SNiPAcard

Block annotations

Block info

genomic range	chr1:204,987,953-204,997,928
block size	9,976 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.246 [-0.256 – 1.049]	**gene(s) hit or close-by**	NFASC
phastCons	μ = 0.011 [0 – 0.048]	**eQTL gene(s)**	–
GERP++	μ = -1.266 [-6.36 – 0.72]	**potentially regulated gene(s)**	LRRN2, NFASC, RP11-383G10.1, TMCC2
CADD score	μ = 5.745 [2.233 – 14.53]	**disease gene(s)**	–

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000391750	1	ENST00000495396, ENST00000503221	NFASC

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000052164	1	ENCP00000006256	NFASC
		ENCP00000006273	TMCC2
		ENCP00000006255	NFASC
		ENCP00000006243	LRRN2
		ENCP00000006265	RP11-383G10.1
		ENCP00000006269	TMCC2
		ENCP00000006247	NFASC

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
NFASC	downstream gene variant	454	ENST00000404076	?	ENSP00000385676	4
NFASC	downstream gene variant	454	ENST00000425360	?	ENSP00000395664	4
NFASC	downstream gene variant	543	ENST00000401399	?	ENSP00000385637	2
NFASC	downstream gene variant	86	ENST00000447819	?	ENSP00000416891	4
NFASC	downstream gene variant	543	ENST00000495396	?	?	2
NFASC	downstream gene variant	2439	ENST00000413225	?	ENSP00000393290	3
NFASC	downstream gene variant	544	ENST00000339876	NM_001005388.2	ENSP00000344786	2
NFASC	downstream gene variant	454	ENST00000404907	?	ENSP00000384061	4
NFASC	downstream gene variant	1887	ENST00000513543	?	ENSP00000425908	3
NFASC	downstream gene variant	454	ENST00000367173	?	ENSP00000356141	4
NFASC	downstream gene variant	544	ENST00000360049	NM_015090.3	ENSP00000353154	2
NFASC	downstream gene variant	1887	ENST00000504476	?	ENSP00000422524	3
NFASC	downstream gene variant	544	ENST00000539706	NM_001160332.1	ENSP00000438614	2
NFASC	downstream gene variant	454	ENST00000430393	NM_001160331.1	ENSP00000415031	4
NFASC	downstream gene variant	2334	ENST00000503221	?	?	1

Putative effect on transcript

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
NFASC	ENST00000539706	NM_001160332.1	ENSP00000438614	3
NFASC	ENST00000360049	NM_015090.3	ENSP00000353154	3
NFASC	ENST00000401399	?	ENSP00000385637	3
NFASC	ENST00000339876	NM_001005388.2	ENSP00000344786	3

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
NFASC	ENST00000495396	?	3
NFASC	ENST00000503221	?	3