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	HGMD®	1

Disease gene annotation

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

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