SNiPAcard

Block annotations

Block info

genomic range	chr19:51,484,553-51,487,175
block size	2,623 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.287 [-4.422 – 0.646]	**gene(s) hit or close-by**	CTB-147C22.9, KLK7
phastCons	μ = 0.001 [0 – 0.002]	**eQTL gene(s)**	KLK7
GERP++	μ = -0.817 [-6.68 – 2.29]	**potentially regulated gene(s)**	KLK7
CADD score	μ = 4.290 [1.036 – 8.337]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
KLK7	?	ENSG00000169035	sun exposed skin	3.70×10^{-10} (p-value)	GTEx Portal V6	6
KLK7	?	ENSG00000169035	unexposed skin	4.61×10^{-8} (p-value)	GTEx Portal V6	6

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000341941	1	ENST00000304045, ENST00000391807	KLK7

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
KLK7	downstream gene variant	157	ENST00000595638	?	?	4
KLK7	upstream gene variant	5	ENST00000593904	?	ENSP00000470084	1
KLK7	upstream gene variant	104	ENST00000391807	NM_139277.2	ENSP00000375683	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
KLK7	ENST00000595820	NM_005046.3	ENSP00000470538	L	Tta/Cta	1
KLK7	ENST00000391807	NM_139277.2	ENSP00000375683	L	Tta/Cta	1
KLK7	ENST00000304045	NM_001243126.1	ENSP00000304791	L	Tta/Cta	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTB-147C22.9	ENST00000594512	?	?	6
KLK7	ENST00000595820	NM_005046.3	ENSP00000470538	6

KLK7	ENST00000391807	NM_139277.2	ENSP00000375683	5
KLK7	ENST00000593904	?	ENSP00000470084	5
KLK7	ENST00000304045	NM_001243126.1	ENSP00000304791	6
KLK7	ENST00000597707	NM_001207053.1	ENSP00000469950	6

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
KLK7	ENST00000595820	NM_005046.3	ENSP00000470538	2
KLK7	ENST00000593904	?	ENSP00000470084	1
KLK7	ENST00000304045	NM_001243126.1	ENSP00000304791	2
KLK7	ENST00000597707	NM_001207053.1	ENSP00000469950	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
KLK7	ENST00000595638	?	2