Locus 220 (rs1461672)

SNiPAcard

Block annotations

Block info

genomic range	chr11:99,042,908-99,225,548
block size	182,641 bp
variant count	39 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.530 [-3.042 – 0.496]	gene(s) hit or close-by	CNTN5
phastCons	μ = 0.007 [0 – 0.089]	eQTL gene(s)	–
GERP++	μ = -0.741 [-7.09 – 3.04]	potentially regulated gene(s)	–
CADD score	μ = 2.568 [0.042 – 8.437]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
2-hydroxyisobutyrate	9.87×10-7	Metabolomics GWAS Server	24816252	4

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001606326 (promoter flanking region)	1	skin (NHDF-AD)	DNase1
ENSR00000567685 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1, Jund, Cjun
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		A549	DNase1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CNTN5	ENST00000528682	NM_001243270.1	ENSP00000436185	39
CNTN5	ENST00000527185	NM_001243271.1	ENSP00000433575	39
CNTN5	ENST00000524871	NM_014361.3	ENSP00000435637	39
CNTN5	ENST00000528727	?	?	39