

# SNiPACard

## Block annotations

### Block info

genomic range	chr19:8,137,843-8,149,247 <i>e!</i>
block size	11,405 bp
variant count	10 variants

### Basic features

Conservation/deleteriousness		Linked genes	
phyloP	$\mu = -0.143$ [-4.325 – 3.177]	gene(s) hit or close-by	ELAVL1 <i>e!</i> , FBN3 <i>e!</i>
phastCons	$\mu = 0.232$ [0 – 1]	eQTL gene(s)	-
GERP++	$\mu = -0.950$ [-7.86 – 2.27]	potentially regulated gene(s)	-
CADD score	$\mu = 3.837$ [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 <i>e!</i>	missense variant	ENST00000600128 <i>e!</i>	?	ENSP00000470498 <i>e!</i>	H/R	cAc/cGc	?	?	1
FBN3 <i>e!</i>	missense variant	ENST00000270509 <i>e!</i>	NM_032447.3	ENSP00000270509 <i>e!</i>	H/R	cAc/cGc	?	?	1
FBN3 <i>e!</i>	missense variant	ENST00000601739 <i>e!</i>	?	ENSP00000472324 <i>e!</i>	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 <i>e!</i>	downstream gene variant, upstream gene variant	987	ENST00000595036 <i>e!</i>	?	?	4
FBN3 <i>e!</i>	downstream gene variant	2841	ENST00000601281 <i>e!</i>	?	ENSP00000472729 <i>e!</i>	1
FBN3 <i>e!</i>	downstream gene variant	3462	ENST00000598269 <i>e!</i>	?	?	1

## Putative effect on transcript

### Synonymous coding variant

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

### Intron variant

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

FBN3 *e!*      ENST00000595036 *e!*      ?      ?      6

**Non-coding exon variant**

gene	affected transcript	RefSeq id	variant(s)
FBN3 <i>e!</i>	ENST00000595036 <i>e!</i>	?	2

