

Figure S1. Representative plots of the MLPA analysis by Coffalyser.Net showing the probe ratios with 95% confidence intervals as error bars for all exons of the *MSH2* gene. **A.** MLPA analysis of the normal reference sample; **B.** MLPA analysis of the proband showing heterozygous deletion of exons 11-16 in the *MSH2* gene.

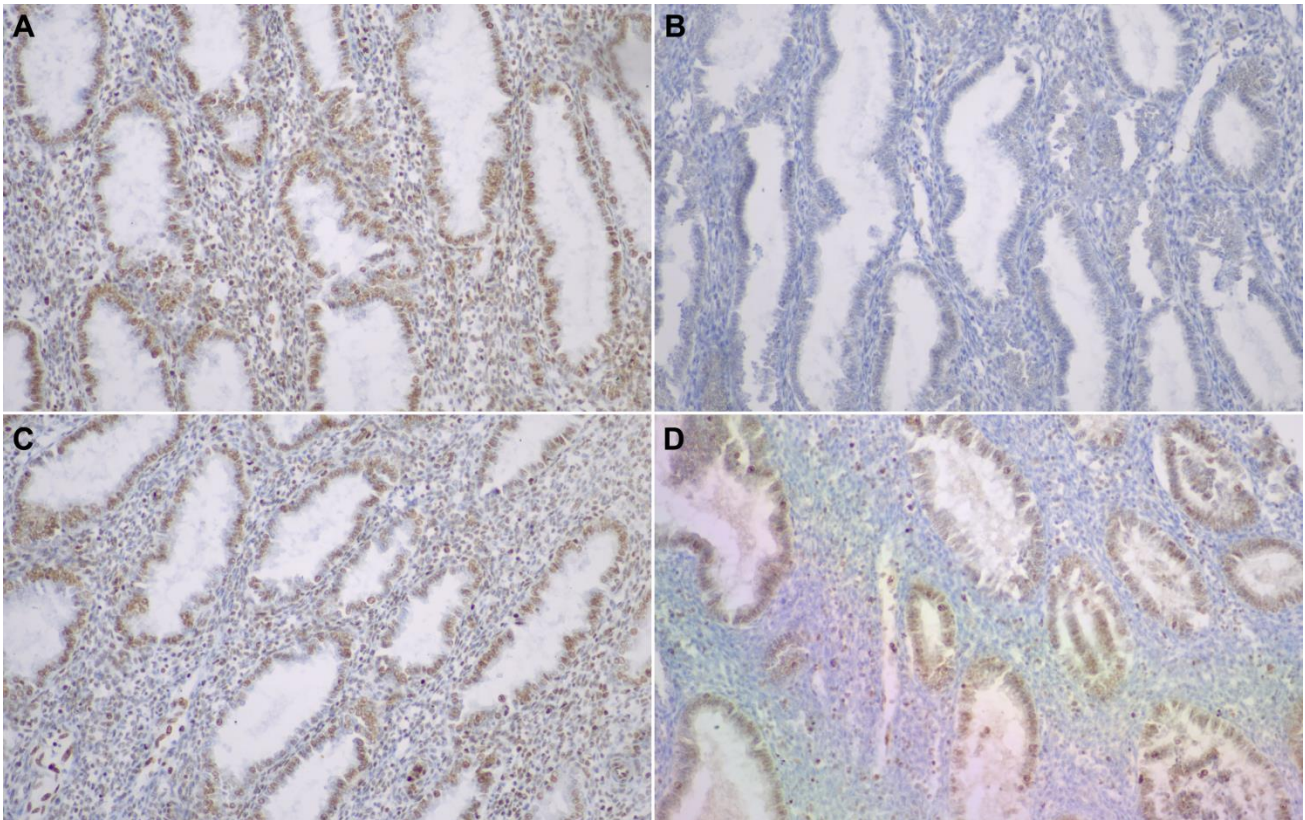


Figure S2. Immunohistochemical analysis of the expression of all four MMR proteins: **A.** MLH1 (antibody M1, ROCHE); **B.** MSH2 (antibody G219-1129, ROCHE); **C.** MSH6 (SP93, ROCHE) and **D.** PMS2 (antibody A16-4, ROCHE).

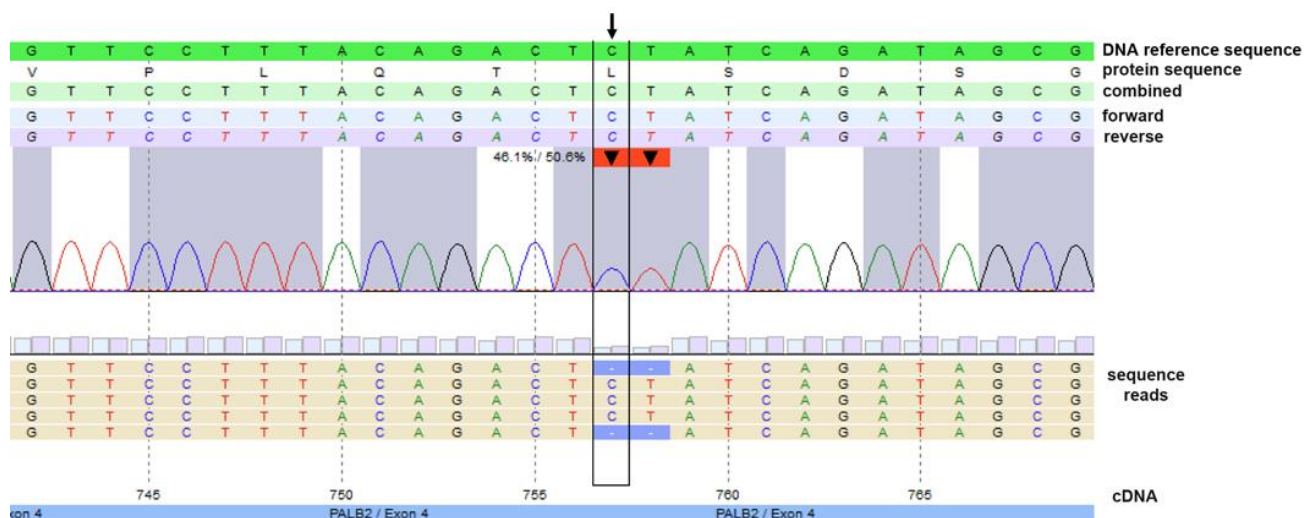


Figure S3. Next generation DNA sequencing pseudo-electropherogram demonstrating heterozygosity for the *c.757_758delCT, p.(Leu253Ilefs*3)* variant in the *PALB2* gene in our patient. Results of forward and reverse DNA sequencing of exon 4 analyzed by SeqNext. The mutation is indicated with a rectangle indicated and an arrow.