Oslo universitetssykehus
Avdeling for medisinsk genetikk

Genpanel for hydrops føtalis

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: **GBA, HBA1, HBA2**

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
AHCY	343	NM_000687.4		1-10	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase OMIM
ALG9	15672	NM_024740.2		1-16	Congenital disorder of glycosylation, type Il OMIM Gillessen-Kaesbach-Nishimura syndrome OMIM
ASAH1	735	NM_177924.5		1-14	Farber lipogranulomatosis OMIM Spinal muscular atrophy with progressive myoclonic epilepsy OMIM
ATP1A2	800	NM_000702.4		1-23	Alternating hemiplegia of childhood 1 OMIM Migraine, familial basilar OMIM Migraine, familial hemiplegic, 2 OMIM

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BRAF	1097	NM_001374258.1	19	1-20	Adenocarcinoma of lung, somatic OMIM Cardiofaciocutaneous syndrome OMIM Colorectal cancer, somatic OMIM LEOPARD syndrome 3 OMIM Melanoma, malignant, somatic OMIM Nonsmall cell lung cancer, somatic OMIM Noonan syndrome 7 OMIM
CBL	1541	NM_005188.4		1-16	?Juvenile myelomonocytic leukemia OMIM Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia OMIM
CCBE1	29426	NM_133459.4		1-11	Hennekam lymphangiectasia-lymphedema syndrome 1 OMIM
CDAN1	1713	NM_138477.4		1-28	Dyserythropoietic anemia, congenital, type Ia OMIM
CTSA	9251	NM_000308.4		2-15	Galactosialidosis OMIM
FLT4	3767	NM_182925.5		1-30	Congenital heart defects, multiple types, 7 OMIM Hemangioma, capillary infantile, somatic OMIM Lymphatic malformation 1 OMIM
FOXC2	3801	NM_005251.3		1	Lymphedema-distichiasis syndrome OMIM Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus OMIM
FOXP3	6106	NM_014009.4		2-12	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked OMIM
GALNS	4122	NM_000512.5		1-14	Mucopolysaccharidosis IVA OMIM

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GBA	4177	NM_000157.4	1-11	1-11	Gaucher disease, perinatal lethal OMIM Gaucher disease, type I OMIM Gaucher disease, type II OMIM Gaucher disease, type III OMIM Gaucher disease, type IIIC OMIM {Lewy body dementia, susceptibility to} OMIM {Parkinson disease, late-onset, susceptibility to} OMIM
GBE1	4180	NM_000158.4		1-16	Glycogen storage disease IV OMIM Polyglucosan body disease, adult form OMIM
GLA	4296	NM_000169.3		1-7	Fabry disease OMIM Fabry disease, cardiac variant OMIM
GLB1	4298	NM_000404.4		1-16	GM1-gangliosidosis, type I OMIM GM1-gangliosidosis, type II OMIM GM1-gangliosidosis, type III OMIM Mucopolysaccharidosis type IVB (Morquio) OMIM
GNPTAB	29670	NM_024312.5		1-21	Mucolipidosis II alpha/beta OMIM Mucolipidosis III alpha/beta OMIM
GUSB	4696	NM_000181.4	11	1-12	Mucopolysaccharidosis VII OMIM
HBA1	4823	NM_000558.5	1-3	1-3	Erythrocytosis 7 OMIM Heinz body anemias, alpha- OMIM Hemoglobin H disease, nondeletional OMIM Methemoglobinemia, alpha type OMIM Thalassemias, alpha- OMIM
HBA2	4824	NM_000517.6	1-3	1-3	Erythrocytosis 7 OMIM Heinz body anemia OMIM Hemoglobin H disease, deletional and nondeletional OMIM Thalassemia, alpha- OMIM

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HRAS	5173	NM_005343.4		2-5	Bladder cancer, somatic OMIM Congenital myopathy with excess of muscle spindles OMIM Costello syndrome OMIM Nevus sebaceous or woolly hair nevus, somatic OMIM Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic OMIM Spitz nevus or nevus spilus, somatic OMIM Thyroid carcinoma, follicular, somatic OMIM
IDUA	5391	NM_000203.5		1-14	Mucopolysaccharidosis Ih OMIM Mucopolysaccharidosis Ih/s OMIM Mucopolysaccharidosis Is OMIM
KLF1	6345	NM_006563.5		1-3	Blood group--Lutheran inhibitor OMIM Dyserythropoietic anemia, congenital, type IV OMIM [Hereditary persistence of fetal hemoglobin] OMIM
KRAS	6407	NM_004985.5	5	2-5	Arteriovenous malformation of the brain, somatic OMIM Bladder cancer, somatic OMIM Breast cancer, somatic OMIM Cardiofaciocutaneous syndrome 2 OMIM Gastric cancer, somatic OMIM Leukemia, acute myeloid, somatic OMIM Lung cancer, somatic OMIM Noonan syndrome 3 OMIM Oculoectodermal syndrome, somatic OMIM Pancreatic carcinoma, somatic OMIM RAS-associated autoimmune leukoproliferative disorder OMIM Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic OMIM

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LARS2	17095	NM_015340.4		3-22	Hydrops, lactic acidosis, and sideroblastic anemia OMIM Perrault syndrome 4 OMIM
LBR	6518	NM_002296.4		2-14	?Reynolds syndrome OMIM Greenberg skeletal dysplasia OMIM Pelger-Huet anomaly OMIM Pelger-Huet anomaly with mild skeletal anomalies OMIM
LIPA	6617	NM_000235.4		2-10	Cholesteryl ester storage disease OMIM Wolman disease OMIM
LZTR1	6742	NM_006767.4		1-21	Noonan syndrome 10 OMIM Noonan syndrome 2 OMIM {Schwannomatosis-2, susceptibility to} OMIM
MAP2K1	6840	NM_002755.4		1-11	Cardiofaciocutaneous syndrome 3 OMIM Melorheostosis, isolated, somatic mosaic OMIM
MAP2K2	6842	NM_030662.4		1-11	Cardiofaciocutaneous syndrome 4 OMIM
MRAS	7227	NM_001085049.3		2-6	Noonan syndrome 11 OMIM
NEU1	7758	NM_000434.4		1-6	Sialidosis, type I OMIM Sialidosis, type II OMIM
NF1	7765	NM_001042492.3	9-11, 13-29, 32-36	1-58	Leukemia, juvenile myelomonocytic OMIM Neurofibromatosis, familial spinal OMIM Neurofibromatosis, type 1 OMIM Neurofibromatosis-Noonan syndrome OMIM Watson syndrome OMIM
NPC1	7897	NM_000271.5		1-25	Niemann-Pick disease, type C1 OMIM Niemann-Pick disease, type D OMIM {Nasopharyngeal carcinoma 1} OMIM

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NRAS	7989	NM_002524.5		2-5	?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic OMIM Colorectal cancer, somatic OMIM Epidermal nevus, somatic OMIM Melanocytic nevus syndrome, congenital, somatic OMIM Neurocutaneous melanosis, somatic OMIM Noonan syndrome 6 OMIM Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic OMIM Thyroid carcinoma, follicular, somatic OMIM
PEX1	8850	NM_000466.3		1-24	Heimler syndrome 1 OMIM Peroxisome biogenesis disorder 1A (Zellweger) OMIM Peroxisome biogenesis disorder 1B (NALD/IRD) OMIM
PEX10	8851	NM_002617.4		1-6	Peroxisome biogenesis disorder 6A (Zellweger) OMIM Peroxisome biogenesis disorder 6B OMIM
PEX11B	8853	NM_003846.3		1-4	Peroxisome biogenesis disorder 14B OMIM
PEX12	8854	NM_000286.3		1-3	Peroxisome biogenesis disorder 3A (Zellweger) OMIM Peroxisome biogenesis disorder 3B OMIM
PEX13	8855	NM_002618.4		1-4	Peroxisome biogenesis disorder 11A (Zellweger) OMIM Peroxisome biogenesis disorder 11B OMIM
PEX14	8856	NM_004565.3		1-9	Peroxisome biogenesis disorder 13A (Zellweger) OMIM
PEX16	8857	NM_004813.4		1-11	Peroxisome biogenesis disorder 8A (Zellweger) OMIM Peroxisome biogenesis disorder 8B OMIM

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PEX19	9713	NM_002857.4		1-8	Peroxisome biogenesis disorder 12A (Zellweger) OMIM
PEX2	9717	NM_000318.3		4	Peroxisome biogenesis disorder 5A (Zellweger) OMIM Peroxisome biogenesis disorder 5B OMIM
PEX26	22965	NM_001127649.3		1-5	Peroxisome biogenesis disorder 7A (Zellweger) OMIM Peroxisome biogenesis disorder 7B OMIM
PEX3	8858	NM_003630.3		1-12	?Peroxisome biogenesis disorder 10B OMIM Peroxisome biogenesis disorder 10A (Zellweger) OMIM
PEX5	9719	NM_001351132.2		2-16	Peroxisome biogenesis disorder 2A (Zellweger) OMIM Peroxisome biogenesis disorder 2B OMIM Rhizomelic chondrodysplasia punctata, type 5 OMIM
PEX6	8859	NM_000287.4		1-17	Heimler syndrome 2 OMIM Peroxisome biogenesis disorder 4A (Zellweger) OMIM Peroxisome biogenesis disorder 4B OMIM
PEX7	8860	NM_000288.4		1-10	Peroxisome biogenesis disorder 9B OMIM Rhizomelic chondrodysplasia punctata, type 1 OMIM
PIEZO1	28993	NM_001142864.4		1-51	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema OMIM Lymphatic malformation 6 OMIM
PMM2	9115	NM_000303.3		1-8	Congenital disorder of glycosylation, type Ia OMIM
PPP1CB	9282	NM_002709.3		1-8	Noonan syndrome-like disorder with loose anagen hair 2 OMIM

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PTPN11	9644	NM_002834.5		1-15	LEOPARD syndrome 1 OMIM Leukemia, juvenile myelomonocytic, somatic OMIM Metachondromatosis OMIM Noonan syndrome 1 OMIM
RAF1	9829	NM_001354689.3		2-18	Cardiomyopathy, dilated, 1NN OMIM LEOPARD syndrome 2 OMIM Noonan syndrome 5 OMIM
RIT1	10023	NM_006912.6		2-6	Noonan syndrome 8 OMIM
RRAS	10447	NM_006270.5		1-6	
RRAS2	17271	NM_012250.6	6	1-6	Noonan syndrome 12 OMIM Ovarian carcinoma
SGPL1	10817	NM_003901.4		2-15	Nephrotic syndrome, type 14 OMIM
SHOC2	15454	NM_007373.4		2-9	Noonan syndrome-like with loose anagen hair 1 OMIM
SLC17A5	10933	NM_012434.5		1-11	Salla disease OMIM Sialic acid storage disorder, infantile OMIM
SMPD1	11120	NM_000543.5		1-6	Niemann-Pick disease, type A OMIM Niemann-Pick disease, type B OMIM
SOS1	11187	NM_005633.4		1-23	?Fibromatosis, gingival, 1 OMIM Noonan syndrome 4 OMIM
SOS2	11188	NM_006939.4		1-23	Noonan syndrome 9 OMIM
SPRED1	20249	NM_152594.3		1-7	Legius syndrome OMIM
TALDO1	11559	NM_006755.2		1-8	Transaldolase deficiency OMIM