# SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| **genomic range** | chr6:31,360,702-31,426,888 |
| **block size** | 66,187 bp |
| **variant count** | 22 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| **phyloP** | μ = -0.290 [-1.89 – 0.318] | **gene(s) hit or close-by** | HCP5, LINC01149, MICA, XXbac-BPG181B23.6, XXbac-BPG181B23.7, Y_RNA |
| **phastCons** | μ = 0.118 [0 – 0.47] | **eQTL gene(s)** | HCG22, HLA-B, LY6G5C, MICA, XXbac-BPG181B23.7 |
| **GERP++** | μ = 0.053 [-0.718 – 0.641] | **potentially regulated gene(s)** | ABHD16A, ABHD16A, ABHD16A, ABHD16A, ABHD16A, ABHD16A, ABHD16A, AIF1, AIF1, AIF1, AIF1, AIF1, AIF1, BAG6, BAG6, BAG6, BAG6, BAG6, BAG6, BAG6, C2, C2, C2, C2, C2, C2, C2, C2, C6orf48, C6orf48, C6orf48, C6orf48, C6orf48, C6orf48, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, HCG22, HCG22, HCG22, HCG22, HCG22, HCG22, HCG22, HCG22, HCG27, HCG27, HCG27, HCG27, HCG27, HCG27, HCG27, HCP5, HCP5, HCP5, HCP5, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, LST1, LST1, LST1, LST1, LST1, LST1, LST1, LST1, LTB, LTB, LTB, LTB, LTB, LTB, LTB, LTB, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, MICA, MICA, MICA, MICA, MICA, MICB, MICB, MICB, MICB, MICB, MICB, MICB, MICB, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, TNF, TNF, TNF, TNF, TNF, TNF, TNF, TNF, TNF, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, VARS, VARS, VARS, VARS, VARS, VARS, VARS, XXbac-BPG299F13.16 |
| **CADD score** | μ = 3.431 [0.17 – 8.268] | **disease gene(s)** | HLA-B, C2, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, TNF, TNF, TNF |

## Trait annotations

### Variant association

| trait | min(p-value) | source DB | source entry/link | | variant(s) |
|---|---|---|---|---|---|
| Behcet syndrome | 1.25×10-6 | dbGaP | pha002888 | dbGaP | 1 |

### Disease gene annotation

| gene | trait | source DB | source entry/link | |
|---|---|---|---|---|
| HLA-B | Takayasu arteritis | OrphaNet | OrphaNet:3287 | orphanet |
| HLA-B | Stevens-Johnson syndrome | OrphaNet | OrphaNet:36426 | orphanet |
| HLA-B | Behcet disease | OrphaNet | OrphaNet:117 | orphanet |
| C2 | Immunodeficiency due to an early component of complement deficiency | OrphaNet | OrphaNet:169147 | orphanet |
| | PSORIASIS 1, SUSCEPTIBILITY TO | OMIM | MIM:177900 | OMIM |
| | PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...] | OMIM | MIM:607507 | OMIM |
| | PSORIASIS 1, SUSCEPTIBILITY TO | OMIM | MIM:177900 | OMIM |
| | PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...] | OMIM | MIM:607507 | OMIM |
| | SEVERE CUTANEOUS ADVERSE REACTION, SUSCEPTIBILITY TOTOXIC EPIDERMAL [...] | OMIM | MIM:608579 | OMIM |
| | HEPATITIS B VIRUS, SUSCEPTIBILITY TO | OMIM | MIM:610424 | OMIM |
| | MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED | OMIM | MIM:611162 | OMIM |
| | PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...] | OMIM | MIM:607507 | OMIM |
| | COMPLEMENT COMPONENT 2 DEFICIENCY | OMIM | MIM:217000 | OMIM |
| | MACULAR DEGENERATION, AGE-RELATED, 1 | OMIM | MIM:603075 | OMIM |

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|---|---|---|---|---|
| | HEPATITIS B VIRUS, SUSCEPTIBILITY TO | OMIM | MIM:610424 | OMIM |
| | MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED | OMIM | MIM:611162 | OMIM |
| | PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...] | OMIM | MIM:607507 | OMIM |
| | HEPATITIS B VIRUS, SUSCEPTIBILITY TO | OMIM | MIM:610424 | OMIM |
| | MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED | OMIM | MIM:611162 | OMIM |
| | PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...] | OMIM | MIM:607507 | OMIM |
| | COMPLEMENT COMPONENT 2 DEFICIENCY | OMIM | MIM:217000 | OMIM |
| | MACULAR DEGENERATION, AGE-RELATED, 1 | OMIM | MIM:603075 | OMIM |
| | HEPATITIS B VIRUS, SUSCEPTIBILITY TO | OMIM | MIM:610424 | OMIM |
| | MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED | OMIM | MIM:611162 | OMIM |
| | PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...] | OMIM | MIM:607507 | OMIM |
| | COMPLEMENT COMPONENT 2 DEFICIENCY | OMIM | MIM:217000 | OMIM |
| | MACULAR DEGENERATION, AGE-RELATED, 1 | OMIM | MIM:603075 | OMIM |
| | COMPLEMENT COMPONENT 2 DEFICIENCY | OMIM | MIM:217000 | OMIM |
| | MACULAR DEGENERATION, AGE-RELATED, 1 | OMIM | MIM:603075 | OMIM |
| | HEPATITIS B VIRUS, SUSCEPTIBILITY TO | OMIM | MIM:610424 | OMIM |
| | MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED | OMIM | MIM:611162 | OMIM |
| | PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...] | OMIM | MIM:607507 | OMIM |
| | PSORIASIS 1, SUSCEPTIBILITY TO | OMIM | MIM:177900 | OMIM |
| C2 | COMPLEMENT COMPONENT 2 DEFICIENCY | OMIM | MIM:217000 | OMIM |
| C2 | MACULAR DEGENERATION, AGE-RELATED, 1 | OMIM | MIM:603075 | OMIM |
| | HEPATITIS B VIRUS, SUSCEPTIBILITY TO | OMIM | MIM:610424 | OMIM |
| | MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED | OMIM | MIM:611162 | OMIM |
| | PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...] | OMIM | MIM:607507 | OMIM |
| TNF | HEPATITIS B VIRUS, SUSCEPTIBILITY TO | OMIM | MIM:610424 | OMIM |
| TNF | MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED | OMIM | MIM:611162 | OMIM |
| TNF | PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...] | OMIM | MIM:607507 | OMIM |
| | COMPLEMENT COMPONENT 2 DEFICIENCY | OMIM | MIM:217000 | OMIM |
| | MACULAR DEGENERATION, AGE-RELATED, 1 | OMIM | MIM:603075 | OMIM |
| | COMPLEMENT COMPONENT 2 DEFICIENCY | OMIM | MIM:217000 | OMIM |
| | MACULAR DEGENERATION, AGE-RELATED, 1 | OMIM | MIM:603075 | OMIM |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| MICA | ? | ENSG00000204520 | atrial appendage | $2.93\times10^{-8}$ (p-value) | GTEx Portal V6 | 13 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | atrial appendage | $1.51\times10^{-7}$ (p-value) | GTEx Portal V6 | 21 |
| MICA | ? | ENSG00000204520 | transformed fibroblasts | $8.78\times10^{-16}$ (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | transformed fibroblasts | $2.90\times10^{-18}$ (p-value) | GTEx Portal V6 | 22 |
| MICA | ? | ENSG00000204520 | blood | $4.17\times10^{-14}$ (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | blood | $1.62\times10^{-33}$ (p-value) | GTEx Portal V6 | 22 |
| MICA | ? | ENSG00000204520 | breast | $2.51\times10^{-10}$ (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | breast | $1.83\times10^{-13}$ (p-value) | GTEx Portal V6 | 22 |
| MICA | ? | ENSG00000204520 | skeletal muscle | $2.44\times10^{-19}$ (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | skeletal muscle | $2.86\times10^{-20}$ (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | putamen | $3.55\times10^{-8}$ (p-value) | GTEx Portal V6 | 21 |
| MICA | ? | ENSG00000204520 | putamen | $4.08\times10^{-7}$ (p-value) | GTEx Portal V6 | 11 |

| | | | | | | |
|---|---|---|---|---|---|---|
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | EBV lymphocytes | 1.99×10-8 (p-value) | GTEx Portal V6 | 22 |
| MICA | ? | ENSG00000204520 | sun exposed skin | 5.53×10-19 (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | sun exposed skin | 1.08×10-20 (p-value) | GTEx Portal V6 | 22 |
| MICA | ? | ENSG00000204520 | unexposed skin | 1.60×10-12 (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | unexposed skin | 3.90×10-11 (p-value) | GTEx Portal V6 | 22 |
| MICA | ? | ENSG00000204520 | aorta | 1.38×10-10 (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | aorta | 1.27×10-13 (p-value) | GTEx Portal V6 | 22 |
| MICA | ? | ENSG00000204520 | left ventricle | 7.94×10-9 (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | left ventricle | 2.20×10-12 (p-value) | GTEx Portal V6 | 22 |
| MICA | ? | ENSG00000204520 | subcutaneous adipocytes | 3.63×10-18 (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | subcutaneous adipocytes | 1.79×10-20 (p-value) | GTEx Portal V6 | 22 |
| MICA | ? | ENSG00000204520 | caudate basal ganglia | 8.43×10-9 (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | caudate basal ganglia | 1.67×10-8 (p-value) | GTEx Portal V6 | 22 |
| MICA | ? | ENSG00000204520 | visceral adipocytes | 2.35×10-12 (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | visceral adipocytes | 2.37×10-11 (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | stomach | 4.11×10-8 (p-value) | GTEx Portal V6 | 5 |
| MICA | ? | ENSG00000204520 | cortex | 9.16×10-12 (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | cortex | 2.89×10-9 (p-value) | GTEx Portal V6 | 21 |
| MICA | ? | ENSG00000204520 | tibial nerve | 3.93×10-13 (p-value) | GTEx Portal V6 | 22 |
| HLA-B | ? | ENSG00000234745 | tibial nerve | 1.12×10-7 (p-value) | GTEx Portal V6 | 12 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | tibial nerve | 7.42×10-17 (p-value) | GTEx Portal V6 | 22 |
| MICA | ? | ENSG00000204520 | esophagus mucosa | 3.85×10-12 (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | esophagus mucosa | 1.01×10-11 (p-value) | GTEx Portal V6 | 22 |
| MICA | ? | ENSG00000204520 | pituitary | 3.36×10-9 (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | pituitary | 1.09×10-9 (p-value) | GTEx Portal V6 | 12 |
| MICA | ? | ENSG00000204520 | testis | 3.08×10-10 (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | testis | 3.09×10-7 (p-value) | GTEx Portal V6 | 22 |
| MICA | ? | ENSG00000204520 | frontal cortex | 4.84×10-9 (p-value) | GTEx Portal V6 | 13 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | pancreas | 3.99×10-10 (p-value) | GTEx Portal V6 | 22 |
| MICA | ? | ENSG00000204520 | pancreas | 1.39×10-7 (p-value) | GTEx Portal V6 | 18 |
| MICA | ? | ENSG00000204520 | muscularis mucosae | 3.04×10-13 (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | muscularis mucosae | 2.96×10-12 (p-value) | GTEx Portal V6 | 22 |
| HCG22 | ? | ENSG00000228789 | muscularis mucosae | 4.78×10-6 (p-value) | GTEx Portal V6 | 7 |
| MICA | ? | ENSG00000204520 | lung | 1.15×10-13 (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | lung | 3.96×10-21 (p-value) | GTEx Portal V6 | 22 |
| MICA | ? | ENSG00000204520 | tibial artery | 1.35×10-16 (p-value) | GTEx Portal V6 | 22 |

| | | | | | | |
|---|---|---|---|---|---|---|
| HLA-B | ? | ENSG00000234745 | tibial artery | 3.93×10-6 (p-value) | GTEx Portal V6 | 4 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | tibial artery | 1.76×10-19 (p-value) | GTEx Portal V6 | 22 |
| MICA | ? | ENSG00000204520 | thyroid | 2.47×10-16 (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | thyroid | 3.66×10-17 (p-value) | GTEx Portal V6 | 22 |
| MICA | ? | ENSG00000204520 | transverse colon | 1.21×10-8 (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | transverse colon | 1.07×10-11 (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | liver | 4.70×10-8 (p-value) | GTEx Portal V6 | 12 |
| MICA | ? | ENSG00000204520 | nucleus accumbens | 1.16×10-7 (p-value) | GTEx Portal V6 | 13 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | coronary artery | 3.81×10-7 (p-value) | GTEx Portal V6 | 4 |
| MICA | ? | ENSG00000204520 | sigmoid colon | 3.24×10-11 (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | sigmoid colon | 4.97×10-10 (p-value) | GTEx Portal V6 | 22 |
| MICA | ? | ENSG00000204520 | gastroesophageal junction | 1.08×10-10 (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | gastroesophageal junction | 8.57×10-12 (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | adrenal gland | 7.92×10-7 (p-value) | GTEx Portal V6 | 4 |
| MICA | ? | ENSG00000204520 | hippocampus | 1.98×10-8 (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG181B23.7 | ? | ENSG00000272221 | hippocampus | 4.35×10-8 (p-value) | GTEx Portal V6 | 22 |
| HCG22 | ENST00000570223 | ILMN_1667229 | blood | 4.45×10-7 (p-value) | MuTHER consortium | 2 |
| HCG22 | ENST00000565192 | | | | | |
| HCG22 | ENST00000615046 | | | | | |
| HCG22 | ENST00000426185 | | | | | |
| HCG22 | ENST00000562344 | | | | | |
| MICA | ENST00000449934 | ILMN_3241692 | monocyte | 1.19×10-10 (p-value) | Fairfax et al. | 2 |
| MICA | ENST00000616296 | | b-cell | 5.10×10-8 (p-value) | Fairfax et al. | 2 |
| MICA | ENST00000421350 | | | | | |
| MICA | ? | ENSG00000204520 | vagina | 5.53×10-7 (p-value) | GTEx Portal V6 | 10 |
| LY6G5C | ENST00000383237 | ILMN_1763467 | blood | 4.51×10-6 (p-value) | Westra et al. | 1 |
| LY6G5C | ENST00000474395 | | | | | |
| LY6G5C | ENST00000375863 | | | | | |
| LY6G5C | ENST00000474678 | | | | | |
| LY6G5C | ENST00000467098 | | | | | |
| LY6G5C | ENST00000460141 | | | | | |

## Putative effect on regulation

### ENCODE promoter-associated DHS

| SNiPA promoter id | variant(s) | associated gene(s) |
|---|---|---|
| ENCP00000048013 | | MICA |
| | | MICA |
| | | HCP5 |
| | | MICA |
| | | HCP5 |
| | | MICA |

| | | | |
|---|---|---|---|
| | | | MICA |
| | | | HCP5 |

## ENCODE promoter-associated distal DHS (Enhancer)

| SNiPA enhancer id | variant(s) | associated SNiPA promoter id | associated gene(s) |
|---|---|---|---|
| ENCE00000444737 | 1 | ENCP00000048058 | BAG6<br>BAG6<br>BAG6<br>BAG6<br>BAG6<br>BAG6<br>BAG6 |
| | | ENCP00000048034 | LTB<br>LTB<br>LTB<br>LTB<br>LTB<br>LTB<br>LTB<br>LTB |
| | | ENCP00000048025 | DDX39B<br>DDX39B<br>DDX39B<br>DDX39B<br>DDX39B<br>DDX39B<br>DDX39B<br>DDX39B |
| | | ENCP00000047992 | HCG27<br>HCG27<br>HCG27<br>HCG27<br>HCG27<br>HCG27<br>HCG27 |
| | | ENCP00000048062 | CSNK2B<br>CSNK2B<br>CSNK2B<br>CSNK2B<br>CSNK2B<br>CSNK2B<br>CSNK2B |
| | | ENCP00000048036 | LST1<br>LST1<br>LST1<br>LST1<br>LST1<br>LST1<br>LST1<br>LST1 |
| | | ENCP00000048043 | AIF1<br>AIF1<br>AIF1<br>AIF1<br>AIF1<br>AIF1 |
| | | ENCP00000048007 | HLA-B<br>HLA-B<br>HLA-B<br>HLA-B<br>HLA-B<br><br>HLA-B |
| | | ENCP00000048063 | GPANK1<br>GPANK1<br>GPANK1<br>GPANK1<br>GPANK1<br>GPANK1<br>GPANK1 |

| | | | |
|---|---|---|---|
| | | ENCP00000047991 | HCG27<br>HCG27<br>HCG27<br>HCG27<br>HCG27<br>HCG27<br>HCG27 |
| | | ENCP00000047975 | HCG22<br>HCG22<br>HCG22<br>HCG22<br>HCG22<br>HCG22<br>HCG22<br>HCG22 |
| | | ENCP00000047995 | XXbac-BPG299F13.16 |
| | | ENCP00000048019 | MICB<br>MICB<br>MICB<br>MICB<br>MICB<br>MICB<br>MICB<br>MICB |
| | | ENCP00000048078 | ABHD16A<br>ABHD16A<br>ABHD16A<br>ABHD16A<br>ABHD16A<br>ABHD16A<br>ABHD16A |
| ENCE00000444880 | 1 | ENCP00000048065 | CSNK2B<br>GPANK1<br>GPANK1<br>GPANK1<br>CSNK2B<br>CSNK2B<br>CSNK2B<br>CSNK2B<br>CSNK2B<br>CSNK2B<br>GPANK1<br>GPANK1<br>GPANK1<br>GPANK1 |
| | | ENCP00000048041 | AIF1<br>AIF1<br>AIF1<br>AIF1<br>AIF1<br>AIF1 |
| | | ENCP00000048069 | LY6G5C<br>LY6G5C<br>LY6G5C<br>LY6G5C<br>LY6G5C<br>LY6G5C<br>LY6G5C |
| | | ENCP00000048050 | PRRC2A<br>PRRC2A<br>PRRC2A<br>PRRC2A<br>PRRC2A<br>PRRC2A<br>PRRC2A |
| | | ENCP00000048017 | HCP5<br>HCP5<br>HCP5<br>HCP5 |
| | | ENCP00000047997 | HLA-C |

| | |
|---|---|
| | HLA-C<br>HLA-C<br>HLA-C<br>HLA-C<br>HLA-C<br>HLA-C |
| ENCP00000048023 | DDX39B<br>DDX39B<br>DDX39B<br>DDX39B<br>DDX39B<br>DDX39B<br>DDX39B<br>DDX39B |
| ENCP00000048104 | VARS<br>VARS<br>VARS<br>VARS<br>VARS<br>VARS<br>VARS |
| ENCP00000048052 | PRRC2A<br>PRRC2A<br>PRRC2A<br>PRRC2A<br>PRRC2A<br>PRRC2A<br>PRRC2A |
| ENCP00000048110 | VARS<br>VARS<br>VARS<br>VARS<br>VARS<br>VARS<br>VARS |
| ENCP00000048109 | VARS<br>VARS<br>VARS<br>VARS<br>VARS<br>VARS<br>VARS |
| ENCP00000048051 | PRRC2A<br>PRRC2A<br>PRRC2A<br>PRRC2A<br>PRRC2A<br>PRRC2A<br>PRRC2A |
| ENCP00000048033 | TNF<br>TNF<br>TNF<br>TNF<br>TNF<br>TNF<br>TNF<br>TNF |
| ENCP00000048035 | LST1<br>LST1<br>LST1<br>LST1<br>LST1<br>LST1<br>LST1<br>LST1 |
| ENCP00000047998 | HLA-C<br>HLA-C<br>HLA-C<br>HLA-C<br>HLA-C<br>HLA-C |

| | | |
|---|---|---|
| | | HLA-C |
| | ENCP00000048116 | C6orf48<br>C6orf48<br>C6orf48<br>C6orf48<br>C6orf48 |
| | ENCP00000048043 | AIF1<br>AIF1<br>AIF1<br>AIF1<br>AIF1<br>AIF1 |
| | ENCP00000048007 | HLA-B<br>HLA-B<br>HLA-B<br>HLA-B<br>HLA-B<br>HLA-B |
| | ENCP00000048048 | PRRC2A<br>PRRC2A<br>PRRC2A<br>PRRC2A<br>PRRC2A<br>PRRC2A<br>PRRC2A |
| | ENCP00000048020 | MICB<br>MICB<br>MICB<br>MICB<br>MICB<br>MICB<br>MICB<br>MICB |
| | ENCP00000048134 | C2<br>C2<br>C2<br>C2<br>C2<br>C2<br>C2<br>C2 |
| | ENCP00000048040 | UQCRHP1<br>UQCRHP1<br>UQCRHP1<br>UQCRHP1<br>UQCRHP1<br>UQCRHP1 |
| | ENCP00000048037 | LST1<br>LST1<br>LST1<br>LST1<br>LST1<br>LST1<br>LST1<br>LST1 |

## Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00000487835 (promoter) | 1 | embryonic stem cell (H1ESC) | DNase1, H3K9ac, H3K36me3, H3K27me3, PolII, Rad21, Egr1, CTCF, Yy1, Tcf12, TAF1, H3K4me2, SP1, USF1, H3K4me3 |
| | | HSMMtube | CTCF, H3K9ac, H3K36me3, H2AZ, H3K27ac, H3K4me2, DNase1 |
| | | blood (K562) | Ini1, Brg1, Egr1, H3K27ac, DNase1, H3K4me3, H3K36me3, H3K4me1, Max, Rad21, E2F6, H3K9ac, USF1, Cmyc, H3K79me2, HEY1, H2AZ, TAF1, Nrsf, Cfos, PolII, H3K4me2, CTCF |
| | | skin (NHDF-AD) | H3K27ac, H3K4me2, H3K9ac, CTCF, H3K4me3, DNase1 |
| | | muscle (HSMM) | H3K36me3, DNase1, H3K9ac, H3K4me2, H2AZ, H3K79me2, CTCF, H3K4me3, H3K27ac |
| | | liver (HepG2) | DNase1, H3K4me3, H3K27me3, Cmyc, CTCF, Gabp, PolII, H3K79me2, Rad21, TAF1, USF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac |
| | | lung (IMR90) | H3K4me3, H3K9ac, CTCF, H3K4me2, DNase1, H3K79me2 |
| | | blood (GM12878) | PolII, H2AZ, DNase1, Egr1, Yy1, Rad21, H3K79me2, H3K4me3, H3K27ac, H3K4me2, CTCF, H3K9ac |

| | | | |
|---|---|---|---|
| | | nervous (NH-A) | DNase1, H3K9ac, H4K20me1, CTCF, H3K4me2, H3K4me3, H3K27ac, H3K36me3 |
| | | skin (NHEK) | H3K36me3, DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac |
| | | NHLF | DNase1, H3K4me3, CTCF, H3K9ac, H3K27ac |
| | | Osteobl | H3K36me3, CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ |
| | | blood (DND-41) | H3K4me1, CTCF, H3K27ac, H4K20me1, H3K9ac, H3K4me3, H3K36me3 |
| | | breast (HMEC) | DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, H3K36me3, CTCF |
| | | cervix (HeLa-S3) | DNase1, PolII, CTCF, Gabp, H3K79me2, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3 |
| | | monocytes (Monocytes-CD14+) | DNase1, H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3 |
| | | endothelium (HUVEC) | H3K36me3, Cjun, H3K4me2, H3K4me3, PolII, CTCF, DNase1 |
| | | A549 | H3K4me3, H3K36me3, H3K27ac, DNase1, CTCF, H3K9ac, H3K4me2 |
| ENSR00001703045 (CTCF binding site) | 1 | monocytes (Monocytes-CD14+) | H3K27me3 |
| | | endothelium (HUVEC) | H3K27me3 |
| | | HSMMtube | H3K27me3 |
| | | blood (DND-41) | H3K27me3 |
| ENSR00001703047 (open chromatin region) | 2 | endothelium (HUVEC) | H3K27me3 |
| | | embryonic stem cell (H1ESC) | H3K36me3 |
| | | A549 | H3K27me3 |

### Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| HCP5 | upstream gene variant | 4071 | ENST00000541196 | ? | ? | 1 |
| HCP5 | upstream gene variant | 28 | ENST00000414046 | ? | ? | 1 |
| HCP5 | upstream gene variant | 4074 | ENST00000460889 | ? | ? | 1 |
| HCP5 | upstream gene variant | 4074 | ENST00000467369 | ? | ? | 1 |
| LINC01149 | downstream gene variant | 2 | ENST00000430364 | ? | ? | 7 |
| MICA | upstream gene variant, downstream gene variant | 278 | ENST00000421350 | ? | ENSP00000402410 | 2 |
| MICA | upstream gene variant, downstream gene variant | 278 | ENST00000449934 | NM_001177519.2 | ENSP00000413079 | 2 |
| MICA | downstream gene variant | 280 | ENST00000616296 | NM_001289152.1, NM_001289153.1, NM_001289154.1 | ENSP00000482382 | 1 |
| XXbac-BPG181B23.6 | downstream gene variant | 3617 | ENST00000440087 | ? | ? | 2 |
| XXbac-BPG181B23.7 | downstream gene variant | 983 | ENST00000606743 | ? | ? | 2 |
| Y_RNA | downstream gene variant | 1478 | ENST00000362462 | ? | ? | 1 |

## Putative effect on transcript

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| HCP5 | ENST00000414046 | ? | ? | 19 |
| LINC01149 | ENST00000430364 | ? | ? | 3 |
| MICA | ENST00000616296 | NM_001289152.1, NM_001289153.1, NM_001289154.1 | ENSP00000482382 | 1 |
| MICA | ENST00000421350 | ? | ENSP00000402410 | 1 |
| MICA | ENST00000449934 | NM_001177519.2 | ENSP00000413079 | 1 |

### 5'-UTR variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| MICA | ENST00000616296 | NM_001289152.1, NM_001289153.1, NM_001289154.1 | ENSP00000482382 | 1 |

### Non-coding exon variant

| gene | affected transcript | RefSeq id | variant(s) |
|---|---|---|---|
| LINC01149 | ENST00000430364 | ? | 2 |