Locus 21 (rs9326248)

SNiPAcard

Block annotations

Block info

genomic range	chr11:116,999,225-117,081,974
block size	82,750 bp
variant count	38 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.268 [-2.147 – 0.86]	gene(s) hit or close-by	AP000892.6, PAFAH1B2, PCSK7, RP11-109L13.1, RP11-109L13.5, SIDT2, TAGLN
phastCons	μ = 0.042 [0 – 0.378]	eQTL gene(s)	CBLN2, PAFAH1B2, PCSK7, RP11-109L13.1, SIDT2, TAGLN
GERP++	μ = -0.957 [-6.23 – 2.63]	potentially regulated gene(s)	PCSK7, RP11-109L13.1, SIK3, TAGLN
CADD score	μ = 3.689 [0.184 – 10.77]	disease gene(s)	CBLN2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
1,6-anhydroglucose	9.23×10^{-5}	Metabolomics GWAS Server	24816252	1

Disease gene annotation

gene	trait	source DB	source entry/link
CBLN2	Heritable pulmonary arterial hypertension	OrphaNet	OrphaNet:275777
CBLN2	Idiopathic pulmonary arterial hypertension	OrphaNet	OrphaNet:275766

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TAGLN	ENST00000525531	ILMN_1778668	blood	2.72×10^{-21} (p-value)	MuTHER consortium	14
TAGLN	ENST00000532870					
TAGLN	ENST00000278968					
TAGLN	ENST00000392951					
TAGLN	ENST00000530649					
TAGLN	ENST00000533863					
TAGLN	ENST00000525531	ILMN_2400935	blood	3.94×10^{-7} (p-value)	MuTHER consortium	14
TAGLN	ENST00000529622					
TAGLN	ENST00000278968					
TAGLN	ENST00000530649					
TAGLN	ENST00000529792					
TAGLN	ENST00000532870					
TAGLN	ENST00000392951					
TAGLN	ENST00000533863					
PCSK7	ENST00000540028	ILMN_1699545	skin	5.96×10^{-6} (p-value)	MuTHER consortium	14
PCSK7	ENST00000529458		blood	5.70×10^{-6} (p-value)	MuTHER consortium	14
PCSK7	ENST00000534529					
PCSK7	ENST00000320934					

PCSK7	ENST00000527037					
TAGLN	ENST00000525531	ILMN_1706783	adipocyte	3.60×10-5 (p-value)	MuTHER consortium	8
TAGLN	ENST00000278968					
RP11-109L13.1	?	ENSG00000254851	atrial appendage	8.55×10-30 (p-value)	GTEx Portal V6	35
PCSK7	?	ENSG00000160613	transformed fibroblasts	2.41×10-13 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	transformed fibroblasts	1.29×10-20 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	breast	8.69×10-32 (p-value)	GTEx Portal V6	35
TAGLN	?	ENSG00000149591	blood	6.45×10-45 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	blood	7.27×10-21 (p-value)	GTEx Portal V6	35
SIDT2	?	ENSG00000149577	blood	1.38×10-11 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	skeletal muscle	1.07×10-26 (p-value)	GTEx Portal V6	35
TAGLN	?	ENSG00000149591	EBV lymphocytes	5.99×10-11 (p-value)	GTEx Portal V6	30
RP11-109L13.1	?	ENSG00000254851	unexposed skin	1.92×10-32 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	sun exposed skin	7.36×10-47 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	ovary	4.30×10-14 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	left ventricle	3.41×10-17 (p-value)	GTEx Portal V6	35
PCSK7	?	ENSG00000160613	aorta	7.38×10-15 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	aorta	8.98×10-38 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	subcutaneous adipocytes	5.16×10-61 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	visceral adipocytes	1.37×10-38 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	stomach	2.76×10-26 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	tibial nerve	1.97×10-48 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	esophagus mucosa	1.49×10-36 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	pituitary	5.84×10-13 (p-value)	GTEx Portal V6	35
TAGLN	?	ENSG00000149591	testis	3.46×10-6 (p-value)	GTEx Portal V6	4
RP11-109L13.1	?	ENSG00000254851	testis	5.08×10-15 (p-value)	GTEx Portal V6	34
SIDT2	?	ENSG00000149577	testis	2.87×10-6 (p-value)	GTEx Portal V6	12
TAGLN	?	ENSG00000149591	pancreas	4.73×10-7 (p-value)	GTEx Portal V6	27
RP11-109L13.1	?	ENSG00000254851	pancreas	2.07×10-9 (p-value)	GTEx Portal V6	32
SIDT2	?	ENSG00000149577	pancreas	1.91×10-7 (p-value)	GTEx Portal V6	25
RP11-109L13.1	?	ENSG00000254851	uterus	1.40×10-11 (p-value)	GTEx Portal V6	34
PCSK7	?	ENSG00000160613	muscularis mucosae	4.73×10-11 (p-value)	GTEx Portal V6	34
RP11-109L13.1	?	ENSG00000254851	muscularis mucosae	1.97×10-41 (p-value)	GTEx Portal V6	35
PCSK7	?	ENSG00000160613	lung	8.44×10-13 (p-value)	GTEx Portal V6	34
RP11-109L13.1	?	ENSG00000254851	lung	3.29×10-45 (p-value)	GTEx Portal V6	35
SIDT2	?	ENSG00000149577	lung	9.70×10-11 (p-value)	GTEx Portal V6	32

PAFAH1B2	?	ENSG00000168092	lung	3.46×10-6 (p-value)	GTEx Portal V6	11
TAGLN	?	ENSG00000149591	lung	6.43×10-6 (p-value)	GTEx Portal V6	2
PCSK7	?	ENSG00000160613	tibial artery	7.26×10-18 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	tibial artery	2.13×10-54 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	thyroid	7.60×10-39 (p-value)	GTEx Portal V6	35
SIDT2	?	ENSG00000149577	thyroid	1.81×10-6 (p-value)	GTEx Portal V6	22
RP11-109L13.1	?	ENSG00000254851	prostate	1.94×10-20 (p-value)	GTEx Portal V6	34
RP11-109L13.1	?	ENSG00000254851	transverse colon	7.57×10-24 (p-value)	GTEx Portal V6	35
SIDT2	ENST00000278951	ILMN_1791912	monocyte	2.57×10-124 (p-value)	Zeller et al.	3
SIDT2	ENST00000532062					
SIDT2	ENST00000324225					
SIDT2	ENST00000431081					
SIDT2	ENST00000620360					
TAGLN	ENST00000525531	ILMN_1778668	monocyte	9.39×10-197 (p-value)	Zeller et al.	3
TAGLN	ENST00000532870					
TAGLN	ENST00000278968					
TAGLN	ENST00000392951					
TAGLN	ENST00000530649					
TAGLN	ENST00000533863					
TAGLN	ENST00000525531	ILMN_2400935	monocyte	4.10×10-99 (p-value)	Zeller et al.	3
TAGLN	ENST00000529622					
TAGLN	ENST00000278968					
TAGLN	ENST00000530649					
TAGLN	ENST00000529792					
TAGLN	ENST00000532870					
TAGLN	ENST00000392951					
TAGLN	ENST00000533863					
RP11-109L13.1	?	ENSG00000254851	terminal ileum	7.72×10-14 (p-value)	GTEx Portal V6	33
RP11-109L13.1	?	ENSG00000254851	spleen	2.70×10-11 (p-value)	GTEx Portal V6	28
PCSK7	?	ENSG00000160613	coronary artery	8.08×10-8 (p-value)	GTEx Portal V6	6
RP11-109L13.1	?	ENSG00000254851	coronary artery	6.64×10-30 (p-value)	GTEx Portal V6	35
PCSK7	?	ENSG00000160613	sigmoid colon	1.18×10-9 (p-value)	GTEx Portal V6	33
RP11-109L13.1	?	ENSG00000254851	sigmoid colon	2.48×10-23 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	vagina	2.61×10-14 (p-value)	GTEx Portal V6	34
TAGLN	ENST00000525531	ILMN_1706783	blood	5.26×10-7 (p-value)	Westra et al.	14
TAGLN	ENST00000278968					
RP11-109L13.1	?	ENSG00000254851	gastroesophageal junction	5.10×10-26 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	adrenal gland	1.03×10-21 (p-value)	GTEx Portal V6	35
PAFAH1B2	?	ENSG00000168092	cerebellum	1.07×10-7 (p-value)	GTEx Portal V6	30
SIDT2	ENST00000278951	ILMN_1791912	monocyte	1.60×10-42 (p-value)	Fairfax et al.	4
SIDT2	ENST00000532062		b-cell	1.35×10-29 (p-value)	Fairfax et al.	4
SIDT2	ENST00000324225					

SIDT2	ENST00000431081						
SIDT2	ENST00000620360						
TAGLN	ENST00000525531	ILMN_2400935	monocyte	9.51×10^{-19} (p-value)	Fairfax et al.	4
TAGLN	ENST00000529622		b-cell	1.46×10^{-14} (p-value)	Fairfax et al.	4
TAGLN	ENST00000278968						
TAGLN	ENST00000530649						
TAGLN	ENST00000529792						
TAGLN	ENST00000532870						
TAGLN	ENST00000392951						
TAGLN	ENST00000533863						

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
CBLN2	ENST00000581425	ILMN_1796069	chr18	monocyte	1.54×10^{-12} (p-value)	Zeller et al.	2
CBLN2	ENST00000585159						
CBLN2	ENST00000269503						

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000014774		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000123647	1	ENCP00000014794	PCSK7
ENCE00000123461	1	ENCP00000014767	SIK3
ENCE00000123486	1	ENCP00000014768	SIK3
ENCE00000123604	1	ENCP00000014791	PCSK7
		ENCP00000014787	TAGLN

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001607349 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, PolII, Rad21, Nrsf, CTCF
		HSMMtube	H3K9ac, H3K4me2, H3K27ac, DNase1
		blood (K562)	ZBTB7A, Rad21, CTCF
		skin (NHDF-AD)	CTCF, H3K4me2
		muscle (HSMM)	H3K4me2, H3K4me3, CTCF
		liver (HepG2)	CTCF
		lung (IMR90)	H3K14ac, H4K8ac, H3K27ac, H3K18ac, H3K4me2, CTCF, H3K56ac, H4K91ac, H3K9ac, H2AK5ac, H3K4ac, H3K36me3, H4K5ac
		blood (GM12878)	CTCF
		nervous (NH-A)	H3K4me2, H3K9ac
		skin (NHEK)	CTCF
		NHLF	H3K27ac, CTCF
		Osteobl	H3K27ac, H3K4me3, H3K4me2, H3K36me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00000569571 (promoter)	4	embryonic stem cell (H1ESC)	DNase1, H3K9ac, CTCF, Yy1, TAF1, H3K4me2, Srf, H3K4me3, H3K36me3, H3K27me3, PolII, Rad21, Jund, TAF7, Nrsf
		HSMMtube	H3K4me2, H3K27ac, H3K36me3, DNase1, H3K9ac
		blood (K562)	ZBTB7A, Rad21, CTCF, H3K36me3

		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	DNase1, H3K79me2, CTCF, H3K4me3, H3K4me2, H3K36me3
		liver (HepG2)	CTCF
		blood (GM12878)	CTCF, PolII
		lung (IMR90)	H3K27ac, H3K79me2, H3K14ac, H4K8ac, DNase1, H3K18ac, CTCF, H3K56ac, H4K91ac, H3K9ac, H2AK5ac, H3K4ac, H3K4me3, H3K36me3, H4K5ac, H3K4me2
		nervous (NH-A)	DNase1, H3K36me3, H3K4me3, H3K4me2, H4K20me1, H3K9ac
		skin (NHEK)	CTCF, DNase1
		NHLF	H3K9ac, CTCF, H3K4me3, DNase1, H3K27ac, H3K36me3
		Osteobl	H3K27ac, H3K4me3, H3K4me2, H3K36me3
		breast (HMEC)	CTCF, H3K4me3
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00000321754	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3, PolII, Rad21, CTCF
(CTCF binding site)		HSMMtube	H3K36me3
		blood (K562)	Rad21, H2AZ, CTCF, H3K36me3
		muscle (HSMM)	H3K36me3
		liver (HepG2)	H3K4me3, H3K4me2, CTCF
		lung (IMR90)	H3K36me3
		blood (GM12878)	PolII, H3K36me3
		nervous (NH-A)	H3K36me3, H4K20me1
		skin (NHEK)	H3K36me3
		NHLF	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00001573939	1	embryonic stem cell (H1ESC)	H3K36me3, PolII, Rad21, CTCF, DNase1
(CTCF binding site)		HSMMtube	H3K36me3, DNase1
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3, H3K27me3
		blood (K562)	CTCF, H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3, CTCF
		A549	H3K36me3
		skin (NHEK)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AP000892.6	upstream gene variant, downstream gene variant	755	ENST00000624094	?	?	7
PAFAH1B2	upstream gene variant	2799	ENST00000304808	?	ENSP00000304006	4
PAFAH1B2	upstream gene variant, downstream gene variant	827	ENST00000419197	NM_001184748.1	ENSP00000388742	6
PAFAH1B2	upstream gene variant, downstream gene variant	1564	ENST00000530272	NM_001184746.1	ENSP00000431365	6
PAFAH1B2	downstream gene variant	146	ENST00000533677	?	?	2
PAFAH1B2	upstream gene variant, downstream gene variant	807	ENST00000526888	?	?	6
PAFAH1B2	upstream gene variant	2888	ENST00000533206	?	?	4
PAFAH1B2	upstream gene variant, downstream gene variant	1279	ENST00000529887	NM_001184747.1	ENSP00000434951	6
PAFAH1B2	upstream gene variant	2773	ENST00000527958	NM_002572.3	ENSP00000435289	4
PCSK7	downstream gene variant	2090	ENST00000320934	NM_004716.2	ENSP00000325917	5
PCSK7	downstream gene variant	2681	ENST00000528973	?	?	1
PCSK7	downstream gene variant	2827	ENST00000534529	?	?	5

PCSK7	downstream gene variant, upstream gene variant	36	ENST00000527861	?	?	6
PCSK7	downstream gene variant	2831	ENST00000540028	?	ENSP00000441944	5
PCSK7	downstream gene variant, upstream gene variant	229	ENST00000531573	?	?	4
PCSK7	downstream gene variant, upstream gene variant	782	ENST00000532810	?	?	5
PCSK7	downstream gene variant, upstream gene variant	581	ENST00000529458	?	?	5
PCSK7	downstream gene variant	156	ENST00000527037	?	?	3
PCSK7	downstream gene variant	1783	ENST00000533135	?	?	1
RP11-109L13.1	upstream gene variant, downstream gene variant	944	ENST00000528464	?	?	11
RP11-109L13.5	downstream gene variant, upstream gene variant	1917	ENST00000525475	?	?	7
SIDT2	upstream gene variant, downstream gene variant	618	ENST00000532960	?	ENSP00000431176	4
SIDT2	upstream gene variant, downstream gene variant	900	ENST00000431081	?	ENSP00000399635	7
SIDT2	upstream gene variant, downstream gene variant	999	ENST00000628876	?	ENSP00000486114	6
SIDT2	upstream gene variant, downstream gene variant	587	ENST00000529441	?	?	6
SIDT2	upstream gene variant, downstream gene variant	1987	ENST00000531353	?	ENSP00000435004	3
SIDT2	upstream gene variant, downstream gene variant	189	ENST00000524988	?	?	5
SIDT2	upstream gene variant, downstream gene variant	1668	ENST00000524842	?	ENSP00000436983	2
SIDT2	upstream gene variant, downstream gene variant	975	ENST00000531255	?	?	5
SIDT2	upstream gene variant, downstream gene variant	1480	ENST00000525339	?	?	3
SIDT2	upstream gene variant, downstream gene variant	2048	ENST00000526813	?	?	4
SIDT2	upstream gene variant, downstream gene variant	912	ENST00000529484	?	?	4
SIDT2	upstream gene variant, downstream gene variant	995	ENST00000528397	?	?	4
SIDT2	upstream gene variant, downstream gene variant	972	ENST00000525065	?	?	4
SIDT2	upstream gene variant, downstream gene variant	904	ENST00000620360	?	ENSP00000482762	7
SIDT2	upstream gene variant, downstream gene variant	1066	ENST00000532062	?	ENSP00000432432	7
SIDT2	upstream gene variant, downstream gene variant	901	ENST00000278951	?	ENSP00000278951	7
SIDT2	upstream gene variant, downstream gene variant	479	ENST00000525478	?	ENSP00000435890	6
SIDT2	upstream gene variant, downstream gene variant	903	ENST00000324225	NM_001040455.1	ENSP00000314023	7
SIDT2	upstream gene variant, downstream gene variant	926	ENST00000530948	?	?	5
SIDT2	upstream gene variant, downstream gene variant	155	ENST00000527654	?	?	4
TAGLN	upstream gene variant, downstream gene variant	1049	ENST00000278968	NM_001001522.1	ENSP00000278968	5
TAGLN	upstream gene variant, downstream gene variant	1216	ENST00000530649	?	ENSP00000431941	7
TAGLN	upstream gene variant, downstream gene variant	812	ENST00000533863	?	?	7
TAGLN	upstream gene variant, downstream gene variant	897	ENST00000529622	?	ENSP00000432380	8
TAGLN	upstream gene variant, downstream gene variant	64	ENST00000532870	?	ENSP00000432282	8
TAGLN	upstream gene variant, downstream gene variant	976	ENST00000392951	NM_003186.3	ENSP00000376678	5
TAGLN	upstream gene variant, downstream gene variant	76	ENST00000529792	?	ENSP00000431862	5
TAGLN	upstream gene variant, downstream gene variant	1037	ENST00000525531	?	ENSP00000432054	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PAFAH1B2	ENST00000530272	NM_001184746.1	ENSP00000431365	7
PAFAH1B2	ENST00000529887	NM_001184747.1	ENSP00000434951	7
PAFAH1B2	ENST00000526888	?	?	7
PAFAH1B2	ENST00000419197	NM_001184748.1	ENSP00000388742	7
PAFAH1B2	ENST00000304808	?	ENSP00000304006	7
PAFAH1B2	ENST00000533206	?	?	7
PAFAH1B2	ENST00000527958	NM_002572.3	ENSP00000435289	7
PCSK7	ENST00000532810	?	?	1
PCSK7	ENST00000320934	NM_004716.2	ENSP00000325917	3
PCSK7	ENST00000540028	?	ENSP00000441944	3
PCSK7	ENST00000534529	?	?	3
PCSK7	ENST00000527037	?	?	3
PCSK7	ENST00000533135	?	?	3
SIDT2	ENST00000525339	?	?	5
SIDT2	ENST00000524842	?	ENSP00000436983	4
SIDT2	ENST00000526813	?	?	1
SIDT2	ENST00000532062	?	ENSP00000432432	3
SIDT2	ENST00000528397	?	?	1
SIDT2	ENST00000531353	?	ENSP00000435004	2
SIDT2	ENST00000525478	?	ENSP00000435890	2
SIDT2	ENST00000431081	?	ENSP00000399635	8
SIDT2	ENST00000278951	?	ENSP00000278951	8
SIDT2	ENST00000530948	?	?	1
SIDT2	ENST00000532960	?	ENSP00000431176	1
SIDT2	ENST00000628876	?	ENSP00000486114	7
SIDT2	ENST00000324225	NM_001040455.1	ENSP00000314023	8
SIDT2	ENST00000620360	?	ENSP00000482762	8
SIDT2	ENST00000529441	?	?	1
TAGLN	ENST00000278968	NM_001001522.1	ENSP00000278968	5
TAGLN	ENST00000530649	?	ENSP00000431941	2
TAGLN	ENST00000392951	NM_003186.3	ENSP00000376678	5
TAGLN	ENST00000529792	?	ENSP00000431862	3
TAGLN	ENST00000525531	?	ENSP00000432054	5

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
PCSK7	ENST00000320934	NM_004716.2	ENSP00000325917	1
PCSK7	ENST00000540028	?	ENSP00000441944	1
SIDT2	ENST00000532062	?	ENSP00000432432	1
SIDT2	ENST00000431081	?	ENSP00000399635	1
SIDT2	ENST00000324225	NM_001040455.1	ENSP00000314023	1
SIDT2	ENST00000620360	?	ENSP00000482762	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
TAGLN	ENST00000532870	?	ENSP00000432282	1
TAGLN	ENST00000530649	?	ENSP00000431941	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AP000892.6	ENST00000624094	?	2
PAFAH1B2	ENST00000533206	?	1
PCSK7	ENST00000529458	?	1
PCSK7	ENST00000528973	?	3
PCSK7	ENST00000534529	?	4
SIDT2	ENST00000529484	?	1
SIDT2	ENST00000531255	?	1
TAGLN	ENST00000533863	?	2