Locus 210 (rs626457)

SNiPAcard

Block annotations

Block info

genomic range	chr1:57,375,718-57,407,484
block size	31,767 bp
variant count	33 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.457 [-3.364 – 2.562]	gene(s) hit or close-by	C8A, C8B
phastCons	μ = 0.031 [0 – 0.981]	eQTL gene(s)	C1orf168
GERP++	μ = -0.849 [-7.05 – 4.16]	potentially regulated gene(s)	C1orf168, DAB1
CADD score	μ = 3.405 [0.041 – 17.59]	disease gene(s)	C8A, C8B

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
C8A	COMPLEMENT COMPONENT 8 DEFICIENCY, TYPE I	OMIM	MIM:613790
C8A	Immunodeficiency due to a late component of complements deficiency	OrphaNet	OrphaNet:169150
C8B	Immunodeficiency due to a late component of complements deficiency	OrphaNet	OrphaNet:169150

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
C1orf168	?	ENSG00000187889	thyroid	4.86×10-6 (p-value)	GTEx Portal V6	30

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000021496	1	ENCP00000002721	DAB1
		ENCP00000002712	C1orf168
ENCE00000021498	2	ENCP00000002721	DAB1
		ENCP00000002712	C1orf168

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001520596 (enhancer)	1	liver (HepG2)	H3K4me1, FOXA1, H3K9ac, H3K27ac
		HSMMtube	H3K27me3
		skin (NHDF-AD)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C8A	downstream gene variant	403	ENST00000361249	NM_000562.2	ENSP00000354458	2
C8B	downstream gene variant	154	ENST00000543257	NM_001278543.1	ENSP00000442548	8
C8B	downstream gene variant	154	ENST00000535057	NM_001278544.1	ENSP00000440113	8

C8B	downstream gene variant	152	ENST00000371237	NM_000066.3	ENSP00000360281	8
C8B	downstream gene variant, upstream gene variant	84	ENST00000465658	?	?	20

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C8A	ENST00000361249	NM_000562.2	ENSP00000354458	4
C8B	ENST00000371237	NM_000066.3	ENSP00000360281	18
C8B	ENST00000535057	NM_001278544.1	ENSP00000440113	18
C8B	ENST00000543257	NM_001278543.1	ENSP00000442548	18
C8B	ENST00000465658	?	?	3

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
C8B	ENST00000371237	NM_000066.3	ENSP00000360281	1
C8B	ENST00000535057	NM_001278544.1	ENSP00000440113	1
C8B	ENST00000543257	NM_001278543.1	ENSP00000442548	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
C8B	ENST00000465658	?	1