SNiPAcard

Block annotations

Block info

genomic range	chr1:169,527,101-169,563,951
block size	36,851 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.550 [-0.777 – 1.877]	**gene(s) hit or close-by**	F5, SELP
phastCons	μ = 0.019 [0.004 – 0.033]	**eQTL gene(s)**	F5
GERP++	μ = -0.890 [-3.92 – 2.14]	**potentially regulated gene(s)**	–
CADD score	μ = 4.322 [3.287 – 5.356]	**disease gene(s)**	F5

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Soluble levels of adhesion molecules	$<4.00\times10^{-61}$	GWAS Catalog	20167578	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Selectin p polymorphism	benign	ClinVar	RCV000014482.1	1
?	HGMD curated	HGMD	CM981792	1
SELECTIN P POLYMORPHISM	OMIM curated	OMIM	MIM:173610	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)
SELP	missense variant	ENST00000367788	?	ENSP00000356762	P/T	Cct/Act	?	?	1
SELP	missense variant	ENST00000367786	?	ENSP00000356760	P/T	Cct/Act	?	?	1
SELP	missense variant	ENST00000426706	?	ENSP00000391694	P/T	Cct/Act	?	?	1
SELP	missense variant	ENST00000263686	NM_003005.3	ENSP00000263686	P/T	Cct/Act	?	?	1
SELP	missense	ENST00000458599	?	ENSP00000399368	P/T	Cct/Act	?	?	1

variant

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
F5	?	ENSG00000198734	esophagus mucosa	4.00×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000547341 (TF binding site)	1	blood (GM12878)	BATF, DNase1, BCL11A
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SELP	upstream gene variant	3143	ENST00000466167	?	?	1

Putative effect on transcript

Intron variant

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