Locus 417 (rs16873418)

SNiPAcard

Block annotations

Block info

genomic range	chr8:106,578,977-106,593,207
block size	14,231 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.260 [-0.31 – 0.853]	gene(s) hit or close-by	ZFPM2
phastCons	μ = 0.110 [0 – 0.982]	eQTL gene(s)	CXCL5
GERP++	μ = 0.121 [-1.7 – 3.49]	potentially regulated gene(s)	–
CADD score	μ = 5.068 [1.506 – 9.561]	disease gene(s)	ZFPM2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Platelet counts	$<4.00\times10^{-17}$	GWAS Catalog	22139419	1
Vascular endothelial growth factor levels	$<5.00\times10^{-23}$	GWAS Catalog	21757650	1

Disease gene annotation

gene	trait	source DB	source entry/link
ZFPM2	CONOTRUNCAL HEART MALFORMATIONS	OMIM	MIM:217095
ZFPM2	46,XY SEX REVERSAL 9	OMIM	MIM:616067
ZFPM2	TETRALOGY OF FALLOT	OMIM	MIM:187500
ZFPM2	DIAPHRAGMATIC HERNIA 3	OMIM	MIM:610187
ZFPM2	TETRALOGY OF FALLOT	DECIPHER	MIM:187500
ZFPM2	DIAPHRAGMATIC HERNIA 3	DECIPHER	MIM:610187
ZFPM2	Congenital diaphragmatic hernia	OrphaNet	OrphaNet:2140
ZFPM2	TETRALOGY OF FALLOT	OrphaNet	OrphaNet:3303

Direct effect on regulation

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
CXCL5	ENST00000296027	ILMN_2171384	chr4	blood	1.28×10^{-7} (p-value)	Westra et al.	1
?	?	ILMN_1884950	chr7	blood	1.23×10^{-7} (p-value)	Westra et al.	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001398577 (enhancer)	1	lung (IMR90)	H3K36me3
		blood (K562)	DNase1
ENSR00001398582 (promoter flanking region)	1	lung (IMR90)	H3K4me2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ZFPM2	ENST00000517361	?	ENSP00000428720	9
ZFPM2	ENST00000511341	?	?	9
ZFPM2	ENST00000520492	?	ENSP00000430757	9
ZFPM2	ENST00000520027	?	ENSP00000428149	9
ZFPM2	ENST00000407775	NM_012082.3	ENSP00000384179	9