PTPN22 C1858T Polymorphism in Women with Endometriosis

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Problem
Endometriosis has been suggested to be an autoimmune disease and recently, an allelic variation of the *PTPN22* (C1858T) gene was revealed to be associated with the development of autoimmunity. The aim of the study was to determine the frequency of the *PTPN22* (C1858T) polymorphism in Brazilian women with endometriosis as compared with controls.

Method of study
Case–control study included 140 women with endometriosis and a control group consisting of 180 healthy fertile women without a history of endometriosis and/or autoimmune diseases from the ABC School of Medicine. The *PTPN22* (C1858T) polymorphism was studied by restriction fragment length polymorphism polymerase chain reaction (RFLP-PCR).

Results
Genotypes CC, CT and TT of *PTPN22* polymorphism presented frequencies of 67.9, 30.0 and 2.1% in the women with endometriosis ($P = 0.008$); 76.2, 19.0 and 4.8% in women with minimal/mild endometriosis ($P = 0.173$); 61.0, 39.0 and 0.0% in women with moderate/severe endometriosis ($P \leq 0.001$) and 82.8, 16.1 and 1.1% in control group. Allele C and T were present in 82.9 and 17.1%; 85.7 and 14.3%; 80.5 and 19.5%; and 90.8 and 9.2% respectively, in women with endometriosis ($P = 0.004$), women with minimal/mild endometriosis ($P = 0.148$), women with moderate/severe endometriosis ($P = 0.002$) and control group.

Conclusion
The data suggest that in Brazilian women polymorphism *PTPN22* (C1858T) may be an important genetic predisposing factor for endometriosis, especially, in advanced disease.

Introduction

Endometriosis is a common disease, defined as the growth of endometrial tissue outside the uterine cavity that often results in a vast array of gynecologic problems including dyspareunia, dysmenorrhea, pelvic pain, and infertility.[1] Numerous hypotheses have been put forward to explain the presence of ectopic