

Abstract ID: - 79

Abstract Topic: - Complex traits and polygenic disorders

Abstract Title: - Clinical significance of FSHR polymorphisms with Polycystic Ovary Syndrome: An association study in the Punjabi Population

Presenting author name: - Mandeep Kaur

Presenting author institute: - Guru Nanak Dev University

Co-authors name: - Sukhjashanpreet Singh, Anupam Kaur

Co-authors institute: - Guru Nanak Dev University, Guru Nanak Dev University

Aims: - The polymorphisms in the follicle-stimulating hormone receptor (FSHR) gene have impact on several aspects of reproductive processes including folliculogenesis, steroidogenesis, and oocyte maturation. Genome-wide association studies (GWAS) have also documented increased polycystic ovary syndrome (PCOS) susceptibility in relation to FSHR polymorphisms. Thus, our study aimed to investigate the association of FSHR polymorphisms (rs2349415 and rs1169782) in Punjabi women.

Methods: - The current investigation comprised a sample size of 600 participants, consisting of 300 females diagnosed with PCOS and 300 females without any known health conditions serving as healthy controls. After obtaining informed consent, pertinent data and blood samples measuring 5 ml were collected from the subjects. The genotyping of FSHR polymorphisms was conducted using the polymerase chain reaction-restriction fragment length polymorphism (PCR-RFLP) technique. Biochemical analysis has been carried out on both groups. The statistical study was conducted using IBM SPSS version 21 in New York, USA.

Results: - The lipid profile, which includes measurements of cholesterol, triglycerides, HDL, LDL, and VLDL levels, along with anthropometric assessments such as body mass index (BMI) and waist-to-hip ratio (WHR), demonstrated a statistically significant difference between PCOS and control group ($p < 0.05$). Total testosterone levels and luteinizing hormone (LH) were found to be significantly higher in PCOS women ($p < 0.05$), however, FSH levels and LH to FSH ratio remained non-significant. The genotypic analysis of the rs2349415 polymorphism demonstrated a significant increase in the mutant genotype (TT) frequency among women with PCOS ($p = 0.002$). Furthermore, the presence of the mutant genotype was found to be associated with a 1.53-fold increased risk for the onset and progression of the disease. While, the distribution of polymorphism rs11692782 remained non-significant among both the groups. It was also noted that FSHR polymorphisms had also a significant impact on the lipid and hormonal profile, as well.

Conclusions: - The present study concludes that rs2349415 polymorphism plays a substantial role in the occurrence of PCOS in the Punjabi population. Additionally, lipid metabolism and hormone levels are severely influenced by SNPs in the FSHR.

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