Case Report

Right Ventricular Thrombus in a 36-Year-Old Man with Factor V Leiden

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Abstract

Factor V Leiden deficiency is the most common hereditary hypercoagulable disease in the United States and involves 5% of the Caucasian population. Up to 30% of patients who present with deep vein thrombosis (DVT) or pulmonary thromboembolism present with this condition. This is a case report of a 36-year-old man who experienced one episode of DVT within the previous year and was admitted to our hospital due to productive coughs and hemoptysis. Paraclinical studies demonstrated a right ventricular thrombus. Additional investigation was done to find the underlying cause. Laboratory tests were positive for Factor V Leiden mutation. Other factors for hypercoagulability states were normal. Given that Factor V Leiden mutation is a life-threatening condition with a relatively high prevalence and considering its thrombogenesis, screening tests are necessary in young patients without obvious reasons for recurrent thrombus formation. It seems that medical noninvasive treatments can be an alternative therapy to surgery when a ventricular thrombus is suspected in these patients.

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Introduction

Cardiac masses are not rare, with their differential diagnoses comprising tumors, thrombi, and inflammatory causes.1 Ventricular thrombi have been reported in patients with the antiphospholipid syndrome2 and other types of hypercoagulable states. They can be a complication of myocardial infarction and deep vein thrombosis (DVT).3

Factor V Leiden deficiency is the most common hereditary hypercoagulable disease in the United States and involves 5% of the Caucasian population.4 Factor V Leiden leads to activated protein C resistance, and it has been demonstrated as a risk factor for venous thrombosis development.5 Approximately one out of 1000 patients will develop DVT or pulmonary thromboembolism each year. Heterozygous Factor V Leiden increases the risk of developing DVT by 5 to 7 fold, while homozygous Factor V Leiden increases the risk of developing clots by 25 to 50 fold.6

Here, we describe a middle-aged male patient, presenting with a cardiac mass as an indicator of the Factor V Leiden deficiency syndrome.
Case Presentation

A 36-year-old male non-smoker was admitted to our clinic with intermittent productive coughs and hemoptysis in the previous 2 months for further evaluation. He mentioned that the coughs had started 7 months previously and that they were relieved with Salbutamol spray, leaving him symptom-free for 2 months. The patient had never complained of chest pain or paroxysmal nocturnal dyspnea and orthopnea before. However, his condition began to deteriorate after 2 months since the previous medication was not effective enough, and he referred to a pulmonologist. In his medical history, DVT in the left leg was noticeable in about 1.5 years previously, which at the time led to his hospital admission for anticoagulant therapy. One week afterward, he was discharged on daily Warfarin and was arranged to have international normalized ratio (INR) follow-up. Unfortunately, due to thyroid hemorrhage, he underwent a partial thyroidectomy and Levothyroxine was added to his medical treatment and the Warfarin dose was decreased. He was under treatment with low-dose Warfarin when he referred to our clinic. In social history, he only stated occasional alcohol intake. There was no history of malignancy, emboli, antiphospholipid antibody syndrome, and other types of hypercoagulable states in his family. In physical examination, no lymphadenopathy was detected and everything else was unremarkable. Primary laboratory data revealed normocytic anemia (hemoglobin = 10.9), with normal serum iron level and total iron-binding capacity. Other routine laboratory tests were within normal ranges. The pulmonary function test demonstrated small airway obstruction with a 19.2% rise in the forced expiratory volume in one second (FEV₁) after short-acting beta-agonist inhalation. Electrocardiography (ECG) showed normal sinus rhythm with a normal axis, and there were no ST-T segment changes. Chest X-ray illustrated a normal pattern (Figure 1).

Echocardiography revealed normal left ventricular size with good systolic function (ejection fraction = 55%), normal right ventricular size and function, no wall motion abnormality, and mild tricuspid regurgitation. The pulmonary artery pressure was estimated at 35 mmHg. A large (36 × 20 mm), echogenic semi-mobile mass was seen in the right ventricle attached to the interventricular septum (Figure 2). Thoracic spiral multi-slice double-contrast computed tomography (CT) demonstrated a filling defect in the right ventricle, measuring 35 mm (Figure 3). No pulmonary artery embolism was detected in CT angiography. Cardiac magnetic resonance imaging (CMR) demonstrated a lobulated mass lesion in the right ventricle (38 × 26 mm), with invasion into the interventricular septum, suggestive of thrombosis according to different sequences. Abnormal enhancement in the right lung, most probably due to pulmonary infarction, was further depicted. The other parameters were normal (Figure 4). Venous Doppler ultrasound of the lower extremities showed no evidence of DVT. For the evaluation of anemia, upper and lower gastrointestinal endoscopy was done, which detected no pathology. Bone marrow aspiration was normal. Paroxysmal nocturnal hematuria (characterized by the triad of hemolytic anemia, hematuria, and thrombosis) was ruled out by the Ham test. The patient was tested for coagulation factor activity, and the screening test was positive for Factor V Leiden. We decided to treat the patient with medical therapy. Anticoagulant therapy was administrated forthwith (Heparin drip and Warfarin). He was discharged with advice for Warfarin consumption and follow-up. After a 2-month follow-up, there was no sign of any mass in echocardiography (Figure 5).

Figure 1. Chest X-ray, showing a normal pattern

Figure 2. Transthoracic echocardiography (right parasternal long-axis view), revealing an echogenic mass (arrow) in the right ventricle attached to the interventricular septum

IVS, Interventricular septum; LV, Left ventricle; RV, Right ventricle
Discussion

Cardiac masses can produce a variety of symptoms consisting of constitutional symptoms (fever, chills, fatigue, malaise, myalgia, arthralgia, and muscle weakness), embolic phenomena (systemic or pulmonary embolism), and cardiac manifestations (arrhythmias, ventricular outflow tract obstruction, valvular dysfunction, dyspnea, orthopnea, paroxysmal nocturnal dyspnea, cough, hemoptysis, and chest pain). For distinction between differential diagnoses, physical examination coupled with appropriate laboratory tests is useful. Nevertheless, echocardiography, chest CT scan with contrast, and CMR with contrast are superior modalities for the characterization of the lesion. Studies have found that about 5% of Caucasians in North America and up to 30% of patients presenting with DVT or pulmonary thromboembolism suffer from Factor V Leiden mutation. Massive pulmonary emboli with bilateral DVT were reported in a 49-year-old with Factor V Leiden mutation. In a study by Sveinsdottir et al., it was concluded that the recurrence of venous thromboemboli was strongly associated with Factor V Leiden mutation in the heterozygous form. Another study suggested screening of Factor V Leiden mutation in patients younger than 50 years suspected of hypercoagulability states. There are, however, other types of hypercoagulability states which can present with cardiac masses. Recently, Nagae et al. reported a right ventricular mass in a 14-year-old female patient presenting with fever, DVT, and chest pain diagnosed with familial Heparin cofactor II deficiency. Other cardiac chambers are also reported to be involved with similar conditions. Corre et al. reported a coronary sinus thrombosis due to Factor V Leiden deficiency. As most patients with mutant genes will manifest the signs of Factor V Leiden deficiency at younger ages, it is suggested that any young patient with a cardiac thrombus should be further evaluated for coagulation proteins and genes. Moreover, as these patients are highly susceptible to thrombus formation, it seems that medical therapy can be an alternative to surgery when a ventricular thrombus is suspected.

Conclusion

Thrombosis is one of the most common causes of right
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ventricular masses. Given the relatively high prevalence of Factor V Leiden mutation and its life-threatening potential, screening tests are necessary in young patients without obvious reasons for recurrent thrombus formation. It seems that medical noninvasive treatments can be an alternative therapy to surgery when a ventricular thrombus is suspected in these patients.

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