Polymorphism of the estrogen receptor β gene is related to infertility and infertility-associated endometriosis

O polimorfismo do gene do receptor β de estrógeno está relacionado com infertilidade e endometriose associada à infertilidade

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ABSTRACT

Objective: To determine the frequency of the estrogen receptor b gene (*ERβ*) +1730 G/A polymorphism in infertile women with and without endometriosis and controls. **Subjects and methods:** Case-control study that included 136 women with endometriosis, 69 women without endometriosis and 209 fertile women as controls. The *ERβ* gene + 1730 G/A polymorphism was identified by RFLP-PCR (Restriction Fragment Length Polymorphism – Polymerase Chain Reaction). **Results:** Genotypes GG, GA and AA of the *ERβ* gene presented frequencies of 60.3%, 38.2% and 1.5%, respectively, in the women with endometriosis ($p < 0.0022$). Of the infertile women without endometriosis, 63.8% presented the normal homozygous genotype GG, 30.4% the GA heterozygous genotype, and 5.8% the homozygous mutated genotype AA ($p < 0.0275$). In the control group, 77.5% presented the normal homozygous genotype GG, 21.1% the heterozygous genotype GA, and 1.4% the homozygous mutated genotype AA. **Conclusion:** The data suggest that the estrogen receptor β gene (*ERβ*) +1730 G/A polymorphism can be associated with risk of infertility and endometriosis-associated infertility. Arq Bras Endocrinol Metab. 2010;54(6):567-71

Keywords

Endometriosis; estrogen receptor β; estrogen; infertility; polymorphism

RESUMO

Objetivo: Determinar a frequência do polimorfismo +1730 G/A do gene do receptor beta de estrógeno (*ERβ*) em mulheres inférteis com e sem endometriose e controles. **Sujeitos e métodos:** Estudo caso-controle que incluiu 136 mulheres com endometriose, 69 mulheres sem endometriose e 209 mulheres férteis como controles. O polimorfismo *ERβ* + 1730 G/A foi identificado por RFLP-PCR (Restriction Fragment Length Polymorphism - Polymerase Chain Reaction). **Resultados:** Os genótipos GG, GA e AA do polimorfismo *ERβ* + 1730 G/A apresentaram frequência de 60,3%, 38,2% e 1,5%, respectivamente, nas mulheres com endometriose ($p = 0,0022$). Das mulheres inférteis sem endometriose, 63,8% apresentaram o genótipo homozigoto normal GG, 30,4% o genótipo heterozigoto GA e 5,8% o genótipo homozigoto mutado AA ($p = 0,0275$). No grupo controle, os genótipos GG, GA e AA apresentaram frequência de 77,5%, 21,1% e 1,4%. **Conclusão:** Os dados sugerem que o polimorfismo *ERβ* +1730G/ pode estar associado ao risco de infertilidade e infertilidade associada à endometriose. Arq Bras Endocrinol Metab. 2010;54(6):567-71

Descritores

Endometriose; receptor β de estrógeno; estrógeno; infertilidade; polimorfismo

INTRODUCTION

Endometriosis is a steroid-dependent condition in which a tissue that is histologically similar to the endometrium with glands and stroma grows outside the uterine cavity and becomes implanted in tissues and organs such as the Fallopian tubes, ovaries, peritoneum,