Reproductive BioMedicine Online
16(3): 416-424

Genetic Alterations of HOXA10 and Their Effect on the Severity of Endometriosis in a Taiwanese Population

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Abstract

Endometriosis is one of the most common gynaecological diseases and evidence has suggested that it may be inherited as a complex genetic trait. HOXA10, a homeobox gene, is expressed in the developing uterus and participates in endometrium development and may contribute to endometriosis. In this study, the HOXA10 gene was analysed in 112 patients with endometriosis and in 54 women without endometriosis, as diagnosed laparoscopically. The entire HOXA10 gene was amplified using polymerase chain reaction followed by single-strand conformation polymorphism analysis and sequencing. Association between the polymorphism and the clinical parameters of endometriosis were examined. There were 7.23% patients with HOXA10 genetic alterations; however, there was no significant increase in the endometriosis patients compared with the controls. Most of these DNA variants were found to be novel mutations that reside within the HOXA10 homeobox domain. Six variants generate amino acid changes in the protein and one harbours a premature stop codon. It was found that patients with HOXA10 polymorphism were associated with a lower serum cancer antigen-125, a lower American Fertility Society score and less severe obliterated cul-de-sac. It is postulated that genetic alterations in the homeobox domain might lead to less specificity for HOXA10 protein binding to a DNA molecule.

Key words：DNA binding; Endometriosis; Homeobox domain; HOXA10; Polymorphism