Locus 209 (rs300756)

SNiPAcard

Block annotations

Block info

genomic range	chr2:233,416-277,995
block size	44,580 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.458 [-0.646 – -0.269]	gene(s) hit or close-by	ACP1, FAM150B, SH3YL1
phastCons	μ = 0.001 [0 – 0.001]	eQTL gene(s)	SH3YL1
GERP++	μ = -3.740 [-6.45 – -1.03]	potentially regulated gene(s)	–
CADD score	μ = 0.765 [0.18 – 1.351]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SH3YL1	?	ENSG00000035115	sun exposed skin	3.77×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
ACP1	1	ENST00000272065 ENST00000272067	hsa-miR-300 hsa-miR-381-3p hsa-miR-520c-3p hsa-miR-520d-3p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ACP1	downstream gene variant	4840	ENST00000405233	?	ENSP00000384307	1
ACP1	downstream gene variant	489	ENST00000413140	?	ENSP00000410331	1
ACP1	downstream gene variant	489	ENST00000463831	?	?	1
ACP1	downstream gene variant	626	ENST00000484464	?	?	1
ACP1	downstream gene variant	2384	ENST00000480874	?	?	1
ACP1	downstream gene variant	734	ENST00000442386	?	ENSP00000389681	1
ACP1	downstream gene variant	2795	ENST00000405364	?	ENSP00000384184	1
ACP1	downstream gene variant	620	ENST00000453390	?	ENSP00000411121	1
FAM150B	downstream gene variant	1934	ENST00000401489	?	ENSP00000385214	1
FAM150B	downstream gene variant	1956	ENST00000344414	?	ENSP00000339565	1
FAM150B	downstream gene variant	1563	ENST00000403610	NM_001002919.2	ENSP00000384604	1
FAM150B	downstream gene variant	1771	ENST00000401503	?	ENSP00000385537	1
FAM150B	downstream gene variant	1934	ENST00000619265	?	ENSP00000478180	1
FAM150B	downstream gene variant	1934	ENST00000405290	?	ENSP00000385672	1
SH3YL1	upstream gene variant	2257	ENST00000472012	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SH3YL1	ENST00000488979	?	?	1
SH3YL1	ENST00000403657	?	ENSP00000385668	1
SH3YL1	ENST00000497051	?	?	1
SH3YL1	ENST00000454318	?	ENSP00000415723	1
SH3YL1	ENST00000475027	?	?	1
SH3YL1	ENST00000405430	?	ENSP00000384269	1
SH3YL1	ENST00000403712	NM_001159597.2	ENSP00000384276	1
SH3YL1	ENST00000468321	?	?	1
SH3YL1	ENST00000488044	?	?	1
SH3YL1	ENST00000481932	?	ENSP00000474411	1
SH3YL1	ENST00000463865	?	?	1
SH3YL1	ENST00000626873	NM_001282687.1	ENSP00000485824	1
SH3YL1	ENST00000356150	NM_015677.3	ENSP00000348471	1
SH3YL1	ENST00000451005	?	ENSP00000416312	1
SH3YL1	ENST00000473104	?	?	1
SH3YL1	ENST00000479739	?	ENSP00000441266	1
SH3YL1	ENST00000403658	NM_001282682.1	ENSP00000383928	1
SH3YL1	ENST00000471948	?	?	1
SH3YL1	ENST00000431160	?	ENSP00000399519	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ACP1	ENST00000272067	NM_007099.3	ENSP00000272067	1
ACP1	ENST00000272065	NM_004300.3	ENSP00000272065	1