Locus 3 (rs11553746)

SNiPAcard

Block annotations

Block info

genomic range	chr2:211,317-296,410
block size	85,094 bp
variant count	48 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.104 [-2.209 – 5.667]	gene(s) hit or close-by	AC079779.4, ACP1, FAM150B, SH3YL1
phastCons	μ = 0.120 [0 – 1]	eQTL gene(s)	ACP1, SH3YL1
GERP++	μ = -0.328 [-5.58 – 5.87]	potentially regulated gene(s)	ACP1, SH3YL1
CADD score	μ = 3.895 [0.08 – 19.46]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
3-(cystein-S-yl)acetaminophen*	4.62×10^{-5}	Metabolomics GWAS Server	24816252	14

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Acid phosphatase 1, soluble, a/b polymorphism of	benign	ClinVar	RCV000014681.1	1
ACID PHOSPHATASE 1, SOLUBLE, A/B POLYMORPHISM OF	OMIM curated	OMIM	MIM:171500	1

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
ACP1	missense variant	ENST00000272065	NM_004300.3	ENSP00000272065	R/Q	cGa/cAa	?	?	1
ACP1	missense variant	ENST00000453390	?	ENSP00000411121	I/T	aTa/aCa	?	?	1
ACP1	missense variant	ENST00000405233	?	ENSP00000384307	I/T	aTa/aCa	?	?	1
ACP1	missense variant	ENST00000407983	NM_001040649.2	ENSP00000385404	I/T	aTa/aCa	?	?	1
ACP1	missense variant	ENST00000272067	NM_007099.3	ENSP00000272067	R/Q	cGa/cAa	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SH3YL1	?	ENSG00000035115	pancreas	4.33×10^{-30} (p-value)	GTEx Portal V6	46
SH3YL1	?	ENSG00000035115	muscularis mucosae	2.30×10^{-11} (p-value)	GTEx Portal V6	46
ACP1	?	ENSG00000143727	muscularis mucosae	1.02×10^{-6} (p-value)	GTEx Portal V6	41
SH3YL1	?	ENSG00000035115	lung	2.20×10^{-6} (p-value)	GTEx Portal V6	41

SH3YL1	?	ENSG00000035115	atrial appendage	5.26×10-12 (p-value)	GTEx Portal V6	46
SH3YL1	?	ENSG00000035115	tibial artery	3.69×10-6 (p-value)	GTEx Portal V6	31
ACP1	?	ENSG00000143727	tibial artery	1.67×10-6 (p-value)	GTEx Portal V6	19
SH3YL1	?	ENSG00000035115	blood	3.57×10-30 (p-value)	GTEx Portal V6	46
SH3YL1	?	ENSG00000035115	thyroid	2.73×10-37 (p-value)	GTEx Portal V6	46
SH3YL1	?	ENSG00000035115	skeletal muscle	2.13×10-13 (p-value)	GTEx Portal V6	46
ACP1	?	ENSG00000143727	skeletal muscle	5.67×10-6 (p-value)	GTEx Portal V6	2
SH3YL1	?	ENSG00000035115	transverse colon	3.79×10-9 (p-value)	GTEx Portal V6	46
SH3YL1	?	ENSG00000035115	unexposed skin	3.10×10-36 (p-value)	GTEx Portal V6	46
SH3YL1	?	ENSG00000035115	sun exposed skin	3.04×10-54 (p-value)	GTEx Portal V6	46
SH3YL1	?	ENSG00000035115	left ventricle	1.07×10-12 (p-value)	GTEx Portal V6	46
SH3YL1	?	ENSG00000035115	subcutaneous adipocytes	4.80×10-8 (p-value)	GTEx Portal V6	44
SH3YL1	?	ENSG00000035115	stomach	5.68×10-7 (p-value)	GTEx Portal V6	40
SH3YL1	?	ENSG00000035115	sigmoid colon	5.87×10-8 (p-value)	GTEx Portal V6	44
SH3YL1	?	ENSG00000035115	tibial nerve	8.72×10-17 (p-value)	GTEx Portal V6	46
SH3YL1	?	ENSG00000035115	esophagus mucosa	3.58×10-75 (p-value)	GTEx Portal V6	46
SH3YL1	?	ENSG00000035115	ovary	7.94×10-7 (p-value)	GTEx Portal V6	37
SH3YL1	ENST00000479739	ILMN_1712231	b-cell	5.27×10-11 (p-value)	Fairfax et al.	5
SH3YL1	ENST00000356150					
SH3YL1	ENST00000463865					
SH3YL1	ENST00000403712					
SH3YL1	ENST00000403657					
SH3YL1	ENST00000473104					
SH3YL1	ENST00000405430					
SH3YL1	ENST00000468321					
SH3YL1	ENST00000626873					
SH3YL1	?	ENSG00000035115	blood	8.90×10-3 (q-value)	SeeQTL DB (HapMap)	8
SH3YL1	?	ENSG00000035115	adrenal gland	1.45×10-6 (p-value)	GTEx Portal V6	27
SH3YL1	?	ENSG00000035115	terminal ileum	1.86×10-6 (p-value)	GTEx Portal V6	1
SH3YL1	?	ENSG00000035115	transformed fibroblasts	1.11×10-5 (p-value)	GTEx Portal V6	1
SH3YL1	?	ENSG00000035115	aorta	8.96×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000024883		
ENCP00000024888		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000234638	1	ENCP00000024888	SH3YL1
		ENCP00000024889	ACP1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001538827 (promoter)	3	embryonic stem cell (H1ESC)	DNase1, H3K4me3, H3K9ac, USF1, H3K4me2, TAF1, Yy1, H3K27ac, TAF7, Egr1, PolII, H3K27me3
		HSMMtube	H3K79me2, H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	Egr1, H3K27ac, Max, H3K9ac, Cmyc, H3K79me2, HEY1, H2AZ, PolII, H3K4me2, H3K36me3, H3K27me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2
		liver (HepG2)	FOXA1, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3, H3K4me2, Yy1, H4K20me1, PolII, H3K79me2, TAF1, H3K4me1, DNase1
		blood (GM12878)	H3K79me2, Yy1, DNase1, H2AZ, PolII, Cmyc, H3K4me3, H3K27ac, H3K36me3, H3K9ac, H3K4me2, ZEB1
		lung (IMR90)	H3K79me2, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K9ac, DNase1
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac
		skin (NHEK)	CTCF, H3K27ac, H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K9ac, H3K4me2, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, Nrf1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, PolII, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cmyc, DNase1, PolII, H3K4me2, H3K4me3, Max
		A549	H3K4me3, H3K4me2, H3K36me3, H3K27ac, DNase1, H3K9ac
ENSR00000675428 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, H3K36me3, CTCF
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K79me2, Rad21, CTCF, H3K36me3
		lung (IMR90)	H3K36me3
		blood (GM12878)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	CTCF
ENSR00001649416 (CTCF binding site)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1, H3K27me3
		skin (NHDF-AD)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
ENSR00000584027 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1, H3K27me3
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
	1		

ACP1	ENST00000272065 ENST00000272067 ENST00000405364 ENST00000413140 ENST00000442386 ENST00000453390 ENST00000463831 ENST00000480874 ENST00000484464	hsa-miR-9-5p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC079779.4	upstream gene variant	1096	ENST00000427831	?	?	3
AC079779.4	upstream gene variant	948	ENST00000450709	?	?	3
ACP1	upstream gene variant, downstream gene variant	251	ENST00000272065	NM_004300.3	ENSP00000272065	7
ACP1	upstream gene variant, downstream gene variant	2370	ENST00000463831	?	?	6
ACP1	upstream gene variant, downstream gene variant	272	ENST00000413140	?	ENSP00000410331	7
ACP1	upstream gene variant, downstream gene variant	248	ENST00000272067	NM_007099.3	ENSP00000272067	7
ACP1	upstream gene variant, downstream gene variant	295	ENST00000439645	?	ENSP00000408596	6
ACP1	upstream gene variant, downstream gene variant	279	ENST00000442386	?	ENSP00000389681	7
ACP1	upstream gene variant, downstream gene variant	584	ENST00000484464	?	?	7
ACP1	upstream gene variant, downstream gene variant	344	ENST00000453390	?	ENSP00000411121	7
ACP1	upstream gene variant, downstream gene variant	279	ENST00000407983	NM_001040649.2	ENSP00000385404	6
ACP1	upstream gene variant, downstream gene variant	298	ENST00000405364	?	ENSP00000384184	5
ACP1	upstream gene variant, downstream gene variant	121	ENST00000484125	?	?	6
ACP1	upstream gene variant, downstream gene variant	315	ENST00000480874	?	?	5
ACP1	upstream gene variant, downstream gene variant	305	ENST00000405233	?	ENSP00000384307	6
FAM150B	downstream gene variant	468	ENST00000463919	?	?	4
FAM150B	downstream gene variant, upstream gene variant	974	ENST00000452023	?	ENSP00000389939	4
FAM150B	downstream gene variant	2926	ENST00000405290	?	ENSP00000385672	1
FAM150B	downstream gene variant	2926	ENST00000619265	?	ENSP00000478180	1
FAM150B	downstream gene variant	2926	ENST00000401489	?	ENSP00000385214	1
FAM150B	downstream gene variant	2948	ENST00000344414	?	ENSP00000339565	1
FAM150B	downstream gene variant	2763	ENST00000401503	?	ENSP00000385537	1
FAM150B	downstream gene variant	2555	ENST00000403610	NM_001002919.2	ENSP00000384604	2
SH3YL1	downstream gene variant, upstream gene variant	387	ENST00000468321	?	?	7
SH3YL1	downstream gene variant, upstream gene variant	2557	ENST00000472861	?	?	6
SH3YL1	downstream gene variant, upstream gene variant	195	ENST00000403712	NM_001159597.2	ENSP00000384276	7
SH3YL1	downstream gene variant, upstream gene variant	162	ENST00000356150	NM_015677.3	ENSP00000348471	7
SH3YL1	downstream gene variant, upstream gene variant	36	ENST00000471948	?	?	11
SH3YL1	downstream gene variant, upstream gene variant	588	ENST00000403657	?	ENSP00000385668	6
SH3YL1	downstream gene variant, upstream gene variant	1661	ENST00000602998	?	?	2
SH3YL1	upstream gene variant	2557	ENST00000402632	?	ENSP00000384910	4
SH3YL1	downstream gene variant, upstream gene variant	2302	ENST00000477707	?	?	6
SH3YL1	downstream gene variant, upstream gene variant	969	ENST00000462719	?	?	6
SH3YL1	downstream gene variant, upstream gene variant	280	ENST00000403658	NM_001282682.1	ENSP00000383928	7
SH3YL1	downstream gene variant	923	ENST00000481932	?	ENSP00000474411	5

SH3YL1	downstream gene variant, upstream gene variant	48	ENST00000405430	?	ENSP00000384269	7
SH3YL1	downstream gene variant, upstream gene variant	438	ENST00000431160	?	ENSP00000399519	8
SH3YL1	downstream gene variant, upstream gene variant	590	ENST00000473104	?	?	6
SH3YL1	downstream gene variant, upstream gene variant	25	ENST00000472012	?	?	7
SH3YL1	downstream gene variant, upstream gene variant	229	ENST00000463865	?	?	6
SH3YL1	downstream gene variant, upstream gene variant	713	ENST00000415368	?	ENSP00000410235	5
SH3YL1	downstream gene variant, upstream gene variant	411	ENST00000451005	?	ENSP00000416312	5
SH3YL1	downstream gene variant, upstream gene variant	53	ENST00000488979	?	?	7
SH3YL1	downstream gene variant, upstream gene variant	1428	ENST00000454318	?	ENSP00000415723	7
SH3YL1	downstream gene variant, upstream gene variant	1382	ENST00000475027	?	?	7
SH3YL1	downstream gene variant, upstream gene variant	1174	ENST00000497051	?	?	5
SH3YL1	downstream gene variant, upstream gene variant	225	ENST00000488044	?	?	10
SH3YL1	downstream gene variant, upstream gene variant	167	ENST00000460974	?	?	6
SH3YL1	downstream gene variant, upstream gene variant	69	ENST00000626873	NM_001282687.1	ENSP00000485824	7
SH3YL1	downstream gene variant, upstream gene variant	1213	ENST00000605370	?	?	6
SH3YL1	downstream gene variant, upstream gene variant	394	ENST00000479739	?	ENSP00000441266	7
SH3YL1	downstream gene variant, upstream gene variant	2133	ENST00000465733	?	?	6

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
ACP1	ENST00000439645	?	ENSP00000408596	D	gaT/gaC	1
ACP1	ENST00000272067	NM_007099.3	ENSP00000272067	D	gaT/gaC	1
SH3YL1	ENST00000626873	NM_001282687.1	ENSP00000485824	S	agC/agT	1
SH3YL1	ENST00000356150	NM_015677.3	ENSP00000348471	S	agC/agT	1
SH3YL1	ENST00000405430	?	ENSP00000384269	S	agC/agT	1

Non-coding exon variant (splice region)

gene	affected transcript	RefSeq id	variant(s)
SH3YL1	ENST00000468321	?	1
SH3YL1	ENST00000463865	?	1
SH3YL1	ENST00000472012	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC079779.4	ENST00000450709	?	?	1
AC079779.4	ENST00000427831	?	?	5
ACP1	ENST00000407983	NM_001040649.2	ENSP00000385404	5
ACP1	ENST00000484125	?	?	2
ACP1	ENST00000484464	?	?	7
ACP1	ENST00000439645	?	ENSP00000408596	6
ACP1	ENST00000272067	NM_007099.3	ENSP00000272067	7

ACP1	ENST00000480874	?	?	7
ACP1	ENST00000442386	?	ENSP00000389681	7
ACP1	ENST00000413140	?	ENSP00000410331	7
ACP1	ENST00000453390	?	ENSP00000411121	6
ACP1	ENST00000272065	NM_004300.3	ENSP00000272065	7
ACP1	ENST00000405364	?	ENSP00000384184	7
ACP1	ENST00000405233	?	ENSP00000384307	5
FAM150B	ENST00000463919	?	?	1
FAM150B	ENST00000405290	?	ENSP00000385672	6
FAM150B	ENST00000619265	?	ENSP00000478180	6
FAM150B	ENST00000452023	?	ENSP00000389939	3
FAM150B	ENST00000344414	?	ENSP00000339565	6
FAM150B	ENST00000401503	?	ENSP00000385537	6
FAM150B	ENST00000401489	?	ENSP00000385214	6
FAM150B	ENST00000403610	NM_001002919.2	ENSP00000384604	6
SH3YL1	ENST00000468321	?	?	25
SH3YL1	ENST00000626873	NM_001282687.1	ENSP00000485824	24
SH3YL1	ENST00000356150	NM_015677.3	ENSP00000348471	24
SH3YL1	ENST00000463865	?	?	25
SH3YL1	ENST00000479739	?	ENSP00000441266	21
SH3YL1	ENST00000465733	?	?	4
SH3YL1	ENST00000403657	?	ENSP00000385668	22
SH3YL1	ENST00000462719	?	?	4
SH3YL1	ENST00000488044	?	?	14
SH3YL1	ENST00000481932	?	ENSP00000474411	2
SH3YL1	ENST00000451005	?	ENSP00000416312	18
SH3YL1	ENST00000431160	?	ENSP00000399519	8
SH3YL1	ENST00000460974	?	?	1
SH3YL1	ENST00000475027	?	?	13
SH3YL1	ENST00000405430	?	ENSP00000384269	24
SH3YL1	ENST00000415368	?	ENSP00000410235	7
SH3YL1	ENST00000473104	?	?	23
SH3YL1	ENST00000488979	?	?	12
SH3YL1	ENST00000402632	?	ENSP00000384910	13
SH3YL1	ENST00000497051	?	?	6
SH3YL1	ENST00000454318	?	ENSP00000415723	13
SH3YL1	ENST00000471948	?	?	12
SH3YL1	ENST00000472012	?	?	7
SH3YL1	ENST00000403658	NM_001282682.1	ENSP00000383928	25
SH3YL1	ENST00000403712	NM_001159597.2	ENSP00000384276	24
SH3YL1	ENST00000472861	?	?	4

Locus 3 (sentinel SNP rs11553746 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

SH3YL1	ENST00000477707	?	?	4

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ACP1	ENST00000442386	?	ENSP00000389681	2
ACP1	ENST00000413140	?	ENSP00000410331	2
ACP1	ENST00000453390	?	ENSP00000411121	1
ACP1	ENST00000405364	?	ENSP00000384184	1
SH3YL1	ENST00000626873	NM_001282687.1	ENSP00000485824	5
SH3YL1	ENST00000356150	NM_015677.3	ENSP00000348471	5
SH3YL1	ENST00000479739	?	ENSP00000441266	4
SH3YL1	ENST00000403657	?	ENSP00000385668	5
SH3YL1	ENST00000481932	?	ENSP00000474411	1
SH3YL1	ENST00000405430	?	ENSP00000384269	5
SH3YL1	ENST00000402632	?	ENSP00000384910	3
SH3YL1	ENST00000403712	NM_001159597.2	ENSP00000384276	5

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SH3YL1	ENST00000356150	NM_015677.3	ENSP00000348471	1
SH3YL1	ENST00000403712	NM_001159597.2	ENSP00000384276	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ACP1	ENST00000484125	?	1
ACP1	ENST00000484464	?	2
ACP1	ENST00000463831	?	2
SH3YL1	ENST00000468321	?	4
SH3YL1	ENST00000602998	?	1
SH3YL1	ENST00000463865	?	5
SH3YL1	ENST00000473104	?	4
SH3YL1	ENST00000471948	?	3