SNiPAcard

Block annotations

Block info

genomic range	chr6:161,201,658-161,276,911
block size	75,254 bp
variant count	36 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.186 [-3.076 – 0.897]	**gene(s) hit or close-by**	RP1-81D8.6
phastCons	μ = 0.067 [0 – 0.519]	**eQTL gene(s)**	–
GERP++	μ = -0.218 [-1.83 – 0.924]	**potentially regulated gene(s)**	AGPAT4, RP11-235G24.2
CADD score	μ = 3.584 [0.031 – 10.36]	**disease gene(s)**	–

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000468570	1	ENCP00000050568	AGPAT4
		ENCP00000050555	RP11-235G24.2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001232628 (open chromatin region)	1	blood (K562)	DNase1
ENSR00001232632 (CTCF binding site)	1	embryonic stem cell (H1ESC)	CTCF, Rad21, Yy1, DNase1
		liver (HepG2)	CTCF
		HSMMtube	DNase1, H3K27me3
		blood (GM12878)	H3K27ac, Rad21, EBF1
		Osteobl	H3K4me2
		blood (K562)	H2AZ, CTCF
		skin (NHEK)	H3K27ac, H3K4me3
ENSR00001232633 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1
		blood (GM12878)	H3K27ac
		blood (K562)	PolII, DNase1, H3K27ac, H3K9ac, H2AZ, H3K4me3, H3K4me2
		skin (NHEK)	H3K27ac
ENSR00001232639 (open chromatin region)	1	A549	H3K27me3
		blood (K562)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP1-81D8.6	downstream gene variant	2404	ENST00000619095	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
RP1-81D8.6	ENST00000619095	?	?	3