

SNiPACard

Block annotations

Block info

genomic range	chr2:152,206,464-152,239,059 <i>e!</i>
block size	32,596 bp
variant count	35 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	$\mu = -0.613$ [-3.041 – 1.071]	gene(s) hit or close-by	MIR4773-2 <i>e!</i> , RN7SL124P <i>e!</i> , TNFAIP6 <i>e!</i>
phastCons	$\mu = 0.086$ [0 – 0.997]	eQTL gene(s)	NMI <i>e!</i> , RBM43 <i>e!</i>
GERP++	$\mu = -0.363$ [-4.58 – 3.46]	potentially regulated gene(s)	TNFAIP6 <i>e!</i>
CADD score	$\mu = 4.274$ [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 <i>e!</i>	missense variant	ENST00000243347 <i>e!</i>	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 <i>e!</i>	ENST00000331426 <i>e!</i>	ILMN_1858599 <i>e!</i>	blood	6.08×10 ⁻⁶ (p-value)	Westra et al. 	8
NMI <i>e!</i>	ENST00000243346 <i>e!</i>	ILMN_1739541 <i>e!</i>	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 <i>e!</i>	1	ENST00000243347 <i>e!</i> , ENST00000460812 <i>e!</i>	TNFAIP6 <i>e!</i>

Regulatory feature cluster

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

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

Variation proximal to gene

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

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TNFAIP6 <i>e!</i>	ENST00000243347 <i>e!</i>	NM_007115.3	ENSP00000243347 <i>e!</i>	26
TNFAIP6 <i>e!</i>	ENST00000460812 <i>e!</i>	?	?	9

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
TNFAIP6 <i>e!</i>	ENST00000243347 <i>e!</i>	NM_007115.3	ENSP00000243347 <i>e!</i>	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
TNFAIP6 <i>e!</i>	ENST00000460812 <i>e!</i>	?	1

