Oslo universitetssykehus
Avdeling for medisinsk genetikk

Mikrooftalmi, anoftalmi og kolobom

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
ABCB6	47	NM_005689.3		1-19	Microphthalmia, isolated, with coloboma 7 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
ALDH1A3	409	NM_000693.3		1-13	Microphthalmia, isolated 8 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

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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
BCOR	20893	NM_017745.5		2-15	Microphthalmia, syndromic 2 OMIM
BMP4	1071	NM_001202.5		3-4	Microphthalmia, syndromic 6 OMIM
CC2D2A	29253	NM_001080522.2		3-38	COACH syndrome OMIM Joubert syndrome 9 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
CHD7	20626	NM_017780.3		2-38	CHARGE syndrome OMIM
CLDN19	2040	NM_148960.2		1-5	Hypomagnesemia 5, renal, with ocular involvement OMIM
CRYAA	2388	NM_000394.3		1-3	Cataract 9, multiple types OMIM
CRYBA4	2396	NM_001886.2		2-6	Cataract 23 OMIM
DPYD	3012	NM_000110.3		1-23	Dihydropyrimidine dehydrogenase deficiency OMIM
EBP	3133	NM_006579.2		2-5	Chondrodysplasia punctata, X-linked dominant OMIM
ERCC1	3433	NM_202001.2		1-8	Cerebrooculofacioskeletal syndrome 4 OMIM

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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
FAM111A	24725	NM_022074.3		3-4	Gracile bone dysplasia OMIM 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 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
FOXE3	3808	NM_012186.2		1	Anterior segment dysgenesis 2, multiple subtypes OMIM
FRAS1	19185	NM_025074.6		1-74	Fraser syndrome 1 OMIM
FREM1	23399	NM_144966.5		3-38	Manitoba oculotrichoanal syndrome OMIM
GDF3	4218	NM_020634.2		1-2	Microphthalmia with coloboma 6 OMIM Microphthalmia, isolated 7 OMIM
GDF6	4221	NM_001001557.3		1-2	Microphthalmia with coloboma 6, digenic OMIM Microphthalmia, isolated 4 OMIM
GJA1	4274	NM_000165.4	2	2	Oculodentodigital dysplasia OMIM

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GRIP1	18708	NM_021150.3		1-24	Fraser syndrome 3 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
HMGB3	5004	NM_005342.3	4-5	2-5	?Microphthalmia, syndromic 13 OMIM
HMX1	5017	NM_018942.2		1-2	Oculoauricular syndrome OMIM
LRP2	6694	NM_004525.2		1-79	Donnai-Barrow syndrome 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
MITF	7105	NM_000248.3		1-9	COMMAD syndrome OMIM
MKS1	7121	NM_017777.3		1-18	Meckel syndrome 1 OMIM
NAA10	18704	NM_003491.3		1-8	?Microphthalmia, syndromic 1 OMIM
NDP	7678	NM_000266.3		2-3	Exudative vitreoretinopathy 2, X-linked OMIM
NHS	7820	NM_198270.3		1-8	Nance-Horan syndrome OMIM
NOTCH2	7882	NM_024408.3	1-4	1-34	Alagille syndrome 2 OMIM
OCRL	8108	NM_000276.3		1-24	Lowe syndrome OMIM
OTX2	8522	NM_001270524.1		2-4	Microphthalmia, syndromic 5 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 Optic nerve hypoplasia OMIM
PDE6D	8788	NM_002601.3		1-5	?Joubert syndrome 22 OMIM
PIGL	8966	NM_004278.3		1-7	CHIME syndrome 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
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
PQBP1	9330	NM_005710.2		1-6	Renpenning 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
RAB3GAP1	17063	NM_012233.2		1-24	Warburg micro syndrome 1 OMIM

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RAB3GAP2	17168	NM_012414.3		1-35	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
RBP4	9922	NM_006744.3		2-6	Microphthalmia, isolated, with coloboma 10 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
SEMA3E	10727	NM_012431.2		1-17	?CHARGE syndrome OMIM
SHH	10848	NM_000193.3		1-3	Microphthalmia with coloboma 5 OMIM
SIX6	10892	NM_007374.2		1-2	Optic disc anomalies with retinal and/or macular dystrophy 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
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
TBX22	11600	NM_001109878.1		2-9	?Abruzzo-Erickson syndrome OMIM
TENM3	29944	NM_001080477.3		2-28	Microphthalmia, isolated, with coloboma 9 OMIM

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TFAP2A	11742	NM_003220.2		1-7	Branchiooculofacial syndrome OMIM
TMEM98	24529	NM_015544.2		3-8	Nanophthalmos 4 OMIM
VAX1	12660	NM_001112704.1		1-3	?Microphthalmia, syndromic 11 OMIM
VSX2	1975	NM_182894.2		1-5	Microphthalmia with coloboma 3 OMIM Microphthalmia, isolated 2 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
ZEB2	14881	NM_014795.3	10	2-10	Mowat-Wilson syndrome OMIM