Locus 335 (rs10860794)

SNiPAcard

Block annotations

Block info

genomic range	chr12:102,216,673-102,225,751
block size	9,079 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.568 [-1.416 – 0.689]	gene(s) hit or close-by	GNPTAB, RNA5SP369
phastCons	μ = 0.002 [0 – 0.014]	eQTL gene(s)	CHPT1, DRAM1
GERP++	μ = 0.015 [-1.61 – 3.03]	potentially regulated gene(s)	CCDC53
CADD score	μ = 3.994 [0.729 – 7.826]	disease gene(s)	GNPTAB

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
GNPTAB	mucolipidosis type III complementation group A (MLIIIA)	DECIPHER	MIM:252600
GNPTAB	mucolipidosis type II (MLII)	DECIPHER	MIM:252500
GNPTAB	MUCOLIPIDOSIS II ALPHA/BETA	OMIM	MIM:252500
GNPTAB	MUCOLIPIDOSIS III ALPHA/BETA	OMIM	MIM:252600
GNPTAB	Mucolipidosis type 3	OrphaNet	OrphaNet:577
GNPTAB	Mucolipidosis type 2	OrphaNet	OrphaNet:576

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CHPT1	?	ENSG00000111666	muscularis mucosae	3.15×10-6 (p-value)	GTEx Portal V6	9
DRAM1	?	ENSG00000136048	muscularis mucosae	2.61×10-12 (p-value)	GTEx Portal V6	9
CHPT1	?	ENSG00000111666	tibial nerve	5.47×10-6 (p-value)	GTEx Portal V6	9
CHPT1	?	ENSG00000111666	transformed fibroblasts	7.69×10-15 (p-value)	GTEx Portal V6	9
CHPT1	?	ENSG00000111666	tibial artery	2.87×10-14 (p-value)	GTEx Portal V6	9
CHPT1	?	ENSG00000111666	thyroid	2.40×10-11 (p-value)	GTEx Portal V6	9
CHPT1	?	ENSG00000111666	skeletal muscle	1.82×10-7 (p-value)	GTEx Portal V6	9
CHPT1	?	ENSG00000111666	sun exposed skin	4.17×10-8 (p-value)	GTEx Portal V6	9
CHPT1	?	ENSG00000111666	aorta	2.91×10-9 (p-value)	GTEx Portal V6	9
CHPT1	?	ENSG00000111666	subcutaneous adipocytes	1.16×10-8 (p-value)	GTEx Portal V6	9
DRAM1	ENST00000258534	ILMN_1669376	blood	2.44×10-8 (p-value)	MuTHER consortium	3
CHPT1	ENST00000229266	ILMN_2202940	adipocyte	4.19×10-6 (p-value)	MuTHER consortium	3
CHPT1	ENST00000549872					
CHPT1	ENST00000552215					

CHPT1	ENST00000552351					
CHPT1	ENST00000549128					
CHPT1	ENST00000549872	230364_at	blood	2.30×10-9 (p-value)	Dixon et al.	2
CHPT1	ENST00000552215					
CHPT1	ENST00000229266	221675_s_at	blood	1.70×10-9 (p-value)	Dixon et al.	2
CHPT1	ENST00000552215					
CHPT1	ENST00000549872					
CHPT1	ENST00000552351					
CHPT1	ENST00000549128					
CHPT1	?	ENSG00000111666	blood	6.59×10-3 (q-value)	SeeQTL DB (HapMap)	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000143578	1	ENCP00000017129	CCDC53
		ENCP00000017128	CCDC53

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001614475 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K36me3
		embryonic stem cell (H1ESC)	H3K4me2, H3K27ac, DNase1
		lung (IMR90)	H3K36me3
		blood (DND-41)	H3K4me1, H3K27ac, H3K36me3
ENSR00001614476 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, Sin3Ak20, CTCF, USF1, DNase1
		blood (DND-41)	H3K4me1, CTCF, H3K27ac
		blood (K562)	USF1, DNase1, Max, CTCF
		skin (NHDF-AD)	DNase1
		cervix (HeLa-S3)	H3K36me3, CTCF
		monocytes (Monocytes-CD14+)	DNase1
		blood (GM12878)	CTCF
		nervous (NH-A)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
GNPTAB	upstream gene variant	1155	ENST00000549940	?	ENSP00000449150	1
GNPTAB	upstream gene variant	1294	ENST00000549165	?	ENSP00000450413	1
GNPTAB	upstream gene variant	1130	ENST00000392919	?	ENSP00000376651	1
GNPTAB	upstream gene variant	1035	ENST00000299314	NM_024312.4	ENSP00000299314	1
RNA5SP369	upstream gene variant	4847	ENST00000411114	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GNPTAB	ENST00000549165	?	ENSP00000450413	8
GNPTAB	ENST00000549940	?	ENSP00000449150	8
GNPTAB	ENST00000392919	?	ENSP00000376651	8
GNPTAB	ENST00000299314	NM_024312.4	ENSP00000299314	8

Locus 335 (sentinel SNP rs10860794 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.