

SNiPacard

Block annotations

Block info

genomic range	chr10:71,949,471-72,006,891 <i>e!</i>
block size	57,421 bp
variant count	15 variants

Basic features

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

Direct effect on regulation

cis-eQTL

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

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

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000077597 <i>e!</i>	1	ENCP00000008751	CEP57L1P1 <i>e!</i>

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors		
ENSR00001424487 <i>e!</i> (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 <i>e!</i>	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 <i>e!</i> 2 (promoter)	embryonic stem cell (H1ESC)	DNase1, USF1, H3K4me3, H3K4me2, TAF1, H3K27me3, PolII, Sin3Ak20, H3K9ac, Yy1
	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 <i>e!</i>	downstream gene variant	194	ENST00000277942 <i>e!</i>	NM_022146.4	ENSP00000277942 <i>e!</i>	3
PPA1 <i>e!</i>	downstream gene variant, upstream gene variant	57	ENST00000460755 <i>e!</i>	?	?	3
PPA1 <i>e!</i>	downstream gene variant	3635	ENST00000625364 <i>e!</i>	?	ENSP00000486162 <i>e!</i>	1
PPA1 <i>e!</i>	downstream gene variant, upstream gene variant	1	ENST00000373230 <i>e!</i>	?	ENSP00000362327 <i>e!</i>	2
PPA1 <i>e!</i>	downstream gene variant	391	ENST00000495346 <i>e!</i>	?	?	1
RP11-367H5.8 <i>e!</i>	upstream gene variant	4613	ENST00000435591 <i>e!</i>	?	?	1

Putative effect on transcript

Intron variant

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

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
PPA1 <i>e!</i>	ENST00000373232 <i>e!</i>	NM_021129.3	ENSP00000362329 <i>e!</i>	1

PPA1 *e!* ENST00000625364 *e!* ? ENSP00000486162 *e!* 1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PPA1 <i>e!</i>	ENST00000610026 <i>e!</i>	?	1
