Locus 379 (rs204888)

SNiPAcard

Block annotations

Block info

genomic range	chr6:32,021,852-32,097,876
block size	76,025 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.132 [-1.693 – 1.09]	gene(s) hit or close-by	ATF6B, FKBPL, TNXB
phastCons	μ = 0.167 [0 – 1]	eQTL gene(s)	ABCF1, EHMT2, LST1, SKIV2L, TNXA
GERP++	μ = -1.180 [-5.78 – 4.69]	potentially regulated gene(s)	CFB, CFB, CFB, CFB, CFB, CFB, CFB, CFB, DDAH2, DDAH2, DDAH2, DDAH2, DDAH2, DDAH2, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, TNXB, TNXB, TNXB, TNXB, TNXB, TNXB, TNXB
CADD score	μ = 10.688 [1.389 – 21.6]	disease gene(s)	SKIV2L, , , , , , CFB, TNXB

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	OMIM	MIM:614602
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
CFB	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
CFB	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
CFB	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	DECIPHER	MIM:615602
SKIV2L	Syndromic diarrhea	OrphaNet	OrphaNet:84064
TNXB	Ehlers-Danlos syndrome due to tenascin-X deficiency	OrphaNet	OrphaNet:230839
TNXB	EHLERS-DANLOS SYNDROME, HYPERMOBILITY TYPE	OrphaNet	OrphaNet:285
CFB	Atypical hemolytic uremic syndrome with B factor anomaly	OrphaNet	OrphaNet:93578

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)

SKIV2L	?	ENSG00000204351	muscularis mucosae	1.17×10^{-11} (p-value)	GTEx Portal V6	6
SKIV2L	?	ENSG00000204351	lung	9.55×10^{-11} (p-value)	GTEx Portal V6	6
SKIV2L	?	ENSG00000204351	sigmoid colon	6.17×10^{-10} (p-value)	GTEx Portal V6	6
SKIV2L	?	ENSG00000204351	blood	2.43×10^{-8} (p-value)	GTEx Portal V6	6
SKIV2L	?	ENSG00000204351	tibial artery	3.15×10^{-11} (p-value)	GTEx Portal V6	6
SKIV2L	?	ENSG00000204351	esophagus mucosa	1.32×10^{-8} (p-value)	GTEx Portal V6	6
SKIV2L	?	ENSG00000204351	aorta	4.81×10^{-8} (p-value)	GTEx Portal V6	4
TNXA	?	ENSG00000248290	subcutaneous adipocytes	6.09×10^{-7} (p-value)	GTEx Portal V6	6
SKIV2L	?	ENSG00000204351	atrial appendage	3.10×10^{-8} (p-value)	GTEx Portal V6	5
SKIV2L	?	ENSG00000204351	tibial nerve	1.85×10^{-7} (p-value)	GTEx Portal V6	2
SKIV2L	?	ENSG00000204351	thyroid	5.06×10^{-9} (p-value)	GTEx Portal V6	5
SKIV2L	?	ENSG00000204351	cerebellar hemisphere	1.71×10^{-8} (p-value)	GTEx Portal V6	5
SKIV2L	?	ENSG00000204351	transverse colon	3.72×10^{-7} (p-value)	GTEx Portal V6	1
SKIV2L	?	ENSG00000204351	unexposed skin	2.03×10^{-7} (p-value)	GTEx Portal V6	2
EHMT2	ENST00000375537	ILMN_1751081	blood	1.80×10^{-5} (p-value)	MuTHER consortium	4
EHMT2	ENST00000375528					
EHMT2	ENST00000461880					
EHMT2	ENST00000478491					
EHMT2	ENST00000494816					
EHMT2	ENST00000480912					
EHMT2	ENST00000395728					
EHMT2	ENST00000375530					
LST1	ENST00000419073	ILMN_1717127	skin	1.73×10^{-5} (p-value)	MuTHER consortium	4
LST1	ENST00000433492					
LST1	ENST00000376089					
LST1	ENST00000376090					
LST1	ENST00000211921					
ABCF1	ENST00000326195	ILMN_1763875	b-cell	2.59×10^{-4} (p-value)	Fairfax et al.	4
ABCF1	ENST00000376545					
ABCF1	ENST00000479542					

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
E2F4	MA0541.1	6	yes	0.000	1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000445099	1	ENCP00000048143	CFB CFB CFB CFB CFB

	CFB
	CFB
	CFB
ENCP00000048138	CFB
	CFB
	CFB
	CFB
	CFB
	CFB
	CFB
ENCP00000048087	DDAH2
	DDAH2
	DDAH2
	DDAH2
	DDAH2
	DDAH2
	DDAH2
ENCP00000048049	PRRC2A
	PRRC2A
	PRRC2A
	PRRC2A
	PRRC2A
	PRRC2A
	PRRC2A
ENCP00000048163	TNXB
	TNXB
	TNXB
	TNXB
	TNXB
	TNXB
	TNXB
	TNXB

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001703098 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
ENSR00000264509 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, TAF1, H3K4me2, SP1, Srf, USF1, H3K9ac, H3K4me3, Yy1, H3K27ac, H3K36me3, CTCF, PolII, Gabp, Sin3Ak20
		HSMMtube	H3K4me3, H3K4me2, H3K9ac, H2AZ, DNase1
		blood (K562)	Max, Srf, H3K9ac, Gabp, USF1, H3K79me2, H2AZ, ELF1, Nrsf, Cfos, PolII, H3K4me2, DNase1, H3K4me3, Egr1, H3K27ac, ZBTB33
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2, H3K9ac, DNase1
		liver (HepG2)	DNase1, ZBTB33, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K27me3, H2AZ, H3K4me1, Gabp, Yy1, Srf, PolII, TAF1, ELF1, USF1
		blood (GM12878)	Tr4, SIX5, PolII, Srf, H2AZ, DNase1, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, ZBTB33, H3K27ac, Cfos, H3K4me2, H3K9ac
		lung (IMR90)	H3K9ac, DNase1, H3K18ac, H3K27ac, H3K4me2, H3K36me3, H3K4me3
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K4me1, H3K4me2, H3K27ac
		breast (HMEC)	H3K4me3, H3K9ac, H3K4me2, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, Gabp, PolII, Nrf1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, DNase1, PolII, Cjun, Max, H3K4me3, H3K4me2
		A549	H3K36me3, DNase1, H3K9ac, H3K4me2, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ATF6B	downstream gene variant, upstream gene variant	1859	ENST00000375203	NM_004381.4	ENSP00000364349	2
ATF6B	upstream gene variant	1346	ENST00000475705	?	?	1

ATF6B	upstream gene variant	945	ENST00000492342	?	?	1
ATF6B	downstream gene variant, upstream gene variant	1846	ENST00000375201	NM_001136153.1	ENSP00000364347	2
ATF6B	upstream gene variant	3755	ENST00000494022	?	?	1
ATF6B	downstream gene variant, upstream gene variant	3474	ENST00000453203	?	ENSP00000393419	2
ATF6B	upstream gene variant	1877	ENST00000495579	?	?	1
ATF6B	upstream gene variant	2332	ENST00000485314	?	?	1
ATF6B	downstream gene variant, upstream gene variant	1881	ENST00000468502	?	?	2
TNXB	upstream gene variant	290	ENST00000479795	?	ENSP00000418248	2
TNXB	upstream gene variant	382	ENST00000486148	?	?	2
TNXB	upstream gene variant	229	ENST00000375244	NM_019105.6	ENSP00000364393	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ATF6B	ENST00000494022	?	?	3
ATF6B	ENST00000375203	NM_004381.4	ENSP00000364349	1
ATF6B	ENST00000485314	?	?	1
ATF6B	ENST00000375201	NM_001136153.1	ENSP00000364347	1
ATF6B	ENST00000495579	?	?	1
TNXB	ENST00000442721	?	ENSP00000389946	3
TNXB	ENST00000479795	?	ENSP00000418248	1
TNXB	ENST00000486148	?	?	1
TNXB	ENST00000375244	NM_019105.6	ENSP00000364393	2
TNXB	ENST00000611016	?	ENSP00000483409	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
FKBPL	ENST00000375156	NM_022110.3	ENSP00000364298	1