Locus 420 (rs2493151)

SNiPAcard

Block annotations

Block info

genomic range	chr1:230,873,488-230,879,511
block size	6,024 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.444 [-2.133 – -0.493]	**gene(s) hit or close-by**	CAPN9, RP11-99J16__A.2
phastCons	μ = 0.000 [0 – 0.001]	**eQTL gene(s)**	–
GERP++	μ = -1.241 [-2.88 – 0.756]	**potentially regulated gene(s)**	–
CADD score	μ = 1.572 [0.279 – 3.222]	**disease gene(s)**	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000554770 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3
		embryonic stem cell (H1ESC)	Rad21
		liver (HepG2)	Rad21
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CAPN9	upstream gene variant	3619	ENST00000271971	NM_006615.2	ENSP00000271971	2
CAPN9	upstream gene variant	3619	ENST00000366666	?	ENSP00000355626	2
CAPN9	upstream gene variant	3650	ENST00000354537	NM_016452.1	ENSP00000346538	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
RP11-99J16__A.2	ENST00000412344	?	?	3