Locus 188 (rs10858881)

SNiPAcard

Block annotations

Block info

genomic range	chr12:89,825,925-89,938,590
block size	112,666 bp
variant count	76 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.255 [-2.445 – 1.215]	gene(s) hit or close-by	CENPCP1, GALNT4, POC1B, POC1B-GALNT4, RP11-734K2.3, RP11-734K2.4
phastCons	μ = 0.052 [0 – 0.745]	eQTL gene(s)	ATP2B1, GALNT4, POC1B, POC1B-GALNT4, RP11-981P6.1
GERP++	μ = -0.521 [-5.92 – 3.23]	potentially regulated gene(s)	–
CADD score	μ = 4.052 [0.12 – 13.8]	disease gene(s)	POC1B

Trait annotations

Disease gene annotation

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

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RP11-981P6.1	?	ENSG00000258302	muscularis mucosae	2.36×10-7 (p-value)	GTEx Portal V6	73
RP11-981P6.1	?	ENSG00000258302	spleen	5.53×10-9 (p-value)	GTEx Portal V6	75
POC1B	?	ENSG00000139323	tibial nerve	1.80×10-12 (p-value)	GTEx Portal V6	76
POC1B	?	ENSG00000139323	transformed fibroblasts	1.33×10-6 (p-value)	GTEx Portal V6	73
POC1B	?	ENSG00000139323	tibial artery	1.41×10-6 (p-value)	GTEx Portal V6	73
RP11-981P6.1	?	ENSG00000258302	tibial artery	7.99×10-8 (p-value)	GTEx Portal V6	75
RP11-981P6.1	?	ENSG00000258302	thyroid	2.97×10-25 (p-value)	GTEx Portal V6	76
RP11-981P6.1	?	ENSG00000258302	pituitary	5.61×10-7 (p-value)	GTEx Portal V6	66
RP11-981P6.1	?	ENSG00000258302	adrenal gland	1.17×10-9 (p-value)	GTEx Portal V6	75
RP11-981P6.1	?	ENSG00000258302	sun exposed skin	8.82×10-7 (p-value)	GTEx Portal V6	74
POC1B	?	ENSG00000139323	aorta	3.08×10-6 (p-value)	GTEx Portal V6	31
POC1B	?	ENSG00000139323	subcutaneous adipocytes	2.15×10-10 (p-value)	GTEx Portal V6	76
GALNT4	ENST00000529983	ILMN_1739297	skin	4.74×10-8 (p-value)	MuTHER consortium	33
POC1B-GALNT4	ENST00000547474		blood	5.22×10-6 (p-value)	MuTHER consortium	33
POC1B-GALNT4	ENST00000548729					
ATP2B1	ENST00000428670	ILMN_1824297	monocyte	6.85×10-15 (p-value)	Zeller et al.	4

ATP2B1	ENST00000359142					
ATP2B1	ENST00000261173					
RP11-981P6.1	?	ENSG00000258302	lung	7.14×10-6 (p-value)	GTEx Portal V6	61
RP11-981P6.1	?	ENSG00000258302	transverse colon	3.61×10-6 (p-value)	GTEx Portal V6	41
RP11-981P6.1	?	ENSG00000258302	cerebellum	3.05×10-6 (p-value)	GTEx Portal V6	60
RP11-981P6.1	?	ENSG00000258302	stomach	3.58×10-7 (p-value)	GTEx Portal V6	67
POC1B	?	ENSG00000139323	breast	5.43×10-6 (p-value)	GTEx Portal V6	1
POC1B-GALNT4	ENST00000547474	ILMN_1739297	monocyte	3.17×10-8 (p-value)	Fairfax et al.	6
GALNT4	ENST00000529983		b-cell	1.62×10-5 (p-value)	Fairfax et al.	6
POC1B-GALNT4	ENST00000548729					
POC1B	?	ENSG00000139323	blood	9.10×10-6 (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
GALNT4	ENST00000529983	ILMN_1739297	chr12	monocyte	4.54×10-12 (p-value)	Zeller et al.	1
POC1B-GALNT4	ENST00000547474						
POC1B-GALNT4	ENST00000548729						

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000434328 (enhancer)	3	embryonic stem cell (H1ESC)	H3K36me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K36me3
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	DNase1
		endothelium (HUVEC)	Cjun
		liver (HepG2)	DNase1
		lung (IMR90)	DNase1
		A549	H3K36me3
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
ENSR00001613708 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H3K36me3
		blood (DND-41)	H3K36me3
ENSR00001613711 (enhancer)	1	liver (HepG2)	H3K4me1
ENSR00001613712 (CTCF binding site)	2	monocytes (Monocytes-CD14+)	H3K36me3
		blood (DND-41)	H3K36me3

ENSR00001613713 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K4me1
ENSR00000434347 (promoter)	3	embryonic stem cell (H1ESC)	DNase1, H4K5ac, H3K27ac, H3K36me3, H3K27me3, PolII, Rad21, Sin3Ak20, H3K9ac, CTCF, Yy1, TAF1, H3K4me2, SP1, Srf, H3K4me3
		HSMMtube	DNase1, H2AZ, H3K4me3, H3K4me2, CTCF
		blood (K562)	H3K27ac, Max, H3K9ac, H2AZ, TAF1, PolII, H3K4me2, CTCF, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me2, H3K9ac, DNase1, H3K4me3, CTCF
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K4me3, CTCF, H2AZ
		liver (HepG2)	H3K79me2, Rad21, TAF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, H3K4me3, DNase1, PolII, Gabp, CTCF, Srf
		lung (IMR90)	H2BK120ac, H3K9ac, CTCF, H3K4ac, H3K4me3, H3K36me3, DNase1, H4K8ac, H3K27ac, H3K18ac, H3K4me2, H4K5ac
		blood (GM12878)	DNase1, Gabp, ELF1, Rad21, H3K79me2, H3K4me3, H3K27ac, H3K4me2, CTCF, H3K9ac, H2AZ, Srf, PolII, Tr4
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, DNase1
		skin (NHEK)	H3K4me2, H3K4me3, CTCF, H3K36me3, DNase1, H3K9ac
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	CTCF, H3K36me3, H3K4me2, H3K4me3, H2AZ, H3K27ac
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, CTCF, H3K4me2, H3K4me1
		breast (HMEC)	H3K4me3, H3K4me2, CTCF
		cervix (HeLa-S3)	H3K4me2, H3K9ac, H3K27ac, PolII, H3K4me3, DNase1, H3K36me3, CTCF, Gabp
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Max, H3K4me2, H3K4me3, H3K9ac, PolII, CTCF, DNase1
		A549	H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1, H3K36me3, H3K27ac
ENSR00001465661 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	CTCF
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		lung (IMR90)	CTCF
		blood (GM12878)	Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF
ENSR00000245284 (TF binding site)	1	embryonic stem cell (H1ESC)	DNase1
		blood (K562)	Gata2, DNase1, Egr1, PolII
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	450	ENST00000533085	?	?	6
GALNT4	downstream gene variant, upstream gene variant	94	ENST00000529983	NM_003774.4	ENSP00000436604	9
POC1B	downstream gene variant	2282	ENST00000549304	?	?	1
POC1B	downstream gene variant, upstream gene variant	387	ENST00000547274	?	ENSP00000449648	11
POC1B	downstream gene variant	131	ENST00000551819	?	?	2
POC1B	downstream gene variant, upstream gene variant	414	ENST00000539190	?	?	9
POC1B	upstream gene variant	1992	ENST00000546740	?	?	2
POC1B	downstream gene variant, upstream gene variant	407	ENST00000546830	?	ENSP00000449256	11
POC1B	downstream gene variant, upstream gene variant	226	ENST00000552563	?	?	10

	variant					
POC1B	upstream gene variant	383	ENST00000313546	NM_172240.2	ENSP00000323302	8
POC1B	upstream gene variant	2688	ENST00000549591	?	?	1
POC1B	upstream gene variant	407	ENST00000547496	?	ENSP00000447437	8
POC1B	upstream gene variant	586	ENST00000549504	?	ENSP00000450118	8
POC1B	upstream gene variant	228	ENST00000549035	NM_001199777.1	ENSP00000447916	8
POC1B	upstream gene variant	176	ENST00000393179	?	ENSP00000376877	8
POC1B	upstream gene variant	410	ENST00000548715	?	ENSP00000449945	8
POC1B-GALNT4	downstream gene variant, upstream gene variant	145	ENST00000547474	?	ENSP00000447754	10
POC1B-GALNT4	downstream gene variant, upstream gene variant	209	ENST00000548729	NM_001199782.1, NM_001199781.1	ENSP00000447852	9
RP11-734K2.3	downstream gene variant, upstream gene variant	325	ENST00000623264	?	?	8
RP11-734K2.4	upstream gene variant	754	ENST00000605233	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
POC1B	ENST00000551819	?	?	2
POC1B	ENST00000548715	?	ENSP00000449945	52
POC1B	ENST00000539190	?	?	29
POC1B	ENST00000549035	NM_001199777.1	ENSP00000447916	51
POC1B	ENST00000547274	?	ENSP00000449648	20
POC1B	ENST00000546740	?	?	23
POC1B	ENST00000313546	NM_172240.2	ENSP00000323302	52
POC1B	ENST00000549504	?	ENSP00000450118	51
POC1B	ENST00000393179	?	ENSP00000376877	51
POC1B	ENST00000552563	?	?	26
POC1B	ENST00000547496	?	ENSP00000447437	52
POC1B	ENST00000546830	?	ENSP00000449256	17
POC1B	ENST00000549304	?	?	4
POC1B-GALNT4	ENST00000548729	NM_001199782.1, NM_001199781.1	ENSP00000447852	4
POC1B-GALNT4	ENST00000547474	?	ENSP00000447754	2
RP11-734K2.4	ENST00000605233	?	?	20

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
GALNT4	ENST00000529983	NM_003774.4	ENSP00000436604	2
POC1B-GALNT4	ENST00000548729	NM_001199782.1, NM_001199781.1	ENSP00000447852	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-734K2.4	ENST00000605233	?	6

Locus 188 (sentinel SNP rs10858881 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.