

POLYMORPHISMS OF ESTROGEN SYNTHESIS AND METABOLISM-RELATED GENES AND THE RISK OF ENDOMETRIOSIS

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Purpose: Endometriosis is associated with genetic factors. The aim of this study was to investigate the relationship between single nucleotide polymorphisms (SNPs) of genes involved in estrogen synthesis and metabolism and the risk of susceptibility to endometriosis.

Materials and methods: Three hundred patients with endometriosis and 337 healthy controls were recruited. Single nucleotide polymorphisms (SNPs) of genes involved in estrogen synthesis and metabolism was determined by using matrix-assisted laser desorption-ionization time-of-flight mass spectrometry (MALDI-TOF MS). A total of 35 non-synonymous SNPs (resulting in a change of amino acid) were studied. Relationship between non-synonymous SNP and the risk of endometriosis was analyzed.

Results: Of 35 non-synonymous SNPs (resulting in a change of amino acid) in estrogen synthesis and metabolism-related genes studied, only a homozygous polymorphism of FSHR gene at position 680 (680Asn/Asn) was demonstrated associated with a significantly increased risk of endometriosis ($P = 0.002$; odds ratio 1.6, 95% CI 1.2-2.3). In addition, the A/A homozygous genotype of polymorphism of FSHR (680Asn/Asn) combined with the G/G homozygous genotype of polymorphism of HSD17B3 (289Gly/Gly) were found related to an increased risk of endometriosis ($P = 0.00002$; odds ratio 2.2, 95% CI 1.5-3.2).

Conclusion: The results of this study suggest that the FSHR gene polymorphism might modulate the risk of endometriosis in Taiwanese Chinese women.