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Utility of circulating cell-free DNA in assessing microsatellite instability and loss of heterozygosity in breast cancer using human identification approach

Norah Alsharhan
Saudi Food and Drug Authority, Saudi Arabia

The diagnostic and prognostic utility of circulating cell-free DNA (cfDNA) in breast cancer (BC) patients was recently reported. Here, we investigated the use of cfDNA to examine microsatellite instability (MSI) and loss of heterozygosity (LOH) for early BC diagnosis. cfDNA and genomic DNA from 41 female BC patients and 40 healthy controls were quantified using NanoDrop spectrophotometry and real-time PCR. The stability of genomic and cfDNA was assessed using a high-resolution AmpFlSTR MiniFiler human identification kit. Significant increases in cfDNA plasma concentrations were observed in BC patients compared to controls. The genotype distribution of the eight autosomal short tandem repeat (STR) loci D7S820, D13S317, D21S11, D2S1338, D18S51, D16S539, FGA, and CSF1PO were in Hardy–Weinberg equilibrium. Significant differences in the allele frequencies of D7S820 allele-8, D21S11 allele-29, allele-30.2, allele-32.2, and CSF1PO allele-11 were seen between BC patients and controls. LOH and MSI were detected in 36.6% of the cfDNA of patients compared to genomic DNA. This study highlights the utility of plasma-derived cfDNA for earlier, less invasive, and cost-effective cancer diagnosis and molecular stratification. It also highlights the potential value of cfDNA in molecular profiling and biomarkers discovery in precision and forensic medicine.

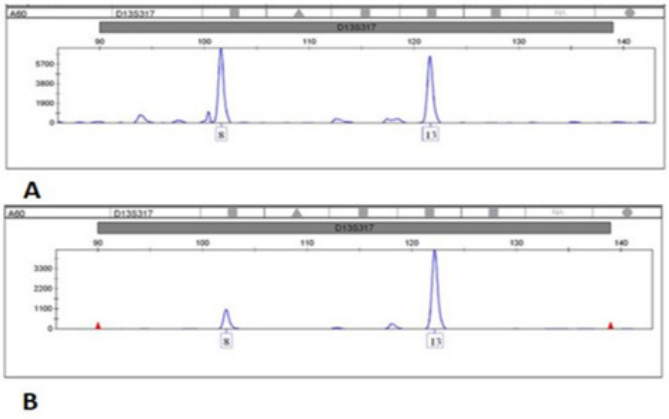

Figure 1. Loss of Heterozygosity observed at locus D13S317. Panels (**A**) represents normal genotypes in genomic DNA, while Panel (**B**) is a representative of MSI in cfDNA.

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nourahh4@hotmail.com