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# Personalized treatment selection in a patient with symptomatic hereditary thrombophilia and hyperhomocysteinemia

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**Abstract.** The article illustrates the clinical observation of a patient with recurrent thrombosis of the lower extremities and hyperhomocysteinemia, has been selected personalized therapy based on the results of molecular genetic surveys.

**Key words:** personalized treatment, thrombophilia, hyperhomocysteinemia, molecular diagnostics.

Introduction. The research of genes' polymorphisms of cardiovascular diseases' susceptibility gets great practical meaning in modern stage of molecular medicine development. Last decade was marked by significant growth of research, dedicated to hyperhomocysteinemia (HHC) as one of the reasons which causes the development of thrombophilia [1, 2]. Homocysteine (HC) has an evident toxic effect that is developed, first of all, in violation of endothelial function, that's why the increasing of HC level in blood has evident atherogenic and thrombophilic effects. The last ones are caused by toxic HC metabolites which damage vessels' endothelium, baring subendothelial matrix and smooth muscle cells, that stimulates thrombocyte aggregation and thrombogenesis [3].

Objective: The illustration of possibility of personalized pathogenetic therapy selection in a patient with hereditary thrombophilia and HHC, according to the results of biochemical and molecular-genetic analysis.

Materials and methods. The clinical observation of 40-years-old patient P. was analyzed, who has been in treatment at GI «IOUS named after V.T.Zaitsev of NAMSU» with a diagnose: acute reccurent deep vein thrombosis (DVT) of right lower extremity.

Results. The patient arrived to the clinic with implications of acute recurrent DVT of right lower extremity, proved by the results of ultrasound duplex phleboangioscanning of lower extremities with conducting of compression tests. Genealogical analysis detected the burdeness of genealogical cardiovascular pathology from maternal side. Laboratory clinical blood test, urine test and general biochemical tests didn't reveal peculiarities. At conducting of additional biochemical blood test, the increasing of HC was revealed to the level of 40,79 mcmol/l (while the norm is 5,46-16,2) mcmol/l.

Taking into account the recurrence of acute DVT of right lower extremity in a

patient with HHC and burdeness of genealogical cardiovascular pathology, the patient was advised to have genes' molecular examination, associated with thrombophilia development. At molecular examination of venous blood samples by PCR method, it was detected that the patient has homozygote compound polymorphisms of G20210A A/A gene F2-prothrombine and A222V (677 C/T) gene methylentetrahydrofolate reductase (MTHFR), as well as heterozygote carriage of Leyden mutation (F5 G1691A gene).

Conclusion. It is known that in normal state, a carrier of Leyden mutation may not have thrombosisoses. Thrombosisoses are developed at the presence of risk factors [4]: HHC, MTHFR and prothombine gene mutation, as in the case of our patient. Variant A of G20210A polymorphism leads to increased gene expression and is a risk marker of thrombosisoses and cardiac infarction development. MTHFR ferment catalyses 5,10- methylentetrahydrofolate recovery into в 5- methylentetrahydrofolate, which is an active form of folic acid, that is necessary for methionine formation from HC and further – S-adenosyl methionine, which plays the key role in DNA methylation process. The presence of thrombophilia which is peculiar to forms, associated with homozygosis by MTHFR polymorphism, 677 C/T, needs an individual correction and basic therapy.

Apart from standard antithrombotic therapy, it is compulsory for patients with symptomatic HHC to keep the special diet with the consumption restriction of products, enriched with methionine, and the inclusion of products, enriched with cofactors of folate cycle (folic acid, Vitamin B6 and B12, betaine).

Taking into account the detected changes, the patient was prescribed above mentioned diet, wearing of elastic compression stockings, anticoagulant therapy and venotonics. As it was shown in further supervision in dynamics, performed therapy led to normalization of HC level in blood, that may be considered as a hope for fixed remission in a patient with HHC and heridatory form of thrombophilia.

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