Locus 12 (rs3117600)

SNiPAcard

Block annotations

Block info

genomic range	chr4:74,729,616-74,734,668
block size	5,053 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.100 [-3.83 – 0.025]	gene(s) hit or close-by	CXCL1
phastCons	μ = 0.000 [0 – 0]	eQTL gene(s)	PF4V1
GERP++	μ = -1.685 [-5.79 – 2.96]	potentially regulated gene(s)	AC112518.3, AFM, CXCL6, EREG, MTHFD2L
CADD score	μ = 2.606 [0.082 – 7.402]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PF4V1	?	ENSG00000109272	blood	4.49×10-6 (p-value)	GTEx Portal V6	7
PF4V1	?	ENSG00000109272	breast	4.04×10-6 (p-value)	GTEx Portal V6	7
PF4V1	ENST00000226524	ILMN_1745522	monocyte	3.56×10-17 (p-value)	Zeller et al.	4
PF4V1	?	ENSG00000109272	transformed fibroblasts	8.50×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000382197	1	ENCP00000041379	MTHFD2L
		ENCP00000041366	CXCL6
		ENCP00000041383	EREG
		ENCP00000041357	AFM
		ENCP00000041361	AC112518.3

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001432039 (CTCF binding site)	2	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		blood (K562)	Rad21, CTCF
		blood (DND-41)	CTCF
		skin (NHDF-AD)	DNase1
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	CTCF, Rad21
		lung (IMR90)	H3K27me3
		blood (GM12878)	CTCF
		A549	CTCF
		skin (NHEK)	CTCF
ENSR00001685779 (enhancer)	1	monocytes (Monocytes-CD14+)	DNase1
		lung (IMR90)	H3K27me3

ENSR00001242853 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K4me3, H3K4me2, DNase1
		skin (NHEK)	H3K4me1, H3K9ac, H3K4me2, CTCF, DNase1
ENSR00001242854 (promoter)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, H3K4me2, H3K4me3, DNase1
		HSMMtube	H2AZ, H3K27me3
		blood (K562)	CTCF, Rad21, H3K9ac, H2AZ, H3K4me2, H3K4me3, H3K27me3
		skin (NHDF-AD)	H3K4me3, H3K4me2
		muscle (HSMM)	H3K4me3, H2AZ
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3, H3K4me3
		blood (GM12878)	H3K27me3
		nervous (NH-A)	H3K4me2, H3K27me3
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1, H3K4me1
		NHLF	H3K9ac, H3K4me3
		Osteobl	H3K4me2, H3K4me3, H2AZ
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K27ac, H3K9ac, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K4me3, PolII
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CXCL1	upstream gene variant	442	ENST00000509101	?	?	7
CXCL1	upstream gene variant	453	ENST00000395761	NM_001511.3	ENSP00000379110	7