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DISTRIBUTION CHARACTERISTICS OF ALLELES AND GENOTYPES OF THE PPAR α rs4253778 POLYMORPHISM IN GROUPS ASSOCIATED WITH INFERTILITY

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Abstract

The etiology of infertility is multifactorial, and genetic factors, including polymorphisms of genes involved in metabolic and hormonal processes, play an important role in determining individual characteristics of reproductive function. The PPAR α gene is involved in the regulation of fatty acid metabolism and energy homeostasis, and its rs4253778 polymorphism is of particular interest in assessing genetic characteristics associated with reproductive health.

Aim of the study

To determine the frequencies of alleles and genotypes of the PPAR α rs4253778 polymorphism in patients presenting with infertility and to comparatively assess them against the corresponding parameters of the control group.

Study population

A total of 217 women were included in the study. The main group consisted of 110 patients, while the control group included 107 women. The main group was further divided according to the effectiveness of assisted reproductive technologies into two subgroups: IVF/ICSI (+) — 59 women and IVF/ICSI (–) — 51 women. Genotyping of the PPAR α rs4253778 polymorphism was performed in all participants.



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Results

Analysis of the overall distribution of the PPAR α rs4253778 polymorphism showed that the G allele was detected at a frequency of 78.64% and the C allele at 21.36% in the main group. In the control group, the corresponding frequencies were 78.04% and 21.96%, respectively.

The genotype distribution in the main group was as follows: G/G — 61.82%, G/C — 33.64%, and C/C — 4.55%. In the control group, the frequencies were 61.68% for G/G, 32.71% for G/C, and 5.61% for C/C.

No statistically significant difference was found in the frequency of the G allele between the main and control groups ($\chi^2=0.0$; $p=0.85$; RR=1.0; 95% CI: 0.65–1.57; OR=1.0; 95% CI: 0.66–1.64). Similarly, no statistically significant difference was observed for the C allele ($\chi^2=0.0$; $p=0.85$; RR=1.0; 95% CI: 0.63–1.56; OR=1.0; 95% CI: 0.61–1.52).

Comparative analysis of genotype frequencies also revealed no statistically significant differences. For the G/G genotype, $\chi^2=0.0$; $p=0.99$; RR=1.0; 95% CI: 0.59–1.70; OR=1.0; 95% CI: 0.58–1.74. For the G/C genotype, $\chi^2=0.0$; $p=0.84$; RR=1.0; 95% CI: 0.60–1.77; OR=1.0; 95% CI: 0.59–1.84. No statistically significant difference was identified for the C/C genotype either ($\chi^2=0.1$; $p=0.85$; RR=0.8; 95% CI: 0.22–2.96; OR=0.8; 95% CI: 0.24–2.70).

Conclusion

A predominance of the G allele and G/G genotype of the PPAR α rs4253778 polymorphism was identified in both the main and control groups. However, the differences in allele and genotype frequencies between the main and control groups were not statistically significant. The obtained findings do not confirm a direct association between the PPAR α rs4253778 polymorphism and the presence of infertility in the studied groups.