SNiPAcard

Block annotations

Block info

genomic range	chr19:51,540,143-51,546,893
block size	6,751 bp
variant count	17 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.622 [-3.722 – 0.941]	**gene(s) hit or close-by**	CTC-518B2.10, CTC-518B2.9, KLK12
phastCons	μ = 0.031 [0 – 0.142]	**eQTL gene(s)**	CTC-518B2.9, KLK12
GERP++	μ = -0.100 [-1.91 – 0.889]	**potentially regulated gene(s)**	–
CADD score	μ = 2.159 [0.022 – 12.37]	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
dihomo-linolenate (20:3n3 or n6)	6.95×10^{-5}	Metabolomics GWAS Server	24816252	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CTC-518B2.9	?	ENSG00000268906	esophagus mucosa	5.39×10^{-13} (p-value)	GTEx Portal V6	16
KLK12	?	ENSG00000186474	esophagus mucosa	1.36×10^{-16} (p-value)	GTEx Portal V6	16
KLK12	?	ENSG00000186474	terminal ileum	1.45×10^{-6} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTC-518B2.10	upstream gene variant	170	ENST00000601280	?	?	11
CTC-518B2.9	downstream gene variant	2551	ENST00000594910	?	?	6
KLK12	upstream gene variant	2024	ENST00000530943	?	ENSP00000434339	6
KLK12	upstream gene variant	2024	ENST00000529888	NM_145895.1	ENSP00000434036	6
KLK12	upstream gene variant	1994	ENST00000250351	NM_019598.2	ENSP00000250351	6
KLK12	upstream gene variant	1994	ENST00000319590	NM_145894.1	ENSP00000324181	6
KLK12	upstream gene variant	2024	ENST00000531374	?	ENSP00000436121	6
KLK12	upstream gene variant	1656	ENST00000526824	?	ENSP00000434604	6
KLK12	upstream gene variant	2145	ENST00000525263	?	ENSP00000436458	6