Locus 348 (rs3134906)

SNiPAcard

Block annotations

Block info

genomic range	chr7:142,503,071-142,506,685
block size	3,615 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.812 [-1.269 – -0.306]	gene(s) hit or close-by	TRBV30
phastCons	μ = 0.000 [0 – 0.001]	eQTL gene(s)	EPHB6, TRBV30
GERP++	μ = -2.489 [-5.14 – -0.256]	potentially regulated gene(s)	TRBC2, TRBJ2-1, TRBJ2-2, TRBJ2-2P, TRBJ2-4, TRBJ2-5, TRBJ2-6, TRBJ2-7, TRBV10-2, TRBV22-1, TRBV24-1, TRBV25-1, TRBV27, TRBV28, TRBV29-1, TRBV3-1, TRBV4-1, TRBV4-2, TRBV5-1, TRBV5-3, TRBV7-1, TRBV7-3, TRBV7-6, TRBV7-7, TRBV8-1, TRBVA, TRPV5, TRPV5
CADD score	μ = 2.120 [0.315 – 4.347]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
EPHB6	ENST00000486511	ILMN_1802646	adipocyte	2.99×10-6 (p-value)	MuTHER consortium	2
EPHB6	ENST00000616380					
EPHB6	ENST00000611578					
EPHB6	ENST00000411471					
EPHB6	ENST00000617632					
EPHB6	ENST00000614832					
EPHB6	ENST00000476059					
EPHB6	ENST00000619012					
EPHB6	ENST00000471581					
EPHB6	ENST00000422643					
?	?	ILMN_1823172	blood	1.07×10-6 (p-value)	Westra et al.	3
TRBV30	?	ENSG00000237254	blood	2.45×10-7 (p-value)	GTEx Portal V6	5
?	?	ILMN_1822307	b-cell	2.90×10-5 (p-value)	Fairfax et al.	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000500137	1	ENCP00000054229	TRBVA
		ENCP00000054196	TRBV4-2
		ENCP00000054234	TRBV28
		ENCP00000054225	TRBV22-1
		ENCP00000054193	TRBV5-1
		ENCP00000054242	TRBC2
		ENCP00000054233	TRBV27
		ENCP00000054191	TRBV3-1

		ENCP00000054240	TRBJ2-1 TRBJ2-2 TRBJ2-2P
		ENCP00000054192	TRBV4-1
		ENCP00000054203	TRBV7-6
		ENCP00000054228	TRBV25-1
		ENCP00000054221	TRBV8-1
		ENCP00000054227	TRBV24-1
		ENCP00000054212	TRBV10-2
		ENCP00000054247	TRPV5 TRPV5
ENCE00000500138	1	ENCP00000054191	TRBV3-1
		ENCP00000054229	TRBVA
		ENCP00000054196	TRBV4-2
		ENCP00000054234	TRBV28
		ENCP00000054240	TRBJ2-1 TRBJ2-2 TRBJ2-2P
		ENCP00000054192	TRBV4-1
		ENCP00000054203	TRBV7-6
		ENCP00000054225	TRBV22-1
		ENCP00000054228	TRBV25-1
		ENCP00000054193	TRBV5-1
		ENCP00000054221	TRBV8-1
		ENCP00000054242	TRBC2
		ENCP00000054233	TRBV27
		ENCP00000054247	TRPV5 TRPV5
ENCE00000500143	1	ENCP00000054229	TRBVA
		ENCP00000054196	TRBV4-2
		ENCP00000054234	TRBV28
		ENCP00000054225	TRBV22-1
		ENCP00000054193	TRBV5-1
		ENCP00000054236	TRBV29-1
		ENCP00000054242	TRBC2
		ENCP00000054233	TRBV27
		ENCP00000054191	TRBV3-1
		ENCP00000054240	TRBJ2-1 TRBJ2-2 TRBJ2-2P
		ENCP00000054192	TRBV4-1
		ENCP00000054203	TRBV7-6
		ENCP00000054228	TRBV25-1
		ENCP00000054221	TRBV8-1
		ENCP00000054212	TRBV10-2
		ENCP00000054227	TRBV24-1
		ENCP00000054247	TRPV5 TRPV5
ENCE00000500145	1	ENCP00000054229	TRBVA
		ENCP00000054234	TRBV28
		ENCP00000054225	TRBV22-1
		ENCP00000054195	TRBV7-1
		ENCP00000054193	TRBV5-1
		ENCP00000054236	TRBV29-1
		ENCP00000054241	TRBJ2-5 TRBJ2-4 TRBJ2-6

	TRBJ2-7
ENCP00000054242	TRBC2
ENCP00000054233	TRBV27
ENCP00000054191	TRBV3-1
ENCP00000054240	TRBJ2-1 TRBJ2-2 TRBJ2-2P
ENCP00000054192	TRBV4-1
ENCP00000054203	TRBV7-6
ENCP00000054228	TRBV25-1
ENCP00000054221	TRBV8-1
ENCP00000054219	TRBV7-3
ENCP00000054200	TRBV7-7
ENCP00000054217	TRBV5-3
ENCP00000054227	TRBV24-1
ENCP00000054212	TRBV10-2
ENCP00000054247	TRPV5 TRPV5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001718692 (enhancer)	2	endothelium (HUVEC)	PolII
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	DNase1
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K36me3
		A549	H3K27me3
ENSR00000635620 (promoter flanking region)	3	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3, DNase1, Egr1, FOSL1, Max, PU1, ETS1
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K36me3
		cervix (HeLa-S3)	DNase1, H3K27ac
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K36me3, Cmyc, PolII, H3K27ac, H3K4me1, DNase1, Cjun
		blood (GM12878)	H3K9ac, H3K4me2, PU1, PolII, BCLAF1, BATF, MEF2A, H3K4me1, ETS1, Tcf12, DNase1, Egr1, H3K4me3, BCL11A, Pax5, H3K27ac, ZEB1
		A549	CTCF, H3K27me3
		nervous (NH-A)	DNase1, H3K9ac, H3K27ac

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
TRBV30	downstream gene variant	3585	ENST00000417977	?	ENSP00000397118	1