Locus 156 (rs8396)

SNiPAcard

Block annotations

Block info

genomic range	chr4:159,621,250-159,648,866
block size	27,617 bp
variant count	16 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.438 [-2.93 – 2.416]	**gene(s) hit or close-by**	ETFDH, PPID, U3
phastCons	μ = 0.111 [0 – 1]	**eQTL gene(s)**	ETFDH, PPID, U3
GERP++	μ = -1.497 [-7.63 – 2.64]	**potentially regulated gene(s)**	–
CADD score	μ = 4.455 [0.066 – 18.16]	**disease gene(s)**	ETFDH

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Blood metabolite ratios	$<2.00\times10^{-57}$	GWAS Catalog	24816252	1
Blood metabolite levels	$<1.00\times10^{-31}$	GWAS Catalog	24816252	1
Metabolic traits	$<6.00\times10^{-15}$	GWAS Catalog	21886157	1
Metabolite levels	$<4.00\times10^{-24}$	GWAS Catalog	20037589	1
decanoylcarnitine	8.69×10^{-38}	Metabolomics GWAS Server	24816252	5

Disease gene annotation

gene	trait	source DB	source entry/link
ETFDH	MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY	OMIM	MIM:231680
ETFDH	glutaric aciduria type 2C	DECIPHER	MIM:231680
ETFDH	Multiple acyl-CoA dehydrogenation deficiency, mild type	OrphaNet	OrphaNet:394532
ETFDH	Multiple acyl-CoA dehydrogenation deficiency, severe neonatal type	OrphaNet	OrphaNet:394529

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)
PPID	missense variant	ENST00000307720	NM_005038.2	ENSP00000303754	I/L	Ata/Cta	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ETFDH	ENST00000307738	ILMN_1758034	skin	4.25×10^{-10} (p-value)	MuTHER consortium	6
ETFDH	ENST00000511912		adipocyte	3.72×10^{-20} (p-value)	MuTHER consortium	6
ETFDH	ENST00000506422					
PPID	?	ENSG00000171497	pancreas	7.06×10^{-7} (p-value)	GTEx Portal V6	12

U3	?	ENSG00000271817	pancreas	4.72×10-6 (p-value)	GTEx Portal V6	1
PPID	?	ENSG00000171497	muscularis mucosae	1.79×10-8 (p-value)	GTEx Portal V6	15
U3	?	ENSG00000271817	muscularis mucosae	8.28×10-7 (p-value)	GTEx Portal V6	12
ETFDH	?	ENSG00000171503	muscularis mucosae	2.95×10-6 (p-value)	GTEx Portal V6	2
U3	?	ENSG00000271817	lung	7.56×10-9 (p-value)	GTEx Portal V6	16
ETFDH	?	ENSG00000171503	atrial appendage	1.58×10-8 (p-value)	GTEx Portal V6	16
U3	?	ENSG00000271817	transformed fibroblasts	1.07×10-10 (p-value)	GTEx Portal V6	16
PPID	?	ENSG00000171497	transformed fibroblasts	1.14×10-14 (p-value)	GTEx Portal V6	16
ETFDH	?	ENSG00000171503	tibial artery	1.13×10-9 (p-value)	GTEx Portal V6	16
U3	?	ENSG00000271817	tibial artery	2.61×10-12 (p-value)	GTEx Portal V6	16
PPID	?	ENSG00000171497	tibial artery	2.37×10-6 (p-value)	GTEx Portal V6	6
U3	?	ENSG00000271817	breast	1.62×10-11 (p-value)	GTEx Portal V6	16
U3	?	ENSG00000271817	thyroid	9.17×10-7 (p-value)	GTEx Portal V6	10
PPID	?	ENSG00000171497	thyroid	2.76×10-6 (p-value)	GTEx Portal V6	13
ETFDH	?	ENSG00000171503	skeletal muscle	7.75×10-40 (p-value)	GTEx Portal V6	16
PPID	?	ENSG00000171497	transverse colon	1.32×10-8 (p-value)	GTEx Portal V6	16
U3	?	ENSG00000271817	sun exposed skin	1.47×10-11 (p-value)	GTEx Portal V6	16
PPID	?	ENSG00000171497	sun exposed skin	1.51×10-10 (p-value)	GTEx Portal V6	16
ETFDH	?	ENSG00000171503	left ventricle	4.25×10-13 (p-value)	GTEx Portal V6	16
PPID	?	ENSG00000171497	left ventricle	1.75×10-10 (p-value)	GTEx Portal V6	16
U3	?	ENSG00000271817	subcutaneous adipocytes	1.07×10-19 (p-value)	GTEx Portal V6	16
PPID	?	ENSG00000171497	subcutaneous adipocytes	6.66×10-10 (p-value)	GTEx Portal V6	16
U3	?	ENSG00000271817	visceral adipocytes	4.40×10-8 (p-value)	GTEx Portal V6	13
PPID	?	ENSG00000171497	visceral adipocytes	3.13×10-7 (p-value)	GTEx Portal V6	14
PPID	?	ENSG00000171497	stomach	7.84×10-8 (p-value)	GTEx Portal V6	14
ETFDH	?	ENSG00000171503	stomach	2.05×10-7 (p-value)	GTEx Portal V6	1
U3	?	ENSG00000271817	tibial nerve	2.56×10-17 (p-value)	GTEx Portal V6	16
PPID	?	ENSG00000171497	tibial nerve	2.96×10-19 (p-value)	GTEx Portal V6	16
U3	?	ENSG00000271817	esophagus mucosa	2.03×10-7 (p-value)	GTEx Portal V6	16
PPID	?	ENSG00000171497	esophagus mucosa	5.64×10-11 (p-value)	GTEx Portal V6	16
ETFDH	?	ENSG00000171503	adrenal gland	5.58×10-8 (p-value)	GTEx Portal V6	14
U3	?	ENSG00000271817	unexposed skin	6.54×10-6 (p-value)	GTEx Portal V6	2
U3	?	ENSG00000271817	aorta	3.30×10-6 (p-value)	GTEx Portal V6	6
U3	?	ENSG00000271817	testis	9.31×10-9 (p-value)	GTEx Portal V6	12
U3	?	ENSG00000271817	gastroesophageal junction	5.99×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001253032 (CTCF binding site)	2	embryonic stem cell (H1ESC)	H3K36me3, CTCF, Rad21
		HSMMtube	CTCF
		skin (NHDF-AD)	H3K36me3, CTCF
		muscle (HSMM)	CTCF, H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3, CTCF
		nervous (NH-A)	H3K36me3
		skin (NHEK)	H3K36me3
		Osteobl	CTCF, H3K36me3
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	H3K36me3
ENSR00001436804 (promoter)	2	embryonic stem cell (H1ESC)	DNase1, H3K4me2, H3K9ac, H3K4me3, TAF1, Yy1, H3K36me3, H3K27me3, CTCF, PolII, Rad21, Egr1, Gabp, TAF7, H3K27ac
		HSMMtube	H3K4me2, H3K27ac, H3K4me3, H2AZ, DNase1, H3K9ac, H3K79me2
		blood (K562)	H3K27ac, Max, E2F6, H3K9ac, Gabp, H3K79me2, HEY1, H2AZ, TAF1, ELF1, Cmyc, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, H3K4me3, DNase1
		muscle (HSMM)	H3K9ac, H3K36me3, H3K4me2, H3K27ac, H3K4me3, H2AZ, H3K79me2
		liver (HepG2)	ELF1, H2AZ, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3, TAF1, H3K79me2, Gabp, Sin3Ak20, PolII, DNase1
		lung (IMR90)	H4K8ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K9ac
		blood (GM12878)	DNase1, PolII, H2AZ, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	H3K27ac, H3K9ac, H3K4me3
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2
		breast (HMEC)	H3K4me2, H3K27ac, H3K4me3, H3K9ac
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, PolII, H3K4me3, Gabp
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cmyc, DNase1, PolII, H3K4me3, H3K4me2, Max
		A549	H3K9ac, DNase1, H3K27ac, H3K36me3, H3K4me2, H3K4me3

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
ETFDH	1	ENST00000511912	hsa-miR-144-3p hsa-miR-340-5p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ETFDH	downstream gene variant	42	ENST00000511912	NM_004453.3	ENSP00000426638	5
ETFDH	downstream gene variant	1096	ENST00000507475	?	ENSP00000422735	2
ETFDH	downstream gene variant	63	ENST00000307738	NM_001281738.1, NM_001281737.1	ENSP00000303552	4
ETFDH	downstream gene variant	94	ENST00000506422	?	?	4
PPID	downstream gene variant, upstream gene variant	18	ENST00000507213	?	ENSP00000421221	7
PPID	downstream gene variant, upstream gene variant	472	ENST00000307720	NM_005038.2	ENSP00000303754	4
PPID	downstream gene variant, upstream gene	96	ENST00000512699	?	ENSP00000423207	5

	variant					
U3	upstream gene variant	593	ENST00000607547	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ETFDH	ENST00000307738	NM_001281738.1, NM_001281737.1	ENSP00000303552	3
ETFDH	ENST00000511912	NM_004453.3	ENSP00000426638	3
ETFDH	ENST00000506422	?	?	3
PPID	ENST00000307720	NM_005038.2	ENSP00000303754	6
PPID	ENST00000512699	?	ENSP00000423207	4

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ETFDH	ENST00000511912	NM_004453.3	ENSP00000426638	1
PPID	ENST00000307720	NM_005038.2	ENSP00000303754	1
PPID	ENST00000512699	?	ENSP00000423207	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
PPID	ENST00000307720	NM_005038.2	ENSP00000303754	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
U3	ENST00000607547	?	1