SNiPAcard

Block annotations

Block info

genomic range	chr4:123,748,616-123,757,748
block size	9,133 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.001 [-0.06 – 0.059]	**gene(s) hit or close-by**	AC021205.1, FGF2, RP11-170N16.2
phastCons	μ = 0.000 [0 – 0]	**eQTL gene(s)**	CETN4P, FGF2
GERP++	μ = 1.940 [1.7 – 2.18]	**potentially regulated gene(s)**	–
CADD score	μ = 8.498 [5.337 – 11.66]	**disease gene(s)**	–

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CR109896	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CETN4P	?	ENSG00000224786	skeletal muscle	8.37×10^{-11} (p-value)	GTEx Portal V6	2
FGF2	?	ENSG00000138685	testis	1.41×10^{-6} (p-value)	GTEx Portal V6	2
FGF2	?	ENSG00000138685	tibial artery	2.49×10^{-5} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001248693 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3, H3K27me3, CTCF, PolII, Rad21, Jund, TAF7, Sin3Ak20, H3K27ac, Yy1, HDAC2, Tcf12, TAF1, H3K4me2, SP1, H4K5ac, Srf, USF1, H3K9ac, H3K4me3
		HSMMtube	H3K9ac, H3K4me2
		blood (K562)	Max, H3K9ac, USF1, H3K79me2, H2AZ, H3K4me2, DNase1, H3K4me3, H3K27ac, Egr1, PolII
		skin (NHDF-AD)	H3K36me3, CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2, H3K9ac, H3K36me3
		liver (HepG2)	H3K4me3, H3K4me2, H3K27me3, DNase1
		blood (GM12878)	EBF1, PolII, Srf, MEF2A, H2AZ, DNase1, H3K4me3, NFKB, H3K4me2
		lung (IMR90)	CTCF, H3K56ac, H3K79me2, H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	H3K4me2, H3K4me3, H3K27me3
		NHLF	H3K4me3, DNase1, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, Cjun, H3K9ac, Jund, H3K4me2, H3K27ac, TAF1, PolII, H3K4me3, H3K79me2, H3K36me3
		monocytes (Monocytes-CD14+)	H3K27me3, H3K4me3

		endothelium (HUVEC)	H3K36me3, DNase1, PolII, H3K4me2, H3K4me3
		A549	H3K4me3, H3K4me2, H3K9ac, H3K27ac, H3K36me3
ENSR00001434812 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3
		blood (K562)	H3K36me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K36me3
		cervix (HeLa-S3)	Nrsf
		monocytes (Monocytes-CD14+)	H3K27me3
		lung (IMR90)	H3K79me2, H3K36me3
		A549	H3K4me2, H3K36me3, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC021205.1	downstream gene variant	371	ENST00000517260	?	?	1
RP11-170N16.2	upstream gene variant	3750	ENST00000468797	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FGF2	ENST00000608478	?	ENSP00000477134	2
FGF2	ENST00000614010	NM_002006.4	ENSP00000478620	2
FGF2	ENST00000264498	?	ENSP00000264498	2