Locus 133 (rs1800973)

SNiPAcard

Block annotations

Block info

genomic range	chr12:69,711,929-69,744,014
block size	32,086 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.680 [-1.08 – 2.441]	gene(s) hit or close-by	LYZ, RP11-1143G9.4
phastCons	μ = 0.538 [0.095 – 0.981]	eQTL gene(s)	–
GERP++	μ = 2.821 [-0.298 – 5.94]	potentially regulated gene(s)	LYZ
CADD score	μ = 8.722 [0.794 – 16.65]	disease gene(s)	LYZ

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM060322	1

Disease gene annotation

gene	trait	source DB	source entry/link
LYZ	AMYLOIDOSIS, FAMILIAL VISCERAL	OMIM	MIM:105200
LYZ	Familial renal amyloidosis due to lysozyme variant	OrphaNet	OrphaNet:93561

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)
LYZ	missense variant	ENST00000548839	?	ENSP00000449969	N/T	aAc/aCc	?	?	1
LYZ	missense variant	ENST00000549690	?	ENSP00000449898	N/T	aAc/aCc	?	?	1
LYZ	missense variant	ENST00000261267	NM_000239.2	ENSP00000261267	N/T	aAc/aCc	?	?	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000127685	1	ENST00000549690, ENST00000261267	LYZ

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000653867 (promoter flanking region)	1	HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H2AZ, PU1, DNase1
		blood (DND-41)	H3K27me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K27me3

	liver (HepG2)	H3K4me3, PolII, H3K79me2, DNase1, TAF1, p300, H3K4me2, FOXA1, H3K9ac, H3K27ac, H3K36me3
	lung (IMR90)	H3K27me3
	A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-1143G9.4	downstream gene variant	3259	ENST00000548900	?	?	1