Locus 164 (rs2289252)

SNiPAcard

Block annotations

Block info

genomic range	chr4:187,199,005-187,213,883
block size	14,879 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.748 [-2.809 – 0.512]	**gene(s) hit or close-by**	F11, F11-AS1
phastCons	μ = 0.001 [0 – 0.004]	**eQTL gene(s)**	IRF2
GERP++	μ = -1.683 [-6.41 – 0.594]	**potentially regulated gene(s)**	–
CADD score	μ = 1.126 [0.034 – 2.484]	**disease gene(s)**	F11

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Activated partial thromboplastin time	$<6.00\times10^{-43}$	GWAS Catalog	22703881	1
bradykinin, des-arg(9)	9.31×10^{-5}	Metabolomics GWAS Server	24816252	2

Disease gene annotation

gene	trait	source DB	source entry/link
F11	FACTOR XI DEFICIENCY	OMIM	MIM:612416
F11	Congenital factor XI deficiency	OrphaNet	OrphaNet:329

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IRF2	ENST00000502750	ILMN_2090607	monocyte	2.92×10^{-4} (p-value)	Fairfax et al.	1
IRF2	ENST00000393593					
IRF2	ENST00000505067					

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
F11	downstream gene variant	2954	ENST00000492972	?	ENSP00000424479	2
F11	downstream gene variant	3026	ENST00000452239	?	ENSP00000397401	2
F11	downstream gene variant	3242	ENST00000514715	?	?	2
F11	upstream gene variant, downstream gene variant	1362	ENST00000503841	?	?	3
F11	upstream gene variant, downstream gene variant	543	ENST00000264691	?	ENSP00000264691	2
F11	downstream gene variant	3949	ENST00000264692	?	ENSP00000264692	1
F11	downstream gene variant	3949	ENST00000403665	NM_000128.3	ENSP00000384957	1
F11-AS1	downstream gene variant	999	ENST00000505103	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
F11	ENST00000452239	?	ENSP00000397401	2
F11	ENST00000264692	?	ENSP00000264692	4
F11	ENST00000403665	NM_000128.3	ENSP00000384957	4
F11	ENST00000264691	?	ENSP00000264691	1
F11-AS1	ENST00000505103	?	?	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
F11-AS1	ENST00000505103	?	1