Locus 342 (rs2523987)

SNiPAcard

Block annotations

Block info

genomic range	chr6:30,075,103-30,103,360
block size	28,258 bp
variant count	27 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.710 [-5.32 – 0.731]	gene(s) hit or close-by	SNORA48, TRIM31, TRIM31-AS1, TRIM40
phastCons	μ = 0.018 [0 – 0.413]	eQTL gene(s)	BTN3A2, BTN3A3, HCG22, HCG4P11, HIST1H3I, HLA-A, HLA-C, HLA-DQA1, HLA-DQB1, HLA-F, HLA-F-AS1, HLA-G, HLA-H, HLA-J, HLA-K, HLA-U, HLA-V, IER3, LINC00243, PPP1R11, PPP1R18, TRIM31, TRIM31-AS1, ZFP57, ZNRD1-AS1
GERP++	μ = -0.849 [-5.14 – 1.74]	potentially regulated gene(s)	TRIM31, TRIM31, TRIM31, TRIM31, TRIM31, TRIM31, TRIM31, TRIM31
CADD score	μ = 4.371 [0.332 – 13.43]	disease gene(s)	HLA-DQB1, ZFP57, HLA-A

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
HLA-DQB1	Bullous pemphigoid	OrphaNet	OrphaNet:703
HLA-DQB1	Narcolepsy-cataplexy	OrphaNet	OrphaNet:2073
HLA-DQB1	Narcolepsy without cataplexy	OrphaNet	OrphaNet:83465
ZFP57	Transient neonatal diabetes mellitus	OrphaNet	OrphaNet:99886
HLA-A	Birdshot chorioretinopathy	OrphaNet	OrphaNet:179
ZFP57	DIABETES MELLITUS, TRANSIENT NEONATAL, 1	OMIM	MIM:601410
ZFP57	Diabetes Mellitus, 6q24-Related Transient Neonatal	DECIPHER	MIM:601410

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)
TRIM31	missense variant	ENST00000376734	NM_007028.3	ENSP00000365924	I/V	Att/Gtt	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
?	?	ILMN_1654987	blood	6.16×10-6 (p-value)	MuTHER consortium	2
			adipocyte	4.86×10-6 (p-value)		
TRIM31	ENST00000376734	ILMN_1748685	skin	6.26×10-7 (p-value)	MuTHER consortium	17
TRIM31	ENST00000484583					
TRIM31	ENST00000471695					
TRIM31	ENST00000468264					
HLA-V	ENST00000446817	ILMN_1660923	adipocyte	6.57×10-7 (p-value)	MuTHER consortium	18

HLA-V	ENST00000429037		blood	3.42×10-6 (p-value)	MuTHER consortium	18
HCG22	ENST00000570223	ILMN_1667229	blood	2.42×10-5 (p-value)	MuTHER consortium	1
HCG22	ENST00000565192					
HCG22	ENST00000615046					
HCG22	ENST00000426185					
HCG22	ENST00000562344					
HLA-J	?	ENSG00000204622	lung	4.30×10-12 (p-value)	GTEx Portal V6	27
TRIM31-AS1	?	ENSG00000231226	lung	6.84×10-6 (p-value)	GTEx Portal V6	1
HLA-H	?	ENSG00000206341	lung	2.51×10-6 (p-value)	GTEx Portal V6	13
TRIM31	?	ENSG00000204616	lung	3.26×10-8 (p-value)	GTEx Portal V6	8
HLA-U	?	ENSG00000228078	lung	6.51×10-6 (p-value)	GTEx Portal V6	2
HLA-G	?	ENSG00000204632	lung	8.97×10-7 (p-value)	GTEx Portal V6	3
ZFP57	?	ENSG00000204644	lung	8.97×10-7 (p-value)	GTEx Portal V6	1
ZNRD1-AS1	?	ENSG00000204623	tibial nerve	1.75×10-10 (p-value)	GTEx Portal V6	26
HLA-U	?	ENSG00000228078	tibial nerve	3.89×10-6 (p-value)	GTEx Portal V6	18
HLA-F-AS1	?	ENSG00000214922	blood	1.11×10-9 (p-value)	GTEx Portal V6	27
LINC00243	?	ENSG00000214894	blood	4.32×10-9 (p-value)	GTEx Portal V6	22
HLA-J	?	ENSG00000204622	blood	2.28×10-6 (p-value)	GTEx Portal V6	12
HLA-H	?	ENSG00000206341	blood	3.17×10-7 (p-value)	GTEx Portal V6	20
ZFP57	?	ENSG00000204644	blood	3.49×10-7 (p-value)	GTEx Portal V6	2
HLA-K	?	ENSG00000230795	blood	3.41×10-6 (p-value)	GTEx Portal V6	2
ZNRD1-AS1	?	ENSG00000204623	tibial artery	6.13×10-9 (p-value)	GTEx Portal V6	26
HLA-U	?	ENSG00000228078	tibial artery	2.93×10-9 (p-value)	GTEx Portal V6	26
HLA-J	?	ENSG00000204622	tibial artery	7.46×10-8 (p-value)	GTEx Portal V6	24
ZFP57	?	ENSG00000204644	tibial artery	1.09×10-5 (p-value)	GTEx Portal V6	1
HLA-J	?	ENSG00000204622	thyroid	2.05×10-7 (p-value)	GTEx Portal V6	26
ZNRD1-AS1	?	ENSG00000204623	thyroid	1.18×10-5 (p-value)	GTEx Portal V6	12
ZFP57	?	ENSG00000204644	thyroid	1.78×10-8 (p-value)	GTEx Portal V6	18
PPP1R18	?	ENSG00000146112	thyroid	8.21×10-8 (p-value)	GTEx Portal V6	2
HLA-J	?	ENSG00000204622	skeletal muscle	2.77×10-11 (p-value)	GTEx Portal V6	27
HLA-U	?	ENSG00000228078	skeletal muscle	5.12×10-6 (p-value)	GTEx Portal V6	2
HLA-H	?	ENSG00000206341	skeletal muscle	4.48×10-6 (p-value)	GTEx Portal V6	13
HLA-F	?	ENSG00000204642	cerebellum	5.51×10-6 (p-value)	GTEx Portal V6	2
HLA-J	?	ENSG00000204622	cerebellum	3.63×10-6 (p-value)	GTEx Portal V6	5
HCG4P11	?	ENSG00000225864	cerebellum	9.28×10-6 (p-value)	GTEx Portal V6	1
HLA-H	?	ENSG00000206341	transformed fibroblasts	1.75×10-6 (p-value)	GTEx Portal V6	7
ZFP57	?	ENSG00000204644	transformed fibroblasts	1.94×10-5 (p-value)	GTEx Portal V6	1
HLA-J	?	ENSG00000204622	breast	1.76×10-7 (p-value)	GTEx Portal V6	15

?	?	ILMN_1679389	b-cell	3.71×10-5 (p-value)	Fairfax et al.	3
IER3	ENST00000376377	ILMN_1682717	monocyte	1.18×10-4 (p-value)	Fairfax et al.	6
IER3	ENST00000259874					
HIST1H3I	ENST00000616365	ILMN_2207865	b-cell	2.37×10-4 (p-value)	Fairfax et al.	1
PPP1R11	ENST00000376765	ILMN_1747598	blood	1.31×10-6 (p-value)	Westra et al.	7
PPP1R11	ENST00000376769					
PPP1R11	ENST00000376763					
PPP1R11	ENST00000376773					
PPP1R11	ENST00000376758					
PPP1R11	ENST00000376772					
ZNRD1-AS1	?	ENSG00000204623	sun exposed skin	7.33×10-6 (p-value)	GTEx Portal V6	2
HLA-J	?	ENSG00000204622	sun exposed skin	3.65×10-7 (p-value)	GTEx Portal V6	24
ZFP57	?	ENSG00000204644	sun exposed skin	2.39×10-6 (p-value)	GTEx Portal V6	2
HLA-J	?	ENSG00000204622	left ventricle	3.77×10-6 (p-value)	GTEx Portal V6	2
HLA-J	?	ENSG00000204622	aorta	4.32×10-7 (p-value)	GTEx Portal V6	6
HLA-H	?	ENSG00000206341	aorta	1.43×10-5 (p-value)	GTEx Portal V6	1
HLA-U	?	ENSG00000228078	aorta	9.62×10-7 (p-value)	GTEx Portal V6	17
ZFP57	?	ENSG00000204644	aorta	2.15×10-8 (p-value)	GTEx Portal V6	18
?	?	ILMN_1666078	monocyte	2.04×10-21 (p-value)	Zeller et al.	3
?	?	ILMN_1671054	monocyte	1.92×10-32 (p-value)	Zeller et al.	3
BTN3A2	ENST00000396934	ILMN_1676528	monocyte	1.37×10-25 (p-value)	Zeller et al.	3
BTN3A2	ENST00000508906					
BTN3A2	ENST00000356386					
?	?	ILMN_1820787	monocyte	4.51×10-17 (p-value)	Zeller et al.	3
BTN3A2	ENST00000524459	ILMN_1700067	monocyte	8.40×10-22 (p-value)	Zeller et al.	3
BTN3A2	ENST00000528541					
BTN3A3	ENST00000467524					
BTN3A3	ENST00000482451					
BTN3A3	ENST00000494393					
BTN3A2	ENST00000527417					
BTN3A3	ENST00000244519					
BTN3A2	ENST00000528222					
BTN3A3	ENST00000474790					
BTN3A2	ENST00000377708					
BTN3A3	ENST00000496719					
BTN3A2	ENST00000396948					
BTN3A2	ENST00000527422					
BTN3A2	ENST00000356386					
?	?	ILMN_1883997	monocyte	1.50×10-36 (p-value)	Zeller et al.	3
?	?	ILMN_1679389	monocyte	4.26×10-12 (p-value)	Zeller et al.	1
ZNRD1-AS1	?	ENSG00000204623	spleen	1.62×10-6 (p-value)	GTEx Portal V6	11
HLA-J	?	ENSG00000204622	esophagus mucosa	9.67×10-6 (p-value)	GTEx Portal V6	12
ZFP57	?	ENSG00000204644	esophagus mucosa	1.51×10-8 (p-value)	GTEx Portal V6	17
ZNRD1-AS1	?	ENSG00000204623	muscularis mucosae	1.12×10-5 (p-value)	GTEx Portal V6	4

ZNRD1-AS1	?	ENSG00000204623	subcutaneous adipocytes	1.34×10^{-5} (p-value)	GTEx Portal V6	2
ZFP57	?	ENSG00000204644	subcutaneous adipocytes	1.16×10^{-5} (p-value)	GTEx Portal V6	1
HLA-A	?	ENSG00000206503	brain	4.30×10^{-2} (q-value)	SeeQTL DB (Myers et al.)	1
ZFP57	?	ENSG00000204644	visceral adipocytes	7.76×10^{-6} (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
BTN3A2	ENST00000524682	209846_s_at	chr6	blood	7.60×10^{-9} (p-value)	Dixon et al.	1
BTN3A2	ENST00000604202						
BTN3A2	ENST00000396934						
BTN3A2	ENST00000532294						
BTN3A2	ENST00000508906						
BTN3A2	ENST00000377708						
BTN3A2	ENST00000527422						
BTN3A2	ENST00000531055						
BTN3A2	ENST00000396948						
BTN3A2	ENST00000356386						
HLA-DQB1	ENST00000399082	209480_at	chr6	blood	1.80×10^{-9} (p-value)	Dixon et al.	3
HLA-DQB1	ENST00000399084						
HLA-DQB1	ENST00000434651						
HLA-DQB1	ENST00000460185						
HLA-DQB1	ENST00000399079						
HLA-DQB1	ENST00000443574						
HLA-DQB1	ENST00000374943						
HLA-DQB1	ENST00000487676						
HLA-DQA1	ENST00000374949	213831_at	chr6	blood	7.10×10^{-8} (p-value)	Dixon et al.	1
HLA-DQA1	ENST00000496318						
HLA-DQA1	ENST00000460633						
HLA-DQA1	ENST00000482745						
HLA-DQA1	ENST00000343139						
HLA-C	ENST00000470363	216526_x_at	chr6	blood	9.60×10^{-8} (p-value)	Dixon et al.	1
HLA-C	ENST00000383329						
HLA-C	ENST00000487245						
HLA-C	ENST00000376228						
HLA-C	ENST00000466892						
HLA-C	ENST00000376237						
HLA-C	ENST00000620806						
?	?	ILMN_1752592	chr6	monocyte	6.28×10^{-21} (p-value)	Zeller et al.	3
HLA-C	ENST00000470363	ILMN_2150787	chr6	monocyte	3.65×10^{-13} (p-value)	Zeller et al.	3
HLA-C	ENST00000383329						
HLA-C	ENST00000487245						
HLA-C	ENST00000376228						
HLA-C	ENST00000376237						
HLA-C	ENST00000620806						
HLA-C	ENST00000466892						

Putative effect on regulation

ENCODE promoter-associated DHS

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

ENCP00000047880	TRIM31
	TRIM31
	TRIM31
	TRIM31
	TRIM31
	TRIM31
	TRIM31

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000487603 (promoter flanking region)	5	embryonic stem cell (H1ESC)	H3K27me3
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		muscle (HSMM)	H3K27me3
		liver (HepG2)	H3K4me1, H3K4me2
		lung (IMR90)	H3K27me3
		blood (GM12878)	EBF1
		nervous (NH-A)	H3K27me3
		skin (NHEK)	H3K27me3
		NHLF	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		A549	H3K4me3
ENSR00001702952 (enhancer)	2	NHLF	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		muscle (HSMM)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K4me1, H3K4me2
		lung (IMR90)	H3K27me3
		nervous (NH-A)	H3K27me3
		skin (NHEK)	H3K27me3
ENSR00001702953 (CTCF binding site)	4	monocytes (Monocytes-CD14+)	H3K27me3
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SNORA48	upstream gene variant	2617	ENST00000516419	?	?	1
TRIM31	downstream gene variant, upstream gene variant	510	ENST00000480808	?	?	17
TRIM31	upstream gene variant	30	ENST00000485864	?	?	19
TRIM31	downstream gene variant, upstream gene variant	22	ENST00000493404	?	?	22
TRIM31	upstream gene variant	2299	ENST00000468264	?	?	9
TRIM31	upstream gene variant	1669	ENST00000471695	?	?	3
TRIM31	upstream gene variant	1886	ENST00000484583	?	?	1
TRIM31	upstream gene variant	73	ENST00000376734	NM_007028.3	ENSP00000365924	16
TRIM31-AS1	downstream gene variant	187	ENST00000440874	?	?	10
TRIM40	upstream gene variant	525	ENST00000396581	NM_001286633.1	ENSP00000379826	1
TRIM40	upstream gene variant	541	ENST00000489892	?	?	1

TRIM40	upstream gene variant	1427	ENST00000307859	NM_138700.4	ENSP00000308310	1
TRIM40	upstream gene variant	1148	ENST00000376724	?	ENSP00000365914	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
TRIM31	ENST00000376734	NM_007028.3	ENSP00000365924	3	3	3

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TRIM31	ENST00000376734	NM_007028.3	ENSP00000365924	8
TRIM31	ENST00000485864	?	?	6
TRIM31	ENST00000468264	?	?	1
TRIM31	ENST00000480808	?	?	5
TRIM31	ENST00000493404	?	?	3
TRIM31-AS1	ENST00000440874	?	?	15

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
TRIM31	ENST00000485864	?	4
TRIM31	ENST00000480808	?	4
TRIM31	ENST00000493404	?	2