Locus 222 (rs4803378)

SNiPAcard

Block annotations

Block info

genomic range	chr19:41,333,441-41,392,490
block size	59,050 bp
variant count	19 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.487 [-1.433 – 1.377]	gene(s) hit or close-by	CTC-490E21.10, CTC-490E21.11, CTC-490E21.12, CYP2A6, CYP2A7, CYP2F2P, CYP2G1P
phastCons	μ = 0.050 [0 – 0.484]	eQTL gene(s)	CYP2B7P
GERP++	μ = -0.607 [-4.56 – 0.89]	potentially regulated gene(s)	–
CADD score	μ = 2.379 [0.27 – 8.637]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CYP2B7P	?	ENSG00000256612	lung	4.19×10^{-6} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001419215 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1, H3K27me3
		Osteobl	H3K4me2, H3K27me3
		blood (K562)	Nrsf, H3K4me3, H3K4me2, H3K4me1, H2AZ, USF1, DNase1, Gata2, H3K27ac, Max, H3K9ac
		skin (NHDF-AD)	DNase1
		breast (HMEC)	DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1, Max, Cmyc
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	USF1, H3K4me1, HNF4A, FOXA1, DNase1
		lung (IMR90)	DNase1, H3K36me3
		A549	H3K27me3, DNase1
		nervous (NH-A)	DNase1
ENSR00001648169 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K27me3, DNase1
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTC-490E21.10	downstream gene variant, upstream gene variant	396	ENST00000596135	?	?	7
CTC-490E21.11	upstream gene variant, downstream gene variant	475	ENST00000623128	?	?	11
CYP2A6	downstream gene variant, upstream gene variant	30	ENST00000599960	?	?	7
CYP2A6	downstream gene variant	1024	ENST00000301141	NM_000762.5	ENSP00000301141	3

CYP2A6	downstream gene variant	91	ENST00000600495	?	ENSP00000472905	4
CYP2A6	downstream gene variant	40	ENST00000596719	?	?	2
CYP2A6	downstream gene variant, upstream gene variant	930	ENST00000610301	?	ENSP00000477899	5
CYP2A7	upstream gene variant	388	ENST00000301146	NM_000764.2	ENSP00000301146	3
CYP2A7	upstream gene variant	3050	ENST00000598264	?	?	1
CYP2A7	upstream gene variant	388	ENST00000291764	NM_030589.2	ENSP00000291764	3
CYP2A7	upstream gene variant	909	ENST00000594332	?	ENSP00000470933	3
CYP2F2P	upstream gene variant	764	ENST00000437853	?	?	3
CYP2G1P	upstream gene variant	4241	ENST00000252909	?	?	2
CYP2G1P	upstream gene variant	4634	ENST00000597833	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTC-490E21.10	ENST00000596135	?	?	4
CTC-490E21.12	ENST00000601627	?	ENSP00000469533	19
CYP2A6	ENST00000596719	?	?	3
CYP2A6	ENST00000301141	NM_000762.5	ENSP00000301141	4
CYP2A6	ENST00000610301	?	ENSP00000477899	4
CYP2A6	ENST00000600495	?	ENSP00000472905	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CYP2A6	ENST00000596719	?	1