RetNet

Gene panel

Gene panel information

Gene panel	**RetNet**
Version	8
Total genes	343
Activation date	Tuesday 14 january 2025
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ABCA4	99.3 %	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	98.57 %	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	99.98 %	613599	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 (3), Autosomal recessive
ACBD5	99.97 %	616618	Retinal dystrophy with leukodystrophy, 618863 (3), Autosomal recessive
ACO2	99.99 %	100850	Optic atrophy 9, 616289 (3), Autosomal dominant, Autosomal recessive; Infantile cerebellar-retinal degeneration, 614559 (3), Autosomal recessive
ADAM9	99.89 %	602713	Cone-rod dystrophy 9, 612775 (3), Autosomal recessive
ADAMTS18	99.99 %	607512	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458 (3), Autosomal recessive
ADGRV1	99.8 %	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	99.83 %	607945	*No OMIM phenotypes*
AFG3L2	99.97 %	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	99.98 %	615900	Retinitis pigmentosa 75, 617023 (3), Autosomal recessive
AGPAT3	100 %	614794	*No OMIM phenotypes*
AHI1	99.86 %	608894	Joubert syndrome 3, 608629 (3), Autosomal recessive
AHR	99.79 %	600253	?Retinitis pigmentosa 85, 618345 (3), Autosomal recessive
AIPL1	100 %	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	99.95 %	609523	Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive
ALMS1	99.9 %	606844	Alstrom syndrome, 203800 (3), Autosomal recessive
ALPK1	99.92 %	607347	ROSAH syndrome, 614979 (3), Autosomal dominant
AMACR	100 %	604489	Alpha-methylacyl-CoA racemase deficiency, 614307 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 4, 214950 (3), Autosomal recessive
ARHGEF18	99.97 %	616432	Retinitis pigmentosa 78, 617433 (3), Autosomal recessive

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ARL13B	99.53 %	608922	Joubert syndrome 8, 612291 (3), Autosomal recessive
ARL2BP	99.89 %	615407	Retinitis pigmentosa 82 with or without situs inversus, 615434 (3), Autosomal recessive
ARL3	99.98 %	604695	Retinitis pigmentosa 83, 618173 (3), Autosomal dominant; Joubert syndrome 35, 618161 (3), Autosomal recessive
ARL6	99.9 %	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	100 %	611313	{Macular degeneration, age-related, 8}, 613778 (3)
ARSG	100 %	610008	Usher syndrome, type IV, 618144 (3), Autosomal recessive
ASRGL1	99.84 %	609212	*No OMIM phenotypes*
ATF6	98.19 %	605537	Achromatopsia 7, 616517 (3), Autosomal recessive
ATOH7	100 %	609875	Persistent hyperplastic primary vitreous, autosomal recessive, 221900 (3), Autosomal recessive
BBIP1	99.99 %	613605	Bardet-Biedl syndrome 18, 615995 (3), Autosomal recessive
BBS1	100 %	209901	Bardet-Biedl syndrome 1, 209900 (3), Digenic recessive, Autosomal recessive
BBS10	99.98 %	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
BBS12	100 %	610683	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive
BBS2	99.9 %	606151	Retinitis pigmentosa 74, 616562 (3), Autosomal recessive; Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive
BBS4	99.88 %	600374	Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive
BBS5	99 %	603650	Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive
BBS7	99.42 %	607590	Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive
BBS9	99.75 %	607968	Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive
BCOR	99.97 %	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant
BEST1	99.86 %	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	100 %	608752	Retinal degeneration, late-onset, autosomal dominant, 605670 (3), Autosomal dominant
C2	99.99 %	613927	C2 deficiency, 217000 (3), Autosomal recessive; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3), Digenic dominant
C3	100 %	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	100 %	114760	*No OMIM phenotypes*
CABP4	100 %	608965	Cone-rod synaptic disorder, congenital nonprogressive, 610427 (3), Autosomal recessive
CACNA1F	99.94 %	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	99.99 %	608171	Retinal cone dystrophy 4, 610478 (3), Autosomal recessive
CAPN5	99.88 %	602537	Vitreoretinopathy, neovascular inflammatory, 193235 (3), Autosomal dominant

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CC2D2A	99.95 %	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	98.46 %	605139	*No OMIM phenotypes*
CDH23	99.95 %	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	99.98 %	114021	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 (3), Autosomal recessive; Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 (3), Autosomal recessive
CDHR1	99.72 %	609502	Macular dystrophy, retinal, 613660 (3), Autosomal recessive; Cone-rod dystrophy 15, 613660 (3), Autosomal recessive; Retinitis pigmentosa 65, 613660 (3), Autosomal recessive
CEP162	99.73 %	610201	*No OMIM phenotypes*
CEP164	99.99 %	614848	Nephronophthisis 15, 614845 (3), Autosomal recessive
CEP19	99.99 %	615586	Morbid obesity and spermatogenic failure, 615703 (3), Autosomal recessive
CEP250	99.93 %	609689	Cone-rod dystrophy and hearing loss 2, 618358 (3), Autosomal recessive
CEP290	98.1 %	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	99.95 %	617110	Cone-rod dystrophy and hearing loss, 617236 (3), Autosomal recessive
CERKL	99.91 %	608381	Retinitis pigmentosa 26, 608380 (3), Autosomal recessive
CFAP20	99.96 %	617906	*No OMIM phenotypes*
CFAP410	100 %	603191	Retinal dystrophy with macular staphyloma, 617547 (3), Autosomal recessive; Spondylometaphyseal dysplasia, axial, 602271 (3), Autosomal recessive
CFAP418	100 %	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	99.97 %	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	99.12 %	134370	{Macular degeneration, age-related, 4}, 610698 (3), Autosomal dominant; 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	96.46 %	300390	Choroideremia, 303100 (3), X-linked
CLCC1	93.17 %	617539	Retinitis pigmentosa 32, 609913 (3), Autosomal recessive
CLCN2	100 %	600570	Leukoencephalopathy with ataxia, 615651 (3), Autosomal recessive; Hyperaldosteronism, familial, type II, 605635 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 (3), Autosomal dominant; {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 (3), Autosomal dominant
CLEC3B	99.94 %	187520	Macular dystrophy, retinal, 4, 619977 (3), Autosomal dominant
CLN3	99.92 %	607042	Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive
CLN5	100 %	608102	Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CLRN1	99.99 %	606397	Usher syndrome, type 3A, 276902 (3), Autosomal recessive; Retinitis pigmentosa 61, 614180 (3)
CLUAP1	99.95 %	616787	*No OMIM phenotypes*
CNGA1	99.6 %	123825	Retinitis pigmentosa 49, 613756 (3), Autosomal recessive
CNGA3	99.95 %	600053	Achromatopsia 2, 216900 (3), Autosomal recessive
CNGB1	97.53 %	600724	Retinitis pigmentosa 45, 613767 (3), Autosomal recessive
CNGB3	99.96 %	605080	Achromatopsia 3, 262300 (3), Autosomal recessive
CNNM4	99.93 %	607805	Jalili syndrome, 217080 (3), Autosomal recessive
COL11A1	90.72 %	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	99.99 %	120328	Knobloch syndrome, type 1, 267750 (3), Autosomal recessive; Glaucoma, primary closed-angle, 618880 (3), Autosomal dominant
COL2A1	99.87 %	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	99.91 %	120210	Stickler syndrome, type IV, 614134 (3), Autosomal recessive; ?Epiphyseal dysplasia, multiple, 6, 614135 (3), Autosomal dominant
COL9A2	98.76 %	120260	Epiphyseal dysplasia, multiple, 2, 600204 (3), Autosomal dominant; ?Stickler syndrome, type V, 614284 (3), Autosomal recessive
COL9A3	99.99 %	120270	{Intervertebral disc disease, susceptibility to}, 603932 (3); Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 (3), Autosomal dominant; Stickler syndrome, type VI, 620022 (3), Autosomal recessive
COQ2	99.9 %	609825	{Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal dominant, Autosomal recessive; Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive
COQ5	99.95 %	616359	?Coenzyme Q10 deficiency, primary, 9, 619028 (3), Autosomal recessive
COQ8B	99.94 %	615567	Nephrotic syndrome, type 9, 615573 (3), Autosomal recessive
CRB1	99.73 %	604210	Leber congenital amaurosis 8, 613835 (3), Autosomal recessive; Retinitis pigmentosa-12, 600105 (3), Autosomal recessive; Pigmented paravenous chorioretinal atrophy, 172870 (3), Autosomal dominant
CRX	99.96 %	602225	Leber congenital amaurosis 7, 613829 (3); Cone-rod retinal dystrophy-2, 120970 (3), Autosomal dominant
CSPP1	98.31 %	611654	Joubert syndrome 21, 615636 (3), Autosomal recessive
CTC1	100 %	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
CTNNA1	99.98 %	116805	Macular dystrophy, patterned, 2, 608970 (3), Autosomal dominant

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CTNNB1	99.95 %	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)
CTNND1	99.92 %	601045	Blepharocheilodontic syndrome 2, 617681 (3), Autosomal dominant
CTSD	100 %	116840	Ceroid lipofuscinosis, neuronal, 10, 610127 (3), Autosomal recessive
CWC27	99.67 %	617170	Retinitis pigmentosa with or without skeletal anomalies, 250410 (3), Autosomal recessive
CYP4V2	99.98 %	608614	Bietti crystalline corneoretinal dystrophy, 210370 (3), Autosomal recessive
DHDDS	98.65 %	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	99.97 %	605584	Retinitis pigmentosa 84, 618220 (3), Autosomal recessive
DMD	99.76 %	300377	Becker muscular dystrophy, 300376 (3), X-linked recessive; Cardiomyopathy, dilated, 3B, 302045 (3), X-linked; Duchenne muscular dystrophy, 310200 (3), X-linked recessive
DNAJC17	100 %	616844	*No OMIM phenotypes*
DRAM2	96.06 %	613360	Cone-rod dystrophy 21, 616502 (3), Autosomal recessive
DYNC2H1	99.66 %	603297	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091 (3), Digenic recessive, Autosomal recessive
DYNC2I2	99.98 %	613363	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633 (3), Autosomal recessive
EFEMP1	99.68 %	601548	Doyne honeycomb degeneration of retina, 126600 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type ID, 620780 (3), Autosomal recessive; Glaucoma 1, open angle, H, 611276 (3), Autosomal dominant
ELOVL1	100 %	611813	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527 (3), Autosomal dominant, Autosomal recessive
ELOVL4	99.91 %	605512	Spinocerebellar ataxia 34, 133190 (3), Autosomal dominant; Stargardt disease 3, 600110 (3), Autosomal dominant; Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457 (3), Autosomal recessive
ENSA	99.72 %	603061	*No OMIM phenotypes*
ERCC6	99.6 %	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), Somatic mutation, Autosomal dominant
ESPN	100 %	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	100 %	602238	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763 (3), Autosomal recessive
EYS	99.86 %	612424	Retinitis pigmentosa 25, 602772 (3), Autosomal recessive
FAM161A	99.77 %	613596	Retinitis pigmentosa 28, 606068 (3), Autosomal recessive

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FBLN5	100 %	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; ?Cutis laxa, autosomal dominant 2, 614434 (3), Autosomal dominant
FBN2	99.9 %	612570	Macular degeneration, early-onset, 616118 (3), Autosomal dominant; Contractural arachnodactyly, congenital, 121050 (3), Autosomal dominant
FLVCR1	99.91 %	609144	Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive
FRMD7	99.97 %	300628	Nystagmus, infantile periodic alternating, X-linked, 310700 (3), X-linked; Nystagmus 1, congenital, X-linked, 310700 (3), X-linked
FSCN2	100 %	607643	Retinitis pigmentosa 30, 607921 (3)
FZD4	100 %	604579	Retinopathy of prematurity, 133780 (3), Autosomal dominant; Exudative vitreoretinopathy 1, 133780 (3), Autosomal dominant
GDF6	100 %	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
GFAP	99.99 %	137780	Alexander disease, 203450 (3), Autosomal dominant
GNAT1	100 %	139330	Night blindness, congenital stationary, autosomal dominant 3, 610444 (3), Autosomal dominant; Night blindness, congenital stationary, type 1G, 616389 (3), Autosomal recessive
GNAT2	99.87 %	139340	Achromatopsia 4, 613856 (3)
GNB3	100 %	139130	Night blindness, congenital stationary, type 1H, 617024 (3), Autosomal recessive; {Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial
GNPTG	100 %	607838	Mucolipidosis III gamma, 252605 (3), Autosomal recessive
GPATCH11	99.79 %		*No OMIM phenotypes*
GPR143	99.6 %	300808	Ocular albinism, type I, Nettleship-Falls type, 300500 (3), X-linked; Nystagmus 6, congenital, X-linked, 300814 (3), X-linked recessive
GPR179	99.99 %	614515	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565 (3), Autosomal recessive
GRID2	99.97 %	602368	Spinocerebellar ataxia, autosomal recessive 18, 616204 (3), Autosomal recessive
GRK1	99.99 %	180381	Oguchi disease-2, 613411 (3)
GRM6	100 %	604096	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270 (3), Autosomal recessive
GRN	100 %	138945	Frontotemporal dementia 2, 607485 (3), Autosomal dominant, Autosomal recessive; Aphasia, primary progressive, 607485 (3), Autosomal dominant, Autosomal recessive; Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive
GUCA1A	100 %	600364	Cone-rod dystrophy 14, 602093 (3), Autosomal dominant; Cone dystrophy-3, 602093 (3), Autosomal dominant
GUCA1B	99.99 %	602275	Retinitis pigmentosa 48, 613827 (3), Autosomal dominant
GUCY2D	100 %	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	99.93 %	610453	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 (3), Autosomal recessive; Retinitis pigmentosa 73, 616544 (3), Autosomal recessive

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HK1	99.97 %	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	99.99 %	617221	Retinitis pigmentosa 92, 619614 (3), Autosomal recessive
HMCN1	99.44 %	608548	{Macular degeneration, age-related, 1}, 603075 (3), Autosomal dominant
HMX1	100 %	142992	Oculoauricular syndrome, 612109 (3), Autosomal recessive
HTRA1	100 %	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	99.98 %	601149	Retinitis pigmentosa 90, 619007 (3), Autosomal recessive
IDH3B	100 %	604526	Retinitis pigmentosa 46, 612572 (3), Autosomal recessive
IFT140	100 %	614620	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive; Retinitis pigmentosa 80, 617781 (3), Autosomal recessive
IFT172	99.98 %	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	100 %	615870	Bardet-Biedl syndrome 19, 615996 (3), Autosomal recessive
IFT43	99.97 %	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
IFT74	99.71 %	608040	Bardet-Biedl syndrome 22, 617119 (3), Autosomal recessive; Spermatogenic failure 58, 619585 (3), Autosomal recessive; Joubert syndrome 40, 619582 (3), Autosomal recessive
IFT81	94.64 %	605489	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895 (3), Autosomal recessive
IFT88	98.26 %	600595	*No OMIM phenotypes*
IMPDH1	92.77 %	146690	Retinitis pigmentosa 10, 180105 (3), Autosomal dominant; Leber congenital amaurosis 11, 613837 (3), Autosomal dominant
IMPG1	99.93 %	602870	Macular dystrophy, vitelliform, 4, 616151 (3), Autosomal dominant, Autosomal recessive; Retinitis pigmentosa 91, 153870 (3), Autosomal dominant
IMPG2	99.97 %	607056	Retinitis pigmentosa 56, 613581 (3), Autosomal recessive; Macular dystrophy, vitelliform, 5, 616152 (3), Autosomal dominant
INPP5E	99.85 %	613037	Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome, 610156 (3), Autosomal recessive; Joubert syndrome 1, 213300 (3), Autosomal recessive
INVS	99.94 %	243305	Nephronophthisis 2, infantile, 602088 (3), Autosomal recessive
IQCB1	99.72 %	609237	Senior-Loken syndrome 5, 609254 (3), Autosomal recessive
ITM2B	99.85 %	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	100 %	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	99.99 %	603208	Snowflake vitreoretinal degeneration, 193230 (3), Autosomal dominant; Leber congenital amaurosis 16, 614186 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
KCNV2	99.97 %	607604	Retinal cone dystrophy 3B, 610356 (3), Autosomal recessive
KIAA1549	99.98 %	613344	Retinitis pigmentosa 86, 618613 (3), Autosomal recessive
KIF11	99.84 %	148760	Microcephaly with or without chorioretinopathy, lymphedema, or impaired intellectual development, 152950 (3), Autosomal dominant
KIF3B	99.99 %	603754	Retinitis pigmentosa 89, 618955 (3), Autosomal dominant
KIZ	99.98 %	615757	Retinitis pigmentosa 69, 615780 (3), Autosomal recessive
KLHL7	99.95 %	611119	Retinitis pigmentosa 42, 612943 (3), Autosomal dominant; PERCHING syndrome, 617055 (3), Autosomal recessive
LAMA1	99.98 %	150320	Poretti-Boltshauser syndrome, 615960 (3), Autosomal recessive
LAMB2	99.99 %	150325	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 (3), Autosomal recessive; Pierson syndrome, 609049 (3), Autosomal recessive
LAMP2	98.95 %	309060	Danon disease, 300257 (3), X-linked dominant
LCA5	99.89 %	611408	Leber congenital amaurosis 5, 604537 (3), Autosomal recessive
LIG3	99.99 %	600940	Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780 (3), Autosomal recessive
LRAT	100 %	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	100 %	615004	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058 (3), Autosomal recessive
LRP2	99.86 %	600073	Donnai-Barrow syndrome, 222448 (3), Autosomal recessive
LRP5	99.95 %	603506	Osteopetrosis, autosomal dominant 1, 607634 (3), Autosomal dominant; [Bone mineral density variability 1], 601884 (3), Autosomal dominant; Polycystic liver disease 4 with or without kidney cysts, 617875 (3), Autosomal dominant; Endosteal hyperostosis, 144750 (3), Autosomal dominant; Osteoporosis-pseudoglioma syndrome, 259770 (3), Autosomal recessive; Exudative vitreoretinopathy 4, 601813 (3), Autosomal dominant, Autosomal recessive
LRRC32	100 %	137207	Cleft palate, proliferative retinopathy, and developmental delay, 619074 (3), Autosomal recessive
LZTFL1	100 %	606568	Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive
MAK	99.47 %	154235	Retinitis pigmentosa 62, 614181 (3), Autosomal recessive
MAPKAPK3	99.97 %	602130	?Macular dystrophy, patterned, 3, 617111 (3), Autosomal dominant
MED12	99.94 %	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	98.9 %	604705	Retinitis pigmentosa 38, 613862 (3), Autosomal recessive
MFRP	100 %	606227	Microphthalmia, isolated 5, 611040 (3), Autosomal recessive; Nanophthalmos 2, 609549 (3)
MFSD8	99.7 %	611124	Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive
MIEF1	99.99 %	615497	Optic atrophy 14, 620550 (3), Autosomal dominant
MIR204	100 %	610942	Retinal dystrophy and iris coloboma with or without cataract, 616722 (3), Autosomal dominant
MKKS	100 %	604896	McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive; Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive
MKS1	99.92 %	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MMACHC	99.98 %	609831	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive
MPDZ	99.86 %	603785	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219 (3), Autosomal recessive
MSTO1	76.34 %	617619	Myopathy, mitochondrial, and ataxia, 617675 (3), Autosomal dominant, Autosomal recessive
MTRFR	99.87 %	613541	Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive
MTTP	99.92 %	157147	Abetalipoproteinemia, 200100 (3), Autosomal recessive
MVK	99.97 %	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant; Mevalonic aciduria, 610377 (3), Autosomal recessive
MYO7A	99.96 %	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	99.86 %	608025	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive; Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive
NDP	99.98 %	300658	Exudative vitreoretinopathy 2, X-linked, 305390 (3), X-linked recessive, X-linked dominant; Norrie disease, 310600 (3), X-linked recessive
NEK2	95.09 %	604043	?Retinitis pigmentosa 67, 615565 (3), Autosomal recessive
NEUROD1	100 %	601724	{Type 2 diabetes mellitus, susceptibility to}, 125853 (3), Autosomal dominant; Maturity-onset diabetes of the young 6, 606394 (3)
NMNAT1	99.93 %	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	99.05 %	607100	Joubert syndrome 4, 609583 (3), Autosomal recessive; Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive
NPHP3	99.89 %	608002	Nephronophthisis 3, 604387 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive; Meckel syndrome 7, 267010 (3), Autosomal recessive
NPHP4	99.98 %	607215	Senior-Loken syndrome 4, 606996 (3), Autosomal recessive; Nephronophthisis 4, 606966 (3), Autosomal recessive
NR2E3	100 %	604485	Retinitis pigmentosa 37, 611131 (3), Autosomal dominant, Autosomal recessive; Enhanced S-cone syndrome, 268100 (3), Autosomal recessive
NRL	100 %	162080	Retinitis pigmentosa 27, 613750 (3), Autosomal dominant; Retinal degeneration, autosomal recessive, clumped pigment type (3)
NYX	100 %	300278	Night blindness, congenital stationary (complete), 1A, X-linked, 310500 (3), X-linked recessive
OAT	90.17 %	613349	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870 (3), Autosomal recessive
OFD1	99.68 %	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
OPN1LW	71.75 %	300822	Blue cone monochromacy, 303700 (3), X-linked recessive; Colorblindness, protan, 303900 (3), X-linked
OPN1MW	30.23 %	300821	Colorblindness, deutan, 303800 (3), X-linked; Blue cone monochromacy, 303700 (3), X-linked recessive
OPN1SW	99.97 %	613522	Colorblindness, tritan, 190900 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
OTX2	100 %	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	99.93 %	610341	Myopia, high, with cataract and vitreoretinal degeneration, 614292 (3), Autosomal recessive
PANK2	99.99 %	606157	Neurodegeneration with brain iron accumulation 1, 234200 (3), Autosomal recessive
PAX2	99.99 %	167409	Glomerulosclerosis, focal segmental, 7, 616002 (3), Autosomal dominant; Papillorenal syndrome, 120330 (3), Autosomal dominant
PCARE	100 %	613425	Retinitis pigmentosa 54, 613428 (3), Autosomal recessive
PCDH12	100 %	605622	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280 (3), Autosomal recessive
PCDH15	99.71 %	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	100 %	123695	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940 (3), Autosomal recessive; Lipodystrophy, congenital generalized, type 5, 620680 (3), Autosomal recessive
PDE6A	99.98 %	180071	Retinitis pigmentosa 43, 613810 (3), Autosomal recessive
PDE6B	100 %	180072	Retinitis pigmentosa-40, 613801 (3), Autosomal recessive; Night blindness, congenital stationary, autosomal dominant 2, 163500 (3), Autosomal dominant
PDE6C	99.91 %	600827	Cone dystrophy 4, 613093 (3), Autosomal recessive
PDE6G	100 %	180073	Retinitis pigmentosa 57, 613582 (3), Autosomal recessive
PDE6H	99.97 %	601190	Retinal cone dystrophy 3, 610024 (3), Autosomal dominant, Autosomal recessive; Achromatopsia 6, 610024 (3), Autosomal dominant, Autosomal recessive
PDSS1	95.7 %	607429	Coenzyme Q10 deficiency, primary, 2, 614651 (3), Autosomal recessive
PEX1	98.8 %	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	100 %	170993	Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive
PEX6	99.99 %	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	99.72 %	601757	Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive; Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive
PGK1	99.93 %	311800	Phosphoglycerate kinase 1 deficiency, 300653 (3), X-linked recessive
PHYH	100 %	602026	Refsum disease, 266500 (3), Autosomal recessive
PLA2G5	99.91 %	601192	[Fleck retina, familial benign], 228980 (3), Autosomal recessive
PLK4	99.89 %	605031	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171 (3), Autosomal recessive
PNPLA6	99.99 %	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	100 %	614784	Cone-rod dystrophy 20, 615973 (3), Autosomal recessive
POC5	99.85 %	617880	*No OMIM phenotypes*

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
POMGNT1	99.69 %	606822	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3, 613151 (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	97.48 %	600722	Ceroid lipofuscinosis, neuronal, 1, 256730 (3), Autosomal recessive
PRCD	100 %	610598	Retinitis pigmentosa 36, 610599 (3)
PRDM13	99.99 %	616741	Pontocerebellar hypoplasia, type 17, 619909 (3), Autosomal recessive; Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761 (3), Autosomal recessive
PROM1	99.97 %	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	99.73 %	607301	Retinitis pigmentosa 18, 601414 (3), Autosomal dominant
PRPF31	99.99 %	606419	Retinitis pigmentosa 11, 600138 (3), Autosomal dominant
PRPF4	99.96 %	607795	Retinitis pigmentosa 70, 615922 (3), Autosomal dominant
PRPF6	99.98 %	613979	Retinitis pigmentosa 60, 613983 (3), Autosomal dominant
PRPF8	99.98 %	607300	Retinitis pigmentosa 13, 600059 (3), Autosomal dominant
PRPH2	99.98 %	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	99.95 %	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
PYGM	99.96 %	608455	McArdle disease, 232600 (3), Autosomal recessive
RAB28	99.97 %	612994	Cone-rod dystrophy 18, 615374 (3), Autosomal recessive
RAX2	100 %	610362	Retinitis pigmentosa 95, 620102 (3), Autosomal recessive; Cone-rod dystrophy 11, 610381 (3), Autosomal dominant; ?Macular degeneration, age-related, 6, 613757 (3)
RBP3	99.99 %	180290	?Retinitis pigmentosa 66, 615233 (3), Autosomal recessive
RBP4	99.99 %	180250	Microphthalmia, isolated, with coloboma 10, 616428 (3), Autosomal dominant; Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147 (3), Autosomal recessive
RCBTB1	99.99 %	607867	Retinal dystrophy with or without extraocular anomalies, 617175 (3), Autosomal recessive
RD3	100 %	180040	Leber congenital amaurosis 12, 610612 (3), Autosomal recessive
RDH11	99.99 %	607849	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108 (3), Autosomal recessive
RDH12	99.98 %	608830	Leber congenital amaurosis 13, 612712 (3), Autosomal dominant, Autosomal recessive
RDH5	100 %	601617	Fundus albipunctatus, 136880 (3), Autosomal dominant, Autosomal recessive
REEP6	100 %	609346	Retinitis pigmentosa 77, 617304 (3), Autosomal recessive
RGR	99.79 %	600342	Retinitis pigmentosa 44, 613769 (3)

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
RGS9	99.9 %	604067	Prolonged electroretinal response suppression 1, 608415 (3), Autosomal recessive
RGS9BP	100 %	607814	Prolonged electroretinal response suppression 2, 620344 (3), Autosomal recessive
RHO	100 %	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	99.97 %	606630	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970 (3), Autosomal recessive
RLBP1	99.99 %	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	99.95 %	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	100 %	180721	Retinitis pigmentosa 7, digenic form, 608133 (3), Digenic dominant, Autosomal dominant, Autosomal recessive
RP1	99.98 %	603937	Retinitis pigmentosa 1, 180100 (3), Autosomal dominant, Autosomal recessive
RP1L1	100 %	608581	Occult macular dystrophy, 613587 (3), Autosomal dominant; Retinitis pigmentosa 88, 618826 (3), Autosomal recessive
RP2	99.58 %	300757	Retinitis pigmentosa 2, 312600 (3), X-linked
RP9	99.83 %	607331	?Retinitis pigmentosa 9, 180104 (3), Autosomal dominant
RPE65	98.99 %	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	94.45 %	312610	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 (3), X-linked; Cone-rod dystrophy, X-linked, 1, 304020 (3), X-linked recessive; Retinitis pigmentosa 3, 300029 (3), X-linked; Macular degeneration, X-linked atrophic, 300834 (3), X-linked recessive
RPGRIP1	99.95 %	605446	Cone-rod dystrophy 13, 608194 (3), Autosomal recessive; Leber congenital amaurosis 6, 613826 (3), Autosomal recessive
RPGRIP1L	96.35 %	610937	Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; ?COACH syndrome 3, 619113 (3), Autosomal recessive
RS1	99.92 %	300839	Retinoschisis, 312700 (3), X-linked recessive
RTN4IP1	99.98 %	610502	Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732 (3), Autosomal recessive
SAG	99.98 %	181031	Retinitis pigmentosa 47, autosomal recessive, 613758 (3), Autosomal recessive; Retinitis pigmentosa 96, autosomal dominant, 620228 (3), Autosomal dominant; Oguchi disease-1, 258100 (3), Autosomal recessive
SAMD11	100 %	616765	*No OMIM phenotypes*
SAMD7	99.99 %	620493	Macular dystrophy with or without cone dysfunction, 620762 (3), Autosomal recessive
SCAPER	99.75 %	611611	Intellectual developmental disorder and retinitis pigmentosa, 618195 (3), Autosomal recessive
SCLT1	95.17 %	611399	*No OMIM phenotypes*
SDCCAG8	100 %	613524	Senior-Loken syndrome 7, 613615 (3), Autosomal recessive; Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive
SEMA4A	99.81 %	607292	Retinitis pigmentosa 35, 610282 (3), Autosomal recessive; Cone-rod dystrophy 10, 610283 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SF3B2	99.98 %	605591	Craniofacial microsomia, 164210 (3), Autosomal dominant
SGSH	100 %	605270	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900 (3), Autosomal recessive
SLC24A1	99.92 %	603617	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830 (3), Autosomal recessive
SLC25A46	99.88 %	610826	Neuropathy, hereditary motor and sensory, type VIB, 616505 (3), Autosomal recessive; Pontocerebellar hypoplasia, type 1E, 619303 (3), Autosomal recessive
SLC37A3	99.97 %	619137	*No OMIM phenotypes*
SLC4A7	99.88 %	603353	*No OMIM phenotypes*
SLC66A1	99.95 %	614760	*No OMIM phenotypes*
SLC6A6	99.98 %	186854	Hypotaurinemic retinal degeneration and cardiomyopathy, 145350 (3), Autosomal recessive
SLC7A14	99.99 %	615720	Retinitis pigmentosa 68, 615725 (3), Autosomal recessive
SNRNP200	99.6 %	601664	Retinitis pigmentosa 33, 610359 (3), Autosomal dominant
SPATA7	99.8 %	609868	Leber congenital amaurosis 3, 604232 (3), Autosomal recessive; Retinitis pigmentosa 94, variable age at onset, autosomal recessive, 604232 (3), Autosomal recessive
SPTLC1	99.74 %	605712	Amyotrophic lateral sclerosis 27, juvenile, 620285 (3), Autosomal dominant; Neuropathy, hereditary sensory and autonomic, type IA, 162400 (3), Autosomal dominant
SRD5A3	99.94 %	611715	Kahrizi syndrome, 612713 (3), Autosomal recessive; Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive
SSBP1	100 %	600439	Optic atrophy 13 with retinal and foveal abnormalities, 165510 (3), Autosomal dominant
STN1	99.88 %	613128	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341 (3), Autosomal recessive
STX3	100 %	600876	Retinal dystrophy and microvillus inclusion disease, 619446 (3), Autosomal recessive; Diarrhea 12, with microvillus atrophy, 619445 (3), Autosomal recessive
SUMF1	99.95 %	607939	Multiple sulfatase deficiency, 272200 (3), Autosomal recessive
TBC1D32	99.75 %	615867	*No OMIM phenotypes*
TEAD1	99.97 %	189967	Sveinsson chorioretinal atrophy, 108985 (3), Autosomal dominant
THRB	99.82 %	190160	Thyroid hormone resistance, autosomal recessive, 274300 (3), Autosomal recessive; Thyroid hormone resistance, 188570 (3), Autosomal dominant; Thyroid hormone resistance, selective pituitary, 145650 (3), Autosomal dominant
TIMM8A	100 %	300356	Mohr-Tranebjaerg syndrome, 304700 (3), X-linked recessive
TIMP3	100 %	188826	Sorsby fundus dystrophy, 136900 (3), Autosomal dominant
TINF2	100 %	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant
TLCD3B	98.95 %	615175	Cone-rod dystrophy 22, 619531 (3), Autosomal recessive
TLR3	99.99 %	603029	{HIV1 infection, resistance to}, 609423 (3); {Immunodeficiency 83, susceptibility to viral infections}, 613002 (3), Autosomal dominant, Autosomal recessive
TLR4	99.99 %	603030	*No OMIM phenotypes*
TMEM216	99.98 %	613277	Joubert syndrome 2, 608091 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive
TMEM218	99.94 %	619285	Joubert syndrome 39, 619562 (3), Autosomal recessive
TMEM231	88.88 %	614949	Joubert syndrome 20, 614970 (3), Autosomal recessive; Meckel syndrome 11, 615397 (3), Autosomal recessive
TMEM237	99.3 %	614423	Joubert syndrome 14, 614424 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TOPORS	99.99 %	609507	Retinitis pigmentosa 31, 609923 (3), Autosomal dominant
TPP1	99.99 %	607998	Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive
TRAF3IP1	99.96 %	607380	Senior-Loken syndrome 9, 616629 (3), Autosomal recessive
TREX1	100 %	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	100 %	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 (3), Autosomal recessive
TRNT1	99.97 %	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	99.96 %	603576	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216 (3), Autosomal recessive
TSPAN12	99.89 %	613138	Exudative vitreoretinopathy 5, 613310 (3), Autosomal dominant
TTC21B	99.5 %	612014	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive; Nephronophthisis 12, 613820 (3), Autosomal dominant, Autosomal recessive
TTC8	99.67 %	608132	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
TTLL5	99.95 %	612268	Cone-rod dystrophy 19, 615860 (3), Autosomal recessive
TTPA	99.88 %	600415	Ataxia with isolated vitamin E deficiency, 277460 (3), Autosomal recessive
TUB	100 %	601197	?Retinal dystrophy and obesity, 616188 (3), Autosomal recessive
TUBB4B	100 %	602660	Leber congenital amaurosis with early-onset deafness, 617879 (3), Autosomal dominant
TUBGCP4	99.8 %	609610	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335 (3), Autosomal recessive
TUBGCP6	100 %	610053	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270 (3), Autosomal recessive
TULP1	99.99 %	602280	Leber congenital amaurosis 15, 613843 (3), Autosomal recessive; Retinitis pigmentosa 14, 600132 (3), Autosomal recessive
UBAP1L	99.99 %		*No OMIM phenotypes*
UNC119	100 %	604011	Cone-rod dystrophy 24, 620342 (3), Autosomal dominant; ?Immunodeficiency 13, 615518 (3), Autosomal dominant
USH1C	99.99 %	605242	Usher syndrome, type 1C, 276904 (3), Autosomal recessive; Deafness, autosomal recessive 18A, 602092 (3), Autosomal recessive
USH1G	100 %	607696	Usher syndrome, type 1G, 606943 (3), Autosomal recessive
USH2A	99.88 %	608400	Usher syndrome, type 2A, 276901 (3), Autosomal recessive; Retinitis pigmentosa 39, 613809 (3), Autosomal recessive
USP45	99.56 %	618439	?Leber congenital amaurosis 19, 618513 (3), Autosomal recessive
VCAN	99.99 %	118661	Wagner syndrome 1, 143200 (3), Autosomal dominant
VPS13B	99.9 %	607817	Cohen syndrome, 216550 (3), Autosomal recessive
VWA8	99.8 %	617509	?Retinitis pigmentosa 97, 620422 (3), Autosomal dominant
WDPCP	99.87 %	613580	Bardet-Biedl syndrome 15, 615992 (3), Autosomal recessive; Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
WDR19	99.8 %	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
WHRN	99.96 %	607928	Deafness, autosomal recessive 31, 607084 (3), Autosomal recessive; Usher syndrome, type 2D, 611383 (3), Autosomal recessive
ZNF408	99.99 %	616454	Retinitis pigmentosa 72, 616469 (3), Autosomal recessive; ?Exudative vitreoretinopathy 6, 616468 (3), Autosomal dominant
ZNF423	98.94 %	604557	Nephronophthisis 14, 614844 (3), Autosomal dominant, Autosomal recessive; Joubert syndrome 19, 614844 (3), Autosomal dominant, Autosomal recessive

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2024-09-05**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

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.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.