Locus 337 (rs1513890)

SNiPAcard

Block annotations

Block info

genomic range	chr4:76,830,353-76,840,698
block size	10,346 bp
variant count	61 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.564 [-3.774 – 0.968]	gene(s) hit or close-by	NAAA
phastCons	μ = 0.071 [0 – 0.993]	eQTL gene(s)	CDKL2, NAAA, PPEF2
GERP++	μ = 0.145 [-3.31 – 3.69]	potentially regulated gene(s)	ART3, CXCL9, SDAD1
CADD score	μ = 3.814 [0.197 – 13.39]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PPEF2	?	ENSG00000156194	tibial nerve	3.33×10-6 (p-value)	GTEx Portal V6	56
NAAA	?	ENSG00000138744	tibial nerve	4.32×10-6 (p-value)	GTEx Portal V6	2
CDKL2	?	ENSG00000138769	tibial nerve	6.76×10-6 (p-value)	GTEx Portal V6	1
PPEF2	?	ENSG00000156194	cerebellum	3.98×10-9 (p-value)	GTEx Portal V6	60
PPEF2	?	ENSG00000156194	subcutaneous adipocytes	1.07×10-6 (p-value)	GTEx Portal V6	58
NAAA	?	ENSG00000138744	subcutaneous adipocytes	8.70×10-6 (p-value)	GTEx Portal V6	1
PPEF2	?	ENSG00000156194	cerebellar hemisphere	7.72×10-6 (p-value)	GTEx Portal V6	1
NAAA	?	ENSG00000138744	unexposed skin	8.29×10-6 (p-value)	GTEx Portal V6	4

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000382796	2	ENCP00000041424	CXCL9
		ENCP00000041435	ART3
ENCE00000382774	2	ENCP00000041420	SDAD1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001432205 (promoter flanking region)	2	monocytes (Monocytes-CD14+)	H3K27ac
		muscle (HSMM)	DNase1
ENSR00001685887 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K36me3
		blood (K562)	H3K27me3
ENSR00001243211 (CTCF binding site)	3	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF

		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	DNase1, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	CTCF
		A549	CTCF, H3K36me3
ENSR00001685888 (promoter flanking region)	5	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, H3K36me3
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF, Rad21
		lung (IMR90)	CTCF, DNase1
		blood (GM12878)	Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF, H3K36me3
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	CTCF
		A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
NAAA	downstream gene variant	227	ENST00000286733	NM_014435.3	ENSP00000286733	32
NAAA	downstream gene variant	50	ENST00000507956	?	ENSP00000427641	26
NAAA	downstream gene variant	154	ENST00000511606	?	?	34
NAAA	downstream gene variant	256	ENST00000602782	?	ENSP00000473575	24
NAAA	downstream gene variant	94	ENST00000513045	?	ENSP00000423397	34
NAAA	downstream gene variant	10	ENST00000505594	?	ENSP00000426977	24

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
NAAA	ENST00000286733	NM_014435.3	ENSP00000286733	25
NAAA	ENST00000507956	?	ENSP00000427641	12
NAAA	ENST00000511606	?	?	25
NAAA	ENST00000602782	?	ENSP00000473575	17
NAAA	ENST00000513045	?	ENSP00000423397	25

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
NAAA	ENST00000286733	NM_014435.3	ENSP00000286733	10
NAAA	ENST00000507956	?	ENSP00000427641	6

NAAA	ENST00000505594	?	ENSP00000426977	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
NAAA	ENST00000511606	?	2