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ASSOCIATION OF THADA, LHCGR, DENND1A, CYP17A1, AND FSHR GENE POLYMORPHISMS IN INFERTILE MALAY FEMALES WITH PCOS

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INTRODUCTION

Polycystic ovary syndrome (PCOS) is the most common endocrinopathy in reproductive-aged women, affecting 5-20% of females. There is a wide heterogeneity and complexity in clinical manifestations and metabolic complications. Therefore, several genes and environmental factors are implicated in the pathogenesis of this disease. Genome-wide association studies (GWAS) identified several susceptible loci with substantial ethnic variations. Our study aimed to investigate the association of *THADA*, *LHCGR*, *DENND1A*, *CYP17A1*, and *FSHR* polymorphisms in Malay patients with PCOS and infertility.

METHODOLOGY

Sixty patients with PCOS and infertility (case group) and 70 infertile females due to other abnormalities (control group) were recruited from infertility clinics at HPUPM, Serdang, Kajang, and Putrajaya hospitals. Clinical data and biochemical results were collected and analysed. High-resolution melting analysis followed by sequencing were used for SNPs (single nucleotide polymorphisms) detection in case and control groups.

RESULT

Weight and the LH, FSH, D21 progesterone, and testosterone levels were significantly elevated in patients with PCOS compared to the control group.

CONCLUSION

The study suggests that SNPs in *DENND1A* gene (rs10818854, rs2479106 and rs10986105), *THADA* (rs13429458), *LHCGR* (rs13405728), *FSHR* (rs6165, rs6166) and *CYP17A1* (rs743572) are associated with PCOS in infertile Malay females.