Locus 388 (rs6687487)

SNiPAcard

Block annotations

Block info

genomic range	chr1:23,060,951-23,066,767
block size	5,817 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.019 [-1.222 – 0.84]	gene(s) hit or close-by	EPHB2
phastCons	μ = 0.031 [0 – 0.123]	eQTL gene(s)	EPHB2
GERP++	μ = 1.218 [-1.12 – 3.76]	potentially regulated gene(s)	C1QA, C1QA, C1QB, C1QB
CADD score	μ = 6.540 [0.389 – 15.18]	disease gene(s)	EPHB2, C1QA, C1QB

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
EPHB2	PROSTATE CANCER/BRAIN CANCER SUSCEPTIBILITY	OMIM	MIM:603688
EPHB2	PROSTATE CANCER	OMIM	MIM:176807
C1QA	C1q DEFICIENCY	OMIM	MIM:613652
C1QB	C1q DEFICIENCY	OMIM	MIM:613652
EPHB2	Familial prostate cancer	OrphaNet	OrphaNet:1331
C1QA	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147
C1QB	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
EPHB2	?	ENSG00000133216	blood	4.78×10^{-8} (p-value)	GTEx Portal V6	5
EPHB2	ENST00000400191	ILMN_2367384	monocyte	1.48×10^{-17} (p-value)	Fairfax et al.	1
EPHB2	ENST00000400191	ILMN_1785061	monocyte	1.93×10^{-10} (p-value)	Fairfax et al.	1
EPHB2	ENST00000400191	ILMN_1785061	monocyte	7.42×10^{-17} (p-value)	Zeller et al.	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000008694	1	ENCP00000001089	C1QA C1QA
		ENCP00000001092	C1QB C1QB

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001518245	2	monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac

(promoter flanking region)		NHLF	DNase1
		Osteobl	H3K27ac
		blood (K562)	H3K27me3
ENSR00001518246	1	NHLF	DNase1
(promoter flanking region)		HSMMtube	DNase1
		Osteobl	H3K27ac, H3K4me2
		blood (K562)	DNase1, H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		endothelium (HUVEC)	DNase1, Cjun, H3K4me1
		lung (IMR90)	H4K5ac, DNase1, H3K18ac, H3K27ac, H3K4me2
		nervous (NH-A)	DNase1, H3K27ac, H3K9ac
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
EPHB2	ENST00000400191	?	ENSP00000383053	5
EPHB2	ENST00000374632	NM_004442.6	ENSP00000363763	5
EPHB2	ENST00000544305	?	ENSP00000444174	5
EPHB2	ENST00000374630	NM_017449.3	ENSP00000363761	5