Locus 157 (rs1126478)

SNiPAcard

Block annotations

Block info

genomic range	chr3:46,484,261-46,504,316
block size	20,056 bp
variant count	24 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.587 [-4.779 – 1.614]	gene(s) hit or close-by	LTF
phastCons	μ = 0.035 [0 – 0.761]	eQTL gene(s)	CCR3
GERP++	μ = -1.107 [-10.6 – 3.49]	potentially regulated gene(s)	CCR5, LTF
CADD score	μ = 3.650 [0.001 – 10.49]	disease gene(s)	LTF, CCR5

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM096382	1

Disease gene annotation

gene	trait	source DB	source entry/link
LTF	SPECIFIC GRANULE DEFICIENCY	OMIM	MIM:245480
CCR5	WEST NILE VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610379
CCR5	DIABETES MELLITUS, INSULIN-DEPENDENT, 22	OMIM	MIM:612522
CCR5	HUMAN IMMUNODEFICIENCY VIRUS TYPE 1, SUSCEPTIBILITY TO	OMIM	MIM:609423

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)
LTF	missense variant	ENST00000431944	?	ENSP00000395234	K/R	aAa/aGa	tolerated	benign	1
LTF	missense variant	ENST00000417439	?	ENSP00000405546	K/R	aAa/aGa	tolerated	benign	1
LTF	missense variant	ENST00000443496	?	ENSP00000397427	K/R	aAa/aGa	tolerated	benign	1
LTF	missense variant	ENST00000426532	NM_001199149.1	ENSP00000405719	K/R	aAa/aGa	tolerated	benign	1
LTF	missense variant	ENST00000231751	NM_002343.4	ENSP00000231751	K/R	aAa/aGa	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CCR3	ENST00000545097	ILMN_1763322	blood	5.43×10^{-7} (p-value)	Westra et al.	2
CCR3	ENST00000452454					
CCR3	ENST00000395942					
CCR3	ENST00000357422					

CCR3	ENST00000395940
CCR3	ENST00000475150

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000607402	1	ENST00000443496, ENST00000417439, ENST00000426532, ENST00000231751	LTF

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000036255		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000337735	1	ENCP00000036250	CCR5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001362955 (CTCF binding site)	2	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K4me1, CTCF, H3K27ac
		skin (NHDF-AD)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	CTCF
		liver (HepG2)	Rad21, CTCF
		lung (IMR90)	H3K27me3
		A549	CTCF
		skin (NHEK)	CTCF
ENSR00001362956 (CTCF binding site)	1	NHLF	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		cervix (HeLa-S3)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	CTCF
ENSR00001362957 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	CTCF, H3K27me3
		blood (K562)	H3K27me3, CTCF, DNase1
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF
		lung (IMR90)	H3K27me3, CTCF
		blood (GM12878)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	H3K27me3, CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF

	A549		CTCF, H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
LTF	downstream gene variant	579	ENST00000431944	?	ENSP00000395234	2
LTF	upstream gene variant	865	ENST00000493056	?	?	12
LTF	downstream gene variant, upstream gene variant	1552	ENST00000462667	?	?	6
LTF	downstream gene variant	2282	ENST00000498301	?	?	2

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
LTF	ENST00000417439	?	ENSP00000405546	N	aaT/aaC	1
LTF	ENST00000443496	?	ENSP00000397427	N	aaT/aaC	1
LTF	ENST00000426532	NM_001199149.1	ENSP00000405719	N	aaT/aaC	1
LTF	ENST00000231751	NM_002343.4	ENSP00000231751	N	aaT/aaC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LTF	ENST00000431944	?	ENSP00000395234	6
LTF	ENST00000478874	?	?	24
LTF	ENST00000417439	?	ENSP00000405546	23
LTF	ENST00000443496	?	ENSP00000397427	23
LTF	ENST00000426532	NM_001199149.1	ENSP00000405719	23
LTF	ENST00000462667	?	?	1
LTF	ENST00000231751	NM_002343.4	ENSP00000231751	23
LTF	ENST00000498301	?	?	6

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
LTF	ENST00000498301	?	1