Oslo universitetssykehus
Avdeling for medisinsk genetikk

Fedme

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
ALMS1	428	NM_015120.4	17-21	1-23	Alstrom syndrome OMIM
ARL6	13210	NM_001278293.1		2-8	Bardet-Biedl syndrome 3 OMIM Retinitis pigmentosa 55 OMIM
BBS1	966	NM_024649.4		1-17	Bardet-Biedl syndrome 1 OMIM
BBS10	26291	NM_024685.3		1-2	Bardet-Biedl syndrome 10 OMIM
BBS12	26648	NM_152618.2		2	Bardet-Biedl syndrome 12 OMIM
BBS2	967	NM_031885.3		1-17	Bardet-Biedl syndrome 2 OMIM Retinitis pigmentosa 74 OMIM
BBS4	969	NM_033028.4		1-16	Bardet-Biedl syndrome 4 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
BBS5	970	NM_152384.3		1-12	Bardet-Biedl syndrome 5 OMIM
BBS7	18758	NM_176824.2		1-19	Bardet-Biedl syndrome 7 OMIM
BBS9	30000	NM_198428.2		2-23	Bardet-Biedl syndrome 9 OMIM
CEP19	28209	NM_032898.4		2-3	Morbid obesity and spermatogenic failure OMIM
CEP290	29021	NM_025114.3	54	2-54	?Bardet-Biedl syndrome 14 OMIM Joubert syndrome 5 OMIM Leber congenital amaurosis 10 OMIM Meckel syndrome 4 OMIM Senior-Loken syndrome 6 OMIM
CPE	2303	NM_001873.2		1-9	Intellectual developmental disorder and hypogonadotropic hypogonadism OMIM
GNAS	4392	NM_016592.4		1	ACTH-independent macronodular adrenal hyperplasia OMIM McCune-Albright syndrome, somatic, mosaic OMIM Osseous heteroplasia, progressive OMIM Pituitary adenoma 3, multiple types, somatic OMIM Pseudohypoparathyroidism Ia OMIM Pseudohypoparathyroidism Ib OMIM Pseudohypoparathyroidism Ic OMIM Pseudopseudohypoparathyroidism OMIM
INPP5E	21474	NM_019892.5		1-10	Joubert syndrome 1 OMIM Mental retardation, truncal obesity, retinal dystrophy, and micropenis OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
KSR2	18610	NM_173598.4		1-20	
LEP	6553	NM_000230.3		2-3	Obesity, morbid, due to leptin deficiency OMIM
LEPR	6554	NM_002303.5		3-20	Obesity, morbid, due to leptin receptor deficiency OMIM
MC4R	6932	NM_005912.2		1	Obesity (BMIQ20) OMIM
MKKS	7108	NM_170784.2		3-6	Bardet-Biedl syndrome 6 OMIM McKusick-Kaufman syndrome OMIM
MKS1	7121	NM_001165927.1		1-18	Bardet-Biedl syndrome 13 OMIM Joubert syndrome 28 OMIM Meckel syndrome 1 OMIM
MYT1L	7623	NM_015025.4		6-25	Mental retardation, autosomal dominant 39 OMIM
NTRK2	8032	NM_006180.4		4-21	Developmental and epileptic encephalopathy 58 OMIM Obesity, hyperphagia, and developmental delay OMIM
PCSK1	8743	NM_000439.4		1-14	Obesity with impaired prohormone processing OMIM
PHF6	18145	NM_032458.3		2-10	Borjeson-Forssman-Lehmann syndrome OMIM
POMC	9201	NM_001035256.1		3-4	{Obesity, early-onset, susceptibility to} OMIM Obesity, adrenal insufficiency, and red hair due to POMC deficiency OMIM
SDCCAG8	10671	NM_006642.5		1-18	Bardet-Biedl syndrome 16 OMIM Senior-Loken syndrome 7 OMIM
SH2B1	30417	NM_015503.2		1-9	
SIM1	10882	NM_005068.2		1-11	

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
TTC8	20087	NM_198309.3		2-15	Bardet-Biedl syndrome 8 OMIM
TUB	12406	NM_003320.4		1-13	?Retinal dystrophy and obesity OMIM