Locus 94 (rs3803884)

SNiPAcard

Block annotations

Block info

genomic range	chr17:48,907,545-48,940,422
block size	32,878 bp
variant count	39 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.454 [-6.153 – 4.074]	gene(s) hit or close-by	RP11-506D12.5, RPL5P33, TOB1, TOB1-AS1, WFIKKN2
phastCons	μ = 0.116 [0 – 1]	eQTL gene(s)	TOB1
GERP++	μ = -1.209 [-7.88 – 5.84]	potentially regulated gene(s)	ABCC3, ACSF2, ANKRD40, CACNA1G, LUC7L3, MYCBPAP, NME2, RP11-294J22.5, RP11-700H6.1, RP11-94C24.8, RSAD1, SPATA20, TOB1, WFIKKN2, XYLT2
CADD score	μ = 4.953 [0.02 – 21.9]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TOB1	ENST00000499247	ILMN_1672004	skin	2.36×10-6 (p-value)	MuTHER consortium	14
TOB1	ENST00000509385		blood	4.66×10-6 (p-value)	MuTHER consortium	14
TOB1	ENST00000268957					
?	?	228834_at	blood	9.00×10-19 (p-value)	Dixon et al.	6

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000023247		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000209627	2	ENCP00000023246	RP11-294J22.5
ENCE00000209535	1	ENCP00000023224	SPATA20
ENCE00000209342	1	ENCP00000023207	RSAD1
		ENCP00000023253	RP11-700H6.1
		ENCP00000023223	SPATA20
		ENCP00000023195	ACSF2
		ENCP00000023218	RP11-94C24.8
		ENCP00000023220	SPATA20
		ENCP00000023250	TOB1
		ENCP00000023222	SPATA20
		ENCP00000023205	RSAD1
		ENCP00000023271	NME2
		ENCP00000023187	XYLT2
		ENCP00000023237	ABCC3

		ENCP00000023241	ANKRD40
		ENCP00000023188	XYLT2
		ENCP00000023226	CACNA1G
		ENCP00000023197	ACSF2
		ENCP00000023196	ACSF2
		ENCP00000023213	MYCBPAP
		ENCP00000023228	CACNA1G
		ENCP00000023242	LUC7L3
		ENCP00000023233	ABCC3
ENCE00000209343	1	ENCP00000023207	RSAD1
		ENCP00000023253	RP11-700H6.1
		ENCP00000023223	SPATA20
		ENCP00000023195	ACSF2
		ENCP00000023218	RP11-94C24.8
		ENCP00000023220	SPATA20
		ENCP00000023250	TOB1
		ENCP00000023222	SPATA20
		ENCP00000023205	RSAD1
		ENCP00000023271	NME2
		ENCP00000023237	ABCC3
		ENCP00000023187	XYLT2
		ENCP00000023241	ANKRD40
		ENCP00000023188	XYLT2
		ENCP00000023226	CACNA1G
		ENCP00000023197	ACSF2
		ENCP00000023196	ACSF2
		ENCP00000023228	CACNA1G
		ENCP00000023213	MYCBPAP
		ENCP00000023242	LUC7L3
		ENCP00000023233	ABCC3
ENCE00000209722	2	ENCP00000023270	NME2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001348919 (enhancer)	1	liver (HepG2)	H3K27me3
		blood (K562)	H2AZ
ENSR00001639631 (enhancer)	1	monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	DNase1
		embryonic stem cell (H1ESC)	DNase1
		liver (HepG2)	H3K27me3, DNase1
		blood (K562)	CTCF
ENSR00001639632 (enhancer)	3	embryonic stem cell (H1ESC)	CTCF, Rad21, DNase1
		HSMMtube	CTCF
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, H3K4me1, CTCF, H3K27me3, DNase1
		blood (GM12878)	CTCF
		lung (IMR90)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	CTCF

		A549	CTCF
ENSR00001348925 (promoter flanking region)	3	embryonic stem cell (H1ESC)	USF1
		HSMMtube	H3K27ac, DNase1
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K27ac
		cervix (HeLa-S3)	DNase1, H3K27ac
		liver (HepG2)	USF1, H3K4me1, H3K4me2, HNF4A, H3K27me3, DNase1
		skin (NHEK)	DNase1
ENSR00001639636 (enhancer)	1	endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K4me1
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-506D12.5	downstream gene variant, upstream gene variant	52	ENST00000572491	?	?	20
RPL5P33	upstream gene variant, downstream gene variant	634	ENST00000487833	?	?	12
TOB1	downstream gene variant	2373	ENST00000499247	NM_005749.3, NM_001243885.1	ENSP00000427695	3
TOB1	downstream gene variant	308	ENST00000509385	?	?	3
TOB1	downstream gene variant	3130	ENST00000268957	NM_001243877.1	ENSP00000268957	2
TOB1-AS1	upstream gene variant	3698	ENST00000523470	?	?	1
TOB1-AS1	upstream gene variant	4087	ENST00000514358	?	?	1
TOB1-AS1	upstream gene variant	3618	ENST00000416263	?	?	1
WFIKKN2	upstream gene variant, downstream gene variant	22	ENST00000426127	?	ENSP00000405889	18
WFIKKN2	upstream gene variant, downstream gene variant	370	ENST00000311378	NM_175575.5	ENSP00000311184	16

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
TOB1	ENST00000499247	NM_005749.3, NM_001243885.1	ENSP00000427695	K	aaA/aaG	1
TOB1	ENST00000268957	NM_001243877.1	ENSP00000268957	K	aaA/aaG	1
WFIKKN2	ENST00000311378	NM_175575.5	ENSP00000311184	S	agT/agC	1
WFIKKN2	ENST00000426127	?	ENSP00000405889	S	agT/agC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
WFIKKN2	ENST00000311378	NM_175575.5	ENSP00000311184	9
WFIKKN2	ENST00000426127	?	ENSP00000405889	9

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
WFIKKN2	ENST00000311378	NM_175575.5	ENSP00000311184	1