

## Block annotations

### Block info

genomic range	chr1:66,054,995-66,058,801 <i>e!</i>
block size	3,807 bp
variant count	2 variants

### Basic features

Conservation/deleteriousness		Linked genes	
phyloP	$\mu = -0.479$ [-0.627 -- -0.331]	gene(s) hit or close-by	LEPR <i>e!</i>
phastCons	$\mu = 0.000$ [0 - 0]	eQTL gene(s)	-
GERP++	$\mu = 0.661$ [-0.258 - 1.58]	potentially regulated gene(s)	-
CADD score	$\mu = 3.693$ [2.605 - 4.781]	disease gene(s)	LEPR <i>e!</i>

## Trait annotations

### Disease gene annotation

gene	trait	source DB	source entry/link
LEPR <i>e!</i>	LEPTIN RECEPTOR DEFICIENCY	OMIM	MIM:614963 
LEPR <i>e!</i>	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 <i>e!</i>	ENST00000371060 <i>e!</i>	NM_001003679.3	ENSP00000360099 <i>e!</i>	2
LEPR <i>e!</i>	ENST00000344610 <i>e!</i>	NM_001198688.1	ENSP00000340884 <i>e!</i>	2
LEPR <i>e!</i>	ENST00000349533 <i>e!</i>	NM_002303.5	ENSP00000330393 <i>e!</i>	2
LEPR <i>e!</i>	ENST00000371059 <i>e!</i>	NM_001198687.1, NM_001003680.3	ENSP00000360098 <i>e!</i>	2
LEPR <i>e!</i>	ENST00000616738 <i>e!</i>	NM_001198689.1	ENSP00000483390 <i>e!</i>	2
LEPR <i>e!</i>	ENST00000406510 <i>e!</i>	?	ENSP00000384025 <i>e!</i>	2
LEPR <i>e!</i>	ENST00000462765 <i>e!</i>	?	?	2
LEPR <i>e!</i>	ENST00000371058 <i>e!</i>	?	ENSP00000360097 <i>e!</i>	2

