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TITLE

MULTIPLE THROMBOPHILIA GENE DEFECTS IN PATIENTS WITH RECURRENT PREGNANCY LOSS

AUTHOR/S

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ABSTRACT

Context. In the structure of miscarriage recurrent pregnancy loss (RPL) takes about 5%. Recently, RPL was considered as a typical multifactorial disease that is a result of combined expression of functionally weakened variants of many genes in the background of adverse external and internal factors.

Objective: to determine the frequency and to establish the role of combinations allelic variants of thrombophilia and endothelial dysfunction genes in the development of RPL.

Methods. This prospective, case-control study looked at the association between multiple hereditary thrombophilia gene defects and RPL. With PCR were detected genetic polymorphisms of coagulation factors and fibrinolysis (1691 G?A F5 Leiden, 20210 G?A prothrombin gene, -675 5G/4G PAI-1, 455 G?A fibrinogen ?), endothelial dysfunction (677 C?T MTHFR). Statistical analysis was performed.

Patient(s): 109 pregnant women with RPL and 34 healthy pregnant women in 1st trimester of pregnancy (control group) were examined and tested for the presence of genes polymorphisms.

Main Outcome Measure(s): abnormal genes polymorphisms that potentiate effect of each other through the same mechanisms or combinations, which take part in different pathogenesis mechanisms of miscarriage, significantly increase the risk of RPL occurrence.

Result(s): The simultaneous existence of two or more pathologic polymorphisms was identified in 80.7% with RPL vs. in 23.5% in control group, $p < 0.001$ (OR=13.6, 95% CI 5.4-34.3), three pathological polymorphisms increases chance of RPL development in 6.36-fold ($p < 0.05$, 95% CI 1.4-28.2), four - in 13.05 times ($p < 0.01$, 95% CI 0.76-223.2).

Conclusions: multiple thrombophilia gene defects (two or more) were diagnosed in 80.7% women with RPL. It was found that combination of abnormal polymorphisms in -675 5G/4G PAI-1 and 455 G ?A fibrinogen ? genes were more frequent in group with RPL.

INSTITUTE

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