Locus 349 (rs8106657)

SNiPAcard

Block annotations

Block info

genomic range	chr19:7,779,733-7,789,796
block size	10,064 bp
variant count	11 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.795 [-1.99 – 0.075]	gene(s) hit or close-by	CLEC4G
phastCons	μ = 0.023 [0 – 0.077]	eQTL gene(s)	CD209, CLEC4G, FCER2, MCEMP1, TGFBR3L, XAB2
GERP++	μ = -0.107 [-0.858 – 0.13]	potentially regulated gene(s)	C19orf45, CCL25
CADD score	μ = 2.398 [0.127 – 5.545]	disease gene(s)	CD209

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CD209	DENGUE VIRUS, SUSCEPTIBILITY TODENGUE FEVER, SUSCEPTIBILITY TO, [...]	OMIM	MIM:614371
CD209	MYCOBACTERIUM TUBERCULOSIS, SUSCEPTIBILITY TOMYCOBACTERIUM TUBERCULOSIS, [...]	OMIM	MIM:607948

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TGFBR3L	?	ENSG00000260001	prostate	3.22×10-7 (p-value)	GTEx Portal V6	3
XAB2	?	ENSG00000076924	left ventricle	3.84×10-6 (p-value)	GTEx Portal V6	1
FCER2	?	ENSG00000104921	tibial nerve	3.82×10-6 (p-value)	GTEx Portal V6	1
FCER2	?	ENSG00000104921	subcutaneous adipocytes	9.02×10-7 (p-value)	GTEx Portal V6	2
FCER2	?	ENSG00000104921	visceral adipocytes	3.72×10-6 (p-value)	GTEx Portal V6	1
CLEC4G	ENST00000620128	ILMN_2193817	monocyte	4.97×10-7 (p-value)	Fairfax et al.	1
CLEC4G	ENST00000328853					
MCEMP1	ENST00000333598	ILMN_1762713	monocyte	7.18×10-5 (p-value)	Fairfax et al.	1
MCEMP1	ENST00000597445					
MCEMP1	ENST00000598851					
CLEC4G	ENST00000620128	ILMN_2193817	monocyte	7.61×10-45 (p-value)	Zeller et al.	1
CLEC4G	ENST00000328853					
CD209	ENST00000315599	ILMN_1676372	monocyte	1.36×10-17 (p-value)	Zeller 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)
ENCE00000222798	1	ENCP00000024006	C19orf45
ENCE00000223049	1	ENCP00000024014	CCL25

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CLEC4G	downstream gene variant	4048	ENST00000620128	?	ENSP00000483109	1
CLEC4G	downstream gene variant	4047	ENST00000328853	NM_198492.3, NM_001244856.1	ENSP00000327599	1