SNiPAcard

Block annotations

Block info

genomic range	chr5:156,507,427-156,547,031
block size	39,605 bp
variant count	49 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.820 [-4.17 – 0.597]	**gene(s) hit or close-by**	CTB-120L21.1, HAVCR2
phastCons	μ = 0.018 [0 – 0.449]	**eQTL gene(s)**	HAVCR1, HAVCR2, MED7
GERP++	μ = -0.242 [-7.18 – 2.35]	**potentially regulated gene(s)**	ADAM19, CTB-120L21.1, CTB-4E7.1, HAVCR2
CADD score	μ = 2.684 [0.011 – 10.38]	**disease gene(s)**	–

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)
HAVCR2	missense variant	ENST00000307851	NM_032782.4	ENSP00000312002	R/L	cGg/cTg	tolerated	benign	1
HAVCR2	missense variant	ENST00000524219	?	ENSP00000430328	R/L	cGg/cTg	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HAVCR1	?	ENSG00000113249	visceral adipocytes	6.97×10^{-9} (p-value)	GTEx Portal V6	47
HAVCR1	?	ENSG00000113249	lung	8.25×10^{-11} (p-value)	GTEx Portal V6	47
HAVCR1	?	ENSG00000113249	tibial nerve	8.25×10^{-10} (p-value)	GTEx Portal V6	47
HAVCR1	?	ENSG00000113249	subcutaneous adipocytes	1.56×10^{-10} (p-value)	GTEx Portal V6	47
HAVCR2	ENST00000307851	ILMN_1693826	blood	5.47×10^{-9} (p-value)	MuTHER consortium	22
MED7	ENST00000524289	ILMN_2229170	b-cell	5.84×10^{-5} (p-value)	Fairfax et al.	4
MED7	ENST00000420343					
MED7	ENST00000286317					
HAVCR1	?	ENSG00000113249	coronary artery	4.35×10^{-6} (p-value)	GTEx Portal V6	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000681538	1	ENST00000307851, ENST00000521665, ENST00000524219	HAVCR2

ENCODE promoter-associated distal DHS (Enhancer)

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

ENCE00000429834	1	ENCP00000046421	HAVCR2
		ENCP00000046422	HAVCR2
ENCE00000429824	1	ENCP00000046420	CTB-120L21.1
ENCE00000430024	1	ENCP00000046430	CTB-4E7.1
		ENCP00000046453	ADAM19

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001294841 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		blood (GM12878)	PU1, PolII, H2AZ, H3K27ac, H3K4me2
ENSR00001699242 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K27ac, DNase1
		blood (GM12878)	H2AZ
		blood (K562)	H3K27me3
ENSR00001294842 (enhancer)	1	blood (GM12878)	H3K27ac

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTB-120L21.1	upstream gene variant, downstream gene variant	63	ENST00000517708	?	?	20
HAVCR2	downstream gene variant, upstream gene variant	151	ENST00000307851	NM_032782.4	ENSP00000312002	9
HAVCR2	downstream gene variant, upstream gene variant	435	ENST00000522593	?	ENSP00000430873	9
HAVCR2	downstream gene variant, upstream gene variant	1648	ENST00000521665	?	?	9
HAVCR2	downstream gene variant, upstream gene variant	179	ENST00000522902	?	?	22
HAVCR2	downstream gene variant	1606	ENST00000524219	?	ENSP00000430328	5
HAVCR2	downstream gene variant, upstream gene variant	446	ENST00000517358	?	?	24

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTB-120L21.1	ENST00000517708	?	?	15
HAVCR2	ENST00000307851	NM_032782.4	ENSP00000312002	26
HAVCR2	ENST00000522593	?	ENSP00000430873	26
HAVCR2	ENST00000524219	?	ENSP00000430328	37
HAVCR2	ENST00000521665	?	?	21

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
HAVCR2	ENST00000307851	NM_032782.4	ENSP00000312002	2

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
HAVCR2	ENST00000307851	NM_032782.4	ENSP00000312002	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CTB-120L21.1	ENST00000517708	?	4
HAVCR2	ENST00000521665	?	5

HAVCR2	ENST00000522902	?	3