A rare cause of venous thrombosis: Congenital absence (agenesis) of the inferior vena cava

Kuldeep Singh MD FACS¹, James Poliquin MD², Guy Syversten DO³, Douglas O Kohler MD⁴

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Deep vein thrombosis is a frequent finding in otherwise healthy, young adults who are diagnosed with congenital absence (agenesis) of the inferior vena cava – a rare anomaly. This condition is best diagnosed

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Key Words: *Agenesis; Anomaly; Fibrosed*

Deep vein thrombosis (DVT) is a frequent finding in otherwise healthy, young adults who are diagnosed with congenital absence (agenesis) of the inferior vena cava (AIVC) – a rare anomaly (1-3). This condition is best diagnosed by computed tomography (CT) angiography with venous phase imaging, and is managed using anticoagulation, percutaneous catheter-directed mechanical thrombectomy and thrombolysis. To illustrate this phenomenon, a case is presented.

Congenital anomalies of the inferior vena cava (IVC) are present in 0.3% to 0.5% of young, healthy adults (4,5). The most common congenital abnormalities of the IVC are duplication and retroaortic left renal vein. AIVC has an incidence of 0.0005% to 1% in the general population (6). During weeks 6 to 10 of gestation, anomalies of the IVC may occur. DVT associated with AIVC may be present in more than 5% of the cases (7).

Ruggeri et al (7) suggested and Chee et al (8) confirmed that patients with IVC malformations have a significant risk of developing DVT. Reduced venous flow, venous hypertension and thrombophilia may play important roles in the development of DVT in these cases.

Causes of DVT are multifactorial. Congenital risk factors are extremely rare, but AIVC is emerging as a significant risk factor that may contribute to DVT in healthy, young adults.

DVT associated with AIVC is best diagnosed with colour venous Doppler ultrasound and CT angiography or magnetic resonance imaging. This condition is successfully managed with anticoagulation, catheter-directed thrombectomy and thrombolysis. We present a case of DVT with underlying congenital AIVC.

CASE PRESENTATION

A 28-year-old Caucasian man presented to the emergency department of Mary Mount Hospital (Ohio, USA) with sudden but significant swelling of his left lower extremity of less than 24 h duration. It was associated with mild discomfort, tingling and dark discolouration of the left leg. He denied trauma or other risk factors of DVT.

His father had hypertension and diabetes. A physical examination was essentially unremarkable, except for the marked swelling and purplish discolouration of the entire left lower extremity, extending from the foot to the groin. No neurovascular deficit was present. No varicose veins or ulcers were noted on the left leg. There was mild tenderness on palpation of the left calf and medial thigh area. Moderate left ankle edema was also present.

A colour venous duplex scan was negative for DVT but suggested a diminished flow rate of the left iliac vein, indicating compression (May-Thurner syndrome, in which the left iliac vein is compressed by the right iliac artery) (9). A contrast-enhanced CT scan of the abdomen and pelvis revealed a fibrosed IVC (Figure 1), dilated azygos and hemiazygos veins (Figure 2), and moderate distension of the left ureteropelvic junction. Both kidneys were of normal shape, size and density. There was no evidence of DVT.

The patient was admitted to the hospital for vascular and urology consultations, and received anticoagulation therapy with low molecular weight heparin due to a strong clinical suspicion of DVT. A vascular surgeon requested a repeat venous Doppler ultrasound the next morning which, surprisingly, demonstrated new thrombi in the left distal external iliac vein, left common femoral vein, proximal superficial femoral vein and the ascending lumbar vein (Figure 3), along with sluggish flow in the distal left femoral vein. CT angiography with venous phase imaging confirmed all of the above findings consistent with DVT as documented on venous ultrasound. It also revealed an almost totally occluded fibrosed IVC, except for a short segment involving the hepatic segment suprarenally. There was also enlargement of both the common and external iliac veins, which drain into innumerable perirenal, paralumbar and retroperitoneal veins. Both renal veins, with collaterals, were occluded.

A urologist advised no further management of the left ureteropelvic junction dilation, which was caused by peripelvic cysts. Blood studies revealed leukocytosis and a positive thrombophilia screen (high D-dimer, antithrombin assay, factor VIII and fibrinogen levels).

¹Department of Emergency Medicine, Mary Mount Hospital; ²Department of Vascular Surgery, Cleveland Clinic Hospital; ³Department of Radiology; ⁴Department of Medicine, Mary Mount Hospital, Garfield Heights, Ohio, USA

Correspondence: Dr Kuldeep Singh, 4804 Pearl Road, Cleveland, Ohio 44109, USA. E-mail drsinghmd@yahoo.com

Figure 1) Fibrosed inferior vena cava (IVC)

The patient was started on low molecular weight heparin on admission merely on suspicion of DVT, which was confirmed on a second venous duplex scan on day 2 of his admission, followed by mechanical thrombectomy and catheter-directed thrombolysis of all venous thrombotic areas over the next three consecutive days. The results were checked by venography, which confirmed that all the thrombotic veins were patent. Anticoagulation therapy with heparin was continued, and warfarin was added before discharge. He is presently doing very well and has returned to work.

DISCUSSION

Congenital AIVC is an uncommon anomaly, although it has been reported in the literature (1-3). AIVC is of paramount interest because it is often associated with DVT, especially in young, healthy individuals without other DVT risk factors. IVC anatomical abnormalities are developmental disorders that can occur at six to 10 weeks of gestation. The infrahepatic IVC develops from three pairs of embryonic veins; namely, the post-cardinal, subcardinal and supracardinal veins. AIVC is probably not a true embryonic anomaly, but rather a chronic perinatal thrombosis of the IVC resulting in chronic occlusion with a fibrosed cord-like structure, similar to what we have described in the present case. IVC anomalies are commonly associated with DVT and renal anomalies, as discussed by Gayer et al (10). DVT occurs in one of 1000 cases, and is associated with risk factors.

Ruggeri et al (7) presented four cases of AIVC over five years in a young population; our case was consistent with this population. Chee et al (8) also described similar findings in a case series. The prevalence of AIVC ranges from 0.3% to 0.5% in normal, healthy individuals, and is 5.3% in young adults with DVT-associated risk factors (7).

Several imaging modalities are currently available to identify AIVC anomalies and DVT. CT angiography with venous phase imaging or magnetic resonance venography is the best option. Venography is reserved for venous mapping before mechanical thrombectomy and thrombolysis, or to check the efficacy of these procedures as described in the present case.

Few hypotheses regarding how AIVC contributes to the formation of DVT have been discussed in the literature; chronic venous stasis, impaired venous return, and venous hypertension due to absent main venous pathways and associated thrombophilia may be contributing factors. Gayer et al (10)

Figure 2) Dilated azygos and hemiazygos veins

Figure 3) Thrombosis of the ascending lumbar vein

recommended that all IVC anomalies be screened for venous thrombosis carefully; the present case illustrates this point very clearly.

Thrombosis of the ascending lumbar vein (Figure 3) in this patient was an important contributing factor in the formation of DVT. This was the main collateral vein in the present case, and occlusion of this critical vein led to significant DVT.

It is also postulated that multiple emboli from DVT in the common and superficial femoral vein may migrate through the hemiazygos/azygos systems to the pulmonary circulation, resulting in fatal embolization.

Catheter-directed mechanical thrombectomy and thrombolysis have been described in the literature (11). We used these techniques in the present case, with excellent results. All patients with suspected or proven DVT due to AIVC should be put on anticoagulation therapy immediately and should be on lifelong warfarin therapy to reduce the risk of recurrent DVT. Associated renal anomalies should be managed accordingly. Surgical intervention at present has a very limited role in ameliorating these IVC anomalies.

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