

SNiPACard

Block annotations

Block info

genomic range	chr5:96,163,905-96,171,829 <i>e!</i>
block size	7,925 bp
variant count	10 variants

Basic features

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

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)					
ERAP2 <i>e!</i>	ENST00000513084 <i>e!</i>	ILMN_1743145 <i>e!</i>	blood	6.15×10 ⁻⁵ (p-value)	MuTHER consortium 	7					
ERAP2 <i>e!</i>	ENST00000512869 <i>e!</i>										
ERAP2 <i>e!</i>	ENST00000379904 <i>e!</i>										
ERAP2 <i>e!</i>	ENST00000437043 <i>e!</i>										
LNPEP <i>e!</i>	ENST00000395770 <i>e!</i>	ILMN_1814737 <i>e!</i>	blood	4.47×10 ⁻⁵ (p-value)	Westra et al. 	7					
LNPEP <i>e!</i>	ENST00000231368 <i>e!</i>										
CAST <i>e!</i>	ENST00000511782 <i>e!</i>	ILMN_1783627 <i>e!</i>	blood	4.26×10 ⁻⁶ (p-value)	Westra et al. 	7					
CAST <i>e!</i>	ENST00000511049 <i>e!</i>										
CAST <i>e!</i>	ENST00000395813 <i>e!</i>										
CAST <i>e!</i>	ENST00000421689 <i>e!</i>										
CAST <i>e!</i>	ENST00000506811 <i>e!</i>										
CAST <i>e!</i>	ENST00000510156 <i>e!</i>										
CAST <i>e!</i>	ENST00000507487 <i>e!</i>										
CAST <i>e!</i>	ENST00000341926 <i>e!</i>										
CAST <i>e!</i>	ENST00000508117 <i>e!</i>										
CAST <i>e!</i>	ENST00000508830 <i>e!</i>										
CAST <i>e!</i>	ENST00000338252 <i>e!</i>										
CAST <i>e!</i>	ENST00000505143 <i>e!</i>										
CAST <i>e!</i>	ENST00000514055 <i>e!</i>										
RP11-254I22.3 <i>e!</i>	?						ENSG00000249746 <i>e!</i>	testis	1.12×10 ⁻⁵ (p-value)	GTEX Portal V6 	7
ERAP2 <i>e!</i>	?						ENSG00000164308 <i>e!</i>	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 <i>e!</i>	1	ENCP00000044982	LIX1 <i>e!</i>

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTD-2260A17.2 <i>e!</i>	ENST00000504056 <i>e!</i>	?	?	10
CTD-2260A17.2 <i>e!</i>	ENST00000502262 <i>e!</i>	?	?	10
CTD-2260A17.2 <i>e!</i>	ENST00000501338 <i>e!</i>	?	?	10

