



*SH3BP2* (NM\_003023.4) - cDNA + Protein - 2025-04-02

CAGCCGGGTG ACCCAGGCCG AGGCCGGCAG AAGACAGCCT GATGCCTTGA -212  
AGACTTCCTC TTGCACTTTT GTTGGAGGGT GCTGGTTTGC TAAAAGCAGA -162  
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CAGCTCAGTT GCCCAGACTG GAGAGCAGTG GCCAATCATA GCTTACTGCC -62  
TCCTGGAAct CCTGGCTCAA TCGATCCTCC TGGATAAGCC TCCTCCGGGT -12  
ACTATAGCTT **CATGCGGGCT** GAAGAGATGC ATTGGCCTGT CCCTATGAAG 39  
MetAlaAla GluGluMetH isTrpProVa lProMetLys 13

GCCATTGGTG CCCAGAACCT GCTAACCATG CCTGGGGGCG TGGCCAAGGC 89  
AlaIleGlyA laGlnAsnLe uLeuThrMet ProGlyGlyV alAlaLysAl 30

TGGCTACCTG CACAAGAAGG GCGGTACCCA GCTGCAGCTG CTGAAATGGC 139  
aGlyTyrLeu HisLysLysG lyGlyThrGl nLeuGlnLeu LeuLysTrpP 47

CCCTGCGCTT TGTCATCATC CACAAACGCT GCGTCTACTA CTTCAAGAGT 189 **408delC**  
roLeuArgPh eValIleIle HisLysArgC ysValTyrTy rPheLysSer 63

AGCACCTCTG CCTCCCCGCA GGGCGCCTTC TCCCTGAGTG GCTATAACCG 239 **R80Q**  
SerThrSerA laSerProGl nGlyAlaPhe SerLeuSerG lyTyrAsnAr 80

GGTGATGCGG GCGGCTGAGG AGACCACGTC CAACAACGTT TTCCCCTTCA 289  
gValMetArg AlaAlaGluG luThrThrSe rAsnAsnVal PheProPheL 97

AGATCATCCA TATCAGCAAG AAGCACCGCA CGTGGTTCTT CTCGGCCTCC 339 T107M

ysIleIleHi sIleSerLys LysHisArgT hrTrpPhePh eSerAlaSer 113

TCCGAGGAGG AGCGCAAGAG CTGGATGGCC TTGCTGCGCA GGGAGATTGG 389

SerGluGluG luArgLysSe rTrpMetAla LeuLeuArgA rgGluIleGl 130

CCACTTCCAC GAAAAGAAAG ACCTGCCCTT GGACACCAGC GACTCCAGCT 439

yHisPheHis GluLysLysA spLeuProLe uAspThrSer AspSerSerS 147

CGGACACAGA CAGCTTCTAC GGCGCAGTTG AGCGGCCTGT GGATATCAGC 489 A155V

erAspThrAs pSerPheTyr GlyAlaValG luArgProVa lAspIleSer 163

CTTTCCCCGT ACCCCACGGA CAATGAAGAC TATGAGCACG ACGATGAGGA 539

LeuSerProT yrProThrAs pAsnGluAsp TyrGluHisA spAspGluAs 180

TGACTCCTAC CTGGAGCCTG ACTCCCCGGA GCCCGGAAGG CTTGAGGATG 589

pAspSerTyr LeuGluProA spSerProGl uProGlyArg LeuGluAspA 197

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LysProAlaP heSerAspMe tProArgAla HisSerPheT hrSerLysGl 230

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GCTGAGCCTT GCCCCAGGGT ACCTGCTACC CCCCGAAGGA TGAGCGATCC 839  
AlaGluProC ysProArgVa lProAlaThr ProArgArgM etSerAspPr 280

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oProLeuSer ThrMetProT hrAlaProGl yLeuArgLys ProProCysP 297

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heArgGluSe rAlaSerPro SerProGluP roTrpThrPr oGlyHisGly 313

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AlaCysSerT hrSerSerAl aAlaIleMet AlaThrAlaT hrSerArgAs 330

CTGTGACAAA CTCAAGTCCT TCCACCTGTC CCCCCGAGGA CCACCCACAT 1039  
nCysAspLys LeuLysSerP heHisLeuSe rProArgGly ProProThrS 347

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GluGluAspP roProArgGl uAlaAlaMet ProGlyLeuP heValProPr 380

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oValAlaPro ArgProProA laLeuLysLe uProValPro GluAlaMetA 397

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laArgProAl aValLeuPro ArgProGluL ysProGlnLe uProHisLeu 413

CAGCGATCAC CCCCCGATGG GCAGAGTTTC AGGAGCTTCT CCTTTGAAAA 1289 [c.1243C>T](#) [R415P](#) [R415Q](#) [P418T](#) [P418L](#) [P418H](#) [P418R](#) [P418Ha](#) [D419N](#) [D419Y](#) [D419G](#) [G420R\\_G>C](#)  
[G420R\\_G>A](#) [G420E](#)

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spTyrGluLy sValProLeu ProAsnSerV alPheValAs nThrThrGlu 463

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SerCysGluV alGluArgLe uPheLysAla ThrSerProA rgGlyGluPr 480

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alLeuValVa lTrpAspGlu ThrSerAsnL ysValArgAs nTyrArgIle 513

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PheGluLysA spSerLysPh eTyrLeuGlu GlyGluValL euPheValSe 530

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CAGTCCATGT GGCTGCCAGG CCAAGGCAGT CACAGGGGCC CTGACCCCAG \*53  
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GGACGAAGGG ACTCTGTTGC CCCACACTAA CTTGCCCTGT CCCAATCCCA \*253  
GAAACCCAGG ACCAAGCTGT GCCTGGGCTC CAAGGACAGG AACACTGGTC \*303  
CCCCATCAC ACTCACCCCT AAGTGGGCTG GGAGCCAGGC AGGGCCAGGG \*353  
CAGCTGGGTG GGGGCCGGGG CTGGCCCTGG GACCCCAGG AACGCTAAGA \*403  
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*SH3BP2* (NM\_003023.4) - cDNA + Protein - 2025-04-02

