SNiPAcard

Block annotations

Block info

genomic range	chr3:56,849,749-56,849,749
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	1.794	**gene(s) hit or close-by**	ARHGEF3
phastCons	1	**eQTL gene(s)**	–
GERP++	4.93	**potentially regulated gene(s)**	ARHGEF3, CCDC66
CADD score	13.91	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Platelet counts	$<6.00\times10^{-24}$	GWAS Catalog	24026423	1
Mean platelet volume	$<9.00\times10^{-34}$	GWAS Catalog	24026423	1
Platelet counts	$<1.00\times10^{-19}$	GWAS Catalog	23263863	1
Platelet counts	$<3.00\times10^{-54}$	GWAS Catalog	22139419	1
Mean platelet volume	$<3.00\times10^{-69}$	GWAS Catalog	22139419	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000341329	1	ENCP00000036876	ARHGEF3
		ENCP00000036866	CCDC66
		ENCP00000036864	CCDC66
		ENCP00000036861	CCDC66

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ARHGEF3	ENST00000477833	?	ENSP00000418984	1
ARHGEF3	ENST00000481422	?	?	1
ARHGEF3	ENST00000468466	?	ENSP00000418469	1
ARHGEF3	ENST00000477440	?	?	1
ARHGEF3	ENST00000496106	NM_001289698.1	ENSP00000420420	1
ARHGEF3	ENST00000338458	NM_001128615.1	ENSP00000341071	1
ARHGEF3	ENST00000486829	?	ENSP00000420633	1
ARHGEF3	ENST00000498517	?	?	1

ARHGEF3	ENST00000473779	?	ENSP00000420402	1
ARHGEF3	ENST00000468727	?	ENSP00000417087	1