Locus 2 (rs4129267)

SNiPAcard

Block annotations

Block info

genomic range	chr1:154,395,839-154,428,283
block size	32,445 bp
variant count	27 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.413 [-2.774 – 1.935]	gene(s) hit or close-by	IL6R
phastCons	μ = 0.041 [0 – 0.875]	eQTL gene(s)	AQP10, IL6R, POU5F1P4, UBE2Q1
GERP++	μ = -0.744 [-5.29 – 2.48]	potentially regulated gene(s)	C1orf189, IL6R, UBAP2L
CADD score	μ = 3.576 [0.199 – 12.65]	disease gene(s)	IL6R

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Cerebrospinal fluid levels of Alzheimer's disease-related proteins	<6.00×10-63	GWAS Catalog	25340798	1
Fibrinogen	<6.00×10-27	GWAS Catalog	23969696	1
Asthma	<2.00×10-8	GWAS Catalog	21907864	1
C-reactive protein levels	<2.00×10-48	GWAS Catalog	21300955	1
C-reactive protein	<2.00×10-14	GWAS Catalog	19567438	1
Protein quantitative trait loci	<2.00×10-57	GWAS Catalog	18464913	1
PULMONARY FUNCTION	<7.00×10-6	GWAS Catalog	17903307	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CS034756	1

Disease gene annotation

gene	trait	source DB	source entry/link
IL6R	INTERLEUKIN 6, SERUM LEVEL OF, QUANTITATIVE TRAIT LOCUS	OMIM	MIM:614752
IL6R	SOLUBLE INTERLEUKIN-6 RECEPTOR, SERUM LEVEL OF, QUANTITATIVE [...]	OMIM	MIM:614689

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
AQP10	ENST00000324978	ILMN_2090004	blood	3.32×10-6 (p-value)	Westra et al.	1
AQP10	ENST00000484864					
UBE2Q1	ENST00000292211	ILMN_1776325	blood	5.52×10-7 (p-value)	Westra et al.	6
IL6R	ENST00000368485	ILMN_1754753	blood	1.45×10-6 (p-value)	Westra et al.	2
IL6R	ENST00000344086					
IL6R	?	ENSG00000160712	transverse colon	2.66×10-6 (p-value)	GTEx Portal V6	18
IL6R	?	ENSG00000160712	blood	1.18×10-6 (p-value)	GTEx Portal V6	19

IL6R	?	ENSG00000160712	tibial artery	6.60×10-6 (p-value)	GTEx Portal V6	16
POU5F1P4	?	ENSG00000237872	tibial artery	2.41×10-5 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000004651		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000037853	1	ENCP00000004625	C1orf189
ENCE00000037890	1	ENCP00000004651	IL6R
		ENCP00000004650	IL6R
		ENCP00000004633	UBAP2L

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000545357 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3, DNase1
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1, H3K9ac, H3K4me2, H3K27ac, H3K4me1
		muscle (HSMM)	DNase1
		liver (HepG2)	FOSL2, Jund, FOXA2, USF1, H3K4me1, p300, H3K4me2, FOXA1, H3K9ac, H3K27ac, Cmyc, DNase1
		lung (IMR90)	DNase1, H3K18ac, H3K27ac, H3K36me3
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K27me3
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K36me3, DNase1, H3K4me1
		NHLF	DNase1, H3K4me1, H3K27ac, H3K36me3
		Osteobl	H3K27ac, H3K4me2
		blood (DND-41)	H3K27me3
		breast (HMEC)	DNase1, H3K27ac, H3K9ac, H3K4me1
		cervix (HeLa-S3)	DNase1, H3K36me3, H3K9ac, Jund, H3K4me2, H3K27ac, H3K4me1, Max, Cmyc
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K9ac, H4K20me1, H3K36me3
		endothelium (HUVEC)	Cjun, Max, H3K4me1, DNase1
		A549	H3K4me3, H3K4me2, DNase1, H3K27ac
ENSR00000545365 (promoter flanking region)	2	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3, DNase1
		blood (K562)	H3K27me3, Egr1, Max
		skin (NHDF-AD)	H3K4me1, DNase1, H3K9ac, H3K27ac
		muscle (HSMM)	DNase1
		liver (HepG2)	H3K36me3, DNase1
		lung (IMR90)	DNase1, H3K36me3
		nervous (NH-A)	DNase1, H3K27me3
		skin (NHEK)	DNase1, H3K4me1, H3K27ac, H3K9ac, H3K36me3
		NHLF	DNase1, H3K27ac, H3K36me3
		Osteobl	H3K27ac, H3K4me2
		blood (DND-41)	H3K27me3
		breast (HMEC)	DNase1, H3K27ac
		cervix (HeLa-S3)	DNase1, Jund, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K9ac, H3K36me3
		endothelium (HUVEC)	Cjun
ENSR00000545369 (promoter flanking region)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	H3K27me3, DNase1
		blood (K562)	Egr1, CTCF, DNase1, H3K27me3
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	CTCF

		lung (IMR90)	H3K27me3, H3K36me3, CTCF
		nervous (NH-A)	DNase1
		skin (NHEK)	CTCF, H3K36me3
		NHLF	DNase1, H3K36me3
		Osteobl	H3K4me2
		blood (DND-41)	H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	H3K36me3, H3K27ac, H4K20me1
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF
ENSR00000545370	4	NHLF	H3K36me3, DNase1, H3K4me1, H3K27ac
(promoter flanking region)		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		lung (IMR90)	DNase1, H3K18ac, H3K27ac, H3K4me2, H3K27me3, H3K36me3
		blood (GM12878)	Tcf12, DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K36me3
ENSR00000545373	2	monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K36me3
(promoter flanking region)		HSMMtube	H3K27me3
		lung (IMR90)	H3K36me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHEK)	H3K4me1, H3K36me3
		skin (NHDF-AD)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IL6R	upstream gene variant	67	ENST00000507256	?	?	3
IL6R	upstream gene variant	321	ENST00000502679	?	?	2
IL6R	upstream gene variant, downstream gene variant	473	ENST00000515190	?	ENSP00000423036	3
IL6R	downstream gene variant	1399	ENST00000622330	NM_001206866.1	ENSP00000477739	4
IL6R	downstream gene variant	360	ENST00000512471	?	ENSP00000423184	4
IL6R	downstream gene variant	3001	ENST00000476006	?	ENSP00000423668	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL6R	ENST00000507256	?	?	23
IL6R	ENST00000476006	?	ENSP00000423668	23
IL6R	ENST00000344086	NM_181359.2	ENSP00000340589	27
IL6R	ENST00000368485	NM_000565.3	ENSP00000357470	27
IL6R	ENST00000512471	?	ENSP00000423184	3
IL6R	ENST00000515190	?	ENSP00000423036	23
IL6R	ENST00000622330	NM_001206866.1	ENSP00000477739	6
IL6R	ENST00000502679	?	?	1