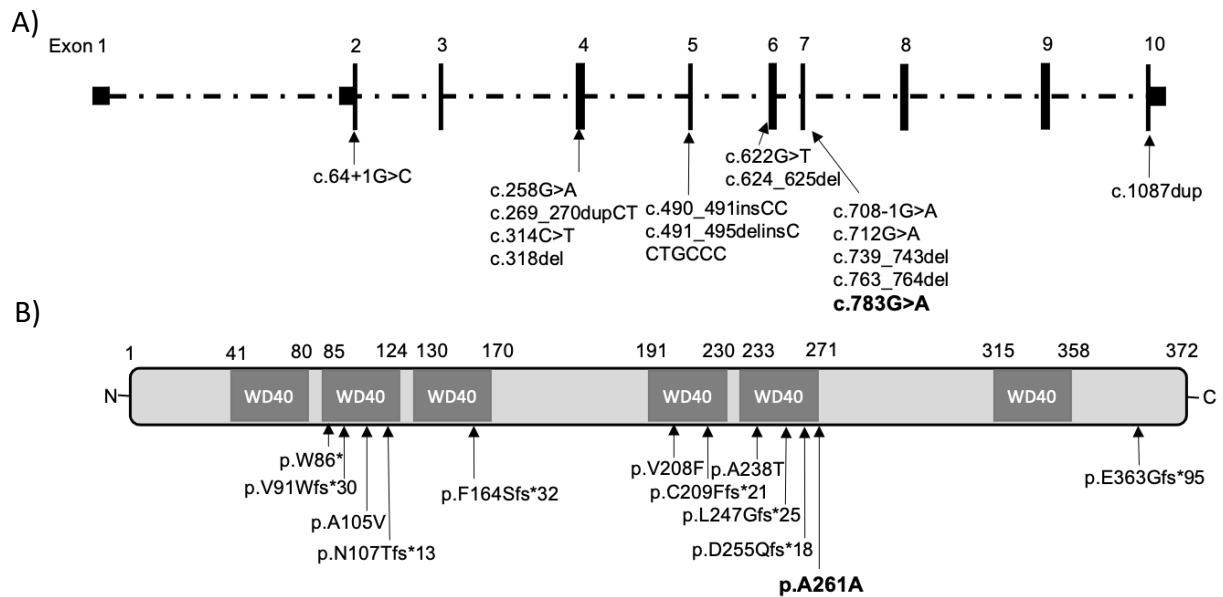


Supplementary figure 2:



A) Nucleotide positions of identified causative variants relative to ARPC1B coding exons (NM_005720.3). Two splice site, 3 missense, 1 nonsense and 8 frameshift variants have been identified until today. Variant in bold is the identified variant in present study. B) ARPC1B has 6 WD40 repeat domains forming a β -propeller required for Arp2/3 complex function. At the protein level there is no amino-acid change, however the sequencing analysis of the cDNA analysis products indicated that the variant creates a stop codon and therefore a truncated 236 amino acid protein NP_005711.1:p.(Val237*).