

Identification of mutations in *BRCA1* exon 11 of breast cancer confirmed patients and in at risk individuals

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Breast cancer is the commonest malignancy among females. Of the risk factors leading to develop breast cancer, germline mutations and sequence variance of breast cancer susceptibility genes play a major role. This study was conducted to identify germline mutations in exon 11 of *BRCA1* breast cancer susceptibility gene. Exon 11 of *BRCA1* is a common region of mutations in breast cancer patients and also in individuals with a family history of breast cancer. The study consists of a total of 43 breast cancer patients with family history (N=12) and without a family history (N=31). About 26 currently unaffected individuals with first or second degree relatives having breast cancer and 25 controls (those who are without any personnel or family history of any cancer) were also included. Self-administered questionnaires were given to the participants to evaluate the personal and family histories of breast and other cancers. Blood samples were collected for detection of mutations in exon 11 of *BRCA1* gene. Following genomic DNA extraction, mutation detection was performed using polymerase chain reaction by amplifying exon 11 which consist of 3426 base pairs that represents 60% of the coding sequence. Upon amplification, PCR products which showed abnormal migration patterns in gel electrophoresis were sequenced.

Mean age of the onset for 43 women diagnosed with breast cancer in this study was 52.256±9.70 years. Mean age at diagnosis of breast cancer patients with family history was 55.08 ± 6.788 and without family history was 51.16 ±10.51. Furthermore, abnormal migration patterns in PCR products were observed in breast cancer confirmed patients and in at risk individuals upon gel electrophoresis. PCR products which show the size variations were subjected for direct sequencing. Seventeen sequence variations were found in the exon 11 of three sequenced samples. G3135delC/exon 11 was observed. Previously reported six polymorphisms were also identified in these sequences. Results obtained from this study confirmed the presence of single nucleotide polymorphisms in the exon 11 of *BRCA1* gene of the individuals included in this study.

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