Locus 433 (rs2228467)

SNiPAcard

Block annotations

Block info

genomic range	chr3:42,906,116-42,906,116
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	2.284	gene(s) hit or close-by	ACKR2, CYP8B1, KRBOX1, RP11-136C24.3, RP11-141M3.6
phastCons	0.985	eQTL gene(s)	–
GERP++	5.52	potentially regulated gene(s)	–
CADD score	10.24	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Cerebrospinal fluid levels of Alzheimer's disease-related proteins	$<4.00\times10^{-18}$	GWAS Catalog	25340798	1
Monocyte count	$<2.00\times10^{-7}$	GWAS Catalog	23314186	1

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)
ACKR2	missense variant	ENST00000493193	?	ENSP00000476581	A/V	gCg/gTg	?	?	1
ACKR2	missense variant	ENST00000442925	?	ENSP00000396150	A/V	gCg/gTg	?	?	1
ACKR2	missense variant	ENST00000497921	?	ENSP00000476901	A/V	gCg/gTg	?	?	1
ACKR2	missense variant	ENST00000494619	?	ENSP00000477157	A/V	gCg/gTg	?	?	1
ACKR2	missense variant	ENST00000422265	NM_001296.4	ENSP00000416996	A/V	gCg/gTg	?	?	1
ACKR2	missense variant	ENST00000273145	?	ENSP00000273145	A/V	gCg/gTg	?	?	1
ACKR2	missense variant	ENST00000492609	?	ENSP00000477475	A/V	gCg/gTg	?	?	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ACKR2	downstream gene variant	90	ENST00000463699	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)

CYP8B1	ENST00000437102	?	ENSP00000404499	1
KRBOX1	ENST00000426937	?	ENSP00000413859	1
RP11-136C24.3	ENST00000451200	?	?	1
RP11-141M3.6	ENST00000487368	?	?	1
RP11-141M3.6	ENST00000471537	?	?	1
RP11-141M3.6	ENST00000496604	?	?	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ACKR2	ENST00000460855	?	1
ACKR2	ENST00000498111	?	1