Locus 7 (rs12459419)

SNiPAcard

Block annotations

Block info

genomic range	chr19:51,727,962-51,737,991
block size	10,030 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.887 [-2.575 – 0.29]	gene(s) hit or close-by	CD33
phastCons	μ = 0.145 [0 – 0.744]	eQTL gene(s)	CD33, SIGLEC22P, SIGLECL1
GERP++	μ = -1.199 [-5.72 – 0.919]	potentially regulated gene(s)	SIGLEC12
CADD score	μ = 4.703 [0.881 – 14.88]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Alzheimer's disease (late onset)	$<3.00\times10^{-6}$	GWAS Catalog	24162737	1
Alzheimer's disease (late onset)	$<2.00\times10^{-9}$	GWAS Catalog	21460841	1

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)
CD33	missense variant	ENST00000262262	NM_001772.3	ENSP00000262262	V/A	gTc/gCc	?	?	1
CD33	missense variant	ENST00000391796	NM_001177608.1	ENSP00000375673	V/A	gTc/gCc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CD33	ENST00000262262	ILMN_1747622	blood	1.88×10^{-7} (p-value)	MuTHER consortium	3
CD33	ENST00000600557					
CD33	ENST00000601785					
CD33	ENST00000421133					
CD33	ENST00000598473					
SIGLEC22P	?	ENSG00000268849	lung	1.91×10^{-7} (p-value)	GTEx Portal V6	6
SIGLECL1	?	ENSG00000179213	testis	1.57×10^{-8} (p-value)	GTEx Portal V6	6

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)

ENCE00000232891	1	ENCP00000024645	SIGLEC12

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000217710 (promoter)	1	HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K4me3, H3K4me2, PolII, H2AZ, Gabp, H3K9ac, PU1, DNase1
		cervix (HeLa-S3)	PolII, H3K9ac, Gabp, H3K27me3, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		liver (HepG2)	H3K27me3, Gabp
		blood (GM12878)	Gabp
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CD33	upstream gene variant	392	ENST00000262262	NM_001772.3	ENSP00000262262	1
CD33	upstream gene variant	358	ENST00000436584	?	ENSP00000403331	1
CD33	upstream gene variant	358	ENST00000421133	NM_001082618.1	ENSP00000410126	1
CD33	upstream gene variant	425	ENST00000601785	?	?	1
CD33	upstream gene variant	745	ENST00000598473	?	?	2
CD33	upstream gene variant	392	ENST00000391796	NM_001177608.1	ENSP00000375673	1
CD33	upstream gene variant	4531	ENST00000600557	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CD33	ENST00000436584	?	ENSP00000403331	5
CD33	ENST00000391796	NM_001177608.1	ENSP00000375673	4
CD33	ENST00000601785	?	?	5
CD33	ENST00000262262	NM_001772.3	ENSP00000262262	4
CD33	ENST00000421133	NM_001082618.1	ENSP00000410126	5
CD33	ENST00000598473	?	?	4

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CD33	ENST00000601785	?	1