Oslo universitetssykehus
Avdeling for medisinsk genetikk

Netthinnesykdommer

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).

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
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
ACO2	118	NM_001098.2		1-18	?Optic atrophy 9 OMIM Infantile cerebellar-retinal degeneration OMIM
ADAM9	216	NM_003816.2		1-22	Cone-rod dystrophy 9 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
ADAMTS18	17110	NM_199355.3		1-23	Microcornea, myopic chorioretinal atrophy, and telecanthus OMIM
AGBL5	26147	NM_021831.5		2-15	Retinitis pigmentosa 75 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
ALDH1A3	409	NM_000693.3		1-13	Microphthalmia, isolated 8 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
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
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
B3GALNT2	28596	NM_152490.4		1-12	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
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
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

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
BFSP1	1040	NM_001195.4		1-8	Cataract 33, multiple types OMIM
BLOC1S3	20914	NM_212550.4		2	Hermansky-Pudlak syndrome 8 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
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
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

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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
CHM	1940	NM_000390.3		1-15	Choroideremia OMIM
CIB2	24579	NM_006383.3		1-6	Usher syndrome, type IJ 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

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CNGB1	2151	NM_001297.4		2-33	Retinitis pigmentosa 45 OMIM
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
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
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
CSPP1	26193	NM_024790.6		1-29	Joubert syndrome 21 OMIM
CTNNA1	2509	NM_001903.4		2-18	Macular dystrophy, patterned, 2 OMIM
CYP4V2	23198	NM_207352.3		1-11	Bietti crystalline corneoretinal dystrophy OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
DFNB31	16361	NM_015404.3		1-12	Usher syndrome, type 2D 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
DRAM2	28769	NM_178454.5		3-9	Cone-rod dystrophy 21 OMIM
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
EMC1	28957	NM_015047.2		1-23	Cerebellar atrophy, visual impairment, and psychomotor retardation OMIM
EYS	21555	NM_001142800.1	12	4-43	Retinitis pigmentosa 25 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
FLVCR1	24682	NM_014053.3		1-10	Ataxia, posterior column, with retinitis pigmentosa 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
FZD4	4042	NM_012193.3		1-2	Retinopathy of prematurity OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
GBA2	18986	NM_020944.2		1-17	Spastic paraplegia 46, autosomal recessive OMIM
GDF6	4221	NM_001001557.3		1-2	Leber congenital amaurosis 17 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
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
GPR98	17416	NM_032119.3		1-90	Usher syndrome, type 2C OMIM Usher syndrome, type 2C, GPR98/PDZD7 digenic 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
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

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
HARS	4816	NM_002109.5		1-13	Usher syndrome type 3B OMIM
HGSNAT	26527	NM_152419.2		1-18	Retinitis pigmentosa 73 OMIM
HK1	4922	NM_000188.2		1-18	Retinitis pigmentosa 79 OMIM
HMX1	5017	NM_018942.2		1-2	Oculoauricular syndrome OMIM
IDH3B	5385	NM_006899.4		1-12	Retinitis pigmentosa 46 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
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
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

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JAG1	6188	NM_000214.2		1-26	Alagille syndrome 1 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
KIF11	6388	NM_004523.3		1-22	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation OMIM
KLHL7	15646	NM_001031710.2		1-11	Retinitis pigmentosa 42 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
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
LZTFL1	6741	NM_020347.3		1-10	Bardet-Biedl syndrome 17 OMIM

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MAB21L2	6758	NM_006439.4		1	Microphthalmia/coloboma and skeletal dysplasia syndrome OMIM
MAK	6816	NM_001242957.2		1-14	Retinitis pigmentosa 62 OMIM
MERTK	7027	NM_006343.2		1-19	Retinitis pigmentosa 38 OMIM
MFSD8	28486	NM_152778.2		2-13	Ceroid lipofuscinosis, neuronal, 7 OMIM Macular dystrophy with central cone involvement 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
MVK	7530	NM_000431.3		2-11	Mevalonic aciduria OMIM
MYO7A	7606	NM_000260.3		2-49	Usher syndrome, type 1B 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
NMNAT1	17877	NM_022787.3		2-5	Leber congenital amaurosis 9 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

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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
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
OFD1	2567	NM_003611.2		1-23	?Retinitis pigmentosa 23 OMIM
OPN1SW	1012	NM_001708.2		1-5	Colorblindness, tritan 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
PAX6	8620	NM_000280.4		4-13	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

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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
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
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

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PHYH	8940	NM_006214.3		1-9	Refsum disease OMIM
PITPNM3	21043	NM_031220.3		1-20	Cone-rod dystrophy 5 OMIM
PLA2G5	9038	NM_000929.2		2-5	[Fleck retina, familial benign] 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
POMGNT1	19139	NM_017739.3		2-22	Retinitis pigmentosa 76 OMIM
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
PPT1	9325	NM_000310.3		1-9	Ceroid lipofuscinosis, neuronal, 1 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

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PRPF8	17340	NM_006445.3		2-43	Retinitis pigmentosa 13 OMIM
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
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
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
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

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
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
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
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

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RPGRIP1L	29168	NM_015272.4		2-27	Joubert syndrome 7 OMIM Meckel syndrome 5 OMIM
RS1	10457	NM_000330.3		1-6	Retinoschisis OMIM
SAG	10521	NM_000541.4		2-16	Oguchi disease-1 OMIM Retinitis pigmentosa 47 OMIM
SDCCAG8	10671	NM_006642.4		1-18	Bardet-Biedl syndrome 16 OMIM Senior-Loken syndrome 7 OMIM
SEMA4A	10729	NM_022367.3		2-15	Cone-rod dystrophy 10 OMIM Retinitis pigmentosa 35 OMIM
SLC24A1	10975	NM_004727.2		2-10	Night blindness, congenital stationary (complete), 1D, autosomal recessive OMIM
SLC25A46	25198	NM_138773.3		1-8	Neuropathy, hereditary motor and sensory, type VIB OMIM
SLC38A8	32434	NM_001080442.2		1-10	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis 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
SPATA7	20423	NM_018418.4		1-12	Leber congenital amaurosis 3 OMIM Retinitis pigmentosa, juvenile, autosomal recessive OMIM
TEAD1	11714	NM_021961.5		3-13	Sveinsson chorioretinal atrophy OMIM
TIMM8A	11817	NM_004085.3	2	1-2	Mohr-Tranebjaerg syndrome OMIM

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TIMP3	11822	NM_000362.4		1-5	Sorsby fundus dystrophy OMIM
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
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
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
TTPA	12404	NM_000370.3		1-5	Ataxia with isolated vitamin E deficiency OMIM

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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
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
VCAN	2464	NM_004385.4		2-15	Wagner syndrome 1 OMIM
VPS13B	2183	NM_017890.4		2-62	Cohen syndrome 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
WFS1	12762	NM_006005.3		2-8	Wolfram syndrome 1 OMIM Wolfram-like syndrome, autosomal dominant OMIM
ZNF408	20041	NM_024741.2		1-5	Retinitis pigmentosa 72 OMIM
ZNF423	16762	NM_015069.4		1-8	Joubert syndrome 19 OMIM
ZNF513	26498	NM_144631.5		1-4	?Retinitis pigmentosa 58 OMIM