SNiPAcard

Block annotations

Block info

genomic range	chr19:51,612,262-51,618,167
block size	5,906 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.815 [-3.587 – 0.21]	**gene(s) hit or close-by**	CTU1
phastCons	μ = 0.054 [0.001 – 0.184]	**eQTL gene(s)**	SIGLEC9
GERP++	μ = -0.193 [-1.14 – 0.362]	**potentially regulated gene(s)**	SHANK1
CADD score	μ = 3.017 [0.628 – 7.501]	**disease gene(s)**	SHANK1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
SHANK1	AUTISM	DECIPHER	MIM:209850 OMIM

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SIGLEC9	?	ENSG00000129450	cerebellar hemisphere	6.90×10-7 (p-value)	GTEx Portal V6	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000232477	1	ENCP00000024630	SHANK1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001419940 (promoter)	1	embryonic stem cell (H1ESC)	Gabp, Sin3Ak20, SIX5, H3K4me2, H3K4me3, DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	Max, H3K27ac, H3K9ac, H3K4me3, DNase1, H3K36me3, CTCF, H3K4me2, PolII, Cmyc, CTCFL, ELF1, H2AZ, ETS1, Gabp
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K4me3, H2AZ
		liver (HepG2)	DNase1, H3K4me3, Cmyc, H3K27ac, H3K9ac, H3K4me2, ELF1, PolII, Gabp
		blood (GM12878)	ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, EBF1, SIX5, PolII, H2AZ, ETS1, DNase1, Cmyc, Gabp
		lung (IMR90)	H3K4me2, H3K4me3, DNase1
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K4me3, DNase1, H3K4me2, H3K9ac
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K4me3, H3K27ac, H2AZ, H3K4me2
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, PolII, Max, H3K4me3, H3K79me2, Gabp, Cmyc

	monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
	endothelium (HUVEC)	Max, H3K4me3, PolII, Cmyc, DNase1
	A549	H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00000217695 2 (CTCF binding site)	embryonic stem cell (H1ESC)	Rad21, CTCF
	HSMMtube	H3K27me3
	blood (K562)	CTCF, H3K27me3
	blood (DND-41)	H3K27me3
	skin (NHDF-AD)	CTCF
	breast (HMEC)	CTCF
	cervix (HeLa-S3)	CTCF
	monocytes (Monocytes-CD14+)	H3K4me1
	endothelium (HUVEC)	H3K27me3
	lung (IMR90)	H3K27me3
	A549	H3K27me3
	skin (NHEK)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTU1	upstream gene variant	635	ENST00000421832	NM_145232.3	ENSP00000390011	6