

ADDITIONAL FILE 4

Sequence of extended 5' UTR ends of the known *CLC-5* transcripts (type 3, 4 and alternative 4) and sequence of the newly identified *CLCN5* 5'UTR variants (types 6-11). The alternative splice sites are indicated (*).

mRNA variant 3

EXTENDED
EXON 1a (242 bp) GCTGGTCCAAAGGTCTGGTCACGTGACTCCAACAGTGACATCACATTCTGACCGCCAGGG
AACACTGGTAGTTCTTTAGGTGGCGTTTTGTTGCTGTGAAAGGCAGAGAATGCAGCAAGTG
CCCTTCTAAAGCACTTCAGACTGGGGCTTCTTTTGTAAAAGCTCCCCAACCTGAATGACC
AAATGCGGGCTGCCAAGGTTATTTCTTCTTGGAGGCAGAAAAAAGAAAACTGTGATTGA
AG
EXON 2 (152 bp) AGGACAAGTCGTACAATGGTGGAGGAATAGGTTCTTCAAATAGGATCATGGACTTCTTGG
AGGAGCCAATCCCTGGTGTAGGGACCTATGATGATTCAATACAATTGATTGGGTGAGAG
AGAAGTCTCGAGACCGGGATAGGCACCGAGAG

mRNA variant 4 and alternative variant 4

ATGATTTGGACTCTGCAGAGCTTTTTCTTTCAAATGTGCACCTTGCCTGTGAAAGGCATG
CTGACTTAGGTATTCAGAAGACTTACAATTGGTAAACCAATTGTAAGTACAGCTTGGGGT
GTTCTTCAAACCTTTCCCATGACATTGCTAAAGTTGTCTGTTTGGAGTTTACAGTGAAG
ATGGGGATGCAGATGGGAGGAGAGGGAGTCAGCTCAGGCCCACTTTGGCCTCCTCTGCAG
AGGGATTCATTACCAAACAGTCTGGAGGAAGCCTTGTCCCTGCCAAGGTTGGTGTGCAA
ATGTGAGCTTTGGCTGGGTGAATTAATGATGGCATTACTCTGCACTTTTTCCCAAGTGTG
CATTTGCATAGATTCCCTCAGTACTGTTTTGTATTTTGTGTTAATAGCAAAAATCTTATTC
CATCAGATGATGTTGCTGAAATATTTTGAATCAAGTTTAAAGCCTTGTGTGAGTATAGCTT
GTGAGTTTAAAACCTTCTCCCCCATTTCCACCGTTTCATCAAACAAATTTTGGTTCAGCTG
ATATATACTGATTCTTGGATATGTGCTAGACACTGTAGAGAAGGGAACGATGAATCAGAC
EXENDED
EXON 1b (1001 bp) ATCAGGCTACCAGCAAAGAGCTTGTAGTCTAGAAAAGGAAGATAAAGCATGCACAAAAGTA
ATTGTAATAGAAGTACCCAAAGCAAAGTAGATAAGGGACTATGATGGGTGGGCAGGTGGA
TATGTTTCTTGCAGTTGAAGGATCAGGGAGGGACATTTTCATCACTGAATCATACTGTT
CTTTAAATCTCTGCTTGTAAATCTTAGACTGTGCTGACATACTTAGAGTTCCCTACAAGCC
TGTTACATCAGAGATCAAGTCTTATACCTATTGAAGGGAAATTTGTACATCCAGCCAAC
TAACTCGTGAAGAGGCCACATGCTTTATTTGGAGTCCCCAGCCAGGCAAAAACCACTT
CATAGGATTGAATTATTGCCTTATAAATGTAAAATGGTTCT
TGATGTGATATGGCTGCAAGTGCCTTTGACCCTTTTGTCTCCCTCCATAAACTGAAATA
EXON 1b (244 bp) TGATGTGATATGGCTGCAAGTGCCTTTGACCCTTTTGTCTCCCTCCATAAACTGAAATA
CCTAAGCTGCTCCAACCTCCTTTTTGTCTTTTGTTCATAAATCCTTTCCATTGCACAT
CAACTCCTGTCTCTCTTTGTACTGTCACTCTCATCTGTTGCTTTCCATTACACTGCCTT
TAGCCACTCATCATTTTGTGCCTACACCACAGAAACCTCTGAATGTAATGGATGTTCTTA
CCAG*
GTGACTGTATTCTTTATCAACCCCACTGTAGTCATCTGATAGTTAAGGGCCCGCCTT
INTRON 1-2 (134 bp) TTGGAACCAAAAAGAATGTATTAATAAATGTTAAGAATTTCTGAAGTACTAATGTCTATTT
CTTGTTCAATACAG
AGGACAAGTCGTACAATGGTGGAGGAATAGGTTCTTCAAATAGGATCATGGACTTCTTGG
EXON 2 (152 bp) AGGAGCCAATCCCTGGTGTAGGGACCTATGATGATTCAATACAATTGATTGGGTGAGAG
AGAAGTCTCGAGACCGGGATAGGCACCGAGAG

mRNA variants 6 and 7

ATGATTTGGACTCTGCAGAGCTTTTTCTTTCAAATGTGCACCTTGCCTGTGAAAGGCATG
CTGACTTAGGTATTCAGAAGACTTACAATTGGTAAACCAATT*GTAAGTACAGCTTGGGGT

EXON c (287 bp) GTTCCTTCAAACCTTCCCATGACATTGCTAAAGTTGTCTGTTTGGAGTTTACAGTGAAG
 ATGGGGATGCAGATGGGAGGAGAGGGAGTCAGCTCAGGCCACTTGGCCCTCCTCTGCAG
 AGGGATTCATTACCAAACAGTCTGGAGGAAGCCCTGTCCCTGCCAAG
 EXON 2 (152 bp) AGGACAAGTCGTACAATGGTGGAGGAATAGTTCTTCAAATAGGATCATGGACTTCTTGG
 AGGAGCCAATCCCTGGTGTAGGGACCTATGATGATTTCAAATACAATTGATTGGGTGAGAG
 AGAAGTCTCGAGACCGGGATAGGCACCGAGAG

mRNA variants 8 and 9

EXON I (197 bp) GATACTCGCGAGGCGCCCGTCACATGAGCCGCGGCCCTGCTTCGCTCCCCCTCCTCCCC
 GGCTGGGCTGTGTGAATGAAGCTCTGCCGGCTACGTGGGGCGCTCACTCAACTTGGTGTCT
 CTGGACGCCAGAGCCGACCGAGCGCGCTGCCACCGGCGGGACACGGGCTCCGCCGCT
 CCGGACCTCGGCGACAG
 AGCAAATCAGTTGCCTGGAGTCCAGTGAAGTTGTACCTGTGCCCGTGCATAACGTCA
 EXON II (335 bp) GTGGAAGCTGCCCCGA *GTAAGTGAGGAAATCTAGAAGTTTGGAGCATAAAAAACCAGC
 ACGAATCAACTTTACATAAACTTCATACATTTATTAGCAACTGCTCGGGTTAACTTCAGG
 GATGTTAAATTTTCGCCTAGTCCCACGGGACGATGATCATTACTTCTTAACTGCACGCGAA
 ATAGGAGAAGAAAAAGGCAAATCAAACAGACTGTCCCAGGTCTCAGAGAAGTCATCGG
 AGCGCTTTTTCCTGCTGAATTCACCCTAGGAGTTT
 EXON III (144 bp) GTTTGGGGCTTTAGCCCCCTGGAGAAAACAGGGCCACATAGCTGCCTTTCTATCAAGTCT
 CCCTACAAAAGTGAAGGGCTCTGGGAAAACAGCCTGTGACCCAGCGTGGGTGAAGATA
 GGATCAAGATGGCCATGTGGCAGG
 EXON IV (147 bp) GTGCCATGGATAACAGAGGCTTTCAGCAGGGGAGTTTTAGTAGCTTCCAGAACAGCTCCA
 GTGATGAAGACCTGATGGACATTCCAGCAACCGCTATGGATTTCTCCATGAGAGATGATG
 TTCTCCCTTAGACCGAGAAGTAGGAG
 EXON VI (194 bp) AGCCAGGAGGGATTCTTAGATCTTCACTGTGAGAAGTTGGTGGGGTTTCTGGATGTAGAG
 CTTTGGTGTGGGAGCTCCCCAAGACCGTGGTCCCCAAGAGTTTCTTGTACTCACGCCAG
 TCCACACTCAGCCTTCAGCAGCTTGTTCAGTTGCCATTTGTGTTTCTACCAGCTCCAGC
 CATTCTACTCCAG
 EXON 2 (152 bp) AGGACAAGTCGTACAATGGTGGAGGAATAGTTCTTCAAATAGGATCATGGACTTCTTGG
 AGGAGCCAATCCCTGGTGTAGGGACCTATGATGATTTCAAATACAATTGATTGGGTGAGAG
 AGAAGTCTCGAGACCGGGATAGGCACCGAGAG

mRNA variants 10 and 11

EXON I (197 bp) GATACTCGCGAGGCGCCCGTCACATGAGCCGCGGCCCTGCTTCGCTCCCCCTCCTCCCC
 GGCTGGGCTGTGTGAATGAAGCTCTGCCGGCTACGTGGGGCGCTCACTCAACTTGGTGTCT
 CTGGACGCCAGAGCCGACCGAGCGCGCTGCCACCGGCGGGACACGGGCTCCGCCGCT
 CCGGACCTCGGCGACAG
 AGCAAATCAGTTGCCTGGAGTCCAGTGAAGTTGTACCTGTGCCCGTGCATAACGTCA
 EXON II (335 bp) GTGGAAGCTGCCCCGA *GTAAGTGAGGAAATCTAGAAGTTTGGAGCATAAAAAACCAGC
 ACGAATCAACTTTACATAAACTTCATACATTTATTAGCAACTGCTCGGGTTAACTTCAGG
 GATGTTAAATTTTCGCCTAGTCCCACGGGACGATGATCATTACTTCTTAACTGCACGCGAA
 ATAGGAGAAGAAAAAGGCAAATCAAACAGACTGTCCCAGGTCTCAGAGAAGTCATCGG
 AGCGCTTTTTCCTGCTGAATTCACCCTAGGAGTTT
 EXON III (144 bp) GTTTGGGGCTTTAGCCCCCTGGAGAAAACAGGGCCACATAGCTGCCTTTCTATCAAGTCT
 CCCTACAAAAGTGAAGGGCTCTGGGAAAACAGCCTGTGACCCAGCGTGGGTGAAGATA
 GGATCAAGATGGCCATGTGGCAGG
 EXON IV (147 bp) GTGCCATGGATAACAGAGGCTTTCAGCAGGGGAGTTTTAGTAGCTTCCAGAACAGCTCCA
 GTGATGAAGACCTGATGGACATTCCAGCAACCGCTATGGATTTCTCCATGAGAGATGATG
 TTCTCCCTTAGACCGAGAAGTAGGAG
 EXON V (131 bp) TGCCACCAAAGTGTCTGCCAAAGTGCCAAAGTGGCTATACCATTTTGCATTCTACCAGC
 TATGAATGAGAGTTCTGTAGTTTGCATCCTTGGTATTGTCAGTGTTTTGGATTTTACG
 CATTCTAATAG
 EXON VI (194 bp) AGCCAGGAGGGATTCTTAGATCTTCACTGTGAGAAGTTGGTGGGGTTTCTGGATGTAGAG
 CTTTGGTGTGGGAGCTCCCCAAGACCGTGGTCCCCAAGAGTTTCTTGTACTCACGCCAG
 TCCACACTCAGCCTTCAGCAGCTTGTTCAGTTGCCATTTGTGTTTCTACCAGCTCCAGC
 CATTCTACTCCAG
 EXON 2 (152 bp) AGGACAAGTCGTACAATGGTGGAGGAATAGTTCTTCAAATAGGATCATGGACTTCTTGG
 AGGAGCCAATCCCTGGTGTAGGGACCTATGATGATTTCAAATACAATTGATTGGGTGAGAG
 AGAAGTCTCGAGACCGGGATAGGCACCGAGAG

