# Locus 263 (rs28367631)

SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| **genomic range** | chr6:31,250,801-31,271,716 |
| **block size** | 20,916 bp |
| **variant count** | 127 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| **phyloP** | μ = -0.758 [-4.394 – 2.486] | **gene(s) hit or close-by** | HLA-C, RPL3P2, USP8P1, WASF5P, XXbac-BPG248L24.10, XXbac-BPG248L24.13 |
| **phastCons** | μ = 0.063 [0 – 0.998] | **eQTL gene(s)** | C4B, CCHCR1, CD248, DDR1, GPBAR1, HCG22, HCG27, HLA-B, HLA-C, MICA, MICB, PLOD3, POU5F1, PSORS1C1, PSORS1C2, PSORS1C3, TCF19, TMEM154, TRAPPC1, WASF5P, XXbac-BPG248L24.10, XXbac-BPG248L24.12, XXbac-BPG299F13.17 |
| **GERP++** | μ = -0.144 [-1.75 – 0.718] | **potentially regulated gene(s)** | CCHCR1, CCHCR1, CCHCR1, CCHCR1, CCHCR1, CCHCR1, DDAH2, DDAH2, DDAH2, DDAH2, DDAH2, DDAH2, DDAH2, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, SFTA2, SFTA2, SFTA2, SFTA2, SFTA2, SFTA2, WASF5P, WASF5P, WASF5P, WASF5P, WASF5P, WASF5P, WASF5P |
| **CADD score** | μ = 2.579 [0.016 – 17.05] | **disease gene(s)** | HLA-B, C4B, PLOD3 |

## Trait annotations

### Variant association

| trait | min(p-value) | source DB | source entry/link | variant(s) |
|---|---|---|---|---|
| Protein quantitative trait loci | <4.00×10-6 | GWAS Catalog | 18464913 | 1 |

### Disease gene annotation

| gene | trait | source DB | source entry/link |
|---|---|---|---|
| HLA-B | Takayasu arteritis | OrphaNet | OrphaNet:3287 |
| HLA-B | Stevens-Johnson syndrome | OrphaNet | OrphaNet:36426 |
| HLA-B | Behcet disease | OrphaNet | OrphaNet:117 |
| C4B | Immunodeficiency due to an early component of complement deficiency | OrphaNet | OrphaNet:169147 |
| PLOD3 | Connective tissue disorder due to lysyl hydroxylase-3 deficiency | OrphaNet | OrphaNet:300284 |
| C4B | COMPLEMENT COMPONENT 4B DEFICIENCY | OMIM | MIM:614379 |
| C4B | BLOOD GROUP, CHIDO/RODGERS SYSTEM | OMIM | MIM:614374 |
| C4B | SYSTEMIC LUPUS ERYTHEMATOSUS | OMIM | MIM:152700 |
| PLOD3 | BONE FRAGILITY WITH CONTRACTURES, ARTERIAL RUPTURE, AND [...] | OMIM | MIM:612394 |
| PLOD3 | LYSYL HYDROXYLASE 3 DEFICIENCY | DECIPHER | MIM:612394 |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| HCG22 | ENST00000570223 | ILMN_1667229 | blood | 5.04×10-5 (p-value) | MuTHER consortium | 1 |
| HCG22 | ENST00000565192 | | | | | |
| HCG22 | ENST00000615046 | | | | | |
| HCG22 | ENST00000426185 | | | | | |
| HCG22 | ENST00000562344 | | | | | |
| HCG27 | ENST00000383331 | ILMN_1746436 | adipocyte | 6.00×10-27 (p-value) | MuTHER consortium | 21 |

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|---|---|---|---|---|---|---|
| HCG27 | ENST00000415276 | | skin | 1.40×10-13 (p-value) | MuTHER consortium | 21 |
| DDR1 | ENST00000376567 | ILMN_1812262 | blood | 3.85×10-7 (p-value) | MuTHER consortium | 20 |
| DDR1 | ENST00000376570 | | | | | |
| DDR1 | ENST00000418800 | | | | | |
| DDR1 | ENST00000376569 | | | | | |
| DDR1 | ENST00000452441 | | | | | |
| DDR1 | ENST00000324771 | | | | | |
| DDR1 | ENST00000482873 | | | | | |
| DDR1 | ENST00000376568 | | | | | |
| DDR1 | ENST00000446312 | | | | | |
| DDR1 | ENST00000454612 | | | | | |
| HCG27 | ? | ENSG00000206344 | muscularis mucosae | 2.64×10-14 (p-value) | GTEx Portal V6 | 104 |
| HLA-C | ? | ENSG00000204525 | muscularis mucosae | 5.82×10-6 (p-value) | GTEx Portal V6 | 22 |
| HCG27 | ? | ENSG00000206344 | lung | 4.59×10-9 (p-value) | GTEx Portal V6 | 104 |
| PSORS1C3 | ? | ENSG00000204528 | lung | 1.12×10-7 (p-value) | GTEx Portal V6 | 95 |
| HLA-C | ? | ENSG00000204525 | lung | 2.82×10-7 (p-value) | GTEx Portal V6 | 99 |
| XXbac-BPG248L24.12 | ? | ENSG00000271581 | lung | 2.21×10-6 (p-value) | GTEx Portal V6 | 1 |
| PSORS1C3 | ? | ENSG00000204528 | atrial appendage | 1.61×10-9 (p-value) | GTEx Portal V6 | 100 |
| HLA-C | ? | ENSG00000204525 | atrial appendage | 4.10×10-7 (p-value) | GTEx Portal V6 | 86 |
| HCG27 | ? | ENSG00000206344 | atrial appendage | 1.72×10-6 (p-value) | GTEx Portal V6 | 4 |
| HCG27 | ? | ENSG00000206344 | transformed fibroblasts | 4.37×10-11 (p-value) | GTEx Portal V6 | 104 |
| HLA-C | ? | ENSG00000204525 | transformed fibroblasts | 4.61×10-13 (p-value) | GTEx Portal V6 | 104 |
| C4B | ? | ENSG00000224389 | transformed fibroblasts | 2.95×10-6 (p-value) | GTEx Portal V6 | 18 |
| XXbac-BPG248L24.12 | ? | ENSG00000271581 | transformed fibroblasts | 2.07×10-7 (p-value) | GTEx Portal V6 | 2 |
| PSORS1C3 | ? | ENSG00000204528 | blood | 4.75×10-11 (p-value) | GTEx Portal V6 | 104 |
| HLA-C | ? | ENSG00000204525 | blood | 4.64×10-15 (p-value) | GTEx Portal V6 | 104 |
| XXbac-BPG248L24.12 | ? | ENSG00000271581 | blood | 6.84×10-10 (p-value) | GTEx Portal V6 | 104 |
| XXbac-BPG299F13.17 | ? | ENSG00000272501 | blood | 3.88×10-13 (p-value) | GTEx Portal V6 | 104 |
| POU5F1 | ? | ENSG00000204531 | blood | 1.09×10-9 (p-value) | GTEx Portal V6 | 104 |
| HCG27 | ? | ENSG00000206344 | blood | 2.38×10-7 (p-value) | GTEx Portal V6 | 13 |
| HCG27 | ? | ENSG00000206344 | tibial artery | 7.32×10-13 (p-value) | GTEx Portal V6 | 104 |
| PSORS1C3 | ? | ENSG00000204528 | tibial artery | 4.66×10-8 (p-value) | GTEx Portal V6 | 100 |
| HLA-C | ? | ENSG00000204525 | tibial artery | 9.94×10-12 (p-value) | GTEx Portal V6 | 104 |
| XXbac-BPG248L24.12 | ? | ENSG00000271581 | tibial artery | 7.57×10-7 (p-value) | GTEx Portal V6 | 52 |
| HLA-B | ? | ENSG00000234745 | tibial artery | 1.09×10-5 (p-value) | GTEx Portal V6 | 5 |
| HCG27 | ? | ENSG00000206344 | thyroid | 1.80×10-15 (p-value) | GTEx Portal V6 | 104 |
| XXbac-BPG248L24.10 | ? | ENSG00000229836 | thyroid | 1.19×10-12 (p-value) | GTEx Portal V6 | 104 |
| HLA-C | ? | ENSG00000204525 | thyroid | 8.55×10-10 (p-value) | GTEx Portal V6 | 104 |
| XXbac-BPG248L24.12 | ? | ENSG00000271581 | thyroid | 2.30×10-8 (p-value) | GTEx Portal V6 | 90 |
| HLA-B | ? | ENSG00000234745 | thyroid | 7.33×10-8 (p-value) | GTEx Portal V6 | 96 |

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|---|---|---|---|---|---|---|
| WASF5P | ? | ENSG00000231402 | thyroid | 1.99×10-6 (p-value) | GTEx Portal V6 | 96 |
| PSORS1C3 | ? | ENSG00000204528 | thyroid | 1.58×10-6 (p-value) | GTEx Portal V6 | 62 |
| HCG27 | ? | ENSG00000206344 | skeletal muscle | 5.07×10-11 (p-value) | GTEx Portal V6 | 104 |
| HLA-C | ? | ENSG00000204525 | skeletal muscle | 3.36×10-13 (p-value) | GTEx Portal V6 | 104 |
| XXbac-BPG248L24.12 | ? | ENSG00000271581 | skeletal muscle | 1.36×10-9 (p-value) | GTEx Portal V6 | 102 |
| HCG27 | ? | ENSG00000206344 | transverse colon | 1.86×10-11 (p-value) | GTEx Portal V6 | 104 |
| HLA-C | ? | ENSG00000204525 | transverse colon | 1.19×10-6 (p-value) | GTEx Portal V6 | 27 |
| XXbac-BPG248L24.12 | ? | ENSG00000271581 | transverse colon | 3.36×10-6 (p-value) | GTEx Portal V6 | 1 |
| HLA-C | ? | ENSG00000204525 | liver | 1.46×10-7 (p-value) | GTEx Portal V6 | 81 |
| HCG27 | ? | ENSG00000206344 | sun exposed skin | 9.23×10-11 (p-value) | GTEx Portal V6 | 104 |
| PSORS1C3 | ? | ENSG00000204528 | sun exposed skin | 1.22×10-9 (p-value) | GTEx Portal V6 | 103 |
| HLA-C | ? | ENSG00000204525 | sun exposed skin | 1.05×10-7 (p-value) | GTEx Portal V6 | 96 |
| XXbac-BPG248L24.12 | ? | ENSG00000271581 | sun exposed skin | 2.06×10-8 (p-value) | GTEx Portal V6 | 23 |
| POU5F1 | ? | ENSG00000204531 | sun exposed skin | 6.68×10-11 (p-value) | GTEx Portal V6 | 103 |
| PSORS1C3 | ? | ENSG00000204528 | unexposed skin | 3.01×10-7 (p-value) | GTEx Portal V6 | 88 |
| HLA-C | ? | ENSG00000204525 | unexposed skin | 7.04×10-7 (p-value) | GTEx Portal V6 | 94 |
| POU5F1 | ? | ENSG00000204531 | unexposed skin | 5.17×10-8 (p-value) | GTEx Portal V6 | 102 |
| HCG27 | ? | ENSG00000206344 | unexposed skin | 1.41×10-6 (p-value) | GTEx Portal V6 | 9 |
| PSORS1C3 | ? | ENSG00000204528 | left ventricle | 1.05×10-6 (p-value) | GTEx Portal V6 | 62 |
| HCG27 | ? | ENSG00000206344 | aorta | 6.79×10-7 (p-value) | GTEx Portal V6 | 87 |
| HLA-C | ? | ENSG00000204525 | aorta | 7.00×10-12 (p-value) | GTEx Portal V6 | 104 |
| HCG27 | ? | ENSG00000206344 | subcutaneous adipocytes | 6.73×10-17 (p-value) | GTEx Portal V6 | 104 |
| HLA-C | ? | ENSG00000204525 | subcutaneous adipocytes | 4.19×10-10 (p-value) | GTEx Portal V6 | 104 |
| XXbac-BPG248L24.12 | ? | ENSG00000271581 | subcutaneous adipocytes | 3.75×10-8 (p-value) | GTEx Portal V6 | 11 |
| HCG27 | ? | ENSG00000206344 | stomach | 5.12×10-8 (p-value) | GTEx Portal V6 | 104 |
| PSORS1C3 | ? | ENSG00000204528 | stomach | 3.67×10-7 (p-value) | GTEx Portal V6 | 93 |
| HCG27 | ? | ENSG00000206344 | visceral adipocytes | 1.20×10-7 (p-value) | GTEx Portal V6 | 101 |
| HLA-C | ? | ENSG00000204525 | visceral adipocytes | 3.81×10-7 (p-value) | GTEx Portal V6 | 83 |
| XXbac-BPG248L24.12 | ? | ENSG00000271581 | visceral adipocytes | 3.09×10-7 (p-value) | GTEx Portal V6 | 84 |
| PSORS1C3 | ? | ENSG00000204528 | spleen | 7.06×10-7 (p-value) | GTEx Portal V6 | 86 |
| HCG27 | ? | ENSG00000206344 | nucleus accumbens | 9.32×10-8 (p-value) | GTEx Portal V6 | 88 |
| HCG27 | ? | ENSG00000206344 | sigmoid colon | 1.10×10-7 (p-value) | GTEx Portal V6 | 86 |
| HLA-C | ? | ENSG00000204525 | sigmoid colon | 3.69×10-7 (p-value) | GTEx Portal V6 | 70 |
| HCG27 | ? | ENSG00000206344 | tibial nerve | 3.74×10-18 (p-value) | GTEx Portal V6 | 104 |
| HLA-C | ? | ENSG00000204525 | tibial nerve | 1.16×10-10 (p-value) | GTEx Portal V6 | 104 |
| XXbac-BPG248L24.12 | ? | ENSG00000271581 | tibial nerve | 8.13×10-6 (p-value) | GTEx Portal V6 | 1 |
| HLA-B | ? | ENSG00000234745 | tibial nerve | 6.97×10-6 (p-value) | GTEx Portal V6 | 9 |

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|---|---|---|---|---|---|---|
| HLA-B | ? | ENSG00000234745 | tibial nerve | 6.97×10-6 (p-value) | GTEx Portal V6 | 9 |
| PSORS1C3 | ? | ENSG00000204528 | esophagus mucosa | 8.85×10-10 (p-value) | GTEx Portal V6 | 103 |
| PSORS1C2 | ? | ENSG00000204538 | esophagus mucosa | 3.96×10-8 (p-value) | GTEx Portal V6 | 104 |
| HLA-C | ? | ENSG00000204525 | esophagus mucosa | 2.95×10-11 (p-value) | GTEx Portal V6 | 104 |
| TCF19 | ? | ENSG00000137310 | esophagus mucosa | 5.99×10-11 (p-value) | GTEx Portal V6 | 104 |
| CCHCR1 | ? | ENSG00000204536 | esophagus mucosa | 7.87×10-7 (p-value) | GTEx Portal V6 | 93 |
| POU5F1 | ? | ENSG00000204531 | esophagus mucosa | 5.23×10-12 (p-value) | GTEx Portal V6 | 103 |
| XXbac-BPG248L24.12 | ? | ENSG00000271581 | esophagus mucosa | 1.20×10-6 (p-value) | GTEx Portal V6 | 3 |
| ? | ? | ILMN_1883997 | blood | 4.92×10-6 (p-value) | Westra et al. | 1 |
| MICB | ENST00000538442 | ILMN_1708006 | blood | 9.19×10-7 (p-value) | Westra et al. | 13 |
| MICB | ENST00000399150 | | | | | |
| MICB | ENST00000252229 | | | | | |
| HCG27 | ? | ENSG00000206344 | adrenal gland | 2.17×10-7 (p-value) | GTEx Portal V6 | 38 |
| HCG27 | ? | ENSG00000206344 | testis | 2.07×10-7 (p-value) | GTEx Portal V6 | 87 |
| HLA-C | ? | ENSG00000204525 | testis | 3.85×10-8 (p-value) | GTEx Portal V6 | 96 |
| PSORS1C1 | ? | ENSG00000204540 | testis | 3.06×10-6 (p-value) | GTEx Portal V6 | 4 |
| HCG27 | ? | ENSG00000206344 | pancreas | 2.72×10-6 (p-value) | GTEx Portal V6 | 1 |
| HLA-C | ? | ENSG00000204525 | EBV lymphocytes | 2.10×10-6 (p-value) | GTEx Portal V6 | 1 |
| CCHCR1 | ? | ENSG00000204536 | blood | 1.78×10-2 (q-value) | SeeQTL DB (HapMap) | 8 |
| HLA-C | ENST00000470363 | ILMN_2150787 | monocyte | 2.06×10-20 (p-value) | Zeller et al. | 4 |
| HLA-C | ENST00000383329 | | | | | |
| HLA-C | ENST00000487245 | | | | | |
| HLA-C | ENST00000376228 | | | | | |
| HLA-C | ENST00000376237 | | | | | |
| HLA-C | ENST00000620806 | | | | | |
| HLA-C | ENST00000466892 | | | | | |
| ? | ? | ILMN_1721113 | monocyte | 3.03×10-30 (p-value) | Zeller et al. | 4 |
| TCF19 | ENST00000496421 | 223274_at | blood | 3.20×10-8 (p-value) | Dixon et al. | 2 |
| TCF19 | ENST00000376257 | | | | | |
| TCF19 | ENST00000376255 | | | | | |
| HLA-C | ? | ENSG00000204525 | gastroesophageal junction | 3.29×10-6 (p-value) | GTEx Portal V6 | 2 |
| HCG27 | ? | ENSG00000206344 | caudate basal ganglia | 1.53×10-6 (p-value) | GTEx Portal V6 | 4 |
| PSORS1C3 | ENST00000412143 | ILMN_3309634 | b-cell | 2.61×10-8 (p-value) | Fairfax et al. | 6 |
| MICA | ENST00000449934 | ILMN_3241692 | monocyte | 9.22×10-5 (p-value) | Fairfax et al. | 5 |
| MICA | ENST00000616296 | | | | | |
| MICA | ENST00000421350 | | | | | |
| HCG27 | ? | ENSG00000206344 | breast | 2.13×10-6 (p-value) | GTEx Portal V6 | 52 |
| XXbac-BPG299F13.17 | ? | ENSG00000272501 | frontal cortex | 7.70×10-7 (p-value) | GTEx Portal V6 | 1 |

## *trans*-eQTL

| gene | transcript | probe | chromosome | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|---|
| PLOD3 | ENST00000223127 | ILMN_1774836 | chr7 | blood | 1.47×10-18 (p-value) | Westra et al. | 1 |

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|---|---|---|---|---|---|---|---|
| PLOD3 | ENST00000454310 | | | | | | |
| TMEM154 | ENST00000304385 | ILMN_2088124 | chr4 | blood | $2.87\times10^{-25}$ (p-value) | Westra et al. | 1 |
| CD248 | ENST00000311330 | ILMN_1726589 | chr11 | blood | $1.06\times10^{-8}$ (p-value) | Westra et al. | 1 |
| GPBAR1 | ENST00000519574 | ILMN_2316386 | chr2 | blood | $2.65\times10^{-8}$ (p-value) | Westra et al. | 1 |
| GPBAR1 | ENST00000479077 | | | | | | |
| GPBAR1 | ENST00000521462 | | | | | | |
| GPBAR1 | ENST00000522678 | | | | | | |
| TRAPPC1 | ENST00000303731 | ILMN_1716913 | chr17 | blood | $3.28\times10^{-7}$ (p-value) | Westra et al. | 1 |
| TRAPPC1 | ENST00000575639 | | | | | | |
| TRAPPC1 | ENST00000571739 | | | | | | |
| TRAPPC1 | ENST00000571947 | | | | | | |
| TRAPPC1 | ENST00000572656 | | | | | | |
| TRAPPC1 | ENST00000573989 | | | | | | |
| TRAPPC1 | ENST00000540486 | | | | | | |

## Putative effect on regulation

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

| SNiPA enhancer id | variant(s) | associated SNiPA promoter id | associated gene(s) |
|---|---|---|---|
| ENCE00000444768 | 1 | ENCP00000047982 | CCHCR1<br>CCHCR1<br>CCHCR1<br>CCHCR1<br>CCHCR1<br>CCHCR1 |
| | | ENCP00000048000 | WASF5P<br>WASF5P<br>WASF5P<br>WASF5P<br>WASF5P<br>WASF5P<br>WASF5P |
| | | ENCP00000047981 | CCHCR1<br>CCHCR1<br>CCHCR1<br>CCHCR1<br>CCHCR1<br>CCHCR1 |
| ENCE00000444720 | 2 | ENCP00000047970 | SFTA2<br>SFTA2<br>SFTA2<br>SFTA2<br>SFTA2<br>SFTA2 |
| ENCE00000444721 | 6 | ENCP00000047970 | SFTA2<br>SFTA2<br>SFTA2<br>SFTA2<br>SFTA2<br>SFTA2 |
| ENCE00000445080 | 6 | ENCP00000048049 | PRRC2A<br>PRRC2A<br>PRRC2A<br>PRRC2A<br>PRRC2A<br>PRRC2A<br>PRRC2A |
| | | ENCP00000048087 | DDAH2<br>DDAH2<br>DDAH2<br>DDAH2 |

| | | | | | | |
|---|---|---|---|---|---|---|
| | | | | DDAH2<br>DDAH2<br>DDAH2 | | |

### Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| RPL3P2 | downstream gene variant | 1505 | ENST00000413027 | ? | ? | 17 |
| USP8P1 | downstream gene variant | 4270 | ENST00000494673 | ? | ? | 9 |
| WASF5P | downstream gene variant, upstream gene variant | 206 | ENST00000428639 | ? | ? | 24 |
| XXbac-BPG248L24.10 | downstream gene variant | 3876 | ENST00000421191 | ? | ? | 3 |
| XXbac-BPG248L24.13 | downstream gene variant, upstream gene variant | 17 | ENST00000539514 | ? | ? | 16 |

## Putative effect on transcript

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| HLA-C | ENST00000620806 | ? | ENSP00000480331 | 127 |
| XXbac-BPG248L24.13 | ENST00000539514 | ? | ? | 90 |

### Non-coding exon variant

| gene | affected transcript | RefSeq id | variant(s) |
|---|---|---|---|
| WASF5P | ENST00000428639 | ? | 1 |
| XXbac-BPG248L24.13 | ENST00000539514 | ? | 5 |