Supplementary Table 3: rs915854 functional annotations

*SNiPA*card – rs915854

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

rs915854 (alias rs59918064)

position / outlink		allele info	
physical position	chr21: 44,466,766	alleles	C/T
genetic position [cM]	57.37	frequencies	0.541/0.459
outlink	e!	non-reference allele	T

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.437	gene(s) hit or close-by	–
phastCons	0.004	eQTL gene(s)	PKNOX1 e!, WDR4 e!
GERP++	0.53	potentially regulated gene(s)	–
CADD score	4.2	disease gene(s)	–
SnpEff effect impact	modifier		

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	statistic (type)	source
PKNOX1 e!	?	ENSG00000160199 e!	tibial nerve	1.87×10^{-7} (p-value)	GTEx Portal V6
WDR4 e!	?	ENSG00000160193 e!	esophagus mucosa	6.87×10^{-6} (p-value)	GTEx Portal V6

Putative effect on regulation

Regulatory feature cluster

element id	tissue/cell	factors
ENSR00000614382 e! (promoter flanking region)	embryonic stem cell (H1ESC)	PolII, Rad21
	HSMMtube	DNase1
	Osteobl	H3K4me2
	blood (DND-41)	H3K4me1, H3K27ac, H3K9ac
	skin (NHDF-AD)	DNase1
	cervix (HeLa-S3)	DNase1, Max, H3K27ac, H3K9ac
	monocytes (Monocytes-CD14+)	H3K4me1
	liver (HepG2)	H3K4me2