

Block annotations

Block info

genomic range	chr22:33,242,162-33,242,162 <i>e!</i>
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 <i>e!</i> , SYN3 <i>e!</i> , TIMP3 <i>e!</i>
phastCons	0.008	eQTL gene(s)	-
GERP++	1.6	potentially regulated gene(s)	-
CADD score	2.879	disease gene(s)	TIMP3 <i>e!</i>

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TIMP3 <i>e!</i>	FUNDUS DYSTROPHY, PSEUDOINFLAMMATORY, OF SORSBY	OMIM	MIM:136900 
TIMP3 <i>e!</i>	Sorsby's Fundus dystrophy	OrphaNet	OrphaNet:59181 

Putative effect on transcript

Intron variant

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

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP1-309I22.2 <i>e!</i>	ENST00000624184 <i>e!</i>	?	1

