Locus 440 (rs10503709)

SNiPAcard

Block annotations

Block info

genomic range	chr8:21,437,574-21,479,725
block size	42,152 bp
variant count	94 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.257 [-2.032 – 1.342]	gene(s) hit or close-by	–
phastCons	μ = 0.053 [0 – 0.999]	eQTL gene(s)	GFRA2
GERP++	μ = -0.220 [-5.17 – 3.73]	potentially regulated gene(s)	FAM160B2, FGF17, NPM2
CADD score	μ = 2.968 [0.028 – 20.2]	disease gene(s)	FGF17

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
erythronate*	4.06×10-5	Metabolomics GWAS Server	24816252	2

Disease gene annotation

gene	trait	source DB	source entry/link
FGF17	HYPOGONADOTROPIC HYPOGONADISM 20 WITH OR WITHOUT ANOSMIA	OMIM	MIM:615270
FGF17	Kallmann Syndrome	OrphaNet	OrphaNet:478
FGF17	Normosmic congenital hypogonadotropic hypogonadism	OrphaNet	OrphaNet:432

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
GFRA2	?	ENSG00000168546	gastroesophageal junction	1.84×10-16 (p-value)	GTEx Portal V6	93
GFRA2	?	ENSG00000168546	muscularis mucosae	6.70×10-31 (p-value)	GTEx Portal V6	93
GFRA2	?	ENSG00000168546	stomach	4.20×10-10 (p-value)	GTEx Portal V6	93
GFRA2	?	ENSG00000168546	atrial appendage	2.58×10-12 (p-value)	GTEx Portal V6	93
GFRA2	?	ENSG00000168546	sigmoid colon	4.15×10-12 (p-value)	GTEx Portal V6	93
GFRA2	?	ENSG00000168546	transverse colon	1.00×10-7 (p-value)	GTEx Portal V6	87
GFRA2	?	ENSG00000168546	esophagus mucosa	3.25×10-17 (p-value)	GTEx Portal V6	93

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000509140	1	ENCP00000055225	FGF17
		ENCP00000055220	NPM2

ENCE00000509287	1	ENCP00000055235	FAM160B2
		ENCP00000055236	FAM160B2
ENCE00000509141	1	ENCP00000055225	FGF17
		ENCP00000055220	NPM2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001439442 (open chromatin region)	1	A549	H3K4me3
ENSR00001720509 (open chromatin region)	4	embryonic stem cell (H1ESC)	Rad21
		A549	DNase1
		blood (K562)	H3K27me3
ENSR00001387343 (promoter flanking region)	1	cervix (HeLa-S3)	H3K27me3
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHEK)	H3K4me1, DNase1