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Case Report

Agenesis of Inferior Vena Cava Associated with Acute Bilateral Common Iliac Vein Thrombosis and Hypoplastic Right Kidney (KILT Syndrome) in a 54-Year-Old Female

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Abstract

Background and Objective: **Agenesis of the inferior vena cava (AIVC) is a rare anomaly (<1%) with known association with renal anomalies, and an uncommon cause of deep venous thrombosis (DVT). Previous reported cases were young (mostly under 30 years), and predominantly male (82%), but the present case was a 54-year-old female with a thrombophilia sibling, presenting with acute onset DVT.**

Material and Method: **A 54-year-old female presented with right lower quadrant pain and swelling of both thighs, right more than left, for 12 consecutive days. There was no history of smoking, alcohol drinking, oral contraceptive use, or other underlying diseases. The patient had one brother with thrombophilia. A CT scan of the abdomen demonstrated absence of the entire inferior vena cava (IVC), and bilateral common iliac vein thrombosis with hypoplastic right kidney (KILT syndrome). Assessment for thrombophilia found presence of lupus anticoagulant antibody. The patient was treated with low molecular weight heparin (LMWH), and planned lifelong oral anticoagulant.**

Conclusion: **AIVC can be found very rarely in female patients over the age of 40 years, with positive family history of thrombophilia. Anticoagulants is the recommended treatment.**

Keywords: **Agenesis of inferior vena cava, deep vein thrombosis**

Introduction

The most common congenital abnormality of the inferior vena cava (IVC) is duplication and retro-aortic left renal vein[1]. Agenesis of inferior vena cava (AIVC) is rare, recognized to be associated with deep vein thrombosis (DVT), and is present in 0.3% to 0.5% of young healthy adults[2,3]. It may not be recognized until later in life, usually as an incidental finding. AIVC has a known association with renal anomalies mainly confined to the right kidney. Previous cases of AIVC reported in the literature are usually younger than 30 years, and are predominantly male (82%)[4]. The present report described a very rare case of a 54-year-old female patient with AIVC.

Case Report

A 54-year-old female presented with right lower quadrant pain, bilateral upper thigh swelling, on the right more than left, with heaviness of both limbs for 12

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consecutive days. She also complained of thigh tingling, but no discoloration of skin of both lower limbs. She denied a history trauma, or previous illnesses or medications. Her brother (a 52-year-old male) was being treated for ahypercoagulable state for more than 10 years, and had a mid-left foot amputation 2 years ago from progressive iliofemoral venous thrombosis and arterial insufficiency. On examination of the patient, no erythema, warmth, varicose vein or ulcers was present. She was admitted to the hospital for evaluation of the cause of right lower quadrant pain and right thigh swelling.

The CT scan of the abdomen and pelvis showed extremely small-caliber inferior vena cava, with markedly prominent bilateral ascending lumbar veins, suggestive of AIVC. There were enlarged bilateral common iliac and external iliac veins, with concentric intraluminal filling defects, suggestive of acute venous thrombosis, which was more severe on the right side. A small-sized right kidney (6.2 cm), with a small but patent right renal artery, was seen, also with renal parenchymal enhancement and excretory function (Figures 1 to 4). Her chest X-ray was normal.

An investigation for thrombophilia was performed, including tests for the presence of antithrombinlll, protein C, protein S and lupus anticoagulant antibody. The result was positive for lupus anticoagulant antibody. The patient received systemic administration of low molecular weight heparin (LMWH), enoxaparin 40 mg subcutaneously

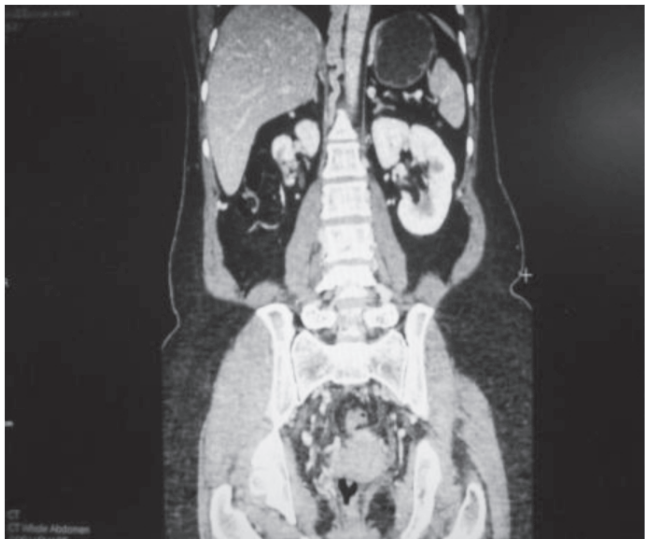

Figure 1 Small size of right kidney

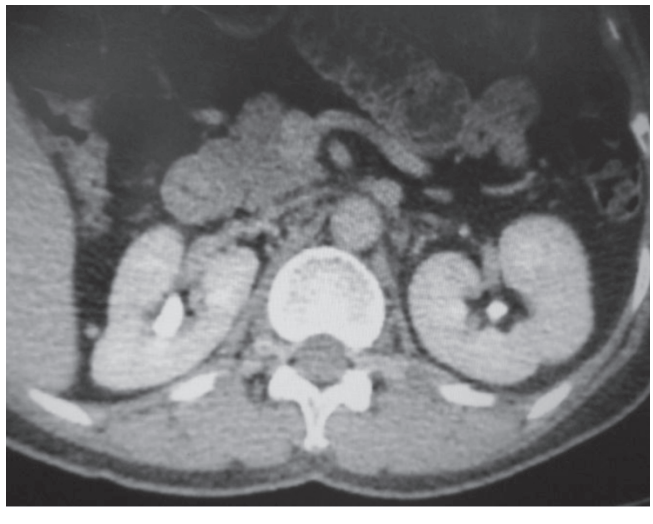

Figure 3 Agenesis of inferior vena cava

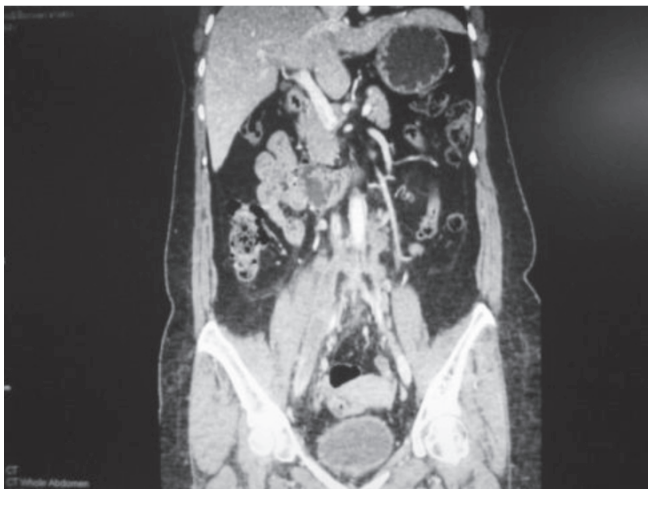

Figure 2 Thrombosis of bilateral common iliac veins

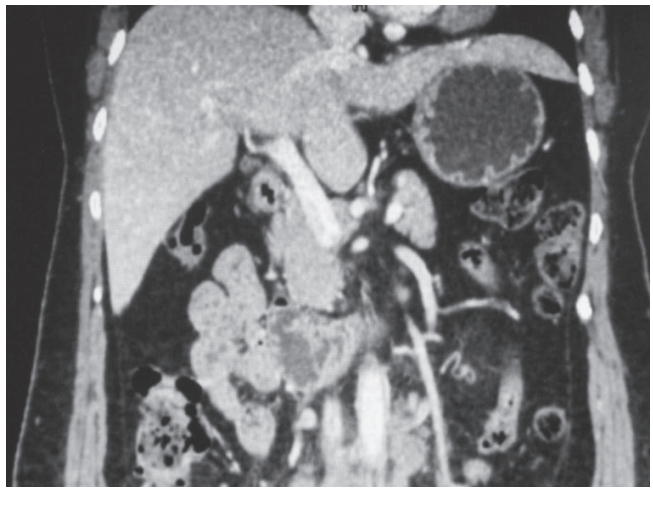

Figure 4 Hepatic veins drain directly to right atrium

twice daily, followed by an oral anticoagulant, warfarin sodium. Graduated stocking with ankle pressure 20 to 30 mmHg (thigh high) was also applied. Symptoms of right lower quadrant pain and heaviness of both legs gradually improved two days after start of treatment. LMWH was continued for 7 days, and warfarin was used to maintain a therapeutic international normalized ratio (INR) between 2 to 3, for life.

Discussion

The prevalence of dysgenesis of inferior vena cava in the general population is approximately 1 %[5]. The overall prevalence of AIVC is very low (0.6%) and can be found in 5% of patients aged under 30 years with deep vein thrombosis[6]. AIVC is often referred to in three different forms, i.e.,

1. Absence of the suprarenal inferior vena cava, where hepatic venous flow drains directly into the right atrium, and the blood from the infrarenal inferior vena cava returns to the heart through the azygos and hemiazygos veins[7,8,9].
2. Absence of the infrarenal inferior vena cava with preservation of the suprarenal segment;
3. Absence of the entire inferior vena cava.

The etiology of these developmental failures is unclear and remains controversial according to the current literature. Inferior vena cava abnormalities have been identified as a possible congenital risk factor for deep vein thrombosis[10,11,12,13]. Garcia -Fuster et al. in their prospective study of 116 patients under the age of 50 years reported that inferior vena cava anomalies were present in 16.2% of patients with iliac vein thrombosis, and vena cava malformations was considered a risk factor in 5.1% of the population studied[1]. Sagban et al. found anomalies of right kidney in 6% of patients with inferior vena cava agenesis, while hypoplastic left kidney was found in 2.7%[14].

In patients with suspected vena cava anomalies, abdominal ultrasound with venous duplex scan of leg should be performed. A contrast-enhanced CT scan or MRI of the abdomen should be done following abdominal sonography[15]. The ultrasound can identify collateral venous circulation, but cannot specify the exact type of inferior vena cava anomaly. Lambert et al. reported 10 cases of deep vein thrombosis associated with inferior vena cava agenesis and reviewed 62 published cases until 2010[15], and states that the ultrasound could not establish the diagnosis of AIVC. Venography is reserved for venous mapping for post-interventional reevaluation[16]. Hypercoagulability studies should also be done.

In 10% of patients with agenesis of inferior vena cava, other organ malformations can coexist, such as renal and pulmonary atrophy, heart valve defects, or asplenia syndromes[17]. The combination of deep vein thrombosis with inferior vena cava agenesis and kidney abnormalities is known as the "KILT syndrome" (kidney anomaly, inferior vena cava anomaly, and leg thrombosis)[18]. In the present case study, the diagnosis was established via abdominal CT, in which the absence of entire inferior vena cava, bilateral iliac vein thrombosis, and hypoplastic right kidney were found. In addition, thrombophilia studies found positive lupus anticoagulant antibody.

Conclusions

AIVC associated with deep vein thrombosis, a rare condition, is usually seen in younger patients, in contrast to patients with classical deep vein thrombosis, and can be difficult to diagnosis due to lack of specific symptom and signs. The present report details an even more rare case of an older, 54-year-old female patient with AIVC, and provides information on the diagnosis and treatment of such a condition.

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บทคัดย่อ **ความผิดปกติแต่กำเนิดชนิดบกพร่องในการสร้างหลอดเลือดดำ inferior vena cava ร่วมกับการเกิดลิ่มเลือดในหลอดเลือดดำส่วนที่อยู่เหนือต่อขาหนีบทั้ง 2 ข้าง (Bilateral Common Iliac Veins Thrombosis) และมีขนาดไตข้างขวาเล็กกว่าปกติ ในผู้หญิงอายุ 54 ปี เรียกกลุ่มอาการนี้ว่า KILT syndrome**

นายแพทย์อนุวัชช์ จันทร์ทิพย์
กลุ่มงานศัลยกรรม โรงพยาบาลลำปาง

วัตถุประสงค์ : ความผิดปกติแต่กำเนิดชนิดบกพร่องในการสร้างหลอดเลือดดำ inferior vena cava (AIVC) เป็นความผิดปกติที่พบได้ค่อนข้างน้อย ประมาณ 0.0005%-1% และมีความผิดปกติร่วมกับไต โดยตรวจพบการมีลิ่มเลือดในหลอดเลือดดำของขา (Deep Vein Thrombosis, DVT) ได้ไม่บ่อย จากรายงานในวารสารทางการแพทย์ที่ผ่านมามักพบในผู้ป่วยอายุน้อย โดยส่วนใหญ่น้อยกว่า 30 ปี และมักพบในผู้ป่วยเพศชายประมาณ 81.9% ในรายงานนี้ตรวจพบกลุ่มอาการนี้ในผู้ป่วยหญิงอายุ 54 ปี ซึ่งมาด้วยอาการลิ่มเลือดในหลอดเลือดดำของขาร่วมกับตรวจพบประวัติการแข็งตัวของเลือดมากกว่าปกติ (thrombophilia) ของน้องชาย

วิธีการ: ผู้ป่วยหญิงอายุ 54 ปี มีอาการปวดท้องด้านล่างขวา ร่วมกับมีการบวมบริเวณต้นขาทั้ง 2 ข้าง, ข้างขวาบวมมากกว่าข้างซ้าย, เป็นมา 12 วันก่อนมาโรงพยาบาล ผู้ป่วยไม่มีประวัติสูบบุหรี่, ดื่มสุรา, การใช้ยาคุมกำเนิด หรือประวัติโรคประจำตัวอื่น ๆ แต่มีประวัติคนในครอบครัวเป็นโรคที่มีการแข็งตัวของเลือดมากกว่าปกติ (thrombophilia) การตรวจภาพถ่ายรังสีคอมพิวเตอร์พบว่าผู้ป่วยไม่มีหลอดเลือดดำ inferior vena cava ร่วมกับการมีลิ่มเลือดในหลอดเลือดดำเหนือต่อระดับขาหนีบขาทั้ง 2 ข้าง และมีไตข้างขวาขนาดเล็กกว่าปกติ การตรวจเลือดหาภาวะผิดปกติเกี่ยวกับการแข็งตัวของเลือดพบมีผลเป็นบวกของการทดสอบ lupus anticoagulant ซึ่งได้ทำการรักษาผู้ป่วยรายนี้โดยใช้ยา low molegular weight heparin (LMWH) และใช้ยาต้านการแข็งตัวของเลือด (warfarin) หลังจากนั้นเพื่อลดการกลับเป็นซ้ำ

สรุปผลการศึกษา: ความผิดปกติแต่กำเนิดชนิดบกพร่องในการสร้างหลอดเลือดดำ inferior vena cava ตรวจพบได้น้อยมากในผู้ป่วยหญิงที่มีอายุมากกว่า 40 ปี ที่มีพี่น้องเป็นโรคที่มีการแข็งตัวของเลือดมากกว่าปกติ การรักษาโรคในผู้ป่วยกลุ่มอาการนี้สามารถใช้ยา LMWH อย่างได้ผล