SNiPAcard

Block annotations

Block info

genomic range	chr6:169,562,593-169,598,082
block size	35,490 bp
variant count	24 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.515 [-2.942 – 0.962]	**gene(s) hit or close-by**	RP11-417E7.1, RP11-417E7.2
phastCons	μ = 0.035 [0 – 0.384]	**eQTL gene(s)**	RP11-417E7.2
GERP++	μ = -0.601 [-3.58 – 1.47]	**potentially regulated gene(s)**	RP1-137D17.1, RP11-417E7.1, RP3-495K2.3, THBS2, WDR27, XXyac-YX65C7_A.2
CADD score	μ = 2.299 [0.198 – 6.035]	**disease gene(s)**	THBS2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
THBS2	INTERVERTEBRAL DISC DISEASE	OMIM	MIM:603932

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RP11-417E7.2	?	ENSG00000261039	aorta	9.21×10^{-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)
ENCE00000470093	1	ENCP00000050698	RP1-137D17.1
		ENCP00000050686	RP3-495K2.3
		ENCP00000050695	THBS2
ENCE00000470094	1	ENCP00000050689	RP11-417E7.1
		ENCP00000050698	RP1-137D17.1
		ENCP00000050686	RP3-495K2.3
		ENCP00000050695	THBS2
ENCE00000470180	1	ENCP00000050690	XXyac-YX65C7_A.2
ENCE00000470279	1	ENCP00000050700	WDR27

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001233874 (promoter flanking region)	1	NHLF	H3K36me3, H3K27ac, DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H2AZ, H3K4me2, DNase1
		Osteobl	H3K27ac, H3K36me3

		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K4me3, H3K4me1, DNase1, H3K9ac, H3K27ac, H3K4me2
		muscle (HSMM)	H3K36me3, H3K27ac, DNase1
		breast (HMEC)	DNase1
		cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	Cjun, DNase1
		liver (HepG2)	DNase1, H3K27me3
		lung (IMR90)	H4K91ac, H3K4ac, H3K36me3, H4K5ac, H3K27ac, DNase1
		nervous (NH-A)	DNase1, H3K9ac, H3K27ac
		A549	DNase1
		skin (NHEK)	H3K4me1, DNase1
ENSR00001502797 (promoter flanking region)	2	NHLF	H3K36me3, H3K27ac, H3K9ac, DNase1
		HSMMtube	DNase1, H3K27ac, H3K9ac
		Osteobl	H3K27ac, H3K36me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K27ac
		liver (HepG2)	H3K27me3
		lung (IMR90)	H4K91ac, H2BK120ac, H3K4ac, H3K36me3, H4K5ac, H3K27ac, DNase1
		nervous (NH-A)	H3K27ac, DNase1, H3K9ac
ENSR00001502798 (promoter flanking region)	5	NHLF	DNase1, H3K27ac
		embryonic stem cell (H1ESC)	H3K36me3, DNase1
		HSMMtube	DNase1, H3K27ac
		Osteobl	H3K27ac, H3K36me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K27ac, H3K9ac, DNase1, H3K4me1
		muscle (HSMM)	DNase1, H3K36me3, H3K27ac
		liver (HepG2)	H3K27me3
		nervous (NH-A)	DNase1, H3K9ac
ENSR00001502800 (promoter flanking region)	1	cervix (HeLa-S3)	H3K27me3
		HSMMtube	DNase1, H3K36me3, H3K27ac
		liver (HepG2)	H3K27me3
		blood (K562)	H3K27me3, Gabp, H2AZ
		muscle (HSMM)	DNase1, H3K36me3, H3K27ac
ENSR00001233884 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1, H3K27ac, H3K9ac, H3K4me1
		Osteobl	H3K27ac
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K27ac, DNase1, H3K4me1
		muscle (HSMM)	DNase1, H3K9ac, H3K27ac
		cervix (HeLa-S3)	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
ENSR00001502806 (open chromatin region)	2	muscle (HSMM)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-417E7.1	upstream gene variant	197	ENST00000449466	?	?	1
RP11-417E7.2	downstream gene variant, upstream gene variant	201	ENST00000564830	?	?	10

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
RP11-417E7.1	ENST00000439703	?	?	1
RP11-417E7.2	ENST00000564830	?	?	3

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-417E7.2	ENST00000564830	?	1