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Prospective study of *MTHFR* genetic polymorphisms as a possible etiology of male infertility

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ABSTRACT. The aim of this study was to explore the relationship between 2 genetic polymorphisms of the methylenetetrahydrofolate reductase gene (*MTHFR*), C677T and A1298C, and determine the long-term reproductive outcome in infertile men. This was a prospective study conducted in an andrology clinic. Men with a 1-year history of infertility were assessed for the *MTHFR* polymorphisms at a 5-year follow-up. We compared the *MTHFR* C677T and A1298C polymorphisms by polymerase chain reaction-restriction fragment length polymorphism between men who did and did not bear children during follow-up. Of the 215 men who were infertile at 1 year, 82 (38.1%) remained infertile and 133 (61.9%) achieved natural conception during the 5-year follow-up, with the highest rate in the first year (32.6%). The *MTHFR* 677TT genotype (homozygote) was associated with a substantially increased risk of infertility during follow-up [odds

ratio (OR) = 10.242; 95% confidence interval (CI) = 1.257-83.464] relative to the *MTHFR* 677CC genotype (wild-type). Risk of infertility was not increased by the *MTHFR* A1298C polymorphism alone, but was increased by the combination of polymorphisms *MTHFR* C677T and *MTHFR* A1298C (OR = 11.818; 95%CI = 1.415-98.674). The homozygous *MTHFR* C677T genotype was a risk factor for male infertility during 5-year follow-up, whereas a correlation between *MTHFR* A1298C and infertility was not observed. The *MTHFR* C677T and *MTHFR* A1298C polymorphisms had additive effects on male infertility.

Key words: Gene polymorphism; Male infertility; Prospective study; Methylenetetrahydrofolate reductase

