# Locus 446 (rs8051517)

SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| genomic range | chr16:68,428,089-68,428,089 |
| block size | 1 bp |
| variant count | 1 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| phyloP | -0.856 | gene(s) hit or close-by | SMPD3 |
| phastCons | 0 | eQTL gene(s) | ATP6V0D1, DUS2, TRADD |
| GERP++ | -1.04 | potentially regulated gene(s) | – |
| CADD score | 2.866 | disease gene(s) | – |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| DUS2 | ? | ENSG00000167264 | blood | $7.29\times10^{-6}$ (p-value) | GTEx Portal V6 | 1 |
| DUS2 | ENST00000432752 | ILMN_1811650 | monocyte | $3.56\times10^{-14}$ (p-value) | Zeller et al. | 1 |
| DUS2 | ENST00000565263 | | | | | |
| DUS2 | ENST00000561965 | | | | | |
| DUS2 | ENST00000565980 | | | | | |
| DUS2 | ENST00000358896 | | | | | |
| ATP6V0D1 | ENST00000568620 | ILMN_1795826 | monocyte | $6.15\times10^{-13}$ (p-value) | Zeller et al. | 1 |
| ATP6V0D1 | ENST00000563064 | | | | | |
| ATP6V0D1 | ENST00000567694 | | | | | |
| ATP6V0D1 | ENST00000602876 | | | | | |
| ATP6V0D1 | ENST00000561852 | | | | | |
| ATP6V0D1 | ENST00000290949 | | | | | |
| ATP6V0D1 | ENST00000567170 | | | | | |
| ATP6V0D1 | ENST00000564615 | | | | | |
| ATP6V0D1 | ENST00000565835 | | | | | |
| ATP6V0D1 | ENST00000426604 | | | | | |
| ATP6V0D1 | ENST00000563305 | | | | | |
| TRADD | ENST00000486556 | ILMN_1765851 | monocyte | $4.94\times10^{-13}$ (p-value) | Zeller et al. | 1 |
| TRADD | ENST00000566247 | | | | | |
| TRADD | ENST00000563348 | | | | | |
| TRADD | ENST00000345057 | | | | | |

## Putative effect on transcript

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| SMPD3 | ENST00000561749 | ? | ENSP00000457236 | 1 |
| SMPD3 | ENST00000566723 | ? | ? | 1 |
| SMPD3 | ENST00000219334 | NM_018667.3 | ENSP00000219334 | 1 |