Locus 315 (rs9295123)

SNiPAcard

Block annotations

Block info

genomic range	chr6:160,570,013-160,578,271
block size	8,259 bp
variant count	14 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.253 [-1.372 – 1.03]	**gene(s) hit or close-by**	SLC22A1
phastCons	μ = 0.007 [0 – 0.023]	**eQTL gene(s)**	MAS1, SLC22A1
GERP++	μ = -0.266 [-7.06 – 1.96]	**potentially regulated gene(s)**	–
CADD score	μ = 2.965 [0.586 – 6.336]	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
isobutyrylcarnitine	2.40×10^{-14}	Metabolomics GWAS Server	24816252	11
Lipoproteins	7.06×10^{-5}	dbGaP	pha003079	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MAS1	?	ENSG00000130368	ovary	7.01×10^{-7} (p-value)	GTEx Portal V6	3
SLC22A1	?	ENSG00000175003	tibial artery	2.64×10^{-6} (p-value)	GTEx Portal V6	12

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001232535 (CTCF binding site)	1	blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SLC22A1	upstream gene variant	1001	ENST00000478607	?	?	7

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SLC22A1	ENST00000324965	NM_153187.1	ENSP00000318103	14
SLC22A1	ENST00000460902	?	ENSP00000439274	14
SLC22A1	ENST00000366963	NM_003057.2	ENSP00000355930	14

SLC22A1	ENST00000539263	?	ENSP00000443245	14
SLC22A1	ENST00000457470	?	ENSP00000409557	14
SLC22A1	ENST00000478607	?	?	2

Locus 315 (sentinel SNP rs9295123 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.