The actual role of the mutations associated with hereditary thrombophilia in recurrent pregnancy loss.

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Context: Single nucleotide polymorphisms of the genes coding for coagulation factors are cause of congenital thrombophilia which might lead to recurrent miscarriages and fetal loss in advanced pregnancy. The most frequent reasons of thrombophilia are the following: factor V Leiden (FLM), mutation G2021A of prothrombin gene (PT), and C667T of 5,10-methylenetetrahydrofolate reductase gene (MTHFR).

Objective: The aim of this study was to estimate the prevalence of FLM and M385T mutation, PT gene G2021A and A19911G mutations, C677T and A1298C MTHFR gene mutations among women with unexplained recurrent pregnancy loss (RPL).

Methods and Patients: a group of 136 women was analyzed, who experienced at least 2 unexplained RPL. Other possible causes of miscarriage were excluded. 106 healthy women having at least two uncomplicated pregnancies and delivered healthy children constituted a control group. Each patient were examined for 6 polymorphisms. Genome DNA isolation from lymphocyte was performed with commercial assay BloodMini firmy A&A Biotechnology Gdynia. Each polymorphism was genotyped with use of Taq-Man Genotyping Assay and thermal cycler

MOM: Results may suggest protective role of C667T MTHFR gene polymorphism against RPL.

Results: Prevalence of FLM was higher among patients with RPL compared to control group (5.9% vs 3.8%, p=0.55). Prevalence of following mutations: M385T factor V polymorphism, mutations G20210A i A19911G PT gene and A1298C MTHFR gene mutation did not differ between groups. The frequency of heterozygote 667TT MTHFR gene careers mutation was higher in control group compared to study group (15.1% vs 6.6%, p= 0.03).

Conclusion: In patients who experienced intrauterine fetal demise or unexplained RPL diagnostic approach should be individualized and inherited thrombophilia screening should be considered, regardless of negative history of thrombosis.

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