4. GENETIC ASPECTS OF THROMBOPHILIA IN PREGNANCY

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Introduction. Thrombophilia represents a condition that increases blood coagulability and the risk of clots. Physiological changes during pregnancy, such as excess fibrinogen synthesis and increased concentration of coagulation factors in the third trimester of pregnancy, can lead to thrombotic phenomena. The thrombophilia impact is intensified in pregnant women with specific variations in the genes responsible for coagulation.

Aim of study. Evaluation of the genetic aspects of thrombophilia in obstetric complications.

Methods and materials. A literature review was done, the search motors being PubMed, Google Scholar, ScienceDirect. Out of the 250 articles submitted, 89 met the research criteria.

Results. An important number of genetic variations with thrombophilia potential in pregnant women have been reported and correlated with: premature birth, intrauterine death of the fetus, premature detachment of the placenta, eclampsia, etc. There have been two groups of hereditary thrombophilia described: through hypomorphic mutations, for example antithrombin III deficiency, protein C and S deficiency; through hypermorphic mutations - factor V Leiden mutation, G20210A mutation in the prothrombin gene. Also, the involvement of the PAI-1 gene and more recently the MTHFR gene has been proven. The most common mutations (heterozygous F5 gene mutation G1691A and prothrombin gene mutation G20210A) are associated with a moderate to low risk of developing thrombophilia, while the rarest types like homozygous F5 and F2 gene mutations, antithrombin III, protein C or protein S deficiency, have a more considerable and higher risk.

Conclusion. Up to day the causes of pregnancy complications were attributed to various pathologies, infections or considered an incidence. Today, though, the evaluation of genetic aspects is required, as it can influence the evolution of the pregnancy too.