

mir-126 rs4636297 and TGFβRI rs334348 functional gene variants are associated with susceptibility to endometriosis and its severity.

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Abstract

microRNAs (miRNAs) are negative regulators in a variety of cellular processes that occur in endometriosis. Therefore, functional polymorphisms in miRNA and miRNA binding sites may affect gene expression and contribute to susceptibility of endometriosis. In this study, we evaluated the association of two miRNA related polymorphisms, mir-126 rs4636297 and TGFβRI rs334348, with endometriosis risk and its severity. This case-control study was done on 157 endometriosis patients and 252 healthy women as a control group. Tetra amplification refractory mutation system-polymerase chain reaction (tetra-ARMS PCR) was designed to determine the polymorphisms. Our finding showed significant differences in genotype frequency of mir-126 rs4636297 between the groups ($\chi^2 = 6.26$, $p = 0.044$). A significant protection against endometriosis was found for mir-126 rs4636297 in allele (G versus A allele: OR = 0.695, 95% CI = 0.519-0.931, $p = 0.015$) and genotype (GG versus AA genotype: OR = 0.451, 95% CI = 0.233-0.873, $p = 0.018$). Significant association was also observed between the A allele and severity of endometriosis (OR = 0.478, 95% CI = 0.297-0.768, $p = 0.002$). Moreover, we found a significant association between AA genotype with the risk of endometriosis (OR = 0.493, 95% CI = 0.250-0.970, $p = 0.041$) and its severity (OR = 0.240, 95% CI = 0.065-0.883, $p = 0.032$) regarding TGFβRI rs334348 polymorphism. These findings suggest that, for the first time, mir-126 rs4636297 and TGFβRI rs334348 polymorphisms may influence individual's susceptibility to endometriosis and its severity.

KEYWORDS:

Endometriosis; TGF-βRI; mir-126; polymorphism; severity