SNiPAcard

Block annotations

Block info

genomic range	chr4:76,852,839-76,985,488
block size	132,650 bp
variant count	62 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.292 [-4.482 – 2.568]	**gene(s) hit or close-by**	AC110615.1, AC112719.2, ART3, CXCL11, CXCL9, NAAA, RP11-630D6.1, RP11-630D6.5, SDAD1
phastCons	μ = 0.119 [0 – 1]	**eQTL gene(s)**	ART3, CXCL10, CXCL11, CXCL9, NAAA, SCARB2
GERP++	μ = -0.578 [-10.3 – 4.28]	**potentially regulated gene(s)**	CCDC158, NAAA, NUP54, USO1
CADD score	μ = 4.078 [0.03 – 18.35]	**disease gene(s)**	SCARB2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Heart rate	1.52×10^{-5}	dbGaP	pha001394	dbGaP	1

Disease gene annotation

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

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)
NAAA	missense variant	ENST00000505594	?	ENSP00000426977	I/V	Atc/Gtc	?	?	1
NAAA	missense variant	ENST00000602782	?	ENSP00000473575	I/V	Atc/Gtc	?	?	1
NAAA	missense variant	ENST00000286733	NM_014435.3	ENSP00000286733	I/V	Atc/Gtc	?	?	1
NAAA	missense variant	ENST00000507956	?	ENSP00000427641	I/V	Atc/Gtc	?	?	1
NAAA	missense variant	ENST00000507187	?	ENSP00000423142	I/V	Atc/Gtc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ART3	?	ENSG00000156219	pancreas	6.72×10^{-15} (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	pancreas	9.25×10^{-12} (p-value)	GTEx Portal V6	62

CXCL11	?	ENSG00000169248	pancreas	1.02×10-6 (p-value)	GTEx Portal V6	24
NAAA	?	ENSG00000138744	muscularis mucosae	1.64×10-18 (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	lung	6.09×10-21 (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	transformed fibroblasts	8.78×10-15 (p-value)	GTEx Portal V6	62
SCARB2	?	ENSG00000138760	transformed fibroblasts	1.06×10-6 (p-value)	GTEx Portal V6	6
NAAA	?	ENSG00000138744	blood	1.63×10-22 (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	breast	2.40×10-15 (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	tibial artery	7.45×10-23 (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	thyroid	6.38×10-15 (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	skeletal muscle	6.09×10-27 (p-value)	GTEx Portal V6	62
CXCL10	?	ENSG00000169245	EBV lymphocytes	2.93×10-9 (p-value)	GTEx Portal V6	42
NAAA	?	ENSG00000138744	transverse colon	1.61×10-11 (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	blood	1.05×10-6 (q-value)	SeeQTL DB (HapMap)	16
CXCL10	?	ENSG00000169245	blood	7.80×10-4 (q-value)	SeeQTL DB (HapMap)	12
NAAA	?	ENSG00000138744	sun exposed skin	3.52×10-24 (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	unexposed skin	5.99×10-17 (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	aorta	6.37×10-22 (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	subcutaneous adipocytes	4.54×10-18 (p-value)	GTEx Portal V6	62
?	?	ILMN_2067890	monocyte	1.57×10-17 (p-value)	Zeller et al.	9
NAAA	ENST00000286733	ILMN_2391512	monocyte	2.13×10-19 (p-value)	Zeller et al.	9
NAAA	ENST00000602782					
NAAA	ENST00000513045					
NAAA	ENST00000507956					
NAAA	ENST00000505594					
NAAA	ENST00000511606					
?	?	ILMN_1668605	monocyte	4.68×10-15 (p-value)	Zeller et al.	5
?	?	ILMN_2285568	monocyte	4.21×10-87 (p-value)	Zeller et al.	9
NAAA	?	ENSG00000138744	caudate basal ganglia	1.41×10-8 (p-value)	GTEx Portal V6	52
NAAA	?	ENSG00000138744	stomach	3.34×10-13 (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	visceral adipocytes	2.50×10-21 (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	sigmoid colon	1.05×10-9 (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	coronary artery	6.63×10-10 (p-value)	GTEx Portal V6	62
ART3	?	ENSG00000156219	tibial nerve	3.51×10-9 (p-value)	GTEx Portal V6	60
NAAA	?	ENSG00000138744	tibial nerve	1.59×10-11 (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	esophagus mucosa	8.68×10-25 (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	gastroesophageal junction	2.64×10-12 (p-value)	GTEx Portal V6	62

NAAA	?	ENSG00000138744	adrenal gland	1.07×10-16 (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	left ventricle	2.30×10-9 (p-value)	GTEx Portal V6	60
NAAA	?	ENSG00000138744	atrial appendage	9.88×10-9 (p-value)	GTEx Portal V6	57
NAAA	?	ENSG00000138744	ovary	9.96×10-7 (p-value)	GTEx Portal V6	43
NAAA	?	ENSG00000138744	spleen	1.51×10-7 (p-value)	GTEx Portal V6	45
NAAA	?	ENSG00000138744	pituitary	3.06×10-7 (p-value)	GTEx Portal V6	56
NAAA	?	ENSG00000138744	frontal cortex	2.61×10-7 (p-value)	GTEx Portal V6	42
NAAA	?	ENSG00000138744	liver	2.36×10-7 (p-value)	GTEx Portal V6	32
NAAA	ENST00000286733	ILMN_2391512	blood	7.55×10-28 (p-value)	MuTHER consortium	21
NAAA	ENST00000602782					
NAAA	ENST00000513045					
NAAA	ENST00000507956					
NAAA	ENST00000505594					
NAAA	ENST00000511606					
CXCL9	ENST00000264888	ILMN_1745356	blood	1.93×10-6 (p-value)	MuTHER consortium	16
CXCL10	ENST00000306602	ILMN_1791759	blood	3.78×10-35 (p-value)	MuTHER consortium	21
NAAA	ENST00000513045	ILMN_2391512	monocyte	8.05×10-13 (p-value)	Fairfax et al.	3
NAAA	ENST00000602782		b-cell	1.98×10-6 (p-value)	Fairfax et al.	3
NAAA	ENST00000286733					
NAAA	ENST00000507956					
NAAA	ENST00000505594					
NAAA	ENST00000511606					
?	?	ILMN_1668605	monocyte	1.09×10-12 (p-value)	Fairfax et al.	6
			b-cell	5.29×10-6 (p-value)		
NAAA	ENST00000602782	214765_s_at	blood	2.10×10-22 (p-value)	Dixon et al.	7
NAAA	ENST00000286733					
NAAA	ENST00000513045					
NAAA	ENST00000507956					
NAAA	ENST00000505594					
NAAA	ENST00000511606					
NAAA	ENST00000286733	227135_at	blood	2.50×10-21 (p-value)	Dixon et al.	7
NAAA	ENST00000511606					
CXCL10	ENST00000306602	204533_at	blood	2.40×10-18 (p-value)	Dixon et al.	7
NAAA	?	ENSG00000138744	cerebellum	9.13×10-6 (p-value)	GTEx Portal V6	3
?	?	ILMN_1668605	liver	2.89×10-15 (p-value)	Innocenti et al.	1
CXCL10	ENST00000306602	ILMN_1791759	blood	3.55×10-5 (p-value)	Westra et al.	9
NAAA	?	ENSG00000138744	uterus	4.78×10-7 (p-value)	GTEx Portal V6	12
NAAA	?	ENSG00000138744	anterior cingulate cortex	1.32×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000382695	1	ENCP00000041438	NUP54
		ENCP00000041452	CCDC158
		ENCP00000041415	NAAA
ENCE00000382651	1	ENCP00000041411	USO1
ENCE00000382654	1	ENCP00000041411	USO1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001432208 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me3, PolII, Gabp, Sin3Ak20, H3K9ac, Yy1, TAF1, H3K4me2
		HSMMtube	H3K79me2, H3K9ac, H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	H3K79me2, HEY1, H2AZ, TAF1, H3K4me2, H3K36me3, DNase1, H3K4me3, Cmyc, Gabp, PolII, Max, H3K9ac
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K79me2, DNase1, H3K36me3, H3K9ac, H3K4me2, H3K4me3
		liver (HepG2)	Gabp, PolII, H3K79me2, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, DNase1
		blood (GM12878)	H3K79me2, H3K4me3, H3K4me2, H3K9ac, ELF1, Gabp, DNase1, Cmyc, PolII, H2AZ
		lung (IMR90)	DNase1, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K9ac
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, H3K27ac, DNase1
		skin (NHEK)	DNase1, H3K4me3, H3K4me2, H3K27ac, H3K9ac
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, PolII, Gabp, H3K9ac, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, H3K79me2
		monocytes (Monocytes-CD14+)	H3K36me3, H3K9ac, H3K27ac, H3K4me2, DNase1, H3K4me3, H3K27me3
		endothelium (HUVEC)	Max, DNase1, H3K4me2, H3K4me3, H3K9ac, PolII, Cmyc
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K36me3
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
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
ENSR00001685900 (promoter flanking region)	1	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

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC110615.1	upstream gene variant, downstream gene variant	122	ENST00000625176	?	ENSP00000485507	6
AC112719.2	upstream gene variant	1189	ENST00000629900	?	ENSP00000487114	2
ART3	upstream gene variant	1561	ENST00000513353	?	ENSP00000421345	2
ART3	upstream gene variant	2474	ENST00000504914	?	ENSP00000421431	2
ART3	upstream gene variant	1577	ENST00000341029	NM_001130017.2	ENSP00000343843	2
ART3	upstream gene variant	1577	ENST00000510669	?	?	2
ART3	upstream gene variant	1626	ENST00000513122	?	ENSP00000422287	2
CXCL11	downstream gene variant, upstream gene variant	721	ENST00000306621	NM_005409.4, NM_001302123.1	ENSP00000306884	4
CXCL11	downstream gene variant, upstream gene variant	595	ENST00000503860	?	ENSP00000425819	4
CXCL9	upstream gene variant	2135	ENST00000264888	NM_002416.1	ENSP00000354901	2
NAAA	upstream gene variant	3046	ENST00000505594	?	ENSP00000426977	1
NAAA	upstream gene variant	2333	ENST00000507956	?	ENSP00000427641	1
NAAA	downstream gene variant, upstream gene variant	653	ENST00000503636	?	?	14
NAAA	upstream gene variant	2255	ENST00000286733	NM_014435.3	ENSP00000286733	1
NAAA	upstream gene variant	5	ENST00000602782	?	ENSP00000473575	8
NAAA	downstream gene variant, upstream gene variant	8	ENST00000507940	?	?	16
NAAA	downstream gene variant, upstream gene variant	168	ENST00000507187	?	ENSP00000423142	11
RP11-630D6.1	downstream gene variant, upstream gene variant	591	ENST00000477971	?	?	3
RP11-630D6.5	upstream gene variant, downstream gene variant	1333	ENST00000501239	?	?	3
SDAD1	downstream gene variant	833	ENST00000503411	?	ENSP00000422368	1
SDAD1	downstream gene variant	1148	ENST00000395711	NM_001288983.1	ENSP00000379061	3
SDAD1	downstream gene variant, upstream gene variant	479	ENST00000515836	?	?	4
SDAD1	downstream gene variant	768	ENST00000395710	?	ENSP00000379060	3
SDAD1	downstream gene variant	778	ENST00000356260	NM_001288984.1, NM_018115.3	ENSP00000348596	3
SDAD1	downstream gene variant, upstream gene variant	164	ENST00000502543	?	?	13
SDAD1	downstream gene variant, upstream gene variant	2587	ENST00000513089	?	?	2
SDAD1	downstream gene variant, upstream gene variant	1721	ENST00000507396	?	?	4

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
	?			V	gtA/gtG	1

NAAA	ENST00000505594		ENSP00000426977			
NAAA	ENST00000507956	?	ENSP00000427641	V	gtA/gtG	1
NAAA	ENST00000507187	?	ENSP00000423142	V	gtA/gtG	1
NAAA	ENST00000286733	NM_014435.3	ENSP00000286733	V	gtA/gtG	1
SDAD1	ENST00000395711	NM_001288983.1	ENSP00000379061	D	gaT/gaC	1
SDAD1	ENST00000356260	NM_001288984.1, NM_018115.3	ENSP00000348596	D	gaT/gaC	1

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
NAAA	ENST00000505594	?	ENSP00000426977	1
NAAA	ENST00000507940	?	?	1
NAAA	ENST00000507956	?	ENSP00000427641	1
NAAA	ENST00000507187	?	ENSP00000423142	1
NAAA	ENST00000286733	NM_014435.3	ENSP00000286733	1

Non-coding exon variant (splice region)

gene	affected transcript	RefSeq id	variant(s)
NAAA	ENST00000507940	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC110615.1	ENST00000625176	?	ENSP00000485507	1
ART3	ENST00000510669	?	?	19
ART3	ENST00000504914	?	ENSP00000421431	19
ART3	ENST00000513122	?	ENSP00000422287	19
ART3	ENST00000341029	NM_001130017.2	ENSP00000343843	19
ART3	ENST00000513353	?	ENSP00000421345	19
CXCL11	ENST00000503860	?	ENSP00000425819	3
CXCL11	ENST00000306621	NM_005409.4, NM_001302123.1	ENSP00000306884	2
CXCL9	ENST00000264888	NM_002416.1	ENSP00000354901	1
NAAA	ENST00000505594	?	ENSP00000426977	20
NAAA	ENST00000507940	?	?	6
NAAA	ENST00000507956	?	ENSP00000427641	20
NAAA	ENST00000507187	?	ENSP00000423142	10
NAAA	ENST00000602782	?	ENSP00000473575	14
NAAA	ENST00000286733	NM_014435.3	ENSP00000286733	20
RP11-630D6.5	ENST00000501239	?	?	4
SDAD1	ENST00000395711	NM_001288983.1	ENSP00000379061	13
SDAD1	ENST00000356260	NM_001288984.1, NM_018115.3	ENSP00000348596	13
SDAD1	ENST00000504975	?	?	5
SDAD1	ENST00000514710	?	ENSP00000427443	5
SDAD1	ENST00000503411	?	ENSP00000422368	5
SDAD1	ENST00000395710	?	ENSP00000379060	13
SDAD1	ENST00000513089	?	?	7
SDAD1	ENST00000502543	?	?	5

SDAD1	ENST00000507396	?	?	3
SDAD1	ENST00000515836	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CXCL11	ENST00000306621	NM_005409.4, NM_001302123.1	ENSP00000306884	1
NAAA	ENST00000507187	?	ENSP00000423142	4
SDAD1	ENST00000395710	?	ENSP00000379060	1

Non-coding exon variant

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