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Abstract Title: - Association Analysis of Multidrug Resistance Gene ABCB1 in Breast Cancer patients of Punjab.

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Aims: - The aim of this study is to find out the frequency distribution of genetic polymorphisms of ABCB1 (rs1128503 and rs1045642) gene in breast cancer patients and controls and the association of these genotypes, if any, with susceptibility to breast cancer.

Methods: - The present case control study was conducted on the population of Punjab (North India). The cases included 300 breast cancer confirmed patients and the control group included (n=300) healthy female volunteers from general public after taking clearance from Institutional Ethical Committee (IEC) of Punjabi University, Patiala. Genomic DNA was isolated from whole blood collected in EDTA coated tubes by employing the standard inorganic (Salting Out) extraction method (Miller et al, 1988). PCR-RFLP technique was used for the amplification of the target region of the gene by using a single pair of primers. Genotyping of the PCR products and digested fragments obtained after RFLP was done by agarose gel electrophoresis. For PCR products, 1% gel was made and for the restriction digestion products, 3.5% gel was made. After separation, the bands representing products/ fragments were visualized under UV light for genotyping.

Results: - The frequency distribution of CC, CT, and TT genotypes of rs1045642 (C3435T) among breast cancer patients were 34%, 46.67% and 19.33% and among controls were 38.67%, 45.67% and 15.67% respectively. The frequencies of CC, CT and TT genotypes of rs1128503 (C1236T) among cases were 28%, 45.67% and 26.33% and among controls were 28.33%, 42% and 29.67%, respectively. The frequency of the wild allele (C) and mutant allele (T) of rs1045642 among cases were 50.83% and 49.17% and among controls were 49.33% and 50.67%, respectively. For association analysis, the contingent chi square test did not show any statistically significant differences between cases and controls for rs1045642 and rs1128503 gene polymorphisms neither at genotypic level and nor at allelic level.

Conclusions: - We found no evidence that reveals any statistically significant association of C3435T and C1236T polymorphisms with susceptibility to breast cancer in Punjab, India.

Keywords: - The frequency distribution of CC, CT, and TT genotypes of rs1045642 (C3435T) among breast cancer patients were 34%, 46.67% and 19.33% and among controls were 38.67%, 45.67% and 15.67% respectively. The frequencies of CC, CT and TT genotypes of rs1128503 (C1236T) among cases were 28%, 45.67% and 26.33% and among controls were 28.33%, 42% and 29.67%, respectively. The frequency of the wild allele (C) and mutant allele (T) of rs1045642 among cases was 57.33% and 42.67% and among cases were 50.83% and 49.17% and among controls were 49.33% and 50.67%, respectively. For association analysis, the contingent chi square test did not show any statistically significant differences between cases and controls for rs1045642 and rs1128503 gene polymorphisms neither at genotypic level and nor at allelic level.