# SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| **genomic range** | chr6:32,218,120-32,776,921 |
| **block size** | 558,802 bp |
| **variant count** | 30 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| **phyloP** | μ = -0.450 [-3.618 – 0.444] | **gene(s) hit or close-by** | BTNL2, C6orf10, HLA-DOB, HLA-DRA, HLA-DRB9, HNRNPA1P2, RNU6-603P, XXbac-BPG154L12.4, XXbac-BPG154L12.5, XXbac-BPG246D15.9 |
| **phastCons** | μ = 0.127 [0 – 0.923] | **eQTL gene(s)** | C4A, C4B, CYP21A1P, CYP21A2, HCP5, HLA-C, HLA-DMA, HLA-DMB, HLA-DPB1, HLA-DQA1, HLA-DQB1, HLA-DQB1-AS1, HLA-DQB2, HLA-DRB5, HLA-DRB6, LY6G6C, MICB, NOTCH4, PSMB9, RNF5, STK19, STK19B, TAP2, TNXA, TNXB, VARS2, WASF5P, XXbac-BPG246D15.9, XXbac-BPG248L24.13 |
| **GERP++** | μ = -0.271 [-5.03 – 1.82] | **potentially regulated gene(s)** | BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, C2, C2, C2, C2, C2, C2, C2, C2, C2, C2, EGFL8, EGFL8, EGFL8, EGFL8, EGFL8, EGFL8, EGFL8, EGFL8, HCG23, HCG23, HCG23, HCG23, HCG23, HCG23, HCG23, HCG23, HLA-DRB6, XXbac-BPG254F23.6, XXbac-BPG254F23.6, XXbac-BPG254F23.6, XXbac-BPG254F23.6, XXbac-BPG254F23.6, XXbac-BPG254F23.6 |
| **CADD score** | μ = 2.492 [0.026 – 15.24] | **disease gene(s)** | C4B, C4A, CYP21A2, VARS2, , , , , , , , , , C2, , , , , , , BTNL2, HLA-DRA, TAP2, TNXB, HLA-DPB1, HLA-DQB1 |

## Trait annotations

### Variant association

| trait | min(p-value) | source DB | source entry/link | | variant(s) |
|---|---|---|---|---|---|
| Lupus erythematosus, systemic | $2.21\times10^{-7}$ | dbGaP | pha002867 | dbGaP | 1 |
| Diabetes mellitus, type 1 | $2.48\times10^{-103}$ | dbGaP | pha002862 | dbGaP | 1 |
| Lupus erythematosus, systemic | $4.01\times10^{-18}$ | dbGaP | pha002848 | dbGaP | 1 |

### Disease gene annotation

| gene | trait | source DB | source entry/link | |
|---|---|---|---|---|
| C4B | COMPLEMENT COMPONENT 4B DEFICIENCY | OMIM | MIM:614379 | OMIM |
| C4B | BLOOD GROUP, CHIDO/RODGERS SYSTEM | OMIM | MIM:614374 | OMIM |
| C4B | SYSTEMIC LUPUS ERYTHEMATOSUS | OMIM | MIM:152700 | OMIM |
| C4A | COMPLEMENT COMPONENT 4A DEFICIENCY | OMIM | MIM:614380 | OMIM |
| C4A | BLOOD GROUP, CHIDO/RODGERS SYSTEM | OMIM | MIM:614374 | OMIM |
| C4A | SYSTEMIC LUPUS ERYTHEMATOSUS | OMIM | MIM:152700 | OMIM |
| C4A | COMPLEMENT COMPONENT 4, PARTIAL DEFICIENCY OF | OMIM | MIM:120790 | OMIM |
| CYP21A2 | ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE [...] | OMIM | MIM:201910 | OMIM |
| VARS2 | COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20 | OMIM | MIM:615917 | OMIM |
| | SARCOIDOSIS, SUSCEPTIBILITY TO, 2 | OMIM | MIM:612387 | OMIM |
| | COMPLEMENT COMPONENT 2 DEFICIENCY | OMIM | MIM:217000 | OMIM |
| | MACULAR DEGENERATION, AGE-RELATED, 1 | OMIM | MIM:603075 | OMIM |
| | SARCOIDOSIS, SUSCEPTIBILITY TO, 2 | OMIM | MIM:612387 | OMIM |
| | SARCOIDOSIS, SUSCEPTIBILITY TO, 2 | OMIM | MIM:612387 | 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 |

| | | | | |
|---|---|---|---|---|
| | COMPLEMENT COMPONENT 2 DEFICIENCY | OMIM | MIM:217000 | OMIM |
| | MACULAR DEGENERATION, AGE-RELATED, 1 | OMIM | MIM:603075 | OMIM |
| | SARCOIDOSIS, SUSCEPTIBILITY TO, 2 | OMIM | MIM:612387 | OMIM |
| C2 | COMPLEMENT COMPONENT 2 DEFICIENCY | OMIM | MIM:217000 | OMIM |
| C2 | MACULAR DEGENERATION, AGE-RELATED, 1 | OMIM | MIM:603075 | OMIM |
| | SARCOIDOSIS, SUSCEPTIBILITY TO, 2 | OMIM | MIM:612387 | OMIM |
| | COMPLEMENT COMPONENT 2 DEFICIENCY | OMIM | MIM:217000 | OMIM |
| | MACULAR DEGENERATION, AGE-RELATED, 1 | OMIM | MIM:603075 | OMIM |
| | SARCOIDOSIS, SUSCEPTIBILITY TO, 2 | OMIM | MIM:612387 | OMIM |
| | SARCOIDOSIS, SUSCEPTIBILITY TO, 2 | OMIM | MIM:612387 | OMIM |
| | COMPLEMENT COMPONENT 2 DEFICIENCY | OMIM | MIM:217000 | OMIM |
| | MACULAR DEGENERATION, AGE-RELATED, 1 | OMIM | MIM:603075 | OMIM |
| BTNL2 | SARCOIDOSIS, SUSCEPTIBILITY TO, 2 | OMIM | MIM:612387 | OMIM |
| HLA-DRA | HEPATITIS B VIRUS, SUSCEPTIBILITY TO | OMIM | MIM:610424 | OMIM |
| TAP2 | BARE LYMPHOCYTE SYNDROME, TYPE I | OMIM | MIM:604571 | OMIM |
| C4B | Immunodeficiency due to an early component of complement deficiency | OrphaNet | OrphaNet:169147 | orphanet |
| TNXB | Ehlers-Danlos syndrome due to tenascin-X deficiency | OrphaNet | OrphaNet:230839 | orphanet |
| TNXB | EHLERS-DANLOS SYNDROME, HYPERMOBILITY TYPE | OrphaNet | OrphaNet:285 | orphanet |
| C4A | Immunodeficiency due to an early component of complement deficiency | OrphaNet | OrphaNet:169147 | orphanet |
| C4A | Behcet disease | OrphaNet | OrphaNet:117 | orphanet |
| CYP21A2 | Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form | OrphaNet | OrphaNet:315306 | orphanet |
| CYP21A2 | Nonclassic congenital adrenal hyperplasia due to 21-hydroxylase deficiency | OrphaNet | OrphaNet:95698 | orphanet |
| CYP21A2 | Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, simple virilizing form | OrphaNet | OrphaNet:315311 | orphanet |
| HLA-DPB1 | Chronic berylliosis | OrphaNet | OrphaNet:133 | orphanet |
| HLA-DPB1 | Granulomatosis with polyangiitis | OrphaNet | OrphaNet:900 | orphanet |
| C2 | Immunodeficiency due to an early component of complement deficiency | OrphaNet | OrphaNet:169147 | orphanet |
| BTNL2 | Sarcoidosis | OrphaNet | OrphaNet:797 | orphanet |
| HLA-DQB1 | Bullous pemphigoid | OrphaNet | OrphaNet:703 | orphanet |
| HLA-DQB1 | Narcolepsy-cataplexy | OrphaNet | OrphaNet:2073 | orphanet |
| HLA-DQB1 | Narcolepsy without cataplexy | OrphaNet | OrphaNet:83465 | orphanet |
| HLA-DRA | Graham Little-Piccardi-Lassueur syndrome | OrphaNet | OrphaNet:505 | orphanet |
| TAP2 | Immunodeficiency by defective expression of HLA class 1 | OrphaNet | OrphaNet:34592 | orphanet |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| C4A | ? | ENSG00000244731 | pancreas | $5.71\times10^{-13}$ (p-value) | GTEx Portal V6 | 24 |
| RNF5 | ? | ENSG00000204308 | pancreas | $1.41\times10^{-6}$ (p-value) | GTEx Portal V6 | 22 |
| XXbac-BPG248L24.13 | ? | ENSG00000256166 | pancreas | $3.28\times10^{-7}$ (p-value) | GTEx Portal V6 | 5 |
| HLA-DMA | ? | ENSG00000204257 | pancreas | $2.02\times10^{-11}$ (p-value) | GTEx Portal V6 | 24 |
| WASF5P | ? | ENSG00000231402 | pancreas | $4.68\times10^{-7}$ (p-value) | GTEx Portal V6 | 5 |
| CYP21A1P | ? | ENSG00000204338 | pancreas | $2.84\times10^{-7}$ (p-value) | GTEx Portal V6 | 14 |
| HLA-DQB2 | ? | ENSG00000232629 | muscularis mucosae | $2.71\times10^{-7}$ (p-value) | GTEx Portal V6 | 19 |
| C4A | ? | ENSG00000244731 | muscularis mucosae | $2.15\times10^{-19}$ (p-value) | GTEx Portal V6 | 25 |
| HLA-C | ? | ENSG00000204525 | muscularis mucosae | $3.72\times10^{-7}$ (p-value) | GTEx Portal V6 | 4 |
| CYP21A1P | ? | ENSG00000204338 | muscularis mucosae | $2.74\times10^{-12}$ (p-value) | GTEx Portal V6 | 25 |
| HLA-DQB2 | ? | ENSG00000232629 | lung | $7.65\times10^{-7}$ (p-value) | GTEx Portal V6 | 20 |

| | | | | | | |
|---|---|---|---|---|---|---|
| C4A | ? | ENSG00000244731 | lung | 4.85×10-17 (p-value) | GTEx Portal V6 | 25 |
| HLA-C | ? | ENSG00000204525 | lung | 1.72×10-8 (p-value) | GTEx Portal V6 | 4 |
| CYP21A1P | ? | ENSG00000204338 | lung | 5.42×10-10 (p-value) | GTEx Portal V6 | 25 |
| HLA-DQB2 | ? | ENSG00000232629 | atrial appendage | 6.02×10-8 (p-value) | GTEx Portal V6 | 19 |
| C4A | ? | ENSG00000244731 | atrial appendage | 6.06×10-16 (p-value) | GTEx Portal V6 | 25 |
| CYP21A1P | ? | ENSG00000204338 | atrial appendage | 1.27×10-9 (p-value) | GTEx Portal V6 | 25 |
| HLA-DMA | ? | ENSG00000204257 | transformed fibroblasts | 1.88×10-20 (p-value) | GTEx Portal V6 | 24 |
| HLA-DMB | ? | ENSG00000242574 | transformed fibroblasts | 5.14×10-13 (p-value) | GTEx Portal V6 | 21 |
| C4B | ? | ENSG00000224389 | transformed fibroblasts | 1.54×10-15 (p-value) | GTEx Portal V6 | 25 |
| C4A | ? | ENSG00000244731 | transformed fibroblasts | 2.95×10-17 (p-value) | GTEx Portal V6 | 25 |
| HLA-C | ? | ENSG00000204525 | transformed fibroblasts | 3.17×10-8 (p-value) | GTEx Portal V6 | 4 |
| MICB | ? | ENSG00000204516 | transformed fibroblasts | 9.47×10-6 (p-value) | GTEx Portal V6 | 1 |
| HLA-DMA | ? | ENSG00000204257 | tibial artery | 2.07×10-9 (p-value) | GTEx Portal V6 | 20 |
| C4A | ? | ENSG00000244731 | tibial artery | 2.30×10-13 (p-value) | GTEx Portal V6 | 25 |
| HLA-C | ? | ENSG00000204525 | tibial artery | 7.01×10-10 (p-value) | GTEx Portal V6 | 4 |
| CYP21A1P | ? | ENSG00000204338 | tibial artery | 1.84×10-12 (p-value) | GTEx Portal V6 | 25 |
| C4A | ? | ENSG00000244731 | breast | 1.15×10-14 (p-value) | GTEx Portal V6 | 25 |
| HLA-C | ? | ENSG00000204525 | breast | 9.99×10-10 (p-value) | GTEx Portal V6 | 4 |
| CYP21A1P | ? | ENSG00000204338 | breast | 6.62×10-13 (p-value) | GTEx Portal V6 | 25 |
| HLA-DQB2 | ? | ENSG00000232629 | breast | 1.26×10-7 (p-value) | GTEx Portal V6 | 10 |
| STK19B | ? | ENSG00000250535 | breast | 6.19×10-7 (p-value) | GTEx Portal V6 | 1 |
| C4B | ? | ENSG00000224389 | blood | 3.97×10-17 (p-value) | GTEx Portal V6 | 25 |
| C4A | ? | ENSG00000244731 | blood | 2.06×10-16 (p-value) | GTEx Portal V6 | 25 |
| HLA-C | ? | ENSG00000204525 | blood | 3.69×10-8 (p-value) | GTEx Portal V6 | 4 |
| CYP21A2 | ? | ENSG00000231852 | blood | 3.10×10-7 (p-value) | GTEx Portal V6 | 23 |
| CYP21A1P | ? | ENSG00000204338 | blood | 8.43×10-16 (p-value) | GTEx Portal V6 | 25 |
| HLA-DMA | ? | ENSG00000204257 | thyroid | 1.77×10-18 (p-value) | GTEx Portal V6 | 25 |
| C4A | ? | ENSG00000244731 | thyroid | 4.56×10-21 (p-value) | GTEx Portal V6 | 25 |
| HLA-C | ? | ENSG00000204525 | thyroid | 1.36×10-6 (p-value) | GTEx Portal V6 | 4 |
| CYP21A1P | ? | ENSG00000204338 | thyroid | 2.70×10-18 (p-value) | GTEx Portal V6 | 25 |
| HLA-DQB2 | ? | ENSG00000232629 | thyroid | 9.43×10-7 (p-value) | GTEx Portal V6 | 12 |
| HLA-DRB6 | ? | ENSG00000229391 | thyroid | 6.75×10-6 (p-value) | GTEx Portal V6 | 3 |
| HLA-DQA1 | ? | ENSG00000196735 | skeletal muscle | 6.40×10-8 (p-value) | GTEx Portal V6 | 24 |
| C4A | ? | ENSG00000244731 | skeletal muscle | 5.08×10-20 (p-value) | GTEx Portal V6 | 25 |
| HLA-DRB5 | ? | ENSG00000198502 | skeletal muscle | 6.10×10-7 (p-value) | GTEx Portal V6 | 20 |
| HLA-C | ? | ENSG00000204525 | skeletal muscle | 1.19×10-13 (p-value) | GTEx Portal V6 | 4 |
| RNF5 | ? | ENSG00000204308 | skeletal muscle | 4.88×10-10 (p-value) | GTEx Portal V6 | 24 |

| | | | | | | |
|---|---|---|---|---|---|---|
| HLA-DRB6 | ? | ENSG00000229391 | skeletal muscle | $3.08\times10^{-8}$ (p-value) | GTEx Portal V6 | 22 |
| CYP21A1P | ? | ENSG00000204338 | skeletal muscle | $6.94\times10^{-7}$ (p-value) | GTEx Portal V6 | 20 |
| HLA-DQB2 | ? | ENSG00000232629 | skeletal muscle | $1.40\times10^{-6}$ (p-value) | GTEx Portal V6 | 9 |
| HLA-DQB1-AS1 | ? | ENSG00000223534 | skeletal muscle | $2.02\times10^{-6}$ (p-value) | GTEx Portal V6 | 7 |
| HLA-DMA | ? | ENSG00000204257 | skeletal muscle | $5.10\times10^{-10}$ (p-value) | GTEx Portal V6 | 9 |
| C4A | ? | ENSG00000244731 | transverse colon | $1.71\times10^{-12}$ (p-value) | GTEx Portal V6 | 25 |
| HLA-C | ? | ENSG00000204525 | transverse colon | $6.93\times10^{-7}$ (p-value) | GTEx Portal V6 | 4 |
| CYP21A1P | ? | ENSG00000204338 | transverse colon | $1.98\times10^{-6}$ (p-value) | GTEx Portal V6 | 1 |
| C4A | ? | ENSG00000244731 | EBV lymphocytes | $9.01\times10^{-10}$ (p-value) | GTEx Portal V6 | 21 |
| HLA-C | ? | ENSG00000204525 | EBV lymphocytes | $3.18\times10^{-6}$ (p-value) | GTEx Portal V6 | 4 |
| C4A | ? | ENSG00000244731 | unexposed skin | $4.36\times10^{-16}$ (p-value) | GTEx Portal V6 | 25 |
| CYP21A2 | ? | ENSG00000231852 | unexposed skin | $9.86\times10^{-7}$ (p-value) | GTEx Portal V6 | 18 |
| CYP21A1P | ? | ENSG00000204338 | unexposed skin | $5.73\times10^{-10}$ (p-value) | GTEx Portal V6 | 24 |
| MICB | ? | ENSG00000204516 | unexposed skin | $2.73\times10^{-6}$ (p-value) | GTEx Portal V6 | 16 |
| HLA-DMA | ? | ENSG00000204257 | unexposed skin | $1.94\times10^{-6}$ (p-value) | GTEx Portal V6 | 1 |
| C4A | ? | ENSG00000244731 | sun exposed skin | $4.91\times10^{-17}$ (p-value) | GTEx Portal V6 | 25 |
| HLA-C | ? | ENSG00000204525 | sun exposed skin | $3.99\times10^{-8}$ (p-value) | GTEx Portal V6 | 4 |
| NOTCH4 | ? | ENSG00000204301 | sun exposed skin | $2.52\times10^{-9}$ (p-value) | GTEx Portal V6 | 21 |
| HLA-DMA | ? | ENSG00000204257 | sun exposed skin | $1.82\times10^{-15}$ (p-value) | GTEx Portal V6 | 25 |
| C4B | ? | ENSG00000224389 | sun exposed skin | $8.56\times10^{-14}$ (p-value) | GTEx Portal V6 | 24 |
| CYP21A2 | ? | ENSG00000231852 | sun exposed skin | $1.11\times10^{-10}$ (p-value) | GTEx Portal V6 | 25 |
| MICB | ? | ENSG00000204516 | sun exposed skin | $5.07\times10^{-8}$ (p-value) | GTEx Portal V6 | 21 |
| CYP21A1P | ? | ENSG00000204338 | sun exposed skin | $2.27\times10^{-13}$ (p-value) | GTEx Portal V6 | 25 |
| HLA-DMA | ? | ENSG00000204257 | liver | $4.11\times10^{-13}$ (p-value) | GTEx Portal V6 | 25 |
| STK19B | ? | ENSG00000250535 | liver | $4.66\times10^{-8}$ (p-value) | GTEx Portal V6 | 21 |
| C4A | ? | ENSG00000244731 | liver | $3.14\times10^{-14}$ (p-value) | GTEx Portal V6 | 25 |
| HLA-DQB2 | ? | ENSG00000232629 | left ventricle | $7.06\times10^{-8}$ (p-value) | GTEx Portal V6 | 21 |
| C4A | ? | ENSG00000244731 | left ventricle | $6.97\times10^{-17}$ (p-value) | GTEx Portal V6 | 25 |
| CYP21A1P | ? | ENSG00000204338 | left ventricle | $1.10\times10^{-9}$ (p-value) | GTEx Portal V6 | 25 |
| NOTCH4 | ? | ENSG00000204301 | left ventricle | $7.25\times10^{-8}$ (p-value) | GTEx Portal V6 | 24 |
| C4A | ? | ENSG00000244731 | aorta | $7.08\times10^{-16}$ (p-value) | GTEx Portal V6 | 25 |
| HLA-C | ? | ENSG00000204525 | aorta | $2.49\times10^{-7}$ (p-value) | GTEx Portal V6 | 4 |
| CYP21A2 | ? | ENSG00000231852 | aorta | $1.41\times10^{-6}$ (p-value) | GTEx Portal V6 | 12 |
| CYP21A1P | ? | ENSG00000204338 | aorta | $1.34\times10^{-8}$ (p-value) | GTEx Portal V6 | 24 |
| MICB | ? | ENSG00000204516 | aorta | $1.44\times10^{-8}$ (p-value) | GTEx Portal V6 | 22 |
| NOTCH4 | ? | ENSG00000204301 | aorta | $3.98\times10^{-6}$ (p-value) | GTEx Portal V6 | 1 |
| HLA-DQB2 | ? | ENSG00000232629 | subcutaneous adipocytes | $5.08\times10^{-11}$ (p-value) | GTEx Portal V6 | 24 |

| | | | | | | |
|---|---|---|---|---|---|---|
| C4A | ? | ENSG00000244731 | subcutaneous adipocytes | $1.31\times10^{-17}$ (p-value) | GTEx Portal V6 | 25 |
| HLA-DRB5 | ? | ENSG00000198502 | subcutaneous adipocytes | $3.29\times10^{-7}$ (p-value) | GTEx Portal V6 | 23 |
| HLA-C | ? | ENSG00000204525 | subcutaneous adipocytes | $1.50\times10^{-8}$ (p-value) | GTEx Portal V6 | 4 |
| NOTCH4 | ? | ENSG00000204301 | subcutaneous adipocytes | $3.45\times10^{-7}$ (p-value) | GTEx Portal V6 | 21 |
| C4B | ? | ENSG00000224389 | subcutaneous adipocytes | $2.32\times10^{-6}$ (p-value) | GTEx Portal V6 | 10 |
| CYP21A2 | ? | ENSG00000231852 | subcutaneous adipocytes | $4.22\times10^{-8}$ (p-value) | GTEx Portal V6 | 25 |
| CYP21A1P | ? | ENSG00000204338 | subcutaneous adipocytes | $1.25\times10^{-14}$ (p-value) | GTEx Portal V6 | 25 |
| HLA-DQB2 | ? | ENSG00000232629 | visceral adipocytes | $4.43\times10^{-10}$ (p-value) | GTEx Portal V6 | 24 |
| C4A | ? | ENSG00000244731 | visceral adipocytes | $5.89\times10^{-11}$ (p-value) | GTEx Portal V6 | 24 |
| HLA-C | ? | ENSG00000204525 | visceral adipocytes | $5.84\times10^{-7}$ (p-value) | GTEx Portal V6 | 4 |
| CYP21A1P | ? | ENSG00000204338 | visceral adipocytes | $2.87\times10^{-9}$ (p-value) | GTEx Portal V6 | 24 |
| NOTCH4 | ? | ENSG00000204301 | visceral adipocytes | $3.60\times10^{-9}$ (p-value) | GTEx Portal V6 | 22 |
| C4A | ? | ENSG00000244731 | stomach | $3.95\times10^{-16}$ (p-value) | GTEx Portal V6 | 25 |
| C4A | ? | ENSG00000244731 | spleen | $3.84\times10^{-8}$ (p-value) | GTEx Portal V6 | 15 |
| C4A | ? | ENSG00000244731 | tibial nerve | $6.98\times10^{-23}$ (p-value) | GTEx Portal V6 | 25 |
| HLA-C | ? | ENSG00000204525 | tibial nerve | $6.43\times10^{-8}$ (p-value) | GTEx Portal V6 | 4 |
| CYP21A1P | ? | ENSG00000204338 | tibial nerve | $1.34\times10^{-13}$ (p-value) | GTEx Portal V6 | 25 |
| C4A | ? | ENSG00000244731 | sigmoid colon | $1.97\times10^{-9}$ (p-value) | GTEx Portal V6 | 24 |
| CYP21A1P | ? | ENSG00000204338 | sigmoid colon | $6.24\times10^{-7}$ (p-value) | GTEx Portal V6 | 1 |
| LY6G6C | ? | ENSG00000204421 | esophagus mucosa | $4.94\times10^{-9}$ (p-value) | GTEx Portal V6 | 23 |
| C4A | ? | ENSG00000244731 | esophagus mucosa | $4.61\times10^{-16}$ (p-value) | GTEx Portal V6 | 25 |
| HLA-C | ? | ENSG00000204525 | esophagus mucosa | $2.15\times10^{-9}$ (p-value) | GTEx Portal V6 | 4 |
| CYP21A1P | ? | ENSG00000204338 | esophagus mucosa | $1.69\times10^{-10}$ (p-value) | GTEx Portal V6 | 24 |
| MICB | ? | ENSG00000204516 | esophagus mucosa | $9.26\times10^{-9}$ (p-value) | GTEx Portal V6 | 23 |
| HLA-DQB1 | ? | ENSG00000179344 | esophagus mucosa | $1.49\times10^{-6}$ (p-value) | GTEx Portal V6 | 6 |
| HLA-DMA | ? | ENSG00000204257 | esophagus mucosa | $2.46\times10^{-6}$ (p-value) | GTEx Portal V6 | 1 |
| C4A | ? | ENSG00000244731 | gastroesophageal junction | $1.02\times10^{-12}$ (p-value) | GTEx Portal V6 | 24 |
| CYP21A1P | ? | ENSG00000204338 | gastroesophageal junction | $1.06\times10^{-8}$ (p-value) | GTEx Portal V6 | 24 |
| HLA-DQB2 | ? | ENSG00000232629 | gastroesophageal junction | $8.86\times10^{-7}$ (p-value) | GTEx Portal V6 | 5 |
| C4A | ? | ENSG00000244731 | cerebellar hemisphere | $5.31\times10^{-8}$ (p-value) | GTEx Portal V6 | 22 |
| TNXB | ? | ENSG00000168477 | adrenal gland | $2.46\times10^{-7}$ (p-value) | GTEx Portal V6 | 15 |
| C4A | ? | ENSG00000244731 | adrenal gland | $5.08\times10^{-8}$ (p-value) | GTEx Portal V6 | 19 |
| STK19 | ? | ENSG00000204344 | adrenal gland | $2.32\times10^{-7}$ (p-value) | GTEx Portal V6 | 20 |
| STK19B | ? | ENSG00000250535 | adrenal gland | $7.79\times10^{-12}$ (p-value) | GTEx Portal V6 | 25 |
| TNXA | ? | ENSG00000248290 | adrenal gland | $5.38\times10^{-10}$ (p-value) | GTEx Portal V6 | 21 |
| CYP21A1P | ? | ENSG00000204338 | adrenal gland | $2.89\times10^{-8}$ (p-value) | GTEx Portal V6 | 22 |
| C4A | ? | ENSG00000244731 | testis | $3.99\times10^{-17}$ (p-value) | GTEx Portal V6 | 25 |

| | | | | | | |
|---|---|---|---|---|---|---|
| CYP21A1P | ? | ENSG00000204338 | testis | 1.92×10-12 (p-value) | GTEx Portal V6 | 25 |
| HLA-DMA | ? | ENSG00000204257 | cerebellum | 2.83×10-9 (p-value) | GTEx Portal V6 | 20 |
| C4A | ? | ENSG00000244731 | cerebellum | 4.80×10-8 (p-value) | GTEx Portal V6 | 20 |
| CYP21A1P | ? | ENSG00000204338 | cerebellum | 8.19×10-8 (p-value) | GTEx Portal V6 | 20 |
| C4A | ? | ENSG00000244731 | frontal cortex | 1.94×10-8 (p-value) | GTEx Portal V6 | 19 |
| NOTCH4 | ENST00000474612 | ILMN_1711157 | adipocyte | 1.20×10-9 (p-value) | MuTHER consortium | 7 |
| NOTCH4 | ENST00000375023 | | | | | |
| NOTCH4 | ENST00000491215 | | | | | |
| HCP5 | ENST00000414046 | ILMN_1803945 | skin | 5.66×10-6 (p-value) | MuTHER consortium | 6 |
| HCP5 | ENST00000541196 | | adipocyte | 3.41×10-6 (p-value) | MuTHER consortium | 6 |
| HLA-DPB1 | ENST00000418931 | ILMN_1749070 | skin | 4.66×10-9 (p-value) | MuTHER consortium | 7 |
| HLA-DPB1 | ENST00000416804 | | blood | 3.81×10-10 (p-value) | MuTHER consortium | 7 |
| | | | adipocyte | 1.57×10-8 (p-value) | MuTHER consortium | 7 |
| MICB | ENST00000538442 | ILMN_1708006 | skin | 3.44×10-9 (p-value) | MuTHER consortium | 6 |
| MICB | ENST00000399150 | | | | | |
| MICB | ENST00000252229 | | | | | |
| PSMB9 | ENST00000467593 | ILMN_1798233 | blood | 1.55×10-7 (p-value) | MuTHER consortium | 4 |
| PSMB9 | ENST00000464863 | | | | | |
| VARS2 | ENST00000321897 | ILMN_1737585 | monocyte | 2.87×10-8 (p-value) | Fairfax et al. | 3 |
| VARS2 | ENST00000625423 | | b-cell | 3.50×10-6 (p-value) | Fairfax et al. | 3 |
| VARS2 | ENST00000542001 | | | | | |
| VARS2 | ENST00000541562 | | | | | |
| VARS2 | ENST00000469358 | | | | | |
| VARS2 | ENST00000477288 | | | | | |
| VARS2 | ENST00000473916 | | | | | |
| VARS2 | ENST00000476162 | | | | | |
| HLA-DPB1 | ENST00000418931 | ILMN_1749070 | monocyte | 3.64×10-8 (p-value) | Fairfax et al. | 2 |
| HLA-DPB1 | ENST00000416804 | | b-cell | 2.63×10-8 (p-value) | Fairfax et al. | 2 |
| C4A | ? | ENSG00000244731 | putamen | 2.67×10-8 (p-value) | GTEx Portal V6 | 1 |
| C4A | ? | ENSG00000244731 | caudate basal ganglia | 2.05×10-8 (p-value) | GTEx Portal V6 | 1 |
| C4A | ? | ENSG00000244731 | hypothalamus | 8.80×10-8 (p-value) | GTEx Portal V6 | 2 |
| C4A | ? | ENSG00000244731 | coronary artery | 8.43×10-7 (p-value) | GTEx Portal V6 | 1 |
| C4A | ? | ENSG00000244731 | anterior cingulate cortex | 1.39×10-8 (p-value) | GTEx Portal V6 | 1 |
| TAP2 | ENST00000620123 | ILMN_1759250 | blood | 8.02×10-7 (p-value) | Westra et al. | 1 |
| TAP2 | ENST00000374899 | | | | | |
| XXbac-BPG246D15.9 | ENST00000452392 | | | | | |
| TAP2 | ENST00000464100 | | | | | |
| TAP2 | ENST00000374897 | | | | | |
| HLA-DQB1 | ENST00000399082 | 209480_at | blood | 4.90×10-12 (p-value) | Dixon et al. | 1 |
| HLA-DQB1 | ENST00000399084 | | | | | |
| HLA-DQB1 | ENST00000434651 | | | | | |
| HLA-DQB1 | ENST00000460185 | | | | | |
| HLA-DQB1 | ENST00000399079 | | | | | |
| HLA-DQB1 | ENST00000443574 | | | | | |

| | | | | | | |
|---|---|---|---|---|---|---|
| HLA-DQB1 | ENST00000374943 | | | | | |
| HLA-DQB1 | ENST00000487676 | | | | | |
| ? | ? | 221491_x_at | blood | $2.80\times10^{-17}$ (p-value) | Dixon et al. | 1 |
| HLA-DQA1 | ENST00000374949 | 213831_at | blood | $1.60\times10^{-10}$ (p-value) | Dixon et al. | 1 |
| HLA-DQA1 | ENST00000496318 | | | | | |
| HLA-DQA1 | ENST00000460633 | | | | | |
| HLA-DQA1 | ENST00000482745 | | | | | |
| HLA-DQA1 | ENST00000343139 | | | | | |

## Putative effect on regulation

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

| SNiPA enhancer id | variant(s) | associated SNiPA promoter id | associated gene(s) |
|---|---|---|---|
| ENCE00000445252 | 1 | ENCP00000048214 | XXbac-BPG254F23.6<br>XXbac-BPG254F23.6<br>XXbac-BPG254F23.6<br>XXbac-BPG254F23.6<br>XXbac-BPG254F23.6<br>XXbac-BPG254F23.6 |
| | | ENCP00000048178 | EGFL8<br>EGFL8<br>EGFL8<br>EGFL8<br>EGFL8<br>EGFL8<br>EGFL8 |
| | | ENCP00000048131 | C2<br>C2<br>C2<br>C2<br>C2<br>C2<br>C2<br>C2 |
| | | ENCP00000048199 | HCG23<br>HCG23<br>HCG23<br>HCG23<br>HCG23<br>HCG23<br>HCG23<br>HCG23 |
| | | ENCP00000048206 | HLA-DRB6 |
| | | ENCP00000048201 | BTNL2<br>BTNL2<br>BTNL2<br>BTNL2<br>BTNL2<br>BTNL2<br>BTNL2<br>BTNL2 |

### Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| BTNL2 | upstream gene variant | 2924 | ENST00000374993 | NM_019602.1 | ENSP00000364132 | 2 |
| BTNL2 | upstream gene variant | 2919 | ENST00000454136 | ? | ENSP00000390613 | 2 |
| BTNL2 | upstream gene variant | 2924 | ENST00000465865 | ? | ENSP00000420063 | 2 |
| BTNL2 | downstream gene variant, upstream gene variant | 2919 | ENST00000446536 | ? | ENSP00000388434 | 3 |
| BTNL2 | upstream gene variant | 2924 | ENST00000544175 | ? | ENSP00000443364 | 2 |

| | | | | | | |
|---|---|---|---|---|---|---|
| C6orf10 | upstream gene variant | 1187 | ENST00000533191 | NM_001286474.1 | ENSP00000431199 | 3 |
| C6orf10 | upstream gene variant | 1182 | ENST00000617061 | ? | ENSP00000482001 | 3 |
| C6orf10 | downstream gene variant, upstream gene variant | 1222 | ENST00000532023 | ? | ENSP00000432814 | 4 |
| C6orf10 | downstream gene variant, upstream gene variant | 1215 | ENST00000534588 | ? | ENSP00000432566 | 4 |
| C6orf10 | upstream gene variant | 1187 | ENST00000375015 | ? | ENSP00000364155 | 3 |
| C6orf10 | downstream gene variant, upstream gene variant | 1195 | ENST00000442822 | ? | ENSP00000411164 | 4 |
| C6orf10 | upstream gene variant | 1222 | ENST00000527965 | NM_001286475.1 | ENSP00000435103 | 3 |
| C6orf10 | upstream gene variant | 1215 | ENST00000375007 | ? | ENSP00000364146 | 3 |
| C6orf10 | upstream gene variant | 1215 | ENST00000447241 | NM_006781.4 | ENSP00000415517 | 3 |
| C6orf10 | upstream gene variant | 1182 | ENST00000612031 | ? | ENSP00000480403 | 3 |
| HLA-DOB | downstream gene variant | 3619 | ENST00000488325 | ? | ENSP00000436618 | 1 |
| HLA-DOB | downstream gene variant | 3903 | ENST00000475235 | ? | ? | 1 |
| HLA-DOB | downstream gene variant | 3619 | ENST00000438763 | NM_002120.3 | ENSP00000390020 | 1 |
| HLA-DRA | downstream gene variant | 1612 | ENST00000374982 | ? | ENSP00000364121 | 2 |
| HLA-DRA | downstream gene variant | 1612 | ENST00000395388 | NM_019111.4 | ENSP00000378786 | 2 |
| HLA-DRB9 | downstream gene variant | 2606 | ENST00000449413 | ? | ? | 2 |
| HNRNPA1P2 | upstream gene variant, downstream gene variant | 1697 | ENST00000432375 | ? | ? | 2 |
| RNU6-603P | downstream gene variant | 4802 | ENST00000411403 | ? | ? | 1 |
| XXbac-BPG154L12.4 | upstream gene variant | 3590 | ENST00000425033 | ? | ? | 1 |
| XXbac-BPG154L12.5 | upstream gene variant | 3163 | ENST00000611838 | ? | ? | 2 |
| XXbac-BPG246D15.9 | downstream gene variant | 4623 | ENST00000452392 | ? | ENSP00000391806 | 1 |

## Putative effect on transcript

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| BTNL2 | ENST00000454136 | ? | ENSP00000390613 | 1 |
| BTNL2 | ENST00000374993 | NM_019602.1 | ENSP00000364132 | 1 |
| BTNL2 | ENST00000544175 | ? | ENSP00000443364 | 1 |
| BTNL2 | ENST00000465865 | ? | ENSP00000420063 | 1 |
| C6orf10 | ENST00000534588 | ? | ENSP00000432566 | 2 |
| C6orf10 | ENST00000533191 | NM_001286474.1 | ENSP00000431199 | 8 |
| C6orf10 | ENST00000375007 | ? | ENSP00000364146 | 8 |
| C6orf10 | ENST00000527965 | NM_001286475.1 | ENSP00000435103 | 8 |
| C6orf10 | ENST00000375015 | ? | ENSP00000364155 | 8 |
| C6orf10 | ENST00000447241 | NM_006781.4 | ENSP00000415517 | 8 |
| C6orf10 | ENST00000442822 | ? | ENSP00000411164 | 8 |
| C6orf10 | ENST00000532023 | ? | ENSP00000432814 | 2 |
| C6orf10 | ENST00000617061 | ? | ENSP00000482001 | 8 |

| | | | | |
|---|---|---|---|---|
| C6orf10 | ENST00000612031 | ? | ENSP00000480403 | 8 |
| HLA-DRB9 | ENST00000449413 | ? | ? | 1 |
| XXbac-BPG154L12.4 | ENST00000425033 | ? | ? | 3 |
| XXbac-BPG154L12.5 | ENST00000611838 | ? | ? | 10 |

## Non-coding exon variant

| gene | affected transcript | RefSeq id | variant(s) |
|---|---|---|---|
| XXbac-BPG154L12.4 | ENST00000425033 | ? | 1 |