

## Thrombophilic Mutations in Tunisian Patients with Recurrent Pregnancy Loss

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**Abstract :** Pregnancy is a hypercoagulable state which causing a defective maternal haemostatic response and leading to thrombosis of the uteroplacental vasculature, that might cause pregnancy complications as recurrent pregnancy loss (RPL). Since heritable Thrombophilic defects are associated with increased thrombosis, their prevalence was evaluated in patients with special emphasis on combinations of the above pathologies. Especially, Factor V Leiden (FVL) G1691A, methylene tetra hydro folate reductase (MTHFR) C677T, and factor II (FII) G20210A mutations are three important causes of thrombophilia, which might be related to recurrent pregnancy loss (RPL). In this study we evaluated the presence of these three mutations [factor V Leiden (FVL), prothrombin G20210A (PTG) and methylenetetrahydrofolate reductase (MTHFR) C677T] amongst 35 Tunisian women with more than 2 miscarriages, referred to our genetic counseling. DNA was extracted from peripheral blood samples and PCR-RFLP was performed for the molecular diagnosis of each mutation. Factor V Leiden and Prothrombin mutation were detected respectively in 5.7% and 2.9% of women with particular history of early fetal loss and thrombotic events. Despite the lack of strength of this study, we insist that testing for the most inherited thrombophilia (FVL and FII mutation) should be performed in women with RPL in the context of thrombotic events. Multi-centre collaboration is necessary to clarify the real impact of thrombotic molecular defects on the pregnancy outcome, to ascertain the effect of thrombophilia on recurrent pregnancy loss and then to evaluate the appropriate therapeutic approach.

**Keywords :** thrombophilia, recurrent pregnancy loss, factor V Leiden, prothrombin G20210A, methylene tetra hydro folate reductase

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