Locus 165 (rs6033)

SNiPAcard

Block annotations

Block info

genomic range	chr1:169,513,583-169,521,853
block size	8,271 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.657 [-0.932 – 2.883]	gene(s) hit or close-by	F5
phastCons	μ = 0.363 [0 – 1]	eQTL gene(s)	–
GERP++	μ = -0.075 [-10.6 – 3.18]	potentially regulated gene(s)	–
CADD score	μ = 8.529 [0.002 – 16.68]	disease gene(s)	F5

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM045144	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	T/M	aCg/aTg	?	?	1
F5	missense variant	ENST00000367797	NM_000130.4	ENSP00000356771	T/M	aCg/aTg	?	?	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000671446 (CTCF binding site)	2	embryonic stem cell (H1ESC)	CTCF, DNase1, Rad21
		HSMMtube	CTCF, DNase1
		blood (K562)	H3K27me3, DNase1, Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	DNase1, CTCF

		liver (HepG2)	H3K36me3, Rad21, FOXA1, CTCF
		lung (IMR90)	CTCF
		blood (GM12878)	Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF, H3K36me3
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	DNase1, CTCF
ENSR00001525847 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	CTCF, H3K27me3, DNase1
		blood (K562)	CTCF
		blood (DND-41)	CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	CTCF
		liver (HepG2)	Rad21, CTCF, H3K36me3
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	CTCF
		A549	CTCF
		nervous (NH-A)	CTCF, H3K27me3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
F5	ENST00000367796	?	ENSP00000356770	3	3	3
F5	ENST00000367797	NM_000130.4	ENSP00000356771	3	3	3

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
F5	ENST00000367796	?	ENSP00000356770	5
F5	ENST00000367797	NM_000130.4	ENSP00000356771	5