

Block annotations

Block info

genomic range	chr17:34,303,312-34,306,470 <i>e!</i>
block size	3,159 bp
variant count	7 variants

Basic features

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

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001344901 <i>e!</i> (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 <i>e!</i> (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 <i>e!</i> (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 <i>e!</i>	downstream gene variant	3857	ENST00000614009 <i>e!</i>	? ?	ENSP00000483186 <i>e!</i>	1
CCL14 <i>e!</i>	downstream gene variant	3860	ENST00000618404 <i>e!</i>	NM_032963.3	ENSP00000481023 <i>e!</i>	1
CCL14 <i>e!</i>	downstream gene variant	4253	ENST00000620991 <i>e!</i>	? ?	ENSP00000484818 <i>e!</i>	1
CCL14 <i>e!</i>	downstream gene variant	4225	ENST00000622526 <i>e!</i>	NM_032962.4	ENSP00000479097 <i>e!</i>	1
CCL15-CCL14 <i>e!</i>	downstream gene variant	4222	ENST00000610751 <i>e!</i>	? ?	ENSP00000481940 <i>e!</i>	1
CCL15-CCL14 <i>e!</i>	downstream gene variant	4222	ENST00000616694 <i>e!</i>	? ?	ENSP00000481402 <i>e!</i>	1

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

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL16 <i>e!</i>	ENST00000611905 <i>e!</i>	NM_004590.2	ENSP00000478024 <i>e!</i>	6
CCL16 <i>e!</i>	ENST00000610493 <i>e!</i>	?	ENSP00000478934 <i>e!</i>	6
CCL16 <i>e!</i>	ENST00000613642 <i>e!</i>	?	ENSP00000478592 <i>e!</i>	5

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL16 <i>e!</i>	ENST00000611905 <i>e!</i>	NM_004590.2	ENSP00000478024 <i>e!</i>	2
CCL16 <i>e!</i>	ENST00000610493 <i>e!</i>	?	ENSP00000478934 <i>e!</i>	2

