



RBCK1 (NM_031229.4) - cDNA + Protein - 2024-12-21

ACTTTCACCTT TCTCTTCCGC CGAAGCCGCT CCCCTTGCGA AGAACTGGGG -410 [c.ex1_ex4del](#)
CCTCCCGGGA GGAGAGAGGG CTTTGCCTTG AAACCCGGGA CGCCAGGGGC -360
GCTCCCGCAA GTGGGGGTCC TCCGGGACTT GGAACGCCCC GGCTGGGTGG -310
TGTCCGGGCG TCCTTTCCCC GCTTCTTCCC ACCTCGGCTG GTCCCGTTTC -260
CTCCTGCGCC CAGTGC GGAC CTGTCTCGGC GCCCGCTGCC CTCTCACC GC -210
CCCACGCAGG ATCCCGGCCT GGTCAACGGG CAGTGTGATG CTTCCCGACT -160
GCCGCGGGGA CAGCGAGGCA CACACAGGGC TTGGGCGCG CCGGAGGCCA -110
CACGGCCTGG CTGAGTTGCT CCTGGTCTCC CGCCTCTCCC AGGCGACCCG -60
GAGGTAGCAT TTCCAGGAG GCACGGTCCC CCCAGGGGG ATGGGCACAG -10
CCACGCCAGA **TGGACGAGAA** **GACCAAGAAA** **GCAGAGGAAA** **TGGCCCTGAG** 41
M etAspGluLy sThrLysLys AlaGluGluM etAlaLeuSe 14

CCTCACCCGA **G**CAGTGGCGG GCGGGGATGA ACAGGTGGCA ATGAAGTGTG 91 [c.52G>C](#)
rLeuThrArg AlaValAlaG lyGlyAspGl uGlnValAla MetLysCysA 31

CCATCTGGCT GGCAGAGCAA CGGGTGCCCC **T**GAGTGTGCA ACTGAAGCCT 141 [L41fsX7](#)
laIleTrpLe uAlaGluGln ArgValProL euSerValGl nLeuLysPro 47

GAGGTCTCCC CAACGCAGGA CATCAGGCTG TGGGTGAGCG TGGAGGATGC 191
GluValSerP roThrGlnAs pIleArgLeu TrpValSerV alGluAspAl 64

TCAGATGCAC ACCGTCACCA TCTGGCTCAC AGTGCGCCCT GATATGACAG 241

aGlnMetHis ThrValThrI leTrpLeuTh rValArgPro AspMetThrV 81

TGGCGTCTCT CAAGGACATG GTTTTTCTGG ACTATGGCTT CCCACCAGTC 291

alAlaSerLe uLysAspMet ValPheLeuA spTyrGlyPh eProProVal 97

TTGCAGCAGT GGGTGATTGG GCAGCGGCTG GCACGAGACC AGGAGACCCT 341

LeuGlnGlnT rpValIleGl yGlnArgLeu AlaArgAspG lnGluThrLe 114

GCACTCCCAT GGGGTGCGGC AGAATGGGGA CAGTGCCTAC CTCTATCTGC 391 [N122H](#)

uHisSerHis GlyValArgG lnAsnGlyAs pSerAlaTyr LeuTyrLeuL 131

TGTCAGCCCG CAACACCTCC CTCAACCCTC AGGAGCTGCA GCGGGAGCGG 441

euSerAlaAr gAsnThrSer LeuAsnProG lnGluLeuGl nArgGluArg 147

CAGCTGCGGA TGCTGGAAGA TCTGGGCTTC AAGGACCTCA CGCTGCAGCC 491 [c.456+1G>C](#)

GlnLeuArgM etLeuGluAs pLeuGlyPhe LysAspLeuT hrLeuGlnPr 164

GCGGGGCCCT CTGGAGCCAG GCCCCCCAA GCCCCGGGTC CCCCAGGAAC 541 [c.494delG](#)

oArgGlyPro LeuGluProG lyProProLy sProGlyVal ProGlnGluP 181

CCGGACGGGG GCAGCCAGAT GCAGTGCCTG AGCCCCACC GGTGGGCTGG 591 [Q185X](#)

roGlyArgGl yGlnProAsp AlaValProG luProProPr oValGlyTrp 197

CAGTGCCCGG GGTGCACCTT CATCAACAAG CCCACGCGGC CTGGCTGTGA 641

GlnCysProG lyCysThrPh eIleAsnLys ProThrArgP roGlyCysGl 214

GATGTGCTGC CGGGCGCGCC CCGAGCCCTA CCAGGTCCCC GCCTCATACC 691 [Q231Sfs*45](#)

uMetCysCys ArgAlaArgP roGluAlaTy rGlnValPro AlaSerTyrG 231

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 lnProAspGl uGluGluArg AlaArgLeuA laGlyGluGl uGluAlaLeu 247

CGTCAGTACC AGCAGCGGAA GCAGCAGCAG CAGGAGGGGA ACTACCTGCA 791 Q222X
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GCACGTCCAG CTGGACCAGA GGAGCCTGGT GCTGAACACG GAGCCCGCCG 841 Q267* L273Pfs*27
 nHisValGln LeuAspGlnA rgSerLeuVa lLeuAsnThr GluProAlaG 281

AGTGCCCGT GTGCTACTCG GTGCTGGCGC CCGGCGAGGC CGTTGGTGCTG 891 V295L
 luCysProVa lCysTyrSer ValLeuAlaP roGlyGluAl aValValLeu 297

CGTGAGTGTC TGCACACCTT CTGCAGGGAG TGCCTGCAGG GCACCATCCG 941 c.896_899del C305F
 ArgGluCysL euHisThrPh eCysArgGlu CysLeuGlnG lyThrIleAr 314

CAACAGCCAG GAGGCGGAGG TCTCCTGCCC CTTCATTGAC AACACCTACT 991 A319V
 gAsnSerGln GluAlaGluV alSerCysPr oPheIleAsp AsnThrTyrS 331

CGTGCTCGGG CAAGCTGCTG GAGAGGGAGA TCAAGGCGCT CCTGACCCCT 1041
 erCysSerGl yLysLeuLeu GluArgGluI leLysAlaLe uLeuThrPro 347

GAGGATTACC AGCGATTTCT AGACCTGGGC ATCTCCATTG CTGAAAACCG 1091 c.1054C>T
 GluAspTyrG lnArgPheLe uAspLeuGly ileSerIleA laGluAsnAr 364

CAGTGCC TTC AGCTACCATT GCAAGACCCC AGATTGCAAG GGATGGTGCT 1141
 gSerAlaPhe SerTyrHisC ysLysThrPr oAspCysLys GlyTrpCysP 381

TCTTTGAGGA TGATGTCA~~A~~T GAGTTCACCT GCCCTGTGTG TTTCCACGTC 1191 [c.1160A>G](#)

hePheGluAs pAspValAsn GluPheThrC ysProValCy sPheHisVal 397

AACTGCCTGC TCTGCAAGGC CATCCATGAG CAGATGAACT GCAAGGAGTA 1241

AsnCysLeuL euCysLysAl aIleHisGlu GlnMetAsnC ysLysGluTy 414

TCAGGAGGAC CTGGCCCTGC GGGCTCAGAA CGATGTGGCT GCCCGGCAGA 1291

rGlnGluAsp LeuAlaLeuA rgAlaGlnAs nAspValAla AlaArgGlnT 431

CGACAGAGAT GCTGAAGGTG ATGCTGCAGC AGGGCGAGGC CATGCGCTGC 1341

hrThrGluMe tLeuLysVal MetLeuGlnG lnGlyGluAl aMetArgCys 447

CCCCAGTGCC AGATCGTGGT ACAGAAGAAG GACGGCTGCG ACTGGATCCG 1391

ProGlnCysG lnIleValVa lGlnLysLys AspGlyCysA spTrpIleAr 464

CTGCACCGTC TGCCACA~~C~~CG AGATCTGCTG GGTCAACAAG GGCCACGCT 1441 [T470S](#) [E471K](#)

gCysThrVal CysHisThrG luIleCysTr pValThrLys GlyProArgT 481

GGGGCCCTGG GGGCCCAGGA GAC~~A~~CCAGCG GGGGCTGCCG CTGCAGGGTA 1491 [T489Pfs*9](#)

rpGlyProGl yGlyProGly AspThrSerG lyGlyCysAr gCysArgVal 497

AATGGGATTC CTTGCCACCC AAGCTGTCTAG ~~A~~ACTGCCACT GAGCTAAAGA *8 [N508Pfs*4](#)

AsnGlyIleP roCysHisPr oSerCysGln AsnCysHisS top

TGGTGGGGCC ACATGCTGAC CCAGCCCCAC ATCCACATTC TGTTAGAATG *58

TAGCTCAGGG AGCTTCGTGG ACGGCCTTGC TTGCTGTAGC GTTGTAGGGG *108

CCCTGCCTGC ACTGCGGTTG TCCACGGTCA CATCTGCCCC AGTGCCTTTG *158

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CAGCCCGTCA GGTCCGGCTC TCGTCTCCC TCTCTGCAGC CTGTGTAAGC *508
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GAATGAGGCT GTGACTGAGC ACTGGGACCT TTCTACCAGA TGTGGACCCC *708
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CGGGAGGGGA AGTCTTGCCA CTCCTGCTCC CTTTTGACCT CTCAGCAGGC *858
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AGTATCGATT CAGCAGTATT TACTAGAACC CACTCTGTGC TGGTCGGAGG *1008
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CCCCTCTCTT GCCAGGGCCT GGCTGGTGT CTCTCAGGAG CCTGGGCATG *1558
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GGCAACATCG TTTACCACAT TAAAATCTAG ATGCCCTGCT TCTCTGAAA *1708

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RBCK1 (NM_031229.4) - cDNA + Protein - 2024-12-21

