Locus 47 (rs45602433)

SNiPAcard

Block annotations

Block info

genomic range	chr9:117,793,413-117,835,899
block size	42,487 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.608 [-0.255 – 1.75]	gene(s) hit or close-by	TNC
phastCons	μ = 0.057 [0 – 0.211]	eQTL gene(s)	–
GERP++	μ = 1.703 [-1.46 – 3.39]	potentially regulated gene(s)	–
CADD score	μ = 8.642 [3.163 – 18.2]	disease gene(s)	TNC

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TNC	DEAFNESS, AUTOSOMAL DOMINANT 56	OMIM	MIM:615629
TNC	Autosomal dominant nonsyndromic sensorineural deafness type DFNA	OrphaNet	OrphaNet:90635

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)
TNC	missense variant	ENST00000341037	?	ENSP00000339553	H/R	cAt/cGt	?	?	1
TNC	missense variant	ENST00000537320	?	ENSP00000443478	H/R	cAt/cGt	?	?	1
TNC	missense variant	ENST00000542877	?	ENSP00000442242	H/R	cAt/cGt	?	?	1
TNC	missense variant	ENST00000535648	?	ENSP00000438152	H/R	cAt/cGt	?	?	1
TNC	missense variant	ENST00000350763	NM_002160.3	ENSP00000265131	H/R	cAt/cGt	?	?	1
TNC	missense variant	ENST00000423613	?	ENSP00000411406	H/R	cAt/cGt	?	?	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001474309 (promoter flanking region)	1	NHLF	DNase1
		HSMMtube	H3K27ac, DNase1
		Osteobl	H3K36me3, H3K27ac
		blood (DND-41)	H3K27me3
		cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K4me1, H3K27me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3, DNase1
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
TNC	downstream gene variant	4968	ENST00000498724	?	?	1
TNC	downstream gene variant	3545	ENST00000473855	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TNC	ENST00000423613	?	ENSP00000411406	3
TNC	ENST00000460345	?	?	1
TNC	ENST00000542877	?	ENSP00000442242	3
TNC	ENST00000544972	?	ENSP00000445380	3
TNC	ENST00000476680	?	?	2
TNC	ENST00000537320	?	ENSP00000443478	3
TNC	ENST00000535648	?	ENSP00000438152	3
TNC	ENST00000341037	?	ENSP00000339553	3
TNC	ENST00000498724	?	?	2
TNC	ENST00000350763	NM_002160.3	ENSP00000265131	3

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
TNC	ENST00000498724	?	1
TNC	ENST00000473855	?	1