Locus 430 (rs300703)

SNiPAcard

Block annotations

Block info

genomic range	chr2:92,999-286,812
block size	193,814 bp
variant count	21 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.867 [-4.969 – 0.641]	gene(s) hit or close-by	AC079779.4, AC079779.7, ACP1, FAM150B, SH3YL1
phastCons	μ = 0.037 [0 – 0.676]	eQTL gene(s)	AC079779.4, AC079779.7, SH3YL1
GERP++	μ = -0.316 [-2.61 – 1.65]	potentially regulated gene(s)	AC093326.2, AC105393.2, SH3YL1, TMEM18
CADD score	μ = 3.032 [0.03 – 10.34]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SH3YL1	?	ENSG00000035115	muscularis mucosae	9.02×10^{-7} (p-value)	GTEx Portal V6	18
AC079779.7	?	ENSG00000227061	testis	1.95×10^{-7} (p-value)	GTEx Portal V6	17
SH3YL1	?	ENSG00000035115	left ventricle	5.23×10^{-6} (p-value)	GTEx Portal V6	1
AC079779.4	?	ENSG00000228643	thyroid	1.90×10^{-5} (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000024884		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000234694	1	ENCP00000024903	TMEM18
		ENCP00000024897	AC105393.2
		ENCP00000024899	AC093326.2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001538823 (open chromatin region)	1	A549	H3K27me3
		blood (K562)	H3K27me3
		muscle (HSMM)	DNase1

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
ACP1	2	ENST00000272065 ENST00000272067 ENST00000405233 ENST00000405364 ENST00000413140 ENST00000442386 ENST00000453390	hsa-miR-216b-5p hsa-miR-9-5p

ENST00000463831
ENST00000480874
ENST00000484464

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC079779.7	downstream gene variant	388	ENST00000437798	?	?	2
ACP1	upstream gene variant, downstream gene variant	440	ENST00000439645	?	ENSP00000408596	3
ACP1	upstream gene variant	870	ENST00000484125	?	?	1
ACP1	upstream gene variant, downstream gene variant	1649	ENST00000405364	?	ENSP00000384184	2
ACP1	upstream gene variant, downstream gene variant	1503	ENST00000463831	?	?	2
ACP1	upstream gene variant, downstream gene variant	1602	ENST00000272065	NM_004300.3	ENSP00000272065	2
ACP1	upstream gene variant, downstream gene variant	1599	ENST00000272067	NM_007099.3	ENSP00000272067	2
ACP1	upstream gene variant, downstream gene variant	1935	ENST00000484464	?	?	2
ACP1	upstream gene variant, downstream gene variant	1630	ENST00000442386	?	ENSP00000389681	2
ACP1	upstream gene variant, downstream gene variant	1695	ENST00000453390	?	ENSP00000411121	2
ACP1	upstream gene variant, downstream gene variant	1623	ENST00000413140	?	ENSP00000410331	2
ACP1	upstream gene variant, downstream gene variant	1297	ENST00000480874	?	?	2
ACP1	upstream gene variant, downstream gene variant	1656	ENST00000405233	?	ENSP00000384307	2
ACP1	upstream gene variant, downstream gene variant	513	ENST00000407983	NM_001040649.2	ENSP00000385404	3
FAM150B	downstream gene variant	5000	ENST00000463919	?	?	1
FAM150B	downstream gene variant	3021	ENST00000619265	?	ENSP00000478180	1
FAM150B	downstream gene variant	3021	ENST00000401489	?	ENSP00000385214	1
FAM150B	downstream gene variant	2858	ENST00000401503	?	ENSP00000385537	1
FAM150B	downstream gene variant	3043	ENST00000344414	?	ENSP00000339565	1
FAM150B	downstream gene variant	2172	ENST00000452023	?	ENSP00000389939	1
FAM150B	downstream gene variant	3021	ENST00000405290	?	ENSP00000385672	1
FAM150B	downstream gene variant	2650	ENST00000403610	NM_001002919.2	ENSP00000384604	1
SH3YL1	downstream gene variant	4428	ENST00000460974	?	?	1
SH3YL1	downstream gene variant	4664	ENST00000454318	?	ENSP00000415723	1
SH3YL1	downstream gene variant	3679	ENST00000602998	?	?	1
SH3YL1	downstream gene variant, upstream gene variant	484	ENST00000473104	?	?	2
SH3YL1	upstream gene variant	2978	ENST00000605370	?	?	1
SH3YL1	upstream gene variant	2562	ENST00000479739	?	ENSP00000441266	1
SH3YL1	downstream gene variant	1873	ENST00000471948	?	?	1
SH3YL1	downstream gene variant	2632	ENST00000488044	?	?	1
SH3YL1	downstream gene variant	4847	ENST00000626873	NM_001282687.1	ENSP00000485824	1
SH3YL1	downstream gene variant	4618	ENST00000475027	?	?	1
SH3YL1	downstream gene variant	4851	ENST00000405430	?	ENSP00000384269	1
SH3YL1	downstream gene variant	3504	ENST00000415368	?	ENSP00000410235	1
SH3YL1	downstream gene variant, upstream gene variant	1947	ENST00000403657	?	ENSP00000385668	2
SH3YL1	downstream gene variant	145	ENST00000402632	?	ENSP00000384910	3
SH3YL1	downstream gene variant, upstream gene variant	1874	ENST00000481932	?	ENSP00000474411	3
SH3YL1	downstream gene variant	4614	ENST00000488079	?	?	1

SH3YL1	downstream gene variant	4614	ENST00000488979	?	?	1
SH3YL1	downstream gene variant	1389	ENST00000431160	?	ENSP00000399519	1
SH3YL1	downstream gene variant, upstream gene variant	2427	ENST00000497051	?	?	2
SH3YL1	downstream gene variant	4856	ENST00000356150	NM_015677.3	ENSP00000348471	1
SH3YL1	downstream gene variant	4834	ENST00000403712	NM_001159597.2	ENSP00000384276	1
SH3YL1	downstream gene variant	4847	ENST00000463865	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC079779.4	ENST00000427831	?	?	1
AC079779.4	ENST00000450709	?	?	1
ACP1	ENST00000484464	?	?	2
ACP1	ENST00000405364	?	ENSP00000384184	1
ACP1	ENST00000272065	NM_004300.3	ENSP00000272065	2
ACP1	ENST00000463831	?	?	1
ACP1	ENST00000272067	NM_007099.3	ENSP00000272067	2
ACP1	ENST00000480874	?	?	1
ACP1	ENST00000442386	?	ENSP00000389681	2
ACP1	ENST00000413140	?	ENSP00000410331	2
ACP1	ENST00000453390	?	ENSP00000411121	2
FAM150B	ENST00000452023	?	ENSP00000389939	1
FAM150B	ENST00000401489	?	ENSP00000385214	2
FAM150B	ENST00000619265	?	ENSP00000478180	2
FAM150B	ENST00000405290	?	ENSP00000385672	2
FAM150B	ENST00000401503	?	ENSP00000385537	2
FAM150B	ENST00000403610	NM_001002919.2	ENSP00000384604	2
FAM150B	ENST00000344414	?	ENSP00000339565	2
FAM150B	ENST00000463919	?	?	1
SH3YL1	ENST00000405430	?	ENSP00000384269	9
SH3YL1	ENST00000463865	?	?	9
SH3YL1	ENST00000415368	?	ENSP00000410235	2
SH3YL1	ENST00000488044	?	?	8
SH3YL1	ENST00000403657	?	ENSP00000385668	8
SH3YL1	ENST00000479739	?	ENSP00000441266	8
SH3YL1	ENST00000402632	?	ENSP00000384910	5
SH3YL1	ENST00000468321	?	?	9
SH3YL1	ENST00000462719	?	?	1
SH3YL1	ENST00000473104	?	?	8
SH3YL1	ENST00000475027	?	?	8
SH3YL1	ENST00000403712	NM_001159597.2	ENSP00000384276	9

SH3YL1	ENST00000488979	?	?	8
SH3YL1	ENST00000451005	?	ENSP00000416312	8
SH3YL1	ENST00000471948	?	?	8
SH3YL1	ENST00000431160	?	ENSP00000399519	7
SH3YL1	ENST00000497051	?	?	6
SH3YL1	ENST00000626873	NM_001282687.1	ENSP00000485824	9
SH3YL1	ENST00000403658	NM_001282682.1	ENSP00000383928	9
SH3YL1	ENST00000454318	?	ENSP00000415723	8
SH3YL1	ENST00000356150	NM_015677.3	ENSP00000348471	9
SH3YL1	ENST00000472012	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ACP1	ENST00000405233	?	ENSP00000384307	1
SH3YL1	ENST00000402632	?	ENSP00000384910	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SH3YL1	ENST00000405430	?	ENSP00000384269	1
SH3YL1	ENST00000415368	?	ENSP00000410235	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
SH3YL1	ENST00000465733	?	1
SH3YL1	ENST00000471948	?	1
SH3YL1	ENST00000472861	?	1
SH3YL1	ENST00000477707	?	1