SNiPAcard

Block annotations

Block info

genomic range	chr7:83,224,626-83,287,607
block size	62,982 bp
variant count	24 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.337 [-2.51 – 0.531]	**gene(s) hit or close-by**	SEMA3E
phastCons	μ = 0.018 [0 – 0.12]	**eQTL gene(s)**	SEMA3E
GERP++	μ = -0.310 [-3.15 – 2.13]	**potentially regulated gene(s)**	–
CADD score	μ = 2.926 [0.263 – 7.555]	**disease gene(s)**	SEMA3E

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
1-eicosadienoylglycerophosphocholine*	9.31×10^{-5}	Metabolomics GWAS Server	24816252	1

Disease gene annotation

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

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SEMA3E	?	ENSG00000170381	gastroesophageal junction	2.51×10^{-7} (p-value)	GTEx Portal V6	20
SEMA3E	?	ENSG00000170381	muscularis mucosae	8.25×10^{-10} (p-value)	GTEx Portal V6	23
SEMA3E	?	ENSG00000170381	breast	2.96×10^{-8} (p-value)	GTEx Portal V6	22
SEMA3E	?	ENSG00000170381	subcutaneous adipocytes	2.84×10^{-9} (p-value)	GTEx Portal V6	23
SEMA3E	?	ENSG00000170381	thyroid	1.32×10^{-12} (p-value)	GTEx Portal V6	24
SEMA3E	?	ENSG00000170381	tibial nerve	8.70×10^{-6} (p-value)	GTEx Portal V6	4
SEMA3E	?	ENSG00000170381	transformed fibroblasts	5.95×10^{-7} (p-value)	GTEx Portal V6	9
SEMA3E	?	ENSG00000170381	esophagus mucosa	1.91×10^{-6} (p-value)	GTEx Portal V6	2

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001561427 (promoter)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, H3K4me2, USF1, H3K4me3, DNase1
		HSMMtube	H3K4me2, H3K27me3
		Osteobl	H3K27me3, H3K4me2

blood (DND-41)	H3K27me3
skin (NHDF-AD)	H3K4me3, H3K4me2
breast (HMEC)	H3K4me2, H3K4me3
cervix (HeLa-S3)	H3K27me3
monocytes (Monocytes-CD14+)	H3K27me3
endothelium (HUVEC)	H3K36me3, H3K27me3
liver (HepG2)	H3K27me3
nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, DNase1
A549	H3K4me3, H3K4me2, H3K9ac, H3K27me3
skin (NHEK)	H3K4me2, H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SEMA3E	upstream gene variant	101	ENST00000453333	?	ENSP00000415184	3
SEMA3E	upstream gene variant	2987	ENST00000442159	?	ENSP00000412867	2
SEMA3E	upstream gene variant	4879	ENST00000427262	NM_001178129.1	ENSP00000405052	1
SEMA3E	upstream gene variant	567	ENST00000307792	NM_012431.2	ENSP00000303212	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SEMA3E	ENST00000307792	NM_012431.2	ENSP00000303212	19
SEMA3E	ENST00000427262	NM_001178129.1	ENSP00000405052	18
SEMA3E	ENST00000453333	?	ENSP00000415184	19
SEMA3E	ENST00000442159	?	ENSP00000412867	7

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SEMA3E	ENST00000307792	NM_012431.2	ENSP00000303212	1