Locus 427 (rs6808835)

SNiPAcard

Block annotations

Block info

genomic range	chr3:46,275,570-46,482,980
block size	207,411 bp
variant count	90 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.456 [-4.588 – 1.248]	gene(s) hit or close-by	CCR2, CCR3, CCR5, CCRL2, LTF, RP11-24F11.2, UQCRC2P1
phastCons	μ = 0.052 [0 – 0.59]	eQTL gene(s)	CCR3, LIMD1, LIMD1-AS1, LRRC2
GERP++	μ = -0.526 [-10.7 – 3.83]	potentially regulated gene(s)	AC104304.1, ALS2CL, CCR1, CCR2, CCR3, CCR5, CCR9, CCRL2, CXCR6, LRRC2, LTF, LZTFL1, PRSS44, PRSS50, RP11-24F11.2, XCR1
CADD score	μ = 3.969 [0.056 – 10.62]	disease gene(s)	LZTFL1, CCR5, LTF, CCR2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Cerebrospinal fluid levels of Alzheimer's disease-related proteins	$<2.00\times10^{-13}$	GWAS Catalog	25340798	1

Disease gene annotation

gene	trait	source DB	source entry/link
LZTFL1	BARDET-BIEDL SYNDROME 17	OMIM	MIM:615994
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
LTF	SPECIFIC GRANULE DEFICIENCY	OMIM	MIM:245480
CCR2	HUMAN IMMUNODEFICIENCY VIRUS TYPE 1, SUSCEPTIBILITY TO	OMIM	MIM:609423
LZTFL1	BARDET-BIEDL SYNDROME	OrphaNet	OrphaNet:110

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)
CCRL2	missense variant	ENST00000399036	NM_003965.4	ENSP00000381994	M/V	Atg/Gtg	?	?	1
CCRL2	missense variant	ENST00000357392	NM_001130910.1	ENSP00000349967	M/V	Atg/Gtg	?	?	1
CCRL2	missense variant	ENST00000400880	?	ENSP00000383677	M/V	Atg/Gtg	?	?	1
CCRL2	missense variant	ENST00000400882	?	ENSP00000383678	M/V	Atg/Gtg	?	?	1
CCRL2	missense variant	ENST00000433848	?	ENSP00000414957	M/V	Atg/Gtg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LIMD1-AS1	?	ENSG00000230530	tibial nerve	5.71×10-6 (p-value)	GTEx Portal V6	23
LIMD1	?	ENSG00000144791	tibial nerve	9.94×10-7 (p-value)	GTEx Portal V6	48
LRRC2	?	ENSG00000163827	transformed fibroblasts	4.59×10-8 (p-value)	GTEx Portal V6	84
LIMD1-AS1	?	ENSG00000230530	tibial artery	9.71×10-6 (p-value)	GTEx Portal V6	8
LIMD1	?	ENSG00000144791	tibial artery	2.16×10-6 (p-value)	GTEx Portal V6	31
CCR3	?	ENSG00000183625	blood	6.66×10-6 (p-value)	GTEx Portal V6	8

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000607398	1	ENST00000443496, ENST00000417439, ENST00000493056, ENST00000426532, ENST00000478874, ENST00000231751	LTF

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000036249		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000337650	1	ENCP00000036251	RP11-24F11.2 CCRL2
		ENCP00000036271	ALS2CL
		ENCP00000036243	CCR3
		ENCP00000036252	CCRL2
		ENCP00000036239	XCR1
		ENCP00000036242	CCR1
ENCE00000337509	1	ENCP00000036231	LZTFL1
		ENCP00000036227	LZTFL1
		ENCP00000036250	CCR5
		ENCP00000036230	CCR9
		ENCP00000036232	LZTFL1
		ENCP00000036234	CXCR6
ENCE00000337805	1	ENCP00000036249	CCR5
ENCE00000337856	1	ENCP00000036254	LTF
ENCE00000337664	1	ENCP00000036251	RP11-24F11.2 CCRL2
		ENCP00000036243	CCR3
		ENCP00000036271	ALS2CL
		ENCP00000036252	CCRL2
		ENCP00000036239	XCR1
		ENCP00000036242	CCR1
ENCE00000337563	3	ENCP00000036231	LZTFL1
		ENCP00000036250	CCR5
		ENCP00000036230	CCR9
		ENCP00000036232	LZTFL1
		ENCP00000036234	CXCR6
ENCE00000337711	1	ENCP00000036261	LRRC2

		ENCP00000036273	PRSS50
		ENCP00000036248	CCR2
		ENCP00000036263	AC104304.1
		ENCP00000036277	PRSS44
		ENCP00000036262	AC104304.1
		ENCP00000036244	CCR3
ENCE00000337859	1	ENCP00000036254	LTF
ENCE00000337713	1	ENCP00000036261	LRRC2
		ENCP00000036273	PRSS50
		ENCP00000036248	CCR2
		ENCP00000036263	AC104304.1
		ENCP00000036277	PRSS44
		ENCP00000036262	AC104304.1
		ENCP00000036244	CCR3
ENCE00000337671	1	ENCP00000036243	CCR3
		ENCP00000036271	ALS2CL
		ENCP00000036239	XCR1
		ENCP00000036242	CCR1
ENCE00000337715	2	ENCP00000036261	LRRC2
		ENCP00000036248	CCR2
		ENCP00000036263	AC104304.1
		ENCP00000036262	AC104304.1
		ENCP00000036277	PRSS44
		ENCP00000036244	CCR3
		ENCP00000036258	LTF
		ENCP00000036273	PRSS50

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001478830 (CTCF binding site)	1	cervix (HeLa-S3)	H3K27me3
		embryonic stem cell (H1ESC)	CTCF, Rad21
		endothelium (HUVEC)	H3K36me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
ENSR00001362902 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		blood (K562)	H3K27me3
ENSR00001362911 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K27ac, H3K4me1
		lung (IMR90)	H3K27me3
		blood (GM12878)	H3K4me1
		blood (DND-41)	H3K4me1, H3K27ac
		blood (K562)	H3K27me3
		skin (NHEK)	DNase1
ENSR00001674590 (enhancer)	1	NHLF	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K4me1, H3K27ac
		muscle (HSMM)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K27ac, H3K4me1
		blood (GM12878)	PU1, H3K4me1, H3K27ac
		lung (IMR90)	H3K27me3
		A549	H3K27me3
ENSR00001362921 (promoter flanking region)	1	HSMMtube	H3K27me3, DNase1
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1, H3K27me3

		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1, H3K27me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K4me3, H3K36me3
		endothelium (HUVEC)	H3K27me3
		blood (GM12878)	BATF, H3K4me1
		lung (IMR90)	H3K27me3
		A549	H3K27me3
		nervous (NH-A)	H3K27me3
ENSR00001478841 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, H3K27me3
		blood (K562)	H3K27me3, CTCF
		skin (NHDF-AD)	H3K27me3
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	H3K27me3
		nervous (NH-A)	H3K27me3
		skin (NHEK)	CTCF
		Osteobl	H3K27me3
		blood (DND-41)	H3K4me1, CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, H3K27me3
		A549	H3K27me3
ENSR00001478842 (promoter flanking region)	1	endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		lung (IMR90)	H3K27me3
		nervous (NH-A)	H3K27me3
		blood (DND-41)	H3K4me1, H3K27ac
		blood (K562)	H3K27me3
ENSR00001674606 (enhancer)	3	HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K4me1, H3K27ac
		blood (K562)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K27me3
		nervous (NH-A)	H3K27me3
ENSR00001478843 (promoter flanking region)	3	embryonic stem cell (H1ESC)	DNase1, H3K27me3
		blood (K562)	H3K27me3, DNase1
		blood (DND-41)	H3K4me1, H3K27ac
		skin (NHDF-AD)	H3K27me3
		breast (HMEC)	DNase1, H3K27ac
		cervix (HeLa-S3)	PolII, DNase1
		monocytes (Monocytes-CD14+)	DNase1
		endothelium (HUVEC)	Cjun, DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1
ENSR00001674607 (CTCF binding site)	1	cervix (HeLa-S3)	H3K27ac, DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	Max, DNase1, H3K27me3
		blood (DND-41)	H3K27ac, H3K4me1
		nervous (NH-A)	H3K27me3
ENSR00001362934 (promoter flanking region)	1	cervix (HeLa-S3)	PolII, H3K27ac, DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		lung (IMR90)	H3K27me3
		nervous (NH-A)	H3K27me3
		blood (K562)	H3K27me3, DNase1, Max
		blood (DND-41)	H3K4me1, H3K27ac
ENSR00001362946	1	HSMMtube	H3K27me3

(enhancer)		liver (HepG2)	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	DNase1, H2AZ
		blood (DND-41)	H3K4me1
ENSR00001362947 (open chromatin region)	3	embryonic stem cell (H1ESC)	DNase1
		blood (K562)	H3K27me3
ENSR00001362952 (open chromatin region)	2	A549	H3K27me3
		blood (K562)	H3K27me3
		skin (NHEK)	DNase1, H3K4me1, H3K4me2, H3K27ac
ENSR00001674619 (enhancer)	1	lung (IMR90)	H3K27me3
		blood (DND-41)	H3K4me1, H3K27ac
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CCR2	downstream gene variant	1779	ENST00000445132	NM_001123396.1	ENSP00000399285	1
CCR2	downstream gene variant	276	ENST00000292301	NM_001123041.2	ENSP00000292301	1
CCR2	downstream gene variant	269	ENST00000400888	?	ENSP00000383681	1
CCR2	downstream gene variant	2509	ENST00000465202	?	?	1
CCR2	downstream gene variant	679	ENST00000421659	?	ENSP00000396736	2
CCR3	upstream gene variant	2491	ENST00000457243	?	ENSP00000401822	2
CCR3	upstream gene variant	2461	ENST00000395940	NM_178329.2, NM_001837.3	ENSP00000379271	2
CCR3	upstream gene variant	2411	ENST00000545097	NM_178328.1, NM_001164680.1	ENSP00000441600	2
CCR3	upstream gene variant	2491	ENST00000484025	?	?	2
CCR3	upstream gene variant	2490	ENST00000452454	?	ENSP00000389336	2
CCR3	upstream gene variant	3681	ENST00000395942	?	ENSP00000379273	1
CCR3	upstream gene variant	1943	ENST00000475150	?	?	2
CCR5	upstream gene variant, downstream gene variant	684	ENST00000292303	NM_001100168.1, NM_000579.3	ENSP00000292303	4
CCR5	upstream gene variant	2500	ENST00000445772	?	ENSP00000404881	2
CCRL2	upstream gene variant, downstream gene variant	489	ENST00000400882	?	ENSP00000383678	15
CCRL2	upstream gene variant, downstream gene variant	411	ENST00000495870	?	?	16
CCRL2	upstream gene variant, downstream gene variant	488	ENST00000357392	NM_001130910.1	ENSP00000349967	15
CCRL2	upstream gene variant, downstream gene variant	488	ENST00000399036	NM_003965.4	ENSP00000381994	16
CCRL2	upstream gene variant, downstream gene variant	279	ENST00000400880	?	ENSP00000383677	15
CCRL2	upstream gene variant, downstream gene variant	662	ENST00000433848	?	ENSP00000414957	15
CCRL2	upstream gene variant, downstream gene variant	121	ENST00000441909	?	?	19
LTF	downstream gene variant	2014	ENST00000231751	NM_002343.4	ENSP00000231751	4
LTF	downstream gene variant	2388	ENST00000426532	NM_001199149.1	ENSP00000405719	4
LTF	downstream gene variant	2570	ENST00000443496	?	ENSP00000397427	3
LTF	downstream gene variant	2562	ENST00000417439	?	ENSP00000405546	3
LTF	downstream gene variant	1695	ENST00000493056	?	?	5
LTF	downstream gene variant	1756	ENST00000478874	?	?	5

RP11-24F11.2	downstream gene variant, upstream gene variant	1314	ENST00000451485	?	?	9
UQCRC2P1	downstream gene variant, upstream gene variant	1366	ENST00000417777	?	?	2

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
CCR2	ENST00000445132	NM_001123396.1	ENSP00000399285	T	acA/acG	1
CCRL2	ENST00000357392	NM_001130910.1	ENSP00000349967	G	ggT/ggG	1
CCRL2	ENST00000400882	?	ENSP00000383678	G	ggT/ggG	1
CCRL2	ENST00000433848	?	ENSP00000414957	G	ggT/ggG	1
CCRL2	ENST00000400880	?	ENSP00000383677	G	ggT/ggG	1
CCRL2	ENST00000399036	NM_003965.4	ENSP00000381994	G	ggT/ggG	1

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
LTF	ENST00000417439	?	ENSP00000405546	1
LTF	ENST00000443496	?	ENSP00000397427	1
LTF	ENST00000426532	NM_001199149.1	ENSP00000405719	1
LTF	ENST00000231751	NM_002343.4	ENSP00000231751	1
LTF	ENST00000493056	?	?	1
LTF	ENST00000478874	?	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCR2	ENST00000292301	NM_001123041.2	ENSP00000292301	1
CCR2	ENST00000400888	?	ENSP00000383681	1
CCR3	ENST00000452454	?	ENSP00000389336	6
CCR3	ENST00000457243	?	ENSP00000401822	6
CCR3	ENST00000484025	?	?	6
CCR3	ENST00000545097	NM_178328.1, NM_001164680.1	ENSP00000441600	6
CCR3	ENST00000357422	?	ENSP00000350003	11
CCR3	ENST00000395940	NM_178329.2, NM_001837.3	ENSP00000379271	6
CCRL2	ENST00000441909	?	?	5
LTF	ENST00000417439	?	ENSP00000405546	3
LTF	ENST00000443496	?	ENSP00000397427	3
LTF	ENST00000426532	NM_001199149.1	ENSP00000405719	3
LTF	ENST00000231751	NM_002343.4	ENSP00000231751	3
LTF	ENST00000493056	?	?	1
LTF	ENST00000478874	?	?	1
RP11-24F11.2	ENST00000451485	?	?	30

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)

gene	affected transcript	RefSeq id	protein	variant(s)
CCRL2	ENST00000357392	NM_001130910.1	ENSP00000349967	1
CCRL2	ENST00000400882	?	ENSP00000383678	1
CCRL2	ENST00000399036	NM_003965.4	ENSP00000381994	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CCR2	ENST00000465202	?	1