

SNiPacard

Block annotations

Block info

| | |
|---------------|---------------------------------------|
| genomic range | chr16:72,747,685-72,800,567 <i>e!</i> |
| block size | 52,883 bp |
| variant count | 5 variants |

Basic features

| Conservation/deleteriousness | | Linked genes | |
|------------------------------|-------------------------------|-------------------------------|------------------------|
| phyloP | $\mu = 0.129$ [-2.24 – 1.711] | gene(s) hit or close-by | RP5-991G20.1 <i>e!</i> |
| phastCons | $\mu = 0.206$ [0.001 – 1] | eQTL gene(s) | - |
| GERP++ | $\mu = 0.692$ [-0.565 – 3.93] | potentially regulated gene(s) | C16orf47 <i>e!</i> |
| CADD score | $\mu = 7.917$ [0.325 – 17.48] | disease gene(s) | - |

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

| SNiPA enhancer id | variant(s) | associated SNiPA promoter id | associated gene(s) |
|---------------------------|------------|------------------------------|--------------------|
| ENCE00000193764 <i>e!</i> | 1 | ENCP00000021721 | C16orf47 <i>e!</i> |

Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|--|------------|-----------------------------|--|
| ENSR00000509797 <i>e!</i> (TF binding site) | 1 | embryonic stem cell (H1ESC) | POU5F1, Nanog, BCL11A, H3K4me2, USF1, DNase1 |

Putative effect on transcript

Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|------------------------|---------------------------|-----------|---------|------------|
| RP5-991G20.1 <i>e!</i> | ENST00000563328 <i>e!</i> | ? | ? | 5 |

