Oslo universitetssykehus
Avdeling for medisinsk genetikk

Genpanel for hjernekanalopatier

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 affisert av segmentale duplikasjoner.

** Transkriptets kodende ekson.

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
ADCY5	236	NM_183357.2		1-21	Dyskinesia, familial, with facial myokymia OMIM
ATP1A2	800	NM_000702.4		1-23	Alternating hemiplegia of childhood 1 OMIM Migraine, familial basilar OMIM Migraine, familial hemiplegic, 2 OMIM
ATP1A3	801	NM_001256214.2		1-23	Alternating hemiplegia of childhood 2 OMIM Dystonia-12 OMIM
ATP7B	870	NM_000053.3		1-21	Wilson disease OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
CACNA1A	1388	NM_001127222.1		1-47	Episodic ataxia, type 2 OMIM Migraine, familial hemiplegic, 1 OMIM Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia OMIM
CACNB4	1404	NM_001145798.1		1-13	Episodic ataxia, type 5 OMIM {Epilepsy, idiopathic generalized, susceptibility to, 9} OMIM {Epilepsy, juvenile myoclonic, susceptibility to, 6} OMIM
CHRNA2	1956	NM_000742.3		2-7	Epilepsy, nocturnal frontal lobe, type 4 OMIM
CHRNA4	1958	NM_000744.6		1-6	Epilepsy, nocturnal frontal lobe, 1 OMIM
CHRNB2	1962	NM_000748.3		1-6	Epilepsy, nocturnal frontal lobe, 3 OMIM
DEPDC5	18423	NM_014662.5		2-42	Epilepsy, familial focal, with variable foci 1 OMIM
GLRA1	4326	NM_001146040.1		1-9	Hyperekplexia 1 OMIM
GLRB	4329	NM_000824.4		2-10	Hyperekplexia 2 OMIM
KCNA1	6218	NM_000217.3		2	Episodic ataxia/myokymia syndrome OMIM
KCNJ2	6263	NM_000891.2		2	Andersen syndrome OMIM
KCNK18	19439	NM_181840.1		1-3	{Migraine, with or without aura, susceptibility to, 13} OMIM
KCNMA1	6284	NM_002247.3		1-27	Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy OMIM {Epilepsy, idiopathic generalized, susceptibility to, 16} OMIM
KCNQ2	6296	NM_172106.1		1-16	Myokymia OMIM Seizures, benign neonatal, 1 OMIM

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KCNQ3	6297	NM_004519.4		1-15	Seizures, benign neonatal, 2 OMIM
KCNT1	18865	NM_001272003.1		1-31	Epilepsy nocturnal frontal lobe, 5 OMIM
NPRL2	24969	NM_006545.4		1-11	Epilepsy, familial focal, with variable foci 2 OMIM
NPRL3	14124	NM_001243247.1		5-15	Epilepsy, familial focal, with variable foci 3 OMIM
PNKD	9153	NM_015488.5		1-10	Paroxysmal nonkinesigenic dyskinesia 1 OMIM
PRRT2	30500	NM_001256442.1		2-3	Convulsions, familial infantile, with paroxysmal choreoathetosis OMIM Episodic kinesigenic dyskinesia 1 OMIM Seizures, benign familial infantile, 2 OMIM
SCN1A	10585	NM_001165963.1		1-26	Developmental and epileptic encephalopathy 6B, non-Dravet OMIM Dravet syndrome OMIM Febrile seizures, familial, 3A OMIM Generalized epilepsy with febrile seizures plus, type 2 OMIM Migraine, familial hemiplegic, 3 OMIM
SCN2A	10588	NM_001040142.1		2-27	Episodic ataxia, type 9 OMIM Seizures, benign familial infantile, 3 OMIM
SCN3A	10590	NM_001081677.1		3-28	Epilepsy, familial focal, with variable foci 4 OMIM
SCN8A	10596	NM_014191.3		2-27	?Myoclonus, familial, 2 OMIM Seizures, benign familial infantile, 5 OMIM
SLC1A3	10941	NM_004172.4		2-10	Episodic ataxia, type 6 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
SLC2A1	11005	NM_006516.3		1-10	Dystonia 9 OMIM GLUT1 deficiency syndrome 1, infantile onset, severe OMIM GLUT1 deficiency syndrome 2, childhood onset OMIM Stomatin-deficient cryohydrocytosis with neurologic defects OMIM {Epilepsy, idiopathic generalized, susceptibility to, 12} OMIM
SLC6A5	11051	NM_004211.5		1-16	Hyperekplexia 3 OMIM
SPR	11257	NM_003124.4		1-3	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency OMIM