Oslo universitetssykehus
Avdeling for medisinsk genetikk

Synsnervesykdommer

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
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
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
ALG3	23056	NM_005787.5		1-9	Congenital disorder of glycosylation, type Id OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
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
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
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
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
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
CISD2	24212	NM_001008388.4	3	1-3	Wolfram syndrome 2 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
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
EIF2B2	3258	NM_014239.3		1-8	Leukoencephalopathy with vanishing white matter OMIM
GJA1	4274	NM_000165.4	2	2	Oculodentodigital dysplasia, autosomal recessive OMIM
KIF7	30497	NM_198525.2		2-19	Acrocallosal 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
MFN2	16877	NM_014874.3		3-19	Hereditary motor and sensory neuropathy VIA OMIM
MPDU1	7207	NM_004870.3		1-7	Congenital disorder of glycosylation, type If OMIM
NR2F1	7975	NM_005654.5		1-3	Bosch-Boonstra-Schaaf optic atrophy syndrome OMIM
OPA1	8140	NM_015560.2		1-28	Optic atrophy 1 OMIM
OPA3	8142	NM_025136.3		1-2	Optic atrophy 3 with cataract OMIM
PAX6	8620	NM_000280.4		4-13	Optic nerve hypoplasia OMIM
PEX6	8859	NM_000287.3		1-17	Peroxisome biogenesis disorder 4A (Zellweger) OMIM
PRPS1	9462	NM_002764.3	7	1-7	Arts syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
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
RTN4IP1	18647	NM_032730.5		1-9	Optic atrophy 10 with or without ataxia, mental retardation, and seizures OMIM
SIX6	10892	NM_007374.2		1-2	Optic disc anomalies with retinal and/or macular dystrophy 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
SOX2	11195	NM_003106.3	1	1	Microphthalmia, syndromic 3 OMIM Optic nerve hypoplasia and abnormalities of the central nervous system OMIM
SPG7	11237	NM_003119.3		1-17	Spastic paraplegia 7, autosomal recessive OMIM
STT3B	30611	NM_178862.2		1-16	?Congenital disorder of glycosylation, type Ix OMIM
TBC1D20	16133	NM_144628.3		1-8	Warburg micro syndrome 4 OMIM
TMEM126A	25382	NM_032273.3		2-5	Optic atrophy 7 OMIM
VPS13B	2183	NM_017890.4		2-62	Cohen syndrome OMIM
WFS1	12762	NM_006005.3		2-8	Wolfram syndrome 1 OMIM Wolfram-like syndrome, autosomal dominant OMIM