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THE ROLE OF THE CYP17A1 RS743572 PROMOTER POLYMORPHISM IN THE DEVELOPMENT OF POLYCYSTIC OVARY SYNDROME IN WOMEN OF REPRODUCTIVE AGE

[1]Yuldashev U. K.,
[2]Musakhodjaeva D. A.,
[2]Azizova Z. Sh.
1 Samarkand State Medical University
2 Institute of Immunology and Human Genomics,
Academy of Sciences of the Republic of Uzbekistan

Ovarian hyperandrogenism is a major pathogenetic component of PCOS, a disorder characterized by the coexistence of ovulatory dysfunction, androgen excess, and metabolic disturbances. This phenotype is driven by impaired steroidogenesis in ovarian theca cells and reduced tissue sensitivity to insulin and steroid hormones, while the marked phenotypic heterogeneity of PCOS necessitates clarification of its molecular genetic basis, including variation in steroidogenic genes (Goodarzi et al., 2011; Rosenfield and Ehrmann, 2016).

The CYP17A1 gene at 10q24.32 encodes cytochrome P450 17A1, a 17α hydroxylase 17,20 lyase that catalyzes key steps in androgen biosynthesis via the generation of DHEA and androstenedione. In the ovary, CYP17A1 is predominantly expressed in theca cells and regulates androgen production as estradiol precursors during folliculogenesis and ovulation (Miller, 2005). The rs743572 −34T>C promoter polymorphism may alter transcriptional regulation, including increased gene expression through creation of an Sp1 binding site in vitro, which may be accompanied by enhanced androgen synthesis (Carey et al., 1994).

Accordingly, rs743572 is considered a functionally relevant promoter variant of CYP17A1 that may contribute to hyperandrogenic manifestations and PCOS phenotype formation, supporting its inclusion in genetic profiling panels for women with ovarian hyperandrogenism.

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Objective

To evaluate the association of the CYP17A1 rs743572 −34T>C promoter polymorphism with the presence of PCOS in women of reproductive age in a case control study

Materials and Methods

Women of reproductive age were examined and allocated to a PCOS group n=114 and a control group n=80. PCOS was confirmed using clinical laboratory and ultrasound criteria. Genotyping of the CYP17A1 rs743572 −34T>C polymorphism was performed, followed by calculation of allele and genotype frequencies. Differences in distributions were assessed using the chi square test, and odds ratios with 95% confidence intervals were calculated. Statistical significance was set at $p<0.05$

Results

Analysis of allele and genotype distributions of the CYP17A1 rs743572 −34T>C variant revealed significant differences between women with PCOS and controls. The T allele predominated in controls 89.38% 143/160 and was less frequent in the PCOS group 71.49% 163/228, consistent with a protective association OR 0.30 95% CI 0.17–0.53, $\chi2$ 16.98, $p<0.0001$. In contrast, the C allele was more common in PCOS 28.51% 65/228 than in controls 10.62% 17/160 and was associated with increased odds of PCOS OR 3.35 95% CI 1.88–5.99

Genotype results were concordant with the allele based findings. The T/T genotype was more frequent among controls 81.25% than among women with PCOS 50.0%, supporting a protective effect OR 0.23 95% CI 0.12–0.45, $\chi2$ 18.35, $p<0.0001$. Conversely, the T/C genotype was enriched in the PCOS group 42.98% compared with controls 16.25% and was associated with increased odds of PCOS OR 3.89 95% CI 1.93–7.83, $\chi2$ 14.24, $p<0.0001$. The C/C genotype was uncommon 7.02% in PCOS and 2.5% in controls and did not differ significantly between groups $\chi2$ 1.15, $p=0.284$. Although OR 2.94 95% CI 0.61–14.25 suggested a possible trend toward increased risk, the wide confidence interval including 1.0 did not support a statistically significant association

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Overall, the C allele and the T/C genotype were associated with higher odds of PCOS, whereas the T allele and the T/T genotype showed a protective association in this cohort

Conclusion

The CYP17A1 rs743572 promoter polymorphism was associated with PCOS. The C allele and the T/C genotype were linked to increased odds of the disorder, while the T/T genotype showed a protective pattern. The lack of a significant association for the C/C genotype is likely related to its low frequency and the limited number of carriers. The observed associations are consistent with a role of rs743572 in hyperandrogenism through altered transcriptional activity of CYP17A1 and increased androgen biosynthesis, supporting inclusion of this variant in genetic profiling panels for PCOS