Locus 70 (rs6138458)

SNiPAcard

Block annotations

Block info

genomic range	chr20:24,899,905-24,977,586
block size	77,682 bp
variant count	61 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.131 [-4.848 – 0.469]	gene(s) hit or close-by	APMAP, CST7, RNU6-1257P, RP4-568C11.4
phastCons	μ = 0.048 [0 – 0.647]	eQTL gene(s)	APMAP, CST7, NINL
GERP++	μ = -0.368 [-8.11 – 1.91]	potentially regulated gene(s)	RP5-1025A1.2
CADD score	μ = 2.517 [0.005 – 11.69]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
APMAP	?	ENSG00000101474	pancreas	1.50×10^{-9} (p-value)	GTEx Portal V6	61
APMAP	?	ENSG00000101474	testis	1.36×10^{-7} (p-value)	GTEx Portal V6	59
APMAP	?	ENSG00000101474	transverse colon	1.12×10^{-7} (p-value)	GTEx Portal V6	50
CST7	?	ENSG00000077984	blood	4.25×10^{-19} (p-value)	GTEx Portal V6	61
APMAP	?	ENSG00000101474	blood	4.94×10^{-7} (p-value)	GTEx Portal V6	8
CST7	?	ENSG00000077984	lung	2.84×10^{-7} (p-value)	GTEx Portal V6	17
NINL	?	ENSG00000101004	subcutaneous adipocytes	1.06×10^{-5} (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000291173	1	ENCP00000031115	RP5-1025A1.2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000399319 (CTCF binding site)	2	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	DNase1, H3K27me3, CTCF
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	DNase1, Rad21, CTCF, H3K27me3
		lung (IMR90)	H3K27me3, CTCF
		blood (GM12878)	Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	H3K27me3, CTCF, H3K4me2
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF

		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF, DNase1
		A549	CTCF, H3K27me3
ENSR00001664641 (CTCF binding site)	3	embryonic stem cell (H1ESC)	H3K27me3, DNase1
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		A549	H3K27me3
		blood (DND-41)	H3K36me3
ENSR00001664642 (enhancer)	2	cervix (HeLa-S3)	H3K27me3
		liver (HepG2)	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K4me1, H3K4me2
ENSR00001664643 (promoter flanking region)	2	embryonic stem cell (H1ESC)	CTCF, Rad21, DNase1
		HSMMtube	H3K27me3
		Osteobl	CTCF
		blood (K562)	H3K27me3, CTCF
		muscle (HSMM)	CTCF
		breast (HMEC)	CTCF, H3K4me2, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K27me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	CTCF, H3K27me3
		blood (GM12878)	CTCF
		A549	H3K27me3, CTCF
		skin (NHEK)	CTCF, H3K4me2, H3K4me1, DNase1
ENSR00000399320 (CTCF binding site)	5	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, H3K27me3
		blood (K562)	H3K27me3, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF, H3K27me3
		blood (GM12878)	CTCF, Rad21
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF, H3K4me2, H3K4me1, DNase1
		Osteobl	H3K27me3, CTCF
		blood (DND-41)	H3K4me1, CTCF
		breast (HMEC)	CTCF, H3K4me2, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K27me3
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF, H3K27me3
ENSR00001448170 (enhancer)	2	HSMMtube	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF, H3K4me1, H3K27ac
		blood (K562)	CTCF, H3K27me3
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	DNase1
		breast (HMEC)	H3K4me2
		cervix (HeLa-S3)	H3K27me3
		liver (HepG2)	CTCF
		blood (GM12878)	CTCF
		A549	H3K27me3, CTCF
		skin (NHEK)	CTCF, H3K4me2
ENSR00001448175 (promoter)	1	embryonic stem cell (H1ESC)	H3K27me3, PolII, Rad21, Gabp, TAF7, TAF1, H3K4me2, H3K9ac, H3K4me3, DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	HEY1, H2AZ, ELF1, PolII, H3K4me2, CTCF, DNase1, H3K4me3, Max, H3K9ac, Gabp, Cmyc
		skin (NHDF-AD)	H3K4me2, H3K9ac, DNase1, H3K4me3, CTCF
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ
		liver (HepG2)	DNase1, Gabp, PolII, H3K79me2, Rad21, TAF1, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, CTCF, H3K27ac, Cmyc, H3K4me3, H3K27me3
		lung (IMR90)	DNase1, H4K8ac, H3K27ac, H3K4me2, H4K5ac, H3K4me3, H3K9ac, CTCF

		blood (GM12878)	H2AZ, DNase1, Cmyc, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, PolII, H3K27ac, H3K4me2, CTCF, H3K9ac
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, DNase1, H3K27ac
		skin (NHEK)	CTCF, H3K9ac, H3K4me2, DNase1, H3K4me3
		NHLF	H3K27ac, H3K9ac, CTCF, H3K4me3, DNase1
		Osteobl	CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, CTCF, H3K4me2, H3K4me1
		breast (HMEC)	CTCF, H3K4me2, H3K9ac, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K9ac, DNase1, PolII, Gabp, H3K4me3, TAF1, H3K27ac, CTCF, H3K4me2
		monocytes (Monocytes-CD14+)	H3K27ac, H3K9ac, H3K4me3, CTCF, DNase1
		endothelium (HUVEC)	Max, H3K4me3, H3K4me2, PolII, CTCF, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac, CTCF, DNase1, H3K27ac
ENSR00000399325 e! (CTCF binding site)	3	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, H3K4me2, H3K27me3, DNase1
		blood (K562)	H3K27me3, Rad21, CTCF
		skin (NHDF-AD)	H3K4me2, DNase1, H3K4me3, CTCF
		muscle (HSMM)	H3K4me2, CTCF
		liver (HepG2)	Rad21, H3K4me1, H2AZ, H3K4me2, H3K9ac, CTCF, H3K4me3, DNase1
		blood (GM12878)	H3K9ac, DNase1, Pbx3, H2AZ, Yy1, Rad21, H3K4me2, CTCF
		lung (IMR90)	H4K8ac, H3K4me2, H4K5ac, H3K9ac, CTCF
		nervous (NH-A)	H3K4me2, CTCF, H3K9ac
		skin (NHEK)	CTCF, H3K4me3, H3K4me2, DNase1
		NHLF	CTCF
		Osteobl	CTCF, H3K4me2, H3K27ac
		blood (DND-41)	H3K9ac, CTCF, H3K4me2, H3K4me1, H3K4me3
		breast (HMEC)	H3K4me3, H3K27ac, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, CTCF, H3K4me2
		monocytes (Monocytes-CD14+)	H3K9ac, DNase1, CTCF, H3K27ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, CTCF, H3K4me2, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac, CTCF, DNase1, H3K27ac
ENSR00001664649 e! (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, CTCF, Rad21
		HSMMtube	CTCF, H3K27me3, DNase1
		blood (K562)	Rad21, CTCF, H3K27me3
		skin (NHDF-AD)	H3K4me2, DNase1, H3K4me3, CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, H3K4me1, H3K4me2, CTCF, DNase1
		lung (IMR90)	H3K4me2, CTCF
		blood (GM12878)	CTCF, Rad21, Yy1, Pbx3, DNase1
		nervous (NH-A)	H3K4me2, CTCF
		skin (NHEK)	CTCF, DNase1
		NHLF	CTCF
		Osteobl	CTCF, H3K4me2
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	CTCF, DNase1
		A549	H3K4me3, CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
APMAP e!	downstream gene variant, upstream gene variant	154	ENST00000217456 e!	NM_020531.2	ENSP00000217456 e!	10
APMAP e!	downstream gene variant, upstream gene variant	491	ENST00000451442 e!	?	ENSP00000395874 e!	10
CST7 e!	upstream gene variant, downstream gene variant	32	ENST00000480798 e!	NM_003650.3	ENSP00000420384 e!	2
RNU6-1257P e!	downstream gene variant	4470	ENST00000384625 e!	?	?	1
RP4-568C11.4 e!	upstream gene variant, downstream gene variant	34	ENST00000620459 e!	?	?	14

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
APMAP	ENST00000217456	NM_020531.2	ENSP00000217456	2
APMAP	ENST00000451442	?	ENSP00000395874	2
CST7	ENST00000480798	NM_003650.3	ENSP00000420384	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
APMAP	ENST00000217456	NM_020531.2	ENSP00000217456	1
APMAP	ENST00000451442	?	ENSP00000395874	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP4-568C11.4	ENST00000620459	?	3