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Frequency & Heterozygosity Assessment of STR Markers Linked to BRCA1 Gene Applicable for Linkage Analysis in PND & PGD

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Abstract

Introduction Breast cancer is the most common cancer in women and the second most common cause of cancer mortality in women, after lung cancer (Siegel, Miller et al. 2015). BRCA1 and BRCA2 are major genes related to hereditary breast cancer and are inherited in autosomal dominant form. Finding the causative mutated gene is very important and can help in prenatal diagnosis (PND) and pre-implantation Genetic Diagnosis (PGD) for high risk families. Linkage studies with the help of STR markers are very helpful to track the possible mutated gene. These markers also help in detecting "Allele Drop out" (ADO) in PGD procedure. This study aimed to find polymorphic STR markers linked to the BRCA1 gene for use in linkage studies in extended pedigrees and also in of PND & PGD studies.

Materials and Methods: 50 unrelated individuals were genotyped to assess the allele frequencies, heterozygosity of the selected markers. Polymorphic STR markers were selected from Tandem Repeats Finder and Sequence-based Estimation of Repeat Variability databases (3, 4) (Legendre, Pochet et al. 2007) websites (Benson 1999). Suitable primers were designed to be to set up a multiplex-PCR reaction. Genotyping of each individual were performed using fragment analysis by ABI Genetic Analyzer 3130.Statistical analysis was performed using GenAlEx6.03.

Results: Our results showed that the heterozygosity of selected markers were between 41%-95%.

Totally, 43 alleles were observed. The highest heterozygosity was observed for D17SDBRCA17.41and the lowest for D17SDBRCA126.04. 6, 3,5 ,7 ,3 ,10,9 alleles were seen for D17SUBRCA118.85, D17SUBRCA115.53, D17SUBRCA113.07, D17SUBRCA18.06, D17SUBRCA11.89, D17SDBRCA17.41, D17SDBRCA126.04 respectively.

Conclusions: Using these markers in a multiplex-PCR can be a simple, cheap, and fast tool for finding the possible mutated gene for cancer in the families under investigation. The next step would be direct sequencing of candidate gene. It is also very helpful for finding carrier members of the family.