



**Supplementary Figure S2.** Multiplex-ligation-dependent probe amplification (MLPA) analysis result shown as relative signal intensities in percent of each MLPA probe in the patient compared to 6 controls demonstrates a 25% (1 of 4 copies) reduction of *PMS2* and *PMS2CL* exon 12 sequences (unspecific: G +P; black solid frame). Gene (G) and pseudogene (P) specific probes and their location in relation to *PMS2* exon 12 are shown in green and purple, respectively (schematic illustration, bottom). Probe labels are indicated below the plot. Red lines indicate relative signal intensity thresholds for calling variants.