Locus 384 (rs41265665)

SNiPAcard

Block annotations

Block info

genomic range	chr4:74,331,266-74,483,608
block size	152,343 bp
variant count	52 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.421 [-3.218 – 1.469]	gene(s) hit or close-by	AFM, RASSF6, RP11-622A1.1, RP11-622A1.2
phastCons	μ = 0.062 [0 – 0.979]	eQTL gene(s)	RASSF6
GERP++	μ = -0.510 [-6.47 – 3.2]	potentially regulated gene(s)	AFM, AFP
CADD score	μ = 3.743 [0.044 – 14.92]	disease gene(s)	AFP

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
AFP	ALPHA-FETOPROTEIN, HEREDITARY PERSISTENCE OF	OMIM	MIM:615970
AFP	ALPHA-FETOPROTEIN DEFICIENCY	OMIM	MIM:615969
AFP	Congenital deficiency in alpha-fetoprotein	OrphaNet	OrphaNet:168612
AFP	Hereditary persistence of alpha-fetoprotein	OrphaNet	OrphaNet:168615

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)
AFM	missense variant	ENST00000226355	NM_001133.2	ENSP00000226355	H/R	cAt/cGt	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RASSF6	ENST00000395777	ILMN_1657381	b-cell	5.47×10^{-18} (p-value)	Fairfax et al.	1
RASSF6	ENST00000307439					
RASSF6	ENST00000512591					

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000041356		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000382154	1	ENCP00000041353	AFP

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001242774 (enhancer)	1	skin (NHEK)	H3K36me3
ENSR00001432007 (promoter flanking region)	6	embryonic stem cell (H1ESC)	DNase1
		blood (DND-41)	H3K4me1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		breast (HMEC)	H3K27ac, H3K4me1
		cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	DNase1
		lung (IMR90)	DNase1
		blood (GM12878)	Cjun, H3K4me3, PU1, PolII, BATF, DNase1, IRF4, BCL11A, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1, H3K27ac, H3K4me2
ENSR00001685745 (CTCF binding site)	4	embryonic stem cell (H1ESC)	DNase1
		blood (DND-41)	H3K4me1
		skin (NHDF-AD)	DNase1
		breast (HMEC)	H3K27ac, H3K4me1
		muscle (HSMM)	DNase1
		liver (HepG2)	DNase1
		lung (IMR90)	DNase1
		blood (GM12878)	PU1, PolII, BATF, DNase1, IRF4, BCL11A, H3K27ac, H3K4me2, Cjun
		skin (NHEK)	H3K4me1, H3K27ac, H3K4me2
ENSR00001685748 (enhancer)	1	A549	H3K36me3
		breast (HMEC)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AFM	upstream gene variant, downstream gene variant	219	ENST00000505794	?	?	5
AFM	upstream gene variant, downstream gene variant	184	ENST00000226355	NM_001133.2	ENSP00000226355	10
RP11-622A1.1	upstream gene variant, downstream gene variant	3438	ENST00000511117	?	?	3
RP11-622A1.2	upstream gene variant, downstream gene variant	1361	ENST00000508147	?	?	6
RP11-622A1.2	upstream gene variant, downstream gene variant	1326	ENST00000504368	?	?	6
RP11-622A1.2	upstream gene variant, downstream gene variant	1562	ENST00000502291	?	?	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AFM	ENST00000505794	?	?	3
AFM	ENST00000226355	NM_001133.2	ENSP00000226355	10
RASSF6	ENST00000307439	NM_177532.4	ENSP00000303877	15
RASSF6	ENST00000342081	NM_201431.2	ENSP00000340578	15
RASSF6	ENST00000335049	NM_001270392.1	ENSP00000335582	15
RASSF6	ENST00000512591	?	?	1
RASSF6	ENST00000395777	NM_001270391.1	ENSP00000379123	15
RP11-622A1.2	ENST00000508147	?	?	8
RP11-622A1.2	ENST00000502291	?	?	8
RP11-622A1.2	ENST00000504368	?	?	9

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AFM	ENST00000505794	?	2
RP11-622A1.2	ENST00000502291	?	1