Locus 11 (rs26496)

SNiPAcard

Block annotations

Block info

genomic range	chr5:96,123,055-96,151,968
block size	28,914 bp
variant count	25 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.605 [-2.841 – 0.73]	gene(s) hit or close-by	AC008906.1, CTD-2260A17.1, CTD-2260A17.2, CTD-2260A17.3, ERAP1
phastCons	μ = 0.067 [0 – 0.895]	eQTL gene(s)	CTD-2260A17.1, CTD-2260A17.3, ERAP1, ERAP2, LNPEP
GERP++	μ = -0.570 [-10.1 – 2.31]	potentially regulated gene(s)	–
CADD score	μ = 2.945 [0.098 – 9.842]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Ankylosing spondylitis	$<5.00\times10^{-12}$	GWAS Catalog	20062062	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM0911244	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ERAP1	?	ENSG00000164307	pancreas	4.21×10^{-23} (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	pancreas	4.98×10^{-6} (p-value)	GTEx Portal V6	3
ERAP1	?	ENSG00000164307	muscularis mucosae	2.83×10^{-8} (p-value)	GTEx Portal V6	24
ERAP1	?	ENSG00000164307	lung	2.55×10^{-33} (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	lung	8.96×10^{-9} (p-value)	GTEx Portal V6	24
CTD-2260A17.3	?	ENSG00000272109	lung	1.00×10^{-13} (p-value)	GTEx Portal V6	25
CTD-2260A17.3	?	ENSG00000272109	atrial appendage	2.69×10^{-6} (p-value)	GTEx Portal V6	6
ERAP1	?	ENSG00000164307	atrial appendage	1.23×10^{-6} (p-value)	GTEx Portal V6	5
ERAP1	?	ENSG00000164307	transformed fibroblasts	1.87×10^{-38} (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	transformed fibroblasts	2.19×10^{-28} (p-value)	GTEx Portal V6	25
ERAP1	?	ENSG00000164307	blood	1.62×10^{-45} (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	blood	3.22×10^{-17} (p-value)	GTEx Portal V6	25
CTD-2260A17.3	?	ENSG00000272109	blood	4.44×10^{-11} (p-value)	GTEx Portal V6	25
ERAP1	?	ENSG00000164307	breast	2.06×10^{-10} (p-value)	GTEx Portal V6	24
CTD-2260A17.3	?	ENSG00000272109	breast	7.84×10^{-7} (p-value)	GTEx Portal V6	21

ERAP2	?	ENSG00000164308	breast	6.95×10-6 (p-value)	GTEx Portal V6	1
ERAP1	?	ENSG00000164307	tibial artery	6.71×10-8 (p-value)	GTEx Portal V6	24
CTD-2260A17.3	?	ENSG00000272109	tibial artery	1.01×10-6 (p-value)	GTEx Portal V6	21
ERAP1	?	ENSG00000164307	thyroid	3.20×10-10 (p-value)	GTEx Portal V6	24
CTD-2260A17.3	?	ENSG00000272109	thyroid	9.13×10-6 (p-value)	GTEx Portal V6	5
ERAP1	?	ENSG00000164307	skeletal muscle	9.90×10-22 (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	skeletal muscle	9.45×10-15 (p-value)	GTEx Portal V6	25
CTD-2260A17.3	?	ENSG00000272109	skeletal muscle	1.31×10-11 (p-value)	GTEx Portal V6	25
ERAP2	?	ENSG00000164308	skeletal muscle	7.63×10-7 (p-value)	GTEx Portal V6	4
ERAP1	?	ENSG00000164307	putamen	8.32×10-7 (p-value)	GTEx Portal V6	20
ERAP1	?	ENSG00000164307	EBV lymphocytes	2.08×10-15 (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	EBV lymphocytes	2.47×10-8 (p-value)	GTEx Portal V6	24
ERAP1	?	ENSG00000164307	transverse colon	1.92×10-18 (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	transverse colon	3.63×10-7 (p-value)	GTEx Portal V6	5
ERAP1	?	ENSG00000164307	liver	1.32×10-8 (p-value)	GTEx Portal V6	25
ERAP1	?	ENSG00000164307	sun exposed skin	8.36×10-25 (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	sun exposed skin	5.11×10-8 (p-value)	GTEx Portal V6	24
CTD-2260A17.3	?	ENSG00000272109	sun exposed skin	4.99×10-8 (p-value)	GTEx Portal V6	24
ERAP1	?	ENSG00000164307	unexposed skin	1.54×10-18 (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	unexposed skin	3.03×10-7 (p-value)	GTEx Portal V6	23
CTD-2260A17.3	?	ENSG00000272109	unexposed skin	1.23×10-8 (p-value)	GTEx Portal V6	25
ERAP2	?	ENSG00000164308	unexposed skin	1.00×10-5 (p-value)	GTEx Portal V6	1
ERAP1	?	ENSG00000164307	left ventricle	4.06×10-11 (p-value)	GTEx Portal V6	25
ERAP1	?	ENSG00000164307	subcutaneous adipocytes	4.70×10-18 (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	subcutaneous adipocytes	5.21×10-8 (p-value)	GTEx Portal V6	24
CTD-2260A17.3	?	ENSG00000272109	subcutaneous adipocytes	5.24×10-10 (p-value)	GTEx Portal V6	25
ERAP1	?	ENSG00000164307	caudate basal ganglia	8.05×10-7 (p-value)	GTEx Portal V6	20
ERAP1	?	ENSG00000164307	stomach	2.22×10-15 (p-value)	GTEx Portal V6	24
ERAP1	?	ENSG00000164307	visceral adipocytes	2.96×10-10 (p-value)	GTEx Portal V6	25
ERAP2	?	ENSG00000164308	visceral adipocytes	1.33×10-9 (p-value)	GTEx Portal V6	24
CTD-2260A17.3	?	ENSG00000272109	visceral adipocytes	3.80×10-8 (p-value)	GTEx Portal V6	25
ERAP1	?	ENSG00000164307	spleen	2.61×10-7 (p-value)	GTEx Portal V6	16
ERAP1	?	ENSG00000164307	coronary artery	1.88×10-7 (p-value)	GTEx Portal V6	23
ERAP1	?	ENSG00000164307	sigmoid colon	7.72×10-13 (p-value)	GTEx Portal V6	25
ERAP1	?	ENSG00000164307	tibial nerve	3.34×10-20 (p-value)	GTEx Portal V6	25

CTD-2260A17.3	?	ENSG00000272109	tibial nerve	1.80×10^{-6} (p-value)	GTEx Portal V6	23
ERAP1	?	ENSG00000164307	esophagus mucosa	2.25×10^{-25} (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	esophagus mucosa	2.32×10^{-7} (p-value)	GTEx Portal V6	24
CTD-2260A17.3	?	ENSG00000272109	esophagus mucosa	1.50×10^{-10} (p-value)	GTEx Portal V6	25
ERAP1	?	ENSG00000164307	gastroesophageal junction	6.19×10^{-7} (p-value)	GTEx Portal V6	23
ERAP1	?	ENSG00000164307	adrenal gland	4.65×10^{-13} (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	adrenal gland	1.73×10^{-6} (p-value)	GTEx Portal V6	11
CTD-2260A17.3	?	ENSG00000272109	testis	1.49×10^{-14} (p-value)	GTEx Portal V6	25
ERAP1	?	ENSG00000164307	frontal cortex	1.45×10^{-7} (p-value)	GTEx Portal V6	23
ERAP2	ENST00000513084	ILMN_1743145	skin	1.67×10^{-9} (p-value)	MuTHER consortium	9
ERAP2	ENST00000512869		blood	7.98×10^{-11} (p-value)	MuTHER consortium	9
ERAP2	ENST00000379904		adipocyte	1.05×10^{-9} (p-value)	MuTHER consortium	9
ERAP2	ENST00000437043					
ERAP1	ENST00000296754	ILMN_1752145	blood	1.25×10^{-5} (p-value)	MuTHER consortium	1
ERAP1	ENST00000296754	214012_at	blood	6.80×10^{-8} (p-value)	Dixon et al.	2
ERAP1	ENST00000443439	209788_s_at	blood	2.70×10^{-32} (p-value)	Dixon et al.	2
ERAP2	ENST00000513084	ILMN_1743145	monocyte	3.89×10^{-7} (p-value)	Fairfax et al.	2
ERAP2	ENST00000512869		b-cell	1.82×10^{-7} (p-value)	Fairfax et al.	2
ERAP2	ENST00000379904					
ERAP2	ENST00000437043					
ERAP1	ENST00000296754	ILMN_1752145	monocyte	3.32×10^{-6} (p-value)	Fairfax et al.	2
ERAP2	?	ENSG00000164308	blood	2.05×10^{-4} (q-value)	SeeQTL DB (HapMap)	3
LNPEP	ENST00000395770	ILMN_1814737	blood	7.60×10^{-6} (p-value)	Westra et al.	9
LNPEP	ENST00000231368					
CTD-2260A17.3	?	ENSG00000272109	aorta	1.22×10^{-5} (p-value)	GTEx Portal V6	2
ERAP2	ENST00000513084	ILMN_1743145	monocyte	6.82×10^{-23} (p-value)	Zeller et al.	2
ERAP2	ENST00000512869					
ERAP2	ENST00000379904					
ERAP2	ENST00000437043					
ERAP1	ENST00000443439	ILMN_2336220	monocyte	1.80×10^{-23} (p-value)	Zeller et al.	2
ERAP1	ENST00000514604					
ERAP1	ENST00000296754					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001412052 (enhancer)	1	blood (K562)	Max
ENSR00001285989 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K4me3
		embryonic stem cell (H1ESC)	DNase1
		blood (GM12878)	H3K27ac, H3K4me2, H3K9ac, H3K4me3, DNase1, PolII, H2AZ, H3K4me1
		blood (K562)	H2AZ
		A549	H3K27me3

blood (DND-41)	H3K9ac, H3K27ac, H3K4me1
skin (NHDF-AD)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC008906.1	upstream gene variant, downstream gene variant	7	ENST00000629418	?	?	9
CTD-2260A17.1	downstream gene variant	1353	ENST00000602972	?	?	6
CTD-2260A17.1	downstream gene variant	1353	ENST00000512856	?	?	6
CTD-2260A17.2	downstream gene variant	1954	ENST00000501338	?	?	1
CTD-2260A17.2	downstream gene variant	2100	ENST00000502262	?	?	1
CTD-2260A17.2	downstream gene variant	1958	ENST00000504056	?	?	1
CTD-2260A17.3	upstream gene variant	1195	ENST00000606656	?	?	7
CTD-2260A17.3	upstream gene variant	2509	ENST00000606346	?	?	7
ERAP1	downstream gene variant, upstream gene variant	1217	ENST00000503921	?	ENSP00000427025	6
ERAP1	downstream gene variant, upstream gene variant	2524	ENST00000508227	?	ENSP00000422631	6
ERAP1	upstream gene variant	592	ENST00000514604	?	?	6
ERAP1	upstream gene variant	4165	ENST00000443439	NM_001040458.1, NM_001198541.1	ENSP00000406304	1
ERAP1	upstream gene variant	1576	ENST00000507859	?	?	5
ERAP1	downstream gene variant, upstream gene variant	138	ENST00000503311	?	?	14
ERAP1	downstream gene variant, upstream gene variant	382	ENST00000507154	?	ENSP00000421697	8
ERAP1	upstream gene variant	3974	ENST00000296754	NM_016442.3	ENSP00000296754	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
ERAP1	ENST00000296754	NM_016442.3	ENSP00000296754	A	gcT/gcC	1
ERAP1	ENST00000443439	NM_001040458.1, NM_001198541.1	ENSP00000406304	A	gcT/gcC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTD-2260A17.2	ENST00000502262	?	?	1
CTD-2260A17.2	ENST00000504056	?	?	1
CTD-2260A17.2	ENST00000501338	?	?	1
ERAP1	ENST00000296754	NM_016442.3	ENSP00000296754	22
ERAP1	ENST00000508227	?	ENSP00000422631	7
ERAP1	ENST00000507859	?	?	6
ERAP1	ENST00000503311	?	?	2
ERAP1	ENST00000443439	NM_001040458.1, NM_001198541.1	ENSP00000406304	22
ERAP1	ENST00000503921	?	ENSP00000427025	11

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)

AC008906.1	ENST00000629418	?	1