

SNiPAcad

Block annotations

Block info


genomic range	chr3:186,431,070-186,458,322 <i>e!</i>
block size	27,253 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	$\mu = 0.023$ [-0.404 – 0.53]	gene(s) hit or close-by	KNG1 <i>e!</i> , RP11-573D15.8 <i>e!</i> , RP11-795A2.3 <i>e!</i>
phastCons	$\mu = 0.002$ [0 – 0.008]	eQTL gene(s)	–
GERP++	$\mu = -0.121$ [-1.03 – 0.6]	potentially regulated gene(s)	–
CADD score	$\mu = 2.264$ [0.571 – 4.553]	disease gene(s)	KNG1 <i>e!</i>

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
bradykinin, des-arg(9)	1.37×10 ⁻¹²	Metabolomics GWAS Server	24816252 	2

Disease gene annotation

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

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
KNG1 <i>e!</i>	upstream gene variant	4067	ENST00000447445 <i>e!</i>	NM_001166451.1	ENSP00000396025 <i>e!</i>	1
KNG1 <i>e!</i>	upstream gene variant	4050	ENST00000265023 <i>e!</i>	NM_001102416.2	ENSP00000265023 <i>e!</i>	1
KNG1 <i>e!</i>	upstream gene variant	3995	ENST00000287611 <i>e!</i>	NM_000893.3	ENSP00000287611 <i>e!</i>	1
RP11-573D15.8 <i>e!</i>	downstream gene variant	3968	ENST00000599314 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	downstream gene variant	1167	ENST00000354642 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	downstream gene variant	721	ENST00000629126 <i>e!</i>	?	?	3
RP11-573D15.8 <i>e!</i>	downstream gene variant	1379	ENST00000629734 <i>e!</i>	?	?	3
RP11-573D15.8 <i>e!</i>	upstream gene variant	2714	ENST00000626649 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	downstream gene variant	678	ENST00000626306 <i>e!</i>	?	?	2
RP11-573D15.8 <i>e!</i>	downstream gene variant	4417	ENST00000627551 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	downstream gene variant	1112	ENST00000628020 <i>e!</i>	?	?	3
RP11-573D15.8 <i>e!</i>	downstream gene variant	3189	ENST00000631299 <i>e!</i>	?	?	2
RP11-573D15.8 <i>e!</i>	downstream gene variant	3191	ENST00000609726 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	downstream gene variant	3191	ENST00000626520 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	downstream gene variant	3172	ENST00000596632 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	downstream gene variant	3191	ENST00000626978 <i>e!</i>	?	?	1

RP11-573D15.8 <i>e!</i>	downstream gene variant	3191	ENST00000625690 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	downstream gene variant	3175	ENST00000596329 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	downstream gene variant	1288	ENST00000628831 <i>e!</i>	?	?	3
RP11-573D15.8 <i>e!</i>	downstream gene variant	3191	ENST00000609652 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	downstream gene variant	3191	ENST00000628291 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	downstream gene variant	3191	ENST00000625869 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	downstream gene variant	3170	ENST00000626586 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	downstream gene variant	3191	ENST00000629201 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	downstream gene variant	3191	ENST00000628928 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	downstream gene variant	3191	ENST00000629954 <i>e!</i>	?	?	1
RP11-795A2.3 <i>e!</i>	downstream gene variant	4707	ENST00000413467 <i>e!</i>	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
KNG1 <i>e!</i>	ENST00000287611 <i>e!</i>	NM_000893.3	ENSP00000287611 <i>e!</i>	4
KNG1 <i>e!</i>	ENST00000447445 <i>e!</i>	NM_001166451.1	ENSP00000396025 <i>e!</i>	4
KNG1 <i>e!</i>	ENST00000265023 <i>e!</i>	NM_001102416.2	ENSP00000265023 <i>e!</i>	4
RP11-573D15.8 <i>e!</i>	ENST00000625386 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000628190 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000627919 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000627268 <i>e!</i>	?	?	4
RP11-573D15.8 <i>e!</i>	ENST00000630178 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000630331 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000626633 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000628858 <i>e!</i>	?	?	4
RP11-573D15.8 <i>e!</i>	ENST00000628831 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	ENST00000628601 <i>e!</i>	?	?	4
RP11-573D15.8 <i>e!</i>	ENST00000627830 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000626151 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000626306 <i>e!</i>	?	?	2
RP11-573D15.8 <i>e!</i>	ENST00000629106 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000628020 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	ENST00000628505 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000628728 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000630864 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000630726 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	ENST00000627015 <i>e!</i>	?	?	4
RP11-573D15.8 <i>e!</i>	ENST00000625741 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000354642 <i>e!</i>	?	?	4
RP11-573D15.8 <i>e!</i>	ENST00000627551 <i>e!</i>	?	?	4

RP11-573D15.8 <i>e!</i>	ENST00000627551 <i>e!</i>	?	?	4
RP11-573D15.8 <i>e!</i>	ENST00000599314 <i>e!</i>	?	?	4
RP11-573D15.8 <i>e!</i>	ENST00000625303 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000627469 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000625710 <i>e!</i>	?	?	4
RP11-573D15.8 <i>e!</i>	ENST00000626845 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000628253 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000629126 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	ENST00000625839 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000631299 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	ENST00000629451 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000629734 <i>e!</i>	?	?	1

