Locus 421 (rs5994176)

SNiPAcard

Block annotations

Block info

genomic range	chr22:17,616,715-17,618,164
block size	1,450 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.697 [-1.53 – -0.194]	**gene(s) hit or close-by**	AC006946.15, CECR5
phastCons	μ = 0.002 [0 – 0.005]	**eQTL gene(s)**	CECR6
GERP++	μ = -0.386 [-2.83 – 1.56]	**potentially regulated gene(s)**	–
CADD score	μ = 4.606 [3.14 – 5.743]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CECR6	?	ENSG00000183307	sun exposed skin	4.38×10^{-8} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC006946.15	downstream gene variant	3721	ENST00000441544	?	?	1
CECR5	downstream gene variant	237	ENST00000155674	NM_017829.5	ENSP00000155674	3
CECR5	downstream gene variant	248	ENST00000477157	?	?	3
CECR5	downstream gene variant	308	ENST00000486462	?	?	3
CECR5	downstream gene variant	305	ENST00000399852	?	ENSP00000382745	3
CECR5	downstream gene variant	246	ENST00000336737	NM_033070.2	ENSP00000337358	3