SNiPAcard

Block annotations

Block info

genomic range	chr9:5,505,822-5,516,067
block size	10,246 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.079 [-1.593 – 0.836]	**gene(s) hit or close-by**	PDCD1LG2
phastCons	μ = 0.008 [0 – 0.023]	**eQTL gene(s)**	PDCD1LG2
GERP++	μ = 0.014 [-5.63 – 2.98]	**potentially regulated gene(s)**	KIAA2026, RP11-574F11.3
CADD score	μ = 3.835 [0.078 – 7.74]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PDCD1LG2	?	ENSG00000197646	muscularis mucosae	3.64×10^{-16} (p-value)	GTEx Portal V6	7
PDCD1LG2	?	ENSG00000197646	visceral adipocytes	3.12×10^{-15} (p-value)	GTEx Portal V6	7
PDCD1LG2	?	ENSG00000197646	lung	1.69×10^{-14} (p-value)	GTEx Portal V6	7
PDCD1LG2	?	ENSG00000197646	tibial nerve	4.38×10^{-11} (p-value)	GTEx Portal V6	7
PDCD1LG2	?	ENSG00000197646	transformed fibroblasts	1.40×10^{-9} (p-value)	GTEx Portal V6	7
PDCD1LG2	?	ENSG00000197646	thyroid	1.98×10^{-16} (p-value)	GTEx Portal V6	7
PDCD1LG2	?	ENSG00000197646	skeletal muscle	3.56×10^{-6} (p-value)	GTEx Portal V6	2
PDCD1LG2	?	ENSG00000197646	left ventricle	1.06×10^{-8} (p-value)	GTEx Portal V6	7
PDCD1LG2	?	ENSG00000197646	aorta	5.40×10^{-9} (p-value)	GTEx Portal V6	7
PDCD1LG2	?	ENSG00000197646	subcutaneous adipocytes	2.82×10^{-11} (p-value)	GTEx Portal V6	7
PDCD1LG2	?	ENSG00000197646	atrial appendage	3.21×10^{-6} (p-value)	GTEx Portal V6	1
PDCD1LG2	?	ENSG00000197646	sigmoid colon	2.43×10^{-6} (p-value)	GTEx Portal V6	1
PDCD1LG2	?	ENSG00000197646	breast	1.59×10^{-6} (p-value)	GTEx Portal V6	1
PDCD1LG2	?	ENSG00000197646	gastroesophageal junction	9.59×10^{-8} (p-value)	GTEx Portal V6	1
PDCD1LG2	?	ENSG00000197646	adrenal gland	3.52×10^{-6} (p-value)	GTEx Portal V6	1
PDCD1LG2	?	ENSG00000197646	tibial artery	3.29×10^{-6} (p-value)	GTEx Portal V6	5
PDCD1LG2	?	ENSG00000197646	coronary artery	4.73×10^{-6} (p-value)	GTEx Portal V6	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)

ENCE00000536431	1	ENCP00000058187	RP11-574F11.3
ENCE00000536432	1	ENCP00000058198	KIAA2026
		ENCP00000058187	RP11-574F11.3

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001299543 (promoter)	1	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K4me2, DNase1
		skin (NHDF-AD)	H3K9ac, DNase1, H3K4me3, H3K4me1, H3K4me2, H3K27ac
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac
		liver (HepG2)	DNase1, H3K27me3
		lung (IMR90)	H3K4me2, H3K56ac, H4K5ac, H3K36me3, H3K4me3, H3K4ac, H2BK120ac, H2BK20ac, H2AK5ac, H3K9ac, H4K91ac, DNase1, H4K8ac, H3K23ac, H3K14ac, H2BK12ac, H3K27ac, H3K18ac
		blood (GM12878)	BCL11A, H3K27ac, H3K4me2, H3K9ac, H3K4me3, H3K79me2, IRF4, Cjun, PU1, BATF, DNase1
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K27ac
		skin (NHEK)	DNase1, H3K4me1, H3K4me2, H3K4me3, H3K36me3
		NHLF	DNase1, H3K4me3, H3K27ac
		Osteobl	H3K27ac, H3K4me3, H3K4me2, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, Jund, Nrsf, H3K27ac, Cjun
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K4me3
		endothelium (HUVEC)	Cjun, Max, DNase1, H3K4me2, H3K9ac, H3K4me1, H3K27ac, PolII
		A549	DNase1, H3K4me2, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PDCD1LG2	upstream gene variant	4748	ENST00000397747	NM_025239.3	ENSP00000380855	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PDCD1LG2	ENST00000397747	NM_025239.3	ENSP00000380855	5

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
PDCD1LG2	ENST00000397747	NM_025239.3	ENSP00000380855	1