Oslo universitetssykehus
Avdeling for medisinsk genetikk

Hyperammonemi

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: **CA5A**

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
ACADM	89	NM_000016.5		1-12	Medium-chain acyl-CoA dehydrogenase deficiency OMIM
ACADVL	92	NM_000018.3		1-20	Very long-chain acyl-CoA dehydrogenase deficiency OMIM
ALDH18A1	9722	NM_002860.3		2-18	?-1-pyrroline-5-carboxylate synthase deficiency, cutis laxa phenotype OMIM Autosomal recessive cutis laxa type 3A; autosomal dominant cutis laxa type 3 OMIM ?-1-pyrroline-5-carboxylate synthase deficiency, spastic paraplegia phenotype OMIM Autosomal dominant spastic paraplegia type 9A; autosomal recessive spastic paraplegia type 9B OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
ARG1	19168	NM_000045.3		1-8	Arginase deficiency OMIM Argininemia OMIM
ASL	746	NM_000048.3		2-17	Argininosuccinate lyase deficiency OMIM Argininosuccinase deficiency; argininosuccinic aciduria OMIM
ASS1	758	NM_000050.4		3-16	Argininosuccinate synthetase deficiency OMIM Citrullinemia OMIM
AUH	890	NM_001698.2		1-10	3-methylglutaconyl-CoA+hydratase deficiency OMIM 3-methylglutaconic aciduria type 1 OMIM
BCKDHA	986	NM_000709.3		1-9	Branched-chain ketoacid dehydrogenase E1? deficiency OMIM Maple syrup urine disease type 1a OMIM
BCKDHB	987	NM_183050.3		1-10	Branched-chain ketoacid dehydrogenase E1? deficiency OMIM Maple syrup urine disease type 1b OMIM
CA5A	1377	NM_001739.1	1-7	1-7	Carbonic anhydrase VA deficiency OMIM
CPS1	2323	NM_001875.4		1-38	Carbamoylphosphate synthetase I deficiency OMIM
CPT1A	2328	NM_001876.3		2-19	Carnitine palmitoyltransferase 1A deficiency OMIM
CPT2	2330	NM_000098.2		1-5	Carnitine palmitoyltransferase 2 deficiency OMIM
DBT	2698	NM_001918.3		1-11	Dihydrolipoyl transacylase deficiency OMIM Maple syrup urine disease type 2; branched-chain ketoacid dehydrogenase E2 deficiency OMIM
ETFA	3481	NM_000126.3		1-12	Electron transfer flavoprotein ? subunit deficiency OMIM Glutaric acidemia type 2A; multiple acyl-CoA dehydrogenase deficiency type 2A OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
ETFB	3482	NM_001985.2		1-6	Electron transfer flavoprotein ? subunit deficiency OMIM Glutaric acidemia type 2B; multiple acyl-CoA dehydrogenase deficiency type 2B OMIM
ETFDH	3483	NM_004453.3		1-13	Electron transfer flavoprotein dehydrogenase deficiency OMIM Glutaric acidemia type 2C; multiple acyl-CoA dehydrogenase deficiency type 2C OMIM
GLUD1	4575	NM_005271.4	2-4, 13	1-13	Glutamate dehydrogenase superactivity OMIM Hyperinsulinism-hyperammonemia syndrome; familial hyperinsulinemic hypoglycemia type 6 OMIM
HADHA	4801	NM_000182.4		1-20	Trifunctional protein ? subunit deficiency OMIM Long-chain hydroxyacyl-CoA dehydrogenase or complete mitochondrial trifunctional protein deficiency OMIM
HADHB	4803	NM_000183.2		2-16	Trifunctional protein ? subunit deficiency OMIM Complete mitochondrial trifunctional protein deficiency OMIM Isolated mitochondrial+long-chain+ketoacyl-CoA thiolase deficiency OMIM
HLCS	4976	NM_000411.7		4-12	Holocarboxylase synthetase deficiency OMIM
HMGCL	5005	NM_000191.2		1-9	3-Hydroxy-3-methylglutaryl-CoA+lyase+deficiency OMIM Hydroxymethylglutaric aciduria OMIM
IVD	6186	NM_002225.3		1-12	Isovaleryl-CoA dehydrogenase deficiency OMIM Isovaleric acidemia OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
MLYCD	7150	NM_012213.2		1-5	Malonyl-CoA decarboxylase deficiency OMIM Malonic aciduria OMIM
MMAA	18871	NM_172250.2		2-7	Methylmalonic aciduria, cblA type OMIM
MMAB	19331	NM_052845.3		1-9	Methylmalonic aciduria, cblB type OMIM
MUT	7526	NM_000255.3		2-13	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency OMIM
NAGS	17996	NM_153006.2		1-7	N-acetylglutamate synthase deficiency OMIM
OAT	8091	NM_000274.3		2-10	Ornithine aminotransferase deficiency OMIM Gyrate atrophy of choroid and retina OMIM
OTC	8512	NM_000531.5		1-10	Ornithine transcarbamylase deficiency OMIM
PC	8636	NM_000920.3		3-22	Pyruvate carboxylase deficiency OMIM
PCCA	8653	NM_000282.3		1-24	Propionic acidemia due to propionyl-CoA carboxylase ? subunit deficiency OMIM
PCCB	8654	NM_000532.4		1-15	Propionic acidemia due to propionyl-CoA carboxylase ? subunit deficiency OMIM
POLG	9179	NM_002693.2		2-23	Mitochondrial DNA polymerase g catalytic subunit deficiency OMIM Mitochondrial DNA depletion syndrome type 1; Alpers-Huttenlocher syndrome; mitochondrial recessive ataxia syndrome (MIRAS); arPEO type 1; adPEO type 1 OMIM
PYGM	9726	NM_005609.3		1-20	Muscle glycogen phosphorylase deficiency OMIM Glycogen storage disease type 5; McArdle disease OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
SERAC1	21061	NM_032861.3		2-17	SERAC1 deficiency OMIM 3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome (MEGDEL) OMIM
SLC22A5	10969	NM_003060.3		1-10	Primary carnitine deficiency OMIM
SLC25A13	10983	NM_014251.2		1-18	Citrin deficiency OMIM
SLC25A15	10985	NM_014252.3	2, 6-7	2-7	Mitochondrial ornithine transporter deficiency OMIM Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome; ornithine translocase deficiency OMIM
SLC25A20	1421	NM_000387.5		1-9	Carnitine-acylcarnitine translocase deficiency OMIM
SLC7A7	11065	NM_001126106.2		3-11	Lysinuric protein intolerance OMIM Dibasic aminoaciduria type 2 OMIM
TMEM70	26050	NM_017866.5		1-3	Transmembrane protein 70 deficiency OMIM