

## **ASSOCIATION BETWEEN MTHFR MUTATION AND** ANTIPHOSPHOLIPID ANTIBODIES WITH POOR OVARIAN RESERVE

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## INTRODUCTION

**Introduction:** The expression of the 5,10-methylentetrahydrofolate reductase (MTHFR) gene in human oocytes and

preimplantation embryos suggests that the MTHFR gene is involved in folliculogenesis and female reproduction (Shahrokhi

et al., 2017) and the MTHFR gene variants are associated with lower ovarian reserve, diminished response to follicular

stimulation and a reduced chance of live birth rate after IVF (Laanpere et al., 2010). Objectives: Our study evaluates the

incidence of MTHFR mutations (C677T and A1298C) and anti-phosphatidylserine (aPS) and anti-phosphatidylethanolamine

(aPE) of infertile patients with an Anti-Müllerian Hormone (AMH) less than 0.8 ng/ml.

Material and Methods: This is a prospective observational cohort study of infertile patients undergoing infertility investigation in the period of January 2019 to March 2020. A total of 98 patients with an AMH level under 0.8 ng/ml were included in the study, with less than 38 years of age. However, 50 patients had all the work out exams complete to be included in the study.



In the study group (AMH less than 0.8), 37 patients out of 50 had a level of anti-phosphatidylserine greater than 20.0

(IgG, M, A), which gives an incidence of 75% of aPS/aPE in this group of patients. Fourty six out of the 50 patients

included in the study tested for mutations in the MTHFR and 27 had it positive, which gives an incidence of 59%.

### **CONCLUSION**

We observed a high incidence of mutation in the MTHFR enzime in patients with very low AMH (less than 0.8 ng/ml) under

38 years of age. We also observed a high incidence of acquired thrombophilya (aPS/aPE) in this group of patients.

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