RBCK1 (NM_031229.4) - cDNA + Protein - 2025-04-02

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ACTTTCACTT TCTCTTCCGC CGAAGCCGCT CCCCTTGCGA AGAACTGGGG -410 c.ex1_ex4del
CCTCCCGGGA GGAGAGAGGG CTTTGCCTTG AAACCCGGGA CGCCAGGGGC -360
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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
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GAGGTCTCCC CAACGCAGGA CATCAGGCTG TGGGTGAGCG TGGAGGATGC 191
GluValSerP roThrGlnAs pIleArgLeu TrpValSerV alGluAspAl 64

TCAGATGCAC ACCGTCACCA TCTGGCTCAC AGTGCGCCCT GATATGACAG 241
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alAlaSerLe uLysAspMet ValPheLeuA spTyrGlyPh eProProVal 97

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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
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CAGTGCCCCG GGTGCACCTT CATCAACAAG CCCACGCGGC CTGGCTGTGA 641
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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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CAACAGCCAG GAGGCGGAGG TCTCCTGCCC CTTCATTGAC AACACCTACT 991  A319V
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GluAspTyrG lnArgPheLe uAspLeuGly IleSerIleA laGluAsnAr 364

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gSerAlaPhe SerTyrHisC ysLysThrPr oAspCysLys GlyTrpCysP 381
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AACTGCCTGC TCTGCAAGGC CATCCATGAG CAGATGAACT GCAAGGAGTA 1241
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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 TGCCACACCG AGATCTGCTG GGTCACCAAG GGCCCACGCT 1441 T470S E471K
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GGGGCCCTGG GGGCCCAGGA GACACCAGCG GGGGCTGCCG CTGCAGGGTA 1491 T489Pfs*9
rpGlyProGl yGlyProGly AspThrSerG lyGlyCysAr gCysArgVal 497

AATGGGATTC CTTGCCACCC AAGCTGTCAG AACTGCCACT GAGCTAAAGA *8   N508Pfs*4
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RBCK1 (NM_031229.4) - cDNA + Protein - 2025-04-02

nfevers