Locus 33 (rs7221769)

SNiPAcard

Block annotations

Block info

genomic range	chr17:34,327,923-34,338,835
block size	10,913 bp
variant count	44 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.596 [-2.574 – 0.251]	gene(s) hit or close-by	CCL15, CCL15-CCL14, CCL23, RP11-104J23.1, RP11-104J23.2
phastCons	μ = 0.145 [0.001 – 0.938]	eQTL gene(s)	–
GERP++	μ = -0.086 [-2.21 – 0.158]	potentially regulated gene(s)	CCL14, CCL14, CCL15, CCL15, CCL16, CCL16
CADD score	μ = 4.547 [0.08 – 16.42]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
?	?	ENSG00000256515	blood	1.82×10^{-2} (q-value)	SeeQTL DB (HapMap)	2
?	?	ENSG00000006075	blood	4.84×10^{-2} (q-value)	SeeQTL DB (HapMap)	1
?	?	ENSG00000129277	blood	1.08×10^{-2} (q-value)	SeeQTL DB (HapMap)	2
?	?	ENSG00000205021	blood	1.16×10^{-2} (q-value)	SeeQTL DB (HapMap)	2

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
CTCF	MA0139.1	1	no	0.000	1

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000022625		CCL15

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000204082	1	ENCP00000022625	CCL15 CCL15
		ENCP00000022623	CCL16 CCL16
ENCE00000204075	2	ENCP00000022624	CCL14 CCL14
		ENCP00000022622	CCL16 CCL16

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001344912 (promoter)	2	cervix (HeLa-S3)	H3K27me3
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3

		liver (HepG2)	H3K4me3, PolII, DNase1, RXRA, H3K4me1, p300, H3K4me2, FOXA1, H3K9ac, H3K27ac, H3K27me3
		blood (DND-41)	H3K27me3
		A549	H3K27me3
		blood (K562)	H3K27me3
ENSR00001344915	4	embryonic stem cell (H1ESC)	CTCF, Rad21, DNase1
(CTCF binding site)		Osteobl	CTCF
		blood (K562)	CTCF, Rad21, Max, H3K27me3
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	CTCF
		lung (IMR90)	CTCF
		A549	CTCF, H3K27me3
		skin (NHEK)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CCL15	upstream gene variant	2514	ENST00000614368	?	ENSP00000484262	11
CCL15	upstream gene variant	143	ENST00000617897	NM_032965.4	ENSP00000484078	19
CCL15-CCL14	upstream gene variant	377	ENST00000616694	?	ENSP00000481402	16
CCL15-CCL14	upstream gene variant	447	ENST00000610751	?	ENSP00000481940	15
CCL23	downstream gene variant	1262	ENST00000613876	?	ENSP00000479076	18
CCL23	downstream gene variant	1263	ENST00000615050	NM_005064.4	ENSP00000481357	18
CCL23	downstream gene variant	1261	ENST00000612516	NM_145898.2	ENSP00000484748	18
RP11-104J23.1	upstream gene variant, downstream gene variant	20	ENST00000619334	?	?	9
RP11-104J23.1	upstream gene variant, downstream gene variant	41	ENST00000617328	?	?	19
RP11-104J23.2	upstream gene variant	709	ENST00000617747	?	?	19

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL15	ENST00000617897	NM_032965.4	ENSP00000484078	2
CCL15-CCL14	ENST00000616694	?	ENSP00000481402	1
CCL15-CCL14	ENST00000610751	?	ENSP00000481940	1
RP11-104J23.1	ENST00000619334	?	?	34
RP11-104J23.1	ENST00000617328	?	?	3

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL15	ENST00000617897	NM_032965.4	ENSP00000484078	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-104J23.1	ENST00000619334	?	1
RP11-104J23.1	ENST00000617328	?	8

Locus 33 (sentinel SNP rs7221769 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.