Locus 189 (rs3848570)

SNiPAcard

Block annotations

Block info

genomic range	chr19:8,137,843-8,149,247
block size	11,405 bp
variant count	10 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.143 [-4.325 – 3.177]	gene(s) hit or close-by	ELAVL1, FBN3
phastCons	μ = 0.232 [0 – 1]	eQTL gene(s)	–
GERP++	μ = -0.950 [-7.86 – 2.27]	potentially regulated gene(s)	–
CADD score	μ = 3.837 [0.226 – 22.4]	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)
FBN3	missense variant	ENST00000600128	?	ENSP00000470498	H/R	cAc/cGc	?	?	1
FBN3	missense variant	ENST00000270509	NM_032447.3	ENSP00000270509	H/R	cAc/cGc	?	?	1
FBN3	missense variant	ENST00000601739	?	ENSP00000472324	H/R	cAc/cGc	?	?	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FBN3	downstream gene variant, upstream gene variant	987	ENST00000595036	?	?	4
FBN3	downstream gene variant	2841	ENST00000601281	?	ENSP00000472729	1
FBN3	downstream gene variant	3462	ENST00000598269	?	?	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
FBN3	ENST00000270509	NM_032447.3	ENSP00000270509	H	caT/caC	1
FBN3	ENST00000600128	?	ENSP00000470498	H	caT/caC	1
FBN3	ENST00000601739	?	ENSP00000472324	H	caT/caC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ELAVL1	ENST00000351593	?	ENSP00000264073	10
FBN3	ENST00000270509	NM_032447.3	ENSP00000270509	9
FBN3	ENST00000600128	?	ENSP00000470498	9
FBN3	ENST00000601739	?	ENSP00000472324	9

FBN3	ENST00000595036	?	?	6

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
FBN3	ENST00000595036	?	2