Locus 120 (rs149313)

SNiPAcard

Block annotations

Block info

genomic range	chr5:96,057,882-96,070,205
block size	12,324 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.582 [-3.14 – 0.626]	**gene(s) hit or close-by**	CAST
phastCons	μ = 0.011 [0 – 0.051]	**eQTL gene(s)**	CAST, CTC-506B8.1, CTD-2260A17.1, ERAP1
GERP++	μ = 0.491 [0.225 – 1.08]	**potentially regulated gene(s)**	–
CADD score	μ = 4.473 [2.207 – 8.12]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ERAP1	?	ENSG00000164307	pancreas	1.38×10-7 (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	muscularis mucosae	2.46×10-9 (p-value)	GTEx Portal V6	5
CAST	?	ENSG00000153113	muscularis mucosae	2.49×10-10 (p-value)	GTEx Portal V6	5
ERAP1	?	ENSG00000164307	lung	1.19×10-7 (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	lung	6.30×10-8 (p-value)	GTEx Portal V6	3
CAST	?	ENSG00000153113	lung	1.78×10-10 (p-value)	GTEx Portal V6	5
ERAP1	?	ENSG00000164307	tibial nerve	1.85×10-5 (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	tibial nerve	8.33×10-9 (p-value)	GTEx Portal V6	5
CAST	?	ENSG00000153113	tibial nerve	7.19×10-11 (p-value)	GTEx Portal V6	5
ERAP1	?	ENSG00000164307	transformed fibroblasts	6.20×10-7 (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	transformed fibroblasts	1.87×10-8 (p-value)	GTEx Portal V6	5
CAST	?	ENSG00000153113	transformed fibroblasts	1.87×10-10 (p-value)	GTEx Portal V6	5
ERAP1	?	ENSG00000164307	blood	6.95×10-15 (p-value)	GTEx Portal V6	5
CTC-506B8.1	?	ENSG00000249180	blood	6.38×10-10 (p-value)	GTEx Portal V6	5
CAST	?	ENSG00000153113	blood	2.32×10-12 (p-value)	GTEx Portal V6	5
CTC-506B8.1	?	ENSG00000249180	tibial artery	2.72×10-10 (p-value)	GTEx Portal V6	5
CAST	?	ENSG00000153113	tibial artery	4.85×10-8 (p-value)	GTEx Portal V6	5
ERAP1	?	ENSG00000164307	esophagus mucosa	9.80×10-7 (p-value)	GTEx Portal V6	2
CTC-506B8.1	?	ENSG00000249180	thyroid	5.78×10-6 (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	thyroid	4.83×10-7 (p-value)	GTEx Portal V6	2
ERAP1	?	ENSG00000164307	skeletal muscle	6.41×10-15 (p-value)	GTEx Portal V6	5
CTD-2260A17.1	?	ENSG00000248734	skeletal muscle	1.45×10-7 (p-value)	GTEx Portal V6	5

ERAP1	?	ENSG00000164307	EBV lymphocytes	2.27×10-6 (p-value)	GTEx Portal V6	1
ERAP1	?	ENSG00000164307	transverse colon	2.42×10-6 (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	aorta	1.98×10-6 (p-value)	GTEx Portal V6	5
ERAP1	?	ENSG00000164307	subcutaneous adipocytes	2.15×10-6 (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	subcutaneous adipocytes	1.11×10-7 (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	subcutaneous adipocytes	9.10×10-7 (p-value)	GTEx Portal V6	1
CTD-2260A17.1	?	ENSG00000248734	testis	5.12×10-7 (p-value)	GTEx Portal V6	4
CAST	?	ENSG00000153113	adrenal gland	3.31×10-6 (p-value)	GTEx Portal V6	1
ERAP1	ENST00000296754	ILMN_1752145	blood	4.72×10-9 (p-value)	MuTHER consortium	1
ERAP1	ENST00000443439	ILMN_2336220	monocyte	3.15×10-23 (p-value)	Zeller et al.	1
ERAP1	ENST00000514604					
ERAP1	ENST00000296754					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001285973 (promoter flanking region)	1	NHLF	DNase1, H3K36me3
		embryonic stem cell (H1ESC)	SP1, DNase1
		blood (K562)	DNase1
		skin (NHDF-AD)	H3K36me3, DNase1
		breast (HMEC)	H3K4me2, H3K27ac, DNase1
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3, H4K20me1
		lung (IMR90)	H3K36me3
		blood (GM12878)	H3K36me3
		nervous (NH-A)	H3K36me3, DNase1
		A549	H3K36me3
		skin (NHEK)	DNase1, H3K36me3, H3K4me2, H3K27ac, H3K4me1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CAST	upstream gene variant	3751	ENST00000348386	?	?	1
CAST	downstream gene variant	2214	ENST00000514845	?	ENSP00000422612	2
CAST	upstream gene variant, downstream gene variant	3360	ENST00000504532	?	?	3
CAST	downstream gene variant	3372	ENST00000506811	?	ENSP00000427092	2
CAST	upstream gene variant	1675	ENST00000515063	?	?	2
CAST	downstream gene variant	3338	ENST00000514055	?	ENSP00000421230	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CAST	ENST00000510156	?	ENSP00000422325	5
CAST	ENST00000395813	?	ENSP00000379158	5
CAST	ENST00000514055	?	ENSP00000421230	3

CAST	ENST00000514055	?	ENSP00000421230	3
CAST	ENST00000511049	?	ENSP00000421130	5
CAST	ENST00000309190	NM_001284212.1, NM_173060.3	ENSP00000312523	5
CAST	ENST00000421689	?	ENSP00000396558	5
CAST	ENST00000511097	?	ENSP00000422951	5
CAST	ENST00000506811	?	ENSP00000427092	3
CAST	ENST00000509903	?	ENSP00000426946	5
CAST	ENST00000512620	?	ENSP00000426422	5
CAST	ENST00000338252	NM_001190442.1	ENSP00000343421	5
CAST	ENST00000508117	?	ENSP00000422466	5
CAST	ENST00000508608	?	ENSP00000422677	5
CAST	ENST00000508830	?	ENSP00000425721	5
CAST	ENST00000511782	?	ENSP00000423638	5
CAST	ENST00000513666	?	?	5
CAST	ENST00000507487	?	?	3
CAST	ENST00000505143	?	ENSP00000422957	5
CAST	ENST00000395812	NM_001042440.3	ENSP00000379157	5
CAST	ENST00000514845	?	ENSP00000422612	3
CAST	ENST00000341926	?	ENSP00000339914	5
CAST	ENST00000508197	?	ENSP00000422831	5
CAST	ENST00000504465	NM_001284213.1	ENSP00000425670	5
CAST	ENST00000510756	?	ENSP00000422176	5
CAST	ENST00000348386	?	?	2