

CYP19 VNTR POLYMORPHISM A RISK FACTOR IN ENDOMETRIOSIS**Syarif T Hidayat¹, Sultana MH Faradz², NP Noor-Pramono¹**

¹Division Fertility Endocrinology Reproduction, Department of Obstetric and Gynecology. ²Center for Biomedical Research, Faculty of Medicine Diponegoro University, Semarang, Indonesia

Abstract

Endometriosis is defined by the presence of endometrial glands and stroma outside of the uterine cavity. Although the exact aetiology and pathogenesis is unclear, both environmental and genetic factors have been implicated in the disease. The role of genetic factors has been supported by familial and twin studies, however the exact genes that play a role in the susceptibility of development and progression of endometriosis are unknown. Preliminary reports have suggested that specific alleles of a TTTA repeat which is located ~80 nucleotides downstream from intron 4 of the human aromatase gene (*CYP19*) may be associated with differences in oestrogen-dependent disease risk. The objective of this study is to investigate whether polymorphisms of *CYP19* genes are associated with the risk of endometriosis.

We analysed the frequency and distribution of a tetranucleotide (TTTA) tandem repeat polymorphism in intron 4 of the *CYP19* gene. This cross-sectional study was done in Department of Obstetrics and Gynecology, Faculty of Medicine Diponegoro University, Semarang, Indonesia. The patients were classified into two groups defined as the endometriosis ($n = 20$) or normally women group ($n = 20$). The mean (\pm SD) ages were 26.3 ± 8.1 and 24.4 ± 7.7 years, and the mean body mass index (BMI) was 20.9 ± 3.2 and 22.4 ± 3.2 kg/m² respectively. The endometriosis group was classified into four different stages i.e. stages I and II were classified as non-severe, while stages III and IV were classified as severe. Diagnosis of endometriosis confirmed by laparoscopy or laparotomy and histological findings. Controls consisted of 20 Javanese healthy women with no history of gynaecological disease. Interventions given were surgical, laparoscopic, and histological examination. Main Outcome Measure(s) is *CYP19* polymorphism.

The distribution of the TTTA repeat polymorphism of *CYP19* were not significantly different between the groups ($p=0.557$). The *CYP19* VNTR, located in intron 4 (TTTA)₁₀ allele homozygote increased the risk for endometriosis development (relative risks [RR], 2.1; 95% confidence interval [95% CI], 1.51 to 2.95). The results suggest that the (TTTA)₁₀ polymorphism of the *CYP19* gene is weakly associated with the susceptibility of endometriosis in a Javanese population.

Key Words : Endometriosis, polymorphism, *CYP19*