Locus 62 (rs12651314)

SNiPAcard

Block annotations

Block info

genomic range	chr4:15,749,703-15,756,270
block size	6,568 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.788 [-2.591 – 0.219]	gene(s) hit or close-by	RP11-442P12.2
phastCons	μ = 0.005 [0 – 0.019]	eQTL gene(s)	BST1, CD38, RP11-115L11.1
GERP++	μ = 0.212 [-2.99 – 2.39]	potentially regulated gene(s)	–
CADD score	μ = 3.061 [2.042 – 4.102]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CD38	ENST00000502843	ILMN_2233783	adipocyte	9.04×10-6 (p-value)	MuTHER consortium	4
CD38	ENST00000226279					
RP11-115L11.1	?	ENSG00000214846	transformed fibroblasts	3.48×10-7 (p-value)	GTEx Portal V6	4
CD38	?	ENSG00000004468	thyroid	6.54×10-6 (p-value)	GTEx Portal V6	2
BST1	ENST00000382346	ILMN_1770161	monocyte	8.49×10-5 (p-value)	Fairfax et al.	1
BST1	ENST00000265016					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001237252 (promoter flanking region)	2	NHLF	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	Cjun, DNase1
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		nervous (NH-A)	H3K27ac, DNase1
		blood (DND-41)	H3K4me1, H3K36me3, H3K4me3, H3K9ac, H3K27ac, CTCF
ENSR00001683638 (CTCF binding site)	1	NHLF	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	Cjun, DNase1
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27ac, CTCF, H3K4me1, H3K9ac, H3K4me3
		nervous (NH-A)	H3K27ac, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-442P12.2	downstream gene variant	4498	ENST00000513715	?	?	1