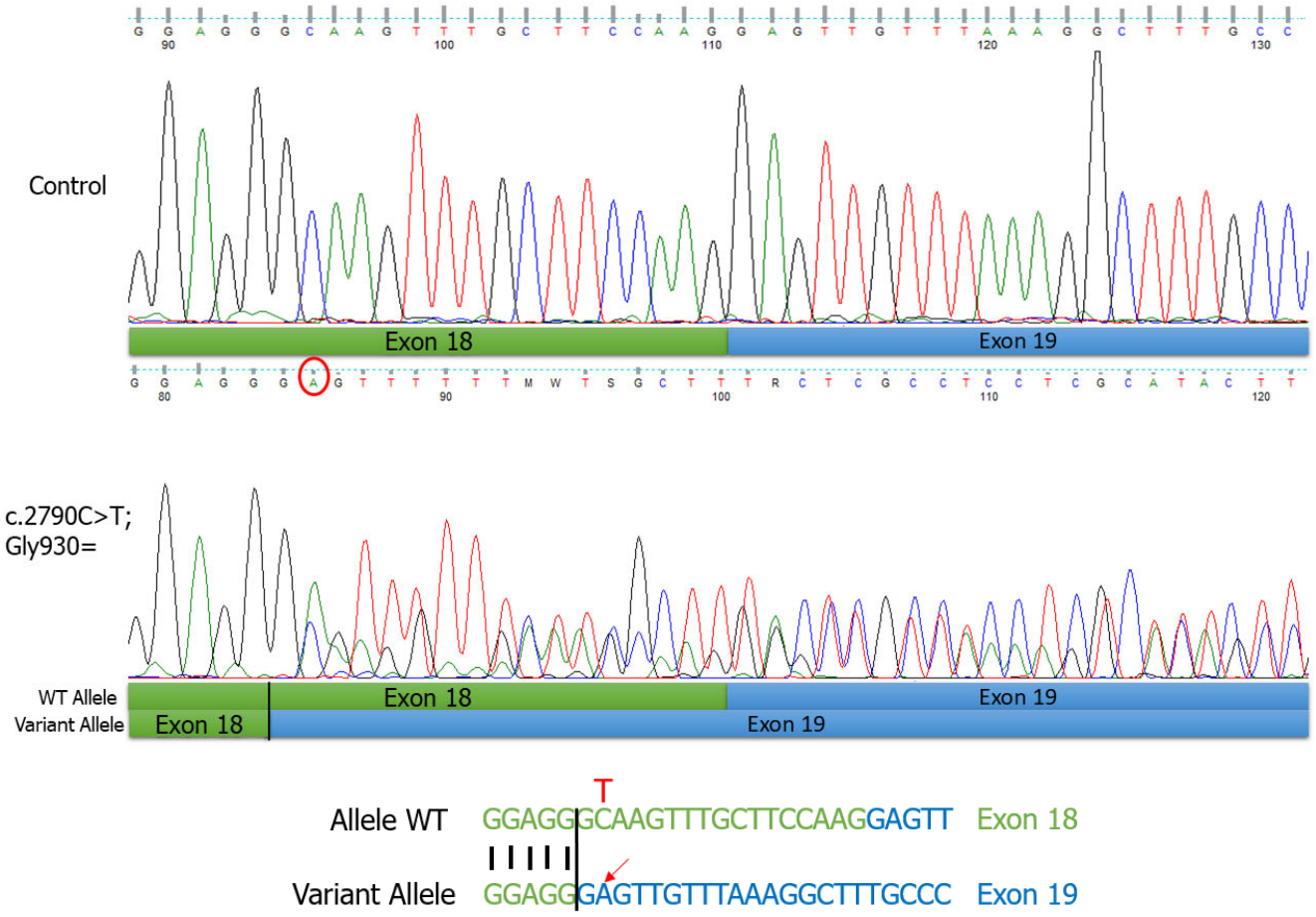

Figure S1. Sanger sequencing of the patient harboring the c.2790C>T; Gly930= synonymous variant in RECQL5 at cDNA level (lower figure) and a control sample (upper figure). The circle and arrow in red indicate the point where the change is located. The C>T change generates a GT cryptic splicing donor site at the end of exon 18, leading to the cut before these two nucleotide and generating a loss of the last 17 nucleotides of the exon and the generation of a premature stop codon in the penultimate coding exon of the gene (p.(Lys931Serfs*14).