Locus 52 (rs10935480)

SNiPAcard

Block annotations

Block info

genomic range	chr3:98,402,969-98,434,493
block size	31,525 bp
variant count	10 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.517 [-1.412 – 0.415]	gene(s) hit or close-by	ST3GAL6-AS1
phastCons	μ = 0.007 [0 – 0.029]	eQTL gene(s)	CPOX, DCBLD2, ST3GAL6, ST3GAL6-AS1
GERP++	μ = -0.989 [-5.16 – 0.372]	potentially regulated gene(s)	–
CADD score	μ = 3.151 [0.104 – 9.904]	disease gene(s)	CPOX

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CPOX	COPROPORPHYRIA, HEREDITARY	OMIM	MIM:121300
CPOX	Hereditary coproporphyria	OrphaNet	OrphaNet:79273

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
ST3GAL6-AS1	splice donor variant	ENST00000475816	?	?					1
ST3GAL6-AS1	splice donor variant	ENST00000488132	?	?					1
ST3GAL6-AS1	splice donor variant	ENST00000461931	?	?					1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ST3GAL6-AS1	?	ENSG00000239445	pancreas	6.23×10^{-8} (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	muscularis mucosae	2.39×10^{-17} (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	lung	2.20×10^{-10} (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	atrial appendage	3.43×10^{-28} (p-value)	GTEx Portal V6	10
CPOX	?	ENSG00000080819	transformed fibroblasts	1.28×10^{-6} (p-value)	GTEx Portal V6	6
ST3GAL6	?	ENSG00000064225	transformed fibroblasts	8.59×10^{-14} (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	transformed fibroblasts	3.69×10^{-12} (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	breast	4.14×10^{-14} (p-value)	GTEx Portal V6	10
ST3GAL6	?	ENSG00000064225	tibial artery	2.47×10^{-8} (p-value)	GTEx Portal V6	9

ST3GAL6-AS1	?	ENSG00000239445	tibial artery	3.76×10-13 (p-value)	GTEx Portal V6	10
DCBLD2	?	ENSG00000057019	thyroid	9.35×10-7 (p-value)	GTEx Portal V6	10
ST3GAL6	?	ENSG00000064225	thyroid	3.05×10-7 (p-value)	GTEx Portal V6	6
ST3GAL6-AS1	?	ENSG00000239445	thyroid	7.65×10-21 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	skeletal muscle	9.61×10-18 (p-value)	GTEx Portal V6	10
ST3GAL6	?	ENSG00000064225	skeletal muscle	4.66×10-6 (p-value)	GTEx Portal V6	5
ST3GAL6-AS1	?	ENSG00000239445	putamen	8.30×10-10 (p-value)	GTEx Portal V6	9
ST3GAL6-AS1	?	ENSG00000239445	liver	1.01×10-7 (p-value)	GTEx Portal V6	9
ST3GAL6-AS1	?	ENSG00000239445	sun exposed skin	1.25×10-16 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	unexposed skin	2.64×10-9 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	aorta	2.81×10-14 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	left ventricle	8.49×10-28 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	subcutaneous adipocytes	1.15×10-16 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	caudate basal ganglia	3.17×10-11 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	visceral adipocytes	2.24×10-12 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	cortex	2.18×10-12 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	coronary artery	6.20×10-10 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	sigmoid colon	3.28×10-17 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	tibial nerve	1.74×10-22 (p-value)	GTEx Portal V6	10
ST3GAL6	?	ENSG00000064225	esophagus mucosa	1.18×10-15 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	esophagus mucosa	6.81×10-14 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	anterior cingulate cortex	6.57×10-13 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	gastroesophageal junction	3.08×10-11 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	adrenal gland	1.34×10-7 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	cerebellar hemisphere	1.14×10-12 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	testis	5.41×10-12 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	hypothalamus	2.14×10-13 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	cerebellum	1.45×10-12 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	frontal cortex	1.51×10-13 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	EBV lymphocytes	2.90×10-6 (p-value)	GTEx Portal V6	8
ST3GAL6-AS1	?	ENSG00000239445	hippocampus	3.18×10-9 (p-value)	GTEx Portal V6	8
ST3GAL6-AS1	?	ENSG00000239445	stomach	2.88×10-7 (p-value)	GTEx Portal V6	5
ST3GAL6-AS1	?	ENSG00000239445	transverse colon	8.03×10-6 (p-value)	GTEx Portal V6	3
ST3GAL6-AS1	?	ENSG00000239445	nucleus accumbens	4.55×10-7 (p-value)	GTEx Portal V6	3

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001676910 (enhancer)	1	HSMMtube	H3K27me3
		Osteobl	H3K27me3
		lung (IMR90)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ST3GAL6-AS1	downstream gene variant	617	ENST00000475816	?	?	3
ST3GAL6-AS1	downstream gene variant	615	ENST00000488132	?	?	3
ST3GAL6-AS1	downstream gene variant	1590	ENST00000461931	?	?	3