Locus 419 (rs1046668)

SNiPAcard

Block annotations

Block info

genomic range	chr2:152,206,464-152,239,059
block size	32,596 bp
variant count	35 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.613 [-3.041 – 1.071]	gene(s) hit or close-by	MIR4773-2, RN7SL124P, TNFAIP6
phastCons	μ = 0.086 [0 – 0.997]	eQTL gene(s)	NMI, RBM43
GERP++	μ = -0.363 [-4.58 – 3.46]	potentially regulated gene(s)	TNFAIP6
CADD score	μ = 4.274 [0.025 – 14.86]	disease gene(s)	–

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)
TNFAIP6	missense variant	ENST00000243347	NM_007115.3	ENSP00000243347	R/Q	cGa/cAa	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RBM43	ENST00000331426	ILMN_1858599	blood	6.08×10^{-6} (p-value)	Westra et al.	8
NMI	ENST00000243346	ILMN_1739541	blood	7.25×10^{-5} (p-value)	Westra et al.	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000505495	1	ENST00000243347, ENST00000460812	TNFAIP6

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001547521 (promoter flanking region)	2	NHLF	DNase1, H3K27ac
		HSMMtube	DNase1
		Osteobl	H3K27ac, H3K4me2
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		breast (HMEC)	DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	PolII
		monocytes (Monocytes-CD14+)	DNase1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
ENSR00000304445 (enhancer)	2	Osteobl	H3K27ac
		skin (NHDF-AD)	DNase1

ENSR00000600272 (promoter flanking region)	4	NHLF	DNase1
		embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K27me3, DNase1
		Osteobl	H3K27ac, H3K4me2
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		lung (IMR90)	DNase1
		blood (GM12878)	BATF
		A549	DNase1
		nervous (NH-A)	DNase1
ENSR00001657792 (enhancer)	2	muscle (HSMM)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MIR4773-2	upstream gene variant, downstream gene variant	244	ENST00000585225	?	?	11
RN7SL124P	upstream gene variant, downstream gene variant	119	ENST00000498656	?	?	16
TNFAIP6	upstream gene variant, downstream gene variant	1707	ENST00000243347	NM_007115.3	ENSP00000243347	5
TNFAIP6	upstream gene variant, downstream gene variant	691	ENST00000460812	?	?	12

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TNFAIP6	ENST00000243347	NM_007115.3	ENSP00000243347	26
TNFAIP6	ENST00000460812	?	?	9

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
TNFAIP6	ENST00000243347	NM_007115.3	ENSP00000243347	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
TNFAIP6	ENST00000460812	?	1