

<b>RetNet panel</b>		
<b>versie</b>	v7 (324 genen)	Centrum voor Medische Genetica Gent
<b>Gene</b>	<b>OMIM gene ID</b>	<b>Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern</b>
<i>ABCA4</i>	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
<i>ABCC6</i>	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
<i>ABHD12</i>	613599	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 (3), Autosomal recessive
<i>ACBD5</i>	616618	Retinal dystrophy with leukodystrophy, 618863 (3), Autosomal recessive
<i>ACO2</i>	100850	?Optic atrophy 9, 616289 (3), Autosomal recessive; Infantile cerebellar-retinal degeneration, 614559 (3), Autosomal recessive
<i>ADAM9</i>	602713	Cone-rod dystrophy 9, 612775 (3), Autosomal recessive
<i>ADAMTS18</i>	607512	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458 (3), Autosomal recessive
<i>ADGRV1</i>	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
<i>ADIPOR1</i>	607945	No OMIM phenotype
<i>AFG3L2</i>	604581	Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Optic atrophy 12, 618977 (3), Autosomal dominant; Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant
<i>AGBL5</i>	615900	Retinitis pigmentosa 75, 617023 (3), Autosomal recessive
<i>AHI1</i>	608894	Joubert syndrome 3, 608629 (3), Autosomal recessive
<i>AHR</i>	600253	?Retinitis pigmentosa 85, 618345 (3), Autosomal recessive
<i>AIPL1</i>	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
<i>ALDH3A2</i>	609523	Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive
<i>ALMS1</i>	606844	Alstrom syndrome, 203800 (3), Autosomal recessive
<i>ALPK1</i>	607347	ROSAH syndrome, 614979 (3), Autosomal dominant

<i>AMACR</i>	604489	Alpha-methylacyl-CoA racemase deficiency, 614307 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 4, 214950 (3), Autosomal recessive
<i>ARHGEF18</i>	616432	Retinitis pigmentosa 78, 617433 (3), Autosomal recessive
<i>ARL13B</i>	608922	Joubert syndrome 8, 612291 (3), Autosomal recessive
<i>ARL2BP</i>	615407	Retinitis pigmentosa with or without situs inversus, 615434 (3), Autosomal recessive
<i>ARL3</i>	604695	Retinitis pigmentosa 83, 618173 (3), Autosomal dominant; Joubert syndrome 35, 618161 (3), Autosomal recessive
<i>ARL6</i>	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
<i>ARMS2</i>	611313	{Macular degeneration, age-related, 8}, 613778 (3)
<i>ARSG</i>	610008	Usher syndrome, type IV, 618144 (3), Autosomal recessive
<i>ASRGL1</i>	609212	No OMIM phenotype
<i>ATF6</i>	605537	Achromatopsia 7, 616517 (3), Autosomal recessive
<i>ATOH7</i>	609875	Persistent hyperplastic primary vitreous, autosomal recessive, 221900 (3), Autosomal recessive
<i>BBIP1</i>	613605	?Bardet-Biedl syndrome 18, 615995 (3), Autosomal recessive
<i>BBS1</i>	209901	Bardet-Biedl syndrome 1, 209900 (3), Digenic recessive, Autosomal 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	Retinitis pigmentosa 74, 616562 (3), Autosomal recessive; Bardet-Biedl syndrome 2, 615981 (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>BCOR</i>	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant
<i>BEST1</i>	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); Vitreoretinopathy, 193220 (3), Autosomal dominant; Bestrophinopathy, autosomal recessive, 611809 (3)
<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), Digenic dominant
<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>CA4</i>	114760	No OMIM phenotype

<i>CABP4</i>	608965	Cone-rod synaptic disorder, congenital nonprogressive, 610427 (3), Autosomal recessive
<i>CACNA1F</i>	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
<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 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
<i>CCT2</i>	605139	No OMIM phenotype
<i>CDH23</i>	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
<i>CDH3</i>	114021	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 (3), Autosomal recessive; Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 (3), Autosomal recessive
<i>CDHR1</i>	609502	Cone-rod dystrophy 15, 613660 (3), Autosomal recessive; Retinitis pigmentosa 65, 613660 (3), Autosomal recessive
<i>CEP162</i>	610201	No OMIM phenotype
<i>CEP164</i>	614848	Nephronophthisis 15, 614845 (3), Autosomal recessive
<i>CEP19</i>	615586	Morbid obesity and spermatogenic failure, 615703 (3), Autosomal recessive
<i>CEP250</i>	609689	Cone-rod dystrophy and hearing loss 2, 618358 (3), Autosomal recessive
<i>CEP290</i>	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
<i>CEP78</i>	617110	Cone-rod dystrophy and hearing loss, 617236 (3), Autosomal recessive
<i>CERKL</i>	608381	Retinitis pigmentosa 26, 608380 (3), Autosomal recessive
<i>CFAP20</i>	617906	No OMIM phenotype
<i>CFAP410</i>	603191	Retinal dystrophy with macular staphyloma, 617547 (3), Autosomal recessive; Spondylometaphyseal dysplasia, axial, 602271 (3), Autosomal recessive
<i>CFAP418</i>	614477	Retinitis pigmentosa 64, 614500 (3), Autosomal recessive; Cone-rod dystrophy 16, 614500 (3), Autosomal recessive; Bardet-Biedl syndrome 21, 617406 (3), Autosomal recessive

<i>CFB</i>	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
<i>CFH</i>	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
<i>CHM</i>	300390	Choroideremia, 303100 (3), X-linked
<i>CLCC1</i>	617539	Retinitis pigmentosa 32, 609913 (3), Autosomal recessive
<i>CLN3</i>	607042	Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive
<i>CLN5</i>	608102	Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive
<i>CLRN1</i>	606397	Usher syndrome, type 3A, 276902 (3), Autosomal recessive; Retinitis pigmentosa 61, 614180 (3)
<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
<i>CNNM4</i>	607805	Jalili syndrome, 217080 (3), Autosomal recessive
<i>COL11A1</i>	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)
<i>COL18A1</i>	120328	Knobloch syndrome, type 1, 267750 (3), Autosomal recessive; Glaucoma, primary closed-angle, 618880 (3), Autosomal dominant
<i>COL2A1</i>	120140	?Vitreo-retinopathy 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
<i>COL9A1</i>	120210	Stickler syndrome, type IV, 614134 (3); ?Epiphyseal dysplasia, multiple, 6, 614135 (3), Autosomal dominant

<i>COQ2</i>	609825	{Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal dominant, Autosomal recessive; Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive
<i>COQ5</i>	616359	?Coenzyme Q10 deficiency, primary, 9, 619028 (3), Autosomal recessive
<i>CRB1</i>	604210	Leber congenital amaurosis 8, 613835 (3), Autosomal recessive; Retinitis pigmentosa-12, 600105 (3), Autosomal recessive; Pigmented paravenous chorioretinal atrophy, 172870 (3), Autosomal dominant
<i>CRX</i>	602225	Leber congenital amaurosis 7, 613829 (3); Cone-rod retinal dystrophy-2, 120970 (3), Autosomal dominant
<i>CSPP1</i>	611654	Joubert syndrome 21, 615636 (3), Autosomal recessive
<i>CTC1</i>	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
<i>CTNNA1</i>	116805	Macular dystrophy, patterned, 2, 608970 (3), Autosomal dominant
<i>CTNNA1</i>	116806	Exudative vitreoretinopathy 7, 617572 (3), Autosomal dominant; Pilomatrixoma, 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)
<i>CTSD</i>	116840	Ceroid lipofuscinosis, neuronal, 10, 610127 (3), Autosomal recessive
<i>CWC27</i>	617170	Retinitis pigmentosa with or without skeletal anomalies, 250410 (3), Autosomal recessive
<i>CYP4V2</i>	608614	Bietti crystalline corneoretinal dystrophy, 210370 (3), Autosomal recessive
<i>DHDDS</i>	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
<i>DHX38</i>	605584	Retinitis pigmentosa 84, 618220 (3), Autosomal recessive
<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>DYNC2H1</i>	603297	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091 (3), Digenic recessive, Autosomal recessive
<i>DYNC2I2</i>	613363	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633 (3), Autosomal recessive
<i>EFEMP1</i>	601548	Doyme honeycomb degeneration of retina, 126600 (3), Autosomal dominant
<i>ELOVL1</i>	611813	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527 (3), Autosomal dominant, Autosomal recessive
<i>ELOVL4</i>	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

<i>ENSA</i>	603061	No OMIM phenotype
<i>ERCC6</i>	609413	UV-sensitive syndrome 1, 600630 (3), Autosomal recessive; Cerebroculofacioskeletal 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
<i>ESPN</i>	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
<i>EXOSC2</i>	602238	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763 (3), Autosomal recessive
<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 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
<i>FBN2</i>	612570	Macular degeneration, early-onset, 616118 (3), Autosomal dominant; Contractural arachnodactyly, congenital, 121050 (3), Autosomal dominant
<i>FLVCR1</i>	609144	Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive
<i>FRMD7</i>	300628	Nystagmus, infantile periodic alternating, X-linked, 310700 (3), X-linked; Nystagmus 1, congenital, X-linked, 310700 (3), X-linked
<i>FSCN2</i>	607643	Retinitis pigmentosa 30, 607921 (3)
<i>FZD4</i>	604579	Retinopathy of prematurity, 133780 (3), Autosomal dominant; Exudative vitreoretinopathy 1, 133780 (3), Autosomal dominant
<i>GDF6</i>	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
<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	Night blindness, congenital stationary, type 1H, 617024 (3), Autosomal recessive; {Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial

<i>GNPTG</i>	607838	Mucopolysaccharidose III gamma, 252605 (3), Autosomal recessive
<i>GPR143</i>	300808	Ocular albinism, type I, Nettleship-Falls type, 300500 (3), X-linked; Nystagmus 6, congenital, X-linked, 300814 (3), X-linked recessive
<i>GPR179</i>	614515	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565 (3), Autosomal recessive
<i>GRID2</i>	602368	Spinocerebellar ataxia, autosomal recessive 18, 616204 (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>GRN</i>	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
<i>GUCA1A</i>	600364	Cone-rod dystrophy 14, 602093 (3), Autosomal dominant; Cone dystrophy-3, 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, 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
<i>HGSNAT</i>	610453	Mucopolysaccharidose type IIIC (Sanfilippo C), 252930 (3), Autosomal recessive; Retinitis pigmentosa 73, 616544 (3), Autosomal recessive
<i>HK1</i>	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
<i>HKDC1</i>	617221	Retinitis pigmentosa 92, 619614 (3), Autosomal recessive
<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	{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
<i>IDH3A</i>	601149	Retinitis pigmentosa 90, 619007 (3), Autosomal recessive
<i>IDH3B</i>	604526	Retinitis pigmentosa 46, 612572 (3), Autosomal recessive
<i>IFT140</i>	614620	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive; Retinitis pigmentosa 80, 617781 (3), Autosomal recessive

<i>IFT172</i>	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
<i>IFT27</i>	615870	Bardet-Biedl syndrome 19, 615996 (3), Autosomal recessive
<i>IFT43</i>	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
<i>IFT81</i>	605489	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895 (3), Autosomal recessive
<i>IFT88</i>	600595	No OMIM phenotype
<i>IMPDH1</i>	146690	Retinitis pigmentosa 10, 180105 (3), Autosomal dominant; Leber congenital amaurosis 11, 613837 (3), Autosomal dominant
<i>IMPG1</i>	602870	Macular dystrophy, vitelliform, 4, 616151 (3), Autosomal dominant, Autosomal recessive; Retinitis pigmentosa 91, 153870 (3), Autosomal dominant
<i>IMPG2</i>	607056	Retinitis pigmentosa 56, 613581 (3), Autosomal recessive; Macular dystrophy, vitelliform, 5, 616152 (3), Autosomal dominant
<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	?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
<i>JAG1</i>	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
<i>KCNJ13</i>	603208	Snowflake vitreoretinal degeneration, 193230 (3), Autosomal dominant; Leber congenital amaurosis 16, 614186 (3), Autosomal recessive
<i>KCNV2</i>	607604	Retinal cone dystrophy 3B, 610356 (3), Autosomal recessive
<i>KIAA1549</i>	613344	Retinitis pigmentosa 86, 618613 (3), Autosomal recessive
<i>KIF11</i>	148760	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950 (3), Autosomal dominant
<i>KIF3B</i>	603754	Retinitis pigmentosa 89, 618955 (3), Autosomal dominant
<i>KIZ</i>	615757	Retinitis pigmentosa 69, 615780 (3), Autosomal recessive
<i>KLHL7</i>	611119	Retinitis pigmentosa 42, 612943 (3), Autosomal dominant; PERCHING syndrome, 617055 (3), Autosomal recessive
<i>LAMA1</i>	150320	Poretti-Boltshauser syndrome, 615960 (3), Autosomal recessive
<i>LAMB2</i>	150325	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 (3); Pierson syndrome, 609049 (3), Autosomal recessive



<i>LCA5</i>	611408	Leber congenital amaurosis 5, 604537 (3), Autosomal recessive
<i>LIG3</i>	600940	Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780 (3), Autosomal recessive
<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>LRP2</i>	600073	Donnai-Barrow syndrome, 222448 (3), Autosomal recessive
<i>LRP5</i>	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)
<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>MED12</i>	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
<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	Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive
<i>MIEF1</i>	615497	No OMIM phenotype
<i>MIR204</i>	610942	?Retinal dystrophy and iris coloboma with or without cataract, 616722 (3), Autosomal dominant
<i>MKKS</i>	604896	McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive; Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive
<i>MKS1</i>	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive
<i>MMACHC</i>	609831	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive

<i>MSTO1</i>	617619	Myopathy, mitochondrial, and ataxia, 617675 (3), Autosomal dominant, Autosomal recessive
<i>MTRFR</i>	613541	Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive
<i>MTTP</i>	157147	{Metabolic syndrome, protection against}, 605552 (3), Autosomal dominant; Abetalipoproteinemia, 200100 (3), Autosomal recessive
<i>MVK</i>	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant; Mevalonic aciduria, 610377 (3), Autosomal recessive
<i>MYO7A</i>	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
<i>NBAS</i>	608025	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive; Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive
<i>NDP</i>	300658	Exudative vitreoretinopathy 2, X-linked, 305390 (3), X-linked recessive, X-linked dominant; Norrie disease, 310600 (3), X-linked recessive
<i>NEK2</i>	604043	?Retinitis pigmentosa 67, 615565 (3), Autosomal recessive
<i>NEUROD1</i>	601724	{Type 2 diabetes mellitus, susceptibility to}, 125853 (3), Autosomal dominant; Maturity-onset diabetes of the young 6, 606394 (3)
<i>NMNAT1</i>	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
<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	Nephronophthisis 3, 604387 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive; Meckel syndrome 7, 267010 (3), Autosomal recessive
<i>NPHP4</i>	607215	Senior-Loken syndrome 4, 606996 (3), Autosomal recessive; Nephronophthisis 4, 606966 (3), Autosomal recessive
<i>NR2E3</i>	604485	Retinitis pigmentosa 37, 611131 (3), Autosomal dominant, Autosomal recessive; Enhanced S-cone syndrome, 268100 (3), Autosomal recessive
<i>NR2F1</i>	132890	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722 (3), Autosomal dominant
<i>NRL</i>	162080	Retinitis pigmentosa 27, 613750 (3), Autosomal dominant; Retinal degeneration, autosomal recessive, clumped pigment type (3)
<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	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
<i>OPA1</i>	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
<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	Colorblindness, deutan, 303800 (3), X-linked; Blue cone monochromacy, 303700 (3), X-linked recessive
<i>OPN1SW</i>	613522	Colorblindness, tritan, 190900 (3), Autosomal dominant
<i>OTX2</i>	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
<i>P3H2</i>	610341	Myopia, high, with cataract and vitreoretinal degeneration, 614292 (3), Autosomal recessive
<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>PCARE</i>	613425	Retinitis pigmentosa 54, 613428 (3)
<i>PCDH15</i>	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
<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	Retinitis pigmentosa-40, 613801 (3), Autosomal recessive; Night blindness, congenital stationary, autosomal dominant 2, 163500 (3), Autosomal dominant
<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	Retinal cone dystrophy 3, 610024 (3), Autosomal dominant, Autosomal recessive; Achromatopsia 6, 610024 (3), Autosomal dominant, Autosomal recessive
<i>PDSS1</i>	607429	Coenzyme Q10 deficiency, primary, 2, 614651 (3), Autosomal recessive

<i>PEX1</i>	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
<i>PEX2</i>	170993	Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive
<i>PEX6</i>	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
<i>PEX7</i>	601757	Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive; Peroxisome biogenesis disorder 9B, 614879 (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	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
<i>POC1B</i>	614784	Cone-rod dystrophy 20, 615973 (3), Autosomal recessive
<i>POC5</i>	617880	No OMIM phenotype
<i>POMGNT1</i>	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
<i>PPT1</i>	600722	Ceroid lipofuscinosis, neuronal, 1, 256730 (3), Autosomal recessive
<i>PRCD</i>	610598	Retinitis pigmentosa 36, 610599 (3)
<i>PRDM13</i>	616741	Pontocerebellar hypoplasia, type 17, 619909 (3), Autosomal recessive; Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761 (3), Autosomal recessive
<i>PROM1</i>	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
<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
<i>PRPH2</i>	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
<i>PRPS1</i>	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
<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>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 dominant, Autosomal recessive
<i>RDH5</i>	601617	Fundus albipunctatus, 136880 (3), Autosomal dominant, Autosomal recessive
<i>REEP6</i>	609346	Retinitis pigmentosa 77, 617304 (3), Autosomal recessive
<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), Autosomal dominant; Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 (3), Autosomal dominant, Autosomal recessive; Retinitis punctata albescens, 136880 (3), Autosomal dominant, Autosomal recessive
<i>RIMS2</i>	606630	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970 (3), Autosomal recessive

<i>RLBP1</i>	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
<i>RNU4ATAC</i>	601428	Roifman syndrome, 616651 (3), Autosomal recessive; Lowry-Wood syndrome, 226960 (3), Autosomal recessive; Microcephalic osteodysplastic primordial dwarfism, type I, 210710 (3), Autosomal recessive
<i>ROM1</i>	180721	Retinitis pigmentosa 7, digenic form, 608133 (3), Digenic dominant, Autosomal dominant, Autosomal recessive
<i>RP1</i>	603937	Retinitis pigmentosa 1, 180100 (3), Autosomal dominant, Autosomal recessive
<i>RP1L1</i>	608581	Occult macular dystrophy, 613587 (3), Autosomal dominant; Retinitis pigmentosa 88, 618826 (3), Autosomal recessive
<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	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
<i>RPGR</i>	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
<i>RPGRIP1</i>	605446	Cone-rod dystrophy 13, 608194 (3), Autosomal recessive; Leber congenital amaurosis 6, 613826 (3), Autosomal recessive
<i>RPGRIP1L</i>	610937	Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; ?COACH syndrome 3, 619113 (3), Autosomal recessive
<i>RS1</i>	300839	Retinoschisis, 312700 (3), X-linked recessive
<i>RTN4IP1</i>	610502	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732 (3), Autosomal recessive
<i>SAG</i>	181031	Retinitis pigmentosa 47, 613758 (3); Oguchi disease-1, 258100 (3), Autosomal recessive
<i>SAMD11</i>	616765	No OMIM phenotype
<i>SAMD7</i>	No OMIM gene	No OMIM phenotype
<i>SCAPER</i>	611611	Intellectual developmental disorder and retinitis pigmentosa, 618195 (3), Autosomal recessive
<i>SCLT1</i>	611399	No OMIM phenotype
<i>SDCCAG8</i>	613524	Senior-Loken syndrome 7, 613615 (3), Autosomal recessive; Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive
<i>SEMA4A</i>	607292	Retinitis pigmentosa 35, 610282 (3), Autosomal dominant, Autosomal recessive; Cone-rod dystrophy 10, 610283 (3), Autosomal recessive
<i>SF3B2</i>	605591	Craniofacial microsomia, 164210 (3), 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; Pontocerebellar hypoplasia, type 1E, 619303 (3), Autosomal recessive
<i>SLC37A3</i>	619137	No OMIM phenotype
<i>SLC4A7</i>	603353	No OMIM phenotype
<i>SLC66A1</i>	614760	No OMIM phenotype
<i>SLC6A6</i>	186854	Hypotaurinemic retinal degeneration and cardiomyopathy, 145350 (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), Autosomal recessive; Retinitis pigmentosa 94, variable age at onset, autosomal recessive, 604232 (3), Autosomal recessive
<i>SPTLC1</i>	605712	Neuropathy, hereditary sensory and autonomic, type IA, 162400 (3), Autosomal dominant
<i>SRD5A3</i>	611715	Kahrizi syndrome, 612713 (3), Autosomal recessive; Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive
<i>SSBP1</i>	600439	Optic atrophy 13 with retinal and foveal abnormalities, 165510 (3), Autosomal dominant
<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>TINF2</i>	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant
<i>TLCD3B</i>	615175	Cone-rod dystrophy 22, 619531 (3), Autosomal recessive
<i>TLR3</i>	603029	{HIV1 infection, resistance to}, 609423 (3); {Immunodeficiency 83, susceptibility to viral infections}, 613002 (3), Autosomal dominant, Autosomal recessive
<i>TLR4</i>	603030	Macular degeneration, age-related, 10, 611488 (2)
<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>TMEM218</i>	619285	Joubert syndrome 39, 619562 (3), Autosomal recessive
<i>TMEM231</i>	614949	Joubert syndrome 20, 614970 (3), Autosomal recessive; Meckel syndrome 11, 615397 (3), Autosomal recessive
<i>TMEM237</i>	614423	Joubert syndrome 14, 614424 (3), Autosomal recessive
<i>TOPORS</i>	609507	Retinitis pigmentosa 31, 609923 (3)
<i>TPP1</i>	607998	Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive
<i>TRAF3IP1</i>	607380	Senior-Loken syndrome 9, 616629 (3), Autosomal recessive

<i>TREX1</i>	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
<i>TRIM32</i>	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 (3), Autosomal recessive
<i>TRNT1</i>	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
<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>TLL5</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>TUBB4B</i>	602660	Leber congenital amaurosis with early-onset deafness, 617879 (3), Autosomal dominant
<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	?Immunodeficiency 13, 615518 (3), Autosomal dominant; ?Cone-rod dystrophy (3)
<i>USH1C</i>	605242	Usher syndrome, type 1C, 276904 (3), Autosomal recessive; Deafness, autosomal recessive 18A, 602092 (3), Autosomal recessive
<i>USH1G</i>	607696	Usher syndrome, type 1G, 606943 (3), Autosomal recessive
<i>USH2A</i>	608400	Usher syndrome, type 2A, 276901 (3), Autosomal recessive; Retinitis pigmentosa 39, 613809 (3)
<i>USP45</i>	618439	?Leber congenital amaurosis 19, 618513 (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	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
<i>WFS1</i>	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
<i>WHRN</i>	607928	Deafness, autosomal recessive 31, 607084 (3), Autosomal recessive; Usher syndrome, type 2D, 611383 (3), Autosomal recessive
<i>ZNF408</i>	616454	Retinitis pigmentosa 72, 616469 (3), Autosomal recessive; ?Exudative vitreoretinopathy 6, 616468 (3), Autosomal dominant
<i>ZNF423</i>	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.