

The MTHFR C677T Polymorphism Screening: A Challenge in Recurrent Pregnancy Loss

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Abstract : Introduction: Recurrent pregnancy loss (RPL) defined as two or more pregnancy losses, is a serious clinical problem. Methylenetetrahydrofolate reductase (MTHFR) polymorphisms, commonly the variant C677T is recognized as an inherited thrombophilia which might affect embryonic development and pregnancy success and cause pregnancy complications as RPL. Material and Methods DNA was extracted from peripheral blood samples and PCR-RFLP was performed for the molecular diagnosis of the C677T MTHFR polymorphism among 70 patients (35 couples) with more than 2 fetal losses. Aims and Objective: The aim of this study is to determine the frequency of MTHFR C677T among Tunisian couples with RPL and to critically analyze the available literature on the importance of MTHFR polymorphism testing in the management of RPL. Result and comments: No C677T mutation was detected in the carriers of RPL. This result would be related to sample size and to different criteria (number of abortion), - The association between MTHFR polymorphisms and pregnancy complications has been reported but with controversial results. - A lack of evidence for MTHFR polymorphism testing previously recommended by ACMG (American College of Medical medicine). Our study highlights the importance of screening of MTHFR polymorphism since the real impact of such thrombotic molecular defect on the pregnancy outcome is evident. - Folic supplementation of these patients during pregnancy can prevent such complications and lead to a successful pregnancy outcome.

Keywords : methylenetetrahydrofolate reductase, C677T, recurrent pregnancy loss, genetic testing

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