Locus 169 (rs4950937)

SNiPAcard

Block annotations

Block info

genomic range	chr1:203,170,892-203,194,186
block size	23,295 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.235 [-1.349 – 4.997]	gene(s) hit or close-by	CHIT1
phastCons	μ = 0.184 [0 – 1]	eQTL gene(s)	KRT8P29
GERP++	μ = -0.102 [-3.04 – 2.59]	potentially regulated gene(s)	–
CADD score	μ = 5.938 [0.591 – 16.99]	disease gene(s)	CHIT1

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Chitotriosidase deficiency	pathogenic	ClinVar	RCV000010134.1	1
?	HGMD curated	HGMD	CM065066	1
CHITOTRIOSIDASE DEFICIENCY	OMIM curated	OMIM	MIM:600031	1

Disease gene annotation

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

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CHIT1	missense variant	ENST00000503786	?	ENSP00000421617	S/G	Agc/Ggc	?	?	1
CHIT1	missense variant	ENST00000491855	?	ENSP00000423778	S/G	Agc/Ggc	?	?	1
CHIT1	missense variant	ENST00000367229	NM_001270509.1, NM_003465.2	ENSP00000356198	S/G	Agc/Ggc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
KRT8P29	?	ENSG00000236430	tibial nerve	7.68×10-6 (p-value)	GTEx Portal V6	6

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors

ENSR00001527715 (promoter flanking region)	3	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
ENSR00001527717 (enhancer)	1	HSMMtube	H3K4me1, H3K27ac, DNase1
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		muscle (HSMM)	H3K27ac, DNase1
ENSR00001527722 (open chromatin region)	1	endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (GM12878)	H3K27me3
		A549	H3K27me3
		blood (K562)	H3K27me3
		skin (NHEK)	H3K27me3
		muscle (HSMM)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CHIT1	downstream gene variant	3506	ENST00000491855	?	ENSP00000423778	1
CHIT1	downstream gene variant	3784	ENST00000503786	?	ENSP00000421617	1
CHIT1	downstream gene variant	2476	ENST00000513472	?	?	2
CHIT1	downstream gene variant	3506	ENST00000255427	NM_001256125.1	ENSP00000255427	1
CHIT1	upstream gene variant	2759	ENST00000479483	?	?	2
CHIT1	downstream gene variant	2915	ENST00000367229	NM_001270509.1, NM_003465.2	ENSP00000356198	1
CHIT1	upstream gene variant	2748	ENST00000506427	?	?	1

Putative effect on transcript

Intron variant

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

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CHIT1	ENST00000484834	?	1
CHIT1	ENST00000513472	?	1

CHIT1	ENST00000513472	?	1