Locus 289 (rs6665631)

SNiPAcard

Block annotations

Block info

genomic range	chr1:8,971,030-8,980,813
block size	9,784 bp
variant count	13 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.162 [-1.676 – 0.529]	**gene(s) hit or close-by**	RP3-477M7.5, RP3-477M7.6
phastCons	μ = 0.022 [0 – 0.112]	**eQTL gene(s)**	–
GERP++	μ = -0.042 [-0.804 – 0.774]	**potentially regulated gene(s)**	–
CADD score	μ = 2.705 [0.013 – 8.856]	**disease gene(s)**	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000530162 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K36me3, DNase1, H3K27ac
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	Max, H3K27me3
		breast (HMEC)	H3K27ac, H3K4me1, H3K4me2
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	USF1, H3K4me1
		nervous (NH-A)	DNase1
ENSR00001576724 (enhancer)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	CTCF
		blood (K562)	CTCF, Rad21, H3K27me3
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, FOXA1, CTCF
		blood (GM12878)	Yy1, Rad21, CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	DNase1, CTCF
		A549	CTCF
ENSR00000074592 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	CTCF
		blood (K562)	Rad21, H3K27me3, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, FOXA1, CTCF
		blood (GM12878)	Yy1, Rad21, CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF

cervix (HeLa-S3)	CTCF
monocytes (Monocytes-CD14+)	CTCF
endothelium (HUVEC)	DNase1, CTCF
A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP3-477M7.5	upstream gene variant	3227	ENST00000424862	?	?	2
RP3-477M7.6	downstream gene variant	1067	ENST00000431968	?	?	6