

Association of CYP11A1 rs4077582 (C/T) Gene Polymorphism on Ovarian Steroidogenesis in Women with Polycystic Ovary Syndrome

Dr. Priya Alva

KS Hegde Medical Academy, Nitte (Deemed to be University), Mangalore, Karnataka, India

Abstract

One of the most common multifactorial endocrinological disorder is polycystic ovary syndrome (PCOS) that affects 5-10% of reproductive aged females worldwide. CYP11A1 is considered an imperative biomarker which is critical for the activity of enzymes involved in androgen metabolism. Any polymorphism taking place may alter its expression conferring risk for the development of PCOS. The present study aimed to analyze SNP rs4077582 of CYP11A1 with PCOS from South India. The current case control study included 150 PCOS females and 150 aged matched healthy controls. Polymerase chain reaction (PCR) and Sanger sequencing was used for genotypic analysis. Association analysis of anthropometric, biochemical and hormonal parameters with PCOS and controls was done. Mutant carrier allele was the predominant pattern observed for both PCOS and controls. No significant differences were observed in the genotypic and allele frequency distribution of SNP rs4077582 of CYP11A1. The LH levels in the PCOS group differed between the three genotypes for rs4077582, as did the FSH levels in general and recessive genotype models, with high LH and low FSH levels observed in the mutant carrier allele. These findings suggest that the mutant T allele of rs4077582 may be linked to an increased risk of PCOS. The study was the first of its kind in South India on providing baseline data of anthropometric, biochemical and hormonal parameters on PCOS genetics.

Keywords

Polycystic ovary syndrome, CYP11A1, rs4077582, single nucleotide polymorphism.