Locus 239 (rs2412776)

SNiPAcard

Block annotations

Block info

genomic range	chr4:57,941,160-57,952,521
block size	11,362 bp
variant count	12 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.191 [-1.557 – 1.293]	gene(s) hit or close-by	IGFBP7, UBE2CP3
phastCons	μ = 0.163 [0 – 0.97]	eQTL gene(s)	–
GERP++	μ = -0.177 [-9.01 – 5.22]	potentially regulated gene(s)	POLR2B
CADD score	μ = 6.193 [0.746 – 18.99]	disease gene(s)	IGFBP7

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
IGFBP7	RETINAL ARTERIAL MACROANEURYSM WITH SUPRAVALVULAR PULMONIC STENOSIS;	DECIPHER	MIM:614224
IGFBP7	RETINAL ARTERIAL MACROANEURYSM WITH SUPRAVALVULAR PULMONIC [...]	OMIM	MIM:614224
IGFBP7	Familial retinal arterial macroaneurysm	OrphaNet	OrphaNet:284247

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000380606	2	ENCP00000041130	POLR2B

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001241735 (promoter flanking region)	5	NHLF	DNase1
		HSMMtube	H3K36me3, DNase1
		Osteobl	H3K27ac, H3K36me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K36me3
		breast (HMEC)	DNase1, H3K27ac
		cervix (HeLa-S3)	Max, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		lung (IMR90)	DNase1, H3K36me3
		nervous (NH-A)	H3K36me3, DNase1
		A549	H3K36me3
		skin (NHEK)	DNase1, H3K36me3
ENSR00001685382 (enhancer)	2	Osteobl	H3K36me3
		nervous (NH-A)	H3K36me3
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
UBE2CP3	downstream gene variant	1862	ENST00000502715	?	?	6

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IGFBP7	ENST00000514062	NM_001253835.1	ENSP00000486293	12
IGFBP7	ENST00000295666	NM_001553.2	ENSP00000295666	12