The aim of this study was to analyze the presence of MTHFR (methylene tetrahydrofolate reductase) mutations and their correlation with infertility.

We investigated forty-four infertile women (aged 27 to 41 years) diagnosed with PCOS according to Rotterdam criteria; n=8 of the study women were with primary infertility (PI) and n=36 of them were women with a history of recurrent miscarriages (RM). We determined two most frequent mutations in the MTHFR gene: A1298C/C677T and we established serum homocysteine concentration. The main outcomes measures: clinical pregnancy rate in the presence of MTHFR mutation. Moreover homocysteine concentration was correlated with the presence of MTHFR mutation and pregnancy outcome in the study groups.

The presence of the mutation in the MTHFR gene was confirmed in 75% patients with PI vs. 83% patients with RM. In the group with RM we observed more frequent incidence C677T gene mutation (60% vs 34%) and higher homocysteine level (8 vs 6 µmol/L) compared to PI. We found no statistical differences between the presence of MTHFR mutation and the pregnancy outcome, p<0.05. Moreover, we found no statistically significant differences in the levels of homocysteine concentration for particular MTHFR mutations, p<0.05.

It seems that the presence of the mutation in MTHFR gene is not a prognostic factor for pregnancy outcome. However, a more frequent presence of the mutation in the study group in comparison to the general population suggests its possible relationship with infertility and recurrent miscarriages. Moreover, the presence of MTHFR mutation does not influence the level of serum homocystine concentration.