Genetic polymorphisms in patients with endometriosis: an analytical study in Goiânia (Central West of Brazil)

K.S.F. Silva¹ and K.K.V.O. Moura²

¹Laboratório de Genética e Biologia Molecular, Universidade Federal de Goiás, Goiânia, GO, Brasil
²Departamento de Biomedicina, Pontifícia Universidade Católica de Goiás, Goiânia, GO, Brasil

Corresponding author: K.S.F. Silva
E-mail: smallbinho@hotmail.com

Received November 25, 2015
Accepted April 15, 2016
Published May 25, 2016
DOI http://dx.doi.org/10.4238/gmr.15028135

ABSTRACT. In healthy women, intra- and extracellular controls prevent the attachment and proliferation of ectopic endometrial cells. During endometriosis, abnormalities in these control mechanisms permit the survival of endometrial cells, their subsequent attachment to the peritoneal cavity, and disease progression. These abnormal cells cause invasion of tissues and induce an inflammatory response. Several genetic, immunological, and environmental factors contribute to this complex process. In this study we examined 6 polymorphisms for 6 different genes (p53; estrogen receptor β; progesterone receptor; GSTM1; GSTT1; CYP1A1). We obtained polymorphic genotype frequencies of all genes for 50 patients and analyzed them using the Fisher exact test or G test. Initially, we analyzed the genes in groups of 2, followed by 3. We found a significant association between polymorphisms in 6 pairs of genes (p53-ERβ showed 5.9-times higher frequency in the experimental group, p53-GSTM1 showed 2.39 times higher, 65.5% patients showed p53-CYP1A1 polymorphism, ERβ-PROGINS showed
3.0-times higher frequency, while 31.25% patients showed GSTM1-PROGINS and GSTT1-CYP1A1 polymorphism. Positive results were found in 15 situations when genes were analyzed in groups of 3; the most significant result corresponded to polymorphisms of p53, ERβ and GSTM1 seen in 20%; PROGINS, ERβ and GSTM1 in 18%; and p53, ERβ and PROGINS in 12% patients. The results indicate that the presence of polymorphisms in more than one endometriosis-related gene is associated with onset of disease and progression. Future studies should focus on these genes to understand their inter-relationships and explore the possibility of developing new diagnostic techniques based on molecular markers.

**Key words:** p53; Estrogen receptor β; Progesterone receptor; GSTM1; GSTT1; CYP1A1