SNiPAcard

Block annotations

Block info

genomic range	chr13:113,780,041-113,781,942
block size	1,902 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.869 [-1.9 – 0.01]	**gene(s) hit or close-by**	F10, F10-AS1, KARSP2
phastCons	μ = 0.001 [0 – 0.002]	**eQTL gene(s)**	F10, F7
GERP++	μ = -0.050 [-1.29 – 0.635]	**potentially regulated gene(s)**	–
CADD score	μ = 3.276 [1.392 – 4.335]	**disease gene(s)**	F10, F7

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
F10	FACTOR X DEFICIENCY	OMIM	MIM:227600
F7	FACTOR VII DEFICIENCY	OMIM	MIM:227500
F10	Congenital factor X deficiency	OrphaNet	OrphaNet:328
F7	Congenital factor VII deficiency	OrphaNet	OrphaNet:327

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
F10	?	ENSG00000126218	lung	1.30×10-6 (p-value)	GTEx Portal V6	1
F10	?	ENSG00000126218	transformed fibroblasts	4.84×10-8 (p-value)	GTEx Portal V6	3
F7	?	ENSG00000057593	sun exposed skin	1.02×10-5 (p-value)	GTEx Portal V6	1
F7	?	ENSG00000057593	unexposed skin	1.63×10-8 (p-value)	GTEx Portal V6	3
F10	?	ENSG00000126218	aorta	7.31×10-8 (p-value)	GTEx Portal V6	3
F10	?	ENSG00000126218	tibial artery	4.98×10-8 (p-value)	GTEx Portal V6	3
F10	?	ENSG00000126218	thyroid	3.89×10-6 (p-value)	GTEx Portal V6	3

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
F10-AS1	downstream gene variant	527	ENST00000424635	?	?	3
KARSP2	upstream gene variant	2788	ENST00000415696	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
F10	ENST00000483537	?	?	3
F10	ENST00000477269	?	?	3
F10	ENST00000410083	?	ENSP00000386320	3
F10	ENST00000409306	?	ENSP00000387092	3
F10	ENST00000375559	NM_000504.3	ENSP00000364709	3
F10	ENST00000375551	?	ENSP00000364701	3