SNiPAcard

Block annotations

Block info

genomic range	chr4:74,735,524-74,837,932
block size	102,409 bp
variant count	53 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.290 [-3.136 – 1.95]	**gene(s) hit or close-by**	CXCL1, CXCL1P, HNRNPA1P55
phastCons	μ = 0.088 [0 – 0.99]	**eQTL gene(s)**	CXCL6, PF4
GERP++	μ = -0.443 [-6.02 – 3.73]	**potentially regulated gene(s)**	AFP, ALB, RP11-622A1.1
CADD score	μ = 3.025 [0.058 – 10.27]	**disease gene(s)**	ALB, AFP

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ALB	HYPERTHYROXINEMIA, FAMILIAL DYSALBUMINEMIC	OMIM	MIM:615999
ALB	ANALBUMINEMIA	OMIM	MIM:616000
AFP	ALPHA-FETOPROTEIN, HEREDITARY PERSISTENCE OF	OMIM	MIM:615970
AFP	ALPHA-FETOPROTEIN DEFICIENCY	OMIM	MIM:615969
ALB	Congenital analbuminemia	OrphaNet	OrphaNet:86816
AFP	Congenital deficiency in alpha-fetoprotein	OrphaNet	OrphaNet:168612
AFP	Hereditary persistence of alpha-fetoprotein	OrphaNet	OrphaNet:168615

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PF4	?	ENSG00000163737	transformed fibroblasts	4.04×10^{-12} (p-value)	GTEx Portal V6	30
PF4	?	ENSG00000163737	tibial artery	1.00×10^{-6} (p-value)	GTEx Portal V6	3
CXCL6	?	ENSG00000124875	ovary	2.91×10^{-6} (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000382127	1	ENCP00000041354	AFP
		ENCP00000041348	ALB
		ENCP00000041359	RP11-622A1.1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001242854 (promoter)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, H3K4me2, H3K4me3, DNase1
		HSMMtube	H2AZ, H3K27me3
		blood (K562)	CTCF, Rad21, H3K9ac, H2AZ, H3K4me2, H3K4me3, H3K27me3

		skin (NHDF-AD)	H3K4me3, H3K4me2
		muscle (HSMM)	H3K4me3, H2AZ
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3, H3K4me3
		blood (GM12878)	H3K27me3
		nervous (NH-A)	H3K4me2, H3K27me3
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1, H3K4me1
		NHLF	H3K9ac, H3K4me3
		Osteobl	H3K4me2, H3K4me3, H2AZ
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K27ac, H3K9ac, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K4me3, PolII
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K36me3
ENSR00001242858 (enhancer)	1	monocytes (Monocytes-CD14+)	DNase1, H3K27ac
		endothelium (HUVEC)	Cjun
		HSMMtube	H3K27me3
		lung (IMR90)	H3K27me3
ENSR00001685780 (open chromatin region)	1	lung (IMR90)	H3K27me3
ENSR00001242860 (promoter flanking region)	1	cervix (HeLa-S3)	DNase1
		HSMMtube	H3K27me3
		blood (K562)	DNase1
		blood (DND-41)	H3K27me3
ENSR00001432046 (enhancer)	1	embryonic stem cell (H1ESC)	Rad21
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		nervous (NH-A)	H3K27me3
		muscle (HSMM)	DNase1
ENSR00001432047 (promoter flanking region)	1	HSMMtube	H3K27me3, DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00001685781 (enhancer)	1	HSMMtube	H3K27me3
ENSR00001685782 (CTCF binding site)	1	NHLF	DNase1
		nervous (NH-A)	DNase1
		blood (K562)	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00001432049 (promoter flanking region)	3	NHLF	DNase1
		HSMMtube	H3K27me3
		nervous (NH-A)	DNase1
		blood (K562)	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00001685783 (CTCF binding site)	1	NHLF	DNase1
		HSMMtube	H3K27me3
		skin (NHDF-AD)	DNase1
ENSR00001432050 (promoter flanking region)	1	NHLF	DNase1
		HSMMtube	H3K27me3
		blood (K562)	DNase1
		nervous (NH-A)	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00001432054 (promoter flanking region)	2	NHLF	DNase1
		HSMMtube	H3K27me3, DNase1
		Osteobl	H3K27ac
		skin (NHDF-AD)	DNase1
		breast (HMEC)	DNase1
		muscle (HSMM)	DNase1, H3K36me3
		cervix (HeLa-S3)	DNase1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1

		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1, DNase1
ENSR00001432057 (CTCF binding site)	1	embryonic stem cell (H1ESC)	CTCF, Rad21
		Osteobl	CTCF
		blood (K562)	Rad21
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		lung (IMR90)	CTCF
		A549	CTCF
ENSR00001242869 (enhancer)	1	embryonic stem cell (H1ESC)	CTCF, Rad21
		HSMMtube	H3K27me3
		Osteobl	CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	CTCF
		lung (IMR90)	CTCF
		A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CXCL1	downstream gene variant	2957	ENST00000509101	?	?	2
CXCL1	downstream gene variant	1892	ENST00000395761	NM_001511.3	ENSP00000379110	2
CXCL1P	downstream gene variant, upstream gene variant	2721	ENST00000502804	?	?	3
HNRNPA1P55	downstream gene variant, upstream gene variant	667	ENST00000505935	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CXCL1	ENST00000509101	?	?	1
CXCL1	ENST00000395761	NM_001511.3	ENSP00000379110	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
HNRNPA1P55	ENST00000505935	?	1