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DISTRIBUTION CHARACTERISTICS AND COMPARATIVE ANALYSIS OF ALLELES AND GENOTYPES OF THE PPAR α rs4253778 GENE POLYMORPHISM

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Abstract

The course of reproductive processes and the effectiveness of assisted reproductive technologies may be influenced not only by clinical and hormonal factors but also by genetic characteristics. The PPAR α gene is involved in the regulation of lipid metabolism and energy homeostasis; therefore, studying the distribution of alleles and genotypes of its rs4253778 polymorphism is of particular importance for identifying genetic factors associated with reproductive health.

Aim of the study

To determine the distribution characteristics of alleles and genotypes of the rs4253778 polymorphism of the PPAR α gene in women presenting with infertility and undergoing assisted reproductive technologies, and to comparatively assess their distribution according to IVF/ICSI outcomes.

Study population

A total of 217 women were included in the study. The main group consisted of 110 women with infertility, while the control group included 107 women. According to IVF/ICSI outcomes, the main group was divided into two subgroups: IVF/ICSI (+) — 59 women and IVF/ICSI (–) — 51 women. Allele



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and genotype frequencies of the PPAR α rs4253778 polymorphism were analyzed in all examined women.

Results

Analysis of allele frequencies 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 frequencies were 78.04% for the G allele and 21.96% for the C allele. Thus, a clear predominance of the G allele was observed in both groups.

In the main group, the distribution of genotypes was as follows: G/G — 61.82%, G/C — 33.64%, and C/C — 4.55%. In the control group, the corresponding frequencies were 61.68%, 32.71%, and 5.61%, respectively. Thus, genotype frequencies were almost identical in the main and control groups, with the G/G genotype being the most prevalent genotype.

No statistically significant difference was found between the main and control groups in the frequency of the G allele ($\chi^2=0.0$; $p=0.85$; 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 genotypes also revealed no statistically significant differences between the groups. 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 found 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).

When the subgroups were analyzed according to IVF/ICSI outcomes, the frequency of the G allele in the IVF/ICSI (+) group was 83.9%, while that of the



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C allele was 16.1%. In the IVF/ICSI (–) group, the G and C alleles were detected at frequencies of 72.55% and 27.45%, respectively. In the control group, the corresponding frequencies were 78.04% and 21.96%.

Regarding genotype distribution, the IVF/ICSI (+) group had G/G, G/C, and C/C genotype frequencies of 71.19%, 25.42%, and 3.39%, respectively. In the IVF/ICSI (–) group, the corresponding frequencies were 50.98%, 43.14%, and 5.88%. In the control group, G/G, G/C, and C/C genotypes were recorded at frequencies of 61.68%, 32.71%, and 5.61%, respectively.

Comparative analysis between the IVF/ICSI (+) and control groups showed no statistically significant difference in the frequency of the G allele ($\chi^2=1.6$; $p=0.49$; $RR=1.1$; 95% CI: 0.48–2.40; $OR=1.5$; 95% CI: 0.82–2.63). No statistically significant difference was observed for the C allele either ($\chi^2=1.6$; $p=0.49$; $RR=0.9$; 95% CI: 0.65–1.32; $OR=0.7$; 95% CI: 0.38–1.23).

The frequency of the G/G genotype in the IVF/ICSI (+) group was 71.19%, which was higher than the 50.98% observed in the IVF/ICSI (–) group. Conversely, the G/C genotype was more frequent in the IVF/ICSI (–) group (43.14%) than in the IVF/ICSI (+) group (25.42%). However, the differences in genotype frequencies between the IVF/ICSI (+) and control groups were not statistically significant: $p=0.47$ for G/G, $p=0.56$ for G/C, and $p=0.73$ for C/C.

The findings demonstrated that the overall allele and genotype distributions of the PPAR α rs4253778 polymorphism did not differ significantly between the main and control groups. At the same time, a relatively higher frequency of the G allele and G/G genotype was observed among women with successful IVF/ICSI outcomes. Since this tendency was not statistically confirmed, further studies are required to assess its potential role as an independent prognostic factor.



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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. The G allele accounted for 78.64% in the main group and 78.04% in the control group, while the G/G genotype was recorded at frequencies of 61.82% and 61.68%, respectively.

No statistically significant differences in allele or genotype frequencies were identified between the main and control groups. The p-values were 0.85 for the G allele, 0.85 for the C allele, 0.99 for the G/G genotype, 0.84 for the G/C genotype, and 0.85 for the C/C genotype.

Analysis according to IVF/ICSI outcomes showed a relatively higher frequency of the G allele and G/G genotype in the IVF/ICSI (+) group. However, because the differences between the IVF/ICSI (+) and control groups were not statistically significant, the independent prognostic significance of the PPAR α rs4253778 polymorphism was not confirmed. Nevertheless, the observed tendencies indicate the need for further investigation into the potential association of this polymorphism with reproductive outcomes.