Locus 285 (rs2523710)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,450,909-31,450,909
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.625	gene(s) hit or close-by	–
phastCons	0.005	eQTL gene(s)	AGER, DXO, HLA-B, LST1, MICB, PSMB8
GERP++	0.207	potentially regulated gene(s)	–
CADD score	5.078	disease gene(s)	PSMB8, HLA-B

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
PSMB8	AUTOINFLAMMATION, LIPODYSTROPHY, AND DERMATOSIS SYNDROME	OMIM	MIM:256040
PSMB8	Nakajo syndrome	DECIPHER	MIM:256040
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426
HLA-B	Behcet disease	OrphaNet	OrphaNet:117
PSMB8	CANDLE syndrome	OrphaNet	OrphaNet:325004
PSMB8	JMP syndrome	OrphaNet	OrphaNet:324999
PSMB8	Nakajo-Nishimura syndrome	OrphaNet	OrphaNet:2615

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LST1	ENST00000339530	ILMN_1718936	blood	1.37×10-6 (p-value)	MuTHER consortium	1
LST1	ENST00000464526					
LST1	ENST00000303757					
LST1	ENST00000419073					
LST1	ENST00000376093					
LST1	ENST00000396112					
LST1	ENST00000490742					
LST1	ENST00000433492					
LST1	ENST00000438075					
LST1	ENST00000464044					
LST1	ENST00000396101					
LST1	ENST00000376099	ILMN_1688373	blood	4.99×10-5 (p-value)	MuTHER consortium	1
LST1	ENST00000464526					
LST1	ENST00000339530					
LST1	ENST00000419073					
LST1	ENST00000376110					
LST1	ENST00000376086					
LST1	ENST00000433492					
LST1	ENST00000438075					

LST1	ENST00000464044					
LST1	ENST00000376090					
LST1	ENST00000396101					
LST1	ENST00000376092					
LST1	ENST00000303757					
LST1	ENST00000376093					
LST1	ENST00000376089					
LST1	ENST00000396112					
LST1	ENST00000418507					
LST1	ENST00000376096					
LST1	ENST00000490742					
LST1	ENST00000211921					
LST1	ENST00000460834					
MICB	ENST00000538442	ILMN_1708006	skin	4.40×10-8 (p-value)	MuTHER consortium	1
MICB	ENST00000399150					
MICB	ENST00000252229					
MICB	?	ENSG00000204516	lung	3.70×10-6 (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	transformed fibroblasts	3.43×10-7 (p-value)	GTEx Portal V6	1
HLA-B	?	ENSG00000234745	tibial artery	3.49×10-6 (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	tibial artery	8.97×10-9 (p-value)	GTEx Portal V6	1
DXO	ENST00000474587	ILMN_1726990	monocyte	4.95×10-7 (p-value)	Fairfax et al.	1
DXO	ENST00000473976					
DXO	ENST00000491327					
DXO	ENST00000480240					
DXO	ENST00000477826					
DXO	ENST00000498357					
DXO	ENST00000337523					
DXO	ENST00000460058					
DXO	ENST00000492946					
DXO	ENST00000375349					
DXO	ENST00000375356					
DXO	ENST00000487914					
DXO	ENST00000478221					
DXO	ENST00000485557					
AGER	ENST00000375070	ILMN_2257015	monocyte	2.29×10-4 (p-value)	Fairfax et al.	1
AGER	ENST00000375056					
AGER	ENST00000450110					
AGER	ENST00000438221					
AGER	ENST00000375065					
AGER	ENST00000375076					
AGER	ENST00000473619					
AGER	ENST00000484849					
AGER	ENST00000488669					
AGER	ENST00000375069					
AGER	ENST00000375055					
PSMB8	ENST00000490613	ILMN_1747195	monocyte	1.27×10-5 (p-value)	Fairfax et al.	1
PSMB8	ENST00000374881					
PSMB8	ENST00000484003					
PSMB8	ENST00000395339					
PSMB8	ENST00000374882					
MICB	?	ENSG00000204516	esophagus mucosa	1.80×10-10 (p-value)	GTEx Portal V6	1

MICB	?	ENSG00000204516	cerebellar hemisphere	1.90×10-7 (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	sun exposed skin	1.49×10-6 (p-value)	GTEx Portal V6	1