Locus 96 (rs5030062)

SNiPAcard

Block annotations

Block info

genomic range	chr3:186,454,180-186,459,927
block size	5,748 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.444 [-0.367 – 1.999]	gene(s) hit or close-by	KNG1, RP11-573D15.8
phastCons	μ = 0.155 [0 – 0.667]	eQTL gene(s)	–
GERP++	μ = 0.739 [-0.875 – 5.61]	potentially regulated gene(s)	–
CADD score	μ = 5.384 [0.567 – 14.24]	disease gene(s)	KNG1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Plasma renin activity levels	$<9.00\times10^{-9}$	GWAS Catalog	25477429	1
Blood metabolite levels	$<6.00\times10^{-13}$	GWAS Catalog	24816252	1
Activated partial thromboplastin time	$<2.00\times10^{-203}$	GWAS Catalog	22703881	1
Activated partial thromboplastin time	$<1.00\times10^{-21}$	GWAS Catalog	20303064	1
bradykinin, des-arg(9)	5.94×10^{-13}	Metabolomics GWAS Server	24816252	4

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM112211	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 transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
KNG1	missense variant	ENST00000265023	NM_001102416.2	ENSP00000265023	I/T	aTa/aCa	tolerated	benign	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-573D15.8	downstream gene variant	1586	ENST00000628928	?	?	4
RP11-573D15.8	downstream gene variant	1586	ENST00000609652	?	?	4
RP11-573D15.8	downstream gene variant	71	ENST00000631299	?	?	5

RP11-573D15.8	downstream gene variant	1586	ENST00000609726	?	?	4
RP11-573D15.8	downstream gene variant	25	ENST00000628831	?	?	1
RP11-573D15.8	downstream gene variant	1586	ENST00000625869	?	?	4
RP11-573D15.8	downstream gene variant	1567	ENST00000596632	?	?	4
RP11-573D15.8	downstream gene variant	116	ENST00000629734	?	?	1
RP11-573D15.8	downstream gene variant	1586	ENST00000625690	?	?	4
RP11-573D15.8	downstream gene variant	1586	ENST00000628291	?	?	4
RP11-573D15.8	downstream gene variant	1586	ENST00000626520	?	?	4
RP11-573D15.8	downstream gene variant	1565	ENST00000626586	?	?	4
RP11-573D15.8	downstream gene variant	1570	ENST00000596329	?	?	4
RP11-573D15.8	downstream gene variant	1586	ENST00000629201	?	?	4
RP11-573D15.8	downstream gene variant	1586	ENST00000626978	?	?	4
RP11-573D15.8	downstream gene variant	1586	ENST00000629954	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
KNG1	ENST00000287611	NM_000893.3	ENSP00000287611	9
KNG1	ENST00000447445	NM_001166451.1	ENSP00000396025	9
KNG1	ENST00000265023	NM_001102416.2	ENSP00000265023	8
RP11-573D15.8	ENST00000627919	?	?	9
RP11-573D15.8	ENST00000629126	?	?	9
RP11-573D15.8	ENST00000628190	?	?	9
RP11-573D15.8	ENST00000629106	?	?	9
RP11-573D15.8	ENST00000625386	?	?	9
RP11-573D15.8	ENST00000626633	?	?	9
RP11-573D15.8	ENST00000627015	?	?	9
RP11-573D15.8	ENST00000626306	?	?	9
RP11-573D15.8	ENST00000626151	?	?	9
RP11-573D15.8	ENST00000627268	?	?	9
RP11-573D15.8	ENST00000599314	?	?	9
RP11-573D15.8	ENST00000630178	?	?	9
RP11-573D15.8	ENST00000626845	?	?	9
RP11-573D15.8	ENST00000628601	?	?	9
RP11-573D15.8	ENST00000630331	?	?	9
RP11-573D15.8	ENST00000625741	?	?	9
RP11-573D15.8	ENST00000628858	?	?	9
RP11-573D15.8	ENST00000627469	?	?	9
RP11-573D15.8	ENST00000627830	?	?	9
RP11-573D15.8	ENST00000627551	?	?	9

RP11-573D15.8	ENST00000354642	?	?	9
RP11-573D15.8	ENST00000628020	?	?	9
RP11-573D15.8	ENST00000630864	?	?	9
RP11-573D15.8	ENST00000625710	?	?	9
RP11-573D15.8	ENST00000628505	?	?	9
RP11-573D15.8	ENST00000629451	?	?	9
RP11-573D15.8	ENST00000625303	?	?	9
RP11-573D15.8	ENST00000629734	?	?	8
RP11-573D15.8	ENST00000625839	?	?	9
RP11-573D15.8	ENST00000628253	?	?	9
RP11-573D15.8	ENST00000628831	?	?	8
RP11-573D15.8	ENST00000628728	?	?	9
RP11-573D15.8	ENST00000631299	?	?	4

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-573D15.8	ENST00000629126	?	2
RP11-573D15.8	ENST00000628020	?	2
RP11-573D15.8	ENST00000629734	?	1