Locus 161 (rs2284367)

SNiPAcard

Block annotations

Block info

genomic range	chr11:34,468,936-34,516,856
block size	47,921 bp
variant count	20 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.298 [-3.369 – 2.261]	**gene(s) hit or close-by**	AL137224.1, CAT, ELF5
phastCons	μ = 0.007 [0 – 0.123]	**eQTL gene(s)**	ABTB2, CAT
GERP++	μ = -0.916 [-8.18 – 3]	**potentially regulated gene(s)**	CAPRIN1, NAT10
CADD score	μ = 4.821 [0.258 – 15.84]	**disease gene(s)**	CAT

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM025323	1

Disease gene annotation

gene	trait	source DB	source entry/link
CAT	ACATALASEMIA	OMIM	MIM:614097
CAT	ACATALASEMIA	OrphaNet	OrphaNet:926

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CAT	?	ENSG00000121691	pancreas	2.40×10^{-10} (p-value)	GTEx Portal V6	20
CAT	ENST00000241052	ILMN_1651705	skin	2.67×10^{-5} (p-value)	MuTHER consortium	10
CAT	ENST00000534710		blood	2.44×10^{-8} (p-value)	MuTHER consortium	10
CAT	?	ENSG00000121691	muscularis mucosae	4.69×10^{-9} (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	lung	4.41×10^{-10} (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	atrial appendage	3.33×10^{-13} (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	transformed fibroblasts	8.63×10^{-26} (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	blood	7.56×10^{-13} (p-value)	GTEx Portal V6	20
ABTB2	?	ENSG00000166016	blood	3.68×10^{-7} (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	breast	3.41×10^{-12} (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	tibial artery	1.02×10^{-12} (p-value)	GTEx Portal V6	20
CAT	ENST00000241052	ILMN_1651705	monocyte	2.30×10^{-14} (p-value)	Fairfax et al.	10
CAT	ENST00000534710		b-cell	1.88×10^{-12} (p-value)	Fairfax et al.	10
CAT	?	ENSG00000121691	thyroid	2.57×10^{-17} (p-value)	GTEx Portal V6	20

CAT	?	ENSG00000121691	skeletal muscle	1.76×10-12 (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	EBV lymphocytes	7.71×10-8 (p-value)	GTEx Portal V6	16
CAT	?	ENSG00000121691	transverse colon	7.97×10-9 (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	blood	4.72×10-5 (q-value)	SeeQTL DB (HapMap)	7
CAT	?	ENSG00000121691	ovary	4.17×10-7 (p-value)	GTEx Portal V6	17
CAT	?	ENSG00000121691	sun exposed skin	6.01×10-16 (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	unexposed skin	4.61×10-7 (p-value)	GTEx Portal V6	18
CAT	?	ENSG00000121691	aorta	3.71×10-8 (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	left ventricle	9.50×10-13 (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	subcutaneous adipocytes	3.93×10-12 (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	stomach	2.66×10-6 (p-value)	GTEx Portal V6	5
CAT	?	ENSG00000121691	visceral adipocytes	1.66×10-7 (p-value)	GTEx Portal V6	16
CAT	?	ENSG00000121691	tibial nerve	1.18×10-12 (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	pituitary	2.88×10-8 (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	esophagus mucosa	4.10×10-12 (p-value)	GTEx Portal V6	20
CAT	ENST00000241052	201432_at	blood	3.80×10-13 (p-value)	Dixon et al.	5
CAT	ENST00000534710					
CAT	ENST00000241052	211922_s_at	blood	1.20×10-9 (p-value)	Dixon et al.	5
CAT	?	ENSG00000121691	testis	1.09×10-9 (p-value)	GTEx Portal V6	20
CAT	ENST00000241052	ILMN_1651705	monocyte	1.61×10-26 (p-value)	Zeller et al.	2
CAT	ENST00000534710					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000102675	1	ENCP00000011718	NAT10
		ENCP00000011715	CAPRIN1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001603033 (enhancer)	1	cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3, H3K4me1
		liver (HepG2)	H3K36me3
		Osteobl	H3K36me3
		skin (NHEK)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL137224.1	downstream gene variant, upstream gene variant	1272	ENST00000579311	?	?	3
CAT	upstream gene variant, downstream gene variant	1947	ENST00000528104	?	?	4

CAT	upstream gene variant, downstream gene variant	459	ENST00000525707	?	?	2
CAT	upstream gene variant	1724	ENST00000534710	?	?	3
CAT	upstream gene variant, downstream gene variant	1185	ENST00000530343	?	?	5
ELF5	downstream gene variant	2463	ENST00000532417	?	ENSP00000436386	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
CAT	ENST00000241052	NM_001752.3	ENSP00000241052	D	gaT/gaC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CAT	ENST00000525707	?	?	8
CAT	ENST00000530343	?	?	5
CAT	ENST00000241052	NM_001752.3	ENSP00000241052	14
ELF5	ENST00000528709	?	?	5
ELF5	ENST00000429939	NM_001243080.1	ENSP00000407589	5
ELF5	ENST00000620316	NM_001243081.1	ENSP00000484521	5
ELF5	ENST00000312319	NM_198381.1	ENSP00000311010	5
ELF5	ENST00000257832	NM_001422.3	ENSP00000257832	5
ELF5	ENST00000532417	?	ENSP00000436386	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CAT	ENST00000525707	?	1
CAT	ENST00000530343	?	1