Locus 288 (rs3856930)

SNiPAcard

Block annotations

Block info

genomic range	chr3:186,431,070-186,458,322
block size	27,253 bp
variant count	5 variants

Basic features

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

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	HIGH MOLECULAR WEIGHT KININOGEN DEFICIENCY	OMIM	MIM:228960
KNG1	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	upstream gene variant	4067	ENST00000447445	NM_001166451.1	ENSP00000396025	1
KNG1	upstream gene variant	4050	ENST00000265023	NM_001102416.2	ENSP00000265023	1
KNG1	upstream gene variant	3995	ENST00000287611	NM_000893.3	ENSP00000287611	1
RP11-573D15.8	downstream gene variant	3968	ENST00000599314	?	?	1
RP11-573D15.8	downstream gene variant	1167	ENST00000354642	?	?	1
RP11-573D15.8	downstream gene variant	721	ENST00000629126	?	?	3
RP11-573D15.8	downstream gene variant	1379	ENST00000629734	?	?	3
RP11-573D15.8	upstream gene variant	2714	ENST00000626649	?	?	1
RP11-573D15.8	downstream gene variant	678	ENST00000626306	?	?	2
RP11-573D15.8	downstream gene variant	4417	ENST00000627551	?	?	1
RP11-573D15.8	downstream gene variant	1112	ENST00000628020	?	?	3
RP11-573D15.8	downstream gene variant	3189	ENST00000631299	?	?	2
RP11-573D15.8	downstream gene variant	3191	ENST00000609726	?	?	1
RP11-573D15.8	downstream gene variant	3191	ENST00000626520	?	?	1
RP11-573D15.8	downstream gene variant	3172	ENST00000596632	?	?	1
RP11-573D15.8	downstream gene variant	3191	ENST00000626978	?	?	1

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

Putative effect on transcript

Intron variant

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

RP11-573D15.8	ENST00000627551	?	?	4
RP11-573D15.8	ENST00000599314	?	?	4
RP11-573D15.8	ENST00000625303	?	?	5
RP11-573D15.8	ENST00000627469	?	?	5
RP11-573D15.8	ENST00000625710	?	?	4
RP11-573D15.8	ENST00000626845	?	?	5
RP11-573D15.8	ENST00000628253	?	?	5
RP11-573D15.8	ENST00000629126	?	?	1
RP11-573D15.8	ENST00000625839	?	?	5
RP11-573D15.8	ENST00000631299	?	?	1
RP11-573D15.8	ENST00000629451	?	?	5
RP11-573D15.8	ENST00000629734	?	?	1