

SNiPACard

Block annotations

Block info

| | |
|---------------|--------------------------------------|
| genomic range | chr1:40,203,722-40,226,046 <i>e!</i> |
| block size | 22,325 bp |
| variant count | 20 variants |


































Basic features












| Conservation/deleteriousness | | Linked genes | |
|------------------------------|--------------------------------|-------------------------------|--|
| phyloP | $\mu = -0.180$ [-1.636 – 0.37] | gene(s) hit or close-by | BMP8B <i>e!</i> , PPIE <i>e!</i> |
| phastCons | $\mu = 0.043$ [0 – 0.341] | eQTL gene(s) | BMP8B <i>e!</i> , OXCT2 <i>e!</i> , PPIE <i>e!</i> |
| GERP++ | $\mu = -0.163$ [-5.61 – 2.6] | potentially regulated gene(s) | MACF1 <i>e!</i> , PPIE <i>e!</i> |
| CADD score | $\mu = 4.840$ [0.066 – 12.78] | disease gene(s) | – |

Direct effect on regulation

cis-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|-----------------|---------------------------|---------------------------|-------------------------|----------------------------------|--------------------|------------|
| PPIE <i>e!</i> | ? | ENSG00000084072 <i>e!</i> | atrial appendage | 8.35×10 ⁻³⁰ (p-value) | GTEx Portal V6 | 20 |
| BMP8B <i>e!</i> | ? | ENSG00000116985 <i>e!</i> | transformed fibroblasts | 6.30×10 ⁻¹¹ (p-value) | GTEx Portal V6 | 20 |
| PPIE <i>e!</i> | ? | ENSG00000084072 <i>e!</i> | transformed fibroblasts | 2.17×10 ⁻³² (p-value) | GTEx Portal V6 | 20 |
| PPIE <i>e!</i> | ? | ENSG00000084072 <i>e!</i> | breast | 1.32×10 ⁻⁴⁴ (p-value) | GTEx Portal V6 | 20 |
| PPIE <i>e!</i> | ? | ENSG00000084072 <i>e!</i> | blood | 8.23×10 ⁻⁵¹ (p-value) | GTEx Portal V6 | 20 |
| PPIE <i>e!</i> | ENST00000497370 <i>e!</i> | ILMN_2326737 <i>e!</i> | monocyte | 3.23×10 ⁻¹⁸ (p-value) | Fairfax et al. | 1 |
| PPIE <i>e!</i> | ENST00000372835 <i>e!</i> | | b-cell | 4.24×10 ⁻²⁴ (p-value) | Fairfax et al. | 1 |
| PPIE <i>e!</i> | ENST00000495526 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000470018 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000480169 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000324379 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000475350 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000474804 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000372830 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000482751 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000356511 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ? | ENSG00000084072 <i>e!</i> | skeletal muscle | 2.42×10 ⁻¹⁹ (p-value) | GTEx Portal V6 | 20 |
| PPIE <i>e!</i> | ? | ENSG00000084072 <i>e!</i> | putamen | 1.32×10 ⁻¹¹ (p-value) | GTEx Portal V6 | 20 |
| PPIE <i>e!</i> | ? | ENSG00000084072 <i>e!</i> | blood | 2.01×10 ⁻⁴ (q-value) | SeeQTL DB (HapMap) | 6 |
| PPIE <i>e!</i> | ? | ENSG00000084072 <i>e!</i> | EBV lymphocytes | 1.64×10 ⁻¹³ (p-value) | GTEx Portal V6 | 20 |
| PPIE <i>e!</i> | ? | ENSG00000084072 <i>e!</i> | unexposed skin | 6.98×10 ⁻⁵¹ (p-value) | GTEx Portal V6 | 20 |
| BMP8B <i>e!</i> | ? | ENSG00000116985 <i>e!</i> | sun exposed skin | 1.15×10 ⁻⁷ (p-value) | GTEx Portal V6 | 20 |
| PPIE <i>e!</i> | ? | ENSG00000084072 <i>e!</i> | sun exposed skin | 8.53×10 ⁻⁸⁶ (p-value) | GTEx Portal V6 | 20 |
| PPIE <i>e!</i> | ? | ENSG00000084072 <i>e!</i> | ovary | 9.46×10 ⁻²² (p-value) | GTEx Portal V6 | 20 |
| PPIE <i>e!</i> | ? | ENSG00000084072 <i>e!</i> | left ventricle | 9.03×10 ⁻¹¹ (p-value) | GTEx Portal V6 | 20 |

| | | | | | | |
|-----------------|---|---------------------------|---------------------------|----------------------------------|--|----|
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | aorta | 4.36×10 ⁻⁴² (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | subcutaneous adipocytes | 6.11×10 ⁻⁸² (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | caudate basal ganglia | 2.43×10 ⁻¹⁴ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | visceral adipocytes | 4.07×10 ⁻⁵⁰ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | stomach | 6.63×10 ⁻³⁹ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | cortex | 1.80×10 ⁻¹¹ (p-value) | GTEX Portal V6  | 20 |
| OXCT2 <i>e!</i> | ? | ENSG00000198754 <i>e!</i> | tibial nerve | 2.10×10 ⁻¹⁰ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | tibial nerve | 1.91×10 ⁻⁶⁹ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | pituitary | 4.54×10 ⁻¹⁹ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | esophagus mucosa | 8.49×10 ⁻⁶³ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | testis | 4.29×10 ⁻⁴³ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | hypothalamus | 9.76×10 ⁻¹¹ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | frontal cortex | 4.38×10 ⁻⁷ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | uterus | 4.53×10 ⁻¹³ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | pancreas | 3.18×10 ⁻³⁴ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | muscularis mucosae | 1.08×10 ⁻³⁶ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | lung | 4.70×10 ⁻⁷⁶ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | tibial artery | 5.45×10 ⁻⁵⁰ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | thyroid | 4.69×10 ⁻⁷³ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | prostate | 3.73×10 ⁻²¹ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | transverse colon | 1.09×10 ⁻³¹ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | liver | 1.18×10 ⁻¹⁸ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | terminal ileum | 3.83×10 ⁻²³ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | nucleus accumbens | 1.11×10 ⁻¹¹ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | spleen | 6.43×10 ⁻²⁴ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | coronary artery | 2.58×10 ⁻²² (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | sigmoid colon | 3.11×10 ⁻¹⁹ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | vagina | 2.89×10 ⁻¹⁷ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | gastroesophageal junction | 6.49×10 ⁻²¹ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | cerebellar hemisphere | 4.63×10 ⁻¹⁰ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | adrenal gland | 1.45×10 ⁻¹⁸ (p-value) | GTEX Portal V6  | 20 |
| PPIE <i>e!</i> | ? | ENSG0000084072 <i>e!</i> | cerebellum | 1.45×10 ⁻¹⁰ (p-value) | GTEX Portal V6  | 20 |
| BMP8B <i>e!</i> | ? | ENSG00000116985 <i>e!</i> | cerebellum | 1.45×10 ⁻⁷ (p-value) | GTEX Portal V6  | 20 |

| | | | | | | |
|-----------------|---------------------------|---------------------------|-------------|----------------------------------|---|----|
| PPIE <i>e!</i> | ? | ENSG00000084072 <i>e!</i> | hippocampus | 1.14×10 ⁻¹⁰ (p-value) | GTEx Portal V6  | 20 |
| PPIE <i>e!</i> | ENST00000497370 <i>e!</i> | ILMN_2326737 <i>e!</i> | skin | 7.60×10 ⁻¹⁸ (p-value) | MuTHER consortium  | 4 |
| PPIE <i>e!</i> | ENST00000495526 <i>e!</i> | | blood | 6.33×10 ⁻³⁵ (p-value) | MuTHER consortium  | 4 |
| PPIE <i>e!</i> | ENST00000372835 <i>e!</i> | | adipocyte | 5.02×10 ⁻¹⁰ (p-value) | MuTHER consortium  | 4 |
| PPIE <i>e!</i> | ENST00000470018 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000480169 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000324379 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000475350 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000474804 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000372830 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000482751 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000356511 <i>e!</i> | | | | | |
| BMP8B <i>e!</i> | ENST00000372827 <i>e!</i> | ILMN_2123415 <i>e!</i> | blood | 7.55×10 ⁻¹³ (p-value) | MuTHER consortium  | 4 |
| PPIE <i>e!</i> | ENST00000497370 <i>e!</i> | ILMN_1680341 <i>e!</i> | skin | 9.11×10 ⁻⁷⁵ (p-value) | MuTHER consortium  | 4 |
| PPIE <i>e!</i> | ENST00000495526 <i>e!</i> | | blood | 2.84×10 ⁻³⁵ (p-value) | MuTHER consortium  | 4 |
| PPIE <i>e!</i> | ENST00000372835 <i>e!</i> | | adipocyte | 3.73×10 ⁻³⁸ (p-value) | MuTHER consortium  | 4 |
| PPIE <i>e!</i> | ENST00000470213 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000324379 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000475350 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000474804 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000482751 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000372830 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000356511 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000497370 <i>e!</i> | ILMN_2326737 <i>e!</i> | monocyte | 3.68×10 ⁻⁶⁴ (p-value) | Zeller et al.  | 2 |
| PPIE <i>e!</i> | ENST00000495526 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000372835 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000470018 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000480169 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000324379 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000475350 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000474804 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000372830 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000482751 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000356511 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000497370 <i>e!</i> | ILMN_1680341 <i>e!</i> | monocyte | 5.74×10 ⁻⁵² (p-value) | Zeller et al.  | 2 |
| PPIE <i>e!</i> | ENST00000495526 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000372835 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000470213 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000324379 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000475350 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000474804 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000482751 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000372830 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000356511 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000475350 <i>e!</i> | 202494_at <i>e!</i> | blood | 4.50×10 ⁻¹⁶ (p-value) | Dixon et al.  | 1 |
| PPIE <i>e!</i> | ENST00000497370 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000372835 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000495526 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000482751 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000470213 <i>e!</i> | | | | | |
| PPIE <i>e!</i> | ENST00000324379 <i>e!</i> | | | | | |

Putative effect on regulation

FANTOM5 expressed promoter

| SNiPA promoter id | variant(s) | associated transcript(s) | gene |
|---------------------------|------------|--|----------------|
| FFCP00000418809 <i>e!</i> | 1 | ENST00000497370 <i>e!</i> , ENST00000495526 <i>e!</i> , ENST00000372835 <i>e!</i> , ENST00000456578 <i>e!</i> , ENST00000480169 <i>e!</i> , ENST00000470213 <i>e!</i> , ENST00000324379 <i>e!</i> , ENST00000475350 <i>e!</i> , ENST00000474804 <i>e!</i> , ENST00000482751 <i>e!</i> , ENST00000372830 <i>e!</i> , ENST00000485507 <i>e!</i> , ENST00000356511 <i>e!</i> | PPIE <i>e!</i> |

ENCODE promoter-associated DHS

| SNiPA promoter id | variant(s) | associated gene(s) |
|---------------------------|------------|--------------------|
| ENCP00000001965 <i>e!</i> | | |


ENCODE promoter-associated distal DHS (Enhancer)

| SNiPA enhancer id | variant(s) | associated SNiPA promoter id | associated gene(s) |
|---------------------------|------------|---|---|
| ENCE00000015243 <i>e!</i> | 1 | ENCP00000001935 ENCP00000001943 ENCP00000001939 | MACF1 <i>e!</i> MACF1 <i>e!</i> MACF1 <i>e!</i> |

Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|--|------------|---|--|
| ENSR00001579074 <i>e!</i> (CTCF binding site) | 1 | embryonic stem cell (H1ESC) liver (HepG2) lung (IMR90) blood (K562) muscle (HSMM) | H3K27me3 H3K27me3 DNase1, H4K8ac H2AZ DNase1 |
| ENSR00000534967 <i>e!</i> (promoter) | 3 | embryonic stem cell (H1ESC) HSMMtube blood (K562) skin (NHDF-AD) muscle (HSMM) liver (HepG2) blood (GM12878) lung (IMR90) nervous (NH-A) skin (NHEK) NHLF Osteobl blood (DND-41) breast (HMEC) cervix (HeLa-S3) monocytes (Monocytes-CD14+) endothelium (HUVEC) A549 | H3K36me3, H3K27me3, PolII, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3, DNase1 H3K9ac, H3K4me2, H3K4me3, H2AZ, DNase1 Yy1, H3K27ac, Max, H3K9ac, Gabp, H3K79me2, HEY1, H2AZ, TAF1, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3 H3K4me2, H3K9ac, DNase1, H3K4me3 H2AZ, H3K4me3, H3K4me2, H3K9ac, H3K36me3, DNase1 H3K27me3, DNase1, H3K4me3, H3K9ac, Gabp, PolII, TAF1 PolII, H2AZ, DNase1, Yy1, Gabp, H3K79me2, H3K4me3, H3K27ac, H3K9ac DNase1, H4K8ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K9ac H3K36me3, H3K4me3, H3K4me2, H3K9ac H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1 DNase1, H3K4me3, H3K9ac, H3K27ac H2AZ, H3K27ac, H3K4me3, H3K4me2, H3K36me3 H3K4me3, H3K9ac H3K27ac, H3K4me3, H3K9ac, H3K4me2 H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, Gabp, PolII, H3K36me3, DNase1 DNase1, H3K4me2, H3K27ac, H3K36me3, H3K27me3, H3K4me3 H3K36me3, Cmyc, H3K4me3, PolII H3K4me3, H3K4me2, H3K9ac, DNase1, H3K36me3 |

Variation in RISC binding site

| gene | variant(s) | affected transcript(s) | targeting miRNA(s) |
|----------------|------------|---|--|
| PPIE <i>e!</i> | 3 | ENST00000324379 <i>e!</i> ENST00000356511 <i>e!</i> ENST00000372830 <i>e!</i> ENST00000372835 <i>e!</i> ENST00000470213 <i>e!</i> ENST00000475350 <i>e!</i> ENST00000482751 <i>e!</i> ENST00000495526 <i>e!</i> ENST00000497370 <i>e!</i> | hsa-miR-411-5p  |

Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|-----------------|--|---------------|---------------------------|----------------|---------------------------|------------|
| BMP8B <i>e!</i> | downstream gene variant | 1787 | ENST00000372827 <i>e!</i> | NM_001720.3 | ENSP00000361915 <i>e!</i> | 3 |
| BMP8B <i>e!</i> | downstream gene variant | 3177 | ENST00000397360 <i>e!</i> | ? | ENSP00000380518 <i>e!</i> | 1 |
| PPIE <i>e!</i> | upstream gene variant | 149 | ENST00000372830 <i>e!</i> | NM_001195007.1 | ENSP00000361918 <i>e!</i> | 3 |
| PPIE <i>e!</i> | upstream gene variant, downstream gene variant | 134 | ENST00000456578 <i>e!</i> | ? | ? | 5 |
| PPIE <i>e!</i> | upstream gene variant, downstream gene variant | 136 | ENST00000495526 <i>e!</i> | ? | ENSP00000434989 <i>e!</i> | 6 |
| PPIE <i>e!</i> | upstream gene variant | 133 | ENST00000356511 <i>e!</i> | NM_203456.2 | ENSP00000348904 <i>e!</i> | 3 |
| PPIE <i>e!</i> | upstream gene variant, downstream gene variant | 149 | ENST00000497370 <i>e!</i> | ? | ENSP00000433475 <i>e!</i> | 6 |
| PPIE <i>e!</i> | upstream gene variant, downstream gene variant | 149 | ENST00000372835 <i>e!</i> | ? | ENSP00000361925 <i>e!</i> | 6 |
| PPIE <i>e!</i> | upstream gene variant, downstream gene variant | 238 | ENST00000480169 <i>e!</i> | ? | ? | 10 |
| PPIE <i>e!</i> | upstream gene variant, downstream gene variant | 127 | ENST00000485507 <i>e!</i> | ? | ? | 6 |
| PPIE <i>e!</i> | upstream gene variant, downstream gene variant | 149 | ENST00000470213 <i>e!</i> | ? | ENSP00000431714 <i>e!</i> | 6 |
| PPIE <i>e!</i> | upstream gene variant, downstream gene variant | 138 | ENST00000482751 <i>e!</i> | ? | ? | 6 |
| PPIE <i>e!</i> | upstream gene variant, downstream gene variant | 130 | ENST00000324379 <i>e!</i> | NM_006112.3 | ENSP00000312769 <i>e!</i> | 6 |
| PPIE <i>e!</i> | downstream gene variant | 662 | ENST00000470018 <i>e!</i> | ? | ENSP00000436689 <i>e!</i> | 7 |
| PPIE <i>e!</i> | upstream gene variant, downstream gene variant | 156 | ENST00000475350 <i>e!</i> | ? | ENSP00000434888 <i>e!</i> | 6 |
| PPIE <i>e!</i> | upstream gene variant | 825 | ENST00000467741 <i>e!</i> | ? | ? | 1 |
| PPIE <i>e!</i> | upstream gene variant, downstream gene variant | 152 | ENST00000474804 <i>e!</i> | ? | ENSP00000432396 <i>e!</i> | 6 |

Putative effect on transcript

Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|----------------|---------------------------|----------------|---------------------------|------------|
| PPIE <i>e!</i> | ENST00000482751 <i>e!</i> | ? | ? | 13 |
| PPIE <i>e!</i> | ENST00000497370 <i>e!</i> | ? | ENSP00000433475 <i>e!</i> | 13 |
| PPIE <i>e!</i> | ENST00000475350 <i>e!</i> | ? | ENSP00000434888 <i>e!</i> | 13 |
| PPIE <i>e!</i> | ENST00000372835 <i>e!</i> | ? | ENSP00000361925 <i>e!</i> | 13 |
| PPIE <i>e!</i> | ENST00000485507 <i>e!</i> | ? | ? | 3 |
| PPIE <i>e!</i> | ENST00000456578 <i>e!</i> | ? | ? | 3 |
| PPIE <i>e!</i> | ENST00000470018 <i>e!</i> | ? | ENSP00000436689 <i>e!</i> | 7 |
| PPIE <i>e!</i> | ENST00000495526 <i>e!</i> | ? | ENSP00000434989 <i>e!</i> | 13 |
| PPIE <i>e!</i> | ENST00000474804 <i>e!</i> | ? | ENSP00000432396 <i>e!</i> | 10 |
| PPIE <i>e!</i> | ENST00000480169 <i>e!</i> | ? | ? | 4 |
| PPIE <i>e!</i> | ENST00000372830 <i>e!</i> | NM_001195007.1 | ENSP00000361918 <i>e!</i> | 17 |
| PPIE <i>e!</i> | ENST00000470213 <i>e!</i> | ? | ENSP00000431714 <i>e!</i> | 13 |
| PPIE <i>e!</i> | ENST00000356511 <i>e!</i> | NM_203456.2 | ENSP00000348904 <i>e!</i> | 17 |
| PPIE <i>e!</i> | ENST00000324379 <i>e!</i> | NM_006112.3 | ENSP00000312769 <i>e!</i> | 13 |

3'-UTR variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|-----------------|---------------------------|-------------|---------------------------|------------|
| BMP8B <i>e!</i> | ENST00000372827 <i>e!</i> | NM_001720.3 | ENSP00000361915 <i>e!</i> | 1 |

PPIE *e!* ENST00000495526 *e!* ? ENSP00000434989 *e!* 5

Non-coding exon variant

| gene | affected transcript | RefSeq id | variant(s) |
|----------------|---------------------------|-----------|------------|
| PPIE <i>e!</i> | ENST00000480169 <i>e!</i> | ? | 1 |

