

# SNiPACard

## Block annotations

### Block info


genomic range	chr6:31,446,466-31,452,103 <i>e!</i>
block size	5,638 bp
variant count	4 variants

### Basic features



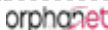
Conservation/deleteriousness		Linked genes	
<b>phyloP</b>	$\mu = -0.604$ [-1.421 -- 0.238]	<b>gene(s) hit or close-by</b>	HCP5 <i>e!</i>
<b>phastCons</b>	$\mu = 0.004$ [0.001 – 0.006]	<b>eQTL gene(s)</b>	ATP6V1G2 <i>e!</i> , HCG27 <i>e!</i> , HLA-B <i>e!</i> , HLA-C <i>e!</i> , LST1 <i>e!</i> , LY6G5C <i>e!</i> , MICA <i>e!</i> , MICB <i>e!</i> , PSORS1C1 <i>e!</i> , PSORS1C2 <i>e!</i> , XXbac-BPG181B23.7 <i>e!</i> , XXbac-BPG248L24.12 <i>e!</i>
<b>GERP++</b>	$\mu = -0.018$ [-0.737 – 0.458]	<b>potentially regulated gene(s)</b>	CCHCR1 <i>e!</i> , CCHCR1 <i>e!</i> , CCHCR1 <i>e!</i> , CCHCR1 <i>e!</i> , CCHCR1 <i>e!</i> , CCHCR1 <i>e!</i> , LY6G6D <i>e!</i> , LY6G6D <i>e!</i> , LY6G6D <i>e!</i> , LY6G6D <i>e!</i> , LY6G6D <i>e!</i> , LY6G6D <i>e!</i> , LY6G6E <i>e!</i> , LY6G6E <i>e!</i> , LY6G6E <i>e!</i> , LY6G6E <i>e!</i> , LY6G6E <i>e!</i> , LY6G6E <i>e!</i> , SLC44A4 <i>e!</i> , SLC44A4 <i>e!</i> , SLC44A4 <i>e!</i> , SLC44A4 <i>e!</i> , SLC44A4 <i>e!</i> , SLC44A4 <i>e!</i> , SLC44A4 <i>e!</i> , SLC44A4 <i>e!</i> , WASF5P <i>e!</i> , WASF5P <i>e!</i> , WASF5P <i>e!</i> , WASF5P <i>e!</i> , WASF5P <i>e!</i> , WASF5P <i>e!</i> , WASF5P <i>e!</i>
<b>CADD score</b>	$\mu = 2.082$ [0.871 – 4.161]	<b>disease gene(s)</b>	HLA-B <i>e!</i>

## Trait annotations

### Variant association






trait	min(p-value)	source DB	source entry/link	variant(s)
gamma-glutamylglutamine	$2.92 \times 10^{-5}$	Metabolomics GWAS Server	24816252 	1

### Disease gene annotation

gene	trait	source DB	source entry/link
HLA-B <i>e!</i>	Takayasu arteritis	OrphaNet	OrphaNet:3287 
HLA-B <i>e!</i>	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426 
HLA-B <i>e!</i>	Behcet disease	OrphaNet	OrphaNet:117 

## Direct effect on regulation

### cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PSORS1C1 <i>e!</i>	ENST00000481450 <i>e!</i>	ILMN_1799669 <i>e!</i>	skin	$2.59 \times 10^{-5}$ (p-value)	MuTHER consortium 	1
PSORS1C1 <i>e!</i>	ENST00000552747 <i>e!</i>					
PSORS1C1 <i>e!</i>	ENST00000547221 <i>e!</i>					
PSORS1C1 <i>e!</i>	ENST00000259881 <i>e!</i>					
PSORS1C1 <i>e!</i>	ENST00000479581 <i>e!</i>					
ATP6V1G2 <i>e!</i>	ENST00000481998 <i>e!</i>	ILMN_1654541 <i>e!</i>	blood	$7.59 \times 10^{-6}$ (p-value)	MuTHER consortium 	2
ATP6V1G2 <i>e!</i>	ENST00000303892 <i>e!</i>		skin	$2.90 \times 10^{-5}$ (p-value)	MuTHER consortium 	2
ATP6V1G2 <i>e!</i>	ENST00000483170 <i>e!</i>					
ATP6V1G2 <i>e!</i>	ENST00000376151 <i>e!</i>					
MICB <i>e!</i>	ENST00000538442 <i>e!</i>	ILMN_1708006 <i>e!</i>	skin	$1.42 \times 10^{-9}$ (p-value)	MuTHER consortium 	2
MICB <i>e!</i>	ENST00000399150 <i>e!</i>					
MICB <i>e!</i>	ENST00000252229 <i>e!</i>					
HCG27 <i>e!</i>	ENST00000383331 <i>e!</i>	ILMN_1746436 <i>e!</i>	adipocyte	$3.10 \times 10^{-5}$ (p-value)	MuTHER consortium 	1
HCG27 <i>e!</i>	ENST00000415276 <i>e!</i>					

XXbac-BPG248L24.12	e!	?	ENSG00000271581	e!	lung	7.99×10 <sup>-6</sup> (p-value)	GTEEx Portal V6		1		
MICA	e!	?	ENSG00000204520	e!	lung	2.96×10 <sup>-6</sup> (p-value)	GTEEx Portal V6		1		
XXbac-BPG181B23.7	e!	?	ENSG00000272221	e!	lung	2.36×10 <sup>-6</sup> (p-value)	GTEEx Portal V6		1		
XXbac-BPG248L24.12	e!	?	ENSG00000271581	e!	transformed fibroblasts	1.29×10 <sup>-5</sup> (p-value)	GTEEx Portal V6		1		
MICA	e!	?	ENSG00000204520	e!	transformed fibroblasts	1.06×10 <sup>-8</sup> (p-value)	GTEEx Portal V6		3		
XXbac-BPG181B23.7	e!	?	ENSG00000272221	e!	transformed fibroblasts	7.57×10 <sup>-6</sup> (p-value)	GTEEx Portal V6		1		
XXbac-BPG248L24.12	e!	?	ENSG00000271581	e!	blood	4.00×10 <sup>-7</sup> (p-value)	GTEEx Portal V6		4		
XXbac-BPG248L24.12	e!	?	ENSG00000271581	e!	thyroid	1.30×10 <sup>-6</sup> (p-value)	GTEEx Portal V6		4		
XXbac-BPG181B23.7	e!	?	ENSG00000272221	e!	thyroid	1.17×10 <sup>-6</sup> (p-value)	GTEEx Portal V6		1		
XXbac-BPG248L24.12	e!	?	ENSG00000271581	e!	skeletal muscle	1.19×10 <sup>-8</sup> (p-value)	GTEEx Portal V6		4		
MICB	e!	?	ENSG00000204516	e!	blood	1.47×10 <sup>-2</sup> (q-value)	SeeQTL DB (HapMap)		1		
MICA	e!	?	ENSG00000204520	e!	sun exposed skin	1.48×10 <sup>-9</sup> (p-value)	GTEEx Portal V6		4		
XXbac-BPG248L24.12	e!	?	ENSG00000271581	e!	sun exposed skin	1.96×10 <sup>-6</sup> (p-value)	GTEEx Portal V6		1		
XXbac-BPG181B23.7	e!	?	ENSG00000272221	e!	sun exposed skin	3.64×10 <sup>-9</sup> (p-value)	GTEEx Portal V6		4		
PSORS1C1	e!	?	ENSG00000204540	e!	sun exposed skin	4.04×10 <sup>-7</sup> (p-value)	GTEEx Portal V6		4		
MICB	e!	?	ENSG00000204516	e!	sun exposed skin	6.17×10 <sup>-6</sup> (p-value)	GTEEx Portal V6		2		
XXbac-BPG248L24.12	e!	?	ENSG00000271581	e!	left ventricle	2.30×10 <sup>-6</sup> (p-value)	GTEEx Portal V6		1		
MICA	e!	?	ENSG00000204520	e!	left ventricle	1.79×10 <sup>-6</sup> (p-value)	GTEEx Portal V6		3		
XXbac-BPG248L24.12	e!	?	ENSG00000271581	e!	subcutaneous adipocytes	1.88×10 <sup>-6</sup> (p-value)	GTEEx Portal V6		4		
PSORS1C1	e!	?	ENSG00000204540	e!	subcutaneous adipocytes	2.65×10 <sup>-6</sup> (p-value)	GTEEx Portal V6		3		
HLA-C	e!		ENST00000470363	e!	ILMN_2150787	e!	monocyte	1.54×10 <sup>-16</sup> (p-value)	Zeller et al.		1
HLA-C	e!		ENST00000383329	e!							
HLA-C	e!		ENST00000487245	e!							
HLA-C	e!		ENST00000376228	e!							
HLA-C	e!		ENST00000376237	e!							
HLA-C	e!		ENST00000620806	e!							
HLA-C	e!		ENST00000466892	e!							
PSORS1C1	e!	?	ENSG00000204540	e!	tibial nerve	5.92×10 <sup>-9</sup> (p-value)	GTEEx Portal V6		4		
PSORS1C2	e!	?	ENSG00000204538	e!	esophagus mucosa	7.70×10 <sup>-8</sup> (p-value)	GTEEx Portal V6		4		
XXbac-BPG248L24.12	e!	?	ENSG00000271581	e!	esophagus mucosa	2.02×10 <sup>-9</sup> (p-value)	GTEEx Portal V6		4		
PSORS1C1	e!	?	ENSG00000204540	e!	esophagus mucosa	9.36×10 <sup>-9</sup> (p-value)	GTEEx Portal V6		4		
LY6G5C	e!		ENST00000383237	e!	ILMN_1763467	e!	blood	1.29×10 <sup>-4</sup> (p-value)	Westra et al.		1
LY6G5C	e!		ENST00000474395	e!							
LY6G5C	e!		ENST00000375863	e!							
LY6G5C	e!		ENST00000474678	e!							
LY6G5C	e!		ENST00000467098	e!							
LY6G5C	e!		ENST00000460141	e!							
ATP6V1G2	e!		ENST00000481998	e!	ILMN_1654541	e!	blood	1.05×10 <sup>-4</sup> (p-value)	Westra et al.		1
ATP6V1G2	e!		ENST00000303892	e!							
ATP6V1G2	e!		ENST00000483170	e!							
ATP6V1G2	e!		ENST00000376151	e!							

LST1 <i>e!</i>	ENST00000419073 <i>e!</i>	ILMN_1717127 <i>e!</i>	blood	7.89×10 <sup>-5</sup> (p-value)	Westra et al. <i>!</i>	1
LST1 <i>e!</i>	ENST00000433492 <i>e!</i>					
LST1 <i>e!</i>	ENST00000376089 <i>e!</i>					
LST1 <i>e!</i>	ENST00000376090 <i>e!</i>					
LST1 <i>e!</i>	ENST00000211921 <i>e!</i>					
XXbac-BPG181B23.7 <i>e!</i>	?	ENSG00000272221 <i>e!</i>	testis	8.41×10 <sup>-8</sup> (p-value)	GTE Portal V6 <i>!</i>	4
HLA-B <i>e!</i>	?	ENSG00000234745 <i>e!</i>	cerebellum	1.63×10 <sup>-6</sup> (p-value)	GTE Portal V6 <i>!</i>	1
XXbac-BPG181B23.7 <i>e!</i>	?	ENSG00000272221 <i>e!</i>	tibial artery	1.16×10 <sup>-6</sup> (p-value)	GTE Portal V6 <i>!</i>	3
PSORS1C1 <i>e!</i>	?	ENSG00000204540 <i>e!</i>	unexposed skin	1.03×10 <sup>-6</sup> (p-value)	GTE Portal V6 <i>!</i>	2
XXbac-BPG248L24.12 <i>e!</i>	?	ENSG00000271581 <i>e!</i>	breast	2.78×10 <sup>-6</sup> (p-value)	GTE Portal V6 <i>!</i>	1

### Putative effect on regulation

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

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444780 <i>e!</i>	1	ENCP00000048082	LY6G6E <i>e!</i> LY6G6E <i>e!</i> LY6G6E <i>e!</i> LY6G6D <i>e!</i> LY6G6D <i>e!</i> LY6G6D <i>e!</i> LY6G6E <i>e!</i> LY6G6D <i>e!</i> LY6G6D <i>e!</i> LY6G6E <i>e!</i> LY6G6E <i>e!</i> LY6G6E <i>e!</i> LY6G6D <i>e!</i> LY6G6D <i>e!</i>
		ENCP00000047982	CCHCR1 <i>e!</i> CCHCR1 <i>e!</i> CCHCR1 <i>e!</i> CCHCR1 <i>e!</i> CCHCR1 <i>e!</i> CCHCR1 <i>e!</i>
		ENCP00000048120	SLC44A4 <i>e!</i> SLC44A4 <i>e!</i> SLC44A4 <i>e!</i> SLC44A4 <i>e!</i> SLC44A4 <i>e!</i> SLC44A4 <i>e!</i> SLC44A4 <i>e!</i>
		ENCP00000048000	WASF5P <i>e!</i> WASF5P <i>e!</i> WASF5P <i>e!</i> WASF5P <i>e!</i> WASF5P <i>e!</i> WASF5P <i>e!</i> WASF5P <i>e!</i>
		ENCP00000047981	CCHCR1 <i>e!</i> CCHCR1 <i>e!</i> CCHCR1 <i>e!</i> CCHCR1 <i>e!</i> CCHCR1 <i>e!</i> CCHCR1 <i>e!</i>

#### Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
------	--------------	---------------	------------	-----------	---------	------------

HCP5 *e!*

downstream gene variant

1183

ENST00000467369 *e!*

?

?

2

---

