SNiPAcard

Block annotations

Block info

genomic range	chr3:3,136,700-3,141,600
block size	4,901 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.028 [-0.759 – 1.12]	**gene(s) hit or close-by**	IL5RA, SNORA43
phastCons	μ = 0.097 [0 – 0.571]	**eQTL gene(s)**	–
GERP++	μ = -0.506 [-4.22 – 3.09]	**potentially regulated gene(s)**	–
CADD score	μ = 2.925 [0.581 – 11.03]	**disease gene(s)**	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
IL5RA	missense variant	ENST00000383846	NM_175728.2	ENSP00000373358	V/I	Gtt/Att	?	?	1
IL5RA	missense variant	ENST00000418488	?	ENSP00000388858	V/I	Gtt/Att	?	?	1
IL5RA	missense variant	ENST00000445701	?	ENSP00000398117	V/I	Gtt/Att	?	?	1
IL5RA	missense variant	ENST00000430514	NM_175724.2	ENSP00000400400	V/I	Gtt/Att	?	?	1
IL5RA	missense variant	ENST00000311981	NM_175725.2	ENSP00000309196	V/I	Gtt/Att	?	?	1
IL5RA	missense variant	ENST00000256452	NM_000564.4	ENSP00000256452	V/I	Gtt/Att	?	?	1
IL5RA	missense variant	ENST00000456302	NM_175727.2	ENSP00000392059	V/I	Gtt/Att	?	?	1
IL5RA	missense variant	ENST00000446632	NM_175726.3	ENSP00000412209	V/I	Gtt/Att	?	?	1
IL5RA	missense variant	ENST00000438560	NM_001243099.1	ENSP00000390753	V/I	Gtt/Att	?	?	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IL5RA	downstream gene variant	833	ENST00000445701	?	ENSP00000398117	4
IL5RA	downstream gene variant	1816	ENST00000427088	?	ENSP00000391274	3
SNORA43	upstream gene variant	2997	ENST00000517240	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL5RA	ENST00000438560	NM_001243099.1	ENSP00000390753	5
IL5RA	ENST00000418488	?	ENSP00000388858	5
IL5RA	ENST00000456302	NM_175727.2	ENSP00000392059	5

IL5RA	ENST00000430514	NM_175724.2	ENSP00000400400	5
IL5RA	ENST00000311981	NM_175725.2	ENSP00000309196	5
IL5RA	ENST00000256452	NM_000564.4	ENSP00000256452	5
IL5RA	ENST00000383846	NM_175728.2	ENSP00000373358	5
IL5RA	ENST00000446632	NM_175726.3	ENSP00000412209	5
IL5RA	ENST00000445701	?	ENSP00000398117	1