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Research Article

Basilar Artery Thrombosis in a Young Patient with Factor V Leiden Mutation and Patent Foramen Ovale: A Case Report

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Abstract

Introduction: Basilar artery stroke is a rare but devastating condition that requires swift diagnosis and treatment. Factor V Leiden is a hypercoagulability mutation primarily associated with venous thromboembolism, but is rarely associated with arterial atherothrombotic events such as ischemic stroke. In young patients, the presence of a PFO and prothrombotic mutation can increase the risk of cerebrovascular accidents.

Case Presentation: A 32-year-old female presented with symptoms of basilar artery thrombosis and was diagnosed through physical examination and relevant investigations. Swift initiation of IV tPA and thrombectomy procedure, as well as multidisciplinary team approach to patient care, were crucial in managing the patient's condition.

Discussion and Conclusion: The rarity of inherited thrombophilias in association with arterial thromboembolic events highlights the need for further research to better understand the etiology of such events in young patients. Considering all risk factors, including PFO and prothrombotic mutations, can help prevent cerebrovascular accidents in young patients.

Keywords: Basilar Artery Thrombosis; Deep Vein Thrombosis (DVT); Pulmonary Embolism

Introduction

Basilar artery stroke is a rare, yet clinically devastating condition. The artery primarily supplies the brainstem. Symptoms can range from focal neurologic deficits such as focal paralysis and paresthesias to a complete “locked-in syndrome.” During a suspected brainstem stroke event, basilar artery occlusion needs to be ruled out swiftly to prevent significant morbidity and mortality [1].

Factor V Leiden mutation is a hypercoagulability mutation associated with recurrent venous thromboembolism events, most commonly deep vein thrombosis (DVT), pulmonary embolism, and recurrent pregnancy losses [2]. In fact, carriers of this mutation have a four-fold increment of risk in venous thromboembolism [3]. However, arterial atherothrombotic events, including ischemic stroke in a patient with Factor V mutation, let alone a heterozygous one, is an extremely rare incident which warrants further investigation into the etiology.

Presence of both PFO and a prothrombotic mutation are minor risk factors for cerebrovascular accidents but can have an additive effect when present simultaneously in a patient, especially in young patients [3].

We present the case of a 32 year-old female with a basilar artery stroke with a heterozygous factor V leiden mutation.

Case Presentation

The patient was a 32 year-old Caucasian female who presented to the Emergency Department (ED) with sudden onset slurred speech, left arm drift, and left leg numbness and weakness. Dizziness and inability to focus was reported by the patient. Her past medical history was unremarkable except for a heterozygous Factor V Leiden mutation. Social history indicated lack of alcohol and tobacco use. Family history revealed an unspecified clotting disorder in the mother of the patient.

On physical examination, the patient was diaphoretic with alert mental status. Neurological examination revealed slurring of speech, and decreased sensation to light touch on the left lower extremity along with weakness in the same limb. Pupils and extraocular movements were intact. The NIH Stroke scale was calculated to be 4. Cardiovascular, Gastrointestinal and Respiratory examinations were insignificant.

Relevant investigations included non-contrast CT scan of the head showed right cerebellar hypodensities suggestive of infarcts. Additionally, CT angiography of the head and neck revealed distal basilar artery occlusion/vasospasm for a segment of 8mm with patent bilateral PCAs.

Interventional Neurology was consulted and recommended an MRI, which indicated a left superior cerebellar infarct and a thrombus in the basilar artery (Refer to Figures 1 and 2). An official diagnosis of basilar artery thrombosis was made and intervention was planned. IV tPA was initiated swiftly along with a thrombectomy procedure (Refer to Figures 3 and 4). Patient was admitted to the ICU after the procedure.

Patient had stable vitals during her stay in the ICU over the next five days. Initially, she had weakness in her right limbs along with slurred speech which was monitored with repeat CT scans. Patient progressed towards recovery with steady improvement in right limb weakness and slurred speech with the help of a multidisciplinary team of speech, occupational, and physical therapists along with a vast team of physicians that included neurologists, cardiologist, critical care physician, and a hematologist.

Lack of certainty regarding the etiology of the stroke provoked the physicians to perform a Transesophageal Echocardiography (TEE) with agitated saline study. The procedure indicated the presence of a Patent Foramen Ovale (PFO), as per suspicion.

Patient was de-escalated from the ICU to a lower level of inpatient care after five days with a prompt discharge of the patient. On discharge, the patient had significantly improved neurological deficits and was prescribed an intensive physical therapy regimen on her road to recovery.

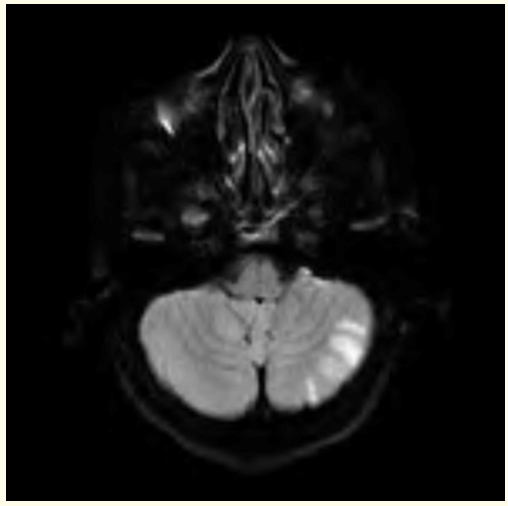

Figure 1: DWI noted Left Cerebellar infarct.

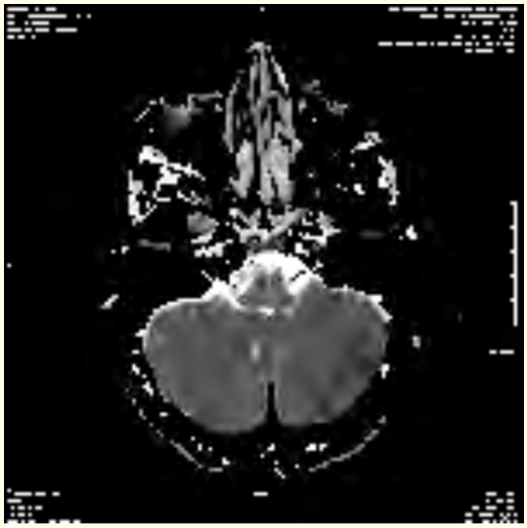

Figure 2: ADC confirmed Left Cerebellar infarct.

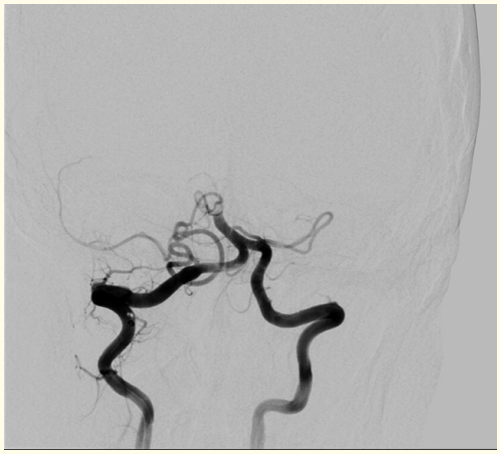

Figure 3: Basilar artery occlusion noted pre-thrombectomy.

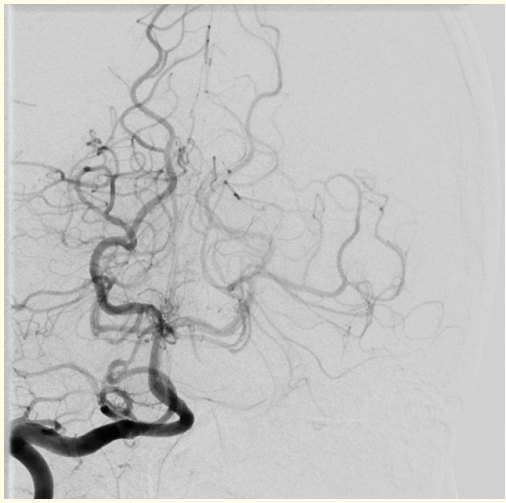

Figure 4: Basilar artery patent flow post-thrombectomy.

Discussion

Factor V Leiden mutation is most commonly associated with Venous thromboembolism. Limited data exists for association of inherited thrombophilias with arterial thromboembolic events but it is definitely a possibility. Basilar artery is a part of the posterior circulation and an occlusion of this vessel is associated with significant morbidity and mortality [4] as evident by the statistic that the mortality rate of this condition can be as high as 74% to 100% if timely recanalization and thrombectomy is not done [5]. Thrombosis of the artery accounts for only 1% of all ischemic strokes [6]. Due to the pressing nature of this condition, and its associated high rates of mortality, all risk factors need to be taken into account to prevent this from occurring in the first place.

This patient is young with no significant risk factors that put her at risk of an ischemic stroke. Obesity, smoking, alcohol abuse, high blood pressure, high cholesterol, type 2 diabetes, cardiovascular disease, and other major risk factors were all ruled out in this patient. Toxicological analysis showed no significant illicit drug usage. Further evaluation into the history revealed an important finding which was the presence of a heterozygous Factor V Leiden Mutation. A meta-analysis of polymorphism in the Factor V Leiden gene showed increased susceptibility to ischemic strokes, especially in young adults (OR 1.84; 95% CI: 1.47 to 2.30) [7]. More specifically, patients who are heterozygous carriers of this mutation had an increased chance of stroke with an OR of 1.23 (95% CI, 1.05–1.45), as specified in this study8. Additionally, individuals with homozygous factor V Leiden mutation had a significantly increased risk of acute ischemic stroke (OR 2.24; 95% CI: 1.26–4.71), as expected [8].

Further investigation into the case prompted the detection of a Patent Foramen Ovale. Presence of a PFO in this patient further strengthened the pathophysiologic process of ischemic stroke in this patient with minimal risk factors.

Purpose of this case report is to discuss possible albeit rare medical emergencies regarding patients with inherited thrombophilias. Acute Basilar artery thrombosis can be an extremely morbid, if not fatal, condition, where chances of a complete recovery are rare. Guidelines regarding the management of thrombophilic conditions need to be updated to prevent such disastrous consequences. All factors related to enhancing thrombogenesis need to be addressed promptly at time of diagnosis. This may include closure of Patent Foramen Ovale and anticoagulation concomitantly, which is associated with decreased overall rates of recurrent ischemic strokes (45% overall relative risk reduction as compared to patients with anticoagulation alone) in patients with an initial cryptogenic episode (RESPECT trial) [9]. Similar findings were observed in the CLOSE trial, supporting the need of PFO closure in patients who had suffered from cryptogenic strokes [10].

Extensive randomized control trial (RCT) data is required to make decisions regarding prescription of medical anticoagulation

therapy and placement of IVC filters, in patients with heterozygous thrombophilias. All of these interventions need to be carefully tailored to each patient and planned promptly to optimize clinical outcomes and decrease morbid consequences related to these mutations.

Conclusion

Patients with Factor V Leiden mutations, heterozygous or homozygous, need to undergo a thorough risk stratification of thromboembolic events. This includes prompt identification of a PFO to prevent paradoxical embolization induced adverse events, as seen in the case above. Updates regarding the guidelines for management of patients with similar thrombophilias, including the usage of anticoagulants in heterozygous patients is also essential.

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