Locus 30 (rs33995560)

SNiPAcard

Block annotations

Block info

genomic range	chr17:34,303,312-34,306,470
block size	3,159 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.168 [-0.463 – 0.853]	**gene(s) hit or close-by**	CCL14, CCL15-CCL14, CCL16, CTB-186H2.3
phastCons	μ = 0.141 [0 – 0.78]	**eQTL gene(s)**	–
GERP++	μ = 0.991 [-0.722 – 3.54]	**potentially regulated gene(s)**	–
CADD score	μ = 4.572 [1.506 – 8.195]	**disease gene(s)**	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001344901 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		Osteobl	CTCF
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	DNase1, H3K4me1, CTCF
		blood (GM12878)	CTCF, Rad21
		A549	CTCF
		skin (NHEK)	CTCF
ENSR00001638275 (enhancer)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	H3K27me3
		Osteobl	CTCF
		blood (K562)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		liver (HepG2)	H3K4me1
		blood (GM12878)	CTCF, Rad21
		A549	CTCF, H3K27me3
ENSR00001638276 (enhancer)	3	cervix (HeLa-S3)	H3K27me3
		HSMMtube	H3K27me3
		liver (HepG2)	H3K4me1
		blood (K562)	H3K27me3
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CCL14	downstream gene variant	3857	ENST00000614009	?	ENSP00000483186	1
CCL14	downstream gene variant	3860	ENST00000618404	NM_032963.3	ENSP00000481023	1
CCL14	downstream gene variant	4253	ENST00000620991	?	ENSP00000484818	1
CCL14	downstream gene variant	4225	ENST00000622526	NM_032962.4	ENSP00000479097	1
CCL15-CCL14	downstream gene variant	4222	ENST00000610751	?	ENSP00000481940	1
CCL15-CCL14	downstream gene variant	4222	ENST00000616694	?	ENSP00000481402	1

CCL16	downstream gene variant, upstream gene variant	217	ENST00000613642	?	ENSP00000478592	2
CCL16	downstream gene variant	223	ENST00000611905	NM_004590.2	ENSP00000478024	1
CCL16	downstream gene variant	292	ENST00000610493	?	ENSP00000478934	1
CTB-186H2.3	upstream gene variant	4222	ENST00000617687	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL16	ENST00000611905	NM_004590.2	ENSP00000478024	6
CCL16	ENST00000610493	?	ENSP00000478934	6
CCL16	ENST00000613642	?	ENSP00000478592	5

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL16	ENST00000611905	NM_004590.2	ENSP00000478024	2
CCL16	ENST00000610493	?	ENSP00000478934	2