Locus 185 (rs35030851)

SNiPAcard

Block annotations

Block info

genomic range	chr9:27,196,743-27,214,579
block size	17,837 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.279 [-1.679 – 1.68]	gene(s) hit or close-by	RNA5SP280, RP11-179D22.1, TEK
phastCons	μ = 0.412 [0 – 0.995]	eQTL gene(s)	–
GERP++	μ = -0.588 [-6.48 – 4.64]	potentially regulated gene(s)	TEK
CADD score	μ = 7.173 [0.031 – 17.78]	disease gene(s)	TEK

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TEK	VENOUS MALFORMATIONS, MULTIPLE CUTANEOUS AND MUCOSAL	DECIPHER	MIM:600195
TEK	VENOUS MALFORMATIONS, MULTIPLE CUTANEOUS AND MUCOSAL	OMIM	MIM:600195
TEK	Mucocutaneous venous malformations	OrphaNet	OrphaNet:2451

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)
TEK	missense variant	ENST00000519080	?	ENSP00000428337	L/V	Ttg/Gtg	?	?	1
TEK	missense variant	ENST00000406359	NM_001290077.1	ENSP00000383977	L/V	Ttg/Gtg	?	?	1
TEK	missense variant	ENST00000519097	?	ENSP00000430686	L/V	Ttg/Gtg	?	?	1
TEK	missense variant	ENST00000380036	NM_000459.3	ENSP00000369375	L/V	Ttg/Gtg	?	?	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000865827	1	ENST00000406359, ENST00000519097, ENST00000519080, ENST00000380036	TEK

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RNA5SP280	upstream gene variant, downstream gene variant	799	ENST00000411230	?	?	4
RP11-179D22.1	upstream gene variant, downstream gene variant	856	ENST00000422804	?	?	5
TEK	downstream gene variant	119	ENST00000519080	?	ENSP00000428337	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
TEK	ENST00000380036	NM_000459.3	ENSP00000369375	Q	caA/caG	1
TEK	ENST00000406359	NM_001290077.1	ENSP00000383977	Q	caA/caG	1
TEK	ENST00000519097	?	ENSP00000430686	Q	caA/caG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TEK	ENST00000380036	NM_000459.3	ENSP00000369375	7
TEK	ENST00000615002	?	ENSP00000480251	7
TEK	ENST00000406359	NM_001290077.1	ENSP00000383977	7
TEK	ENST00000519097	?	ENSP00000430686	7
TEK	ENST00000519080	?	ENSP00000428337	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
TEK	ENST00000615002	?	ENSP00000480251	2