# Locus 127 (rs2297363)

SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| genomic range | chr6:160,476,945-160,513,793 |
| block size | 36,849 bp |
| variant count | 9 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| phyloP | μ = -0.604 [-2.574 – 0.77] | gene(s) hit or close-by | IGF2R, RP11-288H12.3 |
| phastCons | μ = 0.001 [0 – 0.006] | eQTL gene(s) | IGF2R, RP11-288H12.3 |
| GERP++ | μ = -1.707 [-6.01 – 1.86] | potentially regulated gene(s) | LPA, RP1-276N6.2 |
| CADD score | μ = 2.880 [0.501 – 6.396] | disease gene(s) | – |

## Trait annotations

### Variant association

| trait | min(p-value) | source DB | source entry/link | variant(s) |
|---|---|---|---|---|
| isobutyrylcarnitine | $5.24\times10^{-9}$ | Metabolomics GWAS Server | 24816252 | 8 |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| RP11-288H12.3 | ? | ENSG00000213073 | ovary | $2.23\times10^{-6}$ (p-value) | GTEx Portal V6 | 1 |
| RP11-288H12.3 | ? | ENSG00000213073 | blood | $2.66\times10^{-14}$ (p-value) | GTEx Portal V6 | 9 |
| RP11-288H12.3 | ? | ENSG00000213073 | transformed fibroblasts | $4.66\times10^{-8}$ (p-value) | GTEx Portal V6 | 8 |
| IGF2R | ? | ENSG00000197081 | aorta | $4.15\times10^{-6}$ (p-value) | GTEx Portal V6 | 8 |
| RP11-288H12.3 | ? | ENSG00000213073 | thyroid | $1.78\times10^{-6}$ (p-value) | GTEx Portal V6 | 8 |
| RP11-288H12.3 | ? | ENSG00000213073 | EBV lymphocytes | $6.38\times10^{-7}$ (p-value) | GTEx Portal V6 | 6 |

## Putative effect on regulation

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

| SNiPA enhancer id | variant(s) | associated SNiPA promoter id | associated gene(s) |
|---|---|---|---|
| ENCE00000468394 | 1 | ENCP00000050541 | RP1-276N6.2 |
| | | ENCP00000050544 | LPA |

### Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00001232516 (open chromatin region) | 1 | embryonic stem cell (H1ESC) | H3K36me3, DNase1 |
| | | lung (IMR90) | H3K36me3 |
| | | blood (GM12878) | H3K36me3 |
| | | blood (K562) | H3K36me3 |
| | | skin (NHEK) | H3K36me3 |
| | | breast (HMEC) | H3K36me3 |

### Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| IGF2R | downstream gene variant | 2956 | ENST00000487607 | ? | ? | 1 |
| IGF2R | upstream gene variant | 3686 | ENST00000475584 | ? | ? | 1 |
| IGF2R | upstream gene variant, downstream gene variant | 2199 | ENST00000475834 | ? | ENSP00000473880 | 3 |
| RP11-288H12.3 | upstream gene variant | 662 | ENST00000366273 | ? | ? | 2 |
| RP11-288H12.3 | upstream gene variant | 321 | ENST00000569097 | ? | ? | 2 |

## Putative effect on transcript

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| IGF2R | ENST00000356956 | NM_000876.2 | ENSP00000349437 | 9 |
| IGF2R | ENST00000487607 | ? | ? | 1 |
| IGF2R | ENST00000475834 | ? | ENSP00000473880 | 3 |

### 3'-UTR variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| IGF2R | ENST00000475834 | ? | ENSP00000473880 | 1 |