Locus 436 (rs4745216)

SNiPAcard

Block annotations

Block info

genomic range	chr9:75,714,313-75,789,730
block size	75,418 bp
variant count	21 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.458 [-2.261 – 0.537]	gene(s) hit or close-by	ANXA1
phastCons	μ = 0.037 [0 – 0.534]	eQTL gene(s)	ANXA1
GERP++	μ = -1.387 [-9 – 2.1]	potentially regulated gene(s)	ANXA1
CADD score	μ = 3.607 [0.004 – 11.09]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Schizophrenia, bipolar disorder and depression (combined)	$<3.00\times10^{-6}$	GWAS Catalog	20713499	1
2-methoxyacetaminophen sulfate*	5.59×10^{-6}	Metabolomics GWAS Server	24816252	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ANXA1	?	ENSG00000135046	blood	1.37×10^{-3} (q-value)	SeeQTL DB (HapMap)	3

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
CJUN	MA0489.1	10	no	0.000	1
CFOS	MA0476.1	6	no	0.000	1
JUND	MA0491.1	6	no	0.000	1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000543116	1	ENCP00000058973	ANXA1
		ENCP00000058972	ANXA1
		ENCP00000058974	ANXA1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001305737 (promoter flanking region)	3	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		Osteobl	H3K27ac

		blood (DND-41)	H3K4me1, H3K27ac
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K36me3
		breast (HMEC)	H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, Jund, H3K27ac
		endothelium (HUVEC)	DNase1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		A549	H3K4me3, DNase1, H3K36me3
		skin (NHEK)	H3K27ac, DNase1
ENSR00001305741	2	embryonic stem cell (H1ESC)	DNase1
(promoter flanking region)		HSMMtube	H3K9ac, H3K4me2, H3K27ac, DNase1
		blood (K562)	DNase1, H3K27ac, MEF2A, H3K9ac, H2AZ, H3K4me3, H3K4me2
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H3K27ac, H3K4me2, H3K9ac, DNase1
		liver (HepG2)	H3K27me3
		blood (GM12878)	PolII, H2AZ, H3K4me3, H3K27ac, H3K9ac
		lung (IMR90)	DNase1, H3K4me2, H3K4me3
		nervous (NH-A)	H3K4me2, H3K4me3, H3K27ac, DNase1, H3K9ac
		skin (NHEK)	DNase1, H3K4me1, H3K27ac, H3K9ac, H3K4me2
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K4me2, H3K27ac
		blood (DND-41)	H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	DNase1, H3K27ac, H3K9ac, H3K4me2
		cervix (HeLa-S3)	H3K9ac, DNase1, H3K4me2, H3K27ac, TAF1, PolII
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K9ac, H3K27ac, PolII, H3K4me2, DNase1, Cjun
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac
ENSR00001730160	1	cervix (HeLa-S3)	CTCF, DNase1, Cjun, Jund
(CTCF binding site)		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		A549	H3K4me3, H3K4me2, DNase1
ENSR00001471274	1	cervix (HeLa-S3)	Jund, DNase1, CTCF, Cjun
(promoter flanking region)		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		A549	H3K4me3, H3K4me2, DNase1
		skin (NHEK)	DNase1, H3K27ac, H3K4me2
		breast (HMEC)	DNase1, H3K27ac
ENSR00001305755	1	embryonic stem cell (H1ESC)	DNase1
(promoter)		HSMMtube	H3K36me3, DNase1, H3K4me3, H3K27ac, H3K4me2, H3K79me2, H3K9ac
		blood (K562)	PolII, H3K4me2, H3K36me3, H3K4me3, H2AZ, H3K79me2, Jund, Ini1, Brg1, DNase1, H3K27ac, H3K9ac
		skin (NHDF-AD)	H3K36me3, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	DNase1, H3K36me3, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		blood (GM12878)	IRF4, BATF, PU1, Cjun
		lung (IMR90)	DNase1, H3K79me2, H3K4me2, H3K36me3, H3K4me3, H3K9ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	DNase1, H3K27ac, H3K9ac, PolII, H3K4me2, H3K4me3, H3K36me3
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K27ac, H3K4me3, H3K4me2, H3K36me3
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me1
		breast (HMEC)	H3K4me2, H3K36me3, DNase1, H3K27ac, H3K4me3, H3K9ac
		cervix (HeLa-S3)	DNase1, H3K36me3, PolII, Cjun, H3K9ac, Jund, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, Cfos
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	Cjun, DNase1, H3K4me2, H3K4me3, H3K9ac, PolII, H3K36me3
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00001305768	1	embryonic stem cell (H1ESC)	USF1, DNase1
(CTCF binding site)		HSMMtube	H3K36me3
		blood (K562)	PolII, Brg1
		skin (NHDF-AD)	H3K36me3
		muscle (HSMM)	H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	PolII, H3K36me3

skin (NHEK)	PolII, H3K36me3
NHLF	H3K36me3
Osteobl	H3K36me3
blood (DND-41)	H3K36me3
breast (HMEC)	H3K36me3
cervix (HeLa-S3)	H3K36me3, PolII
monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
endothelium (HUVEC)	H3K36me3
A549	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ANXA1	upstream gene variant, downstream gene variant	744	ENST00000257497	NM_000700.1	ENSP00000257497	5
ANXA1	upstream gene variant, downstream gene variant	2167	ENST00000415424	?	ENSP00000414013	5
ANXA1	downstream gene variant	744	ENST00000376911	?	ENSP00000366109	3
ANXA1	upstream gene variant, downstream gene variant	169	ENST00000456643	?	ENSP00000412489	4
ANXA1	upstream gene variant, downstream gene variant	744	ENST00000491192	?	?	5
ANXA1	upstream gene variant, downstream gene variant	2216	ENST00000468639	?	?	5
ANXA1	downstream gene variant	3268	ENST00000495713	?	?	1

Putative effect on transcript

5'-UTR variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
ANXA1	ENST00000257497	NM_000700.1	ENSP00000257497	1
ANXA1	ENST00000415424	?	ENSP00000414013	1
ANXA1	ENST00000456643	?	ENSP00000412489	1

Non-coding exon variant (splice region)

gene	affected transcript	RefSeq id	variant(s)
ANXA1	ENST00000468639	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ANXA1	ENST00000257497	NM_000700.1	ENSP00000257497	3
ANXA1	ENST00000376911	?	ENSP00000366109	3
ANXA1	ENST00000456643	?	ENSP00000412489	1
ANXA1	ENST00000495713	?	?	3
ANXA1	ENST00000489109	?	?	3