SNiPAcard

Block annotations

Block info

genomic range	chr11:14,038,621-14,121,039
block size	82,419 bp
variant count	36 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.230 [-2.046 – 1.303]	**gene(s) hit or close-by**	SPON1
phastCons	μ = 0.054 [0 – 0.999]	**eQTL gene(s)**	SPON1
GERP++	μ = 0.078 [-5.51 – 3.28]	**potentially regulated gene(s)**	COPB1, RP11-21L19.1, RP11-98J9.1, RP11-98J9.3, RRAS2, SPON1
CADD score	μ = 4.253 [0.29 – 19.18]	**disease gene(s)**	RRAS2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
RRAS2	OVARIAN CANCEROVARIAN CANCER, EPITHELIAL, INCLUDED	OMIM	MIM:167000

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SPON1	ENST00000576479	ILMN_1791890	liver	$<2.2\times10^{-16}$ (p-value)	Innocenti et al.	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000098968	1	ENCP00000011250	SPON1
ENCE00000098923	1	ENCP00000011259	COPB1
		ENCP00000011252	RRAS2
		ENCP00000011246	RP11-98J9.3
		ENCP00000011251	RP11-21L19.1
		ENCP00000011245	RP11-98J9.1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000558962 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3, Egr1
		skin (NHDF-AD)	DNase1
		cervix (HeLa-S3)	H3K27me3
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	DNase1
		lung (IMR90)	DNase1

		nervous (NH-A)	DNase1
ENSR00001568447 (promoter flanking region)	1	HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		cervix (HeLa-S3)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K27me3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SPON1	ENST00000576479	NM_006108.3	ENSP00000460236	36