Locus 403 (rs3135063)

SNiPAcard

Block annotations

Block info

genomic range	chr4:3,287,052-3,291,401
block size	4,350 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.490 [-0.944 – -0.147]	gene(s) hit or close-by	RGS12
phastCons	μ = 0.001 [0 – 0.004]	eQTL gene(s)	HTT-AS
GERP++	μ = -0.385 [-0.721 – 0.158]	potentially regulated gene(s)	AC141928.1, ADD1, ADRA2C, HGFAC, LRPAP1, MFSD10, RGS12, SH3BP2
CADD score	μ = 2.217 [0.588 – 5.087]	disease gene(s)	LRPAP1, SH3BP2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
LRPAP1	Extreme myopia;Myopia 23, autosomal recessive	DECIPHER	MIM:615431
SH3BP2	CHERUBISM	OMIM	MIM:118400
LRPAP1	MYOPIA 23, AUTOSOMAL RECESSIVE	OMIM	MIM:615431
SH3BP2	CHERUBISM	OrphaNet	OrphaNet:184
LRPAP1	Rare isolated myopia	OrphaNet	OrphaNet:98619

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HTT-AS	?	ENSG00000251075	aorta	8.73×10^{-8} (p-value)	GTEx Portal V6	3

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000370653	1	ENCP00000040189	RGS12
		ENCP00000040129	SH3BP2
		ENCP00000040203	ADRA2C
		ENCP00000040193	HGFAC
		ENCP00000040188	RGS12
		ENCP00000040179	RGS12
		ENCP00000040176	RGS12
		ENCP00000040198	LRPAP1
		ENCP00000040142	ADD1
		ENCP00000040191	HGFAC
		ENCP00000040132	SH3BP2
ENCE00000370721	1	ENCP00000040141	SH3BP2

	ENCP00000040154	MFSD10
	ENCP00000040155	MFSD10
	ENCP00000040183	RGS12
	ENCP00000040202	AC141928.1
	ENCP00000040153	MFSD10
	ENCP00000040140	SH3BP2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001235104 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3, H3K4me2
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1, H3K4me2
		breast (HMEC)	H3K4me2
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me3
		liver (HepG2)	PolII, H3K79me2, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, DNase1
		lung (IMR90)	H3K27me3
		blood (GM12878)	PU1, H2AZ, H3K4me1, DNase1, H3K4me2
		nervous (NH-A)	H3K27me3, DNase1
		skin (NHEK)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RGS12	upstream gene variant	3354	ENST00000506631	?	?	1