SNiPAcard

Block annotations

Block info

genomic range	chr3:186,422,367-186,450,863
block size	28,497 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.131 [-1.563 – 0.542]	**gene(s) hit or close-by**	KNG1, RP11-573D15.8, RP11-795A2.3
phastCons	μ = 0.003 [0 – 0.006]	**eQTL gene(s)**	–
GERP++	μ = -2.976 [-9.97 – 0.235]	**potentially regulated gene(s)**	–
CADD score	μ = 2.767 [1.095 – 4.152]	**disease gene(s)**	KNG1

Trait annotations

Disease gene annotation

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

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	M/I	atG/atT	?	?	1
KNG1	missense variant	ENST00000287611	NM_000893.3	ENSP00000287611	M/I	atG/atT	?	?	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	3166	ENST00000628020	?	?	2
RP11-573D15.8	upstream gene variant	1472	ENST00000625877	?	?	1
RP11-573D15.8	downstream gene variant	1415	ENST00000628858	?	?	1
RP11-573D15.8	downstream gene variant	3433	ENST00000629734	?	?	1
RP11-573D15.8	downstream gene variant	1304	ENST00000627015	?	?	1
RP11-573D15.8	downstream gene variant	1556	ENST00000626306	?	?	2
RP11-573D15.8	downstream gene variant	1092	ENST00000625710	?	?	1
RP11-573D15.8	downstream gene variant	2775	ENST00000629126	?	?	2
RP11-573D15.8	downstream gene variant	1088	ENST00000627268	?	?	1
RP11-573D15.8	downstream gene variant	3342	ENST00000628831	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
KNG1	ENST00000287611	NM_000893.3	ENSP00000287611	2
KNG1	ENST00000265023	NM_001102416.2	ENSP00000265023	2
KNG1	ENST00000447445	NM_001166451.1	ENSP00000396025	3
RP11-573D15.8	ENST00000626151	?	?	3
RP11-573D15.8	ENST00000628190	?	?	4
RP11-573D15.8	ENST00000628253	?	?	4
RP11-573D15.8	ENST00000627919	?	?	4
RP11-573D15.8	ENST00000625386	?	?	4
RP11-573D15.8	ENST00000626649	?	?	1
RP11-573D15.8	ENST00000629451	?	?	4
RP11-573D15.8	ENST00000626633	?	?	4
RP11-573D15.8	ENST00000628601	?	?	3
RP11-573D15.8	ENST00000630178	?	?	4
RP11-573D15.8	ENST00000627469	?	?	4
RP11-573D15.8	ENST00000627268	?	?	2
RP11-573D15.8	ENST00000627830	?	?	4
RP11-573D15.8	ENST00000630331	?	?	4
RP11-573D15.8	ENST00000630864	?	?	4
RP11-573D15.8	ENST00000625839	?	?	4
RP11-573D15.8	ENST00000628505	?	?	4
RP11-573D15.8	ENST00000628728	?	?	4
RP11-573D15.8	ENST00000629106	?	?	4
RP11-573D15.8	ENST00000354642	?	?	3
RP11-573D15.8	ENST00000625741	?	?	4
RP11-573D15.8	ENST00000599314	?	?	3
RP11-573D15.8	ENST00000628858	?	?	2
RP11-573D15.8	ENST00000627551	?	?	3
RP11-573D15.8	ENST00000630726	?	?	1
RP11-573D15.8	ENST00000625303	?	?	4
RP11-573D15.8	ENST00000627015	?	?	2
RP11-573D15.8	ENST00000626845	?	?	4
RP11-573D15.8	ENST00000625710	?	?	2
RP11-795A2.3	ENST00000413467	?	?	1