Case report - Cardiac general

Extensive arterial thrombosis in a patient with factor V Leiden mutation

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Abstract

Ascending aorta and aortic arch thrombosis is rare in a young man with no risk factor. Here, we report the case of a young male patient with factor V Leiden mutation who developed ascending aorta and aortic arch thrombosis and subsequent emboli. © 2010 Published by European Association for Cardio-Thoracic Surgery. All rights reserved.

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1. Introduction

Ascending aorta and aortic arch thrombosis is an uncommon presentation in an otherwise healthy young man. The role of factor V Leiden mutation in causing venous thrombosis is well-recognized; however, its effect on arterial thromboembolism needs to be elucidated [1]. Herein, we report a young man with factor V Leiden mutation who developed lower limb arterial, ascending aorta and aortic arch thrombosis and subsequent emboli to the eye.

2. Case presentation

A 26-year-old man with progressive pains in his upper and lower extremities during the previous 1.5 years underwent Doppler sonography of the lower extremities, which demonstrated bilateral occlusion of superficial femoral arteries, popliteal arteries, and posterior tibialis arteries as well as right peroneal artery, although the venous flow was normal. Thoracic MR angiography revealed a mass in the ascending aorta and anterior aortic arch. The patient was, thereafter, referred to our center.

The patient had developed visual field loss in the upper half of the left eye three days before presentation. He was a non-smoker and his previous history was not significant.

Transesophageal echocardiography (TEE) showed a mass above the sinotubular junction extending into the aortic arch (Fig. 1). Multi-sliced computed tomography (CT) angiography also revealed a mass (Fig. 2a).

The patient had a heterozygote mutation of factor V Leiden. He was negative for anticoagulins and lupus anticoagulant. Protein C and S levels could not be assessed, because the patient was on warfarin.

In the operating room, epiaortic echocardiography was performed to find a safe site for cannulation (Videos 1 and 2). Considering that the mass in the aorta was mobile and had a thin stalk, there was a risk of detachment in placing the patient on the cardiopulmonary bypass. First, two purse string sutures were placed on the left carotid and the innominate artery and then a tape was placed over the two arteries. The femoral artery and the right atrium were, thereafter, cannulated. The flow of the left carotid artery and innominate artery was started at 700 ml/min and immediately afterwards the two neck vessels were clamped at their origin with the aorta and were separated from the systemic flow to prevent emboli to the cerebral vessels. At the same time, the patient was placed on normothermic cardiopulmonary bypass. The cannula for the retrograde cardioplegia was placed, the flow was reduced, and when the perfusion was stopped complete arrest was done. The aorta was opened as fast as possible, and the mobile mass with extension into the left subclavian artery was evacuated before the part of the aorta adhering to the thrombi could be removed and repaired with a pericardial patch (Figs. 2b,c). At the same time, air suction from the descending aorta was performed through the femoral artery. After the aorta was repaired, the flow was commenced and the clamps were removed. The body circularly arrest was about 10 min. The patient was removed from the pump. He was extubated 6 h later.

The pathology report showed infiltration of acute and chronic inflammatory cells, collection of foam cells and deposition of cholesterol cleft, indicating focal atherosclerosis. The other fragment, a blood coagulum, consisted of fibrin material infiltrated by acute and chronic inflammatory cells.

The patient was discharged on warfarin with an International Normalized Ratio (INR) of 2.8.
3. Discussion

In cases of embolism of unknown origin in young patients without any previous medical condition, TEE and CT angiography can have potential importance. Without these new modalities, thrombi in the ascending aorta and aortic arch could remain unrecognized [2]. Aortic thrombus usually occurs on previous aortic pathologies, including an atherosclerotic plaque, aortic aneurysm or dissection [3].

Atherosclerosis of the aortic arch occurs in the elderly and can be a source of thrombus formation and emboli [3]. Thrombus formation in the aortic arch in a young patient with no previous health problem necessitates extensive thrombophilia evaluation. The optimal treatment of this presentation has yet to be explained, and conservative management with anticoagulant puts the patient at risk of recurrent embolism from the thrombi in the aortic arch [3]. Conservative management was, therefore, not favored by our surgeon.

The thrombophilia work-up led to the diagnosis of factor V Leiden mutation. Metsvaht et al. reported aortic arch thrombi formation in a neonate with factor V Leiden mutation. In a literature review, they found 12 other cases of aortic arch thrombi formation in neonates, 11 of which had not been studied for factor V Leiden and the one case studied was negative [4]. In adults, cases of floating thrombi in the ascending aorta and subsequent embolism have been reported; no known recognizable coagulation abnormalities have, however, been reported for some of these cases [5]. Cases of aortic arch thrombosis have also been reported in adults [6].

Factor V Leiden mutation is the most common heritable cause of thrombophilia. In cases of heterozygote factor V Leiden mutation, which occurs at younger age, trauma, oral contraceptives and pregnancy have a much higher prevalence [7]. The role of factor V Leiden in arterial thromboembolism is less clear. Ng et al. reported another case of catastrophic heterozygote factor V Leiden mutation resulting in recurrent arterial thromboembolism and subsequently bilateral lower limb amputation; the original site of the thrombosis was not recognized [1].

Synergistic effect of factor V Leiden mutation and other prothrombotic conditions, such as high homocystine levels, protein C and S deficiency, oral contraceptives and pregnancy in causing arterial thrombosis has previously been described. Page et al. reported a case of acute popliteal artery thrombosis due to heterozygote factor V Leiden
Video 1. Epiaortic echocardiography demonstrating the mass attached to the ascending aorta.

Video 2. Epiaortic echocardiography demonstrating the mass in the origin of the left subclavian artery.

mutation and hyperhomocysteinemia; they concluded that factor V Leiden in association with other prothrombotic factors might predispose one to arterial thrombosis [8]. The patient’s thrombophilia led to aortic arch thrombosis and subsequent emboli to the eye. Bessero et al. [9] reported four patients with heterozygote factor V Leiden mutation who developed visual field defect due to arterial thrombosis; two of these patients had no other coagulation abnormality other than factor V Leiden mutation.

Further investigation is required to determine whether this mutation would be enough to cause arterial thrombosis.

References