Locus 238 (rs12520053)

SNiPAcard

Block annotations

Block info

genomic range	chr5:96,163,905-96,171,829
block size	7,925 bp
variant count	10 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.824 [-3.443 – 0.636]	**gene(s) hit or close-by**	CTD-2260A17.2
phastCons	μ = 0.005 [0 – 0.021]	**eQTL gene(s)**	CAST, ERAP2, LNPEP, RP11-254I22.3
GERP++	μ = -0.669 [-4.04 – 0.895]	**potentially regulated gene(s)**	LIX1
CADD score	μ = 3.809 [1.094 – 7.845]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ERAP2	ENST00000513084	ILMN_1743145	blood	6.15×10^{-5} (p-value)	MuTHER consortium	7
ERAP2	ENST00000512869					
ERAP2	ENST00000379904					
ERAP2	ENST00000437043					
LNPEP	ENST00000395770	ILMN_1814737	blood	4.47×10^{-5} (p-value)	Westra et al.	7
LNPEP	ENST00000231368					
CAST	ENST00000511782	ILMN_1783627	blood	4.26×10^{-6} (p-value)	Westra et al.	7
CAST	ENST00000511049					
CAST	ENST00000395813					
CAST	ENST00000421689					
CAST	ENST00000506811					
CAST	ENST00000510156					
CAST	ENST00000507487					
CAST	ENST00000341926					
CAST	ENST00000508117					
CAST	ENST00000508830					
CAST	ENST00000338252					
CAST	ENST00000505143					
CAST	ENST00000514055					
RP11-254I22.3	?	ENSG00000249746	testis	1.12×10^{-5} (p-value)	GTEx Portal V6	7
ERAP2	?	ENSG00000164308	thyroid	2.63×10^{-5} (p-value)	GTEx Portal V6	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000417175	1	ENCP00000044982	LIX1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTD-2260A17.2	ENST00000504056	?	?	10
CTD-2260A17.2	ENST00000502262	?	?	10
CTD-2260A17.2	ENST00000501338	?	?	10