Locus 205 (rs3828922)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,708,147-31,741,490
block size	33,344 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.734 [-3.279 – 0.364]	gene(s) hit or close-by	CLIC1, MSH5, MSH5-SAPCD1, RNU6-850P, SAPCD1, SAPCD1-AS1, VARS, VWA7
phastCons	μ = 0.013 [0 – 0.066]	eQTL gene(s)	AIF1, ATF6B, BAG6, C6orf48, DXO, HSPA1B, LY6G5C, LY6G6C, NELFE, PRRC2A, SKIV2L, SNORD52, TNF
GERP++	μ = -1.588 [-4.48 – 1.66]	potentially regulated gene(s)	–
CADD score	μ = 4.579 [0.264 – 7.987]	disease gene(s)	SKIV2L, TNF

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	DECIPHER	MIM:615602
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	OMIM	MIM:614602
TNF	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
TNF	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
TNF	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
SKIV2L	Syndromic diarrhea	OrphaNet	OrphaNet:84064

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LY6G5C	?	ENSG00000204428	caudate basal ganglia	2.18×10^{-7} (p-value)	GTEx Portal V6	6
DXO	ENST00000474587	ILMN_1726990	blood	3.95×10^{-10} (p-value)	MuTHER consortium	5
DXO	ENST00000491327		adipocyte	1.65×10^{-5} (p-value)	MuTHER consortium	5
DXO	ENST00000473976					
DXO	ENST00000480240					
DXO	ENST00000498357					
DXO	ENST00000477826					
DXO	ENST00000492946					
DXO	ENST00000375349					
DXO	ENST00000337523					
DXO	ENST00000460058					
DXO	ENST00000375356					
DXO	ENST00000487914					
DXO	ENST00000478221					
DXO	ENST00000485557					
PRRC2A	ENST00000376033	ILMN_2408179	blood	1.95×10^{-7} (p-value)	MuTHER consortium	5
PRRC2A	ENST00000487089					

PRRC2A	ENST00000376007					
PRRC2A	ENST00000487839					
PRRC2A	ENST00000462617					
PRRC2A	ENST00000482441					
PRRC2A	ENST00000492691					
HSPA1B	ENST00000375650	ILMN_1660436	skin	3.11×10^{-6} (p-value)	MuTHER consortium	5
AIF1	ENST00000337917	ILMN_1792473	blood	8.51×10^{-10} (p-value)	MuTHER consortium	5
AIF1	ENST00000466820					
AIF1	ENST00000497362					
AIF1	ENST00000376059					
AIF1	ENST00000376049					
LY6G5C	?	ENSG00000204428	lung	6.94×10^{-7} (p-value)	GTEx Portal V6	6
C6orf48	?	ENSG00000204387	tibial nerve	6.53×10^{-8} (p-value)	GTEx Portal V6	6
LY6G5C	?	ENSG00000204428	blood	1.15×10^{-11} (p-value)	GTEx Portal V6	6
C6orf48	?	ENSG00000204387	blood	2.20×10^{-7} (p-value)	GTEx Portal V6	6
LY6G6C	?	ENSG00000204421	esophagus mucosa	2.20×10^{-14} (p-value)	GTEx Portal V6	6
TNF	ENST00000449264	ILMN_1728106	blood	5.59×10^{-5} (p-value)	Westra et al.	5
NELFE	ENST00000492185	ILMN_1765532	blood	8.69×10^{-6} (p-value)	Westra et al.	5
NELFE	ENST00000488426					
NELFE	ENST00000481121					
NELFE	ENST00000375429					
SKIV2L	ENST00000484835	ILMN_1666512	blood	9.00×10^{-5} (p-value)	Westra et al.	5
SKIV2L	ENST00000375394					
SKIV2L	ENST00000474839					
SKIV2L	ENST00000470453					
SKIV2L	ENST00000465703					
SKIV2L	ENST00000491994					
SKIV2L	ENST00000471818					
SKIV2L	ENST00000483553					
SKIV2L	ENST00000485349					
C6orf48	?	ENSG00000204387	testis	9.76×10^{-7} (p-value)	GTEx Portal V6	6
C6orf48	?	ENSG00000204387	unexposed skin	1.45×10^{-7} (p-value)	GTEx Portal V6	6
?	?	ILMN_1770787	b-cell	4.66×10^{-5} (p-value)	Fairfax et al.	3
?	?	ILMN_2044471	b-cell	1.41×10^{-4} (p-value)	Fairfax et al.	3
C6orf48	ENST00000375638	ILMN_2391765	monocyte	5.43×10^{-10} (p-value)	Fairfax et al.	3
C6orf48	ENST00000375635		b-cell	1.36×10^{-11} (p-value)	Fairfax et al.	3
C6orf48	ENST00000395788					
C6orf48	ENST00000614363					
C6orf48	ENST00000375640					
SNORD52	ENST00000364884					
C6orf48	ENST00000375641					
C6orf48	ENST00000395789					
C6orf48	ENST00000375639					
C6orf48	ENST00000375633					
C6orf48	ENST00000375642					
ATF6B	ENST00000375201	ILMN_3194911	b-cell	5.63×10^{-5} (p-value)	Fairfax et al.	3
ATF6B	ENST00000375203					

?	?	ILMN_2325394	monocyte	1.41×10-13 (p-value)	Zeller et al.	1
C6orf48	ENST00000375638	ILMN_2391765	monocyte	5.44×10-25 (p-value)	Zeller et al.	1
C6orf48	ENST00000375635					
C6orf48	ENST00000395788					
C6orf48	ENST00000614363					
C6orf48	ENST00000375640					
C6orf48	ENST00000395789					
C6orf48	ENST00000375641					
SNORD52	ENST00000364884					
C6orf48	ENST00000375639					
C6orf48	ENST00000375633					
C6orf48	ENST00000375642					
BAG6	?	ENSG00000204463	tibial artery	7.03×10-6 (p-value)	GTEx Portal V6	1
LY6G5C	?	ENSG00000204428	thyroid	3.04×10-6 (p-value)	GTEx Portal V6	2
LY6G5C	?	ENSG00000204428	sun exposed skin	7.88×10-7 (p-value)	GTEx Portal V6	2
LY6G5C	?	ENSG00000204428	nucleus accumbens	4.64×10-7 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000487898 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3, H3K4me3, PolII, Rad21, Jund, Nrsf, Sin3Ak20, CTCF, H3K27ac, ATF3, Yy1, H3K4me1, TAF1, H3K4me2, SP1, USF1, H3K9ac
		HSMMtube	H3K9ac, DNase1, H3K36me3, H2AZ, H3K4me3, H3K27ac, H3K4me2
		blood (K562)	H3K4me3, Jund, H3K79me2, HEY1, H2AZ, TAF1, ELF1, Cjun, Cfos, Cmyc, PolII, H4K20me1, H3K4me2, CTCF, H3K4me1, H3K36me3, H3K9ac, E2F6, Yy1, Ini1, Brg1, Egr1, FOSL1, H3K27ac, Max, SP2, ZBTB7A, DNase1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac, CTCF, H3K4me1, H3K36me3
		muscle (HSMM)	H2AZ, H3K79me2, H3K4me3, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	DNase1, H3K4me2, HNF4A, FOXA1, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3, H3K36me3, p300, H3K4me1, FOSL2, Jund, HNF4G, FOXA2, PolII, HDAC2, H3K79me2, TAF1, RXRA, ELF1
		lung (IMR90)	H3K9ac, H3K4ac, DNase1, H3K79me2, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3
		blood (GM12878)	H3K27ac, Cfos, H3K4me2, H3K9ac, H3K36me3, H3K4me3, H3K79me2, PolII, H2AZ, H3K4me1, DNase1, CTCF, Yy1, ELF1
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	H3K4me2, H3K4me3, H3K36me3, DNase1, H3K9ac, H3K27ac, H3K4me1, CTCF
		NHLF	H3K27ac, H3K36me3, DNase1, H3K4me3, H3K9ac
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K4me2, CTCF, H3K27ac, H3K9ac, H3K4me3, H3K36me3, H3K4me1
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, H3K36me3, CTCF
		cervix (HeLa-S3)	DNase1, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, H3K79me2, Gabp, PolII, Cmyc, CTCF, H3K36me3, H3K9ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, DNase1, Cmyc, CTCF, PolII, H3K27ac, Cjun, Max, H3K4me2, H3K4me3, H3K9ac, H3K4me1
		A549	H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CLIC1	upstream gene variant	3803	ENST00000375784	NM_001288.4	ENSP00000364940	1
CLIC1	upstream gene variant	3798	ENST00000375779	?	ENSP00000364934	1
CLIC1	upstream gene variant	607	ENST00000395892	?	ENSP00000379229	3
CLIC1	upstream gene variant	3825	ENST00000616760	NM_001287594.1	ENSP00000479808	1
CLIC1	upstream gene variant	3052	ENST00000375780	NM_001287593.1	ENSP00000364935	1

MSH5	upstream gene variant	884	ENST00000497269	?	ENSP00000419131	2
MSH5	downstream gene variant	924	ENST00000463094	?	?	2
MSH5	upstream gene variant	722	ENST00000429846	?	ENSP00000406849	1
MSH5	downstream gene variant	170	ENST00000425703	?	ENSP00000402842	2
MSH5	upstream gene variant, downstream gene variant	631	ENST00000450148	?	ENSP00000394971	3
MSH5	downstream gene variant	317	ENST00000494646	?	?	1
MSH5	upstream gene variant	876	ENST00000463144	?	ENSP00000419648	1
MSH5	downstream gene variant	1356	ENST00000468602	?	?	1
MSH5	upstream gene variant	1433	ENST00000494458	?	?	1
MSH5	downstream gene variant	988	ENST00000467319	?	?	1
MSH5	downstream gene variant	989	ENST00000468136	?	?	1
MSH5-SAPCD1	upstream gene variant	2506	ENST00000425424	?	ENSP00000413372	1
MSH5-SAPCD1	upstream gene variant	1954	ENST00000476085	?	ENSP00000435414	1
MSH5-SAPCD1	upstream gene variant	230	ENST00000491552	?	?	1
RNU6-850P	upstream gene variant	3437	ENST00000516934	?	?	1
SAPCD1	upstream gene variant	3130	ENST00000494299	?	?	1
SAPCD1	upstream gene variant	2309	ENST00000433778	?	ENSP00000408918	1
SAPCD1	upstream gene variant	2506	ENST00000415669	NM_001039651.1	ENSP00000411948	1
SAPCD1-AS1	downstream gene variant	3820	ENST00000419679	?	?	1
VARS	downstream gene variant	3805	ENST00000470953	?	?	1
VARS	downstream gene variant	3805	ENST00000375663	NM_006295.2	ENSP00000364815	1
VWA7	downstream gene variant	2346	ENST00000497645	?	?	1
VWA7	upstream gene variant	706	ENST00000487013	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
MSH5	ENST00000375740	NM_025259.5	ENSP00000364892	5
MSH5	ENST00000463094	?	?	2
MSH5	ENST00000482280	?	?	4
MSH5	ENST00000425703	?	ENSP00000402842	2
MSH5	ENST00000423982	?	ENSP00000406352	5
MSH5	ENST00000375703	NM_172165.3	ENSP00000364855	5
MSH5	ENST00000375755	NM_002441.4	ENSP00000364908	5
MSH5	ENST00000450148	?	ENSP00000394971	2
MSH5	ENST00000375750	NM_172166.3	ENSP00000364903	5
MSH5	ENST00000463144	?	ENSP00000419648	4
MSH5	ENST00000497269	?	ENSP00000419131	2
MSH5	ENST00000395853	?	ENSP00000379194	1
MSH5	ENST00000484309	?	ENSP00000420232	1
MSH5-SAPCD1	ENST00000493662	?	ENSP00000417871	5

MSH5-SAPCD1	ENST00000498473	?	ENSP00000419220	1
VWA7	ENST00000467576	?	?	1
VWA7	ENST00000375688	NM_025258.2	ENSP00000364840	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MSH5	ENST00000423982	?	ENSP00000406352	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
MSH5	ENST00000463094	?	1