Locus 385 (rs3745542)

SNiPAcard

Block annotations

Block info

genomic range	chr19:51,587,288-51,587,893
block size	606 bp
variant count	3 variants

Basic features

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

Putative effect on regulation

Regulatory feature cluster

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

Variation proximal to gene

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

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
KLK14	ENST00000156499	?	ENSP00000156499	1
KLK14	ENST00000391802	NM_022046.4	ENSP00000375678	1