

INK2J00048R_DNMT1_Y511C_A03_AA
5599 bp

| DNMT1 |
| :--- |
| DNMT1-202 |
| CACTGGAGATAAAGAGTTTGAGACCAGCCTGCTACACATAGTGAAACCCCGTCCCTACTAAAAATACAACAATTAGCCGGGCATG |
| GTGACCTCTATTTCTCAAACTCTGGTCGGACGATGTGTATCACTTTGGGGCAGGGATGATTTTTATGTTGTTAATCGGCCCGTAC |

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| DNMT1 |
| ---: |
| DNMT1-202 |

ATTCTCTCATTGCCTCGTGCCTGATCTGAAGTCTGCACGAAGACCCGCCTTCACGGCTTAGCTGGTAAGCATGTGCTTTGTTTCC
 TAAGAGAGTAACGGAGCACGGACTAGACTTCAGACGTGCTTCTGGGCGGAAGTGCCGAATCGACCATTCGTACACGAAACAAAGG DNMT1

DNMT1-202

TGTCTAGTGTGTACTGTAAGCACGGTCACCTGTGTCCCATCGACACCGGCCTCATCGAGAAGAATATCGAACTCTTCTTTTCTGG
 ACAGATCACACATGACATTCGTGCCAGTGGACACAGGGTAGCTGTGGCCGGAGTAGCTCTTCTTATAGCTTGAGAAGAAAAGACC


TTCAGCAAAACCAATCTATGATGATGACCCATCTCTTGAAGGTAAGGAATAGTCCGGGATTATGTTTGGGGCACACTTTAAAAAC
 AAGTCGTTTTGGTTAGATACTACTACTGGGTAGAGAACTTCCATTCCTTATCAGGCCCTAATACAAACCCCGTGTGAAATTTTTG


AGCCAGGCAGGGTGGCTCACATCTGTAATCCTAGCACTTTGGGGGTCTGAGGCCAGAGGATCACTTGAGCCCGGGAGTTTGAGAC
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| DNMT1 |
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| DNMT1-202 |




CAGGAGCAGCTGTAGGCCAAGTCATTGTGATGACTGAGGAGCACCCGACCTGGCTGATGGGATGTCTGCTGAGTGGGCGCTTAGA بнبץبץ GTCCTCGTCGACATCCGGTTCAGTAACACTACTGACTCCTCGTGGGCTGGACCGACTACCCTACAGACGACTCACCCGCGAATCT
 DNMT1-202

GCGCCTCCTGGGTTTGGGATGTGAGTCAGAGCCTTTAGTCCATTTCCTTTTTCTGCTCTAGGTGGTGTTAATGGCAAAAATCTTG بН+中 $+\boldsymbol{+}$ CGCGGAGGACCCAAACCCTACACTCAGTCTCGGAAATCAGGTAAAGGAAAAAGACGAGATCCACCACAATTACCGTTTTTAGAAC


GCCCCATAAATGAATGGTGGATCACTGGCTTTGATGGAGGTGAAAAGGCCCTCATCGGCTTCAGCACCTGTAAGTGTGTGGCCCA
 CGGGGTATTTACTTACCACCTAGTGACCGAAACTACCTCCACTTTTCCGGGAGTAGCCGAAGTCGTGGACATTCACACACCGGGT


TCATAGGCTGGCCGGGGTCTGAAAGGGGCCTTCATGTTCTCCTTCCTGGGGGCTGACGGGGCTCTGGTGGGAATTCTCAGCAGGC

AGTATCCGACCGGCCCCAGACTTTCCCCGGAAGTACAAGAGGAAGGACCCCCGACTGCCCCGAGACCACCCTTAAGAGTCGTCCG


PCR Forward
TTAGTTATGACTGGCTCTGGAACTG
TTGCAGAAGGCCATGTGACTGGGAACCTTAGCAGGTTCAGTTGGGGTAGCCTCTTGTGTTAGTTATGACTGGCTCTGGAACTGAC

AACGTCTTCCGGTACACTGACCCTTGGAATCGTCCAAGTCAACCCCATCGGAGAACACAATCAATACTGACCGAGACCTTGACTG




GCATGGTGGTGCATACCTGTAATCCCAGCTACTTGGGAGGCTATGGCAGGAGAATCACGTGAACCCAGCAGACAGAGGTTGCAGT
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DNMT1
DNMT1-202


GAGCCGAGATCACGCCACTGCCCTCCAGCCTGGGTGACGGAGCAAGACTCTGTCCTCCCCTGAAAAAAAAAAAAGAGCAGGTCTT بץبץبץب CTCGGCTCTAGTGCGGTGACGGGAGGTCGGACCCACTGCCTCGTTCTGAGACAGGAGGGGACTTTTTTTTTTTTCTCGTCCAGAA


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DNMT1
DNMT1-202

DNMT1-202

| Sanger Sequencing |
| :--- |
| ACATTTGGGTACGGGATGAC |

CTGGCTCAGACAGGCTTCTTCAGAACAAGCCAGCTATGATGTGTTGTGCCCTATGTTTCTGACATTTGGGTACGGGATGACTTTT
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DNMT1
DNMT1-202

CTCTGTTCACCCCAGCATTTGCCGAATACATTCTGATGGATCCCAGTCCCGAGTATGCGCCCATATTTGGGCTGATGCAGGAGAA
 GAGACAAGTGGGGTCGTAAACGGCTTATGTAAGACTACCTAGGGTCAGGGCTCATACGCGGGTATAAACCCGACTACGTCCTCTT



Donor Template SNV -> REV


GATCTACATCAGCAAGATTGTGGTGGAGTTCCTGCAGAGCAATTCCGACTCGACCTATGAGGACCTGATCAACAAGATCGAGGTA
 CTAGATGTAGTCGTTCTAACACCACCTCAAGGACGTCTCGTTAAGGCTGAGCTGGATACTCCTGGACTAGTTGTTCTAGCTCCAT

DNMT1
DNMT1-202


Donor Template SNV -> REV

AGAGATCGAGGGTCCTCAGCATCCGGGATTCCCACTGGAAACTTGCCTTCAGAACCAGCAGACACTGTTCTTCAGTTGGATTTAG
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| DNMT1 |
| ---: |
| DNMT1-202 |

DNMT1-202

GCCAGTTTGGCTTAAGCATGAGAGAAACCTGTTCTCTTTCAAGACCACGGTTCCTCCTTCTGGCCTCAACTTGAACCGCTTCACA
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TGAAGACTGGAGACCGGGGAGGGTAGAGCATGGCCCACATCCTCTGTCCCAGTCCTCTGAGATGCTGGAACCTCTCCCGTAGGCG
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 DNMT1-202


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DNMT1-202

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GAAGAATTGGGTAGACCTCTTACCCTTCCCTTAAAATAAAGAGAGTGAATGGTATAAAGATTACTCCAACTTATAACGAAAAAAA




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DNMT1-202

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AAAAAACATTTTGTAGATGGGATTTTGCCATGTTGCCTAGGCTGGTCTTGAATGCCTGAACTCAAGCAGTCCTCCCGCCTTAGCA世нНبН ب TTTTTTGTAAAACATCTACCCTAAAACGGTACAACGGATCCGACCAGAACTTACGGACTTGAGTTCGTCAGGAGGGCGGAATCGT


TCCTGAGTTGCCGGGACTGCAGGAATTTGTCACACGCCTGGCTAATTTTAATTTTTTGTAGAGTTGGGGGTCTTAATATGTTGCC بץبץبץ
AGGACTCAACGGCCCTGACGTCCTTAAACAGTGTGCGGACCGATTAAAATTAAAAAACATCTCAACCCCCAGAATTATACAACGG


CAGGCTGGTCTtGAACTCTTGGACTCAAGTGATCCATCTACCTCTGCTTCAGCCTCCCAATGTACTGGTTCAGGTATGAGCCACC

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DNMT1-202
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 GCCGGAAGACTTTATGACCCTAATGTCCGTACTTGGTGACACGGGTCGGGACTAAACGGAAAAATTACGGTAAAAAATCCACACA

| DNMT1 |
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| DNMT1-202 |$\gg$



tGTGTGGGACAGCACCAGGATTCCTTCGTTAGGCATTGTCTCAGGACCTGTCCCTGTTATGAAGAAAACAGCCCCGGTTGGTCTT
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tCGATGATAACATCCCAGAGATGCCGTCACCCAAAAAAATGCACCAGGGGAAGAAGAAGAAACAGAACAAGAATCGCATCTCTTG



GGTCGGAGAAGCCGTCAAGGTAACCCTTGGAGCCCCTTGGTCAGCTCACTGCCATGTTCCCAATAAGCAGGTTG HبН H CCAGCCTCTTCGGCAGTTCCATTGGGAACCTCGGGGAACCAGTCGAGTGACGGTACAAGGGTTATTCGTCCAAC
DNMT1

/note $\quad=\quad$| gene ENSG00000130816 |
| :--- |
|  |
| Protein coding |

DNMT1-201
/note $\quad=$ primary transcript ENST00000340748

| DNMT1-202 |  |
| :--- | :--- |
| /note | $=$ primary transcript ENST00000359526 |


| DNMT1-227 |  |
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| /note | $=\quad$ primary transcript ENST000000592705 |

## DNMT1-229

/note

DNMT1-230
/note $=$ primary transcript ENST00000676610

| DNMT1-231 | $=\quad$primary transcript ENST000000676820 <br> Retained intron |
| :--- | :--- |
| /note |  |

DNMT1-232
/note $\quad=$ primary transcript ENST00000676868
Retained intron
DNMT1-233
/note

DNMT1-236
/note

DNMT1-237
/note $\quad=\quad$ primary transcript ENST000000677616 Nonsense mediated decay

DNMT1-238
/note

DNMT1-239
/note
DNMT1-240
/note $\quad=\quad$ primary transcript ENST00000677783 Retained intron

DNMT1-241
/note
DNMT1-242
/note

DNMT1-246
/note

DNMT1-247
/note $\quad=\quad$ primary transcript ENST00000678804
DNMT1-251
/note $\quad=\quad$ primary transcript ENST00000679103
DNMT1-252
$=$ primary transcript ENST00000679313
DNMT1-203
/note
= primary transcript ENST00000592705 Nonsense mediated decay
$=$ primary transcript ENST00000676604 protein_coding_CDS_not_defined Retained intron
$=$ primary transcript ENST00000677013 Nonsense mediated decay
$=$ primary transcript ENST00000677250 Nonsense mediated decay
$=$ primary transcript ENST00000677634 Nonsense mediated decay
$=$ primary transcript ENST00000677685 Nonsense mediated decay
$=$ primary transcript ENST00000677946
= primary transcript ENST00000678024 Retained intron
= primary transcript ENST00000678694 Retained intron
$=$ primary transcript ENST00000679103

1 .. 5599
5599 bp
$\square \quad \rightarrow \quad$ prim_transcript

1 .. $5599 \quad 5599 \mathrm{bp} \quad \square \quad \rightarrow \quad$ prim_transcript

1 .. $5599 \quad 5599 \mathrm{bp} \quad \square \quad \rightarrow \quad$ prim_transcript

1 .. $55995599 \mathrm{bp} \quad \square \quad \rightarrow \quad$ prim_transcript

1 .. 5599

1 .. 5599

1 .. 5599

1 .. 5599

1 .. 5599

1 .. 5599
5599 bp
$\square \quad \rightarrow$
prim_transcript

1 .. $55995599 \mathrm{bp} \quad \square \quad \rightarrow \quad$ prim_transcript

1 .. $5599 \quad 5599 \mathrm{bp} \square \quad \rightarrow \quad$ prim_transcript

1 .. $5599 \quad 5599 \mathrm{bp} \square \quad \rightarrow \quad$ prim_transcript

1 .. 5599

1 .. 5599
5599 bp
$\square \quad \rightarrow \quad$ prim_transcript

1 .. $55995599 \mathrm{bp} \square \quad \rightarrow \quad$ prim_transcript

1 .. 2278

Feature
Location
Size
/codon_start $=1$
/note $\quad=$ coding sequence ENSP00000345739
/translation $\quad=$ VYCKHGHLCPIDTGLIEKNIELFFSGSAKPIYDDDPSLE,,GGVNGKNLGPINEWWITGFDGGEKALIGFST,,SFAEYILMDPSPEYAPIFGLMQE KIYISKIVVEFLQSNSDSTYEDLINKIE,,TTVPPSGLNLNRFTEDSLLRHAQFVVEQVESYDEAGDSDEQPIFLTPCMRDLIKLAGVTLGQR,,R A QARRQTIRHSTREKDRGPTKATTTKLVYQIFDTFFAEQIEKDDREDKENAFKRRRCGVCE,,VCQQPECGKCKACKDMVKFGGSGRSKQAC QERR,,CPNMAMKEADDDEEVDDNIPEMPSPKKMHQGKKKKQNKNRISWVGEAVK 328 amino acids $=37.1 \mathrm{kDa}$
DNMT1-202
7 segments $=985 \mathrm{bp}$
/codon_start $=1$
/note $\quad=$ coding sequence ENSP00000352516
/translation $\quad=$ VYCKHGHLCPIDTGLIEKNIELFFSGSAKPIYDDDPSLE,,GGVNGKNLGPINEWWITGFDGGEKALIGFST,,SFAEYILMDPSPEYAPIFGLMQE KIYISKIVVEFLQSNSDSTYEDLINKIE,,TTVPPSGLNLNRFTEDSLLRHAQFVVEQVESYDEA GDSDEQPIFLTPCMRDLIKLAGVTLGQR,,R A QARRQTIRHSTREKDRGPTKATTTKLVYQIFDTFFAEQIEKDDREDKENAFKRRRCGVCE,,VCQQPECGKCKACKDMVKFGGSGRSKQAC QERR,,CPNMAMKEADDDEEVDDNIPEMPSPKKMHQGKKKKQNKNRISWVGEAVK 328 amino acids $=37.1 \mathrm{kDa}$

## DNMT1-230

433 .. $5544 \quad 5112$ bp $\quad \rightarrow \quad$ CDS

* 7 segments $=985 \mathrm{bp}$
/codon_start = 1
/note $\quad=$ coding sequence ENSP00000504236
/translation $\quad=\quad$ VYCKHGHLCPIDTGLIEKNIELFFSGSAKPIYDDDPSLE,,GGVNGKNLGPINEWWITGFDGGEKALIGFST,,SFAEYILMDPSPEYAPIFGLMQE KIYISKIVVEFLQSNSDSTYEDLINKIE,,TTVPPSGLNLNRFTEDSLLRHAQFVVEQVESYDEAGDSDEQPIFLTPCMRDLIKLAGVTLGQR,,R AQARRQTIRHSTREKDRGPTKATTTKLVYQIFDTFFAEQIEKDDREDKENAFKRRRCGVCE,,VCQQPECGKCKACKDMVKFGGSGRSKQAC QERR,,CPNMAMKEADDDEEVDDNIPEMPSPKKMHQ GKKKKQNKNRISWV GEAVK 328 amino acids $=37.1 \mathrm{kDa}$


Donor Template SNV -> REV 1860 .. $1959 \quad 100 \mathrm{bp} \quad \square \quad \mapsto \quad$ misc_feature
PAM 1919 .. $1921 \quad 3 \mathrm{bp} \quad \square \quad \mapsto \quad$ misc_feature



