

Block annotations

Block info









genomic range	chr6:31,450,909-31,450,909 <i>e!</i>
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 <i>e!</i> , DXO <i>e!</i> , HLA-B <i>e!</i> , LST1 <i>e!</i> , MICB <i>e!</i> , PSMB8 <i>e!</i>
GERP++	0.207	potentially regulated gene(s)	-
CADD score	5.078	disease gene(s)	PSMB8 <i>e!</i> , HLA-B <i>e!</i>



Trait annotations

Disease gene annotation



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

Direct effect on regulation

cis-eQTL

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

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

MICB <i>e!</i> ?	ENSG00000204516 <i>e!</i>	cerebellar hemisphere	1.90×10 ⁻⁷ (p-value)	GTEX Portal V6 	1
MICB <i>e!</i> ?	ENSG00000204516 <i>e!</i>	sun exposed skin	1.49×10 ⁻⁶ (p-value)	GTEX Portal V6 	1
