Locus 163 (rs12938)

SNiPAcard

Block annotations

Block info

genomic range	chr1:169,653,795-169,666,871
block size	13,077 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.008 [-2.659 – 0.256]	**gene(s) hit or close-by**	C1orf112, SELL
phastCons	μ = 0.040 [0 – 0.199]	**eQTL gene(s)**	C1orf112, RN7SL333P, SELL
GERP++	μ = -1.263 [-6.06 – 1.21]	**potentially regulated gene(s)**	–
CADD score	μ = 3.621 [0.664 – 7.649]	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Hemoglobin a, glycosylated	6.33×10^{-5}	dbGaP	pha002881	dbGaP	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
C1orf112	?	ENSG00000000460	cerebellar hemisphere	6.03×10^{-6} (p-value)	GTEx Portal V6	5
RN7SL333P	?	ENSG00000239494	tibial nerve	4.85×10^{-6} (p-value)	GTEx Portal V6	1
C1orf112	?	ENSG00000000460	tibial nerve	4.94×10^{-6} (p-value)	GTEx Portal V6	4
C1orf112	?	ENSG00000000460	atrial appendage	3.68×10^{-8} (p-value)	GTEx Portal V6	5
RN7SL333P	?	ENSG00000239494	tibial artery	3.30×10^{-6} (p-value)	GTEx Portal V6	5
C1orf112	?	ENSG00000000460	thyroid	7.29×10^{-7} (p-value)	GTEx Portal V6	5
SELL	ENST00000236147	ILMN_1724422	blood	1.85×10^{-13} (p-value)	MuTHER consortium	3
SELL	ENST00000497295					
SELL	?	ENSG00000188404	blood	5.74×10^{-7} (p-value)	GTEx Portal V6	4
SELL	ENST00000497295	ILMN_1724422	monocyte	2.11×10^{-15} (p-value)	Fairfax et al.	1
SELL	ENST00000236147		b-cell	1.57×10^{-8} (p-value)	Fairfax et al.	1
SELL	?	ENSG00000188404	blood	5.79×10^{-5} (q-value)	SeeQTL DB (HapMap)	2

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
SELL	ENST00000236147	ILMN_1724422	chr1	monocyte	3.58×10^{-29} (p-value)	Zeller et al.	1
SELL	ENST00000497295						

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000547356 (promoter flanking region)	3	cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K4me3, H3K36me3
		embryonic stem cell (H1ESC)	H3K27me3
		HSMMtube	H3K27me3
		blood (GM12878)	H3K27ac, H3K4me1
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SELL	downstream gene variant	41	ENST00000497295	?	?	1
SELL	downstream gene variant	3765	ENST00000463108	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C1orf112	ENST00000498289	?	?	5
SELL	ENST00000236147	NM_000655.4	ENSP00000236147	4
SELL	ENST00000497295	?	?	3

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SELL	ENST00000236147	NM_000655.4	ENSP00000236147	1