Locus 393 (rs10473230)

SNiPAcard

Block annotations

Block info

genomic range	chr5:40,959,959-40,986,374
block size	26,416 bp
variant count	17 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.031 [-2.44 – 3.368]	gene(s) hit or close-by	C7, RP11-301A5.2
phastCons	μ = 0.032 [0 – 0.34]	eQTL gene(s)	C6
GERP++	μ = -0.511 [-7.22 – 4.05]	potentially regulated gene(s)	C7, C7
CADD score	μ = 5.222 [0.192 – 14.47]	disease gene(s)	C7, C6

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
C7	COMPLEMENT COMPONENT 7 DEFICIENCY	OMIM	MIM:610102
C6	COMPLEMENT COMPONENT 6 DEFICIENCY	OMIM	MIM:612446
C7	Immunodeficiency due to a late component of complements deficiency	OrphaNet	OrphaNet:169150
C6	Immunodeficiency due to a late component of complements deficiency	OrphaNet	OrphaNet:169150

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)
C7	missense variant	ENST00000313164	NM_000587.2	ENSP00000322061	S/T	Tct/Act	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
C6	ENST00000263413	ILMN_1688242	adipocyte	4.98×10^{-5} (p-value)	MuTHER consortium	1
C6	ENST00000337836					

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000043959		C7

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000407549	1	ENCP00000043958	C7
			C7

	ENCP00000043961	C7 C7

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001692910 (enhancer)	1	embryonic stem cell (H1ESC)	H3K27me3
ENSR00001408896 (enhancer)	2	cervix (HeLa-S3)	H3K27me3
		NHLF	DNase1
		HSMMtube	H3K27me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C7	upstream gene variant, downstream gene variant	163	ENST00000464864	?	?	7
C7	upstream gene variant, downstream gene variant	1145	ENST00000486779	?	?	6
C7	downstream gene variant	926	ENST00000513922	?	?	2
C7	downstream gene variant	1579	ENST00000494960	?	?	5
C7	downstream gene variant	16	ENST00000313164	NM_000587.2	ENSP00000322061	4
RP11-301A5.2	downstream gene variant	625	ENST00000504890	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C7	ENST00000513922	?	?	1
C7	ENST00000486779	?	?	2
C7	ENST00000313164	NM_000587.2	ENSP00000322061	10
C7	ENST00000494960	?	?	3
C7	ENST00000464864	?	?	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
C7	ENST00000313164	NM_000587.2	ENSP00000322061	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
C7	ENST00000486779	?	1