CASE REPORTS

Congenital agenesis of inferior vena cava as a rare cause of deep vein thrombosis: Case report and review of the literature

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ABSTRACT

Inferior vena cava agenesis (IVCA) is an uncommon cause of deep vein thrombosis (DVT). We report the case of a 26-year-old Caucasian female with unilateral iliofemoral DVT. On further evaluation, CT abdomen revealed diminutive inferior vena cava consistent with IVCA. We have used systemic anticoagulation and catheter mediated thrombolysis with success in this case. Physicians should suspect IVCA in patients with extensive DVT especially in younger individuals.

Key Words: Deep vein thrombosis, Inferior vena cava agenesis, Catheter directed thrombolysis

1. INTRODUCTION

Inferior vena cava agenesis (IVCA) is a rare congenital anomaly and an uncommon cause of deep vein thrombosis (DVT) with an estimated prevalence of 0.0005% to 1% in general population.[1] IVCA is usually reported as an incidental finding and accounts for approximately 5% of cases of unprovoked DVT in individuals under 30 years of age.[2] We report a case of IVCA presenting with unilateral extensive iliofemoral DVT. The present case illustrates the diagnosis and management of this condition and emphasizes the need to consider IVC anomalies in the differential diagnosis of DVT especially in a young patient.

2. CASE PRESENTATION

A 26-year-old female was admitted to the hospital with a sudden onset of low back pain and left lower extremity swelling of less than 24 hrs duration. The swelling was associated with pain, numbness and discoloration. She was on intermittent oral hormonal contraceptive pill (OCP) for 6 years prior to presentation. She denied any trauma or recent prolonged immobilization. She was a non-smoker with no personal or family history of thrombophilia or thromboembolic events. On admission, her vitals were pulse rate 90/min, blood pressure 121/72 mmHg, respiration rate 14/min, temperature 36.9°C. Physical examination revealed diffuse swelling and mottling in her left lower limb extending from upper thigh to the foot. There was mild tenderness and warmth in the affected limb compared to the normal limb. No injuries/varicosities/neurovascular deficits were noted. Her biochemical and hematological investigations were normal except for mild normocytic anemia with hemoglobin 10.6 g/dl, MCV 87.4 fL, Hematocrit 32.5%. Duplex ultrasound of the left lower extremity showed DVT involving the left common femoral, femoral, popliteal and peroneal veins. A

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contrast CT scan of the pelvis was done to look for the proximal extension of the clot. It revealed thrombosis of the left common iliac, left external and internal iliac veins with a diminutive IVC and dilated gonadal veins and lumbar collaterals. This raised concern for a more proximal IVC obstruction. A congenital absence of IVC with multiple collaterals distal to the liver with prominent azygous and hemiazygous circulation was confirmed on contrast-enhanced CT scan of the abdomen (see Figure 1).

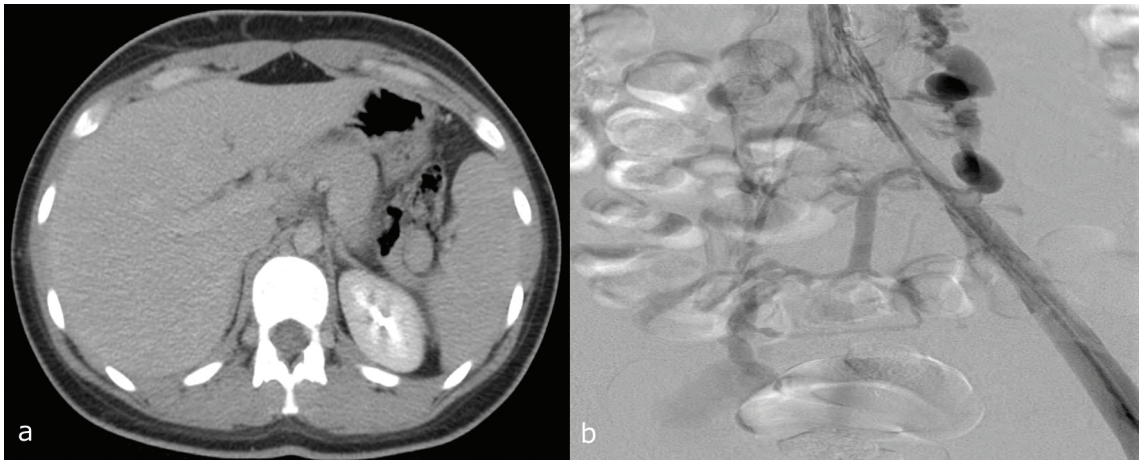

Figure 1. a. Congenitally absent IVC. Note the absence of IVC on the right side; b. Digital subtraction imaging shows thrombosed left iliac vein which does not communicate with the gonadal veins and lower extremity blood flow appears to continue through the azygos, hemizygous and multiple collaterals

Treatment with therapeutic low molecular weight heparin was started promptly on admission. Given her extensive clot burden, a catheter-directed thrombolysis was planned. Patient received continuous thrombolysis with tissue plasminogen activator (tPA) and heparin via the Craig McNamara infusion catheter and a 6 French sheath, respectively. A follow-up venogram demonstrated complete resolution of iliofemoral venous thrombosis with markedly improved flow through the left leg and through the collaterals. No procedural complications were noted and the patient was discharged in a stable condition. She was recommended 6 months of anticoagulation with rivaroxaban, discontinuation of the OCP with the use of two forms of non-pharmacological contraception. Her follow up hospital visit after 2 weeks of discharge showed complete resolution of symptoms with no significant concerns. The patient was receptive to regular follow-ups.

3. DISCUSSION

The incidence of venous thrombosis is 1 in 1,000 individuals per year in developed countries.[3] Virchow's triad describes three main causes of thrombosis hypercoagulability, endothelial injury, and venous stasis.[3] The congenital risk factors for thrombosis include deficiencies of protein C, protein S, antithrombin III and gene mutations of prothrombin and factor V Leiden. The acquired risk factors are surgery, prolonged immobilization, trauma, pregnancy, puerperium, OCP, malignancy and lupus anticoagulant.[3] The dynamic interplay of these risk factors induce a state of hypercoagulability and venous stasis, thereby resulting in thrombosis.[3–5] Recently IVC anomalies have been identified as a possible congenital risk factor for DVT.[2,4–7] The estimated prevalence of IVC anomalies is 0.07%-8.7% in the general population.[6] Inferior vena cava develops between 6^{th} to 8^{th} week of embryogenesis from a composite structure formed by sequential appearance, anastomosis and regression of three pairs of primitive veins the posterior cardinal, the subcardinal and the supracardinal veins.[8] Any aberrations in this process result in IVC anomalies which include double IVC, hypoplastic segments of IVC, agenesis of IVC among others.[8] However, there is a school of thought which diverges from this notion and considers chronic occlusion as a consequence of perinatal thrombosis to be the cause of IVC agenesis.[8] Generally, individuals with IVCA develop extensive collateral circulation as a means to compensate for the anomaly. The inadequate blood return through these collaterals results in increased venous blood pressure, which subsequently predisposes to venous thrombosis.[1,4–6]

IVCA should be suspected in any young individual presenting with extensive venous thrombosis[1,2,4–7] . Chee *et al*. noted in their case series that up to 5% to 6.7% of young adults (aged between 20-40 years) presenting with DVT had an IVC anomaly, which was much higher than the expected 0.5%.[4] Ruggeri *et al.* diagnosed four cases of IVCA in patients under 30 years with idiopathic DVT over a 5-year

period and estimated that IVCA is present in about 5% of cases of DVT in young adults.[2] Garcia-Fuster *et al*. in their prospective study of 116 patients under 50 years reported that IVC anomalies were present in 16.2% of the patients with iliac vein thrombosis and cava malformation represented a risk factor in 5.1% of the population studied.[7] It is believed that IVC anomalies have been underdiagnosed and may be much more common than anticipated.[2,4] This may be due to the fact that venous ultrasound, most widely used modality for diagnosing DVT is insufficient to detect these anomalies[1,2,5] Contrast enhanced CT scan or MRI studies should be employed in suspected patients.[1,2,5] This is especially effective in the cases involving thrombosis of proximal veins and collaterals masquerading as a malignant mass posing a diagnostic challenge.[5] Venogram is reserved for venous mapping during thrombolysis and for checking post procedural efficacy.[9] Hypercoagulability studies are also recommended;[3–5] however, its relevance is uncertain as it did not modify the patient management in majority of the cases.[1]

Since IVCA associated DVT is a rare condition, no specific therapeutic approaches have been developed. The treatment strategy is individualized. Goal of the therapy is to prevent the progression and recurrence of thrombosis. Long-term anticoagulation, elastic stockings and avoidance of risk factors like prolonged immobilization and OCPs have been shown to be of benefit.[1,6] Development of thrombosis in this case is possibly unprovoked since the patient had tolerated intermittent OCP over 6 years without any occurrence of DVT. Six months of systemic anticoagulation was recommended following the discontinuation of the OC pill. In extensive iliofemoral DVT, thrombus removal plus optimal anticoagulation has shown significant improvement in the outcome when compared to anticoagulation alone.[10] Catheter directed intra thrombus delivery of tPA accelerates thrombolysis by preventing neutralization of plasminogen activators by the plasminogen activator inhibitor and also protects the active enzyme plasmin from circulating anti-plasmins.[10] This reduces the overall dose and duration of therapy thereby increasing the efficacy of clot lysis and decreasing the complications when compared to systemic thrombolysis. Catheter directed thrombolysis was also shown to effectively mitigate the clot burden by promoting complete clot resolution, significant thrombus free survival and reduced post-thrombotic morbidity.[10] Patient underwent catheter-mediated thrombolysis in conjunction with systemic anticoagulation with excellent results (see Figure 2). Literature review of similar cases disclosed successful use of thrombolysis coupled with anticoagulation.[9,11–14] Further studies are warranted to reinforce the role of thrombus removal and its long-term benefit in the treatment of iliofemoral DVT.

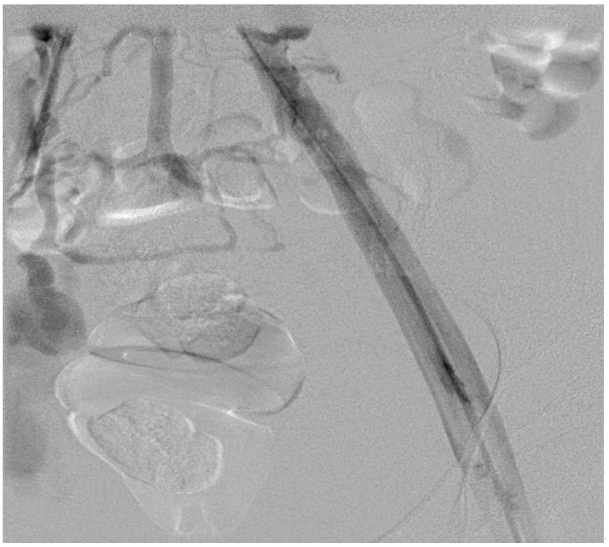

Figure 2. Venogram performed through the left popliteal vein sheath and through the infusion catheter demonstrated complete resolution of iliofemoral venous thrombus with marked improvement in flow through the left leg and through the azygos and pelvic collaterals

4. Conclusion

It is reasonable to consider IVCA as a differential diagnosis in young patients presenting with extensive DVT with no typical risk factors. Contrast enhanced CT scan or MRI studies should be considered in diagnosing IVC anomalies. Thrombus removal with optimal anticoagulation has shown to improve the overall outcome in cases of widespread DVT. Avoidance of thrombosis risk factors and use of elastic stockings should be strongly recommended in all DVT patients.

Conflicts of Interest Disclosure

The authors have declared no conflicts of interest.

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