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Case Report

Left Ventricular Clot due to Wild-Type Homozygous Factor V Leiden Mutation: A Case Report

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ABSTRACT

Factor V Leiden (FVL) mutation has been identified as a frequent risk factor for life-threatening venous thromboembolic events. We describe a 39-year-old man admitted with sudden-onset blurred vision and mild lower-limb paresthesia with a positive activated protein C resistance test. The polymerase chain reaction confirmed wild-type homozygous FVL mutation. Transthoracic echocardiography revealed a highly elongated mobile mass attached to the akinetic cardiac apex, which extended to the left ventricular outflow tract. The thrombosis was removed by thrombectomy through aortotomy. The patient was in good clinical condition at the last follow-up. ***(Iranian Heart Journal 2015; 16(3): 57-59)***

Keywords: ■Factor V Leiden ■Mutation ■Thrombosis

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Factor V Leiden (FVL) mutation has been identified as an important risk factor for venous thromboembolism. It occurs at an earlier age when FVL mutation is homozygous.[1, 2] Based on recent clinical reports, FVL mutation can be frequently associated with increased risks of primary or recurrent thromboembolic events, during other predisposing conditions or in the presence of other genetic and acquired abnormalities of anticoagulation.[3, 4] On the basis of these findings, screening programs for the mutation of this factor should be proposed. This screening approach would be able to facilitate the inhibition of excessive clotting in the cardiac vessels and chambers and, thus, effectively prevent stroke or heart attack.

In the current report, we describe wild-type homozygous FVL mutation in a patient.

Case Report

A 39-year-old man referred to our hospital complaining of blurred vision and unilateral pins-and-needles sensation in his lower extremities. Ophthalmologic examination was normal. Neurological examination revealed paresthesia in both lower limbs below the knees. There was no muscle weakness or deep tendon reflex abnormalities. Family and drug history were unremarkable. Chest radiography was normal. Cardiovascular investigations,

including 24-hour electrocardiography, revealed normal sinus rhythm but with low-amplitude T waves in the inferior and V3-V6 leads as well as coved ST segments in V3 and V4 leads.

Hematological examination was normal with a serum hemoglobin level of 15.1 g/dL, platelet count of 213×10^3/L (with giant platelets), and white blood cell count of 8600/ mm^3. Partial thromboplastin time was 33 sec and prothrombin time was 12 sec (INR= 1). The serum total cholesterol level was 214 mg/dL. Other biochemical tests — including liver function tests, erythrocyte sedimentation rate, and thyroid function tests — were all within the normal range. With respect to the inflammatory biomarkers, the results of the antinuclear antibody test and anti-neutrophil cytoplasmic antibodies were negative. However, the test for activated protein C resistance was positive.

Transthoracic echocardiography revealed a highly elongated mobile mass (7.3×1.3 cm) attached to the akinetic apex and extended to left ventricular outflow tract. This mass most probably showed a thrombosis. Echocardiography evaluation also demonstrated a near normal left ventricular ejection fraction (45 – 50%) and normal left ventricular volume. Coronary angiography revealed no diseased vessels or abnormalities. Due to the positive result of activated protein C resistance, the polymerase chain reaction (PCR) analysis was performed and it confirmed a mutant wild-type homozygous state.

With the primary diagnosis of left ventricular clot, the patient underwent aortotomy. After mid-sternotomy and pericardiotomy, the intrapericardial ascending aorta was exposed. Cardiopulmonary bypass was initiated, and cardiac arrest was induced by a cold antegrade administration of blood cardioplegia. Thrombectomy was performed using catheters through the ascending aortotomy.

The patient's blurred vision and paresthesia improved after 1 – 2 days during his hospitalization. The patient was discharged in good general condition, and warfarin was prescribed with the recommendation for routine follow-up visits.

DISCUSSION

Left ventricular clot is a potentially life-threatening complication that can be related to left ventricular dysfunction. This condition requires urgent treatment to prevent coronary and cerebral events as well as systemic embolic phenomena. Left ventricular clot formation is frequently detected by echocardiography.

Left ventricular clot is commonly caused by hypercoagulability mainly due to a deficiency of natural anticoagulant protein C or protein S. Under normal circumstances, the activation of proteins C and S in the coagulation cascade acts to limit the clotting extent by cleaving and degrading factor V.

FVL is an autosomal dominant condition that exhibits incomplete dominancy and results in a factor V variant, which is not as easily degraded by activated protein C. The appearance of a mutation in exon 10 of the coding gene of this protein results in the replacement of arginine at amino acid position 506 by glutamine. This mutation has been detected in about 20 to 50% of patients presenting with venous thrombosis.[5-7] FVL mutation can be associated with a tendency toward idiopathic venous thromboembolisms, arterial thrombosis, juvenile stroke, and myocardial infarction in young ages. Therefore, suspicion of FVL, as a potential cause for any thrombotic event, should be made in any person with a family history of venous thrombosis or its underlying risk factors — including female gender, pregnancy, and oral contraceptives consumption.

Blood test is initially used as a screening test to determine activated protein C resistance,

which can be caused by the presence of FVL. By this test, discrimination of the heterozygous or homozygous forms of FVL is also facilitated. This differentiation is very important because it has been revealed that heterozygous FVL can increase the risk of developing a first vein clotting by 5 to 7 fold, while in the homozygous type, the rate of this risk has been estimated higher at about 25 to 50-fold.[8]

In the current report, we described a young patient who developed left ventricular clot and concurrently had a positive activated protein C resistance test. This positive finding guided us toward considering probable FVL mutation, which was finally confirmed with PCR analysis. Our patient suffered from a wild-type homozygous FVL mutation, which predisposed him to thromboembolic events manifested by left ventricular clot formation.

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