

Block annotations

Block info


genomic range	chr19:51,802,140-51,802,650 <i>e!</i>
block size	511 bp
variant count	2 variants

Basic features





Conservation/deleteriousness		Linked genes	
phyloP	$\mu = -1.125$ [-1.962 – -0.287]	gene(s) hit or close-by	-
phastCons	$\mu = 0.003$ [0 – 0.005]	eQTL gene(s)	CD33 <i>e!</i> , ETFB <i>e!</i>
GERP++	$\mu = -0.726$ [-1.13 – -0.323]	potentially regulated gene(s)	-
CADD score	$\mu = 1.387$ [1.232 – 1.542]	disease gene(s)	ETFB <i>e!</i>

Trait annotations

Variant association



trait	min(p-value)	source DB	source entry/link	variant(s)
oleate (18:1n9)	3.00×10 ⁻⁵	Metabolomics GWAS Server	24816252 	2

Disease gene annotation

gene	trait	source DB	source entry/link
ETFB <i>e!</i>	MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY	OMIM	MIM:231680 
ETFB <i>e!</i>	glutaric aciduria type 2B	DECIPHER	MIM:231680 
ETFB <i>e!</i>	Multiple acyl-CoA dehydrogenation deficiency, mild type	OrphaNet	OrphaNet:394532 
ETFB <i>e!</i>	Multiple acyl-CoA dehydrogenation deficiency, severe neonatal type	OrphaNet	OrphaNet:394529 

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CD33 <i>e!</i>	ENST00000262262 <i>e!</i>	ILMN_1747622 <i>e!</i>	blood	6.28×10 ⁻⁷ (p-value)	Westra et al. 	1
CD33 <i>e!</i>	ENST00000600557 <i>e!</i>					
CD33 <i>e!</i>	ENST00000601785 <i>e!</i>					
CD33 <i>e!</i>	ENST00000421133 <i>e!</i>					
CD33 <i>e!</i>	ENST00000598473 <i>e!</i>					
ETFB <i>e!</i>	ENST00000596253 <i>e!</i>	ILMN_1729374 <i>e!</i>	blood	8.37×10 ⁻⁵ (p-value)	Westra et al. 	2
ETFB <i>e!</i>	ENST00000593992 <i>e!</i>					
ETFB <i>e!</i>	ENST00000309244 <i>e!</i>					

