Oslo universitetssykehus
Avdeling for medisinsk genetikk

Lysosomale avleiringssykdommer inkludert mukopolysakkaridoser og nevronale ceroide lipofuscinoser

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: **GBA**

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
AGA	318	NM_000027.3		1-9	Aspartylglucosaminidase deficiency OMIM Aspartylglucosaminuria OMIM
AP5Z1	22197	NM_014855.2		1-17	Adaptor-related protein complex 5 ?-1 subunit deficiency OMIM Autosomal recessive spastic paraplegia type 48 OMIM
ARSA	713	NM_000487.5		1-8	Arylsulfatase A deficiency OMIM Metachromatic leukodystrophy OMIM
ARSB	714	NM_000046.4		1-8	N-acetylgalactosamine 4-sulfatase deficiency OMIM Mucopolysaccharidosis type 6; Maroteaux-Lamy syndrome; arylsulfatase B deficiency OMIM

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ASAH1	735	NM_177924.4		1-14	Acid ceramidase deficiency, inflammatory phenotype OMIM Farber disease OMIM Acid ceramidase deficiency, primary neurologic phenotype OMIM Spinal muscular atrophy with progressive myoclonic epilepsy OMIM
ATP13A2	30213	NM_022089.3		1-29	ATP13A2 deficiency OMIM Neuronal ceroid lipofuscinosis type 12; Kufor-Rakeb syndrome; Parkinson disease type 9; autosomal recessive spastic paraplegia type 78 OMIM
CLN3	2074	NM_001042432.1		2-16	Neuronal ceroid lipofuscinosis type 3 OMIM Vogt-Spielmeyer disease OMIM
CLN5	2076	NM_006493.2		1-4	Neuronal ceroid lipofuscinosis type 5 OMIM
CLN6	2077	NM_017882.2		1-7	Neuronal ceroid lipofuscinosis type 6 OMIM Autosomal recessive Kufs disease type A OMIM
CLN8	2079	NM_018941.3		2-3	Neuronal ceroid lipofuscinosis type 8 OMIM
CTNS	2518	NM_004937.2		3-12	Cystinosin deficiency OMIM Cystinosis OMIM
CTSA	9251	NM_000308.3		1-15	Cathepsin A deficiency OMIM Galactosialidosis OMIM
CTSC	2528	NM_001814.5		1-7	Cathepsin C deficiency OMIM Papillon-Lefevre syndrome; Haim-Munk syndrome OMIM
CTSD	2529	NM_001909.4		1-9	Cathepsin D deficiency OMIM Neuronal ceroid lipofuscinosis type 10 OMIM
CTSF	2531	NM_003793.3		1-13	Cathepsin F deficiency OMIM Autosomal recessive Kufs disease type B OMIM

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CTSK	2536	NM_000396.3		2-8	Cathepsin K deficiency OMIM Pycnodysostosis OMIM
DNAJC5	16235	NM_025219.2		2-5	Neuronal ceroid lipofuscinosis type 4 (Parry type) OMIM Autosomal dominant Kufs disease OMIM
FUCA1	4006	NM_000147.4		1-8	?-fucosidase deficiency OMIM ?-fucosidosis OMIM
GAA	4065	NM_000152.4		2-20	?-glucosidase deficiency OMIM Glycogen storage disease type 2; Pompe disease OMIM
GALC	4115	NM_000153.3		1-17	?-galactosylceramidase deficiency OMIM Globoid cell leukodystrophy; Krabbe disease OMIM
GALNS	4122	NM_000512.4		1-14	N-acetylgalactosamine 6-sulfatase deficiency OMIM Mucopolysaccharidosis type 4A; Morquio syndrome type A OMIM
GBA	4177	NM_001005741.2	2-12	2-12	Glucocerebrosidase deficiency OMIM Gaucher disease OMIM
GLA	4296	NM_000169.2		1-7	?-Galactosidase A deficiency OMIM Fabry disease OMIM
GLB1	4298	NM_000404.3		1-16	?-galactosidase deficiency, GM1 gangliosidosis phenotype OMIM ?-galactosidase deficiency, Morquio syndrome phenotype OMIM Mucopolysaccharidosis type 4B OMIM
GM2A	4367	NM_000405.4		1-4	GM2 activator protein deficiency OMIM GM2 gangliosidosis, AB variant OMIM
GNE	23657	NM_001128227.2		1-12	UDP-N-acetylglucosamine-2-epimerase/N-acetylmannosamine kinase deficiency OMIM GNE myopathy; Nonaka myopathy OMIM UDP-N-acetylglucosamine-2-epimerase/N-acetylmannosamine kinase superactivity OMIM Sialuria OMIM

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GNPTAB	29670	NM_024312.4		1-21	UDP-N-acetylglucosamine-1-phosphotransferase ?/? subunit deficiency OMIM Mucolipidosis type 2, I-cell disease (severe); mucolipidosis type 3 ?/?, pseudo-Hurler polydystrophy (milder) OMIM
GNPTG	23026	NM_032520.4		1-11	UDP-N-acetylglucosamine-1-phosphotransferase ? subunit deficiency OMIM Mucolipidosis type 3 ? OMIM
GNS	4422	NM_002076.3		1-14	N-acetylglucosamine 6-sulfatase deficiency OMIM Mucopolysaccharidosis type 3D; Sanfilippo syndrome type D OMIM
GRN	4601	NM_002087.3		2-13	Progranulin deficiency OMIM Frontotemporal lobar degeneration with TDP-43 inclusions+(dominant); neuronal ceroid lipofuscinosis type 11 (recessive) OMIM
GUSB	4696	NM_000181.3	11	1-12	?-glucuronidase deficiency OMIM Mucopolysaccharidosis type 7; Sly syndrome OMIM
HEXA	4878	NM_000520.5		1-14	?-hexosaminidase ?-subunit deficiency OMIM GM2 gangliosidosis, B variant (Tay-Sachs disease) OMIM
HEXB	4879	NM_000521.3		1-14	?-hexosaminidase ?-subunit deficiency OMIM GM2 gangliosidosis, O variant (Sandhoff disease) OMIM
HGSNAT	26527	NM_152419.2		1-18	Heparan-?-glucosaminide N-acetyltransferase deficiency OMIM Mucopolysaccharidosis type 3C, Sanfilippo syndrome type C (severe), retinitis pigmentosa type 73 (milder) OMIM

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HYAL1	5320	NM_153281.1		4-6	Hyaluronidase deficiency OMIM Mucopolysaccharidosis type 9; Natowicz syndrome OMIM
IDS	5389	NM_000202.7	2-3	1-9	Iduronate sulfatase deficiency OMIM Mucopolysaccharidosis type 2; Hunter syndrome OMIM
IDUA	5391	NM_000203.4		1-14	?-iduronidase deficiency OMIM Mucopolysaccharidosis type 1H, Hurler syndrome (severe); mucopolysaccharidosis type 1S, Scheie syndrome (milder) OMIM
KCTD7	21957	NM_153033.4		1-4	Neuronal ceroid lipofuscinosis type 14 OMIM Progressive myoclonic epilepsy type 3 OMIM
LAMP2	6501	NM_002294.2		1-9	Lysosome-associated membrane protein 2 deficiency OMIM Danon disease OMIM
LIPA	6617	NM_000235.3		2-10	Lysosomal acid lipase deficiency OMIM Wolman disease (severe); cholesteryl ester storage disease (milder) OMIM
MAN2B1	6826	NM_000528.3		1-24	Alpha-B-mannosidase deficiency OMIM Alpha-mannosidosis OMIM
MANBA	6831	NM_005908.3		1-17	Beta-A-mannosidase deficiency OMIM Beta-mannosidosis OMIM
MCOLN1	13356	NM_020533.2		1-14	Mucolipin 1 deficiency OMIM Mucolipidosis type 4 OMIM
MFSD8	28486	NM_152778.2		2-13	Neuronal ceroid lipofuscinosis type 7 OMIM Macular dystrophy with central cone involvement (milder) OMIM
NAGA	7631	NM_000262.2		1-9	?-N-acetylgalactosaminidase deficiency OMIM Schindler disease; Kanzaki disease (milder) OMIM

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NAGLU	7632	NM_000263.3		1-6	N-acetylglucosaminidase deficiency OMIM Mucopolysaccharidosis type 3B; Sanfilippo syndrome type B OMIM
NEU1	7758	NM_000434.3		1-6	?-neuraminidase deficiency OMIM Sialidosis OMIM
NPC1	7897	NM_000271.4		1-25	Niemann-Pick disease type C1 OMIM
NPC2	14537	NM_006432.4		1-5	Niemann-Pick disease type C2 OMIM
PPT1	9325	NM_000310.3		1-9	Palmitoyl-protein thioesterase 1 deficiency OMIM Neuronal ceroid lipofuscinosis type 1; Santavuori-Haltia disease OMIM
PSAP	9498	NM_002778.3		1-14	Atypical Gaucher disease due to saposin C deficiency OMIM Atypical Krabbe disease due to saposin A deficiency OMIM Metachromatic leukodystrophy due to saposin B deficiency OMIM Combined saposin deficiency OMIM Prosaposin deficiency OMIM
RAB7A	9788	NM_004637.5		2-6	RAB7 deficiency OMIM Charcot-Marie-Tooth disease type 2B OMIM
SCARB2	1665	NM_005506.3		1-12	Glucocerebrosidase receptor deficiency OMIM Progressive myoclonic epilepsy type 4; action myoclonus-renal failure syndrome OMIM
SGSH	10818	NM_000199.4		1-8	Heparan N-sulfatase deficiency OMIM Mucopolysaccharidosis type 3A; Sanfilippo syndrome type A OMIM
SLC17A5	10933	NM_012434.4		1-11	Sialin deficiency OMIM Infantile sialic acid storage disease (severe); Salla disease (milder) OMIM

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SMPD1	11120	NM_000543.4		1-6	Acid sphingomyelinase deficiency OMIM Niemann-Pick type A (severe); Niemann-Pick type B (milder) OMIM
SPG11	11226	NM_025137.3		1-40	Spatacsin deficiency OMIM Autosomal recessive spastic paraplegia type 11; axonal Charcot-Marie-Tooth disease type 2X;+juvenile amyotrophic lateral sclerosis type 5 OMIM
SUMF1	20376	NM_182760.3		1-9	Formyl-glycine generating enzyme deficiency OMIM Multiple sulfatase deficiency OMIM
TPP1	2073	NM_000391.3		1-13	Tripeptidyl-peptidase 1 deficiency OMIM Neuronal ceroid lipofuscinosis type 2, Jansky-Bielchowsky disease (severe), autosomal recessive spinocerebellar ataxia type 7 (milder) OMIM
VPS33A	18179	NM_022916.5		1-13	Mucopolysaccharidosis-plus syndrome OMIM