

Block annotations

Block info



genomic range	chr3:186,393,547-186,397,474 <i>e!</i>
block size	3,928 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	$\mu = -0.544$ [-1.333 – -0.092]	gene(s) hit or close-by	HRG <i>e!</i> , RP11-573D15.8 <i>e!</i>
phastCons	$\mu = 0.000$ [0 – 0.001]	eQTL gene(s)	-
GERP++	$\mu = 0.212$ [-1.91 – 2.35]	potentially regulated gene(s)	-
CADD score	$\mu = 5.246$ [2.169 – 15.47]	disease gene(s)	HRG <i>e!</i>

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
HRG <i>e!</i>	THROMBOPHILIA DUE TO HISTIDINE-RICH GLYCOPROTEIN DEFICIENCY	OMIM	MIM:613116 
HRG <i>e!</i>	Hereditary thrombophilia due to congenital histidine-rich (poly-L) glycoprotein deficiency	OrphaNet	OrphaNet:217467 

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)
HRG <i>e!</i>	missense variant	ENST00000232003 <i>e!</i>	NM_000412.3	ENSP00000232003 <i>e!</i>	C/R	Tgt/Cgt	?	?	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
HRG <i>e!</i>	downstream gene variant	596	ENST00000495413 <i>e!</i>	?	?	2
HRG <i>e!</i>	downstream gene variant	587	ENST00000232003 <i>e!</i>	NM_000412.3	ENSP00000232003 <i>e!</i>	2
RP11-573D15.8 <i>e!</i>	upstream gene variant	1240	ENST00000626006 <i>e!</i>	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HRG <i>e!</i>	ENST00000495413 <i>e!</i>	?	?	3
HRG <i>e!</i>	ENST00000232003 <i>e!</i>	NM_000412.3	ENSP00000232003 <i>e!</i>	2
RP11-573D15.8 <i>e!</i>	ENST00000627469 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000626649 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000625877 <i>e!</i>	?	?	5

RP11-573D15.8 <i>e!</i>	ENST00000625386 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000630331 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000628190 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000630726 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000630315 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000626633 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000630178 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000629106 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000627830 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000625741 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000628253 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000625303 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000629451 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000626845 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000628505 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000625839 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000630864 <i>e!</i>	?	?	5
RP11-573D15.8 <i>e!</i>	ENST00000628728 <i>e!</i>	?	?	5

#### Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
HRG <i>e!</i>	ENST00000495413 <i>e!</i>	?	1

