CASE REPORT

Congenital atresia of the inferior vena cava and antithrombin III deficiency in a young adult: compounding risk factors for deep vein thrombosis

Laura Muscianese, Ronald R Seese, William Graham, James H Williams

University of California, Irvine,
Orange, California, USA

Correspondence to
Laura Muscianese,
lmuscian@uci.edu

Accepted 9 January 2015

SUMMARY

Atresia, or absence, of the inferior vena cava (AIVC) is a rare, usually fortuitous finding on advanced imaging that predisposes patients to deep venous thrombosis (DVT). In young adults who lack predisposing risk factors but nonetheless develop extensive or bilateral DVTs, AIVC should be considered. We describe a case of a previously healthy 17-year-old male patient who developed an extensive renal vein thrombus due to the absence of the superior portion of his IVC compounded with previously undiagnosed antithrombin III deficiency. We discuss the diagnosis and management of this complicated condition.

BACKGROUND

Atresia, or absence, of the inferior vena cava (AIVC) is an uncommon anomaly associated with complete or partial IVC absence. Compared with normal IVC anatomy, AIVC increases risk of deep venous thrombosis (DVT) 10-fold by diminishing cardiac return and promoting abdominal and lower extremity venous stasis.¹ When a young (<30 years) and otherwise healthy adult presents with an extensive or bilateral DVT, it is imperative to consider AIVC as a potential contributing cause and to employ advanced imaging for further investigation.² We describe a patient presenting with unilateral flank pain who, after further workup, was found to have AIVC and antithrombin III (ATIII) deficiency, a rare thrombogenic combination,¹ with extensive thrombosis of the bilateral renal veins, infrarenal portions of the IVC, and bilateral iliac veins.

CASE PRESENTATION

A 17-year-old male patient was referred to our emergency room with a 2-week history of right flank pain. The patient denied symptoms of genitourinary, renal or coagulation abnormalities. He had exercised vigorously the day before pain began but denied trauma. His medical history was unremarkable; pertinent family history included exercise-induced haematuria in a sibling. Vital signs and physical examination were within normal limits.

INVESTIGATIONS

Laboratory results revealed marked proteinuria (30 mg) on urinalysis, elevated 24 h urine proteins (496 mg/24 h), normal serum creatinine (1.1 mg/dL) and low blood albumin protein (2.2 mg). A hypercoagulable panel demonstrated decreased

ATIII enzyme activity (66% of expected). Considering this patient's age in the presence of a possible ATIII deficiency-induced renal vein thrombosis, we chose to employ abdominal and pelvic contrast-enhanced CT. Interestingly, these studies revealed a shortened and narrowed suprarenal IVC with collateralisation from dilated hemiazygos and azygos veins (figures 1 and 2). Right renal vein thrombosis was noted to extend past the infrarenal IVC into iliac veins bilaterally. Initially, lower extremity duplex ultrasound found no evidence of distal venous thrombosis. Abdominal and pelvic MRI and MR angiography confirmed the CT findings without evidence of arteriovenous malformation. Additionally, while AIVC is associated with congenital cardiac anomalies, an echocardiogram with bubble study in this patient was normal.³

TREATMENT

Despite 24 h of anticoagulation, the patient developed extension of his pre-existing thrombus into the right lower extremity with near occlusion noted in the great saphenous vein, common femoral vein, deep femoral vein, femoral vein, popliteal vein and tibioperoneal trunk. We proceeded with pharmacomechanical catheter-directed thrombolysis (PCDT) and balloon angioplasty to re-establish venous drainage through the IVC, bilateral iliac veins, femoral veins, and corresponding collateral vessels. During this procedure, guide wires were inserted into the popliteal veins using micropuncture technique and advanced under ultrasound guidance into the IVC. Both infusion catheters were then positioned with the cranial tip at the level of the juxtarenal IVC and a 10 mg loading dose of tissue plasminogen activator (TPA) was pulse sprayed into the catheters. Subsequently, TPA infusion was initiated at 1.2 mg/h through each infusion catheter. The patient tolerated this procedure with minimal blood loss and was transferred to the surgical intensive care unit (SICU) for monitoring overnight. The next day during reassessment, there was a slight decrease in thrombus burden noted within the IVC, bilateral iliac veins, right common femoral vein and right femoral vein following overnight thrombolysis. At this point mechanical thrombectomy was performed in the IVC, right iliac vein, right common femoral vein and right femoral vein with improved flow noted. The infusion catheters were then repositioned with the cranial tip of the right-sided catheter placed in the right common femoral vein and the cranial tip of the left-sided

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To cite: Muscianese L, Seese RR, Graham W, et al. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2014-205729

Figure 1 Radiographic findings of atresia of inferior vena cava (IVC). (A) Contrast-enhanced CT demonstrating extensive thrombosis and tortuosity of the right renal vein (arrowhead); (B) absence of the suprarenal IVC with thrombus filling the right renal vein (arrow); (C) dilated right renal venous collaterals (arrowhead) and extensive bilateral thrombus of the renal veins (arrows); (D and E), gadolinium-enhanced angio-MRI demonstrating tortuous renal collateral vasculature (arrowhead) with thrombus filling bilateral renal veins (arrows) and extensive thrombus in the narrowed portion of the IVC (arrowhead) continuing into bilateral iliac veins (dashed arrow). Dilated azygos vein providing collateral flow is also seen (arrow).

infusion catheter placed at the juxtarenal IVC. Thrombolysis with TPA was continued at a rate of 1.2 mg/h and the patient was again monitored overnight in the SICU. Once more, the patient returned for reassessment and at this point venography showed moderate improvement of the thrombus in the IVC, bilateral iliac veins, right common femoral vein and right femoral vein. Balloon angioplasty of a long segment of the right femoral vein was performed, re-establishing patency of the

vessel. The persistent thrombus remaining in the IVC and bilateral iliac veins suggested that the thrombus in those areas was chronic (figure 3). Throughout this process, the patient was anticoagulated with heparin, which was titrated to achieve a partial thromboplastin time of 50–80 s. Additionally, intermittent pneumatic compression devices and class II graduated compression stockings were maintained on the leg and thigh throughout hospitalisation to maximise venous flow.

Following these procedures, the patient was optimised on conservative management with ATIII concentrate replacement and continuation of heparin for anticoagulation. Despite residual thrombus burden, the patient demonstrated symptomatic improvement. Subsequently, he was transitioned to warfarin and discharged to home.

OUTCOME AND FOLLOW-UP

The patient has been followed as an outpatient for the past 1.5 years on an ATIII concentrate and warfarin anticoagulation regimen. It was recommended that the patient continue to utilise compression stockings for at least 6 months as an outpatient; however, he was persistently non-compliant. The initial follow-up appointment was 1 month following discharge, at which point the patient's symptoms had resolved. Prior to this appointment he underwent a duplex ultrasound of his abdomen, pelvis and bilateral lower extremities, which demonstrated improvement of the residual thrombus located in the IVC and bilateral iliac vessels. Additionally, there was no recurrence noted in the patent right femoral vein. Over the past 1.5 years, the patient has not experienced symptoms concerning for new DVTs or progression of his residual thrombus burden. He has been compliant with anticoagulation as well as ATIII concentrate. The patient's ATIII levels are checked semi-annually to assure proper

Figure 2 Diagram demonstrating the atypical venous anatomy in our patient. 1, inferior vena cava (IVC); 2, narrowed hepatic IVC with an absent suprahepatic IVC; 3, dilated and tortuous right renal vein filled with thrombus; 4, dilated renal collateral veins; 5, dilated and tortuous left renal vein partially thrombosed; 6, dilated azygos vein; 7, aorta; and 8, dilated hemiazygos vein.

Figure 3 Prone-positioned venograms following pharmacomechanical catheter-directed thrombolysis. (A) Cranial ends of catheters placed at the juxtarenal inferior vena cava (IVC) with contrast illustrating residual thrombus in the IVC and bilateral iliac vessels. (B) Right femoral vein with contrast demonstrating vessel patency following pharmacomechanical catheter-directed thrombolysis and balloon angioplasty.

ATIII concentrate replacement. Furthermore, he is now screened for progression of the residual thrombus annually with duplex ultrasounds of his abdomen, pelvis and lower extremities.

DISCUSSION

AIVC affects 0.4% of the general population and 5% of patients with DVT.¹ Despite risks of increased stasis, many young adults with AIVC remain asymptomatic until their third decade or until another risk factor (ie, estrogen-containing contraceptives, pregnancy, cancer, smoking, high-impact exercise, hypercoagulability, immobilisation or ageing) is encountered. Our patient exhibited two risk factors for thrombosis: ATIII deficiency and AIVC. In the context of prior vigorous exercise, these factors may have led to layering of new thrombus, thereby yielding the patient's acute presentation. Thus, in young adult patients with non-specific pain in the lower back or abdomen, it is important to consider thrombosis secondary to AIVC as a potential perpetrator, after common diagnoses have been ruled out.

Most patients with an AIVC can be treated successfully with long-term anticoagulation while avoiding or correcting risk factors that increase thrombogenic potential. Life-long

anticoagulation is typically required, as recurrent thrombosis occurs with discontinued therapy. Additionally, aggressive PCDT has been shown to significantly decrease thrombus burden in patients with AIVC with acute DVTs.³ In patients with ATIII deficiency and AIVC who present with acute DVT the treatment of choice involves the correction of modifiable thrombogenic risk factors, compression stocking placement to increase venous return, long-term anticoagulation, and the implementation of PCDT to restore vessel patency.

IVC anomalies represent rare but relevant conditions that in the presence of other thrombogenic risk factors, can prematurely precipitate DVTs. In young adults (<30 years) with extensive or bilateral DVTs, AIVC must be considered as a possible yet uncommon diagnosis.

Learning points

- ▶ Consider absent inferior vena cava (IVC) as a potential cause of deep vein thrombosis in young adults (<30 years) with extensive or bilateral thrombosis.
- ▶ Absent IVC acts synergistically with other clotting risk factors leading to premature presentation of deep vein thrombosis.
- ▶ If a hypercoagulable state is elucidated during workup, there should be a heightened awareness for IVC anomalies, as 30% of these anomalies are associated with hypercoagulable states.⁴
- ▶ The treatment of choice for symptomatic antithrombin III deficiency in the presence of atresia of the IVC includes correction of modifiable thrombogenic risk factors, compression stocking placement, long-term anticoagulation, and the implementation of pharmacomechanical catheter-directed thrombolysis.

Funding National Institutes of Health (grant numbers T32-GM0862 and FMH095432A (R.R.S)).

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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