SNiPAcard

Block annotations

Block info

genomic range	chr1:66,030,445-66,053,063
block size	22,619 bp
variant count	16 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.094 [-2.963 – 1.639]	**gene(s) hit or close-by**	LEPR, RP11-430H12.2
phastCons	μ = 0.089 [0 – 1]	**eQTL gene(s)**	LEPR, LEPROT
GERP++	μ = -0.541 [-6.47 – 2.49]	**potentially regulated gene(s)**	RP11-430H12.2
CADD score	μ = 3.503 [0.504 – 9.95]	**disease gene(s)**	LEPR

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
3-carboxy-4-methyl-5-propyl-2-furanpropanoate (CMPF)	4.14×10^{-5}	Metabolomics GWAS Server	24816252	4
Body mass index	9.45×10^{-5}	dbGaP	pha003009	1

Disease gene annotation

gene	trait	source DB	source entry/link
LEPR	LEPTIN RECEPTOR DEFICIENCY	OMIM	MIM:614963
LEPR	Obesity due to leptin receptor gene deficiency	OrphaNet	OrphaNet:179494

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LEPR	ENST00000616738	ILMN_2234956	monocyte	3.09×10^{-17} (p-value)	Zeller et al.	4
LEPR	ENST00000371060					
LEPROT	ENST00000488747	ILMN_1661537	blood	1.52×10^{-6} (p-value)	Westra et al.	1
LEPROT	ENST00000475108					
LEPROT	ENST00000613538					
LEPROT	ENST00000371065					

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000002892		

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000670220 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		embryonic stem cell (H1ESC)	CTCF, Rad21
		endothelium (HUVEC)	H3K36me3, CTCF

	liver (HepG2)	Rad21, CTCF
	lung (IMR90)	CTCF
	blood (K562)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
LEPR	upstream gene variant	784	ENST00000371058	?	ENSP00000360097	1
RP11-430H12.2	downstream gene variant, upstream gene variant	2814	ENST00000426714	?	?	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LEPR	ENST00000371058	?	ENSP00000360097	15
LEPR	ENST00000616738	NM_001198689.1	ENSP00000483390	16
LEPR	ENST00000406510	?	ENSP00000384025	16
LEPR	ENST00000344610	NM_001198688.1	ENSP00000340884	16
LEPR	ENST00000349533	NM_002303.5	ENSP00000330393	16
LEPR	ENST00000462765	?	?	16
LEPR	ENST00000371059	NM_001198687.1, NM_001003680.3	ENSP00000360098	16
LEPR	ENST00000371060	NM_001003679.3	ENSP00000360099	16

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-430H12.2	ENST00000426714	?	1