

SNiPACard

Block annotations

Block info




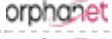

| | |
|---------------|------------------------------------|
| genomic range | chr4:3,287,052-3,291,401 <i>e!</i> |
| block size | 4,350 bp |
| variant count | 3 variants |

Basic features

| Conservation/deleteriousness | | Linked genes | |
|------------------------------|----------------------------------|-------------------------------|---|
| phyloP | $\mu = -0.490$ [-0.944 -- 0.147] | gene(s) hit or close-by | RGS12 <i>e!</i> |
| phastCons | $\mu = 0.001$ [0 - 0.004] | eQTL gene(s) | HTT-AS <i>e!</i> |
| GERP++ | $\mu = -0.385$ [-0.721 - 0.158] | potentially regulated gene(s) | AC141928.1 <i>e!</i> , ADD1 <i>e!</i> , ADRA2C <i>e!</i> , HGFAC <i>e!</i> , LRPAP1 <i>e!</i> , MFSD10 <i>e!</i> , RGS12 <i>e!</i> , SH3BP2 <i>e!</i> |
| CADD score | $\mu = 2.217$ [0.588 - 5.087] | disease gene(s) | LRPAP1 <i>e!</i> , SH3BP2 <i>e!</i> |


Trait annotations

Disease gene annotation

| gene | trait | source DB | source entry/link |
|------------------|---|-----------|--|
| LRPAP1 <i>e!</i> | Extreme myopia;Myopia 23, autosomal recessive | DECIPHER | MIM:615431  |
| SH3BP2 <i>e!</i> | CHERUBISM | OMIM | MIM:118400  |
| LRPAP1 <i>e!</i> | MYOPIA 23, AUTOSOMAL RECESSIVE | OMIM | MIM:615431  |
| SH3BP2 <i>e!</i> | CHERUBISM | OrphaNet | OrphaNet:184  |
| LRPAP1 <i>e!</i> | Rare isolated myopia | OrphaNet | OrphaNet:98619  |

Direct effect on regulation

cis-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|------------------|------------|---------------------------|--------|---------------------------------|--|------------|
| HTT-AS <i>e!</i> | ? | ENSG00000251075 <i>e!</i> | aorta | 8.73×10^{-8} (p-value) | GTEx Portal V6  | 3 |

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

| SNiPA enhancer id | variant(s) | associated SNiPA promoter id | associated gene(s) |
|---------------------------|------------|------------------------------|--------------------|
| ENCE00000370653 <i>e!</i> | 1 | ENCP00000040189 | RGS12 <i>e!</i> |
| | | ENCP00000040129 | SH3BP2 <i>e!</i> |
| | | ENCP00000040203 | ADRA2C <i>e!</i> |
| | | ENCP00000040193 | HGFAC <i>e!</i> |
| | | ENCP00000040188 | RGS12 <i>e!</i> |
| | | ENCP00000040179 | RGS12 <i>e!</i> |
| | | ENCP00000040176 | RGS12 <i>e!</i> |
| | | ENCP00000040198 | LRPAP1 <i>e!</i> |
| | | ENCP00000040142 | ADD1 <i>e!</i> |
| | | ENCP00000040191 | HGFAC <i>e!</i> |
| | | ENCP00000040132 | SH3BP2 <i>e!</i> |
| ENCE00000370721 <i>e!</i> | 1 | ENCP00000040141 | SH3BP2 <i>e!</i> |

| | |
|-----------------|----------------------|
| ENCP00000040154 | MFSD10 <i>e!</i> |
| ENCP00000040155 | MFSD10 <i>e!</i> |
| ENCP00000040183 | RGS12 <i>e!</i> |
| ENCP00000040202 | AC141928.1 <i>e!</i> |
| ENCP00000040153 | MFSD10 <i>e!</i> |
| ENCP00000040140 | SH3BP2 <i>e!</i> |

Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|------------|-----------------------------|--|
| ENSR00001235104 <i>e!</i> (promoter flanking region) | 1 | embryonic stem cell (H1ESC) | DNase1, H3K27me3 |
| | | H5MMtube | H3K27me3 |
| | | Osteobl | H3K27me3, H3K4me2 |
| | | blood (DND-41) | H3K27me3 |
| | | blood (K562) | H3K27me3 |
| | | skin (NHDF-AD) | DNase1, H3K4me2 |
| | | breast (HMEC) | H3K4me2 |
| | | monocytes (Monocytes-CD14+) | DNase1, H3K4me1, H3K4me3 |
| | | liver (HepG2) | PolII, H3K79me2, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, DNase1 |
| | | lung (IMR90) | H3K27me3 |
| | | blood (GM12878) | PU1, H2AZ, H3K4me1, DNase1, H3K4me2 |
| | | nervous (NH-A) | H3K27me3, DNase1 |
| | | skin (NHEK) | H3K27me3 |

Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|-----------------|-----------------------|---------------|---------------------------|-----------|---------|------------|
| RGS12 <i>e!</i> | upstream gene variant | 3354 | ENST00000506631 <i>e!</i> | ? | ? | 1 |

