Locus 287 (rs4655537)

SNiPAcard

Block annotations

Block info

genomic range	chr1:66,054,995-66,058,801
block size	3,807 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.479 [-0.627 – -0.331]	gene(s) hit or close-by	LEPR
phastCons	μ = 0.000 [0 – 0]	eQTL gene(s)	–
GERP++	μ = 0.661 [-0.258 – 1.58]	potentially regulated gene(s)	–
CADD score	μ = 3.693 [2.605 – 4.781]	disease gene(s)	LEPR

Trait annotations

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

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LEPR	ENST00000371060	NM_001003679.3	ENSP00000360099	2
LEPR	ENST00000344610	NM_001198688.1	ENSP00000340884	2
LEPR	ENST00000349533	NM_002303.5	ENSP00000330393	2
LEPR	ENST00000371059	NM_001198687.1, NM_001003680.3	ENSP00000360098	2
LEPR	ENST00000616738	NM_001198689.1	ENSP00000483390	2
LEPR	ENST00000406510	?	ENSP00000384025	2
LEPR	ENST00000462765	?	?	2
LEPR	ENST00000371058	?	ENSP00000360097	2