Locus 17 (rs4525)

SNiPAcard

Block annotations

Block info

genomic range	chr1:169,469,142-169,513,436
block size	44,295 bp
variant count	41 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.242 [-2.54 – 1.334]	gene(s) hit or close-by	F5
phastCons	μ = 0.038 [0 – 0.602]	eQTL gene(s)	–
GERP++	μ = -0.719 [-5.59 – 2.81]	potentially regulated gene(s)	RP1-206D15.5, SELL, SELP
CADD score	μ = 2.836 [0.001 – 9.022]	disease gene(s)	F5

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM101722	1

Disease gene annotation

gene	trait	source DB	source entry/link
F5	STROKE, ISCHEMIC	OMIM	MIM:601367
F5	THROMBOPHILIA DUE TO ACTIVATED PROTEIN C RESISTANCE	OMIM	MIM:188055
F5	PREGNANCY LOSS, RECURRENT, SUSCEPTIBILITY TO, 1	OMIM	MIM:614389
F5	FACTOR V DEFICIENCY	OMIM	MIM:227400
F5	BUDD-CHIARI SYNDROME	OMIM	MIM:600880
F5	Congenital factor V deficiency	OrphaNet	OrphaNet:326
F5	Cerebral sinovenous thrombosis	OrphaNet	OrphaNet:329217
F5	BUDD-CHIARI SYNDROME	OrphaNet	OrphaNet:131
F5	East Texas bleeding disorder	OrphaNet	OrphaNet:391320

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)
F5	missense variant	ENST00000367796	?	ENSP00000356770	3	3			3
F5	missense variant	ENST00000367797	NM_000130.4	ENSP00000356771	3	3			3

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000043595	1	ENCP00000005439	RP1-206D15.5
ENCE00000043639	1	ENCP00000005449	SELL
		ENCP00000005442	SELP

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000547323 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	Rad21, H3K4me1, CTCF
		blood (K562)	H2AZ
ENSR00001525842 (promoter flanking region)	1	cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	CTCF, H3K4me2, Rad21
		blood (K562)	DNase1, H3K9ac, H2AZ, H3K4me3
ENSR00001525843 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		breast (HMEC)	DNase1
		cervix (HeLa-S3)	DNase1, Cjun, Jund
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K4me1, H3K36me3
		nervous (NH-A)	DNase1
ENSR00000547326 (promoter flanking region)	1	cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K36me3
		muscle (HSMM)	DNase1
ENSR00000287070 (promoter flanking region)	4	embryonic stem cell (H1ESC)	Rad21, Jund, ATF3, Tcf12, BCL11A, H3K4me2, SP1, USF1, DNase1
		HSMMtube	DNase1
		Osteobl	H3K27me3
		blood (DND-41)	H3K4me1
		muscle (HSMM)	DNase1
		breast (HMEC)	DNase1
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K4me2
ENSR00001587730 (CTCF binding site)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1, Cjun, Jund, Cfos
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1, H3K4me1
ENSR00000547331 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1, Cfos, Jund, Cjun
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1, H3K4me1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
F5	downstream gene variant	1454	ENST00000367796	?	ENSP00000356770	3

F5	downstream gene variant, upstream gene variant	1454	ENST00000495481	?	?		7
F5	downstream gene variant	1454	ENST00000367797	NM_000130.4	ENSP00000356771		3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
F5	ENST00000367796	?	ENSP00000356770	4	4	4
F5	ENST00000367797	NM_000130.4	ENSP00000356771	4	4	4

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
F5	ENST00000367796	?	ENSP00000356770	25
F5	ENST00000495481	?	?	1
F5	ENST00000367797	NM_000130.4	ENSP00000356771	25