Genetic Analysis of CYP11A1 Gene with Polycystic Ovary Syndrome from North India

Authors : Ratneev Kaur, Tajinder Kaur, Anupam Kaur

Abstract : Introduction: Polycystic Ovary Syndrome (PCOS) is a heterogenous disorder of endocrine system among women of reproductive age. PCOS is characterized by hyperandrogenism, anovulation, polycystic ovaries, hirsutism, obesity, and hyperinsulinemia. Several pathways are implicated in its etiology including the metabolic pathway of steroid hormone synthesis regulatory pathways. PCOS is an androgen excess disorder, genes operating in steroidogenesis may alter pathogenesis of PCOS. The cytochrome P450scc is a cholesterol side chain cleavage enzyme coded by CYP11A1 gene and catalyzes conversion of cholesterol to pregnenolone, the initial and rate-limiting step in steroid hormone synthesis. It is postulated that polymorphisms in this gene may play an important role in the regulation of CYP11A1 expression and leading to increased or decreased androgen production. The present study will be the first study from north India to best of our knowledge, to analyse the association of CYP11A1 (rs11632698) polymorphism in women suffering from PCOS. Methodology: The present study was approved by ethical committee of Guru Nanak Dev University in consistent with declaration of Helsinki. A total of 300 samples (150 PCOS cases and 150 controls) were recruited from Hartej hospital, for the present study. Venous blood sample (3ml) was withdrawn from women diagnosed with PCOS by doctor, according to Rotterdam 2003 criteria and from healthy age matched controls only after informed consent and detailed filled proforma. For molecular genetics analysis, blood was stored in EDTA vials. After DNA isolation by organic method, PCR-RFLP approach was used for genotyping and association analysis of rs11632698 polymorphism. Statistical analysis was done to check for significance of selected polymorphism with PCOS. Results: In 150 PCOS cases, the frequency of AA, AG and GG genotype was found to be 48%, 35%, and 13% compared to 62%, 27% and 8% in 150 controls. The major allele (A) and minor allele (G) frequency was 68% and 32% in cases and 78% and 22% in controls. Minor allele frequency was higher in cases as compared to controls, as well as the distribution of genotype was observed to be statistically significant (x^2=6.525, p=0.038). Odds ratio in dominant, co-dominant and recessive models observed was 1.81 (p=0.013), 1.54 (p=0.012) and 1.77 (p=0.132) respectively. Conclusion: The present study showed statistically significant association of rs11632698 with PCOS (p=0.038) in North Indian women.

Keywords : polycystic ovary syndrome, CYP11A1, rs11632698, hyperandrogenism

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