**CYP2C19** polymorphism increases the risk of endometriosis

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**Abstract**

Purpose Estrogen metabolizing gene mutations can be associated with defective hormonal signaling leading to disease processes. Endometriosis is an estrogen dependent that can be influenced by defective signaling in the estrogen pathway.

Objectives To evaluate the association of A/G 85952 CYP2C19 and A/G 937 HSD17B1 gene polymorphisms with endometriosis through the investigation of a large Brazilian sample of women with endometriosis and a fertile control group.

Methods Five hundred women with endometriosis and 500 women without endometriosis were tested for CYP2C19 and HSD17B1 polymorphisms, by TaqMan Real Time PCR. The results were statistically analyzed by chi-square, logistic regression and tested for Hardy-Weinberg equilibrium.

Results The comparison of genotype and allelic frequency of CYP2C19 polymorphism (rs11592737) in patients with endometriosis and control group showed a statistically significant difference (p=0.0203) and for the HSD17B1 polymorphism (rs605059) differences were not significant (p=0.0687). Comparing the stages I/II and III/IV endometriosis with the control group for the CYP2C19 we observed p=0.0133 and p=0.0564, respectively, and for HSD17B1 the values for p=0.4319 and p=0.0667.

Conclusion We observed that CYP2C19 polymorphism is associated with endometriosis in Brazilian women and can be considered a potential biomarker of the disease.

Keywords Endometriosis · CYP2C19 · HSD17B1 · Infertility · Molecular biomarker

**Introduction**

Endometriosis is a steroid-dependent condition recognized as the most common cause of female infertility [1, 2]. The disease is defined by the presence of endometrial glands and/or stroma outside the uterine cavity.

The estrogen-dependent growth of endometrial tissue is mediated by aromatase, that is the key of local estrogen biosynthesis, promoting the conversion of androstenedione to estrone and from testosterone to estradiol [3].

Genetic polymorphisms in genes associated to the estrogen synthesis pathway, as receptors and metabolizing enzymes of the hormone have been associated to interindividual variation in the levels of circulating estrogen [4].

CYP2C19 is an important gene of cytochrome p450 family and encodes an aromatase associated by the estrogen metabolism, including the conversion of estradiol in estrone and the