Locus 183 (rs11957503)

SNiPAcard

Block annotations

Block info

genomic range	chr5:35,827,646-35,883,804
block size	56,159 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.527 [-1.238 – 0.295]	gene(s) hit or close-by	CTD-2113L7.1, IL7R
phastCons	μ = 0.001 [0 – 0.005]	eQTL gene(s)	–
GERP++	μ = -0.467 [-2.75 – 1.27]	potentially regulated gene(s)	–
CADD score	μ = 2.893 [0.346 – 5.497]	disease gene(s)	IL7R

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Breast neoplasms	6.57×10-5	dbGaP	pha002853	dbGaP	1

Disease gene annotation

gene	trait	source DB	source entry/link	
IL7R	T-B+ severe combined immunodeficiency due to IL-7Ralpha deficiency	OrphaNet	OrphaNet:169154	orphanet
IL7R	OMENN SYNDROME	OrphaNet	OrphaNet:39041	orphanet

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001278142 (promoter flanking region)	4	NHLF	DNase1, H3K27ac
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K27ac
		cervix (HeLa-S3)	Cjun, Jund, H3K27ac, H3K4me1, Cfos, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	DNase1
		endothelium (HUVEC)	Cjun
		liver (HepG2)	DNase1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTD-2113L7.1	upstream gene variant	526	ENST00000510433	?	?	3
IL7R	downstream gene variant	3471	ENST00000303115	NM_002185.3	ENSP00000306157	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL7R	ENST00000303115	NM_002185.3	ENSP00000306157	1
IL7R	ENST00000514217	?	ENSP00000427688	1
IL7R	ENST00000511031	?	?	1
IL7R	ENST00000511982	?	ENSP00000425309	1
IL7R	ENST00000506850	?	ENSP00000421207	1