

SNiPacard

Block annotations

Block info

genomic range	chr19:51,587,288-51,587,893 <i>e!</i>
block size	606 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	$\mu = -2.387$ [-3.363 – -1.015]	gene(s) hit or close-by	KLK14 <i>e!</i>
phastCons	$\mu = 0.000$ [0 – 0.001]	eQTL gene(s)	–
GERP++	$\mu = -0.833$ [-2.23 – 1.69]	potentially regulated gene(s)	–
CADD score	$\mu = 4.350$ [0.518 – 7.659]	disease gene(s)	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000348484 <i>e!</i> (open chromatin region)	1	embryonic stem cell (H1ESC) A549 blood (K562)	DNase1 H3K27me3 H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
KLK14 <i>e!</i>	upstream gene variant	141	ENST00000156499 <i>e!</i>	?	ENSP00000156499 <i>e!</i>	2
KLK14 <i>e!</i>	upstream gene variant	141	ENST00000391802 <i>e!</i>	NM_022046.4	ENSP00000375678 <i>e!</i>	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
KLK14 <i>e!</i>	ENST00000156499 <i>e!</i>	?	ENSP00000156499 <i>e!</i>	1
KLK14 <i>e!</i>	ENST00000391802 <i>e!</i>	NM_022046.4	ENSP00000375678 <i>e!</i>	1

