Locus 139 (rs1455434)

SNiPAcard

Block annotations

Block info

genomic range	chr19:41,214,932-41,291,872
block size	76,941 bp
variant count	30 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.573 [-3.842 – 1.533]	gene(s) hit or close-by	ADCK4, C19orf54, ITPKC, MIA, MIA-RAB4B, RAB4B, RAB4B-EGLN2, RNU6-195P, SNRPA
phastCons	μ = 0.034 [0 – 0.546]	eQTL gene(s)	C19orf54, MIA
GERP++	μ = -1.528 [-9.93 – 1.94]	potentially regulated gene(s)	C19orf54
CADD score	μ = 5.053 [0.217 – 14.91]	disease gene(s)	ADCK4, ITPKC

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ADCK4	NEPHROTIC SYNDROME, TYPE 9	OMIM	MIM:615573
ITPKC	KAWASAKI DISEASE	OMIM	MIM:611775
ADCK4	Familial idiopathic steroid-resistant nephrotic syndrome with focal segmental hyalinosis	OrphaNet	OrphaNet:93213

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
C19orf54	?	ENSG00000188493	thyroid	7.69×10-8 (p-value)	GTEx Portal V6	21
MIA	?	ENSG00000261857	tibial nerve	1.84×10-7 (p-value)	GTEx Portal V6	23
C19orf54	?	ENSG00000188493	blood	2.44×10-8 (q-value)	SeeQTL DB (HapMap)	8

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
EGR1	MA0162.2	5	yes	0.000	1

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000330481	1	ENST00000470681, ENST00000469741	C19orf54

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000024392		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000228995	1	ENCP00000024393	C19orf54

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001648159 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00000346640 (promoter)	4	embryonic stem cell (H1ESC)	H3K36me3, CTCF, PolII, p300, Sin3Ak20, H3K27ac, TAF1, H3K4me2, SP1, USF1, H3K9ac, H3K4me3, DNase1
		HSMMtube	H3K27ac, H3K4me2, DNase1, H2AZ, H3K4me3, H3K9ac, H3K79me2
		blood (K562)	H3K9ac, Gabp, H3K79me2, H2AZ, ELF1, Cfos, PolII, H3K4me2, H3K27me3, H3K36me3, DNase1, H3K4me3, Max, H3K27ac, Egr1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H3K4me3, DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac, H3K79me2, H2AZ
		liver (HepG2)	Gabp, PolII, H3K79me2, TAF1, ELF1, USF1, H3K4me1, H3K4me2, HNF4A, H3K9ac, H3K27ac, H3K4me3, H3K36me3, H3K27me3, DNase1
		blood (GM12878)	H3K27ac, H3K4me2, H3K9ac, H3K4me3, H3K79me2, Pbx3, Sin3Ak20, PolII, H2AZ, DNase1, Gabp, ELF1
		lung (IMR90)	DNase1, H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K36me3, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2, H3K36me3
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K9ac, Jund, H3K4me2, H3K27ac, H3K4me3, H3K79me2, Gabp, PolII
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cjun, Max, H3K4me3, H3K4me2, H3K9ac, H3K4me1, H3K27ac, PolII, DNase1, Cmyc
		A549	H3K9ac, H3K4me2, H3K4me3, DNase1, H3K27ac, H3K36me3
ENSR00000346651 (promoter)	5	embryonic stem cell (H1ESC)	TAF1, H3K4me2, SP1, USF1, H3K9ac, H3K4me3, Yy1, ATF3, H4K5ac, H3K36me3, H3K27me3, PolII, TAF7, Sin3Ak20, H3K27ac, DNase1
		HSMMtube	H3K36me3, H3K4me2, H3K9ac, H3K27ac, DNase1, H2AZ, H3K4me3
		blood (K562)	Yy1, Egr1, H3K27ac, Nfe2, E2F6, H3K9ac, USF1, Cmyc, H3K79me2, HEY1, H2AZ, TAF1, ELF1, Max, Cfos, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K36me3, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2
		liver (HepG2)	H3K4me1, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K36me3, USF1, ELF1, Gabp, Yy1, PolII, H3K79me2, TAF1, DNase1
		blood (GM12878)	H3K36me3, H3K4me1, DNase1, Yy1, ELF1, H3K79me2, H3K4me3, USF1, H3K27ac, H4K20me1, Cfos, H3K4me2, H2AZ, PolII, Tr4, Cmyc, H3K9ac
		lung (IMR90)	H4K5ac, H3K36me3, H3K4me3, H3K4ac, H3K9ac, H4K91ac, H3K4me2, H3K27ac, DNase1, H4K8ac, H3K79me2
		nervous (NH-A)	DNase1, H3K36me3, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K36me3, H3K4me3, H3K4me2, PolII, H3K9ac, H3K27ac
		NHLF	H3K36me3, H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2, H3K36me3
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H4K20me1, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	H3K4me2, H3K4me1, H3K36me3, H3K9ac, H3K4me3, DNase1, H3K27ac
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, H3K4me1, TAF1, Max, H3K4me3, H3K79me2, Gabp, PolII, Cmyc, H3K36me3
		monocytes (Monocytes-CD14+)	H3K4me3, H3K36me3, H3K9ac, DNase1, H3K4me1, H3K27ac
		endothelium (HUVEC)	H3K36me3, DNase1, Cmyc, PolII, H3K27ac, Max, H3K4me3, H3K9ac
		A549	H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00001648164 (promoter)	1	Osteobl	H3K27me3
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ADCK4	upstream gene variant	1207	ENST00000594084	?	ENSP00000473189	2
ADCK4	upstream gene variant	1379	ENST00000594490	?	ENSP00000471310	3

ADCK4	downstream gene variant, upstream gene variant	943	ENST00000596357 ?	ENSP00000472925	4
ADCK4	upstream gene variant	930	ENST00000600080 ?	ENSP00000473017	3
ADCK4	downstream gene variant, upstream gene variant	401	ENST00000600707 ?	ENSP00000472978	5
ADCK4	upstream gene variant	916	ENST00000601967 ?	ENSP00000470916	3
ADCK4	upstream gene variant	2529	ENST00000324464 NM_024876.3	ENSP00000315118	2
ADCK4	upstream gene variant	2518	ENST00000594720 ?	ENSP00000470876	2
ADCK4	downstream gene variant, upstream gene variant	398	ENST00000593723 ?	ENSP00000472775	5
ADCK4	upstream gene variant	941	ENST00000243583 NM_001142555.2	ENSP00000243583	3
ADCK4	upstream gene variant	1229	ENST00000601304 ?	ENSP00000472519	3
ADCK4	upstream gene variant	929	ENST00000595254 ?	ENSP00000470894	3
ADCK4	upstream gene variant	908	ENST00000593544 ?	?	3
ADCK4	upstream gene variant	229	ENST00000596455 ?	?	4
ADCK4	upstream gene variant	47	ENST00000599643 ?	ENSP00000471192	4
ADCK4	upstream gene variant	2529	ENST00000601451 ?	?	2
C19orf54	upstream gene variant	93	ENST00000598352 ?	ENSP00000473024	5
C19orf54	upstream gene variant	22	ENST00000597507 ?	ENSP00000472022	3
C19orf54	downstream gene variant, upstream gene variant	168	ENST00000598485 ?	ENSP00000469330	4
C19orf54	downstream gene variant, upstream gene variant	649	ENST00000470681 ?	ENSP00000471114	3
C19orf54	downstream gene variant, upstream gene variant	145	ENST00000596940 ?	ENSP00000472820	6
C19orf54	upstream gene variant	104	ENST00000600139 ?	ENSP00000472948	5
C19orf54	downstream gene variant	4111	ENST00000594163 ?	?	1
C19orf54	downstream gene variant, upstream gene variant	61	ENST00000378313 NM_198476.3	ENSP00000367564	5
C19orf54	downstream gene variant	4941	ENST00000596809 ?	ENSP00000469807	1
C19orf54	downstream gene variant, upstream gene variant	588	ENST00000469741 ?	ENSP00000471589	3
ITPKC	upstream gene variant	952	ENST00000263370 NM_025194.2	ENSP00000263370	3
ITPKC	upstream gene variant	106	ENST00000597003 ?	ENSP00000472655	3
MIA	upstream gene variant, downstream gene variant	190	ENST00000593317 ?	?	7
MIA	upstream gene variant, downstream gene variant	349	ENST00000601159 ?	ENSP00000471439	5
MIA	downstream gene variant	3612	ENST00000594436 NM_001202553.1	ENSP00000470129	4
MIA	upstream gene variant, downstream gene variant	346	ENST00000597140 ?	ENSP00000470641	5
MIA	upstream gene variant, downstream gene variant	176	ENST00000263369 NM_006533.3	ENSP00000263369	5
MIA	upstream gene variant, downstream gene variant	63	ENST00000597784 ?	ENSP00000469499	5
MIA	upstream gene variant, downstream gene variant	34	ENST00000597600 ?	ENSP00000472982	3
MIA-RAB4B	upstream gene variant	1712	ENST00000600729 ?	ENSP00000472384	1
RAB4B	upstream gene variant	478	ENST00000598430 ?	?	5
RAB4B	upstream gene variant	1615	ENST00000357052 NM_016154.4	ENSP00000349560	3
RAB4B	upstream gene variant	1705	ENST00000378307 ?	ENSP00000367557	3
RAB4B	upstream gene variant	3781	ENST00000595728 ?	ENSP00000471803	2
RAB4B	upstream gene variant, downstream gene variant	1082	ENST00000600078 ?	?	5
RAB4B	upstream gene variant	1774	ENST00000602173 ?	ENSP00000471504	3
RAB4B	upstream gene variant	1646	ENST00000602069 ?	?	3

RAB4B	upstream gene variant	1612	ENST00000594800	?	ENSP00000470246	3
RAB4B	upstream gene variant	2443	ENST00000597476	?	?	3
RAB4B-EGLN2	upstream gene variant	3775	ENST00000596216	?	?	2
RAB4B-EGLN2	upstream gene variant	1638	ENST00000601949	?	?	3
RAB4B-EGLN2	upstream gene variant	1667	ENST00000594136	?	ENSP00000469872	3
RNU6-195P	upstream gene variant	864	ENST00000411352	?	?	2
SNRPA	upstream gene variant, downstream gene variant	405	ENST00000599570	?	?	5
SNRPA	downstream gene variant	2669	ENST00000598452	?	?	2
SNRPA	upstream gene variant, downstream gene variant	157	ENST00000600456	?	ENSP00000471230	6
SNRPA	upstream gene variant, downstream gene variant	401	ENST00000599362	?	ENSP00000472258	5
SNRPA	upstream gene variant, downstream gene variant	68	ENST00000601393	?	ENSP00000472355	5
SNRPA	upstream gene variant, downstream gene variant	397	ENST00000601253	?	ENSP00000469224	5
SNRPA	upstream gene variant	133	ENST00000598923	?	?	4
SNRPA	upstream gene variant	398	ENST00000597353	?	ENSP00000472449	3
SNRPA	upstream gene variant, downstream gene variant	167	ENST00000601545	?	ENSP00000470534	4
SNRPA	upstream gene variant, downstream gene variant	389	ENST00000243563	NM_004596.4	ENSP00000243563	4
SNRPA	upstream gene variant, downstream gene variant	891	ENST00000596860	?	?	4

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
C19orf54	ENST00000378313	NM_198476.3	ENSP00000367564	L	Ttg/Ctg	1
ITPKC	ENST00000263370	NM_025194.2	ENSP00000263370	A	gcG/gcA	1
MIA	ENST00000597784	?	ENSP00000469499	P	ccT/ccC	1
MIA	ENST00000263369	NM_006533.3	ENSP00000263369	P	ccT/ccC	1
MIA	ENST00000594436	NM_001202553.1	ENSP00000470129	P	ccT/ccC	1
MIA	ENST00000597140	?	ENSP00000470641	P	ccT/ccC	1
MIA-RAB4B	ENST00000600729	?	ENSP00000472384	P	ccT/ccC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ADCK4	ENST00000596357	?	ENSP00000472925	1
ADCK4	ENST00000324464	NM_024876.3	ENSP00000315118	4
ADCK4	ENST00000601451	?	?	4
ADCK4	ENST00000594490	?	ENSP00000471310	2
ADCK4	ENST00000601304	?	ENSP00000472519	2
ADCK4	ENST00000596455	?	?	2
ADCK4	ENST00000593544	?	?	2
ADCK4	ENST00000600080	?	ENSP00000473017	2
ADCK4	ENST00000599643	?	ENSP00000471192	2
ADCK4	ENST00000595254	?	ENSP00000470894	2
ADCK4	ENST00000594720	?	ENSP00000470876	4

ADCK4	ENST00000601967	?	ENSP00000470916	2
ADCK4	ENST00000243583	NM_001142555.2	ENSP00000243583	2
ADCK4	ENST00000600707	?	ENSP00000472978	1
ADCK4	ENST00000594084	?	ENSP00000473189	4
C19orf54	ENST00000596809	?	ENSP00000469807	5
C19orf54	ENST00000470681	?	ENSP00000471114	3
C19orf54	ENST00000597507	?	ENSP00000472022	2
C19orf54	ENST00000598485	?	ENSP00000469330	2
C19orf54	ENST00000469741	?	ENSP00000471589	3
ITPKC	ENST00000597003	?	ENSP00000472655	3
ITPKC	ENST00000263370	NM_025194.2	ENSP00000263370	7
MIA	ENST00000597784	?	ENSP00000469499	2
MIA	ENST00000263369	NM_006533.3	ENSP00000263369	2
MIA	ENST00000601159	?	ENSP00000471439	2
MIA	ENST00000594436	NM_001202553.1	ENSP00000470129	3
MIA	ENST00000597140	?	ENSP00000470641	2
MIA	ENST00000597600	?	ENSP00000472982	1
MIA-RAB4B	ENST00000600729	?	ENSP00000472384	9
RAB4B	ENST00000600078	?	?	4
RAB4B	ENST00000602173	?	ENSP00000471504	6
RAB4B	ENST00000597476	?	?	6
RAB4B	ENST00000602069	?	?	6
RAB4B	ENST00000378307	?	ENSP00000367557	6
RAB4B	ENST00000594800	?	ENSP00000470246	6
RAB4B	ENST00000357052	NM_016154.4	ENSP00000349560	6
RAB4B	ENST00000595728	?	ENSP00000471803	6
RAB4B-EGLN2	ENST00000596216	?	?	6
RAB4B-EGLN2	ENST00000594136	?	ENSP00000469872	6
RAB4B-EGLN2	ENST00000601949	?	?	6
SNRPA	ENST00000600456	?	ENSP00000471230	2
SNRPA	ENST00000598923	?	?	4
SNRPA	ENST00000601545	?	ENSP00000470534	5
SNRPA	ENST00000599570	?	?	3
SNRPA	ENST00000601393	?	ENSP00000472355	4
SNRPA	ENST00000601253	?	ENSP00000469224	3
SNRPA	ENST00000243563	NM_004596.4	ENSP00000243563	5
SNRPA	ENST00000597353	?	ENSP00000472449	5
SNRPA	ENST00000598452	?	?	1
SNRPA	ENST00000599362	?	ENSP00000472258	3

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
C19orf54	ENST00000596809	?	ENSP00000469807	3
C19orf54	ENST00000470681	?	ENSP00000471114	1
C19orf54	ENST00000469741	?	ENSP00000471589	1
MIA	ENST00000594436	NM_001202553.1	ENSP00000470129	1
SNRPA	ENST00000243563	NM_004596.4	ENSP00000243563	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RAB4B	ENST00000598430	?	1
SNRPA	ENST00000599570	?	1