Locus 128 (rs8071756)

SNiPAcard

Block annotations

Block info

genomic range	chr17:1,574,342-1,587,212
block size	12,871 bp
variant count	14 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.511 [-1.525 – 0.101]	**gene(s) hit or close-by**	PRPF8, RP11-961A15.3
phastCons	μ = 0.077 [0 – 0.922]	**eQTL gene(s)**	–
GERP++	μ = -0.848 [-3.44 – 1.67]	**potentially regulated gene(s)**	–
CADD score	μ = 6.553 [1.41 – 16]	**disease gene(s)**	PRPF8

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
PRPF8	RETINITIS PIGMENTOSA 13	OMIM	MIM:600059
PRPF8	Retinitis pigmentosa	OrphaNet	OrphaNet:791

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001338852 (promoter)	1	embryonic stem cell (H1ESC)	SP1, H3K36me3, PolII, H3K9ac, H3K27ac, Yy1, TAF1, H3K4me2, H3K4me3, DNase1
		HSMMtube	H3K4me2, H3K9ac, H3K27ac, DNase1, H3K36me3, H2AZ, H3K4me3
		blood (K562)	H3K4me3, DNase1, H3K36me3, H3K4me2, PolII, Cfos, H3K79me2, Cmyc, H3K9ac, Max
		skin (NHDF-AD)	DNase1, H3K9ac, H3K4me2, H3K4me3
		muscle (HSMM)	DNase1, H3K9ac, H2AZ, H3K79me2, H3K4me3, H3K27ac
		liver (HepG2)	H2AZ, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3, H3K36me3, DNase1, H3K4me1, TAF1, H3K79me2, Gabp, Yy1, PolII
		lung (IMR90)	H3K9ac, H3K4me3, H3K36me3, H3K4me2, DNase1, H4K8ac, H3K79me2, H3K27ac
		blood (GM12878)	H3K9ac, H3K36me3, H3K4me2, Cfos, H3K27ac, Pbx3, PolII, H2AZ, DNase1, Cmyc, Yy1, Gabp, H3K79me2, H3K4me3
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, DNase1
		skin (NHEK)	H3K4me1, H3K27ac, H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K27ac, H2AZ, H3K4me3, H3K36me3, H3K4me2
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K4me1, H3K4me2, H3K27ac, H4K20me1
		breast (HMEC)	DNase1, H3K4me3, H3K4me2, H3K36me3
		cervix (HeLa-S3)	H3K4me3, H3K79me2, Gabp, PolII, H3K36me3, DNase1, TAF1, H3K27ac, H3K9ac, Jund, H3K4me2
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	PolII, DNase1, H3K36me3, H3K9ac, H3K4me2, Max, H3K4me3
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PRPF8	downstream gene variant	358	ENST00000571346	?	?	2
PRPF8	downstream gene variant, upstream gene variant	806	ENST00000573716	?	?	10
PRPF8	downstream gene variant, upstream gene variant	987	ENST00000574217	?	?	10
PRPF8	downstream gene variant	142	ENST00000577001	?	ENSP00000458151	5

PRPF8	downstream gene variant, upstream gene variant	11	ENST00000576958	?	?	8
PRPF8	downstream gene variant, upstream gene variant	1620	ENST00000574728	?	?	5
PRPF8	downstream gene variant, upstream gene variant	364	ENST00000576407	?	?	10
RP11-961A15.3	upstream gene variant	808	ENST00000615078	?	?	3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
PRPF8	ENST00000572621	?	ENSP00000460348	3	3	3
PRPF8	ENST00000304992	NM_006445.3	ENSP00000304350	3	3	3
PRPF8	ENST00000577001	?	ENSP00000458151	3	3	3

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
PRPF8	ENST00000572621	?	ENSP00000460348	1
PRPF8	ENST00000304992	NM_006445.3	ENSP00000304350	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PRPF8	ENST00000572621	?	ENSP00000460348	10
PRPF8	ENST00000304992	NM_006445.3	ENSP00000304350	10
PRPF8	ENST00000576958	?	?	3
PRPF8	ENST00000577001	?	ENSP00000458151	6
PRPF8	ENST00000571346	?	?	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PRPF8	ENST00000574728	?	1
PRPF8	ENST00000574217	?	1
PRPF8	ENST00000576958	?	1