SNiPAcard

Block annotations

Block info

genomic range	chr3:186,365,143-186,396,645
block size	31,503 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.461 [-0.432 – 2.533]	**gene(s) hit or close-by**	FETUB, HRG, RP11-573D15.8
phastCons	μ = 0.011 [0 – 0.027]	**eQTL gene(s)**	–
GERP++	μ = -1.116 [-5.26 – 2.97]	**potentially regulated gene(s)**	–
CADD score	μ = 3.494 [1.577 – 6.299]	**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	OMIM
HRG	Hereditary thrombophilia due to congenital histidine-rich (poly-L) glycoprotein deficiency	OrphaNet	OrphaNet:217467	orphanet

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001381859 (promoter flanking region)	1	cervix (HeLa-S3)	DNase1, Jund, H3K4me1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHEK)	H3K4me1, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FETUB	downstream gene variant	2442	ENST00000488561	?	?	1
HRG	downstream gene variant	4058	ENST00000468154	?	?	1
HRG	downstream gene variant	616	ENST00000232003	NM_000412.3	ENSP00000232003	1
HRG	downstream gene variant	625	ENST00000495413	?	?	1
RP11-573D15.8	downstream gene variant	708	ENST00000629451	?	?	2
RP11-573D15.8	downstream gene variant	1266	ENST00000630726	?	?	2
RP11-573D15.8	upstream gene variant	4536	ENST00000455926	?	?	1
RP11-573D15.8	downstream gene variant	4412	ENST00000625839	?	?	1
RP11-573D15.8	downstream gene variant, upstream gene variant	765	ENST00000625826	?	?	3
RP11-573D15.8	downstream gene variant	4485	ENST00000628728	?	?	1
RP11-573D15.8	downstream gene variant	647	ENST00000627469	?	?	2
RP11-573D15.8	upstream gene variant	4338	ENST00000626006	?	?	1
RP11-573D15.8	upstream gene variant	4532	ENST00000428501	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FETUB	ENST00000265029	NM_014375.2	ENSP00000265029	2
FETUB	ENST00000382134	?	ENSP00000371569	2
FETUB	ENST00000435961	?	ENSP00000393851	2
FETUB	ENST00000382136	?	ENSP00000371571	2
FETUB	ENST00000450521	?	ENSP00000404288	2
FETUB	ENST00000420570	?	ENSP00000405438	2
FETUB	ENST00000431018	?	ENSP00000396581	2
HRG	ENST00000232003	NM_000412.3	ENSP00000232003	1
HRG	ENST00000495413	?	?	1
RP11-573D15.8	ENST00000627469	?	?	2
RP11-573D15.8	ENST00000628190	?	?	4
RP11-573D15.8	ENST00000626649	?	?	4
RP11-573D15.8	ENST00000428501	?	?	2
RP11-573D15.8	ENST00000625877	?	?	2
RP11-573D15.8	ENST00000626633	?	?	4
RP11-573D15.8	ENST00000625839	?	?	1
RP11-573D15.8	ENST00000630331	?	?	4
RP11-573D15.8	ENST00000625386	?	?	4
RP11-573D15.8	ENST00000625303	?	?	4
RP11-573D15.8	ENST00000628253	?	?	4
RP11-573D15.8	ENST00000626845	?	?	4
RP11-573D15.8	ENST00000630315	?	?	4
RP11-573D15.8	ENST00000455926	?	?	2
RP11-573D15.8	ENST00000630178	?	?	4
RP11-573D15.8	ENST00000626006	?	?	1
RP11-573D15.8	ENST00000629106	?	?	4
RP11-573D15.8	ENST00000627830	?	?	2
RP11-573D15.8	ENST00000630864	?	?	4
RP11-573D15.8	ENST00000630726	?	?	2
RP11-573D15.8	ENST00000628505	?	?	4
RP11-573D15.8	ENST00000629451	?	?	2
RP11-573D15.8	ENST00000625741	?	?	4
RP11-573D15.8	ENST00000628728	?	?	1