Locus 20 (rs12141375)

SNiPAcard

Block annotations

Block info

genomic range	chr1:203,173,094-203,193,134
block size	20,041 bp
variant count	14 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.357 [-3.015 – 0.381]	**gene(s) hit or close-by**	CHIT1
phastCons	μ = 0.005 [0 – 0.039]	**eQTL gene(s)**	ADORA1, CHIT1
GERP++	μ = -1.325 [-4.67 – 2.4]	**potentially regulated gene(s)**	–
CADD score	μ = 3.377 [0.095 – 11.08]	**disease gene(s)**	CHIT1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CHIT1	CHITOTRIOSIDASE DEFICIENCY	OMIM	MIM:614122

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ADORA1	?	ENSG00000163485	transformed fibroblasts	1.36×10^{-6} (p-value)	GTEx Portal V6	12
CHIT1	?	ENSG00000133063	blood	1.71×10^{-8} (p-value)	GTEx Portal V6	14
CHIT1	?	ENSG00000133063	cortex	3.81×10^{-7} (p-value)	GTEx Portal V6	8

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001527715 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1, CTCF, Rad21
		HSMMtube	H3K27ac, DNase1
		Osteobl	H3K4me2, H3K27ac
		blood (DND-41)	H3K27me3
		blood (K562)	CTCF, H3K27me3
		skin (NHDF-AD)	DNase1
		breast (HMEC)	DNase1
		muscle (HSMM)	H3K4me1, H3K27ac, H3K36me3, DNase1
		cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K4me1, DNase1
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	DNase1, H3K4me1, H3K18ac, H3K27ac, H4K5ac
		nervous (NH-A)	DNase1
		A549	H3K27me3
ENSR00001527718 (open chromatin region)	2	endothelium (HUVEC)	H3K27me3
		blood (K562)	H3K27me3
		muscle (HSMM)	H3K27ac, H3K9ac, H3K36me3, DNase1

ENSR00001527719 (open chromatin region)	1	endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
		muscle (HSMM)	H3K27ac, DNase1
ENSR00001589716 (enhancer)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21
		HSMMtube	CTCF
		blood (K562)	CTCF, H3K27me3
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	CTCF, H3K27me3
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, H3K27me3, DNase1
		monocytes (Monocytes-CD14+)	H3K27me3, DNase1, CTCF
		endothelium (HUVEC)	H3K27me3
		A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CHIT1	downstream gene variant, upstream gene variant	609	ENST00000506427	?	?	3
CHIT1	downstream gene variant	1032	ENST00000255427	NM_001256125.1	ENSP00000255427	2
CHIT1	downstream gene variant	593	ENST00000484834	?	?	4
CHIT1	downstream gene variant, upstream gene variant	601	ENST00000479483	?	?	6
CHIT1	downstream gene variant	1032	ENST00000491855	?	ENSP00000423778	2
CHIT1	downstream gene variant	441	ENST00000367229	NM_001270509.1, NM_003465.2	ENSP00000356198	2
CHIT1	downstream gene variant	1310	ENST00000503786	?	ENSP00000421617	2
CHIT1	downstream gene variant	1454	ENST00000513472	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CHIT1	ENST00000506427	?	?	1
CHIT1	ENST00000255427	NM_001256125.1	ENSP00000255427	3
CHIT1	ENST00000484834	?	?	4
CHIT1	ENST00000479483	?	?	1
CHIT1	ENST00000491855	?	ENSP00000423778	3
CHIT1	ENST00000367229	NM_001270509.1, NM_003465.2	ENSP00000356198	3
CHIT1	ENST00000503786	?	ENSP00000421617	3
CHIT1	ENST00000513472	?	?	1