

Rare case of a kidney and inferior vena cava abnormalities with extensive lower extremity deep vein thrombosis in a young healthy male

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Abstract

Kidney and inferior vena cava (IVC) abnormalities with extensive deep vein thrombosis (DVT) is a very rare cause of DVT and has a diverse clinical presentation. Computed tomography (CT) angiography is the gold standard for diagnosis and treatment including thrombectomy, thrombolysis and systemic anticoagulation. We present a rare case of active young healthy male admitted with acute onset of right lower extremity pain and swelling who was found to have extensive DVT on doppler ultrasound. CT abdomen showed extensive clot burden involving right common femoral vein extending into internal and external iliac veins associated with IVC hypoplasia and hypoplastic left kidney. Patient underwent urgent thrombectomy, catheter directed thrombolysis and was discharged home in stable condition on oral anticoagulation.

Introduction

Kidney and inferior vena cava abnormalities with leg thrombosis (KILT) syndrome is most commonly seen as an incidentally finding on imaging studies especially in a young population. It is a rare cause of deep vein thrombosis with very few case reports in literature so far. We present a case of 27-year-old young healthy male admitted with right leg pain who was found to have extensive deep vein thrombosis (DVT) of right lower extremity extending into the common femoral vein, pelvic and abdominal veins with kidney and inferior vena cava abnormalities.

Case Report

A 27-year-old male with no significant past medical history was admitted with right lower extremity pain and swelling that started a week ago. Patient first noted swelling in the right ankle that was associated with pain in the calf and thigh. Pain was described as progressively worsening with tightness and heaviness in the affected leg that was exacerbated with ambulation. Patient endorsed recent short duration of immobility involving a 4 hour road trip but denied any recent surgery, prior history of thrombosis, weight changes, new bone pain, or history of easy bruising or bleeding. No known history of any congenital abnormalities, intra uterine growth restriction, oligohydramnios and no pregnancy related complications was reported.

Vitals on admission included blood pressure B.P 133/98 mm Hg, pulse rate of 100 beats per minutes, respiratory rate 18 per minute, temperature of 97.8 F, and normal oxygen saturation on room air. Physical examination was significant for right lower extremity swelling most prominent at the thigh with mild erythema and calf tenderness. Laboratory showed normal blood counts, liver and kidney functions.

Patient initially received full dose of Lovenox that was switched to low molecular weight heparin infusion. CT scan of chest, abdomen and pelvis was obtained to evaluate potential etiology of thrombosis and extent of DVT. CT scan ruled out pulmonary embolus and revealed an extensive DVT of the right common femoral vein, left common iliac vein, left internal and external iliac veins. There was an incidental finding of congenital inferior vena cava (IVC) anomaly, with no intrahepatic portion of inferior vena cava, and a hypoplastic left kidney (Figure 1A-C). Further evaluation of the right lower extremity with doppler ultrasound revealed acute thrombus occluding the common femoral, profunda femoral and popliteal veins (Figure 1D). Left lower extremity doppler study was negative for DVT. CT scan showed continuation of azygous vein into the superior vena cava via azygous arch which has been highlighted in detail in (Figure 2A-E). There was no significant venous collaterals found on CT scan after discussion with the radiologist.

Hypercoagulable work up was ordered. Factor V Leiden, prothrombin mutation and anticardiolipin antibody were negative. Testing for protein C, protein S, antithrombin III, lupus anticoagulant was deferred because of difficulty of interpretation of these results in the setting of an acute venous thromboembolism and systemic

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anticoagulation. Patient was found to have extensive thrombosis of the pelvic veins on angiogram. Based on CT scan and angiogram findings, our patients has congenital anomaly of inferior vena cava with no intrahepatic portion of the IVC, and hepatic veins drain into the azygous venous system which continues into the superior vena cava which can sometimes mimic with chronic inferior vena cava occlusion with venous thrombosis usually associated with significant venous collaterals. The patient was subsequently treated with direct thrombolysis, mechanical thrombectomy and balloon angioplasty (Figure 3A-C). Post thrombolysis catheter showed contrast flow from left iliac vein to right iliac vein and lumbar vein with no inferior vena cava seen (Figure 3D). Oncology recommended life-long anticoagulation because of unprovoked DVT, extent of thrombosis with multiple risk factors including IVC abnormalities. Patient was started on rivaroxaban on discharge and instructed to use elastic stockings, and to avoid excessive exercise, immobilization and smoking.

Discussion

KILT syndrome was first reported by

Van Veen *et al.*¹ It is a triad of kidney and IVC abnormalities and leg thrombosis. Only few case report studies have been reported in the literature. Inferior vena cava hypoplasia is an uncommon congenital cause of ilio-femoral DVT especially in young patients, accounting for 5% cases of DVT under 30 years old² and having a prevalence rate of % in the general population.³ IVC embryogenesis is very complex and involves confluence of three sets of the fetal veins, the posterior cardinal, sub cardinal, and supracardinal veins. There are four different parts to the IVC, which are hepatic, supra renal, renal and infra renal. The possible etiology of IVC atresia/hypoplasia is felt to be due to intrauterine or perinatal thrombosis of IVC, resulting in occlusion and disappearance or hypoplasia of IVC.⁴ The most common congenital anomalies are duplication of IVC, IVC continued with intra thoracic azygous vein, left sided IVC, IVC agenesis and retro aortic left renal vein. IVC abnormalities have a reported prevalence of 0.07-8.5% in the general population. IVC hypoplasia/agenesis usually results in the extensive collateral formation which results in the slow venous flow from the lower extremity ended up with the increased venous pressure, venous stasis and recurrent DVT.⁴

Agenesis of IVC has already been reported in the literature to be associated with extensive DVT.⁵ Duciu *et al.*⁶ first reported a case of KILT syndrome in siblings. DVT can be either provoked or unprovoked. Factors that are involved in provoked DVT include recent surgery, prolonged immobilization, trauma, pregnancy, obesity, smoking, recent travel, lupus anticoagulant, use of oral contraceptive pills and malignancy. If there are no obvious predisposing factors present, then DVT is most likely unprovoked which can be due to the absence of protein C, Protein S, antithrombin III, factor V/prothrombin gene mutation or congenital IVC anomalies. Most of the patients are asymptomatic prior to diagnosis and, if symptomatic, clinical presentation can be diverse including lower limb pain and swelling, loin pain, hematuria, back pain, and lower extremity ulcers.⁷ Pulmonary embolus is rarely seen with KILT syndrome, because the embolus is usually trapped in the azygous vein, which prevents propagation to the pulmonary circulation.

The typical presentation of KILT syndrome are in male patients presenting with extensive or bilateral DVT in the setting of physical exertion and associated with physical exertion. Shaline *et al.* reported a case of IVC atresia presenting with DVT associated in the setting of physical exertion.⁸ The

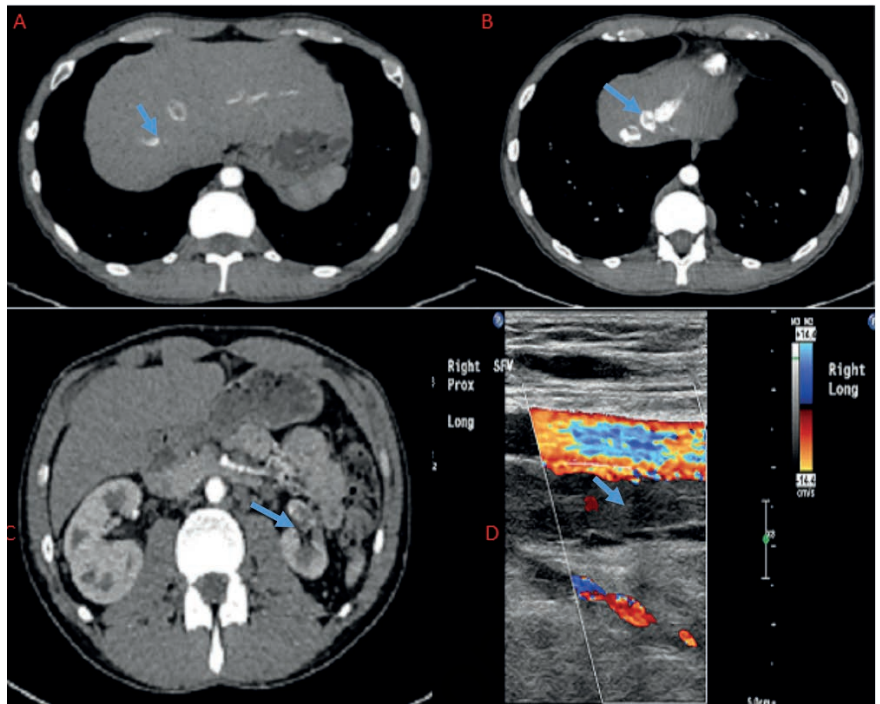


Figure 1. (A-C) Computed tomography scan of the abdomen showed absent intra-hepatic portion of IVC and confluence of hepatic veins, hypoplastic left kidney and absent both hepatic and renal portions of the inferior vena cava (arrows). (D) Doppler ultrasound clot in the deep veins of right leg (arrow).

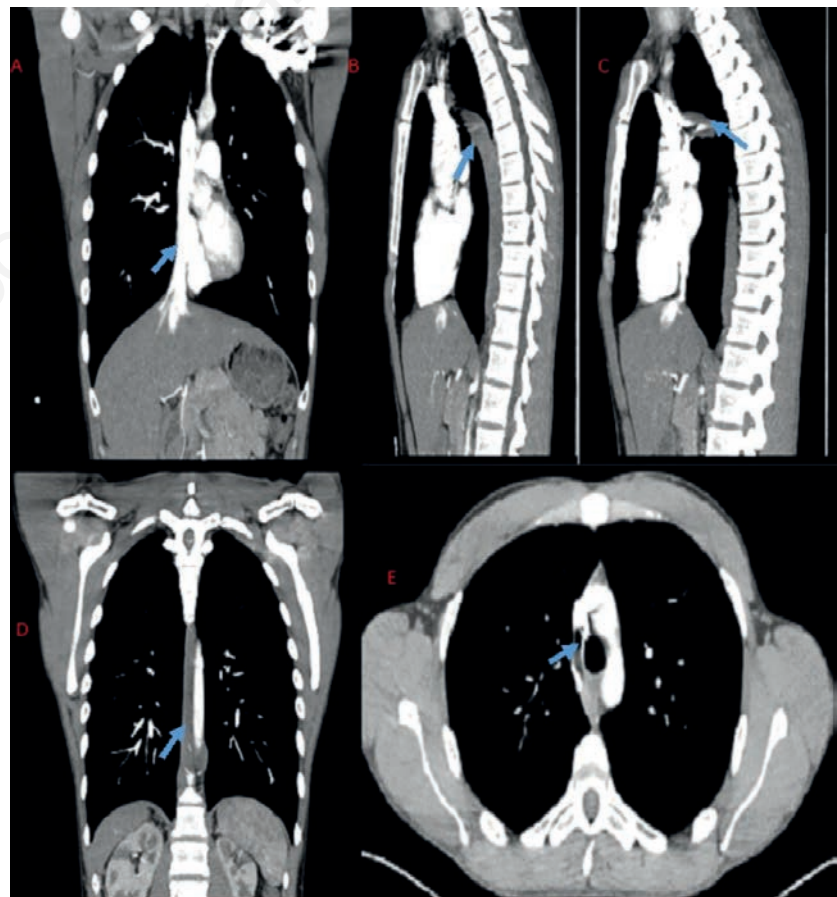


Figure 2. Computed tomography scan demonstrates azygous continuation from the hepatic veins into the superior vena cava as continuation of the hepatic veins (arrows).

DVT commonly involves distal IVC, common, internal and external iliac and femoral veins. Our patient was physically very active and CT scan showed DVT of the right common femoral vein through common iliac vein and the left common iliac vein extending into the left internal and external iliac veins. Studies have also reported association of agenesis of the infra renal IVC agenesis with renal anomalies. The most common abnormalities are renal agenesis, hypoplasia and aplasia. Renal anomalies usually involve the right kidney since, during embryogenesis, venous return from the right metanephros goes to IVC but the venous return from the left renal vein goes to gonadal vein.⁹ Although less common, cases of the left kidney involvement

have also been reported as seen in our patient.¹⁰

Usually, patients with unexplained or unprovoked DVT required hypercoagulable work up which could be the possible explanation of extensive DVT but sometimes, it can be associated with congenital IVC anomalies. Only few cases of DVT due to congenital IVC agenesis and associated heterozygous Factor V Leiden mutation have been reported in the literature.¹¹ Therefore, we did hypercoagulable work up to rule out any associated cause. Our patient's hypercoagulable work up included factor V Leiden, prothrombin mutation and anticardiolipin antibody which all were negative. Doppler ultrasound, CT venography or angiography and MRI angiogram are the

different diagnostic modalities that may be used to diagnose DVT. CT venography is the gold standard to diagnose DVT but it has been replaced by regular ultrasound due to invasive in nature. Further evaluation with these modalities is recommended in the setting of unprovoked DVT to evaluate for IVC agenesis and/or KILT syndrome.

Since this is a rare entity, there are no standard treatment guidelines available. Anticoagulation therapy and compression stockings are likely to relieve symptoms in patients with congenital IVC anomalies and symptomatic IVC related thrombosis. Studies have shown good results with combination of thrombectomy/thrombolysis and anticoagulation.^{12,13} However, the length of further duration of anticoagulation varies with the case as per American College of Chest Physician guidelines,¹⁴ if there is secondary cause of thrombosis without thrombophilia, then oral anticoagulation should be continued for 3 months. If there is co-existence or causative thrombophilia, then cause is non-reversible like IVC anomaly that needs lifetime anticoagulation to prevent recurrent thrombosis.¹⁵ Catheter directed thrombolysis with tissue plasminogen activator is an effective approach which lyses the clot and results in immediate resolution of symptoms. Additionally, this method prevents inactivation of plasminogen activators and its usual complications.^{13,16} Commonly reported complication of catheter directed thrombolysis are major bleeding, minor bleeding, fever, hematoma, pain, pulmonary embolism and death.^{17,18}

Our patient underwent thrombectomy, catheter directed thrombolysis and systemic anticoagulation. Long term anticoagulation, use of elastic stockings and avoidance of oral contraceptive pills, excessive exercise, prolong immobilization and smoking should be recommended in such patients. There is a high risk of DVT recurrence in such patients because of poor venous return and venous stasis owing to hypoplastic IVC.

Conclusions

This case highlights the importance of a work up for a congenital venous anomaly with or without kidney hypoplasia in a young healthy male, who presented with an extensive unprovoked lower extremity DVT in the setting of negative hypercoagulable work up. If any congenital IVC anomaly is found then catheter directed thrombolysis, thrombectomy, and systemic anticoagulation represent effective treatment.

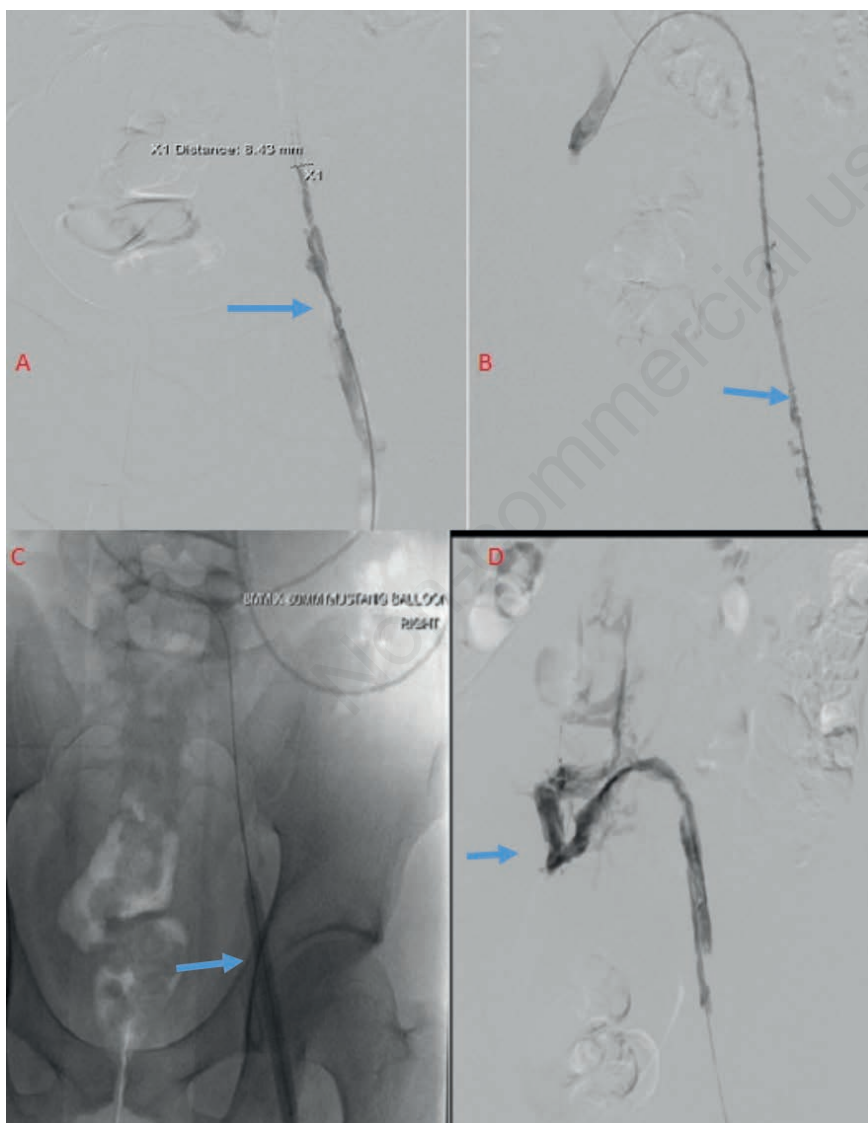


Figure 3. (A-B) Extensive thrombosis of the lower extremity (arrow); (C-D) Fluoro images showing balloon angioplasty and catheter course from left iliac vein to right iliac vein with no inferior vena cava (arrows).

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