Locus 229 (rs429017)

SNiPAcard

Block annotations

Block info

genomic range	chr5:41,263,091-41,266,229
block size	3,139 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.503 [-2.33 – 0.563]	gene(s) hit or close-by	C6
phastCons	μ = 0.014 [0 – 0.112]	eQTL gene(s)	C7, PLCXD3
GERP++	μ = -0.409 [-6.2 – 3.32]	potentially regulated gene(s)	PLCXD3
CADD score	μ = 4.149 [0.977 – 9.545]	disease gene(s)	C7, C6

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link	
C7	COMPLEMENT COMPONENT 7 DEFICIENCY	OMIM	MIM:610102	OMIM
C6	COMPLEMENT COMPONENT 6 DEFICIENCY	OMIM	MIM:612446	OMIM
C7	Immunodeficiency due to a late component of complements deficiency	OrphaNet	OrphaNet:169150	orphanet
C6	Immunodeficiency due to a late component of complements deficiency	OrphaNet	OrphaNet:169150	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PLCXD3	?	ENSG00000182836	muscularis mucosae	2.99×10^{-7} (p-value)	GTEx Portal V6	9
C7	?	ENSG00000112936	skeletal muscle	9.97×10^{-8} (p-value)	GTEx Portal V6	9
PLCXD3	?	ENSG00000182836	tibial nerve	2.08×10^{-7} (p-value)	GTEx Portal V6	9
PLCXD3	?	ENSG00000182836	sun exposed skin	5.99×10^{-6} (p-value)	GTEx Portal V6	9
PLCXD3	?	ENSG00000182836	lung	1.57×10^{-5} (p-value)	GTEx Portal V6	6

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000407607	1	ENCP00000043972	PLCXD3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C6	upstream gene variant	1551	ENST00000263413	NM_001115131.2	ENSP00000263413	9