Locus 444 (rs12079353)

SNiPAcard

Block annotations

Block info

genomic range	chr1:159,530,662-159,582,487
block size	51,826 bp
variant count	47 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.526 [-3.513 – 0.931]	gene(s) hit or close-by	APCS, OR10AE1P, OR10J6P, RP11-740C1.2
phastCons	μ = 0.043 [0 – 0.289]	eQTL gene(s)	CXCL10, CXCL11
GERP++	μ = -1.045 [-7.3 – 2.62]	potentially regulated gene(s)	APCS, CADM3, IGSF8, OR10J1, OR10J2P, OR10J5, OR10J6P, OR10J8P, RP11-550P17.5, SLAMF8
CADD score	μ = 2.474 [0.219 – 7.616]	disease gene(s)	–

Direct effect on regulation

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
CXCL10	?	ENSG00000169245	chr4	brain	5.31×10^{-5} (q-value)	SeeQTL DB (Myers et al.)	1
CXCL11	?	ENSG00000169248	chr4	brain	7.53×10^{-6} (q-value)	SeeQTL DB (Myers et al.)	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000040241	1	ENCP00000005063	OR10J1
		ENCP00000005060	OR10J8P
		ENCP00000005066	OR10J5
		ENCP00000005055	OR10J2P
ENCE00000040220	1	ENCP00000005069	OR10J6P
		ENCP00000005065	RP11-550P17.5
		ENCP00000005076	SLAMF8
ENCE00000040180	1	ENCP00000005050	CADM3
		ENCP00000005068	APCS
		ENCP00000005097	IGSF8

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000546120 (enhancer)	3	endothelium (HUVEC)	DNase1
		blood (DND-41)	H3K27me3
ENSR00001525176 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27ac, DNase1
		Osteobl	H3K27ac
		blood (DND-41)	H3K27me3
		muscle (HSMM)	H3K27ac, H3K4me2, H3K9ac, DNase1
		cervix (HeLa-S3)	Cjun
		endothelium (HUVEC)	Cmyc, H3K27ac, H3K4me1, Cjun, DNase1
		lung (IMR90)	DNase1, H3K4ac, H2BK120ac, H4K91ac

A549	DNase1, H3K27ac, H3K4me3
nervous (NH-A)	H3K9ac, H3K27ac, DNase1
skin (NHEK)	DNase1, H3K4me1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
APCS	upstream gene variant, downstream gene variant	100	ENST00000255040	NM_001639.3	ENSP00000255040	8
OR10AE1P	downstream gene variant, upstream gene variant	112	ENST00000606962	?	?	13
OR10J6P	upstream gene variant, downstream gene variant	118	ENST00000289731	?	?	8
RP11-740C1.2	downstream gene variant	2672	ENST00000435442	?	?	4

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
APCS	ENST00000255040	NM_001639.3	ENSP00000255040	P	ccA/ccG	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
OR10AE1P	ENST00000606962	?	1