Locus 68 (rs4693167)

SNiPAcard

Block annotations

Block info

genomic range	chr4:88,400,924-88,431,892
block size	30,969 bp
variant count	37 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.799 [-3.224 – 0.94]	gene(s) hit or close-by	SPARCL1
phastCons	μ = 0.045 [0 – 0.942]	eQTL gene(s)	HSD17B11, HSD17B13, NUDT9, PPM1K, RP11-529H2.2, SPARCL1
GERP++	μ = 0.298 [-4.01 – 4.25]	potentially regulated gene(s)	RP11-529H2.1, SPARCL1
CADD score	μ = 3.588 [0.322 – 10.4]	disease gene(s)	PPM1K

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
PPM1K	MAPLE SYRUP URINE DISEASE, MILD VARIANT	OMIM	MIM:615135
PPM1K	Intermediate maple syrup urine disease	OrphaNet	OrphaNet:268162

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)
SPARCL1	missense variant	ENST00000458304	?	ENSP00000406251	A/D	gCt/gAt	?	benign	1
SPARCL1	missense variant	ENST00000418378	NM_001128310.1	ENSP00000414856	2	2			2
SPARCL1	missense variant	ENST00000434434	?	ENSP00000416971	2	2			2
SPARCL1	missense variant	ENST00000282470	NM_004684.4	ENSP00000282470	2	2			2
SPARCL1	missense variant	ENST00000535835	?	ENSP00000438188	2	2			2
SPARCL1	missense variant	ENST00000509407	?	ENSP00000423483	2	2			2
SPARCL1	missense variant	ENST00000512317	?	ENSP00000423448	2	2			2
SPARCL1	missense variant	ENST00000543631	?	ENSP00000444832	A/D	gCt/gAt	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SPARCL1	ENST00000282470	ILMN_2218208	adipocyte	1.24×10-6 (p-value)	MuTHER consortium	7
SPARCL1	ENST00000503414					
SPARCL1	ENST00000418378					
SPARCL1	ENST00000282470	ILMN_1795251	adipocyte	1.65×10-6 (p-value)	MuTHER consortium	7
SPARCL1	ENST00000503414					
SPARCL1	ENST00000418378					

NUDT9	ENST00000302174	ILMN_1680239	blood	5.72×10-6 (p-value)	Westra et al.	6
NUDT9	ENST00000473942					
NUDT9	ENST00000440591					
HSD17B11	ENST00000508413	ILMN_1675117	blood	5.44×10-7 (p-value)	Westra et al.	4
HSD17B11	ENST00000358290					
HSD17B11	?	ENSG00000198189	tibial nerve	3.20×10-6 (p-value)	GTEx Portal V6	25
SPARCL1	?	ENSG00000152583	sun exposed skin	5.36×10-7 (p-value)	GTEx Portal V6	26
HSD17B11	?	ENSG00000198189	esophagus mucosa	1.01×10-7 (p-value)	GTEx Portal V6	36
SPARCL1	?	ENSG00000152583	subcutaneous adipocytes	5.55×10-6 (p-value)	GTEx Portal V6	14
HSD17B13	?	ENSG00000170509	thyroid	2.57×10-6 (p-value)	GTEx Portal V6	12
PPM1K	ENST00000608933	ILMN_2070043	b-cell	6.14×10-4 (p-value)	Fairfax et al.	1
PPM1K	ENST00000295908					
HSD17B11	?	ENSG00000198189	pancreas	5.48×10-6 (p-value)	GTEx Portal V6	3
RP11-529H2.2	?	ENSG00000255723	pancreas	1.05×10-5 (p-value)	GTEx Portal V6	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000660191	1	ENST00000503414, ENST00000541496	SPARCL1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000384981	1	ENCP00000041684	RP11-529H2.1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001244943 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF, DNase1, H3K4me2
		muscle (HSMM)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	CTCF
		blood (GM12878)	CTCF
		skin (NHEK)	CTCF
ENSR00001244946 (promoter flanking region)	4	cervix (HeLa-S3)	DNase1
		muscle (HSMM)	DNase1
ENSR00001686554 (CTCF binding site)	2	cervix (HeLa-S3)	DNase1
		muscle (HSMM)	DNase1
ENSR00001244947 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		liver (HepG2)	CTCF
		blood (K562)	CTCF
		skin (NHEK)	CTCF
ENSR00001432957 (promoter flanking region)	8	NHLF	H3K27ac, DNase1
		embryonic stem cell (H1ESC)	CTCF, DNase1
		HSMMtube	DNase1
		Osteobl	H3K4me2, H3K27ac

blood (K562)	CTCF
skin (NHDF-AD)	H3K4me1, DNase1, H3K4me2, H3K27ac
muscle (HSMM)	H3K4me1, H2AZ, H3K27ac, H3K4me2, DNase1
endothelium (HUVEC)	CTCF
nervous (NH-A)	DNase1
skin (NHEK)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SPARCL1	downstream gene variant	2545	ENST00000535835	?	ENSP00000438188	1
SPARCL1	downstream gene variant	2504	ENST00000434434	?	ENSP00000416971	1
SPARCL1	downstream gene variant	49	ENST00000543631	?	ENSP00000444832	2
SPARCL1	downstream gene variant	529	ENST00000458304	?	ENSP00000406251	2
SPARCL1	downstream gene variant	2568	ENST00000512317	?	ENSP00000423448	1
SPARCL1	downstream gene variant	2497	ENST00000541496	?	ENSP00000445678	1
SPARCL1	downstream gene variant	2497	ENST00000509407	?	ENSP00000423483	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SPARCL1	ENST00000543631	?	ENSP00000444832	31
SPARCL1	ENST00000503414	?	ENSP00000422903	35
SPARCL1	ENST00000282470	NM_004684.4	ENSP00000282470	35
SPARCL1	ENST00000512317	?	ENSP00000423448	31
SPARCL1	ENST00000418378	NM_001128310.1	ENSP00000414856	35
SPARCL1	ENST00000458304	?	ENSP00000406251	31
SPARCL1	ENST00000541496	?	ENSP00000445678	31
SPARCL1	ENST00000535835	?	ENSP00000438188	31
SPARCL1	ENST00000434434	?	ENSP00000416971	31
SPARCL1	ENST00000509407	?	ENSP00000423483	31

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SPARCL1	ENST00000503414	?	ENSP00000422903	2
SPARCL1	ENST00000541496	?	ENSP00000445678	2