

# SNiPACard

## Block annotations

### Block info

genomic range	chr4:185,571,123-185,608,591 <i>e!</i>
block size	37,469 bp
variant count	89 variants

### Basic features

Conservation/deleteriousness		Linked genes	
phyloP	$\mu = -0.622$ [-3.857 – 1.623]	gene(s) hit or close-by	CASP3 <i>e!</i> , PRIMPOL <i>e!</i>
phastCons	$\mu = 0.027$ [0 – 0.937]	eQTL gene(s)	CASP3 <i>e!</i> , CENPU <i>e!</i> , PRIMPOL <i>e!</i>
GERP++	$\mu = -0.734$ [-7.23 – 3.23]	potentially regulated gene(s)	ACSL1 <i>e!</i> , IRF2 <i>e!</i> , PRIMPOL <i>e!</i> , RP11-162O12.2 <i>e!</i> , RP11-242J7.1 <i>e!</i> , RP11-386B13.3 <i>e!</i> , RP11-701P16.5 <i>e!</i>
CADD score	$\mu = 3.236$ [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 <i>e!</i>	missense variant	ENST00000508001 <i>e!</i>	?	ENSP00000424639 <i>e!</i>	N/S	aAt/aGt	?	?	1

## Direct effect on regulation

### cis-eQTL

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

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

### Putative effect on regulation

#### FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000640933 <i>e!</i>	1	ENST00000515774 <i>e!</i> , ENST00000314970 <i>e!</i> , ENST00000509002 <i>e!</i> , ENST00000503752 <i>e!</i> , ENST00000512834 <i>e!</i>	PRIMPOL <i>e!</i>

#### ENCODE promoter-associated distal DHS (Enhancer)

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

#### Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001690770 <i>e!</i> (promoter)	1	embryonic stem cell (H1ESC)	DNase1, CTCF, PolII, Rad21, TAF7, H3K27ac, TAF1, H3K4me2, H3K9ac, H3K4me3
		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 <i>e!</i>	4 (promoter flanking region)	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 <i>e!</i>	1 (enhancer)	endothelium (HUVEC) H3K36me3
	Osteobl	H3K36me3
	lung (IMR90)	H3K36me3
	A549	H3K36me3
	blood (DND-41)	H3K36me3
	skin (NHEK)	H3K36me3
	muscle (HSMM)	DNase1
ENSR00001690773 <i>e!</i>	5 (enhancer)	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 <i>e!</i>	2 (promoter flanking region)	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 <i>e!</i>	2	ENST00000314970 <i>e!</i>	hsa-miR-134-5p
		ENST00000503752 <i>e!</i>	hsa-miR-302a-3p
		ENST00000506278 <i>e!</i>	hsa-miR-302b-3p
		ENST00000508001 <i>e!</i>	hsa-miR-302c-3p
		ENST00000509002 <i>e!</i>	hsa-miR-302d-3p
		ENST00000512834 <i>e!</i>	hsa-miR-329-3p
		ENST00000515152 <i>e!</i>	hsa-miR-362-3p
		ENST00000515774 <i>e!</i>	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 <i>e!</i>	upstream gene variant	522	ENST00000447121 <i>e!</i>	?	ENSP00000407142 <i>e!</i>	6
CASP3 <i>e!</i>	upstream gene variant	514	ENST00000393588 <i>e!</i>	?	ENSP00000377213 <i>e!</i>	6
CASP3 <i>e!</i>	upstream gene variant	522	ENST00000517513 <i>e!</i>	?	ENSP00000428372 <i>e!</i>	6
CASP3 <i>e!</i>	upstream gene variant	460	ENST00000523916 <i>e!</i>	NM_032991.2	ENSP00000428929 <i>e!</i>	6
CASP3 <i>e!</i>	upstream gene variant	494	ENST00000308394 <i>e!</i>	NM_004346.3	ENSP00000311032 <i>e!</i>	6
CASP3 <i>e!</i>	upstream gene variant	1377	ENST00000393585 <i>e!</i>	?	ENSP00000377210 <i>e!</i>	3
CASP3 <i>e!</i>	upstream gene variant	1377	ENST00000613118 <i>e!</i>	?	ENSP00000478339 <i>e!</i>	3
PRIMPOL <i>e!</i>	downstream gene variant	278	ENST00000506278 <i>e!</i>	?	ENSP00000423409 <i>e!</i>	19
PRIMPOL <i>e!</i>	downstream gene variant	277	ENST00000515152 <i>e!</i>	?	ENSP00000422199 <i>e!</i>	9
PRIMPOL <i>e!</i>	downstream gene variant	980	ENST00000509538 <i>e!</i>	?	?	2
PRIMPOL <i>e!</i>	upstream gene variant	506	ENST00000508001 <i>e!</i>	?	ENSP00000424639 <i>e!</i>	10
PRIMPOL <i>e!</i>	upstream gene variant	3931	ENST00000512658 <i>e!</i>	?	?	1

## Putative effect on transcript

## Synonymous coding variant

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

## Intron variant

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

## 3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
PRIMPOL <i>e!</i>	ENST00000506278 <i>e!</i>	?	ENSP00000423409 <i>e!</i>	1

## Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PRIMPOL <i>e!</i>	ENST00000509538 <i>e!</i>	?	1

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