Locus 57 (rs17284969)

SNiPAcard

Block annotations

Block info

genomic range	chr7:83,004,981-83,063,088
block size	58,108 bp
variant count	30 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.184 [-2.023 – 2.024]	gene(s) hit or close-by	AC079987.2, SEMA3E
phastCons	μ = 0.063 [0 – 1]	eQTL gene(s)	CHI3L2
GERP++	μ = -0.468 [-4.54 – 3.12]	potentially regulated gene(s)	SEMA3E
CADD score	μ = 3.790 [0.019 – 22.4]	disease gene(s)	SEMA3E

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link	
SEMA3E	CHARGE SYNDROME	OrphaNet	OrphaNet:138	orphanet

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
SEMA3E	missense variant	ENST00000427262	NM_001178129.1	ENSP00000405052	P/R	cCa/cGa	?	?	1
SEMA3E	missense variant	ENST00000307792	NM_012431.2	ENSP00000303212	P/R	cCa/cGa	?	?	1
SEMA3E	missense variant	ENST00000442159	?	ENSP00000412867	P/R	cCa/cGa	?	?	1

Direct effect on regulation

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
CHI3L2	?	ENSG00000064886	chr1	blood	2.70×10^{-2} (q-value)	SeeQTL DB (HapMap)	2

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000798976	1	ENST00000427262, ENST00000307792	SEMA3E

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001715349 (enhancer)	1	HSMMtube	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC079987.2	upstream gene variant	4757	ENST00000420181	?	?	1
SEMA3E	downstream gene variant	3322	ENST00000442159	?	ENSP00000412867	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SEMA3E	ENST00000427262	NM_001178129.1	ENSP00000405052	29
SEMA3E	ENST00000307792	NM_012431.2	ENSP00000303212	29
SEMA3E	ENST00000442159	?	ENSP00000412867	5