Locus 367 (rs1403694)

SNiPAcard

Block annotations

Block info

genomic range	chr3:186,437,296-186,439,998
block size	2,703 bp
variant count	21 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.876 [-3.124 – 0.439]	gene(s) hit or close-by	KNG1, RP11-573D15.8
phastCons	μ = 0.009 [0 – 0.152]	eQTL gene(s)	KNG1
GERP++	μ = -0.343 [-6.02 – 1.81]	potentially regulated gene(s)	RP11-48F14.1
CADD score	μ = 2.657 [0.161 – 8.36]	disease gene(s)	KNG1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
bradykinin, des-arg(9)	2.21×10^{-5}	Metabolomics GWAS Server	24816252	1

Disease gene annotation

gene	trait	source DB	source entry/link
KNG1	HIGH MOLECULAR WEIGHT KININOGEN DEFICIENCY	OMIM	MIM:228960
KNG1	Congenital high-molecular-weight kininogen deficiency	OrphaNet	OrphaNet:483

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
KNG1	?	ENSG00000113889	frontal cortex	2.73×10^{-9} (p-value)	GTEx Portal V6	20
KNG1	?	ENSG00000113889	cortex	1.16×10^{-6} (p-value)	GTEx Portal V6	13

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000365623	2	ENCP00000039492	RP11-48F14.1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001381866 (open chromatin region)	2	blood (K562)	H3K27me3, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-573D15.8	upstream gene variant	1003	ENST00000630726	?	?	21

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
RP11-573D15.8	ENST00000628601	?	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
KNG1	ENST00000265023	NM_001102416.2	ENSP00000265023	21
KNG1	ENST00000447445	NM_001166451.1	ENSP00000396025	21
KNG1	ENST00000287611	NM_000893.3	ENSP00000287611	21
RP11-573D15.8	ENST00000626633	?	?	21
RP11-573D15.8	ENST00000630331	?	?	21
RP11-573D15.8	ENST00000625303	?	?	21
RP11-573D15.8	ENST00000629106	?	?	21
RP11-573D15.8	ENST00000625386	?	?	21
RP11-573D15.8	ENST00000628190	?	?	21
RP11-573D15.8	ENST00000626845	?	?	21
RP11-573D15.8	ENST00000627919	?	?	21
RP11-573D15.8	ENST00000629451	?	?	21
RP11-573D15.8	ENST00000630178	?	?	21
RP11-573D15.8	ENST00000630864	?	?	21
RP11-573D15.8	ENST00000627830	?	?	21
RP11-573D15.8	ENST00000625741	?	?	21
RP11-573D15.8	ENST00000626151	?	?	21
RP11-573D15.8	ENST00000627469	?	?	21
RP11-573D15.8	ENST00000354642	?	?	21
RP11-573D15.8	ENST00000627551	?	?	21
RP11-573D15.8	ENST00000628601	?	?	21
RP11-573D15.8	ENST00000628253	?	?	21
RP11-573D15.8	ENST00000628505	?	?	21
RP11-573D15.8	ENST00000625839	?	?	21
RP11-573D15.8	ENST00000599314	?	?	21
RP11-573D15.8	ENST00000628728	?	?	21

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-573D15.8	ENST00000628601	?	2