SNiPAcard

Block annotations

Block info

genomic range	chr12:94,620,432-94,634,913
block size	14,482 bp
variant count	11 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.040 [-1.413 – 2.23]	**gene(s) hit or close-by**	PLXNC1
phastCons	μ = 0.166 [0 – 1]	**eQTL gene(s)**	UBE2N
GERP++	μ = 0.078 [-7.06 – 3.51]	**potentially regulated gene(s)**	–
CADD score	μ = 7.383 [2.948 – 17.47]	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
ibuprofen	2.09×10^{-5}	Metabolomics GWAS Server	24816252	2

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)
PLXNC1	missense variant	ENST00000258526	NM_005761.2	ENSP00000258526	S/R	agT/agG	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
UBE2N	?	ENSG00000177889	transformed fibroblasts	2.12×10^{-5} (p-value)	GTEx Portal V6	3

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001614108 (enhancer)	1	cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	DNase1, H3K4me1, p300, FOXA1, H3K27ac
		HSMMtube	H3K27me3
		blood (GM12878)	H3K36me3
		Osteobl	H3K27me3
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PLXNC1	downstream gene variant	2273	ENST00000546659	?	?	2

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
PLXNC1	ENST00000258526	NM_005761.2	ENSP00000258526	N	aaT/aaC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PLXNC1	ENST00000258526	NM_005761.2	ENSP00000258526	9

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PLXNC1	ENST00000546659	?	1