Locus 100 (rs2518136)

SNiPAcard

Block annotations

Block info

genomic range	chr3:186,335,941-186,341,830
block size	5,890 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.386 [-1.316 – 0.255]	gene(s) hit or close-by	AHSG, RP11-573D15.8
phastCons	μ = 0.000 [0 – 0.001]	eQTL gene(s)	–
GERP++	μ = 0.464 [-3.04 – 3.81]	potentially regulated gene(s)	–
CADD score	μ = 3.943 [0.153 – 11.52]	disease gene(s)	–

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Calcium oxalate urolithiasis	association	ClinVar	RCV000128583.1 ClinVar	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001486982 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, Yy1, DNase1
		HSMMtube	DNase1, H3K27me3, CTCF
		blood (K562)	CTCF, Rad21, H3K27me3
		skin (NHDF-AD)	H3K4me2, DNase1, H3K4me3, CTCF
		muscle (HSMM)	H3K4me2, CTCF, H2AZ, DNase1
		liver (HepG2)	DNase1, H3K36me3, CTCF, Rad21, PolII, H4K20me1
		blood (GM12878)	DNase1, H2AZ, Rad21, H3K4me2, CTCF
		lung (IMR90)	DNase1, H3K4me2, H3K27me3, CTCF
		nervous (NH-A)	CTCF, H3K4me2, DNase1
		skin (NHEK)	DNase1, CTCF, H3K4me3, H3K4me2
		NHLF	CTCF, DNase1
		Osteobl	H2AZ, H3K4me2, CTCF, H3K27me3
		blood (DND-41)	H3K27me3, CTCF
		breast (HMEC)	CTCF, H3K4me2, DNase1
		cervix (HeLa-S3)	Max, CTCF, H3K27me3, DNase1, H3K4me2
		monocytes (Monocytes-CD14+)	CTCF, H3K27me3
		endothelium (HUVEC)	H3K36me3, CTCF, H3K27me3, DNase1
		A549	H3K27me3, DNase1, CTCF, H3K4me3, H3K4me2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AHSG	downstream gene variant	163	ENST00000478441	?	?	3
AHSG	downstream gene variant	2286	ENST00000411641	NM_001622.2	ENSP00000393887	3
AHSG	downstream gene variant	2304	ENST00000273784	?	ENSP00000273784	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AHSG	ENST00000273784	?	ENSP00000273784	3
AHSG	ENST00000411641	NM_001622.2	ENSP00000393887	3
RP11-573D15.8	ENST00000625386	?	?	6
RP11-573D15.8	ENST00000630178	?	?	6
RP11-573D15.8	ENST00000628505	?	?	6