessive

RTN4IP1	610502	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732 (3), Autosomal recessive
SAG	181031	Oguchi disease-1, 258100 (3), Autosomal recessive; Retinitis pigmentosa 47, 613758 (3)
SAMD11	616765	No OMIM phenotype
SAMD7	No OMIM gene	No OMIM phenotype
SCAPER	611611	Intellectual developmental disorder and retinitis pigmentosa, 618195 (3), Autosomal recessive
SCLT1	611399	No OMIM phenotype
SDCCAG8	613524	Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive; Senior-Loken syndrome 7, 613615 (3)
SEMA4A	607292	Cone-rod dystrophy 10, 610283 (3), Autosomal recessive; Retinitis pigmentosa 35, 610282 (3), Autosomal recessive, Autosomal dominant
SF3B2	605591	No OMIM phenotype
SLC24A1	603617	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830 (3), Autosomal recessive
SLC25A46	610826	Neuropathy, hereditary motor and sensory, type VIB, 616505 (3), Autosomal recessive
SLC6A6	186854	No OMIM phenotype
SLC7A14	615720	Retinitis pigmentosa 68, 615725 (3), Autosomal recessive
SNRNP200	601664	Retinitis pigmentosa 33, 610359 (3), Autosomal dominant
SPATA7	609868	Retinitis pigmentosa, juvenile, autosomal recessive, 604232 (3); Leber congenital amaurosis 3, 604232 (3)
SPP2	602637	No OMIM phenotype
TEAD1	189967	Sveinsson chorioretinal atrophy, 108985 (3), Autosomal dominant
TIMM8A	300356	Mohr-Tranebjaerg syndrome, 304700 (3), X-linked recessive
TIMP3	188826	Sorsby fundus dystrophy, 136900 (3), Autosomal dominant
TLR3	603029	{HIV1 infection, resistance to}, 609423 (3); {Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 2}, 613002 (3), Autosomal recessive, Autosomal dominant
TLR4	603030/611488	-/Macular degeneration, age-related, 10, 611488 (2)
TMEM126A	612988	Optic atrophy 7, 612989 (3), Autosomal recessive
TMEM216	613277	Meckel syndrome 2, 603194 (3), Autosomal recessive; Joubert syndrome 2, 608091 (3), Autosomal recessive
TMEM237	614423	Joubert syndrome 14, 614424 (3), Autosomal recessive
TOPORS	609507	Retinitis pigmentosa 31, 609923 (3)
TREX1	606609	{Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Vasculopathy, retinal, with cerebral leukodystrophy, 192315 (3), Autosomal dominant; Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal recessive, Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant

TRIM32	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 (3), Autosomal recessive
TRNT1	612907	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 (3), Autosomal recessive; Retinitis pigmentosa and erythrocytic microcytosis, 616959 (3), Autosomal recessive
TRPM1	603576	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216 (3)
TSPAN12	613138	Exudative vitreoretinopathy 5, 613310 (3), Autosomal dominant
TTC8	608132	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
TTLL5	612268	Cone-rod dystrophy 19, 615860 (3), Autosomal recessive
TTPA	600415	Ataxia with isolated vitamin E deficiency, 277460 (3), Autosomal recessive
TUB	601197	?Retinal dystrophy and obesity, 616188 (3), Autosomal recessive
TUBGCP4	609610	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335 (3), Autosomal recessive
TUBGCP6	610053	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270 (3), Autosomal recessive
TULP1	602280	Retinitis pigmentosa 14, 600132 (3), Autosomal recessive; Leber congenital amaurosis 15, 613843 (3), Autosomal recessive
UNC119	604011	?Immunodeficiency 13, 615518 (3), Autosomal dominant; ?Cone-rod dystrophy (3)
USH1C	605242	Deafness, autosomal recessive 18A, 602092 (3), Autosomal recessive; Usher syndrome, type 1C, 276904 (3), Autosomal recessive
USH1G	607696	Usher syndrome, type 1G, 606943 (3), Autosomal recessive
USH2A	608400	Retinitis pigmentosa 39, 613809 (3); Usher syndrome, type 2A, 276901 (3), Autosomal recessive
VCAN	118661	Wagner syndrome 1, 143200 (3), Autosomal dominant
WDPCP	613580	?Bardet-Biedl syndrome 15, 615992 (3), Autosomal recessive; ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 (3), Autosomal recessive
WDR19	608151	Nephronophthisis 13, 614377 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; ?Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive
WFS1	606201	?Cataract 41, 116400 (3), Autosomal dominant; Deafness, autosomal dominant 6/14/38, 600965 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent, association with}, 125853 (3), Autosomal dominant; Wolfram-like syndrome, autosomal dominant, 614296 (3), Autosomal dominant; Wolfram syndrome 1, 222300 (3), Autosomal recessive

WHRN	607928	Deafness, autosomal recessive 31, 607084 (3), Autosomal recessive; Usher syndrome, type 2D, 611383 (3), Autosomal recessive
ZNF408	616454	?Exudative vitreoretinopathy 6, 616468 (3), Autosomal dominant; Retinitis pigmentosa 72, 616469 (3), Autosomal recessive
ZNF423	604557	Nephronophthisis 14, 614844 (3), Autosomal recessive, Autosomal dominant; Joubert syndrome 19, 614844 (3), Autosomal recessive, Autosomal dominant
ZNF513	613598	?Retinitis pigmentosa 58, 613617 (3), Autosomal recessive

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.
Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.
OMIM release used for OMIM disease identifiers and descriptions: Sep 01, 2020

Possible phenotype mapping keys
(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known
(2) the disorder has been placed on the map by linkage; no mutation has been found
(3) the molecular basis for the disorder is known; a mutation has been found in the gene
(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).
Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).
A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.