Locus 391 (rs6842047)

SNiPAcard

Block annotations

Block info

genomic range	chr4:187,122,904-187,179,486
block size	56,583 bp
variant count	19 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.796 [-2.537 – 0.27]	gene(s) hit or close-by	CYP4V2, KLKB1
phastCons	μ = 0.029 [0 – 0.386]	eQTL gene(s)	CYP4V2, KLKB1
GERP++	μ = -1.076 [-8.01 – 2.8]	potentially regulated gene(s)	F11, FAT1
CADD score	μ = 2.122 [0.111 – 6.993]	disease gene(s)	F11, CYP4V2, KLKB1

Trait annotations

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)
CYP4V2	?	ENSG00000145476	caudate basal ganglia	1.15×10^{-6} (p-value)	GTEx Portal V6	2
CYP4V2	?	ENSG00000145476	pancreas	4.50×10^{-13} (p-value)	GTEx Portal V6	18
CYP4V2	?	ENSG00000145476	stomach	1.94×10^{-6} (p-value)	GTEx Portal V6	5
CYP4V2	?	ENSG00000145476	lung	1.59×10^{-10} (p-value)	GTEx Portal V6	18
KLKB1	?	ENSG00000164344	tibial nerve	5.79×10^{-13} (p-value)	GTEx Portal V6	11
CYP4V2	?	ENSG00000145476	blood	5.18×10^{-12} (p-value)	GTEx Portal V6	18
CYP4V2	?	ENSG00000145476	breast	2.79×10^{-7} (p-value)	GTEx Portal V6	6
CYP4V2	?	ENSG00000145476	esophagus mucosa	1.17×10^{-13} (p-value)	GTEx Portal V6	18
CYP4V2	?	ENSG00000145476	thyroid	7.67×10^{-18} (p-value)	GTEx Portal V6	18
CYP4V2	?	ENSG00000145476	skeletal muscle	4.60×10^{-10} (p-value)	GTEx Portal V6	17
CYP4V2	?	ENSG00000145476	liver	1.13×10^{-6} (p-value)	GTEx Portal V6	4
CYP4V2	?	ENSG00000145476	sun exposed skin	1.72×10^{-9} (p-value)	GTEx Portal V6	18
CYP4V2	?	ENSG00000145476	unexposed skin	4.57×10^{-9} (p-value)	GTEx Portal V6	18
CYP4V2	?	ENSG00000145476	subcutaneous adipocytes	2.19×10^{-7} (p-value)	GTEx Portal V6	16

CYP4V2	ENST00000502665	ILMN_2054607	monocyte	2.98×10-21 (p-value)	Zeller et al.	4
CYP4V2	ENST00000507209					
KLKB1	ENST00000511406	ILMN_1800468	adipocyte	2.44×10-5 (p-value)	MuTHER consortium	4
KLKB1	ENST00000511608					
KLKB1	ENST00000264690					
KLKB1	ENST00000513864					
CYP4V2	ENST00000502665	ILMN_2054607	blood	6.23×10-7 (p-value)	MuTHER consortium	5
CYP4V2	ENST00000507209					
CYP4V2	?	ENSG00000145476	adrenal gland	2.13×10-6 (p-value)	GTEx Portal V6	2
CYP4V2	ENST00000507209	ILMN_2054607	monocyte	2.13×10-12 (p-value)	Fairfax et al.	3
CYP4V2	ENST00000502665					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000399558	1	ENCP00000043224	FAT1
		ENCP00000043223	FAT1
		ENCP00000043226	FAT1
		ENCP00000043217	F11
		ENCP00000043225	FAT1
		ENCP00000043222	FAT1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001256081 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, USF1, Yy1, CTCF, Rad21
		Osteobl	CTCF
		blood (K562)	Egr1, Max, CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	DNase1, CTCF, Rad21
		lung (IMR90)	CTCF
		blood (GM12878)	Yy1, Rad21, CTCF
		A549	CTCF, DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CYP4V2	downstream gene variant	204	ENST00000378802	NM_207352.3	ENSP00000368079	10
CYP4V2	upstream gene variant, downstream gene variant	331	ENST00000513354	?	?	10
CYP4V2	downstream gene variant	964	ENST00000507209	?	?	6
CYP4V2	upstream gene variant, downstream gene variant	519	ENST00000502665	?	?	7
KLKB1	upstream gene variant	2414	ENST00000513864	?	ENSP00000424469	2
KLKB1	upstream gene variant	1334	ENST00000428196	?	ENSP00000412366	2
KLKB1	upstream gene variant	2298	ENST00000264690	NM_000892.3	ENSP00000264690	2
KLKB1	upstream gene variant, downstream gene variant	40	ENST00000511406	?	?	3
KLKB1	upstream gene variant	2403	ENST00000446598	?	ENSP00000415563	2

KLKB1	upstream gene variant	2403	ENST00000446598	?	ENSP00000415363 2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CYP4V2	ENST00000507209	?	?	8
CYP4V2	ENST00000502665	?	?	7
CYP4V2	ENST00000378802	NM_207352.3	ENSP00000368079	4
CYP4V2	ENST00000513354	?	?	4
KLKB1	ENST00000264690	NM_000892.3	ENSP00000264690	2
KLKB1	ENST00000511608	?	ENSP00000426629	17
KLKB1	ENST00000511406	?	?	2
KLKB1	ENST00000513864	?	ENSP00000424469	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
KLKB1	ENST00000264690	NM_000892.3	ENSP00000264690	1
KLKB1	ENST00000511608	?	ENSP00000426629	1
KLKB1	ENST00000513864	?	ENSP00000424469	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CYP4V2	ENST00000507209	?	5
CYP4V2	ENST00000502665	?	4
CYP4V2	ENST00000513354	?	1