Locus 74 (rs4986791)

SNiPAcard

Block annotations

Block info

genomic range	chr9:120,467,576-120,475,602
block size	8,027 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.319 [-0.896 – 2.147]	gene(s) hit or close-by	RNU6-1082P, TLR4
phastCons	μ = 0.011 [0 – 0.051]	eQTL gene(s)	BRINP1, TLR4
GERP++	μ = -0.045 [-5.56 – 4.64]	potentially regulated gene(s)	–
CADD score	μ = 5.743 [1.266 – 12.31]	disease gene(s)	TLR4

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Clinvar: phenotype not specified	not provided	ClinVar	RCV000086999.1	1
Endotoxin hyporesponsiveness	pathogenic	ClinVar	RCV000007043.1	1
Colorectal cancer, susceptibility to	risk factor	ClinVar	RCV000007042.1	1
Macular degeneration, age-related, 10, susceptibility to	risk factor	ClinVar	RCV000007041.1	1
Endotoxin hyporesponsiveness	pathogenic	ClinVar	RCV000007040.1	1
?	HGMD curated	HGMD	CM001362	1
?	HGMD curated	HGMD	CM001361	1
ENDOTOXIN HYPORESPONSIVENESS	OMIM curated	OMIM	MIM:603030	2

Disease gene annotation

gene	trait	source DB	source entry/link
TLR4	TOLL-LIKE RECEPTOR 4	OMIM	MIM:603030
TLR4	MACULAR DEGENERATION, AGE-RELATED, 10	OMIM	MIM:611488
TLR4	Behcet disease	OrphaNet	OrphaNet:117

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)
TLR4	missense variant	ENST00000355622	NM_138554.4	ENSP00000363089	2	2			2
TLR4	missense variant	ENST00000394487	NM_003266.3	ENSP00000377997	2	2			2

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TLR4	ENST00000394487	ILMN_1706217	blood	6.25×10^{-6} (p-value)	Westra et al.	6

TLR4	ENST00000355622						
TLR4	ENST00000472304						
TLR4	ENST00000394487	ILMN_1706217	blood	7.20×10^{-5} (p-value)	MuTHER consortium	2	
TLR4	ENST00000355622						
TLR4	ENST00000472304						
BRINP1	ENST00000265922	ILMN_1741603	monocyte	7.17×10^{-5} (p-value)	Fairfax et al.	1	

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001312883 (promoter)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1, H3K4me2, H3K9ac
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		breast (HMEC)	H3K4me3, H3K4me2
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, H3K4me3, PolII
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	PolII, Cmyc, H3K27ac, H3K9ac, H3K4me3, Cjun, DNase1, H3K4me2
		liver (HepG2)	DNase1
		blood (GM12878)	PU1
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RNU6-1082P	downstream gene variant, upstream gene variant	724	ENST00000364574	?	?	6
TLR4	upstream gene variant, downstream gene variant	103	ENST00000490685	?	?	8

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TLR4	ENST00000355622	NM_138554.4	ENSP00000363089	6
TLR4	ENST00000472304	?	?	6
TLR4	ENST00000394487	NM_003266.3	ENSP00000377997	6

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RNU6-1082P	ENST00000364574	?	1
TLR4	ENST00000472304	?	3