Locus 14 (rs17482078)

SNiPAcard

Block annotations

Block info

genomic range	chr5:96,118,866-96,161,069
block size	42,204 bp
variant count	23 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.139 [-5.113 – 4.515]	gene(s) hit or close-by	AC008906.1, CAST, CTD-2260A17.1, CTD-2260A17.2, CTD-2260A17.3, ERAP1
phastCons	μ = 0.153 [0 – 1]	eQTL gene(s)	CAST, CTC-506B8.1, CTD-2260A17.1, ERAP1
GERP++	μ = 0.119 [-5.73 – 5.78]	potentially regulated gene(s)	CTD-2215E18.2, ERAP1, ERAP2, LIX1, LNPEP
CADD score	μ = 5.730 [0.159 – 21.1]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Behcet's disease	<4.00×10-8	GWAS Catalog	23291587	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM0911245	1

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
ERAP1	missense variant	ENST00000443439	NM_001040458.1, NM_001198541.1	ENSP00000406304	3	3			3
ERAP1	missense variant	ENST00000296754	NM_016442.3	ENSP00000296754	3	3			3

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ERAP1	ENST00000296754	ILMN_1752145	blood	3.69×10-17 (p-value)	MuTHER consortium	10
			adipocyte	5.78×10-8 (p-value)	MuTHER consortium	10
ERAP1	?	ENSG00000164307	caudate basal ganglia	3.10×10-8 (p-value)	GTEx Portal V6	23
ERAP1	?	ENSG00000164307	skeletal muscle	2.85×10-21 (p-value)	GTEx Portal V6	23
CTD-2260A17.1	?	ENSG00000248734	skeletal muscle	1.88×10-10 (p-value)	GTEx Portal V6	23
ERAP1	?	ENSG00000164307	putamen	2.52×10-6 (p-value)	GTEx Portal V6	2

ERAP1	?	ENSG00000164307	EBV lymphocytes	6.40×10-7 (p-value)	GTEx Portal V6	2
CTD-2260A17.1	?	ENSG00000248734	ovary	3.35×10-8 (p-value)	GTEx Portal V6	22
ERAP1	?	ENSG00000164307	blood	7.47×10-7 (p-value)	GTEx Portal V6	4
ERAP1	ENST00000296754	ILMN_1752145	monocyte	7.42×10-8 (p-value)	Fairfax et al.	3
ERAP1	ENST00000296754	214012_at	blood	1.40×10-22 (p-value)	Dixon et al.	1
ERAP1	ENST00000443439	209788_s_at	blood	9.40×10-17 (p-value)	Dixon et al.	1
CTC-506B8.1	?	ENSG00000249180	subcutaneous adipocytes	8.39×10-6 (p-value)	GTEx Portal V6	9
ERAP1	ENST00000443439	ILMN_2336220	monocyte	8.06×10-37 (p-value)	Zeller et al.	3
ERAP1	ENST00000514604					
ERAP1	ENST00000296754					
ERAP1	ENST00000296754	ILMN_1752145	monocyte	9.66×10-24 (p-value)	Zeller et al.	3
CAST	ENST00000511782	ILMN_1783627	blood	2.96×10-6 (p-value)	Westra et al.	1
CAST	ENST00000511049					
CAST	ENST00000395813					
CAST	ENST00000421689					
CAST	ENST00000506811					
CAST	ENST00000510156					
CAST	ENST00000507487					
CAST	ENST00000341926					
CAST	ENST00000508117					
CAST	ENST00000508830					
CAST	ENST00000338252					
CAST	ENST00000505143					
CAST	ENST00000514055					

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
CJUN	MA0489.1	1	no	0.000	1
CJUN	MA0303.1	2	no	0.000	1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000417173	1	ENCP00000044982	LIX1
ENCE00000417059	1	ENCP00000044978	LNPEP
		ENCP00000044966	ERAP1
		ENCP00000044987	CTD-2215E18.2
		ENCP00000044971	ERAP2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001285985 (promoter flanking region)	1	NHLF	H3K36me3
		embryonic stem cell (H1ESC)	DNase1
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	H3K4me1
		cervix (HeLa-S3)	DNase1

monocytes (Monocytes-CD14+)	DNase1, H3K36me3
endothelium (HUVEC)	H3K36me3, Cjun, DNase1
liver (HepG2)	H3K36me3
blood (GM12878)	H3K36me3
lung (IMR90)	H3K36me3
nervous (NH-A)	H3K36me3
A549	H3K36me3
skin (NHEK)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC008906.1	upstream gene variant, downstream gene variant	261	ENST00000629418	?	?	9
CAST	downstream gene variant	3567	ENST00000510098	?	ENSP00000427195	2
CTD-2260A17.1	upstream gene variant, downstream gene variant	507	ENST00000512856	?	?	6
CTD-2260A17.1	upstream gene variant, downstream gene variant	507	ENST00000602972	?	?	6
CTD-2260A17.3	upstream gene variant	630	ENST00000606346	?	?	6
CTD-2260A17.3	upstream gene variant	190	ENST00000606656	?	?	5
ERAP1	upstream gene variant	1981	ENST00000512852	?	ENSP00000425381	4
ERAP1	downstream gene variant, upstream gene variant	100	ENST00000507859	?	?	7
ERAP1	downstream gene variant	2331	ENST00000508227	?	ENSP00000422631	3
ERAP1	downstream gene variant	1467	ENST00000503921	?	ENSP00000427025	3
ERAP1	downstream gene variant	177	ENST00000507154	?	ENSP00000421697	6
ERAP1	downstream gene variant, upstream gene variant	161	ENST00000503311	?	?	10
ERAP1	upstream gene variant	966	ENST00000514604	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTD-2260A17.1	ENST00000602972	?	?	2
CTD-2260A17.1	ENST00000512856	?	?	2
CTD-2260A17.2	ENST00000501338	?	?	2
CTD-2260A17.2	ENST00000502262	?	?	2
CTD-2260A17.2	ENST00000504056	?	?	2
CTD-2260A17.3	ENST00000606656	?	?	1
ERAP1	ENST00000508227	?	ENSP00000422631	6
ERAP1	ENST00000443439	NM_001040458.1, NM_001198541.1	ENSP00000406304	18
ERAP1	ENST00000296754	NM_016442.3	ENSP00000296754	18
ERAP1	ENST00000507154	?	ENSP00000421697	2
ERAP1	ENST00000503921	?	ENSP00000427025	9
ERAP1	ENST00000507859	?	?	3
ERAP1	ENST00000514604	?	?	4
ERAP1	ENST00000503311	?	?	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)

CTD-2260A17.1	ENST00000602972	?	1
CTD-2260A17.1	ENST00000512856	?	1
ERAP1	ENST00000507859	?	2
ERAP1	ENST00000514604	?	4

Locus 14 (sentinel SNP rs17482078 + 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.