

CLINICAL SCIENCE

The progens progesterone receptor gene polymorphism is not related to endometriosis-associated infertility or to idiopathic infertility

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OBJECTIVE: This study aimed to determine the frequency of the PROGINS polymorphism in women with endometriosis-associated infertility, in infertile women without endometriosis and in controls.

INTRODUCTION: The human progesterone receptor gene has two isoforms that modulate the biological action of progesterone: isoform A, which is capable of inhibiting the activation of the estrogen receptors, and isoform B, which has the capacity to activate the estrogen receptors. Several polymorphisms have been described for this gene, among which one stands out: a polymorphism named PROGINS, which has been speculated to be related to the genesis of endometriosis by several studies with conflicting results.

METHODS: This was a prospective study that included 148 patients with endometriosis-associated infertility, 50 idiopathic infertile patients and 179 fertile women as controls. The PROGINS polymorphism was studied by PCR.

RESULTS: Genotypes P1P1, P1P2 and P2P2 (P2 representing the PROGINS polymorphism) of the progesterone receptor gene presented frequencies of 93.9%, 5.4% and 0.7%, respectively, in the women with endometriosis-associated infertility ($p=0.2101$, OR = 0.51, 95% CI = 0.24-1.09); 94.4%, 4.2% and 1.4%, respectively, in the patients with minimal/mild endometriosis ($p=0.2725$, OR = 0.53, 95% CI = 0.20-1.43); 93.5%, 6.5% and 0%, respectively, among the patients with moderate/severe endometriosis ($p=0.3679$, OR = 0.49, 95% CI = 0.18-1.31); 86.0%, 14.0% and 0%, respectively, in idiopathic infertile women ($p=0.8146$, OR = 1.10, 95% CI = 0.46-2.63); and 88.3%, 10.6% and 1.1%, respectively, in the control group.

CONCLUSION: The data suggest that PROGINS is not related either to endometriosis-associated infertility or to idiopathic infertility in the population studied.

KEYWORDS: endometriosis; polymorphism; PROGINS; progesterone receptor gene; infertility.

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INTRODUCTION

Endometriosis is a common disease defined as the growth of endometrial tissue outside the uterine cavity and results in a vast array of gynecological problems including dyspareunia, dysmenorrhea, pelvic pain and infertility.^{1,2} Several studies have revealed a large number of genetic markers related to immune, neuroendocrine and reproductive functions present in high frequency among patients with endometriosis, indicating associations between the development of endometriosis and genetic polymorphisms.³⁻⁶

Progesterone is a potent antagonist of estrogen-induced proliferation in the endometrium and may play a pivotal role in the pathogenesis of endometriosis. The human progesterone receptor gene is located at chromosome 11q22-23 and has two isoforms that modulate the biological action of progesterone: isoform A, which is capable of inhibiting the activation of the estrogen receptors, and isoform B, which has the capacity to activate the estrogen receptors.⁷ Several polymorphisms have been described for this gene, among which one stands out: a polymorphism named PROGINS, which arises due to the insertion of an *Alu* element into intron G between exons 7 and 8 of isoform A of the *PR* gene, resulting in an increase of 306 bp in the gene product.⁸

Wieser et al.⁹ studied 95 women with endometriosis and 107 women without endometriosis and concluded that the PROGINS polymorphism is associated with susceptibility to endometriosis. Similarly, Lattuada et al.¹⁰ studied the

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