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SUSCEPTIBILITY TO PCOS IN SOUTH INDIAN WOMEN

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Objective: Polycystic Ovary Syndrome (PCOS) is a heterogeneous multifactorial endocrine metabolic disorder. In addition to hyperandrogenism, acne, hirsutism, obesity, oligoanovulation and infertility, insulin resistance is also a common feature in women of PCOS. Tumor suppressor genes (TSGs) perform essential function in the maintenance of genomic stability and regulatory pathways influencing the activity of several replication and transcription factors. The main aim of this study was to investigate the association of single nucleotide polymorphisms (SNPs) of *TP53, BRCA1and BRCA2* genes with the susceptibility to PCOS in South Indian women.

Study Design: Present study investigated association between *TP53 gene (rs1042522 G/C), BRCA1 (rs71361504 –/GTT, rs3092986 T/C) and BRCA2 (rs206118 A/G)* and, SNPs and PCOS risk. Genotyping of TSGs was carried out on DNA from PCOS patients (n=110) and controls (n=130) of South Indian origin by polymerase chain reaction (PCR) and confirmed by sequencing analysis. Haplotype frequencies for multiple loci and the standardized disequilibrium coefficient (D') for pairwise linkage disequilibrium (LD) were assessed by Haploview Software.

Results: Our results showed significant increase in frequencies of *TP53 (rs1042522 G/C), BRCA1 (rs71361504 –/GTT, rs3092986 T/C)* genotypes and alleles in patients compared to controls. In addition, the frequency of the *C/T (P*=0.002) and *A/C (P*=0.012) haplotype was also significantly elevated in patients. But *BRCA2 (rs206118 A/G)* did not show significant association with PCOS.

Conclusion: the *TP53 and BRCA1* and may constitute an inheritable risk factor for PCOS in South Indian women.

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