

# SNiPAcord

## Block annotations

### Block info



genomic range	chr6:31,380,034-31,391,401 <i>e!</i>
block size	11,368 bp
variant count	2 variants

### Basic features

Conservation/deleteriousness		Linked genes	
phyloP	$\mu = 0.000$ [-]	gene(s) hit or close-by	HCP5 <i>e!</i> , MICA <i>e!</i>
phastCons	$\mu = 0.000$ [-]	eQTL gene(s)	MICA <i>e!</i> , MICB <i>e!</i> , PSORS1C2 <i>e!</i> , TNXA <i>e!</i> , TNXB <i>e!</i> , XXbac-BPG181B23.7 <i>e!</i>
GERP++	$\mu = -1.225$ [-2.45 - 0]	potentially regulated gene(s)	-
CADD score	$\mu = 4.632$ [4.233 - 5.031]	disease gene(s)	TNXB <i>e!</i>

## Trait annotations

### Variant association














trait	min(p-value)	source DB	source entry/link	variant(s)
HIV-1 susceptibility	<4.00×10 <sup>-11</sup>	GWAS Catalog	23935489 	1
HIV-1 control	<1.00×10 <sup>-34</sup>	GWAS Catalog	21051598 	1





































### Disease gene annotation

gene	trait	source DB	source entry/link
TNXB <i>e!</i>	Ehlers-Danlos syndrome due to tenascin-X deficiency	OrphaNet	OrphaNet:230839 
TNXB <i>e!</i>	EHLERS-DANLOS SYNDROME, HYPERMOBILITY TYPE	OrphaNet	OrphaNet:285 

## Direct effect on regulation

### cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MICA <i>e!</i>	?	ENSG00000204520 <i>e!</i>	pancreas	2.01×10 <sup>-12</sup> (p-value)	GTEx Portal V6 	2
XXbac-BPG181B23.7 <i>e!</i>	?	ENSG00000272221 <i>e!</i>	pancreas	3.75×10 <sup>-7</sup> (p-value)	GTEx Portal V6 	1
MICA <i>e!</i>	?	ENSG00000204520 <i>e!</i>	muscularis mucosae	3.13×10 <sup>-10</sup> (p-value)	GTEx Portal V6 	2
XXbac-BPG181B23.7 <i>e!</i>	?	ENSG00000272221 <i>e!</i>	muscularis mucosae	6.11×10 <sup>-13</sup> (p-value)	GTEx Portal V6 	2
MICB <i>e!</i>	?	ENSG00000204516 <i>e!</i>	muscularis mucosae	4.75×10 <sup>-7</sup> (p-value)	GTEx Portal V6 	2
MICA <i>e!</i>	?	ENSG00000204520 <i>e!</i>	lung	1.75×10 <sup>-14</sup> (p-value)	GTEx Portal V6 	2
XXbac-BPG181B23.7 <i>e!</i>	?	ENSG00000272221 <i>e!</i>	lung	6.14×10 <sup>-14</sup> (p-value)	GTEx Portal V6 	2
MICB <i>e!</i>	?	ENSG00000204516 <i>e!</i>	lung	3.14×10 <sup>-8</sup> (p-value)	GTEx Portal V6 	2
XXbac-BPG181B23.7 <i>e!</i>	?	ENSG00000272221 <i>e!</i>	atrial appendage	6.38×10 <sup>-10</sup> (p-value)	GTEx Portal V6 	2
MICA <i>e!</i>	?	ENSG00000204520 <i>e!</i>	transformed fibroblasts	6.61×10 <sup>-14</sup> (p-value)	GTEx Portal V6 	2
XXbac-BPG181B23.7 <i>e!</i>	?	ENSG00000272221 <i>e!</i>	transformed fibroblasts	2.25×10 <sup>-16</sup> (p-value)	GTEx Portal V6 	2
MICB <i>e!</i>	?	ENSG00000204516 <i>e!</i>	transformed fibroblasts	4.12×10 <sup>-7</sup> (p-value)	GTEx Portal V6 	1
MICA <i>e!</i>	?	ENSG00000204520 <i>e!</i>	blood	1.23×10 <sup>-11</sup> (p-value)	GTEx Portal V6 	2

XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i> blood	7.53×10 <sup>-13</sup> (p-value)	GTEx Portal V6 	2
MICB <i>e!</i> ?	ENSG00000204516 <i>e!</i> blood	1.78×10 <sup>-6</sup> (p-value)	GTEx Portal V6 	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i> breast	1.18×10 <sup>-8</sup> (p-value)	GTEx Portal V6 	2
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i> breast	3.41×10 <sup>-11</sup> (p-value)	GTEx Portal V6 	2
MICB <i>e!</i> ?	ENSG00000204516 <i>e!</i> breast	8.43×10 <sup>-7</sup> (p-value)	GTEx Portal V6 	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i> tibial artery	4.75×10 <sup>-14</sup> (p-value)	GTEx Portal V6 	2
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i> tibial artery	1.80×10 <sup>-17</sup> (p-value)	GTEx Portal V6 	2
MICB <i>e!</i> ?	ENSG00000204516 <i>e!</i> tibial artery	7.33×10 <sup>-8</sup> (p-value)	GTEx Portal V6 	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i> thyroid	7.64×10 <sup>-14</sup> (p-value)	GTEx Portal V6 	2
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i> thyroid	1.88×10 <sup>-15</sup> (p-value)	GTEx Portal V6 	2
MICB <i>e!</i> ?	ENSG00000204516 <i>e!</i> thyroid	2.47×10 <sup>-9</sup> (p-value)	GTEx Portal V6 	2
PSORS1C2 <i>e!</i> ?	ENSG00000204538 <i>e!</i> thyroid	2.21×10 <sup>-6</sup> (p-value)	GTEx Portal V6 	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i> skeletal muscle	7.03×10 <sup>-10</sup> (p-value)	GTEx Portal V6 	2
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i> skeletal muscle	9.61×10 <sup>-11</sup> (p-value)	GTEx Portal V6 	2
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i> prostate	2.29×10 <sup>-8</sup> (p-value)	GTEx Portal V6 	2
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i> prostate	8.17×10 <sup>-12</sup> (p-value)	GTEx Portal V6 	2
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i> transverse colon	3.92×10 <sup>-9</sup> (p-value)	GTEx Portal V6 	2
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i> transverse colon	1.18×10 <sup>-8</sup> (p-value)	GTEx Portal V6 	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i> sun exposed skin	9.48×10 <sup>-16</sup> (p-value)	GTEx Portal V6 	2
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i> sun exposed skin	6.43×10 <sup>-16</sup> (p-value)	GTEx Portal V6 	2
MICB <i>e!</i> ?	ENSG00000204516 <i>e!</i> sun exposed skin	6.95×10 <sup>-9</sup> (p-value)	GTEx Portal V6 	2
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i> unexposed skin	5.77×10 <sup>-9</sup> (p-value)	GTEx Portal V6 	2
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i> unexposed skin	1.22×10 <sup>-10</sup> (p-value)	GTEx Portal V6 	2
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i> aorta	4.89×10 <sup>-8</sup> (p-value)	GTEx Portal V6 	2
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i> aorta	2.71×10 <sup>-9</sup> (p-value)	GTEx Portal V6 	2
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i> left ventricle	1.81×10 <sup>-11</sup> (p-value)	GTEx Portal V6 	2
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i> left ventricle	6.68×10 <sup>-15</sup> (p-value)	GTEx Portal V6 	2
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i> subcutaneous adipocytes	5.34×10 <sup>-14</sup> (p-value)	GTEx Portal V6 	2
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i> subcutaneous adipocytes	1.04×10 <sup>-12</sup> (p-value)	GTEx Portal V6 	2
MICB <i>e!</i> ?	ENSG00000204516 <i>e!</i> subcutaneous adipocytes	1.29×10 <sup>-9</sup> (p-value)	GTEx Portal V6 	2
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i> terminal ileum	5.11×10 <sup>-8</sup> (p-value)	GTEx Portal V6 	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i> terminal ileum	3.42×10 <sup>-7</sup> (p-value)	GTEx Portal V6 	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i> caudate basal ganglia	1.06×10 <sup>-6</sup> (p-value)	GTEx Portal V6 	2
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i> stomach	2.56×10 <sup>-14</sup> (p-value)	GTEx Portal V6 	2
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i> stomach	2.18×10 <sup>-11</sup> (p-value)	GTEx Portal V6 	2
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i> visceral adipocytes	4.85×10 <sup>-10</sup> (p-value)	GTEx Portal V6 	2

XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	visceral adipocytes	3.15×10 <sup>-11</sup> (p-value)	GTEx Portal V6 <i>!M</i>	2
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i>	spleen	1.65×10 <sup>-6</sup> (p-value)	GTEx Portal V6 <i>!M</i>	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	sigmoid colon	3.37×10 <sup>-7</sup> (p-value)	GTEx Portal V6 <i>!M</i>	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i>	coronary artery	6.58×10 <sup>-7</sup> (p-value)	GTEx Portal V6 <i>!M</i>	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	coronary artery	2.18×10 <sup>-8</sup> (p-value)	GTEx Portal V6 <i>!M</i>	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i>	tibial nerve	8.74×10 <sup>-17</sup> (p-value)	GTEx Portal V6 <i>!M</i>	2
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	tibial nerve	3.76×10 <sup>-13</sup> (p-value)	GTEx Portal V6 <i>!M</i>	2
MICB <i>e!</i> ?	ENSG00000204516 <i>e!</i>	tibial nerve	1.52×10 <sup>-9</sup> (p-value)	GTEx Portal V6 <i>!M</i>	2
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i>	esophagus mucosa	3.15×10 <sup>-11</sup> (p-value)	GTEx Portal V6 <i>!M</i>	2
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	esophagus mucosa	4.80×10 <sup>-12</sup> (p-value)	GTEx Portal V6 <i>!M</i>	2
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i>	gastroesophageal junction	1.40×10 <sup>-8</sup> (p-value)	GTEx Portal V6 <i>!M</i>	2
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	gastroesophageal junction	3.56×10 <sup>-7</sup> (p-value)	GTEx Portal V6 <i>!M</i>	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i>	adrenal gland	9.32×10 <sup>-10</sup> (p-value)	GTEx Portal V6 <i>!M</i>	2
TNXB <i>e!</i> ?	ENSG00000168477 <i>e!</i>	adrenal gland	2.07×10 <sup>-6</sup> (p-value)	GTEx Portal V6 <i>!M</i>	1
TNXA <i>e!</i> ?	ENSG00000248290 <i>e!</i>	adrenal gland	3.08×10 <sup>-7</sup> (p-value)	GTEx Portal V6 <i>!M</i>	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i>	testis	1.12×10 <sup>-8</sup> (p-value)	GTEx Portal V6 <i>!M</i>	2
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	testis	3.86×10 <sup>-12</sup> (p-value)	GTEx Portal V6 <i>!M</i>	2
MICB <i>e!</i>	ENST00000538442 <i>e!</i>	ILMN_1708006 <i>e!</i> skin	8.50×10 <sup>-10</sup> (p-value)	MuTHER consortium <i>!M</i>	1
MICB <i>e!</i>	ENST00000399150 <i>e!</i>	blood	3.34×10 <sup>-6</sup> (p-value)	MuTHER consortium <i>!M</i>	1
MICB <i>e!</i>	ENST00000252229 <i>e!</i>				
MICB <i>e!</i>	ENST00000399150 <i>e!</i>	ILMN_1708006 <i>e!</i> monocyte	3.63×10 <sup>-6</sup> (p-value)	Fairfax et al. <i>!M</i>	1
MICB <i>e!</i>	ENST00000538442 <i>e!</i>	b-cell	4.35×10 <sup>-9</sup> (p-value)	Fairfax et al. <i>!M</i>	1
MICB <i>e!</i>	ENST00000252229 <i>e!</i>				

### Putative effect on transcript

#### Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HCP5 <i>e!</i>	ENST00000414046 <i>e!</i> ?		?	2
MICA <i>e!</i>	ENST00000616296 <i>e!</i>	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382 <i>e!</i>	1
MICA <i>e!</i>	ENST00000449934 <i>e!</i>	NM_001177519.2	ENSP00000413079 <i>e!</i>	1
MICA <i>e!</i>	ENST00000421350 <i>e!</i> ?		ENSP00000402410 <i>e!</i>	1

