# Locus 405 (rs6050091)

SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| genomic range | chr20:24,788,418-24,897,155 |
| block size | 108,738 bp |
| variant count | 60 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| phyloP | μ = -0.308 [-4.928 – 0.939] | gene(s) hit or close-by | – |
| phastCons | μ = 0.066 [0 – 0.743] | eQTL gene(s) | ABHD12 |
| GERP++ | μ = -0.366 [-3.16 – 0.823] | potentially regulated gene(s) | ACSS1, CST7, PYGB |
| CADD score | μ = 3.159 [0.037 – 15.94] | disease gene(s) | ABHD12 |

## Trait annotations

### Disease gene annotation

| gene | trait | source DB | source entry/link |
|---|---|---|---|
| ABHD12 | POLYNEUROPATHY, HEARING LOSS, ATAXIA, RETINITIS PIGMENTOSA, AND [...] | OMIM | MIM:612674 |
| ABHD12 | Polyneuropathy - hearing loss - ataxia - retinitis pigmentosa - cataract | OrphaNet | OrphaNet:171848 |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| ABHD12 | ENST00000465694 | ILMN_1745116 | monocyte | 6.09×10-4 (p-value) | Fairfax et al. | 1 |
| ABHD12 | ENST00000376542 | | | | | |

## Putative effect on regulation

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

| SNiPA enhancer id | variant(s) | associated SNiPA promoter id | associated gene(s) |
|---|---|---|---|
| ENCE00000291041 | 1 | ENCP00000031107 | CST7 |
| ENCE00000291312 | 1 | ENCP00000031129 | PYGB |
| ENCE00000291043 | 1 | ENCP00000031112 | ACSS1 |
| | | ENCP00000031111 | ACSS1 |
| | | ENCP00000031107 | CST7 |

### Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00000399317 (CTCF binding site) | 1 | cervix (HeLa-S3) | H3K27me3 |
| | | monocytes (Monocytes-CD14+) | H3K27me3 |
| | | HSMMtube | H3K27me3 |
| | | liver (HepG2) | H3K27me3 |
| | | lung (IMR90) | H3K27me3 |
| | | Osteobl | H3K27me3 |
| ENSR00000399318 (open chromatin region) | 1 | embryonic stem cell (H1ESC) | H3K36me3, H3K27me3, DNase1 |
| | | A549 | H3K4me3, H3K27me3 |