Locus 340 (rs2147190)

SNiPAcard

Block annotations

Block info

genomic range	chr13:92,031,820-92,058,888
block size	27,069 bp
variant count	13 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.417 [-2.725 – 1.103]	**gene(s) hit or close-by**	AL138714.1, GPC5
phastCons	μ = 0.010 [0 – 0.12]	**eQTL gene(s)**	GPC5
GERP++	μ = -1.921 [-8.41 – 1.94]	**potentially regulated gene(s)**	–
CADD score	μ = 3.144 [0.08 – 6.815]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
GPC5	?	ENSG00000179399	caudate basal ganglia	2.25×10-6 (p-value)	GTEx Portal V6	4
GPC5	?	ENSG00000179399	lung	4.24×10-6 (p-value)	GTEx Portal V6	3

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001511261 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	CTCF
		blood (K562)	CTCF, Rad21
		muscle (HSMM)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	CTCF
		liver (HepG2)	CTCF
		blood (GM12878)	CTCF, Rad21
		lung (IMR90)	CTCF
ENSR00001511262 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3, Nrsf, TAF1, H3K4me2, H3K4me3, H3K27me3, DNase1
		HSMMtube	H3K27me3, DNase1
		blood (K562)	H3K4me3, DNase1, H3K27me3, H3K4me2, Nrsf, H2AZ, H3K4me1, H3K9ac, Max, Egr1
		skin (NHDF-AD)	H3K27me3
		muscle (HSMM)	H3K27me3
		liver (HepG2)	H3K4me3, H3K4me2, H3K27me3, DNase1
		blood (GM12878)	H3K4me2, H3K4me3, H2AZ, DNase1, PolII, Nrsf
		lung (IMR90)	H3K27me3
		nervous (NH-A)	H3K27me3, H3K4me2
		skin (NHEK)	H3K27me3, H3K4me3, H3K4me2, DNase1
		NHLF	H3K27me3
		Osteobl	H3K27me3, H3K4me2
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K4me2, H3K4me3
		cervix (HeLa-S3)	DNase1, H3K27ac, TAF1, PolII, H3K4me3, H3K79me2, H3K4me2, Nrsf, H3K9ac
		monocytes (Monocytes-CD14+)	H3K4me3, H3K27me3, H3K4me2, H3K4me1
		endothelium (HUVEC)	H3K36me3, H3K27me3, DNase1
		A549	H3K4me3, H3K4me2, H3K27me3

ENSR00001511263 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	DNase1, H3K27me3
		blood (K562)	DNase1, CTCF, Rad21
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		Osteobl	CTCF
		blood (DND-41)	H3K27me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	H3K79me2, CTCF
		endothelium (HUVEC)	CTCF
		A549	H3K27me3, CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL138714.1	downstream gene variant, upstream gene variant	29	ENST00000408499	?	?	5
GPC5	upstream gene variant	526	ENST00000377067	NM_004466.4	ENSP00000366267	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GPC5	ENST00000377067	NM_004466.4	ENSP00000366267	3