SNiPAcard

Block annotations

Block info

genomic range	chr3:50,562,042-51,196,423
block size	634,382 bp
variant count	12 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.302 [-2.21 – 0.611]	**gene(s) hit or close-by**	DOCK3, MAPKAPK3, ST13P14
phastCons	μ = 0.054 [0 – 0.294]	**eQTL gene(s)**	C3orf62, HYAL3, MAPKAPK3, NAT6, RPL29, USP4, WDR6
GERP++	μ = -0.348 [-8.17 – 2]	**potentially regulated gene(s)**	–
CADD score	μ = 4.951 [0.255 – 7.854]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HYAL3	?	ENSG00000186792	prostate	5.22×10^{-6} (p-value)	GTEx Portal V6	1
MAPKAPK3	ENST00000446044	ILMN_1732452	blood	9.73×10^{-5} (p-value)	Westra et al.	1
MAPKAPK3	ENST00000357955					
MAPKAPK3	ENST00000621469					
NAT6	?	ENSG00000243477	coronary artery	3.52×10^{-6} (p-value)	GTEx Portal V6	3
WDR6	ENST00000615452	ILMN_1669484	monocyte	2.08×10^{-6} (p-value)	Fairfax et al.	1
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000395474					
WDR6	ENST00000610967					
WDR6	ENST00000608424					
WDR6	ENST00000492780					
WDR6	ENST00000420783					
USP4	ENST00000351842	ILMN_1798712	monocyte	2.75×10^{-7} (p-value)	Fairfax et al.	1
USP4	ENST00000483212					
C3orf62	ENST00000343010					
USP4	ENST00000485450					
USP4	ENST00000351842	ILMN_1798712	monocyte	1.00×10^{-17} (p-value)	Zeller et al.	1
USP4	ENST00000483212					
USP4	ENST00000485450					
C3orf62	ENST00000343010					
RPL29	?	ENSG00000162244	liver	6.15×10^{-6} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001363979 (promoter flanking region)	1	NHLF	DNase1
		HSMMtube	DNase1
		Osteobl	H3K27ac

		blood (K562)	H3K36me3
		blood (DND-41)	H4K20me1, H3K36me3
		skin (NHDF-AD)	H3K4me1, DNase1
		muscle (HSMM)	DNase1, H3K4me1, H3K27ac, H3K36me3
		breast (HMEC)	H3K36me3
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1, H3K36me3, DNase1
ENSR00001364012 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		lung (IMR90)	CTCF
		blood (GM12878)	Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ST13P14	upstream gene variant	1868	ENST00000474095	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
DOCK3	ENST00000266037	NM_004947.4	ENSP00000266037	9
MAPKAPK3	ENST00000457064	?	ENSP00000402045	1
MAPKAPK3	ENST00000446044	NM_001243926.1	ENSP00000396467	1
MAPKAPK3	ENST00000357955	?	ENSP00000350639	1
MAPKAPK3	ENST00000430409	?	ENSP00000410970	1
MAPKAPK3	ENST00000621469	NM_001243925.1, NM_004635.4	ENSP00000478922	1