

# Abstract

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## **MTHFR C677T polymorphism associates with unexplained infertile male factors.**

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**PURPOSE:** To determine whether 5,10-methylenetetrahydrofolate reductase (MTHFR C677T and A1298C) genotype is associated with male infertility.

**METHODS:** Analysis of cytogenetic, Y chromosomal microdeletion assay (Yq), and the C677T and A1298C polymorphisms of the MTHFR gene by pyrosequencing and PCR-Restriction Fragment Length Polymorphism (RFLP) method. SAS 8.1 assessed the statistical risk of MTHFR genotype.

**RESULTS:** The homozygous (T/T) C677T polymorphism of the MTHFR gene was present at a statistically high significance in unexplained infertile men with normal karyotype, instead at no significance in explained infertile men with chromosomal abnormality or Y chromosome deletion. There was no statistically significance of A1298C variation in infertile males.

**CONCLUSIONS:** The MTHFR 677TT genotype may be a genetic risk factor for male infertility, especially with severe OAT and non-obstructive azoospermia in unexplained infertile males.

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