

ADDITIONAL FILE 5

RT/PCR analysis of the *CLCN5* 5'UTR isoforms in different human tissues. **A)** common region of all isoforms (580 bp). **B)** mRNA variant 1 (645 bp) and mRNA variant 2 (386 bp). **C)** mRNA variant 3 (512 bp). **D)** mRNA variant 4 (586 bp) and mRNA alternative variant 4 (720 bp). **E)** mRNA variant 6 (417 bp) and variant 7 (602 bp). In colon is present a further PCR 782 bp long fragment which, once sequenced, was found to correspond to exon c 467 bp long, probably originated from a further alternative splicing of exon 1b1. The longest amplicon (1694 bp) represents the mRNA alternative variant 4. **F)** mRNA variant 8 (584 bp), 9 (715 bp), 10 (843 bp), and mRNA variant 11 (974 bp). **G)** Housekeeping gene GAPDH (983 bp).

