

عنوان مقاله:

Investigation of the FSHR, CYP11, and INSR Mutations and Polymorphisms in Iranian Infertile Women with Polycystic (Ovary Syndrome (PCOS

محل انتشار:

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خلاصه مقاله:

Introduction: polycystic ovary syndrome (PCOS) is the most common cause of ovarian dysfunction associated with infertility, Oligomenorrhea or amenorrhea, hirsutism, acne, and obesity. A large body of evidence unraveled, three major groups of genes play critical roles in underlying PCOS molecular mechanism. The aim of this study is to investigate critical exonic variant of FSHR, CYP11, and INSR and determine the functionality of these mutations in Iranian patients with PCOS. **Materials and methods:** In this case-control study, 130 patients with PCOS who referred to the Vali-e-Asr Hospital with infertility were included. 3 ml peripheral blood was taken from the participants for DNA extraction. PCR was conducted for each gene and the PCR product was genotyped by sequencing. **Results:** The data showed that there were two polymorphisms in INSR genes which did not change the protein sequences; these alterations can also be considered as a single nucleotide polymorphism (SNP). Moreover, any exonic variant has not been detected in CYP11B1. Whereas, two missense mutation have been detected in FSHR gene including p.Ala307Thr and p. Asn680Ser. It has been shown that the polymorphisms of the FSHR gene affect the hormone response in the ovaries. Our data demonstrated that the FSHR mutations frequencies were higher in the patients with PCOS rather than control people (without any infertility complication) significantly. **Conclusion:** Altogether, our data showed that the polymorphisms of FSHR were significantly associated with PCOS in Iranian infertile women. Further studies with larger sample sizes are needed to be performed in order to explore the strength of the association

کلمات کلیدی:

CYP11, FSHR, Infertile, INSR, PCOS, Polymorphisms

