

SNiPacard

Block annotations

Block info

genomic range	chr19:7,758,154-7,759,715 <i>e!</i>
block size	1,562 bp
variant count	12 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	$\mu = -0.585$ [-2.035 – 0.325]	gene(s) hit or close-by	FCER2 <i>e!</i>
phastCons	$\mu = 0.005$ [0 – 0.02]	eQTL gene(s)	FCER2 <i>e!</i>
GERP++	$\mu = -0.886$ [-3.83 – 1.32]	potentially regulated gene(s)	CCL25 <i>e!</i>
CADD score	$\mu = 2.513$ [0.088 – 6.7]	disease gene(s)	-

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FCER2 <i>e!</i>	?	ENSG00000104921 <i>e!</i>	blood	1.13×10 ⁻⁷ (p-value)	GTEx Portal V6 <i>!m</i>	11
FCER2 <i>e!</i>	ENST00000597921 <i>e!</i>	ILMN_1662451 <i>e!</i>	b-cell	3.36×10 ⁻⁵ (p-value)	Fairfax et al. <i>!m</i>	1
FCER2 <i>e!</i>	ENST00000346664 <i>e!</i>					
FCER2 <i>e!</i>	ENST00000598803 <i>e!</i>					
FCER2 <i>e!</i>	ENST00000597934 <i>e!</i>					
FCER2 <i>e!</i>	ENST00000597312 <i>e!</i>					
FCER2 <i>e!</i>	ENST00000360067 <i>e!</i>					

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
EBF1	MA0154.2	1	no	0.009	1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000223034 <i>e!</i>	1	ENCP00000024014	CCL25 <i>e!</i>

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000342882 <i>e!</i> (TF binding site)	2	embryonic stem cell (H1ESC) blood (GM12878) blood (K562) A549	H3K36me3, H3K27me3 Tr4, Yy1, PolII, DNase1, H3K79me2, H3K4me3, Pax5, H3K4me2, H3K36me3 H3K27me3 H3K27me3
ENSR00000342883 <i>e!</i> (TF binding site)	3	embryonic stem cell (H1ESC) blood (GM12878) A549 blood (K562) skin (NHEK)	H3K27me3 H3K4me2, H3K36me3, EBF1, PolII, DNase1, H3K79me2, H3K4me3, H3K27ac H3K27me3 H3K27me3 H3K27me3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FCER2 <i>e!</i>	ENST00000597921 <i>e!</i>	?	ENSP00000471974 <i>e!</i>	12
FCER2 <i>e!</i>	ENST00000593418 <i>e!</i>	?	ENSP00000472067 <i>e!</i>	12
FCER2 <i>e!</i>	ENST00000360067 <i>e!</i>	NM_001207019.2	ENSP00000353178 <i>e!</i>	12
FCER2 <i>e!</i>	ENST00000346664 <i>e!</i>	NM_002002.4, NM_001220500.1	ENSP00000264072 <i>e!</i>	12
FCER2 <i>e!</i>	ENST00000597934 <i>e!</i>	?	?	12
FCER2 <i>e!</i>	ENST00000598803 <i>e!</i>	?	?	12
FCER2 <i>e!</i>	ENST00000597312 <i>e!</i>	?	?	12

