SNiPAcard

Block annotations

Block info

genomic range	chr1:66,062,325-66,170,362
block size	108,038 bp
variant count	191 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.388 [-3.75 – 2.308]	**gene(s) hit or close-by**	LEPR
phastCons	μ = 0.082 [0 – 0.971]	**eQTL gene(s)**	–
GERP++	μ = -0.270 [-5.29 – 4.15]	**potentially regulated gene(s)**	RP11-397C12.1
CADD score	μ = 3.009 [0.046 – 16.78]	**disease gene(s)**	LEPR

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Fasting plasma glucose (childhood)	$<5.00\times10^{-8}$	GWAS Catalog	25223902	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Leptin receptor polymorphism	benign	ClinVar	RCV000009050.1	1
?	HGMD curated	HGMD	CM025915	1
LEPTIN RECEPTOR POLYMORPHISM	OMIM curated	OMIM	MIM:601007	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 transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
LEPR	missense variant	ENST00000349533	NM_002303.5	ENSP00000330393	N/K	aaC/aaG	?	?	1
LEPR	missense variant	ENST00000371058	?	ENSP00000360097	N/K	aaC/aaG	?	?	1
LEPR	missense variant	ENST00000616738	NM_001198689.1	ENSP00000483390	N/K	aaC/aaG	?	?	1
LEPR	missense variant	ENST00000371060	NM_001003679.3	ENSP00000360099	N/K	aaC/aaG	?	?	1
LEPR	missense variant	ENST00000344610	NM_001198688.1	ENSP00000340884	N/K	aaC/aaG	?	?	1
LEPR	missense variant	ENST00000371059	NM_001198687.1, NM_001003680.3	ENSP00000360098	N/K	aaC/aaG	?	?	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000023340	1	ENCP00000002897	RP11-397C12.1
ENCE00000023461	1	ENCP00000002897	RP11-397C12.1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001521219 (open chromatin region)	2	blood (K562)	H3K36me3
		muscle (HSMM)	DNase1
ENSR00001580863 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	CTCF
		lung (IMR90)	CTCF
ENSR00001521220 (CTCF binding site)	2	blood (K562)	H3K36me3
ENSR00001580864 (CTCF binding site)	2	cervix (HeLa-S3)	H3K27me3
		A549	H3K27me3
ENSR00001521221 (enhancer)	2	cervix (HeLa-S3)	H3K27me3
		A549	H3K27me3
ENSR00001521223 (CTCF binding site)	7	endothelium (HUVEC)	H3K27me3
		embryonic stem cell (H1ESC)	Rad21
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
LEPR	downstream gene variant	2903	ENST00000406510	?	ENSP00000384025	6
LEPR	upstream gene variant, downstream gene variant	98	ENST00000471762	?	?	8
LEPR	downstream gene variant	183	ENST00000616738	NM_001198689.1	ENSP00000483390	7
LEPR	downstream gene variant	649	ENST00000344610	NM_001198688.1	ENSP00000340884	4
LEPR	downstream gene variant	1	ENST00000462765	?	?	4
LEPR	downstream gene variant	649	ENST00000371058	?	ENSP00000360097	4
LEPR	downstream gene variant	164	ENST00000371060	NM_001003679.3	ENSP00000360099	7
LEPR	downstream gene variant	820	ENST00000371059	NM_001198687.1, NM_001003680.3	ENSP00000360098	5
LEPR	downstream gene variant	1099	ENST00000349533	NM_002303.5	ENSP00000330393	6

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LEPR	ENST00000371058	?	ENSP00000360097	27
LEPR	ENST00000349533	NM_002303.5	ENSP00000330393	37
LEPR	ENST00000371060	NM_001003679.3	ENSP00000360099	36
LEPR	ENST00000616738	NM_001198689.1	ENSP00000483390	36
LEPR	ENST00000371059	NM_001198687.1, NM_001003680.3	ENSP00000360098	31
LEPR	ENST00000462765	?	?	14
LEPR	ENST00000406510	?	ENSP00000384025	38

LEPR	ENST00000344610	NM_001198688.1	ENSP00000340884	27
LEPR	ENST00000471762	?	?	4

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
LEPR	ENST00000349533	NM_002303.5	ENSP00000330393	8
LEPR	ENST00000371060	NM_001003679.3	ENSP00000360099	2
LEPR	ENST00000616738	NM_001198689.1	ENSP00000483390	2