Locus 171 (rs9787249)

SNiPAcard

Block annotations

Block info

genomic range	chr1:40,203,722-40,226,046
block size	22,325 bp
variant count	20 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.180 [-1.636 – 0.37]	gene(s) hit or close-by	BMP8B, PPIE
phastCons	μ = 0.043 [0 – 0.341]	eQTL gene(s)	BMP8B, OXCT2, PPIE
GERP++	μ = -0.163 [-5.61 – 2.6]	potentially regulated gene(s)	MACF1, PPIE
CADD score	μ = 4.840 [0.066 – 12.78]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PPIE	?	ENSG00000084072	atrial appendage	8.35×10^{-30} (p-value)	GTEx Portal V6	20
BMP8B	?	ENSG00000116985	transformed fibroblasts	6.30×10^{-11} (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	transformed fibroblasts	2.17×10^{-32} (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	breast	1.32×10^{-44} (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	blood	8.23×10^{-51} (p-value)	GTEx Portal V6	20
PPIE	ENST00000497370	ILMN_2326737	monocyte	3.23×10^{-18} (p-value)	Fairfax et al.	1
PPIE	ENST00000372835		b-cell	4.24×10^{-24} (p-value)	Fairfax et al.	1
PPIE	ENST00000495526					
PPIE	ENST00000470018					
PPIE	ENST00000480169					
PPIE	ENST00000324379					
PPIE	ENST00000475350					
PPIE	ENST00000474804					
PPIE	ENST00000372830					
PPIE	ENST00000482751					
PPIE	ENST00000356511					
PPIE	?	ENSG00000084072	skeletal muscle	2.42×10^{-19} (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	putamen	1.32×10^{-11} (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	blood	2.01×10^{-4} (q-value)	SeeQTL DB (HapMap)	6
PPIE	?	ENSG00000084072	EBV lymphocytes	1.64×10^{-13} (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	unexposed skin	6.98×10^{-51} (p-value)	GTEx Portal V6	20
BMP8B	?	ENSG00000116985	sun exposed skin	1.15×10^{-7} (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	sun exposed skin	8.53×10^{-86} (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	ovary	9.46×10^{-22} (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	left ventricle	9.03×10^{-11} (p-value)	GTEx Portal V6	20

PPIE	?	ENSG00000084072	aorta	4.36×10-42 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	subcutaneous adipocytes	6.11×10-82 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	caudate basal ganglia	2.43×10-14 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	visceral adipocytes	4.07×10-50 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	stomach	6.63×10-39 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	cortex	1.80×10-11 (p-value)	GTEx Portal V6	20
OXCT2	?	ENSG00000198754	tibial nerve	2.10×10-10 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	tibial nerve	1.91×10-69 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	pituitary	4.54×10-19 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	esophagus mucosa	8.49×10-63 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	testis	4.29×10-43 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	hypothalamus	9.76×10-11 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	frontal cortex	4.38×10-7 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	uterus	4.53×10-13 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	pancreas	3.18×10-34 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	muscularis mucosae	1.08×10-36 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	lung	4.70×10-76 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	tibial artery	5.45×10-50 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	thyroid	4.69×10-73 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	prostate	3.73×10-21 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	transverse colon	1.09×10-31 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	liver	1.18×10-18 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	terminal ileum	3.83×10-23 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	nucleus accumbens	1.11×10-11 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	spleen	6.43×10-24 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	coronary artery	2.58×10-22 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	sigmoid colon	3.11×10-19 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	vagina	2.89×10-17 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	gastroesophageal junction	6.49×10-21 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	cerebellar hemisphere	4.63×10-10 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	adrenal gland	1.45×10-18 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	cerebellum	1.45×10-10 (p-value)	GTEx Portal V6	20
BMP8B	?	ENSG00000116985	cerebellum	1.45×10-7 (p-value)	GTEx Portal V6	20

PPIE	?	ENSG00000084072	hippocampus	1.14×10^{-10} (p-value)	GTEx Portal V6	20
PPIE	ENST00000497370	ILMN_2326737	skin	7.60×10^{-18} (p-value)	MuTHER consortium	4
PPIE	ENST00000495526		blood	6.33×10^{-35} (p-value)	MuTHER consortium	4
PPIE	ENST00000372835		adipocyte	5.02×10^{-10} (p-value)	MuTHER consortium	4
PPIE	ENST00000470018					
PPIE	ENST00000480169					
PPIE	ENST00000324379					
PPIE	ENST00000475350					
PPIE	ENST00000474804					
PPIE	ENST00000372830					
PPIE	ENST00000482751					
PPIE	ENST00000356511					
BMP8B	ENST00000372827	ILMN_2123415	blood	7.55×10^{-13} (p-value)	MuTHER consortium	4
PPIE	ENST00000497370	ILMN_1680341	skin	9.11×10^{-75} (p-value)	MuTHER consortium	4
PPIE	ENST00000495526		blood	2.84×10^{-35} (p-value)	MuTHER consortium	4
PPIE	ENST00000372835		adipocyte	3.73×10^{-38} (p-value)	MuTHER consortium	4
PPIE	ENST00000470213					
PPIE	ENST00000324379					
PPIE	ENST00000475350					
PPIE	ENST00000474804					
PPIE	ENST00000482751					
PPIE	ENST00000372830					
PPIE	ENST00000356511					
PPIE	ENST00000497370	ILMN_2326737	monocyte	3.68×10^{-64} (p-value)	Zeller et al.	2
PPIE	ENST00000495526					
PPIE	ENST00000372835					
PPIE	ENST00000470018					
PPIE	ENST00000480169					
PPIE	ENST00000324379					
PPIE	ENST00000475350					
PPIE	ENST00000474804					
PPIE	ENST00000372830					
PPIE	ENST00000482751					
PPIE	ENST00000356511					
PPIE	ENST00000497370	ILMN_1680341	monocyte	5.74×10^{-52} (p-value)	Zeller et al.	2
PPIE	ENST00000495526					
PPIE	ENST00000372835					
PPIE	ENST00000470213					
PPIE	ENST00000324379					
PPIE	ENST00000475350					
PPIE	ENST00000474804					
PPIE	ENST00000482751					
PPIE	ENST00000372830					
PPIE	ENST00000356511					
PPIE	ENST00000475350	202494_at	blood	4.50×10^{-16} (p-value)	Dixon et al.	1
PPIE	ENST00000497370					
PPIE	ENST00000372835					
PPIE	ENST00000495526					
PPIE	ENST00000482751					
PPIE	ENST00000470213					
PPIE	ENST00000324379					

BMP8B	ENST00000372827	235275_at	blood	1.60×10-17 (p-value)	Dixon et al.	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000418809	1	ENST00000497370, ENST00000495526, ENST00000372835, ENST00000456578, ENST00000480169, ENST00000470213, ENST00000324379, ENST00000475350, ENST00000474804, ENST00000482751, ENST00000372830, ENST00000485507, ENST00000356511	PPIE

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000001965		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000015243	1	ENCP00000001935	MACF1
		ENCP00000001943	MACF1
		ENCP00000001939	MACF1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001579074 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	DNase1, H4K8ac
		blood (K562)	H2AZ
		muscle (HSMM)	DNase1
ENSR00000534967 (promoter)	3	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, PolII, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3, DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	Yy1, H3K27ac, Max, H3K9ac, Gabp, H3K79me2, HEY1, H2AZ, TAF1, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me2, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	H3K27me3, DNase1, H3K4me3, H3K9ac, Gabp, PolII, TAF1
		blood (GM12878)	PolII, H2AZ, DNase1, Yy1, Gabp, H3K79me2, H3K4me3, H3K27ac, H3K9ac
		lung (IMR90)	DNase1, H4K8ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K9ac
		nervous (NH-A)	H3K36me3, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2, H3K36me3
		blood (DND-41)	H3K4me3, H3K9ac
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, Gabp, PolII, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K36me3, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cmyc, H3K4me3, PolII
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K36me3

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
PPIE	3	ENST00000324379 ENST00000356511 ENST00000372830 ENST00000372835 ENST00000470213 ENST00000475350 ENST00000482751 ENST00000495526 ENST00000497370	hsa-miR-411-5p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
BMP8B	downstream gene variant	1787	ENST00000372827	NM_001720.3	ENSP00000361915	3
BMP8B	downstream gene variant	3177	ENST00000397360	?	ENSP00000380518	1
PPIE	upstream gene variant	149	ENST00000372830	NM_001195007.1	ENSP00000361918	3
PPIE	upstream gene variant, downstream gene variant	134	ENST00000456578	?	?	5
PPIE	upstream gene variant, downstream gene variant	136	ENST00000495526	?	ENSP00000434989	6
PPIE	upstream gene variant	133	ENST00000356511	NM_203456.2	ENSP00000348904	3
PPIE	upstream gene variant, downstream gene variant	149	ENST00000497370	?	ENSP00000433475	6
PPIE	upstream gene variant, downstream gene variant	149	ENST00000372835	?	ENSP00000361925	6
PPIE	upstream gene variant, downstream gene variant	238	ENST00000480169	?	?	10
PPIE	upstream gene variant, downstream gene variant	127	ENST00000485507	?	?	6
PPIE	upstream gene variant, downstream gene variant	149	ENST00000470213	?	ENSP00000431714	6
PPIE	upstream gene variant, downstream gene variant	138	ENST00000482751	?	?	6
PPIE	upstream gene variant, downstream gene variant	130	ENST00000324379	NM_006112.3	ENSP00000312769	6
PPIE	downstream gene variant	662	ENST00000470018	?	ENSP00000436689	7
PPIE	upstream gene variant, downstream gene variant	156	ENST00000475350	?	ENSP00000434888	6
PPIE	upstream gene variant	825	ENST00000467741	?	?	1
PPIE	upstream gene variant, downstream gene variant	152	ENST00000474804	?	ENSP00000432396	6

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PPIE	ENST00000482751	?	?	13
PPIE	ENST00000497370	?	ENSP00000433475	13
PPIE	ENST00000475350	?	ENSP00000434888	13
PPIE	ENST00000372835	?	ENSP00000361925	13
PPIE	ENST00000485507	?	?	3
PPIE	ENST00000456578	?	?	3
PPIE	ENST00000470018	?	ENSP00000436689	7
PPIE	ENST00000495526	?	ENSP00000434989	13
PPIE	ENST00000474804	?	ENSP00000432396	10
PPIE	ENST00000480169	?	?	4
PPIE	ENST00000372830	NM_001195007.1	ENSP00000361918	17
PPIE	ENST00000470213	?	ENSP00000431714	13
PPIE	ENST00000356511	NM_203456.2	ENSP00000348904	17
PPIE	ENST00000324379	NM_006112.3	ENSP00000312769	13

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
BMP8B	ENST00000372827	NM_001720.3	ENSP00000361915	1

PPIE	ENST00000495526	?	ENSP00000434989	5

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PPIE	ENST00000480169	?	1