

RetNet panel		
versie	V5 (259 genen)	Centrum voor Medische Genetica Gent
Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
<i>ABCA4</i>	601691	Cone-rod dystrophy 3, 604116 (3); Fundus flavimaculatus, 248200 (3), Autosomal recessive; {Macular degeneration, age-related, 2}, 153800 (3), Autosomal dominant; Retinal dystrophy, early-onset severe, 248200 (3), Autosomal recessive; Retinitis pigmentosa 19, 601718 (3), Autosomal recessive; Stargardt disease 1, 248200 (3), Autosomal recessive
<i>ABCC6</i>	603234	Arterial calcification, generalized, of infancy, 2, 614473 (3), Autosomal recessive; Pseudoxanthoma elasticum, 264800 (3), Autosomal recessive; Pseudoxanthoma elasticum, forme fruste, 177850 (3), Autosomal dominant
<i>ABHD12</i>	613599	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 (3), Autosomal recessive
<i>ACBD5</i>	616618	No OMIM phenotype
<i>ADAM9</i>	602713	Cone-rod dystrophy 9, 612775 (3)
<i>ADAMTS18</i>	607512	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458 (3), Autosomal recessive
<i>ADGRA3 (GPR125)</i>	612303	No OMIM phenotype
<i>ADGRV1</i>	602851	?Febrile seizures, familial, 4, 604352 (3), Autosomal dominant; Usher syndrome, type 2C, 605472 (3), Autosomal recessive, Digenic dominant; Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 (3), Autosomal recessive, Digenic dominant
<i>ADIPOR1</i>	607945	No OMIM phenotype
<i>AGBL5</i>	615900	Retinitis pigmentosa 75, 617023 (3), Autosomal recessive
<i>AHI1</i>	608894	Joubert syndrome 3, 608629 (3), Autosomal recessive
<i>AIPL1</i>	604392	Cone-rod dystrophy, 604393 (3), Autosomal recessive; Leber congenital amaurosis 4, 604393 (3), Autosomal recessive; Retinitis pigmentosa, juvenile, 604393 (3), Autosomal recessive
<i>ALMS1</i>	606844	Alstrom syndrome, 203800 (3), Autosomal recessive
<i>ARL2BP</i>	615407	Retinitis pigmentosa with or without situs inversus, 615434 (3), Autosomal recessive
<i>ARL3</i>	604695	No OMIM phenotype
<i>ARL6</i>	608845	{Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Autosomal recessive, Digenic recessive; Bardet-Biedl syndrome 3, 600151 (3), Autosomal recessive; ?Retinitis pigmentosa 55, 613575 (3)
<i>ARMS2</i>	611313	{Macular degeneration, age-related, 8}, 613778 (3)

<i>ASRGL1</i>	No OMIM gene	No OMIM phenotype
<i>ATF6</i>	605537	Achromatopsia 7, 616517 (3), Autosomal recessive
<i>ATXN7</i>	607640	Spinocerebellar ataxia 7, 164500 (3), Autosomal dominant
<i>BBIP1</i>	613605	?Bardet-Biedl syndrome 18, 615995 (3), Autosomal recessive
<i>BBS1</i>	209901	Bardet-Biedl syndrome 1, 209900 (3), Autosomal recessive, Digenic recessive
<i>BBS10</i>	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
<i>BBS12</i>	610683	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive
<i>BBS2</i>	606151	Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive; Retinitis pigmentosa 74, 616562 (3), Autosomal recessive
<i>BBS4</i>	600374	Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive
<i>BBS5</i>	603650	Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive
<i>BBS7</i>	607590	Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive
<i>BBS9</i>	607968	Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive
<i>BEST1</i>	607854	Bestrophinopathy, autosomal recessive, 611809 (3); Macular dystrophy, vitelliform, 2, 153700 (3), Autosomal dominant; Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 (3), Autosomal dominant; Retinitis pigmentosa, concentric, 613194 (3); Retinitis pigmentosa-50, 613194 (3); Vitreoretinopathy, 193220 (3), Autosomal dominant
<i>C12orf65</i>	613541	Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive; Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive
<i>C1QTNF5</i>	608752	Retinal degeneration, late-onset, autosomal dominant, 605670 (3), Autosomal dominant
<i>C2</i>	613927	C2 deficiency, 217000 (3), Autosomal recessive; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3)
<i>C21orf2</i>	603191	Retinal dystrophy with macular staphyloma, 617547 (3), Autosomal recessive; Spondylometaphyseal dysplasia, axial, 602271 (3), Autosomal recessive
<i>C2orf71</i>	613425	Retinitis pigmentosa 54, 613428 (3)
<i>C3</i>	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)
<i>C8orf37</i>	614477	Bardet-Biedl syndrome 21, 617406 (3), Autosomal recessive; Cone-rod dystrophy 16, 614500 (3), Autosomal recessive; Retinitis pigmentosa 64, 614500 (3), Autosomal recessive
<i>CA4</i>	114760	Retinitis pigmentosa 17, 600852 (3), Autosomal dominant
<i>CABP4</i>	608965	Cone-rod synaptic disorder, congenital nonprogressive, 610427 (3), Autosomal recessive
<i>CACNA1F</i>	300110	Aland Island eye disease, 300600 (3), X-linked; Cone-rod dystrophy, X-linked, 3, 300476 (3), X-linked recessive; Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 (3), X-linked

<i>CACNA2D4</i>	608171	Retinal cone dystrophy 4, 610478 (3), Autosomal recessive
<i>CAPN5</i>	602537	Vitreoretinopathy, neovascular inflammatory, 193235 (3), Autosomal dominant
<i>CC2D2A</i>	612013	COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive
<i>CDH23</i>	605516	Deafness, autosomal recessive 12, 601386 (3), Autosomal recessive; {Pituitary adenoma, susceptibility to}, 617540 (3), Autosomal dominant; Usher syndrome, type 1D, 601067 (3), Autosomal recessive, Digenic recessive; Usher syndrome, type 1D/F digenic, 601067 (3), Autosomal recessive, Digenic recessive
<i>CDH3</i>	114021	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 (3), Autosomal recessive; Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 (3), Autosomal recessive
<i>CDHR1</i>	609502	Cone-rod dystrophy 15, 613660 (3), Autosomal recessive; Retinitis pigmentosa 65, 613660 (3), Autosomal recessive
<i>CEP164</i>	614848	Nephronophthisis 15, 614845 (3), Autosomal recessive
<i>CEP250</i>	609689	No OMIM phenotype
<i>CEP290</i>	610142	?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Joubert syndrome 5, 610188 (3), Autosomal recessive; Leber congenital amaurosis 10, 611755 (3); Meckel syndrome 4, 611134 (3), Autosomal recessive; Senior-Loken syndrome 6, 610189 (3), Autosomal recessive
<i>CERKL</i>	608381	Retinitis pigmentosa 26, 608380 (3)
<i>CFB</i>	138470	?Complement factor B deficiency, 615561 (3); {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 (3), Autosomal dominant; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3)
<i>CFH</i>	134370	Basal laminar drusen, 126700 (3), Autosomal dominant; Complement factor H deficiency, 609814 (3), Autosomal recessive, Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 (3), Autosomal recessive, Autosomal dominant; {Macular degeneration, age-related, 4}, 610698 (3)
<i>CHM</i>	300390	Choroideremia, 303100 (3), X-linked dominant
<i>CIB2</i>	605564	Deafness, autosomal recessive 48, 609439 (3), Autosomal recessive; Usher syndrome, type IJ, 614869 (3), Autosomal recessive
<i>CLN3</i>	607042	Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive
<i>CLRN1</i>	606397	Retinitis pigmentosa 61, 614180 (3); Usher syndrome, type 3A, 276902 (3), Autosomal recessive
<i>CLUAP1</i>	616787	No OMIM phenotype
<i>CNGA1</i>	123825	Retinitis pigmentosa 49, 613756 (3)
<i>CNGA3</i>	600053	Achromatopsia 2, 216900 (3), Autosomal recessive
<i>CNGB1</i>	600724	Retinitis pigmentosa 45, 613767 (3), Autosomal recessive

<i>CNGB3</i>	605080	Achromatopsia 3, 262300 (3), Autosomal recessive; Macular degeneration, juvenile, 248200 (3), Autosomal recessive
<i>CNNM4</i>	607805	Jalili syndrome, 217080 (3), Autosomal recessive
<i>COL11A1</i>	120280	Fibrochondrogenesis 1, 228520 (3), Autosomal recessive; {Lumbar disc herniation, susceptibility to}, 603932 (3); Marshall syndrome, 154780 (3), Autosomal dominant; Stickler syndrome, type II, 604841 (3), Autosomal dominant
<i>COL2A1</i>	120140	Achondrogenesis, type II or hypochondrogenesis, 200610 (3), Autosomal dominant; Avascular necrosis of the femoral head, 608805 (3), Autosomal dominant; Czech dysplasia, 609162 (3), Autosomal dominant; Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 (3), Autosomal dominant; Kniest dysplasia, 156550 (3), Autosomal dominant; Legg-Calve-Perthes disease, 150600 (3), Autosomal dominant; Osteoarthritis with mild chondrodysplasia, 604864 (3), Autosomal dominant; Platyspondylic skeletal dysplasia, Torrance type, 151210 (3), Autosomal dominant; SED congenita, 183900 (3), Autosomal dominant; SMED Strudwick type, 184250 (3), Autosomal dominant; Spondyloepiphyseal dysplasia, Stanescu type, 616583 (3), Autosomal dominant; Spondyloperipheral dysplasia, 271700 (3), Autosomal dominant; Stickler syndrome, type I, nonsyndromic ocular, 609508 (3), Autosomal dominant; Stickler syndrome, type I, 108300 (3), Autosomal dominant; Vitreoretinopathy with phalangeal epiphyseal dysplasia (3)
<i>COL9A1</i>	120210	?Epiphyseal dysplasia, multiple, 6, 614135 (3), Autosomal dominant; Stickler syndrome, type IV, 614134 (3)
<i>CRB1</i>	604210	Leber congenital amaurosis 8, 613835 (3); Pigmented paravenous chorioretinal atrophy, 172870 (3), Autosomal dominant; Retinitis pigmentosa-12, autosomal recessive, 600105 (3), Autosomal recessive
<i>CRX</i>	602225	Cone-rod retinal dystrophy-2, 120970 (3), Autosomal dominant; Leber congenital amaurosis 7, 613829 (3)
<i>CSPP1</i>	611654	Joubert syndrome 21, 615636 (3), Autosomal recessive
<i>CTNNA1</i>	116805	Macular dystrophy, patterned, 2, 608970 (3), Autosomal dominant
<i>CYP4V2</i>	608614	Bietti crystalline corneoretinal dystrophy, 210370 (3), Autosomal recessive
<i>DHDDS</i>	608172	Retinitis pigmentosa 59, 613861 (3), Autosomal recessive
<i>DHX38</i>	605584	No OMIM phenotype
<i>DMD</i>	300377	Becker muscular dystrophy, 300376 (3), X-linked recessive; Cardiomyopathy, dilated, 3B, 302045 (3), X-linked; Duchenne muscular dystrophy, 310200 (3), X-linked recessive
<i>DRAM2</i>	613360	Cone-rod dystrophy 21, 616502 (3), Autosomal recessive
<i>DTHD1</i>	616979	No OMIM phenotype
<i>EFEMP1</i>	601548	Doyme honeycomb degeneration of retina, 126600 (3), Autosomal dominant

<i>ELOVL4</i>	605512	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 (3), Autosomal recessive; Spinocerebellar ataxia 34, 133190 (3), Autosomal dominant; Stargardt disease 3, 600110 (3), Autosomal dominant
<i>EMC1</i>	616846	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875 (3), Autosomal recessive
<i>ERCC6</i>	609413	Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive; {Lung cancer, susceptibility to}, 211980 (3), Autosomal recessive; {Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); Premature ovarian failure 11, 616946 (3), Autosomal dominant; UV-sensitive syndrome 1, 600630 (3), Autosomal recessive
<i>EXOSC2</i>	602238	No OMIM phenotype
<i>EYS</i>	612424	Retinitis pigmentosa 25, 602772 (3), Autosomal recessive
<i>FAM161A</i>	613596	Retinitis pigmentosa 28, 606068 (3)
<i>FBLN5</i>	604580	Cutis laxa, autosomal dominant 2, 614434 (3); Cutis laxa, autosomal recessive, type IA, 219100 (3), Autosomal recessive; Macular degeneration, age-related, 3, 608895 (3), Autosomal dominant; Neuropathy, hereditary, with or without age-related macular degeneration, 608895 (3), Autosomal dominant
<i>FLVCR1</i>	609144	Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive
<i>FSCN2</i>	607643	Retinitis pigmentosa 30, 607921 (3)
<i>FZD4</i>	604579	Exudative vitreoretinopathy 1, 133780 (3), Autosomal dominant; Retinopathy of prematurity, 133780 (3), Autosomal dominant
<i>GDF6</i>	601147	Klippel-Feil syndrome 1, autosomal dominant, 118100 (3), Autosomal dominant; Leber congenital amaurosis 17, 615360 (3), Autosomal recessive; Microphthalmia with coloboma 6, digenic, 613703 (3), Autosomal dominant; Microphthalmia, isolated 4, 613094 (3)
<i>GNAT1</i>	139330	Night blindness, congenital stationary, autosomal dominant 3, 610444 (3), Autosomal dominant; ?Night blindness, congenital stationary, type 1G, 616389 (3), Autosomal recessive
<i>GNAT2</i>	139340	Achromatopsia 4, 613856 (3)
<i>GNB3</i>	139130	{Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial; Night blindness, congenital stationary, type 1H, 617024 (3), Autosomal recessive
<i>GNPTG</i>	607838	Mucopolipidosis III gamma, 252605 (3), Autosomal recessive
<i>GPR179</i>	614515	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565 (3), Autosomal recessive
<i>GRK1</i>	180381	Oguchi disease-2, 613411 (3)
<i>GRM6</i>	604096	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270 (3), Autosomal recessive

<i>GUCA1A</i>	600364	Cone dystrophy-3, 602093 (3), Autosomal dominant; Cone-rod dystrophy 14, 602093 (3), Autosomal dominant
<i>GUCA1B</i>	602275	Retinitis pigmentosa 48, 613827 (3)
<i>GUCY2D</i>	600179	Cone-rod dystrophy 6, 601777 (3), Autosomal dominant; Leber congenital amaurosis 1, 204000 (3), Autosomal recessive
<i>HARS</i>	142810	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 (3), Autosomal dominant; Usher syndrome type 3B, 614504 (3), Autosomal recessive
<i>HGSNAT</i>	610453	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 (3), Autosomal recessive; Retinitis pigmentosa 73, 616544 (3), Autosomal recessive
<i>HK1</i>	142600	Hemolytic anemia due to hexokinase deficiency, 235700 (3), Autosomal recessive; Neuropathy, hereditary motor and sensory, Russe type, 605285 (3), Autosomal recessive; Retinitis pigmentosa 79, 617460 (3), Autosomal dominant
<i>HMCN1</i>	608548	{Macular degeneration, age-related, 1}, 603075 (3), Autosomal dominant
<i>HMX1</i>	142992	Oculoauricular syndrome, 612109 (3), Autosomal recessive
<i>HTRA1</i>	602194	CARASIL syndrome, 600142 (3), Autosomal recessive; Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 (3), Autosomal dominant; {Macular degeneration, age-related, 7}, 610149 (3); {Macular degeneration, age-related, neovascular type}, 610149 (3)
<i>IDH3B</i>	604526	Retinitis pigmentosa 46, 612572 (3)
<i>IFT140</i>	614620	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive
<i>IFT172</i>	607386	Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive
<i>IFT27</i>	615870	?Bardet-Biedl syndrome 19, 615996 (3), Autosomal recessive
<i>IMPDH1</i>	146690	Leber congenital amaurosis 11, 613837 (3); Retinitis pigmentosa 10, 180105 (3), Autosomal dominant
<i>IMPG1</i>	602870	Macular dystrophy, vitelliform, 4, 616151 (3), Autosomal dominant
<i>IMPG2</i>	607056	Macular dystrophy, vitelliform, 5, 616152 (3), Autosomal dominant; Retinitis pigmentosa 56, 613581 (3), Autosomal recessive
<i>INPP5E</i>	613037	Joubert syndrome 1, 213300 (3), Autosomal recessive; Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 (3), Autosomal recessive
<i>INVS</i>	243305	Nephronophthisis 2, infantile, 602088 (3), Autosomal recessive
<i>IQCB1</i>	609237	Senior-Loken syndrome 5, 609254 (3), Autosomal recessive
<i>ITM2B</i>	603904	Dementia, familial British, 176500 (3), Autosomal dominant; Dementia, familial Danish, 117300 (3), Autosomal dominant; ?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 (3), Autosomal dominant

<i>JAG1</i>	601920	Alagille syndrome 1, 118450 (3), Autosomal dominant; ?Deafness, congenital heart defects, and posterior embryotoxon (3); Tetralogy of Fallot, 187500 (3), Autosomal dominant
<i>KCNJ13</i>	603208	Leber congenital amaurosis 16, 614186 (3), Autosomal recessive; Snowflake vitreoretinal degeneration, 193230 (3), Autosomal dominant
<i>KCNV2</i>	607604	Retinal cone dystrophy 3B, 610356 (3), Autosomal recessive
<i>KIAA1549</i>	613344	No OMIM phenotype
<i>KIF11</i>	148760	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950 (3), Autosomal dominant
<i>KIZ</i> (<i>PLK1S1</i>)	615757	Retinitis pigmentosa 69, 615780 (3), Autosomal recessive
<i>KLHL7</i>	611119	Cold-induced sweating syndrome 3, 617055 (3); Retinitis pigmentosa 42, 612943 (3), Autosomal dominant
<i>LAMA1</i>	150320	Poretti-Boltshauser syndrome, 615960 (3), Autosomal recessive
<i>LCA5</i>	611408	Leber congenital amaurosis 5, 604537 (3)
<i>LRAT</i>	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
<i>LRIT3</i>	615004	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058 (3), Autosomal recessive
<i>LRP5</i>	603506	[Bone mineral density variability 1], 601884 (3), Autosomal dominant; Exudative vitreoretinopathy 4, 601813 (3), Autosomal recessive, Autosomal dominant; Hyperostosis, endosteal, 144750 (3), Autosomal dominant; Osteopetrosis, autosomal dominant 1, 607634 (3), Autosomal dominant; Osteoporosis-pseudoglioma syndrome, 259770 (3), Autosomal recessive; {Osteoporosis}, 166710 (3), Autosomal dominant; Osteosclerosis, 144750 (3), Autosomal dominant; van Buchem disease, type 2, 607636 (3), Autosomal dominant
<i>LZTFL1</i>	606568	Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive
<i>MAK</i>	154235	Retinitis pigmentosa 62, 614181 (3), Autosomal recessive
<i>MAPKAPK3</i>	602130	?Macular dystrophy, patterned, 3, 617111 (3), Autosomal dominant
<i>MERTK</i>	604705	Retinitis pigmentosa 38, 613862 (3), Autosomal recessive
<i>MFN2</i>	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
<i>MFRP</i>	606227	Microphthalmia, isolated 5, 611040 (3), Autosomal recessive; Nanophthalmos 2, 609549 (3)
<i>MFSD8</i>	611124	Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive; Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive
<i>MIR204</i>	610942	?Retinal dystrophy and iris coloboma with or without cataract, 616722 (3), Autosomal dominant

<i>MKKS</i>	604896	Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive; McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive
<i>MKS1</i>	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive
<i>MTTP</i>	157147	Abetalipoproteinemia, 200100 (3), Autosomal recessive; {Metabolic syndrome, protection against}, 605552 (3), Autosomal dominant
<i>MVK</i>	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Mevalonic aciduria, 610377 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant
<i>MYO7A</i>	276903	Deafness, autosomal dominant 11, 601317 (3), Autosomal dominant; Deafness, autosomal recessive 2, 600060 (3), Autosomal recessive; Usher syndrome, type 1B, 276900 (3), Autosomal recessive
<i>NBAS</i>	608025	Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive; Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive
<i>NDP</i>	300658	Exudative vitreoretinopathy 2, X-linked, 305390 (3); Norrie disease, 310600 (3), X-linked recessive
<i>NEK2</i>	604043	?Retinitis pigmentosa 67, 615565 (3), Autosomal recessive
<i>NEUROD1</i>	601724	{Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant; Maturity-onset diabetes of the young 6, 606394 (3)
<i>NMNAT1</i>	608700	Leber congenital amaurosis 9, 608553 (3), Autosomal recessive
<i>NPHP1</i>	607100	Joubert syndrome 4, 609583 (3), Autosomal recessive; Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive
<i>NPHP3</i>	608002	Meckel syndrome 7, 267010 (3), Autosomal recessive; Nephronophthisis 3, 604387 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive
<i>NPHP4</i>	607215	Nephronophthisis 4, 606966 (3), Autosomal recessive; Senior-Loken syndrome 4, 606996 (3), Autosomal recessive
<i>NR2E3</i>	604485	Enhanced S-cone syndrome, 268100 (3), Autosomal recessive; Retinitis pigmentosa 37, 611131 (3), Autosomal recessive, Autosomal dominant
<i>NR2F1</i>	132890	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722 (3), Autosomal dominant
<i>NRL</i>	162080	Retinal degeneration, autosomal recessive, clumped pigment type (3); Retinitis pigmentosa 27, 613750 (3), Autosomal dominant
<i>NYX</i>	300278	Night blindness, congenital stationary (complete), 1A, X-linked, 310500 (3), X-linked recessive
<i>OAT</i>	613349	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870 (3), Autosomal recessive

<i>OFD1</i>	300170	Joubert syndrome 10, 300804 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive
<i>OPA1</i>	605290	Behr syndrome, 210000 (3), Autosomal recessive; {Glaucoma, normal tension, susceptibility to}, 606657 (3); ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 (3); Optic atrophy 1, 165500 (3), Autosomal dominant; Optic atrophy plus syndrome, 125250 (3), Autosomal dominant
<i>OPA3</i>	606580	3-methylglutaconic aciduria, type III, 258501 (3), Autosomal recessive; Optic atrophy 3 with cataract, 165300 (3), Autosomal dominant
<i>OPN1LW</i>	300822	Blue cone monochromacy, 303700 (3), X-linked recessive; Colorblindness, protan, 303900 (3), X-linked
<i>OPN1MW</i>	300821	Blue cone monochromacy, 303700 (3), X-linked recessive; Colorblindness, deutan, 303800 (3), X-linked
<i>OPN1SW</i>	613522	Colorblindness, tritan, 190900 (3), Autosomal dominant
<i>OTX2</i>	600037	Microphthalmia, syndromic 5, 610125 (3), Autosomal dominant; Pituitary hormone deficiency, combined, 6, 613986 (3), Autosomal dominant; Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 (3), Autosomal dominant
<i>PANK2</i>	606157	HARP syndrome, 607236 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 1, 234200 (3), Autosomal recessive
<i>PAX2</i>	167409	Glomerulosclerosis, focal segmental, 7, 616002 (3), Autosomal dominant; Papillorenal syndrome, 120330 (3), Autosomal dominant
<i>PCDH15</i>	605514	Deafness, autosomal recessive 23, 609533 (3), Autosomal recessive; Usher syndrome, type 1D/F digenic, 601067 (3), Autosomal recessive, Digenic recessive; Usher syndrome, type 1F, 602083 (3), Autosomal recessive
<i>PCYT1A</i>	123695	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940 (3), Autosomal recessive
<i>PDE6A</i>	180071	Retinitis pigmentosa 43, 613810 (3)
<i>PDE6B</i>	180072	Night blindness, congenital stationary, autosomal dominant 2, 163500 (3), Autosomal dominant; Retinitis pigmentosa-40, 613801 (3), Autosomal recessive
<i>PDE6C</i>	600827	Cone dystrophy 4, 613093 (3), Autosomal recessive
<i>PDE6G</i>	180073	Retinitis pigmentosa 57, 613582 (3), Autosomal recessive
<i>PDE6H</i>	601190	Achromatopsia 6, 610024 (3), Autosomal recessive, Autosomal dominant; Retinal cone dystrophy 3, 610024 (3), Autosomal recessive, Autosomal dominant
<i>PDZD7</i>	612971	{Retinal disease in Usher syndrome type IIA, modifier of}, 276901 (3), Autosomal recessive; Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 (3), Autosomal recessive, Digenic dominant

<i>PEX1</i>	602136	Heimler syndrome 1, 234580 (3), Autosomal recessive; Peroxisome biogenesis disorder 1A (Zellweger), 214100 (3), Autosomal recessive; Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 (3), Autosomal recessive
<i>PEX2</i>	170993	Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive
<i>PEX7</i>	601757	Peroxisome biogenesis disorder 9B, 614879 (3); Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive
<i>PGK1</i>	311800	Phosphoglycerate kinase 1 deficiency, 300653 (3), X-linked recessive
<i>PHYH</i>	602026	Refsum disease, 266500 (3), Autosomal recessive
<i>PITPNM3</i>	608921	Cone-rod dystrophy 5, 600977 (3), Autosomal dominant
<i>PLA2G5</i>	601192	[Fleck retina, familial benign], 228980 (3), Autosomal recessive
<i>PLK4</i>	605031	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171 (3), Autosomal recessive
<i>PNPLA6</i>	603197	Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive
<i>POC1B</i>	614784	Cone-rod dystrophy 20, 615973 (3), Autosomal recessive
<i>POMGNT1</i>	606822	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 (3), Autosomal recessive; 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
<i>PRCD</i>	610598	Retinitis pigmentosa 36, 610599 (3)
<i>PRDM13</i>	616741	No OMIM phenotype
<i>PROM1</i>	604365	Cone-rod dystrophy 12, 612657 (3); Macular dystrophy, retinal, 2, 608051 (3), Autosomal dominant; Retinitis pigmentosa 41, 612095 (3), Autosomal recessive; Stargardt disease 4, 603786 (3)
<i>PRPF3</i>	607301	Retinitis pigmentosa 18, 601414 (3), Autosomal dominant
<i>PRPF31</i>	606419	Retinitis pigmentosa 11, 600138 (3), Autosomal dominant
<i>PRPF4</i>	607795	Retinitis pigmentosa 70, 615922 (3), Autosomal dominant
<i>PRPF6</i>	613979	Retinitis pigmentosa 60, 613983 (3), Autosomal dominant
<i>PRPF8</i>	607300	Retinitis pigmentosa 13, 600059 (3), Autosomal dominant

		Choroidal dystrophy, central areolar 2, 613105 (3), Autosomal dominant; Leber congenital amaurosis 18, 608133 (3), Autosomal recessive, Autosomal dominant; Macular dystrophy, patterned, 1, 169150 (3), Autosomal dominant; Macular dystrophy, vitelliform, 3, 608161 (3), Autosomal dominant; Retinitis pigmentosa 7 and digenic, 608133 (3), Autosomal recessive, Autosomal dominant; Retinitis punctata albescens, 136880 (3), Autosomal recessive, Autosomal dominant
<i>PRPH2</i>	179605	
<i>PRPS1</i>	311850	Arts syndrome, 301835 (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; Phosphoribosylpyrophosphate synthetase superactivity, 300661 (3), X-linked recessive
<i>RAB28</i>	612994	Cone-rod dystrophy 18, 615374 (3), Autosomal recessive
<i>RAX2</i>	610362	Cone-rod dystrophy 11, 610381 (3), Autosomal dominant; ?Macular degeneration, age-related, 6, 613757 (3)
<i>RB1</i>	614041	Bladder cancer, somatic, 109800 (3); Osteosarcoma, somatic, 259500 (3); Retinoblastoma, 180200 (3), Autosomal dominant, Somatic mutation; Retinoblastoma, trilateral, 180200 (3), Autosomal dominant, Somatic mutation; Small cell cancer of the lung, somatic, 182280 (3)
<i>RBP3</i>	180290	?Retinitis pigmentosa 66, 615233 (3), Autosomal recessive
<i>RBP4</i>	180250	Microphthalmia, isolated, with coloboma 10, 616428 (3), Autosomal dominant; Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147 (3), Autosomal recessive
<i>RCBTB1</i>	607867	Retinal dystrophy with or without extraocular anomalies, 617175 (3), Autosomal recessive
<i>RD3</i>	180040	Leber congenital amaurosis 12, 610612 (3), Autosomal recessive
<i>RDH11</i>	607849	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108 (3), Autosomal recessive
<i>RDH12</i>	608830	Leber congenital amaurosis 13, 612712 (3), Autosomal recessive
<i>RDH5</i>	601617	Fundus albipunctatus, 136880 (3), Autosomal recessive, Autosomal dominant
<i>RGR</i>	600342	Retinitis pigmentosa 44, 613769 (3)
<i>RGS9</i>	604067	Bradyopsia, 608415 (3)
<i>RGS9BP</i>	607814	Bradyopsia, 608415 (3)
<i>RHO</i>	180380	Night blindness, congenital stationary, autosomal dominant 1, 610445 (3); Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 (3), Autosomal recessive, Autosomal dominant; Retinitis punctata albescens, 136880 (3), Autosomal recessive, Autosomal dominant
<i>RIMS1</i>	606629	Cone-rod dystrophy 7, 603649 (3)

<i>RLBP1</i>	180090	Bothnia retinal dystrophy, 607475 (3), Autosomal recessive; Fundus albipunctatus, 136880 (3), Autosomal recessive, Autosomal dominant; Newfoundland rod-cone dystrophy, 607476 (3); Retinitis punctata albescens, 136880 (3), Autosomal recessive, Autosomal dominant
<i>ROM1</i>	180721	Retinitis pigmentosa 7, digenic, 608133 (3), Autosomal recessive, Autosomal dominant
<i>RP1</i>	603937	Retinitis pigmentosa 1, 180100 (3), Autosomal recessive, Autosomal dominant
<i>RP1L1</i>	608581	Occult macular dystrophy, 613587 (3), Autosomal dominant
<i>RP2</i>	300757	Retinitis pigmentosa 2, 312600 (3), X-linked
<i>RP9</i>	607331	?Retinitis pigmentosa 9, 180104 (3), Autosomal dominant
<i>RPE65</i>	180069	Leber congenital amaurosis 2, 204100 (3), Autosomal recessive; Retinitis pigmentosa 20, 613794 (3), Autosomal recessive
<i>RPGR</i>	312610	Cone-rod dystrophy, X-linked, 1, 304020 (3), X-linked; Macular degeneration, X-linked atrophic, 300834 (3), X-linked recessive; Retinitis pigmentosa 3, 300029 (3); Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 (3)
<i>RPGRIP1</i>	605446	Cone-rod dystrophy 13, 608194 (3); Leber congenital amaurosis 6, 613826 (3)
<i>RPGRIP1L</i>	610937	COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive
<i>RS1</i>	300839	Retinoschisis, 312700 (3), X-linked dominant
<i>RTN4IP1</i>	610502	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732 (3), Autosomal recessive
<i>SAG</i>	181031	Oguchi disease-1, 258100 (3), Autosomal recessive; Retinitis pigmentosa 47, 613758 (3)
<i>SDCCAG8</i>	613524	Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive; Senior-Loken syndrome 7, 613615 (3)
<i>SEMA4A</i>	607292	Cone-rod dystrophy 10, 610283 (3), Autosomal recessive; Retinitis pigmentosa 35, 610282 (3), Autosomal recessive, Autosomal dominant
<i>SLC24A1</i>	603617	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830 (3), Autosomal recessive
<i>SLC25A46</i>	610826	Neuropathy, hereditary motor and sensory, type VIB, 616505 (3), Autosomal recessive
<i>SLC7A14</i>	615720	Retinitis pigmentosa 68, 615725 (3), Autosomal recessive
<i>SNRNP200</i>	601664	Retinitis pigmentosa 33, 610359 (3), Autosomal dominant
<i>SPATA7</i>	609868	Leber congenital amaurosis 3, 604232 (3); Retinitis pigmentosa, juvenile, autosomal recessive, 604232 (3)
<i>SPP2</i>	602637	No OMIM phenotype
<i>TEAD1</i>	189967	Sveinsson chorioretinal atrophy, 108985 (3), Autosomal dominant
<i>TIMM8A</i>	300356	Mohr-Tranebjaerg syndrome, 304700 (3), X-linked recessive
<i>TIMP3</i>	188826	Sorsby fundus dystrophy, 136900 (3), Autosomal dominant

<i>TLR3</i>	603029	{HIV1 infection, resistance to}, 609423 (3); {Herpes simplex encephalitis, susceptibility to, 2}, 613002 (3)
<i>TLR4</i>	603030	No OMIM phenotype
<i>TMEM126A</i>	612988	Optic atrophy 7, 612989 (3), Autosomal recessive
<i>TMEM216</i>	613277	Joubert syndrome 2, 608091 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive
<i>TMEM237</i>	614423	Joubert syndrome 14, 614424 (3), Autosomal recessive
<i>TOPORS</i>	609507	Retinitis pigmentosa 31, 609923 (3)
<i>TREX1</i>	606609	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal recessive, Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Vasculopathy, retinal, with cerebral leukodystrophy, 192315 (3), Autosomal dominant
<i>TRIM32</i>	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, type 2H, 254110 (3), Autosomal recessive
<i>TRNT1</i>	612907	Retinitis pigmentosa and erythrocytic microcytosis, 616959 (3), Autosomal recessive; Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 (3), Autosomal recessive
<i>TRPM1</i>	603576	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216 (3)
<i>TSPAN12</i>	613138	Exudative vitreoretinopathy 5, 613310 (3), Autosomal dominant
<i>TTC8</i>	608132	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
<i>TTL5</i>	612268	Cone-rod dystrophy 19, 615860 (3), Autosomal recessive
<i>TTPA</i>	600415	Ataxia with isolated vitamin E deficiency, 277460 (3), Autosomal recessive
<i>TUB</i>	601197	?Retinal dystrophy and obesity, 616188 (3), Autosomal recessive
<i>TUBGCP4</i>	609610	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335 (3), Autosomal recessive
<i>TUBGCP6</i>	610053	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270 (3), Autosomal recessive
<i>TULP1</i>	602280	Leber congenital amaurosis 15, 613843 (3), Autosomal recessive; Retinitis pigmentosa 14, 600132 (3), Autosomal recessive
<i>UNC119</i>	604011	?Cone-rod dystrophy (3); ?Immunodeficiency 13, 615518 (3), Autosomal dominant
<i>USH1C</i>	605242	Deafness, autosomal recessive 18A, 602092 (3), Autosomal recessive; Usher syndrome, type 1C, 276904 (3), Autosomal recessive
<i>USH1G</i>	607696	Usher syndrome, type 1G, 606943 (3), Autosomal recessive
<i>USH2A</i>	608400	Retinitis pigmentosa 39, 613809 (3); Usher syndrome, type 2A, 276901 (3), Autosomal recessive
<i>VCAN</i>	118661	Wagner syndrome 1, 143200 (3), Autosomal dominant

<i>WDPCP</i>	613580	?Bardet-Biedl syndrome 15, 615992 (3), Autosomal recessive; ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 (3), Autosomal recessive
<i>WDR19</i>	608151	?Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; Nephronophthisis 13, 614377 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive
<i>WFS1</i>	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 syndrome, 222300 (3), Autosomal recessive; Wolfram-like syndrome, autosomal dominant, 614296 (3), Autosomal dominant
<i>WHRN</i> (<i>DFNB31</i>)	607928	Deafness, autosomal recessive 31, 607084 (3), Autosomal recessive; Usher syndrome, type 2D, 611383 (3), Autosomal recessive
<i>ZNF408</i>	616454	?Exudative vitreoretinopathy 6, 616468 (3), Autosomal dominant; Retinitis pigmentosa 72, 616469 (3), Autosomal recessive
<i>ZNF423</i>	604557	Joubert syndrome 19, 614844 (3), Autosomal recessive, Autosomal dominant; Nephronophthisis 14, 614844 (3), Autosomal recessive, Autosomal dominant
<i>ZNF513</i>	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: June 06, 2017

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.