Inferior Vena Cava Agenesis as Cause of Pulmonary Embolism: Case Report and Literature Review

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ABSTRACT

Introduction: Inferior vena cava agenesis (IVCA) is a very uncommon vascular condition, occurring in about 0.0005% to 1% of the population. It is often overlooked as a potential cause of deep venous thrombosis and pulmonary embolism, particularly in younger patients.

Clinical Case: A 25-year-old Hispanic woman presented to the emergency department with leg swelling and pain. She later developed shortness of breath and chest pain, prompting her transfer to the intensive care unit. Chest and abdominal computed tomographic angiography confirmed a left pulmonary embolism, absence of the infrarenal vena cava, and a thrombus in the left superficial iliac and femoral veins. She was treated with thrombolytic therapy followed by a successful thrombectomy.

Discussion: IVCA is a rare vascular condition caused by either a developmental failure of the inferior vena cava or thrombosis with secondary resorption. This leads to a complex collateral venous system with slower blood flow, increasing the risk of stasis and thrombus formation. IVCA should be considered in young patients with recurrent thromboembolism who lack typical risk factors. Diagnosis is best achieved through computed tomography or magnetic resonance angiography. Management usually involves long-term anticoagulation, with surgery reserved for select cases.

Conclusions: IVCA is an uncommon cause of thromboembolism events. It is important to consider this diagnosis in young patients after immune, genetic, or traumatic issues have been ruled out, as prompt treatment can help prevent serious health risks.

INTRODUCTION

Inferior vena cava (IVC) abnormalities are rare conditions that affect up to 4% of the total population.[1] One of the unique characteristics noted for this vessel is agenesis, a vascular condition that affects about 0.0005% to 1% of the population. Interestingly, it accounts for up to 5% of deep venous thrombosis (DVT) cases in younger patients.[2] Two hypotheses explain this abnormality. The first involves an embryological issue between the 4th and 8th weeks of gestation, affecting the development of the IVC. The second suggests it results from intrauterine or perinatal thrombosis, causing the vessel's obstruction and resorption. This condition may occur alone or alongside other abnormalities, such as dextrocardia, heart defects, or spleen malformations.[1,3] The best way to diagnose this condition is through vascular evaluation using tomography or magnetic resonance studies, though these methods are not commonly applied for patients with DVT.[4]

We report the case of a female patient who experienced DVT and pulmonary embolism, ultimately leading to a diagnosis of infrarenal vena cava agenesis (IVCA).

CASE PRESENTATION

A 25-year-old Hispanic woman presented to the emergency department with severe, sharp pain in her left thigh, along with swelling that gradually affected her entire leg. Her medical history included schizophrenia, managed with 50 mg of clozapine daily. She had no history of surgery, fractures, immobilization, or previ-

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Table 1. Patient's Lab Results

Test	Result	Control
Chloride	102.5 mEq/L	98 – 106 mEq/L
Potassium	3.98 mEq/L	3.3 – 5.1 mEq/L
Sodium	139 mEq/L	136 – 145 mEq/L
Creatinine	0.77 mg/dL	0.51 – 0.95 mg/dL
Ureic nitrogen	23.6 mg/dL	6 – 20 mg/dL
Hemogram		
Leucocytes	10.77 x 10^3/uL	4.5 – 11.3 x10^3/uL
Neutrophils	8220 x 10^3/uL	2.25 – 8.48 x10^3/uL
Lymphocytes	1340 x 10^3/uL	0.9 – 4.52 x10^3/uL
Monocytes	990 x 10^3/uL	0 – 1.24 x10^3/uL
Hemoglobin	12 g/dL	12.3 – 15.3 g/dL
Hematocrit	36.9%	35% – 47%
Platelets	319.000 x mm^3	150 000 – 450 000 x mm^3
Reticulocytes	6.93%	0.5% x 2.0%
Blood smear test	Mormocytic and normochromic erythrocytes, platelets with normal shape and number, leukocytes with normal features, shape, and number	–
Fibrinogen	245 mg/dL	200 – 397 mg/dL
PTT	31.5 seconds	25.1 – 36.5 mg/dL
INR	1.08	–
Prothrombin time	12.4 seconds	9.1 – 12.5 seconds
D-dimer	47 046 ng/mL	0 – 499 mg/dL
Haptoglobin	287.62 mg/dL	30 – 200 mg/dL
Total bilirubin	0.32 mg/dL	0 – 1.0 mg/dL
Direct bilirubin	0.18 mg/dL	0 – 1.3 mg/dL
Lupus anticoagulant	Negative	–
β-2 glycoprotein IgG	0.7 UA/mL	Negative < 10 UA/L Positive > 20UA/L
β-2 Glycoprotein IgM	0.00 UA/mL	Negative < 10 UA/L Positive ≥ 10UA/L
Cardiolipin antibodies IgM	0.0 MPL U/mL	Negative < 10 MPL U/mL Positive > 10 MPL U/mL
Cardiolipin antibodies IgG	1.3 GLP U/mL	Negative < 10 GPL U/mL Positive > 20 GPL U/mL
Anti-La/SSB antibodies	0.00 UA/mL	Negative < 10 UA/L Positive ≥ 10UA/L
Anti RNP antibodies	0.00 UA/mL	Negative < 10 UA/L Positive ≥ 10UA/L
Anti-Ro/SSA antibodies	0.00 UA/mL	Negative < 10 UA/L Positive ≥ 10UA/L
Anti-Sm antibodies	0.00 UA/mL	Negative < 10 UA/L Positive ≥ 10UA/L
Anti-DNA antibodies	Negative	Negative < 1/10

Abbreviations: PTT, partial thromboplastin time; INR, international normalized ratio; RNP, ribonucleoprotein.

ous symptoms, including lower limb edema, chest pain, palpitations, or dyspnea.

On examination, she had tachycardia, but other vital signs were normal. Her heart and lungs sounded normal, and her abdomen showed no signs of vascular issues. The left leg was swollen from the groin down, with visible veins on the upper back of the thigh. Both Homans and Pratt's signs were positive.

With a clinical suspicion of DVT, the patient was admitted for further testing. (See blood test results in Table 1.) An arterial and venous duplex ultrasound revealed a thrombus in the femoropopliteal region, leading to the initiation of treatment with low molecular weight heparin. During her hospital stay, she experienced chest pain and shortness of breath, prompting transfer to the intensive care unit, where she was stabilized. A transthoracic echocardiogram showed a normal left ventricular ejection fraction (58%) and normal systolic and diastolic function, with a low probability of pulmonary hypertension. No intracavitary thrombus was detected.

A chest and abdominal angiogram revealed the absence of the infrarenal portion of the inferior vena cava with multiple collateral vessels; acute thrombosis in the superficial femoral, common iliac, and superficial iliac veins; and a flow defect in the proximal left renal vein (Figure 1 and 2A). It also showed a left pulmonary embolism without heart involvement (Figure 2B).

With that diagnosis, we performed thrombolysis using intravenous alteplase through a Fountain catheter (Squirt Fluid Dispensing System). After the procedure, we started a continuous dose of heparin for anticoagulant therapy. However, evaluation indicated minimal reperfusion following the thrombolysis, so we proceeded with thromboaspiration using an 8F catheter (Penumbra CAT8TORQ85 Indigo Catheter 8.0Fr. Torq Tip, 85 cm). Following that, we infused heparin and plasminogen intravenously, which resulted in complete reperfusion of the affected vessels. Postoperative recovery went smoothly, chest pain resolved, and, after checking for any autoimmune or coagulation issues, the patient was discharged with a plan for long-term rivaroxaban therapy and compression socks. Six months later, during a follow-up at the outpatient clinic, she showed no signs of new embolism episodes.

DISCUSSION

DVT occurs when a clot forms in a deep vein. VTE affects up to 1 million people annually in the United States, with higher rates in older adults and women. The mortality rate is 10.6% in the first 30 days and up to 23% annually.[5] Most patients under 30 typically are evaluated for acquired risk factors, such as cancer, surgery, immobility, estrogen use, pregnancy, and certain medical conditions – ie, obesity, venous insufficiency, rhumatologic diseases, macrovascular arterial disease, and antiphospholipid antibody syndrome – which explain 95% of cases.[6]

IVCA accounts for 6% of IVC malformations. Though most

Table 2. Publications Presenting IVCA as Cause of Pulmonary Embolism or Renal Vein Thrombosis

Authors	Sex/Age	Main features and outcome
Van Laethem et al[2]	F/30 yo	Patient with history of bariatric surgery presented to ED with edema of right lower extremity and NYHA Class II dyspnea. D-dimer was within normal values and chest CT scan showed bilateral pulmonary embolism and unusual dilation of azygos vein and paraortic varicose networks. Abdominal angiography CT showed IVCA with paralumbar varicose networks draining into azygos vein. This CT also showed thromboses in primitive iliac veins bilaterally and in right superficial and common femoral veins. Patient was managed with rivaroxaban considering risk of impaired absorption of vitamin K antagonists, along with elastic compresion. Patient had no recurrence during 2 years follow up.
Ramos-Aranda et al[10]	M/30 yo	Patient had history of DVT in left leg 5 weeks prior to hospital admission due to severe dyspnea. Chest CT scan showed bilateral pulmonary embolism. VDU showed DVT in left leg at the femoropopliteal segment. LMWH was started as treatment. Abdominal CT scan showed extensive thrombosis of the femoropopliteal segment, suprarenal IVCA and duplicated IVC, with azygous continuation of a left retro-aortic renal vein. After venogram confirming the thrombosis, UET was performed along with alteplase infusion. Balloon angioplasty was required due to residual thrombosis of the left iliac vein with full resolution. Patient was managed with rivaroxaban and had no recurrence within 6-month follow-up.
Skeik et al[13]	M/23 yo	Patient presented to ED with 6 days of right lower quadrant pain, initially mild ache that progressed to severe pain. History was unremarkable. Physical examination revealed tenderness to palpation in right flank and lower quadrant. Blood tests were significant only for slight leukocytosis (12.400 x mL). Abdominal CT revealed IVCA below the infrahepatic segment with large retroperitoneal collateral veins draining into azygos and hemiazygos systems and a thrombus in right renal vein. Management included LMWH and warfarin started later. Patient was discharged when proper INR. No follow-up information was available.

Abbreviations: ED, emergency department; NYHA, New York Heart Association Functional Classification; CT, computed tomography; IVC, inferior vena cava; IVCA, inferior vena cava agenesis; DVT, deep vein thrombosis; VDU, venous doppler ultrasound; LMWH:, low molecular weight heparin; UET, ultrasound enhanced thrombolysis; INR, international normalized ratio.

patients are asymptomatic due to collateral vein development, VTE is common due to inadequate venous drainage, leading to stasis and clot formation.[7]

IVCA often is discovered incidentally,[8] and typically, it is diagnosed due to complications such as embolism. Symptoms may include abdominal or back pain, caused by the collateral venous system compensating for the absent IVC. If the lower extremity drainage is compromised, DVT signs such as leg edema, discomfort, and skin discoloration may occur. Though rare, pulmonary embolism is the most concerning complication.[2,9,10]

IVCA should be suspected in young patients without typical DVT risk factors or with recurrent thrombosis.[6,11] Diagnosis is best made using abdominal contrast magnetic resonance imaging, though CT and venography are also effective.[4,9,12]

Due to the low incidence of this condition, there is no standard treatment. Thrombolytic therapy via catheter is effective, while thrombectomy is used for large clots or failed thrombolysis. Outpatient management often includes anticoagulation and compression therapy, with Xa inhibitors favored for their lower bleed-

Figure 1. Coronal Computed Tomographic Angiography Showing in Both Frames the Absence of the Infrarenal Inferior Vena Cava (red arrow)

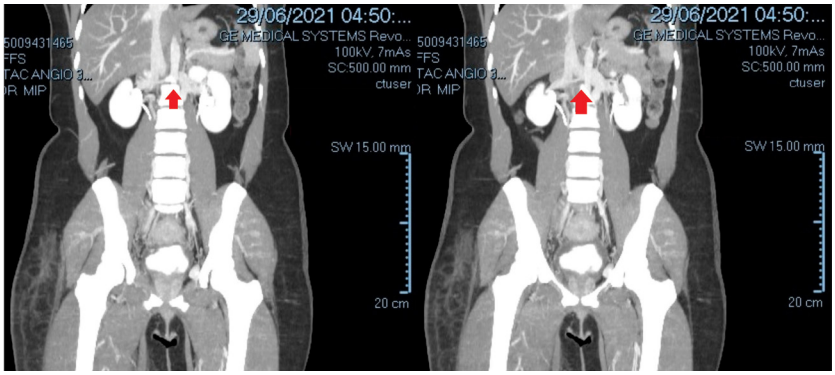
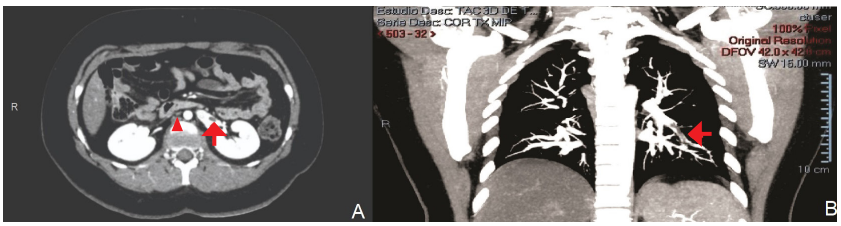


Figure 2. Computed Tomographic (CT) Angiography



A. Transversal CT angiography, showing absence of the infrarenal inferior vena cava (red arrowhead), and filling defect on the arrival of the gonadal vein to the left renal vein, suggesting thrombosis (red arrow).
B. Chest CT angiography, showing filling defects in segmental and lobar sections in the basal region of the left lung due to a pulmonary embolism (red arrow).

ing risk.[14] However, thrombosis recurrence is common within 2 years.[9,10,12,15] Surgical treatment is reserved for those who cannot take anticoagulants or experience a high rate of recurrence. Procedures may include IVC replacement or bypass, with minimal complications reported. Endovascular management with a WALLSTENT endoprosthesis (Boston Scientific) has shown success, with no recurrence after 15 months.[9,12,16]

CONCLUSIONS

IVCA is an uncommon condition that usually presents with DVT. In younger patients who have typical DVT risk factors or lack significant immune or genetic findings, this condition should be considered. It is important to tailor the management approach based on the patient's specific medical situation and comfort.

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