Oslo universitetssykehus
Avdeling for medisinsk genetikk

Øyesykdommer

Genpanel, versjon v01

* Enkelte genomiske regioner har lav eller ingen sekvensdekning ved eksomsekvensering. Dette skyldes at de har stor likhet med andre områder i genomet, slik at spesifikk gjenkjennelse av disse områdene og påvisning av varianter i disse områdene, blir vanskelig og upålitelig. Disse genetiske regionene har vi identifisert ved å benytte USCS segmental duplication hvor områder større enn 1 kb og ≥90% likhet med andre regioner i genomet , gjenkjennes (https://genome.ucsc.edu).

For noen gener ligger alle ekson i områder med segmentale duplikasjoner, så disse vil ikke bli analysert: **ACTB, ACTG1, GJA1**

Vi gjør oppmerksom på at ved identifiseringav ekson oppstrøms for startkodon kan eksonnummereringen endres uten at transkript ID endres.

Avdelingens websider har en full oversikt over områder som er utelatt pga. segmentale duplikasjoner.

** Transkriptets kodende ekson.

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
ABCA4	34	NM_000350.2		1-50	Cone-rod dystrophy 3 OMIM Fundus flavimaculatus OMIM Retinal dystrophy, early-onset severe OMIM Retinitis pigmentosa 19 OMIM Stargardt disease 1 OMIM
ABCB6	47	NM_005689.3		1-19	Microphthalmia, isolated, with coloboma 7 OMIM
ABCC6	57	NM_001171.5	1-9	1-31	Pseudoxanthoma elasticum OMIM
ABHD12	15868	NM_001042472.2		1-13	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract OMIM

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ACO2	118	NM_001098.2		1-18	?Optic atrophy 9 OMIM Infantile cerebellar-retinal degeneration OMIM
ACTB	132	NM_001101.3	2-6	2-6	Baraitser-Winter syndrome 1 OMIM
ACTG1	144	NM_001614.3	2-6	2-6	Baraitser-Winter syndrome 2 OMIM
ADAM9	216	NM_003816.2		1-22	Cone-rod dystrophy 9 OMIM
ADAMTS10	13201	NM_030957.3		3-26	Weill-Marchesani syndrome 1, recessive OMIM
ADAMTS17	17109	NM_139057.3		1-22	Weill-Marchesani-like syndrome OMIM
ADAMTS18	17110	NM_199355.3		1-23	Microcornea, myopic chorioretinal atrophy, and telecanthus OMIM
ADAMTSL4	19706	NM_019032.5		3-19	Ectopia lentis et pupillae OMIM Ectopia lentis, isolated, autosomal recessive OMIM
AGBL1	26504	NM_152336.3		1-25	Corneal dystrophy, Fuchs endothelial, 8 OMIM
AGBL5	26147	NM_021831.5		2-15	Retinitis pigmentosa 75 OMIM
AGK	21869	NM_018238.3	16	2-16	Cataract 38, autosomal recessive OMIM Sengers syndrome OMIM
AHI1	21575	NM_017651.4		3-28	Joubert syndrome 3 OMIM
AIPL1	359	NM_014336.4		1-6	Cone-rod dystrophy OMIM Leber congenital amaurosis 4 OMIM Retinitis pigmentosa, juvenile OMIM
ALDH18A1	9722	NM_002860.3		2-18	Cutis laxa, autosomal dominant 3 OMIM Cutis laxa, autosomal recessive, type IIIA OMIM

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ALDH1A3	409	NM_000693.3		1-13	Microphthalmia, isolated 8 OMIM
ALG13	30881	NM_001099922.2		1-27	?Congenital disorder of glycosylation, type Is OMIM Epileptic encephalopathy, early infantile, 36 OMIM
ALG2	23159	NM_033087.3		1-2	?Congenital disorder of glycosylation, type Ii OMIM
ALG3	23056	NM_005787.5		1-9	Congenital disorder of glycosylation, type Id OMIM
ALMS1	428	NM_015120.4	17-21	1-23	Alstrom syndrome OMIM
AP3B1	566	NM_003664.4		1-27	Hermansky-Pudlak syndrome 2 OMIM
AP3D1	568	NM_001261826.1		1-32	?Hermansky-Pudlak syndrome 10 OMIM
ARHGEF18	17090	NM_015318.3		2-20	Retinitis pigmentosa 78 OMIM
ARL13B	25419	NM_182896.2		1-10	Joubert syndrome 8 OMIM
ARL2BP	17146	NM_012106.3		1-6	Retinitis pigmentosa with or without situs inversus OMIM
ARL6	13210	NM_177976.3		3-9	?Retinitis pigmentosa 55 OMIM Bardet-Biedl syndrome 3 OMIM
ASB10	17185	NM_001142459.1		1-5	Glaucoma 1, open angle, F OMIM
ATF6	791	NM_007348.3		1-16	Achromatopsia 7 OMIM
ATOH7	13907	NM_145178.3		1	Persistent hyperplastic primary vitreous, autosomal recessive OMIM
ATXN7	10560	NM_000333.3		3-13	Spinocerebellar ataxia 7 OMIM

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B3GALNT2	28596	NM_152490.4		1-12	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11 OMIM
B3GALTL	20207	NM_194318.3		1-15	Peters-plus syndrome OMIM
B3GNT1	15685	NM_006876.2		1-2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13 OMIM
BBIP1	28093	NM_001195306.1		2-4	?Bardet-Biedl syndrome 18 OMIM
BBS1	966	NM_024649.4		1-17	Bardet-Biedl syndrome 1 OMIM
BBS10	26291	NM_024685.3		1-2	Bardet-Biedl syndrome 10 OMIM
BBS12	26648	NM_152618.2		2	Bardet-Biedl syndrome 12 OMIM
BBS2	967	NM_031885.3		1-17	Bardet-Biedl syndrome 2 OMIM Retinitis pigmentosa 74 OMIM
BBS4	969	NM_033028.4		1-16	Bardet-Biedl syndrome 4 OMIM
BBS5	970	NM_152384.2		1-12	Bardet-Biedl syndrome 5 OMIM
BBS7	18758	NM_176824.2		1-19	Bardet-Biedl syndrome 7 OMIM
BBS9	30000	NM_198428.2		2-23	Bardet-Biedl syndrome 9 OMIM
BCOR	20893	NM_017745.5		2-15	Microphthalmia, syndromic 2 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
BEST1	12703	NM_004183.3		2-11	Bestrophinopathy, autosomal recessive OMIM Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma OMIM Retinitis pigmentosa, concentric OMIM Retinitis pigmentosa-50 OMIM Vitreoretinochoroidopathy OMIM
BFSP1	1040	NM_001195.4		1-8	Cataract 33, multiple types OMIM
BFSP2	1041	NM_003571.3		1-7	Cataract 12, multiple types OMIM
BLOC1S3	20914	NM_212550.4		2	Hermansky-Pudlak syndrome 8 OMIM
BLOC1S6	8549	NM_012388.3		1-5	?Hermansky-pudlak syndrome 9 OMIM
BMP4	1071	NM_001202.5		3-4	Microphthalmia, syndromic 6 OMIM
C10orf11	23405	NM_032024.4		1-6	Albinism, oculocutaneous, type VII OMIM
C12orf65	26784	NM_152269.4		2-3	Combined oxidative phosphorylation deficiency 7 OMIM
C1QTNF5	14344	NM_015645.4		14-15	Retinal degeneration, late-onset, autosomal dominant OMIM
C21orf2	1260	NM_004928.2		1-7	Retinal dystrophy with macular staphyloma OMIM Spondylometaphyseal dysplasia, axial OMIM
C8orf37	27232	NM_177965.3		1-6	Bardet-Biedl syndrome 21 OMIM Cone-rod dystrophy 16 OMIM Retinitis pigmentosa 64 OMIM
CA4	1375	NM_000717.4		1-8	Retinitis pigmentosa 17 OMIM
CABP4	1386	NM_145200.3		1-6	Cone-rod synaptic disorder, congenital nonprogressive OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
CACNA1F	1393	NM_005183.3		1-48	Aland Island eye disease OMIM Cone-rod dystrophy, X-linked, 3 OMIM Night blindness, congenital stationary (incomplete), 2A, X-linked OMIM
CACNA2D4	20202	NM_172364.4		1-38	Retinal cone dystrophy 4 OMIM
CAPN5	1482	NM_004055.4		2-13	Vitreoretinopathy, neovascular inflammatory OMIM
CBS	1550	NM_000071.2		3-17	Homocystinuria, B6-responsive and nonresponsive types OMIM
CC2D2A	29253	NM_001080522.2		3-38	COACH syndrome OMIM Joubert syndrome 9 OMIM
CDH23	13733	NM_022124.5		2-68	Usher syndrome, type 1D OMIM Usher syndrome, type 1D/F digenic OMIM
CDHR1	14550	NM_033100.3		1-17	Cone-rod dystrophy 15 OMIM Retinitis pigmentosa 65 OMIM
CEP104	24866	NM_014704.3		2-22	Joubert syndrome 25 OMIM
CEP164	29182	NM_014956.4		3-33	Nephronophthisis 15 OMIM
CEP290	29021	NM_025114.3	54	2-54	?Bardet-Biedl syndrome 14 OMIM Joubert syndrome 5 OMIM Leber congenital amaurosis 10 OMIM Meckel syndrome 4 OMIM Senior-Loken syndrome 6 OMIM
CEP78	25740	NM_032171.2		1-15	Cone-rod dystrophy and hearing loss OMIM
CERKL	21699	NM_001030311.2		1-14	Retinitis pigmentosa 26 OMIM
CHD7	20626	NM_017780.3		2-38	CHARGE syndrome OMIM
CHM	1940	NM_000390.3		1-15	Choroideremia OMIM

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CHMP4B	16171	NM_176812.4		1-5	Cataract 31, multiple types OMIM
CHN1	1943	NM_001822.5		1-13	Duane retraction syndrome 2 OMIM
CHRDL1	29861	NM_001143981.1		2-12	Megalocornea 1, X-linked OMIM
CHST6	6938	NM_021615.4		3	Macular corneal dystrophy OMIM
CIB2	24579	NM_006383.3		1-6	Usher syndrome, type IJ OMIM
CISD2	24212	NM_001008388.4	3	1-3	Wolfram syndrome 2 OMIM
CLDN19	2040	NM_148960.2		1-5	Hypomagnesemia 5, renal, with ocular involvement OMIM
CLN3	2074	NM_001042432.1		2-16	Ceroid lipofuscinosis, neuronal, 3 OMIM
CLN5	2076	NM_006493.2		1-4	Ceroid lipofuscinosis, neuronal, 5 OMIM
CLN6	2077	NM_017882.2		1-7	Ceroid lipofuscinosis, neuronal, 6 OMIM Ceroid lipofuscinosis, neuronal, Kufs type, adult onset OMIM
CLN8	2079	NM_018941.3		2-3	Ceroid lipofuscinosis, neuronal, 8 OMIM Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant OMIM
CLRN1	12605	NM_174878.2		1-3	Retinitis pigmentosa 61 OMIM Usher syndrome, type 3A OMIM
CNGA1	2148	NM_000087.3		4-11	Retinitis pigmentosa 49 OMIM
CNGA3	2150	NM_001298.2		2-8	Achromatopsia 2 OMIM
CNGB1	2151	NM_001297.4		2-33	Retinitis pigmentosa 45 OMIM

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CNGB3	2153	NM_019098.4		1-18	Achromatopsia 3 OMIM
CNNM4	105	NM_020184.3		1-7	Jalili syndrome OMIM
COL11A1	2186	NM_001854.3		1-67	Fibrochondrogenesis 1 OMIM Marshall syndrome OMIM Stickler syndrome, type II OMIM
COL18A1	2195	NM_130445.3		1-43	Knobloch syndrome, type 1 OMIM
COL2A1	2200	NM_001844.4		1-54	Stickler sydrome, type I, nonsyndromic ocular OMIM Stickler syndrome, type I OMIM Vitreoretinopathy with phalangeal epiphyseal dysplasia
COL4A1	2202	NM_001845.5		1-52	?Retinal arteries, tortuosity of OMIM
COL8A2	2216	NM_005202.3		3-4	Corneal dystrophy, Fuchs endothelial, 1 OMIM Corneal dystrophy, posterior polymorphous 2 OMIM
COL9A1	2217	NM_001851.4		1-38	?Epiphyseal dysplasia, multiple, 6 OMIM Stickler syndrome, type IV OMIM
COL9A2	2218	NM_001852.3		1-32	?Stickler syndrome, type V OMIM Epiphyseal dysplasia, multiple, 2 OMIM
CPAMD8	23228	NM_015692.3	16-17	1-42	Anterior segment dysgenesis 8 OMIM
CRB1	2343	NM_201253.2		1-12	Leber congenital amaurosis 8 OMIM Pigmented paravenous chorioretinal atrophy OMIM Retinitis pigmentosa-12, autosomal recessive OMIM
CRX	2383	NM_000554.5		2-4	Cone-rod retinal dystrophy-2 OMIM Leber congenital amaurosis 7 OMIM
CRYAA	2388	NM_000394.3		1-3	Cataract 9, multiple types OMIM

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CRYAB	2389	NM_001885.2		2-4	Cataract 16, multiple types OMIM
CRYBA1	2394	NM_005208.4		1-6	Cataract 10, multiple types OMIM
CRYBA2	2395	NM_057093.1		1-4	?Cataract 42 OMIM
CRYBA4	2396	NM_001886.2		2-6	Cataract 23 OMIM
CRYBB1	2397	NM_001887.3		2-6	Cataract 17, multiple types OMIM
CRYBB2	2398	NM_000496.2	4-6	2-6	Cataract 3, multiple types OMIM
CRYBB3	2400	NM_004076.4		2-6	Cataract 22 OMIM
CRYGB	2409	NM_005210.3		1-3	Cataract 39, multiple types, autosomal dominant OMIM
CRYGC	2410	NM_020989.3		1-3	Cataract 2, multiple types OMIM
CRYGD	2411	NM_006891.3		1-3	Cataract 4, multiple types OMIM
CRYGS	2417	NM_017541.3		1-3	Cataract 20, multiple types OMIM
CSPP1	26193	NM_024790.6		1-29	Joubert syndrome 21 OMIM
CTDP1	2498	NM_004715.4		1-13	Congenital cataracts, facial dysmorphism, and neuropathy OMIM
CTNNA1	2509	NM_001903.4		2-18	Macular dystrophy, patterned, 2 OMIM
CYP1B1	2597	NM_000104.3		2-3	Anterior segment dysgenesis 6, multiple subtypes OMIM Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset OMIM

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CYP27A1	2605	NM_000784.3		1-9	Cerebrotendinous xanthomatosis OMIM
CYP4V2	23198	NM_207352.3		1-11	Bietti crystalline corneoretinal dystrophy OMIM
DCN	2705	NM_001920.4		2-8	Corneal dystrophy, congenital stromal OMIM
DDX58	19102	NM_014314.3		1-18	Singleton-Merten syndrome 2 OMIM
DFNB31	16361	NM_015404.3		1-12	Usher syndrome, type 2D OMIM
DHCR7	2860	NM_001360.2		3-9	Smith-Lemli-Opitz syndrome OMIM
DHDDS	20603	NM_024887.3		2-9	Retinitis pigmentosa 59 OMIM
DNAI2	18744	NM_023036.4		2-13	Ciliary dyskinesia, primary, 9, with or without situs inversus OMIM
DPAGT1	2995	NM_001382.3		1-9	Congenital disorder of glycosylation, type Ij OMIM
DPM1	3005	NM_003859.2		1-9	Congenital disorder of glycosylation, type Ie OMIM
DPM2	3006	NM_003863.3		1-4	Congenital disorder of glycosylation, type Iu OMIM
DPYD	3012	NM_000110.3		1-23	Dihydropyrimidine dehydrogenase deficiency OMIM
DRAM2	28769	NM_178454.5		3-9	Cone-rod dystrophy 21 OMIM
DTNBP1	17328	NM_032122.4		1-10	Hermansky-Pudlak syndrome 7 OMIM
EBP	3133	NM_006579.2		2-5	Chondrodysplasia punctata, X-linked dominant OMIM MEND syndrome OMIM
EDN3	3178	NM_207034.2		1-5	Waardenburg syndrome, type 4B OMIM

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EFEMP1	3218	NM_001039348.2		3-12	Doyne honeycomb degeneration of retina OMIM
EIF2B2	3258	NM_014239.3		1-8	Leukoencephalopathy with vanishing white matter OMIM
ELOVL4	14415	NM_022726.3		1-6	Stargardt disease 3 OMIM
ELP4	1171	NM_019040.4		1-10	?Aniridia 2 OMIM
EMC1	28957	NM_015047.2		1-23	Cerebellar atrophy, visual impairment, and psychomotor retardation OMIM
EPG5	29331	NM_020964.2		1-44	Vici syndrome OMIM
EPHA2	3386	NM_004431.4		1-17	Cataract 6, multiple types OMIM
ERCC1	3433	NM_202001.2		1-8	Cerebrooculofacioskeletal syndrome 4 OMIM
ERCC2	3434	NM_000400.3		1-23	?Cerebrooculofacioskeletal syndrome 2 OMIM Trichothiodystrophy 1, photosensitive OMIM
ERCC3	3435	NM_000122.1		1-15	Trichothiodystrophy 2, photosensitive OMIM Xeroderma pigmentosum, group B OMIM
ERCC5	3437	NM_000123.3		1-15	Cerebrooculofacioskeletal syndrome 3 OMIM
ERCC6	3438	NM_000124.3		2-21	Cerebrooculofacioskeletal syndrome 1 OMIM Cockayne syndrome, type B OMIM De Sanctis-Cacchione syndrome OMIM
ERCC8	3439	NM_000082.3		1-12	Cockayne syndrome, type A OMIM
EYA1	3519	NM_000503.5		3-18	Anterior segment anomalies with or without cataract OMIM

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EYS	21555	NM_001142800.1	12	4-43	Retinitis pigmentosa 25 OMIM
FAM111A	24725	NM_022074.3		3-4	Gracile bone dysplasia OMIM Kenny-Caffey syndrome, type 2 OMIM
FAM126A	24587	NM_032581.3		2-11	Leukodystrophy, hypomyelinating, 5 OMIM
FAM161A	25808	NM_032180.2		1-6	Retinitis pigmentosa 28 OMIM
FBLN5	3602	NM_006329.3		1-11	Macular degeneration, age-related, 3 OMIM
FBN1	3603	NM_000138.4		2-66	Acromicric dysplasia OMIM Ectopia lentis, familial OMIM Marfan syndrome OMIM MASS syndrome OMIM
FKRP	17997	NM_024301.4		4	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5 OMIM Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5 OMIM
FKTN	3622	NM_001079802.1		3-11	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4 OMIM
FLVCR1	24682	NM_014053.3		1-10	Ataxia, posterior column, with retinitis pigmentosa OMIM
FOXC1	3800	NM_001453.2		1	Anterior segment dysgenesis 3, multiple subtypes OMIM Axenfeld-Rieger syndrome, type 3 OMIM
FOXC2	3801	NM_005251.2		1	Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus OMIM

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FOXE3	3808	NM_012186.2		1	Anterior segment dysgenesis 2, multiple subtypes OMIM Cataract 34, multiple types OMIM
FOXL2	1092	NM_023067.3		1	Blepharophimosis, epicanthus inversus, and ptosis, type 1 OMIM Blepharophimosis, epicanthus inversus, and ptosis, type 2 OMIM
FRAS1	19185	NM_025074.6		1-74	Fraser syndrome 1 OMIM
FREM1	23399	NM_144966.5		3-38	Manitoba oculotrichoanal syndrome OMIM
FRMD7	8079	NM_194277.2		1-12	Nystagmus 1, congenital, X-linked OMIM
FSCN2	3960	NM_001077182.2		1-5	Retinitis pigmentosa 30 OMIM
FTL	3999	NM_000146.3		1-4	Hyperferritinemia-cataract syndrome OMIM
FYCO1	14673	NM_024513.3		2-18	Cataract 18, autosomal recessive OMIM
FZD4	4042	NM_012193.3		1-2	Retinopathy of prematurity OMIM
GALK1	4118	NM_000154.1		1-8	Galactokinase deficiency with cataracts OMIM
GALT	4135	NM_000155.3		1-11	Galactosemia OMIM
GBA2	18986	NM_020944.2		1-17	Spastic paraplegia 46, autosomal recessive OMIM
GCNT2	4204	NM_001491.2		1-3	Cataract 13 with adult i phenotype OMIM
GDF3	4218	NM_020634.2		1-2	Microphthalmia with coloboma 6 OMIM Microphthalmia, isolated 7 OMIM

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GDF6	4221	NM_001001557.3		1-2	Leber congenital amaurosis 17 OMIM Microphthalmia with coloboma 6, digenic OMIM Microphthalmia, isolated 4 OMIM
GFER	4236	NM_005262.2		1-3	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay OMIM
GJA1	4274	NM_000165.4	2	2	Oculodentodigital dysplasia OMIM Oculodentodigital dysplasia, autosomal recessive OMIM
GJA3	4277	NM_021954.3		2	Cataract 14, multiple types OMIM
GJA8	4281	NM_005267.4		2	Cataract 1, multiple types OMIM
GMPPB	22932	NM_021971.2		1-9	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14 OMIM
GNAT1	4393	NM_144499.2		1-8	Night blindness, congenital stationary, autosomal dominant 3 OMIM
GNAT2	4394	NM_005272.3		1-8	Achromatopsia 4 OMIM
GNB3	4400	NM_002075.3		2-10	Night blindness, congenital stationary, type 1H OMIM
GNPAT	4416	NM_014236.3		1-16	Rhizomelic chondrodysplasia punctata, type 2 OMIM
GNPTG	23026	NM_032520.4		1-11	Fine corneal opacities OMIM
GPR143	20145	NM_000273.2		1-9	Nystagmus 6, congenital, X-linked OMIM Ocular albinism, type I, Nettleship-Falls type OMIM
GPR179	31371	NM_001004334.3		1-11	Night blindness, congenital stationary (complete), 1E, autosomal recessive OMIM

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GPR98	17416	NM_032119.3		1-90	Usher syndrome, type 2C OMIM Usher syndrome, type 2C, GPR98/PDZD7 digenic OMIM
GRIP1	18708	NM_021150.3		1-24	Fraser syndrome 3 OMIM
GRK1	10013	NM_002929.2		1-7	Oguchi disease-2 OMIM
GRM6	4598	NM_000843.3		1-10	Night blindness, congenital stationary (complete), 1B, autosomal recessive OMIM
GSN	4620	NM_000177.4		1-17	Amyloidosis, Finnish type OMIM
GTF2H5	21157	NM_207118.2		2-3	Trichothiodystrophy 3, photosensitive OMIM
GUCA1A	4678	NM_000409.4		3-6	Cone dystrophy-3 OMIM Cone-rod dystrophy 14 OMIM
GUCA1B	4679	NM_002098.5		1-4	Retinitis pigmentosa 48 OMIM
GUCY2D	4689	NM_000180.3		2-19	Cone-rod dystrophy 6 OMIM Leber congenital amaurosis 1 OMIM
HARS	4816	NM_002109.5		1-13	Usher syndrome type 3B OMIM
HCCS	4837	NM_005333.4		2-7	Linear skin defects with multiple congenital anomalies 1 OMIM
HESX1	4877	NM_003865.2		1-4	Septooptic dysplasia OMIM
HGSNAT	26527	NM_152419.2		1-18	Retinitis pigmentosa 73 OMIM
HK1	4922	NM_000188.2		1-18	Retinitis pigmentosa 79 OMIM
HMGB3	5004	NM_005342.3	4-5	2-5	?Microphthalmia, syndromic 13 OMIM
HMX1	5017	NM_018942.2		1-2	Oculoauricular syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
HOXB1	5111	NM_002144.3		1-2	Facial paresis, hereditary congenital, 3 OMIM
HPS1	5163	NM_000195.4	4-6	3-20	Hermansky-Pudlak syndrome 1 OMIM
HPS3	15597	NM_032383.4		1-17	Hermansky-Pudlak syndrome 3 OMIM
HPS4	15844	NM_022081.5		2-14	Hermansky-Pudlak syndrome 4 OMIM
HPS5	17022	NM_181507.1		2-23	Hermansky-Pudlak syndrome 5 OMIM
HPS6	18817	NM_024747.5		1	Hermansky-Pudlak syndrome 6 OMIM
HSF4	5227	NM_001538.3		3-15	Cataract 5, multiple types OMIM
IARS2	29685	NM_018060.3		1-23	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia OMIM
IDH3B	5385	NM_006899.4		1-12	Retinitis pigmentosa 46 OMIM
IFIH1	18873	NM_022168.3		1-16	Singleton-Merten syndrome 1 OMIM
IFT140	29077	NM_014714.3		3-31	Retinitis pigmentosa 80 OMIM
IFT172	30391	NM_015662.2		1-48	Retinitis pigmentosa 71 OMIM
IFT27	18626	NM_006860.4		1-7	?Bardet-Biedl syndrome 19 OMIM
IFT43	29669	NM_052873.2		1-8	Retinitis pigmentosa 85 OMIM
IMPDH1	6052	NM_000883.3		1-17	Leber congenital amaurosis 11 OMIM Retinitis pigmentosa 10 OMIM
IMPG1	6055	NM_001563.3		1-17	Macular dystrophy, vitelliform, 4 OMIM

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IMPG2	18362	NM_016247.3		1-19	Macular dystrophy, vitelliform, 5 OMIM Retinitis pigmentosa 56 OMIM
INPP5E	21474	NM_019892.5		1-10	Joubert syndrome 1 OMIM Mental retardation, truncal obesity, retinal dystrophy, and micropenis OMIM
INPP5K	33882	NM_016532.3		1-12	Muscular dystrophy, congenital, with cataracts and intellectual disability OMIM
IQCB1	28949	NM_001023570.3		3-15	Senior-Loken syndrome 5 OMIM
ITM2B	6174	NM_021999.4		1-6	?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities OMIM
ITPR1	6180	NM_002222.5		3-58	Gillespie syndrome OMIM
JAG1	6188	NM_000214.2		1-26	Alagille syndrome 1 OMIM
JAM3	15532	NM_032801.4		1-9	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts OMIM
KAT6B	17582	NM_012330.3		3-18	SBBYSS syndrome OMIM
KCNJ13	6259	NM_002242.4		2-3	Leber congenital amaurosis 16 OMIM Snowflake vitreoretinal degeneration OMIM
KCNV2	19698	NM_133497.3		1-2	Retinal cone dystrophy 3B OMIM
KERA	6309	NM_007035.3		2-3	Cornea plana 2, autosomal recessive OMIM
KIF11	6388	NM_004523.3		1-22	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation OMIM

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KIF21A	19349	NM_017641.3		1-37	Fibrosis of extraocular muscles, congenital, 1 OMIM Fibrosis of extraocular muscles, congenital, 3B OMIM
KIF7	30497	NM_198525.2		2-19	Acrocallosal syndrome OMIM Joubert syndrome 12 OMIM
KLHL7	15646	NM_001031710.2		1-11	Retinitis pigmentosa 42 OMIM
KMT2D	7133	NM_003482.3		1-54	Kabuki syndrome 1 OMIM
KRT12	6414	NM_000223.3		1-8	Meesmann corneal dystrophy OMIM
KRT3	6440	NM_057088.2		1-9	Meesmann corneal dystrophy OMIM
LAMA1	6481	NM_005559.3		1-63	Poretti-Boltshauser syndrome OMIM
LARGE	6511	NM_004737.5		3-16	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6 OMIM Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6 OMIM
LCA5	31923	NM_181714.3		3-9	Leber congenital amaurosis 5 OMIM
LCAT	6522	NM_000229.1		1-6	Fish-eye disease OMIM Norum disease OMIM
LEMD2	21244	NM_181336.3		1-9	Cataract 46, juvenile-onset OMIM
LEPREL1	19317	NM_018192.3		1-15	Myopia, high, with cataract and vitreoretinal degeneration OMIM
LIM2	6610	NM_030657.3		2-5	Cataract 19, multiple types OMIM
LMX1B	6654	NM_002316.3		1-8	Nail-patella syndrome OMIM

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LRAT	6685	NM_004744.4		2-3	Leber congenital amaurosis 14 OMIM Retinal dystrophy, early-onset severe OMIM Retinitis pigmentosa, juvenile OMIM
LRIT3	24783	NM_198506.4		1-4	Night blindness, congenital stationary (complete), 1F, autosomal recessive OMIM
LRP2	6694	NM_004525.2		1-79	Donnai-Barrow syndrome OMIM
LRP5	6697	NM_002335.3	1, 3-9	1-23	Exudative vitreoretinopathy 4 OMIM
LSS	6708	NM_001001438.2		1-22	Cataract 44 OMIM
LTBP2	6715	NM_000428.2		1-36	?Weill-Marchesani syndrome 3, recessive OMIM Glaucoma 3, primary congenital, D OMIM Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma OMIM
LYST	1968	NM_000081.3		3-53	Chediak-Higashi syndrome OMIM
LZTFL1	6741	NM_020347.3		1-10	Bardet-Biedl syndrome 17 OMIM
MAB21L2	6758	NM_006439.4		1	Microphthalmia/coloboma and skeletal dysplasia syndrome OMIM
MAF	6776	NM_005360.4		1-2	Ayme-Gripp syndrome OMIM Cataract 21, multiple types OMIM
MAFB	6408	NM_005461.4		1	Duane retraction syndrome 3 OMIM
MAK	6816	NM_001242957.2		1-14	Retinitis pigmentosa 62 OMIM
MAN2B1	6826	NM_000528.3		1-24	Mannosidosis, alpha-, types I and II OMIM

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MAPKAPK3	6888	NM_001243926.1		4-13	RP
MED25	28845	NM_030973.3		1-18	Basel-Vanagait-Smirin-Yosef syndrome OMIM
MERTK	7027	NM_006343.2		1-19	Retinitis pigmentosa 38 OMIM
MFN2	16877	NM_014874.3		3-19	Hereditary motor and sensory neuropathy VIA OMIM
MFSD8	28486	NM_152778.2		2-13	Ceroid lipofuscinosis, neuronal, 7 OMIM Macular dystrophy with central cone involvement OMIM
MIP	7103	NM_012064.3		1-4	Cataract 15, multiple types OMIM
MITF	7105	NM_000248.3		1-9	COMMAD syndrome OMIM Tietz albinism-deafness syndrome OMIM
MKKS	7108	NM_018848.3		3-6	Bardet-Biedl syndrome 6 OMIM
MKS1	7121	NM_017777.3		1-18	Bardet-Biedl syndrome 13 OMIM Joubert syndrome 28 OMIM Meckel syndrome 1 OMIM
MPDU1	7207	NM_004870.3		1-7	Congenital disorder of glycosylation, type If OMIM
MSMO1	10545	NM_006745.4		2-6	Microcephaly, congenital cataract, and psoriasiform dermatitis OMIM
MTTP	7467	NM_000253.3		2-19	Abetalipoproteinemia with RP in the oldest patients OMIM
MVK	7530	NM_000431.3		2-11	Mevalonic aciduria OMIM
MYH9	7579	NM_002473.5		2-41	Fechtner syndrome OMIM
MYO7A	7606	NM_000260.3		2-49	Usher syndrome, type 1B OMIM

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MYOC	7610	NM_000261.1		1-3	Glaucoma 1A, primary open angle OMIM
NAA10	18704	NM_003491.3		1-8	?Microphthalmia, syndromic 1 OMIM
NBAS	15625	NM_015909.3		1-52	Short stature, optic nerve atrophy, and Pelger-Huet anomaly OMIM
NDP	7678	NM_000266.3		2-3	Exudative vitreoretinopathy 2, X-linked OMIM Norrie disease OMIM
NEK2	7745	NM_002497.3		1-8	?Retinitis pigmentosa 67 OMIM
NF2	7773	NM_000268.3		1-16	Neurofibromatosis, type 2 OMIM
NHS	7820	NM_198270.3		1-8	Cataract 40, X-linked OMIM Nance-Horan syndrome OMIM
NMNAT1	17877	NM_022787.3		2-5	Leber congenital amaurosis 9 OMIM
NOTCH2	7882	NM_024408.3	1-4	1-34	Alagille syndrome 2 OMIM
NPHP1	7905	NM_000272.3		1-20	Joubert syndrome 4 OMIM Nephronophthisis 1, juvenile OMIM Senior-Loken syndrome-1 OMIM
NPHP3	7907	NM_153240.4		1-27	Meckel syndrome 7 OMIM Nephronophthisis 3 OMIM
NPHP4	19104	NM_015102.4		2-30	Senior-Loken syndrome 4 OMIM
NR2F1	7975	NM_005654.5		1-3	Bosch-Boonstra-Schaaf optic atrophy syndrome OMIM
NRL	8002	NM_006177.3		2-3	Retinal degeneration, autosomal recessive, clumped pigment type Retinitis pigmentosa 27 OMIM
NUS1	21042	NM_138459.4	5	1-5	?Congenital disorder of glycosylation, type 1aa OMIM

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NYX	8082	NM_022567.2		1-2	Night blindness, congenital stationary (complete), 1A, X-linked OMIM
OAT	8091	NM_000274.3		2-10	Gyrate atrophy of choroid and retina with or without ornithinemia OMIM
OCA2	8101	NM_000275.2		2-24	Albinism, brown oculocutaneous OMIM Albinism, oculocutaneous, type II OMIM
OCRL	8108	NM_000276.3		1-24	Lowe syndrome OMIM
OFD1	2567	NM_003611.2		1-23	?Retinitis pigmentosa 23 OMIM
OPA1	8140	NM_015560.2		1-28	Optic atrophy 1 OMIM Optic atrophy plus syndrome OMIM
OPA3	8142	NM_025136.3		1-2	Optic atrophy 3 with cataract OMIM
OPN1SW	1012	NM_001708.2		1-5	Colorblindness, tritan OMIM
OPTN	17142	NM_021980.4		2-14	Glaucoma 1, open angle, E OMIM
OTX2	8522	NM_001270524.1		2-4	Microphthalmia, syndromic 5 OMIM Retinal dystrophy, early-onset, with or without pituitary dysfunction OMIM
OVOL2	15804	NM_021220.3		1-4	Corneal dystrophy, posterior polymorphous, 1 OMIM
PANK2	15894	NM_153638.3		1-7	Neurodegeneration with brain iron accumulation 1 OMIM
PAX2	8616	NM_003987.4		1-11	Glomerulosclerosis, focal segmental, 7 OMIM
PAX3	8617	NM_181457.3		1-8	Waardenburg syndrome, type 1 OMIM Waardenburg syndrome, type 3 OMIM

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PAX6	8620	NM_000280.4		4-13	?Coloboma of optic nerve OMIM ?Coloboma, ocular OMIM Aniridia OMIM Anterior segment dysgenesis 5, multiple subtypes OMIM Cataract with late-onset corneal dystrophy OMIM Optic nerve hypoplasia OMIM
PCDH15	14674	NM_033056.3		2-33	Usher syndrome, type 1D/F digenic OMIM Usher syndrome, type 1F OMIM
PCYT1A	8754	NM_005017.3		3-10	Spondylometaphyseal dysplasia with cone-rod dystrophy OMIM
PDE6A	8785	NM_000440.2		1-22	Retinitis pigmentosa 43 OMIM
PDE6B	8786	NM_000283.3		1-22	Retinitis pigmentosa-40 OMIM
PDE6C	8787	NM_006204.3		1-22	Cone dystrophy 4 OMIM
PDE6D	8788	NM_002601.3		1-5	?Joubert syndrome 22 OMIM
PDE6G	8789	NM_002602.3		2-4	Retinitis pigmentosa 57 OMIM
PDE6H	8790	NM_006205.2		2-4	Retinal cone dystrophy 3 OMIM
PDZD7	26257	NM_024895.4		2-10	Usher syndrome, type IIC, GPR98/PDZD7 digenic OMIM {Retinal disease in Usher syndrome type IIA, modifier of} OMIM
PEX1	8850	NM_000466.2		1-24	Heimler syndrome 1 OMIM Peroxisome biogenesis disorder 1A (Zellweger) OMIM Peroxisome biogenesis disorder 1B (NALD/IRD) OMIM
PEX11B	8853	NM_003846.2		1-4	?Peroxisome biogenesis disorder 14B OMIM

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PEX12	8854	NM_000286.2		1-3	Peroxisome biogenesis disorder 3A (Zellweger) OMIM Peroxisome biogenesis disorder 3B OMIM
PEX13	8855	NM_002618.3		1-4	Peroxisome biogenesis disorder 11A (Zellweger) OMIM
PEX16	8857	NM_004813.2		1-11	Peroxisome biogenesis disorder 8A (Zellweger) OMIM Peroxisome biogenesis disorder 8B OMIM
PEX2	9717	NM_000318.2		4	Peroxisome biogenesis disorder 5A (Zellweger) OMIM Peroxisome biogenesis disorder 5B OMIM
PEX26	22965	NM_017929.5		2-6	Peroxisome biogenesis disorder 7A (Zellweger) OMIM Peroxisome biogenesis disorder 7B OMIM
PEX3	8858	NM_003630.2		1-12	?Peroxisome biogenesis disorder 10B OMIM Peroxisome biogenesis disorder 10A (Zellweger) OMIM
PEX5	9719	NM_001131025.1		2-16	Peroxisome biogenesis disorder 2A (Zellweger) OMIM Peroxisome biogenesis disorder 2B OMIM
PEX6	8859	NM_000287.3		1-17	Peroxisome biogenesis disorder 4A (Zellweger) OMIM
PEX7	8860	NM_000288.3		1-10	Peroxisome biogenesis disorder 9B OMIM
PGK1	8896	NM_000291.3		1-11	Phosphoglycerate kinase 1 deficiency OMIM
PHOX2A	691	NM_005169.3		1-3	Fibrosis of extraocular muscles, congenital, 2 OMIM
PHYH	8940	NM_006214.3		1-9	Refsum disease OMIM

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PIGL	8966	NM_004278.3		1-7	CHIME syndrome OMIM
PIK3R1	8979	NM_181523.2		2-16	SHORT syndrome OMIM
PIKFYVE	23785	NM_015040.3		2-42	Corneal fleck dystrophy OMIM
PITPNM3	21043	NM_031220.3		1-20	Cone-rod dystrophy 5 OMIM
PITX2	9005	NM_153427.2		3-5	Anterior segment dysgenesis 4 OMIM Axenfeld-Rieger syndrome, type 1 OMIM Ring dermoid of cornea OMIM
PITX3	9006	NM_005029.3		2-4	Anterior segment dysgenesis 1, multiple subtypes OMIM Cataract 11, multiple types OMIM Cataract 11, syndromic OMIM
PLA2G5	9038	NM_000929.2		2-5	[Fleck retina, familial benign] OMIM
PLG	9071	NM_000301.3	1-5	1-19	Dysplasminogenemia OMIM Plasminogen deficiency, type I OMIM
PLK4	11397	NM_014264.4		1-16	Microcephaly and chorioretinopathy, autosomal recessive, 2 OMIM
PMM2	9115	NM_000303.2		1-8	Congenital disorder of glycosylation, type Ia OMIM
PNPLA6	16268	NM_006702.4		3-35	?Laurence-Moon syndrome OMIM
POC1B	30836	NM_172240.2		1-12	Cone-rod dystrophy 20 OMIM
POLR1C	20194	NM_203290.3		1-9	Treacher Collins syndrome 3 OMIM
POLR1D	20422	NM_015972.3		1-2	Treacher Collins syndrome 2 OMIM
POMGNT1	19139	NM_017739.3		2-22	Retinitis pigmentosa 76 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
POMK	26267	NM_032237.4		4-5	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12 OMIM Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12 OMIM
POMT1	9202	NM_007171.3		2-20	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1 OMIM
POMT2	19743	NM_013382.5		1-21	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2 OMIM
PORCN	17652	NM_203475.2		2-15	Focal dermal hypoplasia OMIM
PPT1	9325	NM_000310.3		1-9	Ceroid lipofuscinosis, neuronal, 1 OMIM
PQBP1	9330	NM_005710.2		1-6	Renpenning syndrome OMIM
PRDM5	9349	NM_018699.3		1-16	Brittle cornea syndrome 2 OMIM
PROM1	9454	NM_006017.2		1-26	Cone-rod dystrophy 12 OMIM Retinitis pigmentosa 41 OMIM Stargardt disease 4 OMIM Macular Dystrophy, retinal, 2 OMIM
PRPF3	17348	NM_004698.3		2-16	Retinitis pigmentosa 18 OMIM
PRPF31	15446	NM_015629.3		2-14	Retinitis pigmentosa 11 OMIM
PRPF4	17349	NM_004697.4		1-14	Retinitis pigmentosa 70 OMIM
PRPF8	17340	NM_006445.3		2-43	Retinitis pigmentosa 13 OMIM

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PRPH2	9942	NM_000322.4		1-3	Choroidal dystrophy, central areolar 2 OMIM Leber congenital amaurosis 18 OMIM Retinitis pigmentosa 7 and digenic OMIM Retinitis punctata albescens OMIM
PRPS1	9462	NM_002764.3	7	1-7	Arts syndrome OMIM
PRSS56	39433	NM_001195129.1		1-13	Microphthalmia, isolated 6 OMIM
PXDN	14966	NM_012293.2		1-23	Anterior segment dysgenesis 7, with sclerocornea OMIM
RAB18	14244	NM_021252.4		1-7	Warburg micro syndrome 3 OMIM
RAB28	9768	NM_004249.3		1-7	Cone-rod dystrophy 18 OMIM
RAB3GAP1	17063	NM_012233.2		1-24	Warburg micro syndrome 1 OMIM
RAB3GAP2	17168	NM_012414.3		1-35	Martsolf syndrome OMIM Warburg micro syndrome 2 OMIM
RARB	9865	NM_000965.4		1-8	Microphthalmia, syndromic 12 OMIM
RAX	18662	NM_013435.2		1-3	Microphthalmia, isolated 3 OMIM
RAX2	18286	NM_032753.3		2-3	Cone-rod dystrophy 11 OMIM
RBP3	9921	NM_002900.2		1-4	?Retinitis pigmentosa 66 OMIM
RBP4	9922	NM_006744.3		2-6	Microphthalmia, isolated, with coloboma 10 OMIM Retinal dystrophy, iris coloboma, and comedogenic acne syndrome OMIM
RCBTB1	18243	NM_018191.3		3-13	Retinal dystrophy with or without extraocular anomalies OMIM

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RD3	19689	NM_183059.2		2-3	Leber congenital amaurosis 12 OMIM
RDH11	17964	NM_001252650.1		1-6	?Retinal dystrophy, juvenile cataracts, and short stature syndrome OMIM
RDH12	19977	NM_152443.2		3-9	Leber congenital amaurosis 13 OMIM
RDH5	9940	NM_002905.3		2-5	Fundus albipunctatus OMIM
REEP6	30078	NM_138393.2		1-5	Retinitis pigmentosa 77 OMIM
RGR	9990	NM_001012720.1		1-7	Retinitis pigmentosa 44 OMIM
RGS9	10004	NM_003835.3		1-19	Bradyopsia OMIM
RHO	10012	NM_000539.3		1-5	Retinitis pigmentosa 4, autosomal dominant or recessive OMIM Retinitis punctata albescens OMIM
RIMS1	17282	NM_014989.5		1-34	Cone-rod dystrophy 7 OMIM
RLBP1	10024	NM_000326.4		3-9	Bothnia retinal dystrophy OMIM Newfoundland rod-cone dystrophy OMIM Retinitis punctata albescens OMIM
ROBO3	13433	NM_022370.3		1-28	Gaze palsy, familial horizontal, with progressive scoliosis, 1 OMIM
ROM1	10254	NM_000327.3		1-3	Retinitis pigmentosa 7, digenic OMIM
RP1	10263	NM_006269.1		2-4	Retinitis pigmentosa 1 OMIM
RP1L1	15946	NM_178857.5		2-4	Occult macular dystrophy OMIM
RP2	10274	NM_006915.2		1-5	Retinitis pigmentosa 2 OMIM

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RP9	10288	NM_203288.1		1-6	?Retinitis pigmentosa 9 OMIM
RPE65	10294	NM_000329.2		1-14	Leber congenital amaurosis 2 OMIM Retinitis pigmentosa 20 OMIM
RPGR	10295	NM_000328.2		1-19	Cone-rod dystrophy, X-linked, 1 OMIM Retinitis pigmentosa 3 OMIM Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness OMIM
RPGRIP1	13436	NM_020366.3		1-24	Cone-rod dystrophy 13 OMIM Leber congenital amaurosis 6 OMIM
RPGRIP1L	29168	NM_015272.4		2-27	Joubert syndrome 7 OMIM Meckel syndrome 5 OMIM
RS1	10457	NM_000330.3		1-6	Retinoschisis OMIM
RTN4IP1	18647	NM_032730.5		1-9	Optic atrophy 10 with or without ataxia, mental retardation, and seizures OMIM
SAG	10521	NM_000541.4		2-16	Oguchi disease-1 OMIM Retinitis pigmentosa 47 OMIM
SALL2	10526	NM_005407.2		1-2	?Coloboma, ocular, autosomal recessive OMIM
SALL4	15924	NM_020436.4		1-4	Duane-radial ray syndrome OMIM
SBF2	2135	NM_030962.3		1-40	Charcot-Marie-Tooth disease, type 4B2 OMIM
SC5D	10547	NM_006918.4		2-5	Lathosterolosis OMIM
SDCCAG8	10671	NM_006642.4		1-18	Bardet-Biedl syndrome 16 OMIM Senior-Loken syndrome 7 OMIM
SEC23A	10701	NM_006364.3		2-20	Craniolenticulosutural dysplasia OMIM
SEMA3E	10727	NM_012431.2		1-17	?CHARGE syndrome OMIM

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SEMA4A	10729	NM_022367.3		2-15	Cone-rod dystrophy 10 OMIM Retinitis pigmentosa 35 OMIM
SH3PXD2B	29242	NM_001017995.2		1-13	Frank-ter Haar syndrome OMIM
SHH	10848	NM_000193.3		1-3	Microphthalmia with coloboma 5 OMIM
SIL1	24624	NM_022464.4		2-10	Marinesco-Sjogren syndrome OMIM
SIPA1L3	23801	NM_015073.2		3-22	?Cataract 45 OMIM
SIX6	10892	NM_007374.2		1-2	Optic disc anomalies with retinal and/or macular dystrophy OMIM
SLC16A12	23094	NM_213606.3		3-8	Cataract 47, juvenile, with microcornea OMIM
SLC24A1	10975	NM_004727.2		2-10	Night blindness, congenital stationary (complete), 1D, autosomal recessive OMIM
SLC24A5	20611	NM_205850.2		1-9	Albinism, oculocutaneous, type VI OMIM
SLC25A46	25198	NM_138773.3		1-8	Neuropathy, hereditary motor and sensory, type VIB OMIM
SLC2A1	11005	NM_006516.2		1-10	Stomatin-deficient cryohydrocytosis with neurologic defects OMIM
SLC33A1	95	NM_004733.3	6	1-6	Congenital cataracts, hearing loss, and neurodegeneration OMIM
SLC38A8	32434	NM_001080442.2		1-10	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis OMIM
SLC45A2	16472	NM_016180.4		1-7	Albinism, oculocutaneous, type IV OMIM

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SLC4A11	16438	NM_032034.3		1-19	Corneal dystrophy, Fuchs endothelial, 4 OMIM Corneal endothelial dystrophy and perceptive deafness OMIM Corneal endothelial dystrophy, autosomal recessive OMIM
SLC7A14	29326	NM_020949.2		2-8	Retinitis pigmentosa 68 OMIM
SNRNP200	30859	NM_014014.4		1-45	Retinitis pigmentosa 33 OMIM
SOX10	11190	NM_006941.3		2-4	Waardenburg syndrome, type 2E, with or without neurologic involvement OMIM Waardenburg syndrome, type 4C OMIM
SOX2	11195	NM_003106.3	1	1	Microphthalmia, syndromic 3 OMIM Optic nerve hypoplasia and abnormalities of the central nervous system OMIM
SPATA7	20423	NM_018418.4		1-12	Leber congenital amaurosis 3 OMIM Retinitis pigmentosa, juvenile, autosomal recessive OMIM
SPG7	11237	NM_003119.3		1-17	Spastic paraplegia 7, autosomal recessive OMIM
SRD5A3	25812	NM_024592.4	4-5	1-5	Kahrizi syndrome OMIM
STRA6	30650	NM_022369.3		2-19	Microphthalmia, isolated, with coloboma 8 OMIM Microphthalmia, syndromic 9 OMIM
STT3B	30611	NM_178862.2		1-16	?Congenital disorder of glycosylation, type Ix OMIM
TACSTD2	11530	NM_002353.2		1	Corneal dystrophy, gelatinous drop-like OMIM
TBC1D20	16133	NM_144628.3		1-8	Warburg micro syndrome 4 OMIM

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TBX22	11600	NM_001109878.1		2-9	?Abruzzo-Erickson syndrome OMIM
TCF4	11634	NM_001083962.1		2-19	Corneal dystrophy, Fuchs endothelial, 3 OMIM Pitt-Hopkins syndrome OMIM
TCOF1	11654	NM_001135243.1		1-26	Treacher Collins syndrome 1 OMIM
TDRD7	30831	NM_014290.2		2-17	Cataract 36 OMIM
TEAD1	11714	NM_021961.5		3-13	Sveinsson chorioretinal atrophy OMIM
TEK	11724	NM_000459.4		1-23	Glaucoma 3, primary congenital, E OMIM
TENM3	29944	NM_001080477.3		2-28	Microphthalmia, isolated, with coloboma 9 OMIM
TFAP2A	11742	NM_003220.2		1-7	Branchiooculofacial syndrome OMIM
TGFBI	11771	NM_000358.2		1-17	Corneal dystrophy, Avellino type OMIM Corneal dystrophy, epithelial basement membrane OMIM Corneal dystrophy, Groenouw type I OMIM Corneal dystrophy, lattice type I OMIM Corneal dystrophy, lattice type IIIA OMIM Corneal dystrophy, Reis-Bucklers type OMIM Corneal dystrophy, Thiel-Behnke type OMIM
TIMM8A	11817	NM_004085.3	2	1-2	Mohr-Tranebjaerg syndrome OMIM
TIMP3	11822	NM_000362.4		1-5	Sorsby fundus dystrophy OMIM
TMEM126A	25382	NM_032273.3		2-5	Optic atrophy 7 OMIM

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TMEM216	25018	NM_001173990.2		1-5	Joubert syndrome 2 OMIM Meckel syndrome 2 OMIM
TMEM237	14432	NM_001044385.2		1-12	Joubert syndrome 14 OMIM
TMEM67	28396	NM_153704.5		1-28	COACH syndrome OMIM Joubert syndrome 6 OMIM Meckel syndrome 3 OMIM Nephronophthisis 11 OMIM
TMEM98	24529	NM_015544.2		3-8	Nanophthalmos 4 OMIM
TOPORS	21653	NM_005802.4		1-3	Retinitis pigmentosa 31 OMIM
TPP1	2073	NM_000391.3		1-13	Ceroid lipofuscinosis, neuronal, 2 OMIM
TRAF3IP1	17861	NM_015650.3		1-17	Senior-Loken syndrome 9 OMIM
TREX1	12269	NM_033629.4		2	Vasculopathy, retinal, with cerebral leukodystrophy OMIM
TRIM32	16380	NM_012210.3		2	?Bardet-Biedl syndrome 11 OMIM
TRIM44	19016	NM_017583.5		1-5	?Aniridia 3 OMIM
TRNT1	17341	NM_182916.2		2-8	Retinitis pigmentosa and erythrocytic microcytosis OMIM
TRPM1	7146	NM_002420.5		2-27	Night blindness, congenital stationary (complete), 1C, autosomal recessive OMIM
TSPAN12	21641	NM_012338.3		2-8	Exudative vitreoretinopathy 5 OMIM
TTC8	20087	NM_198309.3		2-15	?Retinitis pigmentosa 51 OMIM Bardet-Biedl syndrome 8 OMIM
TTLL5	19963	NM_015072.4		2-32	Cone-rod dystrophy 19 OMIM

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TTPA	12404	NM_000370.3		1-5	Ataxia with isolated vitamin E deficiency OMIM
TUB	12406	NM_003320.4		1-13	?Retinal dystrophy and obesity OMIM
TUBGCP4	16691	NM_014444.4		1-18	Microcephaly and chorioretinopathy, autosomal recessive, 3 OMIM
TUBGCP6	18127	NM_020461.3		1-25	Microcephaly and chorioretinopathy, autosomal recessive, 1 OMIM
TULP1	12423	NM_003322.5		1-15	Leber congenital amaurosis 15 OMIM Retinitis pigmentosa 14 OMIM
TYR	12442	NM_000372.4	4-5	1-5	Albinism, oculocutaneous, type IA OMIM Albinism, oculocutaneous, type IB OMIM Waardenburg syndrome/albinism, digenic OMIM
TYRP1	12450	NM_000550.2		2-8	Albinism, oculocutaneous, type III OMIM
UBIAD1	30791	NM_013319.2		1-2	Corneal dystrophy, Schnyder type OMIM
UNC45B	14304	NM_173167.3		1-19	?Cataract 43 OMIM
USH1C	12597	NM_005709.3		1-21	Deafness, autosomal recessive 18A OMIM Usher syndrome, type 1C OMIM
USH1G	16356	NM_173477.4		1-3	Usher syndrome, type 1G OMIM
USH2A	12601	NM_206933.2		2-72	Retinitis pigmentosa 39 OMIM Usher syndrome, type 2A OMIM
VAX1	12660	NM_001112704.1		1-3	?Microphthalmia, syndromic 11 OMIM
VCAN	2464	NM_004385.4		2-15	Wagner syndrome 1 OMIM
VIM	12692	NM_003380.3		2-10	Cataract 30, pulverulent OMIM

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VPS13B	2183	NM_017890.4		2-62	Cohen syndrome OMIM
VSX1	12723	NM_014588.5		1-5	Keratoconus 1 OMIM
VSX2	1975	NM_182894.2		1-5	Microphthalmia with coloboma 3 OMIM Microphthalmia, isolated 2 OMIM
WDPCP	28027	NM_015910.5		1-18	?Bardet-Biedl syndrome 15 OMIM ?Congenital heart defects, hamartomas of tongue, and polysyndactyly OMIM
WDR19	18340	NM_025132.3		1-36	Nephronophthisis 13 OMIM Senior-Loken syndrome 8 OMIM
WDR36	30696	NM_139281.2		1-23	Glaucoma 1, open angle, G OMIM
WFS1	12762	NM_006005.3		2-8	?Cataract 41 OMIM Wolfram syndrome 1 OMIM Wolfram-like syndrome, autosomal dominant OMIM
XYLT2	15517	NM_022167.3		1-11	Spondyloocular syndrome OMIM
YAP1	16262	NM_001130145.2		1-9	Coloboma, ocular OMIM Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation OMIM
ZEB1	11642	NM_030751.5		1-9	Corneal dystrophy, Fuchs endothelial, 6 OMIM Corneal dystrophy, posterior polymorphous, 3 OMIM
ZEB2	14881	NM_014795.3	10	2-10	Mowat-Wilson syndrome OMIM
ZNF408	20041	NM_024741.2		1-5	Retinitis pigmentosa 72 OMIM
ZNF423	16762	NM_015069.4		1-8	Joubert syndrome 19 OMIM

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ZNF469	23216	NM_001127464.2		1-2	Brittle cornea syndrome 1 OMIM
ZNF513	26498	NM_144631.5		1-4	?Retinitis pigmentosa 58 OMIM