Locus 118 (rs12134133)

SNiPAcard

Block annotations

Block info

genomic range	chr1:207,411,912-207,529,576
block size	117,665 bp
variant count	79 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.314 [-2.969 – 1.497]	gene(s) hit or close-by	CD55, RP11-6J21.2
phastCons	μ = 0.070 [0 – 0.985]	eQTL gene(s)	CD55, CR1
GERP++	μ = -0.380 [-6.12 – 3.97]	potentially regulated gene(s)	C4BPAP2, C4BPB, IL20, PIGR, RP11-164O23.5
CADD score	μ = 3.646 [0.01 – 11.79]	disease gene(s)	CD55

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
threonine	2.20×10^{-5}	Metabolomics GWAS Server	24816252	8
4-acetaminophen sulfate	8.26×10^{-5}	Metabolomics GWAS Server	24816252	1

Disease gene annotation

gene	trait	source DB	source entry/link
CD55	BLOOD GROUP, CROMER SYSTEM	OMIM	MIM:613793

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CD55	?	ENSG00000196352	adrenal gland	3.70×10^{-8} (p-value)	GTEx Portal V6	78
CD55	?	ENSG00000196352	tibial artery	1.85×10^{-8} (p-value)	GTEx Portal V6	78
CD55	?	ENSG00000196352	transformed fibroblasts	6.82×10^{-6} (p-value)	GTEx Portal V6	2
CR1	ENST00000367051	ILMN_1767193	blood	2.76×10^{-5} (p-value)	MuTHER consortium	16
CR1	ENST00000367052					
CR1	ENST00000400960					
CR1	ENST00000367049					
CR1	ENST00000529814					
CR1	ENST00000367053					
?	?	ILMN_1742601	blood	3.62×10^{-5} (p-value)	MuTHER consortium	9

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000053173	1	ENCP00000006362	PIGR
		ENCP00000006355	IL20

ENCE00000053309	1	ENCP00000006375	RP11-164O23.5
		ENCP00000006373	C4BPB
		ENCP00000006371	C4BPB
ENCE00000053321	1	ENCP00000006373	C4BPB
ENCE00000053363	1	ENCP00000006378	C4BPAP2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001590164 (enhancer)	1	cervix (HeLa-S3)	DNase1, H3K27ac
		HSMMtube	H3K27me3
		skin (NHEK)	H3K4me1
ENSR00000551826 (promoter flanking region)	10	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K4me2, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, Cjun, PolII, Jund, H3K4me2, H3K27ac, Max, Cmyc
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K4me1, FOXA1, H3K27ac, DNase1
		A549	H3K4me3
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1, H3K4me1, H3K27ac, H3K4me2
ENSR00000551828 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		blood (K562)	CTCF, Rad21
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, DNase1, CTCF
		liver (HepG2)	Rad21, CTCF
		lung (IMR90)	CTCF
		skin (NHEK)	CTCF
ENSR00000551831 (promoter flanking region)	4	cervix (HeLa-S3)	DNase1
		embryonic stem cell (H1ESC)	DNase1
		endothelium (HUVEC)	Cjun, DNase1, H3K4me2, H3K4me1, H3K27ac
		nervous (NH-A)	DNase1
		skin (NHDF-AD)	DNase1
ENSR00001528072 (CTCF binding site)	1	Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
ENSR00000551833 (enhancer)	1	cervix (HeLa-S3)	H3K36me3
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1, H3K4me3
ENSR00000288774 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		blood (K562)	H2AZ
ENSR00000288776 (promoter flanking region)	5	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		HSMMtube	DNase1
		blood (K562)	Brg1, DNase1, H3K27ac, Max, Junb, PU1, MEF2A, Jund, Cmyc, H2AZ, Cjun
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H2AZ, DNase1
		liver (HepG2)	H3K4me2
		blood (GM12878)	H2AZ, PU1
		nervous (NH-A)	DNase1, H3K4me2
		skin (NHEK)	DNase1
		NHLF	DNase1
		Osteobl	H3K27me3, H3K4me2
		blood (DND-41)	H3K27me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me2
		cervix (HeLa-S3)	DNase1, Cjun, PolII, H3K9ac, Jund, H3K4me2, H3K27ac, TAF1, Max, Cfos
		monocytes (Monocytes-CD14+)	H3K27ac, H3K4me1, DNase1

		endothelium (HUVEC)	Cjun, Max, DNase1, H3K4me2, H3K27ac, PolII
		A549	H3K4me3, H3K4me2, DNase1
ENSR00001590170 (CTCF binding site)	1	cervix (HeLa-S3)	DNase1, Jund, H3K4me2, H3K27ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1
		embryonic stem cell (H1ESC)	H3K36me3, DNase1
		nervous (NH-A)	DNase1
		blood (K562)	Brg1, DNase1, Junb, MEF2A, Jund, Cjun
ENSR00000551839 (promoter)	2	embryonic stem cell (H1ESC)	PolII, Rad21, Jund, TAF7, Sin3Ak20, CTCF, H3K27ac, Yy1, TAF1, H3K4me2, USF1, H3K9ac, H3K4me3, DNase1, H3K27me3, H3K36me3
		HSMMtube	H3K9ac, H3K4me2, H2AZ, H3K27me3, DNase1
		blood (K562)	H3K79me2, HEY1, H2AZ, TAF1, Cmyc, PolII, H3K4me2, CTCF, H3K36me3, DNase1, H3K4me3, H3K9ac, PU1, H3K27ac, Max
		skin (NHDF-AD)	H3K4me2, H3K9ac, DNase1, H3K4me3, CTCF, H3K27ac
		muscle (HSMM)	H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, DNase1
		liver (HepG2)	DNase1, H3K27me3, H3K4me3, PolII, TAF1, H2AZ, H3K4me2, H3K9ac, Cmyc
		lung (IMR90)	DNase1, H3K4me2, H3K4me3
		blood (GM12878)	PolII, H2AZ, DNase1, CTCF, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, DNase1
		skin (NHEK)	CTCF, H3K27ac, H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H3K27me3, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K27me3, H3K9ac, H3K4me2, H3K4me1
		breast (HMEC)	H3K4me2, H3K9ac, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, H3K79me2, PolII, Cmyc, CTCF, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, DNase1, Cmyc, PolII, H3K27ac, Cjun, Max, H3K4me2, H3K4me3, H3K9ac
		A549	DNase1, H3K27ac, H3K36me3, H3K9ac, H3K4me2, H3K4me3
ENSR00001590172 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, PolII, Rad21, TAF7, Sin3Ak20, H3K27ac, Yy1, TAF1, H3K4me2, DNase1, H3K9ac, H3K4me3
		HSMMtube	H3K9ac, H3K4me2, H2AZ
		blood (K562)	H3K27ac, Max, H3K9ac, Cmyc, H3K79me2, HEY1, H2AZ, PolII, H3K4me2, H3K36me3, H3K4me3
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, DNase1, H3K4me3, H3K36me3
		muscle (HSMM)	H3K4me2, H3K9ac, H3K36me3, H3K27ac, H2AZ, H3K4me3
		liver (HepG2)	H3K4me2, H3K9ac, H3K4me3, H3K27me3, DNase1
		lung (IMR90)	H3K4me2, H3K36me3, H3K4me3
		blood (GM12878)	PolII, H2AZ, DNase1, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3
		NHLF	H3K27ac, H3K9ac, H3K4me3
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, H3K9ac, H3K27me3
		breast (HMEC)	H3K4me2, H3K9ac, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K79me2, PolII, H3K36me3, H3K4me3, H3K27ac, H3K9ac, DNase1, H3K4me2
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII, H3K36me3
		A549	H3K4me3, H3K4me2, H3K9ac, H3K27ac, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CD55	upstream gene variant, downstream gene variant	457	ENST00000343420	?	ENSP00000340631	2
CD55	upstream gene variant	174	ENST00000367064	NM_000574.3	ENSP00000356031	1
CD55	upstream gene variant	399	ENST00000367067	?	ENSP00000356034	1
CD55	upstream gene variant	1107	ENST00000465534	?	?	3
CD55	upstream gene variant, downstream gene variant	353	ENST00000488171	?	?	3
CD55	upstream gene variant	1107	ENST00000476590	?	?	3
CD55	upstream gene variant	309	ENST00000391921	?	ENSP00000375788	1
CD55	upstream gene variant	367	ENST00000314754	NM_001114752.1	ENSP00000316333	1
CD55	upstream gene variant, downstream gene variant	198	ENST00000367063	NM_001300902.1	ENSP00000356030	3

RP11-6J21.2	upstream gene variant, downstream gene variant	1110	ENST00000417084	?	?	10

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
RP11-6J21.2	ENST00000417084	?	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CD55	ENST00000343420	?	ENSP00000340631	3
CD55	ENST00000314754	NM_001114752.1	ENSP00000316333	10
CD55	ENST00000367064	NM_000574.3	ENSP00000356031	10
CD55	ENST00000367067	?	ENSP00000356034	10
CD55	ENST00000367063	NM_001300902.1	ENSP00000356030	3
CD55	ENST00000488171	?	?	3
CD55	ENST00000391921	?	ENSP00000375788	10
CD55	ENST00000465534	?	?	8
CD55	ENST00000476590	?	?	8
RP11-6J21.2	ENST00000417084	?	?	48