Association of the estrogen receptor-β gene 
RsaI and AluI polymorphisms with human idiopathic thin endometrium

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ABSTRACT. The aim of this study was to investigate the potential correlation between polymorphisms of the estrogen receptor (ER)-β gene (RsaI and AluI) and ethnic Han Chinese human idiopathic thin endometrium. A total of 120 patients with idiopathic thin endometrium and 120 sterility patients with normal endometrium thickness (controls) were included in the study. RsaI and AluI polymorphisms of the ER-β gene were analyzed with polymerase chain reaction-restriction fragment length polymorphism. The distribution of polymorphisms, genotype, allele, and haplotype was compared between the 2 groups. A significant difference in the RsaI genotype was observed between idiopathic thin endometrium patients and controls. The distribution of the R allele of the RsaI polymorphism in idiopathic thin endometrium patients was 37.1% compared with 48.3% in controls. The odds ratio was 0.630 (95%
confidence interval = 0.438 to 0.907, P = 0.013). No significant difference in the genotype of the AluI polymorphism was found between the two groups. The linkage disequilibrium between Rsal and AluI haploids was not significant in either group, with D’ values of 0.2036 and 0.0685 in the idiopathic thin endometrium patients and controls, respectively. The results of this study suggest a potential role of the ER-β gene polymorphisms in the etiology of idiopathic atrophic endometrium and the R allele as a potential protective factor in ethnic Han Chinese.

**Key words:** Endometrium; Estrogen receptor; Allele; Gene polymorphism