Locus 202 (rs1003645)

SNiPAcard

Block annotations

Block info

genomic range	chr17:34,319,442-34,351,034
block size	31,593 bp
variant count	27 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.338 [-2.377 – 0.716]	gene(s) hit or close-by	CCL15, CCL15-CCL14, CCL23, CTB-186H2.3, RP11-104J23.1, RP11-104J23.2
phastCons	μ = 0.102 [0 – 0.89]	eQTL gene(s)	CCL14, CCL15-CCL14, CCL23
GERP++	μ = 0.013 [-4.46 – 2.02]	potentially regulated gene(s)	–
CADD score	μ = 4.586 [0.258 – 15.76]	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)
CCL23	missense variant	ENST00000615050	NM_005064.4	ENSP00000481357	M/V	Atg/Gtg	tolerated	benign	1
CCL23	missense variant	ENST00000612516	NM_145898.2	ENSP00000484748	M/V	Atg/Gtg	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CCL23	ENST00000613876	ILMN_1764030	skin	1.79×10^{-16} (p-value)	MuTHER consortium	9
CCL23	ENST00000615050		adipocyte	2.68×10^{-10} (p-value)	MuTHER consortium	9
CCL23	ENST00000613876	ILMN_1686109	skin	2.24×10^{-19} (p-value)	MuTHER consortium	9
CCL23	ENST00000615050		adipocyte	1.16×10^{-10} (p-value)	MuTHER consortium	9
CCL23	ENST00000612516					
CCL14	ENST00000622526	ILMN_1740609	adipocyte	2.06×10^{-9} (p-value)	MuTHER consortium	9
CCL15-CCL14	ENST00000616694					
CCL15-CCL14	ENST00000610751					
CCL14	ENST00000618404					
CCL14	ENST00000614009					
CCL14	ENST00000620991					
CCL23	ENST00000613876	ILMN_1764030	blood	3.54×10^{-6} (p-value)	Westra et al.	10
CCL23	ENST00000615050					

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)

CCL15	downstream gene variant, upstream gene variant	473	ENST00000617897	NM_032965.4	ENSP00000484078	4
CCL15	downstream gene variant, upstream gene variant	364	ENST00000614368	?	ENSP00000484262	5
CCL15-CCL14	upstream gene variant	960	ENST00000610751	?	ENSP00000481940	1
CCL15-CCL14	upstream gene variant	890	ENST00000616694	?	ENSP00000481402	1
CCL23	downstream gene variant, upstream gene variant	224	ENST00000612516	NM_145898.2	ENSP00000484748	9
CCL23	downstream gene variant, upstream gene variant	218	ENST00000615050	NM_005064.4	ENSP00000481357	9
CCL23	downstream gene variant, upstream gene variant	261	ENST00000613876	?	ENSP00000479076	9
CTB-186H2.3	downstream gene variant	2136	ENST00000612584	?	?	2
RP11-104J23.1	upstream gene variant, downstream gene variant	1029	ENST00000619334	?	?	7
RP11-104J23.1	upstream gene variant, downstream gene variant	225	ENST00000617328	?	?	7
RP11-104J23.2	upstream gene variant, downstream gene variant	353	ENST00000617747	?	?	9

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL15	ENST00000617897	NM_032965.4	ENSP00000484078	2
CCL15-CCL14	ENST00000616694	?	ENSP00000481402	6
CCL15-CCL14	ENST00000610751	?	ENSP00000481940	6
CCL23	ENST00000612516	NM_145898.2	ENSP00000484748	6
CCL23	ENST00000615050	NM_005064.4	ENSP00000481357	6
RP11-104J23.1	ENST00000619334	?	?	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL15	ENST00000614368	?	ENSP00000484262	2
CCL23	ENST00000613876	?	ENSP00000479076	1