Locus 92 (rs16971802)

SNiPAcard

Block annotations

Block info

genomic range	chr17:34,311,387-34,350,776
block size	39,390 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.338 [0.241 – 0.521]	gene(s) hit or close-by	CCL14, CCL15, CCL15-CCL14, CCL16, CTB-186H2.3
phastCons	μ = 0.140 [0.053 – 0.304]	eQTL gene(s)	–
GERP++	μ = 1.229 [0.158 – 4.05]	potentially regulated gene(s)	–
CADD score	μ = 5.712 [1.539 – 13.93]	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)
CCL14	missense variant	ENST00000614009	?	ENSP00000483186	E/K	Gag/Aag	?	?	1
CCL14	missense variant	ENST00000622526	NM_032962.4	ENSP00000479097	E/K	Gag/Aag	?	?	1
CCL14	missense variant	ENST00000618404	NM_032963.3	ENSP00000481023	E/K	Gag/Aag	?	?	1
CCL14	missense variant	ENST00000620991	?	ENSP00000484818	E/K	Gag/Aag	?	?	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CCL14	upstream gene variant	4965	ENST00000618404	NM_032963.3	ENSP00000481023	1
CCL14	upstream gene variant	4719	ENST00000614009	?	ENSP00000483186	1
CCL15	downstream gene variant	4717	ENST00000614368	?	ENSP00000484262	1
CCL16	upstream gene variant	2855	ENST00000611905	NM_004590.2	ENSP00000478024	1
CCL16	upstream gene variant	2890	ENST00000610493	?	ENSP00000478934	1
CTB-186H2.3	upstream gene variant, downstream gene variant	1234	ENST00000612584	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL15-CCL14	ENST00000610751	?	ENSP00000481940	2
CCL15-CCL14	ENST00000616694	?	ENSP00000481402	2
CTB-186H2.3	ENST00000617687	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)

CCL15-CCL14	ENST00000610751	?	ENSP00000481940	1
CCL15-CCL14	ENST00000616694	?	ENSP00000481402	1

Locus 92 (sentinel SNP rs16971802 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.