```
GTTTATGAGA AACAGTGTGT TTCAGAGAGG CTGTACCAGA ATTAACTCTG -204

CTCAGAGTTA GATTTGCTGG TCTTAAAGTA CTTTTCCTCT TTAAGATAAA -154

AGAAGTTCTT CTAAATCAGG AATGGATTGA AATCTAATGA ACCGAAACTT -104

TGGGTACTTC GGCCTTCAAG GGGCTCCTTT ATTGAGAATC AATGTCTTCT -54

CCTAGGTAAT TGATCACCCT AGACCCAGGG ACACCCAATT CATCGTAATC -4

ATCATGAATA ATCAAAAGT GGTAGCTGTG CTACTGCAAG AGTGCAAGCA 47
  MetAsnA  snGlnLysVa lValAlaVal LeuLeuGlnG luCysLysGl 16


AGTGCTGGAT CAGCTCTTGT TGGAAGCGCC AGATGTGTCG GAAGAGGACA 97
nValLeuAsp GlnLeuLeuL euGluAlaPr oAspValSer GluGluAspL 33


AGAGCGAGGA CCAGCGCTGC AGAGCTTTAC TCCCCAGCGA GTTAAGGACC 147
ysSerGluAs pGlnArgCys ArgAlaLeuL euProSerGl uLeuArgThr 49


CTGATCCAGG AGGCAAAGGA AATGAAGTGG CCCTTCGTGC CTGAAAAGTG 197
LeuIleGlnG luAlaLysGl uMetLysTrp ProPheValP roGluLysTr 66


GCAGTACAAA CAAGCCGTGG GCCCAGAGGA CAAAACAAAC CTGAAGGATG 247
pGlnTyrLys GlnAlaValG lyProGluAs pLysThrAsn LeuLysAspV 83


TGATTGGCGC CGGGTTGCAG CAGTTACTGG CGTCCCTGAG GGCCTCCATC 297
alIleGlyAl aGlyLeuGln GlnLeuLeuA laSerLeuAr gAlaSerIle 99
```

```
CTCGCTCGGG ACTGTGCGGC TGCGGCGGCT ATTGTGTTCT TGGTGGACCG 347
LeuAlaArgA spCysAlaAl aAlaAlaAla IleValPheL euValAspAr 116

GTTCCTGTAT GGGCTCGACG TCTCTGGAAA ACTTCTGCAG GTCGCCAAAG 397
gPheLeuTyr GlyLeuAspV alSerGlyLy sLeuLeuGln ValAlaLysG 133

GTCTCCACAA GTTGCAGCCA GCCACGCCAA TTGCCCCGCA GGTGGTTATT 447
lyLeuHisLy sLeuGlnPro AlaThrProI leAlaProGl nValValIle 149

CGCCAAGCCC GAATCTCCGT GAACTCAGGA AAACTTTTAA AAGCAGAGTA 497
ArgGlnAlaA rgIleSerVa lAsnSerGly LysLeuLeuL ysAlaGluTy 166

TATTCTGAGC AGTCTAATAA GCAACAATGG AGCAACGGGT ACCTGGCTGT 547
rIleLeuSer SerLeuIleS erAsnAsnGl yAlaThrGly ThrTrpLeuT 183

ACAGAAATGA AAGTGACAAG GTCCTGGTGC AGTCGGTCTG TATACAGATC 597
yrArgAsnGl uSerAspLys ValLeuValG lnSerValCy sIleGlnIle 199

AGAGGGCAGA TTCTGCAAAA GCTGGGGATG TGGTACGAAG CAGCAGAGTT 647
ArgGlyGlnI leLeuGlnLy sLeuGlyMet TrpTyrGluA laAlaGluLe 216

AATATGGGCC TCCATTGTAG GATATTTGGC ACTTCCTCAG CCGGATAAAA 697
uIleTrpAla SerIleValG lyTyrLeuAl aLeuProGln ProAspLysL 233

AGGGCCTCTC CACGTCGCTA GGTATACTGG CAGACATCTT TGTTTCCATG 747    T237M
ysGlyLeuSe rThrSerLeu GlyIleLeuA laAspIlePh eValSerMet 249
```

AGCAAGAACG ATT<u>A</u>TGAAAA GTTTAAAAAC AATCCACAAA TTAATTTGAG 797   Y254C

SerLysAsnA spTyrGluLy sPheLysAsn AsnProGlnI leAsnLeuSe 266

CCTGCTGAAG GAGTTTGACC ACCATTTGCT GT<u>C</u>CGCTGCA GAAGCCTGCA 847   S277F

rLeuLeuLys GluPheAspH isHisLeuLe uSerAlaAla GluAlaCysL 283

AGCTGGCAGC TGCCTTCAGT GCCTATACGC CGCTCTTCGT GCTCACAGCT 897

ysLeuAlaAl aAlaPheSer AlaTyrThrP roLeuPheVa lLeuThrAla 299

GTGAATATCC GTGGCACGTG TTTATTGTCC TACAGTAGTT CAAATGACTG 947

ValAsnIleA rgGlyThrCy sLeuLeuSer TyrSerSerS erAsnAspCy 316

TCCTCCAGAA TTGAAAAACT TACATCTGTG TGAAGCCAAA GAGGCCTTTG 997

sProProGlu LeuLysAsnL euHisLeuCy sGluAlaLys GluAlaPheG 333

AGATTGGCCT CCTCACCAAG AGAGAT<u>G</u>ATG AGCCTGTTAC TGGAAAACAG 1047   c.1024G>C

luIleGlyLe uLeuThrLys ArgAspAspG luProValTh rGlyLysGln 349

GAGCTTCACA GCTTTGTCAA AGCTGCTTTC GGTCTCACCA CAGTGCACAG 1097

GluLeuHisS erPheValLy sAlaAlaPhe GlyLeuThrT hrValHisAr 366

AAGGCTCCAT GGGGAGACAG GGACGGTCCA TGCAGCAAGT CAGCTCTGTA 1147

gArgLeuHis GlyGluThrG lyThrValHi sAlaAlaSer GlnLeuCysL 383

AGGAAGCAAT GGGGAAGCTG TACAATTTCA GCACTTCCTC CAGAAGTCAG 1197

ysGluAlaMe tGlyLysLeu TyrAsnPheS erThrSerSe rArgSerGln 399

GACAGAGAAG CTCTGTCTCA AGAAGTTATG TCTGTGATTG CCCAGGTGAA 1247

```
AspArgGluA laLeuSerGl nGluValMet SerValIleA laGlnValLy 416


GGAACATTTA CAAGTTCAAA GCTTCTCAAA TGTAGATGAC AGATCTTATG 1297
sGluHisLeu GlnValGlnS erPheSerAs nValAspAsp ArgSerTyrV 433


TTCCCGAGAG TTTCGAGTGC AGGTTGGATA AACTTATCTT GCATGGGCAA 1347
alProGluSe rPheGluCys ArgLeuAspL ysLeuIleLe uHisGlyGln 449


GGGGATTTCC AAAAAATCCT TGACACCTAT TCACAGCACC ATACTTCGGT 1397
GlyAspPheG lnLysIleLe uAspThrTyr SerGlnHisH isThrSerVa 466


GTGTGAAGTA TTTGAAAGTG ATTGTGGAAA CAACAAAAAT GAACAGAAAG 1447
lCysGluVal PheGluSerA spCysGlyAs nAsnLysAsn GluGlnLysA 483


ATGCAAAAAC AGGAGTCTGC ATCACTGCTC TAAAAACAGA AATAAAAAAC 1497
spAlaLysTh rGlyValCys IleThrAlaL euLysThrGl uIleLysAsn 499


ATAGATACTG TGAGTACTAC TCAAGAAAAG CCACATTGTC AAAGAGACAC 1547
IleAspThrV alSerThrTh rGlnGluLys ProHisCysG lnArgAspTh 516


AGGAATATCT TCCTCCCTAA TGGGTAAGAA TGTTCAGAGG GAACTCAGAA 1597
rGlyIleSer SerSerLeuM etGlyLysAs nValGlnArg GluLeuArgA 533


GGGGAGGAAG GAGAAACTGG ACCCATTCTG ATGCATTTCG AGTCTCCTTG 1647
rgGlyGlyAr gArgAsnTrp ThrHisSerA spAlaPheAr gValSerLeu 549


GATCAAGATG TGGAGACTGA GACTGAGCCA TCGGACTACA GCAATGGTGA 1697
AspGlnAspV alGluThrGl uThrGluPro SerAspTyrS erAsnGlyGl 566
```

GGGAGCTGTT TTCAACAAGT CTCTGAGTGG CAGCCAGACT TCCAGTGCTT 1747

uGlyAlaVal PheAsnLysS erLeuSerGl ySerGlnThr SerSerAlaT 583

GGAGCAACTT ATCAGGGTTT AGTTCCTCTG CAAGCTGGGA GGAAGTGAAT 1797

rpSerAsnLe uSerGlyPhe SerSerSerA laSerTrpGl uGluValAsn 599

TATCACGTTG ACGACAGGTC AGCCAGAAAA GAGCCTGGCA AAGAACATCT 1847

TyrHisValA spAspArgSe rAlaArgLys GluProGlyL ysGluHisLe 616

GGTGGACACT CAGTGTTCCA CTGCCTTGTC TGAGGAGCTA GAGAATGACA 1897

uValAspThr GlnCysSerT hrAlaLeuSe rGluGluLeu GluAsnAspA 633

GGGAAGGCAG AGCTATGCAT TCATTGCATT CACAGCTTCA TGATCTCTCT 1947

rgGluGlyAr gAlaMetHis SerLeuHisS erGlnLeuHi sAspLeuSer 649

CTTCAGGAAC CCAACAATGA CAATTTGGAG CCTTCTCAAA ATCAGCCACA 1997

LeuGlnGluP roAsnAsnAs pAsnLeuGlu ProSerGlnA snGlnProGl 666

GCAACAGATG CCCTTGACAC CCTTCTCGCC TCATAATACC CCAGGCATTT 2047

nGlnGlnMet ProLeuThrP roPheSerPr oHisAsnThr ProGlyIleP 683

TCTTGGCCCC TGGTGCAGGG CTTCTAGAAG GAGCTCCAGA AGGTATCCAG 2097

heLeuAlaPr oGlyAlaGly LeuLeuGluG lyAlaProGl uGlyIleGln 699

GAAGTCAGAA ATATGGGACC CAGAAATACT TCTGCTCACT CCAGACCCTC 2147

GluValArgA snMetGlyPr oArgAsnThr SerAlaHisS erArgProSe 716

ATATCGTTCT GCTTCTTGGT CTTCTGATTC TGGTAGGCCC AAGAATATGG 2197
rTyrArgSer AlaSerTrpS erSerAspSe rGlyArgPro LysAsnMetG 733

GCACACATCC TTCAGTCCAA AAAGAAGAAG CCTTTGAAAT AATTGTTGAG 2247
lyThrHisPr oSerValGln LysGluGluA laPheGluIl eIleValGlu 749

TTTCCAGAAA CCAACTGCGA TGTCAAAGAC AGGCAGGGGA AAGAGCAGGG 2297
PheProGluT hrAsnCysAs pValLysAsp ArgGlnGlyL ysGluGlnGl 766

AGAAGAAATT AGTGAAAGAG GCGCAGGCCC TACATTTAAA GCTAGTCCCT 2347
yGluGluIle SerGluArgG lyAlaGlyPr oThrPheLys AlaSerProS 783

CCTGGGTTGA CCCAGAAGGA GAAACAGCAG AAAGCACTGA AGATGCACCC 2397
erTrpValAs pProGluGly GluThrAlaG luSerThrGl uAspAlaPro 799

TTAGACTTTC ACAGGGTCCT GCACAATTCT CTGGGAAACA TTTCCATGCT 2447
LeuAspPheH isArgValLe uHisAsnSer LeuGlyAsnI leSerMetLe 816

GCCATGTAGC TCCTTCACCC CTAATTGGCC TGTTCAAAAT CCTGACTCCA 2497
uProCysSer SerPheThrP roAsnTrpPr oValGlnAsn ProAspSerA 833

GAAAAAGTGG TGGCCCAGTC GCAGAGCAGG GCATCGACCC TGATGCCTCC 2547
rgLysSerGl yGlyProVal AlaGluGlnG lyIleAspPr oAspAlaSer 849

ACAGTGGATG AGGAGGGGCA ACTGCTCGAC AGCATGGATG TTCCCTGCAC 2597
ThrValAspG luGluGlyGl nLeuLeuAsp SerMetAspV alProCysTh 866

AAATGGGCAC GGCTCTCATA GACTGTGCAT TCTGAGACAG CCGCCTGGTC 2647

rAsnGlyHis GlySerHisA rgLeuCysIl eLeuArgGln ProProGlyG 883

AGAGGGCGGA GACCCCCAAT TCCTCTGTAA GCGGTAACAT CCTCTTCCCT 2697
lnArgAlaGl uThrProAsn SerSerValS erGlyAsnIl eLeuPhePro 899

GTCCTCAGCG AGGACTGCAC TACCACAGAG GAAGGAAATC AGCCTGGAAA 2747
ValLeuSerG luAspCysTh rThrThrGlu GluGlyAsnG lnProGlyAs 916

CATGCTAAAC TGCAGCCAGA ACTCCAGCTC ATCCTCAGTG TGGTGGCTGA 2797   c.2770T>C
nMetLeuAsn CysSerGlnA snSerSerSe rSerSerVal TrpTrpLeuL 933

AATCACCTGC ATTTTCCAGT GGTTCTTCTG AGGGGGACAG CCCTTGGTCC 2847
ysSerProAl aPheSerSer GlySerSerG luGlyAspSe rProTrpSer 949

TATCTGAATT CCAGTGGGAG TTCTTGGGTT TCATTGCCGG GAAAGATGAG 2897
TyrLeuAsnS erSerGlySe rSerTrpVal SerLeuProG lyLysMetAr 966

GAAAGAGATC CTTGAGGCTC GCACCTTGCA ACCTGATGAC TTTGAAAAGC 2947
gLysGluIle LeuGluAlaA rgThrLeuGl nProAspAsp PheGluLysL 983

TGTTGGCAGG AGTGAGGCAT GATTGGCTGT TTCAGAGACT AGAGAATACG 2997
euLeuAlaGl yValArgHis AspTrpLeuP heGlnArgLe uGluAsnThr 999

GGGGTTTTTA AGCCCAGTCA ACTCCACCGA GCACATAGTG CTCTTTTGTT 3047
GlyValPheL ysProSerGl nLeuHisArg AlaHisSerA laLeuLeuLe 1016

AAAATATTCA AAAAAATCTG AACTGTGGAC GGCCCAGGAA ACTATTGTCT 3097
uLysTyrSer LysLysSerG luLeuTrpTh rAlaGlnGlu ThrIleValT 1033

```
ATTTGGGGGA CTACTTGACT GTGAAGAAAA AAGGCAGACA AAGAAATGCT 3147
yrLeuGlyAs pTyrLeuThr ValLysLysL ysGlyArgGl nArgAsnAla 1049


TTTTGGGTTC ATCATCTTCA TCAAGAAGAA ATTCTGGGGA GGTATGTTGG 3197
PheTrpValH isHisLeuHi sGlnGluGlu IleLeuGlyA rgTyrValGl 1066


GAAAGACTAT AAGGAGCAGA AGGGGCTCTG GCACCACTTC ACTGATGTGG 3247
yLysAspTyr LysGluGlnL ysGlyLeuTr pHisHisPhe ThrAspValG 1083


AGCGACAGAT GACCGCACAG CACTATGTGA CAGAATTTAA CAAGAGACTC 3297
luArgGlnMe tThrAlaGln HisTyrValT hrGluPheAs nLysArgLeu 1099


TATGAACAAA ACATTCCCAC CCAGATATTC TACATCCCAT CCACAATACT 3347
TyrGluGlnA snIleProTh rGlnIlePhe TyrIleProS erThrIleLe 1116


ACTGATTTTA GAGGACAAGA CAATAAAGGG ATGTATCAGT GTGGAGCCTT 3397
uLeuIleLeu GluAspLysT hrIleLysGl yCysIleSer ValGluProT 1133


ACATACTGGG AGAATTTGTA AAATTGTCAA ATAACACGAA AGTGGTGAAA 3447
yrIleLeuGl yGluPheVal LysLeuSerA snAsnThrLy sValValLys 1149


ACAGAATACA AAGCCACAGA ATATGGCTTG GCCTATGGCC ATTTTTCTTA 3497
ThrGluTyrL ysAlaThrGl uTyrGlyLeu AlaTyrGlyH isPheSerTy 1166


TGAGTTTTCT AATCATAGAG ATGTTGTGGT CGATTTACAA GGTTGGGTAA 3547
rGluPheSer AsnHisArgA spValValVa lAspLeuGln GlyTrpValT 1183
```

CCGGTAATGG AAAAGGACTC ATCTACCTCA CAGATCCCCA GATTCACTCC 3597
hrGlyAsnGl yLysGlyLeu IleTyrLeuT hrAspProGl nIleHisSer 1199

GTTGATCAGA AAGTTTTCAC TACCAATTTT GGAAAGAGAG GAATTTTTA 3647
ValAspGlnL ysValPheTh rThrAsnPhe GlyLysArgG lyIlePheTy 1216

CTTCTTTAAT AACCAGCATG TGGAATGTAA TGAAATCTGC CATCGTCTTT 3697
rPhePheAsn AsnGlnHisV alGluCysAs nGluIleCys HisArgLeuS 1233

CTTTGACTAG ACCTTCAATG GAGAAACCAT GCACATAGAA TACGGCACAG *12
erLeuThrAr gProSerMet GluLysProC ysThrStop

TCTGGTCCTT TGGGGCTTGG GCAGGGCCGT GACACAGGTT CTGGCCAATG *62
ATTTGCAAGA GGAATTGATC AGTATCACTT TAAGTCCTGC ATTTAATTGG *112
CAGCACAAGA TCCTGCAGAG CCTCTTTCCC TCTGCCACAG TTATCAAGAA *162
TGGGTCAGGA GACCGCTGCT TCTGGGCATA AGTCCTGCAA GGAAAGCAAC *212
ATGGAAAACA GCCCCAACTC ACCCATGAGG GATGAAAAGC ACTCTTGAGA *262
AAGGCATGTG TTGTTTAAGC CATTGAGATT TTAGAGCTTT TTGTCACTAT *312
CTGTCAAGAC TGATACTACT GGGGCTTTTC CTATTGATTT GGGAGTTCTT *362
TACATATTAA AAAAATGTGA GCCTTTGTGA TACGAATTCA ATTTGTTTTC *412
CTGTCTTTTG ACATTTGACT TTGCATAAAA GTTTATCTGT GCATAATTTT *462
ATATGTAGTT GAATTCATCA ATCTTTTATT TTGTATGGCT TTTTGGTTAT *512
GTATAATACT TAGATCCTCC TTATACTCTG AGTTCTTTC TTTTTAATTC *562
TCCTGTATTT CCTTCTAGTA TAATTAAATC TGTAAAAAGT AAGATGGAAG *612
AGTGGTACAG TTTTCTTTAT CCAGTCTGTC CTTGATGGGC ATTTAGGTAG *662
ACTGGATAAA GAAAATGTGG TACATATACA CCATGGAACA CTATGTGTAT *712
TAATCCACTC TCACACTGCT ATGAAGAGAT ACCTGAGACT GGGTAATTTA *762
GAAAGAAAAG AGGTTTAATT AACTCACAGT TCCACATGGC TGGGGAGACC *812

```
TCAGGAAACT TACAATCATG GCAGAAGGCA CCTCTTCATA GGGTAGCAGG *862
AGAGAGAATG AGTGCCAGCA GGGGAAATGC CAGATGCTTA TAAAGCCATC *912
AGATCTTGTG AGAATTCATT CACTCTCACG AGAACAGCAT GGGAAAAACT *962
GCCTCAATTA CCTCCTACCA GGTCCTTCCC ATGACACATG GGAATTATGG *1012
GACTACAATT CGAGATGAGA TTTGGGTGGG GACACAAAGC CAAACCATAT *1062
CACAATGTAA CCATAAAAAA GAATGAGATC ATGTCCTTTG CAGGGACATG *1112
GATAGAGCTG GAGGCCATTA TTTTTAGCAA ACTAATGCAA GAACAGAAAA *1162
CTAAATACCA CTTGTTCTCA CTTATAGGTG AGAGCTAAGT GATGAGAGTA *1212
GGTGGACACA TAGAGGGAAC AACACACACC AGGGCTTATC AGAGGGTGGA *1262
CAGTGGGAGG AGGGAGAGGA TCAGGAAAAA TAACTAATGG GTACTAGGCT *1312
GAATACCTGG GTGATGAAGT AATTCGCACA ACAAACCCCC ATGACACAAA *1362
CCTGCACATG TACCCCTGAA CTTAAAATAA AAGTAAAAAA AAAAAATCA
```