Locus 197 (rs17741049)

SNiPAcard

Block annotations

Block info

genomic range	chr8:86,089,292-86,292,870
block size	203,579 bp
variant count	36 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.154 [-3.447 – 1.323]	gene(s) hit or close-by	C8orf59, CA1, CA13, CA3, E2F5, RP11-219B4.3, RP11-219B4.5, RP11-219B4.6, RP11-219B4.7
phastCons	μ = 0.098 [0 – 1]	eQTL gene(s)	LRRCC1
GERP++	μ = -0.634 [-6.53 – 3.79]	potentially regulated gene(s)	CA1, CA3, E2F5
CADD score	μ = 3.878 [0.014 – 14.87]	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
E2F5	missense variant	ENST00000418930	NM_001083588.1	ENSP00000414312	A/G	gCg/gGg	?	?	1
E2F5	missense variant	ENST00000416274	NM_001951.3	ENSP00000398124	A/G	gCg/gGg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LRRCC1	?	ENSG00000133739	esophagus mucosa	5.45×10-6 (p-value)	GTEx Portal V6	33

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
EGR1	MA0341.1	4	yes	0.000	1
EGR1	MA0366.1	4	yes	0.000	1

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000056669		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000521373	1	ENCP00000056670	CA1
ENCE00000521314	1	ENCP00000056657	E2F5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001395450 (promoter)	3	embryonic stem cell (H1ESC)	DNase1, H3K36me3, PolII, Rad21, TAF7, H3K27ac, TAF1, H3K4me2, H3K9ac, H3K4me3
		HSMMtube	H3K4me2, H2AZ, DNase1
		blood (K562)	Egr1, Max, H3K9ac, Cmyc, H2AZ, PolII, H3K4me2, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me2, H3K4me3, DNase1
		muscle (HSMM)	H2AZ, H3K4me3
		liver (HepG2)	Gabp, PolII, H3K79me2, TAF1, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, DNase1
		lung (IMR90)	H3K4me2, H3K4me3
		blood (GM12878)	H2AZ, Tcf12, Cmyc, Egr1, Yy1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, PolII, DNase1
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	CTCF, H3K4me2, H3K4me3, DNase1
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H2AZ, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K4me3
		breast (HMEC)	H3K4me2, H3K4me3, H3K27ac
		cervix (HeLa-S3)	DNase1, H3K4me3, PolII, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, DNase1, PolII, H3K4me2, H3K4me3, Max
		A549	H3K36me3, H3K27ac, DNase1, H3K4me3, H3K4me2, H3K9ac
ENSR00001442694 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	CTCF, Rad21
		lung (IMR90)	CTCF
		skin (NHEK)	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	H3K27me3, CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF, H3K27me3
ENSR00001442695 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, H3K27me3
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	CTCF, Rad21
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF, H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C8orf59	downstream gene variant	1417	ENST00000612977	NM_001099671.1, NM_001099670.1	ENSP00000484101	1
C8orf59	downstream gene variant	1432	ENST00000545322	?	ENSP00000444092	1
C8orf59	downstream gene variant	1433	ENST00000611854	?	ENSP00000484811	1

C8orf59	downstream gene variant	1432	ENST00000612809	NM_001099672.1	ENSP00000481745	1
C8orf59	downstream gene variant	1945	ENST00000523281	?	ENSP00000428722	1
C8orf59	downstream gene variant	3743	ENST00000523245	?	?	1
C8orf59	downstream gene variant	1486	ENST00000615697	?	ENSP00000482944	1
C8orf59	downstream gene variant	1432	ENST00000615071	?	?	1
C8orf59	downstream gene variant	1007	ENST00000614462	NM_001293320.1	ENSP00000480167	1
C8orf59	downstream gene variant	1006	ENST00000619594	NM_001099673.1	ENSP00000484492	1
C8orf59	downstream gene variant	4090	ENST00000321777	?	ENSP00000319020	1
CA1	upstream gene variant	2528	ENST00000523022	NM_001738.3, NM_001128830.2, NM_001128831.2, NM_001128829.2	ENSP00000429798	1
CA1	upstream gene variant	2528	ENST00000523858	?	ENSP00000430975	1
CA1	upstream gene variant	2528	ENST00000522814	?	ENSP00000430737	1
CA1	upstream gene variant	2528	ENST00000520990	?	?	1
CA1	upstream gene variant	2528	ENST00000518341	?	?	1
CA1	upstream gene variant	2528	ENST00000517590	?	ENSP00000429843	1
CA1	upstream gene variant	2528	ENST00000519991	?	ENSP00000430543	1
CA1	upstream gene variant	2533	ENST00000521846	?	ENSP00000430471	1
CA1	upstream gene variant	2528	ENST00000522662	?	ENSP00000430372	1
CA1	upstream gene variant	1627	ENST00000626824	?	ENSP00000486171	1
CA1	upstream gene variant	2528	ENST00000524324	?	ENSP00000428923	1
CA1	upstream gene variant	2528	ENST00000519129	?	ENSP00000429688	1
CA1	upstream gene variant	2528	ENST00000520663	?	ENSP00000430571	1
CA1	upstream gene variant	2528	ENST00000520093	?	?	1
CA1	upstream gene variant	2528	ENST00000522579	?	ENSP00000427852	1
CA1	upstream gene variant	1627	ENST00000523953	?	ENSP00000430656	1
CA1	upstream gene variant	2528	ENST00000517429	?	ENSP00000430710	1
CA13	downstream gene variant	3683	ENST00000517298	?	?	1
CA13	upstream gene variant, downstream gene variant	740	ENST00000518392	?	?	6
CA13	upstream gene variant, downstream gene variant	825	ENST00000321764	NM_198584.2	ENSP00000318912	3
CA13	downstream gene variant	1119	ENST00000522631	?	?	2
CA3	upstream gene variant	1127	ENST00000520921	?	ENSP00000429760	2
E2F5	upstream gene variant	978	ENST00000521429	?	ENSP00000428606	1
E2F5	upstream gene variant	978	ENST00000517476	NM_001083589.1	ENSP00000429120	1
E2F5	upstream gene variant	3406	ENST00000521234	?	?	1
E2F5	upstream gene variant	330	ENST00000416274	NM_001951.3	ENSP00000398124	1
E2F5	upstream gene variant	168	ENST00000418930	NM_001083588.1	ENSP00000414312	1
E2F5	upstream gene variant	139	ENST00000256117	?	ENSP00000256117	2
RP11-219B4.3	downstream gene variant, upstream gene variant	49	ENST00000520129	?	?	3

RP11-219B4.5	upstream gene variant	2439	ENST00000546501 ?	?	2
RP11-219B4.5	upstream gene variant	1532	ENST00000549291 ?	?	2
RP11-219B4.6	downstream gene variant	2348	ENST00000551479 ?	?	1
RP11-219B4.7	upstream gene variant	411	ENST00000566000 ?	?	3
RP11-219B4.7	upstream gene variant	16	ENST00000562577 ?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CA1	ENST00000523953	?	ENSP00000430656	3
CA1	ENST00000522579	?	ENSP00000427852	3
CA1	ENST00000519991	?	ENSP00000430543	3
CA1	ENST00000521846	?	ENSP00000430471	3
CA1	ENST00000524324	?	ENSP00000428923	3
CA1	ENST00000523022	NM_001738.3, NM_001128830.2, NM_001128831.2, NM_001128829.2	ENSP00000429798	3
CA1	ENST00000523858	?	ENSP00000430975	3
CA1	ENST00000522814	?	ENSP00000430737	3
CA1	ENST00000519129	?	ENSP00000429688	3
CA1	ENST00000518341	?	?	3
CA1	ENST00000520093	?	?	3
CA1	ENST00000520990	?	?	3
CA1	ENST00000626824	?	ENSP00000486171	3
CA1	ENST00000517590	?	ENSP00000429843	3
CA1	ENST00000520663	?	ENSP00000430571	3
CA1	ENST00000517429	?	ENSP00000430710	3
CA1	ENST00000522662	?	ENSP00000430372	3
CA13	ENST00000517831	?	?	9
CA13	ENST00000517298	?	?	13
CA13	ENST00000522631	?	?	4
CA13	ENST00000321764	NM_198584.2	ENSP00000318912	10
CA3	ENST00000520921	?	ENSP00000429760	1
E2F5	ENST00000521429	?	ENSP00000428606	4
E2F5	ENST00000416274	NM_001951.3	ENSP00000398124	7
E2F5	ENST00000256117	?	ENSP00000256117	7
E2F5	ENST00000518234	?	ENSP00000429669	1
E2F5	ENST00000517476	NM_001083589.1	ENSP00000429120	4
E2F5	ENST00000520225	?	ENSP00000430461	1
E2F5	ENST00000418930	NM_001083588.1	ENSP00000414312	7
RP11-219B4.5	ENST00000546501	?	?	4
RP11-219B4.5	ENST00000549291	?	?	7

RP11-219B4.6	ENST00000551479	?	?	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CA13	ENST00000321764	NM_198584.2	ENSP00000318912	1
E2F5	ENST00000256117	?	ENSP00000256117	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-219B4.3	ENST00000520129	?	1
RP11-219B4.7	ENST00000566000	?	1