Locus 304 (rs722414)

SNiPAcard

Block annotations

Block info

genomic range	chr12:89,826,210-89,940,502
block size	114,293 bp
variant count	94 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.634 [-4.107 – 5.606]	gene(s) hit or close-by	CENPCP1, GALNT4, POC1B, POC1B-GALNT4, RP11-734K2.3, RP11-734K2.4
phastCons	μ = 0.036 [0 – 1]	eQTL gene(s)	ATP2B1, GALNT4, POC1B, POC1B-GALNT4, RP11-981P6.1
GERP++	μ = -0.111 [-9.6 – 4.59]	potentially regulated gene(s)	–
CADD score	μ = 3.623 [0.036 – 20.6]	disease gene(s)	POC1B

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
1-linoleoylglycerophosphoethanolamine*	1.97×10^{-5}	Metabolomics GWAS Server	24816252	13
ascorbate (Vitamin C)	9.10×10^{-5}	Metabolomics GWAS Server	24816252	1

Disease gene annotation

gene	trait	source DB	source entry/link
POC1B	CONE-ROD DYSTROPHY 20	OMIM	MIM:615973

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)
GALNT4	missense variant	ENST00000529983	NM_003774.4	ENSP00000436604	I/T	aTa/aCa	tolerated	benign	1
POC1B-GALNT4	missense variant	ENST00000548729	NM_001199782.1, NM_001199781.1	ENSP00000447852	I/T	aTa/aCa	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
POC1B	?	ENSG00000139323	tibial nerve	1.80×10^{-12} (p-value)	GTEx Portal V6	89
GALNT4	ENST00000529983	ILMN_1739297	skin	7.81×10^{-44} (p-value)	MuTHER consortium	36
POC1B-GALNT4	ENST00000547474		blood	5.59×10^{-68} (p-value)	MuTHER consortium	36
POC1B-GALNT4	ENST00000548729		adipocyte	2.57×10^{-41} (p-value)	MuTHER consortium	36
GALNT4	?	ENSG00000257594	blood	7.61×10^{-4} (q-value)	SeeQTL DB (HapMap)	16
POC1B-GALNT4	ENST00000547474	ILMN_1739297	monocyte	4.40×10^{-58} (p-value)	Fairfax et al.	5

GALNT4 / POC1B-GALNT4	ENST00000529983 / ENST00000548729		b-cell	7.78×10-58 (p-value)	Fairfax et al.	5
GALNT4	?	ENSG00000257594	brain	1.13×10-3 (q-value)	SeeQTL DB (Myers et al.)	10
RP11-981P6.1	?	ENSG00000258302	thyroid	1.24×10-6 (p-value)	GTEx Portal V6	1
ATP2B1	ENST00000428670 / ENST00000359142 / ENST00000261173	ILMN_1824297	blood	7.75×10-6 (p-value)	Westra et al.	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
GALNT4 / POC1B-GALNT4	ENST00000529983 / ENST00000547474 / ENST00000548729	ILMN_1739297	chr12	monocyte	7.49×10-107 (p-value)	Zeller et al.	10

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000434333 (promoter flanking region)	2	embryonic stem cell (H1ESC)	USF1, DNase1
		HSMMtube	H3K27ac, H3K4me2, DNase1, H3K27me3, H3K9ac
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K27ac
		liver (HepG2)	H3K36me3
		lung (IMR90)	DNase1, H3K27ac
		nervous (NH-A)	DNase1
		NHLF	DNase1, H3K27ac
		Osteobl	H3K27ac
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K27ac, H3K4me1, H3K4me2
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	DNase1
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1, H3K4me1, H3K27ac, PolII
		A549	H3K36me3
ENSR00000434334 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		lung (IMR90)	CTCF
		blood (DND-41)	H3K36me3
		breast (HMEC)	CTCF
ENSR00000434337 (promoter flanking region)	1	cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	Cjun
		embryonic stem cell (H1ESC)	DNase1
		blood (GM12878)	BATF, MEF2A, H3K4me2
		nervous (NH-A)	DNase1
		A549	H3K36me3
		blood (DND-41)	H3K36me3, H3K4me1
ENSR00000654046 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	DNase1, CTCF
		blood (K562)	CTCF, Rad21
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF, Rad21
		lung (IMR90)	CTCF
		blood (GM12878)	Rad21, CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF

		Osteobl	CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	CTCF
		A549	CTCF
ENSR00001613709 (enhancer)	1	blood (DND-41)	H3K36me3
ENSR00001613708 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H3K36me3
		blood (DND-41)	H3K36me3
ENSR00001465659 (promoter flanking region)	2	cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		Osteobl	H3K4me2, H3K36me3
		lung (IMR90)	DNase1, H3K4me2
		blood (DND-41)	H3K36me3
		A549	H3K36me3
ENSR00000434344 (promoter flanking region)	1	cervix (HeLa-S3)	DNase1
		blood (DND-41)	H3K36me3
		A549	H3K27ac, DNase1, H3K4me3
ENSR00001465660 (promoter flanking region)	4	embryonic stem cell (H1ESC)	DNase1, H3K36me3
		Osteobl	H3K4me2, H3K36me3
		blood (K562)	H3K36me3
		blood (DND-41)	H3K36me3, H3K4me3, H3K4me2, H3K4me1
		skin (NHDF-AD)	H3K4me3
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K79me2, H3K4me1, H3K4me2, FOXA1
		lung (IMR90)	H3K36me3
		blood (GM12878)	H3K4me3
		A549	H3K36me3, H3K4me2, H3K4me3
		skin (NHEK)	H3K36me3
ENSR00000245284 (TF binding site)	2	embryonic stem cell (H1ESC)	DNase1
		blood (K562)	Gata2, DNase1, Egr1, PolII
ENSR00001613716 (enhancer)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	CTCF
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	DNase1, CTCF
		muscle (HSMM)	DNase1, CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF, H3K27me3
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	CTCF, DNase1
		endothelium (HUVEC)	CTCF
		A549	CTCF
ENSR00001465663 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, CTCF, Rad21
		HSMMtube	DNase1, CTCF
		blood (K562)	DNase1, Rad21, CTCF
		skin (NHDF-AD)	DNase1, CTCF
		muscle (HSMM)	DNase1, CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	DNase1, CTCF
		nervous (NH-A)	CTCF, H3K27me3
		skin (NHEK)	CTCF
		NHLF	CTCF

		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	CTCF
		A549	CTCF
ENSR00001613717 (promoter flanking region)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	DNase1, Rad21, CTCF
		skin (NHDF-AD)	DNase1, CTCF
		muscle (HSMM)	CTCF, DNase1, H3K27ac
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	DNase1, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	CTCF
		A549	CTCF
ENSR00000434348 (promoter flanking region)	1	HSMMtube	DNase1
		A549	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K27ac, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CENPCP1	upstream gene variant, downstream gene variant	158	ENST00000533085	?	?	15
GALNT4	downstream gene variant	230	ENST00000529983	NM_003774.4	ENSP00000436604	14
POC1B	upstream gene variant	2973	ENST00000549591	?	?	1
POC1B	downstream gene variant, upstream gene variant	40	ENST00000549304	?	?	11
POC1B	downstream gene variant	793	ENST00000539190	?	?	9
POC1B	downstream gene variant	2049	ENST00000552563	?	?	3
POC1B	downstream gene variant, upstream gene variant	250	ENST00000551819	?	?	4
POC1B	downstream gene variant	2257	ENST00000546830	?	ENSP00000449256	3
POC1B	upstream gene variant	1519	ENST00000546740	?	?	3
POC1B	downstream gene variant	900	ENST00000547274	?	ENSP00000449648	1
POC1B-GALNT4	downstream gene variant	230	ENST00000548729	NM_001199782.1, NM_001199781.1	ENSP00000447852	14
POC1B-GALNT4	downstream gene variant	162	ENST00000547474	?	ENSP00000447754	9
RP11-734K2.3	downstream gene variant, upstream gene variant	543	ENST00000623264	?	?	7
RP11-734K2.4	upstream gene variant	1913	ENST00000605233	?	?	5

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
POC1B	ENST00000548715	?	ENSP00000449945	1
POC1B	ENST00000313546	NM_172240.2	ENSP00000323302	1
POC1B	ENST00000393179	?	ENSP00000376877	1
POC1B	ENST00000549035	NM_001199777.1	ENSP00000447916	1
POC1B	ENST00000547496	?	ENSP00000447437	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
POC1B	ENST00000551819	?	?	3
POC1B	ENST00000546740	?	?	23
POC1B	ENST00000548715	?	ENSP00000449945	82
POC1B	ENST00000313546	NM_172240.2	ENSP00000323302	82
POC1B	ENST00000547274	?	ENSP00000449648	49
POC1B	ENST00000393179	?	ENSP00000376877	82
POC1B	ENST00000549504	?	ENSP00000450118	82
POC1B	ENST00000549035	NM_001199777.1	ENSP00000447916	82
POC1B	ENST00000552563	?	?	56
POC1B	ENST00000549304	?	?	8
POC1B	ENST00000546830	?	ENSP00000449256	44
POC1B	ENST00000547496	?	ENSP00000447437	82
POC1B	ENST00000539190	?	?	59
POC1B-GALNT4	ENST00000547474	?	ENSP00000447754	1
RP11-734K2.4	ENST00000605233	?	?	5

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
GALNT4	ENST00000529983	NM_003774.4	ENSP00000436604	6
POC1B-GALNT4	ENST00000548729	NM_001199782.1, NM_001199781.1	ENSP00000447852	6

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CENPCP1	ENST00000533085	?	4
RP11-734K2.4	ENST00000605233	?	7