Locus 283 (rs17200067)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,370,722-31,376,989
block size	6,268 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.077 [-0.057 – 0.212]	**gene(s) hit or close-by**	HCP5, MICA, Y_RNA
phastCons	μ = 0.117 [0.001 – 0.232]	**eQTL gene(s)**	–
GERP++	μ = -0.392 [-1.04 – 0.257]	**potentially regulated gene(s)**	–
CADD score	μ = 5.005 [4.24 – 5.771]	**disease gene(s)**	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000487837 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, CTCF, PolII, TAF1, H3K4me2, SP1, USF1, H3K4me3, DNase1
		HSMMtube	DNase1, H3K36me3, H2AZ, H3K27ac, H3K4me2, H3K9ac
		blood (K562)	H3K27ac, Max, E2F6, H3K9ac, USF1, H3K79me2, H2AZ, Cfos, PolII, H3K4me2, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	USF1, H3K4me1, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, DNase1, TAF1, H3K79me2, PolII
		blood (GM12878)	PolII, H2AZ, DNase1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		lung (IMR90)	H3K79me2, H3K4me2, H3K4me3, H3K9ac, DNase1
		nervous (NH-A)	H3K9ac, H4K20me1, H3K4me2, H3K4me3, H3K27ac, H3K36me3, DNase1
		skin (NHEK)	H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K4me1, H3K27ac, H4K20me1, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, H3K36me3
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, PolII, DNase1
		monocytes (Monocytes-CD14+)	H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3, H3K4me2, H3K4me1
		endothelium (HUVEC)	H3K36me3, H3K4me2, H3K4me3, PolII, DNase1
		A549	H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MICA	upstream gene variant	634	ENST00000449934	NM_001177519.2	ENSP00000413079	1
MICA	upstream gene variant	727	ENST00000421350	?	ENSP00000402410	1
Y_RNA	upstream gene variant	695	ENST00000362462	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HCP5	ENST00000414046	?	?	2
MICA	ENST00000449934	NM_001177519.2	ENSP00000413079	1
MICA	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	2

MICA	ENST00000421350	?	ENSP00000402410	1