Oslo universitetssykehus
Avdeling for medisinsk genetikk

Fremre segment dysgenesi

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
ABCB6	47	NM_005689.3		1-19	Microphthalmia, isolated, with coloboma 7 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

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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
ASB10	17185	NM_001142459.1		1-5	Glaucoma 1, open angle, F OMIM
ATOH7	13907	NM_145178.3		1	Persistent hyperplastic primary vitreous, autosomal recessive 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
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
CBS	1550	NM_000071.2		3-17	Homocystinuria, B6-responsive and nonresponsive 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

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CHST6	6938	NM_021615.4		3	Macular corneal dystrophy OMIM
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
CPAMD8	23228	NM_015692.3	16-17	1-42	Anterior segment dysgenesis 8 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
DCN	2705	NM_001920.4		2-8	Corneal dystrophy, congenital stromal OMIM
DDX58	19102	NM_014314.3		1-18	Singleton-Merten syndrome 2 OMIM
DHCR7	2860	NM_001360.2		3-9	Smith-Lemli-Opitz syndrome OMIM
EBP	3133	NM_006579.2		2-5	Chondrodysplasia punctata, X-linked dominant OMIM
ELP4	1171	NM_019040.4		1-10	?Aniridia 2 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
FAM111A	24725	NM_022074.3		3-4	Kenny-Caffey syndrome, type 2 OMIM
FKRP	17997	NM_024301.4		4	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5 OMIM

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FKTN	3622	NM_001079802.1		3-11	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4 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
FOXE3	3808	NM_012186.2		1	Anterior segment dysgenesis 2, multiple subtypes OMIM Cataract 34, multiple types OMIM
GJA1	4274	NM_000165.4	2	2	Oculodentodigital dysplasia OMIM Oculodentodigital dysplasia, autosomal recessive OMIM
GNPTG	23026	NM_032520.4		1-11	Fine corneal opacities OMIM
GSN	4620	NM_000177.4		1-17	Amyloidosis, Finnish type OMIM
HMX1	5017	NM_018942.2		1-2	Oculoauricular syndrome OMIM
IFIH1	18873	NM_022168.3		1-16	Singleton-Merten syndrome 1 OMIM
ITPR1	6180	NM_002222.5		3-58	Gillespie syndrome OMIM
JAG1	6188	NM_000214.2		1-26	Alagille syndrome 1 OMIM
KERA	6309	NM_007035.3		2-3	Cornea plana 2, autosomal recessive OMIM
KRT12	6414	NM_000223.3		1-8	Meesmann corneal dystrophy OMIM
KRT3	6440	NM_057088.2		1-9	Meesmann corneal dystrophy OMIM

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LCAT	6522	NM_000229.1		1-6	Fish-eye disease OMIM Norum disease OMIM
LMX1B	6654	NM_002316.3		1-8	Nail-patella syndrome OMIM
LRP2	6694	NM_004525.2		1-79	Donnai-Barrow syndrome 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
MAF	6776	NM_005360.4		1-2	Ayme-Gripp syndrome OMIM
MED25	28845	NM_030973.3		1-18	Basel-Vanagait-Smirin-Yosef syndrome OMIM
MYOC	7610	NM_000261.1		1-3	Glaucoma 1A, primary open angle OMIM
NOTCH2	7882	NM_024408.3	1-4	1-34	Alagille syndrome 2 OMIM
OPTN	17142	NM_021980.4		2-14	Glaucoma 1, open angle, E OMIM
OVOL2	15804	NM_021220.3		1-4	Corneal dystrophy, posterior polymorphous, 1 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	Peroxisome biogenesis disorder 1A (Zellweger) OMIM Peroxisome biogenesis disorder 1B (NALD/IRD) OMIM

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PEX12	8854	NM_000286.2		1-3	Peroxisome biogenesis disorder 3A (Zellweger) OMIM Peroxisome biogenesis disorder 3B OMIM
PIK3R1	8979	NM_181523.2		2-16	SHORT syndrome OMIM
PIKFYVE	23785	NM_015040.3		2-42	Corneal fleck dystrophy 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
PLG	9071	NM_000301.3	1-5	1-19	Dysplasminogenemia OMIM Plasminogen deficiency, type I OMIM
PORCN	17652	NM_203475.2		2-15	Focal dermal hypoplasia OMIM
PRDM5	9349	NM_018699.3		1-16	Brittle cornea syndrome 2 OMIM
PXDN	14966	NM_012293.2		1-23	Anterior segment dysgenesis 7, with sclerocornea 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
SBF2	2135	NM_030962.3		1-40	Charcot-Marie-Tooth disease, type 4B2 OMIM
SH3PXD2B	29242	NM_001017995.2		1-13	Frank-ter Haar syndrome OMIM
SLC16A12	23094	NM_213606.3		3-8	Cataract 47, juvenile, with microcornea OMIM

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SLC38A8	32434	NM_001080442.2		1-10	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis OMIM
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
TACSTD2	11530	NM_002353.2		1	Corneal dystrophy, gelatinous drop-like OMIM
TBC1D20	16133	NM_144628.3		1-8	Warburg micro syndrome 4 OMIM
TCF4	11634	NM_001083962.1		2-19	Corneal dystrophy, Fuchs endothelial, 3 OMIM Pitt-Hopkins syndrome OMIM
TEK	11724	NM_000459.4		1-23	Glaucoma 3, primary congenital, E 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
TRIM44	19016	NM_017583.5		1-5	?Aniridia 3 OMIM
UBIAD1	30791	NM_013319.2		1-2	Corneal dystrophy, Schnyder type OMIM
VSX1	12723	NM_014588.5		1-5	Keratoconus 1 OMIM

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WDR36	30696	NM_139281.2		1-23	Glaucoma 1, open angle, G OMIM
WFS1	12762	NM_006005.3		2-8	Wolfram syndrome 1 OMIM Wolfram-like syndrome, autosomal dominant OMIM
ZEB1	11642	NM_030751.5		1-9	Corneal dystrophy, Fuchs endothelial, 6 OMIM Corneal dystrophy, posterior polymorphous, 3 OMIM
ZNF469	23216	NM_001127464.2		1-2	Brittle cornea syndrome 1 OMIM