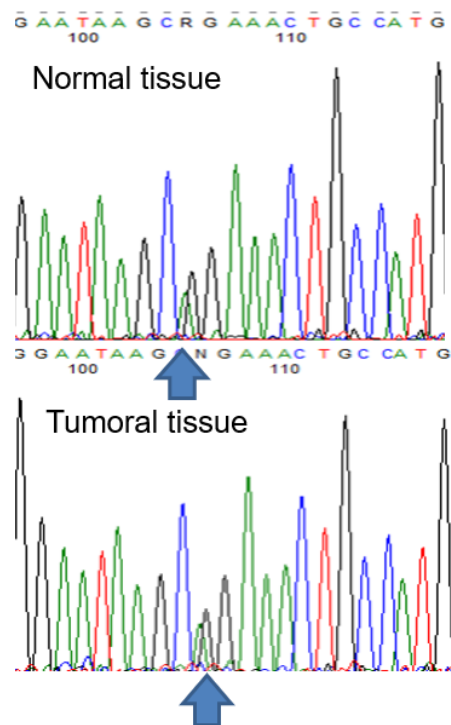


Figure S1: Size analysis with agarose migration of the PCR product for validation of the triplication and High Resolution Melting curve pike to screen this alteration. Lane 1: DNA of index case of F16; Lane 2: DNA of index case of F15; Lane 3: DNA of index case of F17; Lane 4 and 5: Controls. The primers used are the one for the families F15, F16, F17, F18 in table 5.

F17



F16

Normal tissue

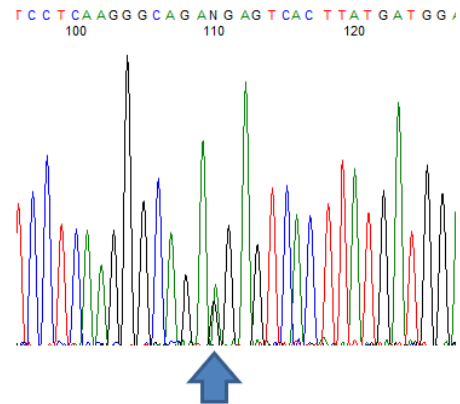


Figure S2: Functional analysis of the consequences of the *BRCA1* triplication. RNA sequencing of the coding region containing a SNP present at the heterozygous state was performed to estimate the allelic imbalance and possibly confirm a possible impact of the event on the *BRCA1* transcription. The variant c.1067A>G was analyzed in two healthy tissues and one tumoral tissue – the RNA quality of the tumor in the second case was too low. No allelic imbalance was observed in those three results.

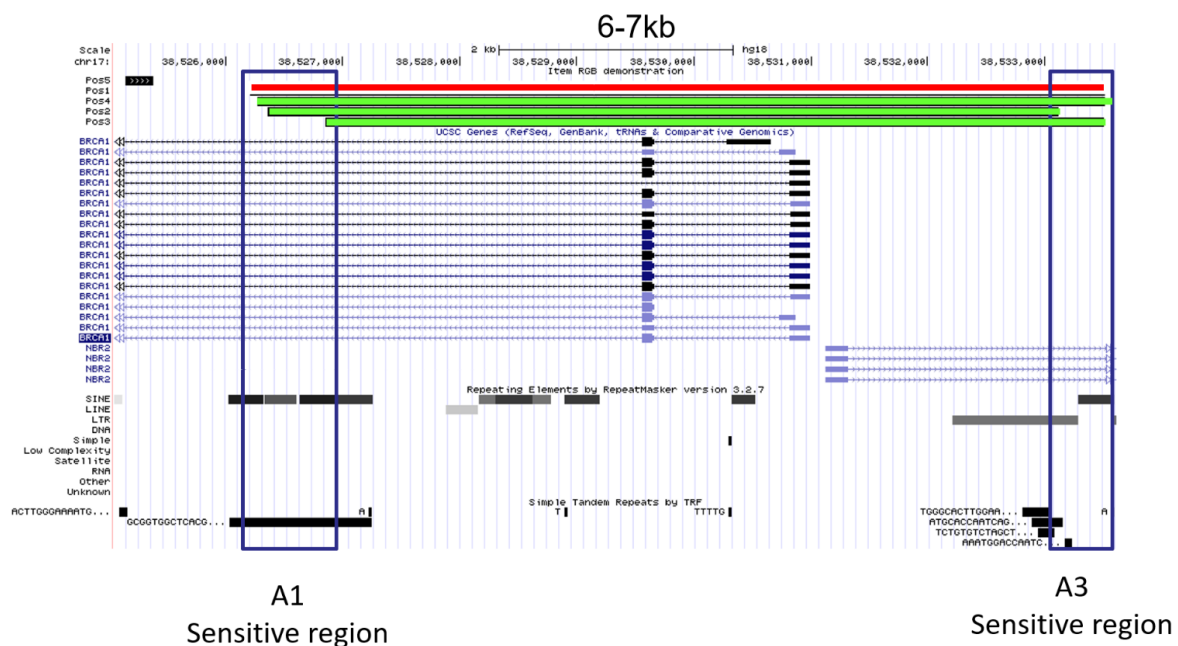


Figure S3: Structural organization of *BRCA1* 5' region to explain the deletion of 5-10kb. Description of the breakpoint for the triplication (red) and the three common deletion of 5-10 kb (green) which identifies sensitive regions with Alu sequences (A1-A3).