

Block annotations

Block info

genomic range	chr9:120,467,576-120,475,602 <i>e!</i>
block size	8,027 bp
variant count	8 variants

Basic features

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

Trait annotations

Variant annotation

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

Disease gene annotation

gene	trait	source DB	source entry/link
TLR4 <i>e!</i>	TOLL-LIKE RECEPTOR 4	OMIM	MIM:603030 <i>OMIM</i> [®]
TLR4 <i>e!</i>	MACULAR DEGENERATION, AGE-RELATED, 10	OMIM	MIM:611488 <i>OMIM</i> [®]
TLR4 <i>e!</i>	Behcet disease	OrphaNet	OrphaNet:117 <i>orphanet</i>

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 <i>e!</i>	missense variant	ENST00000355622 <i>e!</i>	NM_138554.4	ENSP00000363089 <i>e!</i>	2	2			2
TLR4 <i>e!</i>	missense variant	ENST00000394487 <i>e!</i>	NM_003266.3	ENSP00000377997 <i>e!</i>	2	2			2

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TLR4 <i>e!</i>	ENST00000394487 <i>e!</i>	ILMN_1706217 <i>e!</i>	blood	6.25×10 ⁻⁶ (p-value)	Westra et al. <i>IQ</i>	6

TLR4 <i>e!</i>	ENST00000355622 <i>e!</i>						
TLR4 <i>e!</i>	ENST00000472304 <i>e!</i>						
TLR4 <i>e!</i>	ENST00000394487 <i>e!</i>	ILMN_1706217 <i>e!</i>	blood	7.20×10 ⁻⁵ (p-value)	MuTHER consortium <i>!M</i>		2
TLR4 <i>e!</i>	ENST00000355622 <i>e!</i>						
TLR4 <i>e!</i>	ENST00000472304 <i>e!</i>						
BRINP1 <i>e!</i>	ENST00000265922 <i>e!</i>	ILMN_1741603 <i>e!</i>	monocyte	7.17×10 ⁻⁵ (p-value)	Fairfax et al. <i>!M</i>		1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001312883 <i>e!</i>	1 (promoter)	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 <i>e!</i>	downstream gene variant, upstream gene variant	724	ENST00000364574 <i>e!</i>	?	?	6
TLR4 <i>e!</i>	upstream gene variant, downstream gene variant	103	ENST00000490685 <i>e!</i>	?	?	8

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TLR4 <i>e!</i>	ENST00000355622 <i>e!</i>	NM_138554.4	ENSP00000363089 <i>e!</i>	6
TLR4 <i>e!</i>	ENST00000472304 <i>e!</i>	?	?	6
TLR4 <i>e!</i>	ENST00000394487 <i>e!</i>	NM_003266.3	ENSP00000377997 <i>e!</i>	6

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RNU6-1082P <i>e!</i>	ENST00000364574 <i>e!</i>	?	1
TLR4 <i>e!</i>	ENST00000472304 <i>e!</i>	?	3

