

# SNiPacard

## Block annotations

### Block info

genomic range	chr2:102,858,921-102,875,540 <i>e!</i>
block size	16,620 bp
variant count	5 variants

### Basic features

Conservation/deleteriousness		Linked genes	
phyloP	$\mu = 0.169$ [-0.796 – 1.086]	gene(s) hit or close-by	FAM183DP <i>e!</i> , IL1RL2 <i>e!</i>
phastCons	$\mu = 0.010$ [0 – 0.027]	eQTL gene(s)	AC007278.3 <i>e!</i> , IL18R1 <i>e!</i> , IL1RL1 <i>e!</i>
GERP++	$\mu = 0.493$ [-0.539 – 1.48]	potentially regulated gene(s)	-
CADD score	$\mu = 3.754$ [1.939 – 6.629]	disease gene(s)	-

## Direct effect on regulation

### cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IL1RL1 <i>e!</i>	?	ENSG00000115602 <i>e!</i>	lung	2.92×10 <sup>-6</sup> (p-value)	GTEx Portal V6	5
AC007278.3 <i>e!</i>	?	ENSG00000234389 <i>e!</i>	lung	1.49×10 <sup>-5</sup> (p-value)	GTEx Portal V6	1
AC007278.3 <i>e!</i>	?	ENSG00000234389 <i>e!</i>	blood	1.87×10 <sup>-8</sup> (p-value)	GTEx Portal V6	5
IL18R1 <i>e!</i>	?	ENSG00000115604 <i>e!</i>	cerebellum	8.77×10 <sup>-6</sup> (p-value)	GTEx Portal V6	1

## Putative effect on regulation

### Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001544945 <i>e!</i> (promoter flanking region)	1	NHLF HSMMtube Osteobl skin (NHDF-AD) breast (HMEC) muscle (HSMM) monocytes (Monocytes-CD14+) endothelium (HUVEC) liver (HepG2) lung (IMR90) nervous (NH-A) skin (NHEK)	DNase1 DNase1 H3K27ac DNase1 DNase1 DNase1 DNase1, H3K27me3 H3K36me3, Cjun, DNase1 H3K27me3, DNase1 DNase1, H2BK20ac DNase1 H3K4me1, H3K27ac, DNase1

### Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FAM183DP <i>e!</i>	downstream gene variant	442	ENST00000421987 <i>e!</i>	?	?	2
IL1RL2 <i>e!</i>	downstream gene variant	2459	ENST00000264257 <i>e!</i>	NM_003854.2	ENSP00000264257 <i>e!</i>	2
IL1RL2 <i>e!</i>	downstream gene variant	3110	ENST00000441515 <i>e!</i>	?	ENSP00000413348 <i>e!</i>	2
IL1RL2 <i>e!</i>	downstream gene variant	3123	ENST00000481806 <i>e!</i>	?	?	2

