# 10. DEEP VENOUS THROMBOSIS COMPLICATED WITH ACUTE PULMONARY EMBOLISM, A CASE OF A PATIENT WITH PROVEN THROMBOPHILIA

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**Introduction.** Pulmonary thromboembolism is the clinical condition that is the consequence of thrombus embolic obstruction of the pulmonary arteries or its branches, representing a major cardiovascular emergency. Venous thromboembolism (VTE) presents clinically as deep vein thrombosis (DVT) or pulmonary embolism (PE). According to the literature, pulmonary embolism ranks third among acute cardiovascular syndromes, after myocardial infarction and stroke.

**Case statement.** We present the case of a patient hospitalized with complaints of marked dyspnea at rest, acrocyanosis, fatigue. Hemodynamically detected sinus tachycardia (FCC 102 c/min) decreased oxygen saturation (SaO2 85%), BP 130/80 mmHg. High risk was determined when assessing the WELLS score. Laboratory analyzes determined D-Dimers >8.27 mg/l. At echocardiography (echo-cg), dilation of the right parts of the heart is found. In the cavity of the right atrium, hypermobile inhomogeneous formations of large sizes were visualized, with a protrusion in the right ventricle. Venous doppler evaluation detected thrombosis of the popliteal vein on the right. PESI prediction score – intermediate risk. Angio CT reported thrombi in both pulmonary arteries with dimensions of 3.4 cm on the right and 2.0 cm on the left respectively. The evolution of the disease with the presence of thrombi with different localization suggested the determination of genetic polymorphisms where the presence of mutations in the genes responsible for hereditary thrombophilia was confirmed. Treatment with fondaparinux and oral anticoagulant was instituted, with target INR values maintained at 2-3. After the establishment of the diagnosis and the institution of effective treatment, the improvement of the clinical condition was noted, the complete resorption of thrombi from the heart and the lower limb. Echo-CG revealed the return to normal of the right sides of the heart. Complete resolution of thrombi in the pulmonary arteries was demonstrated at CT angio after five months.

**Discussions.** PE is a serious manifestation of VTE with a 90-day mortality rate of approximately 15-20%. One of the common causes of VTE is thrombophilia, which can be of three types: hereditary, acquired, and mixed. Several hereditary mutations/polymorphisms affecting genes encoding factors involved in hemostasis are described. In the patient presented with VTE, we determined four genetic mutations: F2/Prothrombin (coagulation factor II), FGB/Fibrinogen (coagulation factor I), ITGA2–O2/integrin (platelet receptor for collagen), PAI–1/Serpin (tissue plasminogen activator antagonist).

**Conclusion.** Considering the case of the patient in whom venous thrombosis with various localization was demonstrated, we suspected a thrombophilia, later demonstrated by genetic polymorphism analysis and genetic mutations were diagnosed. Consequently, the decision was made for long-term treatment with oral anticoagulants.