SNiPAcard

Block annotations

Block info

genomic range	chr20:43,763,968-43,888,106
block size	124,139 bp
variant count	150 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.456 [-3.331 – 1.497]	**gene(s) hit or close-by**	PI3, RP1-300I2.2, RP1-300I2.3, SEMG1, SEMG2, SLPI
phastCons	μ = 0.073 [0 – 0.822]	**eQTL gene(s)**	DBNDD2, KCNS1, MATN4, PI3, RP3-453C12.8, SEMG1, SLPI, SYS1-DBNDD2, WFDC12, WFDC5
GERP++	μ = -0.394 [-6.16 – 3.49]	**potentially regulated gene(s)**	DBNDD2, PABPC1L, PI3, RP3-453C12.8, RPL5P2, RPS2P7, SEMG2, SPINT3, STK4, STK4, SYS1, WFDC11, WFDC13, WFDC2, WFDC5, WFDC9
CADD score	μ = 3.639 [0.002 – 14.69]	**disease gene(s)**	STK4

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM099561	1

Disease gene annotation

gene	trait	source DB	source entry/link	
STK4	T-CELL IMMUNODEFICIENCY, RECURRENT INFECTIONS, AND AUTOIMMUNITY [...]	OMIM	MIM:614868	
STK4	Combined immunodeficiency due to STK4 deficiency	OrphaNet	OrphaNet:314689	

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
PI3	missense variant	ENST00000243924	NM_002638.3	ENSP00000243924	2	2			2
SEMG2	missense variant	ENST00000372769	NM_003008.2	ENSP00000361855	2	2			2

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MATN4	?	ENSG00000124159	pancreas	1.67×10^{-7} (p-value)	GTEx Portal V6	148
SLPI	?	ENSG00000124107	sun exposed skin	1.26×10^{-14} (p-value)	GTEx Portal V6	150
KCNS1	?	ENSG00000124134	sun exposed skin	1.07×10^{-5} (p-value)	GTEx Portal V6	9
SLPI	?	ENSG00000124107	esophagus mucosa	2.65×10^{-17} (p-value)	GTEx Portal V6	150
SLPI	?	ENSG00000124107	unexposed skin	9.30×10^{-11} (p-value)	GTEx Portal V6	66

WFDC5	?	ENSG00000175121	unexposed skin	2.65×10-6 (p-value)	GTEx Portal V6	10
KCNS1	ENST00000306117	ILMN_1761903	blood	1.92×10-5 (p-value)	Westra et al.	28
DBNDD2	ENST00000372717	ILMN_1730612	skin	3.96×10-7 (p-value)	MuTHER consortium	47
DBNDD2	ENST00000372712					
DBNDD2	ENST00000372720					
DBNDD2	ENST00000372710					
SYS1-DBNDD2	ENST00000475242					
DBNDD2	ENST00000360981					
DBNDD2	ENST00000443296					
DBNDD2	ENST00000372722					
DBNDD2	ENST00000372723					
DBNDD2	ENST00000357275					
SYS1-DBNDD2	ENST00000419593					
WFDC5	ENST00000372789	ILMN_2079042	skin	4.65×10-7 (p-value)	MuTHER consortium	43
WFDC5	ENST00000307971					
KCNS1	?	ENSG00000124134	adrenal gland	3.84×10-6 (p-value)	GTEx Portal V6	41
SEMG1	?	ENSG00000124233	adrenal gland	3.08×10-6 (p-value)	GTEx Portal V6	113
RP3-453C12.8	?	ENSG00000232880	tibial artery	1.01×10-5 (p-value)	GTEx Portal V6	4
PI3	?	ENSG00000124102	blood	5.52×10-6 (p-value)	GTEx Portal V6	2
WFDC12	?	ENSG00000168703	transverse colon	6.65×10-6 (p-value)	GTEx Portal V6	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000296292	1	ENCP00000031705	WFDC5
ENCE00000296259	1	ENCP00000031698	PABPC1L
ENCE00000296165	1	ENCP00000031720	SYS1
		ENCP00000031724	DBNDD2
		ENCP00000031716	SYS1
		ENCP00000031722	RP3-453C12.8
		ENCP00000031733	RPL5P2
ENCE00000296320	2	ENCP00000031707	PI3
ENCE00000296274	1	ENCP00000031741	WFDC11
		ENCP00000031702	STK4 STK4
ENCE00000296369	1	ENCP00000031709	SEMG2
		ENCP00000031740	WFDC9 RPS2P7
		ENCP00000031742	WFDC13
		ENCP00000031734	SPINT3
		ENCP00000031731	WFDC2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000009720 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3, Max, Rad21, Gabp, H2AZ, CTCFL, CTCF

		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Gabp, Rad21, CTCF, H3K27me3, DNase1
		blood (GM12878)	CTCF
		lung (IMR90)	CTCF, H3K27me3
		skin (NHEK)	CTCF, DNase1
		NHLF	CTCF
		Osteobl	H3K27me3, CTCF
		blood (DND-41)	H3K27me3, CTCF
		breast (HMEC)	CTCF, DNase1
		cervix (HeLa-S3)	DNase1, CTCF, Gabp
		monocytes (Monocytes-CD14+)	CTCF, H3K27me3
		endothelium (HUVEC)	H3K36me3, H3K27me3, CTCF
		A549	CTCF, H3K27me3
ENSR00001665870 (open chromatin region)	1	A549	H3K27me3
ENSR00001665871 (open chromatin region)	2	A549	H3K27me3
ENSR00001449109 (promoter flanking region)	2	NHLF	DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1, H3K27me3
		muscle (HSMM)	DNase1
		A549	H3K27me3
		nervous (NH-A)	DNase1
ENSR00001449110 (open chromatin region)	2	endothelium (HUVEC)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K27me3
		breast (HMEC)	DNase1
ENSR00000401582 (TF binding site)	6	embryonic stem cell (H1ESC)	DNase1
		blood (K562)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K27me3
		breast (HMEC)	DNase1
		muscle (HSMM)	DNase1
ENSR00000401583 (TF binding site)	6	blood (K562)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K27me3
ENSR00001665874 (open chromatin region)	1	A549	H3K27me3
		blood (K562)	H3K27me3
ENSR00001449111 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K27me3
		embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	H3K27me3
		A549	H3K27me3
		blood (DND-41)	H3K27me3
ENSR00000649547 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Yy1, CTCF, Rad21, DNase1
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3, CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	CTCF
		blood (GM12878)	CTCF
		skin (NHEK)	CTCF

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
PI3	2	ENST00000243924	hsa-miR-28-5p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PI3	upstream gene variant, downstream gene variant	89	ENST00000243924	NM_002638.3	ENSP00000243924	22
RP1-300I2.2	upstream gene variant, downstream gene variant	1809	ENST00000461248	?	?	8
RP1-300I2.2	upstream gene variant, downstream gene variant	1883	ENST00000486547	?	?	8
RP1-300I2.3	upstream gene variant, downstream gene variant	758	ENST00000422041	?	?	7
SEMG1	upstream gene variant, downstream gene variant	406	ENST00000372781	NM_003007.3	ENSP00000361867	10
SEMG2	upstream gene variant, downstream gene variant	177	ENST00000372769	NM_003008.2	ENSP00000361855	10
SLPI	downstream gene variant, upstream gene variant	791	ENST00000338380	NM_003064.3	ENSP00000342082	4

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
SEMG2	ENST00000372769	NM_003008.2	ENSP00000361855	Q	caG/caA	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PI3	ENST00000243924	NM_002638.3	ENSP00000243924	6
SEMG1	ENST00000372781	NM_003007.3	ENSP00000361867	1
SEMG2	ENST00000372769	NM_003008.2	ENSP00000361855	3

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
PI3	ENST00000243924	NM_002638.3	ENSP00000243924	2

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SEMG1	ENST00000372781	NM_003007.3	ENSP00000361867	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP1-300I2.2	ENST00000486547	?	1
RP1-300I2.2	ENST00000461248	?	1