

INK2J00065_POLG_G737R_E07_AB
29,664 bp
 AGGTGCAGAAGGTCGGTCATTTTCTTCGGTTCGACCTCGGGTTTCGGTCCACAAGACTGAGGGTCGCACCCCCAGGGACGTGGTT

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CCATGAGCCGCCTGCTCTGGAGGAAGGTGGCCGGCGCCACCGTCGGGCCAGGGCCGGTTCCAGCTCCGGGGCGCTGGGTCTCCAG
 GGTACTCGGCGGACGAGACCTCCTTCCACCGGCCGCGGTGGCAGCCCGGTCCCGGCCAAGGTCGAGGCCCCGCGACCCAGAGGTC
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CTCCGTCCCCGCGTCCGACCCCAGCGACGGGCAGCGGCGGCGGCAGCAGCAGCAGCAGCAGCAGCAGCAGCAGCAACAGCAGCCT بץبНبץ GAGGCAGGGGCGCAGGCTGGGGTCGCTGCCCGTCGCCGCCGCCGTCGTCGTCGTCGTCGTCGTCGTCGTCGTCGTTGTCGTCGGA


CAGCAGCCGCAAGTGCTATCCTCGGAGGGCGGGCAGCTGCGGCACAACCCATTGGACATCCAGATGCTCTCGAGAGGGCTGCACG
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AGCAAATCTTCGGGCAAGGAGGGGAGATGCCTGGCGAGGCCGCGGTGCGCCGCAGCGTCGAGCACCTGCAGAAGCACGGGCTCTG

TCGTTTAGAAGCCCGTTCCTCCCCTCTACGGACCGCTCCGGCGCCACGCGGCGTCGCAGCTCGTGGACGTCTTCGTGCCCGAGAC


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CGTGGAGGTCTGCTTGGCAGAGGGAACTTGCCCCACATTGGCGGTGGCCATATCCCCCTCGGCCTGGTAAGTAGGGGCAGGGTTG
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GGGACATAAGCAGGCATGGGGGCCCAGCTTAATAGTTTGTTTCAGTGAACATTTTCTGAGGTCCTGTTACGGGCTGGGTGCTCAC
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GTAGGGAGCGCTGATGTGTTGAATTAGGACTAGACCCCTGTTTATGTGGGACTCACTTTCTGGTGGGAAGATCACAGGCAGTAAG بـبCATCCCTCGCGACTACACAACTTAATCCTGATCTGGGGACAAATACACCCTGAGTGAAAGACCACCCTTCTAGTGTCCGTCATTC


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CCCAGCCTTTCCCATAAACGCCAGACCAACACCCCCCCGCCCCCCTGCCCCCGCCGGCAACGTTTCCAGACGCCCCCTTGAGGTG
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TCCAATCTCTCTTTTCTTTTTAAATAACTTTCTAAGTGCTACCCAAGTTTCTTTTTCAAACAATGATGGCAGTACTGTTTCTCCC ب+ $+\boldsymbol{+}$ AGGTTAGAGAGAAAAGAAAAATTTATTGAAAGATTCACGATGGGTTCAAAGAAAAAGTTTGTTACTACCGTCATGACAAAGAGGG


СтTTTTTTATTCTTCATTCCAGGATTAAAATACTATTTACAACCTTAATGCTTTCAGGCATGGCCAGCAAAAAAGTTGGCAGTTT بHץبН
GAAAAAAATAAGAAGTAAGGTCCTAATTTTATGATAAATGTTGGAATTACGAAAGTCCGTACCGGTCGTTTTTTCAACCGTCAAA


CTTTATTCCTATtGGAAGCTACATCTTTGTAAAGAAAGCTGCGAAATGTTAAATATGCAGTTGAAAATGGTGAAAACATGGCTAA

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CCTGGGAGTCTGACTGGTGGGTGGGGGTGGGTGCCACATGGTGCCCTAATAGCGGTCCACTTTGAACCTGGGCATGCCCCTGCCC HبH H
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GATTCATCCTGTACATGTGTGCAGCAGCATTTGTCATTTTTTTTTTTTTTGCCAATTCAATTAAATCTCCACCTTGGGTTCTGTT Hبץبץ CTAAGTAGGACATGTACACACGTCGTCGTAAACAGTAAAAAAAAAAAAAACGGTTAAGTTAATTTAGAGGTGGAACCCAAGACAA


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AGGGAGCAGTACCTGATCCAGGTAAGGTTCCTGGGGCCAACTGCAGGTTCTGGCATGGGATGGGCCAGGAGCCCTAATCTCAGTG НبН H TCCCTCGTCATGGACTAGGTCCATTCCAAGGACCCCGGTTGACGTCCAAGACCGTACCCTACCCGGTCCTCGGGATTAGAGTCAC
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CAGACAGCCTTGTTGACATGCTCAGGGCCACAGAGCTTTTGAGTGGCAGGGTTGGGGCCAGACCAGATAGCCCTGAAGGCTTTAT
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CTTAAAGGGTGGCTAGGATTTGGGAAGGCAGGCCCCTTACAGGTGATGGGGCAAGCATGAACAAGCATGAGGATTCTGTATTTGG



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TGTTGAAGGCTGTGTGCTGGGAGGGGAGGCTGTTTGAGGAGCTGAGGTGGGGCTGGAGGTCCACACCACCAAGCAGTGGTGGGCT ННبНبН Н ACAACTTCCGACACACGACCCTCCCCTCCGACAAACTCCTCGACTCCACCCCGACCTCCAGGTGTGGTGGTTCGTCACCACCCGA


 CCGGGGTGTCAACGTCGGAGGGAGGAAGGGAAGGGAAAAGAGGAGGAGGAGGAGTCCCAAGGGCGTACGCAAAGGACCTGTGGTA


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 AGgTCGTCGTCGATGGCGAGAAGAACCTCTCCACTCCCCCTCGGGTACACCCTTAGAGACCCCCAGTCACACAAGGACCATGGGC

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CAGTGGTGTCCATCTGGGCCCTGGACTCTGGATTTGACAGAGGCCCTGAAGGGGAGGGCCATGGAGTTGTGCTTGTGTGTCATGT
 gtcaccacaggtagacccgagacctgagacctanactgtctccgagacttcccctcccgatacctcaacacgancacacagtaca





ATtTTTTATGTTATACGTATTATATACTGTATTCTTAAAGTAAGCTAGATAAAAGAAAATGTATTAAGAAAATCATGAGGAGAAA HНН TAAAAAATACAATATGCATAATATATGACATAAGAATTTCATTCGATCTATTTTCTTTTACATAATTCTTTTAGTACTCCTCTTT


ATATGTTTACTATTCATTAGGTGGAAGTGGATCATCATAAAGATATCTATCCTTCACGTTGAGTAGGCTGAGGGCGGGGGTTGGG بнبץب TATACAAATGATAAGTAATCCACCTTCACCTAGTAGTATTTCTATAGATAGGAAGTGCAACTCATCCGACTCCCGCCCCCAACCC


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GACTTTTGAGCTGTGCCATGTCAGTGGCTGGGTTGAGGCTTGCTAAACCAGCTCAGGGATTGGGCCAGTCTTGCCTCCTGTGGTC HبН CTGAAAACTCGACACGGTACAGTCACCGACCCAACTCCGAACGATTTGGTCGAGTCCCTAACCCGGTCAGAACGGAGGACACCAG


ATTTATGGCAGCTCCTGGTGTTTGCCTCCAAGGTGTCCCCACCCAGTGACTCTGGCCGGCATGCTGGAGATGGGTGTCTCCTACC بнبץبץ TAAATACCGTCGAGGACCACAAACGGAGGTTCCACAGGGGTGGGTCACTGAGACCGGCCGTACGACCTCTACCCACAGAGGATGG


TGCCTGTCAACCAGAACTGGGAGCGTTACCTGGCAGAGGCACAGGGCACTTATGAGGAGCTCCAGCGGGAGATGAAGAAGTCGTT
 ACGGACAGTTGGTCTTGACCCTCGCAATGGACCGTCTCCGTGTCCCGTGAATACTCCTCGAGGTCGCCCTCTACTTCTTCAGCAA


GATGGATCTGGCCAATGATGCCTGCCAGCTGCTCTCAGGAGAGAGGTAGCCAGGCCTTGGGTGGGCAGGATCTAGGCAGGGGACT بץبץبץ CTACCTAGACCGGTTACTACGGACGGTCGACGAGAGTCCTCTCTCCATCGGTCCGGAACCCACCCGTCCTAGATCCGTCCCCTGA


GGCAGGTGGGCGGCCTAGCCTTCGGCTTAGCCTTAGCCCTGCCCTAGTGGACTGGCTCTGTAGGTACAAAGAAGACCCCTGGCTC H-H CCGTCCACCCGCCGGATCGGAAGCCGAATCGGAATCGGGACGGGATCACCTGACCGAGACATCCATGTTTCTTCTGGGGACCGAG


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CCTGGGTGAGCCCTGCCCACCCCCAGCAGTGTATCTAGAGTCTACCCTTGCTCCATTCTCAGGACAGCCCTGGTCTGGGTTCTGG +НННبН gGacccactcgagacgagtgaggatcgtcacatagatctcagatgagaicgaggtangagtcctgtcaggaccagacccaigacc

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CACAGAGGCATCATGCACATGTATACTTATTGACCTGCTGCCATTCAGTCACACTGTCTTCCAGTCCTATTCTCATTTGCTCACT
 GTGTCTCCGTAGTACGTGTACATATGAATAACTGGACGACGGTAAGTCAGTGTGACAGAAGGTCAGGATAAGAGTAAACGAGTGA


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 AACGATCAGAGTGGGACCCGAGTTGAGACCACTTGTCGGACGTTCTCTCGTGGGTCTTCCTGACCACAAAGAGATCTCCCCACCC



TTTTGTGTCCTGGCTTCTGACTAGTCCAGTGGGACTGACTTAGTGTCTTTGCTTTTGAAATATTCTTCTAGAGGATTCCATGGGG بHץبН
AAAACACAGGACCGAAGACTGATCAGGTCACCCTGACTGAATCACAGAAACGAAAACTTTATAAGAAGATCTCCTAAGGTACCCC


GTCCTGGCTAAAGCATCCCAGAGGAGGGGATGGCGGCTGTAGGCTGGGGTCACCAGAAAGCCCCAGGGCTTTGGAGGGTGGGTGG



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ACCGGAAGCTCTGCCCCCGGCTAGACGACCCTGCATGGACCCCGGGCCCCAGCCTCCTCAGCCTGCAGATGCGGGTCACACCTAA
 TGGCCTTCGAGACGGGGGCCGATCTGCTGGGACGTACCTGGGGCCCGGGGTCGGAGGAGTCGGACGTCTACGCCCAGTGTGGATT


ACTCATGGCACTTACCTGGGATGGCTTCCCTCTGCACTACTCAGAGCGTCATGGCTGGGGCTACTTGGTGCCTGGGCGGCGGGAC
 TGAGTACCGTGAATGGACCCTACCGAAGGGAGACGTGATGAGTCTCGCAGTACCGACCCCGATGAACCACGGACCCGCCGCCCTG


AACCTGGCCAAGCTGCCGACAGGTACCACCCTGGAGTCAGCTGGGGTGGTCTGCCCCTACAGGTAAGGCTTAGGCCCAGGGGAGG HبН TTGGACCGGTTCGACGGCTGTCCATGGTGGGACCTCAGTCGACCCCACCAGACGGGGATGTCCATTCCGAATCCGGGTCCCCTCC

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| POLG-201 |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| 630 |  |  | 1 | 1 | 635 | 1 |  | 1 | 1 | 640 |  |  | 1 |  |  | 645 | 1 |  |  |  | 650 |
| N | L | A | K | L | P | T | G | T | T | L | E | S | A |  | G | V | V | C | P | Y | R | POLG-201

AAGGGGCTGGAGCCTAGGGACCCCTTCCCCTGGCTGGTCAGCTCAGGCTAGTGGAAAGAGTTTGGGTTCAAGAGTCTGGGTTCAG

TTCCCCGACCTCGGATCCCTGGGGAAGGGGACCGACCAGTCGAGTCCGATCACCTTTCTCAAACCCAAGTTCTCAGACCCAAGTC

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 HبН ب GAGAGTGAGTGAGTGAGTGAGTGAGTGAGAGAGTGAGTGAGTGAGTGAGAGACCCAAGTCCAAAAAAGAAGGTACCGATGGGAAT


CCCTCTGGATCTCAGAGCTCTGGGAGGGAGTATGTTGAGATGTTCACAGTGGGGAGGACTAAAGGCCCTACTCTTGGGCCCAGAA НННН H GGGAGACCTAGAGTCTCGAGACCCTCCCTCATACAACTCTACAAGTGTCACCCCTCCTGATTTCCGGGATGAGAACCCGGGTCTT



GAGTGATAATTATGGGCCAAGCAGTGCTTCCCTTATTTGTTCCCCAAGGAGTCCCATGAGCTAGAATGGTTATCCCCATGTTGTA世بץبН CTCACTATTAATACCCGGTTCGTCACGAAGGGAATAAACAAGGGGTTCCTCAGGGTACTCGATCTTACCAATAGGGGTACAACAT


GTTGACAAAGGCTTGGTTGACTTAAGATCACAGACCCTGAGCTTTAGGCAGGCAGGTGTTGGGGAGAAACTTACAGTGGCCCAGA

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GTCATTTTCAGTTTGTCAATCAATCCCTGTCTAAAACCCGGGAAAGTGCTATCTGGTTCTGCCTCAGAGCTGATTCTGAGGACTA HץНبнبН CAGTAAAAGTCAAACAGTTAGTTAGGGACAGATTTTGGGCCCTTTCACGATAGACCAAGACGGAGTCTCGACTAAGACTCCTGAT

 AGACGGGTCTCGGTAGCTCAGGGACATGTCCTTCGTGACAGAGCTTGTCCCCTTCGTCGTCGACTACGGGGTCCTCCGGCCGGAC


GCGGAGGAGTTCCTGCTCACTGACAATAGTGCCATATGGCAAACGGTGAGGGCAGGCTCTGAACCTGAGCTTTGGGGAGGGGAGG بн+ץبץ CGCCTCCTCAAGGACGAGTGACTGTTATCACGGTATACCGTTTGCCACTCCCGTCCGAGACTTGGACTCGAAACCCCTCCCCTCC

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TCTCTGTATTCCACCCAGGGAAGGGGCAGCCTTTGGGTGGGAGGCTGGCACTGGTGGCTCACCCCAGACTGGCCTGCAGTGTCTG
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AGAACTGGATTACTTAGAAGTGGAGGCTGAGGCCAAGATGGAGAACTTGCGAGCTGCAGTGCCAGGTCAACCCCTAGCTCTGGTG بץНبץبץب+ TCTTGACCTAATGAATCTTCACCTCCGACTCCGGTTCTACCTCTTGAACGCTCGACGTCACGGTCCAGTTGGGGATCGAGACCAC


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Donor Template WT -> SNV
GGTGTGTGGCCCTCACAGACTGCCCGTGGTGGCCCCAAGGACACCCAGCCCAGCTATC
GCAGGTGCTGGAGCAGGGCCTGATATAGGTGTGTGGCCCTCACAGACTGCCCGTGGTGGCCCCAAGGACACCCAGCCCAGCTATC
 CGTCCACGACCTCGTCCCGGACTATATCCACACACCGGGAGTGTCTGACGGGCACCACCGGGGTTCCTGTGGGTCGGGTCGATAG POLG POLG-201


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ACCATGGCAATCGOR Template WT -> SNV

ACCATGGCAATGGACCTTACAACGACGTGGACATCCCTGGCTGCTGGTTTTTCAAGCTGCCTCACAAGGTGTGTCCTGGGTCATG
 TGGTACCGTTACCTGGAATGTTGCTGCACCTGTAGGGACCGACGACCAAAAAGTTCGACGGAGTGTTCCACACAGGACCCAGTAC


GCCTGTCCTGTGGTGTTTCCTCATTCTGCTCAAGGCCCACAGCAGGCCTTCAGAGTGACACACCTGAGACTTTCCTTTTTGTGGG
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GGACAGGACACCACAAAGGA
Sanger Sequencing

AATGACTAGTAGTGGGACAGAGTGTGATTTCAGGCACATACTGTCATCTCTCAGCTTTTGTTTTTCTAATGAAAGTCGGGTGGCA

TTACTGATCATCACCCTGTCTCACACTAAAGTCCGTGTATGACAGTAGAGAGTCGAAAACAAAAAGATTACTTTCAGCCCACCGT


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 TCCCCGTACCACCACCTTAATTTACTGTACCCCGTGCAGCATACAAACCATGCTGTAGACCATGCACTATCCAAAAAGGCTAAAC


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GCGCGTGCGTGATACAATCAGGGATTTGCCTCAGACTGCTGAGGTTCTGGGCTCAGTGTTGGGAGGAGTGCAGGTACTCACGTTG
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CCTACCATTATCGACATTACACCCT PCR Reverse

CCCCTTTGCCAAGGACTTCCTGCCCAAGATGGAGGATGGCACCCTGCAGGCTGGCCCAGGAGGTGCCAGTGGGCCCCGTGCTCTG
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CAGTCCTGGAGGTGGGCAGGTGAGGGCCATGGTGGGATGTGGGATAGATTCTGCTTCCCATGGCTGTGCTGAGCCTCACGTTGTC
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GTAGGGATAGGAGAACTGAGGTGAGGTGGTAGGTCCTAAGGCCAAAGCACCCTGCTAAGACCCATTTCCTTCCCCTGCACCCCAC بН-H CATCCCTATCCTCTTGACTCCACTCCACCATCCAGGATTCCGGTTTCGTGGGACGATTCTGGGTAAAGGAAGGGGACGTGGGGTG


CAGGCACCCCGACTATGATGAGGAAGGCCTCTATGGGGCCATCCTGCCCCAAGTGGTGACTGCCGGCACCATCACTCGCCGGGCT H-H H GTCCGTGGGGCTGATACTACTCCTTCCGGAGATACCCCGGTAGGACGGGGTTCACCACTGACGGCCGTGGTAGTGAGCGGCCCGA

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CACACCAATTGCTGATAGGTGGCCATCTCTGGAACTGTGGAATTTCAGCAGTGCTGTCTGGCATTCTCTAAAGCCATCCCCTCAG
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CAGCCTTTGGGTGGATGACACTGCAGGGCAGGAAGAGCAGGGGCACTGATCTACACAGTAAGACAGCCACTACTGTGGGCATCAG
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CACCGGCTCACACAGCAGGAGGCAGCTGAGAAGGCCCAGCAGATGTACGCTGCCACCAAGGGCCTCCGCTGGTGAGGGTCCCTCT بнبНبн GTGGCCGAGTGTGTCGTCCTCCGTCGACTCTTCCGGGTCGTCTACATGCGACGGTGGTTCCCGGAGGCGACCACTCCCAGGGAGA
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| :---: |
| THGAGAGTGGATCGGGGAGAGGAAGGTGTGGACACATTGGACCAAGGTCCTACTAAACAGGATAACACTGTAAACCAACGAAATA |
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ACCTCCCAGTGGACAGGACTGAGGGTGGCTGGATTTCCCTGCAGGATCTGCGCAAGGTCCAGAGAGAAACTGCAAGGAAGTAAGA ب+ $+\boldsymbol{+}$ TGGAGGGTCACCTGTCCTGACTCCCACCGACCTAAAGGGACGTCCTAGACGCGTTCCAGGTCTCTCTTTGACGTTCCTTCATTCT


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## AGCCGAGCCCTGGAGCCCTCGGCTGTCCAGGAAGAGGTATCTTGCTACCTTTGGAGCATGGGCAGAGGGGCCCCAGGGAGGGCAG

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AAAACTGAAAAGTAGGGGGATCTTCACTTACAAAGTCGAATAAATAAATAAAGATTAAAAATTTTTCAACATCTTCAGGATTTTT

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GACTAGCCTCAATTCGTAAAAAAAGAGTTATTGGGTTTGAAAATGTGAAATACCAAGACTGATCATTGAGGGAAGCAGTGAGGTT بـبCTGATCGGAGTTAAGCATTTTTTTCTCAATAACCCAAACTTTTACACTTTATGGTTCTGACTAGTAACTCCCTTCGTCACTCCAA


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CTTGCACTCTATAGGTGACAGAAAAACAAACTGTGCTTTAAATACCAAACAAGTAAATCAGAAAGCTTATTTTCTATTTAAAATA +Н-HبH GAACGTGAGATATCCACTGTCTTTTTGTTTGACACGAAATTTATGGTTTGTTCATTTAGTCTTTCGAATAAAAGATAAATTTTAT POLG

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СтСтCCTTTTGACCTTAGCATTAAGCTCCTTTGCTCACTTCTGAGCTCTCAGTTACAGTTCTTGAGGTGGCATCCTAACCAATTT
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CCATGCTCTGATGCAGTGCCCTAGAAGGGGAAAGAATTAATGCTCTAACGTGATAAACCTGCTCCAAGGCAGTGGAAATAAAAAG

 TTCCTTTTTTTTCTGAGATAGAAGAGTAGTTTCGGAAACAATCAGTACGAAAGGGGTGAAAGGGTGAGTGTCCTCCATTAATACA

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| :---: |
| TGTTGAAGAAGAGGAAGGAGTCCTCCAAAGTGTAGAGTAATAAGACAGTGGCACCTCCTTTTAGGACTTTTGCTATACTGAGGAT |
| ACAACTTCTTCTCCTTCCTCAGGAGGTTTCACATCTCATTATTCTGTCACCGTGGAGGAAAATCCTGAAAACGATATGACTCCTA |
| $H$ | ACTTATGGGACAAAATGACAAGAGAAGAAGCCATTGACATCAAAATGCTACATGACTCTCCTGAGTGCAGCTGCCAAGTGGGTAA

$+H$
POLG $>$

AGAGAGTGGATGGGGCCAGGTACATTAGAGCCTGGAGTGTGATCATCTGAAGAATGCATGTTCAAAATTCTGAAATCCAGGGCAC
 TCTCTCACCTACCCCGGTCCATGTAATCTCGGACCTCACACTAGTAGACTTCTTACGTACAAGTTTTAAGACTTTAGGTCCCGTG

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| :---: |
| GAGGAACAAACCACAAGGGAAAAGGACAAGACATTGGCATGAATTGAAGGAAGAAGCCTTAACCCAAACTAATGAGTGGCCTTTG |
| CTCCTTGTTTGGTGTTCCCTTTTCCTGTTCTGTAACCGTACTTAACTTCCTTCTTCGGAATTGGGTTTGATTACTCACCGGAAAC |
| $+H$ |

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$\square$ POLG $>$

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POLG
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TGGGTGACAGAATGAGACACAGTCTCAAAAAAAAAAAGGGGGGGGGGGGGAGAGGGGGGATGGCAAACACACAAATGGCCCAGAG + $+\boldsymbol{+} \boldsymbol{+}$ ACCCACTGTCTTACTCTGTGTCAGAGTTTTTTTTTTTCCCCCCCCCCCCCTCTCCCCCCTACCGTTTGTGTGTTTACCGGGTCTC

TCTCTTTTCCTCCATTGTAGTTGTGCTGCTTCAGTTTGCCAGATCTGTCAAGAAGGTAAATGGGAATGGTAGCACATAATAAGAG
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$\square$ POLG

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| :--- |
| TTTAACATTATCTTGGAGATAATTCCAGAGAGCTGAACATTTAGATGTTTATTTTTTTCATATCTCTCCAGTATAAATATATTAT |
| AAATTGTAATAGAACCTCTATTAAGGTCTCTCGACTTGTAAATCTACAAATAAAAAAAGTATAGAGAGGTCATATTTATATAATA |
| $H+H$ | AACCTATTTAGCCAGTCCTCTATtAATGGATATCTTTGTTTCTAATATTTCACTTTTATAAATAATGTTGTGAGGACCATCCTGT HبHبН TTGGATAAATCGGTCAGGAGATAATTACCTATAGAAACAAAGATTATAAAGTGAAAATATTTATTACAACACTCCTGGTAGGACA

POLG

ACACAGGCCTGTATATCCTAAAAGTGAAACTGCTACATCAGAATGGGATTCAGTAGTCATTGCTAAGCCTCTGGAGAGGCTGTAC
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> POLG

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> POLG

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POLG

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$\qquad$

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POLG $>$

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> POLG

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#### Abstract

POLG

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> POLG

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$\square$ POLG $>$

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| TGTCCGACT | $5^{\prime}$ |  |
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| POLG |  |
| :--- | :--- |
| /note $\quad=$ | $\quad$ gene ENSG00000140521 |
|  | Protein coding |


/note
$=$ primary transcript ENST00000637711 protein_coding_CDS_not_defined

| POLG-225 | $1053 \ldots$ | 4654 | $3602 \mathrm{bp} \quad \square$ | $\rightarrow$ | CDS |
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| ( 2 segments $=783 \mathrm{bp}$ |  |  |  |  |  |

/note $\quad=$ coding sequence ENSP00000497242
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| POLG-225 | 1053 | .. | 4654 | 3602 bp | $\square$ | $\rightarrow$ | prim_transcript |  |
| :--- | ---: | ---: | ---: | ---: | ---: | ---: | ---: | ---: |
| /note | $=$ | primary transcript ENST000000650303 |  |  |  |  |  |  |
| POLG-201 |  | 1108 | .. | 18,111 | $17,004 \mathrm{bp}$ | $\square$ | $\rightarrow$ | CDS |

[^0]/note $\quad=$ coding sequence ENSP00000268124
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Feature
Location
Size
$\square$
Type
POLG-202
( 22 segments $=3720 \mathrm{bp}$
/note $\quad=$ coding sequence ENSP00000399851
/translation $\quad=$ MSRLLWRKVAGATVGPGPVPAPGRWVSSSVPASDPSDGQRRRQQQQQQQQQQQQQPQQPQVLSSEGGQLRHNPLDIQMLSRGLHE Q IFGQ GGEMPGEAAVRRSVEHLQKHGLWGQPAVPLPDVELRLPPLYGDNLDQHFRLLAQKQSLPYLEAANLLLQAQLPPKPPAWAWAEG WTRYGPEGEAVPVAIPEERALVFDVEVCLAEGTCPTLAVAISPSAW,,YSWCSQRLVEERYSWTSQLSPADLIPLEVPTGASSPTQRDWQE QLVVGHNVSFDRAHIREQYLIQ,,GSRMRFLDTMSMHMAISGLSSFQRSLWIAAKQGKHKVQPPTKQGQKSQRKARRGPA ,ISSWDWLDI SSVNSLAEVHRLYVGGPPLEKEPRELFVKGTMKDIRENFQ, ,DLMQYCAQDVWATHEVFQQQLPLFLER,,CPHPVTLAGMLEMGVSYLPV NQNWERYLAEAQ GTYEELQREMKKSLMDLANDACQLLSGER,,YKEDPWLWDLEWDLQEFKQKKAKKVKKEPATASKLPIEGAGAPGDPM DQE ,,DLGPCSEEEEFQQDVMARACLQKLKGTTELLPKRPQHLPGHPG,,WYRKLCPRLDDPA WTPGPSLLSLQMRVTPKLMALTWDGFPLH YSERHGWGYLVPGRRDNLAKLPTGTTLESA GVVCPYR,,AIESLYRKHCLEQGKQQLMPQEAGLAEEFLLTDNSAIWQT,,VEELDYLEVEAE AKMENLRAAVPGQPLAL,,TARGGPKDTQPSYHHGNGPYNDVDIPGCWFFKLPHK,,DGNSCNVGSPFAKDFLPKMEDGTLQAGPGGASG PRALEINKMISFWRNAHKRIS,,SQMVVWLPRSALPRAVIR,,HPDYDEEGLYGAILPQVVTAGTITRRAVEPTWLTASNAR,,PDRVGSELKA MVQAPPGYTLVGADVDSQELWIAAVLGDAHFAGMH,,GCTAFGWMTLQGRKSRGTDLHSKTATTVGISREHAKIFNYGRIYGAGQPFAER LLMQFNHRLTQQEAAEKAQQMYAATKGLRW,,YRLSDEGEWLVRELNLPVDRTEGGWISLQDLRKVQRETARK,,SQ WKKWEVVAERA WK



* 22 segments $=3720 \mathrm{bp}$
/note $\quad=$ coding sequence ENSP00000516154
/translation $\quad=$ MSRLLWRKVAGATVGPGPVPAPGRWVSSSVPASDPSDGQRRRQQQQQQQQQQQQQPQQPQVLSSEGGQLRHNPLDIQMLSRGLHE QIFGQ GGEMPGEAAVRRSVEHLQKHGLWGQPAVPLPDVELRLPPLYGDNLDQHFRLLAQKQSLPYLEAANLLLQAQLPPKPPAWAWAEG WTRYGPEGEAVPVAIPEERALVFDVEVCLAEGTCPTLAVAISPSAW,,YSWCSQRLVEERYSWTSQLSPADLIPLEVPTGASSPTQRDWQE QLVVGHNVSFDRAHIREQYLIQ,,GSRMRFLDTMSMHMAISGLSSFQRSLWIAAKQGKHKVQPPTKQGQKSQRKARRGPA ,ISSWDWLDI SSVNSLAEVHRLYVGGPPLEKEPRELFVKGTMKDIRENFQ,,DLMQYCAQDVWATHEVFQQQLPLFLER,,CPHPVTLAGMLEMGVSYLPV NQNWERYLAEAQ GTYEELQREMKKSLMDLANDACQLLSGER,,YKEDPWLWDLEWDLQEFKQKKAKKVKKEPATASKLPIEGAGAPGDPM DQE,,DLGPCSEEEEFQQDVMARACLQKLKGTTELLPKRPQHLPGHPG,,WYRKLCPRLDDPAWTPGPSLLSLQMRVTPKLMALTWDGFPLH YSERHGWGYLVPGRRDNLAKLPTGTTLESA GVVCPYR,,AIESLYRKHCLEQGKQQLMPQEA GLAEEFLLTDNSAIWQT,,VEELDYLEVEAE AKMENLRAAVPGQPLAL,,TARGGPKDTQPSYHHGNGPYNDVDIPGCWFFKLPHK,,DGNSCNVGSPFAKDFLPKMEDGTLQAGPGGASG PRALEINKMISFWRNAHKRIS,,SQMVVWLPRSALPRAVIR,,HPDYDEEGLYGAILPQVVTAGTITRRAVEPTWLTASNAR,,PDRVGSELKA MVQAPPGYTLVGADVDSQELWIAAVLGDAHFAGMH,,GCTAFGWMTLQGRKSRGTDLHSKTATTVGISREHAKIFNYGRIYGAGQPFAER LLMQFNHRLTQQEAAEKAQQMYAATKGLRW,,YRLSDEGEWLVRELNLPVDRTEGGWISLQDLRKVQRETARK,,SQ WKKWEVVAERA WK G彑 VREEDRYRAALALQITNLLTR,,CMFAYKLGLNDLPQSVAFFSAVDIDRCLRKEVTMDCKTPSNPTGMERRYGIPQ,,GEALDIYQIIELTKGS LEKRSQPGP*


## POLG-226

$\begin{aligned} \text { /note }= & \text { primary transcript ENST000000666746 } \\ & \text { Nonsense mediated decay }\end{aligned}$
$\begin{aligned} \text { /note }= & \text { primary transcript ENST000000666746 } \\ & \text { Nonsense mediated decay }\end{aligned}$
POLG-208 $\begin{array}{llllllll}1507 & . . & 18,534 & 17,028 \mathrm{bp} \quad \square \quad \rightarrow \quad \text { prim_transcript }\end{array}$

/note $\quad=$ coding sequence ENSP00000489844
/translation $=$ VCG*QPSRANTRSSPPQSKARSPRGKPEEAQR,,SHPGTGWTSAVSTVWQRCTDFM*GGLP*RRSLENCL*RAP*RTFVRTSR,,T*CSTV PRTCGPPMRFSSSSYRSSWR,,GVPTQ*LWPACWRWVSPTCLSTRTGSVTWQRHRALMRSSSGR*RSR*WIWPMMPASCSQER,,GTKKTP GSGTWSGTCKNLSRRKLRR*RRNQPQPASCPSRGLGPLVIPWIRK,,TSAPAVRRRSFNKMSWPAPACRS*RGPQSSCPSGPSTFLDTL,,D GTGSSAPG*TTLHGPRAPASSACRCGSHLNSWHLPGMASLCTTQSVMAGATWCLGGGTTWPSCRQVPPWSQLGWSAPT,,EPSSPCTGS TVSNRGSSS* CPRRPA WRRSSCSLTIVPYGKR,,*KNWIT*KWRLRPRWRTCELQCQVNP*L*,,LPVVAPRTPSPAITMAMDLTTTWTSLAA GFSSCLTR,,MVIAVMWEAPLPRTSCPRWRMAPCRLAQEVPVGPVLWKSTK*FLSGGTPINVS,,APRWWCGCPGQLCPVL*S,,GTPTMMR KA SMGPSCPKW*LPAPSLAGLWSPHGSPPAMPG,,LTE*AVS*KPWCRPHLATPLWVLMWTPKSCGLQLCLETPTLPACM,,AAQPLGG*H CRAGRA GALIYTVRQPLLWA SA VSMPKSSTTAASMVLGSPLLSAY*CSLTTGSHSRRQLRRPSRCTLPPRASA, ,GIGCRMRASGW*GS*T SQ WT GLRV A GFPCRICARSREKLQG,,SHSGRSGRWLLNGHGRGAQSQKCSISLRALLRLTYHVPRCWAAASAEPWSPRLSRKS,,L*PAV*

POLG-230 GDTGFPR,,VKRWIFTR*LNSPKAP WKNEASLDH
$12,622 \mathrm{bp} \quad \square \quad \rightarrow \quad$ prim_transcript
/note
= primary transcript ENST00000672923 Retained intron
POLG-210 6150 .. $7849 \quad 1700 \mathrm{bp} \quad \square \quad \rightarrow \quad$ prim_transcript
/note $\quad=\quad$ primary transcript ENST000000532363 Retained intron



Feature
Location
Size
= primary transcript ENST00000561894 Protein coding
FANCI-211
/note

FANCI-210
/note
= primary transcript ENST00000565522 protein_coding_CDS_not_defined
FANCI-227
/note
$=$ primary transcript ENST00000696722 Retained intron



[^0]:    * 22 segments $=3720 \mathrm{bp}$

