Locus 404 (rs757158)

SNiPAcard

Block annotations

Block info

genomic range	chr7:94,954,696-94,955,528
block size	833 bp
variant count	2 variants

Basic features

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

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Paraoxonase activity	$<5.00\times10^{-116}$	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	MICROVASCULAR COMPLICATIONS OF DIABETES, SUSCEPTIBILITY TO, [...]	OMIM	MIM:612633
PON1	PARAOXONASE 1	OMIM	MIM:168820
PON2	Amyotrophic lateral sclerosis	OrphaNet	OrphaNet:803
PON1	Amyotrophic lateral sclerosis	OrphaNet	OrphaNet:803

Direct effect on regulation

cis-eQTL

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

ASB4	?	ENSG00000005981	sun exposed skin	4.88×10-15 (p-value)	GTEx Portal V6	2
PON2	?	ENSG00000105854	left ventricle	3.73×10-7 (p-value)	GTEx Portal V6	2
PON2	?	ENSG00000105854	muscularis mucosae	2.07×10-5 (p-value)	GTEx Portal V6	1
PON2	?	ENSG00000105854	esophagus mucosa	2.16×10-5 (p-value)	GTEx Portal V6	1
ASB4	?	ENSG00000005981	ovary	1.24×10-6 (p-value)	GTEx Portal V6	1
ASB4	?	ENSG00000005981	subcutaneous adipocytes	2.18×10-5 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Variation proximal to gene

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