RetNet panel

versie	v6 (323 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; Retinitis pigmentosa 19, 601718 (3), Autosomal recessive; {Macular degeneration, age-related, 2}, 153800 (3), Autosomal dominant; Cone-rod dystrophy 3, 604116 (3), Autosomal recessive; Fundus flavimaculatus, 248200 (3), Autosomal recessive; Stargardt disease 1, 248200 (3), Autosomal recessive
ABCC6	603234	Pseudoxanthoma elasticum, 264800 (3), Autosomal recessive; Arterial calcification, generalized, of infancy, 2, 614473 (3), Autosomal recessive; Pseudoxanthoma elasticum, forme fruste, 177850 (3), Autosomal dominant
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	?Optic atrophy 9, 616289 (3), Autosomal recessive; Infantile cerebellar-retinal degeneration, 614559 (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
ADGRV1	602851	Usher syndrome, type 2C, 605472 (3), Digenic dominant, Autosomal recessive; Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 (3), Digenic dominant, Autosomal recessive; ?Febrile seizures, familial, 4, 604352 (3), Autosomal dominant
ADIPOR1	607945	No OMIM phenotype
AFG3L2	604581	Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Optic atrophy 12, 618977 (3), Autosomal dominant; 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 dominant, Autosomal recessive; Retinitis pigmentosa, juvenile, 604393 (3), Autosomal dominant, Autosomal recessive; Cone-rod dystrophy, 604393 (3), Autosomal dominant, Autosomal recessive
ALDH3A2	609523	Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive
ALMS1	606844	Alstrom syndrome, 203800 (3), Autosomal recessive
ALPK1	607347	ROSAH syndrome, 614979 (3), Autosomal dominant

AMACR	604489	Alpha-methylacyl-CoA racemase deficiency, 614307 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 4, 214950 (3), Autosomal recessive
ARHGEF18	616432	Retinitis pigmentosa 78, 617433 (3), Autosomal recessive
ARL13B	608922	Joubert syndrome 8, 612291 (3), Autosomal recessive
ARL2BP	615407	Retinitis pigmentosa with or without situs inversus, 615434 (3), Autosomal recessive
ARL3	604695	Retinitis pigmentosa 83, 618173 (3), Autosomal dominant; Joubert syndrome 35, 618161 (3), Autosomal recessive
ARL6	608845	Retinitis pigmentosa 55, 613575 (3), Autosomal recessive; {Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Digenic recessive, Autosomal recessive; Bardet-Biedl syndrome 3, 600151 (3), Autosomal recessive
ARMS2	611313	{Macular degeneration, age-related, 8}, 613778 (3)
ARSG	610008	Usher syndrome, type IV, 618144 (3), Autosomal recessive
ASRGL1	609212	No OMIM phenotype
ATF6	605537	Achromatopsia 7, 616517 (3), Autosomal recessive
ATOH7	609875	Persistent hyperplastic primary vitreous, autosomal recessive, 221900 (3), Autosomal recessive
BBIP1	613605	?Bardet-Biedl syndrome 18, 615995 (3), Autosomal recessive
BBS1	209901	Bardet-Biedl syndrome 1, 209900 (3), Digenic recessive, Autosomal recessive
BBS10	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
BBS12	610683	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive
BBS2	606151	Retinitis pigmentosa 74, 616562 (3), Autosomal recessive; Bardet-Biedl syndrome 2, 615981 (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
BCOR	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant
BEST1	607854	Macular dystrophy, vitelliform, 2, 153700 (3), Autosomal dominant; ?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 2, 193220 (3), Autosomal dominant; Retinitis pigmentosa-50, 613194 (3); Retinitis pigmentosa, concentric, 613194 (3); Vitreoretinochoroidopathy, 193220 (3), Autosomal dominant; Bestrophinopathy, autosomal recessive, 611809 (3)
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), Digenic dominant
C3	120700	C3 deficiency, 613779 (3), Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 (3),

		Autosomal dominant; {Macular degeneration, age-related, 9}, 611378 (3)
CA4	114760	No OMIM phenotype
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	COACH syndrome 2, 619111 (3), Autosomal recessive; Retinitis pigmentosa 93, 619845 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive
CCT2	605139	No OMIM phenotype
CDH23	605516	Usher syndrome, type 1D, 601067 (3), Digenic recessive, Autosomal recessive; {Pituitary adenoma 5, multiple types}, 617540 (3), Autosomal dominant; Usher syndrome, type 1D/F digenic, 601067 (3), Digenic recessive, Autosomal recessive; Deafness, autosomal recessive 12, 601386 (3), Autosomal recessive
CDH3	114021	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 (3), Autosomal recessive; Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 (3), Autosomal recessive
CDHR1	609502	Cone-rod dystrophy 15, 613660 (3), Autosomal recessive; Retinitis pigmentosa 65, 613660 (3), Autosomal recessive
CEP162	610201	No OMIM phenotype
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	Leber congenital amaurosis 10, 611755 (3); Joubert syndrome 5, 610188 (3), Autosomal recessive; Senior-Loken syndrome 6, 610189 (3), Autosomal recessive; ?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Meckel syndrome 4, 611134 (3), Autosomal recessive
CEP78	617110	Cone-rod dystrophy and hearing loss, 617236 (3), Autosomal recessive
CERKL	608381	Retinitis pigmentosa 26, 608380 (3), Autosomal recessive
CFAP20	617906	No OMIM phenotype

CFAP410	603191	Retinal dystrophy with macular staphyloma, 617547 (3), Autosomal recessive; Spondylometaphyseal dysplasia, axial, 602271 (3), Autosomal recessive
CFAP418	614477	Retinitis pigmentosa 64, 614500 (3), Autosomal recessive; Cone-rod dystrophy 16, 614500 (3), Autosomal recessive; Bardet-Biedl syndrome 21, 617406 (3), Autosomal recessive
CFB	138470	?Complement factor B deficiency, 615561 (3), Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 (3), Autosomal dominant; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3), Digenic dominant
CFH	134370	{Macular degeneration, age-related, 4}, 610698 (3); Basal laminar drusen, 126700 (3), Autosomal dominant; Complement factor H deficiency, 609814 (3), Autosomal dominant, Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 (3), Autosomal dominant, Autosomal recessive
CHM	300390	Choroideremia, 303100 (3), X-linked
CLCC1	617539	Retinitis pigmentosa 32, 609913 (3), Autosomal recessive
CLN3	607042	Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive
CLN5	608102	Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive
CLRN1	606397	Usher syndrome, type 3A, 276902 (3), Autosomal recessive; Retinitis pigmentosa 61, 614180 (3)
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	Fibrochondrogenesis 1, 228520 (3), Autosomal recessive; 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)
COL18A1	120328	Knobloch syndrome, type 1, 267750 (3), Autosomal recessive; Glaucoma, primary closed-angle, 618880 (3), Autosomal dominant

COL2A1	120140	?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248 (3), Autosomal dominant; Czech dysplasia, 609162 (3), Autosomal dominant; Achondrogenesis, type II or hypochondrogenesis, 200610 (3), Autosomal dominant; Spondyloperipheral dysplasia, 271700 (3), Autosomal dominant; SMED Strudwick type, 184250 (3), Autosomal dominant; ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 (3), Autosomal dominant; SED congenita, 183900 (3), Autosomal dominant; Kniest dysplasia, 156550 (3), Autosomal dominant; Stickler syndrome, type I, nonsyndromic ocular, 609508 (3), Autosomal dominant; Osteoarthritis with mild chondrodysplasia, 604864 (3), Autosomal dominant; Stickler syndrome, type I, 108300 (3), Autosomal dominant; Platyspondylic skeletal dysplasia, Torrance type, 151210 (3), Autosomal dominant; Spondyloepiphyseal dysplasia, Stanescu type, 616583 (3), Autosomal dominant; Avascular necrosis of the femoral head, 608805 (3), Autosomal dominant; Legg-Calve-Perthes disease, 150600 (3), Autosomal dominant
COL9A1	120210	Stickler syndrome, type IV, 614134 (3); ?Epiphyseal dysplasia, multiple, 6, 614135 (3), Autosomal dominant
COQ2	609825	{Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal dominant, Autosomal recessive; Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive
COQ5	616359	?Coenzyme Q10 deficiency, primary, 9, 619028 (3), Autosomal recessive
CRX	602225	Leber congenital amaurosis 7, 613829 (3); Cone-rod retinal dystrophy-2, 120970 (3), Autosomal dominant
CSPP1	611654	Joubert syndrome 21, 615636 (3), Autosomal recessive
CTC1	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
CTNNA1	116805	Macular dystrophy, patterned, 2, 608970 (3), Autosomal dominant
CTNNB1	116806	Exudative vitreoretinopathy 7, 617572 (3), Autosomal dominant; Pilomatricoma, somatic, 132600 (3); Colorectal cancer, somatic, 114500 (3); Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 (3), Autosomal dominant; Medulloblastoma, somatic, 155255 (3); Ovarian cancer, somatic, 167000 (3); Hepatocellular carcinoma, somatic, 114550 (3)
CTSD	116840	Ceroid lipofuscinosis, neuronal, 10, 610127 (3), Autosomal recessive
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	Developmental delay and seizures with or without movement abnormalities, 617836 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1bb, 613861 (3), Autosomal recessive; Retinitis pigmentosa 59, 613861 (3), Autosomal recessive
DHX38	605584	Retinitis pigmentosa 84, 618220 (3), Autosomal recessive
DMD	300377	Becker muscular dystrophy, 300376 (3), X-linked recessive; Cardiomyopathy, dilated, 3B, 302045 (3), X-linked; Duchenne muscular dystrophy, 310200 (3), X-linked recessive
DRAM2	613360	Cone-rod dystrophy 21, 616502 (3), Autosomal recessive
DYNC2H1	603297	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091 (3), Digenic recessive, Autosomal recessive
DYNC2I2	613363	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633 (3), Autosomal 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, Autosomal recessive
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
ENSA	603061	No OMIM phenotype
ERCC6	609413	UV-sensitive syndrome 1, 600630 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; ?De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; {Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); Premature ovarian failure 11, 616946 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Autosomal dominant, Somatic mutation
ESPN	606351	Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006 (3), Autosomal recessive; Deafness, autosomal recessive 36, 609006 (3), Autosomal recessive; ?Usher syndrome, type 1M, 618632 (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	Cutis laxa, autosomal recessive, type IA, 219100 (3), Autosomal recessive; Charcot-Marie-Tooth disease, demyelinating, type 1H, 619764 (3), Autosomal dominant; Macular degeneration, age-related, 3, 608895 (3), Autosomal dominant; Neuropathy, hereditary, with or without age-related macular degeneration,

		608895 (3), Autosomal dominant; ?Cutis laxa, autosomal dominant 2, 614434 (3), Autosomal dominant
FBN2	612570	Macular degeneration, early-onset, 616118 (3), Autosomal dominant; Contractural arachnodactyly, congenital, 121050 (3), Autosomal dominant
FLVCR1	609144	Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive
FRMD7	300628	Nystagmus, infantile periodic alternating, X-linked, 310700 (3), X-linked; Nystagmus 1, congenital, X-linked, 310700 (3), X-linked
FSCN2	607643	Retinitis pigmentosa 30, 607921 (3)
FZD4	604579	Retinopathy of prematurity, 133780 (3), Autosomal dominant; Exudative vitreoretinopathy 1, 133780 (3), Autosomal dominant
GDF6	601147	Microphthalmia with coloboma 6, digenic, 613703 (3), Autosomal dominant; Microphthalmia, isolated 4, 613094 (3); Leber congenital amaurosis 17, 615360 (3), Autosomal recessive; Multiple synostoses syndrome 4, 617898 (3), Autosomal dominant; Klippel-Feil syndrome 1, autosomal dominant, 118100 (3), Autosomal dominant
GNAT1	139330	Night blindness, congenital stationary, autosomal dominant 3, 610444 (3), Autosomal dominant; Night blindness, congenital stationary, type 1G, 616389 (3), Autosomal recessive
GNAT2	139340	Achromatopsia 4, 613856 (3)
GNB3	139130	Night blindness, congenital stationary, type 1H, 617024 (3), Autosomal recessive; {Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial
GNPTG	607838	Mucolipidosis III gamma, 252605 (3), Autosomal recessive
GPR143	300808	Ocular albinism, type I, Nettleship-Falls type, 300500 (3), X-linked; Nystagmus 6, congenital, X-linked, 300814 (3), X-linked 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
GRN	138945	Aphasia, primary progressive, 607485 (3), Autosomal dominant; Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 (3), Autosomal dominant; Ceroid lipofuscinosis, neuronal, 11, 614706 (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 dominant, Autosomal recessive; ?Choroidal dystrophy, central areolar 1, 215500 (3), Autosomal dominant; Leber congenital amaurosis 1, 204000 (3), Autosomal recessive; Night blindness, congenital stationary, type 1I, 618555 (3), Autosomal recessive
HGSNAT	610453	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 (3), Autosomal recessive; Retinitis pigmentosa 73, 616544 (3), Autosomal recessive
HK1	142600	Retinitis pigmentosa 79, 617460 (3), Autosomal dominant; Neuropathy, hereditary motor and sensory, Russe type, 605285 (3), Autosomal recessive; Neurodevelopmental disorder with visual defects and brain anomalies, 618547 (3), Autosomal dominant; Hemolytic anemia due to hexokinase deficiency, 235700 (3), Autosomal recessive
HKDC1	617221	Retinitis pigmentosa 92, 619614 (3), Autosomal recessive
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); CARASIL syndrome, 600142 (3), Autosomal recessive; Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 (3), Autosomal dominant
IDH3A	601149	Retinitis pigmentosa 90, 619007 (3), Autosomal recessive
IDH3B	604526	Retinitis pigmentosa 46, 612572 (3), Autosomal recessive
IFT140	614620	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive; Retinitis pigmentosa 80, 617781 (3), Autosomal recessive
IFT172	607386	Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Bardet-Biedl syndrome 20, 619471 (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; ?Retinitis pigmentosa 81, 617871 (3), Autosomal recessive; Short-rib thoracic dysplasia 18 with polydactyly, 617866 (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	Retinitis pigmentosa 10, 180105 (3), Autosomal dominant; Leber congenital amaurosis 11, 613837 (3), Autosomal dominant
IMPG1	602870	Macular dystrophy, vitelliform, 4, 616151 (3), Autosomal dominant, Autosomal recessive; Retinitis pigmentosa 91, 153870 (3), Autosomal dominant

IMPG2	607056	Retinitis pigmentosa 56, 613581 (3), Autosomal recessive; Macular dystrophy, vitelliform, 5, 616152 (3), Autosomal dominant
INPP5E	613037	Joubert syndrome 1, 213300 (3), Autosomal recessive; Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 (3), Autosomal recessive
INVS	243305	Nephronophthisis 2, infantile, 602088 (3), Autosomal recessive
IQCB1	609237	Senior-Loken syndrome 5, 609254 (3), Autosomal recessive
ITM2B	603904	?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 (3), Autosomal dominant; Dementia, familial British, 176500 (3), Autosomal dominant; Dementia, familial Danish, 117300 (3), Autosomal dominant
JAG1	601920	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 (3), Autosomal dominant; Alagille syndrome 1, 118450 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
KCNJ13	603208	Snowflake vitreoretinal degeneration, 193230 (3), Autosomal dominant; Leber congenital amaurosis 16, 614186 (3), Autosomal recessive
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
LIG3	600940	Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780 (3), Autosomal recessive
LRAT	604863	Leber congenital amaurosis 14, 613341 (3), Autosomal recessive; Retinal dystrophy, early-onset severe, 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
LRP2	600073	Donnai-Barrow syndrome, 222448 (3), Autosomal recessive

LRP5	603506	Osteopetrosis, autosomal dominant 1, 607634 (3), Autosomal dominant; [Bone mineral density variability 1], 601884 (3), Autosomal dominant; {Osteoporosis}, 166710 (3), 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; Exudative vitreoretinopathy 4, 601813 (3), Autosomal dominant, Autosomal recessive; van Buchem disease, type 2, 607636 (3)
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
MED12	300188	Lujan-Fryns syndrome, 309520 (3), X-linked recessive; Ohdo syndrome, X-linked, 300895 (3), X-linked recessive; Hardikar syndrome, 301068 (3), X-linked dominant; Opitz-Kaveggia syndrome, 305450 (3), X-linked recessive
MERTK	604705	Retinitis pigmentosa 38, 613862 (3), Autosomal recessive
MFN2	608507	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 (3), Autosomal recessive; Hereditary motor and sensory neuropathy VIA, 601152 (3), Autosomal dominant
MFRP	606227	Microphthalmia, isolated 5, 611040 (3), Autosomal recessive; Nanophthalmos 2, 609549 (3)
MFSD8	611124	Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive
MIEF1	615497	No OMIM phenotype
MIR204	610942	?Retinal dystrophy and iris coloboma with or without cataract, 616722 (3), Autosomal dominant
MKKS	604896	McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive; Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive
MKS1	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive
MMACHC	609831	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive
MSTO1	617619	Myopathy, mitochondrial, and ataxia, 617675 (3), Autosomal dominant, Autosomal recessive
MTRFR	613541	Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive
MTTP	157147	{Metabolic syndrome, protection against}, 605552 (3), Autosomal dominant; Abetalipoproteinemia, 200100 (3), Autosomal recessive

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; Usher syndrome, type 1B, 276900 (3), Autosomal recessive; Deafness, autosomal dominant 11, 601317 (3), Autosomal dominant
NBAS	608025	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive; Infantile liver failure syndrome 2, 616483 (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	{Type 2 diabetes mellitus, susceptibility to}, 125853 (3), Autosomal dominant; Maturity-onset diabetes of the young 6, 606394 (3)
NMNAT1	608700	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260 (3), Autosomal recessive; Leber congenital amaurosis 9, 608553 (3), Autosomal recessive
NPHP1	607100	Joubert syndrome 4, 609583 (3), Autosomal recessive; Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive
NPHP3	608002	Nephronophthisis 3, 604387 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive; Meckel syndrome 7, 267010 (3), Autosomal recessive
NPHP4	607215	Senior-Loken syndrome 4, 606996 (3), Autosomal recessive; Nephronophthisis 4, 606966 (3), Autosomal recessive
NR2E3	604485	Retinitis pigmentosa 37, 611131 (3), Autosomal dominant, Autosomal recessive; Enhanced S-cone syndrome, 268100 (3), Autosomal recessive
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	Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; Joubert syndrome 10, 300804 (3), X-linked recessive

OPA1	605290	Optic atrophy plus syndrome, 125250 (3), Autosomal dominant; {Glaucoma, normal tension, susceptibility to}, 606657 (3); Optic atrophy 1, 165500 (3), Autosomal dominant; Behr syndrome, 210000 (3), Autosomal recessive; ?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	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 (3), Autosomal dominant; Pituitary hormone deficiency, combined, 6, 613986 (3), Autosomal dominant; Microphthalmia, syndromic 5, 610125 (3), Autosomal dominant
P3H2	610341	Myopia, high, with cataract and vitreoretinal degeneration, 614292 (3), Autosomal recessive
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), Digenic recessive, Autosomal recessive; Deafness, autosomal recessive 23, 609533 (3), Autosomal recessive; Usher syndrome, type 1F, 602083 (3), Autosomal recessive
PCYT1A	123695	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940 (3), Autosomal recessive
PDE6A	180071	Retinitis pigmentosa 43, 613810 (3)
PDE6B	180072	Retinitis pigmentosa-40, 613801 (3), Autosomal recessive; Night blindness, congenital stationary, autosomal dominant 2, 163500 (3), Autosomal dominant
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 dominant, Autosomal recessive; Achromatopsia 6, 610024 (3), Autosomal dominant, Autosomal recessive
PDSS1	607429	Coenzyme Q10 deficiency, primary, 2, 614651 (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 dominant, Autosomal recessive; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive; Heimler syndrome 2, 616617 (3), Autosomal recessive
PEX7	601757	Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive; Peroxisome biogenesis disorder 9B, 614879 (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; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive; Boucher-Neuhauser syndrome, 215470 (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
PPT1	600722	Ceroid lipofuscinosis, neuronal, 1, 256730 (3), Autosomal recessive
PRCD	610598	Retinitis pigmentosa 36, 610599 (3)
PRDM13	616741	Pontocerebellar hypoplasia, type 17, 619909 (3), Autosomal recessive; Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761 (3), Autosomal recessive
PROM1	604365	Macular dystrophy, retinal, 2, 608051 (3), Autosomal dominant; Retinitis pigmentosa 41, 612095 (3), Autosomal recessive; Stargardt disease 4, 603786 (3), Autosomal dominant; Cone-rod

		dystrophy 12, 612657 (3), Autosomal dominant, Autosomal recessive
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; Choroidal dystrophy, central areolar 2, 613105 (3), Autosomal dominant; Retinitis punctata albescens, 136880 (3), Autosomal dominant, Autosomal recessive; Leber congenital amaurosis 18, 608133 (3), Digenic dominant, Autosomal dominant, Autosomal recessive; Macular dystrophy, vitelliform, 3, 608161 (3), Autosomal dominant; Retinitis pigmentosa 7 and digenic form, 608133 (3), Digenic dominant, Autosomal dominant, Autosomal recessive
PRPS1	311850	Arts syndrome, 301835 (3), X-linked recessive; Phosphoribosylpyrophosphate synthetase superactivity, 300661 (3), X-linked recessive; Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 (3), X-linked recessive; Deafness, X-linked 1, 304500 (3), X-linked; Gout, PRPS-related, 300661 (3), X-linked recessive
RAB28	612994	Cone-rod dystrophy 18, 615374 (3), Autosomal recessive
RAX2	610362	Cone-rod dystrophy 11, 610381 (3), Autosomal dominant; ?Macular degeneration, age-related, 6, 613757 (3)
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 dominant, Autosomal recessive
RDH5	601617	Fundus albipunctatus, 136880 (3), Autosomal dominant, Autosomal recessive
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), Autosomal dominant; Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 (3), Autosomal dominant, Autosomal recessive; Retinitis punctata albescens, 136880 (3), Autosomal dominant, Autosomal recessive
RIMS2	606630	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970 (3), Autosomal recessive
RLBP1	180090	Bothnia retinal dystrophy, 607475 (3), Autosomal recessive; Newfoundland rod-cone dystrophy, 607476 (3); Retinitis punctata albescens, 136880 (3), Autosomal dominant, Autosomal recessive; Fundus albipunctatus, 136880 (3), Autosomal dominant, Autosomal recessive
RNU4ATAC	601428	Roifman syndrome, 616651 (3), Autosomal recessive; Lowry-Wood syndrome, 226960 (3), Autosomal recessive; Microcephalic osteodysplastic primordial dwarfism, type I, 210710 (3), Autosomal recessive
ROM1	180721	Retinitis pigmentosa 7, digenic form, 608133 (3), Digenic dominant, Autosomal dominant, Autosomal recessive
RP1	603937	Retinitis pigmentosa 1, 180100 (3), Autosomal dominant, Autosomal recessive
RP1L1	608581	Occult macular dystrophy, 613587 (3), Autosomal dominant; Retinitis pigmentosa 88, 618826 (3), Autosomal recessive
RP2	300757	Retinitis pigmentosa 2, 312600 (3), X-linked
RP9	607331	?Retinitis pigmentosa 9, 180104 (3), Autosomal dominant
RPE65	180069	Retinitis pigmentosa 20, 613794 (3), Autosomal recessive; Retinitis pigmentosa 87 with choroidal involvement, 618697 (3), Autosomal dominant; Leber congenital amaurosis 2, 204100 (3), Autosomal recessive
RPGR	312610	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 (3); Cone-rod dystrophy, X-linked, 1, 304020 (3), X-linked recessive; Retinitis pigmentosa 3, 300029 (3); Macular degeneration, X-linked atrophic, 300834 (3), X-linked recessive
RPGRIP1	605446	Cone-rod dystrophy 13, 608194 (3), Autosomal recessive; Leber congenital amaurosis 6, 613826 (3), Autosomal recessive
RPGRIP1L	610937	Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; ?COACH syndrome 3, 619113 (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	Retinitis pigmentosa 47, 613758 (3); Oguchi disease-1, 258100 (3), Autosomal recessive
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	Senior-Loken syndrome 7, 613615 (3), Autosomal recessive; Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive
SEMA4A	607292	Retinitis pigmentosa 35, 610282 (3), Autosomal dominant, Autosomal recessive; Cone-rod dystrophy 10, 610283 (3), Autosomal recessive
SF3B2	605591	Craniofacial microsomia, 164210 (3), Autosomal dominant
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; Pontocerebellar hypoplasia, type 1E, 619303 (3), Autosomal recessive
SLC37A3	619137	No OMIM phenotype
SLC4A7	603353	No OMIM phenotype
SLC66A1	614760	No OMIM phenotype
SLC6A6	186854	Hypotaurinemic retinal degeneration and cardiomyopathy, 145350 (3), Autosomal recessive
SLC7A14	615720	Retinitis pigmentosa 68, 615725 (3), Autosomal recessive
SNRNP200	601664	Retinitis pigmentosa 33, 610359 (3), Autosomal dominant
SPATA7	609868	Leber congenital amaurosis 3, 604232 (3), Autosomal recessive; Retinitis pigmentosa 94, variable age at onset, autosomal recessive, 604232 (3), Autosomal recessive
SPTLC1	605712	Neuropathy, hereditary sensory and autonomic, type IA, 162400 (3), Autosomal dominant
SRD5A3	611715	Kahrizi syndrome, 612713 (3), Autosomal recessive; Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive
SSBP1	600439	Optic atrophy 13 with retinal and foveal abnormalities, 165510 (3), Autosomal dominant
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
TINF2	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant
TLCD3B	615175	Cone-rod dystrophy 22, 619531 (3), Autosomal recessive
TLR3	603029	{HIV1 infection, resistance to}, 609423 (3); {Immunodeficiency 83, susceptibility to viral infections}, 613002 (3), Autosomal dominant, Autosomal recessive
TLR4	603030	Macular degeneration, age-related, 10, 611488 (2)
TMEM126A	612988	Optic atrophy 7, 612989 (3), Autosomal recessive
TMEM216	613277	Joubert syndrome 2, 608091 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive
TMEM218	619285	Joubert syndrome 39, 619562 (3), Autosomal recessive

TMEM231	614949	Joubert syndrome 20, 614970 (3), Autosomal recessive; Meckel syndrome 11, 615397 (3), Autosomal recessive
TMEM237	614423	Joubert syndrome 14, 614424 (3), Autosomal recessive
TOPORS	609507	Retinitis pigmentosa 31, 609923 (3)
TPP1	607998	Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive
TRAF3IP1	607380	Senior-Loken syndrome 9, 616629 (3), Autosomal recessive
TREX1	606609	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 (3), Autosomal dominant; Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal dominant, Autosomal recessive; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), 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
TUBB4B	602660	Leber congenital amaurosis with early-onset deafness, 617879 (3), Autosomal dominant
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	Leber congenital amaurosis 15, 613843 (3), Autosomal recessive; Retinitis pigmentosa 14, 600132 (3), Autosomal recessive
UNC119	604011	?Immunodeficiency 13, 615518 (3), Autosomal dominant; ?Cone-rod dystrophy (3)
USH1C	605242	Usher syndrome, type 1C, 276904 (3), Autosomal recessive; Deafness, autosomal recessive 18A, 602092 (3), Autosomal recessive
USH1G	607696	Usher syndrome, type 1G, 606943 (3), Autosomal recessive

USH2A	608400	Usher syndrome, type 2A, 276901 (3), Autosomal recessive; Retinitis pigmentosa 39, 613809 (3)
USP45	618439	?Leber congenital amaurosis 19, 618513 (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; Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive; ?Spermatogenic failure 72, 619867 (3), Autosomal recessive
WFS1	606201	Deafness, autosomal dominant 6/14/38, 600965 (3), Autosomal dominant; ?Cataract 41, 116400 (3), Autosomal dominant; Wolfram-like syndrome, autosomal dominant, 614296 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent, association with}, 125853 (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	Retinitis pigmentosa 72, 616469 (3), Autosomal recessive; ?Exudative vitreoretinopathy 6, 616468 (3), Autosomal dominant
ZNF423	604557	Nephronophthisis 14, 614844 (3), Autosomal dominant, Autosomal recessive; Joubert syndrome 19, 614844 (3), Autosomal dominant, 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: August 24, 2022

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.