Locus 193 (rs241890)

SNiPAcard

Block annotations

Block info

genomic range	chr22:33,242,162-33,242,162
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.451	gene(s) hit or close-by	RP1-309I22.2, SYN3, TIMP3
phastCons	0.008	eQTL gene(s)	–
GERP++	1.6	potentially regulated gene(s)	–
CADD score	2.879	disease gene(s)	TIMP3

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TIMP3	FUNDUS DYSTROPHY, PSEUDOINFLAMMATORY, OF SORSBY	OMIM	MIM:136900
TIMP3	Sorsby's fundus dystrophy	OrphaNet	OrphaNet:59181

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SYN3	ENST00000462268	?	?	1
SYN3	ENST00000358763	NM_003490.3, NM_001135774.1, NM_133633.2	ENSP00000351614	1
TIMP3	ENST00000266085	NM_000362.4	ENSP00000266085	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP1-309I22.2	ENST00000624184	?	1