SNiPAcard

Block annotations

Block info

genomic range	chr6:31,464,029-31,472,308
block size	8,280 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.950 [-2.109 – 0.418]	**gene(s) hit or close-by**	MICB, Y_RNA
phastCons	μ = 0.040 [0 – 0.202]	**eQTL gene(s)**	HCG22, HCG27, HCP5, HLA-C, HLA-DRB5, HLA-DRB6, LST1, MICB, MUC22, XXbac-BPG248L24.12
GERP++	μ = -0.208 [-1.33 – 0.13]	**potentially regulated gene(s)**	–
CADD score	μ = 2.638 [0.78 – 7.406]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MICB	?	ENSG00000204516	cerebellar hemisphere	2.83×10^{-6} (p-value)	GTEx Portal V6	7
HCG22	?	ENSG00000228789	visceral adipocytes	1.50×10^{-6} (p-value)	GTEx Portal V6	8
HCG22	?	ENSG00000228789	lung	7.16×10^{-7} (p-value)	GTEx Portal V6	9
MICB	?	ENSG00000204516	cerebellum	5.73×10^{-8} (p-value)	GTEx Portal V6	9
HCP5	?	ENSG00000206337	transformed fibroblasts	2.96×10^{-6} (p-value)	GTEx Portal V6	7
XXbac-BPG248L24.12	?	ENSG00000271581	tibial artery	7.88×10^{-6} (p-value)	GTEx Portal V6	7
MUC22	?	ENSG00000261272	esophagus mucosa	3.50×10^{-6} (p-value)	GTEx Portal V6	7
LST1	ENST00000376099	ILMN_1688373	blood	5.96×10^{-9} (p-value)	MuTHER consortium	3
LST1	ENST00000464526					
LST1	ENST00000339530					
LST1	ENST00000419073					
LST1	ENST00000376110					
LST1	ENST00000376086					
LST1	ENST00000433492					
LST1	ENST00000438075					
LST1	ENST00000464044					
LST1	ENST00000376090					
LST1	ENST00000396101					
LST1	ENST00000376092					
LST1	ENST00000303757					
LST1	ENST00000376093					
LST1	ENST00000376089					
LST1	ENST00000396112					
LST1	ENST00000418507					
LST1	ENST00000376096					
LST1	ENST00000490742					

LST1	ENST00000211921					
LST1	ENST00000460834					
HCG22	ENST00000570223	ILMN_1667229	blood	1.28×10^{-12} (p-value)	MuTHER consortium	3
HCG22	ENST00000565192					
HCG22	ENST00000615046					
HCG22	ENST00000426185					
HCG22	ENST00000562344					
LST1	ENST00000376099	ILMN_2345353	blood	1.62×10^{-8} (p-value)	MuTHER consortium	3
LST1	ENST00000464526					
LST1	ENST00000339530					
LST1	ENST00000419073					
LST1	ENST00000376110					
LST1	ENST00000376086					
LST1	ENST00000433492					
LST1	ENST00000438075					
LST1	ENST00000464044					
LST1	ENST00000376090					
LST1	ENST00000396101					
LST1	ENST00000376092					
LST1	ENST00000303757					
LST1	ENST00000376093					
LST1	ENST00000376089					
LST1	ENST00000376096					
LST1	ENST00000418507					
LST1	ENST00000396112					
LST1	ENST00000490742					
LST1	ENST00000460834					
LST1	ENST00000211921					
MICB	ENST00000538442	ILMN_1708006	skin	8.01×10^{-8} (p-value)	MuTHER consortium	3
MICB	ENST00000399150					
MICB	ENST00000252229					
HCG27	ENST00000383331	ILMN_1746436	blood	1.03×10^{-6} (p-value)	MuTHER consortium	3
HCG27	ENST00000415276					
HLA-C	ENST00000470363	ILMN_2150787	blood	2.10×10^{-5} (p-value)	Westra et al.	1
HLA-C	ENST00000383329					
HLA-C	ENST00000487245					
HLA-C	ENST00000376228					
HLA-C	ENST00000376237					
HLA-C	ENST00000620806					
HLA-C	ENST00000466892					
LST1	ENST00000419073	ILMN_1717127	blood	4.51×10^{-6} (p-value)	Westra et al.	1
LST1	ENST00000433492					
LST1	ENST00000376089					
LST1	ENST00000376090					
LST1	ENST00000211921					
LST1	?	ENSG00000204482	blood	4.68×10^{-2} (q-value)	SeeQTL DB (HapMap)	1
?	?	ILMN_1893395	monocyte	8.32×10^{-30} (p-value)	Zeller et al.	1
LST1	ENST00000339530	ILMN_1718936	monocyte	1.96×10^{-28} (p-value)	Zeller et al.	1
LST1	ENST00000464526					
LST1	ENST00000303757					
LST1	ENST00000419073					
LST1	ENST00000376093					

LST1	ENST00000396112						
LST1	ENST00000490742						
LST1	ENST00000433492						
LST1	ENST00000438075						
LST1	ENST00000464044						
LST1	ENST00000396101						
?	?	ILMN_1721113	monocyte	3.76×10^{-14} (p-value)	Zeller et al.	1	
?	?	ILMN_1883997	monocyte	9.79×10^{-27} (p-value)	Zeller et al.	1	

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
?	?	ILMN_2159694	chr6	monocyte	3.67×10^{-14} (p-value)	Zeller et al.	1
?	?	ILMN_1715169	chr6	monocyte	1.33×10^{-15} (p-value)	Zeller et al.	1
HLA-DRB6	ENST00000437183	ILMN_2066060	chr6	monocyte	1.45×10^{-19} (p-value)	Zeller et al.	1
HLA-DRB6	ENST00000411500						
HLA-DRB6	ENST00000437650						
HLA-DRB5	ENST00000374975	ILMN_1697499	chr6	monocyte	9.17×10^{-25} (p-value)	Zeller et al.	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000487848 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, CTCF, PolII, TAF7, TAF1, H3K4me2, USF1, H3K4me3, DNase1
		HSMMtube	DNase1, H3K27me3, H2AZ, H3K4me2
		blood (K562)	Max, ZBTB7A, CTCF, H3K9ac, H2AZ, TAF1, Cfos, PolII, H3K4me2, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, H3K79me2, H3K4me3, H3K4me2
		liver (HepG2)	PolII, TAF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K4me3, H3K27me3, DNase1
		lung (IMR90)	H3K4me3, H3K4me2
		blood (GM12878)	PolII, H2AZ, DNase1, CTCF, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	H3K4me2, H3K4me3, DNase1, H3K9ac
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1
		NHLF	H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K27ac, H2AZ, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me3, H3K36me3, H3K9ac, H3K27ac, CTCF, H3K4me1
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2, PolII, DNase1
		monocytes (Monocytes-CD14+)	H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K4me3
		endothelium (HUVEC)	DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac, PolII, H3K36me3
		A549	H3K4me3, H3K4me2, H3K9ac
ENSR00000487850 (enhancer)	2	monocytes (Monocytes-CD14+)	H4K20me1
		HSMMtube	H3K27me3
		blood (GM12878)	H3K79me2
		blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MICB	upstream gene variant	760	ENST00000399150	NM_001289161.1	ENSP00000382103	2
MICB	upstream gene variant	789	ENST00000252229	NM_005931.4	ENSP00000252229	2
MICB	upstream gene variant	1404	ENST00000494577	?	?	6
Y_RNA	downstream gene variant, upstream gene variant	437	ENST00000383850	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
MICB	ENST00000538442	NM_001289160.1	ENSP00000442345	9
MICB	ENST00000399150	NM_001289161.1	ENSP00000382103	7
MICB	ENST00000252229	NM_005931.4	ENSP00000252229	7