SNiPAcard

Block annotations

Block info

genomic range	chr6:161,177,237-161,252,795
block size	75,559 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.106 [-1.896 – -0.595]	**gene(s) hit or close-by**	PLG, RP1-81D8.5
phastCons	μ = 0.105 [0 – 0.276]	**eQTL gene(s)**	–
GERP++	μ = 0.088 [-0.465 – 0.687]	**potentially regulated gene(s)**	AGPAT4, PLG, RP1-81D8.4
CADD score	μ = 3.338 [1.084 – 4.486]	**disease gene(s)**	PLG

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Lipoproteins	1.04×10-11	dbGaP	pha003079	dbGaP	1

Disease gene annotation

gene	trait	source DB	source entry/link	
PLG	PLASMINOGEN DEFICIENCY, TYPE ILIGNEOUS CONJUNCTIVITIS, [...]	OMIM	MIM:217090	OMIM
PLG	Hypoplasminogenemia	OrphaNet	OrphaNet:722	orphanet
PLG	Ligneous conjunctivitis	OrphaNet	OrphaNet:97231	orphanet

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000468471	1	ENCP00000050551	PLG
		ENCP00000050546	RP1-81D8.4
ENCE00000468646	1	ENCP00000050568	AGPAT4

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PLG	downstream gene variant	2899	ENST00000308192	NM_000301.3	ENSP00000308938	1
PLG	downstream gene variant	2890	ENST00000461414	?	?	1
PLG	downstream gene variant	3952	ENST00000467466	?	?	1
RP1-81D8.5	upstream gene variant	2721	ENST00000612111	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
RP1-81D8.5	ENST00000612111	?	?	1