Locus 389 (rs2559856)

SNiPAcard

Block annotations

Block info

genomic range	chr12:102,080,976-102,093,459
block size	12,484 bp
variant count	41 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.275 [-3.362 – 1.909]	gene(s) hit or close-by	CHPT1, MYBPC1, RP11-285E23.2
phastCons	μ = 0.029 [0 – 0.492]	eQTL gene(s)	CHPT1, GNPTAB
GERP++	μ = -0.432 [-4.69 – 2.62]	potentially regulated gene(s)	–
CADD score	μ = 2.917 [0.018 – 8.437]	disease gene(s)	GNPTAB, MYBPC1

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
MYBPC1	ARTHROGRYPOSIS, DISTAL, TYPE 1B	OMIM	MIM:614335
MYBPC1	LETHAL CONGENITAL CONTRACTURE SYNDROME 4	OMIM	MIM:614915
GNPTAB	Mucolipidosis type 3	OrphaNet	OrphaNet:577
GNPTAB	Mucolipidosis type 2	OrphaNet	OrphaNet:576
MYBPC1	lethal congenital contracture syndrome type 3	OrphaNet	OrphaNet:137783
MYBPC1	Digitotalar dysmorphism	OrphaNet	OrphaNet:1146

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CHPT1	?	ENSG00000111666	pancreas	2.04×10^{-10} (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	lung	2.66×10^{-10} (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	tibial nerve	5.21×10^{-10} (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	transformed fibroblasts	8.22×10^{-40} (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	blood	2.02×10^{-13} (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	tibial artery	8.25×10^{-33} (p-value)	GTEx Portal V6	41
GNPTAB	?	ENSG00000111670	tibial artery	1.43×10^{-6} (p-value)	GTEx Portal V6	5
CHPT1	?	ENSG00000111666	esophagus mucosa	1.12×10^{-14} (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	thyroid	2.38×10^{-16} (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	gastroesophageal junction	2.55×10^{-6} (p-value)	GTEx Portal V6	2
CHPT1	?	ENSG00000111666	skeletal muscle	6.95×10^{-24} (p-value)	GTEx Portal V6	41

CHPT1	?	ENSG00000111666	transverse colon	1.05×10-9 (p-value)	GTEx Portal V6	39
CHPT1	?	ENSG00000111666	sun exposed skin	1.16×10-25 (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	unexposed skin	1.36×10-12 (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	aorta	4.81×10-17 (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	subcutaneous adipocytes	3.43×10-27 (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	muscularis mucosae	1.26×10-9 (p-value)	GTEx Portal V6	40
CHPT1	?	ENSG00000111666	atrial appendage	1.10×10-7 (p-value)	GTEx Portal V6	37
CHPT1	?	ENSG00000111666	EBV lymphocytes	4.35×10-9 (p-value)	GTEx Portal V6	40
CHPT1	?	ENSG00000111666	left ventricle	6.67×10-8 (p-value)	GTEx Portal V6	35
CHPT1	?	ENSG00000111666	visceral adipocytes	1.23×10-10 (p-value)	GTEx Portal V6	40
CHPT1	?	ENSG00000111666	sigmoid colon	6.14×10-6 (p-value)	GTEx Portal V6	3
CHPT1	?	ENSG00000111666	coronary artery	1.63×10-9 (p-value)	GTEx Portal V6	40
CHPT1	?	ENSG00000111666	cerebellum	4.38×10-8 (p-value)	GTEx Portal V6	36
CHPT1	?	ENSG00000111666	blood	6.05×10-16 (q-value)	SeeQTL DB (HapMap)	7
CHPT1	ENST00000229266	ILMN_2202940	adipocyte	2.25×10-5 (p-value)	MuTHER consortium	7
CHPT1	ENST00000549872					
CHPT1	ENST00000552215					
CHPT1	ENST00000552351					
CHPT1	ENST00000549128					
?	?	ILMN_1729112	monocyte	3.62×10-40 (p-value)	Zeller et al.	4
CHPT1	ENST00000229266	ILMN_2202940	monocyte	7.88×10-17 (p-value)	Zeller et al.	4
CHPT1	ENST00000549872					
CHPT1	ENST00000552215					
CHPT1	ENST00000552351					
CHPT1	ENST00000549128					
CHPT1	ENST00000549872	230364_at	blood	1.30×10-9 (p-value)	Dixon et al.	2
CHPT1	ENST00000552215					
CHPT1	ENST00000229266	221675_s_at	blood	9.00×10-17 (p-value)	Dixon et al.	2
CHPT1	ENST00000552215					
CHPT1	ENST00000549872					
CHPT1	ENST00000552351					
CHPT1	ENST00000549128					
?	?	1559739_at	blood	3.10×10-8 (p-value)	Dixon et al.	1
?	?	ILMN_1729112	monocyte	8.28×10-7 (p-value)	Fairfax et al.	2
			b-cell	3.32×10-4 (p-value)		
CHPT1	?	ENSG00000111666	uterus	5.49×10-7 (p-value)	GTEx Portal V6	3
CHPT1	?	ENSG00000111666	hippocampus	1.23×10-6 (p-value)	GTEx Portal V6	1
CHPT1	?	ENSG00000111666	pituitary	3.42×10-6 (p-value)	GTEx Portal V6	2

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000090464 (promoter)	4	embryonic stem cell (H1ESC)	DNase1, H3K27me3, PolII, Rad21, H3K9ac, Yy1, TAF1, H3K4me2, H3K4me3
		HSMMtube	H3K9ac, H3K4me2, H2AZ
		blood (K562)	TAF1, PolII, H3K4me2, CTCF, DNase1, H3K4me3, H2AZ, H3K79me2, H3K27ac, Max, ZBTB7A, H3K9ac, Cmyc
		skin (NHDF-AD)	H3K4me2, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	DNase1, H3K36me3, H3K4me2, H3K4me3, H2AZ
		liver (HepG2)	PolII, H3K79me2, TAF1, H3K4me1, H3K4me2, H3K9ac, H3K27ac, CTCF, H3K4me3, DNase1
		lung (IMR90)	H3K4me2, H3K4me3
		blood (GM12878)	DNase1, PolII, H2AZ, CTCF, Yy1, H3K4me3, H3K4me2, H3K9ac
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	H3K9ac, H3K4me2, H3K4me3
		NHLF	H3K27ac, H3K4me3, H3K9ac
		Osteobl	H2AZ, H3K4me3, H3K4me2
		blood (DND-41)	H3K27me3, H3K4me3, H3K9ac, H3K4me2
		breast (HMEC)	H3K4me3, H3K4me2
		cervix (HeLa-S3)	DNase1, CTCF, PolII, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K4me3, H3K4me2, PolII, CTCF, DNase1
		A549	H3K36me3, H3K9ac, H3K4me2, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CHPT1	upstream gene variant	71	ENST00000549872	?	ENSP00000448766	26
CHPT1	upstream gene variant	26	ENST00000229266	NM_020244.2	ENSP00000229266	26
CHPT1	upstream gene variant	91	ENST00000549128	?	ENSP00000446994	26
CHPT1	upstream gene variant	479	ENST00000550385	?	?	24
CHPT1	upstream gene variant	253	ENST00000546490	?	?	26
CHPT1	upstream gene variant	91	ENST00000552351	?	ENSP00000447887	26
CHPT1	upstream gene variant	364	ENST00000552215	?	ENSP00000448831	26
MYBPC1	downstream gene variant	1350	ENST00000441232	?	ENSP00000388989	11
MYBPC1	downstream gene variant	1318	ENST00000550501	?	?	11
MYBPC1	downstream gene variant	1583	ENST00000541119	NM_001254720.1	ENSP00000442847	11
MYBPC1	downstream gene variant	1180	ENST00000549145	?	ENSP00000447660	12
MYBPC1	downstream gene variant	1328	ENST00000361466	NM_002465.3	ENSP00000354849	11
MYBPC1	downstream gene variant	1392	ENST00000392934	NM_001254723.1	ENSP00000376665	11
MYBPC1	downstream gene variant	1387	ENST00000549608	?	?	11
MYBPC1	downstream gene variant	1329	ENST00000452455	NM_001254718.1	ENSP00000400908	11
MYBPC1	downstream gene variant	1386	ENST00000547509	?	ENSP00000447362	11
MYBPC1	downstream gene variant	1329	ENST00000551300	?	ENSP00000447116	11
MYBPC1	downstream gene variant	1322	ENST00000361685	NM_206819.2	ENSP00000354845	11
MYBPC1	downstream gene variant	2803	ENST00000550270	NM_206820.2	ENSP00000449702	10
MYBPC1	downstream gene variant	1387	ENST00000547405	NM_001254722.1	ENSP00000448175	11
MYBPC1	downstream gene variant	1583	ENST00000536007	NM_001254721.1	ENSP00000446128	11
MYBPC1	downstream gene variant	1583	ENST00000545503	NM_001254719.1	ENSP00000440034	11
MYBPC1	downstream gene variant	1328	ENST00000553190	NM_206821.2	ENSP00000447900	11
RP11-285E23.2	downstream gene variant, upstream gene variant	18	ENST00000616668	?	?	27

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CHPT1	ENST00000229266	NM_020244.2	ENSP00000229266	2
CHPT1	ENST00000550385	?	?	4
CHPT1	ENST00000552215	?	ENSP00000448831	2
CHPT1	ENST00000549872	?	ENSP00000448766	2
CHPT1	ENST00000546490	?	?	2
CHPT1	ENST00000552351	?	ENSP00000447887	2
CHPT1	ENST00000549128	?	ENSP00000446994	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-285E23.2	ENST00000616668	?	2