

# Hubungan polimorfisme promotor gen reseptor progesteron pada rs544843047 bagian promotor terhadap penyakit endometriosis di Indonesia = Association between progesterone receptor gene polymorphism rs544843047 in promoter region with endometriosis in Indonesia

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## Abstrak

Endometriosis adalah kelainan ginekologis yang ditandai dengan adanya jaringan endometrium yang tumbuh di luar uterus. Penyakit ini bersifat multifaktorial, salah satunya dipengaruhi genetik. Polimorfisme genetik gen reseptor progesteron (PR) diketahui berhubungan dengan penyakit endometriosis. Penelitian ini bertujuan untuk mengetahui hubungan antara polimorfisme gen PR rs544843047 di bagian promotor dengan endometriosis di Indonesia. Penelitian ini menggunakan desain cross sectional, dengan membandingkan 25 jaringan endometriosis dari wanita penderita endometriosis dan 21 jaringan endometrium dari wanita tanpa endometriosis. Molekul DNA dari kedua jenis jaringan diisolasi, diamplifikasi dengan menggunakan metode PCR. Analisis perubahan nukleotida pada gen PR dilakukan dengan metode sequencing. Hasil penelitian menunjukkan frekuensi genotip dan alel pada SNP gen PR rs544843047 adalah genotip TT 100% dan alel T 100%. Penelitian ini menyimpulkan bahwa tidak ada hubungan antara SNP gen PR pada rs544843047 dengan penyakit endometriosis di Indonesia.

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Endometriosis is a gynecological disorder characterized by the presence of endometrial tissues that grow outside the uterus. This disease is multifactorial cause, one of which is influenced by genetics factor, and genetic polymorphism of the Progesterone Receptor (PR) gene is known to be associated with endometriosis. The aim of this study was to determine the relationship between PR gene polymorphism rs544843047 in the promoter and endometriosis in Indonesia. A cross sectional design was used in this study, comparing 25 endometriosis tissues of women with endometriosis and 21 endometrial tissues of women without endometriosis. DNA molecules from both types of tissues were isolated, then amplified using the PCR method. While analysis of nucleotide changes in the PR gene was conducted by sequencing. The results showed that the genotypic and allele frequencies of the PR rs544843047 SNP were 100% TT genotype and 100% T allele. This research concludes that there are no association between SNP PR gene in rs544843047 and endometriosis in Indonesia.