

# SNiPACard

## Block annotations

### Block info

genomic range	chr7:94,954,696-94,955,528 <i>el</i>
block size	833 bp
variant count	2 variants

### Basic features

Conservation/deleteriousness		Linked genes	
phyloP	$\mu = -0.623$ [-0.924 – -0.321]	gene(s) hit or close-by	PON1 <i>el</i>
phastCons	$\mu = 0.001$ [0 – 0.001]	eQTL gene(s)	ASB4 <i>el</i> , PON1 <i>el</i> , PON2 <i>el</i>
GERP++	$\mu = 0.224$ [-0.317 – 0.764]	potentially regulated gene(s)	-
CADD score	$\mu = 3.414$ [2.109 – 4.72]	disease gene(s)	PON1 <i>el</i> , PON2 <i>el</i>

## Trait annotations

### Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Paraoxonase activity	<5.00×10 <sup>-116</sup>	GWAS Catalog	22982463	1

### Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CR000232	1

### Disease gene annotation

gene	trait	source DB	source entry/link
PON1 <i>el</i>	MICROVASCULAR COMPLICATIONS OF DIABETES, SUSCEPTIBILITY TO, [...]	OMIM	MIM:612633
PON1 <i>el</i>	PARAOXONASE 1	OMIM	MIM:168820
PON2 <i>el</i>	Amyotrophic lateral sclerosis	OrphaNet	OrphaNet:803
PON1 <i>el</i>	Amyotrophic lateral sclerosis	OrphaNet	OrphaNet:803

## Direct effect on regulation

### cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PON1 <i>el</i>	?	ENSG00000005421 <i>el</i>	pancreas	2.91×10 <sup>-9</sup> (p-value)	GTEEx Portal V6	2
ASB4 <i>el</i>	?	ENSG00000005981 <i>el</i>	tibial nerve	5.98×10 <sup>-8</sup> (p-value)	GTEEx Portal V6	2
PON2 <i>el</i>	?	ENSG00000105854 <i>el</i>	tibial nerve	6.61×10 <sup>-10</sup> (p-value)	GTEEx Portal V6	2
PON1 <i>el</i>	?	ENSG00000005421 <i>el</i>	pituitary	2.82×10 <sup>-7</sup> (p-value)	GTEEx Portal V6	1
ASB4 <i>el</i>	?	ENSG00000005981 <i>el</i>	thyroid	7.13×10 <sup>-10</sup> (p-value)	GTEEx Portal V6	2
PON2 <i>el</i>	?	ENSG00000105854 <i>el</i>	thyroid	3.32×10 <sup>-7</sup> (p-value)	GTEEx Portal V6	2
PON1 <i>el</i>	?	ENSG00000005421 <i>el</i>	adrenal gland	5.05×10 <sup>-20</sup> (p-value)	GTEEx Portal V6	2
ASB4 <i>el</i>	?	ENSG00000005981 <i>el</i>	unexposed skin	2.84×10 <sup>-9</sup> (p-value)	GTEEx Portal V6	2
PON1 <i>el</i>	?	ENSG00000005421 <i>el</i>	liver	4.60×10 <sup>-11</sup> (p-value)	GTEEx Portal V6	2
ASB4 <i>el</i>	?	ENSG00000005981 <i>el</i>	sun exposed skin	4.86×10 <sup>-13</sup> (p-value)	GTEEx Portal V6	2

ASB4 <i>e!</i> ?	ENSG00000005981 <i>e!</i>	sun exposed skin	4.66×10 <sup>-5</sup> (p-value)	GTEX Portal V6 <i>e!</i>	2
PON2 <i>e!</i> ?	ENSG00000105854 <i>e!</i>	left ventricle	3.73×10 <sup>-7</sup> (p-value)	GTEX Portal V6 <i>e!</i>	2
PON2 <i>e!</i> ?	ENSG00000105854 <i>e!</i>	muscularis mucosae	2.07×10 <sup>-5</sup> (p-value)	GTEX Portal V6 <i>e!</i>	1
PON2 <i>e!</i> ?	ENSG00000105854 <i>e!</i>	esophagus mucosa	2.16×10 <sup>-5</sup> (p-value)	GTEX Portal V6 <i>e!</i>	1
ASB4 <i>e!</i> ?	ENSG00000005981 <i>e!</i>	ovary	1.24×10 <sup>-6</sup> (p-value)	GTEX Portal V6 <i>e!</i>	1
ASB4 <i>e!</i> ?	ENSG00000005981 <i>e!</i>	subcutaneous adipocytes	2.18×10 <sup>-5</sup> (p-value)	GTEX Portal V6 <i>e!</i>	1

### Putative effect on regulation

#### Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PON1 <i>e!</i>	upstream gene variant	677	ENST00000222381 <i>e!</i>	NM_000446.5	ENSP00000222381 <i>e!</i>	2
PON1 <i>e!</i>	upstream gene variant	908	ENST00000433729 <i>e!</i>	?	ENSP00000407359 <i>e!</i>	2

