SNiPAcard

Block annotations

Block info

genomic range	chr1:204,943,947-204,943,991
block size	45 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.410 [-2.081 – 1.261]	**gene(s) hit or close-by**	NFASC
phastCons	μ = 0.500 [0 – 1]	**eQTL gene(s)**	–
GERP++	μ = 1.000 [-3.8 – 5.8]	**potentially regulated gene(s)**	–
CADD score	μ = 8.738 [3.806 – 13.67]	**disease gene(s)**	–

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
NFASC	downstream gene variant	136	ENST00000514644	?	?	2
NFASC	upstream gene variant	4525	ENST00000471392	?	?	2

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
NFASC	ENST00000339876	NM_001005388.2	ENSP00000344786	V	gtA/gtC	1
NFASC	ENST00000404076	?	ENSP00000385676	V	gtA/gtC	1
NFASC	ENST00000513543	?	ENSP00000425908	V	gtA/gtC	1
NFASC	ENST00000403080	NM_001005389.1, NM_001160333.1	ENSP00000384875	V	gtA/gtC	1
NFASC	ENST00000360049	NM_015090.3	ENSP00000353154	V	gtA/gtC	1
NFASC	ENST00000401399	?	ENSP00000385637	V	gtA/gtC	1
NFASC	ENST00000367173	?	ENSP00000356141	V	gtA/gtC	1
NFASC	ENST00000404907	?	ENSP00000384061	V	gtA/gtC	1
NFASC	ENST00000430393	NM_001160331.1	ENSP00000415031	V	gtA/gtC	1
NFASC	ENST00000539706	NM_001160332.1	ENSP00000438614	V	gtA/gtC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
NFASC	ENST00000339876	NM_001005388.2	ENSP00000344786	1
NFASC	ENST00000404076	?	ENSP00000385676	1
NFASC	ENST00000513543	?	ENSP00000425908	1
NFASC	ENST00000404977	?	?	1
NFASC	ENST00000504476	?	ENSP00000422524	1
NFASC	ENST00000403080	NM_001005389.1, NM_001160333.1	ENSP00000384875	1
NFASC	ENST00000360049	NM_015090.3	ENSP00000353154	1

NFASC	ENST00000401399	?	ENSP00000385637	1
NFASC	ENST00000367173	?	ENSP00000356141	1
NFASC	ENST00000404907	?	ENSP00000384061	1
NFASC	ENST00000430393	NM_001160331.1	ENSP00000415031	1
NFASC	ENST00000539706	NM_001160332.1	ENSP00000438614	1
NFASC	ENST00000512826	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
NFASC	ENST00000504476	?	ENSP00000422524	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
NFASC	ENST00000404977	?	1
NFASC	ENST00000512826	?	1