RBCK1 (NM_031229.4) - cDNA + Protein - 2026-02-23

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ACTTTCACTT TCTCTTCCGC CGAAGCCGCT CCCCTTGCGA AGAACTGGGG -410  c.ex1_ex4del
CCTCCCGGGA GGAGAGAGGG CTTTGCCTTG AAACCCGGGA CGCCAGGGGC -360
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CCCACGCAGG ATCCCGGCCT GGTCACCGGG CAGTGTGATG CTTCCCGACT -160
GCCGCGGGGA CAGCGAGGCA CACACAGGGC TTGGGCCGCG CCGGAGGCCA -110
CACGGCCTGG CTGAGTTGCT CCTGGTCTCC CGCCTCTCCC AGGCGACCCG -60
GAGGTAGCAT TTCCCAGGAG GCACGGTCCC CCCCAGGGGG ATGGGCACAG -10
CCACGCCAGA TGGACGAGAA GACCAAGAAA GCAGAGGAAA TGGCCCTGAG 41
         M etAspGluLy sThrLysLys AlaGluGluM etAlaLeuSe 14

CCTCACCCGA GCAGTGGCGG GCGGGGATGA ACAGGTGGCA ATGAAGTGTG 91   c.52G>C
rLeuThrArg AlaValAlaG lyGlyAspGl uGlnValAla MetLysCysA 31

CCATCTGGCT GGCAGAGCAA CGGGTGCCCC TGAGTGTGCA 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
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aGlnMetHis ThrValThrI leTrpLeuTh rValArgPro AspMetThrV 81

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alAlaSerLe uLysAspMet ValPheLeuA spTyrGlyPh eProProVal 97

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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 GCCCCCCAAA GCCCGGGGTC CCCCAGGAAC 541  c.494delG
oArgGlyPro LeuGluProG lyProProLy sProGlyVal ProGlnGluP 181

CCGGACGGGG GCAGCCAGAT GCAGTGCCTG AGCCCCCACC GGTGGGCTGG 591  Q185X
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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

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luCysProVa lCysTyrSer ValLeuAlaP roGlyGluAl aValValLeu 297

CGTGAGTGTC TGCACACCTT CTGCAGGGAG TGCCTGCAGG GCACCATCCG 941 c.896_899del C305F

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gAsnSerGln GluAlaGluV alSerCysPr oPheIleAsp AsnThrTyrS 331

CGTGCTCGGG CAAGCTGCTG GAGAGGGAGA TCAAGGCGCT CCTGACCCCT 1041

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GluAspTyrG lnArgPheLe uAspLeuGly IleSerIleA laGluAsnAr 364

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gSerAlaPhe SerTyrHisC ysLysThrPr oAspCysLys GlyTrpCysP 381

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TCTTTGAGGA TGATGTCAAT 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

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CTGCACCGTC TGCCACACCG AGATCTGCTG GGTCACCAAG GGCCCACGCT 1441  T470S  E471K
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rpGlyProGl yGlyProGly AspThrSerG lyGlyCysAr gCysArgVal 497

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TCCTTCCCTT GGGGCTTGCC GGCCAGACTT CTCTCCCCTG CGGCTCCCAC *208
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RBCK1 (NM_031229.4) - cDNA + Protein - 2026-02-23