

Additional files

Additional file 1

File format: .pdf

Title and description of the data:

Nucleotide sequence alignment of human *PNMT* and rat *Pnmt* genes. Exons of the gene are boxed. mRNA start site is marked with +1. Translation start site is indicated by **ATG** (red/bold) and **red** arrow; **translation end sites** for human and rat are in purple/bold. Seven **SNPs** identified by resequencing *PNMT* in this study are marked bold with grey shading. The sequence of human-specific *AluSp element* in the first intron is marked in italics and its start/end sites are indicated by **black** arrows. Sequences of regulatory elements and transcription factor binding sites (TFBS) are underlined. Experimentally confirmed **rat *Pnmt* expression regulating elements** [6, 59-62] are typed in *italics* on green shading background. Predicted regulatory elements for human *PNMT* [12-13] are typed with regular font (no shading).

hPNMT 1 TTCAGGAAGACCACGGAGATGCCACAGGCAGAATTGGCTTCCCATCTGGGAGATAGGTGG
rPnmt 1 -----CT.CAGCCC..T..G.--..A.T.C..G..C..

hPNMT 61 GGAGACCCTGGCATTTTGACAGCCAGAACCTGGGGTGCTGAGCAGAATCTTCATGCCTGG
rPnmt 33 .A....AAGCA....A..-TGC.AG.C....A.A.CA.CA...G.G.C..GA.G...A..

hPNMT 121 CCTGGCCGCCTTCGGAGGGAAAGCTGGAGGGTTGGGTGCGAGAGGAGTGGGGTCAGAGCCC
rPnmt 92 ..CT...AA..C.CC..T.GGA.A...T.-...AC.--...G..T.G.....G.AG.ATG

hPNMT 181 CTACATCCGCAGGACCCCAAATCGGCTGGGCCCAAGGCCCGGAC-TGCGCT-CCCCGGT
rPnmt 149 -----.....GT..TCT.C.T.TGAAAAA....AAAGT.CG.A.....G..T.T..

hPNMT 239 GGCCCCGGCGGCCCT-CCGCGAATGCGTCCCTGCCCTGCCCAAGCCCTCTGCCCT
rPnmt 201 A.TGG.T.T...AG.A..AA.....T..T....-----A.T.....TT..

hPNMT 298 CACCCGGGTCCGGCGCCGCCCGCAAGTGGCGGGAACAACCCGAACCCGAACCTTCTGTG
rPnmt 246 T..A..A.....T.T--...T..CC...TA.....CTT.....T--.....-A.G

hPNMT 358 CTCGGGAGCCCCAGATAAGCGGCTGGGAACCCGCGGGGCCGCAGGGGAGGCCCGGCTG
rPnmt 297 ..T.CCG.A..T.....A..GCAT.G...-A.....A.....-T.....A..

hPNMT 418 TTCCGCCCGCTAAGTGCATTAG-CACAGCTCACCTCCCCTATCGCGCCTGCCATCGGACG
rPnmt 355 ...T..TT.....AA.....A.C.CA.....G---...T.TG.....AA.T.

hPNMT 477 GGCAGTGCCGCGCCCTGCTCTGGGGCCCCCGGAGCGACCACAGCGGAGGCCGGAACGGAC
rPnmt 412 CT.G.G..A.A.A.....-A.TA...A.-.C...AGA.A.....A...A..G

hPNMT 537 TGTCCTTTCTGGGGCGGGTGGGGAGGGGGTGTGCTGGAGGGCCCGGTGGCATAGCAAC
rPnmt 468AA.GA..A.A.A..C.....-----

hPNMT 597 GGACGAGAGAGCCCTGGAGGAGGGGCGGGGAGGGGGGAGTTGTGTGGCAGTTCTAAGGGAA
rPnmt 499 -----.....T.....T....-----A.....-GA...A.A.G

hPNMT 657 GGGTGGGTGCTGGGACGGGTGTCCGGGAGGGAGGGGAGCCTGGCGGGTCTGGGGCCTCG
rPnmt 534 ...A.TT..T--A.A...AC..C.AGA..G.A....-----

hPNMT 717 TCGCGGAGGGCGCTGCGAGGGGAAACTGGGGAAAGGGCCTAATTCCCCAGTCTCCACCT
rPnmt 570 C..G.A...AT....G..TC.....CA..A.-----T..CTCAG....

hPNMT 777 CGAATCAGGAAAGAGAAGGGGCGGGCTGCTGGGCAAAAAGAGGTGAATGGCTGCGGGGGGGC
rPnmt 614 ----.TG.....AT...A.....A.GCTG..C.-.G.--...T..A...

hPNMT 837 TGGAGAAGAGAGATGGGAGGGGCCGGCCGGGGGTGAGGGGGTCTAAAGATTGTGGGG
rPnmt 667G...TC.-...G.CCA.T.--AA.T.A..CTG....AG.AG.GC.GCACT..A


hPNMT 897 GTGAGGAACTGAGGGTGGGGGCGCCAGAGCGGGACTCGGGGCGGGGACAGGCGAGGCG
rPnmt 724 .C..TC..-----A..C.AT.A...T..A-----...TT.T.A.--C

hPNMT 957 GAGGGCGAGGGCTGCGGGAGCAAGTACGGAGCCGGGGGTGTGGGGGAGCATTGCCCTGC
rPnmt 761 A..A.TA.A.----.A....TGG.-G.A.GAG..AT.T.C.AAA..G..CCCT..A.AT.

hPNMT 1017 AGCCGCCGCCCACTCACCTCCGGTGTGTCTGCAGCCCGGACACTAAGGGAGATGGATGA
rPnmt 816 TC..C..T...G.C.-.G.GC..CC...C.GC..T.A.....CT.....-

hPNMT 1077 ATGGGTGGGGAGGATGCGGCGCACATGGCCCCGGGCGGCTCGGCGGTCAGCTGCCCCCC
rPnmt 874 --.....ACAGA...T....GC.TC...G..TCATCC...A..A.C..C.CA..C.TGT

hPNMT 1137 CACAGCGGA--CCGGTCGGGGCGGGGG---TCGGGCGGTAGAAAAAGGGCCGCGAGGCG
rPnmt 932 G.TG.A..GGT.T..G....G..A..GGAC.CA.TA....T...G..ATG.G...TT



hPNMT 1192 AGCGGGGCACTGGGCGGACCGCGGCGGCAGC**ATG**AGC---GGCGCAGACCGTAGCCCCAA
rPnmt 992A.--A.A.....-..T.AA.A.G.....GA.CGT...T.....CG.AG.A..C
+1 rat exon 1

hPNMT 1249 TCGGGGCGCAGCCCCTGACTCGGCCCGGGCCAGGCGGCGGTGGCTTCGGCCTACCAGCG
rPnmt 1049 ...A..GATG.A.T.....C.A..T.....A.A..C..C.T..T.....

hPNMT 1309 CTTCGAGCCGCGCGCCTACCTCCGCAACAACACTACGCGCCCCCTCGGGGGACCTGTGCAA
rPnmt 1109 ...T.....C..T.....T.....T.....A...

hPNMT 1369 CCCGAACGGCGTTCGGGCGGCTGGAAGCTGCGCTGCTTGGCGCAGACCTTCGCCACCGGTGA
rPnmt 1169 ...TG.T..T..G.....T.....T.....A...A..AGT...T.....)....

hPNMT 1429 GCGGGGAAACTGAGGCACGAGGACAAGAGGTCGTCGGGGAGTGAAAGCAGGCGCAGGG
rPnmt 1229 ..ACT.....A.....A..GC...A..A.A..A.A.....-----G...


hPNMT 1489 AAATAAAAAGAAGGAAAGGGAGACAGACCAGGCGCCTAACAGATGGGGACCAAGAAACAA
rPnmt 1282 .G.G.G...C...A-----T.TGC.C..A.....A.C.A...GC.

hPNMT 1549 GAGATAGCTGAGAGGTGCAAACAGAAGAGAAAAAGGAGCAACATCCCTTAGGAGAGGGGC
rPnmt 1325 ..AC..T---.GT...T.GC.G.C.C.A.C.-----

hPNMT 1609 AGAGGAGAGAGAGGTGGAGAGAGGGGGCGGAGAGTGCTCAGAATTGAGAGCTAAGGTGGG
rPnmt 1354 -----A.....CGA.AG...----...GCTA...G..AT..

hPNMT 1669 GGATGCAGGACAGACTGAGGTGGAGATGCATAGGAGGAAATGGAGGCAGATGTGGGACAG
rPnmt 1387 ...G..-----T.....C.---A.T..T..A..CC.G.

hPNMT 1729 GGGTGAGAAACTCCAGGATTTCTCGCTGAGCCTGGCTGGTAGGTATAGTTGTTTTCTTT
rPnmt 1419 AAT..GA.G...-----A..A.ACAACT..A--.G..CT...

 **Alu sequence**

hPNMT 1789 CTTTTTCTTTATTTTATTTTCAATTTATTTACTTATTTTATTTTATTTTATTTGTTTTGAGAC
rPnmt 1455-----

hPNMT 1849 GGAGTTTCGCTCTTGTTGCCAGGCTGGAGTACAATGGCGCCATCTCGGCTCACTGCAAC
rPnmt 1459 -----.....G.-----


GRE

hPNMT 1909 CTCCGCCTCCCCGGTTCAAGCGATTCTTGCCTCAGCTTCCCTAGTAGCTGGGATTAC
rPnmt 1470 -----...T.A.AG..GAGAGAC..A.....CC-T

hPNMT 1969 AGGCATGCGCCCCCATGCCTGGCTAATTTATTTGTATTTTATTTAGTAGAGACGGGACTTCTC
rPnmt 1499 G..AGA....-----GAGG.G..T....-----

hPNMT 2029 CATGTTGGTCAGGCTGGTCTCGAACTCCCAACCTTAGGATCCACCCACCCCGGCTCCCA
rPnmt 1524 ---.....--..A...-----..

hPNMT 2089 AAGTGCTGGGATTACAGGTGTGAGCCACTGCGCCCGGCCAGTAGGTATAGTCTTCTAGAT
rPnmt 1537 ...A.A-.A.GC.C...-----C..T...

Alu sequence 

hPNMT 2149 GTGAAACCTGAGTCTCAGAGCGGTGAAGTTCCTTCCGAAGGGCAGCCCATGTTGGAGCT
rPnmt 1560 ..C..G---.GA....-----...CA.TA..TT.T.A.-----A.....

hPNMT 2209 GGGTTCAGTCTAACTCTGGGGCCAATGCTTTTTCCAGATGGAGACACATTTGCAGAGGAG
rPnmt 1597 A.T...T---.GTG....AA..CGA.T-----....C.CTG.CA...C-----

hPNMT 2269 AAGGAAGAACTAGAGAGAGGCAGGAGATGCAGGGGAGGGAAGGGTAAGGAGGCAGGGGC
rPnmt 1642 ----...TG..G.G...-----C.C.AG..A.....GT..C.---.....AA.

exon 2

hPNMT 2329 TGCCTGGGCTGGCTGGCACCAGGACCCCTCTTCTCTGCCCTGCCAGGTGAAGTGTCCGG
rPnmt 1688 CC....ATA.-----C.C.....T..A.-..)....G.....T..

hPNMT 2389 ACGCACCCTCATCGACATTGGTTCAGGCCCCACCGTGTACCAGCTGCTCAGTGCCTGCAG
rPnmt 1731 ..AGGT.....T.....C..C.....A.A.....C.....TGC

hPNMT 2449 CCTTTGAGGACATCACCATGACAGATTTCTGGAGGTCAACCGCCAGGAGCTGGGGCC
rPnmt 1791C.....C..T.....G.....A.T

hPNMT 2509 CTGGCTGCAGGAGGAGCCGGGGCCTTCAACTGGAGCATGTACAGCCAACATGCCTGCCT
rPnmt 1851GA....A..A..A.....G.....TG...T....G...T.....

hPNMT 2569 CATTGAGGGCAAGGGGTAAGGACTGGGGGTGAGGGTTGGGGAGGAGGCTTCCCATAGAG
rPnmt 1911 ..C...A......G..A.....C--.G.A.C.TCAC.C.GA.TG.-----G.

hPNMT 2629 TGGCTGGTTG-GGGCAACAGAGGCCTGAGCGT--AGAACAGCCTTGAGCCCTGCCTTGTG
rPnmt 1963 C..G..C.G.A....GGAG...A.....C.CTG.GGT..T.-.....C.....

exon 3

hPNMT 2686 CCTCCTGCACAGGGAATGCTGGCAGGATAAGGAGCGCCAGCTGCGAGCCAGGGTGAAACG
rPnmt 2022 ..C....T....A..G.C.....G..A..A.....C....G.....G..

hPNMT 2746 GGTCCTGCCCATCGACGTGCACCAGCCCCAGCCCCTGGGTGCTGGGAGCCCAGCTCCCCT
rPnmt 2082 A...T.....T..T.....A.....A...TC.G...TG..A....

hPNMT 2806 GCCTGCTGACGCCTGGTCTCTGCCTTCTGCTTGGAGGCTGTGAGCCCAGATCTTGCCAG
rPnmt 2142T.....C.....G....CC.A..

hPNMT 2866 CTTTCAGCGGGCCCTGGACCACATCACCACGCTGCTGAGGCCTGGGGGGCACCTCCTCCT
rPnmt 2202 ..C.G..A...TT..T..T....G.....C....TA.T.....T.

hPNMT 2926 CATCGGGGCCCTGGAGGAGTCGTGGTACCTGGCTGGGGAGGCCAGGCTGACGGTGGTGCC
rPnmt 2262T.....AT.T....T..

hPNMT 2986 AGTGTCTGAGGAGGAGGTGAGGGAGGCCCTGGTGCCTAGTGGCTACAAGGTCCGGGACCT
rPnmt 2322A.....CT.....T..TG.....A....

hPNMT 3046 CCGCACCTATATCATGCCTGCCACCTTCAGACAGGCGTAGATGATGTCAAGGGCGTCTT
rPnmt 2382 T.....C.....C.GC..G..T..G....C.....TA....

hPNMT 3106 CTTCGCCTGGGCTCAGAAGGTTGGGCTGTGAGGGCTGTACCTGGTGCCTGTGGCCCCCA
rPnmt 2442 ...T.....C.....A.A.A.G..CAG.T.-----..AG..T.CA.-----TTG

hPNMT 3166 CCCACCTGGATTCCCTGTTCTTTGAAGTGGCACCTAATAAAGAAATAATACCCTGCCGCT
rPnmt 2490 ..TC...A-.C...T.A.CACCC.....T..C.G.C...A..TAT-

hPNMT 3226 GCGGTCAGTGCTGTGTGGCTCTCCTGGGAAGCAGCAAGGGCCCAGAGATCTGAGTGTC
rPnmt 2548 ---...T.....T....A.....A.AC.CA..G.TA..AAA..GTC..C...-CT

hPNMT 3286 CGGGTAGGGGAGACATTCACCCTAGGCTTTTTTCCAGAAGCTTCCTTGAGGCTAGCATT
rPnmt 2604 TA.AAG.AACCA.TCC.AG.TT.TTCT...GCC.T.....C.T.G...A.GCT..T..

hPNMT 3346 CTGTACCACTCATCTTCCCAAATAAGGAAGGCCAAGGTCAGGGGAGCTCACCGGGCAC
rPnmt 2664G....C.....C...TCC.C.G.-----...C..CAC.CA.GTG..A...G.

hPNMT 3406 CTGCAGTGCACCAGGCACTTGTGATATGCCATTGTGTC
rPnmt 2717 T..TGTG.TT.TC.A.CTGCAG.C.-..A.GCT..AG