Supplementary Table 2: rs2839629 functional annotations

*SNiPA*card – rs2839629

SNP properties – Genome Assembly: grch37, Variant set: 1kgpp3v5, Population: EUR

rs2839629 (alias rs60091962, rs56533768, rs3746944)

position / outlink		allele info	
physical position	chr21: 44,453,022	alleles	G/A
genetic position [cM]	57.37	frequencies	0.545/0.455
outlink	e!	non-reference allele	A

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-2.733	gene(s) hit or close-by	PKNOX1 e!
phastCons	0	eQTL gene(s)	CBS e!, CBS e!, PKNOX1 e!
GERP++	-7.94	potentially regulated gene(s)	–
CADD score	1.122	disease gene(s)	CBS e!, CBS e!
SnpEff effect impact	modifier		

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link	
CBS e!	HOMOCYSTINURIA DUE TO CYSTATHIONINE BETA-SYNTHASE DEFICIENCY	OMIM	MIM:236200	OMIM
CBS e!	HOMOCYSTINURIA DUE TO CYSTATHIONINE BETA-SYNTHASE DEFICIENCY	OMIM	MIM:236200	OMIM
CBS e!	cystathionine beta-synthase deficiency (CBSD)	DECIPHER	MIM:236200	OMIM
CBS e!	cystathionine beta-synthase deficiency (CBSD)	DECIPHER	MIM:236200	OMIM
CBS e!	Classical homocystinuria	OrphaNet	OrphaNet:394	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	statistic (type)	source
CBS e!	ENST00000398165 e!	ILMN_1804735 e!	skin	2.60×10^{-15} (p-value)	MuTHER consortium
CBS e!	ENST00000617706 e!		blood	3.06×10^{-8} (p-value)	MuTHER consortium
CBS e!	ENST00000462349 e!				
CBS e!	ENST00000352178 e!				
CBS e!	ENST00000624406 e!				
CBS e!	ENST00000359624 e!				
CBS e!	ENST00000461686 e!				
CBS e!	ENST00000618024 e!				
CBS e!	ENST00000398168 e!				
CBS e!	ENST00000624691 e!				
CBS e!	ENST00000398158 e!				

PKNOX1	?	ENSG00000160199	tibial nerve	5.59×10^{-8} (p-value)	GTEx Portal V6	

Putative effect on regulation

Variation proximal to gene

gene	variant type	distance	transcript	RefSeq id	protein
PKNOX1	downstream gene variant	4131	ENST00000474336	?	?
PKNOX1	downstream gene variant	2581	ENST00000560448	?	ENSP00000453486
PKNOX1	downstream gene variant	4037	ENST00000607150	?	?
PKNOX1	downstream gene variant	2750	ENST00000607049	?	?
PKNOX1	downstream gene variant	2963	ENST00000557820	?	?
PKNOX1	downstream gene variant	2698	ENST00000558955	?	?

Putative effect on transcript

3'-UTR variant

gene	affected transcript	RefSeq id	protein
PKNOX1	ENST00000291547	NM_004571.4	ENSP00000291547
PKNOX1	ENST00000432907	NM_001286258.1	ENSP00000402243