Oslo universitetssykehus
Avdeling for medisinsk genetikk

Nevrotransmittersykdommer

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
ABAT	23	NM_020686.5		2-16	GABA transaminase deficiency OMIM
ALDH5A1	408	NM_001080.3		1-10	Succinic semialdehyde dehydrogenase deficiency OMIM 4-hydroxybutyric aciduria OMIM
ALDH7A1	877	NM_001182.4		1-18	?-aminoadipic semialdehyde dehydrogenase deficiency OMIM Pyridoxine-dependent epilepsy OMIM
AMT	473	NM_000481.3		1-9	Glycine encephalopathy due to aminomethyltransferase deficiency OMIM Nonketotic hyperglycinemia OMIM
ATAD1	25903	NM_032810.3	10	2-10	Thorase deficiency OMIM Hyperekplexia type 4 OMIM
DBH	2689	NM_000787.3		1-12	Dopamine ?-hydroxylase deficiency OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
DDC	2719	NM_000790.3		2-14	Aromatic L-amino acid decarboxylase deficiency OMIM DOPA decarboxylase deficiency OMIM
DHFR	2861	NM_000791.3	6	1-6	Dihydrofolate reductase deficiency OMIM
DNAJC12	28908	NM_021800.2		1-5	DNAJC12 deficiency OMIM Non-tetrahydrobiopterin-deficient hyperphenylalaninemia OMIM
FOLR1	3791	NM_016725.2		2-5	Folate receptor ? deficiency OMIM Neurodegeneration due to cerebral folate transport deficiency OMIM
GABBR2	4507	NM_005458.7		1-19	GABA type B receptor subunit 2 deficiency OMIM
GABRA1	4075	NM_000806.5		3-11	GABA type A receptor ?1 subunit deficiency OMIM Early infantile epileptic encephalopathy type 19 OMIM
GABRA6	4080	NM_000811.2		1-9	GABA type A receptor ?6 subunit deficiency OMIM
GABRB1	4081	NM_000812.3		1-9	GABA type A receptor ?1 subunit deficiency OMIM Early infantile epileptic encephalopathy type 45 OMIM
GABRB2	4082	NM_000813.2		2-10	GABA type A receptor ?2 subunit deficiency OMIM Infantile or early childhood epileptic encephalopathy type 2 OMIM
GABRB3	4083	NM_000814.5		1-9	GABA type A receptor ?3 subunit deficiency OMIM Early infantile epileptic encephalopathy type 43 OMIM
GABRD	4084	NM_000815.4		1-9	GABA type A receptor ? subunit deficiency OMIM
GABRG2	4087	NM_000816.3		1-9	GABA type A receptor ?2 subunit deficiency OMIM

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GAD1	4092	NM_000817.2		2-17	Glutamate decarboxylase 1 deficiency OMIM
GCH1	4193	NM_000161.2		1-6	Autosomal+recessive GTP cyclohydrolase+1 deficiency OMIM Autosomal+dominant GTP cyclohydrolase+1 deficiency OMIM Segawa syndrome; dystonia type 5A OMIM
GCSH	4208	NM_004483.4		1-5	Glycine encephalopathy due to H protein deficiency OMIM
GLDC	4313	NM_000170.2		1-25	Glycine encephalopathy due to glycine decarboxylase deficiency OMIM Nonketotic hyperglycinemia OMIM
GLRA1	4326	NM_000171.3		1-9	Glycine receptor ?1 subunit deficiency OMIM Hereditary hyperekplexia type 1 OMIM
GLRB	4329	NM_000824.4		2-10	Glycine receptor ? subunit deficiency OMIM Hereditary hyperekplexia type 2 OMIM
GPHN	15465	NM_020806.4		1-23	Gephyrin deficiency OMIM Molybdenum cofactor deficiency type C OMIM
GRIA3	4573	NM_000828.4		1-15	Ionotropic glutamate receptor AMPA type subunit 3 deficiency OMIM Syndromic X-linked mental retardation, Wu type OMIM
GRIA4	4574	NM_000829.3		2-17	Ionotropic glutamate receptor AMPA type subunit 4 dysregulation OMIM Neurodevelopmental disorder with or without seizures and gait abnormalities (NEDSGA) OMIM
GRIN1	4584	NM_007327.3		1-20	Ionotropic glutamate receptor NMDA type subunit 1 dysregulation OMIM Autosomal dominant mental retardation type 8; neurodevelopmental disorder with or without hyperkinetic movements and seizures OMIM

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GRIN2A	4585	NM_000833.4		3-14	Ionotropic glutamate receptor NMDA type subunit 2A dysregulation OMIM
GRIN2B	4586	NM_000834.4		2-13	Ionotropic glutamate receptor NMDA type subunit 2B dysregulation OMIM Early infantile epileptic encephalopathy type 27; autosomal dominant mental retardation type 6 OMIM
GRIN2D	4588	NM_000836.2		2-13	Ionotropic glutamate receptor NMDA type subunit 2D superactivity OMIM Early infantile epileptic encephalopathy type 46 OMIM
GRM1	4593	NM_001278066.1		1-8	Metabotropic glutamate receptor 1 deficiency OMIM Autosomal recessive spinocerebellar ataxia type 13 OMIM Metabotropic glutamate receptor 1 superactivity OMIM Spinocerebellar ataxia type 44 OMIM
GRM6	4598	NM_000843.4		2-11	Metabotropic glutamate receptor 6 deficiency OMIM Congenital stationary night blindness type 1B OMIM
MAOA	6833	NM_000240.3		1-15	Monoamine oxidase A deficiency OMIM Brunner syndrome OMIM
MTHFS	7437	NM_006441.3		1-3	5,10-methenyltetrahydrofolate synthetase deficiency OMIM 5-formyltetrahydrofolate cycloligase deficiency OMIM
PCBD1	8646	NM_000281.3		1-4	Pterin-4-?-carbinolamine dehydratase deficiency+ OMIM Primapterinuria; maturity-onset diabetes of the young (MODY) with hypomagnesemia and renal magnesium loss OMIM
PHGDH	8923	NM_006623.3		1-12	3-phosphoglycerate dehydrogenase deficiency OMIM

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PNPO	30260	NM_018129.3		1-7	Pyridoxamine 5'-phosphate oxidase deficiency OMIM
PSPH	9577	NM_004577.3	8	8,4-7	Phosphoserine+phosphatase+deficiency OMIM
PTS	9689	NM_000317.2		1-6	6-pyruvoyl-tetrahydropterin synthase deficiency OMIM
QDPR	9752	NM_000320.2		1-7	Dihydropteridine reductase deficiency OMIM
SLC18A2	10935	NM_003054.4		2-16	Vesicular monoamine transporter 2 deficiency OMIM
SLC1A2	10940	NM_004171.3		1-11	Astroglial glutamate aspartate transporter deficiency OMIM EAAT2 deficiency; early infantile epileptic encephalopathy type 41 OMIM
SLC1A3	10941	NM_004172.4		2-10	Glutamate aspartate transporter deficiency OMIM EAAT1 deficiency; episodic ataxia type 6 OMIM
SLC6A1	11042	NM_003042.3		3-16	GABA transporter deficiency OMIM Myoclonic-atonic epilepsy OMIM
SLC6A3	11049	NM_001044.4		2-15	Dopamine transporter deficiency OMIM Infantile Parkinsonism-dystonia OMIM
SLC6A5	11051	NM_004211.4		1-16	Glycine transporter 2 deficiency OMIM Hereditary hyperekplexia type 3 OMIM
SLC6A9	11056	NM_001024845.2		2-14	Glycine transporter 1 deficiency OMIM Glycine encephalopathy with normal serum glycine OMIM
SPR	11526	NM_003124.4		1-3	Sepiapterin reductase deficiency OMIM
TH	11782	NM_199292.2		1-14	Tyrosine hydroxylase deficiency OMIM
TRAK1	29947	NM_001042646.2		1-16	Trafficking kinesin-binding protein 1 deficiency OMIM