

# SNiPACard

## Block annotations

### Block info




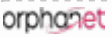
genomic range	chr5:40,959,959-40,986,374 <i>e!</i>
block size	26,416 bp
variant count	17 variants

### Basic features

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

## Trait annotations

### Disease gene annotation

gene	trait	source DB	source entry/link
C7 <i>e!</i>	COMPLEMENT COMPONENT 7 DEFICIENCY	OMIM	MIM:610102 
C6 <i>e!</i>	COMPLEMENT COMPONENT 6 DEFICIENCY	OMIM	MIM:612446 
C7 <i>e!</i>	Immunodeficiency due to a late component of complements deficiency	OrphaNet	OrphaNet:169150 
C6 <i>e!</i>	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 <i>e!</i>	missense variant	ENST00000313164 <i>e!</i>	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 <i>e!</i>	ENST00000263413 <i>e!</i>	ILMN_1688242 <i>e!</i>	adipocyte	4.98×10 <sup>-5</sup> (p-value)	MuTHER consortium 	1
C6 <i>e!</i>	ENST00000337836 <i>e!</i>					

## Putative effect on regulation

### ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000043959 <i>e!</i>	C7 <i>e!</i>	

### ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000407549 <i>e!</i>	1	ENCP00000043958	C7 <i>e!</i> C7 <i>e!</i>

## Regulatory feature cluster

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

## Variation proximal to gene

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

## Putative effect on transcript

## Intron variant

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

## 3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
C7 <i>e!</i>	ENST00000313164 <i>e!</i>	NM_000587.2	ENSP00000322061 <i>e!</i>	2

## Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
C7 <i>e!</i>	ENST00000486779 <i>e!</i>	? ?	1

