



**Figure S1.** Integrative Genomics Viewer (IGV) screen capture showing the alignment of the two *BRCA1* alleles. The paired-end reads obtained from patient II-4 were aligned to the human genome hg19 in the genomic range: chromosome 17, position 41,228,420 to 41,228,608. Chimeric reads containing the softclipped Poly-T reflecting the 3' end and the softclipped 5' start of the *Alu* element are shown (right red stretch and left multicolor stretch). The *Alu* Tandem Duplication Site (TDS) is boxed with green.