

# SNiPacard

## Block annotations

### Block info


genomic range	chr13:92,414,715-92,453,051 <i>e!</i>
block size	38,337 bp
variant count	76 variants

### Basic features

Conservation/deleteriousness		Linked genes	
phyloP	$\mu = -0.632$ [-5.445 – 1.165]	gene(s) hit or close-by	GPC5 <i>e!</i>
phastCons	$\mu = 0.023$ [0 – 0.991]	eQTL gene(s)	-
GERP++	$\mu = -0.480$ [-5.75 – 2.7]	potentially regulated gene(s)	FABP5P4 <i>e!</i>
CADD score	$\mu = 2.705$ [0.015 – 13.41]	disease gene(s)	-

## Trait annotations

### Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
LUNG CANCER	<6.00×10 <sup>-6</sup>	GWAS Catalog	20304703 	1

## Putative effect on regulation

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

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000161614 <i>e!</i>	2	ENCP00000018923	FABP5P4 <i>e!</i>

### Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000518738 <i>e!</i> (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		embryonic stem cell (H1ESC)	Rad21, CTCF
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	Rad21, CTCF
		blood (K562)	CTCF

## Putative effect on transcript

### Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GPC5 <i>e!</i>	ENST00000377067 <i>e!</i>	NM_004466.4	ENSP00000366267 <i>e!</i>	76

