Locus 235 (rs8283)

SNiPAcard

Block annotations

Block info

genomic range	chr6:32,083,300-32,083,300
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.226	gene(s) hit or close-by	ATF6B, TNXB
phastCons	0	eQTL gene(s)	ATF6B, FKBPL, HLA-DRB6, MICB, STK19B, TAP2, TNXA, XXbac-BPG246D15.9
GERP++	3.07	potentially regulated gene(s)	–
CADD score	9.99	disease gene(s)	TAP2, TNXB

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TAP2	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571
TAP2	Immunodeficiency by defective expression of HLA class 1	OrphaNet	OrphaNet:34592
TNXB	Ehlers-Danlos syndrome due to tenascin-X deficiency	OrphaNet	OrphaNet:230839
TNXB	EHLERS-DANLOS SYNDROME, HYPERMOBILITY TYPE	OrphaNet	OrphaNet:285

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TAP2	ENST00000620123	ILMN_1759250	blood	5.92×10^{-5} (p-value)	MuTHER consortium	1
TAP2	ENST00000374899					
XXbac-BPG246D15.9	ENST00000452392					
TAP2	ENST00000464100					
TAP2	ENST00000374897					
MICB	ENST00000538442	ILMN_1708006	skin	2.74×10^{-5} (p-value)	MuTHER consortium	1
MICB	ENST00000399150					
MICB	ENST00000252229					
STK19B	?	ENSG00000250535	muscularis mucosae	2.36×10^{-6} (p-value)	GTEx Portal V6	1
TNXA	?	ENSG00000248290	muscularis mucosae	2.63×10^{-6} (p-value)	GTEx Portal V6	1
TNXA	?	ENSG00000248290	visceral adipocytes	5.27×10^{-7} (p-value)	GTEx Portal V6	1
TNXA	?	ENSG00000248290	tibial nerve	2.04×10^{-7} (p-value)	GTEx Portal V6	1
STK19B	?	ENSG00000250535	esophagus mucosa	4.77×10^{-6} (p-value)	GTEx Portal V6	1
STK19B	?	ENSG00000250535	thyroid	9.02×10^{-10} (p-value)	GTEx Portal V6	1
HLA-DRB6	?	ENSG00000229391	skeletal muscle	1.15×10^{-7} (p-value)	GTEx Portal V6	1
STK19B	?	ENSG00000250535	cerebellum	3.12×10^{-6} (p-value)	GTEx Portal V6	1
ATF6B	?	ENSG00000213676	blood	8.78×10^{-3} (p-value)	SeeQTL DB (HapMap)	1

ATF6B	?	ENSG00000213676	blood	8.78×10-5 (q-value)	SeeQTL DB (HapMap)	1
STK19B	?	ENSG00000250535	sun exposed skin	4.28×10-7 (p-value)	GTEx Portal V6	1
FKBPL	?	ENSG00000204315	sun exposed skin	4.94×10-8 (p-value)	GTEx Portal V6	1
HLA-DRB6	?	ENSG00000229391	sun exposed skin	1.81×10-8 (p-value)	GTEx Portal V6	1
HLA-DRB6	?	ENSG00000229391	subcutaneous adipocytes	3.42×10-7 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
ATF6B	1	ENST00000375201 ENST00000375203 ENST00000494022	hsa-miR-371a-5p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ATF6B	downstream gene variant	1185	ENST00000453203	?	ENSP00000393419	1
ATF6B	downstream gene variant	3254	ENST00000475705	?	?	1
ATF6B	downstream gene variant	4986	ENST00000485314	?	?	1
ATF6B	downstream gene variant	4919	ENST00000495579	?	?	1
ATF6B	downstream gene variant	3308	ENST00000492342	?	?	1
TNXB	upstream gene variant	189	ENST00000442721	?	ENSP00000389946	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ATF6B	ENST00000494022	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ATF6B	ENST00000375201	NM_001136153.1	ENSP00000364347	1
ATF6B	ENST00000375203	NM_004381.4	ENSP00000364349	1