Locus 172 (rs9332701)

SNiPAcard

Block annotations

Block info

genomic range	chr1:169,474,397-169,484,767
block size	10,371 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.887 [-1.839 – 3.613]	gene(s) hit or close-by	F5
phastCons	μ = 0.500 [0.001 – 1]	eQTL gene(s)	–
GERP++	μ = 2.580 [-0.451 – 5.61]	potentially regulated gene(s)	–
CADD score	μ = 12.966 [0.632 – 25.3]	disease gene(s)	F5

Trait annotations

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 transcript

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
F5	ENST00000495481	?	1