SNiPAcard

Block annotations

Block info

genomic range	chr1:206,885,839-206,904,464
block size	18,626 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.577 [-2.101 – 0.376]	**gene(s) hit or close-by**	MAPKAPK2
phastCons	μ = 0.001 [0 – 0.006]	**eQTL gene(s)**	IL19
GERP++	μ = -0.975 [-4.84 – 2.73]	**potentially regulated gene(s)**	IKBKE, IL19
CADD score	μ = 4.853 [0.834 – 12.73]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IL19	ENST00000620365	ILMN_1799575	blood	2.78×10-6 (p-value)	MuTHER consortium	5
IL19	ENST00000270218					
IL19	ENST00000340758					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000052952	1	ENCP00000006333	IKBKE
		ENCP00000006352	IL19

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001040819 (promoter flanking region)	1	NHLF	H3K27ac
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K79me2
		Osteobl	H3K36me3, H3K27ac
		blood (K562)	H3K79me2, H3K4me1, H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K79me2
		cervix (HeLa-S3)	Jund, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H4K20me1
		lung (IMR90)	DNase1, H4K20me1, H3K79me2
		blood (GM12878)	H3K79me2, H3K27ac
		A549	H3K4me3, H3K4me2, DNase1, H3K27ac
ENSR00000551719 (promoter flanking region)	2	HSMMtube	H3K79me2
		blood (K562)	DNase1, H3K79me2, H3K4me1
		muscle (HSMM)	H3K79me2
		monocytes (Monocytes-CD14+)	H4K20me1
		liver (HepG2)	H3K36me3
		lung (IMR90)	H4K20me1, H3K79me2
		blood (GM12878)	Srf, DNase1, H3K79me2, H3K27ac
		skin (NHEK)	H3K36me3
ENSR00000672024	1	embryonic stem cell (H1ESC)	Rad21, CTCF, Tcf12, DNase1
(CTCF binding site)		HSMMtube	DNase1, CTCF

blood (K562)	DNase1, H3K27me3, Rad21, H4K20me1, CTCF
skin (NHDF-AD)	CTCF
muscle (HSMM)	H3K79me2, CTCF, H4K20me1, DNase1
liver (HepG2)	CTCF
lung (IMR90)	H4K20me1, H3K36me3, CTCF
blood (GM12878)	Pax5, CTCF, H3K79me2, Rad21, DNase1, H3K4me1
nervous (NH-A)	H4K20me1, CTCF
skin (NHEK)	CTCF, H3K36me3
NHLF	CTCF
Osteobl	H4K20me1, CTCF
blood (DND-41)	CTCF, H3K36me3
breast (HMEC)	CTCF
cervix (HeLa-S3)	CTCF, DNase1, H3K4me2, H3K4me1
monocytes (Monocytes-CD14+)	CTCF, H4K20me1, H3K36me3
endothelium (HUVEC)	H3K36me3, CTCF
A549	H3K4me2, CTCF, DNase1, H3K27ac

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MAPKAPK2	upstream gene variant, downstream gene variant	897	ENST00000493447	?	?	4
MAPKAPK2	upstream gene variant	169	ENST00000479009	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
MAPKAPK2	ENST00000294981	NM_004759.4	ENSP00000294981	7
MAPKAPK2	ENST00000367103	NM_032960.3	ENSP00000356070	7