

Block annotations

Block info

genomic range	chr3:186,472,816-186,475,666 <i>e!</i>
block size	2,851 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	$\mu = -0.641$ [-1.457 – 0.176]	gene(s) hit or close-by	PSMD10P2 <i>e!</i> , RP11-573D15.8 <i>e!</i>
phastCons	$\mu = 0.250$ [0 – 0.5]	eQTL gene(s)	-
GERP++	$\mu = -1.788$ [-3.68 – 0.104]	potentially regulated gene(s)	-
CADD score	$\mu = 6.903$ [2.027 – 11.78]	disease gene(s)	-

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001381876 <i>e!</i> (promoter flanking region)	1	endothelium (HUVEC) HSMMtube Osteobl blood (K562) nervous (NH-A) skin (NHEK)	H3K27me3 H3K27me3 H3K27me3 H3K27me3 H3K27me3 H3K4me2, H3K9ac, H3K27ac, H3K4me1, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PSMD10P2 <i>e!</i>	upstream gene variant	3227	ENST00000432890 <i>e!</i>	?	?	1
PSMD10P2 <i>e!</i>	upstream gene variant	2816	ENST00000626135 <i>e!</i>	?	?	1
PSMD10P2 <i>e!</i>	upstream gene variant	3137	ENST00000627340 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	upstream gene variant	1272	ENST00000596632 <i>e!</i>	?	?	2
RP11-573D15.8 <i>e!</i>	upstream gene variant	150	ENST00000626586 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	downstream gene variant	1998	ENST00000608173 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	upstream gene variant	358	ENST00000625690 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	upstream gene variant	1563	ENST00000609652 <i>e!</i>	?	?	2
RP11-573D15.8 <i>e!</i>	upstream gene variant	659	ENST00000629201 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	upstream gene variant	209	ENST00000599314 <i>e!</i>	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
RP11-573D15.8 <i>e!</i>	ENST00000625869 <i>e!</i>	?	?	2
RP11-573D15.8 <i>e!</i>	ENST00000596329 <i>e!</i>	?	?	2
RP11-573D15.8 <i>e!</i>	ENST00000625690 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	ENST00000626520 <i>e!</i>	?	?	2
RP11-573D15.8 <i>e!</i>	ENST00000629954 <i>e!</i>	?	?	2

RP11-573D15.8 <i>e!</i>	ENST00000599314 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	ENST00000626586 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	ENST00000626978 <i>e!</i>	?	?	2
RP11-573D15.8 <i>e!</i>	ENST00000629201 <i>e!</i>	?	?	1
RP11-573D15.8 <i>e!</i>	ENST00000628291 <i>e!</i>	?	?	2
RP11-573D15.8 <i>e!</i>	ENST00000608173 <i>e!</i>	?	?	1

