Locus 218 (rs10823500)

SNiPAcard

Block annotations

Block info

genomic range	chr10:71,949,471-72,006,891
block size	57,421 bp
variant count	15 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.160 [-2.43 – 1.545]	**gene(s) hit or close-by**	NPFFR1, PPA1, RP11-367H5.8
phastCons	μ = 0.015 [0 – 0.148]	**eQTL gene(s)**	AIFM2, EIF4EBP2, PPA1, SAR1A, TYSND1
GERP++	μ = -0.574 [-4.12 – 1.88]	**potentially regulated gene(s)**	CEP57L1P1
CADD score	μ = 3.477 [0.062 – 10.56]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PPA1	?	ENSG00000180817	uterus	9.44×10^{-8} (p-value)	GTEx Portal V6	13
PPA1	?	ENSG00000180817	pancreas	6.48×10^{-9} (p-value)	GTEx Portal V6	14
SAR1A	?	ENSG00000079332	muscularis mucosae	2.47×10^{-10} (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	muscularis mucosae	3.13×10^{-19} (p-value)	GTEx Portal V6	14
SAR1A	?	ENSG00000079332	lung	8.34×10^{-11} (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	lung	9.85×10^{-7} (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	atrial appendage	5.31×10^{-16} (p-value)	GTEx Portal V6	14
SAR1A	?	ENSG00000079332	transformed fibroblasts	1.12×10^{-9} (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	transformed fibroblasts	4.94×10^{-7} (p-value)	GTEx Portal V6	13
SAR1A	?	ENSG00000079332	tibial artery	2.18×10^{-18} (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	tibial artery	1.53×10^{-48} (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	blood	2.42×10^{-14} (p-value)	GTEx Portal V6	14
SAR1A	?	ENSG00000079332	thyroid	2.38×10^{-11} (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	thyroid	8.16×10^{-11} (p-value)	GTEx Portal V6	14
SAR1A	?	ENSG00000079332	skeletal muscle	2.68×10^{-6} (p-value)	GTEx Portal V6	11
PPA1	?	ENSG00000180817	skeletal muscle	1.70×10^{-63} (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	unexposed skin	1.39×10^{-11} (p-value)	GTEx Portal V6	14
SAR1A	?	ENSG00000079332	sun exposed skin	1.29×10^{-6} (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	sun exposed skin	2.38×10^{-9} (p-value)	GTEx Portal V6	14
SAR1A	?	ENSG00000079332	aorta	1.20×10^{-7} (p-value)	GTEx Portal V6	13
PPA1	?	ENSG00000180817	aorta	1.49×10^{-25} (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	left ventricle	8.39×10^{-15} (p-value)	GTEx Portal V6	14

SAR1A	?	ENSG00000079332	subcutaneous adipocytes	1.78×10-13 (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	caudate basal ganglia	1.64×10-7 (p-value)	GTEx Portal V6	13
SAR1A	?	ENSG00000079332	visceral adipocytes	1.76×10-7 (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	stomach	4.51×10-12 (p-value)	GTEx Portal V6	14
AIFM2	?	ENSG00000042286	cortex	1.05×10-12 (p-value)	GTEx Portal V6	13
PPA1	?	ENSG00000180817	cortex	4.92×10-7 (p-value)	GTEx Portal V6	12
SAR1A	?	ENSG00000079332	tibial nerve	2.12×10-13 (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	tibial nerve	3.18×10-7 (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	sigmoid colon	2.95×10-9 (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	pituitary	3.66×10-12 (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	gastroesophageal junction	2.42×10-11 (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	testis	1.35×10-8 (p-value)	GTEx Portal V6	14
TYSND1	ENST00000287078	ILMN_2325185	blood	5.01×10-5 (p-value)	Westra et al.	6
TYSND1	ENST00000335494					
TYSND1	ENST00000494143					
TYSND1	ENST00000479086					
EIF4EBP2	ENST00000373218	ILMN_1728083	blood	1.92×10-6 (p-value)	Westra et al.	6
AIFM2	ENST00000307864	ILMN_1712639	blood	9.47×10-7 (p-value)	Westra et al.	1
AIFM2	ENST00000613322					
PPA1	ENST00000610026	ILMN_1805827	monocyte	8.11×10-58 (p-value)	Zeller et al.	1
PPA1	ENST00000373232					
SAR1A	?	ENSG00000079332	esophagus mucosa	3.61×10-6 (p-value)	GTEx Portal V6	2
AIFM2	?	ENSG00000042286	cerebellum	1.25×10-6 (p-value)	GTEx Portal V6	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000077597	1	ENCP00000008751	CEP57L1P1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001424487 (promoter flanking region)	1	NHLF	H3K27ac, H3K4me1, DNase1
		HSMMtube	DNase1
		Osteobl	H3K4me1, H3K4me2, H3K27ac
		blood (K562)	Max, H3K27me3
		skin (NHDF-AD)	H3K4me1, H3K4me3, DNase1
		muscle (HSMM)	H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, H3K4me1, Max
		monocytes (Monocytes-CD14+)	H3K4me1
		lung (IMR90)	DNase1, H3K27ac
		A549	H3K36me3
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1
ENSR00001424491	1	embryonic stem cell (H1ESC)	H3K36me3

(promoter flanking region)		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K79me2, H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	DNase1
ENSR00000356568	2	embryonic stem cell (H1ESC)	DNase1, USF1, H3K4me3, H3K4me2, TAF1, H3K27me3, PolII, Sin3Ak20, H3K9ac, Yy1
(promoter)		HSMMtube	DNase1, H2AZ, H3K4me3, H3K4me2
		blood (K562)	Egr1, H3K27ac, Max, CTCF, H3K9ac, H3K79me2, HEY1, H2AZ, TAF1, Cmyc, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	DNase1, H3K9ac, H3K4me2, H3K4me3
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2, H3K9ac, DNase1
		liver (HepG2)	PolII, H3K79me2, TAF1, USF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, CTCF, Cmyc, H3K4me3, H3K27me3, H3K36me3, DNase1
		blood (GM12878)	PolII, H3K9ac, H3K36me3, H3K4me2, H3K27ac, H2AZ, DNase1, Cmyc, CTCF, Yy1, H3K79me2, H3K4me3, USF1
		lung (IMR90)	DNase1, H3K4me2, H3K36me3, H3K4me3
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K4me2, H3K4me3, DNase1, H3K9ac, H3K27ac
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		breast (HMEC)	H3K4me2, DNase1, H3K27ac, H3K4me3, H3K9ac
		cervix (HeLa-S3)	DNase1, H3K79me2, PolII, Cmyc, CTCF, H3K36me3, H3K4me3, Max, TAF1, Nrf1, H3K9ac, H3K4me2, H3K27ac
		monocytes (Monocytes-CD14+)	H3K4me3, H3K36me3, H3K9ac, H3K27ac, H3K4me2, DNase1
		endothelium (HUVEC)	H3K36me3, Cjun, Max, H3K4me3, H3K9ac, PolII, DNase1, Cmyc
		A549	H3K27ac, H3K36me3, DNase1, H3K9ac, H3K4me3, H3K4me2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
NPFFR1	downstream gene variant	194	ENST00000277942	NM_022146.4	ENSP00000277942	3
PPA1	downstream gene variant, upstream gene variant	57	ENST00000460755	?	?	3
PPA1	downstream gene variant	3635	ENST00000625364	?	ENSP00000486162	1
PPA1	downstream gene variant, upstream gene variant	1	ENST00000373230	?	ENSP00000362327	2
PPA1	downstream gene variant	391	ENST00000495346	?	?	1
RP11-367H5.8	upstream gene variant	4613	ENST00000435591	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PPA1	ENST00000610026	?	?	2
PPA1	ENST00000460755	?	?	4
PPA1	ENST00000373232	NM_021129.3	ENSP00000362329	9
PPA1	ENST00000373230	?	ENSP00000362327	6
PPA1	ENST00000625364	?	ENSP00000486162	7
PPA1	ENST00000495346	?	?	6

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
PPA1	ENST00000373232	NM_021129.3	ENSP00000362329	1

PPA1	ENST00000625364	?	ENSP00000486162	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PPA1	ENST00000610026	?	1