Locus 303 (rs17354650)

SNiPAcard

Block annotations

Block info

genomic range	chr1:20,390,322-20,390,322
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.128	gene(s) hit or close-by	PLA2G5
phastCons	0	eQTL gene(s)	MRTO4, OTUD3
GERP++	0.13	potentially regulated gene(s)	–
CADD score	4.268	disease gene(s)	PLA2G5

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
PLA2G5	FLECK RETINA, FAMILIAL BENIGN	OMIM	MIM:228980
PLA2G5	Familial benign flecked retina	OrphaNet	OrphaNet:363989

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MRTO4	ENST00000330263	ILMN_1689800	monocyte	4.13×10^{-4} (p-value)	Fairfax et al.	1
MRTO4	ENST00000479559					
OTUD3	ENST00000375120	ILMN_1779280	b-cell	3.71×10^{-6} (p-value)	Fairfax et al.	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000531978 (promoter flanking region)	1	NHLF	DNase1
		HSMMtube	DNase1
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PLA2G5	ENST00000465698	?	?	1
PLA2G5	ENST00000489871	?	?	1

PLA2G5	ENST00000469069	?	?	1
PLA2G5	ENST00000498348	?	?	1
PLA2G5	ENST00000486277	?	?	1
PLA2G5	ENST00000460175	?	?	1