Locus 40 (rs9013)

SNiPAcard

Block annotations

Block info

genomic range	chr2:143,778,115-143,810,757
block size	32,643 bp
variant count	28 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.207 [-2.518 – 1.857]	gene(s) hit or close-by	KYNU
phastCons	μ = 0.063 [0 – 0.402]	eQTL gene(s)	–
GERP++	μ = -0.222 [-6.91 – 4.36]	potentially regulated gene(s)	–
CADD score	μ = 4.571 [0.063 – 13.06]	disease gene(s)	KYNU

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
KYNU	HYDROXYKYNURENINURIA	OMIM	MIM:236800
KYNU	Encephalopathy due to hydroxykynureninuria	OrphaNet	OrphaNet:79155

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)
KYNU	missense variant	ENST00000264170	NM_003937.2	ENSP00000264170	E/K	Gaa/Aaa	?	?	1
KYNU	missense variant	ENST00000409512	NM_001199241.1	ENSP00000386731	E/K	Gaa/Aaa	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
?	?	ILMN_1746517	monocyte	2.76×10^{-5} (p-value)	Fairfax et al.	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001657233 (CTCF binding site)	2	embryonic stem cell (H1ESC)	CTCF
		endothelium (HUVEC)	H3K36me3
		HSMMtube	H3K27me3
		A549	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
KYNU	downstream gene variant	166	ENST00000409512	NM_001199241.1	ENSP00000386731	10

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
KYNU	ENST00000264170	NM_003937.2	ENSP00000264170	7
KYNU	ENST00000409512	NM_001199241.1	ENSP00000386731	7

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
KYNU	ENST00000264170	NM_003937.2	ENSP00000264170	20