Locus 230 (rs7439493)

SNiPAcard

Block annotations

Block info

genomic range	chr4:110,649,891-110,658,856
block size	8,966 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.257 [-2.61 – 0.539]	**gene(s) hit or close-by**	CFI, PLA2G12A
phastCons	μ = 0.021 [0 – 0.118]	**eQTL gene(s)**	CASP6, PLA2G12A
GERP++	μ = -0.596 [-4.35 – 1.04]	**potentially regulated gene(s)**	CFI, CFI
CADD score	μ = 2.770 [0.01 – 4.375]	**disease gene(s)**	CFI

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Macular degeneration	1.59×10-6	dbGaP	pha002890	dbGaP	1
Macular degeneration	1.26×10-6	dbGaP	pha002869	dbGaP	1

Disease gene annotation

gene	trait	source DB	source entry/link	
CFI	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 3	OMIM	MIM:612923	OMIM
CFI	COMPLEMENT FACTOR I DEFICIENCY	OMIM	MIM:610984	OMIM
CFI	MACULAR DEGENERATION, AGE-RELATED, 13	OMIM	MIM:615439	OMIM
CFI	Immunodeficiency with factor I anomaly	OrphaNet	OrphaNet:200418	orphanet
CFI	Atypical hemolytic uremic syndrome with I factor anomaly	OrphaNet	OrphaNet:93580	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CASP6	?	ENSG00000138794	cerebellar hemisphere	1.87×10-8 (p-value)	GTEx Portal V6	8
CASP6	?	ENSG00000138794	atrial appendage	1.39×10-6 (p-value)	GTEx Portal V6	7
PLA2G12A	?	ENSG00000123739	tibial nerve	6.77×10-22 (p-value)	GTEx Portal V6	8
CASP6	?	ENSG00000138794	cerebellum	1.45×10-8 (p-value)	GTEx Portal V6	8
PLA2G12A	?	ENSG00000123739	transformed fibroblasts	4.97×10-8 (p-value)	GTEx Portal V6	8
PLA2G12A	?	ENSG00000123739	esophagus mucosa	2.23×10-12 (p-value)	GTEx Portal V6	8
PLA2G12A	?	ENSG00000123739	subcutaneous adipocytes	7.24×10-13 (p-value)	GTEx Portal V6	8
PLA2G12A	?	ENSG00000123739	tibial artery	1.79×10-5 (p-value)	GTEx Portal V6	1
PLA2G12A	?	ENSG00000123739	vagina	2.08×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000388317	1	ENCP00000042049	CFI CFI

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001434100 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, PolII, Rad21, Sin3Ak20, H3K27ac, Yy1, TAF1, H3K4me2, USF1, H3K9ac, H3K4me3
		HSMMtube	H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	H3K27ac, Max, MEF2A, H3K9ac, USF1, H3K79me2, H2AZ, PolII, H3K4me2, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2
		liver (HepG2)	PolII, TAF1, USF1, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K27me3, DNase1
		blood (GM12878)	DNase1, PolII, H2AZ, H3K4me3, USF1, ZEB1, H3K4me2, H3K9ac
		lung (IMR90)	H3K4me2, H3K4me3, H3K9ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	DNase1, H3K27ac, H3K9ac, H3K4me2, H3K4me3
		NHLF	H3K27ac, H3K9ac, H3K4me3
		Osteobl	H3K27ac, H2AZ, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me3, H3K9ac, H3K4me2, H3K4me1
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	H3K4me3, H3K79me2, PolII, DNase1, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	H3K4me2, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Max, H3K4me3, H3K4me2, PolII, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CFI	downstream gene variant	2994	ENST00000394634	NM_000204.3	ENSP00000378130	1
CFI	downstream gene variant	2994	ENST00000504853	?	?	1
CFI	downstream gene variant	3013	ENST00000618244	?	ENSP00000483416	1
CFI	downstream gene variant	4176	ENST00000515512	?	?	1
CFI	downstream gene variant	3024	ENST00000512148	?	ENSP00000427438	1
CFI	downstream gene variant	2996	ENST00000394635	?	ENSP00000378131	1
PLA2G12A	upstream gene variant	1604	ENST00000502283	?	ENSP00000425274	5
PLA2G12A	upstream gene variant	1501	ENST00000243501	NM_030821.4	ENSP00000243501	5
PLA2G12A	upstream gene variant	1622	ENST00000507961	?	ENSP00000424021	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PLA2G12A	ENST00000507961	?	ENSP00000424021	1
PLA2G12A	ENST00000243501	NM_030821.4	ENSP00000243501	1
PLA2G12A	ENST00000502283	?	ENSP00000425274	1