



RBCK1 (NM_031229.4) - cDNA - 2024-05-18

ACTTTCACTT 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 TTGGGCCGCG CCGGAGGCCA -110
CACGGCCTGG CTGAGTTGCT CCTGGTCTCC CGCCTCTCCC AGGCGACCCG -60
GAGGTAGCAT TTCCAGGAG GCACGGTCCC CCCAGGGGG ATGGGCACAG -10
CCACGCCAGA **TGGACGAGAA** **GACCAAGAAA** **GCAGAGGAAA** **TGGCCCTGAG** 41
CCTCACCCGA **G**CAGTGGCGG **G**CGGGGATGA **A**CAGGTGGCA **A**TGAAGTGTG 91 [c.52G>C](#)
CCATCTGGCT **G**GCAGAGCAA **C**GGGTGCC**C** **T**GAGTGTGCA **A**CTGAAGCCT 141 [L41fsX7](#)
GAGGTCTCCC **C**AACGCAGGA **C**ATCAG**G**CTG **T**GGGTGAGCG **T**GGAGGATGC 191
TCAGATGCAC **A**CCGTCACCA **T**CTGGCTCAC **A**GTGCGCCCT **G**ATATGACAG 241
TGGCGTCTCT **C**AAGGACATG **G**TTTTTCTGG **A**CTATGGCTT **C**CCACCAGTC 291
TTGCAGCAGT **G**GGTGATTGG **G**CAGCGGCTG **G**CACGAGACC **A**GGAGACCCT 341
GCACTCCCAT **G**GGGTGCGGC **A**G**A**ATGGGGA **C**AGTGCCTAC **C**TCTATCTGC 391 [N122H](#)
TGTCAGCCCG **C**AACACCTCC **C**TCAACCCTC **A**GGAGCTGCA **G**CGGGAGCGG 441
CAGCTGCGGA **T**GT**G**GAAGA **T**CTGGGCTTC **A**AGGACCTCA **C**GCTGCAGCC 491 [c.456+1G>C](#)
GCGGG**G**CCCT **C**TGGAGCCAG **G**CCCCCAAA **G**CCCCGGGTC **C**CCCAGGAAC 541 [c.494delG](#)
CCGGACGGGG **G****C**AGCCAGAT **G**CAGTGCCTG **A**GCCCCCACC **G**GTGGGCTGG 591 [Q185X](#)
CAGTGCCCCG **G**GTGCACCTT **C**ATCAACAAG **C**CCACGCGGC **C**TGGCTGTGA 641

GATGTGCTGC CGGGCGCGCC CCGAGGCCTA CCAGGTCCCC GCCTCATACC 691 [Q231Sfs*45](#)
 AGCCCGACGA GGAGGAGCGA GCGCGCCTGG CGGGCGAGGA GGAGGCGCTG 741 [P190fs](#) [c.722del](#) [c.727_728insGGCG](#) [c.727G>T](#)
 CGTCAGTACC AGCAGCGGAA GCAGCAGCAG CAGGAGGGGA ACTACCTGCA 791 [Q222X](#)
 GCACGTCCAG CTGGACCAGA GGAGCCTGGT GCTGAACACG GAGCCCGCCG 841 [Q267*](#) [L273Pfs*27](#)
 AGTGCCCCGT GTGCTACTCG GTGCTGGCGC CCGGCGAGGC CGTTGGTGCTG 891 [V295L](#)
 CGTGAGTGTC TGCACACCTT CTGCAGGGAG TGCCTGCAGG GCACCATCCG 941 [c.896_899del](#) [C305F](#)
 CAACAGCCAG GAGGCGGAGG TCTCCTGCCC CTTCAATTGAC AACACCTACT 991 [A319V](#)
 CGTGCTCGGG CAAGCTGCTG GAGAGGGAGA TCAAGGCGCT CCTGACCCCT 1041
 GAGGATTACC AGCGATTTCT AGACCTGGGC ATCTCCATTG CTGAAAACCG 1091 [c.1054C>T](#)
 CAGTGCC TTC AGCTACCATT GCAAGACCCC AGATTGCAAG GGATGGTGCT 1141
 TCTTTGAGGA TGATGTCAAT GAGTTCACCT GCCCTGTGTG TTTCCACGTC 1191 [c.1160A>G](#)
 AACTGCCTGC TCTGCAAGGC CATCCATGAG CAGATGAACT GCAAGGAGTA 1241
 TCAGGAGGAC CTGGCCCTGC GGGCTCAGAA CGATGTGGCT GCCCGGCAGA 1291
 CGACAGAGAT GCTGAAGGTG ATGCTGCAGC AGGGCGAGGC CATGCGCTGC 1341
 CCCCAGTGCC AGATCGTGGT ACAGAAGAAG GACGGCTGCG ACTGGATCCG 1391
 CTGCACCGTC TGCCACACCG AGATCTGCTG GGTACCAAG GGCCACGCT 1441 [T470S](#) [E471K](#)
 GGGGCCCTGG GGGCCCAGGA GACACCAGCG GGGGCTGCCG CTGCAGGGTA 1491 [T489Pfs*9](#)
 AATGGGATTC CTTGCCACCC AAGCTGTGAG AACTGCCACT **G**AGCTAAAGA *8 [N508Pfs*4](#)
 TGGTGGGGCC ACATGCTGAC CCAGCCCCAC ATCCACATTC TGTTAGAATG *58
 TAGCTCAGGG AGCTTCGTGG ACGGCCTTGC TTGCTGTAGC GTTGTAGGGG *108
 CCCTGCCTGC ACTGCGGTTG TCCACGGTCA CATCTGCCCC AGTGCCTTTG *158
 TCCTTCCCTT GGGGCTTGCC GGCCAGACTT CTCTCCCCTG CGGCTCCCAC *208
 CTCTGCCTGA CCCCAGCCTT AAACATAGCC CCTGGCCAGA GGCCTTGCTG *258
 GGTGGAGCCT CTGTGTGACT CCATACTCCT CCCACCACAA CACTCATCTG *308
 TCAAACACCA AGCACTCTCA GCCTCCCCGC CTTCAGCTGT CAGCTTTCTG *358
 GGGCTAACTT CTCTGCCTTT GTGGTTGGAG GCCTGAGGCC TCTTGGAAGT *408
 CTTGCTAACC TGTTCAGAGC CAGGAAGGAG ACTGCACAGT TTTGAAAGCA *458
 CAGCCCGTCA GGTCCGGCTC TCGTCTCCC TCTCTGCAGC CTGTGTAAGC *508

TATTATAATT AAAATGGTTT TCCGGGAAGG GATGAGTGTG ATGTCCTTGA *558
GAGGAAATGA ATGTCCTGGC CTGGGACTCT ACACACAGGC AGGATCCTGA *608
GGTCTCTGGG AACTGCATCA GAAAGTTGAC TTGTCAGTCC ATCTGTGGTA *658
GAATGAGGCT GTGACTGAGC ACTGGGACCT TTCTACCAGA TGTGGACCCC *708
ATGCCCAGCC TCAGGGGCAA GGATGCTCTT GGGTCACCGT CAGCCAGGAC *758
AGGTGGAGTG TGCAGTGTGT CAAGTCTGCA GAGAAGGATG GGCTTAGGGG *808
CGGGAGGGGA AGTCTTGCCA CTCCTGCTCC CTTTTGACCT CTCAGCAGGC *858
ATCTAGGGTT GGCAGGTAGA TAGTTCAAGA AGGAACGAAG CTGCTGCAGT *908
TGAGGGGTGG GGTGTGCCAT CCTATTTTCT CGTCTCAAGC AAGATGGCAC *958
AGTATCGATT CAGCAGTATT TACTAGAACC CACTCTGTGC TGGTCGGAGG *1008
TTACTAAGAC AGGGTCCTGG GATGTTTATT CTCTAAGTCT TTCCTCCGCT *1058
CTGTGACCCA CCCTCCTTCC CCTTTTGAGA TCTGGTATTT GATGCCCAAC *1108
ACATTGTCCA CGCTGTGACG TGACCATCAT CATAGCAGGC AGAGGGCGCC *1158
TCTGCTGCTG AAGGCCTGTG ATTTTGTGGG GAAGGGCCTG TTCTAGCAAC *1208
TGAAAGGCA CTGCCACCTG CCGTTGGATG CCAGGACTCA AGAGCTGGCC *1258
CCAGTCACTG TGCGCAGAGC TGTCTGAGAA TGTGTGAGTG GACTGGGTCC *1308
TTCGGCACTG CCTGCATTGG CTCAGGGCAG TCAACCGTCG CAGAGGATGA *1358
GGGGCACACT CAGGCAGCCT CCCCggccCT GGAGGCAGAA AGGCCCAGGC *1408
AGAACCACTG ACTGGGAGGA AACAGAAAAA GCAGAGGAGA GCCAGGCTGC *1458
AGGCGTGTGG ATGGGACCAG CTCAGGCAGA CGCTGTCTCA TACCACTCT *1508
CCCCTCTCTT GCCAGGGCCT GGCCTGGTGT CTCTCAGGAG CCTGGGCATG *1558
AGACAAAAGC AGAGATTGTT CTCTTGTGGT ACCACAGGCT GTAACCAGTC *1608
CACCCAGTGT TGTTTTAGAA ATTTAAATCG GTTGCCCATC TTTTAAATT *1658
GGCAACATCG TTTACCACAT TAAATCTAG ATGCCCTGCT TCTCTTGAAA *1708

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