Locus 45 (rs12151621)

SNiPAcard

Block annotations

Block info

genomic range	chr2:85,933,003-85,938,407
block size	5,405 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.852 [-2.998 – 0.359]	gene(s) hit or close-by	–
phastCons	μ = 0.020 [0 – 0.116]	eQTL gene(s)	AC012454.4, ATOH8, GGCX, GNLY, MLH3, VAMP8
GERP++	μ = -1.305 [-5.72 – 2.01]	potentially regulated gene(s)	AC105053.3, CAPG, IMMT, MAT2A, MRPL35, POLR1A, PTCD3, SFTPB, SH2D6, TGOLN2, TMEM150A, USP39, VAMP5, VAMP8
CADD score	μ = 2.968 [0.191 – 6.026]	disease gene(s)	GGCX, MLH3, SFTPB

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
GGCX	VITAMIN K-DEPENDENT CLOTTING FACTORS, COMBINED DEFICIENCY OF, [...]	OMIM	MIM:277450
GGCX	PSEUDOXANTHOMA ELASTICUM-LIKE DISORDER WITH MULTIPLE COAGULATION [...]	OMIM	MIM:610842
MLH3	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, TYPE 7	OMIM	MIM:614385
MLH3	COLORECTAL CANCER	OMIM	MIM:114500
SFTPB	SURFACTANT METABOLISM DYSFUNCTION, PULMONARY, 1	OMIM	MIM:265120
SFTPB	RESPIRATORY DISTRESS SYNDROME IN PREMATURE INFANTS	OMIM	MIM:267450
GGCX	Body skin hyperlaxity due to vitamin K-dependent coagulation factor deficiency	OrphaNet	OrphaNet:91135
GGCX	Hereditary combined deficiency of vitamin K-dependent clotting factors	OrphaNet	OrphaNet:98434
MLH3	Hereditary nonpolyposis colon cancer	OrphaNet	OrphaNet:144
SFTPB	Congenital pulmonary alveolar proteinosis	OrphaNet	OrphaNet:264675
SFTPB	Infant acute respiratory distress syndrome	OrphaNet	OrphaNet:70587
SFTPB	Neonatal acute respiratory distress with surfactant metabolism deficiency	OrphaNet	OrphaNet:217563

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
GNLY	?	ENSG00000115523	pancreas	8.36×10^{-9} (p-value)	GTEx Portal V6	6
GNLY	ENST00000489214	ILMN_1708779	adipocyte	6.83×10^{-14} (p-value)	MuTHER consortium	5
GNLY	ENST00000409696					
GNLY	ENST00000534351					
GNLY	ENST00000489980					
GNLY	ENST00000533041					
GNLY	ENST00000482900					
GNLY	ENST00000470974					
GNLY	ENST00000464298					
GNLY	ENST00000263863					
GNLY	ENST00000524600					
GNLY	?	ENSG00000115523	muscularis mucosae	1.78×10^{-14} (p-value)	GTEx Portal V6	6

GNLY	?	ENSG00000115523	lung	1.37×10-11 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	atrial appendage	6.52×10-7 (p-value)	GTEx Portal V6	5
ATOH8	?	ENSG00000168874	blood	5.22×10-20 (p-value)	GTEx Portal V6	6
AC012454.4	?	ENSG00000203363	blood	1.46×10-12 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	blood	9.39×10-24 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	thyroid	6.13×10-11 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	skeletal muscle	4.22×10-6 (p-value)	GTEx Portal V6	5
GNLY	?	ENSG00000115523	prostate	9.77×10-9 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	transverse colon	1.42×10-8 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	sun exposed skin	7.34×10-7 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	aorta	2.64×10-8 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	subcutaneous adipocytes	5.31×10-12 (p-value)	GTEx Portal V6	6
?	?	ILMN_2256295	monocyte	7.03×10-22 (p-value)	Zeller et al.	2
?	?	ILMN_1790692	monocyte	1.26×10-24 (p-value)	Zeller et al.	2
ATOH8	ENST00000469442	ILMN_1654065	monocyte	1.32×10-157 (p-value)	Zeller et al.	2
ATOH8	ENST00000306279					
ATOH8	ENST00000489682					
GNLY	ENST00000489214	ILMN_1708779	monocyte	2.86×10-19 (p-value)	Zeller et al.	2
GNLY	ENST00000409696					
GNLY	ENST00000534351					
GNLY	ENST00000489980					
GNLY	ENST00000533041					
GNLY	ENST00000482900					
GNLY	ENST00000470974					
GNLY	ENST00000464298					
GNLY	ENST00000263863					
GNLY	ENST00000524600					
GNLY	?	ENSG00000115523	visceral adipocytes	1.30×10-7 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	stomach	9.04×10-10 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	spleen	3.83×10-8 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	sigmoid colon	3.04×10-6 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	coronary artery	3.38×10-6 (p-value)	GTEx Portal V6	5
GNLY	?	ENSG00000115523	tibial nerve	1.39×10-6 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	esophagus mucosa	2.80×10-16 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	gastroesophageal junction	2.82×10-7 (p-value)	GTEx Portal V6	6
GGCX	ENST00000233838	ILMN_1758232	blood	1.22×10-4 (p-value)	Westra et al.	1
VAMP8	ENST00000263864	ILMN_2190084	blood	9.93×10-6 (p-value)	Westra et al.	5
VAMP8	ENST00000432071					
VAMP8	ENST00000409760					

ATOH8	ENST00000306279	ILMN_1654065	monocyte	2.74×10-25 (p-value)	Fairfax et al.	1
ATOH8	ENST00000469442					
ATOH8	ENST00000489682					
GNLY	?	ENSG00000115523	terminal ileum	1.09×10-6 (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
MLH3	ENST00000555144	ILMN_2282282	chr14	monocyte	2.52×10-18 (p-value)	Zeller et al.	2
MLH3	ENST00000555499						
MLH3	ENST00000553713						
MLH3	ENST00000556740						
MLH3	ENST00000355774						

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000253865	1	ENCP00000026860	USP39
		ENCP00000026814	TGOLN2
		ENCP00000026873	AC105053.3
		ENCP00000026830	CAPG
		ENCP00000026897	MRPL35
		ENCP00000026853	TMEM150A
		ENCP00000026896	IMMT
ENCE00000253866	2	ENCP00000026854	USP39
		ENCP00000026837	SH2D6
		ENCP00000026840	MAT2A
		ENCP00000026849	TMEM150A
		ENCP00000026888	PTCD3
		ENCP00000026853	TMEM150A
		ENCP00000026863	SFTPB
		ENCP00000026834	SH2D6
		ENCP00000026841	MAT2A
		ENCP00000026864	SFTPB
		ENCP00000026883	POLR1A
		ENCP00000026845	VAMP8
		ENCP00000026858	USP39
		ENCP00000026825	CAPG
		ENCP00000026814	TGOLN2
		ENCP00000026828	CAPG
		ENCP00000026830	CAPG
		ENCP00000026847	VAMP5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000593772 (promoter flanking region)	3	embryonic stem cell (H1ESC)	DNase1, H3K27me3, Rad21, CTCF
		HSMMtube	DNase1, CTCF
		blood (K562)	H3K27me3, DNase1, Rad21, CTCF
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF, H3K27ac, DNase1
		liver (HepG2)	Rad21, CTCF, H3K27me3
		lung (IMR90)	DNase1, CTCF
		blood (GM12878)	H3K4me2, CTCF, H3K27ac, Rad21, H3K4me3, PU1, Srf, BATF, H3K4me1, DNase1

		nervous (NH-A)	CTCF, DNase1
		skin (NHEK)	DNase1, H3K4me1, CTCF
		NHLF	CTCF, DNase1
		Osteobl	CTCF, H3K27ac
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	DNase1, H3K4me1, CTCF
		cervix (HeLa-S3)	Jund, H3K27ac, H3K4me1, CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, H3K27ac, H3K9ac, H3K4me1, DNase1
		endothelium (HUVEC)	DNase1, CTCF
		A549	H3K4me3, CTCF, DNase1
ENSR00000676793 e! (CTCF binding site)	4	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	DNase1, CTCF, H3K27me3
		blood (K562)	H3K27me3, DNase1, Rad21, CTCF
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF, DNase1
		liver (HepG2)	Rad21, CTCF, H3K27me3
		lung (IMR90)	DNase1, CTCF
		blood (GM12878)	DNase1, Rad21, CTCF
		nervous (NH-A)	CTCF, H3K27me3
		skin (NHEK)	DNase1, CTCF
		NHLF	CTCF
		Osteobl	H3K27me3, CTCF
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	H3K9ac, DNase1, H3K4me1, CTCF, H3K27ac
		endothelium (HUVEC)	DNase1, CTCF
		A549	H3K4me3, CTCF, DNase1
ENSR00001544078 e! (enhancer)	1	HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		muscle (HSMM)	DNase1
		monocytes (Monocytes-CD14+)	H3K4me1, H3K27ac
		liver (HepG2)	H3K27me3
		nervous (NH-A)	H3K27me3
		skin (NHEK)	H3K27me3