Locus 416 (rs870825)

SNiPAcard

Block annotations

Block info

genomic range	chr4:185,571,123-185,608,591
block size	37,469 bp
variant count	89 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.622 [-3.857 – 1.623]	gene(s) hit or close-by	CASP3, PRIMPOL
phastCons	μ = 0.027 [0 – 0.937]	eQTL gene(s)	CASP3, CENPU, PRIMPOL
GERP++	μ = -0.734 [-7.23 – 3.23]	potentially regulated gene(s)	ACSL1, IRF2, PRIMPOL, RP11-162O12.2, RP11-242J7.1, RP11-386B13.3, RP11-701P16.5
CADD score	μ = 3.236 [0.009 – 10.91]	disease gene(s)	–

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)
PRIMPOL	missense variant	ENST00000508001	?	ENSP00000424639	N/S	aAt/aGt	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PRIMPOL	?	ENSG00000164306	tibial nerve	2.48×10-8 (p-value)	GTEx Portal V6	72
CENPU	?	ENSG00000151725	tibial nerve	1.81×10-7 (p-value)	GTEx Portal V6	6
PRIMPOL	?	ENSG00000164306	transformed fibroblasts	2.85×10-6 (p-value)	GTEx Portal V6	25
CENPU	?	ENSG00000151725	transformed fibroblasts	2.12×10-7 (p-value)	GTEx Portal V6	3
CENPU	?	ENSG00000151725	tibial artery	2.13×10-9 (p-value)	GTEx Portal V6	81
PRIMPOL	?	ENSG00000164306	breast	2.48×10-8 (p-value)	GTEx Portal V6	71
CASP3	?	ENSG00000164305	blood	1.80×10-8 (p-value)	GTEx Portal V6	51
PRIMPOL	?	ENSG00000164306	thyroid	3.10×10-7 (p-value)	GTEx Portal V6	28
CENPU	?	ENSG00000151725	thyroid	2.30×10-8 (p-value)	GTEx Portal V6	73
CENPU	?	ENSG00000151725	esophagus mucosa	4.40×10-15 (p-value)	GTEx Portal V6	86
PRIMPOL	?	ENSG00000164306	skeletal muscle	1.04×10-8 (p-value)	GTEx Portal V6	75
CENPU	?	ENSG00000151725	skeletal muscle	3.37×10-6 (p-value)	GTEx Portal V6	16
PRIMPOL	?	ENSG00000164306	unexposed skin	6.96×10-9 (p-value)	GTEx Portal V6	74
CENPU	?	ENSG00000151725	unexposed skin	2.46×10-9 (p-value)	GTEx Portal V6	7
PRIMPOL	?	ENSG00000164306	sun exposed skin	2.24×10-6 (p-value)	GTEx Portal V6	25

CENPU	?	ENSG00000151725	sun exposed skin	9.04×10-13 (p-value)	GTEx Portal V6	86
PRIMPOL	?	ENSG00000164306	subcutaneous adipocytes	6.42×10-7 (p-value)	GTEx Portal V6	44
CENPU	?	ENSG00000151725	subcutaneous adipocytes	2.51×10-8 (p-value)	GTEx Portal V6	77
CENPU	ENST00000502461	ILMN_1679438	skin	3.40×10-9 (p-value)	MuTHER consortium	11
CENPU	ENST00000281453					
CASP3	ENST00000393585	ILMN_1731639	blood	1.19×10-7 (p-value)	MuTHER consortium	11
CASP3	ENST00000517513					
CASP3	ENST00000308394					
CASP3	ENST00000613118					
CENPU	?	ENSG00000151725	lung	3.92×10-7 (p-value)	GTEx Portal V6	7
CENPU	ENST00000502461	ILMN_1679438	blood	1.92×10-5 (p-value)	Westra et al.	11
CENPU	ENST00000281453					
CENPU	?	ENSG00000151725	testis	3.50×10-11 (p-value)	GTEx Portal V6	74
CENPU	?	ENSG00000151725	blood	4.39×10-3 (q-value)	SeeQTL DB (HapMap)	3
CASP3	ENST00000393585	ILMN_1731639	monocyte	2.71×10-17 (p-value)	Zeller et al.	6
CASP3	ENST00000517513					
CASP3	ENST00000308394					
CASP3	ENST00000613118					
CENPU	?	ENSG00000151725	EBV lymphocytes	3.08×10-7 (p-value)	GTEx Portal V6	4
CENPU	?	ENSG00000151725	visceral adipocytes	1.55×10-6 (p-value)	GTEx Portal V6	1
CENPU	?	ENSG00000151725	atrial appendage	1.10×10-6 (p-value)	GTEx Portal V6	2
CENPU	?	ENSG00000151725	transverse colon	1.33×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000640933	1	ENST00000515774, ENST00000314970, ENST00000509002, ENST00000503752, ENST00000512834	PRIMPOL

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000398855	1	ENCP00000043126	RP11-242J7.1
ENCE00000398670	2	ENCP00000043153	RP11-386B13.3
		ENCP00000043119	RP11-162O12.2
		ENCP00000043145	ACSL1
		ENCP00000043138	ACSL1
		ENCP00000043147	RP11-701P16.5
		ENCP00000043124	IRF2
		ENCP00000043144	ACSL1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001690770	1	embryonic stem cell (H1ESC)	DNase1, CTCF, PolII, Rad21, TAF7, H3K27ac, TAF1, H3K4me2, H3K9ac, H3K4me3
(promoter)		HSMMtube	H3K9ac, H3K4me2, H3K27ac, H3K4me3, DNase1
		blood (K562)	Egr1, H3K27ac, Max, Rad21, H3K9ac, HEY1, H2AZ, TAF1, PolII, H3K4me2, CTCF, DNase1,

			H3K4me3
		skin (NHDF-AD)	H3K4me2, H3K27ac, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, DNase1
		liver (HepG2)	Gabp, PolII, TAF1, ELF1, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K27me3, DNase1
		lung (IMR90)	CTCF, H3K9ac, H3K4me3, H3K4me2, H3K27ac, DNase1
		blood (GM12878)	PolII, H2AZ, DNase1, CTCF, Egr1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H3K4me3, H3K4me2, H3K27ac, H2AZ
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, PolII, CTCF
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, H3K27ac, H3K9ac, H3K4me3, H3K4me2, PolII, CTCF, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac
ENSR00001438105 (promoter flanking region)	4	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		lung (IMR90)	DNase1
		A549	H3K36me3
ENSR00001438106 (enhancer)	1	endothelium (HUVEC)	H3K36me3
		Osteobl	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		blood (DND-41)	H3K36me3
		skin (NHEK)	H3K36me3
		muscle (HSMM)	DNase1
ENSR00001690773 (enhancer)	5	NHLF	H3K36me3
		HSMMtube	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001255743 (promoter flanking region)	2	monocytes (Monocytes-CD14+)	H3K27ac, DNase1
		blood (K562)	Ini1, Brg1, Gata2, DNase1, Egr1, H3K27ac, H3K4me3, PolII, H3K4me2
		blood (DND-41)	H3K36me3
		nervous (NH-A)	DNase1

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
PRIMPOL	2	ENST00000314970 ENST00000503752 ENST00000506278 ENST00000508001 ENST00000509002 ENST00000512834 ENST00000515152 ENST00000515774	hsa-miR-134-5p hsa-miR-302a-3p hsa-miR-302b-3p hsa-miR-302c-3p hsa-miR-302d-3p hsa-miR-329-3p hsa-miR-362-3p hsa-miR-372-3p hsa-miR-373-3p hsa-miR-494-3p hsa-miR-520a-3p hsa-miR-520b hsa-miR-520c-3p hsa-miR-520d-3p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CASP3	upstream gene variant	522	ENST00000447121	?	ENSP00000407142	6
CASP3	upstream gene variant	514	ENST00000393588	?	ENSP00000377213	6
CASP3	upstream gene variant	522	ENST00000517513	?	ENSP00000428372	6
CASP3	upstream gene variant	460	ENST00000523916	NM_032991.2	ENSP00000428929	6
CASP3	upstream gene variant	494	ENST00000308394	NM_004346.3	ENSP00000311032	6
CASP3	upstream gene variant	1377	ENST00000393585	?	ENSP00000377210	3
CASP3	upstream gene variant	1377	ENST00000613118	?	ENSP00000478339	3
PRIMPOL	downstream gene variant	278	ENST00000506278	?	ENSP00000423409	19
PRIMPOL	downstream gene variant	277	ENST00000515152	?	ENSP00000422199	9
PRIMPOL	downstream gene variant	980	ENST00000509538	?	?	2
PRIMPOL	upstream gene variant	506	ENST00000508001	?	ENSP00000424639	10
PRIMPOL	upstream gene variant	3931	ENST00000512658	?	?	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
PRIMPOL	ENST00000512834	NM_001300768.1	ENSP00000425316	Q	caA/caG	1
PRIMPOL	ENST00000314970	NM_152683.2	ENSP00000313816	Q	caA/caG	1
PRIMPOL	ENST00000503752	?	ENSP00000420860	Q	caA/caG	1
PRIMPOL	ENST00000515774	NM_001300767.1	ENSP00000421913	Q	caA/caG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PRIMPOL	ENST00000509002	?	ENSP00000423353	89
PRIMPOL	ENST00000512834	NM_001300768.1	ENSP00000425316	89
PRIMPOL	ENST00000314970	NM_152683.2	ENSP00000313816	89
PRIMPOL	ENST00000503752	?	ENSP00000420860	89
PRIMPOL	ENST00000515774	NM_001300767.1	ENSP00000421913	89
PRIMPOL	ENST00000506278	?	ENSP00000423409	62
PRIMPOL	ENST00000515152	?	ENSP00000422199	48
PRIMPOL	ENST00000509538	?	?	10
PRIMPOL	ENST00000508001	?	ENSP00000424639	27

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
PRIMPOL	ENST00000506278	?	ENSP00000423409	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PRIMPOL	ENST00000509538	?	1

Locus 416 (sentinel SNP rs870825 + 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.