Locus 73 (rs947409)

SNiPAcard

Block annotations

Block info

genomic range	chr13:47,341,767-47,391,824
block size	50,058 bp
variant count	27 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.443 [-1.875 – 0.869]	gene(s) hit or close-by	ESD
phastCons	μ = 0.008 [0 – 0.077]	eQTL gene(s)	–
GERP++	μ = -0.219 [-3.58 – 4.31]	potentially regulated gene(s)	–
CADD score	μ = 3.585 [0.087 – 11.38]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Iron	3.02×10-4	dbGaP	pha002876 dbGaP	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
?	?	ILMN_1720285	liver	1.26×10-12 (p-value)	Innocenti et al.	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001509548 (CTCF binding site)	1	NHLF	H3K36me3, DNase1
		embryonic stem cell (H1ESC)	H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3
		A549	H3K36me3
ENSR00001509551 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27ac, DNase1
		Osteobl	H3K27ac
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K27ac, H3K36me3, DNase1
		breast (HMEC)	DNase1
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	DNase1

		nervous (NH-A)	DNase1, H3K27ac	
		skin (NHEK)	DNase1	
ENSR00001509552 (enhancer)	1	blood (DND-41)	H3K27me3	
		muscle (HSMM)	DNase1	

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ESD	downstream gene variant, upstream gene variant	395	ENST00000412582	?	ENSP00000391350	6
ESD	downstream gene variant, upstream gene variant	1856	ENST00000378697	?	ENSP00000367969	5
ESD	downstream gene variant, upstream gene variant	1777	ENST00000378720	NM_001984.1	ENSP00000367992	6
ESD	downstream gene variant, upstream gene variant	398	ENST00000471867	?	ENSP00000476193	3
ESD	downstream gene variant	1110	ENST00000495654	?	?	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ESD	ENST00000412582	?	ENSP00000391350	6
ESD	ENST00000378697	?	ENSP00000367969	15
ESD	ENST00000378720	NM_001984.1	ENSP00000367992	15
ESD	ENST00000471867	?	ENSP00000476193	5
ESD	ENST00000495654	?	?	9

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ESD	ENST00000471867	?	ENSP00000476193	2