Locus 325 (rs17412403)

SNiPAcard

Block annotations

Block info

genomic range	chr1:65,931,394-65,966,564
block size	35,171 bp
variant count	20 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.065 [-2.435 – 2.666]	gene(s) hit or close-by	LEPR, RP4-630A11.3
phastCons	μ = 0.077 [0 – 0.995]	eQTL gene(s)	LEPR, LEPROT
GERP++	μ = 0.179 [-3.2 – 2.26]	potentially regulated gene(s)	AK4, DNAJC6, RP4-535B20.1
CADD score	μ = 4.010 [0.187 – 14.56]	disease gene(s)	LEPR, DNAJC6

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
LEPR	LEPTIN RECEPTOR DEFICIENCY	OMIM	MIM:614963
DNAJC6	PARKINSON DISEASE 19, JUVENILE-ONSET	OMIM	MIM:615528
LEPR	Obesity due to leptin receptor gene deficiency	OrphaNet	OrphaNet:179494
DNAJC6	Atypical juvenile parkinsonism	OrphaNet	OrphaNet:391411

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LEPR	?	ENSG00000116678	blood	2.43×10^{-7} (p-value)	GTEx Portal V6	19
LEPR	?	ENSG00000116678	subcutaneous adipocytes	1.63×10^{-7} (p-value)	GTEx Portal V6	20
LEPROT	ENST00000488747	ILMN_1661537	skin	4.64×10^{-11} (p-value)	MuTHER consortium	11
LEPROT	ENST00000475108		blood	7.39×10^{-16} (p-value)	MuTHER consortium	11
LEPROT	ENST00000613538		adipocyte	2.44×10^{-16} (p-value)	MuTHER consortium	11
LEPROT	ENST00000371065					
LEPR	ENST00000349533	ILMN_1696391	adipocyte	4.17×10^{-6} (p-value)	MuTHER consortium	3
LEPR	ENST00000616738					
LEPR	ENST00000371060					
LEPR	ENST00000406510					
LEPR	ENST00000616738	ILMN_2234956	monocyte	3.79×10^{-20} (p-value)	Zeller et al.	6
LEPR	ENST00000371060					
LEPROT	ENST00000488747	ILMN_1661537	monocyte	3.38×10^{-15} (p-value)	Fairfax et al.	2
LEPROT	ENST00000613538		b-cell	2.00×10^{-15} (p-value)	Fairfax et al.	2
LEPROT	ENST00000475108					
LEPROT	ENST00000371065					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000023290	1	ENCP00000002874	RP4-535B20.1
ENCE00000023303	1	ENCP00000002886	DNAJC6
		ENCP00000002877	AK4

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000538286 (promoter flanking region)	5	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1, H3K36me3
		HSMMtube	DNase1
		Osteobl	H3K27ac
		blood (K562)	DNase1, Max, CTCF, H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K4me1
		endothelium (HUVEC)	DNase1
		liver (HepG2)	HNF4G, DNase1, USF1, H3K4me1, p300, H3K4me2, HNF4A, FOXA1, H3K27ac, CTCF
		nervous (NH-A)	DNase1
		A549	H3K4me3
ENSR00000670217 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1
		liver (HepG2)	H3K4me1, FOXA1, H3K27ac
		blood (K562)	H3K27ac, H3K9ac, H3K79me2, PolII, H3K4me2, H3K36me3, H3K4me3
ENSR00001521210 (promoter flanking region)	4	liver (HepG2)	H3K4me1
		Osteobl	H3K4me2
		blood (K562)	H3K4me2, H3K36me3, H3K4me3, PolII, Max, DNase1, H3K27ac, H3K9ac, USF1, Cmyc, H3K79me2
		muscle (HSMM)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP4-630A11.3	upstream gene variant	192	ENST00000429871	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LEPR	ENST00000371059	NM_001198687.1, NM_001003680.3	ENSP00000360098	20
LEPR	ENST00000371060	NM_001003679.3	ENSP00000360099	20
LEPR	ENST00000349533	NM_002303.5	ENSP00000330393	20
LEPR	ENST00000406510	?	ENSP00000384025	20
RP4-630A11.3	ENST00000429871	?	?	3