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**Case Report**

# From rare anomaly to clinical reality; managing venous thrombosis in the setting of congenital inferior vena cava agenesis

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## ABSTRACT

A congenital absence of the inferior vena cava (IVC) is a rare anomaly known to be linked with idiopathic deep venous thrombosis (DVT). This anomaly is more commonly observed in individuals with other congenital cardiac anomalies, with an incidence ranging from 0.6% to 2%. Here we are reporting a rare case of missing IVC presented with extensive bilateral lower extremity and iliac veins DVT. The symptoms responded well to therapeutic doses of heparin. In this article, we will review some other cases of IVC agenesis and different management including medical, interventional, and surgical approaches.

**Keywords:** Inferior vena cava, Congenital agenesis, Deep vein thrombosis

## INTRODUCTION

A congenital absence of the inferior vena cava (IVC) is a rare anomaly known to be linked with idiopathic deep venous thrombosis (DVT), especially in younger individuals.[1] Anomalies in the IVC can manifest as complete absence, partial absence, or duplication of the IVC. These anomalies are more commonly observed in individuals with other congenital cardiac anomalies, with an incidence ranging from 0.6% to 2%. This congenital condition may be identified incidentally or due to symptoms related to associated congenital heart disease, asplenia, polysplenia, congenital kidney anomalies, or deep venous thrombosis.[2] Considering the rarity of this disease there is no standard practice for treating these patients. Therefore, reporting each case is necessary and adds to existing data to come to a common consensus for management.

## CASE REPORT

A 19-year-old female otherwise healthy presented to the emergency department with complaints of a two-day history of bilateral progressive lower extremity swelling. She had a recent low back pain for which she was less ambulated within the last three weeks. She has been on estrogen-component birth control pills for about six months. On the examination, she had significant right lower extremity edema from foot to groin and mild distal edema on the left lower extremity. Initial blood work was significant for elevated D-dimer and CRP. Other laboratory findings were within normal limits.

The US (Figure 1) and CT venogram showed bilateral thrombosis of common, external, and internal iliac veins with extension into the left femoral and right popliteal veins, along with the absence of the IVC below the renal veins. The outflow was provided by collaterals via ovarian and lumbar veins into the retroperitoneal azygos vein (Figure 2). She was placed on the systemic therapeutic dose of anticoagulation with heparin at the dose of 17 units/kg for the first 48 hours then switched to Eliquis 10 mg twice a day for seven days. In 24 hours, symptoms practically resolved. On the hospital day seven, she was discharged on lifelong Eliquis 5 mg twice a day. On her 9-month follow up she did not have significant edema. US and CT were requested and

discussed with the patient however, since she was asymptomatic, she refused more follow-up imaging. Any recurrence will be addressed accordingly based on the severity of the symptoms, the extent of the disease, and medication compliance.

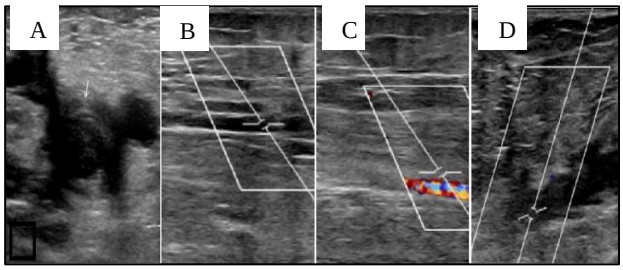


**Figure 1: Right lower extremity ultrasound: A) thrombosis of right common femoral vein. B) thrombosis of the right superficial femoral vein. C) right profounda vein thrombosis. D) right popliteal vein thrombosis.**

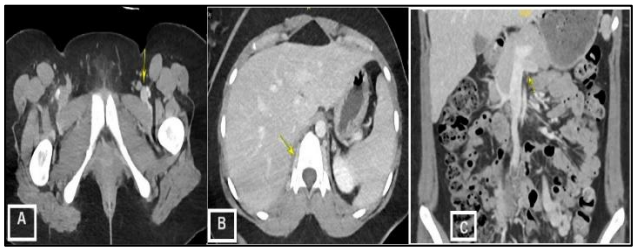


**Figure 2: CT of the abdomen and pelvis: A) left femoral vein thrombosis B) prominent lumbar vein C) prominent left gonadal vein.**

## DISCUSSION

In normal development, the infrahepatic IVC originates from three paired parallel veins that appear sequentially between 4 and 8 weeks of life—the posterior cardinal, subcardinal, and supracardinal veins. The suprahepatic IVC is formed from the upper segment of the right vitelline vein. The infrahepatic/suprarenal IVC arises from the right subcardinal vein. The suprarenal segments of both the right and left supracardinal veins link with the posterior cardinal veins, forming the azygos and hemiazygos systems, respectively. The infrarenal segments vanish on the left side but contribute to the infrarenal part of the IVC on the right side. The posterior cardinal veins typically regress, except for their distal section, which transforms into the iliac confluence and future iliac veins, and their proximal section, which merges with the azygos and hemiazygos veins.[3]

IVC anatomical abnormalities are developmental disorders that can arise between six to 10 weeks of gestation. As mentioned above the infrahepatic IVC originates from three pairs of embryonic veins. An absent IVC is one type of vascular anomaly that results from failure of these paired structures to fuse. It has also been shown that agenesis of the IVC may be caused by a chronic perinatal thrombosis of the IVC, leading to persistent occlusion and the formation of a fibrosed cord-like structure, and not by an embryonic anomaly.[4]

When the IVC is absent, venous blood from its drainage region follows an alternative route to reach the right atrium. This alternate pathway is the azygos vein, which drains the lower portions of the body and terminates in the superior vena cava. With agenesis of the IVC, collateral veins become enlarged to facilitate blood flow to the superior vena cava. While these alternate pathways allow venous drainage from the lower extremities, IVC agenesis (IVCA) can increase the risk of venous hypertension and associated complications, such as thromboembolism.[5]

Congenital anomalies of the inferior vena cava (IVC) occur in approximately 0.3% to 0.5% of young, healthy adults; however, many authors agree an absent IVC may be underreported.[4] The diagnosis can be elucidated by several clues on radiological imaging, such as the presence of well-developed and potentially dilated intra-thoracic hemiazygos and/or azygous continuations. These collateral circulations, along with other retro-peritoneal venous pathways, are typically well-established before symptoms manifest.[6] The most dependable non-invasive methods for detecting IVC anomalies are CT scans with intravenous contrast or magnetic resonance imaging (MRI) 7. Ultrasound scanning alone is often inadequate for diagnosing IVC anomalies. Venography, though more invasive, is highly accurate and beneficial, especially when surgical intervention is planned.[1]

Anomalies involving the IVC are acknowledged but represent uncommon origins of DVT. The occurrence of DVT among individuals aged 40 and above is estimated at 1 in 1000, whereas it is even less frequent in those aged 20–40, with an incidence as low as 1 in 10,000.[8] The causes of DVT are multifaceted and stem from a combination of environmental and genetic factors that influence blood coagulation or flow. In younger individuals presenting with DVT where the primary known risk factors are absent, investigations often focus on identifying potential genetic or congenital factors as the underlying cause.[1]

Ruggeri et al, reported four instances of absent IVC within a five-year span among individuals under 30 years old presenting with idiopathic DVT. This accounted for approximately 5% of idiopathic DVT cases in this age group.[9] Similarly, Fuster and colleagues examined 116 patients younger than 50 years old who had DVT. They discovered that 6 out of 37 patients (16.2%) with iliac vein thrombosis had a venous anomaly. The diagnosis of DVT linked to anomalies of the IVC is typically made using color venous Doppler ultrasound, along with CT angiography or magnetic resonance imaging.[4,10] Additionally, research has indicated that anomalies of the IVC are associated with other conditions, such as

congenital heart disease, asplenia, polysplenia, and congenital kidney anomalies. However, these anomalies are more commonly observed in individuals with other congenital cardiac anomalies, with an incidence ranging from 0.6% to 2%.[11] Commonly associated cardiac defects include dextrocardia, atrial septal defects, atrioventricular canal, or pulmonary stenosis.[12] The presence of congenital heart disease alongside IVC anomalies has been shown to increase the risk of thrombosis and formation of systemic emboli, and thus must be managed effectively.[13]

Currently, there are no guidelines regarding the management of patients with absent IVCs. Studies have demonstrated that only 10.7% of patients exhibited pulmonary embolism (PE), which could be attributed to the abnormal anatomy of the deep veins, reducing the likelihood of thrombi reaching the pulmonary arteries.[14] PE is a rare occurrence as emboli are intercepted in the azygos/hemiazygos system before they reach the pulmonary circulation.[15,16]

The current mainstay treatment approach involves prolonged anticoagulation, although there is limited evidence regarding the optimal duration. Anticoagulation may be prescribed indefinitely to prevent recurrence, or for a minimum of 6 months initially, with a reassessment of patients' bleeding risk before considering further treatment adjustments.[14] It's also recommended to use compression elastic stockings and elevate the limbs concurrently. Patients are advised to avoid risk factors such as excessive physical exertion, prolonged immobilization, or smoking.[15]

Besides conventional anticoagulation methods, modern endovascular techniques such as pharmaco-mechanical thrombectomy (PMT) and catheter-directed thrombolysis (CDT) are becoming more prevalent in the treatment of extensive extremity DVT in both pediatric and adult populations.[16] Multiple authors have reported successful management of acute iliofemoral DVT in cases of IVC absence using catheter-directed pharmaco-mechanical thrombolysis and thrombectomy, resulting in excellent outcomes over 2 to 5 years.[17,18] The combination of pharmaco-mechanical catheter-directed thrombolysis (PCDT) followed by systemic anticoagulation and the utilization of compression stockings seems to be a safe and efficacious approach for the long-term management of patients presenting with acute DVT and IVC agenesis or atresia.[19,20]

The definitive treatment of IVC agenesis is venous reconstruction, which has shown favorable outcomes. Early surgical intervention followed by oral anticoagulation has been suggested in the literature, as it has the potential to reverse the debilitating venous insufficiency associated with IVC agenesis. The first documented case of IVC reconstruction using a polytetrafluoroethylene (PTFE) graft for symptomatic congenital absence of IVC was reported in 2008.[21]

## CONCLUSION

rare; therefore, no standard guideline has been published regarding their management. Treatment approaches vary but commonly involve anticoagulation, endovascular techniques such as pharmaco-mechanical thrombectomy, and occasionally surgical reconstruction in severe cases. Management strategies for patients with absent IVCs aim to prevent recurrence of thrombotic events and alleviate symptoms of venous insufficiency. However, there is a lack of standardized guidelines regarding the optimal duration of anticoagulation. Despite this, modern endovascular interventions have shown promising results in managing acute DVT in the setting of IVC anomalies, with catheter-directed thrombolysis and thrombectomy leading to excellent outcomes over several years.

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