



*SH3BP2* (NM\_003023.4) - cDNA + Protein - 2024-06-20

CAGCCGGGTG ACCCAGGCCG AGGCCGGCAG AAGACAGCCT GATGCCTTGA -212  
AGACTTCCTC TTGCACTTTT GTTGGAGGGT GCTGGTTTGC TAAAAGCAGA -162  
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CAGCTCAGTT GCCCAGACTG GAGAGCAGTG GCCAATCATA GCTTACTGCC -62  
TCCTGGAAC TCTGGCTCAA 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 GCTATAACC**G** 239 **R80Q**  
SerThrSerA laSerProGl nGlyAlaPhe SerLeuSerG lyTyrAsnAr 80

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

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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

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erAspThrAs pSerPheTyr GlyAlaValG luArgProVa lAspIleSer 163

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LeuSerProT yrProThrAs pAsnGluAsp TyrGluHisA spAspGluAs 180

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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

TCCGGGAGAG TGCCAGCCCC AGCCCGGAGC CCTGGACCCC TGGCCACGGG 939 [G313R](#)  
heArgGluSe rAlaSerPro SerProGluP roTrpThrPr oGlyHisGly 313

GCCTGCTCCA CTTCCAGTGC TGCCATCATG GCCACTGCCA CCTCCAGAAA 989  
AlaCysSerT hrSerSerAl aAlaIleMet AlaThrAlaT hrSerArgAs 330

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

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erGluProPr oProValPro AlaAsnLysP roLysPheLe uLysIleAla 363

GAAGAGGACC CCCCAAGGGA GGCAGCCATG CCCGGACTCT TTGTGCCCCC 1139  
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](#)

GlnArgSerP roProAspGl yGlnSerPhe ArgSerPheS erPheGluLy 430

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spTyrGluLy sValProLeu ProAsnSerV alPheValAs nThrThrGlu 463

TCCTGCGAAG TGGAAAGGTT GTTCAAGGCT ACAAGCCCCC GGGGAGAGCC 1439

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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rValGlySer MetValGluH isTyrHisTh rHisValLeu ProSerHisG 547

AGAGCCTGCT GCTGC~~C~~GGCAC CCCTACGGCT ACACTGGGCC TAGG~~T~~GATGG \*3 [R552W](#)

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GCCACACAGA CGGACATGGG CCCACATGGG AGGGTGAGCA GGAGCAAGGC \*103  
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GGACGAAGGG ACTCTGTTGC CCCACACTAA CTTGCCCTGT CCCAATCCCA \*253  
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CAGCTGGGTG GGGGCCGGGG CTGGCCCTGG GACCCCAGG AACGCTAAGA \*403  
CACAGGCTCC AGTAGGGGCT GTTGCCTCCA ATAAAGCAGC AGTGAGCTTT \*453  
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*SH3BP2* (NM\_003023.4) - cDNA + Protein - 2024-06-20

