

Supplementary Table 1. List of primers used to amplify the coding regions of *RAB23*

| Exons | Forward primer | Reverse primer |
|---------------|--------------------------------------|-----------------------------|
| 1 | TGCAATGTGATAATTTCTTGATAAAA | CGAAATCCACTGCAATCAGTT |
| 2 | GGTTTATTGGGACAACCCATC | GAAGAATAATGAACTGGGGTTTT |
| 3 | GGATAAAAGTTGCCCCCATC | TCATTAAAGCCTTCAAAATGAAA |
| 4 | CCTTCCCTGCCAAACTGTAT | TGGAAAACCTTACGAGGATGA |
| 5 | CATTGTACTCAGATACTGCCCAA | TCACTTTTAAATCACATTTCTGAAAGA |
| 6 | AGATGCTAAATTTTGTGGATTTT | TTCAGAGAGCCATTAGGAGCA |
| cDNA | TGAGCCAAATTGTGCTGTTC | GGGCAATAATTTTTCCACCA |
| TaqMan | TTTGGAGCGACAAATTCAAG | TCCAACCTGGAAACTGCTTCA |
| Probe | 5`Fam_CCTCCTGACCTGCAGTGTCCCA_3`Tamra | |

Supp. Table 2. Consensus value for potential 5` and 3` motifs of exon 5 for *RAB23* compiled using HSF software (<http://www.umd.be/HSF/>) [Desmet et al.2009].

| Position | Splice site type | Motif | New potential splice site | Consensus value* |
|----------|------------------|----------------|---------------------------|------------------|
| -12 | Acceptor | ttttattctagTT | ttttattctagTT | 84.19 (WT) |
| -4 | Acceptor | ctagTTTTTAAGTA | ctagttttaagTA | 75.65 |
| 5 | Donor | TAAGTATTT | TAAGtattt | 66.32 |
| 21 | Acceptor | AAATACCTTCAGAA | aaatacctcagAA | 82.38 |
| 40 | Acceptor | AACAACAAATAGCT | aacaacaaatagCT | 66.83 |
| 45 | Acceptor | CAAATAGCTGAGGA | caaatagctgagGA | 69.13 |
| 49 | Donor | TAGCTGAGG | TAGctgagg | 68.09 |
| 52 | Acceptor | CTGAGGATCCAGAA | ctgaggatccagAA | 79.97 |
| 68 | Acceptor | AACGCATTCAAGTA | aacgcattcaagTA | 71.25 |
| 71 | Acceptor | GCATTCAAGTAGTA | gcattcaagtagTA | 70.84 |
| 76 | Donor | CAAGTAGTA | CAAgtagta | 66.43 |
| 78 | Acceptor | AGTAGTAACAAGAT | agtagtaacaagAT | 67.32 |
| 79 | Donor | GTAGTAACA | GTAgtaaca | 68.56 |
| 91 | Donor | TTGgtaagt | TTGgtaagt | 93.27 (WT) |
| 94 | Acceptor | Gtaagtacatagga | gtaagtacatagGA | 74.15 |
| +2 | Donor | Taagtacat | TAAgtacat | 66.47 |

*The consensus value (CV) was set between 0-100. Lowercase nucleotides denote intronic sequences. Uppercase nucleotides represent exonic sequences. WT: wildtype indicates the consensus splice sites.