Locus 311 (rs4356932)

SNiPAcard

Block annotations

Block info

genomic range	chr4:76,922,988-76,997,258
block size	74,271 bp
variant count	265 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.627 [-4.521 – 1.846]	gene(s) hit or close-by	ART3, CXCL10, CXCL11, CXCL9, RP11-630D6.1, RP11-630D6.5
phastCons	μ = 0.100 [0 – 1]	eQTL gene(s)	ART3, CXCL10, CXCL11, NAAA, NUP54, SLC6A20
GERP++	μ = -0.174 [-6.98 – 3.47]	potentially regulated gene(s)	ART3, CCDC158, CXCL11, CXCL9, G3BP2, NAAA, NUP54, PPEF2, RP11-630D6.5, SCARB2, SDAD1, USO1
CADD score	μ = 3.773 [0.088 – 15.97]	disease gene(s)	SLC6A20, SCARB2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
SLC6A20	HYPERGLYCINURIA	OMIM	MIM:138500
SLC6A20	IMINOGLYCINURIA	OMIM	MIM:242600
SCARB2	EPILEPSY, PROGRESSIVE MYOCLONIC, 4, WITH OR WITHOUT RENAL [...]	OMIM	MIM:254900
SLC6A20	IMINOGLYCINURIA	OrphaNet	OrphaNet:42062
SCARB2	Gaucher disease type 1	OrphaNet	OrphaNet:77259
SCARB2	Action myoclonus - renal failure syndrome	OrphaNet	OrphaNet:163696
SCARB2	Unverricht-Lundborg disease	OrphaNet	OrphaNet:308

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
NAAA	ENST00000286733	ILMN_2391512	skin	1.23×10^{-6} (p-value)	MuTHER consortium	55
NAAA	ENST00000602782		blood	2.46×10^{-35} (p-value)	MuTHER consortium	55
NAAA	ENST00000513045		adipocyte	8.04×10^{-14} (p-value)	MuTHER consortium	55
NAAA	ENST00000507956					
NAAA	ENST00000505594					
NAAA	ENST00000511606					
CXCL10	ENST00000306602	ILMN_1791759	blood	4.51×10^{-11} (p-value)	MuTHER consortium	55
NAAA	?	ENSG00000138744	atrial appendage	2.28×10^{-10} (p-value)	GTEx Portal V6	255
NAAA	?	ENSG00000138744	transformed fibroblasts	5.01×10^{-41} (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	blood	3.49×10^{-22} (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	breast	2.46×10^{-17} (p-value)	GTEx Portal V6	256
NAAA	ENST00000513045	ILMN_2391512	monocyte	6.55×10^{-14} (p-value)	Fairfax et al.	10
NAAA	ENST00000602782		b-cell	4.77×10^{-11} (p-value)	Fairfax et al.	10
NAAA	ENST00000286733					

NAAA	ENST00000507956					
NAAA	ENST00000505594					
NAAA	ENST00000511606					
?	?	ILMN_1668605	monocyte	8.79×10-11 (p-value)	Fairfax et al.	10
			b-cell	1.35×10-10 (p-value)		
CXCL10	ENST00000306602	ILMN_1791759	monocyte	3.84×10-4 (p-value)	Fairfax et al.	10
NAAA	?	ENSG00000138744	skeletal muscle	3.24×10-37 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	EBV lymphocytes	6.31×10-8 (p-value)	GTEx Portal V6	250
CXCL10	?	ENSG00000169245	EBV lymphocytes	5.98×10-6 (p-value)	GTEx Portal V6	1
NAAA	?	ENSG00000138744	blood	4.02×10-8 (q-value)	SeeQTL DB (HapMap)	25
CXCL10	?	ENSG00000169245	blood	2.54×10-2 (q-value)	SeeQTL DB (HapMap)	5
NAAA	?	ENSG00000138744	ovary	1.86×10-11 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	sun exposed skin	8.67×10-36 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	unexposed skin	1.46×10-18 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	aorta	2.26×10-20 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	left ventricle	3.49×10-13 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	subcutaneous adipocytes	2.09×10-28 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	stomach	5.02×10-11 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	visceral adipocytes	1.47×10-15 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	cortex	5.43×10-8 (p-value)	GTEx Portal V6	212
ART3	?	ENSG00000156219	tibial nerve	1.44×10-63 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	tibial nerve	2.52×10-24 (p-value)	GTEx Portal V6	256
NUP54	?	ENSG00000138750	tibial nerve	1.17×10-5 (p-value)	GTEx Portal V6	4
NAAA	?	ENSG00000138744	esophagus mucosa	8.13×10-21 (p-value)	GTEx Portal V6	256
ART3	?	ENSG00000156219	pituitary	2.55×10-11 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	pituitary	2.63×10-6 (p-value)	GTEx Portal V6	22
NUP54	?	ENSG00000138750	pituitary	7.59×10-9 (p-value)	GTEx Portal V6	243
NAAA	?	ENSG00000138744	testis	1.89×10-17 (p-value)	GTEx Portal V6	256
ART3	?	ENSG00000156219	pancreas	4.23×10-42 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	pancreas	1.06×10-22 (p-value)	GTEx Portal V6	256
CXCL11	?	ENSG00000169248	pancreas	4.64×10-21 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	muscularis mucosae	4.58×10-30 (p-value)	GTEx Portal V6	256
ART3	?	ENSG00000156219	muscularis mucosae	1.11×10-5 (p-value)	GTEx Portal V6	1
NAAA	?	ENSG00000138744	lung	1.67×10-23 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	tibial artery	9.98×10-31 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	thyroid	2.39×10-13 (p-value)	GTEx Portal V6	256
NUP54	?	ENSG00000138750	thyroid	5.69×10-6 (p-value)	GTEx Portal V6	17

NAAA	?	ENSG00000138744	transverse colon	1.13×10-9 (p-value)	GTEx Portal V6	255
NAAA	?	ENSG00000138744	terminal ileum	7.67×10-8 (p-value)	GTEx Portal V6	233
NAAA	?	ENSG00000138744	coronary artery	5.31×10-15 (p-value)	GTEx Portal V6	256
ART3	?	ENSG00000156219	sigmoid colon	3.42×10-8 (p-value)	GTEx Portal V6	244
NAAA	?	ENSG00000138744	sigmoid colon	2.81×10-13 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	vagina	3.01×10-8 (p-value)	GTEx Portal V6	234
NAAA	?	ENSG00000138744	gastroesophageal junction	1.32×10-12 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	adrenal gland	3.14×10-18 (p-value)	GTEx Portal V6	256
NAAA	ENST00000602782	214765_s_at	blood	1.40×10-23 (p-value)	Dixon et al.	16
NAAA	ENST00000286733					
NAAA	ENST00000513045					
NAAA	ENST00000507956					
NAAA	ENST00000505594					
NAAA	ENST00000511606					
NAAA	ENST00000286733	227135_at	blood	7.30×10-21 (p-value)	Dixon et al.	16
NAAA	ENST00000511606					
CXCL10	ENST00000306602	204533_at	blood	2.60×10-9 (p-value)	Dixon et al.	14
NAAA	?	ENSG00000138744	frontal cortex	5.99×10-7 (p-value)	GTEx Portal V6	91
?	?	ILMN_2067890	monocyte	4.60×10-64 (p-value)	Zeller et al.	14
NAAA	ENST00000286733	ILMN_2391512	monocyte	5.69×10-38 (p-value)	Zeller et al.	14
NAAA	ENST00000602782					
NAAA	ENST00000513045					
NAAA	ENST00000507956					
NAAA	ENST00000505594					
NAAA	ENST00000511606					
?	?	ILMN_1668605	monocyte	2.02×10-35 (p-value)	Zeller et al.	14
?	?	ILMN_2285568	monocyte	1.10×10-33 (p-value)	Zeller et al.	14
CXCL10	ENST00000306602	ILMN_1791759	monocyte	2.69×10-12 (p-value)	Zeller et al.	9
NAAA	?	ENSG00000138744	cerebellum	3.44×10-6 (p-value)	GTEx Portal V6	12
NAAA	?	ENSG00000138744	uterus	4.67×10-7 (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
SLC6A20	ENST00000353278	ILMN_1698585	chr3	monocyte	6.46×10-13 (p-value)	Zeller et al.	11
SLC6A20	ENST00000456124						
SLC6A20	ENST00000358525						

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000655593	1	ENST00000504914, ENST00000513353, ENST00000341029, ENST00000513122	ART3

FFCP00000655602	1	ENST00000504914, ENST00000513353, ENST00000341029, ENST00000513122	ART3

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000041424		
ENCP00000041429		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000382631	1	ENCP00000041405	G3BP2
		ENCP00000041421	SDAD1
		ENCP00000041417	NAAA
		ENCP00000041404	G3BP2
ENCE00000382671	1	ENCP00000041434	ART3
		ENCP00000041413	PPEF2
		ENCP00000041412	USO1
		ENCP00000041414	PPEF2
ENCE00000382734	1	ENCP00000041421	SDAD1
		ENCP00000041417	NAAA
		ENCP00000041440	NUP54
ENCE00000382736	1	ENCP00000041421	SDAD1
		ENCP00000041417	NAAA
ENCE00000382737	2	ENCP00000041422	RP11-630D6.5
		ENCP00000041421	SDAD1
		ENCP00000041443	SCARB2
		ENCP00000041417	NAAA
		ENCP00000041440	NUP54
ENCE00000382672	1	ENCP00000041434	ART3
		ENCP00000041413	PPEF2
		ENCP00000041412	USO1
		ENCP00000041414	PPEF2
ENCE00000382653	2	ENCP00000041411	USO1
		ENCP00000041425	ART3
ENCE00000382696	1	ENCP00000041438	NUP54
		ENCP00000041415	NAAA
		ENCP00000041452	CCDC158
ENCE00000382738	2	ENCP00000041443	SCARB2
		ENCP00000041421	SDAD1
		ENCP00000041417	NAAA

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001685894 (enhancer)	1	HSMMtube	H3K27me3
ENSR00001243218 (promoter flanking region)	2	NHLF	H3K27ac
		embryonic stem cell (H1ESC)	H3K27me3, DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K4me2, H3K27ac
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		breast (HMEC)	H3K27ac, H3K4me2
		blood (GM12878)	H2AZ, H3K27ac, H3K4me2
		lung (IMR90)	DNase1, H3K18ac, H3K27ac, H3K4ac, H2BK120ac, H2BK20ac, H4K91ac, H3K56ac
		skin (NHEK)	DNase1

ENSR00001243224 (promoter flanking region)	2	HSMMtube	H3K27me3
		Osteobl	H3K27me3, H3K4me2
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K4me3
		monocytes (Monocytes-CD14+)	H3K4me3
		endothelium (HUVEC)	H3K4me2
		A549	H3K27me3
		nervous (NH-A)	H3K27me3
		skin (NHEK)	H3K4me2
ENSR00001685897 (enhancer)	2	monocytes (Monocytes-CD14+)	DNase1
		Osteobl	H3K27me3
		A549	H3K27me3
		blood (DND-41)	H3K27me3
ENSR00001243225 (enhancer)	1	endothelium (HUVEC)	Cjun, DNase1
		HSMMtube	H3K27me3
ENSR00001243226 (enhancer)	1	embryonic stem cell (H1ESC)	CTCF, Yy1, DNase1
		endothelium (HUVEC)	H3K36me3
		HSMMtube	H3K27me3
		skin (NHEK)	CTCF
ENSR00001685898 (CTCF binding site)	1	endothelium (HUVEC)	H3K36me3
		embryonic stem cell (H1ESC)	CTCF, Yy1, DNase1
		HSMMtube	H3K27me3
		skin (NHEK)	CTCF
ENSR00001243231 (enhancer)	1	NHLF	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (GM12878)	Cjun, BATF, DNase1
		nervous (NH-A)	H3K27me3
		A549	H3K27me3
ENSR00001432212 (enhancer)	7	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		breast (HMEC)	H3K4me2
		monocytes (Monocytes-CD14+)	H3K4me1
		endothelium (HUVEC)	H3K27me3
		blood (GM12878)	Cjun, BATF, MEF2A, Jund
		nervous (NH-A)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K4me1, H3K4me2
ENSR00001685900 (promoter flanking region)	4	HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		breast (HMEC)	H3K4me2, DNase1
		monocytes (Monocytes-CD14+)	H3K4me1, DNase1
		endothelium (HUVEC)	H3K27me3
		nervous (NH-A)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	DNase1, H3K4me1, H3K4me2
ENSR00001432213 (CTCF binding site)	1	HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		breast (HMEC)	DNase1, H3K4me2
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1
		endothelium (HUVEC)	H3K27me3
		nervous (NH-A)	H3K27me3

	A549	H3K27me3
	skin (NHEK)	DNase1, H3K4me1, H3K4me2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ART3	upstream gene variant	941	ENST00000510423	?	ENSP00000425327	2
ART3	upstream gene variant	352	ENST00000341029	NM_001130017.2	ENSP00000343843	2
ART3	upstream gene variant	336	ENST00000513353	?	ENSP00000421345	2
ART3	upstream gene variant	76	ENST00000504112	?	?	2
ART3	upstream gene variant	1249	ENST00000504914	?	ENSP00000421431	2
ART3	upstream gene variant	401	ENST00000513122	?	ENSP00000422287	2
ART3	upstream gene variant	352	ENST00000510669	?	?	2
ART3	upstream gene variant	76	ENST00000395688	?	?	2
ART3	upstream gene variant	76	ENST00000506667	?	?	2
ART3	upstream gene variant	400	ENST00000355810	NM_001130016.2	ENSP00000348064	2
ART3	upstream gene variant	403	ENST00000513494	?	?	2
ART3	upstream gene variant	400	ENST00000349321	NM_001179.5	ENSP00000304313	2
CXCL10	downstream gene variant, upstream gene variant	452	ENST00000306602	NM_001565.3	ENSP00000305651	21
CXCL11	downstream gene variant, upstream gene variant	38	ENST00000503860	?	ENSP00000425819	73
CXCL11	downstream gene variant, upstream gene variant	208	ENST00000306621	NM_005409.4, NM_001302123.1	ENSP00000306884	46
CXCL9	upstream gene variant	560	ENST00000264888	NM_002416.1	ENSP00000354901	3
RP11-630D6.1	downstream gene variant, upstream gene variant	117	ENST00000477971	?	?	48
RP11-630D6.5	downstream gene variant	2106	ENST00000501239	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ART3	ENST00000513494	?	?	2
ART3	ENST00000341029	NM_001130017.2	ENSP00000343843	261
ART3	ENST00000504112	?	?	1
ART3	ENST00000513353	?	ENSP00000421345	261
ART3	ENST00000504914	?	ENSP00000421431	261
ART3	ENST00000510669	?	?	261
ART3	ENST00000513122	?	ENSP00000422287	261
ART3	ENST00000506667	?	?	1
ART3	ENST00000395688	?	?	1
ART3	ENST00000349321	NM_001179.5	ENSP00000304313	2
ART3	ENST00000510423	?	ENSP00000425327	2
ART3	ENST00000355810	NM_001130016.2	ENSP00000348064	2
CXCL10	ENST00000306602	NM_001565.3	ENSP00000305651	7
CXCL11	ENST00000503860	?	ENSP00000425819	25

CXCL11	ENST00000306621	NM_005409.4, NM_001302123.1	ENSP00000306884	15
CXCL9	ENST00000264888	NM_002416.1	ENSP00000354901	2
RP11-630D6.5	ENST00000501239	?	?	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CXCL10	ENST00000306602	NM_001565.3	ENSP00000305651	2
CXCL11	ENST00000503860	?	ENSP00000425819	1
CXCL11	ENST00000306621	NM_005409.4, NM_001302123.1	ENSP00000306884	11
CXCL9	ENST00000264888	NM_002416.1	ENSP00000354901	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CXCL11	ENST00000503860	?	ENSP00000425819	2
CXCL11	ENST00000306621	NM_005409.4, NM_001302123.1	ENSP00000306884	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-630D6.1	ENST00000477971	?	1