SNiPAcard

Block annotations

Block info

genomic range	chr10:101,798,405-101,814,910
block size	16,506 bp
variant count	11 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.652 [-2.147 – 0.21]	**gene(s) hit or close-by**	CPN1
phastCons	μ = 0.019 [0 – 0.152]	**eQTL gene(s)**	HIF1AN
GERP++	μ = 0.007 [-2.16 – 1.68]	**potentially regulated gene(s)**	–
CADD score	μ = 2.581 [0.293 – 6.817]	**disease gene(s)**	CPN1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CPN1	CARBOXYPEPTIDASE N DEFICIENCY	OMIM	MIM:212070

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HIF1AN	?	ENSG00000166135	transverse colon	4.80×10^{-6} (p-value)	GTEx Portal V6	3

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001426512 (enhancer)	1	embryonic stem cell (H1ESC)	DNase1
		liver (HepG2)	H3K4me1, H3K4me2, HNF4A, H3K27ac
		blood (GM12878)	H2AZ, H3K4me1
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
ENSR00001426513 (CTCF binding site)	2	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, DNase1
		blood (K562)	H3K27me3, Rad21, CTCF
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF
		liver (HepG2)	H3K36me3, Rad21, CTCF
		lung (IMR90)	CTCF, DNase1
		blood (GM12878)	DNase1, H2AZ, Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	DNase1, CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	CTCF, DNase1

		endothelium (HUVEC)	H3K36me3, CTCF
		A549	DNase1, CTCF
ENSR00000360384 (promoter flanking region)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, DNase1
		blood (K562)	H3K27me3, Rad21, CTCF
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF, DNase1
		liver (HepG2)	CTCF, H3K36me3, Rad21
		blood (GM12878)	DNase1, H2AZ, Rad21, CTCF
		lung (IMR90)	DNase1, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF, H3K4me2, H3K9ac, DNase1, H3K27ac
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF, H3K27ac
		cervix (HeLa-S3)	H3K27ac, CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, DNase1
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CPN1	downstream gene variant	2217	ENST00000370418	NM_001308.2	ENSP00000359446	2
CPN1	downstream gene variant	2062	ENST00000441382	?	ENSP00000410895	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CPN1	ENST00000370418	NM_001308.2	ENSP00000359446	9
CPN1	ENST00000441382	?	ENSP00000410895	7