Oslo universitetssykehus
Avdeling for medisinsk genetikk

Linsesykdommer

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: **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
ABHD12	15868	NM_001042472.2		1-13	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract 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

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AGK	21869	NM_018238.3	16	2-16	Cataract 38, autosomal recessive OMIM Sengers syndrome OMIM
ALDH18A1	9722	NM_002860.3		2-18	Cutis laxa, autosomal dominant 3 OMIM Cutis laxa, autosomal recessive, type IIIA OMIM
ALG2	23159	NM_033087.3		1-2	?Congenital disorder of glycosylation, type Ii OMIM
ATOH7	13907	NM_145178.3		1	Persistent hyperplastic primary vitreous, autosomal recessive OMIM
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
BBIP1	28093	NM_001195306.1		2-4	?Bardet-Biedl syndrome 18 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
BFSP1	1040	NM_001195.4		1-8	Cataract 33, multiple types OMIM
BFSP2	1041	NM_003571.3		1-7	Cataract 12, multiple types OMIM
C8orf37	27232	NM_177965.3		1-6	Bardet-Biedl syndrome 21 OMIM Cone-rod dystrophy 16 OMIM Retinitis pigmentosa 64 OMIM

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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
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
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
COL4A1	2202	NM_001845.5		1-52	?Retinal arteries, tortuosity of OMIM
CRYAA	2388	NM_000394.3		1-3	Cataract 9, multiple types OMIM
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

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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
CTDP1	2498	NM_004715.4		1-13	Congenital cataracts, facial dysmorphism, and neuropathy OMIM
CYP27A1	2605	NM_000784.3		1-9	Cerebrotendinous xanthomatosis OMIM
DHCR7	2860	NM_001360.2		3-9	Smith-Lemli-Opitz syndrome OMIM
DPAGT1	2995	NM_001382.3		1-9	Congenital disorder of glycosylation, type Ij OMIM
EBP	3133	NM_006579.2		2-5	MEND 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

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ERCC5	3437	NM_000123.3		1-15	Cerebrooculofacioskeletal syndrome 3 OMIM
EYA1	3519	NM_000503.5		3-18	Anterior segment anomalies with or without cataract OMIM
FAM126A	24587	NM_032581.3		2-11	Leukodystrophy, hypomyelinating, 5 OMIM
FBN1	3603	NM_000138.4		2-66	Ectopia lentis, familial OMIM Marfan syndrome OMIM MASS syndrome OMIM
FOXE3	3808	NM_012186.2		1	Anterior segment dysgenesis 2, multiple subtypes OMIM Cataract 34, multiple types OMIM
FTL	3999	NM_000146.3		1-4	Hyperferritinemia-cataract syndrome OMIM
FYCO1	14673	NM_024513.3		2-18	Cataract 18, autosomal recessive 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
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
GJA3	4277	NM_021954.3		2	Cataract 14, multiple types OMIM
GJA8	4281	NM_005267.4		2	Cataract 1, multiple types OMIM

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GNPAT	4416	NM_014236.3		1-16	Rhizomelic chondrodysplasia punctata, type 2 OMIM
GTF2H5	21157	NM_207118.2		2-3	Trichothiodystrophy 3, photosensitive OMIM
HMX1	5017	NM_018942.2		1-2	Oculoauricular syndrome 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
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
JAM3	15532	NM_032801.4		1-9	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts OMIM
LEMD2	21244	NM_181336.3		1-9	Cataract 46, juvenile-onset OMIM
LIM2	6610	NM_030657.3		2-5	Cataract 19, multiple types OMIM
LRP2	6694	NM_004525.2		1-79	Donnai-Barrow syndrome OMIM
LSS	6708	NM_001001438.2		1-22	Cataract 44 OMIM

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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
MAF	6776	NM_005360.4		1-2	Ayme-Gripp syndrome OMIM Cataract 21, multiple types OMIM
MAN2B1	6826	NM_000528.3		1-24	Mannosidosis, alpha-, types I and II OMIM
MED25	28845	NM_030973.3		1-18	Basel-Vanagait-Smirin-Yosef syndrome OMIM
MIP	7103	NM_012064.3		1-4	Cataract 15, multiple types OMIM
MITF	7105	NM_000248.3		1-9	COMMAD syndrome OMIM
MSMO1	10545	NM_006745.4		2-6	Microcephaly, congenital cataract, and psoriasiform dermatitis OMIM
MVK	7530	NM_000431.3		2-11	Mevalonic aciduria OMIM
MYH9	7579	NM_002473.5		2-41	Fechtner syndrome OMIM
NDP	7678	NM_000266.3		2-3	Exudative vitreoretinopathy 2, X-linked OMIM Norrie disease 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
NOTCH2	7882	NM_024408.3	1-4	1-34	Alagille syndrome 2 OMIM
OAT	8091	NM_000274.3		2-10	Gyrate atrophy of choroid and retina with or without ornithinemia OMIM

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OCRL	8108	NM_000276.3		1-24	Lowe syndrome OMIM
OPA3	8142	NM_025136.3		1-2	Optic atrophy 3 with cataract OMIM
OTX2	8522	NM_001270524.1		2-4	Microphthalmia, syndromic 5 OMIM Retinal dystrophy, early-onset, with or without pituitary dysfunction OMIM
PAX6	8620	NM_000280.4		4-13	?Coloboma of optic nerve OMIM ?Coloboma, ocular OMIM Aniridia OMIM Anterior segment dysgenesis 5, multiple subtypes OMIM Optic nerve hypoplasia 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
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
PEX26	22965	NM_017929.5		2-6	Peroxisome biogenesis disorder 7A (Zellweger) OMIM Peroxisome biogenesis disorder 7B OMIM

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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
PITX2	9005	NM_153427.2		3-5	Anterior segment dysgenesis 4 OMIM Axenfeld-Rieger syndrome, type 1 OMIM
PITX3	9006	NM_005029.3		2-4	Anterior segment dysgenesis 1, multiple subtypes OMIM Cataract 11, multiple types OMIM Cataract 11, syndromic 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
PORCN	17652	NM_203475.2		2-15	Focal dermal hypoplasia 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
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

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RDH11	17964	NM_001252650.1		1-6	?Retinal dystrophy, juvenile cataracts, and short stature syndrome OMIM
SC5D	10547	NM_006918.4		2-5	Lathosterolosis OMIM
SEC23A	10701	NM_006364.3		2-20	Craniolenticulosutural dysplasia OMIM
SIL1	24624	NM_022464.4		2-10	Marinesco-Sjogren syndrome OMIM
SIPA1L3	23801	NM_015073.2		3-22	?Cataract 45 OMIM
SLC16A12	23094	NM_213606.3		3-8	Cataract 47, juvenile, with microcornea 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
SRD5A3	25812	NM_024592.4	4-5	1-5	Kahrizi syndrome OMIM
TBC1D20	16133	NM_144628.3		1-8	Warburg micro syndrome 4 OMIM
TDRD7	30831	NM_014290.2		2-17	Cataract 36 OMIM
TFAP2A	11742	NM_003220.2		1-7	Branchiooculofacial syndrome OMIM
UNC45B	14304	NM_173167.3		1-19	?Cataract 43 OMIM
VIM	12692	NM_003380.3		2-10	Cataract 30, pulverulent OMIM
WFS1	12762	NM_006005.3		2-8	?Cataract 41 OMIM
XYLT2	15517	NM_022167.3		1-11	Spondyloocular syndrome OMIM