Locus 138 (rs1511802)

SNiPAcard

Block annotations

Block info

genomic range	chr4:187,140,276-187,178,300
block size	38,025 bp
variant count	18 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.067 [-1.778 – 1.154]	gene(s) hit or close-by	KLKB1
phastCons	μ = 0.100 [0 – 1]	eQTL gene(s)	CYP4V2, F11
GERP++	μ = -0.431 [-5.17 – 1.7]	potentially regulated gene(s)	KLKB1
CADD score	μ = 2.994 [0.416 – 10.46]	disease gene(s)	F11, CYP4V2, KLKB1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
bradykinin, des-arg(9)	8.22×10^{-33}	Metabolomics GWAS Server	24816252	7

Disease gene annotation

gene	trait	source DB	source entry/link
F11	FACTOR XI DEFICIENCY	OMIM	MIM:612416
CYP4V2	BIETTI CRYSTALLINE CORNEORETINAL DYSTROPHY	OMIM	MIM:210370
F11	Congenital factor XI deficiency	OrphaNet	OrphaNet:329
CYP4V2	Bietti crystalline dystrophy	OrphaNet	OrphaNet:41751
KLKB1	Congenital prekallikrein deficiency	OrphaNet	OrphaNet:749

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
F11	?	ENSG00000088926	muscularis mucosae	2.06×10^{-11} (p-value)	GTEx Portal V6	15
CYP4V2	?	ENSG00000145476	tibial nerve	7.73×10^{-12} (p-value)	GTEx Portal V6	15
F11	?	ENSG00000088926	tibial artery	2.59×10^{-9} (p-value)	GTEx Portal V6	15
CYP4V2	?	ENSG00000145476	thyroid	6.38×10^{-7} (p-value)	GTEx Portal V6	15
F11	?	ENSG00000088926	skeletal muscle	3.43×10^{-8} (p-value)	GTEx Portal V6	7
CYP4V2	?	ENSG00000145476	skeletal muscle	2.18×10^{-7} (p-value)	GTEx Portal V6	6
F11	?	ENSG00000088926	cortex	9.79×10^{-7} (p-value)	GTEx Portal V6	11
F11	?	ENSG00000088926	transverse colon	3.76×10^{-7} (p-value)	GTEx Portal V6	6
CYP4V2	ENST00000507209	ILMN_2054607	monocyte	3.48×10^{-4} (p-value)	Fairfax et al.	1
CYP4V2	ENST00000502665					

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000642205	1	ENST00000511406, ENST00000511608, ENST00000264690, ENST00000513864	KLKB1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
KLKB1	downstream gene variant	3011	ENST00000428196	?	ENSP00000412366	1
KLKB1	downstream gene variant	1603	ENST00000446598	?	ENSP00000415563	4
KLKB1	upstream gene variant, downstream gene variant	229	ENST00000467271	?	?	11

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
KLKB1	ENST00000511608	?	ENSP00000426629	18
KLKB1	ENST00000511406	?	?	17
KLKB1	ENST00000446598	?	ENSP00000415563	1
KLKB1	ENST00000428196	?	ENSP00000412366	1
KLKB1	ENST00000513864	?	ENSP00000424469	17
KLKB1	ENST00000264690	NM_000892.3	ENSP00000264690	17

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
KLKB1	ENST00000467271	?	1