Locus 246 (rs13157656)

SNiPAcard

Block annotations

Block info

genomic range	chr5:40,960,166-40,985,653
block size	25,488 bp
variant count	23 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.153 [-2.108 – 1.183]	gene(s) hit or close-by	C7, RP11-301A5.2
phastCons	μ = 0.069 [0 – 0.496]	eQTL gene(s)	–
GERP++	μ = -0.671 [-4.69 – 3.22]	potentially regulated gene(s)	–
CADD score	μ = 3.816 [0.138 – 11.55]	disease gene(s)	C7

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
C7	COMPLEMENT COMPONENT 7 DEFICIENCY	OMIM	MIM:610102
C7	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	P/T	Cca/Aca	?	?	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001692911 (enhancer)	2	HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
ENSR00001692912 (enhancer)	1	cervix (HeLa-S3)	H3K27me3
		NHLF	DNase1
		lung (IMR90)	DNase1, H3K18ac, H2BK120ac, H4K91ac
		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	252	ENST00000494960	?	?	8
C7	upstream gene variant, downstream gene variant	148	ENST00000486779	?	?	8
C7	upstream gene variant	134	ENST00000513922	?	?	5
C7	upstream gene variant, downstream gene variant	588	ENST00000464864	?	?	10
C7	downstream gene variant	50	ENST00000313164	NM_000587.2	ENSP00000322061	2
RP11-301A5.2	downstream gene variant, upstream gene variant	163	ENST00000504890	?	?	13

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C7	ENST00000494960	?	?	5
C7	ENST00000313164	NM_000587.2	ENSP00000322061	18
C7	ENST00000486779	?	?	10
C7	ENST00000513922	?	?	5

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