Locus 43 (rs1042445)

SNiPAcard

Block annotations

Block info

genomic range	chr3:186,393,547-186,397,474
block size	3,928 bp
variant count	5 variants

Basic features

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

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
HRG	THROMBOPHILIA DUE TO HISTIDINE-RICH GLYCOPROTEIN DEFICIENCY	OMIM	MIM:613116
HRG	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	missense variant	ENST00000232003	NM_000412.3	ENSP00000232003	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	downstream gene variant	596	ENST00000495413	?	?	2
HRG	downstream gene variant	587	ENST00000232003	NM_000412.3	ENSP00000232003	2
RP11-573D15.8	upstream gene variant	1240	ENST00000626006	?	?	4

Putative effect on transcript

Intron variant

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

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

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
HRG	ENST00000495413	?	1