SNiPAcard

Block annotations

Block info

genomic range	chr4:102,710,688-102,756,731
block size	46,044 bp
variant count	49 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.684 [-3.212 – 0.908]	**gene(s) hit or close-by**	BANK1
phastCons	μ = 0.017 [0 – 0.16]	**eQTL gene(s)**	BANK1
GERP++	μ = -0.306 [-4.46 – 2.03]	**potentially regulated gene(s)**	–
CADD score	μ = 2.185 [0.066 – 7.844]	**disease gene(s)**	BANK1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link	
BANK1	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700	OMIM®

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
BANK1	ENST00000428908	ILMN_1661646	blood	2.82×10^{-16} (p-value)	MuTHER consortium	14
BANK1	ENST00000504592					
BANK1	ENST00000444316					
BANK1	ENST00000322953					
BANK1	ENST00000504403					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001433620 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	DNase1, H3K4me1
		HSMMtube	H3K27me3
		Osteobl	H3K4me2
		A549	H3K4me3, H3K4me2, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
BANK1	upstream gene variant	1076	ENST00000322953	NM_017935.4	ENSP00000320509	1
BANK1	upstream gene variant	1199	ENST00000508653	?	ENSP00000422314	1
BANK1	upstream gene variant	1076	ENST00000428908	NM_001127507.2	ENSP00000412748	1
BANK1	upstream gene variant	4428	ENST00000444316	NM_001083907.2	ENSP00000388817	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
BANK1	ENST00000322953	NM_017935.4	ENSP00000320509	48
BANK1	ENST00000508653	?	ENSP00000422314	48
BANK1	ENST00000504592	?	ENSP00000421443	49
BANK1	ENST00000428908	NM_001127507.2	ENSP00000412748	48
BANK1	ENST00000444316	NM_001083907.2	ENSP00000388817	1