Locus 250 (rs10754218)

SNiPAcard

Block annotations

Block info

genomic range	chr1:197,162,526-197,165,267
block size	2,742 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.239 [-2.612 – 0.829]	gene(s) hit or close-by	ZBTB41
phastCons	μ = 0.081 [0 – 0.389]	eQTL gene(s)	CFHR3
GERP++	μ = -0.548 [-4.43 – 1.87]	potentially regulated gene(s)	–
CADD score	μ = 4.600 [0.478 – 14.6]	disease gene(s)	CFHR3

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CFHR3	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CFHR3	?	ENSG00000116785	lung	4.33×10-7 (p-value)	GTEx Portal V6	4

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001527247 (promoter flanking region)	3	cervix (HeLa-S3)	H3K4me2
		embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K27ac, H3K4me2, H3K36me3
		blood (GM12878)	H3K4me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K79me2, H3K4me2
		muscle (HSMM)	H3K36me3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ZBTB41	ENST00000467322	?	?	5
ZBTB41	ENST00000367405	NM_194314.2	ENSP00000356375	5