Locus 449 (rs4239252)

SNiPAcard

Block annotations

Block info

genomic range	chr17:34,087,704-34,261,831
block size	174,128 bp
variant count	53 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.665 [-5.315 – 2.165]	**gene(s) hit or close-by**	7SK, AC015849.12, AC015849.13, AC015849.14, AC015849.16, AC015849.19, AC015849.2, C17orf50, CCL5, HEATR9, LRRC37A9P, LYZL6, MMP28, RDM1, RP1-161P9.5, TAF15
phastCons	μ = 0.071 [0 – 1]	**eQTL gene(s)**	CCL5
GERP++	μ = -0.087 [-6.19 – 4.15]	**potentially regulated gene(s)**	CCL23, CCL23, CCL5, CCL5, RP11-1094M14.1
CADD score	μ = 3.563 [0.191 – 22.3]	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
N-acetylglycine	5.76×10^{-5}	Metabolomics GWAS Server	24816252	2
pyridoxate	9.63×10^{-5}	Metabolomics GWAS Server	24816252	3

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)
C17orf50	missense variant	ENST00000604830	?	ENSP00000474618	D/E	gaC/gaG	?	unknown	1
C17orf50	missense variant	ENST00000603305	?	ENSP00000474048	R/G	Cgc/Ggc	?	unknown	1
C17orf50	missense variant	ENST00000605587	NM_145272.3	ENSP00000475146	D/E	gaC/gaG	tolerated	benign	1
LYZL6	missense variant	ENST00000618542	NM_001199951.1	ENSP00000482955	S/F	tCc/tTc	?	?	1
LYZL6	missense variant	ENST00000615905	NM_020426.2	ENSP00000483897	S/F	tCc/tTc	?	?	1
RDM1	missense variant	ENST00000615378	NM_001163125.1	ENSP00000480131	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000612980	NM_001163120.1	ENSP00000483387	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000619828	NM_001163122.1	ENSP00000483933	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000619876	?	ENSP00000483890	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000615288	?	ENSP00000477869	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000619368	?	ENSP00000478131	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000615024	?	ENSP00000481648	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000617591	NM_001163124.1	ENSP00000479622	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000620284	NM_145654.3	ENSP00000483549	W/C	tgG/tgT	?	?	1

RDM1	missense variant	ENST00000616596	NM_001163121.1	ENSP00000478915	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000619193	?	ENSP00000482981	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000613308	NM_001034836.1	ENSP00000482288	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000618511	?	ENSP00000477995	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000619262	NM_001163130.1	ENSP00000479310	W/C	tgG/tgT	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
?	?	ENSG00000187456	blood	1.75×10^{-5} (q-value)	SeeQTL DB (HapMap)	12
?	?	ENSG00000161570	blood	4.48×10^{-5} (q-value)	SeeQTL DB (HapMap)	7
CCL5	ENST00000603197	ILMN_2098126	blood	1.34×10^{-11} (p-value)	MuTHER consortium	22
CCL5	ENST00000605140					
CCL5	ENST00000603197	ILMN_1773352	blood	4.39×10^{-11} (p-value)	MuTHER consortium	22
CCL5	ENST00000605140					
CCL5	ENST00000605509					
?	?	241885_at	blood	1.00×10^{-9} (p-value)	Dixon et al.	7
CCL5	ENST00000603197	ILMN_2098126	blood	1.56×10^{-5} (p-value)	Westra et al.	12
CCL5	ENST00000605140					

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
?	?	ILMN_1761314	chr20	monocyte	1.82×10^{-13} (p-value)	Zeller et al.	3

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000022616		CCL5

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000204092	1	ENCP00000022627	CCL23 CCL23
ENCE00000203983	1	ENCP00000022610	RP11-1094M14.1
		ENCP00000022616	CCL5 CCL5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001344848 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3, Rad21, TAF7, CTCF, Yy1, H3K4me2, H3K4me3
		HSMMtube	CTCF, H3K27ac, H2AZ, DNase1
		blood (K562)	H3K4me3, DNase1, CTCF, H2AZ, Rad21, Max, H3K27me3
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	H2AZ, CTCF, DNase1
		liver (HepG2)	H3K27me3, DNase1, CTCF, H3K4me2, H3K4me3, Rad21
		blood (GM12878)	DNase1, H2AZ, Rad21, H3K4me3, H3K4me2, CTCF

		lung (IMR90)	H3K4me2, H3K27me3, CTCF
		nervous (NH-A)	DNase1, H3K27me3, H3K4me2, CTCF
		skin (NHEK)	CTCF, DNase1, H3K4me2
		NHLF	DNase1, CTCF
		Osteobl	H3K4me2, CTCF, H3K27me3
		blood (DND-41)	H3K4me1, H3K4me2, CTCF, H3K27ac, H3K9ac, H3K4me3
		breast (HMEC)	H3K4me2, CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	H3K27me3, H3K4me3, DNase1, CTCF, H3K4me2
		endothelium (HUVEC)	DNase1, Max, H3K4me2, CTCF
		A549	CTCF, DNase1, H3K4me2, H3K4me3
ENSR00001638253 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3, Rad21, TAF7, CTCF, Yy1, H3K4me2
		HSMMtube	CTCF, H2AZ, DNase1
		blood (K562)	H3K4me3, DNase1, CTCF, H2AZ, Rad21, Max
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	H2AZ, CTCF
		liver (HepG2)	H3K4me3, Rad21, H3K4me2, CTCF, DNase1
		blood (GM12878)	DNase1, H2AZ, Rad21, H3K4me3, H3K4me2, CTCF
		lung (IMR90)	H3K4me2, CTCF
		nervous (NH-A)	CTCF, H3K4me2, DNase1
		skin (NHEK)	DNase1, CTCF, H3K4me2
		NHLF	DNase1, CTCF
		Osteobl	H3K4me2, CTCF
		blood (DND-41)	H3K9ac, H3K4me3, H3K27ac, CTCF, H3K4me1, H3K4me2
		breast (HMEC)	H3K4me2, CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me2, H3K27me3, H3K4me3
		endothelium (HUVEC)	Max, H3K4me2, CTCF, DNase1
		A549	DNase1, H3K4me3, H3K4me2, CTCF
ENSR00001344864 (promoter flanking region)	1	NHLF	H3K27me3
		embryonic stem cell (H1ESC)	H3K27me3
		HSMMtube	DNase1
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	PolII, DNase1
		lung (IMR90)	H3K27me3
ENSR00001344879 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	DNase1
		HSMMtube	H3K27me3
		blood (GM12878)	MEF2A, DNase1, H3K4me3, H3K27ac, PolII
		blood (DND-41)	H3K27me3
		blood (K562)	H3K4me1, H2AZ, DNase1
ENSR00001638269 (CTCF binding site)	1	cervix (HeLa-S3)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		Osteobl	H3K4me2
		blood (DND-41)	H3K27me3
		blood (K562)	H2AZ

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
TAF15	1	ENST00000603067 ENST00000603393 ENST00000603427 ENST00000603777 ENST00000604360 ENST00000604841 ENST00000604879 ENST00000605197 ENST00000605649 ENST00000605844	hsa-miR-18a-5p

Variation proximal to gene

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
7SK	upstream gene variant, downstream gene variant	4434	ENST00000603504	?	?	2
AC015849.12	downstream gene variant, upstream gene variant	535	ENST00000605738	?	?	2
AC015849.13	upstream gene variant, downstream gene variant	1758	ENST00000603678	?	?	4
AC015849.14	downstream gene variant, upstream gene variant	447	ENST00000605454	?	?	3
AC015849.19	downstream gene variant, upstream gene variant	76	ENST00000603981	?	?	3
AC015849.2	upstream gene variant	438	ENST00000615709	?	?	2
AC015849.2	upstream gene variant	4565	ENST00000605548	?	?	1
C17orf50	upstream gene variant	212	ENST00000604830	?	ENSP00000474618	1
C17orf50	upstream gene variant	225	ENST00000605587	NM_145272.3	ENSP00000475146	1
C17orf50	upstream gene variant	224	ENST00000603305	?	ENSP00000474048	1
CCL5	upstream gene variant	268	ENST00000603197	NM_002985.2	ENSP00000474412	2
CCL5	upstream gene variant	485	ENST00000605509	NM_001278736.1	ENSP00000474141	2
CCL5	upstream gene variant	4331	ENST00000605140	?	ENSP00000475057	1
HEATR9	downstream gene variant, upstream gene variant	356	ENST00000604376	?	?	4
HEATR9	upstream gene variant	3798	ENST00000604400	?	?	2
HEATR9	upstream gene variant	3810	ENST00000603323	?	ENSP00000474391	2
HEATR9	upstream gene variant	3746	ENST00000605080	?	ENSP00000474893	2
HEATR9	upstream gene variant	3766	ENST00000603870	?	ENSP00000473760	2
HEATR9	upstream gene variant	3747	ENST00000603852	?	ENSP00000474899	2
HEATR9	downstream gene variant, upstream gene variant	2260	ENST00000603218	?	ENSP00000474337	3
HEATR9	upstream gene variant	3746	ENST00000604834	NM_152781.2	ENSP00000473941	2
LRRC37A9P	upstream gene variant	1228	ENST00000603850	?	?	8
LYZL6	downstream gene variant	1542	ENST00000615905	NM_020426.2	ENSP00000483897	7
LYZL6	downstream gene variant	1529	ENST00000618542	NM_001199951.1	ENSP00000482955	7
LYZL6	downstream gene variant	1862	ENST00000617692	?	ENSP00000481373	4
MMP28	downstream gene variant, upstream gene variant	1011	ENST00000605424	NM_024302.3	ENSP00000473853	2
MMP28	downstream gene variant, upstream gene variant	969	ENST00000615136	?	ENSP00000483299	2
MMP28	downstream gene variant, upstream gene variant	850	ENST00000612292	?	ENSP00000484562	2
MMP28	upstream gene variant	886	ENST00000612672	NM_001032278.1	ENSP00000483539	1
MMP28	upstream gene variant	793	ENST00000616383	?	?	1
MMP28	upstream gene variant	886	ENST00000619655	?	ENSP00000481051	1
MMP28	upstream gene variant	4410	ENST00000615317	?	ENSP00000482385	1
RDM1	upstream gene variant	721	ENST00000615024	?	ENSP00000481648	5
RDM1	downstream gene variant, upstream gene variant	704	ENST00000615378	NM_001163125.1	ENSP00000480131	12
RDM1	upstream gene variant	234	ENST00000619262	NM_001163130.1	ENSP00000479310	5
RDM1	upstream gene variant	276	ENST00000612980	NM_001163120.1	ENSP00000483387	5
RDM1	upstream gene variant	276	ENST00000616596	NM_001163121.1	ENSP00000478915	5
RDM1	upstream gene variant	721	ENST00000617591	NM_001163124.1	ENSP00000479622	5
RDM1	downstream gene variant, upstream gene variant	250	ENST00000619876	?	ENSP00000483890	12
RDM1	upstream gene variant	276	ENST00000615288	?	ENSP00000477869	5
RDM1	upstream gene variant	721	ENST00000619828	NM_001163122.1	ENSP00000483933	5

RDM1	upstream gene variant	721	ENST00000618511	?	ENSP00000477995	5
RDM1	upstream gene variant	276	ENST00000619193	?	ENSP00000482981	5
RDM1	upstream gene variant	230	ENST00000620284	NM_145654.3	ENSP00000483549	5
RDM1	upstream gene variant	276	ENST00000619368	?	ENSP00000478131	5
RDM1	downstream gene variant, upstream gene variant	263	ENST00000613308	NM_001034836.1	ENSP00000482288	12
RP1-161P9.5	upstream gene variant	2360	ENST00000603816	?	?	1
TAF15	upstream gene variant, downstream gene variant	1528	ENST00000604434	?	?	2
TAF15	upstream gene variant, downstream gene variant	664	ENST00000604360	?	?	4
TAF15	upstream gene variant, downstream gene variant	1552	ENST00000605197	?	?	3
TAF15	upstream gene variant	1501	ENST00000605844	NM_139215.2	ENSP00000474096	1
TAF15	upstream gene variant, downstream gene variant	197	ENST00000604879	?	ENSP00000475044	3
TAF15	upstream gene variant	3439	ENST00000603967	?	ENSP00000474047	1
TAF15	upstream gene variant	1530	ENST00000603777	?	ENSP00000474522	1
TAF15	upstream gene variant, downstream gene variant	2337	ENST00000603346	?	?	3
TAF15	downstream gene variant	2943	ENST00000603067	?	?	1
TAF15	upstream gene variant	2581	ENST00000604694	?	ENSP00000475147	1
TAF15	upstream gene variant	1923	ENST00000604195	?	?	1
TAF15	upstream gene variant	1535	ENST00000604841	NM_003487.3	ENSP00000474609	1
TAF15	upstream gene variant, downstream gene variant	1530	ENST00000605649	?	?	3
TAF15	upstream gene variant, downstream gene variant	703	ENST00000603427	?	ENSP00000474436	4
TAF15	upstream gene variant, downstream gene variant	1263	ENST00000603393	?	ENSP00000474653	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC015849.13	ENST00000603678	?	?	2
AC015849.16	ENST00000604647	?	?	1
AC015849.16	ENST00000604907	?	?	3
AC015849.19	ENST00000603981	?	?	4
AC015849.2	ENST00000605548	?	?	4
CCL5	ENST00000605140	?	ENSP00000475057	3
CCL5	ENST00000603197	NM_002985.2	ENSP00000474412	2
CCL5	ENST00000605509	NM_001278736.1	ENSP00000474141	2
HEATR9	ENST00000605080	?	ENSP00000474893	3
HEATR9	ENST00000603218	?	ENSP00000474337	1
HEATR9	ENST00000604400	?	?	3
HEATR9	ENST00000603852	?	ENSP00000474899	3
HEATR9	ENST00000604834	NM_152781.2	ENSP00000473941	3
HEATR9	ENST00000603870	?	ENSP00000473760	3
HEATR9	ENST00000603323	?	ENSP00000474391	3

MMP28	ENST00000619655	?	ENSP00000481051	4
MMP28	ENST00000612672	NM_001032278.1	ENSP00000483539	1
MMP28	ENST00000605424	NM_024302.3	ENSP00000473853	2
MMP28	ENST00000615317	?	ENSP00000482385	2
MMP28	ENST00000615136	?	ENSP00000483299	2
MMP28	ENST00000612292	?	ENSP00000484562	1
RDM1	ENST00000613308	NM_001034836.1	ENSP00000482288	10
RDM1	ENST00000619876	?	ENSP00000483890	10
RDM1	ENST00000612980	NM_001163120.1	ENSP00000483387	17
RDM1	ENST00000616596	NM_001163121.1	ENSP00000478915	17
RDM1	ENST00000615024	?	ENSP00000481648	17
RDM1	ENST00000615378	NM_001163125.1	ENSP00000480131	10
RDM1	ENST00000617591	NM_001163124.1	ENSP00000479622	17
RDM1	ENST00000619193	?	ENSP00000482981	17
RDM1	ENST00000619262	NM_001163130.1	ENSP00000479310	17
RDM1	ENST00000618511	?	ENSP00000477995	17
RDM1	ENST00000615288	?	ENSP00000477869	17
RDM1	ENST00000619368	?	ENSP00000478131	17
RDM1	ENST00000619828	NM_001163122.1	ENSP00000483933	17
RDM1	ENST00000620284	NM_145654.3	ENSP00000483549	17
TAF15	ENST00000605844	NM_139215.2	ENSP00000474096	12
TAF15	ENST00000604841	NM_003487.3	ENSP00000474609	12
TAF15	ENST00000604879	?	ENSP00000475044	7
TAF15	ENST00000604360	?	?	3
TAF15	ENST00000605649	?	?	4
TAF15	ENST00000603777	?	ENSP00000474522	12
TAF15	ENST00000603346	?	?	3
TAF15	ENST00000603067	?	?	17
TAF15	ENST00000603967	?	ENSP00000474047	5
TAF15	ENST00000604694	?	ENSP00000475147	2
TAF15	ENST00000605197	?	?	4
TAF15	ENST00000603393	?	ENSP00000474653	10
TAF15	ENST00000603427	?	ENSP00000474436	3

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
TAF15	ENST00000603967	?	ENSP00000474047	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL5	ENST00000605140	?	ENSP00000475057	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)

gene	affected transcript	RefSeq id	variant(s)
TAF15	ENST00000604195	?	1